Reproductive pasts, reproductive futures : genetic counseling and its effectiveness / James R. Sorenson, Judith P. Swazey, Norman A. Scotch ; with the assistance of Carole M. Kavanagh, Daryl B. Matthews ; assistant editor, Sue Conde Greene.

Reproductive Pasts Reproductive Futures

Genetic Counseling and Its Effectiveness

James R. Sorenson
Judith P. Swazey
Norman A. Scotch

March of Dimes
Birth Defects Foundation

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The **March of Dimes Birth Defects Foundation** is dedicated to the goal of preventing birth defects and ameliorating their consequences for patients, families, and society.

As part of our efforts to achieve these goals, we sponsor, or participate in, a variety of scientific meetings where all questions relating to birth defects are freely discussed. Through our professional education program we speed the dissemination of information by publishing the proceedings of these and other meetings. From time to time, we also reprint pertinent journal articles to help achieve our goal. Now and then, in the course of these articles or discussions, individual viewpoints may be expressed which go beyond the purely scientific and into controversial matters. It should be noted, therefore, that personal viewpoints about such matters will not be censored but this does not constitute an endorsement of them by the **March of Dimes Birth Defects Foundation.**

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To our reproductive pasts
and their reproductive futures:

Peter
Beth and Woody
Stephen, Ruth, and Kenneth
Meghan

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Foreword

Although birth defects are as old as the human race, early advances in understanding human inheritance were limited. Beginning in 1910, with the rediscovery of the work of the great pioneer Gregor Mendel, there was steady but slow growth in knowledge of the mechanisms of inheritance and in understanding the causes of certain birth defects.

In the last three decades, however, the field of human genetics has virtually exploded. In terms of scientific understanding, research on DNA holds promise of remarkable developments. In addition, there has been significant growth in charting the role of genetics in more and more birth defects and in making increasingly precise estimates of the likelihood that such defects will occur in specific pregnancies. Perhaps the most spectacular technologic advances have involved the development of a wide array of prenatal diagnostic procedures.

This increased understanding of human inheritance, and associated technologies, is unfortunately clinically applicable today to but a minority of the population with and at risk for birth defects. As investigators continue to explore new frontiers, what lies ahead will undoubtedly be pertinent to many more people.

The field of applied human genetics rests on this foundation of rapidly expanding knowledge of inheritance and technologic developments. Clients seeking genetic or birth defects counseling confront many novel issues and concerns, as do the providers of these services. Genetic counseling, a hybrid field slightly more than 25 years old, is expected — by practitioners, clients, and society — to provide expert services with the same effectiveness as other, more developed areas of medicine.

Whatever its basis in knowledge and technology, whatever the expectations of practitioners, clients, and society, genetic counseling is an evolving and expanding service. It exists, it provides services — some better than others, and it touches the lives of thousands of individuals.

This book examines the nature of the services provided today from two points of view — providers' and recipients'. We examine the discrepancy in expectations and assessments of services by these two parties. We try to ascertain which services are good and which are lacking. We also include contextual factors and their influence on services. In our view, a major missing ingredient in the growth of genetic counseling has been a virtual absence of feedback to providers about the services and their effectiveness. In contrast with the laboratory situation, where investigators see with their own eyes the impact of varying procedures, in genetic counseling this important ingredient is often missing. Accordingly, genetic counselors cannot change ineffective practices, and genetic counseling cannot be modified for maximum effectiveness.

The study reported here is designed to provide some of this necessary information, so that providers may develop more effective genetic counseling and clients may find the genetics of the 21st century humane and useful to them in living with their reproductive pasts and achieving their varied reproductive futures.

James R. Sorenson, PhD
Judith P. Swazey, PhD
Norman A. Scotch, PhD

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We also express our appreciation to the clinic directors and staff who participated in this study (see Appendix 3). Their willingness to take on the burdens of the study in addition to their regular activities is a testament to their commitment to the field of genetic counseling.

We would also like to acknowledge the support and encouragement of the March of Dimes in this research. The March of Dimes' role encompassed three activities. First, they provided funding through a series of grants to the research team. Second, they served as initial liaison between the genetic counseling clinics and the researchers, encouraging the participation of the clinics and their staffs in the research project. Third, they gave the research team a single mandate: to evaluate the effectiveness of genetic counseling in the clinics to which they were providing service funds.

Beyond this, the March of Dimes played no role in the design, conduct, or conclusions of the research. Determining what aspects of genetic counseling were to be studied, how they were to be studied, and what constituted effective genetic counseling was solely the responsibility of the research team, which consisted of N.A. Scotch, PhD, J.R. Sorenson, PhD, and J.P. Swazey, PhD, Co-Principal Investigators; D.B. Matthews, MD, PhD, Project Director; Carole M. Kavanagh, MS, Project Coordinator; and M. Griffin and C. Goodman, Research Assistants. Statistical consultation was provided by J. Barrett and computer assistance by Marc Mucatel.

Finally, we want to express our appreciation and gratitude to the more than 2,000 genetic counseling clients who participated in this study. While experiencing life events of major personal importance, they were willing to take the time to assist us. Hopefully, their efforts will be justified.

Chapter 1
Genetic Counseling: Definitions and Goals

INTRODUCTION

In 1981 approximately 3,300,000 babies will be born in the United States. Between 150,000 and 200,000 of these newborns will be diagnosed as having a birth defect, that is, a structural or metabolic disease or disorder that is genetically determined or the result of environmental influence during embryonic or fetal development [1, 2]. These birth defects will range from mild to severe to fatal, and involve symptoms which may be physical or mental, or both. Some defects will be present at birth; others will appear later in life. Some disorders will occur throughout the population, while others will be confined to certain ethnic or social groups. Some birth defects will be treatable, but most will impose a lifetime of limitation on the biologic, psychologic, or intellectual functions of the affected individual. In all, in 1981 some 15,000,000 Americans of all ages will be living under varying levels of handicap due to one or more birth defects [1].

Statistics such as these, imperfect as they are and open to revision as knowledge changes, counter the notion that birth defects are not an important public health problem. Indeed, each individual type of birth defect is rare; but collectively birth defects have major public health significance. Birth defects are today a leading cause of mortality in this country in the early years of life [3]. In addition, because of their chronic nature, they account for sizable amounts of health expenditures [2]. In addition it has been estimated that birth defects, because of their usual early onset, account for a heavier loss or reduction in productive future years than other more widely recognized public health problems such as cardiovascular disease, cancer, and stroke [4].

A second perspective gained from such statistics is that birth defects are a recurring threat to a sizable proportion of each generation of children. While medical science has gained dramatic control over some major health problems, such as certain serious infectious diseases, it appears that knowledge of the causes, treatment, and prevention of birth defects has not progressed as much or as rapidly as needed.

While there is validity to this perspective, it is important to recognize that progress in understanding and controlling birth defects is being made. Significant developments, many with direct clinical applications, have occurred within the past decade. For example, it is possible to provide more and more couples planning to have a child with increasingly precise estimates of the likelihood of any of their children having a birth defect [5]. In some cases, using prenatal diagnosis, it is possible to detect certain disorders in a developing fetus [6]. Progress also has been made in developing effective therapy for some genetic diseases, such as phenylketonuria [7]. And research is identifying more of the factors that play a significant role in the etiology of birth defects, enabling couples to avoid and society to eliminate or control certain factors [8].

What these developments mean today is that the birth of a child with a defect, or concern about the chances of such an event, can set into motion the deployment of medical resources and technology, much of which did not exist just a few years ago. Central to the delivery of such developments is a medical service called birth-defect or genetic counseling. It is not a new service, but is rapidly becoming useful to a larger and larger segment of the childbearing population.

At one time genetic counseling could offer only limited and often incomplete information to a public concerned about birth defects. Such information may have entailed a diagnosis of a child born with a problem, and the provision to parents of some estimate, usually quite tentative, of the likelihood of any future child being born with the same disease or disorder. An understanding of how the tragedy occurred often eluded not only the parents, but medical science as well.

Today genetic counseling is a complex service, encompassing not only better diagnosis and more precise prediction of birth defects, but also improved prognosis, treatment, and sometimes prevention. As its applicability has broadened, so too has its clientele, who seek counseling with various interests and questions about birth defects and face a variety of medical, personal, and social problems.

Genetic counseling has evolved, as have many medical services, not by systematic study of clients* and the design of appropriate professional services, but rather in response to various pressures, concerns, and interests of clients, professionals, and society.

To date there has been no large scale systematic study of genetic counseling or its clients, or whether or not this service is accomplishing the possibilities provided by past and recent developments.

*A variety of labels have been used to designate those who receive genetic counseling, including "patient," "counselee," and "client." We have chosen to use the word "client" in this monograph.

The absence of such information has not gone unnoticed by the providers of genetic counseling. To the contrary, there have been numerous calls by genetic counselors to gather information that will permit the design of more effective services [9, 10]. Moreover, many counselors, within the limits of their own clinic settings, have attempted to provide some information on their clients and the effectiveness of their services [11, 12].

The questions being asked by providers about genetic counseling reflect the present elementary understanding of many aspects of this service. Questions cover a broad spectrum of issues, from what information should be provided to clients, to how best to provide such information and when, to what are the proper objectives of genetic counseling, to what degree these are being accomplished. In addition, there are questions about who should provide counseling and how counselors should be trained. Clearly there is professional interest in better understanding genetic counseling and, as reflected in the efforts of some providers, a realization that clinical impressions, while useful, are only part of the information needed to develop and improve genetic counseling services. As diagnostic, therapeutic, and preventive knowledge about birth defects continue to develop, there will be increased pressure on these services and, accordingly, increased need to make such services more effective.

The study reported here is one effort to provide more systematic knowledge about genetic counseling. The study, initiated and funded by the March of Dimes Birth Defects Foundation, was designed to involve a large number of clients and professionals, so a more general and representative picture of genetic counseling than heretofore available could be developed. To this end, over 2,000 genetic counseling clients, counseled at 47 clinics located throughout the United States, were carefully followed to provide information on them, their questions and concerns, on counselors, their training and experience, and ultimately on the effectiveness of genetic counseling as a clinical service.

Before providing a detailed picture of the study's focus, however, we present a brief discussion of the history of genetic counseling. This discussion provides a context for viewing the scope and focus of the present study.

GENETIC COUNSELING: A BRIEF HISTORY

In the United States, genetic counseling clinics can be traced back at least as far as 1910 to the Eugenics Record Office in Cold Spring Harbor, New York [13]. In the eugenics movement, genetic counseling was a service provided by a variety of professionals, mostly self-proclaimed experts in social reform. Genetic counseling was a service that embraced the eugenics movement's broad social reform mandate and often coercive measures.

After the demise of the eugenics movement in the 1930s, genetic counseling shifted both its institutional focus and its goals. By 1955 there were at least a dozen genetic counseling clinics in this country, with over three-fourths located in academic departments of biology and zoology. Genetic counseling during this era was provided largely by academically respectable professors who were involved not in social reform but in the rapidly evolving science of genetics. Applied human genetics was being used during this period not to remake society, but to enable individuals to achieve some understanding of the genetics of diseases and disorders they were experiencing in their families.

Just over a decade later there were almost 100 genetic counseling clinics in the United States. By this time, another shift in the institutional focus and, to some degree, the functions of this service had occurred. In contrast to 1955, by 1968 over three-fourths of the clinics were located in medical settings, not academic university departments. Genetic counseling had become firmly ensconced in the medical world, and now providers were largely medical professionals, experts in the genetics of human disease and clinical medicine. Applied human genetics in this context began to reflect the values and objectives of the medical world.

Organized medicine, with its clinical rather than social reform or simply educational objectives, remains the primary location of genetic counseling services today. While the precise number of specialized clinics providing this service is not known, by 1980 there were at least 327 clinics in this country, and certainly the number is larger today [14].

Genetic counseling received its name in 1947. Dr. Sheldon Reed, an early genetic counselor, was concerned that the prevailing names for the service, which at that time included "genetic advice" and "genetic hygiene," were too eugenic or socially oriented [15]. In their place he suggested the name genetic counseling to emphasize more the individual, one-to-one relationship and concerns that he felt should exist between a counselor and client.

While the label genetic counseling has become widely accepted, it is neither precise nor does it provide us with an understanding of what actually takes place when a professional and a layman come together for genetic counseling.

During the 20th century, in sum, genetic counseling has been practiced by various types of experts, in different institutional settings, and with shifting emphases and objectives. The types of services or activities that constitute genetic counseling have changed, as has the name of this service.

As the science of human genetics, epidemiologic research, and various technologic developments have extended the scope and complexity of problems to which genetic counseling is relevant, there has been an increased interest in the objectives and effectiveness of this developing medical service.

How effective is genetic counseling given recent developments, and are there ways in which the service can be improved?

No study, however large, can or should attempt to address all the questions one can ask about a medical service. For the present study three topics were deemed important, because a prior understanding of them was considered necessary before efforts to change or expand existing genetic counseling services could be considered or undertaken. These three topics are: 1) the questions and concerns clients bring to counseling, and the degree to which counseling addresses these client-defined needs; 2) the effectiveness of counseling in educating clients; and 3) the impact of counseling on clients' reproductive intentions.

The rationale for the selection of these topics as major foci of the present study is discussed in the following sections. We explore definitions of counseling, as well as previously published studies of the effectiveness of this evolving medical service.

WHAT IS GENETIC COUNSELING?

The question, "What is genetic counseling?" can be answered in at least two ways. First, we can examine existing definitions and from them obtain an understanding of both the methods and the objectives of this medical service. Second, we can answer the question by describing empirically what takes place when this service is provided. The study to be reported here is in part a descriptive study, that is, an empiric answer to the question, "What is genetic counseling?" To define the scope and focus of this empiric study, we necessarily began, however, with an examination of professional views and definitions of genetic counseling.

There are numerous definitions of genetic counseling in the professional literature [16], which vary to some degree in how the process of counseling is defined and what are seen as the proper goals or objectives of genetic counseling. A review of these definitions, particularly the more recent ones, shows that most of them give considerable attention to one or both of two recurring objectives. First, virtually all definitions refer to the goal of client education. While there is some variation in professional views as to what clients are to be educated about, there is considerable agreement that counseling is basically an educative undertaking. Clients should be able to make more informed decisions regarding their birth defect concerns and questions after counseling than they would have been without it. We will return to the question of what clients are to be educated about after examining briefly the second major objective contained in many provider definitions of genetic counseling.

The second frequently cited objective is to provide counsel to clients about their problems and situation. By counseling, these providers mean more than simply client education. They mean helping clients think through their situation, so that they are able to use the information they are given in a constructive fashion. In addition, many advocate helping clients adjust psychologically and socially to their problems.

No single definition of the several available can reflect completely the diverse and varying emphasis given by providers to the educative, counseling, and other aspects of genetic counseling. One definition, however, does seem to capture the spirit of many definitions, and in addition provides some elaboration of the educative and counseling objectives of this service.

This definition of counseling appeared in a December 1974 paper in the *American Journal of Human Genetics* [17]. As formulated by F.C. Fraser and a number of other experts, counseling is:

> ...a communication process which deals with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or the family to (1) comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management; (2) appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

This definition provides a broad and complex mandate for genetic counseling. As defined here it is clear that counseling is aimed at both clients already confronted with a birth defect or genetic disorder and those concerned about the probabilities of a birth defect occurring.

Accomplishment of the tasks outlined in this definition requires considerable knowledge and skills on the part of the professionals involved. Not only would they have to have sophisticated genetic-medical knowledge, but they would also have to have counseling skills. Moreover, the accomplishment of objectives three and four suggests rather in-depth and detailed knowledge of clients, their values, and family aspirations, knowledge that would seem to require social work skills as well.

The definition provides some clarification of the objectives of client education in counseling. Clients are to be educated about the diagnosis, prognosis, and treatment of the medical problem in question. In addition, clients should understand their options for dealing with their problems, as well as

the etiology of the problem and the risks of recurrence or occurrence in a specific relative, who often, of course, is a future child.

Item four in the definition suggests that counselors should act as "decision facilitators" and help clients make their decisions. Finally, item five touches perhaps more than any other on the counseling aspects of this service. The emphasis here is on assisting client and family adjustment to their situation and to any decisions made. It appears that counseling activities, in this definition, are oriented more to social, ie family adjustment, than to psychologic issues such as depression, although there are providers of this service who emphasize more psychologic issues [18].

The definition cited is oriented almost exclusively to the objectives of genetic counseling as a complex clinical encounter requiring multiple professional skills. The paper in which this definition appears does provide some discussion of how each of the various objectives may be accomplished, but the paper remains largely focused on the mission rather than the method of genetic counseling [17].

In focusing on this one definition, it is important to recognize that the professionals involved vary in their orientation to counseling and, presumably, in the delivery of the service [19]. Hence, one cannot speak of genetic counseling as a single, monolithic type of service, wherein a client at one clinic would necessarily receive the same service as at another. Rather, providers undoubtedly vary in how they approach their professional role as counselor, and accordingly the services received by clients will vary. Nevertheless, many of the differences among providers in their approaches to genetic counseling seem to be largely a variation in emphasis on what clients should be taught and the type of counseling, social or psychologic, considered essential to this service [20]. Thus, while only one of numerous definitions, that by Fraser et al provides a useful perspective on major aspects of genetic counseling as viewed by the providers today.

We thus found it useful to refer to this definition in determining in part the focus and scope of the descriptive aspects of our study. In addition, the definition provided us with a starting point in addressing the study's second major function, evaluation of the effectiveness of current genetic counseling services. By identifying client education and counseling as major goals, this definition provides at least two criteria by which we can begin to answer the question, "Is genetic counseling effective?" But as will become apparent, we deemed it important to include other criteria as well.

IS GENETIC COUNSELING EFFECTIVE?

The utility of an evaluation is very much influenced by how one determines what criteria will be used as standards to judge success or failure. In

much health care research it is customary to look primarily, if not exclusively, at professional criteria. Thus, one finds much attention given to professional views, definitions, and objectives.

A second source for specifying criteria to judge the effectiveness of a service also exists. This source, along with professionally designated criteria, may be particularly valuable when a service is new or undergoing significant evolution or change. The source is, of course, the consumers of the service.

While the views of patients or consumers have been included in evaluations of many medical services, this inclusion often has been limited to asking them to globally judge their satisfaction with the service provided. Such judgments are often of such a general nature as to be of little utility.

Because of the elementary state of knowledge about genetic counseling, we considered it essential to utilize client as well as provider criteria in assessing the effectiveness of this service. We did not merely want to gather general opinions of clients as to their satisfaction or dissatisfaction with counseling services. Rather, using what is referred to as a "needs assessment" methodology, we wanted to evaluate the degree to which genetic counseling is meeting the needs of clients coming to this service. This was accomplished by asking clients upon arrival for counseling to indicate the questions and concerns they specifically came to counseling to discuss. As will be seen in Chapter 3, clients came with a broad spectrum of genetic, medical, social, and psychologic questions and concerns.

After counseling, clients were asked to indicate what specific questions and concerns they had discussed with their counselor, and to what degree. This information permitted a comparison between what clients told us before counseling they had come to discuss and what they reported after counseling they in fact had discussed. To the degree that clients discussed the questions and concerns they brought to counseling, counseling may be considered, at least in part, effective in terms of meeting client-defined needs.

Client-defined needs, and whether counseling addresses them, constituted the first set of criteria we employed to assess the effectiveness of genetic counseling. The second set of criteria we employed were professionally defined ones, drawn from professional definitions such as that by Fraser et al, and from previously published studies of genetic counseling.

There have been numerous studies published on the effectiveness of genetic counseling [11–13]. By and large these studies, usually conducted by genetic counselors, have been limited to genetic counseling in a single clinic with a relatively small number of clients. A significant limitation of many if not most of these studies is that they have employed retrospective designs which do not permit rigorous assessment of counseling's impact [21]. That is, they have usually interviewed clients only after counseling, and on the basis of this single observation have made assumptions about the effectiveness of genetic counseling.

Setting aside questions about the methodologic adequacy of these studies, they do tell us something about various populations of clients, and about what providers feel counseling should be achieving.

A review of these studies suggests that the criteria most consistently used by genetic counselors to evaluate the success of their work are, most importantly, the level of client medical-genetic knowledge postcounseling and, secondarily, client reproductive intentions and/or behavior postcounseling.

Assessments of client knowledge have focused almost exclusively on client understanding of their occurrence/recurrence risk. Some effort has also been made to assess client diagnostic knowledge postcounseling. Finally, while there is some discussion of client understanding of the characteristics of a disease and its prognosis, it is usually unclear whether it is the clients, as opposed to the professionals, whose prognostic understanding was studied.

The numerous published studies suggest wide variability in the knowledge of clients after counseling. Some studies report as few as one-fourth of counseling clients knowledgeable after counseling about such things as their risk for having a child with a specific birth defect [28]. Other studies report much higher levels of client knowledge on this issue [29]. Because of retrospective study designs, even where most clients are knowledgeable after counseling, we cannot be sure that clients' knowledge is due to genetic counseling. Most counseling is, as we shall see, tertiary care, which means by the time clients see a genetic counselor they usually have had considerable contact with the medical world. In these contacts they may have learned a great deal about their problem, including the diagnosis and risk. Hence, if clients are knowledgeable, it does not mean they acquired their information in counseling. What these studies do show, nevertheless, is that after counseling clients vary widely in their grasp of the knowledge that many counselors consider essential to "informed" client decision-making.

A second criterion used to assess the efficacy of counseling involves the reproductive intentions and behavior of clients postcounseling. Generally in the published studies counselors have suggested that effective counseling results in not only informed but also "rational" reproductive planning and behavior. In the literature this translates into a) clients wanting to have children or actually having children when they are at low or even moderate risk for a minor disease or one that can be effectively treated, and b) not wanting to have or not having children when they are at a high risk for minor as well as serious disorders that cannot be treated. Such a position views genetic counseling as a form of preventive medicine, suggesting that its ultimate utility resides in preventing the birth of children with more or less predictable serious birth defects.

Several studies to date suggest a strong inverse relationship between the magnitude of risk a couple faces for having a child with a birth defect and their intention of having a child. There is some suggestion also that the more

serious or burdensome a disease, the less likely clients are to plan or have children.

Because of design limitations, it is not possible to be sure that the observed relationships among risk magnitude, burden, and reproductive intentions were influenced by a client's genetic counseling. In addition, in virtually all studies to date assessments of what constitutes a high or low risk or of what is a serious or burdensome disease have been the opinions not of clients, but of professionals. Hence, other than in a very general fashion, we do not really understand how a client's perceptions of risk magnitude and disease characteristics relate to reproductive intentions and behavior, nor how such concerns are influenced by counseling.

Finally, while client knowledge and reproductive intention and behavior constitute the most frequently studied aspects of genetic counseling, some attention, but much less, has been given to the "counseling" aspect of this medical service. Moreover, of the studies that have looked at such issues, most have examined psychologic more than social or familial issues.

For example, there has been some sustained interest in how genetic counseling impacts on parental self-concepts [24, 25] but to our knowledge there has been only one effort to assess the impact of genetic counseling on the family or its adjustment [26].

It is difficult to draw firm conclusions from so few, often very small, studies. They suggest, however, that while genetic counseling can be a psychologically positive event for some parents, for others it does not seem to lessen the risks of elevated levels of marital disruption that may be associated with being the parents of a handicapped child [27, 28].

THE STUDY

Against such a background, the present study was designed focusing on three issues: delineation of client needs, client education, and the impact of counseling on client reproductive intentions.

To our knowledge, no effort has been made to identify the questions and concerns clients bring to counseling, knowledge that should be very useful in designing services that meet not only professional but also client criteria of effective counseling.

Client education has clearly been an issue at the center of genetic counseling, one, however, that has not been as critically assessed as it should be. Accordingly we examine this issue in some detail in our study.

Finally, the impact of genetic counseling on client reproductive decisions is a topic that warrants attention for two reasons. First, there has been much professional and lay interest in the topic, and while genetic counseling is viewed perhaps less today than in the past as a form of preventive medicine, strong interest remains. Second, it will be useful to provide some perspective

for professionals on their role in shaping client reproductive decisions. At the present time we have only a limited understanding of how clients use the information obtained in counseling. A better understanding will help us put genetic counseling in perspective as but one of several factors clients consider in making their reproductive choices.

Of course, there are many additional features of genetic counseling that would be useful to examine. For example, what are the psychologic status and needs of clients in counseling, and does counseling assist clients with their psychologic problems? Also, how does counseling impact on the families of clients, and are there familial problems that need attention? While such topics are important, the scope and methodology of this study were limited, and we only touch on some issues that some may feel should be further explored.

The topics chosen for examination here are, we believe, central to the service of counseling as presently conceived by the professionals involved. We want to add to this an awareness of how clients view this service. Together, discussion of these several issues is necessary if appropriate and more effective professional resources and medical facilities are to be rationally deployed in making genetic counseling as effective a service as possible.

ORGANIZATION OF THE MONOGRAPH

The remainder of this monograph is organized into six chapters. Chapter 2 describes some of the more significant methodologic aspects of the study, while Appendix 3 lists clinics participating in the study and Appendixes 1 and 2 contain copies of the questionnaire instruments. Separate male and female client questionnaires were used in the study. However, since we report primarily data on female clients in this monograph, and since the male and female instruments are virtually identical except for certain questions on pregnancy history and some other minor wording differences, Appendix 2 contains only female client questionnaires.

Chapter 3 provides a description of the clinics, counselors, clients, and counseling sessions on which this report is based. These data should permit the reader to readily grasp the various factors that combine to produce genetic counseling as delivered in the clinics studied here.

Chapter 4 is the first of three chapters focusing specifically on the issues of major concern in this study. This chapter describes the questions and concerns we found clients bringing to counseling, and the degree to which these are discussed in counseling. We also present a "profile" of the questions and concerns of various types of clients seeking counseling. These data provide a commentary on the variety of situations that professionals in counseling can expect to confront.

Chapter 5 looks at client learning in counseling. Here we focus on learning risk and diagnostic information. In addition, we look at how clients' perceptions of such information change, and we draw out some implications that these data may have for professionals in counseling.

Chapter 6 deals with the topic of change and stability in client reproductive intentions. This is an exceedingly complex topic, and the discussion in this monograph provides some insight into the role of genetic counseling in shaping client reproductive intentions.

Finally, Chapter 7 raises the general question of what is meant by genetic counseling and its effectiveness. We return to definitional issues and, in light of our study, offer comments and observations on the current organization of counseling services, as well as raise issues we think warrant further discussion.

In the process of conducting the study considerably more information was gathered than is reported in this monograph. We will publish a number of papers in the future on selected aspects of genetic counseling not included here.

SUMMARY

Genetic counseling has received considerable attention over the past decade from a number of professional groups. For example, ethical issues in the application of genetic knowledge have been discussed extensively by ethicists, theologians, genetic counselors, and social scientists [29]. Similarly, legal scholars and other professionals have explored numerous legal aspects in the provision of genetic services [30].

Many factors contribute to the sustained interest in genetic counseling. Certainly the problems, decisions, and dilemmas confronting clients in genetic counseling have captured interest because of their human dimensions. Many persons receiving genetic counseling are experiencing a series of life events of major significance. Some are reconsidering their desire to be a parent, their willingness to live with a child with a birth defect, and their ability to restructure their lives in terms of events and compromises that many, if not most, had never previously considered. Given such difficult issues there is genuine interest and concern in how people confronting such decisions respond to these challenges, and there is equally strong commitment to develop services that effectively meet the needs and problems of these people.

Along with the human side of such problems is the fact that these clients are confronting issues and dilemmas that are largely novel, not only for each client as an individual, but more importantly for our culture as well. Our culture has not and does not prepare us for parenthood in light of the knowledge and technology of 21st century human genetics. In the past we have had children, not without concern as to their normalcy, but without the ability to make reproductive decisions on the basis of probabilistic statements as to

their normalcy. What kinds of questions do clients have who are facing such decisions? Are there ways of helping them make – and then live with – such decisions?

The professionals delivering genetic counseling services are also confronting novelty. Medical genetics is a relatively new specialty, and training is necessarily limited. Genetic counselors, most often trained as physicians and steeped in the knowledge of human genetics, while being technically proficient recognize the need to better understand genetic counseling as a medical service and to develop counseling-related means to work more effectively with clients and their problems.

No study or discussion can give full due to the complex issues involved in genetic counseling. We hope, however, that the present study can provide some information that will permit those engaged in the provision of genetic counseling services to make "more informed decisions" about the content and process of their work and suggest ways counselors may better go about accomplishing their important task.

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Chapter 2

The Study: Instrumentation, Design, and Methods

INTRODUCTION

With support from the March of Dimes, and the generous cooperation of many professionals and clients, we were able to conduct a study of sufficient size and scope to provide a more detailed view of genetic counseling than previously available. Employing a prospective, longitudinal design, the study eventually involved 2,220 clients, counseled by 205 professionals in 47 clinics located in 25 states and the District of Columbia. Information was gathered not only on clients, but also on their counselors and the clinics in which the counseling was provided.

The conduct of a study of this size and complexity encountered numerous problems and obstacles in its execution. Many of these were solved, but some necessitated changes and a few compromises in what we set out to accomplish.

In this chapter we discuss the more important methodologic features of the study, beginning with a year's pilot work that led to the formal study. We then provide detailed discussions of the study's instruments, design, and participation rates. We conclude by discussing some methodologic issues of special significance in this study.

THE PILOT STUDY

The research began with a year of field work at four genetic counseling centers located at the following universities: Albert Einstein College of Medicine of Yeshiva University, Bronx, NY; Jefferson Medical College of Thomas Jefferson University, Philadelphia, PA; Tufts-New England Medical Center, Boston, MA; and Yale University School of Medicine, New Haven, CT. This pilot phase had two major objectives. First, while we were familiar with the extant literature on genetic counseling, we wanted to broaden and sharpen our understanding of this complex medical encounter by increasing the amount of direct experience and discussion we had had with both professionals and clients. Through interviews and observations, considerable attention was given in this early stage of our research to noting

the language, phrases, and wordings used by professionals and clients with reference to the problems, dilemmas, and decisions they faced in, or subsequent to, genetic counseling. This information proved valuable in adding to our understanding of the views, concerns and assumptions of both professionals and clients, and of the general nature of genetic counseling as a medical activity.

Second, we wanted to explore various ways of gathering information on the issues we saw as needing study. By having access to the staff and clients in four clinics we were able to try various methods of data collection. For example, we looked at the relative merits and costs of interviews versus questionnaires, and the research costs and benefits of contacting clients before their visit to the clinic versus contacting them only after they had arrived at the clinic.

The pilot work led to a series of decisions concerning the design of the study, appropriate instrumentation, and operationally feasible methods of mounting and completing a national study. Moreover, it suggested that information should be gathered on the following four topics: 1) the staffing arrangements and resources of the clinics; 2) training, experiences, and attitudes of the clinics' professional and nonprofessional staff; 3) the nature and volume of cases referred to counseling clinics and their disposition; and 4) the impact of genetic counseling on clients. In this monograph our primary attention is to the fourth aspect of the study, but we use information from the other three study components at various times to provide as complete a description and analysis of genetic counseling as possible.

INSTRUMENTATION

Our major task was to evaluate the effectiveness of genetic counseling. Since we wanted to survey counseling outcomes for a large number of clients at many clinics, the operationally feasible route was utilization of self-administered questionnaires as the main data collection instrument. In addition, as the focus of the study became clear during the pilot work, it was apparent that questionnaires could supply much of the information to be collected for the study.

Considerable effort went into developing and testing questionnaires that would be comprehensible to the variety of clients we found seeking counseling. In deciding on the data to be collected, we limited ourselves to information and attitudes that could most validly and reliably be ascertained through self-administered questionnaires.

While we felt the questionnaire methodology could provide us with much useful information on the selected core aspects of genetic counseling, it was clear during our pilot work that personal interviewing would also be necessary if we wanted to gather more in-depth information on clients, their

problems, and decisions. For example, questionnaires could supply good information on such things as client knowledge and reproductive intentions, but issues such as why clients had certain questions or concerns, why they wanted to discuss certain issues in counseling, and why they made the reproductive decisions they eventually did, were better ascertained through personal interviews.

Accordingly, 190 semistructured interviews were conducted at 12 selected clinics. Observations of counseling sessions were also conducted at these sites. This information provides a rich body of qualitative data unobtainable from survey questionnaire research. We are using in a very limited fashion the information gathered via personal interviews in this monograph. We are preparing a separate monograph that will more fully utilize the information gathered through personal interviews.

DESIGN

To adequately assess the impact of genetic counseling, it was necessary to design a study that was prospective in format; that is, one that initially would collect information from clients prior to their receiving genetic counseling. This design would enable us to ascertain such things as client questions and concerns, level of knowledge, and reproductive intentions before seeing a counselor. Such data could then be collected again from clients after counseling and a comparison made to assess the impact of counseling. Without such a before-after design it is impossible to rigorously assess the impact of counseling, whether one is looking at client needs, education, or reproductive decision making [1].

In conjunction with this prospective feature, we felt it necessary to make the study longitudinal in design as well. That is, we wanted to follow clients not just through the counseling experience itself, but also to look at them later. We suspected that for some clients the full importance of genetic counseling only becomes apparent after they have had time to think through, discuss, and, in general, reflect on their counseling. In addition, by following clients over time we could assess retention of knowledge. Accordingly, we decided to follow up clients right after counseling, and again six months later.

A pre-post counseling design, in addition to providing a prospective and longitudinal assessment of genetic counseling, permitted us to use each client entered into the study as their own control. In other words, the measurement taken on each client as they entered the study served as a comparison against which to assess later measurements on the same client.

According to experimental design theory, it would have been ideal, in addition to using each subject as their own control, to have a control or carefully constructed comparison group against which to compare clients re-

ceiving genetic counseling [2]. During the pilot study we explored the use of several such control or comparison groups. For example, we considered using clients who either cancelled or failed to keep appointments as comparison groups.

While methodologically preferable, we found it impossible to utilize a control or comparison group in the conduct of the study. Regarding a classical "control group," we considered it ethically indefensible. Such a strategy would have necessitated withholding genetic counseling from a random sample of clients for the duration of the study. With respect to various comparison groups, our strategy of using clients who failed to keep counseling appointments and those for whom counseling sessions were cancelled proved operationally infeasible. These cases occurred infrequently, and, additionally, the logistics of locating and following these individuals precluded their use within the resource and time limitations of the study. Moreover, on methodologic grounds we have reservations about using such arbitrarily constructed comparison groups. For example, we suspect that those clients who fail to keep appointments are significantly different from those who keep appointments, and hence it could be very misleading to use "no shows" or even client-cancelled cases for constructing a comparison group.

The absence of a control or comparison group requires some comment. As noted in Chapter 1, we focused on three specific substantive issues in this study, and special effort was made in gathering data on each issue to compensate for the absence of a control or comparison group.

First, regarding our assessment of client-defined needs, clients were asked before counseling for the specific reasons, that is, the questions or concerns, that brought them to counseling. We then asked clients after counseling what they had discussed with their counselor. A comparison was then made between what clients wanted to discuss and what they report they actually discussed. Exclusive of client recall of problems, this strategy which cues clients specifically to their counseling experiences should provide us with a reasonably valid and reliable report of what transpired. Given this cueing procedure, the necessity of a control or comparison group is limited.

Second, of the three topics examined, client education is the one that would appear most likely to require a control or comparison group. More specifically, in the time period that passes between our before and after measures of a client's knowledge, clients could conceivably acquire knowledge from sources other than their own counseling session. Our response to this concern is that it is extremely unlikely that there exists any source, medical or otherwise, from which a person could receive the highly *client-specific* information about their case other than from their counseling session. Hence, it is likely that if we observe a change in client knowledge

from before to after counseling, particularly an increase in accuracy, it can be attributed validly to the counseling experience. Clients seek or are sent to genetic counseling because the information on their case cannot be obtained elsewhere in the medical care system, and it certainly does not exist reliably from other nonmedical sources. Hence, once again, it would appear that a control or comparison group would have limited utility.

Finally, regarding change and stability in reproductive intentions, we again attempted to cue clients specifically to their counseling experience in an effort to obtain a valid assessment of the impact of genetic counseling. We ascertained the clients' before and after reproductive intentions, and also asked clients after counseling to tell us whether or not their counseling per se had changed their reproductive intentions. Thus, clients were cued specifically to the impact of their counseling and, aside from recall problems, we should obtain relatively valid estimates of the impact of counseling on reproductive intentions.

As should be apparent we gave considerable attention to the utility of control or comparison groups in this study. In the final analysis, because ethical and logistic problems made it impossible to employ such groups, we attempted to design compensatory corrections into the study. At appropriate places below, we will comment on this strategy.

Table 2-1 summarizes the overall design of the study. As can be seen, a prospective-longitudinal repeated measures design was employed. After agreeing to participate in the study, clients completed a precounseling questionnaire (T_1). They then received genetic counseling (T_2). Subsequently, both the client and the counselor (or principal counselor, if more than one

TABLE 2-1. Research Design

	Time/Activity			
Study participants	T_1 Client asked to enter study and completes pre-counseling questionnaire/interviews	T_2 Client receives genetic counseling	T_3 Client/counselor complete post-counseling questionnaire/interviews	T_4 Client completes six-month follow-up questionnaire/interview
Female clients	O_{1F}	X	O_{2F}	O_{3F}
Male clients	O_{1M}	X	O_{2M}	O_{3M}
Counselors		X	O_{1C}	

X = activity.
O = questionnaire or interview.

was involved) completed a postcounseling questionnaire (T_3). Six months later the client completed a follow-up questionnaire (T_4).

PROCEDURES

A study of this magnitude and design posed formidable logistic problems. It proved impractical to place research personnel at each clinic, regardless of how desirable this would have been both from the clinic's perspective and from the study's vantage point. Instead, each clinic director designated a person who acted as the "study coordinator" for that clinic. This person coordinated all of the data collection activities for the clinic and served as our source of contact and information when it became apparent that a clinic was having difficulty adhering to the research protocol.

Operationally we designed the study to put as small a burden on each clinic as possible. When clients arrived for counseling they were given a self-explanatory packet of materials about the study. An introductory letter in this packet informed clients about the study, its sponsor, and their possible role in it. We carefully explained to clients that no person at the clinic would have access to their individual questionnaires. To ensure confidentiality, we had clients put their completed forms in envelopes addressed to the study center at Boston University School of Medicine, seal the envelopes, and return them to the study coordinator. Subsequent to this initial contact, the burden of continued client participation fell on the research team at Boston University School of Medicine.

To provide time for clients to complete a precounseling form, clinics were instructed to ask them to arrive one-half hour before their actual counseling session. Separate male and female versions were prepared for couples, and they were instructed to complete these independently.

After completing the forms, clients received genetic counseling. Included in the packet of materials given to clients as they arrived at the clinic was a sealed postcounseling questionnaire which they were instructed not to read before counseling. They were asked to take it home after counseling, complete it, and return it to Boston University School of Medicine within one week. Soon after the counseling session, the counselor completed a post-counseling form provided by the clinic coordinator. This form was collected by the study coordinator, who made sure the identification code numbers for precounseling and counselor postcounseling forms matched. The coordinator then returned these forms to the study center.

Six months after counseling, the research staff mailed clients their follow-up questionnaire, which clients mailed back to Boston University School of Medicine.

As an examination of the client questionnaires in Appendix 2 shows, we asked the same or very similar questions on the pre-, post-, and six-month questionnaires. This permitted us to look at clients across time and assess

consistency or change in knowledge and attitudes. In addition to soliciting necessary medical and genetic information on each case, the counselor post-counseling form had some of the same questions that appeared on the client postcounseling form. This permitted us to compare client and counselor perceptions of genetic counseling.

CLINIC PARTICIPATION RATES

Our clinic target population for the study consisted of 80 clinics receiving service funds from the March of Dimes in 1976. As anticipated, some of these clinics were not able to participate in the client assessment phase of the study. Of the 80 clinics, 47, or 59%, provided us with clients.*

Thirty-one clinics were not able to contribute clients to the study, for various reasons. The primary reasons related to inconvenience. Some clinics were so small that the added burden of the counselor acting as study coordinator proved impossible. In addition, some clinics were undergoing change in either directors or important staff, and this made their situation too fluid to participate. Few centers refused to participate without stating inconvenience as the primary factor.

Clinic organizational information was ascertained on 77, or 96%, of the 80 clinics in the target group. These data permitted us to make a detailed comparison of the 47 clinics providing clients with those that did not.

To assess similarities and differences between clinics contributing and those not contributing clients to the study, we compared the clinics on three sets of criteria: 1) staff, in terms of both size and professional mix; 2) organization of counseling sessions, including such activities as pre- and postcounseling conferences; and 3) services available and diseases or disorders for which counseling was offered.

In terms of clinic staff size and professional mix, no significant differences emerged between clinics contributing and those not contributing clients. Similarly, contributing and noncontributing clinics were indistinguishable in terms of how they organized their counseling sessions and "processed" clients. Looking at services offered, we found that a significant difference emerged for only one of 18 services generally offered at these clinics. Noncontributing clinics were more likely to have social work services available than contributing clinics (42.9 vs 21.3%, $p < .05$). Regarding the diseases and disorders for which counseling was offered, we looked at 22 different conditions, and only one significant difference emerged. Contributing clinics were more likely to provide counseling for Huntington disease than noncontributing clinics (93.6 vs 71.4%, $p < .02$).

*Actually 49, or 61%, of the 80 clinics agreed to provide clients for the study. Because of methodologic concerns over possible instrument effects, 2 clinics were used solely for a methods study, to be reported below. This left 47 clinics contributing to the main client study.

Taken together, these data suggest no consistent pattern of differences for the criteria we examined between clinics contributing and those not contributing clients to our study. We thus conclude that, within the limits of our comparison, and with the exceptions noted, the sample of contributing clinics was not significantly different from the group of noncontributing clinics.

CLIENT PARTICIPATION RATES

Using the method of continuous case ascertainment, the participation of clients in the study was solicited via a study coordinator as clients arrived at a contributing genetic counseling clinic.* In general, coordinators were instructed to try to enlist all new clients who were coming in for genetic counseling *for the first time*, until the clinic had contributed 50 clients to the study, or the clinic had participated for a year, whichever came first. Coordinators were instructed to exclude non-English-speaking clients, clients coming in for diagnostic work only (including amniocentesis), and those who were returning for a follow-up genetic counseling session. Pilot work indicated that within the time and resource constraints of the research project, we would necessarily be limited to these participation rate criteria.†

Using this procedure, a total of 1,773 genetic counseling cases, whether involving a single person, a couple, or a family, were designated as eligible for entry into the study. Of these, 1,369, or 77.2%, were registered into the study.

Table 2-2 reports the reasons for nonentry among the 404 eligible cases not registered into the study. These 404 cases are divided into those asked and those not asked to participate. Specific reasons within each of these categories are given.

As can be seen, 73.9% (299) of the 404 nonentered cases were asked but did not participate. The major reason was that the client chose not to participate. Among the eligible but not asked cases, the major reason was that the client did not speak English well enough, in the judgment of the study coordinator, to participate.

In the pilot work we had become concerned about the possibility of clients being withheld from study participation by their counselor because he or she felt they might be too emotionally upset. In our study only 7 of 1,773 cases were excluded at the request of the counselor. Since our design called for

*For a detailed description and data on problems undergoing multi-institutional review, see Kavanaugh C et al: We shall overcome: Multi-institutional review of a genetic counseling study. IRB 1(2): 1–3, 1979.

†This selection strategy was designed to provide the study with new cases and, among these, the genetic counseling sessions in which most or all of the genetic counseling was provided, if the case was to be seen more than once.

TABLE 2-2. Reasons for Case Nonentry Into Study

Reason for non-entry into study	Reason as percent of all reasons given for case nonentry	Reason as percent of total eligible case population
Cases asked	(73.9)	
Client chose not to participate	46.5	10.6
Client failed to return pre-counseling questionnaire	16.8	3.8
Other	10.6	2.5
Cases not asked	(26.0)	
Client did not speak/write English	10.9	2.5
Insufficient time at clinic	8.2	1.9
Other	6.9	1.6
N	404	1,773

enrolling clients in the study prior to their contact with the genetic counselor, a low number of such exclusions is to be expected. It should be noted, nevertheless, that this low counselor-based exclusion rate suggests that counselors were not systematically excluding "difficult" cases and entering only "easy" ones into the study.

Little information was available about clients who were not registered into the study, and hence we could not make a detailed analysis of what biases, if any, may have been operating to make our client study population different from the client target population. Most of the time, however, we were able to ascertain the medical-genetic problem that had brought nonparticipating clients into counseling.

We compared the ten most frequently mentioned problems in the participating and nonparticipating client populations and found few differences, although the numbers involved for most problems are so small as to make comparisons statistically unreliable. Nevertheless, these data suggest similarity in the rank order of occurrence of problems in the participating and nonparticipating client populations.

In sum, the data comparing entered and nonentered client cases indicate that slightly more than 82% (1,369) of cases asked (1,668) agreed to participate. About 6% of eligible cases were lost due to insufficient time to partici-

pate or because a client did not understand English. It does not appear that there was any significant selective withdrawal of cases from the study by counselors or study coordinators. Finally, proportional representation and rank of disorders in the nonparticipating case population is comparable to that in the participating population. This suggests no significant bias as to the type of cases entered into the study from among the pool of eligible cases.

CLIENT RETENTION RATES

Once clients completed a precounseling questionnaire we were concerned about maintaining as many of them in the study through the six-month follow-up as possible. After counseling, the burden of returning study questionnaires rested with clients. Standard follow-up procedures were employed to maintain the involvement of clients over time.

Table 2-3 reports client retention rates throughout the study, by sex. Retention was over 80% at all times. As can be seen, among the cases entered into the study there were 1,314 female clients and 906 male clients, or a total of 2,220 clients who completed precounseling questionnaires. Of these, 1,099 female clients and 752 male clients returned a postcounseling questionnaire. These numbers constitute a retention rate of 83.6% among females and 83.0% among males.

Because of time and resource limitations, it was not possible to solicit the participation at six months of all clients who had completed the postcounseling questionnaire. Approximately 90% of T_3 participants were asked to take part at six months (T_4). As can be seen in Table 2-3, of those asked, 82.2% of the females and 80.8% of the males did participate.

Retention rates achieved in this study are very satisfactory. A detailed analysis has been performed comparing those clients who stayed in the study through T_4 with those who did not. Clients were compared on a large number of criteria, including age, marital status, household income, religious identity, education, occupational status, diagnosis, and relationship of proband to client. Such comparison should reveal any marked factors that differentiate clients remaining in the study from those who dropped out or were excluded because of project resource limits.

This analysis revealed three statistically different differences between clients who were retained in the study and those who were not. These differences were statistically significant only for female, not male, clients. First, a somewhat larger proportion of female clients who stayed in the study tended to have postgraduate training than those who were lost — 28.5% compared to 18.5% ($p < .05$). In a related fashion, female clients who were retained in the study reported on the average having occupations with slightly higher status than those who dropped out. Finally, female clients who dropped out were more likely (71.1% vs 64.0%, $p < .05$) to report an affected child as the proband in the genetic counseling case.

TABLE 2-3. Client Retention Rates

Client sex	T_1 Questionnaire number entering study	T_2 Number receiving counseling	T_3 Postcounseling questionnaire			T_4* Six-month questionnaire		
			Asked	Participated	Percent participated	Asked	Participated	Percent participated
Female	1,314	1,314	1,314	1,099	83.6	983	808	82.2
Male	906	906	906	752	83.0	678	548	80.8
Total	2,220	2,220	2,220	1,851	83.3	1,661	1,356	81.6

*Due to resource and time limitations 89.7% of those participating at T_3 were asked to participate at T_4.

The differences reported, while statistically significant, we feel are not substantially large, and hence are of limited importance. Thus, for most purposes the clients who dropped out of this longitudinal study generally do not appear to be markedly dissimilar to those who remained.

COUNSELOR PARTICIPATION RATES

As the research design presented in Table 2-1 indicates, for each genetic counseling case entered into the study the professional primarily responsible for providing the counseling was asked to complete a counselor postcounseling questionnaire. Counselors were asked to complete these questionnaires for all 1,369 counseling sessions. They completed 1,350 forms, for a completion rate of 98.6%. Thus we have very complete medical-genetic information on almost all cases, as well as comparative information on how clients and professionals view the same counseling experiences.

SPECIAL METHODOLOGIC CONSIDERATIONS

In a study of the size and complexity of this one, a variety of methodologic issues may take on special importance. In addition to the usual concerns about ascertaining and maintaining client participation rates, three additional issues, two internal and one external to the study, were of concern to us.

Instrument Effects

The greatest special methodologic concern internal to the study resided in the potential for a significant instrument effect. Our pilot work indicated that for some clients, seeing the precounseling questionnaire suggested issues and topics to discuss in counseling that they otherwise might not have thought of or raised. Similarly, some counselors in the pilot phase indicated that they would like to use a copy of the counselor postcounseling questionnaire as a guide in their counseling sessions, perhaps structuring their counseling to fit topics in the questionnaire, rather than following their usual procedures.

If either or both of these instrument effects took place, our very method of assessing genetic counseling would be shaping and potentially altering the counseling process as it would usually take place. Accordingly, we decided to conduct a methodologic assessment of possible instrument effects.

To accomplish this, two clinics were designated "methods study clinics." The data collection period at each of these centers was divided into two phases. In the first phase (one-half of the data collection period), clients were given no precounseling questionnaire, but only a postcounseling form. Also in phase 1, counselors did not fill out a counselor postcounseling questionnaire. This procedure kept the client and the counselor blind as to the study

focus. Thus both parties could participate in the genetic counseling session without the "suggestions" provided in the client pre- and counselor postquestionnaires.

In phase 2, clients received a pre- and a postform, and counselors also began filling out their postforms. This procedure replicated the design of the actual study.

Our pilot work suggested that if the instruments were to have an effect, a major way would be to increase the number of topics that counselor and client discussed during counseling and in so doing increase the duration of counseling sessions. If this is how the instruments were exerting an effect, we should be able to detect it by comparing client data collected in phase 1 of our method study with client data collected in phase 2. If the instruments were shaping counseling behavior, phase 2 clients should have reported discussion of more issues, and counseling sessions should generally have lasted longer than those involving phase 1 clients.

Of course many factors can shape the variety of issues discussed in a genetic counseling session. Thus it was necessary, before assessing any instrument effect, to analyze the comparability of phase 1 and phase 2 clients at the methods study clinics. The only difference that emerged was that phase 2 female clients reported slightly higher mean occupational prestige scores than phase 1 female clients. While the difference was statistically significant, it was small, and hence we do not think of much substantive importance in this situation. Thus, for the purpose of our analysis we assume the phase 1 and phase 2 clients to be highly similar.

An analysis of the number of topics discussed, as well as the duration of counseling sessions, revealed no significant differences between phase 1 and phase 2 counseling sessions at our methods study clinics. While these are not the only ways in which the study instruments could have had an impact, we think they are among the most probable and significant. Hence, on the basis of this analysis we have concluded that, while the instruments may have had some effect, they did not have a strong effect on the "natural" process of counseling, either for clients or for counselors.

Postcounseling Questionnaires

A second internal methodologic concern revolved around the procedure of having clients take the postcounseling questionnaire home to be completed. This raised two possible problems. First, in a prospective study of this kind it is important that the pre- and postmeasures take place as close in time to the event being assessed—in this case genetic counseling—as is feasible. The more removed in time either the pre- or postassessment becomes, the less confident we can be that any observed difference reflects the actual "effect" of genetic counseling, rather than some other event occur-

ring between the two measures. Attainment of the premeasure constituted no problem in this study, since it was ascertained just prior to genetic counseling. We were concerned, however, about the speed with which clients would return their postcounseling forms.

We had originally anticipated that clients could fill out the postcounseling forms in the clinic right after counseling, and hence avoid this methodologic concern, but this strategy proved to be logistically impossible. Accordingly, clients took their postcounseling forms home to complete.

Clients were asked to return postcounseling forms as soon after counseling as feasible, but no later than one week. Clients from whom we did not receive a postcounseling form within 14 days of counseling were contacted by the research team and asked to return their forms.

Fortunately, client response time did not prove to be a significant problem. For female clients, 52.3% completed their postform within one day of counseling, 72.9% within one week, and almost 80% within two weeks. Male clients responded only somewhat more slowly, 40.4% within one day, 65.4% within one week, and nearly three-quarters by two weeks. A few clients took three to four weeks, and sometimes longer, to respond, but for the majority of clients a postcounseling form was completed within seven days of counseling.

Counselors were more diligent, with better than 90% completing forms within a day of the counseling session.

Another concern related to having clients fill out their postcounseling questionnaires at home was the potential for collaboration between spouses in completing forms. However, a methodologic study during the pilot phase of our work indicated that collaboration between spouses on the take-home postcounseling form would probably not be a significant problem. We compared the degree of agreement between ten couples who completed their postcounseling forms in the clinic, where collaboration was not possible, to ten couples who took the form home and returned it to us. We found only a minor increase in agreement in the latter group. Thus, while there may have been some collaboration at home, the increase in agreement, as suggested by this limited assessment, does not appear to be substantial.

Representativeness of the Study Population

A third methodologic issue external to the study concerns the generalizability of our study population and hence our findings. As we have already noted, the 47 clinics providing clients for this study are virtually identical to the clinics which did not, and participating clients do not appear to be significantly different from nonparticipating clients. Thus, we feel reasonably comfortable in generalizing our findings from the specific clients and clinics studied to all 80 target clinics that received service funds from the March of Dimes in 1976.

A second question of generalizability is how representative the 47 clinics providing clients to this study are of all genetic counseling clinics in the United States. Although the mandate of the project was not to study a representative sample of all clinics in the United States, but only those receiving service funds from the March of Dimes, this issue of generalizability does bear on the significance of our findings for clinics outside our target population.

It is exceedingly difficult, if not impossible, to assess how representative the clinics in our study are of all clinics. In the first place there is no exhaustive listing of genetic clinics for the United States. The March of Dimes "International Directory of Genetic Services" is perhaps the most complete listing available, but it is known to be incomplete [3].

Setting aside the issue of how exhaustive this directory is, a review of the publication shows that the only criterion that can reliably be used to judge representativeness is national/regional clinic distribution, a limited criterion at best. Nevertheless, an analysis was performed comparing the national geographic distribution of clinics in the study with those in the "Directory." This analysis revealed that, with the exception of the Midwest, the distribution of clinics in the study is very similar to their distribution nationally. Clinics in the Midwest are underrepresented 1) because they were not as likely to be receiving service funds from the March of Dimes in 1976, and 2) because in one large midwestern state a statewide assessment of counseling was being undertaken at the same time as this study, and that entire state opted not to participate.

SUMMARY

A variety of methodologic considerations have been reviewed in this chapter. First, we examined the prospective-longitudinal design, pointing out special efforts to compensate for the absence of a control or comparison group in this study.

Second, a comparison of clinics providing clients with those not providing clients indicates that the two groups of clinics are comparable in several important ways. This suggests that the findings based on the clinics contributing clients can, for practical purposes, be generalized to the noncontributing clinics.

Likewise, we reported that almost 80% of clients who were asked to enter the study did so. An analysis of those who did not participate revealed that the major reason for nonparticipation was that the client did not want to. Virtually no clients were excluded because of a counselor's concern about the capacity of the client to participate. Finally, a comparison of the diseases or disorders in the participating and nonparticipating client population suggests that the two groups are typified by highly similar medical concerns.

Client retention in the study was highly satisfactory, in excess of 80% at all times. More importantly, a statistical comparison of those clients who were retained through all three data collection periods with those who were not revealed only a few minor differences.

Three methodologic issues were of special concern in this study, but none proved to be of significance. First, most clients returned their postcounseling questionnaires sufficiently soon after counseling to assure us of a reasonable assessment of the impact of counseling. Second, while we had some concern about a possible instrument effect, an analysis of a special methods study revealed no appreciable effect. Finally, while it would be useful to know how representative participating clinics in the study are of all such clinics in the United States, data are not available to make such an assessment. We are confident, however, that the participating clinics are highly similar to the nonparticipating clinics and hence the study results can be generalized to the targeted research population.

With these methodologic considerations in mind, we turn in the next chapter to a description of clinics, counselors, and clients.

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Chapter 3

Clinics, Counselors, and Clients

INTRODUCTION

In this chapter we attempt to turn the sketch of genetic counseling presented in Chapter 1 into a picture by adding more detail and perspective. We look first at the clinics in our study and their genetic counseling sessions, noting how they are organized and the kinds of experiences they provide recipients of genetic counseling. In what type of medical care setting is most counseling in this study done? How long does counseling take? Is it usually accomplished in a single session, or does it require more time? Are clients seen in relative privacy, or is counseling a situation in which numerous people are present?

Having filled in some details about the settings in which counseling occurs, as well as the counseling sessions themselves, we turn our attention to the providers of this service. So far we know very little about them other than they are mostly medical professionals. What type of training and how much experience have these experts had in genetic counseling? More importantly, what do they view as the goals for their counseling, and how do they define their role as a genetic counselor?

Finally, we add to the developing picture some detail and perspective on clients. Do clients usually come alone, or as couples? What is their educational and economic status? What kinds of reproductive experiences do they bring to counseling, and what types of medical problems?

When combined, answers to the above questions help us to develop a more complete picture of genetic counseling as a medical service and human activity. We will still have, however, just a picture, a sort of photograph of the service. A more dynamic perspective will be developed in Chapters 4–6, where we look closely at what happens when clients and counselors meet.

CLINIC SETTINGS

While genetic counseling undoubtedly takes place in numerous settings, including private office practice, much if not most formal counseling today necessarily occurs in medical centers in this country. This is largely because of the limited training of primary care physicians in clinical genetics, as well as the frequent need in counseling for the highly specialized consulting ex-

pertise and technology usually found only in medical centers. Thus, not surprisingly, the genetic counseling we studied was provided most frequently in clinics located in large urban medical centers, often university affiliated.

Clients receiving counseling usually are referred by a physician, who often does not feel professionally prepared or sufficiently up to date to provide the most precise and useful counseling. However, many clients do refer themselves, because of worry about birth defects. Counseling is sought or recommended for numerous reasons, but most often because people have had a child with a birth defect, or because there is some other reason for concern, such as a family history of a birth defect.

What these brief comments suggest is that the genetic counseling examined in this report is not what is ordinarily considered primary medical care. Rather, it can more accurately be conceived of as a tertiary care service that relies on highly trained subspecialists, located in complex medical settings. It is a type of medical encounter to which the client has traveled relatively long distances, and a type of medical situation in which traditional conceptions of the doctor-patient relationship, based chiefly on primary medical care, may not necessarily hold true.

Table 3-1 begins to document what we have been discussing; namely, of the 47 clinics that provided clients for this study, most were located in hospitals. Although not shown in the table, many of these hospitals were affiliated with a medical school or a large medical center.

Very few clinics were located in other types of institutional settings, such as public health agencies or private medical care foundations. In a sense, we have studied the clinics and the professionals who, by virtue of their research and medical center affiliation, are at the forefront of modern medicine in applying knowledge of human genetics in a clinical setting. They are, in several ways, the best that medicine can offer today in its various efforts to treat, predict, and prevent birth defects.

TABLE 3-1. Institutional Location of Genetic Clinics

Type of institution	Percent
Hospitals	72.2
Research institutes	4.3
Public health agencies	4.3
Private foundations	4.3
More than one of above	4.3
Other	10.6
Total	100.0
N	47

Staffing Patterns

The clinics had various staffing patterns. In some clinics the "staff" was a single professional, most often a physician who performed multiple activities ranging from appointment making to client follow-up. Other clinics had staffs with various medical professionals, including nurses, social workers, and genetic associates as well as MD or PhD genetic counselors.

Table 3-2 reports the average number of the most frequent types of medical professionals in each of the 47 clinics. It also reports the percentages of each professional group who were identified by clinic directors as routinely providing counseling.

As can be seen from Table 3-2, genetic counseling clinics have mostly physicians on their staff, and at the other extreme there are relatively few social workers. Similarly, almost all of the physicians and most genetic associates are directly involved in counseling, but few nurses and very few social workers.

As in most medical settings, the clinics studied here were run and directed by physicians. Because of the clinical nature of the work involved, PhDs rarely function independently as counselors. In addition, genetic associates, specifically trained to do genetic counseling, rarely operate independently of a physician since they too lack certain skills, such as diagnostic capabilities, that would enable them to be autonomous, independent genetic counselors. In a sense, thus, physicians are the dominant professionals in counseling, determining the form, flavor, and eventually the effectiveness of genetic counseling [1].

The data reported in Table 3-2 also suggest that the client's clinic experiences in this study could vary considerably. For example, in the smaller clinics clients may have found their visit not markedly different in many re-

TABLE 3-2. Professional Staff Reported to Regularly See Counseling Clients at Clinics

Staff	Average number per clinic	Percent reported to counsel	N
MD	2.8	94.7	131
Genetic associate	0.8	89.4	37
Nurse	0.4	26.3	19
PhD	0.3	56.3	16
Social worker	0.3	6.7	15
Total staff	4.6	78.9	218

spects from a visit to their family physician, other than perhaps having to travel considerable distances to the genetic counseling clinic. Once there, however, they were met by and spent time with one professional in a single clinic site.

For other clients, however, the experience may have been much different. During their visit they may have been greeted by a social worker or genetic associate, who spent time asking questions and soliciting information on a broad spectrum of issues. Subsequent to this, and usually after a waiting period, they met with the counselor and several other "experts" who may or may not have discussed their case with the social worker or genetic associate.

In addition, some clients may have been sent to various specialty clinics located throughout a medical center, seeing several different professionals. In short, there is considerable variation in the professional staffing pattern among the 47 clinics in this study, and this shapes in numerous ways the types of experiences clients have. We will report below how staffing arrangements and variation in the way clinics "process" clients relate to the quality or effectiveness of genetic counseling.

Clinic Services

Table 3-3 reports the wide spectrum of diseases and disorders for which counseling is provided at the clinics in our sample. Regardless of staffing patterns it is apparent that most of the clinics are prepared to provide counseling for a wide array of diseases and disorders.

The various types of services provided in the clinics are summarized in Table 3-4. These have been arranged into four groupings—diagnostic, management, specialty, and support services. The most frequent services provided by the 47 clinics are diagnostic services, including diagnosis of an affected child or adult, prenatal diagnosis, and carrier status testing for disorders such as sickle cell anemia and Tay-Sachs disease.

Just under half of the clinics provide medical management for a disease or disorder. Even fewer provide within their clinic a variety of specialty services relevant to the diagnosis or treatment of many birth defects, such as neurology, orthopedics, or cardiology services. This does not mean that such services are not available when a genetic counseling case warrants them. Rather it means that most often a client will be referred for such services to specialty clinics elsewhere in the same medical institution and occasionally to other institutions.

In terms of support services, we have noted that social workers are not often members of clinic staffs. However, about one-fifth of the clinics make social work services available, presumably by borrowing them from other clinics when necessary. Very few clinics are directly linked to such support services as groups of parents of children with birth defects, and an even

smaller number are directly involved in helping clients handle the special schooling problems that may be associated with an affected child.

The Organization of Counseling Services

In terms of how clinics organized their services, counseling was offered in these 47 clinics for an average of ten hours per week. This average came from a range of four clinics scheduling only one hour per week to two clinics scheduling 40 hours. On average, the clinics reported that they saw about 3.4 new counseling cases per week, and 2.0 follow-up cases. Including other cases, such as those seen for amniocentesis, the estimated total client load per clinic was about ten a week. Thus, on average, somewhat less than an hour is spent with each client seen at these clinics.

Just over a third of the clinics reported routinely sending correspondence to clients before they arrived for counseling. This correspondence often requested medical history information, and sometimes solicited a family or social history as well. The correspondence frequently provided information on the clinic, such as a map, as well as some information about genetic counsel-

TABLE 3-3. Diseases and Disorders for Which Counseling Is Provided at 47 Clinics (percent)

Disease/Disorder	Percent of clinics offering counseling for disorder
Achondroplasia	97.8
Cleft lip and/or palate	93.5
Congenital heart disease	87.0
Cystic fibrosis	76.1
Diabetes	72.7
Down syndrome	100.0
Duchenne muscular dystrophy	87.2
Hemophilia	80.9
Huntington chorea	93.6
Klinefelter syndrome	100.0
Marfan syndrome	100.0
Mental retardation (nonspecific)	97.8
Microcephaly	95.8
Mucopolysaccharidoses	97.8
Multiple congenital anomalies	97.8
Multiple spontaneous abortions	97.8
Neural tube defects	95.8
Neurofibromatosis	97.8
Phenylketonuria	86.9
Sickle cell anemia	80.0
Tay-Sachs disease	97.8
Turner syndrome	100.0
N	47

ing. Over half the clinics, 53%, reported they did not regularly hold preclinic conferences to discuss cases before they were seen at the clinic.

In terms of the genetic counseling session itself, we have seen that they averaged about an hour in length. As reported in Chapter 2, of those entered into this study, a total of 1,851 cases completed their postcounseling questionnaire. Of these, 1,472, or approximately 80%, were cases involving couples, as seen in Table 3-5. When clients came alone, it was a female 91% of the time. Clients rarely brought relatives or friends to counseling.

In terms of the professionals usually present, counseling included professionals other than a counselor. Genetic associates, nurses, or social workers sat in, but rarely participated in the actual counseling. In addition, house staff and genetic fellows frequently attended the counseling sessions as observers, as did medical students in over 80% of the clinics in the study. Genetic counseling for many clients is thus not a private encounter between themselves and their physician. Rather, it often involves several professionals, as well as students, some participating, some simply observing.

Finally, a majority of the clinics (74%) reported holding regular postclinic conferences in which they discussed each case. In addition, most clinics

TABLE 3-4. Services Clinics Report Providing Directly to Clients (percent)

Service	Percent of clinics providing service
Diagnostic services	
Diagnosis of proband	97.9
Amniocentesis counseling	91.5
Other diagnostic tests	89.4
Amniocentesis	61.6
Other prenatal diagnosis	53.2
Tay-Sachs screening	50.2
Sickle cell screening	42.6
Disease management	
Proband management	44.6
Specialty services	
Neurology	17.0
Psychiatry	10.7
Hematology	6.4
Orthopedics	4.3
Cardiology	2.1
Physical therapy	2.1
Support services	
Social work services	21.3
Parents' group	12.7
Schooling for proband	4.3
N	47

(94%) reported some type of client follow-up procedure after counseling. Virtually all clinics (95%) sent a letter to the client's referring physician reporting specifics of the counseling session. Eighty percent also sent a letter to the client with the same or similar information. Six clinics reported that they arranged home follow-up visits.

The picture or, perhaps more accurately, pictures of genetic counseling clinics and counseling sessions that emerge from this digest of our findings suggest that they are variably organized and present clients with a diversity of clinical experiences. These range from private, client-physician consults, to large, group-based counseling. Diagnostic services predominate among the counseling-related activities provided by the clinics. Clinics, on average, schedule about ten hours of counseling sessions per week, with most sessions averaging about one hour.

In sum, just as we could not speak of a single definition of counseling in Chapter 1, neither can we say that there exists a single method of organizing and delivering these services. We will examine below how such variation in the organization and delivery of genetic counseling services is related to the effectiveness of these services.

THE COUNSELORS

In addition to gathering information on each clinic we also requested information from their staff who provide counseling. A total of 205 professionals counseled the cases to be reported in this study. In this section we examine the degrees, training, and experience of these contemporary experts in applied human clinical genetics. In addition we look at the goals these specialists hold for their counseling as well as how they view their role vis-à-vis counseling clients.

Specialization

Table 3-6 reports the highest degrees held by the counselors, arrayed in terms of the frequency of the degree among all counselors. As can be seen, the largest percent of counselors in this study, 58.6%, were physicians. An

TABLE 3-5. Clients in Study (percent)

Client status	Client sex		
	Female	Male	N*
Solo	18.3	1.8	369
Member of couple	41.6	38.6	1,472
N	1,097	744	1,841

*Data were incomplete on 10 cases, reducing table N to 1,841.

additional 7.9% held both the MD and PhD. Thus, about two-thirds of the counselors had medical degrees.

The second largest group of counselors, 19%, held the master level genetic associate degree. Other types of degrees, including the PhD and nursing degrees, were infrequently represented among the counselors in this study. These data support our earlier observation that genetic counseling today is very much a medically based activity, practiced mostly by medically trained professionals.

More detail about the training of the counselors is provided in Table 3-7, which summarizes information on areas of specialization by degree.

TABLE 3-6. Professional Degree of Counselor Staff (percent)*

Degree	Percent	N
MD	58.6	119
Masters	18.7	38
MD/PhD	7.9	16
PhD	5.9	12
RN	2.0	4
Others	6.9	14†
N		203
No response		2
Total		205

*For Tables 3-6 through 3-14, the N's reported represent the number of counselors, of the 205 entering cases into this study, who provided information on the topic reported.
†Includes 7 medical students and 7 unspecified others.

TABLE 3-7. Areas of Specialization, by Type of Professional Degree (percent)

	Professional degree					
Specialty area	MD	MA	MD/PhD	PhD	RN	Other
Pediatrics	67.2	–	50.0 –	–	–	–
Obstetrics-gynecology	6.7	–	– –	–	–	–
Internal medicine	10.9	–	18.8 –	–	–	–
Unspecified	6.7	–	25.0 –	–	–	–
Biology	–	–	– 37.5	8.3	–	–
Genetics	–	78.9	– 31.3	83.3	–	–
Other	8.4	21.1	6.3 31.3	8.3	100.0	100.0
N	119	38	16	12	4	14

Most physicians, including joint MD/PhD degree holders, list pediatrics as their area of specialization. Other medical specialties are infrequently represented. Most master level genetic associates report genetics as their area of specialization, of course, as do most PhD degree holders. In the latter case the training and degree in all probability are much more research oriented than the more clinically oriented training of genetic associates.

The fact that most MD counselors are pediatricians reflects the historic evolution of this subspecialty within medicine. While one might expect obstetricians-gynecologists to be playing a larger role, because of the significant potential for prevention given their position in the reproductive career of most females, pediatricians predominate. Genetics has made its inroad into medicine not so much in a preventive fashion, but through work with children born with birth defects.

Professional Training in Genetics and Counseling

A more comprehensive picture of counselors is reflected in Tables 3-8–11. Tables 3-8 and 9 report the percent of counselors with formal or didactic courses and with clinical or experientially based training in human genetics and counseling. Together these tables show that counselors, within degree groupings, report comparable levels of course work and direct clinical training in human genetics. Conversely, they consistently report more supervised clinical training than formal course work in counseling, master level counselors reporting highly similar levels. With the exception of master level counselors, only a minority of counselors, regardless of degree, had formal course work in counseling. And, with the exception of PhDs, most professionals, however, did report supervised clinical training in counseling. In short, it would appear that most counselors rely primarily upon supervised

TABLE 3-8. Training in Human Genetics, by Type of Professional Degree (percent)

	Type of training			
Degree	Didactic course(s)	Clinical genetics training	Other, including fellowship	N
MD	72.3	77.3	31.1	119
Masters	100.0	100.0	18.5	38
MD/PhD	87.5	87.5	25.0	16
PhD	91.7	91.7	8.3	12
RN*	–	–	–	4
Other	57.1	42.9	28.6	14
Average	78.3	81.3	26.6	203

*N too small for reliable percentage reporting.

clinical experience in developing counseling skills and techniques and less on formal didactic courses. The reverse is true regarding their training in human genetics.

Table 3-10 reports the mean number of years of counseling experience, by degree, of the counselors in the study. There is, in fact, a wide range in years of experience among the counselors in this study. For example, 27% reported less than one year's experience, while 3% reported 20 or more years of experience. Almost 60% reported five or fewer years of counseling experience.

The data in Table 3-10 show that MD and PhD degree counselors have the longest average experiences, while master level genetic associates among the least. We would expect this since genetic associate degrees have been granted only within the past few years. In general, counseling is a service provided by professionals with limited experience, reflecting the recent development and growth of this area of medicine.

Counseling as a Professional Activity

Above we noted that on average the clinics in this study scheduled about ten clinic hours a week. In light of this one can ask how professionals in counseling spend the rest of their professional time.

Table 3-11 shows that the different professionals in counseling vary in the estimated total time spent in professional activities each week. More importantly, we can see that with the exception of master level genetic associates, genetic counseling takes up a minority of the professionals' time. For example, MDs spend approximately 30% of their time in counseling, as do joint MD/PhD degree holders. Genetic counseling is thus not a full-time profession for most of these practitioners. They are, like many of their col-

TABLE 3-9. Training in Counseling, by Type of Professional Degree (percent)

	Type of training			
Degree	Didactic course(s)	Supervised clinical training	Other counseling training	N
MD	27.7	74.8	8.4	119
Masters	89.5	92.1	10.5	38
MD/PhD	12.5	87.5	12.5	16
PhD	8.3	41.7	25.0	12
RN*	–	–	–	4
Other	21.4	57.1	28.6	14
Average	37.4	76.4	11.3	203

*N too small for reliable percentage reporting.

leagues in tertiary medical care settings, engaged in research, teaching, clinical services, and other activities. Genetic counseling is but one of their several professional activities and commitments.

The Counselors' Objectives

As part of the study counselors were asked to indicate the extent to which they endorsed various counseling objectives. These objectives, and the percents of counselors giving various levels of support to each, are reported in Table 3-12.

Counselors could endorse all or none of these goals, to whatever degree they felt it important that their counseling achieve the stated goal. As we can see, the goal most strongly supported was to help clients adjust to and cope with their medical problems. Receiving almost as much support was the goal of removing or lessening client guilt or anxiety. Clearly, from the level of attitudinal endorsement of these goals, counselors have considerable commitment to the "counseling" aspects of genetic counseling.

At the same time it would appear, from the level of support for the third and fourth goals, that disease prevention and helping clients achieve parenting goals have less support. There is very little strong support for population-focused goals, either at the disease or genetic level.

Table 3-13 reports the degree to which the counselors endorsed the va-

TABLE 3-10. Average Number of Years of Counseling Experience, by Type of Professiona' Degree (mean)

	Professional degree					
	MD	Masters	MD/PhD	PhD	RN only*	Other
Average # yrs	6.1	2.9	8.1	7.3	–	2.0
N	117	38	16	12	4	12

*N too small for reliable reporting.

TABLE 3-11. Mean Estimated Time in Selected Professional Activities, by Counselor Degree (hours/week)

	Total professional time	Counseling in clinic	Activities outside clinic			
Degree			Research	Practice	Teaching	N
MD	58.2	17.7	12.0	12.7	7.2	119
Masters	37.3	30.4	2.8	0.6	1.6	38
MD/PhD	63.6	17.9	20.2	12.5	5.8	16
PhD	49.8	19.7	18.1	.2	5.2	12
RN*	–	–	–	–	–	4
Other	42.9	30.0	3.2	1.7	1.3	14

*N too small for reliable statistical reporting.

rious elements comprising the definition of counseling that we discussed in Chapter 1.

There is near universal support for the educative aspects of counseling, as well as helping clients understand their reproductive options. There is only somewhat less support for helping clients adjust, and the least support is given to the definitional elements that put the counselor in the position of being a decision facilitator or helping clients actually act in accordance with their decisions.

In general then, Tables 3-12 and 13 show strong support for the counseling and educative aspects of genetic counseling. They also suggest that counseling, at least in the eyes of the providers in this study, should not be as strongly focused on disease prevention.

Finally, counselors were asked what they viewed as their role in helping clients make decisions. Counselors and others have expressed marked interest in this issue, particularly the role counselors might play in assisting clients in making reproductive decisions [2,3].

Table 3-14 reports the opinions of the counselors on this topic. As can be seen, almost all counselors do not feel that their role includes directly advising or actually telling clients what to do. This does not mean they do not think it useful or important to facilitate decisions, for example, by clarifying

TABLE 3-12. Importance of Six Counseling Goals to Counseling Staff (percent)

Goals	Importance*			N
	High	Moderate	Low	
Helping individuals/couples adjust to and cope with their genetic problems	82.8	17.3	0.0	203
The removal or lessening of patient guilt or anxiety	75.2	24.8	0.0	202
The prevention of disease or abnormality	52.0	46.5	1.5	202
Helping individuals/couples achieve their parenting goals	43.8	55.2	1.0	201
Improvement of the general health and vigor of the population	11.0	62.5	26.5	200
A reduction in the number of carriers of genetic disorders in the population	6.9	48.0	45.0	202

*High—very important; moderate—somewhat important or important; low—not at all important.

TABLE 3-13. Importance of Seven Tasks in Genetic Counseling to Counseling Staff (percent)

Tasks	Importance*		
	High	Moderate	Low
To help the individual or family comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management	94.1	5.4	0.5
To help them understand the options for dealing with the risk of recurrence	91.2	8.4	0.5
To help them appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives	86.3	13.2	0.5
To help them make the best possible adjustment to the disorder in an affected family member	66.2	29.4	4.5
To help them make the best possible adjustment to the risk of recurrence of that disorder	62.7	33.3	4.0
To help them choose the course of action which seems appropriate to them in view of their risk and their family goals	54.4	31.9	13.8
To help them act in accordance with their decision	49.0	41.6	9.3
Total N			204

*Counselors were asked to rate the importance of these tasks on a scale ranging from 0 to 5: 0–2 (low); 3 (moderate); 4–5 (high).

options. Rather, it suggests that they do not feel it appropriate to intervene actively in client decision making.

The information presented in this section provides perspective on the professionals giving genetic counseling. Most are MDs and most of these are pediatricians. Most have both formal course work and supervised clinical training in human genetics, but they rely primarily upon supervised clinical training to acquire their counseling expertise.

Attitudinally, the counselors readily endorse the importance of client education as well as counseling. They are considerably less committed to endorsing disease prevention or achievement of client reproductive goals as important objectives of their counseling. And, while considering it important to help clients understand their options, even facilitating clients' decisions, they do not consider it appropriate to advise or actually tell clients what to do.

TABLE 3-14. Appropriateness of Five Counselor Decision-Making Strategies to Counseling Staff (percent)

Counselor role in client decision making	Appropriateness: Always/ sometimes appropriate	Appropriateness: Rarely/ never appropriate	N
Suggest that while you will not make decisions for patients you will support any they make	98.6	1.5	203
Tell patients that decisions, especially reproductive ones, are theirs alone and refuse to make any for them	93.1	6.9	203
Inform patients what most other people in their situation have done	65.6	34.5	203
Inform patients what you would do if you were in their situation	19.7	80.3	203
Advise patients what they ought to do	13.4	86.6	202

CLIENTS

The people who sought genetic counseling in this study shared certain things in common, yet were very different in some ways. In the final section of this chapter we provide information on who these clients were and the medical problems they brought to counseling.

Sociodemographic Characteristics

Clients can be described in numerous ways. For example, Tables 3-15 through 3-19 provide sociodemographic information. Not surprisingly most of the clients who came to counseling were married. The modal age range for both females and males was 25 to 29, with few clients 35 or older. In terms of education, 4 to 5% of the clients had no more than a Grade 9 education. Men clients tended to have had slightly more formal education than female clients, perhaps related to the fact that males tended to be somewhat older.

Table 3-18 shows total family income, as reported by male clients. Here, as with education, we find wide variability with 19.4% reporting a family income in excess of $25,000, and 24.6% reporting income of $10,000 or less.

The religious identification of clients, by sex, is reported in Table 3-19. Most clients were Protestant, followed by Catholic and Jewish identification.

TABLE 3-15. Marital Status of Study Population (percent)

Marital status	Females	Males
Single	6.1	3.7
Married	86.7	94.4
Divorced	3.4	0.8
Separated	2.6	0.5
Cohabitees	0.7	0.5
Widowed	0.6	0
N	1,088	738

TABLE 3-16. Age of Clients (percent)

Years	Females	Males
Under 20	6.2	1.1
20–24	25.2	16.5
25–29	33.8	33.2
30–34	21.7	31.4
35–39	7.0	9.0
40 or over	6.1	8.8
N	1,046	697

The impression conveyed by these data is one of client diversity regarding several sociodemographic characteristics. While most were married and young, they varied considerably in terms of formal education, income, and religion. We will examine below how these characteristics relate to the impact of counseling.

Reproductive History

Reproductively, the female client population reported an average of 1.9 pregnancies. They had an average of 1.4 live births, 0.4 miscarriages, and 0.1 therapeutic abortions. Not all clients in this study had had a child with a birth defect or had experienced some type of compromised pregnancy or birth outcome when they came for counseling. In fact, as shown in Table

TABLE 3-17. Clients' Highest Level of Education (percent)

Highest level of education	Females	Males
Grade 9 or less	5.3	4.4
Some high school	7.8	4.7
Finished high school	33.7	24.1
Some college or other post-high school training	27.6	25.8
Finished college	11.9	17.6
At least some postgraduate work	13.7	23.4
N	1,084	735

TABLE 3-18. Reported Family Income (percent)

Annual income (dollars)	Study population*
0–10,000	24.8
10,001–15,000	22.0
15,001–20,000	22.8
20,001–25,000	11.1
25,001 +	19.4
N	965

*For married couples the family income reported by males was used.

3-20, 30.2% of the population reported they had never been pregnant, were pregnant for the first time, or had had only normal births.

Table 3-20 is constructed to provide a perspective on the most adverse pregnancy or birth outcomes the population reported at the time of their counseling. In effect, each client is entered only once in the Table, into the most "extreme" adverse pregnancy or birth outcome category they had experienced up to that time. Thus, a client who had a therapeutic abortion with a diagnosed abnormality as well as a normal birth would be placed in the former category. Likewise, a client who had a living affected child and who also had had a normal child and a spontaneous abortion would be placed in the "affected child living" category.

As can be seen, about 13% of the population had experienced therapeutic abortions or spontaneous miscarriages. Almost 14% reported giving birth to one or more children with a defect, but the child had died, and 43% reported

TABLE 3-19. Clients' Religion (percent)

Religion	Females	Males
Protestant	59.0	54.4
Jewish	5.9	7.1
Catholic	27.7	28.7
None	6.5	9.5
Other	0.9	0.3
	100.0%	100.0%
N	1,066	724

TABLE 3-20. Reproductive Experience of Female Client Population (percent)

Outcome	Percent
No pregnancies	18.4
Pregnant for first time	4.8
Only normal births	7.0
Therapeutic abortion	2.1
Spontaneous abortion, miscarriage	6.6
Spontaneous abortion, miscarriage, therapeutic abortion with abnormality	4.5
Birth of affected child now deceased	13.5
Affected child living	43.1
N	1,096

one or more living affected children. As might be expected, this is a population with an extremely high level of compromised pregnancy and birth outcomes.

Medical Concerns

Clients brought a broad spectrum of medical concerns and problems to genetic counseling. Table 3-21 lists all problems and disorders which occurred among 10 or more of the cases entered into the study.

As can be seen, the list includes specific diseases such as Huntington disease, disorders with significant clinical variability such as spina bifida, as well as very general or vague problems such as nonspecific mental retardation and multiple congenital anomalies. In short, clients brought a broad spectrum of disorders and diseases to counseling, a range of problems that clearly requires very sophisticated clinical knowledge.

SUMMARY

The picture of counseling as a medical service that emerges from this discussion is one that would probably emerge if we were examining any other relatively new, rapidly evolving, highly specialized medical service in the United States today. Within the specific confines of clinical genetics, it begins with specialized, relatively small clinics, providing mostly diagnostic services. These clinics schedule an average of 10 clinic hours a week and are staffed predominantly by physicians.

The providers of the services have both formal course work and clinical training in human genetics, but rely mostly on clinical training for their expertise and skill in counseling. As a group these counselors commit more of their time to noncounseling activities, such as research and teaching, than to genetic counseling per se. In addition they consider client education and counseling as very important objectives of their counseling work, more so than disease prevention. While willing to help clients think through their options, they do not view it appropriate to tell clients what to do.

The clients, usually young and married, come to counseling mostly as couples. Rarely do they bring relatives or friends. Their counseling is likely to take place in a setting where various professionals other than the counselor are present. Clients are quite diverse in terms of education and income. As a group they have experienced a very high rate of compromised pregnancy and birth outcomes. Thirty percent reported no prior reproductive problems, but 43% have at least one living child with a birth defect. And, they bring a wide array of diseases and disorders to counseling.

Given such diversity—diversity in clinics, their organization and services; the professionals, their training and experience; and the clients, their charac-

teristics and reproductive experiences—what happens when they all come together? The next three chapters provide some insight into the complexities and impact of genetic counseling as delivered in the clinics in this study.

TABLE 3-21. Problems and Disorders for Which Clients Were Most Frequently Counseled (percent)*

Problems/Disorders	Cases in which problem/disorder diagnosed	Number of times diagnosed
Down syndrome	10.6	116
Multiple congenital anomalies	6.9	75
Mental retardation	3.9	43
Congenital heart defects	2.9	32
Delayed development	2.4	26
Hydrocephalus	2.4	26
Cleft lip and/or cleft palate	2.2	24
Meningomyelocele	2.1	23
Anencephaly	2.0	22
Short stature	2.0	22
Extremity malformations	1.7	19
Turner syndrome	1.7	18
Spina bifida	1.5	16
Drug intake in pregnancy	1.4	15
Neurofibromatosis	1.4	15
Epilepsy/seizures	1.4	15
Tay-Sachs screening	1.3	14
Tuberous sclerosis	1.2	13
Diabetes mellitus	1.2	13
Neural tube defects	1.2	13
Polycystic kidney disease—infantile form	1.2	13
Autosomal abnormalities	1.2	13
Trisomy 13	1.1	12
Microcephaly	1.1	12
Cystic fibrosis	1.0	11
Osteogenesis imperfecta tarda	1.0	11
Huntington disease	1.0	11
Duchenne muscular dystrophy	0.9	10
Trisomy 18	0.9	10
Achondroplasia	0.9	10
Stillbirths	0.9	10
N cases	1,090	

*Data taken from the Counselor Postcounseling Questionnaires. Although we asked the counselors to be as specific as possible in giving a diagnosis, at times they were not. For this reason the list includes some broad categories of problems which may overlap with some of the more specific diagnoses given. If more than one disorder or problem per case was given, the first two mentioned were recorded. For 20.9% of the cases, a second disorder was recorded.

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Chapter 4

Effective Genetic Counseling: Discussing Client Questions and Concerns

INTRODUCTION

Within the past few years a body of empirical research has begun to develop on the dynamics of the physician-patient relationship [1]. Of some interest in this literature has been the degree of agreement between the provider and the patient over the nature of the problem that brought them together [2]. Interest has focused on this problem for several reasons, but one of the more significant of these is the accumulating evidence that the most effective medical care, in terms of such issues as patient compliance, for example, is facilitated when the patients perceive that the provider understands the problem as they do, in essence, when the provider comprehends the patients' view of their problem and why they are seeking care [3].

Several studies have been published which demonstrate that while physicians are generally aware of their patients' medical problem, they seem to have difficulty appreciating the broader social and psychologic context from which a patient views problems, contextual factors that can dramatically affect the patient's response to medical care or advice [4]. Moreover, even when physicians are aware of such issues, some studies suggest that there is a tendency among providers not to incorporate this awareness into their medical consultations. In short, many providers do not appear willing to define their encounter with a patient in terms of the patient's point of view, a position that may seriously compromise the motivation, interest, and trust of the patient in their provider and his or her advice.

With such studies in mind, we felt it important to examine the counselor-counselee relationship in some depth, and to focus in particular on the extent to which counselors were aware of, and respond to, the needs of genetic counseling clients.

While we have some understanding of the ways in which professionals in the field define the scope and content of genetic counseling, we know little about the questions and concerns clients bring to this encounter. Such infor-

mation could be useful in assuring that the necessary resources are available to meet the needs of clients and to train genetic counselors.

Because of these considerations, we gathered information on the questions clients brought to counseling, and assessed the extent to which these questions were discussed, from the client's perspective, in counseling. In essence, we wanted to learn from clients, before they saw their counselor, the reasons they were seeking genetic counseling. Then, after counseling, we wanted to learn from the clients whether or not these concerns and questions had been discussed. To the degree that a goal of counseling is providing a situation in which clients raise and discuss the very questions and concerns that brought them there, such discussion is one way to assess the effectiveness of genetic counseling.

We begin with a description of the questions and concerns we found clients bringing to counseling. In addition to providing descriptive information on the more frequent questions and concerns of all clients, we have developed "profiles" of subsets of the client population, based on their reproductive experiences, which demonstrate how clients' questions and concerns can vary. Third, we report data on the degree to which clients' questions and concerns get discussed in counseling, and factors which appear to facilitate or hinder such discussion. Finally, we examine clients' assessments of their counseling experience six months after they received counseling.

CLIENTS' QUESTIONS AND CONCERNS

During our pilot work we devoted considerable effort to learning from clients the various reasons that had brought them to counseling. Several lists were developed and repeatedly revised. Much attention was given to developing questionnaire items that reflected the views of and were comprehensible to clients.

Our pilot work suggested that the topics listed in Table 4-1 encompassed the most common reasons cited by most clients. Clients were given the option of adding other reasons if they desired, but by the time this list evolved, we felt comfortable that it contained the most commonly recurring questions and concerns of most clients seen in our pilot work.

We have somewhat arbitrarily divided the 15 items we found clients bringing to counseling into two general categories. The first is what we call genetic-medical issues; the second, sociomedical issues. Of the ten items in the first category, two relate more or less to genetics—risk and etiology; five are diagnostically oriented; one focuses on characteristics of a disease or disorder as it develops; and the remaining two are oriented to care and treatment issues. The sociomedical group contains divergent issues, ranging from school and financial concerns, to interpersonal issues in relating to an affected child, other children, or a spouse.

TABLE 4-1. Most Commonly Occurring Questions and Concerns

Genetic-Medical Topics	Sociomedical Topics
Etiology of disorder/disease	Client's feelings about affected child
Chance of having an affected child	School or other special programs
Medical treatment of disorder/disease	Financial costs of disorder/disease
Diagnosis of child	Relationship with other children
Prenatal diagnosis	Relationship with spouse
Diagnosis of client	
Prognosis of disorder/disease	
Care for affected child at home	
Diagnosis of other family member	
Medical status of affected child	

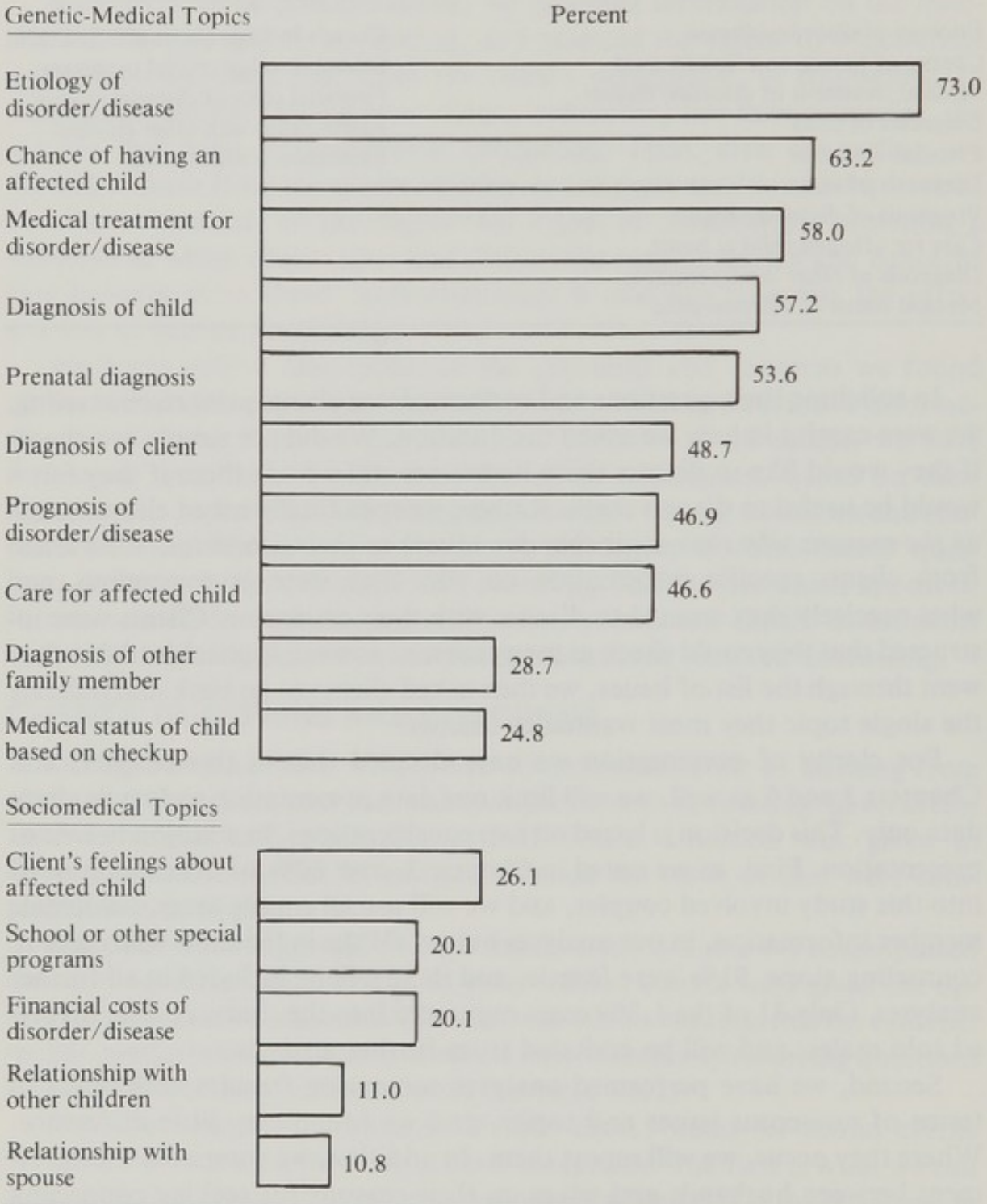
In soliciting their questions and concerns from clients prior to counseling, we were careful in how we asked the question. We did not simply ask clients if they would like to discuss these items, nor did we ask them if they felt it would be useful to discuss them. Rather, we specifically asked clients to tell us *the reasons why they came that day to talk to their counselor.* We wanted from clients specific information on why they were in counseling, and what precisely they wanted to discuss with their counselor. Clients were instructed that they could check as many reasons as were applicable. After they went through the list of issues, we then asked clients to go back and indicate the single topic they most wanted to discuss.

For clarity of presentation we have decided that in this chapter, and Chapters 5 and 6 as well, we will limit our data presentation to female client data only. This decision is based on two considerations, in addition to ease of presentation. First, as we noted in Chapter 3, over 80% of the cases entered into this study involved couples, and we will use all couple cases, via female member information, in our analyses below. Of the individuals who came to counseling alone, 91% were female, and these will be included in all further analyses. Only 31 of the 1,369 cases registered into the study, or 2%, involved solo males, and will be excluded from further analysis.

Second, we have performed analyses comparing females with males in terms of numerous issues and topics, and we found very little difference. Where they occur, we will report them. In addition, we compared the agreement between husbands and wives on their reasons for seeking counseling and found substantial agreement, with wives and husbands agreeing over 80% of the time on various reasons why they were seeking counseling. We will provide an analysis of couples in counseling in later publications.

Table 4-2 reports the percent of female clients who indicated they had come to counseling to discuss each of the 15 items we found most frequently mentioned in our pilot work. We have ordered the individual items within

TABLE 4-2. Percent of Female Clients Saying They Came to Counseling to Discuss Various Topics (N = 1,097)



the two categories in terms of the frequency with which it was checked as being a reason for wanting to talk to a genetic counselor.

As we found in our pilot work, clients brought a variety of genetic, diagnostic, treatment, and sociomedical questions and concerns to genetic counseling. On average, clients brought more diagnostic-genetic than

disease-treatment concerns, and more disease-treatment questions than sociomedical concerns.

Looking first at the genetic-medical set, we see that the question most frequently brought was the etiology of a disorder, followed by the chance of having an(other) affected child. Treatment for a disorder was a question 58% of the female clients brought, and prenatal diagnosis was a concern for almost as many, 54%. Twenty-five percent of the clients came with questions about the medical status of an affected child.

The most frequent sociomedical concern among female clients involved wanting to discuss their feelings about an affected child (26%), with about 20% wanting to discuss schooling or other special programs, as well as the financial cost of raising a child with a birth defect. About 10% wanted to discuss their relationship with their other children or with their spouse.

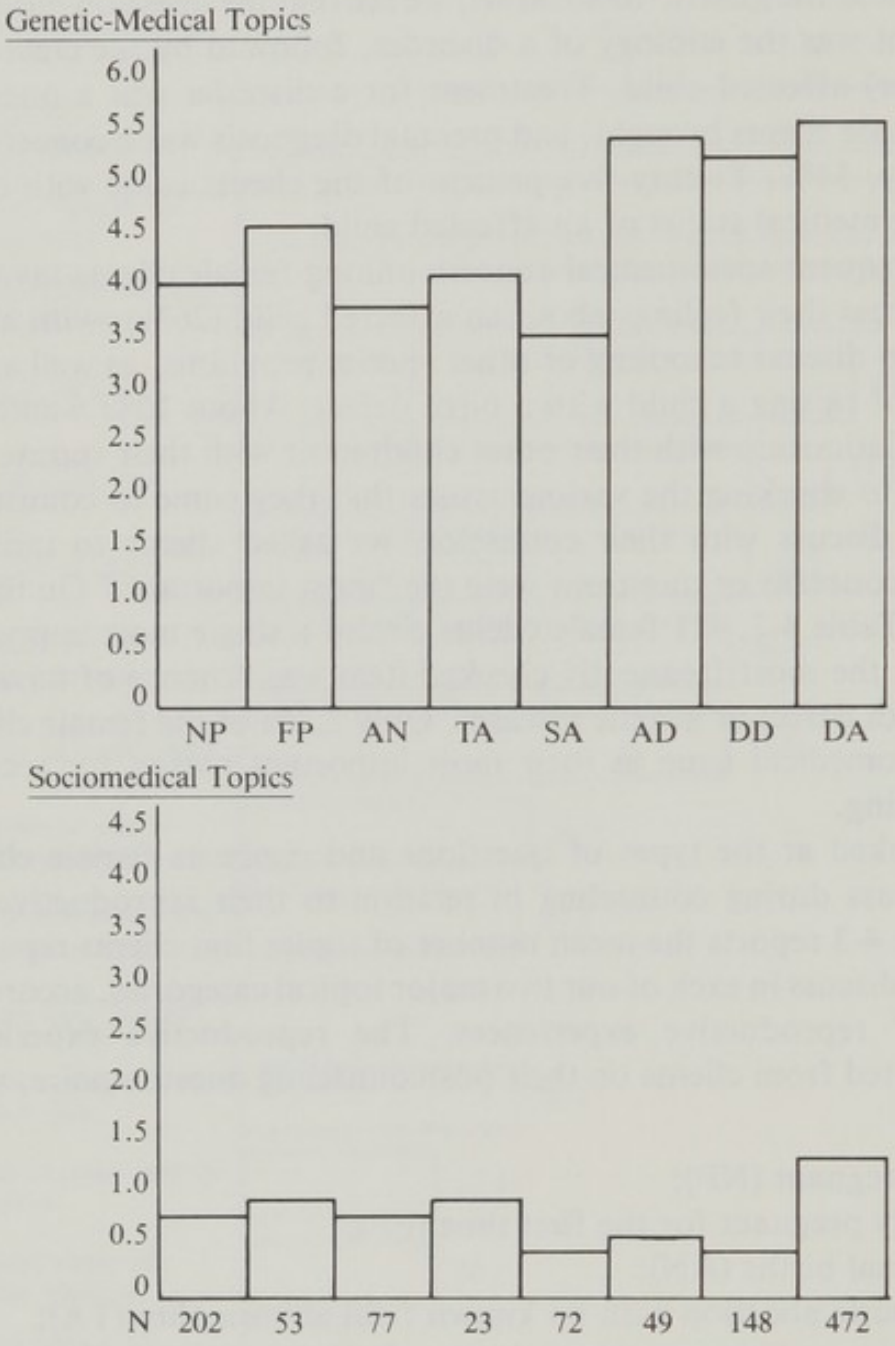
In addition to checking the various issues that they came to counseling specifically to discuss with their counselor, we asked clients to indicate which, if any, concerns or questions were the "most important." On the 15 items listed in Table 4-2, 411 female clients circled a single most important item. Of these, the most frequently checked item was "chance of having a child with a birth defect or genetic disease." Only 2.2% of the female clients checked a sociomedical issue as their most important reason for seeking genetic counseling.

We also looked at the types of questions and concerns female clients wanted to discuss during counseling in relation to their reproductive experience. Table 4-3 reports the mean number of topics that clients reported they wanted to discuss in each of our two major topical categories, according to the clients' reproductive experiences. The reproductive experience categories, elicited from clients on their postcounseling questionnaire, were as follows:

1) Never pregnant (NP);
2) Presently pregnant for the first time (FP);
3) All normal births (AN);
4) Therapeutic abortion with no known fetal abnormality (TA);
5) Spontaneous abortion with no known fetal abnormality (SA);
6) Abortion (therapeutic or spontaneous) with fetal abnormality (AD);
7) Live birth of infant with birth defect, child now deceased (DD); and
8) Live birth of infant with birth defect, child now alive (DA).

As explained in Chapter 3, each client could give multiple responses to the reproductive history/pregnancy outcome questions in their postcounseling questionnaire. In analyzing these data, however, a client was counted only once, in terms of the "worst" outcome she reported.

TABLE 4-3. Mean Number of Topics Client Wanted to Discuss, by Area and Client Reproductive Experience



Key NP, never pregnant; FP, presently pregnant for first time; AN, all normal; TA, therapeutic abortion with no abnormality indicated; SA, spontaneous abortion without apparent defect; AD, abortion with defect; DD, defective child now deceased; DA, defective-alive.

While not shown in Table 4-3, the mean number of topics clients wanted to discuss was 4.9 per client for the genetic-medical category and 0.9 per client for sociomedical concerns. These data, like those in Table 4-2, indicate that topics dealing with etiology, risk, and diagnosis are the most salient for clients.

Within the category of genetic-medical topics, the number of questions or concerns per client ranged from 3.4 for those who had experienced one or

more spontaneous abortions, to 5.6 for clients who reported a living affected child. In short, the data on client questions involving genetic-medical issues suggest that the experience of a birth defect, whether present in an aborted fetus or in a full-term birth, significantly increases the number of medical-genetic questions a client brings to counseling.

Turning to sociomedical topics, it is clear from Table 4-3 that these had markedly less saliency for clients than diagnostic-treatment issues. As we might expect, the greatest number of such questions occurred among clients with a living affected child.

An examination of the most frequent questions of clients reveals that for five of the eight reproductive-experience groups, the most frequently asked question involved etiologic concerns: the cause of the birth defect or disorder. For only two groups —those never pregnant, or pregnant for the first time— did the most frequent question or concern involve a consideration of the occurrence/recurrence risk. It would appear that clients generally are somewhat more interested in knowing why, or rather how, some birth defect occurs than they are in knowing how likely it is to occur, although their interest in the latter is significant also.

The information presented suggests that clients bring an array of questions and concerns to genetic counseling. While the most frequent of these are diagnostic or genetic in nature, there are substantial numbers of clients with disease-treatment questions, and a sizable minority of clients with what we term sociomedical questions and concerns. Clients clearly attach most importance to diagnostic-genetic issues. These data suggest that clinics need a wide array of information, resources, or referral capabilities if counseling is intended to meet the divergent questions and concerns of clients.

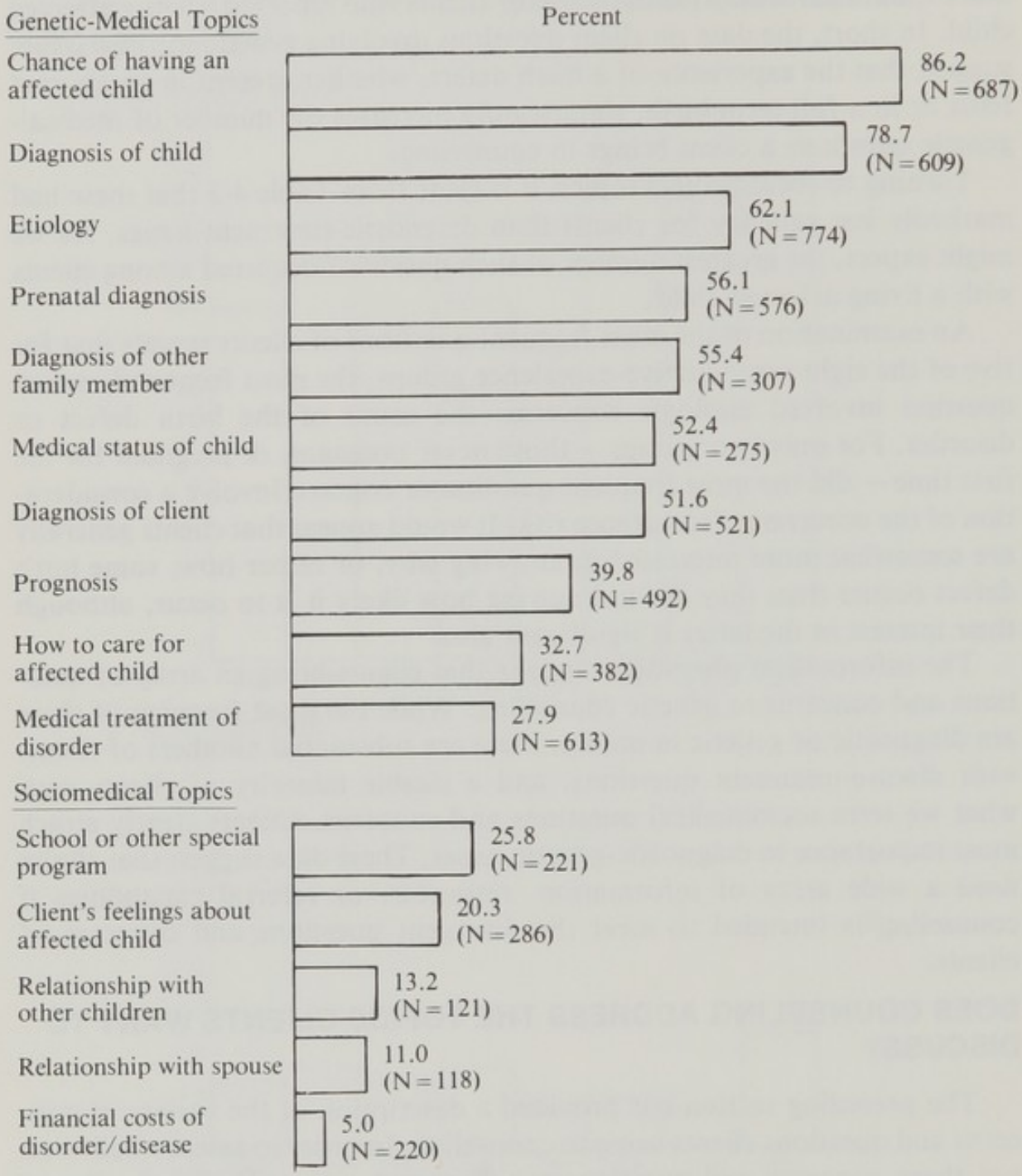
DOES COUNSELING ADDRESS THE TOPICS CLIENTS WANT TO DISCUSS?

The preceding section has provided a description of the variety of concerns and questions clients bring to counseling. In order to assess whether or not these concerns and questions are discussed, we replicated the list of topics provided by the "Registration Questionnaire" (Table 4-1) in the "Post-Counseling Questionnaire." Thus, after counseling clients were asked which of these specific issues they had discussed with their counselor.

For purposes of analysis we have considered the responses "not discussed" and "just mentioned" as *not* meeting a client's need for discussing an issue which the client stated before counseling that he or she had specifically come to discuss. The response "discussed in some depth" seems to be the best indicator of whether an issue was covered during counseling.

In Table 4-4 we have recorded the percent of clients wanting to discuss a topic *before* counseling who report having discussed it *after* counseling.

TABLE 4-4. Percent of Female Clients Wanting to Discuss a Topic Before Counseling Who Reported Having Discussed It in Some Depth During Counseling



N = total number who, on Question 8 of the Registration Questionnaire (Appendix 2), indicated they came to counseling to discuss specific issue. Percent reflects number of these who indicated on Question 18 of the Postcounseling Questionnaire (Appendix 2) that they in fact had discussed the issue in depth. The N's for certain items above may vary slightly from N's suggested in Table 4-2 because calculation of N's for the above table required respondent to answer two questions, not just one as in Table 4-2.

Results were analyzed by client sex, and once again there were virtually no significant differences in the experience of male and female clients. It was also the case that there was very substantial agreement between spouses as to whether an issue was discussed or not, generally in excess of 80%. However,

there was less agreement between clients and counselors over what was discussed. We will explore this latter issue in a subsequent publication. What is important here is the degree to which clients consider their specific questions and concerns discussed, for if they feel they have not been discussed fully, then, at least for the client, they have not been.

Table 4-4 contains considerable information regarding the extent to which client needs, expressed prior to counseling, were viewed by clients as in fact discussed during counseling. For 7 of the 15 items, at least half of the clients who wanted to discuss the issue report that the issue was in fact discussed during their counseling session. For example, 86.2% of the females who wanted to discuss their chances of having an affected child indicated that this topic was discussed in depth. Similarly, 78.7% of clients wanting to discuss the diagnosis of their child report discussing this topic in depth. Also, 62.1% of those who wanted to discuss the etiology of the disease or disorder indicated that this issue was covered in depth.

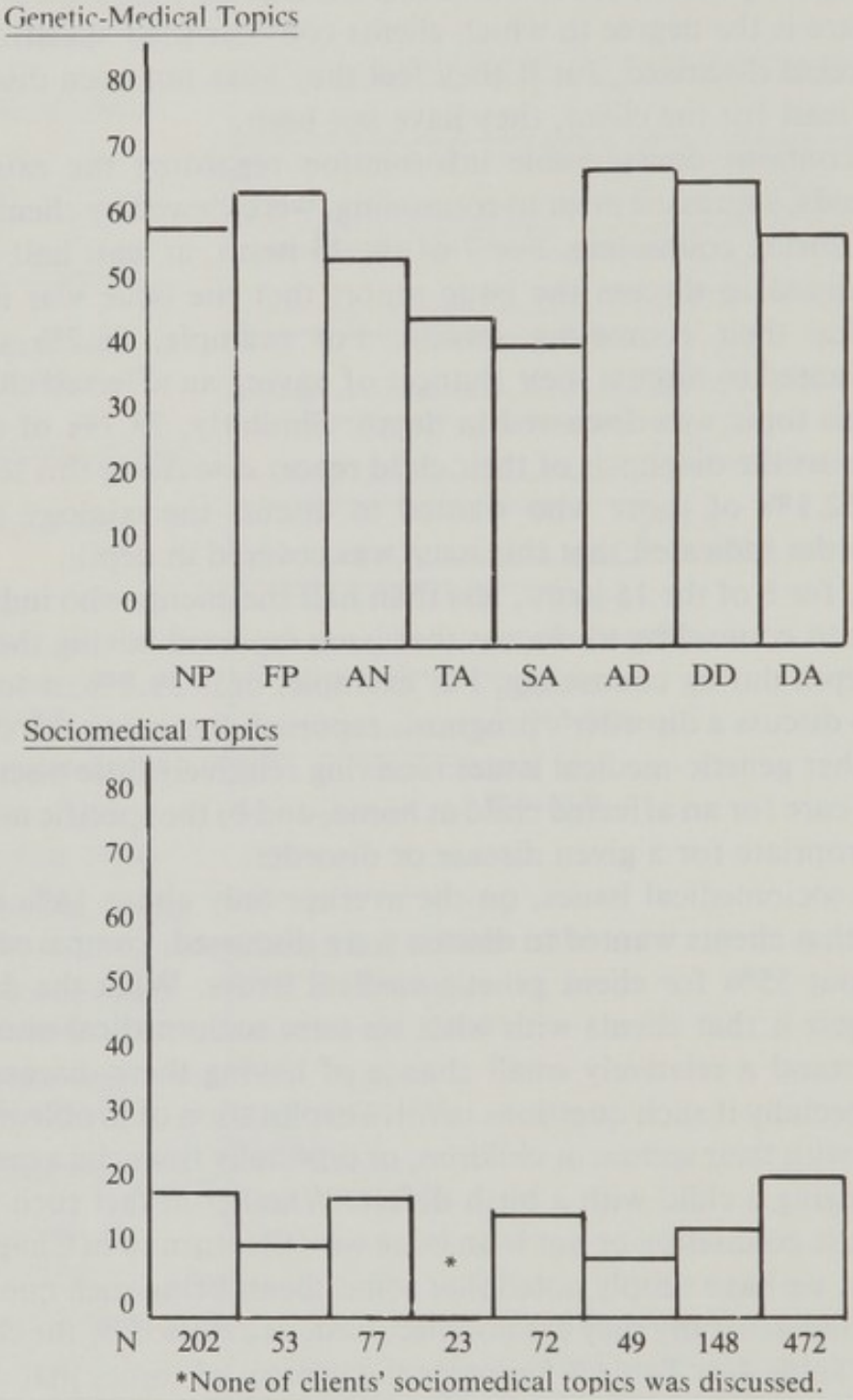
Conversely, for 8 of the 15 items, less than half the clients who indicated they had come to counseling to discuss that issue reported having the issue discussed in depth during counseling. For example, only 39.8% of females who wanted to discuss a disorder's prognosis reported discussing this during counseling. Other genetic-medical issues receiving relatively little discussion were a) how to care for an affected child at home, and b) the specific medical treatment appropriate for a given disease or disorder.

Turning to sociomedical issues, on the average only about 16% of the various issues that clients wanted to discuss were discussed, compared to an average of about 55% for client genetic-medical issues. What the data in Table 4-4 suggest is that clients with what we term sociomedical questions and concerns stand a relatively small chance of having these discussed in counseling, especially if such questions involve exploration of problems they may be having with their spouse or children, or especially financial aspects of treating and raising a child with a birth defect. Whether in fact such issues constitute genetic counseling or not is an issue we will return to in Chapter 7. For the present, we have simply noted that some clients bring such questions and concerns, and generally they are not discussed, as assessed by the clients.

Paralleling Table 4-4, Table 4-5 reports the percent of topics that clients said they wanted to discuss which were in fact discussed in counseling in relation to the clients' reproductive experiences. For all clients, an average of 55% of the genetic-medical topics they wanted to talk about were discussed. The group least likely to report the discussion of such issues are those clients with therapeutic or spontaneous abortion experiences, while the group most likely to have such issues discussed are those with some experience with a defective abortion/miscarriage, or those who are pregnant for the first time.

As for sociomedical topics – with the exception of clients having a therapeutic abortion experience, who report absolutely none of their

TABLE 4-5. Percent of Topics Clients Wanted to Discuss That Were Discussed, by Client Reproductive Experience



*None of clients' sociomedical topics was discussed.

Key NP, never pregnant; FP, presently pregnant for first time; AN, all normal; TA, therapeutic abortion with no abnormality indicated; SA, spontaneous abortion without apparent defect; AD, abortion with defect; DD, defective child now deceased; DA, defective-alive.

sociomedical concerns were discussed, and those with an abortion/miscarriage experience with an indication of an abnormality — there is little variation across the remaining groups in the percent of sociomedical topics discussed. In short, very few were discussed for any group of clients.

The data reported so far in this chapter suggest that clients come with an array of genetic-medical and sociomedical questions and concerns, and on

TABLE 4-6. Agreement Between Topic Client Most Wanted to Discuss and Counselor's Assessment of What That Topic Was*

Column A Topic client most wanted to discuss	How often identified correctly by counselor (%)	Column B Topic most often identified by counselor as being of greatest interest to client	N
Risk for having an affected child	74.5	Risk for having an affected child	411
Prognosis of the disorder	43.1	Prognosis of the disorder	51
Diagnosis of client	26.9	Risk for having an affected child	26
Diagnosis of client's child	19.8	Risk for having an affected child	91
Amniocentesis	14.6	Risk for having an affected child	48
Medical treatment for disorder	12.8	Prognosis of disorder	39
Etiology of the disorder	11.5	Risk for having an affected child	52
Diagnosis of another family member	8.3	Risk for having an affected child	24
School programs for affected child	7.1	Prognosis of disorder	14
How to care for affected child	5.3	Prognosis of disorder	19
Affected child's status	0.0	Prognosis of disorder	11

*Table reports topics which ten or more clients cited as being of most interest to them during counseling.

average, just over half of the former get discussed in depth, while only about 16% of the latter do.

Table 4-6 suggests one reason why clients may be reporting generally low levels of discussion of issues other than diagnosis, etiology, and risks. As part of the "Post-Counseling Questionnaire," clients were asked to indicate what single topic they were most interested in discussing during their counseling. Likewise, counselors were asked to indicate what one topic they thought was of most interest to the client. Table 4-6 reports the frequency of agreement between the client's report and the counselor's assessment. In addition, we report the topic counselors most often perceived as the client's major interest when there is substantial disagreement. We have arranged the topics, not in terms of the frequency with which clients cited them as their most important topic, but rather in terms of the frequency with which the counselor correctly identified the topic as being of most interest to their clients.

TABLE 4-7. Reasons Cited by Female Clients Why All Their Genetic-Medical Questions and Concerns Were Not Discussed

Reason	Percent*
Facts are not known medically	70.5
Counselor had to gather information	36.1
Client did not ask question	15.9
Counselor did not want to discuss	2.5
N	515

*Clients could cite more than one reason.

TABLE 4-8. Reasons Cited by Female Clients Why All Their Sociomedical Questions and Concerns Were Not Discussed

Reason	Percent*
Issue cannot be dealt with medically	57.9
Counselor had to gather information	36.2
Client did not ask question	20.3
Counselor did not want to discuss	3.3
N	480

*Client could cite more than one reason.

As can be seen, the client concern most frequently identified correctly by counselors involved questions about the clients' risk for having an affected child. In the 411 cases where clients identified this topic as the one of most interest to them during counseling, counselors correctly identified it 74.5% of the time. However, after this concern, counselors were usually not able to say what issue or question was of most concern to their clients. For example, counselors correctly identified a client's dominant interest in prognosis only 43.1% of the time, and a client's interest in his or her own diagnosis only 26.9% of the time. Clearly, counselors most of the time do not appear to perceive correctly their clients' major interest. In fact, they were able to do so only in 47% of the cases where a client identified a major concern or question.

Table 4-6 also reports the topics counselors most frequently identified as the one of most interest to their clients for each client concern or question. These data suggest that counselors appear to assume that most of the time the client is most interested in either the risk for having an affected child, or in information concerning the development of a disease or disorder (prognosis).

While these assumptions are frequently correct, they are wrong in an almost equal proportion of the time, suggesting a significant mismatch between what clients say is most important to them and what counselors believe to be the case.

TABLE 4–9. Female Clients Scheduled for Additional Counseling Who Reported Counselor Had to Gather More Information

Scheduled to be seen by counselor again	Type of information to be gathered	
	Genetic-medical (%)	Sociomedical (%)
Yes	60.9	56.9
Maybe	26.4	32.3
No	12.6	10.8
N	174	167

Numerous factors undoubtedly contribute to the observation that a significant proportion of the specific genetic-medical questions and most of the sociomedical concerns of clients were not discussed in depth during counseling, as assessed by the clients. The study did not ascertain either client or counselor assessments of why specific issues were not discussed. However, clients were asked after counseling whether or not they felt their genetic-medical concerns or sociomedical questions had been discussed. If they believed all such questions and concerns had not been discussed, they were asked what factors they felt might have contributed to this. Client responses are reported in Tables 4-7 and 4-8.

Looking at the reasons female clients gave for not obtaining all the genetic-medical information they wanted (Table 4-7), 70.5% said that it was because the information was unavailable – that is, "all the facts are not known to medical science." Another 36.1% reported that the counselor was gathering the information for them, while 15.9% acknowledged that they had not raised the topic or topics about which they wanted information. Only 3% felt that their counselor did not want to discuss a genetic-medical issue in which the client was interested.

Comparably, 20% reported that they had not asked the counselor about a sociomedical question or concern (Table 4-8), while again only 3% said the counselor had not wanted to discuss such a topic. Almost 36% said the counselor was gathering the information they wanted. Lastly, approximately 58% said that their concerns were not addressed because they cannot be dealt with medically – that is, they fall outside of the expertise and capabilities of medical genetics.

We thought it important to see if in situations where clients said additional information was being gathered, whether or not the client was scheduled for an additional counseling session. The data reported in Table 4-9 show that between approximately 57% and 61% of these cases were scheduled for more counseling, while there was some doubt in between a fourth and a third of these cases. This suggests a significant level of follow-up, at least for those cases where more information was being gathered.

Overall, the data in Tables 4-7 and 4-9 indicate that a majority of clients feel that the majority of the topics concerning them prior to counseling were dealt with during counseling to the extent possible in terms of available knowledge or the information possessed by the counselor at the time. Only a very small number felt that their counselor did not want to discuss one or more of the topics concerning them. Finally, there was a group of clients who said they did not get information they wanted because they did not ask the counselor about it. There was follow-up counseling scheduled for many clients when additional information was being gathered, but for clients in other situations, there was only limited follow-up.

To summarize, the data in Tables 4-4 through 4-9 suggest the following: First, counselors have a restricted field of interest compared to the topics clients bring to counseling for discussion. In general, counselor interest appears to focus on diagnosis, etiology, and risk estimation. These three topics are usually well explained by counselors and they are of considerable interest to many clients. However, many clients have a much broader set of interests, including other medical topics, such as disease prognosis and treatment, and sociomedical issues, such as a discussion of personal feelings and economic concerns.

In current practice, most sociomedical issues stand a rather poor chance of being discussed in counseling, and even such medical issues as prognosis and therapy are most often not discussed in depth, according to clients' assessments of what transpires.

It is also the case that when a client's major interest in counseling is anything other than risk estimation or prognosis, this is not likely to be perceived by the counselor. Counselors appear to assume that risk or prognosis are the chief interest of most clients; while they are of substantial interest to many clients, they are not the dominant interests counselors assume them to be.

MEETING CLIENT NEEDS: CLINIC, COUNSELING PROCESS, CLIENT, AND COUNSELOR FACTORS

The preceding sections of this chapter have documented the extent to which clients come to genetic counseling with a variety of questions and concerns, and the degree to which clients report that these issues were discussed in depth during their counseling. In this section we examine the question of what factors may contribute to the discussion of a client's self-defined questions and concerns or needs. We saw in the previous section some factors that play a role, such as the lack of relevant information by the counselor, as well as failure on the part of the client to raise some questions of concern. To examine the issue further, we looked at four categories of variables: the characteristics of clinics, the counseling process, the counselors, and, of course, the clients.

To make this analysis parallel with data presented earlier, we have analyzed separately the discussion of genetic-medical questions and sociomedical topics as organized in Table 4-1. This was also done because the factors which facilitate or hinder the discussion of medical-genetic issues may be different than those which facilitate the discussion of sociomedical topics.

Clinics

We first examined how well the various clinics did in meeting client defined needs. If we could identify clinics that did very well and contrast them with those that did poorly, we might be able to identify clinic organizational features that facilitate the discussion of client-defined questions and concerns.

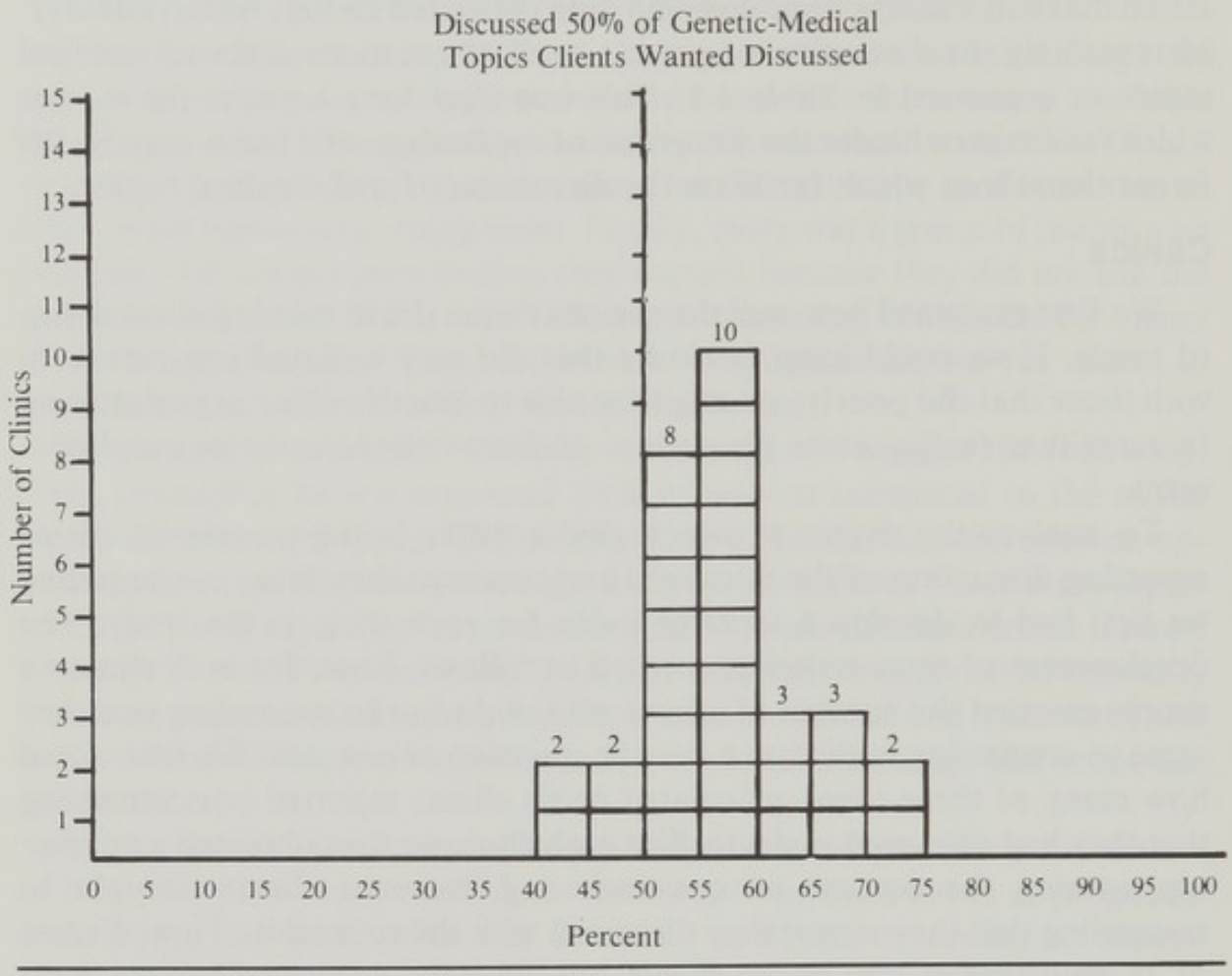
To analyze the degree to which clinics differ in the percent of clients reporting discussion of the questions and concerns they bring to counseling we first had to develop a score or index for each clinic in the study. The development of these scores proceeded as follows. First, for each clinic we simply counted the number of clients who said prior to counseling that they came to counseling to discuss a specific question or concern. We then noted how many of these client-articulated needs clients reported postcounseling that they had discussed in depth. For each clinic we thus obtained a number representing the percent of questions and concerns clients brought to counseling that they report they discussed with the counselor. Thus, a score of 25 would mean 25% of the clients' questions were reported by the clients to be discussed, and a score of 75 would mean 75% of client questions were reported discussed in depth during counseling.

The scores this procedure developed must be interpreted carefully. First, we must keep in mind that when we talk about genetic-medical and sociomedical issues, we mean only those ten- and five-item groupings shown in Table 4-1. Clearly, there are many other genetic-medical and sociomedical issues that could be discussed, but our pilot work suggested the importance of these 15 items.

Second, the score has been constructed such that it assesses a clinic only in terms of the questions and concerns the clinic's clients brought to counseling – that is, a clinic scores high only if clients report discussing topics they wanted to discuss. No "credit" is given for discussing topics clients said they did not want to discuss. At the same time, clinics are not penalized for not discussing topics clients said they did not want to discuss.

Third, the score for each clinic is based not on the number of clients seen by a clinic, but instead on the number of medical and nonmedical topics that a clinic's clients wanted to discuss. Thus, since our measure represents the percent of client-articulated concerns discussed, the score does not control for the average number of questions and concerns clients bring to a particular clinic, nor does it control for the number of clients at a particular clinic.

TABLE 4-10. Distribution of Clinic Scores for Discussing With Clients the Genetic-Medical Topics Clients Wanted Discussed*



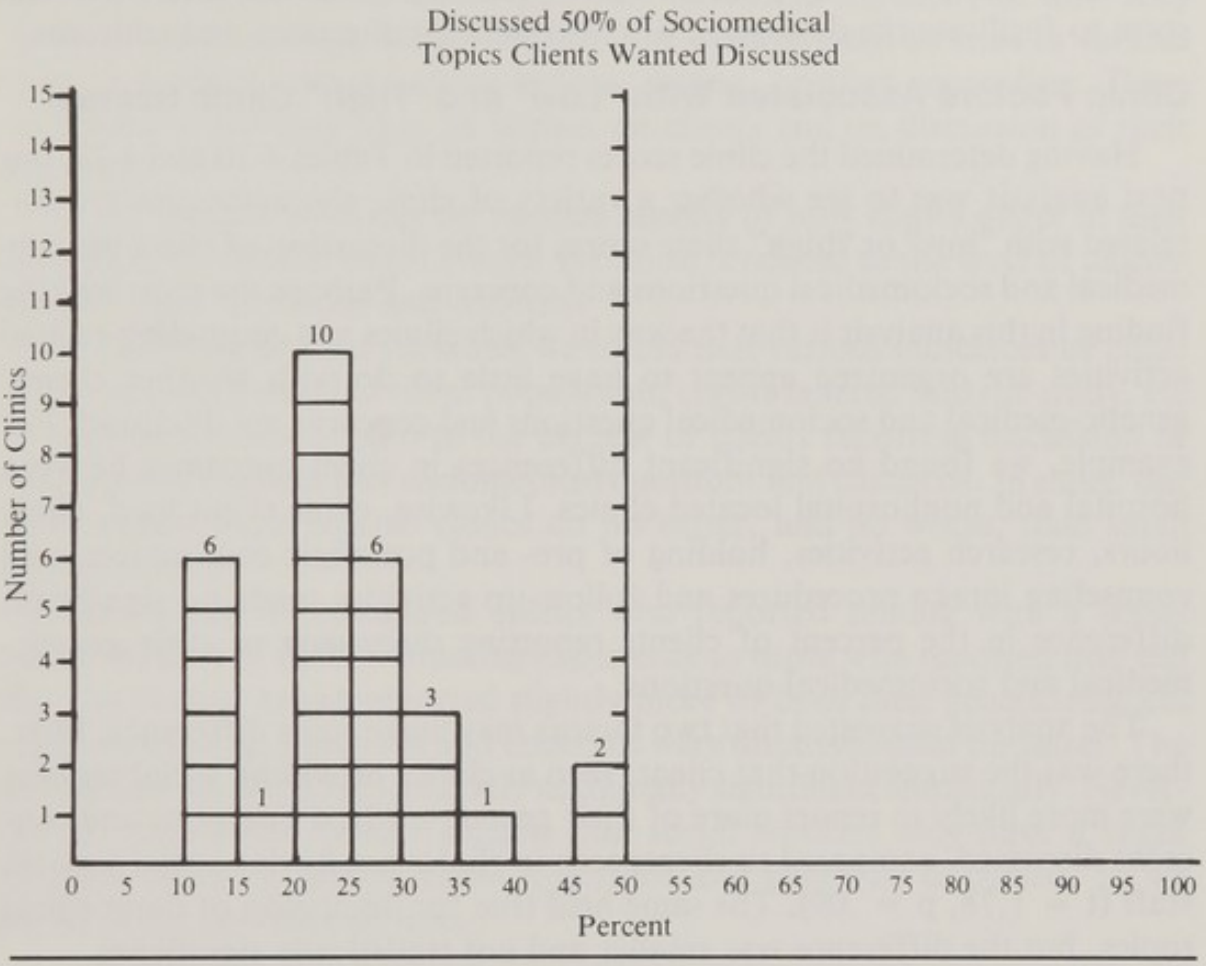
*For number of clinics entering 20 or more clients into study.

Accordingly, we performed an analysis to see if either client volume or client question level could account for the scores clinics obtained. This analysis revealed that neither client volume nor client question level accounted for the clinic's score. The clinics with the lowest scores do not have the largest client populations, nor do they have clients with more concerns or questions than clinics with higher scores.

Finally, it may be recalled from Chapter 3 that clinics varied in the number of clients they entered into the study, some entering very few, while a few entered up to 50 clients. It is the case that the score we develop on the genetic-medical dimension is based on ten items, and the sociomedical score on five items per client. Hence, they should be highly reliable scores, even if a clinic entered only ten clients into the study. Nevertheless, to assure highly reliable scores, we will examine only those clinics which entered 20 or more clients into the study.

Table 4-10 reports the distribution of clinic scores for discussion of client genetic-medical questions and concerns. At four clinics clients reported discussion of 50% or less of their genetic-medical questions and concerns. At the other extreme, at only two clinics did clients report discussion of 70–75% of such questions and concerns. The range of scores thus is from 40–75%, and the norm for discussion is from 55 to 60% of client-specified questions and concerns.

TABLE 4-11. Distribution of Clinic Scores for Discussing With Clients the Sociomedical Topics Clients Wanted Discussed*



*For number of clinics entering 20 or more clients into study.

Table 4-11 reports clinic scores for discussion of sociomedical questions and concerns. It is strikingly apparent that a much lower percent of sociomedical than genetic-medical client questions and concerns were discussed. These tables also show that there is variability among clinics in discussion of client sociomedical questions and concerns as there was in discussion of medical-genetic topics. In Table 4-11 we can see that scores range from a low of 10% to a high of 50% of client sociomedical questions discussed. Scores peak around the 20–25% range.

Clearly, these data show that many client nonmedical questions and concerns are not getting discussed, as we saw earlier. At the same time the tables suggest that clinics vary significantly in their scores. Clearly some clinics do well, while others do very poorly.

It should be noted that there is a statistically significant association between clinics' genetic-medical and sociomedical scores ($r = +.38$, $p = .02$). Within certain limits, this suggests that clinics that score highly on discussion of client genetic-medical questions and concerns also tend to score among the top clinics for discussion of client sociomedical questions and concerns. It is important, of course, to put this observation about the "top" clinics in perspective by recalling the distribution of clinic scores reported in Tables 4-10 and 4-11. Granted this, given that some clinics do comparatively well

and some comparatively poorly, can we identify clinic characteristics that seem to facilitate the discussion of client-defined questions and concerns?

Clinic Factors Associated With "Low" and "High" Clinic Scores

Having determined the clinic scores reported in Tables 4-10 and 4-11, our next analysis was to see whether a variety of clinic characteristics are correlated with "low" or "high" clinic scores for the discussion of client genetic-medical and sociomedical questions and concerns. Perhaps the most striking finding in this analysis is that the way in which clinics and counseling-related activities are organized appear to have little to do with whether clients' genetic-medical and sociomedical questions and concerns are discussed. For example, we found no significant differences in client outcomes between hospital and nonhospital located clinics. Likewise, clinic client load, clinic hours, research activities, holding of pre- and postclinic conferences, and counseling intake procedures and follow-up activities made no significant difference in the percent of clients reporting discussion of their genetic-medical and sociomedical questions.

The analysis suggested that two factors may make some difference. First, there was the suggestion that clients seen at clinics providing social services were more likely to report more of their genetic-medical questions and concerns discussed, compared to clients seen in clinics not having social services staff ($t = 1.78$, $p = .09$). The same held true for discussion of nonmedical topics, but the difference was smaller and not statistically significant.

Second, clients seen at clinics with affiliated parent groups reported a significantly greater number of their medical questions discussed than clients seen at clinics not having a parent group affiliation: ($t = 4.8$, $p = .02$). The same occurred for discussion of nonmedical topics, but the difference was not statistically significant. It may be that the presence of social service staff facilitates the surfacing of client questions and concerns. Likewise, clinic involvement in parent groups may acquaint the professionals with the broad range of questions and concerns of clients, making them more sensitive to their clients' concerns. It could also reflect a generally more open approach to clients by the professionals, an openness that leads to greater recognition and discussion of client problems.

Our analysis, while covering a broad spectrum of clinic characteristics, did not suggest any particular organizational features that seemed to make a large difference in the extent to which clients reported discussion of their questions and concerns. We did not, of course, look at all clinic features, and there could well be clinic characteristics which make an important difference. In this study, however, we were not able to identify any such clinic features.

Counseling Process Factors

While the particular resources available in a clinic may not significantly affect the extent to which clients are able to discuss their questions and con-

cerns, the way in which clinics put these resources together, or how they "process" clients, may. Social workers, for example, can be used in various ways in a clinic, seeing a client before, during, or after counseling. These variations in use may have an impact on clients and on discussion of their concerns.

In this section we focus on various aspects of how clinics differ in their processing of clients, to see if such variation is related to the level of discussion of client questions and concern.

In analyzing process variables we found that various indicators of clinic size — such as estimated client population, clients entered into the study, etc — showed no relationship to the percent of clients reporting discussion of their genetic-medical and sociomedical questions and concerns. In short, our data suggest large volume clinics do no better, and no worse, than small volume clinics.

Also, when we compared clients who reported talking with a social worker as part of their counseling experience to those who reported they did not, the former group reported slightly more of *both* their genetic-medical and sociomedical questions and concerns were discussed than the latter. The differences are not large, and only marginally significant statistically. Nevertheless, the similar results suggest that social workers may have a slight salutory effect in getting client questions and concerns discussed, as we noted in the previous section.

TABLE 4-12. Relationship Between Estimated Length of Counseling Session and Discussion of Client Genetic-Medical and Sociomedical Questions and Concerns

	Estimated duration of counseling session (N = 1,034)*			
Percent of client genetic-medical topics discussed	Under 20 minutes (%)	20–39 minutes (%)	40–59 minutes (%)	60 minutes or more (%)
≤ 25%	40.9	22.2	11.8	11.5
26–74%	35.9	48.8	52.7	50.5
≥ 75%	23.3	29.1	35.5	38.0
	Estimated duration of counseling session (N = 795)†			
Percent of client sociomedical topics discussed	Under 20 minutes (%)	20–39 minutes (%)	40–59 minutes (%)	60 minutes or more (%)
≤ 25%	88.2	66.8	64.5	60.8
26–74%	3.9	13.6	14.2	15.6
≥ 75%	7.9	19.6	21.3	23.6

*$\chi^2 = 50.3$, df = 6, p = .000.
†$\chi^2 = 20.7$, df = 6, p = .002.

When we compared clients who reported talking with a genetic associate as part of their clinic experience to those who did not have such an experience, the former reported somewhat fewer of their genetic medical questions and concerns being discussed in depth in counseling. The difference, while statistically significant ($\chi^2 = 6.2$, $p = .05$), is quite small. No such difference existed for discussion of sociomedical issues.

The strongest factor we found associated with discussion of client questions and concerns was simply *length of time* spent with the counselor (Table 4-12). The data in this table are quite striking. For example, whereas 40.9% of clients reporting a session less than 20 minutes had 25% or less of their medical topics discussed, only 11.5% of those reporting a session of one hour or more had so few of their genetic-medical questions and concerns discussed. For sociomedical topics, when the session was less than 20 minutes in length, only 7.9% of the clients reported that 75% or more of their sociomedical topics were discussed, whereas when the session was one hour or more in length, just under one-fourth reported discussion of as many of their sociomedical questions.

Two important observations can be drawn from Table 4-12. First, there is a striking relationship between the amount of time devoted to counseling and percent of both genetic-medical and sociomedical topics discussed. Second, even when more time was devoted to counseling, there were still significant percents of clients reporting that many of their genetic-medical and sociomedical topics were not discussed in depth. In short, increasing the amount of time devoted to counseling may be instrumental — up to a point. However, it may also be necessary to broaden the knowledge, skill base, or professional resources available in counseling. As we saw, clients frequently reported that the reason their various questions and concerns were not discussed was because the facts were not known or the counselor had to gather information. A different mix of professionals, different training, or resources could significantly alleviate the situation.

Client Characteristics

In attempting to understand what factors facilitated or hindered discussion of client questions or concerns we also examined numerous client characteristics. We have already reported in Table 4-5 variation in the percent of client questions discussed when clients were compared on the basis of reproductive experience. There we saw some variation, especially with respect to discussion of genetic-medical topics.

Many client variables were examined. Of particular interest was the relationship between a client's level of education and discussion of genetic-medical and sociomedical topics. Research on the doctor-patient relationship has shown that patients' level of education commonly is related to

their raising questions and concerns with their doctor: the more educated patients are, the more often they learn and retain what the doctor told them.

With these considerations in mind, we assessed whether clients' level of education was related to their reporting discussion of their medical and nonmedical questions and concerns in genetic counseling. This analysis revealed that there is a small but statistically significant association between a client's educational level and reporting discussion of genetic-medical questions in counseling. For example, of those clients with a high school education or less, 20.5% reported that only 25% or less of their medical questions were discussed. Thirty-one percent reported that 75% or more of their medical concerns were discussed in depth.

Conversely, of clients with a graduate degree or some graduate training, only 11% reported discussion of 25% or less of their genetic-medical questions, and 40% reported that 75% or more of such questions and concerns were discussed. While the differences are statistically significant ($\chi^2 = 11.26$, $p = .05$), they are not large. Thus, while a client's education may make some difference in getting medical questions discussed, it does not make a substantial difference.

Interestingly, when we look at client education and discussion of nonmedical topics in counseling, there is no relationship. The more educated are no more likely than the less educated to get their nonmedical questions and concerns discussed in genetic counseling.

In short, with the exception of client education, which appeared to make only a small difference, of the various client characteristics we examined, none seemed to affect the level of discussion of client questions and concern. To the degree that discussion of only 55% of clients' genetic-medical and only 15% of their sociomedical questions is viewed as problematic, there is a problem in contemporary counseling in meeting client needs.

Counselor Factors

Finally, we turned our attention to how a counselor's training and experience relate to discussion of the questions and concerns clients bring to genetic counseling. We examined a large number of counselor characteristics, including type of degree, medical specialty, experience, and counseling attitudes. It is relatively easy to generate a series of hypotheses about how such factors as training and attitudes toward counseling may condition the nature of genetic counseling, including the degree to which clients discuss a wide array of their questions and concern.

The analysis showed that the type of degree — MD, PhD, MS, etc — that a counselor held was not related significantly to clients reporting discussion of their genetic-medical and sociomedical questions and concerns. We also found that there was no significant difference among medical specialties

with respect to clients reporting discussion of their sociomedical questions and concerns.

In Chapter 3 (see Tables 3-12—14) we examined various attitudes counselors can hold toward their counseling. One could hypothesize that the more favorable counselors' attitudes toward the "counseling" aspect of their work, the more likely their clients would be to report discussion of a larger number of their questions and concerns, genetic as well as sociomedical.

Interestingly, an analysis revealed that the level of commitment a counselor held toward a variety of tasks in counseling, such as helping clients cope, reducing client guilt, assisting clients in comprehending their options in a situation, and so forth, showed no systematic or significant relationship to the degree to which clients report discussing either their genetic-medical or sociomedical questions and concerns. In short, clients seen by counselors strongly committed to the various counseling tasks shown in Tables 3-12—14, tasks which reflect many of the genetic and sociomedical questions of clients, were no more likely to have discussed such concerns than were clients seen by counselors who were less strongly committed to these counseling activities and objectives. This is an important observation, for it suggests that simply knowing one is committed to a particular counseling philosophy or strategy is probably not sufficient to assure that those policies will in fact be expressed in practice. Many factors shape what transpires when a layman and a professional meet, at least in the case of genetic counseling, and even strong commitment to some important counseling objectives is not sufficient enough to assure that one's counseling will reflect these commitments.

The only additional observation our analysis of counselor characteristics disclosed was that counselor training, specifically in counseling techniques, resulted in clients reporting discussion of slightly more of their genetic-medical questions and concerns ($\chi^2 = 6.3$, $p = .04$). Interestingly, this training was not related to discussion of client sociomedical questions and concerns.

Finally, it is reasonable to assume that with experience, a professional should become more adept at understanding their clients and should be able either to sense or actually elicit from the clients their questions and concerns. Accordingly, it would seem that clients who are seen by more experienced counselors might report a larger percent of their questions and concerns discussed than clients seen by less experienced counselors.

We tested this hypothesis by looking at the relationship between the number of years a counselor has been providing counseling and the percent of genetic-medical and sociomedical questions clients reported they discussed. Interestingly, no relationship exists for discussion of either genetic-medical or sociomedical questions or concerns. Apparently, in the case of the counselors in this study, practice not only does not make perfect, it does

not appear to make counselors better at this aspect of genetic counseling, if one uses discussion of client questions and concerns as an index of effective counseling.

In summary, we have examined several classes of variables, in terms of how they relate to the discussion of various genetic-medical and sociomedical questions and concerns clients bring to counseling. We found that clinics vary markedly in degree to which their clients reported discussion of their questions. The only variable we found related to such discussion was simply time spent with clients, but even when an hour or more was spent, many questions were not discussed. Moreover, the way in which clinics processed clients and even the mix of professionals used seemed to make little difference, although social workers seemed to facilitate discussion somewhat. Finally, we looked at both client and counselor characteristics, and found little to understand why some issues are discussed and others are not. Educated clients are only marginally more likely to have their questions discussed. Experienced counselors did not seem to do any better at helping clients discuss issues than inexperienced counselors. Even strong counselor attitudinal commitment to the "counseling" aspects of genetic counseling does not seem to make any significant difference in what gets discussed in counseling. We will discuss these observations further at the end of this chapter.

CLIENTS' SIX-MONTH ASSESSMENT OF THEIR GENETIC COUNSELING EXPERIENCES

In attempting to assess whether genetic counseling meets clients' needs it seemed important to follow-up clients after they had had a chance to reflect on their counseling experience. Several items on the "Six-Month Follow-up Questionnaire" are useful as indicators of how clients feel about counseling after some time has elapsed.

When asked how satisfied they were with their counseling, 78.4% of female clients indicated that they were either satisfied or very satisfied. Just over 7% felt dissatisfied or very dissatisfied. The remaining clients responded that they were only somewhat satisfied with the counseling they had received.

One might expect that if the initial counseling session left clients with questions and concerns, they would want to see a genetic counselor again. Table 4-13 shows that six months after their counseling, 20.8% do indeed want to receive more counseling. It is particularly interesting that just under a third of all clients are not sure whether they want more counseling. It would seem that this group of respondents could be experiencing at least two types of uncertainty. First, there are those who, even six months after counseling, do not know what to make of the information they received in

TABLE 4-13. Female Clients' Desire for More Genetic Counseling Six Months After Initial Counseling Session*

	Females (%)
Yes	20.8
No	47.6
Unsure	31.7
N	790

*Based on the number of clients who responded to the question.

counseling. It is still not clear to them how to use this information, and so they are not sure whether they need to talk to a counselor again about their questions and concerns. The second type of uncertainty which some clients may experience could come from not knowing whether counseling can effectively help with the problems and concerns which they have. These may be things that were not dealt with in their initial counseling session, or perhaps new issues that have arisen as a result of counseling, that is, concerns which have transpired in the six months since counseling.

We can only speculate why a significant number of clients are uncertain about wanting more counseling at six months. We did ask clients wanting to see a counselor why they wanted to. Although the number of responding clients is relatively small, their responses, reported in Table 4-14, are of considerable interest in addressing the question of whether or not counseling meets clients' needs. About 24% of responding females said that either the counselor had not answered all of their questions or that they had not understood some of the information they received during counseling. Similar numbers report not asking all of the questions they had. In addition, 17.1% had forgotten some of the information received during counseling. Clearly, all of these clients still have questions or concerns that either were not dealt with in their initial counseling sessions or were not dealt with in an effective way.

About half of the clients who wanted additional counseling gave other reasons than those provided in Table 4-14. New questions or concerns that had come up since counseling was the most common of these reasons. Many wanted to know more about a disorder, especially whether there were any new findings, such as the development of prenatal or carrier tests. Others had learned new family or medical information that they thought might change or clarify a diagnosis. Still others wanted information about how to care for an affected child, or medical treatment for such a child. Some stated

TABLE 4-14. Client Reasons for Wanting Additional Genetic Counseling*

Reason	Females (%)
I have forgotten some of the information I received during counseling.	17.1
I did not understand some of the information I received during counseling.	14.0
The counselor did not answer all of my questions.	9.8
I did not ask all of the questions I had.	23.2
I have had a child with a birth defect or genetic disorder since the counseling session.	3.0
I am (my wife is) pregnant or trying to become pregnant.	20.1
Other reasons given.	56.1
N	164

*Includes only those clients who indicated they were sure they wanted additional counseling. Each client may have given more than one reason.

TABLE 4-15. Topics Clients Sought Information About After Counseling

Topic	Percent discussed	N
Care of affected child	61.6	177
Etiology	55.0	218
Treatment	51.7	321
Disorder characteristics	48.9	90
Recurrence risk	45.3	64
Prenatal diagnosis	45.1	184
Prognosis	37.1	217

that they had come to recognize that their child had more problems than they at first realized.

Clients at the six-month follow-up were asked what topics they had sought information about since counseling. Table 4-15 presents data on these topics. Just over 60% of the clients who came with questions regarding care for an affected child, questions not discussed in counseling, had sought information from such people as their family doctor, friends, relatives, or even books by the time of our six-month follow-up. Almost as many, percentagewise, sought information on the etiology of a disease and treatment concerns. Interestingly, less than half followed up on questions involving recurrence risks, prenatal diagnosis, and prognosis. It would appear that, other

things being equal, the more pressing the questions, care and treatment concerns, for example, the more likely clients are to exercise some individual initiative in obtaining information sought but not obtained in counseling. It also appears that significant percents of clients do not, at least by six months, obtain information on questions of concern to them, including some very important questions such as recurrence risks and disease prognosis.

SUMMARY

This chapter has examined the variety of medical and nonmedical questions and concerns that clients brought to the genetic counseling clinics in this study. Clearly, clients have a wide variety of questions and concerns, and generally bring more genetic-medical than sociomedical topics to counseling.

Our analysis revealed that clients are more likely to report discussion of their genetic-medical than sociomedical concerns. Also, we found that when asked to identify the major concern of their clients, counselors had difficulty doing so except in those cases where clients wanted to learn their chances for having a child with a birth defect or where clients were interested in obtaining a prognosis.

An analysis of factors that may facilitate or hinder discussion of client questions and concerns revealed that, with one important exception, clinic organizational characteristics and the way in which clinics organize delivery of their counseling services did not appear to make any significant differences. The one clearly important variable was time: the more time counselors spent with clients, the greater the number of genetic-medical and sociomedical topics clients reported they discuss. It would appear that a major factor contributing to the lack of discussion of some client concerns may be that many counseling sessions are just too short. However, other factors must be contributing to this low level of discussion as well, such as lack of knowledge and training or inappropriate professional resources, since even when an hour or more is spent in counseling many questions and concerns remain undiscussed.

Our analysis revealed that the educational level of the client and the training and experience of the counselor made virtually no difference in the extent to which the client reported discussing questions and concerns in counseling.

Several observations can be drawn from the six-month follow-up data. We have seen that six months after their initial counseling session about three-fourths of female clients report being satisfied or very satisfied with counseling, while only a small percentage were dissatisfied. When asked whether they would like to receive more genetic counseling, about half the responding clients say no. Of the remaining clients, just over 30% were unsure whether they would like more counseling or not. It would seem that

these people may still be trying to assimilate the information they received, or may have questions and concerns about which they are not sure counseling can help.

The reasons given by clients who were sure they wanted more counseling reveal that most of these people might benefit from seeing a genetic counselor again. This may be either because questions and concerns which they had brought to counseling had not been effectively dealt with, or because new needs had arisen since counseling.

Finally, as we look at the data reported in this chapter, and consider effective counseling in part to consist of providing a context in which clients can discuss their various questions, worries, and problems, it is clear that there is room for significant improvement in this aspect of genetic counseling. We will return to this topic in Chapter 7.

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Chapter 5

Effective Genetic Counseling: More Informed Clients*

INTRODUCTION

Clients come to genetic counseling with a variety of questions and concerns, among them, questions about medical problems, answers to which they have been unable to obtain elsewhere. As the field of genetic counseling has grown over the past decade different approaches to counseling have been presented and discussed in the clinical genetics literature. Some of these approaches differ because their proponents disagree about the ultimate goals of counseling. Other vary in the emphasis given to certain components of the genetic counseling process. Regardless of such differences, however, there is virtually universal agreement that one of the essential tasks of genetic counseling is client education [2]. Subsequent to counseling, clients should know more of the medical and genetic facts of their particular problem than they did when they first sought this specialized service. Such information can be useful to clients in a number of ways, but a basic goal of counseling is to enable clients to make informed family and reproductive decisions.

While clients come to counseling expecting to have the state of their knowledge changed, they may also experience another type of change as a result of their counseling session — a change in the way in which they perceive the problem or disorder that brought them to counseling. Client attitudes about the risk and burden associated with a disease or disorder may be as important in determining their behavior, as the state of the factual or "rational" knowledge about the problem. In fact, there is some evidence to indicate that factors other than "rational" knowledge may be very important to clients in making future reproductive decisions [3].

*This is a revised and expanded version of "Client Learning of Risk and Diagnosis in Genetic Counseling" [1].

Although clients may not have consciously posed such questions as "What does this disorder mean for me and my family?" and "How will it affect our lives?" these are certainly important issues for them to resolve. Whether, and to what extent, counseling should directly deal with such issues is to some degree more controversial than the basic function of teaching factual information. Nevertheless, we felt that it would be useful, while studying clients' factual learning, to note the impact of counseling on client attitudes and perceptions about the disorder they confront.

As in the previous chapter, we report data primarily on female clients in this chapter. Male client data are generally comparable to female data on all analyses reported in this chapter. Exceptions are noted in the text.

While many types of information may be transmitted during a counseling session, typically the issues of diagnosis and risk are of central interest to both the counselor and the client. Diagnostic knowledge is useful not only because it provides the client with a category or label to attach to their problem, but also because it enables clients to gather additional information. By knowing the name of a disorder a client can discuss the problem with a variety of medical care providers, and even seek additional information about the disease or disorder in a library or bookstore. Knowledge of the risks a client faces, whether for continued miscarriages, abortion, or for a specific disease or disorder is, of course, extremely valuable. It enables clients to put into perspective their problem and to begin to judge, from their own vantage point, whether or not to have children.

It is the case, of course, that "informed family and reproductive decisions" entail considerably more "factual" information than simply diagnostic and risk knowledge. Nevertheless, comprehension of these two central aspects of genetic counseling is necessary if clients are to be fully informed.

For these and other reasons we decided to focus attention in our study on the diagnostic and risk knowledge clients brought to counseling, and the knowledge, correct and incorrect, they took from counseling. If counseling is effective, in a very simple but basic sense, clients must know more, diagnostically and risk wise, after counseling than before.

CLIENT DIAGNOSTIC KNOWLEDGE

Client knowledge of the diagnosis of the disorder or medical problem for which they sought counseling was ascertained before, after, and approximately six months postcounseling. A client's diagnostic knowledge was evaluated by asking the individual to name and/or describe the medical problem or disorder for which she had sought genetic counseling. To assess accuracy, the client's responses were compared to the diagnosis or problem name provided by the genetic counselor. In determining the extent of

agreement between client and counselor, both the name and the description of the problem or disorder provided by the client were taken into account, allowing us the greatest possible insight into the client's understanding of the nature of the condition for which counseling was being obtained.

As noted in Chapter 3, two or more disorders or problems were involved in approximately 20% of the cases entered into this study. In these cases the scoring procedure, discussed below, involved judgments regarding the client's knowledge of both problems or diseases. Very few cases involved three or more problems, and when they did, our scoring procedure was limited to assessing only the first two disorders listed.

The accuracy of clients' knowledge was judged by comparing their diagnoses with the diagnosis the counselor provided after seeing the client. Clients' responses were judged as accurate, marginally accurate, or inaccurate.

For a client's diagnosis to be scored as accurate, the correct name or description of the disorder was required. Differences in terminology were acceptable (for example, a counselor's report of "Down syndrome" with a client's response of "mongolism"), as were client descriptions of a disorder in the absence of its correct name ("missing brain" for "anencephaly").

Marginally accurate diagnoses were those which clients did not provide as specific a diagnosis as the counselor, or where the information provided by the client was correct but too general. For example, a client reporting a "heart disorder" in the face of a counselor's diagnosis of "mitral valve defect" was judged to be marginally accurate.

When clients provided an extremely vague diagnosis, such as merely a "birth defect," or when they gave a garbled response which could not be interpreted, the response was scored as inaccurate. This score was also given when the diagnosis or description given by a client was completely different from that reported by the counselor.

Because of the judgmental nature of designating a client's response into one of these three categories, all comparisons between counselors' and clients' diagnoses were scored by two judges. Using this system, the reliability in scoring clients' diagnoses was found to exceed 90%. In those cases where agreement on scoring could not be reached by the coding staff, the final decision was made by the project coordinator (a genetic associate) and/or the project director (a physician).

Table 5-1 reports the accuracy of female client diagnostic knowledge at the three data gathering periods in this study. As we can see, almost 60% of the female clients came with accurate diagnostic knowledge, and an additional 18% were marginally accurate. Approximately 20% were scored as inaccurate, the largest number of this group giving an incorrect name or description of their disorder or disease. As one looks at client knowledge

TABLE 5-1. Female Clients' Knowledge of Diagnosis Before, Immediately After, and Six Months After Counseling

Client Diagnostic Knowledge Status	Before T_1 (%)	After T_2 (%)	Six Months T_3 (%)
Accurate	57.6	67.6	70.3
Marginally accurate	18.2	16.0	14.8
Inaccurate	19.6	12.0	10.2
(Incorrect)	(8.1)	(7.2)	(4.0)
(Client did not know)	(6.7)	(2.6)	(2.5)
(No client response)	(4.8)	(2.2)	(3.7)
No counselor diagnosis given*	4.5	4.5	4.7
N	1,097	1,097	806

*The primary reason for no diagnosis was that the counselor had to obtain more medical/genetic information.

just after counseling, there is an increase of about 10% in the clients scored as accurate, a slight decrease in the percent marginally accurate, and a drop of about 8% in those inaccurate, most of the latter change due to clients giving correct diagnoses postcounseling when they initially said they did not know.

The relatively high level of accurate diagnostic knowledge prior to counseling (T_1) is not surprising when it is remembered that the genetic counseling studied here was largely referral based, constituting a type of tertiary medical care. As such, we would expect that many clients had already had significant contact with medical providers regarding their problem. In such contacts we would expect many to have acquired some understanding of their problem, including a diagnosis.

Table 5-1 also shows that of those clients contacted at the six-month follow-up, the proportions of accurate, marginally accurate, and inaccurate responses remained quite similar to those obtained immediately after counseling (T_2). There does not appear to be, in other words, any deterioration or significant decay in client diagnostic knowledge across the time period studied.

In short, the data in Table 5-1 show that there is approximately a 10% increase in accurate knowledge in the counseled population but just over 10% remain inaccurate, even though they had been given a diagnosis as part of their genetic counseling.

Table 5-1 provides only partial information on stability and change in client diagnostic knowledge. Table 5-2 reports the data shown in Table 5-1

in a slightly different form, allowing us to examine client diagnostic accuracy after counseling in terms of their knowledge state prior to counseling.

The data presented in Table 5-2, while based on data reported previously, have been computed somewhat differently than shown in Table 5-1. First, since most of the change in client diagnostic knowledge occurs between the before and after period, and client knowledge at six months appears very comparable to postcounseling knowledge, we report only before-after diagnostic knowledge. Second, in most cases the distinction between "accurate" and "marginally accurate" is simply a difference in specificity or degree, not in the quality of knowledge. In most cases, we suspect, clients who can remember the general nature or name of their problem will find this as useful as when the client can be precise. For this reason in Table 5-2 the accurate and marginally accurate categories have been combined to simplify the analysis.

Table 5-2 shows that, of the 847 clients coming into counseling with accurate knowledge, 805, or 95%, had accurate knowledge postcounseling, while 42, or 5%, were scored as inaccurate. This included 5 clients who gave completely incorrect answers, 21 who gave extremely vague or garbled responses, and 16 who left their response category blank.

TABLE 5-2. Comparison of Female Clients' Knowledge of Diagnosis Before and After Counseling*†

Client Diagnostic Knowledge Status		Before T_1		Totals After T_2
		Accurate	Inaccurate	
After T_2	Accurate	95.0	59.7	88.3
	Inaccurate	5.0	40.3	11.7
Totals Before T_1		80.8	19.2	N = 1,048

*Of the 1,097 cases with a female client member entered into the study, a diagnosis was given in 1,048 cases, or 95.5%.

†To test the null hypothesis — that genetic counseling had no effect on client learning — we first categorized clients at T_2 as either changed or not changed from their T_1 knowledge state. Then, looking only at those who changed, we compared the percent who lost knowledge to the percent who gained knowledge. $\chi^2 = 38.5$, df = 1, $p < 0.01$. Hence, we can conclude that genetic counseling resulted in a statistically significant, positive increase in clients' diagnostic knowledge.

Turning to clients who came to counseling with inaccurate knowledge, of those given a diagnosis, 60% reported it accurately after counseling, while 40% (or 81 clients) did not. Of these 81 clients, 10 reported a completely incorrect diagnosis, 44 had an extremely vague or garbled response, and 27 left the response category blank, although the counselor had given them a diagnosis.

In short, genetic counseling, at least from the perspective of informing clients about their diagnosis, appears to educate a substantial proportion of those clients who are ignorant about a diagnosis. However, a full 40% of such clients by our scoring method appear ignorant diagnostically after counseling. At the same time, there does not appear to be any significant drop in the accuracy of diagnostically knowledgeable clients during counseling, although a few do appear less accurate after counseling than before.

CLIENT KNOWLEDGE OF RISK

Clients come to counseling to discuss many issues. One of the most frequent reasons cited by clients in this study was to obtain information about their chance of having a child with a birth defect. Sixty-three percent of female clients said they came to counseling to discuss this issue.

As was the case with diagnostic knowledge, clients' knowledge of the occurrence/recurrence risk for the problem for which counseling was being provided was ascertained at three points in time: immediately before the session, soon after, and six months later. We requested clients to express the risk as numbers or as a percent. Counselors were asked on their postcounseling questionnaire to indicate the precise risk they provided to clients. All client and counselor risk figures were converted to percentages for client/counselor comparisons.

Client knowledge of risk has been termed "accurate" if it agreed with the risk counselors indicated they gave during the session. When the risk given by clients did not exactly correspond with that noted as given by the counselor, the response was scored as "inaccurate." Thus, we set a high standard for assessing the accuracy of client risk knowledge, since even small discrepancies resulted in a client being judged as inaccurate.

While both counselors and clients were asked to provide numeric risk figures, this did not always occur. When a counselor stated that he or she had given the client a particular numeric risk and the client reported a nonnumeric risk (such as "low" or "very high") had been given, the client's response was scored, for the purpose of this analysis, as inaccurate. In all cases where the counselor gave a nonnumeric risk, indicated that no risk was given to the client, or left the question blank, it was not possible to evaluate client knowledge of risk. This was the case even when the client provided numeric risk figures.

Table 5-3 provides a view of client's knowledge of risk at the three points in time at which it was ascertained. Cases where counselors provided us with a nonnumeric risk are included in the group "no counselor numeric risk."

Table 5-3 shows that in almost 57% of all cases in this study, it was not possible to assess the accuracy of a client's risk knowledge, even if they reported a figure. This was due to several factors, including the counselor needing to obtain more information, the counselor not reporting to the study the risk reported to clients, and, in almost 15% of all cases, the counselor reporting a nonnumeric risk to clients. We mean by this that the counselor reported to us that he or she gave the client not a percent or chance figure, but instead a statement as to the magnitude of the risk, that is, the risk was reported as "high," "moderate," or "low." We will return to the topic of nonnumeric risks below.

In addition, analysis of the cases for which no risk was reported showed that among the most commonly represented diagnoses or problems were "multiple miscarriages," "Down syndrome," "multiple congenital anomalies," "delayed development," and "mental retardation." Since all of these conditions are nonmendelian in inheritance pattern and can have multiple etiologies, it would seem that the counselors in these cases simply may not have felt comfortable giving or reporting risks, or may have found it difficult to establish a reliable risk estimate. A great many of the problems for which clients seek genetic counseling fall into this category as shown in Table 3-21.

TABLE 5-3. Female Clients' Knowledge of Risk Before, Immediately After, and Six Months After Counseling

Client Risk Knowledge Status	Before T_1 (%)	After T_2 (%)	Six months T_3 (%)
Accurate	5.6	21.9	19.1
Inaccurate	37.7	21.3	26.8
(Incorrect)	(5.7)	(12.6)	(15.6)
(Client did not know)	(28.6)	(7.2)	(9.1)
(No client response)	(3.4)	(1.5)	(2.1)
No counselor numeric risk given*	56.8	56.8	54.1
Numeric Risk*			
N	1,097	1,097	806

*The counselor provided a nonnumeric risk 14.8% of the time; provided the client with a numeric risk but did not report it for the study in 14.7% of the cases; and in 27.3% of the cases, no risk was given or the counselor failed to indicate whether or not one was.

Table 5-3 shows that at entry to counseling many fewer clients knew their risk than knew their diagnosis, approximately 6% compared to 58%. The data also show that, at the population level, there is a marked increase in the number of clients with accurate risk knowledge from before to after counseling, a change that has decreased only slightly at the six-month follow-up.

An examination of the data shows that there is a large drop in the percent of clients from before to after counseling who say they do not know, and an actual increase in the percent reporting an incorrect risk figure. This undoubtedly reflects, in part, our strict criterion for judging accuracy, namely, complete agreement between the counselor's and client's risks.

Table 5-4, similar in design to Table 5-2, looks at changes in client risk knowledge from before to after counseling in terms of the client's initial knowledge state. As can be seen, among those with accurate risk knowledge before counseling, 83.6% were still accurate after counseling. Among the 16% who were inaccurate, in seven cases the client gave an incorrect response or reported they had not been given a risk or did not remember it, and in three cases they reported a nonnumeric risk or left the response category blank. It is worth emphasizing that in this table we are reporting only those cases where the counselor specifically reported giving the client a numeric risk.

TABLE 5-4. Comparison of Female Clients' Knowledge of Risk Before and After Counseling*†

Client Risk Knowledge Status		Before T_1 Accurate (%)	Before T_1 Inaccurate (%)	Totals After T_2 (%)
After T_2	Accurate	83.6	45.8	49.4
	Inaccurate	16.4	54.2	50.6
Totals Before T_1		12.9	87.1	N = 474

*A numeric risk was given by the counselor to the client in 474 of the 1,097 cases, or 43.2% of the time.

†To test the null hypothesis — that genetic counseling had no effect on client learning — we first categorized clients at T_2 as either changed or not changed from their T_1 knowledge state. Then, looking only at those who changed, we compared the percent who lost knowledge to the percent who gained knowledge. $\chi^2 = 68.4$, df = 1, $p < 0.01$.

Of the clients coming to counseling with inaccurate risk information, 45.8% correctly reported their risk after counseling. However, the majority, 54.2%, did not. Among these 54.2% (or 224 clients), the majority (133 clients or 59%) gave a numerically incorrect answer after counseling, 20 clients said they did not remember the risk given them, 30 reported a nonnumeric risk, and 41 left the response form blank or claimed the counselor had not given them a risk.

There was a good deal of variation in the magnitude of the discrepancies between clients' inaccurate responses and the risk figures reported by their counselors. The errors made by clients who underestimated their risk either before or after counseling ranged from 0.5–50%. Those who overestimated their risk before counseling did so by anywhere from 0.5–88%. After counseling their errors ranged from 0.5–57%.

It is interesting that more clients overestimated than underestimated their risk, both before and after counseling. Before counseling 32% overestimated their risk, while after counseling 24% did. Nineteen percent underestimated their risk prior to counseling, while 13% did so after.

Subsequent to counseling there was also a drop in the magnitude of the errors made by clients compared to before counseling. As noted in Table 5-5, of those clients with incorrect risks, 34.8% underestimated the risk prior to counseling by less than 5%, while after counseling 51% did. For errors of overestimation, 22.5% erred by 5% before, while 52.8% did so after counseling. Thus, there was a movement toward more accurate estimates, from before to after counseling, among both over- and underestimators.

In summary, it would appear that, for a substantial fraction of clients without knowledge of risk or with incorrect risk information, counseling is

TABLE 5-5. Magnitude of Errors in Risk Estimation by Female Clients Before and After Counseling

	Errors in Underestimation		Errors in Overestimation	
Magnitude of Error	Before (%)	After (%)	Before (%)	After (%)
< 5%	34.8	51.0	22.5	52.8
5–10%	26.1	18.4	12.5	11.2
>10%	38.1	30.6	65.0	36.0
N*	23	49	40	89

*Includes only cases in which both the client and the counselor reported numeric risk figures and there was a discrepancy.

effective in teaching numerically accurate risk figures. It should be emphasized that we have used the strictest possible definition of accuracy here. Were we to have employed a less stringent standard, somewhat greater rates of learning would have been observed.

On the other hand, it is also evident from these data that there is considerable room for improvement in informing and educating clients as to their risks. The data also suggest risk figures appear to be somewhat prone to a decrease in precise accuracy with passage of time. Returning to the proposition that genetic counseling should result in fully informed clients, and granted that knowing one's recurrence risk is an important part of being fully informed, there is a need for improving the educative aspects of genetic counseling.

FACTORS ASSOCIATED WITH ACCURATE CLIENT KNOWLEDGE POSTCOUNSELING

Our analysis of the level of accurate client diagnostic and risk knowledge after counseling suggests that efforts are needed both to improve the presentation or teaching of such information, as well as to facilitate client learning of such facts. In an effort to provide some commentary on how this may be accomplished, in this section we look critically at clients who came to counseling with either inaccurate diagnostic or risk knowledge, and try to identify factors that differentiate those who knew these facts after counseling from those who did not. As in the previous chapter, we examine a large number of factors, including characteristics of clinics, the process of counseling, as well as attributes of the counselors and the clients.

Clinics

In Chapter 4 we developed a score for each clinic in terms of the percent of clients who wanted to discuss an issue and who in fact reported after counseling that the topic was discussed. This score enabled us to describe clinics with high scores and those with low scores, and perhaps identify clinic features associated with high scores.

We wanted to use the same procedure in terms of client learning of diagnostic and risk information, but this proved not to be possible statistically for two reasons. First, to study the acquisition of knowledge by clients, we must first exclude from our analysis all those clients who come knowing their diagnosis or risk. While this was no problem for studying the learning of risk figures, it will be recalled that approximately 80% of the clients came to counseling with accurate or marginally accurate diagnostic knowledge. This reduced our sample of "learning eligible" clients to about 200, dispersed among 47 clinics. The number of clients per clinic, on

average, was thus about only 4, too small a number for reliable statistical analysis.

As for client acquisition of risk information, we had the fact that a numeric risk figure was given in only 474 cases entered into the study. This means that on average we had at maximum 10 clients per clinic to study, again a number too small for reliable statistical analysis.

Because of these reasons we did not analyze individual clinic scores; however, we did compute such scores. With the above comments about the unreliability of individual clinic scores in mind, these scores showed marked variations from clinic to clinic in terms of the percent of clients who left the clinic knowing their diagnosis and risk. The individual clinic scores ranged from 0 to 100% of clients who could have learned their diagnosis and actually knew it postcounseling, with 41 of the 47 clinics having a score of 50% or less of such clients knowing their diagnosis postcounseling. The range in clinic scores of client knowledge of risk ranged again from 0 to 100%, with 29 of 47 clinics having a score of 50% or less. These data must be viewed cautiously, and beyond this simple description, we will not provide additional analysis.

While it was not possible to compute reliable individual clinic scores, we could still examine, across all clients and clinics, the factors that were related to client learning. To accomplish this, the following analysis excludes those clients who came to genetic counseling with accurate diagnostic or risk knowledge and retained their accuracy after counseling. Obviously, such people were not "eligible" to learn something they already knew. The groups on which we based our analyses are composed of those who came to counseling not knowing, or with incorrect information about their risk or diagnosis. Also included are the small number of clients who came to counseling with accurate diagnostic or risk knowledge but appeared not to know it postcounseling. All of the clients to be examined were provided with diagnostic or risk information as part of their counseling sessions. Some learned and others did not. How do the clients who could have accurate diagnostic knowledge or risk information postcounseling, but did not, differ from those who did? Were they counseled differently or by different kinds of counselors; were they possibly different kinds of clients to begin with?

From clinic organizational data collected early in the study we knew that there were differences in the way in which clients were processed at various clinics, and variation in the types of professionals with whom they had contact. Aware of these differences and the variation in learning at the participating clinics, we were surprised to find that the clinic structural variables we examined, as well as the way in which clients were processed,

made almost no difference in client learning of risk or diagnostic information. A few minor observations were made, however. For example, of female clients eligible to learn diagnosis, 38.2% of those counseled in a hospital-based clinic attained full diagnostic accuracy after counseling, compared with 27.0% counseled in clinics located outside a hospital environment ($\chi^2 = 3.9$, $p < .05$). While a similar trend was observed for the learning of risk (47.6% of those counseled in a hospital attaining full accuracy vs 39.4% counseled elsewhere), the difference was not statistically significant.

Clinics whose directors report that a social worker was usually present during counseling appear to be more effective in teaching risk than those whose directors report that a social worker is occasionally or never present: of the former group, 60.3% of eligible patients learn the correct risk through counseling vs 44.3% of the latter group ($\chi^2 = 4.4$, $p < .05$). The strength of this finding is bolstered somewhat by the nonstatistically significant trend observed ($\chi^2 = 4.9$, $p = .10$) that clinics providing social services on site are more effective in teaching risk (54.1%) than those whose directors report that social service problems are referred elsewhere (42.1%) or are "not handled" (45.1%). Similar but nonsignificant statistical differences were detected for the learning of diagnostic information. This finding is of particular interest in light of the data reported in Chapter 4 on the positive effects of social work personnel in meeting both clients' genetic-medical and sociomedical needs.

A host of other organizational features of clinics were studied. These variables failed to yield statistically significant or consistent associations with client learning.

Turning to more process-oriented variables, we looked in some detail at whether the handling of clients in different ways seemed to facilitate or hinder client learning. For example, at some clinics clients routinely have contact with non-MD professionals, most commonly genetic associates. Our analysis showed that talking to one of these professionals, in addition to the MD counselor, did not lead significantly to more or less client learning. In addition, whether the client attended counseling alone or was with a spouse or other relative led to no appreciable increase or decrease in client learning.

We also found that clients who reported being helped with their personal concerns in counseling were no more likely to learn their risk and diagnosis than those who were not so helped. This is interesting because we might expect that if not discussed such concerns could interfere with a client's ability to learn the facts about the problem for which they sought counseling. Likewise, when counseling raised new concerns for a client, he or she was as likely to learn risk and diagnostic information as other clients for whom no new concerns were raised. On the other hand, being able to

discuss most or all of one's medical questions and concerns during a counseling session did seem to make a difference in client learning. More specifically, a significantly larger percentage of clients who reported having such discussion learned their diagnosis than did clients who discussed some or none of their medical concerns (56.6% vs 39.7%, $p < .05$). Although not statistically significant, a similar relationship was observed between discussing such concerns and learning risk information (46.5% vs 38.1%, ns).

Finally, the analysis also showed that the amount of time spent in counseling was not related to the accuracy of a client's knowledge. Clients counseled in short periods of time were as likely to know their correct diagnosis and risk as those who participated in lengthy sessions.

In short, our analysis of clinic characteristics and of the processing of counseling clients does not lead to many suggestions as to how to improve client education. There is, however, value in noting that discussion of medical concerns, achieved for about one-half of the study population, seems to facilitate learning. There is also merit in realizing that simply spending more time in counseling will not necessarily lead to better client education. The problem is obviously more complex.

Counselors

Our analysis of counselors and their impact on client learning has produced some interesting results. For example, there was no significant difference in the levels of diagnosis or risk knowledge among clients counseled by the various types of genetic counselors involved in the study. In other words, medical doctors were as effective as PhDs, who were as effective as genetic associates, professionals specifically trained to provide counseling. In other words, the specific professional degree and its associated training did not relate to significant differences in clients' diagnostic and risk knowledge.

An analysis of the various counselor attitudes reported in Chapter 3 regarding their counseling showed that neither the goals nor the orientation of a counselor to his or her counseling was related to client learning. This included counselors who strongly endorsed client education as one of their goals or main objectives in counseling.

Perhaps the most surprising observation from our analysis of counselor characteristics and their relationship to client learning was that established, experienced counselors, those with multiple years of counseling experience, had virtually the same proportion of clients leaving counseling ignorant as to their risk and diagnosis, as did younger, inexperienced counselors. In other words, at least in this situation, more experience does not seem to be related to more effective counseling, if one uses the teaching of risk and diagnosis as an indicator of counseling effective-

ness. This may be due, in part, to the fact that as providers of a type of tertiary medical care, counselors do not usually follow up their clients, nor do they usually have any opportunity, other than their single encounter with a client, to assess how effective they have been as an educator. Genetic counseling, at least that counseling studied here, provides a poor learning environment for the professional. Learning whether one is doing a good or poor job professionally requires feedback, and in the case of one's effectiveness as an educator, feedback from clients. This is problematic given the social organization of most genetic counseling today.

We did look at another type of counselor variable which we thought might bear some relationship to the overall rate of client learning — the counselors' sensitivity to whether or not clients were having difficulty in understanding the information being relayed to them. Since we asked counselors to give us their impressions of each counseling case in addition to providing us with medical-genetic facts, we were able to explore this hypothesis.

Our findings were in fact different with regard to client risk and diagnostic learning. In terms of risk knowledge, counselors reported the same level of professional satisfaction for sessions in which the client did not learn, as for sessions in which the client did learn their risk. Moreover, we found that counselors were not aware of the fact that clients who did not learn their risks were having difficulty with this information.

A different picture emerged in regard to client learning of diagnostic knowledge. Here, counselors were significantly less satisfied with sessions in which clients failed to learn their diagnosis, than in those cases in which diagnostic knowledge was acquired by the client (84.4% vs 47.9%, $p < .05$). As expected, counselors were aware of those clients having trouble understanding the diagnostic information presented during the counseling session (59.0% vs 40.7%, $p < .05$). While far from conclusive, this suggests counselors may be more attuned to educating clients about their diagnosis than about their risk.

In summary, our analysis of counselor characteristics and their relationship to client diagnostic and risk knowledge suggests that no single professional group of providers in this study had a monopoly of clients who learned their diagnostic and risk information. This may raise some questions about how professionals ought to be trained for counseling, especially when we recall the relatively large number of learning eligible clients who failed to acquire accurate diagnostic or risk information in counseling.

It was also the case that counselor attitudes and goals did not predict their effectiveness as educators, nor did the amount of experience a counselor had. Stated differently, the role models selected for educating tomor-

row's genetic counselors cannot be chosen simply on the basis of their opinions about counseling, or their experience; at least our data suggest they should not be, if very high levels of client diagnosis and risk education are important counseling objectives.

Clients

The final set of factors we examined in the attempt to better understand client learning relates to the clients themselves. Much information was gathered on clients, their sociodemographic characteristics, attitudes, and perspectives. We will briefly summarize the results of our analysis.

First, it should be noted that male clients learned the risks given to them in counseling significantly better than females (59.9% vs 44.7%, $p < .05$). However, there was no significant difference between the sexes in terms of learning diagnosis (54.0% vs 49.4%, ns).

As one might suspect, a significant association was found between a client's education and the accuracy of his or her diagnostic and risk knowledge *before* counseling. Approximately 70% of clients with a junior high education or less knew the diagnosis of the problem that brought them to counseling. Ninety-one percent of those with graduate level training had this knowledge ($p < .05$). Similarly, while no clients with a junior high education or less knew their risk prior to counseling, almost 30% of those with graduate training entered counseling with accurate risk knowledge ($p < .05$).

Given the above facts, one might assume that the more educated a client, the more likely they would be to learn the diagnostic and risk information provided by counseling. Surprisingly, this was not the case. Clients with a graduate or college level education who entered counseling not knowing their diagnosis or risk, were no more likely to learn this in counseling than clients with a high school or junior high school level education.

Naturally, clients came to genetic counseling with a diversity of reproductive backgrounds (see Table 3-20). The analysis suggested that those clients who reported any pregnancy experience prior to counseling were somewhat more likely to learn their risk than those who reported none (46.9% vs 36.5%, $p = .10$). Those with a prior pregnancy experience were no more likely to learn their diagnosis, however (50.5% vs 44.7%, ns).

When we looked at the nature of the prior pregnancy experience, we found that clients who had experienced an adverse pregnancy outcome, ranging from stillbirth to the birth of a child with some defect, were more likely to learn both their risk and diagnosis than those who had not (48.4%

vs 38.0%, $p < .05$, and 53.7% vs 39.8%, $p < .05$). Interestingly, clients who were planning a future pregnancy or were pregnant at the time of counseling were no more likely to learn their risk and diagnosis than those who were not.

It has been hypothesized by several counselors that a psychologic reaction, not unlike mourning, takes place after the birth of a defective child [4]. It is argued that if counseling takes place too soon after such a birth — before a person has coped with what has happened — they may not be able to absorb what they are told by the genetic counselor. Because of the general interest in this issue we used our data, to the extent possible, to see if client knowledge acquisition varied in relation to the recency of the birth of a child with a birth defect. Our data were collected such that we could classify parents as within six months of such an event, from six months up to a year, a year to a year and a half, and so forth. Using this classification scheme, we found inconsistent results. Parents who had a child with a birth defect born within two years did not learn their diagnosis as well as parents whose affected child was older (38.6% vs 61.1%, $p < .05$). However, no such relationship held for clients learning risk information. In fact the data, while not statistically significant, showed a reverse trend, with parents having a very young affected child learning their risk better than those with older affected children. Thus, there is, at best, only partial support for this hypothesis in the data we gathered.

Our analysis also revealed that a client who had received genetic counseling prior to entry into this study (for the same or a different problem) was no more likely to learn risk and diagnosis than a client who had no prior counseling experience. Study coordinators at the 47 clinics were instructed to include in the study only clients presenting for their initial counseling session, but in some cases clients who had been seen before at the same clinic or elsewhere were included. Regardless, prior genetic counseling was not related to learning risk and diagnosis. Of course, we do not know what went on in the previous counseling, but this observation suggests that even multiple genetic counseling sessions may not improve client learning as significantly as one might assume.

As discussed in Chapter 4, clients brought to counseling a variety of questions and concerns, some genetic-medical, some more social or psychologic in content. Not surprisingly, our analysis showed that those who specifically stated that they had come to counseling to learn risk and diagnosis were most likely to learn this information correctly than those not so disposed. To be precise, 46.8% of those clients who said, prior to counseling, that they wanted to learn their chances of having an affected child learned their risk, while only 24.0% of those who did not specify this as a reason for seeking counseling did so ($p < .05$). A similar, but nonstatis-

tically significant finding concerned learning diagnostic information. Those who reported that one of their reasons for seeking counseling was to learn whether they (or a relative) had a specific disease or disorder were more likely to learn diagnostic information accurately than clients who did not come to counseling with this question (54.0% vs 43.2%, ns).

Whether a client had inaccurate information or no information at all prior to counseling also influenced learning. Those who reported that they did not know their risk before the counseling session learned it significantly better than did those who came with erroneous risk information (48.4% vs 34.9%, $p < .05$). A similar, but nonstatistically significant relationship was found with respect to learning diagnostic information (63.5% vs 57.3%, ns).

In addition to prior risk and diagnostic information, clients also come to counseling with many attitudes and beliefs about a disorder. The data show that these can affect client learning as well. For example, clients who *believed* that the disorder for which they sought counseling was inherited, whether it was or not, were more likely to learn both their risk and their diagnosis than were clients who did not share this belief. More specifically, 54.3% of those who came to counseling believing that their problem was inherited learned their risk correctly, while only 40.3% of those who did not have this belief did so ($p < .05$). Although not statistically significant, learning differences for diagnostic knowledge were similarly associated with client belief about inheritance (61.1% vs 48.8%, ns).

Finally, the analysis disclosed that there exists a relationship between the learning of risk and diagnostic information and client beliefs about the magnitude of risk involved.

While 41.7% of the clients who thought their risk was 11% or greater learned their diagnosis, 66.6% of those who thought their risk was 4–10% and an equal percent of those who thought their risk was 0–3% learned their diagnosis ($p < .05$). Similarly, 34.7% of those who thought their risk was 11% or higher learned their risk correctly, while 47.3% of those who thought their risk was 4–10% did, and 57.4% of those who thought their risk was 0–3% ($p < .05$). Clearly, the data indicate that the higher the risk a client believed was involved, the less likely he or she was to have learned the correct diagnostic and risk information as reported by the counselor.

Together, these final few observations suggest that prior beliefs, including misinformation, may significantly impede client learning. It may thus be of some value for the counselor to know what clients believe, so as to correct misinformation or point out erroneous beliefs, or consider how such beliefs may relate to a client's receptivity to the information provided in counseling.

Before concluding this chapter we will briefly examine another aspect of counseling, what one may call "attitudinal learning."

CLIENT ATTITUDINAL CHANGES

We have already discussed some of the ways in which a variety of factors, including a client's beliefs about a genetic disorder, can affect the learning of risk and diagnostic information presented during genetic counseling. Now, we wish to consider briefly whether client beliefs and attitudes, specifically those relating to the risk and burden of a disorder, may also change during counseling. Along with the counselor's efforts to teach clients such factual information as risk and diagnosis, another type of education may occur during counseling which results in changes in attitudes or beliefs, a sort of "attitudinal learning."

Clients' interpretations of numeric risk can vary significantly. What to one client is a "high" risk may be to another "moderate" or "low." The same is valid regarding counselors and their attitudes about various risks. Such interpretations, or attitudes about risks, are important, particularly from the client's perspective in that they are one factor that clients unquestionably will consider in making reproductive decisions, in addition, of course, to the "objective" numeric risk and their sense of how burdensome a disease or disorder may be.

For these reasons we examined client and counselor attitudes about risks and disease burdens in some detail, although we will present only part of the information in this monograph.

In an earlier section of this chapter we noted that in approximately 15% of the cases in this study the counselor indicated to the study team that they had reported a nonnumeric risk to their client. That is, instead of reporting a risk as 10%, they told the client the risk was very high, high, or moderate and so forth.

Because such a significant proportion of risks was reported in this fashion, and because we suspect that clients and counselors may not always share the same interpretation or attitude about a given numeric risk, we have prepared Table 5-6.

This table reports the agreement in attitudes about the *same* risk as viewed by the counselor and the client. To construct this table we selected only those cases (N = 222) where the client and counselor agree as to what the numeric risk was. As part of the study clients and counselors were asked how they interpreted the risk faced in each case. Table 5-6 simply reports the agreement and disagreement between counselors and clients in their interpretations of the same risk.

The table shows that, while there is some agreement in counselor-client attitudes about various risks, as reflected in the main table diagonal, in only one cell do more than 50% of counselors and clients agree as to how to interpret the risk. Stated somewhat differently, except where the clients perceived their risk as being low, the perceptions of the counselor were

TABLE 5-6. Comparison of Client and Counselor Attitudes About Clients' Risk*

Client Attitude About Risk	Counselor Attitudes About Clients' Risk					
	VH (%)	H (%)	M (%)	L (%)	VL (%)	N
Very High	18.8	37.5	31.3	12.5	0.0	16
High	35.3	26.5	20.6	11.8	5.9	34
Moderate	8.6	38.6	28.6	21.4	2.9	70
Low	6.5	6.5	29.0	54.8	3.2	62
Very Low	0.0	7.5	17.5	42.5	32.5	40
Total N						222

*Includes only those clients who gave the same numeric risk as their counselors.
Key: VH — very high; H — high; M — moderate; L — low; VL — very low.

more likely to differ from the client's than to agree. In addition, when clients felt their risk to be high, moderate, or very low the majority of counselors perceived the client's risk as being *higher* than the client himself saw it.

There was exact agreement between clients' and counselors' perceptions of risk 36% of the time. Combining categories so that responses which only varied by one degree either way would be considered in agreement, matching interpretations result 83% of the time.

We specifically asked counselors to report how they saw the risk for each particular client because we realized that such individual factors as the nature and seriousness of the disorder involved, a client's family situation, finances, and so forth might play a role in how the counselor perceived the risk. Thus, the data reported in Table 5-6 are an attempt to view similarity in counselor and client interpretation with both of them using as common a framework as possible.

What the data say, in general, is that there is substantial disagreement in how the counselors viewed the risks given clients, compared to how clients viewed these same risks. Such an observation raises questions about the reporting of nonnumeric risks by counselors. Given the variability observed in this study, it may be more useful to report only numeric risk figures to clients, and to let clients assign their own personal interpretations to the risks involved.

However, do, and if so, how do clients' interpretation of risk change during counseling? To answer this question we selected those clients in our

study who both before and after counseling reported both a numeric risk and an interpretation of that risk. We then classified these cases in terms of the groups shown in Table 5-7. First, we noted whether the numeric risk the client reported before counseling (T_1) was greater than, the same as, or less than the numeric risk the client reported after counseling (T_2). In addition, we noted whether the clients' attitudes or interpretation of the risk before counseling was greater than, the same as, or less than that reported after counseling. This classification allows us to see if a) clients' attitudes about their risks stay constant if the numeric risk does, and b) to what degree changes in numeric risk are accompanied by corresponding changes in attitudes about the risk.

Table 5-7 shows that, of the 85 cases where the client reported the *same* numeric risk both before and after counseling, in 50, or 58.8% of these, the client gave the same interpretation before and after counseling. However, in 18.8% of the cases the client attached a lesser interpretation to the same risk after than before counseling, and in 22.4% of the cases the reverse held.

Looking at the other columns, when the clients reported a decreased numeric risk (column one) most clients, 59.5%, showed a similar reduction in their attitude about the significance of the risk. However, 28.6% of the clients maintained the same interpretation although the risk had gone down numerically, and 11.9%, in some apparently complex psychologic process, saw their *lower* numeric risk as even *more* significant attitudinally than they had their higher earlier risk. Column three can be read analogously to column one.

What these data suggest is a significant amount of "slippage" between a client's cognitive knowledge of a risk, their interpretation of that risk, and changes of their interpretations of that risk as it changes. In interpreting

TABLE 5-7. Changes in Clients' Attitudes About Numeric Risk Before and After Counseling*†

	Numeric Risk			
Interpretation of Risks	$T_1 > T_2$ (%)	$T_1 = T_2$ (%)	$T_1 < T_2$ (%)	Total N
$T_1 > T_2$	59.5	18.8	13.6	
$T_1 = T_2$	28.6	58.8	40.9	
$T_1 < T_2$	11.9	22.4	45.5	
N	42	85	22	149

*The numeric risk as reported by the client before and after counseling.
†Clients were asked to rate the numeric risk they reported as very high, high, moderate, low or very low.

these results it must first of all be pointed out that while some of the changes in numeric risk reported by the client may have been large, some were also small. Nevertheless, the data presented in Table 5-7 suggest that there is considerable change in clients' attitudes about their risk during counseling, and the changes do not always correspond "rationally" to what the clients learned about their numeric risk. This suggests that perhaps something else conveyed during counseling may affect a client's perceptions of risk as much as the actual numeric risk information. This might be other factual information about the disorder, perhaps the counselor's own attitudes, or any of a number of factors.

Obviously, the psychology of risk interpretation is complex, and we have only touched on it here [5]. Nevertheless, it should be clear that it is difficult to know how a client will interpret a risk, and it is almost as difficult to predict how they will interpret a change in their numeric risk. In light of this, to reemphasize, it seems necessary to be cautious about supplying clients with judgments or attitudes about risk, rather than actual numeric risks as part of genetic counseling.

Just as clients have attitudes about risks that can be shaped by counseling, so too do they have attitudes about diseases and disorders that may change as a function of counseling. Often such attitudes or judgments are talked about with reference to a client's sense of how burdensome he or she feels a disease or disorder is or might be. As in the case of risk, we would anticipate that such attitudes can play an important role in shaping client reproductive decisions.

In order to assess whether clients' attitudes about the burden of having a child with a particular birth defect or genetic disorder change from pre- to postcounseling, we asked clients both before and after the session how serious a burden they felt a disease or disorder would be for them if they were to have a child with the problem or disease that had brought them to counseling.

Instead of soliciting a single global judgment about the "burdensomeness" of a disease, we conceptualized it in various ways, including the financial, emotional, interpersonal, caring, educational, and medical treatment aspects of having an affected child. Clients were asked to rate on a scale of 1 (no problem) to 4 (very serious) how burdensome they felt it would be to have a child with the problem that concerned them. Client responses, before and after counseling, are reported in Table 5-8.

What stands out immediately is that for all six problems listed there was a statistically significant drop in clients' mean sense of burden from before to after counseling. The change in the mean score was almost the same for each of the six issues. For the clients reported in Table 5-8 counseling is

TABLE 5-8. Female Client's Sense of Disorder Burden Before and After Counseling*

Type of Problem	Before T_1 (mean)	After T_2 (mean)	p†	N
Financial	1.9	1.7	.000	668
Feelings about affected child	1.5	1.2	.000	727
Relationship with spouse	1.0	0.7	.000	741
Caring for affected child at home	1.3	1.0	.000	717
Educating affected child	1.5	1.2	.000	690
Medical care for affected child	1.4	1.1	.000	707

*Clients were asked to express their sense of the future impact of each issue as follows:
4 = very serious
3 = moderately serious
2 = slightly serious
1 = no problem (Reversed from questionnaire for clarity of presentation.)
†T test of significance.

followed by markedly reduced concern about the burdensomeness of the disorders that brought them to counseling.

When we looked at clients' reports about whether or not they specifically discussed the six problems listed in Table 5-8 during their counseling session we obtained some interesting results. Discussing one's feelings about an affected child or about caring for an affected child at home was related to a significantly greater decrease in a client's sense of burden than if these issues were not discussed. For the other four issues however, the clients' sense of burden was lessened to virtually the same degree, regardless of whether the client reported the topic had been specifically discussed during counseling or not.

Thus, even when counseling does not specifically address itself to the possible burdens of a disorder, as conceptualized here, it seems that it may

serve to reassure clients about the burden of different aspects of a disease or disorder. Perhaps simply talking to a concerned or sympathetic professional in a medical setting can lessen certain types of client anxieties.

Whatever the dynamics, we can see from our discussion that not only knowledge but attitudes as well change from before to after counseling. In the next chapter, we will explore what this may mean for client reproductive planning. Now, however, we will draw together some of the major findings reported in this rather detailed chapter.

SUMMARY

This chapter has examined the degree to which clients are more informed after counseling than before, more informed to make decisions that may have profound effects on their individual and familial futures. As we pointed out at the beginning of this chapter there are many considerations that go into making fully informed decisions, and we have looked at only two sets of factors: 1) client knowledge of risks of having a child with a birth defect and their attitudes or interpretation of such risks, and 2) client diagnostic knowledge, and their views on how burdensome such diseases or disorders are.

With respect to client diagnostic and risk knowledge it is clear that while the genetic counseling studied in this project resulted in many clients learning their risk and diagnosis, it is equally clear that there is considerable room for improvement. In short, this chapter has documented a significant gap between what could, or should, be accomplished educationally in counseling, and what in fact is being accomplished. The fact that not all clients who could improve their knowledge do so can be interpreted in several ways, reflected perhaps in the words and phrases throughout this chapter. One may view less than perfect client knowledge as a failure in education, suggesting that the problem lies with the professionals. They are simply poor educators, perhaps well trained in clinical genetics, but not especially skilled at transmitting their knowledge to laymen.

Conversely, some may view the gap that we have documented as the result of poor learning by clients. For various reasons, attributable to clients, not to counselors or the counseling session, clients fail to hear or to retain the information provided by counselors.

It is not necessary to take one side or the other to conclude that there is a problem. And, as we think our analysis suggests, many factors enter into a useful understanding of why we find 53% of the clients given a risk not learning it, and 40% of the clients given a diagnosis not appearing to know it after their counseling. While it is the case that, on average, the percent of patients who learn from their physician in routine medical encounters is not

much different than observed here, one can ask if this is good enough [6]. When one weighs the importance of the family and personal decisions that can be premised on the information learned or not learned in genetic counseling, it seems that an effort should be made to improve this aspect of counseling.

Recognition that there is an educational problem in genetic counseling is important, but more useful is some understanding of what contributes to the problem.

In attempting to provide some indicators of potential use to counselors, we compared clients who learned risk and diagnosis to those who did not learn, on the basis of three groups of variables: 1) characteristics of the genetic counseling experience, 2) counselor characteristics, and 3) client characteristics.

Although we found considerable variation in client learning at different study clinics, most of the clinic and counseling structure and content variables we examined did not make a significant difference in client learning. It is particularly interesting that neither the presence of support professionals nor the amount of time spent with the counselor did so. In regard to the content of the counseling session, neither being helped with personal concerns nor having new concerns raised affected learning appreciably, while discussion of medical concerns did.

We could not look at such hard-to-measure characteristics as the personality and manner of individual counselors; these could well play a role in how well clients learn. While the analysis did not disclose any significant relationship between the counselor characteristics we examined and client learning, three findings did stand out. First, inexperienced counselors were as successful as those with more experience in regard to their ability to teach risk and diagnosis. In addition, there was no appreciable difference in the educative success of counselors with different types of professional training. MDs, PhDs, and masters' level genetic associates had comparable levels of informed and uninformed clients postcounseling. Finally, several findings in our study indicate that the professionals involved in genetic counseling are more attuned to client diagnostic learning than to the learning of risk. In fact, for most cases in which the client failed to learn risk information, the counselor did not perceive that the client was having any difficulty. These findings may shed some light on how such a significant proportion of clients can leave counseling without having learned the risk information presented.

The final set of variables, those relating to individual client characteristics, yielded some interesting results. For example, male clients learned risk information significantly better than females. When we looked at clients' educational backgrounds we found what we expected — those

clients with higher levels of education were far more likely to know their risk and diagnosis prior to counseling. However, when such clients arrived ignorant of their risk and diagnosis they were no more likely to learn in counseling than were less-educated clients. Not unexpectedly, clients who came to counseling wanting to learn their risk and diagnosis were more likely to succeed in doing so than were others. It is interesting, however, that those who had no information before counseling were more likely to learn than clients who had incorrect information. This suggests that reeducating may be a frequent and more difficult task for counselors than simply education.

Turning to the important area of the clients' reproductive history, clients who were pregnant at the time of counseling or who were planning a future pregnancy were no more likely to learn risk and diagnosis than were other clients. However, clients who had had a past pregnancy experience were somewhat more likely to learn their risk than those who had not. More important was the outcome of such a past pregnancy. The data revealed a significant relationship between having had an adverse pregnancy outcome and the learning of both risk and diagnosis. When we tested the hypothesis that counseling which takes place soon after the birth of a defective child may be ineffective, we found only partial support.

Clients come to counseling with certain attitudes or beliefs about a problem or disorder, and the data show that these too can affect client learning. For example, clients who believed that the problem for which they sought counseling was inherited were more likely to learn both risk and diagnostic information than those who did not have such a belief. There was also a significant relationship between the learning of risk and diagnosis and clients' beliefs about the magnitude of their occurrence/recurrence risk. The higher the risk a client *believed* was involved, the less likely he or she was to have learned the *actual* risk and diagnosis as reported by the counselor.

In addition to the learning of such factual information as risk and diagnosis, there is also attitudinal learning which occurs as a result of counseling. In particular, we have looked briefly at how counseling may affect clients' attitudes about the risk and burden of a disorder. When we compared changes in clients' attitudes about their risks before and after counseling, we discovered that, while considerable attitudinal change occurred, such changes did not always correspond to the change in client-reported numeric risks. This suggests that in some cases things other than the numeric risk conveyed during counseling may affect a client's perception of risk. A comparison of clients' postcounseling attitudes about their risk with the attitudes of their counselors' revealed exact agreement only 36% of the time.

Our final set of findings relate to a client's sense of burden about a disorder. We found that following counseling there was a significant decrease in various clients' concerns about the future burdensomeness of the problem that brought them to counseling.

While attitudinal learning about risk and burden may not be a planned part of either the counselor's or the client's agenda prior to counseling, there is ample evidence to indicate that it does occur. There is also evidence that client attitudes can affect the learning of factual information which is the primary focus of counseling, and that likewise the learning of factual information may affect clients' attitudes.

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Chapter 6

Genetic Counseling and Client Reproductive Plans

INTRODUCTION

The role of genetic counseling in client reproductive planning is a topic that has received considerable discussion but to date little systematic empirical analysis. Professionals have expressed interest in how clients use the information received in counseling. This interest has ranged from questions of how to make genetic counseling information more useful to clients who are still active reproductively, to questions about how specific information, such as recurrence risk and perceived disease burden, affects client reproductive plans and behavior [1, 2].

Professional interest in client reproductive planning is reflected in the *American Journal of Human Genetics* definition discussed at length in Chapter 1. From the perspective of this definition genetic counseling should, among other things, "help the individual or the family to . . . understand the options for dealing with the risk of recurrence . . . choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision . . . and help clients make the best possible adjustment . . . to the risk of the recurrence of that disorder" [3].

These several items presumably include such tasks as educating clients about the availability or utility of contraception, abortion, artificial insemination, and prenatal diagnosis. Also listed is helping clients actually choose a course of reproductive action and live in accordance with and adjust to that decision.

We saw in Chapter 3 that, at least attitudinally, there was considerable endorsement among the counselors in this study for these various tasks as important aspects of their genetic counseling. Sizeable numbers of counselors, however, did not give the highest level of endorsement to the role of helping clients actually choose a course of reproductive action.

For this chapter we felt it important, given the state of the field, to provide a general perspective on the role of genetic counseling in client reproductive planning, rather than focus on more narrow issues, such as client reproductive plans in light of recurrence risk and disease burden. This latter issue, while important, goes somewhat beyond the scope of the *American Journal of Human Genetics* definition and seems to place more emphasis on genetic counseling as preventive medicine than as an educative and counseling undertaking. We are preparing a separate publication that will look in detail at genetic counseling as "preventive medicine."

We consequently are approaching our analysis of the role of genetic counseling in client reproductive planning in this chapter from a somewhat different perspective than the evaluative posture developed in Chapters 4 and 5. While there may not be universal agreement among counselors that all client genetic and sociomedical questions and concerns are to be discussed in counseling, it has been possible, as we saw in Chapter 4, to identify the most common client questions and concerns and report which are and are not being discussed in counseling. With respect to client knowledge after counseling, counselors may vary in terms of the knowledge they consider necessary for a client to be "fully informed." But it has been possible, as we saw in Chapter 5, to assess client diagnostic and risk knowledge before and after counseling, and make a judgment as to the effectiveness of counseling. With respect to counseling and client reproductive plans, it is difficult to formulate specific goals and objectives for assessing the effectiveness of genetic counseling. This is so in part because virtually no definitions go beyond the statement that clients should find the information provided in genetic counseling useful in making reproductive plans. That is, they do not specify the type of reproductive decisions clients should make.

With these observations as background, we felt it important to provide descriptive information on two issues regarding genetic counseling and client reproductive planning. The first focuses on how clients define the context surrounding their reproductive planning. More specifically, what factors or considerations do clients say are important to them in making their reproductive plans? How do such considerations vary as a function of the pregnancy and reproductive experience of the clients? Such information should permit a more informed professional perspective on the clients who seek genetic counseling, and how they approach their reproductive decisions and view reproductive planning.

Second, it is the case that some clients come to genetic counseling with reproductive plans they may have held for many years, plans tied to deeply held values and familial expectations. In such cases, how likely is it for counseling, which often lasts less than an hour, to make a significant

difference or change in such plans? On the other hand, some clients come to counseling having recently experienced the birth of a child with a problem. This event may have called into question their family plans, making them uncertain about what to do reproductively. Such clients may be very open to influence by genetic counseling. In short, what role does counseling play in client reproductive planning? How often is it irrelevant, an important source of change, or a reinforcer for reproductive plans that existed prior to counseling?

CLIENT CONSIDERATIONS IN REPRODUCTIVE PLANNING

Most existing publications exploring the reproductive plans of genetic counseling clients have focused almost singly on reproduction and the risk the client faced, and some assessment of the degree of burden imposed by a particular disease or disorder. Often, but not always, both of these considerations were from the perspective of the professionals, not the clients.

It is understandable why professionals might focus on these two considerations. First, of course, they are clearly factors relevant to clients in planning their pregnancies, unless we assume reproductive planning to be totally irrational. Second, information about risk and burden constitutes, to some degree, primary contributions genetic counselors can make to clients and the problems they face. Accordingly, it is not surprising that counselors as professionals are interested in viewing and assessing client behavior in terms of their primary contributions to these clients.

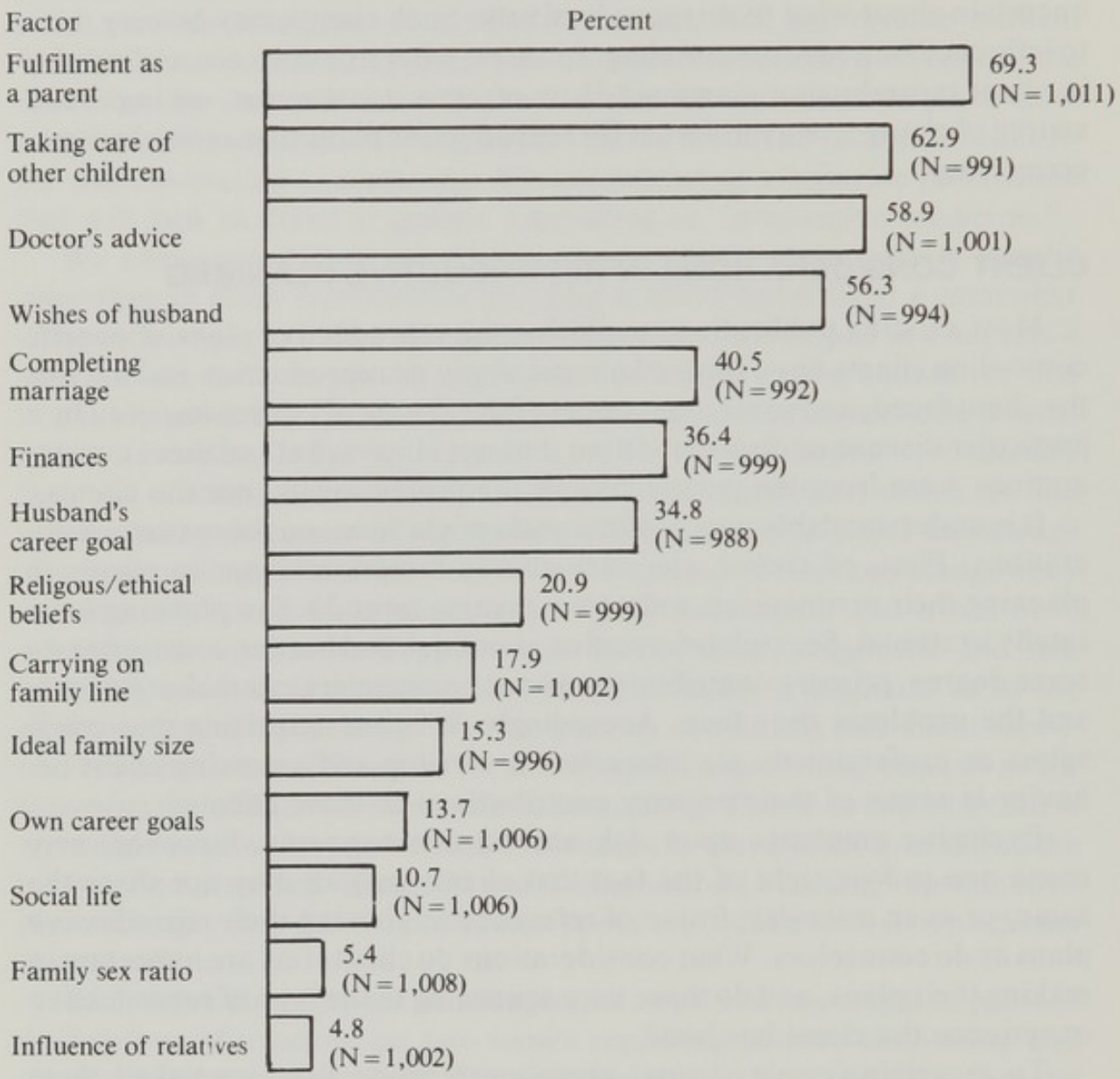
Exclusive emphasis upon risk and burden concerns, however, may cause one to lose sight of the fact that clients may or may not share the same, or even a similar, frame of reference in viewing their reproductive plans as do counselors. What considerations do clients feel are important in making their plans, and do these vary according to the type of reproductive experience the client has had?

To ascertain female clients' views on reproduction, we asked them before their genetic counseling to assess the importance they attached to a variety of possible issues or topics people might consider in making reproductive plans. The list of concerns or factors was developed during the pilot phase of the research in discussions with clients.

Table 6-1 reports the assessments provided by the female clients in this study, ordered from the most frequently cited consideration to the least. In ascertaining client assessments we were careful to solicit the importance the client personally attached to the various considerations, not the importance they felt people in general might or should attach to them.

The list of reproductive considerations covers a broad spectrum of topics. Some considerations, such as fulfillment as a parent, reflect very

TABLE 6–1. Female Clients Rating Factors as Very Important in Their Reproductive Planning



basic personal parenting goals. Other considerations are oriented more toward the family as a social unit, such as completing one's marriage, carrying on one's family line, and achieving a desired family size. Still other items reflect more practical types of considerations, such as taking care of other children, finances, career goals, and the impact on one's social life of

having a child. From the list it is clear that the clients consider a variety of factors in planning their pregnancies, and some considerations are much more likely to be viewed as very important than others.

The data shown in Table 6-1 reveal that of those considerations most often rated very important by clients, fulfillment as a parent was the most frequently cited consideration. A more practical concern, taking care of other children, was cited as very important by almost as many clients. Over half the female clients cited their doctor's advice and the wishes of their husband as being very important in making reproductive plans. In short, looking at the most frequently cited reproductive considerations, most clients cited a mix of personal, practical, and medical as well as interpersonal considerations in making their reproductive plans.

From 21% to 40% of the clients cited completing one's marriage, financial concerns, a spouse's career goals, and religious or ethical concerns as very important in their reproductive planning. Finally, there were six considerations, including carrying on one's family line, and achieving an ideal family size or sex ratio, that were less likely to be rated as very important.

Clearly, clients bring many factors to their reproductive planning, considerations that would seem to make the process of arriving at a specific reproductive plan very complex. The various topics listed in Table 6-1 suggest why, to some degree, we found in Chapter 3 clients bringing an array of sociomedical concerns and questions to counseling for discussion. The data here show that clients consider many issues in their reproductive planning, and not surprisingly they often want to discuss some of these with their counselor.

For the professionals in genetic counseling it is perhaps important to note the importance many clients attached to their "doctor's advice" in reproductive planning. In fact, slightly more clients viewed such advice as being as important as the wishes of their spouse in their reproductive planning. To the degree that clients view the information and counsel received in genetic counseling as part of their "doctor's advice," it thus could play an important role in reproductive planning, a role shared with a variety of other nonmedical considerations.

Client assessments of the importance of the considerations listed in Table 6-1 were also solicited at the six-month follow-up period. To a very substantial degree, clients maintained the same relative ranking of these considerations, suggesting stability in the importance attached to these issues in reproductive planning. The same top four concerns listed in Table 6-1 were the top four concerns at six months. However, concern about taking care of other children had become the most frequently cited as most important, followed by the wishes of one's spouse, fulfillment as a parent, and lastly, a doctor's advice. The relative ordering of the other concerns

listed in Table 6-1 remained unchanged, with the exception that financial concerns became more frequently cited as very important than did completing one's marriage. Thus, while the relative ranking remained largely similar at six months, there was the suggestion that more "practically" oriented concerns were taking on importance for more people, compared to their initial precounseling judgments.

One might expect that factors considered important in making reproductive plans would be different for clients who are pregnant or for those planning a pregnancy. The reality of pregnancy, or the expectations of a pregnancy, may shift the concerns or interests of clients. Information bearing on this is presented in Table 6-2.

Table 6-2 reports the percent of female clients planning a pregnancy, or pregnant at entry into the study, and their ratings of the importance of the factors shown in Table 6-1. Looking first at those planning a pregnancy, we see that the same four issues listed in Table 6-1 — fulfillment as a parent, care of other children, wishes of spouse, and doctor's advice — are the most frequently cited as very important. For clients who were pregnant at entry into the study, the same four items were the most frequently rated of the highest importance.

As for other considerations, the general relative ranking of items is very similar to that reported for the total client population in Table 6-1. Com-

TABLE 6-2. Female Client Reproductive Considerations Rated as Very Important, by Pregnancy and Planned Pregnancy Status

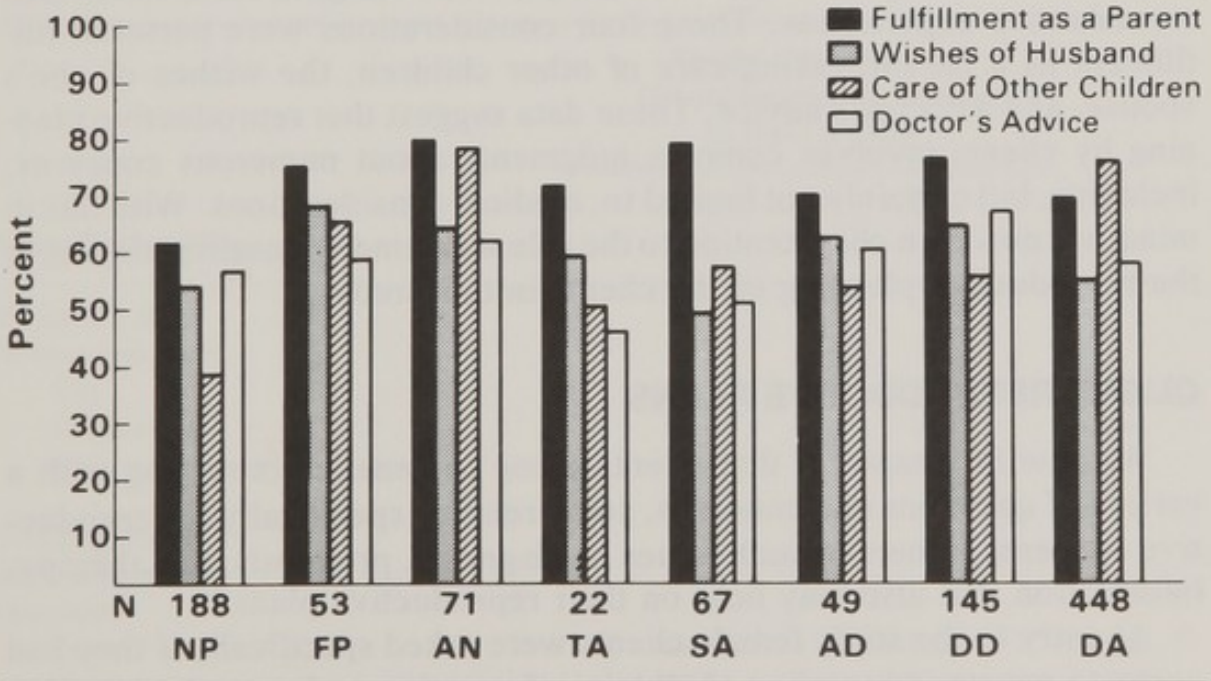
Reproductive Considerations	Pregnancy Planned (%)	Pregnant (%)
Fulfillment as parent	76.4	73.9
Care of other children	61.2	72.5
Wishes of husband	61.1	61.2
Doctor's advice	55.5	56.5
Completing marriage	41.0	45.5
Finances	31.8	41.7
Husband's career goals	39.0	41.5
Religious/ethical beliefs	17.9	16.7
Carrying on family line	17.6	19.7
Ideal family size	6.2	15.8
Own career goals	13.7	13.9
Effects on social life	11.3	10.1
Desire for certain number of boys and girls	6.2	2.9
Influence from relatives	3.9	5.0
N	344	142

pleting one's marriage, financial concerns, and husband's career goals are more likely to be assessed as very important than such issues as the effects of another child on one's social life, the desire for a certain number of boys and girls, or the influence of relatives. It would appear that planning a pregnancy or actually being pregnant does not alter dramatically the general importance attributed to the issues listed in Table 6-1.

Table 6-3 reports clients' rating of the four most frequently cited considerations in Table 6-1 by their reproductive experience. Within groups having different reproductive experiences the ordering of considerations reflects their ordering in Table 6-1. The height of the bar graph reflects the percent of female clients rating the concern as very important, and hence gives us some indication of how the importance of these four considerations varies by the type of reproductive experience the client has had.

Considerable information is presented in Table 6-3 and we will only highlight some of the major observations here. First, as is apparent for every group except one, the factor most frequently cited as important in shaping reproductive decisions is personal fulfillment as a parent. This consideration is particularly important among those who have experienced

TABLE 6-3. Reproductive Consederations Rated Most Important, by Female Client Reproductive Experience



Key NP, never pregnant; FP, presently pregnant for first time; AN, all normal; TA, therapeutic abortion with no abnormality indicated; SA, spontaneous abortion without apparent defect; AD, abortion with defect; DD, defective child now deceased; DA, defective-alive.

spontaneous abortions and multiple miscarriages, perhaps understandably, but it is also very important among all other groups. This consideration is surpassed in importance only for parents with a living child having a birth defect. For these parents the most frequently cited consideration is how having another child could impact on taking care of other children. It is also the case that almost as many parents with normal children rate taking care of other children as very important, as rate personal fulfillment as very important in their reproductive planning.

As observed in previous tables, there is similarity in the percent of clients attaching high importance to the wishes of their spouse and the advice of a doctor in making reproductive plans. The group most likely to attach high importance to a doctor's advice in reproductive planning are those who have had a child with a birth defect, but the child is now deceased. The group least likely to do so are those who have reported therapeutic abortions as their worst pregnancy outcome. The group most likely to attach high importance to the wishes of their spouses are those presently pregnant for the first time, while those with spontaneous abortions and multiple miscarriages are the least likely to rate this consideration as highly important in their reproductive planning.

While the data presented in Tables 6-1–3 can provide only a limited perspective on the factors that clients bring to bear in their reproductive planning, we can see that a relatively broad set of considerations is given attention. In particular, four considerations were rated rather consistently as very important, both before and after counseling, and with different reproductive experiences. These four considerations were personal fulfillment as a parent, taking care of other children, the wishes of one's spouse, and a doctor's advice. These data suggest that reproductive planning by clients involves complex judgments about numerous concerns, including, but certainly not limited to, medical considerations. With this in mind, we now turn our attention to the role we found counseling playing in the reproductive planning of the clients in this study.

CLIENT REPRODUCTIVE PLANS

We saw in Chapter 3 that clients come to genetic counseling with a variety of questions and concerns, some relating specifically to reproductive concerns, others to such issues as diagnosis, prognosis, and therapy, information that also may bear on their reproductive plans.

At entry to the study female clients were asked specifically if they had come to genetic counseling to obtain information to be used in making reproductive plans. Fifty-three percent said they had, while 47% said they

had not. For slightly more than half our study sample then, genetic counseling was approached as a reproductively relevant medical encounter.

As shown in Table 6-4, the group of clients most likely to be approaching counseling to obtain information to make reproductive plans were those clients who were unsure about having a child, either in the short or long term. Conversely, approximately 30% of clients committed to having a child in the next two years or sometime thereafter were not in counseling to get reproductive planning information. And nearly 70% of clients not planning a pregnancy reported they had not approached counseling to obtain information to make a reproductive decision. Certainly clients vary in their orientation to counseling as an encounter to obtain information relevant to reproductive planning.

In Table 6-5 we present the percent of female clients seeking reproductive planning information in light of their pregnancy experience. The group most oriented to using counseling to help them in their pregnancy planning were those clients never pregnant (NP), those with an abortion with some indication of an abnormality (AD), and those who had had a child, now deceased, born with a birth defect (DD). Those least likely to be viewing counseling from a reproductive planning perspective were those pregnant for the first time (FP), those having a living child with a birth defect (DA), and those with only normal children (AN).

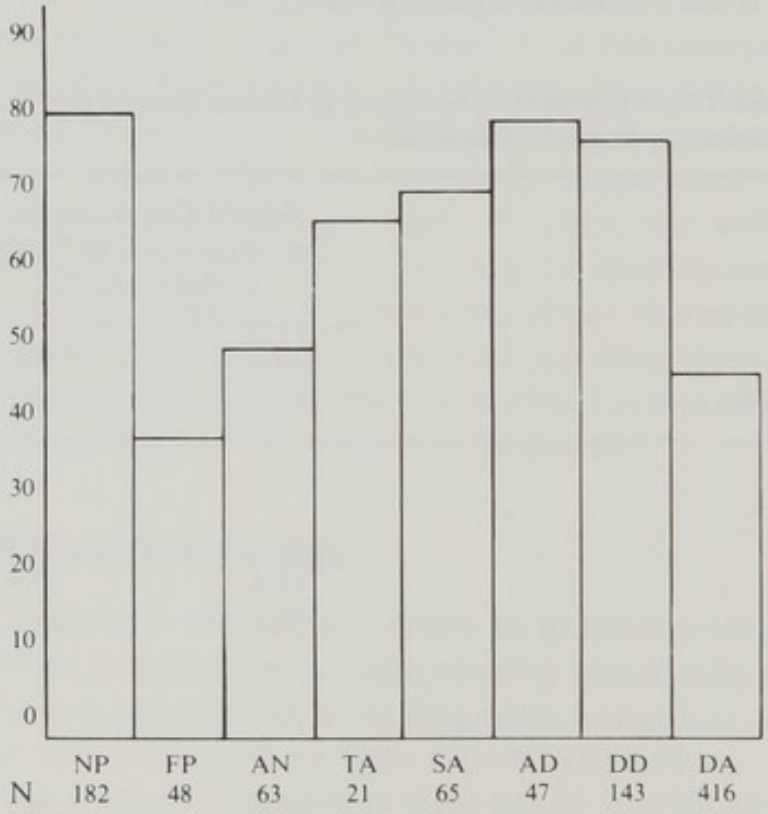
TABLE 6-4. Female Client Interest in Counseling to Obtain Information to Make Reproductive Decision, by Reproductive Plans

	Sought Counseling to Get Information to Decide to Have a Child		
Reproductive Plans	Yes (%)	No (%)	N
To have a child in next two years			
Yes	72.1	27.9	326
Unsure	82.4	17.6	284
No	30.3	69.7	343
Thinking about child after next two years			
Yes	69.8	30.2	281
Unsure	77.7	22.3	337
No	32.9	67.1	322

Finally, Table 6-6 provides a profile of the types of questions and concerns brought by clients who were and were not viewing their counseling as a place to obtain pregnancy planning information. The two groups have some marked differences in the questions and concerns they bring to counseling. It is clear that those oriented to counseling as an encounter in which to obtain reproductive information are very interested in risk, etiology, and prenatal diagnosis information. On the other hand, those not viewing counseling as a source of information for their reproductive planning, while still interested in etiologic information, have marked interest in the diagnosis of their child, therapy, and information on how to care for their child at home. There were smaller differences with respect to sociomedical topics.

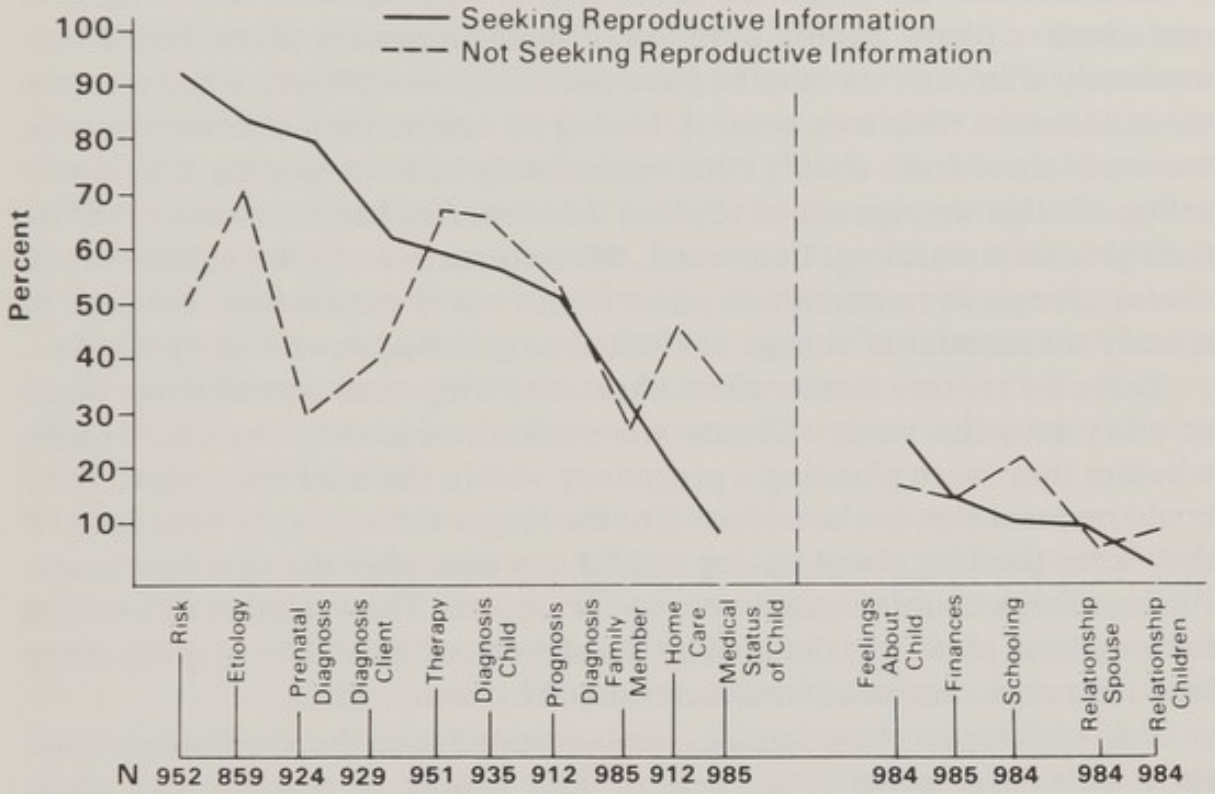
It would appear that the specific questions and concerns clients bring to counseling are shaped, in part, by their views of counseling as an aid in reproductive planning.

TABLE 6–5. Female Clients Seeking Reproductive Planning Information in Counseling, by Reproductive Experience



Key NP, never pregnant; FP, presently pregnant for first time; AN, all normal; TA, therapeutic abortion with no abnormality indicated; SA, spontaneous abortion without apparent defect; AD, abortion with defect; DD, defective child now deceased; DA, defective-alive.

TABLE 6-6. Profile of Questions and Concerns of Clients Seeking and Not Seeking Reproductive Planning Information in Their Counseling



In our pilot work it became clear that ascertainment of client reproductive plans was a difficult task. We explored various ways of obtaining such information. In the process we came to recognize that clients entertained both short- and long-term reproductive plans. However, while short-term plans carried some certainty and commitment, it was evident that reproductive plans beyond the next couple of years were quite speculative for many clients, and they found it difficult to give reasonable judgments about these plans. Because of this, in ascertaining client reproductive plans for this study we solicited both short-term (within two years) and long-term (after two years) reproductive plans. While we will report some data on both short- and long-term plans, as we did in Table 6-4, we will limit much of our subsequent analysis to reproductive plans of clients covering their next two years, because of its greater reliability for clients.

For each client entered into this study, the actual time period spent as a study participant was limited to six months. Resource limitations made it impossible to follow clients for a longer period of time. Because of this, our attention focused on client reproductive plans, not pregnancies or births. Obviously, in a period of six months many clients are not going to have sufficient time to act on their plans. Accordingly, we felt that in a six-month

period we could gauge the impact of counseling more accurately by looking at reproductive plans, rather than at pregnancies or births.

To assess the role of genetic counseling in shaping short- and long-term reproductive plans, clients were asked their pregnancy plans before, immediately after, and six months postcounseling. In addition, since we were using clients as their own control, having no comparison or control group, we ascertained from clients both immediately and six months after counseling whether the reproductive plans they reported had been influenced by their genetic counseling. Combined, this information enables us not only to assess change in reproductive plans in our study population, but also to specify the amount of change attributed to genetic counseling by clients.

Table 6-7 reports female client short- and long-term reproductive plans at entry into the study. Clients were asked, regarding the short term, whether they were planning a pregnancy within the next two years. They could respond yes, no, or unsure. For the longer term, clients were asked if they were thinking about having a child any time after the next two years. Again, clients could respond yes, no, or unsure. The variation in wording for long-term plans was dictated by the aforementioned difficulty reporting long-term reproductive commitments noted by many clients.

Table 6-7 reports how female clients responded to these questions upon their arrival for genetic counseling. This profile of reproductive plans shows that prior to genetic counseling, 28% of the female client population had neither short- nor long-term plans to have a child. Apparently they had completed their reproductive careers. At the other extreme, not quite 16% were planning a child in both the short and long term, while an almost equal amount, 18%, were unsure about both their short- and long-term reproductive plans. The remainder, approximately 37%, while having reproductive

TABLE 6-7. Female Client Short- and Long-Term Reproductive Plans Prior to Genetic Counseling

Are You Thinking About Having Children Any Time After the Next Two Years?	Do You Intend to Have a Child (or Another Child if Now Pregnant) Within the Next Two Years?			Total Long-Term Plans (%)
	Yes (%)	Unsure (%)	No (%)	
Yes	15.8	8.1	4.7	28.6
Unsure	11.4	18.4	5.5	35.3
No	4.9	2.8	28.4	36.1
Total Short-Term Plans	32.1	29.3	38.6	N=1,027

plans, expressed some uncertainty about these plans in either the short or long term.

An examination of clients not planning any additional pregnancies was performed to describe the reasons for their seeking genetic counseling. This analysis revealed what was suggested in Table 6-5. Fully 71% reported they had a living child with a birth defect, a rate markedly higher than for the total population entered into this study (43%). Moreover, the most frequent reasons this group gave for seeking counseling involved learning the etiology of a disease or disorder, presumably in their affected living child; obtaining information about treatment, getting a diagnosis; and obtaining a prognosis. It would appear that the segment of the population not planning pregnancies in either the short or long term is one that, at entry into the study, was very focused on adjusting to and/or coping with a living child with a birth defect.

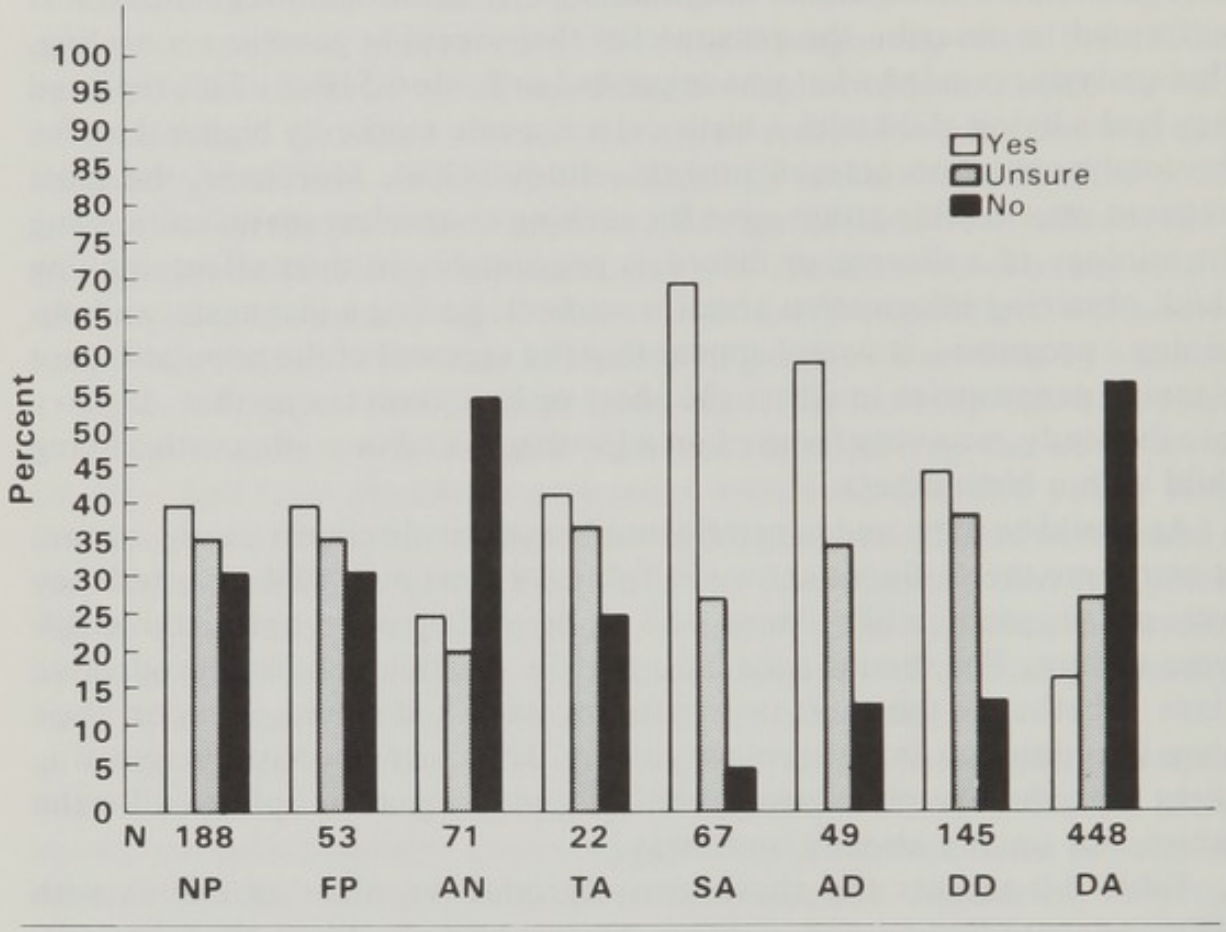
As would be expected, a certain number of female clients were pregnant at entry into the study, as shown in Table 6-8. Just over 83% reported they were not pregnant, while almost 14% reported they were, and just over 3% were unsure. We should note here that in obtaining client reproductive plans, whether in the short or long term, we asked clients to report their plans in addition to any current pregnancy. Thus, our emphasis in assessing client reproductive plans was specifically on pregnancies planned for the future, not on any already underway.

Table 6-9 reports the short-term reproductive plans of clients with different reproductive experiences. As can be seen, there are some dramatic differences in the short-term reproductive plans of these different groups. For example, the group most likely to be planning a pregnancy in the next two years is that group who have experienced spontaneous abortions and multiple miscarriages with no indication of abnormalities (SA). Also, over half of the group with abortions with some indication of an abnormality in the fetus (AD) were planning a pregnancy. Conversely, the

TABLE 6-8. Female Clients Pregnant at Entrance to Study

Pregnancy Status	Percent
Yes	13.5
Unsure	3.4
No	83.1
N	1,094

TABLE 6-9. Female Client Intentions to Have a Child Within Two Years, by Reproductive Experience



Key NP, never pregnant; FP, presently pregnant for first time; AN, all normal; TA, therapeutic abortion with no abnormality indicated; SA, spontaneous abortion without apparent defect; AD, abortion with defect; DD, defective child now deceased; DA, defective-alive.

group least likely to be planning a pregnancy was that with a living affected child (DA), a finding consistent with our discussion above. Also, over half of clients with all normal births (AN) were not planning a pregnancy in the next two years. Finally, the highest level of short-term reproductive uncertainty occurred among those who had given birth to a child, now deceased, with a birth defect (DD). Those clients reporting therapeutic abortions (TA) were also likely to express considerable uncertainty regarding their short-term reproductive plans.

Together, the information in Tables 6-7–6-9 suggests that just under 30% of the female client population in this study indicated prior to counseling that they were not planning pregnancies in either the short or long term. In contrast, some 45% were planning a pregnancy, in either or both the short

and long term, while just under 20% were uncertain. And, clients' reproductive experiences prior to counseling were related to their reproductive plans. Given these reproductive plans, including uncertainty, to what extent did the genetic counseling received by these clients alter, support, or prove irrelevant to their plans?

CHANGES IN CLIENT REPRODUCTIVE PLANS

It is possible for the information, discussion, and counseling clients receive to impact on their reproductive plans in a number of ways. Counseling may, of course, encourage or discourage clients from planning a pregnancy. It may also make them uncertain about what to do reproductively. In addition, counseling may reinforce existing reproductive plans. Or, it may be irrelevant and play no substantial role in either reinforcing or changing client reproductive plans. Focusing on client short-term reproductive plans in this section, we describe how client reproductive plans changed from before to six months following their genetic counseling.

In Chapters 4 and 5 we focused much of our analysis and discussion on pre- and postcounseling assessments, giving limited attention to client knowledge and concerns six months after counseling. We argued that this was appropriate in that the sooner after counseling we assessed client knowledge and reporting of concerns and questions discussed in counseling, the more valid would be our assessment of counseling regarding these issues. Turning attention to the influence of genetic counseling on reproductive plans, we now argue that it is most useful to focus on reproductive plans at six months, not those immediately postcounseling. We think this is so because, as may be recalled, most clients returned their postcounseling questionnaires within a relatively short period of time—one to two weeks—after their counseling. Given the weight and importance of reproductive planning, it may take time for some clients to think through their genetic counseling and what it means for them reproductively. Accordingly we may not be able to assess the impact of counseling on reproductive plans if we examine these plans too soon after counseling. Thus, in the following section we focus most of our analysis on client reproductive plans before and six months after counseling.

Because we felt it necessary to look at clients' reproductive plans through the six-month follow-up, we necessarily had to limit our analysis to those female clients we retained through the six-month period. This reduces our study sample of 1,097 females to 806, or not quite 74% of the original sample. The question arises whether this subset of clients is representative of female clients on whom we have information before counseling.

In Chapter 2 we discussed the issue of study participant retention rates across our data collection periods. This discussion suggested that well-educated female clients were somewhat more likely than less-educated clients to be retained through the six-month follow-up. Nevertheless, we retained in the six-month population a spectrum of more and less well-educated clients, and our population is only slightly biased in terms of educational attainment. In addition and perhaps more central to the issue of reproductive planning, we compared the precounseling female client population with that in the study at the six-month follow-up in terms of 1) short-term reproductive plans, 2) the percent pregnant prior to counseling, and 3) the percent coming to counseling to obtain information to use in their reproductive planning. This anslysis revealed no significant differences between the total study population and those retained through six months. Thus, at least in terms of these three reproductive issues, our six-month sample looks very similar to the total study population.

Of the 806 female clients studied at six months, we ascertained reproductive planning information on 747, or 93%. In this section we are interested in the impact of genetic counseling on change and stability in reproductive plans. Accordingly, it was necessary to remove from the population of 747 clients those who, because of sterilization of either themselves or their spouse, could not have any additional children. Their reproductive plans were not really open to meaningful change and hence they are not of interest here. There were 116 such cases among the 747 clients studied at six months postcounseling. Analysis suggests that 10% of these sterilizations may have been due, in part, to genetic counseling.

In addition, we removed from the six-month follow-up sample all clients who became pregnant between entry into the study and the six-month follow-up. This was necessary because such pregnancies made it impossible to interpret change in client reproductive plans from precounseling to six months postcounseling. A total of 97 clients became pregnant during this period, 12 from the precounseling to immediate postcounseling follow-up, and 85 from the immediate postcounseling period to the six-month follow-up. One of the former and 7 of the latter pregnancies appear attributable, in part, to the counseling the client received.* Seventy-two of these 97 pregnancies were planned and 25 were unplanned.

With sterilized clients and those who became pregnant during the study period removed from the 747, our analysis is based on 534 cases. Table 6-10 presents data on stability and change in these female clients' reproductive plans from before to immediately after counseling. The data are arrayed so we can examine change and stability in light of client's precounseling plans.

*We are preparing for separate publication a detailed analysis of clients who were sterilized or became pregnant during the study.

TABLE 6-10. Changes in Female Client Two-Year Reproductive Plans From Before to After Counseling

	Before Counseling			
Postcounseling	Yes (%)	Unsure (%)	No (%)	Total After (%)
Yes	84.5	22.0	10.1	41.2
Unsure	11.3	69.2	12.7	31.5
No	4.1	8.8	77.2	27.3
Total Before	36.3	34.1	29.6	N=534

T = 92; p = .004.

The table shows that there was a statistically significant shift in the pregnancy plans of clients during this time period. As can be seen, at the population level there was an increase of about 4.9% in planned pregnancies over the percent planned prior to counseling. This translates into an increase of approximately 26 more planned pregnancies after counseling than the 194 planned prior to counseling.

The table also shows that the clients most likely to report changes are those coming into counseling uncertain about what to do reproductively. A majority of these changes are in the direction of planning a pregnancy. Of the clients coming to counseling not planning a pregnancy in the next two years and who change, almost equal percents decided to have a child (10.1%) or became uncertain about their plans (12.7%). Most clients who came planning a pregnancy and who changed became unsure about their reproductive future. It is clear, nevertheless, that even though we have observed a statistically significant positive shift in planned pregnancies, the majority of clients reported the same reproductive plans after as before counseling.

Table 6-11 arrays female client reproductive plans at six months in terms of their precounseling plans. Here too the table shows a statistically significant shift in pregnancy plans, a shift in the same direction as that noted in Table 6-10. There is an increase of about 3.4% in the clients planning a pregnancy at six months compared to the percent with such plans prior to counseling. This amounts to an increase of 18 planned pregnancies from the number planned prior to counseling. There is correspondingly a decrease in the number of clients not planning a pregnancy in the next two years. As before, the group of clients most likely to change are those who came to counseling uncertain about their short-term reproductive plans. At six months, most clients who change from their initial no-pregnancy position report reproductive uncertainty, as is the case among those who came planning a pregnancy and changed their plans.

TABLE 6-11. Changes in Female Client Two-Year Reproductive Plans From Before to Six Months After Counseling

Six Months Postcounseling	Before Counseling			Total Six Months Postcounseling (%)
	Yes (%)	Unsure (%)	No (%)	
Yes	71.4	32.4	9.5	39.7
Unsure	21.1	51.1	27.2	33.1
No	7.7	16.5	63.3	27.2
Total Before	36.3	34.1	29.6	N = 534

T = 147, p = .04.

GENETIC COUNSELING AND ITS INFLUENCE ON CLIENT REPRODUCTIVE PLANS

The information presented in Tables 6-10 and 6-11 reports the total amount of change and stability in client reproductive plans. It is not clear from these data the degree to which we can attribute any or all of the changes to the genetic counseling the clients received. Accordingly, an analysis was performed to describe the influence on six-month reproductive plans attributed to genetic counseling by clients.

In the following discussion we focus our attention on the precounseling to six-month postcounseling period. Our decision for doing this is based on two considerations. First, at the six-month follow-up when we asked clients to indicate whether counseling had influenced their short-term reproductive plans, the question was asked in such a way that it referred to the entire period of time from counseling up to the six-month follow-up, not just the period of time since clients filled out their postcounseling questionnaire. Thus, responses should reflect the influence of counseling whether it occurred during the counseling session or at any time up to the six-month follow-up. Second, and perhaps more important, we anticipated that while some clients would be influenced almost immediately by their counseling others would probably need a period of time to think over the information provided in their counseling before they come to a decision.

Of the 528 clients at six months on whom information was available, 235, or 44%, reported that the genetic counseling they received had influenced their reproductive plans. Conversely, 293, or 56%, said that it had

not. Tables 6-12 and 6-13 present data on the precounseling and six-month reproductive plans of these two groups of clients.

Table 6-12 shows that among those who say their reproductive plans were influenced by counseling there is a statistically significant positive shift in planned pregnancies from precounseling to six months postcounseling. More specifically, while there were 96 planned pregnancies prior to counseling, at six months 117 were planned, an increase of about 22%. The increased pregnancies came almost equally from clients who were unsure or were not planning a pregnancy prior to counseling. Even though influenced by their counseling, the majority of these clients, 58%, report the same reproductive plans at six months that they reported prior to counseling. For these clients, counseling apparently served to reinforce precounseling reproductive plans.

Table 6-13 reports the change and stability in reproductive plans before to six months after counseling among those clients who said that genetic counseling had not influenced their reproductive plans. Here we see no statistically significant net change in the number of clients planning, not planning, or unsure about their plans from before to six months after counseling. This suggests that almost all of the shift in pregnancy plans reported for the total population of clients in Table 6-11 can be attributed to those clients who were influenced by their genetic counseling.

Sixty-five percent of the clients not influenced by their counseling report identical reproductive plans prior to and six months after counseling. However, while 35% report changes over this period, the marginal percentages in Table 6-13 suggest these changes were such as to lead to no significant net shift in the number of planned pregnancies.

TABLE 6-12. Reproductive Plans Before and Six Months After Counseling for Clients Influenced by Their Counseling

	Before Counseling			Total Six Months
Six Months After Counseling	Yes (%)	Unsure (%)	No (%)	After Counseling (%)
Yes	76.0	37.2	20.0	49.8
Unsure	20.8	47.9	37.8	34.9
No	3.1	14.9	42.2	15.3
Total Before	40.9	40.0	19.1	N = 235

T = 70, p = .006.

TABLE 6-13. Reproductive Plans Before and Six Months After Counseling for Clients Not Influenced by Their Counseling

Six Months After Counseling	Before Counseling: Yes (%)	Before Counseling: Unsure (%)	Before Counseling: No (%)	Total Six Months After Counseling (%)
Yes	66.3	27.6	5.6	32.4
Unsure	21.4	55.2	22.2	35.8
No	12.2	17.2	72.2	31.7
Total Before	33.4	29.7	36.9	N = 293

T = 60, p = NS.

Table 6-14 combines Tables 6-12 and 6-13 to permit an overall view of change and stability in influenced and noninfluenced clients from precounseling to six months postcounseling. As noted, in both cases most clients reported the same reproductive plans at six months as they reported prior to genetic counseling. This was slightly truer for those clients not influenced by counseling than for those who were. Among clients influenced, the major impact of counseling was to reinforce precounseling reproductive plans. Of those influenced by their counseling and who changed, the largest number were "encouraged," that is, they either said they were unsure or were not planning a pregnancy in the next two years prior to counseling and at six months reported that they were planning a pregnancy.

Among those clients not influenced by counseling, a plurality of those who changed became reproductively uncertain, while almost equal numbers were planning to have or not have a child in the next two years by the six-month follow-up.

The data in Tables 6-10 to 6-14 report the reproductive intentions or plans of the clients in this study, not actual pregnancies, of course. We did gather information on the contraceptive behavior of these clients at six months, and they reveal a marked relationship between reproductive plans and contraceptive behavior. For example, while 82% of those not planning or unsure about pregnancy in the next two years were using some form of contraception at our six-month follow-up, only 46% of those planning a pregnancy were ($\chi^2 = 64.8$, $p < .001$). Most of those using some form of contraception were using the pill, 33.4%; condom, 17.7%; IUD, 13.0%; or diaphram, 12.6%. Only 13, or 4%, of the clients employing contraception were using a highly unreliable method such as withdrawal or rhythm. Thirty-six percent of those clients who were reproductively

TABLE 6-14. Female Clients' Reproductive Plans Six Months After Counseling and Role of Counseling in Those Plans

	Role of Counseling											
	Influenced Plans (235)						Did Not Influence Plans (293)					
Change-Stability in Reproductive Plans T_1–T_3	No Change			Changed to			No Change			Changed to		
Reproductive Plan at T_3	Yes	Unsure	No	Yes	Unsure	No	Yes	Unsure	No	Yes	Unsure	No
Percent	13.8	8.5	3.6	8.3	7.0	3.2	12.3	9.1	14.8	5.7	8.5	5.1
N	73	45	19	44	37	17	65	48	78	30	45	27

unsure or not planning a pregnancy in the next two years and were not using some form of contraception reported that they had recently had a baby and did not consider themselves fertile. Seventeen percent were sexually inactive and 17% were opposed to birth control.

Among those clients who were planning a child in the next two years and who were not using contraception, 90% said they were trying to get pregnant. Thus, the data suggest that just under half, 45%, of those planning a pregnancy were trying to get pregnant by six-months postcounseling, and the remainder, 55%, were delaying their planned pregnancy. For those not planning a child in the next two years, four out of five were using some form of contraception, usually a highly reliable method. These data suggest that there may ultimately be a significant relationship between the reproductive plans we have charted in the tables above and the actual reproductive behavior of these clients.

An additional analysis was performed looking at those clients who came to counseling specifically seeking information to help them make a reproductive decision. It may be recalled that approximately 53% of the clients entering the study reported this as one of their reasons for obtaining genetic counseling. Our analysis of clients coming to obtain information to assist in reproductive planning revealed virtually identical results to those reported in Tables 6-10 and 6-11. A majority of these clients, 55.5%, said that counseling had been useful reproductively in that it influenced their reproductive decisions. The influence was very similar to that portrayed in Tables 6-13 and 6-14. Most female clients reported the same reproductive intentions at six months as they did prior to counseling. Clients who changed their plans more often planned a child than decided against a pregnancy or became reproductively uncertain.

It is the case that 44.5% of the clients who came to counseling to obtain information to assist in their reproductive planning said that the counseling they received had not influenced their reproductive plans. This suggests that counseling was not as useful in providing reproductively relevant information for these clients as the client might have expected. An analysis of the level of diagnostic and risk knowledge of clients who were influenced and not influenced reproductively by their counseling revealed that they were equally knowledgeable. Nearly 90% of both groups knew the diagnosis of the problem they had discussed in counseling, and of those given a numeric risk in their counseling about 45% of both groups knew what it was at six months. It would appear, thus, that such knowledge does not distinguish between those who were influenced by their counseling reproductively and those who were not.

An additional analysis revealed that clients who claimed to have been reproductively influenced by their counseling were more likely to have

discussed the issues they came to counseling specifically to discuss than clients who were not reproductively influenced. This was especially so with respect to discussion of genetic-medical questions and concerns. Thus, discussion of the clients' questions and concerns is related to how useful clients found counseling to be in planning their reproductive futures. The fact that on average only about 55% of clients' genetic-medical and 16% of their sociomedical questions and concerns were discussed in depth in counseling may help explain why 44% of the clients found their genetic counseling to be unrelated to their reproductive planning, although this was one of their reasons for obtaining counseling.

SUMMARY

This chapter has looked at female client reproductive planning from two perspectives. First, we examined the various factors that clients say are very important in their reproductive planning process. Second, we looked at change and stability in client short-term reproductive plans over the duration of our study period. We will briefly summarize our major findings here.

Our analysis and discussion of the context from which the female clients in this study viewed their reproductive planning suggested several conclusions. Many different types of considerations are reviewed in reproductive planning by clients, some intensely personal, some medical, some practical in nature. Across the six months of the study, and even among various subgroups within our study population, four considerations were the most frequently cited as very important in client reproductive planning. These were 1) fulfillment as a parent, 2) care of other children, 3) wishes of one's spouse, and 4) a doctor's advice. This is obviously a mixed set of considerations to bring together, suggesting that clients employ a complex calculus to arrive at any given decision, a calculus unquestionably influenced by medical criteria, but a calculus set in a context of psychologic as well as social considerations. From a client's perspective, an informed reproductive decision would necessarily entail such considerations.

Clients reported that they considered numerous additional issues in their reproductive planning, such as finances, career goals, and religious or ethical beliefs. These additional factors, less frequently cited as very important than the above four, nevertheless add to the complexity of client reproductive planning.

Turning to the impact of counseling on client reproductive plans, our data suggest that for 56% of the clients in this study followed through six months the counseling they received was viewed as not bearing on their

reproductive plans. For 44% of the clients, counseling was a useful experience, reproductively, in that they report it influenced their reproductive plans. Among these clients, counseling served most often to reinforce reproductive plans existing before counseling. Among those who showed changes and said that counseling influenced them, the largest number reported that they had decided to have a child in the next two years because of their counseling. Of the remainder, most became uncertain, while a minority were discouraged reproductively by their counseling.

Analysis of clients who came to counseling specifically to obtain information to assist in their reproductive planning revealed that clients who were influenced reproductively in their counseling were no more knowledgeable about diagnostic or risk information than those not influenced by their counseling. However, clients who were influenced reported that significantly more of their questions and concerns were discussed in counseling than clients who reported counseling did not influence their reproductive plans. This suggests that it is not so much knowledge per se, not even such information as diagnosis and risk information, that may make genetic counseling invariably useful reproductively for clients. It is also discussion of their various questions and concerns.

The literature on genetic counseling is replete with references to genetic counseling and client decision making [4, 5]. Throughout this chapter we have attempted to avoid the phrase "reproductive decision making" and in its place have used the notion of reproductive planning. To speak of client reproductive decision making gives an image, to some degree, of a point-in-time decision event, with an aura of finality or rigidity. Planning, on the other hand, connotes direction, sometimes considerable commitment, but at the same time a flexibility reflecting the reality of trying to act in a situation of complexity and uncertainty. While undoubtedly some genetic counseling clients do make quick decisions, sometimes in the counseling session itself, it is also the case that some find it requires time to make a decision, to plan their reproductive futures.

To the degree that this is the case, it would appear that achievement of one of the goals in the *American Journal of Human Genetics* definition of genetic counseling—". . . to help the individual or the family . . . choose the course of (reproductive) action which seems appropriate to them in view of their risk and their family goals . . ."—may be a difficult task to accomplish in the counseling session [3]. We saw in Chapter 3 that this goal was the one least likely to be strongly supported by the counselors in this study, although the majority did view it as important for their counseling. In addition, in this study only 25% of the clients reported that they discussed in depth with their counselor the issue of whether or not to have a child. Given the complexity of considerations which we found clients using in

their reproductive planning, as well as the fact that many clients may need some time to think about their reproductive futures, helping clients actually make their reproductive decisions is a task that may go beyond the scope of genetic counselors and the resources currently available for genetic counseling.

What our data do suggest is that counselors can increase the utility of genetic counseling for client reproductive planning by discussing in depth the various questions and concerns clients bring to genetic counseling. Not discussing these issues lessens the impact of genetic counseling on client reproductive planning, as may singular attention by the counselor to educating his or her client about such basic but, from the clients' perspective, perhaps not so central issues as diagnosis and risk information.

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Chapter 7

Medical Genetics and Genetic Counseling

INTRODUCTION

In Chapter 1 we provided statistics on the incidence of birth defects, noting that they constitute a significant public health issue with some 150,000 to 200,000 children born in the United States each year with various diseases and disorders. However, statistics such as these, which focus our attention on affected individuals, provide only a partial understanding of the impact of birth defects. A more complete picture of the significance of birth defects becomes apparent when we realize that each of these children is born into a family—one which may consist of only the new parents and the child, but may sometimes include other children. Of course, these families belong to larger family networks, involving brothers, sisters, uncles, and aunts. In short, looking just at immediate family members, the birth of a child with a defect involves, then, not just the affected child, but families. From this perspective we are not talking about only 150,000 to 200,000 children, but this many families, involving perhaps as many as one-half to three-quarters of a million people.

It is from these families, with their plans, expectations, and hopes for the future, that genetic counseling clients come. As we have seen, they bring with them not only questions about why and how a birth defect happens, or might happen, but questions about what can be done, concerns about how to care for an affected child, and also questions about their families, its resources and relationships within the family. The spectrum of genetic-medical and sociomedical concerns and questions that clients come to counseling to discuss with their genetic counselor reflects this. The variety of these issues raises some basic questions about the role or limits of genetic counseling, the training of its practitioners, and the various responsibilities of both the professionals and the clients in this rapidly evolving medical service.

In this volume we have presented the major features and results of a prospective, longitudinal study of genetic counseling. Our study population, which is described in detail in Chapter 2, consisted of over 1,000 female clients, the 205 professionals whom they saw for genetic counseling, and the 47 clinics in which these medical encounters took place. In defining the scope

and limits of our study we realized that even with substantial resources we could not study all the various facets of genetic counseling we believed should be studied. Accordingly, this study should not be viewed as a comprehensive or global assessment of genetic counseling in all its various ramifications.

Our broad objective in conducting this study was to assess the "effectiveness" of genetic counseling. Given current definitions of the content and goals of genetic counseling, and the limited knowledge of its operation and outcomes provided by previously published studies, our study was designed to address three major topics that we deemed essential to assessing the effectiveness of this medical service. These three topics were: 1) the types of questions and concerns that clients bring to counseling, and the degree to which counseling addresses these client-defined needs or objectives; 2) how well counseling succeeds in educating clients about two of the most salient aspects of a genetic disorder—its diagnosis and recurrence risk; and 3) how counseling impacts on the reproductive plans of its clients.

We will not, in this concluding chapter, recapitulate our findings in detail; they are summarized at the ends of Chapters 4, 5, and 6. Our focus, rather, will be on the question that generated the study: How effective is the service known as genetic or birth defects counseling? In addressing this question we will, in part, explore the use of the term "counseling," and, in light of our data, offer a series of suggestions about the ways that the teaching and counseling aspects of this relatively new medical service could be strengthened.

IS GENETIC COUNSELING EFFECTIVE?

To the question "Is genetic counseling effective?" we would respond that there is no simple answer. Rather, the answer or answers are situationally bound, and must be addressed in terms of understanding why counseling is sought, the objectives of its providers, what the service consists of, and how it impacts on its recipients.

Both clients and those who counsel them, as we have reported, have various objectives or goals for their participation in genetic counseling. It is our view, as we explained in describing the rationale for the design of our study, that the effectiveness of genetic counseling thus must be assessed in multiple ways that relate to the service's meeting the objectives of both its recipients and its providers.

In Chapter 3, we saw that the counselors in our study are strongly committed to what we described as the intertwined educative and counseling aspects of genetic counseling. They strongly endorsed the importance of helping individuals or families understand the diagnosis, etiology, prognosis, management, risk of recurrence, and options for dealing with that risk. They

reported that they were only slightly less strongly committed to the tasks of helping a client adjust to, or cope with, a disorder in a family member or the recurrence risk faced by a client. In terms of educative and counseling tasks, these counselors gave the least support to a role that involves them as decision-facilitators for their clients. In describing this task as one that received the "least support," however, we should note that over 80% of the counselors viewed it as of moderate to high importance. That it was not endorsed as strongly as other counseling objectives may be due to the fact that it can be linked with or imply a counseling role that our counselors did not endorse: that of directly advising or actually telling clients what to do.

As ascertained by our precounseling questionnaire, the objectives of clients in seeking genetic counseling are highly congruent with what counselors see as their most important educative and counseling goals. That is, the clients' questions and concerns deal, most predominantly, with what we termed in Chapter 4 "genetic-medical issues." In terms of the percent of clients who wanted to discuss them, these issues concerned etiology, risk, medical treatment of a disease or disorder, diagnosis, prognosis, and caring for an affected child. Significantly fewer clients stated that they wanted to discuss what we called "sociomedical" issues with their genetic counselor, such as their feelings about an affected child, schooling or other special programs, or the financial costs of a genetic disease. But a sizable number of clients did have such concerns.

We recognize that there are limitations in the use of questionnaire data to ascertain both counselor and client objectives. Our sets of questions to counselors, at best, can give us simple indicators of complex attitudes about their tasks. Similarly, our questions to clients, while more situationally focused than the questions to counselors about their general objectives, necessarily reduce complex feelings, questions, and needs to a uniform checklist of possible responses.

Within these limitations, however, the study's prospective, longitudinal design did enable us to examine the goals and objectives of both counselors and clients in relationship to several aspects of counseling sessions and their outcomes.

In looking at the content of genetic counseling and the various outcomes we have examined in this study, it is apparent that one cannot neatly separate the "educative" and "counseling" functions of this complex medical service. Imparting information, whether regarding a recurrence risk, a diagnosis, or a prognosis, and understanding what that information means to clients is an integral aspect of counseling. Discussing with clients their questions and concerns, whether genetic-medical or sociomedical, as defined by the clients, is also an activity that can counsel, reassure, or advise clients, as well as provide them with information.

From the analysis presented in Chapter 4 it is apparent that clients bring an array of genetic-medical and sociomedical questions and concerns to discuss with their genetic counselor. Our analysis showed that, on average, about 55% of the genetic-medical and only about 16% of the sociomedical questions and concerns clients specifically came to discuss were discussed. It is important in discussing these figures to draw attention to the fact that nearly half—45%—of the genetic-medical questions and concerns clients brought to counseling were not discussed in depth in counseling. Several important medical topics, such as the prognosis of a disease or disorder, and its treatment, were more likely not to be discussed than discussed. Given the congruence of goals for counseling by counselors and clients, it is striking how many issues are not discussed in the counseling session.

In short, genetic counseling, from the perspective of the clients involved, is not addressing a significant number of their genetic-medical concerns, and relatively few of their sociomedical concerns. Of course, some of this information is not known, but some of this information could be better organized and made available to counselors and their clients. To the degree that genetic counseling is defined as an activity which should address the array of questions and concerns clients have, it is necessary to increase the general availability of much genetic-medical and sociomedical information to both counselors and clients.

Making such information available does not ensure that it will be used and transmitted in genetic counseling. More specifically, our analysis suggests that at this time many counselors tend to define the interests of their clients in terms of recurrence risks and diagnostic or prognostic information, even when clients have different interests. Our analysis would suggest that counselors should be more sensitive to the major questions and concerns of their clients, and, to the degree possible, address these questions and concerns with the most useful information available.

Another way of stating the above is that there is frequently a mismatch between the "agenda" the client brings to genetic counseling and that of the professional. As is customary in many contemporary medical encounters, it appears that it is the agenda of the professional, more so than that of the client, that is instrumental in defining what actually transpires. Whose responsibility is it to raise various types of issues for discussion in this medical encounter? While we might assume that counselors would usually cover most genetic-medical concerns of their clients, this is not the case. And, while we might assume that clients would raise not only their genetic-medical concerns, but also their sociomedical concerns, this does not appear to be the case either.

While there is unquestionably a burden on the client to raise his or her concerns, to the degree that genetic counseling involves "counseling," there

appears to be some obligation on the part of the counselor to attempt to explore, elicit, or surface and discuss the concerns of his or her clients. The notion of counseling imparts some professional obligation to provide a context in which clients feel comfortable raising and discussing the issues that are of concern to them.

In examining some of the other major findings of this study, looking first at client knowledge, the fact that 54% of clients given a risk and approximately 40% of clients given a diagnosis were not able to report it almost immediately after counseling is a complex issue involving both problems in the counselors' abilities to educate and the clients' abilities to learn. Our analysis of the various factors related to client diagnostic and risk knowledge after counseling suggests that it would be unproductive to lodge responsibility of the less than perfect knowledge of clients with either the counselors or the clients. We suspect that had we assessed other types of client knowledge we would have obtained similar results, findings we suspect not peculiar to genetic counseling. Regardless, our analysis suggested that genetic counseling as an educative activity should be made substantially more effective, and there are numerous activities counselors can do, as outlined below, involving not only themselves but their clients, that should make counseling more effective.

With respect to genetic counseling and client reproductive planning, our analysis, which focused on reproductive plans, not pregnancies or births, showed that there were significant changes in client reproductive plans over the period of the study and the counseling the clients received was perceived as playing a significant role in many of these changes. Our analysis suggested that the most significant role of counseling was to reinforce reproductive plans clients held upon entrance to counseling. For those clients whose plans changed due to their counseling, it most often led to a significant increase in the number of planned pregnancies, an increase of just over 20%. In addition, we noted in our analysis that about 53% of the study population came to counseling to obtain information to use in planning pregnancies. The remainder did not, and rarely, only 6% of the time, did the latter report that their reproductive plans were influenced by their counseling. Conversely, counseling influenced the plans of 56% of the clients coming to get information to make reproductive decisions and whom we followed for six months. Here again the major role counseling played was to reinforce the reproductive plans clients held prior to counseling and to increase significantly the number of planned pregnancies.

It is difficult, as we discussed in Chapter 6, to say what effective counseling is regarding client reproductive plans. Our data say that about half of those who come to counseling to obtain reproductively relevant information report that they did, in that counseling influenced their plans. However, al-

most half of those approaching counseling to help them make reproductive decisions found it not directly relevant to such planning. It is clear that clients who had an opportunity to explore and discuss the questions and concerns they brought to counseling were significantly more likely to report that counseling had influenced their reproductive plans than those who did not have this opportunity. In short, discussion of a client's agenda significantly increases the utility of counseling for client reproductive planning.

There is, as we have noted at several places in this report, no single or simple answer to the question, "Is genetic counseling effective?" In many respects, an overall assessment of the effectiveness of counseling, at least the counseling we assessed in this study, is confronted with the problem of whether the glass is half full or half empty. That is, about half of the clients who could have learned their risk did, but about half did not. And, over half of the clients who could have learned their diagnosis did, but the remainder did not. In a similar vein, clients report that just over half of their genetic-medical questions and concerns were discussed, but about half were not. The picture for sociomedical concerns and questions was markedly worse however. And, reproductively, just over half of those coming to counseling to obtain information to use in making their reproductive plans reported counseling influenced these plans, but about half did not. Any overall assessment must point to the fact that counseling has been effective for many clients, but ineffective or irrelevant for an almost equal number. In short, while counseling is accomplishing much, there clearly is room for substantial improvement. Given the various problems and limitations we have noted, how can counseling be made more effective?

TOWARD MORE EFFECTIVE GENETIC COUNSELING*

Given the magnitude of the medical, personal, and social problems associated with birth defects and genetic disorders, and the expressed reasons that individuals seek genetic counseling, it is clear to us that genetic counseling is a needed and important facet of medical care delivery services. In this section, we offer a number of suggestions about ways that the educative and counseling components of genetic counseling might be strengthened to increase its effectiveness in terms of meeting the objectives of both its clients and providers.

TEACHING AND LEARNING MEDICAL-GENETIC INFORMATION

Clearly, there is a strong professional consensus that the teaching and learning of diagnostic and risk information is at the center of genetic coun-

*Excerpted and expanded from "Client Learning of Risk and Diagnosis in Genetic Counseling" [1].

seling. Genetic counselors are the medical care providers uniquely qualified to determine and provide such information, and therefore it is essential that they be highly skilled in teaching clients risk and diagnostic information. Our data strongly indicate that there is room for improvement in the interactive processes of counselor teaching and client learning.

In concluding that counseling as an educative encounter is only moderately successful, we recognize that the "learning rates" of the clients in our study appear to be comparable to the amount of learning which takes place in other types of medical encounters [2]. However, when one weighs the importance of the family decisions that can be premised on the information learned or not learned in genetic counseling, it seems that an effort should be made to strengthen this aspect of counseling. This position takes on added importance when we realize that in our study only 40% of the clients who had not learned their risk planned to see a genetic counselor again.

Our comments about what factors contribute to a teaching-learning problem in genetic counseling, and what might be done to mitigate it, are organized according to the three general categories of factors we examined: the structure and process of counseling, counselor characteristics, and client characteristics.

The Provision of Counseling

In terms of client education, two observations stand out from our data on the structure and process of counseling in the clinics we studied. First, the data imply that support professionals, such as social workers and genetic associates, are not making any significant or only very small differences in terms of client education. If facilitating client education is viewed as one of their functions, it would be useful to explore better ways of utilizing such adjunct professionals in the counseling process.

Second, contrary to what one might expect, the study suggests that increasing the time spent with clients does not necessarily improve learning. While spending increased time with clients may have other benefits, such as increased discussion of client questions and concerns, it is not a simple solution to the problem of client education.

Looking at the counseling process, we offer the following three suggestions for increasing the effectiveness of counselor teaching and client learning. First, client knowledge of risk and diagnosis should routinely be ascertained by the counselor at the beginning and end of each session. This strategy would disclose whether the task confronting the counselor is to reinforce existing knowledge, to instill new knowledge, or, more complexly, client reeducation. In addition, ascertaining client knowledge at the end of the session would allow for a test of learning and an opportunity to reinforce correct learning or to correct misinformation.

Second, a variety of postcounseling, client follow-up procedures could be used to reinforce client knowledge of diagnosis and risk. For example, a telephone call to clients by the counselor or other clinic staff a week or two after counseling could reinforce existing knowledge, as well as provide for correction of misinformation or of incorrect changes in a client's understanding. At the same time, such feedback could serve to educate counselors about their effectiveness, as well as other problems clients may be having with the information received in counseling.

Third, our study revealed that 14% of all risks given to clients were nonnumeric risks. That is, the counselor did not give a risk of 0.5% or 25%, but instead told clients they faced a high, moderate, or low risk. We think serious consideration should be given to the merits of providing genetic counseling clients with nonnumeric risks. Since such qualitative statements involve a variety of individual value judgments, people may have very different interpretations of whether a given numeric risk is high, medium, or low.

Counselor Characteristics

Three findings relevant to client education stand out from our data on genetic counselors. First, in terms of the question that has arisen as to who can counsel "best" [3], we have seen that there is no significant difference in the educative abilities of the three major groups of professionals providing counseling—MDs, PhDs, and masters-level genetic associates. Assuming that counselors agree that client education is an essential task, it appears that despite their different educational backgrounds these three types of professionals are equally effective (or ineffective) as educators. This observation suggests that in the training of each of these groups of professionals more attention needs to be given to the issue of client education and techniques for improving learning. Since each of these groups of professionals has particular skills, it would be worthwhile to explore better ways to coordinate these skills to maximize the amount of client learning.

Second, most counselors may be surprised to learn that, in our study, experienced counselors did not educate clients significantly better than their inexperienced counterparts. Since the field of genetic counseling has been expanding rapidly in recent years, one might point to the presence of many relatively inexperienced professionals as a reason for the shortcomings in client education. However, our data indicate that this is not the problem.

The finding that experienced counselors generally are no more effective than inexperienced counselors has at least two implications. It should alert those who have been involved in genetic counseling for some time that they may need to reassess their educational effectiveness. And, it follows that it may be misleading to use those who are established in the field of genetic counseling as clinical role models, at least in terms of client education. The

data do not mean, of course, that there are no experienced counselors who are effective. Rather, they suggest that we cannot simply take experience as an indicator of educational effectiveness.

Third, several observations suggest that counselors, regardless of their training, are more sensitive to a client's diagnostic knowledge than to his or her risk knowledge. While one can debate the relative importance of clients' knowing their diagnosis versus their risk of having a child with a birth defect, it seems clear that if counselors want to improve client learning of risk, they need to become as sensitive to it as they are to whether or not the client knows the appropriate diagnosis.

Client Characteristics

In many respects client characteristics and their relationship to learning are the most complex and interesting of our findings. As one would expect, a client's educational level was found to be related to his or her level of knowledge upon entrance into counseling. Contrary to expectation, however, client educational level was not related to learning during counseling. Thus, counselors should not assume that the well-educated client will have little or no trouble absorbing the information presented during counseling.

Clients who came to counseling to learn their risk and diagnosis were more likely to do so than those who did not come with these objectives. Interestingly, clients who came to counseling without prior knowledge generally learned better than those who came with misinformation. This suggests the importance of assessing what the client knows early in the counseling session. Depending on the client's knowledge state, the counselor's role may not be simply to educate, but the more complex task of reeducation.

Two further observations suggest the role of prior experience in client learning, and also reflect the fact that client beliefs, whether valid or not, can play a significant role in learning. Our data show that clients who had been pregnant were somewhat more likely to learn than those who had not. In addition, if the pregnancy experience was in some way compromised, ranging from a stillbirth to the birth of a child with a problem, this experience seemed to especially sensitize clients to learning. Thus, the counselor might want to make sure that he or she considers the reproductive history of a client as a factor related to educating that client.

Turning to the role of client beliefs, we found that clients who believed that the problem they were being counseled for was inherited (whether it actually was or was not) were more likely to learn than those who did not. Perhaps this belief increases one's sense of responsibility, and in so doing, sensitizes one to learning.

At the same time, clients who believed that they were at moderate or high risks were markedly less likely to have learned than those who believed they

were at lower risk, regardless of the validity of the beliefs. This suggests that perhaps the belief that one is at a high risk is so threatening that learning is suppressed. Once again, we can see the importance of an accurate assessment by the counselor of the client's risk knowledge. If the client erroneously believes his or her risk to be a high one, the counselor's reassurance may enable more accurate learning. In cases where the client's risk is truly high, the counselor should be alerted to the fact that greater educative efforts than usual may be indicated.

Counseling Clients

If one accepts a "broader" definition of genetic counseling as encompassing more than ascertaining and conveying medical-genetic information to clients, it follows that counseling should also deal with what we have termed "sociomedical" topics. These topics were discussed in Chapter 4 in terms of client-defined needs or objectives in seeking counseling. As we saw in Chapter 3, these topics also are an explicit component of what the professionals engaged in counseling see as their task, in terms of such expressed goals as helping individuals or couples adjust to and cope with their genetic problems, removing or lessening guilt or anxiety, and helping clients achieve their parenting goals.

Our study revealed that, in general, while counselors do a fair job of discussing the medical questions clients bring to counseling, they do a poor job of discussing sociomedical issues and problems. For clients, these issues and concerns are neither trivial nor peripheral. Rather, they are centrally important aspects of the genetic problems that have brought them into genetic counseling.

For a counselor to provide such client-centered sociomedical information does not mean either that the counselor should be expected to give advice or solve a client's psychosocial problems. Rather, as is well documented in the literature, a major function for patients of their interaction with physicians is to have a knowledgeable person with whom one can share concerns and gain insight and support.

Given these observations, we suggest that efforts be undertaken to systematically increase and improve the counseling aspects of genetic counseling. This may be achieved through a variety of mechanisms; for example, provide a mechanism by which counselors can learn the clients' questions and concerns prior to counseling. A simple checklist, such as that used in our study, could routinely be completed by clients before counseling, given to the counselor, and used as a basis for discussion. In addition, adequate time should be scheduled for each counseling session to permit discussion of both clients' and professionals' agendas. It would appear that supervised clinical

training by senior medical geneticists is inadequate for learning the substantive content or techniques needed for effective counseling of sociomedical topics, as well as for some genetic-medical topics. Therefore, some training of medical geneticists in counseling techniques is essential.

An alternative strategy for meeting the counseling needs of clients is to utilize other professionals trained specifically in medical-genetic counseling. Some clinics currently have personnel who could perform this function (eg, social workers or genetic associates) if such personnel were so trained and if clinics better utilized a team approach. Other clinics, where counseling services essentially are a one-person operation, would need to add such staff, or, if available, utilize other resources in their institution. Additionally, clinics should better utilize an array of other community resources to help clients cope more effectively with their sociomedical needs: for example, community social service agencies, mental health centers, parents' groups, self-help groups, and various local, state, and federal agencies.

Discussion of prognosis provides an example of a client concern that has both medical and sociomedical components. We found that only about 40% of clients who wanted to discuss the prognosis for a specific disease or disorder report that prognosis was discussed in depth. Given the potentially significant role that information of this kind can play in making informed decisions about raising or having an affected child, we feel that it is a counselor's responsibility to routinely include this topic.

We realize that these suggestions to improve genetic counseling are made without full consideration of the many constraints which exist in the medical care world. We know, for example, that recommending that counselors spend more time with their clients may be difficult, if not impossible, in terms of their client load and the other activities that consume a major portion of their time. Likewise, other strategies we have mentioned are likely to raise the cost of services, and we do not suggest where these additional funds might be found. Further, no matter how much time and training is provided to counselors to improve their expertise, there well may be a limit to the educability of some number of clients. Therefore, to implement these recommendations it would be necessary to identify many constraints and to develop alternative methods for overcoming obstacles in the delivery of genetic counseling. If counseling is not strengthened, however, our data indicate that a significant number of clients will leave counseling not having learned such medical-genetic information as diagnosis and risk, without having had the opportunity to discuss a variety of both genetic-medical and sociomedical concerns and questions they bring to counseling, and without having learned specific methods of dealing with a host of psychologic, social, and economic aspects of the genetic disease with which they have to cope.

COUNSELING AND MEDICAL GENETICS

Genetic counseling, the mother of a child with a chromosomal defect told us, is

> an attempt by geneticists to find the reason why or to explore all of the avenues to see what the cause is. . .or the cause and effect; and also to tell you something about the particular genetic problem, to give you precise information if it is available. I think when I first heard "genetic counseling" I thought I was going to come here and they were going to say "this is what a trisomy twenty is, and this is what you have to look forward to, and on day number 99, this is going to be the result, et cetera." And it's not. I think in this world today, we all sit there and say something is black or white. But there also has always to exist the gray(s).

In the preceding chapters we have presented data about individuals in search of knowledge and help. The information they sought, the problems they confronted, and the kinds of help they wanted, were diverse. But in common, all were sent, or turned on their own initiative, to the same source —a genetic counselor. The goal of those counselors was to help their clients learn about and deal with the medical-genetic and related psychosocial problems that confronted them. But counselors, too, must work with many shades of gray; they often are unable to answer questions of "whether" or "why." Given both inherent limitations in the state of the art of applied human genetics, and in the knowledge and skills of any provider of genetic service, counselors and clients alike must confront and come to terms with many problems of uncertainty. In some instances, clients' questions about risk, etiology, and prognosis can be answered with certainty. In other situations, clients learn, in the words of a parent we interviewed, that "there are no answers." Uncertainty often cannot be removed, nor can it always be reduced. It can, sometimes, be restructured by the ways that counseling helps clients understand and deal with the "grays." Sometimes, counseling unwittingly serves to create new uncertainty for its clientele, raising problems of which those seeking counseling had not anticipated or been aware.

The problems of uncertainty that confront both clients and providers of the service called genetic counseling, a service that deals with the most medically, personally, and socially complex aspects of our "private biology," suggest how difficult it is to define in any simple way what genetic counseling does or should consist of, or how effective it is.

Considering how our data address the question of genetic counseling's effectiveness, and the consequent suggestions we have made about ways that the provision of these services could be strengthened, we end this monograph as we began it: by considering how genetic counseling is or could be defined. How data are interpreted, and the types of recommendations we have made based on them, closely turn upon how one views the nature and goals of genetic counseling.

In Chapter 1 we discussed a widely cited definition of genetic counseling [4], one that incorporates the thrust of many other definitions and provides a more specific elaboration than most of this service's educative and counseling objectives. This definition of genetic counseling is:

> . . .a communication process which deals with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or the family to (1) comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management; (2) appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives; (3) understand the options for dealing with the risk of recurrence; (4) choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision; and (5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

A more succinct but comparable statement about the content and objectives of genetic counseling, which along with the above definition provided a useful starting point in defining the focus and scope of our study, is that provided by the March of Dimes Birth Defects Foundation [5]:

> Genetic counseling provides and interprets medical information based on expanding knowledge of human genetics, the branch of science concerned with heredity. Its major goal is to convey understanding of birth defects to affected families and enable prospective parents to make informed decisions about childbearing.

According to this statement, the content of genetic counseling consists of providing and interpreting medical-genetic information. The goals or objectives of counseling, in turn, are twofold: first, to convey an understanding of birth defects to those who already have an affected child; and second, to enable prospective parents to make informed reproductive decisions.

In terms of this and other comparable statements, what is done in the service called "genetic counseling" by professionals called "counselors" can have either of two major foci. First, this activity can concentrate almost exclusively on technical medical-genetic information; eg diagnosing a proband, determining etiology and risk, assessing the probable or possible prognosis, and conveying this information to clients and, when indicated, a referring physician. This medically focused work is, in fact, what the majority of counselors (eg physicians), by and large, are uniquely equipped to do through their specialized didactic and clinical training in applied human genetics. If this medical-genetic focus is deemed to be the major, if not exclusive, objective of what is termed genetic counseling, what clients should expect to receive from their contact with this area of medical service might bet-

ter be defined as a medical-genetic consult or seeing a medical geneticist.

The word "counselor" in turn implies a broader range of client expectations and professional responsibilities than does "medical-geneticist." The word counsel means to give advice or propose a policy or plan of action or behavior, and this type of activist professional orientation, we would argue, is what most persons expect when they consult a job, marriage, guidance, or other counselor. For reasons that in part are tied to the historical development of applied human genetics, however, the majority of those engaged in genetic counseling feel strongly that "giving advice" to clients about reproductive or other decisions related to genetic disorders is an inappropriate professional role. Nonetheless, even with this more "passive" orientation, counseling connotes dealing with the broad range of problems, sociomedical as well as medical-genetic, that surround genetic disorders. Looking at the March of Dimes' definition of genetic counseling in this context, one can thus view "interpreting medical-genetic information" and enabling parents to make "informed" reproductive decisions as indeed mandating that this service involve a broad-based counseling approach, rather than a more narrow medical-genetic orientation.

To return to the point we emphasized at the beginning of this section, how our data are viewed in terms of addressing the effectiveness of what is termed genetic counseling, depends, in part, on whether the providers and recipients of this service primarily see themselves as engaged in medical-genetics, or, in a more encompassing sense, in genetic counseling. To the extent that most providers of genetic counseling accept a broad definition of their tasks and goals, they assume the authority and the responsibility for providing those who seek their services with both medical-genetic and counseling expertise. To the extent that a narrower definition is accepted, responsibility remains, as a "helping profession," to see that clients are referred to or put in contact with appropriate professional and paraprofessional resources, for the questions and problems clients bring to genetic counseling, whether of a genetic-medical or sociomedical nature, reflect their real life concerns. For clients to make "informed decisions" and to live with their reproductive pasts and plan their reproductive futures, their questions and concerns must be addressed.

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APPENDIX 1
COUNSELOR QUESTIONNAIRES

QUESTIONNAIRE FOR COUNSELORS

The National Foundation – March of Dimes
Genetic Counseling Study
Department of Socio-Medical Sciences
and Community Medicine
Boston University School of Medicine
Boston, Massachusetts 02118

FIRST, WE WOULD LIKE SOME INFORMATION ON YOUR CAREER AS A GENETIC COUNSELOR.

1. Which of the following degrees do you hold? (Check as many as apply)
   ( ) M.D. (specify):
   ( )$_1$Pediatrician ( )$_2$Ob-Gyn ( )$_3$Internist Other (specify) _____
   ( ) Ph.D. (specify field) _____
   ( ) Master's degree (specify field) _____
   ( ) R.N.
   Other (specify field and degree) _____

2. How long have you been providing genetic counseling? _____ year(s)

3. How long have you been providing counseling in this clinic? _____ year(s)

4. What training have you had in human genetics? (Check as many as apply.)
   ( )Didactic course in genetics
   ( ) Supervised clinical training
   Other (specify) _____
   ( ) None

5. What training have you had in counseling techniques? (Check as many as apply.)
   ( ) Didactic course(s) in counseling techniques
   ( ) Supervised clinical training
   Other (specify) _____
   ( ) None

6. If you have had training in counseling techniques, do you think that it has been useful to you?
   ( )$_1$Yes ( )$_2$No

7. If you have not had training in counseling techniques, do you think it would be useful to you?
   ( )$_1$Yes ( )$_2$No

FOLLOWING ARE A SERIES OF QUESTIONS ON YOUR PROFESSIONAL ACTIVITIES.

8. Are particular disorders usually referred to you? ( )$_1$Yes ( )$_2$No

9. IF YES, specify _____

10. How many hours a week do you usually devote to all your professional activities?_____ hours/wk.

11. About how much of your time is spent in genetic counseling-related activities, in this clinic and elsewhere? _____ hours/week

12. Concerning your counseling-related activities in this clinic, approximately how many hours per week do you spend on the following?

a. Seeing patients	_____ hours/week
b. Clerical and administrative work	_____ hours/week
c. Laboratory work	_____ hours/week
d. Other (specify) _____	_____ hours/week
	_____ TOTAL hours/week

13. Approximately how many patients do you counsel per week? _____

14. Concerning your professional time outside this clinic, approximately how many hours per week do you spend on the following?

a. Research	_____	hours/week
b. Practice	_____	hours/week
c. Teaching	_____	hours/week
d. Administration	_____	hours/week
e. Other (specify) _____	_____	hours/week
	_____	TOTAL hours/week

15. What functions do you routinely perform in this clinic? (Check as many as apply.)

Administrative
- ( ) Take initial phone calls
- ( ) Fill out intake forms
- ( ) Obtain medical records
- ( ) Schedule appointments
- ( ) Assign patients to counselors
- ( ) Coordinate clinic activities

Counseling
- ( ) Take patient/family social history
- ( ) Genetic counseling
- ( ) Amniocentesis counseling
- ( ) Counseling for family problems
- ( ) Counseling for personal emotional problems
- ( ) Suggest resources for financial problems
- ( ) Refer to specialists
- ( ) Follow-up to genetic counseling visit (by phone, mail, or in person)

Diagnostic
- ( ) Take pedigree
- ( ) Take medical history
- ( ) Physical examination
- ( ) Refer for diagnostic tests
- ( ) Laboratory work
- ( ) Make diagnosis
- ( ) Perform amniocentesis
- ( ) Participate in genetic screening program

Patient Management
- ( ) Provide medical treatment
- ( ) Physical examination
- ( ) Long-term management
- ( ) Provide information for management
- ( ) Refer to a parents' group
- ( ) Refer to social or educational agencies
- ( ) Refer to specialists
- ( ) Involved in a parents' group
- ( ) Follow-up

Other
- ( ) Attend preclinic conference
- ( ) Attend postclinic conference
- ( ) Participate in satellite or outreach program
- ( ) Provide education for professionals
- ( ) Provide lay education

16. Are there any functions you routinely perform in this clinic that you feel should not be part of your job?

( )$_1$Yes ( )$_2$No

17. **IF YES,** specify _____

_____

18. Are there any functions not now part of your job in this clinic that you feel should be?

( )$_1$Yes ( )$_2$No

19. **IF YES,** specify _____

_____

PROFESSIONALS IN GENETIC COUNSELING HAVE MANY OPINIONS ABOUT COUNSELING. BELOW ARE SOME QUESTIONS DEALING WITH ISSUES OF CURRENT INTEREST TO THOSE INVOLVED IN COUNSELING. WE WOULD LIKE TO HAVE YOUR PERSONAL OPINIONS ON THESE TOPICS.

20. Below are listed 7 tasks that can be involved in genetic counseling. Rate each task from a low of 1 (of little importance) to a high of 5 (very important) in terms of its importance to you as a professional obligation in counseling. Circle the 9 if you think a task is not a professional obligation.

Tasks	Of Little Importance				Very Important	Not an Obligation
a. To help the individual or the family to comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management	1	2	3	4	5	9
b. To help them appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives	1	2	3	4	5	9
c. To help them understand the options for dealing with the risk of recurrence	1	2	3	4	5	9
d. To help them choose the course of action which seems appropriate to them in view of their risk and their family goals	1	2	3	4	5	9
e. To help them act in accordance with their decision	1	2	3	4	5	9
f. To help them make the best possible adjustment to the disorder in an affected family member	1	2	3	4	5	9
g. To help them make the best possible adjustment to the risk of recurrence of that disorder	1	2	3	4	5	9

21. Professionals in genetic counseling may hold one or more goals for their counseling. In your opinion, how important is it that your counseling achieve each of the following: (Please check ☑.)

	Very Important$_1$	*Important*$_2$	*Somewhat Important*$_3$	*Not at all Important*$_4$
a. The prevention of disease or abnormality	☐	☐	☐	☐
b. The removal or lessening of patient guilt or anxiety	☐	☐	☐	☐
c. A reduction in the number of carriers of genetic disorders in the population	☐	☐	☐	☐
d. Improvement of the general health and vigor of the population	☐	☐	☐	☐

e. Helping individuals/couples adjust to and cope with their genetic problems	☐	☐	☐	☐
f. Helping individuals/couples achieve their parenting goals	☐	☐	☐	☐

22. In your opinion, how appropriate do you think it is for you as a professional in counseling to do each of the following:

	Always Appropriate$_1$	*Sometimes Appropriate*$_2$	*Rarely Appropriate*$_3$	*Never Appropriate*$_4$
a. Tell patients that decisions, especially reproductive ones, are theirs alone and refuse to make any for them	☐	☐	☐	☐
b. Suggest that while you will not make decisions for patients you will support any they make	☐	☐	☐	☐
c. Inform patients what most other people in their situation have done	☐	☐	☐	☐
d. Inform patients what you would do if you were in their situation	☐	☐	☐	☐
e. Advise patients what they ought to do	☐	☐	☐	☐

THANK YOU

COUNSELOR POST–COUNSELING QUESTIONNAIRE

Please complete this form immediately after the counseling session and return it to the study coordinator.

THANK YOU

The National Foundation – March of Dimes
Genetic Counseling Study
Department of Socio-Medical Sciences
and Community Medicine
Boston University School of Medicine
Boston, Massachusetts 02118

1. Today's date ____/____/____ (Month / Day / Year)

2. Your name _____ 3. Patient(s) name _____

4. Proband(s) name _____ (If different from patient.) 5. Proband's relationship to patient _____

6. Problem counseled for (be specific) _____

_____

7. For this counseling session, which of the following types of information did you have about or ascertain from the patient(s)? How useful was the information *for counseling this patient(s)*? If you did not have the information, how useful might it have been?

Information	Have info? Yes 1	No 2	How useful was it (or might it have been)? Very Useful 1	Useful 2	Somewhat Useful 3	Not Useful 4	Don't Know 8
Pedigree	☐	☐	☐	☐	☐	☐	☐
Diagnosis	☐	☐	☐	☐	☐	☐	☐
Test Results	☐	☐	☐	☐	☐	☐	☐
Other medical information	☐	☐	☐	☐	☐	☐	☐
Patient's knowledge about problem	☐	☐	☐	☐	☐	☐	☐
Patient/family psychosocial information	☐	☐	☐	☐	☐	☐	☐
Patient's expectations about counseling	☐	☐	☐	☐	☐	☐	☐

8. Were other professionals present during counseling? ( )$_1$Yes ( )$_2$No

9. **IF YES**, did any participate? ( )$_1$Yes ( )$_2$No

10. As part of clinic routine, did (or will) the patient(s) talk to any of the following professional people, *besides* yourself, at this clinic?

Social Worker ( )$_1$Yes ( )$_2$No
Nurse ( ) Yes ( ) No
Physician ( ) Yes ( ) No
Genetic Associate ( ) Yes ( ) No
Other (specify) _____

11. For each topic in the following list, please check ☒ whether it was *not discussed, just mentioned,* or *discussed in some depth* with this patient(s). *THEN* check the *1* item *you felt* it was most important to discuss with this patient and the *1* item you felt was of greatest interest to the patient(s).

If you counseled a couple and there were significant differences in interest, please indicate by using ♀ or ♂ instead of *1* check mark in the last column.

TOPIC	*Check if* Just mentioned 1	Discussed in some depth 2	Not discussed 3	Check *1* item *you* regarded as most important	Check *1* item of greatest interest *to the patient*
Patient's risk for having an affected child$_1$	☐	☐	☐	☐	☐
Whether or not the patient should have a child$_2$	☐	☐	☐	☐	☐
Amniocentesis$_3$	☐	☐	☐	☐	☐
Diagnosis of patient's child$_4$	☐	☐	☐	☐	☐
Diagnosis of another family member$_5$	☐	☐	☐	☐	☐

Diagnosis of patient$_6$	☐	☐	☐	☐	☐	
The etiology of the disorder$_7$	☐	☐	☐	☐	☐	
The prognosis of the disorder$_8$	☐	☐	☐	☐	☐	
Medical treatment for the disorder$_9$	☐	☐	☐	☐	☐	
How to care for an affected child$_{10}$	☐	☐	☐	☐	☐	
Affected child's current status, based on check-up$_{11}$	☐	☐	☐	☐	☐	
School programs or other special programs for an affected child$_{12}$	☐	☐	☐	☐	☐	
Financial costs of the disorder$_{13}$	☐	☐	☐	☐	☐	
Patient's feelings about his/her affected child$_{14}$	☐	☐	☐	☐	☐	
Relationship with spouse$_{15}$	☐	☐	☐	☐	☐	
Relationship with other children in family$_{16}$	☐	☐	☐	☐	☐	
Other (specify)	☐	☐	☐	☐	☐	
	☐	☐	☐	☐	☐	

PLEASE BE SURE TO CHECK THE *1* ITEM YOU MOST WANTED TO DISCUSS, AND THE *1* ITEM THE PATIENT(S) MOST WANTED TO DISCUSS.

12. How much difficulty do you think the patient(s) had understanding *what you told her/him* about the following? If you counseled a couple and there were significant differences in understanding, please indicate by using ♀ or ♂ in the appropriate boxes.

TOPIC	No Trouble 1	A Little Trouble 2	Moderate Trouble 3	A Lot of Trouble 4	Not Discussed 5
Diagnosis	☐	☐	☐	☐	☐
Etiology	☐	☐	☐	☐	☐
Prognosis	☐	☐	☐	☐	☐
Risk of occurrence/recurrence	☐	☐	☐	☐	☐
Burden of abnormal child to parents	☐	☐	☐	☐	☐

13. Did you give this patient (or couple) a recurrence/ occurrence risk?
( )$_1$Yes ( )$_2$No **(IF NO, SKIP TO #17)**

14. **IF YES**, the risk as stated to the patient was_____

15. *In general*, do you consider such a risk to be:
( )$_1$Very high ( )$_2$High ( )$_3$Moderate ( )$_4$Low ( )$_5$Very low

16. For *this patient(s)*, do you consider this risk to be:
( )$_1$Very high ( )$_2$High ( )$_3$Moderate ( )$_4$Low ( )$_5$Very low

17. Will you see this patient(s) again? ( )$_1$Yes ( )$_3$Maybe ( )$_2$No **(IF NO, SKIP TO #19)**

18. **IF YES OR MAYBE**, why will you see them again? (Check as many as apply.)
    ( ) More tests/medical information are needed to complete counseling
    ( ) For routine medical follow-up
    ( ) Patient(s) did not adequately understand information presented in this session
    ( ) Not enough time to deal with all the issues
    Other (specify)_____

19. As a result of this counseling session, have you (or will you) refer the patient(s) to any of the following?
    ( ) Social worker ( ) Parents' group
    ( ) Psychiatrist Other (specify)_____
    ( ) Social service agency ( ) No referrals

20. Will you send a follow-up letter to:
    Patient(s) ( )$_1$Yes ( )$_2$No
    Referring physician ( ) Yes ( ) No

21. In general, how satisfied are you with this counseling session?
    ( )$_1$Very satisfied ( )$_2$Satisfied ( )$_3$Dissatisfied ( )$_4$Very dissatisfied

22. Do you have any comments?_____

THANK YOU

APPENDIX 2
CLIENT QUESTIONNAIRES

FOR FEMALES

REGISTRATION QUESTIONNAIRE

Please complete this form BEFORE your counseling session. Then put it in the WHITE envelope, along with the signed Informed Consent Form.

Please return the envelope to the person at the clinic who gave it to you.

THANK YOU

The National Foundation – March of Dimes
Genetic Counseling Study
Department of Socio-Medical Sciences
and Community Medicine
Boston University School of Medicine
Boston, Massachusetts 02118

FIRST, WE WOULD LIKE SOME GENERAL INFORMATION.

1. Today's date ____ / ____ / ____
   Month Day Year

2. Your name _____

3. Telephone number ( ) _____
   Area Code

4. Address _____
   Street

   _____
   City State

5. Have you had genetic counseling before (either in person or over the phone)?
   ( )$_1$Yes ( )$_2$No ( )$_8$Unsure

(IF NO OR UNSURE, SKIP TO #8)

6. **IF YES**, did the counselor you talked to before work at *this* center?
   ( )$_1$Yes ( )$_2$No

7. Is it the same counselor you now have an appointment to see?
   ( )$_1$Yes ( )$_2$No ( )$_8$Unsure

8. NEXT, WE ARE INTERESTED IN THE REASONS **YOU** CAME TO TALK TO A COUNSELOR. (PLEASE CIRCLE **YES** OR **NO** FOR EACH OF THE FOLLOWING REASONS.)

YES NO To find out what chance I have of having a child with a birth defect or genetic disorder$_1$
IF YES, do you want to find out because (check as many as apply):
( ) I have a birth defect or genetic disorder$_2$
( ) My husband has a birth defect or genetic disorder$_3$
( ) I have had (or my husband has had) an affected child (a child with a birth defect or genetic disorder)$_4$
( ) There is a disorder in my family (or in my husband's family) which may be inherited$_5$
( ) I think I might carry a gene for a specific disorder$_6$
( ) My husband might carry a gene for a specific disorder$_7$
( ) I have had (or my husband has had) drug or x-ray exposure$_8$

YES NO To get information that will help me to decide if I should have a child$_9$

YES NO To find out about tests that can help to tell if an unborn baby will be normal$_{10}$

YES NO To find out if my child (living or not) has (or had) a specific birth defect or genetic disorder$_{11}$

YES NO To find out if a member of my family, other than my child, has a specific genetic disorder$_{12}$

YES NO To find out whether or not I have a specific genetic disorder$_{13}$

YES NO I want to know what a specific birth defect or genetic disorder is like as it develops after birth$_{14}$

YES NO I want to find out why a specific birth defect or genetic disorder happens$_{15}$

YES NO I want to get more facts about the treatment for a specific birth defect or genetic disorder$_{16}$

YES NO I want some information concerning my affected child$_{17}$
IF YES, would you like (check as many as apply):
( ) To get advice or information that will help me care for my child$_{18}$
( ) To get a check-up for my child and find out how she/he is doing$_{19}$
( ) To find out about school programs or other special programs for my child$_{20}$

YES NO I have some personal concerns about having an affected child$_{21}$
IF YES, would you like (check as many as apply):
( ) To find out about the financial costs of a specific birth defect or genetic disorder$_{22}$
( ) To discuss my feelings about my affected child$_{23}$
( ) To discuss my relationship with my husband$_{24}$
( ) To discuss my relationship with my other children$_{25}$

Please go on to the next page.

YES NO I am here because a doctor told me to talk to a genetic counselor$_{26}$

YES NO I do not want to talk to the counselor about anything in particular, but my husband wanted me to come$_{27}$

Other reasons (specify)_____

_____

YES NO I am not exactly sure why I am seeing a counselor.$_{98}$

NOW THAT YOU HAVE GONE THROUGH THIS LIST PLEASE GO BACK AND CIRCLE THE **ONE** MAJOR REASON WHY **YOU** WANT TO SEE A COUNSELOR.

NEXT, WE WOULD LIKE TO ASK YOU ABOUT THE MEDICAL PROBLEM OR DISORDER THAT BROUGHT YOU TO THE GENETIC COUNSELING CLINIC.

9. Please name the medical problem or disorder that brought you to this clinic.

I know it is$_1$ _____

I think it is$_2$ _____

( )$_8$ I cannot name the problem or disorder

10. Please describe the medical problem or disorder that brought you to this clinic. ____ _____

_____

( )$_{98}$ I cannot describe the problem or disorder

11. Are you concerned about having a child with a particular birth defect or genetic disorder?

( )$_1$ Yes ( )$_2$ No

(IF NO, SKIP TO #16)

12. Is the birth defect or disorder about which you are concerned inherited?

( )$_1$ Yes, it is inherited
( )$_2$ No, it is not inherited
( )$_3$ It is sometimes inherited
( )$_8$ I am not sure

13. Do you know what the chances are of your having a child with this birth defect or disorder? (Give numbers or a percent.)

I know the chances are$_1$ _____

I think the chances are$_2$ _____

( )$_3$ The chances are not known to medical science

( )$_8$ I don't know

14. Even though you may not know what the specific chances are, what kind of chance do you think there is of your having a child with this birth defect or disorder? (Please circle.)

Very high$_1$ High$_2$ Moderate$_3$ Low$_4$ Very low$_5$

15. Suppose you were to have a child. Do you think your child would (check one):

( )$_1$ Definitely be normal
( )$_2$ Probably be normal
( )$_3$ Probably have the birth defect or disorder
( )$_4$ Definitely have the birth defect or disorder
( )$_8$ Not sure
( )$_9$ Does not apply

(IF YOU HAVE ANSWERED #15, SKIP TO #19)

16. Even though you may not be concerned about a particular birth defect or disorder, what do you think the chances are of your having a child with a birth defect or genetic disorder? (Give numbers or a percent.)

I know the chances are$_1$ _____

I think the chances are$_2$ _____

( )$_3$ The chances are not known

( )$_8$ I don't know

Please go on to the next page.

17. Even though you may not know what the specific chances are, what kind of chance do you think there is of your having a child with a birth defect or genetic disorder? (Please circle.)

Very high$_1$ High$_2$ Moderate$_3$ Low$_4$ Very low$_5$

18. Suppose you were to have a child. Do you think your child would:

( )$_1$ Definitely be normal
( )$_2$ Probably be normal
( )$_3$ Probably have a birth defect or genetic disorder
( )$_4$ Definitely have a birth defect or genetic disorder
( )$_8$ Not sure
( )$_9$ Does not apply

THE NEXT SECTION ASKS ABOUT YOUR PLANS FOR HAVING A FAMILY.

19. Are you now pregnant?

( )$_1$ Yes ( )$_2$ No ( )$_8$ Unsure

20. Do you intend to have a child (or another child if now pregnant) within the next two years?

( )$_1$ Yes ( )$_2$ No ( )$_8$ Unsure

21. Are you thinking about having children anytime after the next two years?

( )$_1$ Yes ( )$_2$ No ( )$_8$ Unsure

22. Given the circumstances of your life, how many children would you like to have altogether?

_____ Number ( )$_8$ Unsure

23. IF YOU ARE MARRIED, how many children do you think your husband would like to have?

_____ Number ( )$_8$ Unsure

24. As you think about whether or not to have children (or additional children), how important are the following factors to you in your planning? (Please check ☐ one answer for each factor.)

Possible Factors	How Important Is It?					
	Very Important$_1$	*Somewhat Important*$_2$	*Of Little Importance*$_3$	*Of no Importance*$_4$	*Not Applicable*$_9$	*Don't Know*$_8$
My desire to have a certain number of boys and girls	☐	☐	☐	☐	☐	☐
Fulfillment as a parent	☐	☐	☐	☐	☐	☐
Completing my marriage	☐	☐	☐	☐	☐	☐
Carrying on my family line	☐	☐	☐	☐	☐	☐
My view of the ideal family size	☐	☐	☐	☐	☐	☐
The wishes of my husband	☐	☐	☐	☐	☐	☐
Taking care of my other children	☐	☐	☐	☐	☐	☐
Effects on my social life	☐	☐	☐	☐	☐	☐

Please go on to the next page.

Finances	☐	☐	☐	☐	☐	☐
My career goals	☐	☐	☐	☐	☐	☐
Husband's career goals	☐	☐	☐	☐	☐	☐
Influence from relatives	☐	☐	☐	☐	☐	☐
Doctor's advice	☐	☐	☐	☐	☐	☐
Religious or ethical beliefs	☐	☐	☐	☐	☐	☐
Other (specify)	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐

25. Parents who have a child with a birth defect or genetic disorder may experience a number of problems, to varying degrees. We would like to know how you feel about some of these problems, whether or not you have had an affected child.

If you *already have an affected child*, please answer *both Sections A and B*. Section A asks about problems you may be having now. Section B asks whether you think these same problems will be problems to you in the future.

If you *DO NOT* have an affected child, please answer *only Section B*. That is, if you were to have a child with the birth defect or genetic disorder you are concerned about, what types of problems do you think you *might* have, and how serious *might* they be for you?

	SECTION A How serious is this problem now? (Circle a number.)				SECTION B How serious might it be in the future? (Circle a number.)				
Type of Problem:	Very	Moderately	Slightly	No Problem	Very	Moderately	Slightly	No Problem	Don't know
Financial	1	2	3	4	1	2	3	4	8
My own feelings about such a child	1	2	3	4	1	2	3	4	8
Telling my family and friends about having had an affected child	1	2	3	4	1	2	3	4	8
Taking care of my other children	1	2	3	4	1	2	3	4	8
My relationship with my husband	1	2	3	4	1	2	3	4	8
Effects on my social life	1	2	3	4	1	2	3	4	8
Getting medical treatment for my affected child	1	2	3	4	1	2	3	4	8
Taking care of my child at home	1	2	3	4	1	2	3	4	8
Educating my child	1	2	3	4	1	2	3	4	8

Please go on to the next page.

My own emotional problems	1	2	3	4		1	2	3	4	8
Providing care for my child when he/she is an adult	1	2	3	4		1	2	3	4	8

26. Do you have any comments? _____

_____

_____

THANK YOU

FOR FEMALES

POST–COUNSELING QUESTIONNAIRE

Please do not break the seal or look at the form until *after* your appointment.

This questionnaire should be returned in the TAN post-paid envelope within *one week* of your appointment.

THANK YOU

The National Foundation – March of Dimes
Genetic Counseling Study
Department of Socio-Medical Sciences
and Community Medicine
Boston University School of Medicine
Boston, Massachusetts 02118

FIRST, WE WOULD LIKE SOME GENERAL INFORMATION.

1. Today's date____/____/____ (Month / Day / Year) 2. Date you saw counselor____/____/____ (Month / Day / Year)

3. Your name_____ 4. Age_____

5. What is your marital status? ( )$_1$Single ( )$_2$Married ( )$_3$Widowed ( )$_4$Divorced ( )$_5$Separated

6. What is your religion? ( )$_1$Protestant ( )$_2$Jewish ( )$_3$Catholic ( )$_4$None
   Other (specify)_____

7. Please circle the last grade you completed in school:
   Elementary/Secondary$_1$: 1 2 3 4 5 6 7 8 9 10 11 12
   College$_2$: 1 2 3 4 Postgraduate$_3$: 1 2 3 4 more than 4
   Other (specify)_____

8. What is your occupation? (Be as specific as possible.)_____

IF YOU ARE NOT MARRIED, SKIP TO #11.

9. Please circle the last grade your *HUSBAND* completed in school.
   Elementary/Secondary$_1$: 1 2 3 4 5 6 7 8 9 10 11 12
   College$_2$: 1 2 3 4 Postgraduate$_3$: 1 2 3 4 more than 4
   Other (specify)_____

10. What is your *HUSBAND'S* occupation? (Be as specific as possible.)_____

11. Please circle your approximate *TOTAL HOUSEHOLD* income, before taxes.
    0 - \$5,000$_1$ \$5,001 - \$10,000$_2$ \$10,001 - \$15,000$_3$ \$15,001 - \$20,000$_4$
    \$20,001 - \$25,000$_5$ \$25,001 - \$30,000$_6$ above \$30,000$_7$

NEXT, WE WOULD LIKE TO ASK SOME QUESTIONS ABOUT YOUR COUNSELING SESSION.

12. Did any family member or friend sit in on the counseling session with you? ( )$_1$Yes ( )$_2$No

13. **IF YES**, what is their relation to you?
    _____
    _____
    _____

14. *Besides* the genetic counselor, did you talk to any of the following professional people at the clinic about the problem for which you came?

Social worker	( )$_1$Yes ( )$_2$No	Genetic associate	( ) Yes ( ) No
Nurse	( ) Yes ( ) No	Not sure what their	
Doctor	( ) Yes ( ) No	position was	( ) Yes ( ) No

15. About how much time did you spend with the counselor? (Please circle.)
    Under 20 min.$_1$ 20 - 39 min.$_2$ 40 - 59 min.$_3$ An hour or more$_4$

16. What is the *name* of the medical problem or disorder you talked to the counselor about?
    I *know* it was$_1$_____
    I *think* it was$_2$_____
    ( )$_8$ I cannot name the problem or disorder

17. Please *describe* the medical problem or disorder you talked to the counselor about.$_1$
    _____
    _____
    ( )$_{98}$I cannot describe the problem or disorder

Please go on to the next page.

18. Next, we'd like to find out what you and the counselor talked about. Please check ☒ whether each item was *not discussed, just mentioned,* or *discussed in some depth. AFTER* you have done that, please go back and check the *1* item *YOU* most wanted to discuss, and the *1* item that the *counselor* seemed to want to discuss the most.

TOPIC	Check if: Not discussed 1	Just mentioned 2	Discussed in some depth 3	Check *1* item *you* wanted to discuss most	Check *1* item *the counselor* wanted to discuss most
What chance I have of having a child with a birth defect or genetic disorder$_{1}$	☐	☐	☐	☐	☐
Whether or not I should have a child$_{2}$	☐	☐	☐	☐	☐
Tests that can help to tell if an unborn baby will be normal$_{3}$	☐	☐	☐	☐	☐
Whether or not my child (living or not) has (or had) a specific birth defect or genetic disorder$_{4}$	☐	☐	☐	☐	☐
Whether or not a member of my family, other than my child, has a specific genetic disorder$_{5}$	☐	☐	☐	☐	☐
Whether or not I have a specific genetic disorder$_{6}$	☐	☐	☐	☐	☐
Why a specific birth defect or genetic disorder happens$_{7}$	☐	☐	☐	☐	☐
What a specific birth defect or genetic disorder is like as it develops after birth$_{8}$	☐	☐	☐	☐	☐
Medical treatment for a specific birth defect or genetic disorder$_{9}$	☐	☐	☐	☐	☐
How I should care for my affected child$_{10}$	☐	☐	☐	☐	☐
My child had a check-up, and we discussed how he/she is doing$_{11}$	☐	☐	☐	☐	☐
School programs or other special programs for my child$_{12}$	☐	☐	☐	☐	☐
Financial costs of a specific birth defect or genetic disorder$_{13}$	☐	☐	☐	☐	☐
My feelings about my affected child$_{14}$	☐	☐	☐	☐	☐
My relationship with my husband$_{15}$	☐	☐	☐	☐	☐
My relationship with my other children$_{16}$	☐	☐	☐	☐	☐
Other (specify)	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐

PLEASE BE SURE TO CHECK THE *1* ITEM YOU MOST WANTED TO DISCUSS, AND THE *1* ITEM THE COUNSELOR MOST WANTED TO DISCUSS.

NEXT, WE WOULD LIKE TO ASK YOU ABOUT THE MEDICAL PROBLEM OR DISORDER THAT BROUGHT YOU TO THE GENETIC COUNSELING CLINIC.

19. Is the birth defect or disorder you discussed with the counselor inherited?

( )$_{1}$ Yes, it is inherited
( )$_{2}$ No, it is not inherited
( )$_{3}$ It is sometimes inherited
( )$_{8}$ I am not sure

Please go on to the next page.

20. Did the counselor tell you what *your* chances are of having a child with the birth defect or disorder you talked about?

( )$_1$Yes ( )$_2$No **(IF NO, SKIP TO #22)**

21. **IF YES**, what are your chances? (Give numbers or a percent.)

I *know* the chances are$_1$ _____

I *think* the chances are$_2$ _____

( )$_8$ The counselor told me, but I don't remember what the chances are

22. Even though you may *not* know what the specific chances are, what kind of chance do you *think* there is of *your* having a child with this birth defect or disorder? (Please circle.)

Very high$_1$ High$_2$ Moderate$_3$ Low$_4$ Very low$_5$

23. *Suppose* you were to have a child. Do you think your child would (check one):

( )$_1$Definitely be normal
( )$_2$Probably be normal
( )$_3$Probably have the birth defect or disorder
( )$_4$Definitely have the birth defect or disorder
( )$_8$Not sure
( )$_9$Does not apply

24. Parents who have a child with a birth defect or genetic disorder may experience a number of problems, to varying degrees. We would like to know how you feel about some of these problems, whether or not you have had an affected child.

If you *already have an affected child*, please answer *both Sections A and B*. Section A asks about problems you may be having now. Section B asks whether you think these same problems will be problems to you in the future.

If you *DO NOT* have an affected child, please answer *only Section B*. That is, if you were to have a child with the birth defect or genetic disorder you are concerned about, what types of problems do you think you *might* have, and how serious *might* they be for you?

	SECTION A — How serious is this problem now? (Circle a number.)				SECTION B — How serious might it be in the future? (Circle a number.)				
Type of Problem:	Very	Moderately	Slightly	No Problem	Very	Moderately	Slightly	No Problem	Don't know
Financial	1	2	3	4	1	2	3	4	8
My own feelings about such a child	1	2	3	4	1	2	3	4	8
Telling my family and friends about having had an affected child	1	2	3	4	1	2	3	4	8
Taking care of my other children	1	2	3	4	1	2	3	4	8
My relationship with my husband	1	2	3	4	1	2	3	4	8
Effects on my social life	1	2	3	4	1	2	3	4	8
Getting medical treatment for my affected child	1	2	3	4	1	2	3	4	8
Taking care of my child at home	1	2	3	4	1	2	3	4	8
Educating my child	1	2	3	4	1	2	3	4	8

Please go on to the next page.

My own emotional problems	1	2	3	4		1	2	3	4	8
Providing care for my child when he/she is an adult	1	2	3	4		1	2	3	4	8

THE FOLLOWING SECTION ASKS ABOUT **ALL** OF YOUR PAST PREGNANCIES. IF YOU HAVE NEVER BEEN PREGNANT OR ARE PREGNANT FOR THE **FIRST** TIME, **SKIP TO #28.**

25. For each of your past pregnancies, please indicate by a check if the pregnancy resulted in a live birth or not.

Pregnancy number	Live birth$_1$	Stillbirth$_2$	Miscarriage$_3$	Therapeutic abortion$_4$
1	___	___	___	___
2	___	___	___	___
3	___	___	___	___
4	___	___	___	___
5	___	___	___	___
6	___	___	___	___
7	___	___	___	___
8	___	___	___	___
9	___	___	___	___

26. For all live-born children with a birth defect or genetic disorder (or who may have a birth defect or genetic disorder), please name (or describe) the defect or disorder and provide the information requested.

Pregnancy number	Name (or describe) the defect or disorder	Is the child living? Yes$_1$	No$_2$	If living, age	Living with you? Yes$_1$	No$_2$
___	_____	___	___	___	___	___
___	_____	___	___	___	___	___
___	_____	___	___	___	___	___
___	_____	___	___	___	___	___

27. For any pregnancies *not* resulting in a *live* birth, if the baby had a birth defect or genetic disorder, please indicate below.

Pregnancy number	Name (or describe) the birth defect or genetic disorder
___	_____
___	_____
___	_____
___	_____

THE NEXT SECTION ASKS YOU ABOUT YOUR PLANS FOR HAVING A FAMILY.

28. Do you intend to have a child (or another child if now pregnant) within the next two years?
( )$_1$Yes ( )$_2$No ( )$_8$Unsure

29. Has this intention been changed by your counseling session?
( )$_1$Yes ( )$_2$No

Please go on to the next page.

30. Are you thinking about having children anytime after the next two years?
( )$_1$Yes ( )$_2$No ( )$_8$Unsure

31. Have these thoughts about future children been changed by your counseling session?
( )$_1$Yes ( )$_2$No

32. Given the circumstances of your life, how many children would you like to have altogether?
____ ( )$_8$Unsure
Number

33. Has this preferred number been influenced by your counseling session?
( )$_1$Yes ( )$_2$No

34. *IF YOU ARE MARRIED*, how many children do you think your husband would like to have?
____ ( )$_8$Unsure
Number

35. Are you currently using a contraceptive method? ( )$_1$Yes ( )$_2$No

36. **IF YES**, which method?_____

37. **IF NO**, is it because:
( )$_1$ I am pregnant
( )$_2$ I am trying to become pregnant
( )$_3$ I recently had a baby
( )$_4$ I (or my husband) do not believe in using contraceptives
( )$_5$ I (or my husband) have been sterilized
Other (specify)_____

FINALLY, WE WOULD LIKE TO ASK YOU A FEW MORE QUESTIONS ABOUT YOUR COUNSELING SESSION.

38. How clearly did the counselor explain the following topics? (Please check ☒.)

TOPIC	Not clearly 1	Somewhat clearly 2	Moderately clearly 3	Very clearly 4	Not discussed 5
What the birth defect or genetic disorder is	☐	☐	☐	☐	☐
Why a specific birth defect or genetic disorder happens	☐	☐	☐	☐	☐
Information about how a specific birth defect or genetic disorder develops after birth	☐	☐	☐	☐	☐
The chance of having a child with a specific birth defect or genetic disorder	☐	☐	☐	☐	☐
What it is like to raise a child with a specific birth defect or genetic disorder	☐	☐	☐	☐	☐

39. Did the counseling session give you the medical–genetic facts you wanted?
It gave me: ( )$_1$All ( )$_2$Most ( )$_3$Some ( )$_4$None

40. If you did not get **ALL** the facts you wanted, was it because (check as many as apply):
( ) Some of the facts are not known by medical science
( ) The counselor has to gather more facts
( ) The counselor did not want to discuss some facts I asked about
( ) I did not ask about certain facts that I wanted to know

41. Did the counseling session help you with the types of personal concerns that you had?
It helped with: ( )$_1$All ( )$_2$Most ( )$_3$Some ( )$_4$None

Please go on to the next page.

42. If counseling did not help you with **ALL** the personal concerns you had, was it because (check as many as apply):
    ( ) Some of the things I am concerned about can't be dealt with medically
    ( ) The counselor has to get more information for me
    ( ) The counselor did not want to discuss some of the concerns I have
    ( ) I did not ask about some of the concerns I have

43. Did the counseling session raise any new concerns for you? ( )$_1$Yes ( )$_2$No

44. **IF YES**, what are they?_____

45. Would you like to (check one):
    ( )$_1$Talk to this counselor again
    ( )$_2$Talk to some other counselor
    ( )$_8$Not sure if I want to talk to a counselor again
    ( )$_3$Don't want to talk to a counselor again

46. Are you going to see a counselor again? ( )$_1$Yes ( )$_2$No

47. Do you have any comments about your counseling session?_____

THANK YOU

FOR FEMALES

6–MONTH FOLLOW–UP QUESTIONNAIRE

The National Foundation – March of Dimes
Genetic Counseling Study
Department of Socio-Medical Sciences
and Community Medicine
Boston University School of Medicine
Boston, Massachusetts 02118

FIRST, WE WOULD LIKE SOME GENERAL INFORMATION ABOUT YOU AND THE MEDICAL PROBLEM OR DISORDER THAT BROUGHT YOU TO COUNSELING.

1. Your name _____ 2. Today's date ___/___/___ (Month / Day / Year)

3. What is your marital status?
   ( )1 Single ( )2 Married ( )3 Widowed ( )4 Divorced ( )5 Separated

4. Date you last saw a genetic counselor ___/___/___ (Month / Day / Year)

5. What is the *name* of the medical problem or disorder you talked to the counselor about?
   I *know* it was 1 _____
   I *think* it was 2 _____
   ( )8 I cannot name the problem or disorder

6. Please *describe* the medical problem or disorder 1 _____
   _____
   ( )8 I cannot describe the problem or disorder

7. Is the birth defect or disorder you discussed with the counselor inherited?
   ( )1 Yes, it is inherited ( )3 It is sometimes inherited
   ( )2 No, it is not inherited ( )8 I am not sure

8. Did the counselor tell you what *your* chances are of having a child with the birth defect or disorder you talked about?
   ( )1 Yes ( )2 No **(IF NO, SKIP TO #10)**

   9. **IF YES**, what are your chances? (Give numbers or a percent.)
      I *know* the chances are 1 _____
      I *think* the chances are 2 _____
      ( )8 The counselor told me, but I don't remember what the chances are

10. Even though you may *not* know what the specific chances are, what kind of chance do you *think* there is of *your* having a child with this birth defect or disorder? (Please circle.)
    Very high 1 High 2 Moderate 3 Low 4 Very low 5

11. Suppose you were to have a child. Do you think your child would (check one):
    ( )1 Definitely be normal
    ( )2 Probably be normal
    ( )3 Probably have the birth defect or disorder
    ( )4 Definitely have the birth defect or disorder
    ( )8 Not sure
    ( )9 Does not apply

12. Parents who have a child with a birth defect or genetic disorder may experience a number of problems, to varying degrees. We would like to know how you feel about some of these problems, whether or not you have had an affected child.

    If you *already have an affected child*, please answer *both Sections A and B*. Section A asks about problems you may be having now. Section B asks whether you think these same problems will be problems to you in the future.

    If you *DO NOT* have an affected child, please answer *only Section B*. That is, if you were to have a child with the birth defect or genetic disorder you are concerned about, what types of problems do you think you *might* have, and how serious *might* they be for you?

Please go on to the next page.

Type of Problem:	SECTION A: How serious is this problem now? (Circle a number.) Very	Moderately	Slightly	No Problem	SECTION B: How serious might it be in the future? (Circle a number.) Very	Moderately	Slightly	No Problem	Don't know
Financial	1	2	3	4	1	2	3	4	8
My own feelings about such a child	1	2	3	4	1	2	3	4	8
Telling my family and friends about having had an affected child	1	2	3	4	1	2	3	4	8
Taking care of my other children	1	2	3	4	1	2	3	4	8
My relationship with my husband	1	2	3	4	1	2	3	4	8
Effects on my social life	1	2	3	4	1	2	3	4	8
Getting medical treatment for my affected child	1	2	3	4	1	2	3	4	8
Taking care of my child at home	1	2	3	4	1	2	3	4	8
Educating my child	1	2	3	4	1	2	3	4	8
My own emotional problems	1	2	3	4	1	2	3	4	8
Providing care for my child when he/she is an adult	1	2	3	4	1	2	3	4	8

THE FOLLOWING SECTION ASKS YOU ABOUT YOUR PLANS FOR HAVING A FAMILY.

13. Have you had a child in the last six months? (1 Yes (2 No

(IF NO, SKIP TO #16)

14. **IF YES**, is the child living? (1 Yes (2 No

15. Does (did) the child have a birth defect or genetic disorder?
(1 Yes (specify) _____
(2 No (8 Unsure

16. Are you now pregnant? (1 Yes (2 No (8 Unsure

(IF NO, SKIP TO # 18)

17. During this pregnancy, will you have (or have you had) amniocentesis? (Amniocentesis is a test which can help to tell if an unborn baby is normal.)
(1 Yes (2 No (8 Unsure

Please go on to the next page.

18. Do you intend to have a child (or another child if now pregnant) within the next two years?
( )$_1$Yes ( )$_2$No ( )$_8$Unsure **(IF NO, SKIP TO #20)**

19. **IF YES**, will you have amniocentesis? ( )$_1$Yes ( )$_2$No ( )$_8$Unsure

20. Has your decision about having a child within the next two years been influenced by your counseling session? ( )$_1$Yes ( )$_2$No

21. Are you thinking about having children anytime after the next two years?
( )$_1$Yes ( )$_2$No ( )$_8$Unsure

22. Have your thoughts about future children been changed by your counseling session?
( )$_1$Yes ( )$_2$No

23. Given the circumstances of your life, how many children would you like to have altogether?
_____ Number ( )$_8$Unsure

24. Has this preferred number been influenced by your counseling session?
( )$_1$Yes ( )$_2$No

25. **IF YOU ARE MARRIED**, how many children do you think your husband would like to have?
_____ Number ( )$_8$Unsure

26. Are you currently using a contraceptive method? ( )$_1$Yes ( )$_2$No
IF YES, which method?_____
IF NO, is it because:
( )$_1$I am pregnant
( )$_2$I am trying to become pregnant
( )$_3$I recently had a baby
( )$_4$I (or my husband) do not believe in using contraceptives
( )$_5$I (or my husband) have been sterilized
Other (specify)_____

27. As you think about whether or not to have children (or additional children), how important are the following factors to *you* in your planning? (Please check ☒ one answer for each factor.)

Possible Factors	How important is it?					
	Very Important$_1$	Somewhat Important$_2$	Of little Importance$_3$	Of no Importance$_4$	Not Applicable$_9$	Don't Know$_8$
My desire to have a certain number of boys and girls	☐	☐	☐	☐	☐	☐
Fulfillment as a parent	☐	☐	☐	☐	☐	☐
Completing my marriage	☐	☐	☐	☐	☐	☐
Carrying on my family line	☐	☐	☐	☐	☐	☐
My view of the ideal family size	☐	☐	☐	☐	☐	☐
The wishes of my husband	☐	☐	☐	☐	☐	☐
Taking care of my other children	☐	☐	☐	☐	☐	☐
Effects on my social life	☐	☐	☐	☐	☐	☐
Finances	☐	☐	☐	☐	☐	☐
My career goals	☐	☐	☐	☐	☐	☐

Please go on to the next page.

Husband's career goals	☐	☐	☐	☐	☐	☐
Influence from relatives	☐	☐	☐	☐	☐	☐
Doctor's advice	☐	☐	☐	☐	☐	☐
Religious or ethical beliefs	☐	☐	☐	☐	☐	☐
Other (specify)	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐

28. NEXT WE ARE INTERESTED IN WHETHER OR NOT YOU HAVE TRIED TO GET INFORMATION OR ADVICE ABOUT SOME OF THE THINGS YOU MAY HAVE DISCUSSED WITH THE GENETIC COUNSELOR.

For *each* of the topics listed below, please indicate whether or not you have sought information or advice, *outside the genetic counseling clinic.* (From your family doctor, friends, relatives, or books, for example.)

TOPICS	Have you sought information or advice *outside* the clinic? (Check ☒)	
	Yes$_1$	No$_2$
What chance I have of having a child with a specific birth defect or genetic disorder	☐	☐
The problems I would have if I had a child with a specific birth defect or genetic disorder	☐	☐
Whether or not I should have a child	☐	☐
Tests that can help to tell if an unborn baby will be normal	☐	☐
Finding out what the birth defect or genetic disorder is	☐	☐
Why a specific birth defect or genetic disorder happens	☐	☐
What a specific birth defect or genetic disorder is like as it develops after birth	☐	☐
Medical treatment for a specific birth defect or genetic disorder	☐	☐
How I should care for my affected child	☐	☐
Other (specify)	☐	☐

Have you asked for *any* advice or information from parents of children with birth defects or genetic disorders? ( )$_1$ Yes ( )$_2$ No

Please go on to the next page.

Are you *now* a member of any groups or organizations - large or small - that help parents who have children with birth defects or genetic disorders?

( )$_1$ No

IF NO, *why* don't you belong? _____

_____

_____

( )$_2$ Yes. Specify name of group or organization. _____

IF YES, how long have you been a member? _____

About how many times a year does the group or organization hold meetings? _____

About how many meetings a year do you attend? _____

Have you ever been an officer or leader of the group? ( )$_1$ Yes ( )$_2$ No

What have you gained from being in this group? _____

_____

_____

_____

29. Have you discussed with your family physician the genetic counseling you received?
( )$_1$Yes ( )$_2$No **(IF NO, SKIP TO # 31)**

30. **IF YES**, what type of contact was it? (Check as many as apply.)
( ) Letter
( ) Phone call
( ) Office visit

31. Have you been contacted by the genetic counseling clinic since your visit?
( )$_1$Yes ( )$_2$No **(IF NO, SKIP TO # 34)**

32. **IF YES**, what type of contact was it? (Check as many as apply.)
( ) Letter
( ) Phone call (specify from whom) _____
( ) Visit at home (specify by whom) _____

33. What were you contacted about? (Specify.) _____

34. Have *you* contacted the genetic counseling clinic since your visit?
( )$_1$Yes ( )$_2$No **(IF NO, SKIP TO # 36)**

35. **IF YES**, for what reason? _____

_____

_____

FINALLY, WE WOULD LIKE TO ASK YOU A FEW MORE QUESTIONS ABOUT YOUR COUNSELING SESSION.

36. Would you like to receive more genetic counseling? ( )$_1$Yes ( )$_2$No ( )$_8$Unsure

(IF NO OR UNSURE, SKIP TO # 38)

37. **IF YES**, for what reasons? (Check as many as apply.)
( ) I have forgotten some of the information I received during counseling
( ) I did not understand some of the information I received during counseling
( ) The counselor did not answer all of my questions before
( ) I did not ask all of the questions I had before
( ) I have had a child with a birth defect or genetic disorder since the counseling session
( ) I am pregnant or trying to become pregnant

New questions or concerns have come up (specify)_____

_____

Other (specify)_____ _____

38. In general, how satisfied are you with the genetic counseling you received several months ago?
( )$_1$Very satisfied ( )$_2$Satisfied ( )$_3$Somewhat satisfied ( )$_4$Dissatisfied ( )$_5$Very dissatisfied

39. Do you have any comments?_____

_____

_____

_____

THANK YOU

APPENDIX 3
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