The early diagnosis of congenital abnormalities / R.W. Smithells.

THE EARLY DIAGNOSIS OF CONGENITAL ABNORMALITIES

The Early Diagnosis of Congenital Abnormalities

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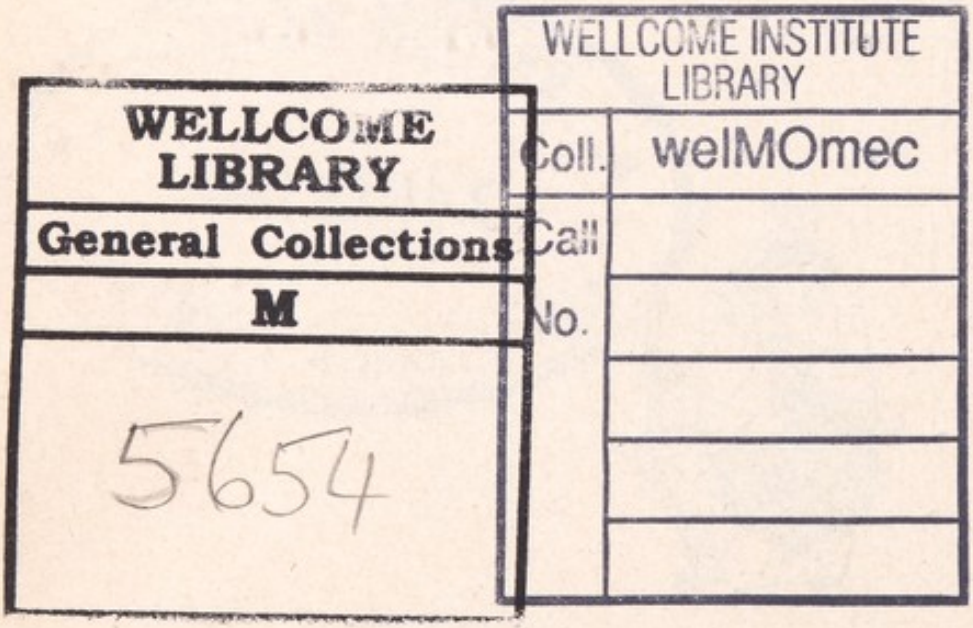

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Foreword

THE fifty per cent. increase, between 1951 and 1960, in the proportion of infants' deaths attributed to congenital malformations is a major challenge. To meet it, general practitioners, local authority doctors, obstetricians and paediatricians have increasingly felt the need of a book from which they can get useful knowledge on the subject.

Dr. Smithells' book is good for reading and for reference; it is small but up to date, comprehensible and comprehensive. A mine of information in which to work, it is enjoyable as well as productive.

The author starts with chapters on two unusual but valuable topics: *Prenatal Diagnosis* and *Diagnosis During Labour*. The obstetrician who reads these will be forewarned and will have neat opportunities to forewarn his paediatric colleague. Chapter 3, on *Examination of the Symptomless Infant*, will reward reading and re-reading. Chapter 4, *Early Symptoms of Congenital Abnormalities*, approaches the problem of the newborn infant who has a symptom, in a way that is rare but right. A clinician writing for doctors at the cotside or clinic, Dr. Smithells considers in turn the baby who is floppy, the baby who is jaundiced, and so on. After all, the baby does not arrive labelled as 'birth injury' or 'Möbius syndrome', or as having a disorder of the nervous system; he does not arrive as having hepatitis or hypothyroidism; he arrives with a symptom, sometimes one which the doctor will have learnt from Chapter 3 to recognize. The doctor has to start from the symptom and, if the best management is to be planned, he needs to learn how to arrive at the cause.

But even the doctor who knows that bubbly breathing or

cyanosis with dextrocardia on the first day of life implies a probable urgent need for surgery, who knows that persisting rapid breathing in the neonate may be due to heart failure, who knows how to recognize Ehlers-Danlos syndrome may still be neglecting one of the most important opportunities we have of preventing life-long emotional disturbance in parents and children. This major clinical responsibility is discussed in Chapter 5 on *Interviewing Parents of Malformed Babies*; it is an opportunity for initiating preventive mental hygiene which, surprisingly, is omitted or dealt with very inadequately in most textbooks.

This is a book to have in the prenatal clinic, in the delivery room and in the lying-in ward; a copy should be kept in the surgery and in the clinic. It will answer emergency problems as well as less urgent problems of diagnosis and management. It is an excellent present to give to doctors; the recipients will find that it is readable and answers their questions, and the donors will like the low price. Dr. Smithells is known to many as a gifted teacher, clinician and research worker. This book makes his talents available to all.

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TO MY WIFE

Preface

THE infant mortality rate in Great Britain is one of the lowest in the world. Social and economic factors are largely responsible, but the medical services have played a part. During the decade 1951 to 1960, the infant mortality rate fell from 30 to 22 per thousand live births. Having regard to the advances in paediatric surgery which, during this time, saved the lives of many malformed babies, a fall in the number of infant deaths attributed to congenital malformations might have been expected. Far from this being the case, there has been a substantial increase in both absolute and relative numbers, as the following table shows:

	1951	*1960*	*Change 1951–1960*
Live births . . .	677,529	785,005	14% increase
Infant deaths . .	20,223	17,118	15% decrease
Infant deaths attributed to malformations .	2,864	3,549	24% increase
Proportion of infant deaths attributed to malformations . .	14%	21%	50% increase

The *deaths* attributed to malformations are only part of the problem. There are also *stillbirths*, 20 to 25 per cent. of which are associated with malformations. More important, the figures of deaths take no account of the *life-long handicaps* which non-lethal abnormalities may bestow

upon the newborn child—and on his family. In some ways the biggest problems of all are presented by these children —children with paralysed legs and sphincters resulting from myelomeningocele, children with fibrocystic disease of the pancreas, mongol children.

Prevention of these anomalies must clearly be our ultimate aim. At present this is only possible in a negative way. We can attempt to protect pregnant women from viruses, from ionizing radiations, and from potentially teratogenic drugs. We can advise parents of the recurrence risks of genetically determined disorders. But even if we do this conscientiously and efficiently, 2 per cent. of all babies will still be born with serious malformations and a further 3 per cent. with lesser abnormalities. Until by epidemiological and experimental research we have learnt a great deal more about causes, we can do very little to promote normality.

Meanwhile, wherever we can, we must make the abnormal infant normal, and where this is not possible, help him and his family to make the best of his handicap and live as full a life as his capacities allow. Sometimes, as in the case of a simple intestinal obstruction, normality is attainable; at other times, as with a mongol, it is not possible at all. Early diagnosis of each and every abnormality will enable the best use to be made of the resources available. In the example of intestinal obstruction, the earlier the diagnosis is made, the better is the infant's chance of survival. With the mongol, early diagnosis may be less vital, but it gives the physician more time in which to plan how best to help the family. Sometimes it is not only the infant whose life may be endangered by a malformation. The prenatal diagnosis of hydrocephalus and conjoined twins, for instance, may greatly help the obstetrician to avoid dangerous hazards.

It is with early diagnosis that this book is concerned. This is interpreted as meaning early in the infant's life as well as early in the course of the disorder. The emphasis is on diagnosis during the four weeks before and the four

weeks after the birth of the baby. The book is intended for those who have the privilege of caring for mothers and their infants during this critical, exciting, and responsible time.

LIVERPOOL, 1963

R. W. SMITHELLS

Acknowledgements

I OWE an incalculable debt to Dr. Ronald Mac Keith, at whose suggestion this book was written. He has given me invaluable guidance at every stage and is responsible for much of what may be found helpful. The shortcomings are my own.

I should like to thank Professor John Hay and Dr. Elton Goldblatt for their advice on the sections on congenital heart disease; Dr. Norah Walker and Dr. Constance Davies for their assistance with some of the radiographs; and Dr. Bryan Hibbard for helpful discussion of some obstetric problems. Friends who have provided material for the illustrations are acknowledged in the text, and I am grateful to Mr. Charles Fitzsimons for most of the photography.

Finally, I wish to acknowledge the help I have been given by my publishers.

Contents

CHAPTER 1

Prenatal Diagnosis

INDICATIONS FOR INCREASED VIGILANCE

THE prenatal recognition of foetal disorders may be of the greatest importance to obstetrician and paediatrician alike. The obstetrician is concerned with the diagnosis of foetal malformations that may affect the course of pregnancy or labour and influence his management of them. The paediatrician also is concerned with disorders initiated *in utero*, notably asphyxia and haemolytic disease.

In some instances a definite prenatal diagnosis of malformation can be made, but more commonly there are circumstances which merely increase the likelihood that a particular pregnancy will end in the birth of an abnormal baby. These factors should increase the vigilance of those responsible for the care of the mother and baby. The most important of them are:

(1) A family history of heritable disorders.
(2) An excess or deficiency of amniotic fluid.
(3) Persistent foetal malpresentation or abnormal attitude.
(4) Maternal ill-health in the first trimester.
(5) Certain drugs taken in the first trimester.
(6) A history of recurrent foetal wastage.
(7) Increasing maternal age and parity.

Although the majority of congenital abnormalities will be revealed by the routine examination of the newborn baby, others may require more detailed examination or laboratory tests to establish the diagnosis. The selection of infants requiring such special investigations often begins in the prenatal period; they may be impracticable or inadvisable as a routine but are, in some cases, highly important.

Heritable disorders

A family history of heritable disorders will clearly prompt a particularly careful search for evidence of the disease in the new baby. Conditions inherited as Mendelian dominants are far less common than recessive disorders because they tend continually to eliminate themselves. Strictly speaking, it is not the diseases that are dominant or recessive but the genes that control them.* These dominant genes may have been inherited from a parent or may have arisen by mutation. If the gene invariably causes serious disease (for example, achondroplasia), there is little opportunity for it to be handed down to another generation, and the majority of such disorders will arise by mutation. Conversely, if the effect of the gene is relatively trivial (aniridia, for instance) or is delayed until after the reproductive period (Huntingdon's chorea), the disease will usually be found in one or other parent. A third alternative is seen when the gene has variable expressivity, causing only minor symptoms in the parent but severe disease in the infant. An example is fragilitas ossium (osteogenesis imperfecta). The infant may be born with multiple fractures and live only a short time. In about half the cases there is a family history of a less severe tendency to fractures, or otosclerosis, or simply blueness of the sclerotics.

* Geneticists seem to be divided as to whether it is the genes or the characters determined by them that are dominant or recessive. Fortunately, there is authoritative support for either view.

In two of these three groups of dominant disorders, therefore, there may be a positive family history which forewarns the clinician. Each child born to a parent with a dominant gene has a 50–50 chance of inheriting the gene and manifesting the disease. However, the birth of a baby with a gene mutation, for example, an achondroplastic infant born to healthy parents, in no way diminishes the probability that future babies will be normal. Unfortunately, even when there is the closest co-operation between obstetrician and paediatrician, significant family illnesses may come to light only in retrospect. An infant with unexplained jaundice may turn out to have a mother whose spleen was removed for hereditary spherocytosis twenty years previously.

Recessive genes are manifest only in the homozygote, who carries a pair of abnormal genes. One gene has been inherited from each of the parents, who are 'normal' heterozygous carriers. It is now possible to detect some heterozygotes by special tests, but these are rarely done except after the arrival of an abnormal baby. It is, therefore, almost impossible to foresee a recessive condition in the first affected child to be born in a family. Once an affected child has been born, the same condition can be expected, on average, in one-quarter of subsequent siblings. The most common recessive gene in Great Britain is that for fibrocystic disease of the pancreas (mucoviscidosis), but in other countries the gene for sickle haemoglobin is far more widespread. This gene persists because the heterozygotes are at an advantage in terms of resistance to malaria and will presumably become less common if malaria is ever eradicated on a global basis. The heterozygotes for fibrocystic disease must also have some advantage over those without the gene, but nobody yet knows what this advantage is.

The inborn errors of metabolism are almost all inherited as Mendelian recessive characters, albinism, phenylketonuria, and galactosaemia being among the most familiar. Congenital adrenal hyperplasia follows the same pattern,

as do most examples of the lipidoses. The birth of one child with any of these conditions will alert the clinician to the possibility of recurrence in subsequent children.

In sex-linked inheritance the abnormal gene is borne on an X chromosome. In the female, this gene is usually recessive, being suppressed by the allelomorph on the other X chromosome. In the male, the Y chromosome is very small so that most of the single X chromosome is unpaired; the abnormal gene is therefore manifest. The most familiar examples of sex-linked abnormalities are haemophilia, Christmas disease, pseudohypertrophic muscular dystrophy, and congenital hypogammaglobulinaemia. There may be a history of these conditions in previous male siblings, in maternal uncles or in the male siblings of female forebears, but it is possible for these genes to be carried silently through generation after generation down the female line. The genes determining the production of glucose-6-phosphate dehydrogenase, deficiency of which causes haemolytic anaemia, and the blood group Xg^a are also situated on the X chromosome.

In many congenital abnormalities there is undoubtedly a genetic factor even though the classic Mendelian patterns may not be seen. This is certainly true of central nervous system malformations, but usually these can be diagnosed without much difficulty. It is equally true of congenital 'dislocation' of the hip, which is more easily overlooked. A history of this condition in any close relative is an indication for meticulous and, if necessary, repeated examination of the baby's hips (*see* p. 99).

Whenever there is a history of abnormality in a previous baby in a family, this abnormality must be excluded in the new infant, whether it is believed to have a genetic basis or not. Few mothers have any knowledge of genetics, and there is always a fear, often unspoken, that a malformation may recur. Mothers who have previously had abnormal babies need assurance, therefore, not only that the new baby is healthy, but that the brain, the heart, the palate, or whatever was malformed in the previous baby, is normal.

Sometimes, as in the case of the lipidoses, only the passage of time can provide this assurance.

Excess or deficiency of amniotic fluid

An excess or deficiency of amniotic fluid is often a warning sign that foetal development is not proceeding normally. There is still much discussion about the precise mechanisms of production and circulation of amniotic fluid. However, there is no doubt that foetal swallowing plays a part in its removal, and that foetal micturition contributes towards its production. Congenital abnormalities that interfere with swallowing tend, therefore, to be associated with polyhydramnios, while those that prevent micturition are often associated with oligohydramnios.

The incidence varies widely in reported series, but, in round figures, polyhydramnios complicates about 1 per cent. of all pregnancies. The proportion of these cases associated with foetal malformation also varies very considerably, but in most series the figure lies between 25 and 40 per cent. (Stevenson, 1960). The proportion associated with multiple pregnancy is about 5 per cent. Polyhydramnios is therefore far more likely to be accompanied by malformation than by multiple pregnancy.

By far the most common malformation to be associated with polyhydramnios is anencephaly. It has been shown by amniography that some anencephalic foetuses do not swallow, but the reason for this is a matter of conjecture. It may be that hyperextension of the neck causes mechanical obstruction of the pharynx, or that the swallowing reflex is faulty. Some anencephalic babies born alive have normal sucking and swallowing reflexes, but it has not yet been shown whether these particular infants are associated with maternal polyhydramnios. Iniencephaly is a less common related malformation, in which the base of the skull and the cervical spine are grossly abnormal. The cerebral hemispheres lie immediately behind the pharynx, and the

'neck' is grossly hyperextended. Polyhydramnios is seen with iniencephaly and with the Klippel-Feil syndrome, which is anatomically related to iniencephaly and is characterized by malformation and fusion of the cervical vertebrae.

The second important group of malformations associated with polyhydramnios is that of the high intestinal obstructions, principally oesophageal atresia; some examples of diaphragmatic hernia and duodenal obstruction also fall into this category. It is in this group that the recognition of polyhydramnios and its significance is so important, for, in contrast with anencephaly, early diagnosis leading to early surgery may save the infant's life. In practice, the diagnosis of polyhydramnios will prompt radiological examination. If this shows neither multiple pregnancy nor foetal malformation, and the baby at birth appears normal, steps should immediately be taken to exclude oesophageal atresia (*see* p. 136).

Polyhydramnios also occurs in association with other malformations, often multiple and apparent at birth, and with hydrops foetalis and maternal diabetes.

Oligohydramnios is far less commonly noted than polyhydramnios, partly because it is less easy to recognize, but it is far more regularly associated with foetal malformation. It should be suspected when, the foetus being alive, the uterus fails to grow at the normal rate. X-ray examination may reveal a cramped appearance of the foetus (Fig. 1). The diagnosis is confirmed by the absence or gross deficiency of liquor amnii when the membranes rupture. If there is no opportunity to make this observation, examination of the foetal membranes will usually reveal amnion nodosum (*see* p. 31). In this condition the amnion is studded with small, yellowish nodules composed of foetal squames.

Foetal abnormalities associated with oligohydramnios are usually those that prevent the passage of urine into the amniotic cavity. These include renal agenesis, obstructive lesions of the urinary tract and polycystic kidneys. In-

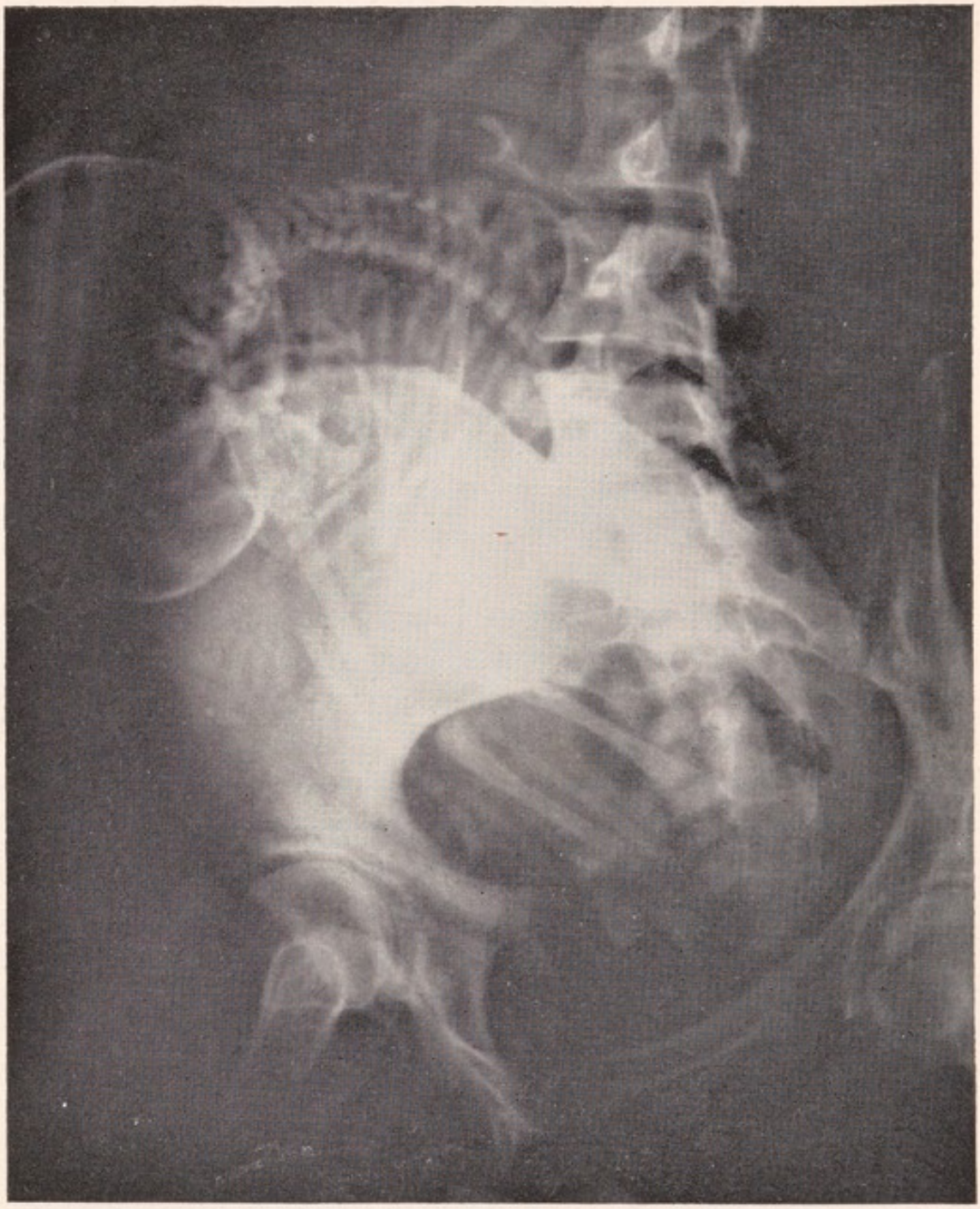


FIG. 1. Oligohydramnios. The foetus appears cramped and presents by the breech. This is commonly seen with renal agenesis.

spection of the face and palpation of the abdomen will help the clinician to make the correct diagnosis (Table 1).

TABLE 1

FOETAL ABNORMALITIES ASSOCIATED WITH OLIGOHYDRAMNIOS

Malformation	*Potter facies* (*see page 53*)	*Kidneys*	*Bladder*
Renal agenesis	Present	Not palpable	Not palpable
Polycystic kidneys	Absent	Very big	Not palpable
Obstruction of the ureters	Absent	Very big	Not palpable
Obstruction of the urethra	Absent	Very big	Distended

Infants with renal agenesis die very soon after birth because of the associated pulmonary hypoplasia, and those with polycystic kidneys a little later because of renal failure. In contrast, babies with obstructive lesions require early surgery. As will be seen from Table 1, it may not be possible to distinguish between polycystic kidneys and bilateral ureteric obstruction except by exploration.

Persistent foetal malpresentation

Abnormal attitude, or persistent foetal malpresentation, is attributable to foetal abnormality only in a minority of cases. It is common with polyhydramnios; it is also common with oligohydramnios, about 50 per cent. of babies with renal agenesis presenting by the breech. Breech presentation is also common in hydrocephalus, the usual explanation being that the larger end of the foetus occupies

the larger end of the uterus (*but see* p. 14). Face presentation is seen with anencephaly and iniencephaly, and, rarely, when a tumour in the front of the neck pushes the head back. Abnormal extension of the neck is sometimes seen in achondroplasia (Fig. 5, p. 19); this not only results in a face presentation but may interfere with swallowing and cause polyhydramnios.

Maternal ill-health

Maternal ill-health during pregnancy may affect the development of the embryo and foetus. Virus infections in the first trimester, especially the first two months, may be responsible for congenital abnormalities. The only unassailable example is rubella, although there is circumstantial evidence incriminating the viruses of influenza and poliomyelitis. A history of rubella, or contact with rubella, in early pregnancy would prompt a particularly careful examination of the infant's heart, eyes, and hearing. Nevertheless, it is worth emphasizing that only a minority of such infants will be abnormal. The incidence of malformations following rubella lies between 15 and 20 per cent., being rather higher when the disease is contracted in the first month and rather lower when it occurs in the third month (Manson, Logan, and Loy, 1960). Furthermore, rubella may be mimicked clinically by viruses of the ECHO group, and at least one study has failed to show any teratogenic effect of ECHO 9 infection (Kleinman *et al.*, 1962).

Another organism which can undoubtedly produce foetal malformations is *Toxoplasma gondii*, but this protozoon invades the embryo without disturbing maternal health. The mother's infection can therefore only be recognized retrospectively by serological tests.

Chronic maternal illnesses may lead to increased rates of abortion and premature birth, but do not appear to cause congenital abnormalities in humans. There is a slight increase in malformations amongst babies born to mothers with diabetes mellitus, and there is good evidence that

maternal prediabetes and hypothyroidism may also predispose to malformation (Hoet, Gommers, and Hoet, 1960). However, the extra risk is so small that it need not influence the handling of the infant. Notwithstanding the possible teratogenic effect of cortisone and allied compounds, a woman taking such drugs in replacement dosage for conditions of adrenal cortical insufficiency (Addison's disease or after adrenalectomy) should have no special risk of giving birth to a malformed child. Myasthenia gravis in the mother will alert the clinician to the possibility of transient neonatal myasthenia, although this is not properly a congenital abnormality (p. 133). Neonatal thyrotoxicosis, presumably caused by maternal thyroid-stimulating hormone, is occasionally seen in the infants of exophthalmic mothers, whether hyperthyroid or not.

There is no evidence to suggest that malnutrition, however gross, predisposes towards foetal malformation, nor has any teratogenic effect of vitamin deficiency or overdosage ever been demonstrated in man. The effect of threatened abortion is more difficult to assess. It has to be remembered that embryogenesis is complete in eight weeks, ending at the tenth week of pregnancy as usually calculated. After this time, disturbances of the placenta may cause foetal death but cannot cause malformation (with the possible exception of atresias). It is certainly within the bounds of possibility that a threatened abortion before this time might lead to malformation, but convincing evidence of an association is still lacking (Thompson and Lein, 1961).

Drugs taken in first trimester

Drugs taken by the mother during the first trimester may cause foetal malformation, and it is useful to make a note at the first prenatal consultation of any drugs that may have been taken up to that time. Unfortunately, memories are short and unreliable, and patients are rarely told the identity of drugs prescribed for them.

Hormonal drugs that have masculinizing effects, including androgens and some progestogens, may disturb the development of the female genitalia. Taken in later pregnancy they may only cause a reversible enlargement of the phallus, but early administration may cause female pseudohermaphroditism (*see* p. 86).

Antithyroid drugs, including iodides, used in the treatment of maternal hyperthyroidism, may cause foetal goitre (with or without evidence of hypothyroidism) in the newborn baby. Such a goitre is occasionally large enough to cause difficulty at delivery and, by compressing the trachea, to interfere with the infant's breathing. It should be borne in mind that iodides are a common ingredient of proprietary remedies for bronchitis and asthma.

The effect of other hormonal drugs is not yet known. Foetal anomalies have been reported after tolbutamide therapy for diabetes (Larsson and Sterky, 1960) and after insulin treatment for psychiatric disease (Wickes, 1954). Malformed babies have also been born to mothers who have taken cortisone or allied drugs during pregnancy for non-adrenal disease (for example, intractable asthma, or skin disorders). There is no doubt that normal embryogenesis can be disturbed by adrenal steroids in the experimental animal. However, the association in human beings is not yet proved and many women, after taking cortisone derivatives throughout their pregnancies, have been delivered of normal babies.

As might be expected, cytotoxic drugs are likely to interfere with the normal growth and differentiation of primitive embryonic cells. This has been shown repeatedly in the experimental animal. In practice, such drugs are reserved for the treatment of malignant disease and a coincident pregnancy is unusual. In any event, the demands of the mother's illness would outweigh any possible risk to the foetus. However, cytotoxic drugs (for example, aminopterin) have sometimes been used to procure abortion. If the attempt is unsuccessful, malformations may be induced. Quinine may be used for the

same purpose and may cause congenital deafness. It is, however, unusual to obtain a history of attempted abortion, even in a retrospective inquiry, and it is unlikely that the clinician will have this information in advance.

A few other drugs deserve brief mention. It is not clear whether streptomycin given to a pregnant woman can damage the foetal auditory pathways, but on *a priori* grounds it seems unlikely. The effect of radio-active drugs will depend upon the nature and dose of the preparation used and the stage of pregnancy at which it is given. Clearly it is wise to avoid the use of such drugs altogether during the first trimester.

This comment is equally applicable to all drugs, except those that are considered strictly necessary to the well-being of the mother. The hypnotic drug thalidomide was withdrawn from the market in November 1961, following evidence that its administration in early pregnancy could lead to severe foetal malformations affecting particularly the limbs, ears, alimentary tract, and heart. Each new drug that becomes available must be regarded as potentially teratogenic until experience has shown it to be safe.

Recurrent foetal wastage

In general, a history of recurrent foetal wastage suggests that it is quite likely to continue. Some malformations, especially those of the nervous system, are more frequently preceded by miscarriages than usual. Much has still to be learnt about the relationship of recurrent foetal wastage to genetic and endocrine factors.

Maternal age and parity

Maternal age and parity affect the likelihood of congenital abnormalities in a general way but, with the exception of mongolism, cannot alert one in any particular direction. The incidence of malformations is highest in

the first pregnancy and from the fifth onwards. Increasing age and parity probably have independent effects but, in practice, are usually inseparable. The chromosomal aberration of non-disjunction, which is responsible for most examples of mongolism, occurs more frequently as maternal age increases (*see* pp. 56 and 174). About 2 per cent. of babies born to mothers over 40 years old are mongols, which may be compared with an overall incidence of one in every 600–700 births. It is important not to confuse this risk of mongol incidence with the risk of mongol recurrence, which is highest in young mothers.

DEFINITIVE PRENATAL DIAGNOSIS

The preceding section described the principal factors that increase the likelihood of foetal abnormality. These tend to put the clinician on his guard and may indicate the need for special investigations of an apparently normal baby. In a limited number of instances it is possible to make a definite diagnosis of malformation before birth, usually by radiological examination but occasionally by other means. In most cases, the special test that reveals the abnormality is prompted by one of the alerting factors, such as disproportion, polyhydramnios, or persistent malpresentation, although sometimes an abnormality is recognized quite fortuitously.

Radiological examination of the foetus

Radiological examination of the pregnant woman's abdomen has been undertaken with circumspection since the suggestion by Stewart *et al.* (1958) that foetal irradiation is associated with an increased incidence of childhood leukaemia, despite the fact that attempts to confirm this work have been unsuccessful (Court Brown, Doll and Hill, 1960). Nevertheless there are circumstances in which the

advantages to be gained by learning more about the foetus or the mother's pelvis outweigh any possible risks of this kind. By limiting the number of exposures, the amount of radiation can be kept at relatively low levels. Amongst the indications for such radiological examinations are: disproportion, persistent abnormal attitude or presentation, polyhydramnios, oligohydramnios, and failure of the uterus to enlarge normally. Any of these may be associated with foetal abnormality. Since only the skeleton of the foetus is demonstrable with X-rays, malformations detectable in this way are almost entirely restricted to those involving the bones. Sometimes, however, a grossly abnormal foetal posture gives a strong hint of a soft tissue abnormality. Thus, hyperextension of a cervical spine may be associated with a large congenital goitre.

Anencephaly is frequently associated with hydramnios, and either this, or the clinician's failure to define a head on abdominal palpation, prompts the taking of an X-ray film. The radiological appearances in this condition are unmistakable (Fig. 2), and the diagnosis in a substantial majority of cases is therefore made before birth.

Hydrocephalus (Fig. 3) is only exceptionally associated with hydramnios, but frequently leads to breech presentation or failure of the head to engage in the pelvis. The frequency of breech presentation is usually attributed to 'the large head occupying the larger end of the uterus'. This hypothesis is not entirely satisfying, because the larger end of the normal foetus (the head) usually occupies the smaller end of the uterus. The inability of the enlarged head to enter the normal pelvis (which is not quite the same thing) may be part of the explanation. Recently, however, obstetricians have formed the opinion that the position of the legs is an important factor in determining the final presentation, and one cannot help recalling that the majority of hydrocephalic infants have an associated myelomeningocele and some degree of paralysis of the legs. They are, therefore, unable to maintain the normal, fully flexed attitude *in utero*, and this may interfere with spontaneous

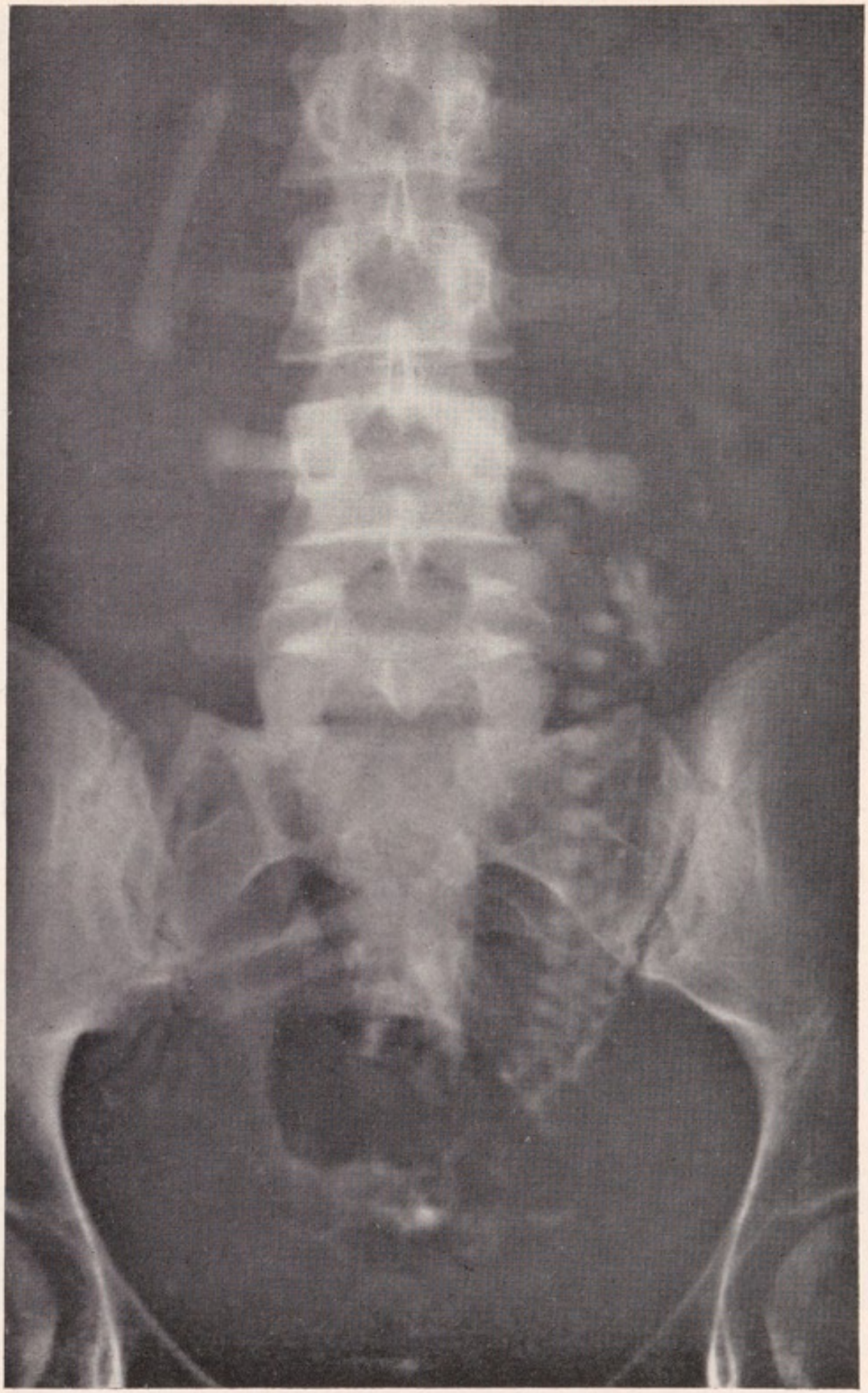


FIG. 2. Anencephaly. The base of the skull is visible but there is no vault.

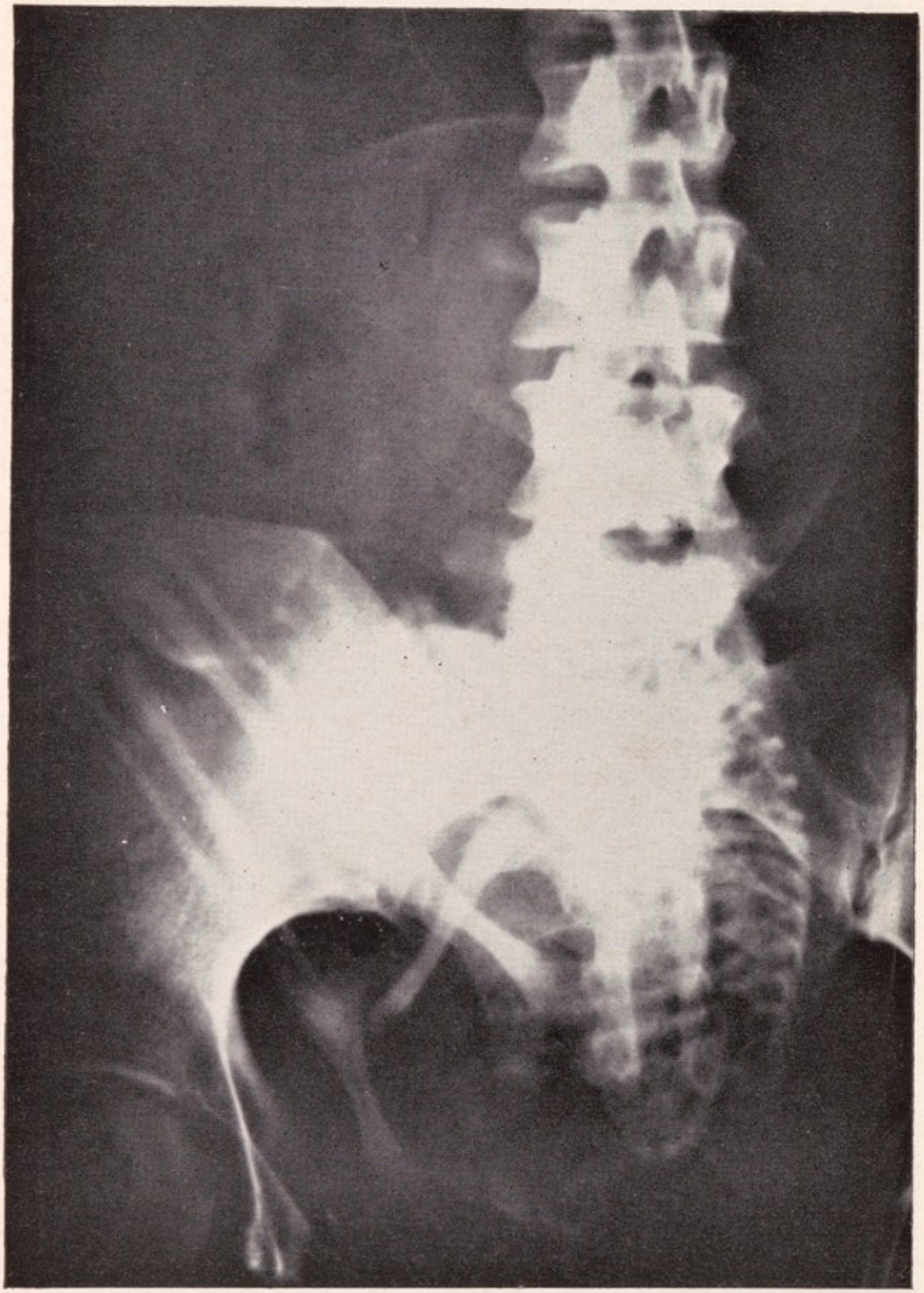


FIG. 3. Hydrocephalus. The head is enormously enlarged. The foetus presents by the breech. The legs are extended.

version in those infants that present by the breech in the earlier months of gestation.

Myelomeningocele can sometimes be diagnosed before birth if the associated deformity of the spine is sufficiently marked. An example is shown in Fig. 4. It must be admitted, however, that in this condition diagnostic errors are made in two directions: sometimes the X-ray appearances give rise to anxiety about the integrity of the spine, which is found at birth to be normal; on other occasions, when a baby is found to have myelomeningocele, retrospective examination of the films will reveal an abnormal angulation of the lumbar spine that had been overlooked. Since most instances of spina bifida are not associated with any of the alerting factors previously mentioned, there is usually no indication to take an X-ray film. Thus, the condition is likely to be diagnosed prenatally only if there is an associated hydrocephalus of sufficient size to cause trouble.

Renal agenesis cannot be diagnosed with absolute certainty before birth, but the circumstantial evidence often leaves little room for doubt. The associated oligohydramnios has already been mentioned and is recognised by a uterus that is small in relation to the length of gestation. The foetus is shown radiographically (Fig. 1) to have a cramped appearance and in about half the cases it presents by the breech. In this condition there is no question of one end of the foetus being too big to engage in the pelvis. It would seem that the position of the foetus in early pregnancy is random and very variable, but as term approaches the majority settle head downwards. The foetus with renal agenesis, instead of being able to manoeuvre round, is virtually in dry dock and must stay where it is. Thus, about half finally present by the vertex and the other half by the breech.

Widespread disorders of the skeletal system may also be picked up on prenatal radiographs. Achondroplasia (Fig. 5) is characterised by short, stumpy limb bones, a small thoracic cage, and a relatively large calvarium.

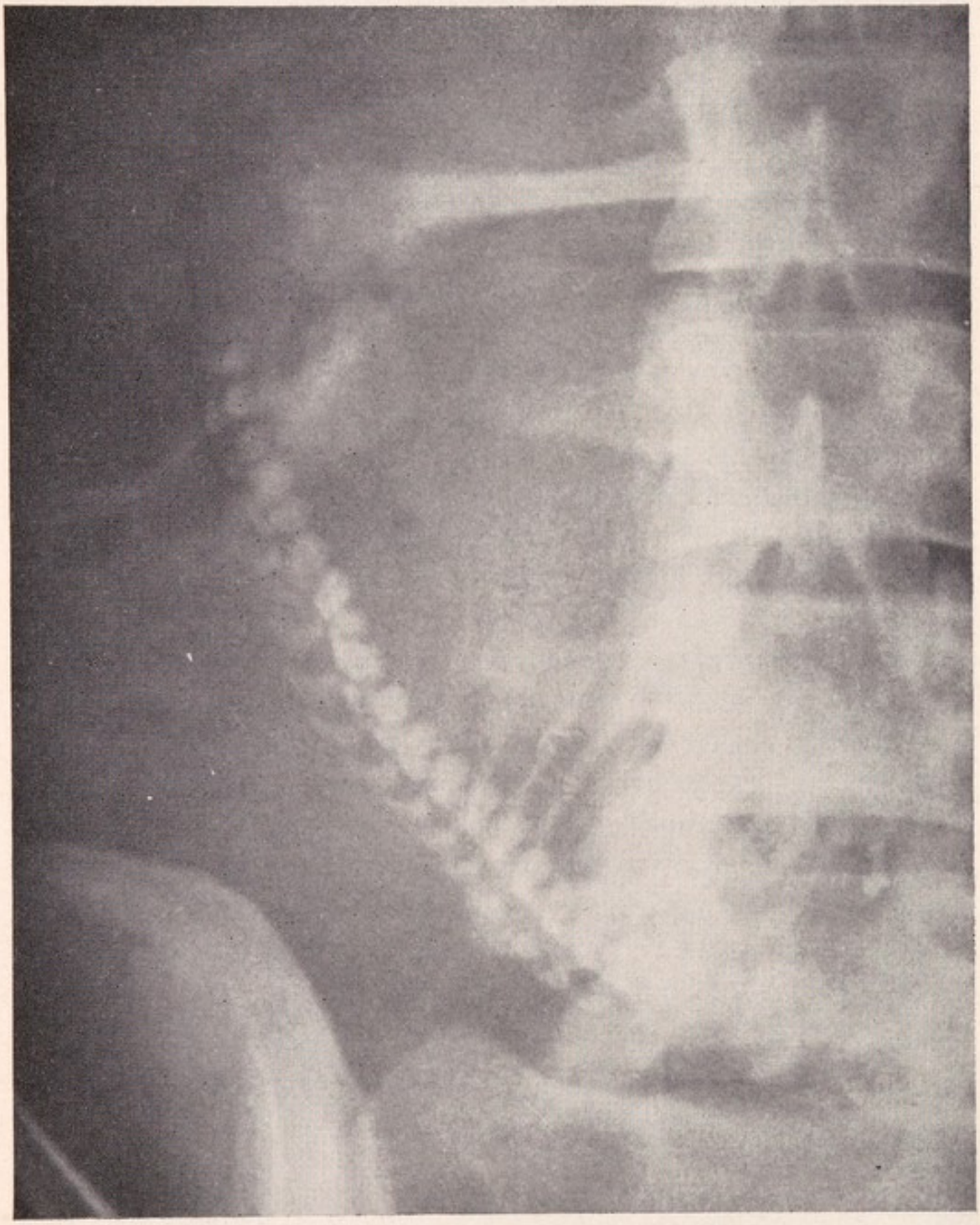


FIG. 4. Myelomeningocele. The foetal spine is seen to be distorted in the lumbar region.

Osteogenesis imperfecta (fragilitas ossium) of the severe, congenital type is superficially similar in that the skull is poorly ossified and the chest rather small, but the diagnostic feature is multiple fractures. These are especially

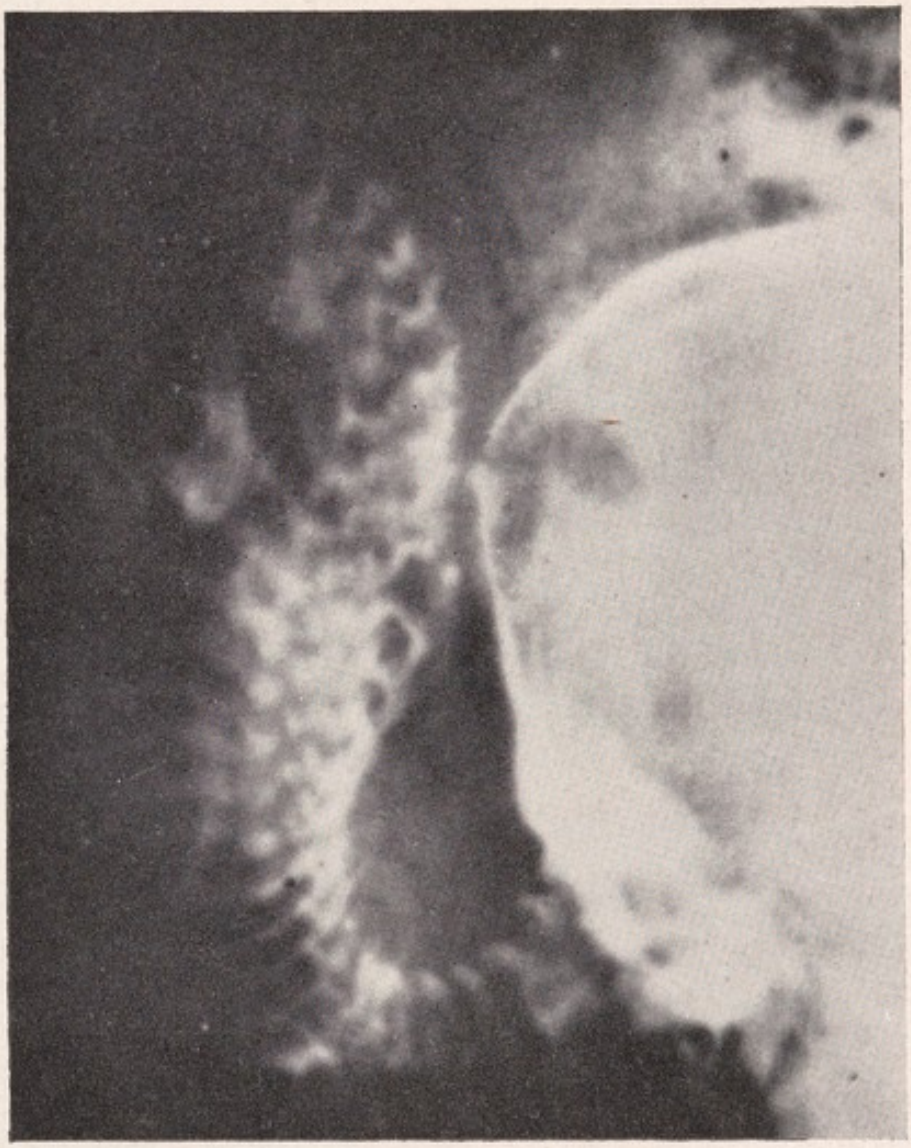


FIG. 5. Achondroplastic foetus. The head is large and the neck hyperextended. The leg bones are short and stumpy.

apparent in the long bones of the limbs and in the ribs; they are often present before birth, and further fractures are likely to be sustained during delivery.

An uncommon condition, illustrated in Fig. 6, is congenital hypophosphatasia. This is an inborn error of metabolism determined by a recessive gene and manifest

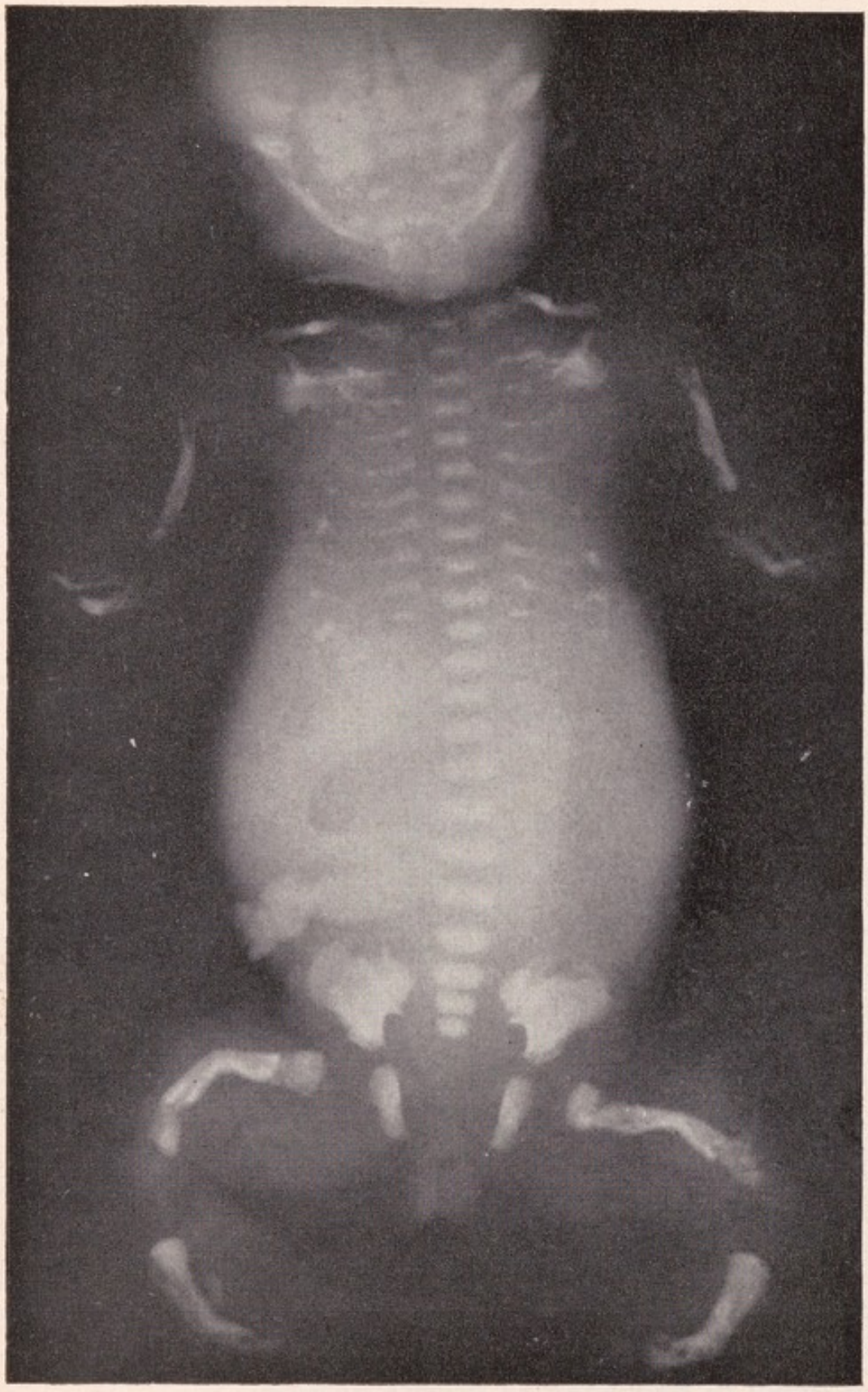


FIG. 6. Hypophosphatasia. In this example the bone changes are diffuse and severe and were detectable on prenatal radiographs. The absence of air from the chest and abdomen suggests that the infant was stillborn. (*By courtesy of Dr. R. M. Todd.*)

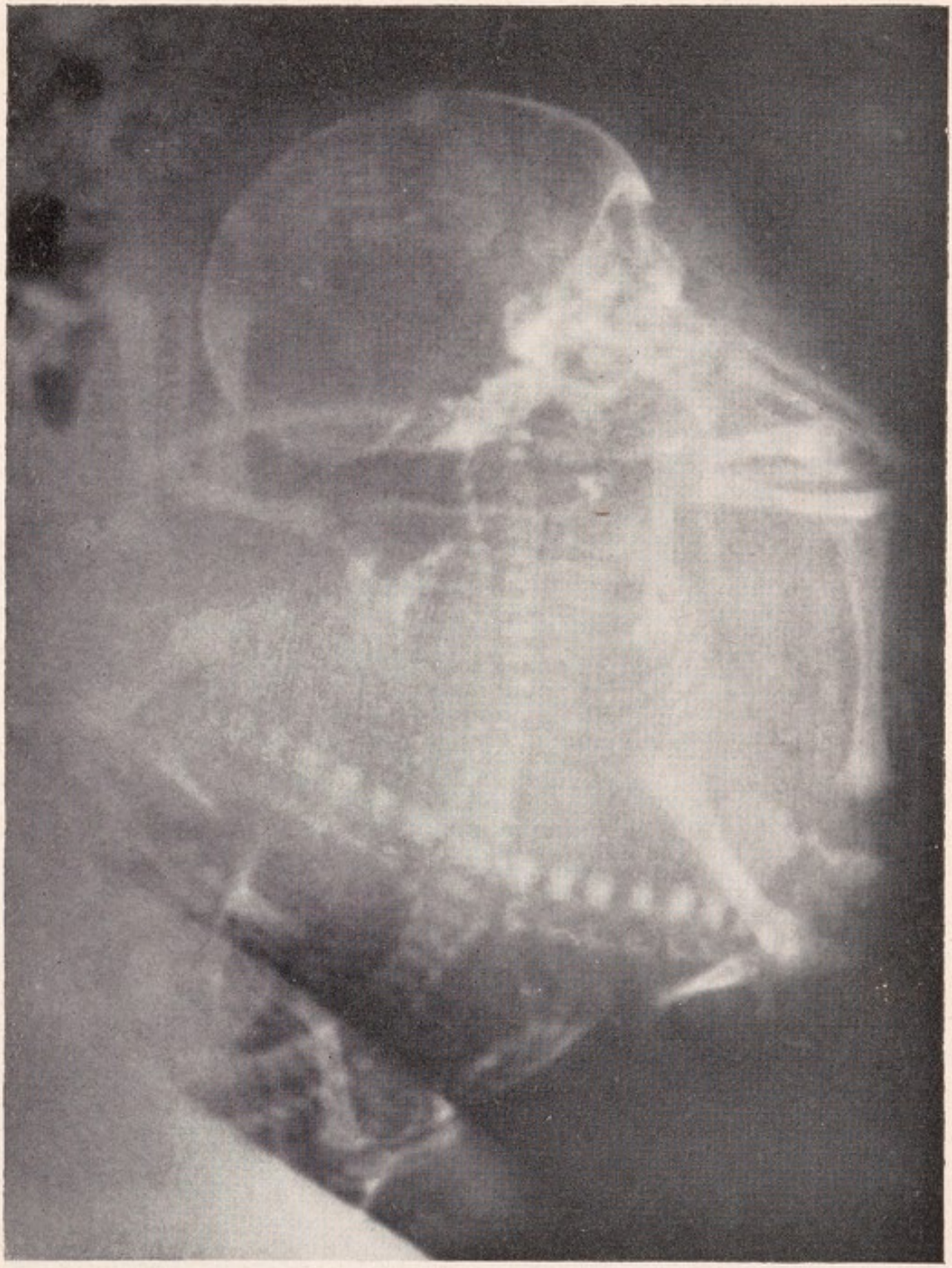


FIG. 7. Conjoined twins. In this case the twins are joined by the spines in the region of the first lumbar vertebrae. (*By courtesy of Mr. C. H. Walsh.*)

by a deficiency of alkaline phosphatase and widespread bone changes which superficially resemble rickets (Lapatsanis and Todd, 1963). There is no particular virtue in diagnosing this disorder before birth, but the radiographic appearances may provide a diagnostic puzzle when seen for the first time. On rare occasions, a radiograph of the foetus has revealed bone disease which would not ordinarily be regarded as a congenital defect. An example is infantile cortical hyperostosis (Caffey's disease), in which gross thickening of bones occurs, involving especially the mandible, ribs, ulna and often other long bones.

Two other groups of congenital abnormalities that may be detected radiologically deserve mention. The first is conjoined twins (Fig. 7). The importance of recognizing this condition before labour begins needs no emphasis. Not only will it have a profound effect upon the management of the delivery, but if there is a possibility of separating the twins subsequently, preparations can be made in advance for intensive investigation and specialized surgery at an appropriate centre.

Finally, a prenatal radiograph will sometimes reveal that there is a major developmental defect of the limbs. During the years 1959–62 many infants were born in Western Europe, Scandinavia, and Australia, with gross defects, sometimes amounting to complete absence of one or more limbs. In numerous instances these malformations were attributable to the drug, thalidomide, a hypnotic, having been taken by the mother during the first trimester. A high proportion of these infants had associated obstructive lesions of the alimentary tract.

In the foetus these lesions had sometimes resulted in polyhydramnios which prompted X-ray examination. In the example illustrated (Fig. 8), the absence of arms was overlooked on the prenatal films, a remarkably easy omission to make, for, as every writer of detective fiction knows, it is far more difficult to appreciate the absence of something normal than the presence of something abnormal. The infant illustrated had duodenal atresia.

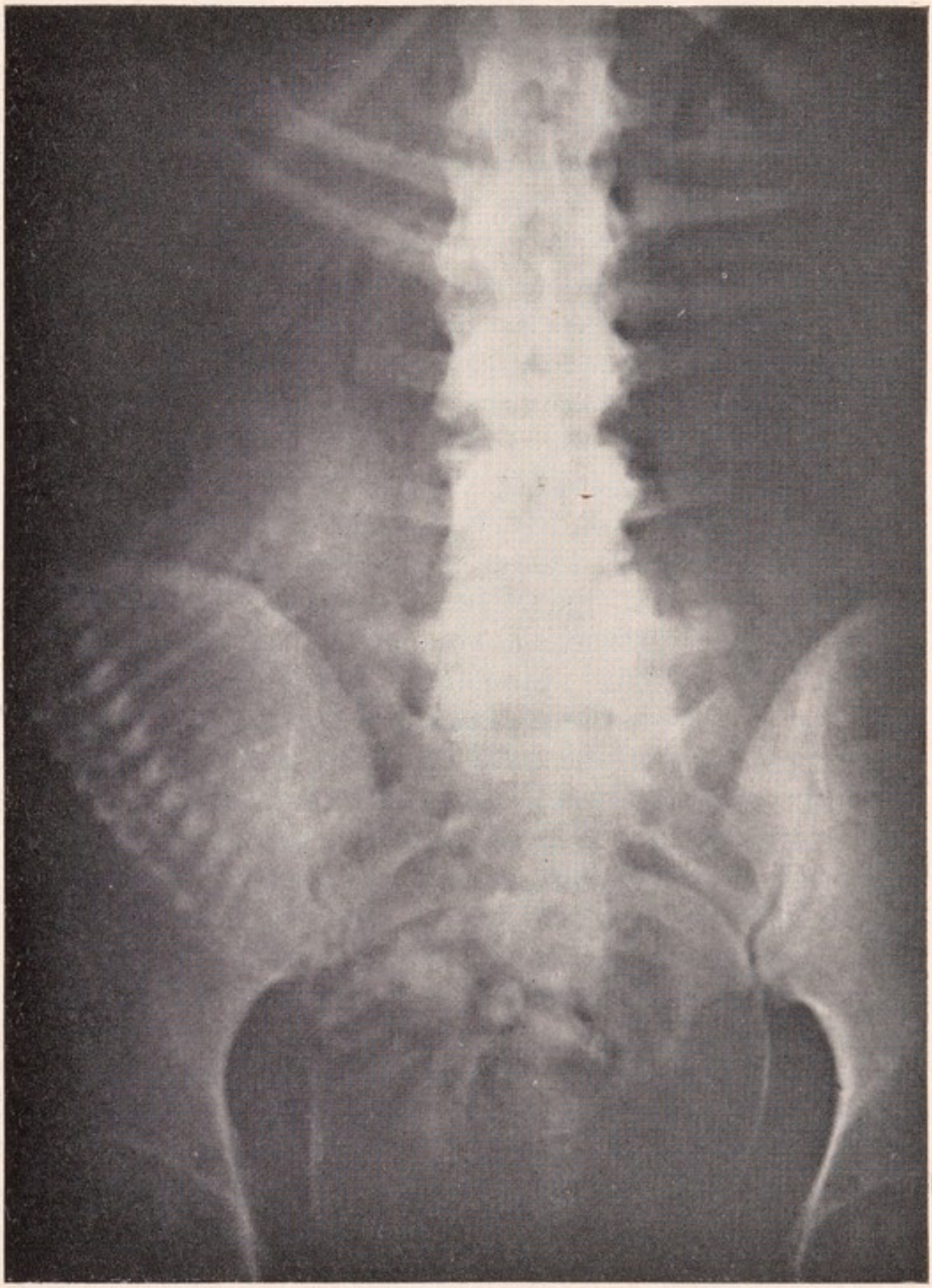


FIG. 8. Congenital absence of arms.

Other methods of examining foetus

Apart from radiology, methods of examining the foetus are very limited.

Auscultation of the foetal heart may reveal a cardiac murmur, but is is unwise to attempt any interpretation of such a murmur until after the baby has been born. Of greater rarity, but more practical importance, is the detection of congenital cardiac arrhythmias. Heart block may be suspected when the foetal heart is regular but the rate is constantly in the region of 40 to 50 beats per minute. It may be possible to confirm the atrio-ventricular dissociation by recording the foetal electrocardiogram. The importance of recognizing the condition is not that it carries any grave risk, but that if the slow foetal heart is not heard until after labour has started, it is likely to be misinterpreted as a sign of foetal distress. This may lead to precipitate intervention not entirely devoid of hazard (Eyton-Jones, 1960). Similarly, paroxysmal tachycardia may give a mistaken impression of foetal distress.

Prenatal sexing of the foetus has been used as a method of definitive diagnostic exclusion, rather than diagnosis (Riis and Fuchs, 1960). When a woman is a known carrier of a sex-linked abnormality, having previously given birth to an affected son, it would be of some prognostic value to know whether a subsequent infant was to be male or female. This is especially true in those countries where the existence of such a carrier state is regarded as adequate grounds for termination of pregnancy. The sex of a foetus can be determined prenatally by examining the nuclear chromatin of foetal squames recovered from the amniotic fluid. The presence of sex chromatin, indicating female sex, would enable the pregnancy to be continued in confidence, although, of course, the girl would have a 50–50 chance of being a carrier.

On the other hand, the establishment of male sex, indicating a 50–50 chance of an affected infant, would give stronger grounds for termination than the overall 1 to 3 risk

where the sex is unknown. The diseases for which this approach might be considered include haemophilia, Christmas disease, congenital hypogammaglobulinaemia, pseudohypertrophic muscular dystrophy, and nephrogenic diabetes insipidus. There is at present little scope for this technique in Great Britain, but the traditional attitude to the termination of pregnancy is perhaps easing a trifle, and a further change in the climate of opinion may provide opportunities for its use.

CHAPTER 2

Diagnosis During Labour

DIAGNOSIS IN THE FIRST AND SECOND STAGES OF LABOUR

MALFORMATIONS may call attention to themselves by interfering with the normal course of labour. During the second stage of labour the average baby fills the average pelvis, and there is remarkably little room to spare. Any substantial enlargement of the whole or of a major part of the foetus is, therefore, likely to cause delay in the second stage, if not complete obstruction. Enlargement of the soft tissues of the foetus needs to be extensive before it will delay delivery, especially when the swelling is of the abdomen, normally one of the slimmest parts of the foetus. By contrast, enlargement of the head need not be very great in order to give rise to trouble because it is already the largest part of the foetus. Furthermore, although a remarkable degree of moulding of the skull bones may take place, the head is relatively incompressible. There seems little doubt that some head moulding occurs within the uterus, that is, before the second stage of labour begins, and that further reduction in head circumference takes place in the vagina.

Enlargement of the head

The head may be enlarged either from within, usually by hydrocephalus and rarely by hydranencephaly (*see* p. 47), or externally by the attachment of a swelling, such as a meningocele. In congenital hydrocephalus the size of the head at birth may be anything from normal to grossly enlarged. If enlargement is extreme, normal engagement of the head is impossible, and the foetus often presents by the breech. These features are easily recognizable on a radiograph. In such cases delivery of the head (often an after-coming head) can be effected only after cerebrospinal fluid has been drained and the skull bones have been allowed to collapse. This can be achieved by paracentesis either *per vaginam* after delivery of the shoulders, or through the mother's abdominal wall after catheterization. In these extreme examples of hydrocephalus, it is clearly helpful to the obstetrician to be aware of the diagnosis before labour begins.

In less severe cases of hydrocephalus, the majority of which are associated with lumbar myelomeningocele, the head may be delivered spontaneously and yet be clearly abnormal at birth. In other instances, the head may be well within the limits of normal size at birth and only enlarge subsequently.

In hydranencephaly, the size of the head at birth is equally variable; it may be normal or be so large as to obstruct labour. The management of this is, of course, the same as that of hydrocephalus. Indeed, unless transillumination is performed, the absence of cerebral hemispheres is likely to be overlooked or discovered only at necropsy.

The commonest swellings on the head are meningoceles and meningoencephaloceles. Usually they are in the occipital region, but they may occur anywhere in the midline between the posterior fontenelle and the root of the nose. Being soft, they do not ordinarily interfere with labour but, if encountered at vaginal examination, they may cause confusion.

Enlargement of the thorax and abdomen

Enlargement of the foetal thorax is extremely rare. Swelling of the abdomen is more common and may be gross. It is not always the result of congenital abnormality, one of the commoner causes being hydrops foetalis. Usually this is due to severe haemolytic disease complicating rhesus incompatibility, but it is not always possible to establish this diagnosis. Congenital chylous ascites has also been described but is a great rarity.

Malformations that can cause abdominal enlargement usually involve the urinary tract. Urethral obstruction may result from complete agenesis, localized atresia at any level, or the presence of urethral valves. Whatever the underlying deformity, there is gross distension of the bladder, dilated and tortuous ureters, and bilateral hydronephrosis. Bilateral ureteric obstruction at the ureterovesical junction causes similar changes in the ureters and kidneys but the bladder is not distended. Congenital polycystic kidneys (Fig. 9) may be large enough to interfere with labour. In the example illustrated, one kidney was haemorrhagic as a result of manipulations that had been necessary to deliver the infant.

A variation on this theme, which is not uncommon and which gives rise to marked distension of the foetal abdomen, is cloacal atresia. In this condition the alimentary and urinary tracts terminate in a common viscus, which represents both rectum and bladder and from which there is no exit orifice. Edith Potter (1961) described an interesting variant in which a urethro-vaginal fistula was associated with vaginal atresia; the urine drained into the genital tract and caused great distension of the vagina and uterus. Distension of the uterus is also seen in hydrometrocolpos. In this condition vaginal atresia is associated with the secretion of large amounts of milky, mucoid fluid.

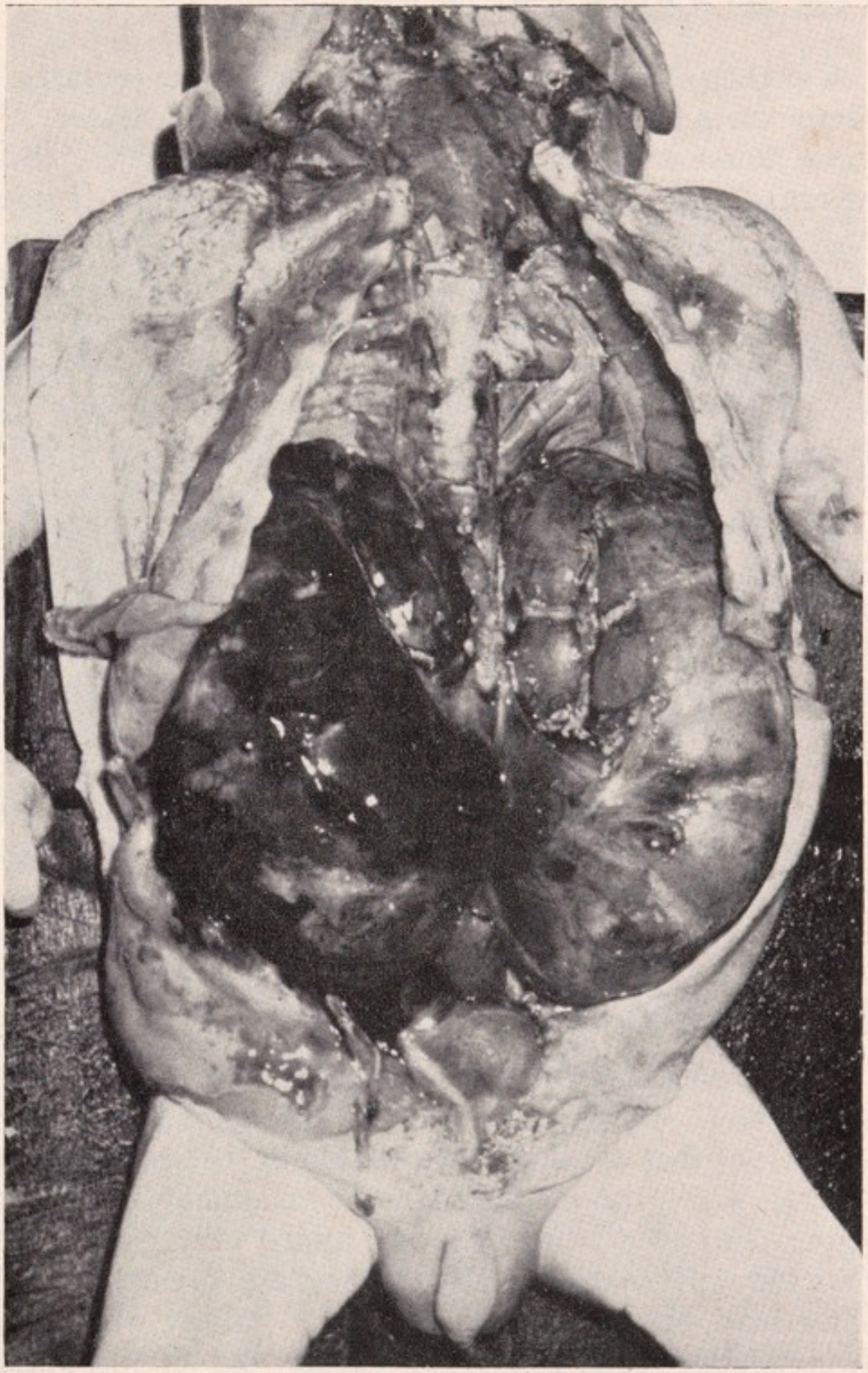


FIG. 9. Congenital polycystic kidneys. Abdominal enlargement caused difficulty at delivery and there is diffuse bruising of the right kidney.

Cysts and tumours

Congenital cysts and tumours may either enlarge the foetus from within or may be attached to the surface. The most familiar cystic swellings are meningoceles, myelomeningoceles, and exomphalos (omphalocele). Tumours may be cystic or solid. The most common cystic tumour is a cystic hygroma (or lymphangioma), which sometimes reaches a very large size. It occurs most frequently in the neck, or over the front or back of the chest. Sometimes diffuse lymphangiectasis affects the whole or a major part of a foetus, and this may also interfere with normal delivery.

The best-known solid tumour is sacral teratoma. These tumours vary from small, sessile swellings to large, pedunculated masses. Because of their position in relation to a flexed foetus, they tend to be towed astern and cause little obstetric difficulty. Other solid tumours at birth are rare; though many different varieties have been described, most are too small to interfere with labour. Wilms' tumour of the kidney may be present at birth but is likely to be diagnosed as polycystic disease until histological sections have been examined.

Conjoined twins

The problem of conjoined twins has already been briefly mentioned. If diagnosis has not been made earlier, the difficulties that are almost inevitable in the second stage of labour will prompt radiographic examination. Craniopagus conjoined twins, who are joined only by the heads, may be delivered vaginally if they are orientated in the same line, but for almost any other variety of conjoined twins spontaneous delivery is usually out of the question if the pregnancy progresses to term or near term.

DIAGNOSIS IN THE THIRD STAGE OF LABOUR

The placenta

Only brief reference need be made to the diagnostic clues that may be detected in the placenta, foetal membranes, and umbilical cord. Examination of the placenta is always a worthwhile exercise, and it is regrettable that this viscus is often disposed of beyond recall, with almost unseemly haste. Both macroscopic and microscopic examination may help to elucidate the cause of an unexpected stillbirth or neonatal death. The foetus and placenta have been united for many months and separated for almost no time at all. Infection acquired *in utero* is likely to affect both. Hydropic changes affect both. A pathologist who is asked to perform a necropsy on an infant that has died before or immediately after birth should certainly expect to examine the placenta and therefore to receive it from his clinical colleagues.

With malformations, examination of the placenta is likely to be rather less rewarding than elsewhere. Congenital abnormalities do not, as a rule, have any effect upon it, although in at least one case the reverse was true. Benson and Joseph (1961) described an infant, born with cardiomegaly and in heart failure, whose illness was found to be due to a chorioangioma of the placental vessels.

The amnion

In every instance in which oligohydramnios has been suspected or proved, confirmatory evidence may be obtained by careful examination of the amnion. This shows the condition of amnion nodosum in which very small, yellowish plaques are seen on the foetal surface of the amnion. Microscopic examination of the plaques shows them to be composed of foetal squames (Figs. 10 and 11). It would seem that the deficiency of liquor

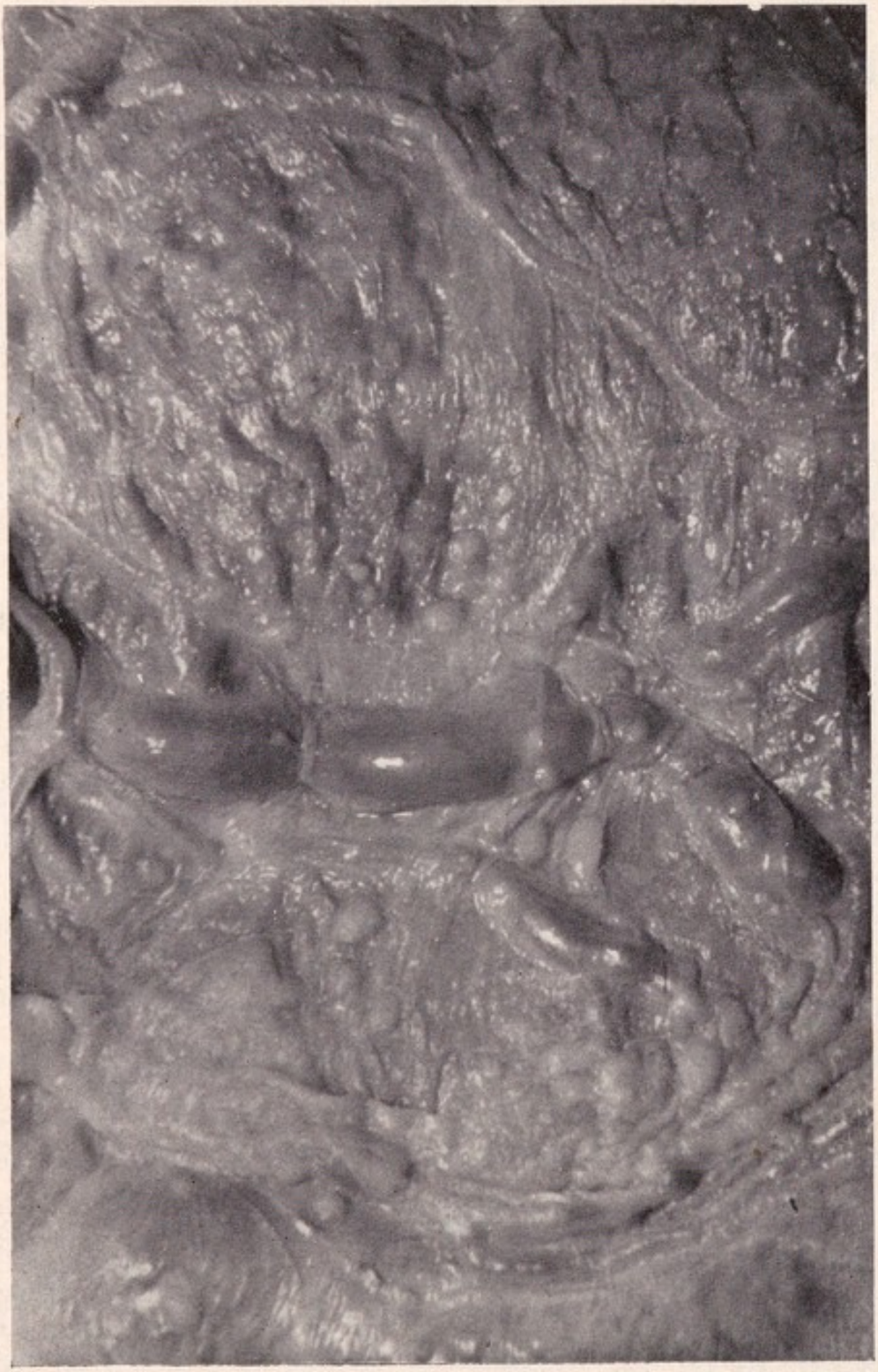


FIG. 10. Amnion nodosum. There are many pale nodules over the amniotic surface of the placenta. There had been oligohydramnios. The infant is shown in Fig. 18 on pages 54 and 55.

amnii has led to intimate contact between the amnion and the foetal skin with the result that the squames, normally shed into the fluid, become adherent to the amnion. It is remarkably difficult to establish the diagnosis of oligohydramnios before birth, and the detection of amnion

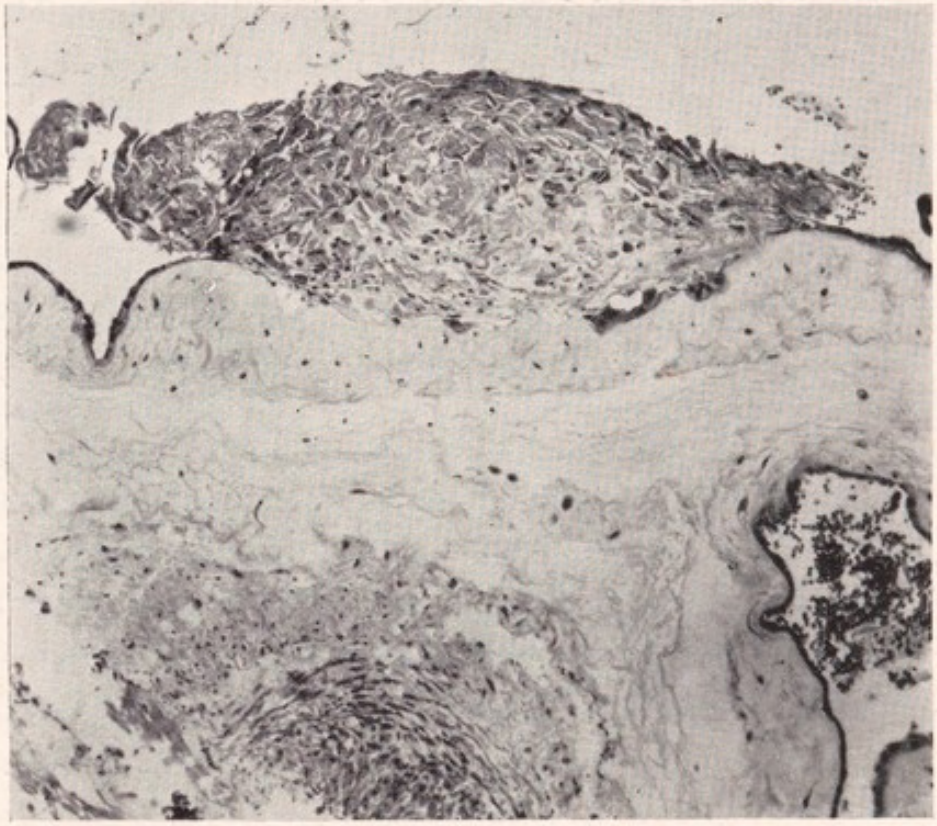


FIG. 11. Amnion nodosum. Section of a nodule showing that it consists of degenerated squames clustered on the surface of the amnion. (×45). (*By courtesy of Professor T. N. A. Jeffcoate and the Editors of the Journal of the Canadian Medical Association.*)

nodosum provides a useful retrospective check. The difficulty arises because, as labour progresses and no liquor appears, it is assumed that the membranes have not ruptured. When eventually the baby is born, and still no liquor is seen, it is assumed that the membranes must have ruptured long before. Sometimes the baby's skin is noticeably dry at birth.

The significance of amnion nodosum is the same as the significance of oligohydramnios, for the two conditions are merely different manifestations of the same phenomenon.

It suggests that the foetus has not micturated *in utero*, and therefore there is either renal agenesis, in which case the facial appearance is usually characteristic, or there is an obstructive lesion of the urinary tract, in which case the abdomen is likely to be distended. Although a deficiency of liquor amnii frequently signifies a deficiency of foetal urine, it may occur with a perfectly healthy baby. Identical abnormalities may occur with normal amounts of liquor. Conversely, the author has seen a case of bilateral renal agenesis associated with polyhydramnios. In the same way oesophageal atresia is usually associated with polyhydramnios, but not always. These observations serve to remind us that there is more than one source of liquor amnii and more than one route for its disposal. The total volume of fluid represents a balance between production and disposal. In general, interference with foetal swallowing tends to shift the equilibrium towards polyhydramnios, and interference with foetal micturition disturbs the balance in the opposite direction. Because the magnitude of the other contributory factors is unknown, however, the unexpected may occur.

Umbilical cord

The umbilical cord should always be examined where it is to be ligated. The inch or two immediately adjacent to the baby may occasionally be seen to contain bowel or, less commonly, a urachal cyst. Larger protrusions of bowel or other abdominal viscera into the cord constitute the condition of exomphalos (omphalocele), which will be obvious at birth. Lesser protrusions should be regarded as small omphaloceles, rather than umbilical hernias, and treated with corresponding urgency. The bowel in the cord often turns out to be a Meckel's diverticulum, and there may be an associated ileal stenosis or atresia.

The significance of a single umbilical artery merits discussion. The vessels are readily distinguishable on the cut end of the cord. The arteries contract tightly and look

very small, and the two lie close together. The vein is of much greater calibre and is situated a little apart from the arteries. Normally the vein is single and the artery double, but inspection of the cut end may show a single artery. It has been found that in about 50 per cent. of babies with single umbilical arteries there are serious internal malformations. The anomaly is not uncommon. Benirschke and Bourne (1960) found 15 examples in 1500 consecutive deliveries—an incidence of 1 per cent. In another paper (Bourne and Benirschke, 1960) they reviewed 113 cases of single umbilical artery and noted an incidence of 7 per cent. in twin pregnancies. In 58 cases there were malformations, mostly multiple, which were incompatible with life in 39 instances. They recommended routine examination of cut cords, but it has to be remembered that many of these malformations are clinically obvious, most are untreatable, and other diagnostic clues, such as polyhydramnios, are often present. However, whenever a single umbilical artery is noted, the baby should be examined with particular care and a close watch kept for symptoms referable to an internal malformation. The extra vigilance thus stimulated may enable a valuable early diagnosis to be made.

CHAPTER 3

Examination of the Symptomless Infant

FROM the foregoing chapters it is clear that only in a small minority of instances can congenital abnormalities be diagnosed prenatally. In a rather larger number of cases one or other of the alerting factors, such as a family history of hereditary disease or polyhydramnios, will have put the clinician on his guard before the infant is born. There remains a very substantial majority of anomalies that cannot be diagnosed or even suspected before birth. Many of these are obvious on the most superficial inspection of the nnewbor infant, and with them this book is not primarily concerned. This chapter is intended to call attention to those abnormalities that may be overlooked or mis-diagnosed by the busy clinician.

The initial examination

Congenital abnormalities that are not apparent at birth will in many cases declare their presence sooner or later by the appearance of symptoms. As a basic principle it is better, whenever possible, to diagnose these conditions before symptoms arise. Great importance, therefore, attaches to the initial examination of every newborn baby. This examination of babies delivered normally at full term should be made not later than 24 hours after birth, and much earlier if the delivery has been abnormal or premature. The purpose of the initial examination is to assess the general health of the baby and to make a particular search for congenital abnormalities, so that where possible

these can be corrected at an early stage by appropriate treatment. In some ways it is analogous to a final inspection at a factory.

It cannot be too strongly stressed that the routine examination of the newborn baby must be carried out with great care and thoroughness, and should not be left, as it so often is, to the busiest and least experienced doctor available. There is a double reason for trying to minimize the risk of overlooking abnormality or disease. The first, which is simply the positive fact of early diagnosis, is obvious. The second is negative and less obvious. An infant that has been examined and declared healthy has been given his passport, so to speak; he is *persona grata*. The newborn infant not yet examined is still on probation. There is likely to be a tendency, subconscious, perhaps, but still present, to pay slightly less attention to minor symptoms in an infant who has been signed up as normal than in one who has not been examined. If an unexamined infant vomits, the doctor will probably be informed. If the vomit occurs after the infant has been declared healthy, there may well be an inclination to wait to see if he vomits again. After all, the doctor said he was normal. It is not suggested that people with medical training would be likely to think this way, but elsewhere there is an exaggerated faith in the scope of routine medical checks, whether of newborn babies, schoolchildren, or business executives. There are many congenital anomalies that cannot be detected by the most competent hands until symptoms develop; but, by instilling false confidence, a hurried or inadequate routine examination may prove more dangerous than no examination at all.

Technique of examination

Each clinician develops his own technique for the routine examination of newborn infants. It needs to be gentle and thorough without exposing the baby to prolonged handling, particularly premature babies. The initial examination of very small prematures often has to be

trimmed to the bare essentials, and a good deal postponed until another day. For example, it matters little if examination of the hips in such an infant is left for a week or two. It does matter if it is altogether forgotten, and this is easily done if the baby goes through a critical period with respiratory distress or deep jaundice. As a standard part of obstetric case notes, it is a help to have a printed form, with headings to act as an *aide-mémoire* and an outline diagram of an infant on which to record abnormal findings. Fig. 12 shows an example of such a case sheet, and there are many others equally satisfactory in use.

Examination of a small infant is most easily done when he is content and relaxed which, for practical purposes, is immediately after a feed. Before a feed he is likely to be crying and, if woken between feeds, he may be resentful. However, in the first twenty-four hours of life, and especially in the first twelve, food may not yet have become his chief concern, and examination is usually possible at almost any time. This is an additional reason for early examination. Auscultation of the heart and palpation of the abdomen and femoral pulses can be carried out satisfactorily only when the infant is quiet, and it is often wise to do these first. Examination of the hips is also more easily performed when the baby is relaxed. On the other hand, auscultation of the lungs and examination of the mouth and palate are often easier when the baby is crying. The only equipment necessary for routine examination of the newborn consists of a warm stethoscope (with an end-piece of plastic or rubber—not cold metal), warm hands, a tape measure and a 'dummy' (pacifier); the latter is usually effective in reducing a noisy baby to temporary silence.

The order in which the various parts of the infant are examined is a matter of personal preference, bearing in mind the remarks that have already been made. A certain amount of opportunism is often necessary. If, for example, a male infant decides to micturate whilst being examined, dissuade the nurse or mother from flinging a nappy over the offending part; stand back and watch.

The infant with a paralysed bladder (usually due to myelomeningocele), or with bladder neck obstruction, or urethral valves cannot pass water with a strong stream. It has been

INFANT

NAME SEX BIRTH WEIGHT LB. OZ.

IDENTITY CHECKED BY GESTATION WEEKS

RELIGION BAPTISED BY ON

CONDITION AT BIRTH AND IMMEDIATE CARE.	DRUGS	DOSE	TIME	INITIALS
NORMAL / O_2 BY MASK / INTUBATION				

SPECIAL INSTRUCTIONS

NORMAL / NURSERY / SORRENTO / INCUBATOR / ALEVAIRE / HIGH HUMIDITY

FIRST EXAMINATION DATE TIME

IMMATURE / MATURE / POST MATURE LENGTH

COLOUR

CRY

MOVEMENTS

TONE

HEAD CIRCUMFERENCE

FONTANELLES & SUTURES

EYES

EARS

MOUTH

HEART

LUNGS

ABDOMEN

FEMORAL PULSES

GENITALIA

ANUS

HIP ABDUCTION

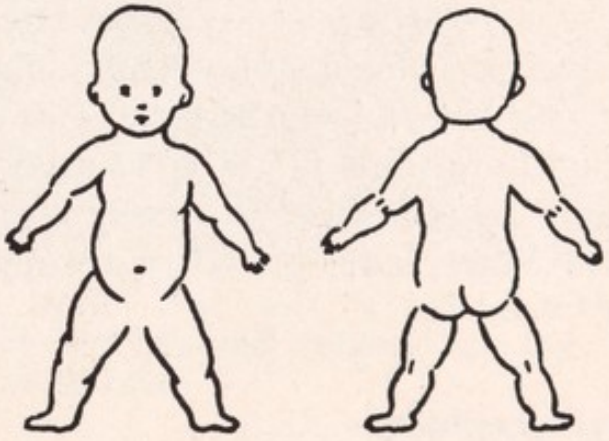


FIG. 12. Case sheet for newborn babies.
(*By courtesy of the East Liverpool Hospital Management Committee.*)

suggested, perhaps with tongue in cheek, that watching micturition should be part of the routine examination of the newborn. Impracticable though this may be as a regular feature, the fortuitous opportunity should never be disregarded.

If some kind of record sheet is used, the examiner need never be afraid of allowing himself to be side-tracked from his routine; it will always remind him of what has not yet been done. The order in which the following sections are arranged is not intended to imply that the infant should be examined in the same way.

EXAMINATION OF THE HEAD

The purpose of examining the head is to detect abnormalities in size, shape and ossification of the skull, and any subcutaneous swelling or defect of the overlying skin.

If the hand and the tape-measure detect any abnormality of the skull, additional information may be obtained by transillumination and radiographic examination. Transillumination must be carried out in a dark room. If there are no facilities for blacking out a room, a large cupboard can be used perfectly satisfactorily. A strong flashlight (not a pen-torch) should be fitted with a cuff of sponge rubber or plastic foam extending about 1·25 cm. ($\frac{1}{2}$ in.) beyond the end of the torch (*see* Fig. 13). This permits a snug, light-tight fit to be secured on the curved surface of the infant's head. Normally around the cuff is seen a red halo which fades off fairly abruptly within half an inch or so.

Hydrocephalus

Hydrocephalus occurs as an isolated abnormality but is unfortunately far more often associated with myelomeningocele. If the head has enlarged appreciably before the baby is born, it will interfere with normal delivery. In this event, surgical intervention to reduce the size of the head is often unavoidable, and the infant is stillborn. In liveborn babies the size of the head may well be within the range of normal, although the shape is often abnormal. The whole brow may be prominent, and the eyes are often

turned downward, constantly or intermittently. Alternatively, there may be a prominent bulge in the centre of the forehead overlying a wide metopic suture.

The size of the fontanelles in normal newborn babies is very variable. The anterior fontanelle is sometimes almost

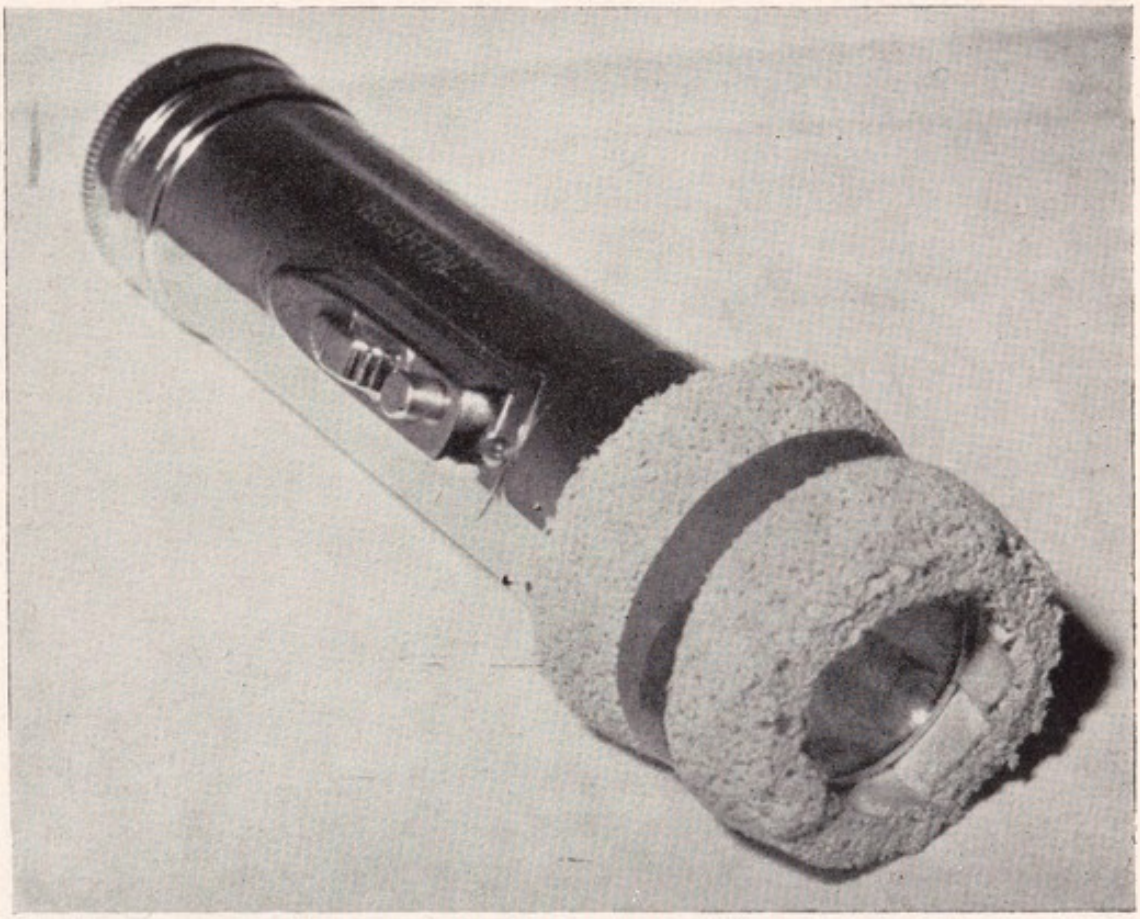


FIG. 13. Transillumination torch. The cuff of sponge-rubber projects beyond the end of the flashlight and makes a light-tight fit on the head.

completely occluded by overlapping of the parietal and frontal bones, and this may take some days to return to normal. In the hydrocephalic, this fontanelle is usually noticeably large at birth, extending at the corners into wide sagittal, coronal and metopic sutures. The frontal bones may be separated as far down as the root of the nose.

The posterior fontanelle is normally very small at birth, and its size is more constant than that of the anterior fontanelle because far less moulding takes place around it. In the hydrocephalic it is almost invariably enlarged,

frequently strikingly so, whilst the anterior fontanelle is within the limits of normal. For this reason it is important to make a practice of feeling both fontanelles. The width of the sutures is usually proportional to the size of the anterior fontanelle, but is not necessarily related to the size of the head. Extremely wide sutures may be felt in a head that has a circumference within the range of normal; these are not necessarily abnormal.

The third fontanelle can easily cause confusion. This is a small fontanelle situated in the sagittal suture about two inches in front of the posterior fontanelle, for which it is often mistaken. A third fontanelle is a perfectly normal finding and may remain open for several months. It is said to be particularly common in mongols, although it is certainly common in healthy infants and is of no significance. If the sagittal suture is wide, the third fontanelle may be represented by notches on opposing edges of the parietal bones. In extreme cases, which are rare, a bony defect in the parietal bones (so-called parietal foramina) may persist throughout life.

The tension of the fontanelles can be judged only when the infant is quiet and, preferably, asleep. There is much to be said for feeling the fontanelles at the very beginning of the examination, before the baby is removed from the cot. Increased fontanelle tension in a newborn baby almost always signifies hydrocephalus, although acquired lesions such as subdural haematoma and meningitis are responsible in rare instances. It is, however, important to appreciate that a normal fontanelle tension is entirely compatible with a diagnosis of hydrocephalus. An increased volume of cerebrospinal fluid may be accommodated by a corresponding reduction in the volume of brain tissue, or by expansion of the head, or by both. While increase in the amount of cerebrospinal fluid usually leads to increase in the size of the head, this may not occur if there is atrophy of the brain.

If the tension of the fontanelles and the size and shape of the head are all normal, hydrocephalus may be over-

looked. There are, however, other diagnostic clues: the veins of the scalp are often prominent, especially in the temporal regions; the surface of the cranial bones may feel irregular, these external corrugations corresponding to depressions on the inner table of the skull caused by

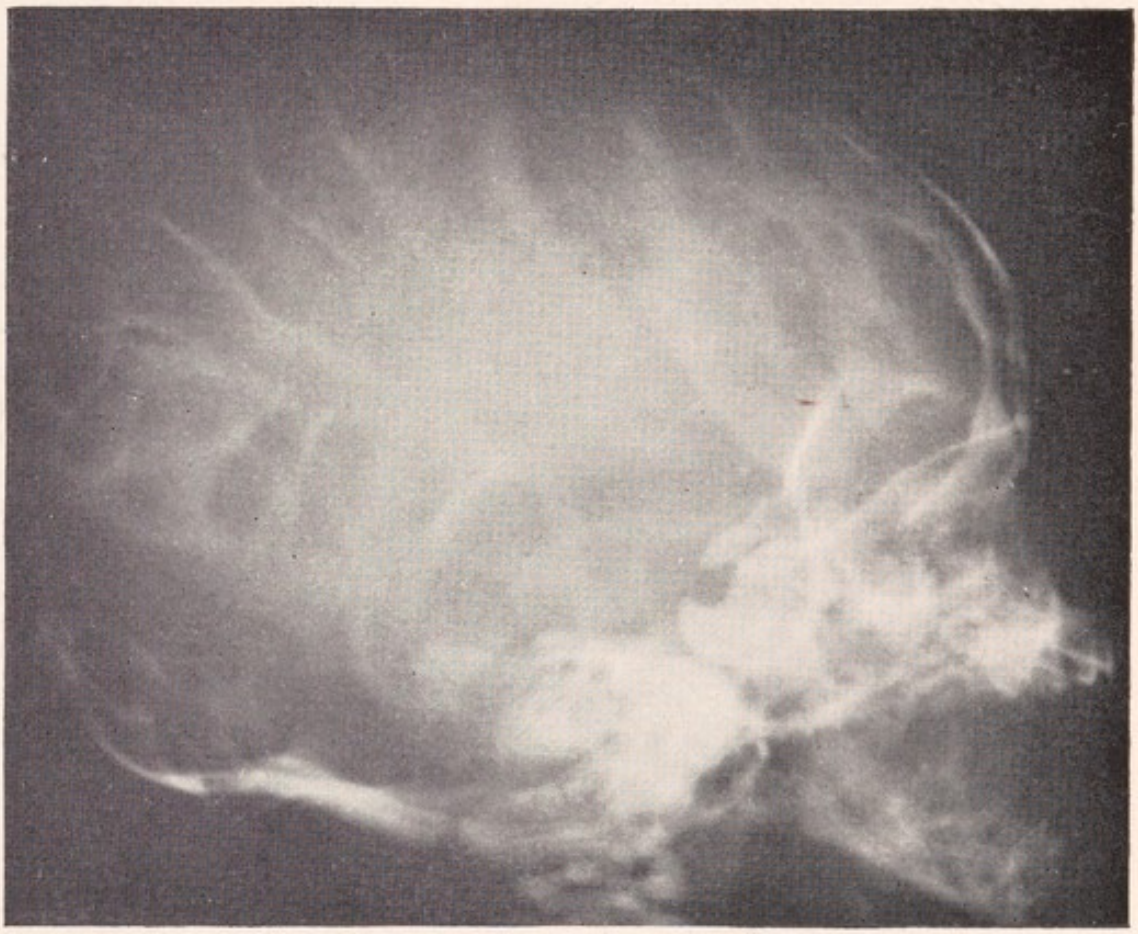


FIG. 14. Lacunar skull.

pressure of the cerebral gyri; the bone is considerably thinner than normal, and the appearances of lacunar skull are seen on a radiograph (Fig. 14).

In the absence of all these signs, the only way in which hydrocephalus can be recognized reasonably early is by confirming that the head is growing with abnormal rapidity by serial measurements of the head circumference. Whatever the circumference of a head at birth, its rate of growth tends to parallel closely the percentile lines shown on the charts (Fig. 15). Some infants with myelomeningocele have a normal head circumference at birth, but serial

measurements reveal an abnormally steep rise of the growth curve when plotted on a percentile chart. It has been shown by pneumoencephalography that these babies have in fact dilated ventricles at birth (Lorber, 1961). Conversely, those with normal ventricles at birth do not

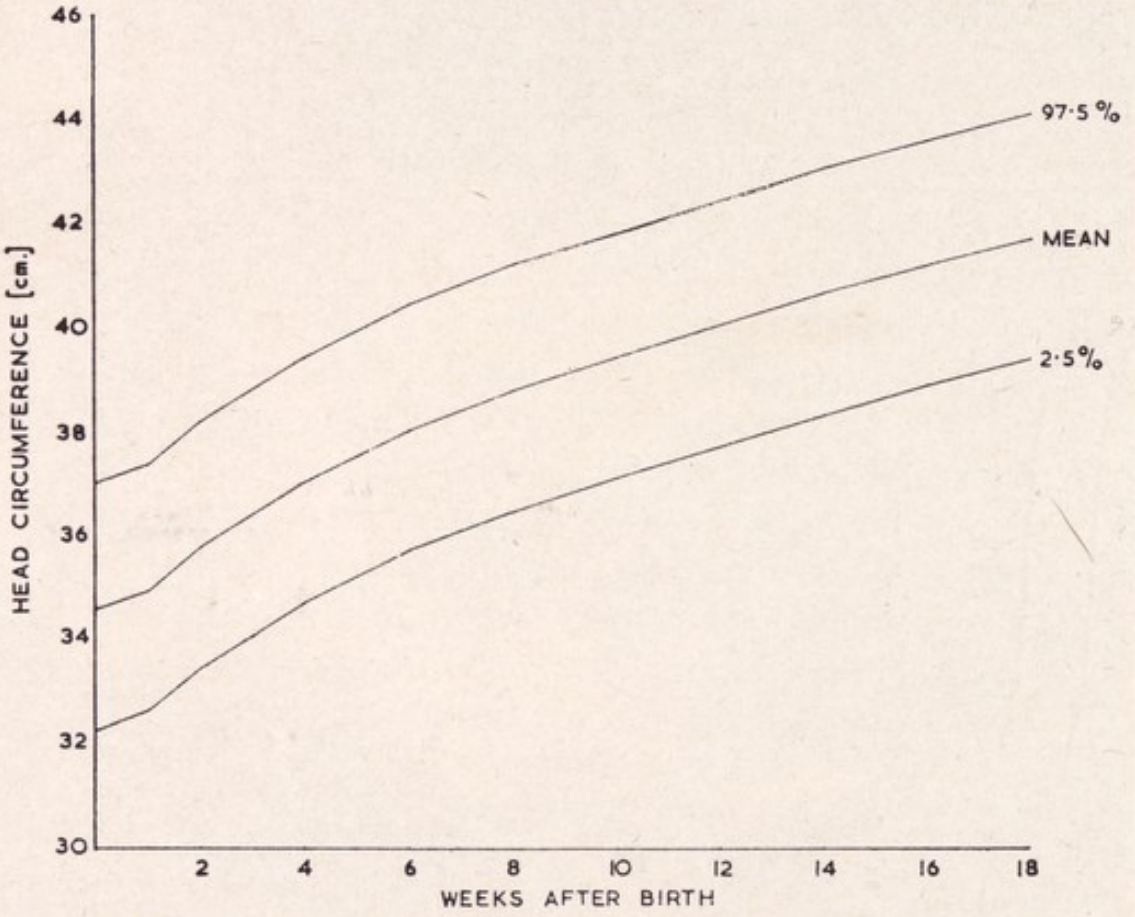


FIG. 15 (*a*). Head circumference/age chart for full-term babies.

develop hydrocephalus later, unless there is some complicating factor such as meningitis. This observation appears to put an end to the theory that repair of a myelomeningocele can precipitate hydrocephalus.

The features that have been described are characteristic of hydrocephalus, but many of them are shared by other disorders. When there is clinical doubt about the diagnosis, the following possibilities are worth considering:

(1) The skull bones (especially the parietals) in some normal infants are so thin that they can be indented with the thumb, the depressions springing out again when

pressure is released. This sign is not necessarily pathological. Again, some normal infants are born with a head circumference of 37 cm. (15 in.) or even 38 cm. (15½ in.).

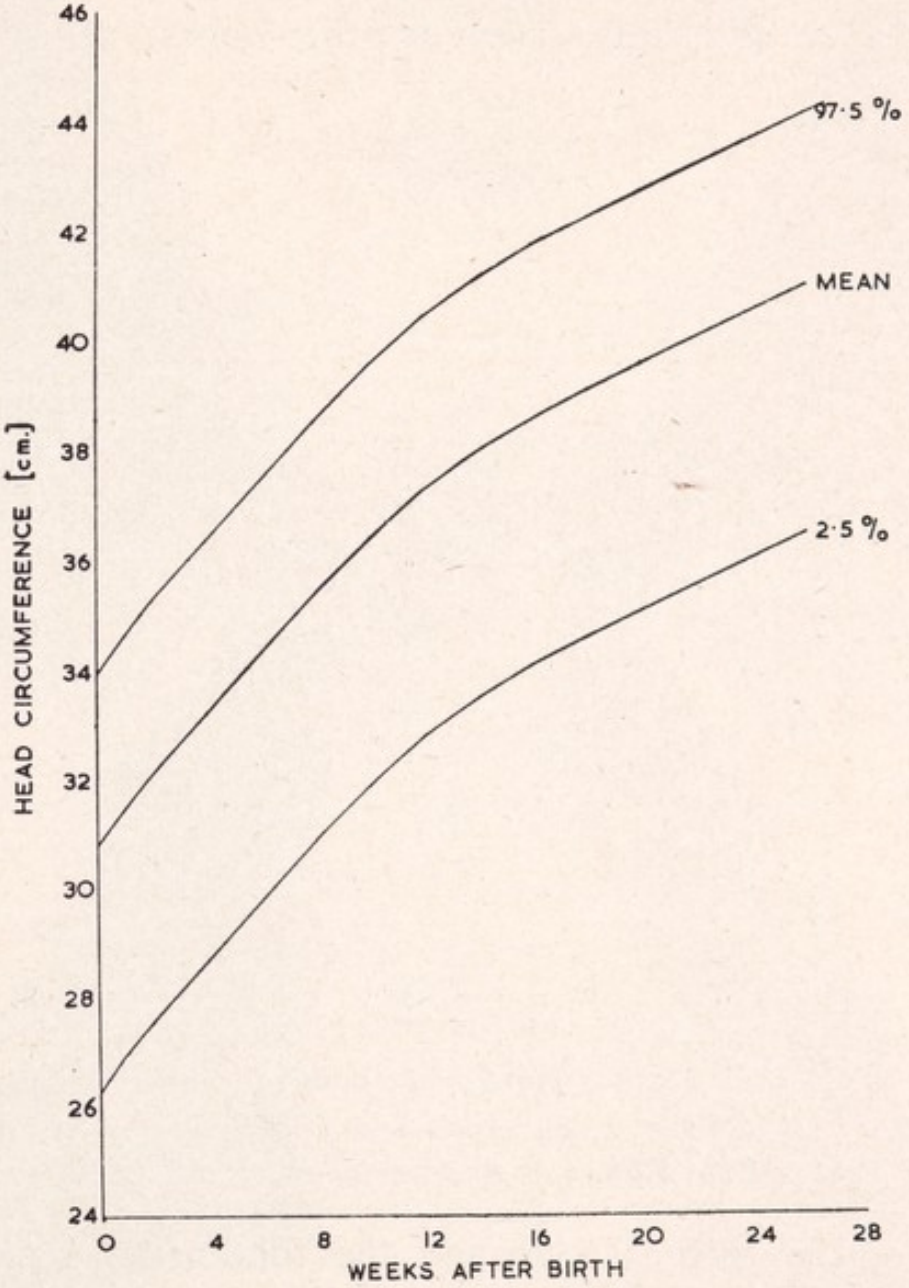


FIG. 15 (*b*). Head circumference/age chart for premature babies.

If serial measurements are made the rate of growth is found to be normal, and the anterior fontanelle frequently closes rather earlier than usual.

(2) The head in premature babies is large in proportion to the body and poorly ossified. In very small infants, weighing 2 lb. or less, this disparity may be so great as to

suggest hydrocephalus. Measurement, reference to normal standards and continued observation will settle any misgivings on this point.

(3) There are some congenital defects of ossification that involve the skull as well as other bones; the cranium

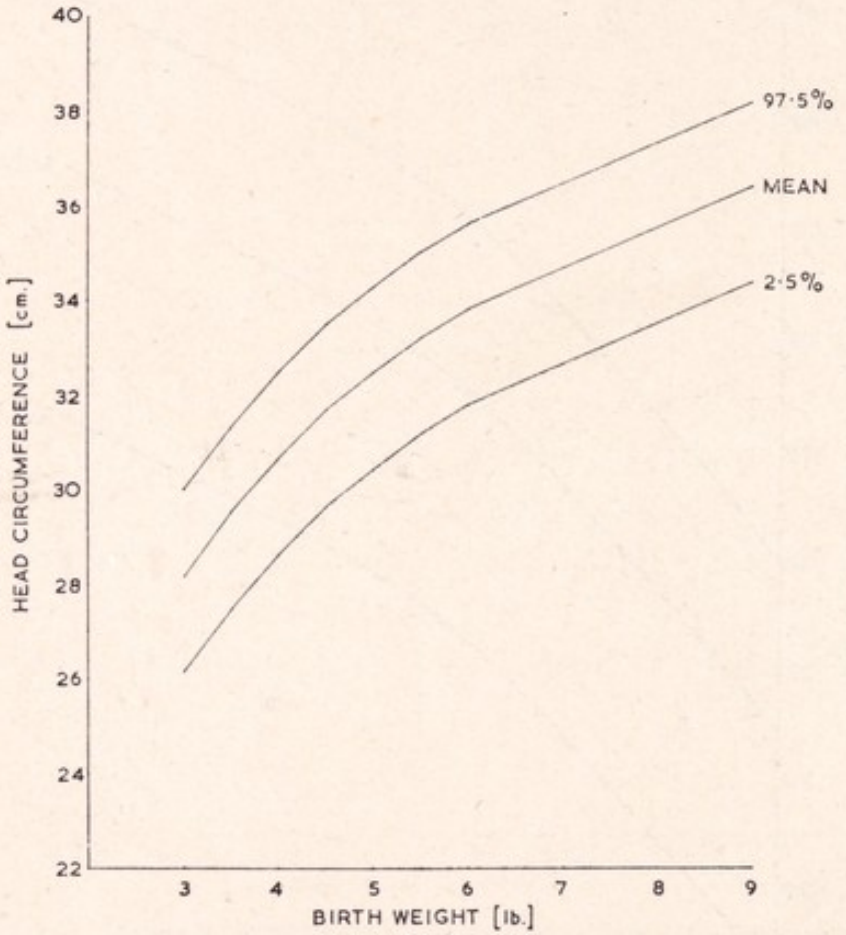


FIG. 15 (*c*). Head circumference relative to birth weight.
(*Figs.* 15 (*a*), (*b*) *and* (*c*) *are reproduced by courtesy of Dr. Eileen O'Neill and the Editors of Archives of Disease in Childhood.*)

is thin, the sutures wide and the fontanelles large. In achondroplasia the vault of the skull may be large, but the short arms and legs and small thorax should leave no diagnostic doubt. In osteogenesis imperfecta involving the skull, fractures of the long bones are usually present at birth and can be detected clinically and radiologically. A third condition, cleidocranial dysostosis, can be diagnosed readily provided it is borne in mind. The discovery of incomplete ossification in a skull of normal size should

prompt palpation of the clavicles. In cleidocranial dysostosis they are nearly always absent in whole or in part, and this can be confirmed radiologically (Fig. 16).

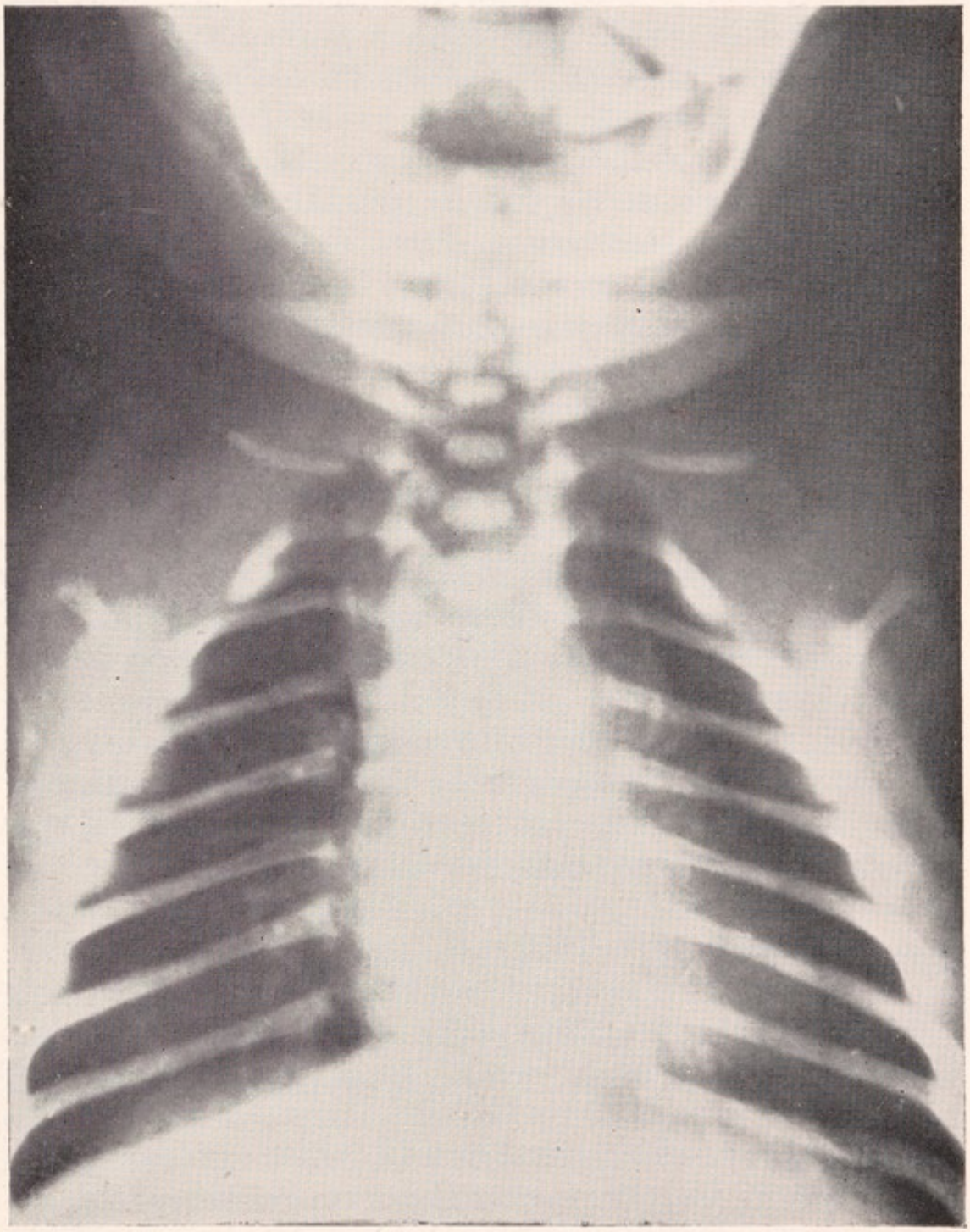


FIG. 16. Cleidocranial dysostosis. The clavicles are rudimentary.

The disorder is commonly present in one or other parent but sometimes arises as the result of a mutant gene.

(4) Hydranencephaly is a relatively rare malformation in

which the skull is well formed but the cerebral hemispheres fail to develop or, more probably, first develop and then degenerate. The space which should have been occupied by the cerebrum is filled with cerebrospinal fluid. The size of the head is usually greater than normal and increases with abnormal rapidity. Infants with this condition lack even the primitive reflexes shown by the live-born anencephalic and die within a few days. The diagnosis can readily be made during life by transillumination of the skull. The entire calvarium shines red, reminiscent of a transilluminated hydrocele. If the light is applied to the temporal region it may be seen shining through from the opposite side. This degree of translucency is not seen in hydrocephalus even when there is marked thinning of the cerebral cortex. Early diagnosis enables the clinician to give an accurate prognosis and prevents the raising of false therapeutic hopes.

(5) Macrocephaly is an extremely rare malformation in which the brain and skull are abnormally large at birth and grow abnormally fast. Mental deterioration is said to be inevitable.

(6) Congenital absence of the corpus callosum may be associated with enlargement of the head. Pneumoencephalograms show, in the anteroposterior view, a gap between the cerebral hemispheres.

Microcephaly

Microcephaly, an abnormally small head, is far less common than hydrocephalus. In a maternity unit of average size, with 1000 to 2000 deliveries annually, hydrocephalus will be seen several times a year but microcephaly only once in several years. The term microcephaly has led to much confusion. Literally translated, it means 'a small head' and is sometimes used in reference to any mentally retarded child or adult with a head circumference below the lower limits of normal for the age. Used in this sense the term has little meaning, for a small head is a

common finding in mental retardation of very varied aetiology. Most mental defectives have a head of normal size at birth but because some perinatal or postnatal event has interrupted brain development, subsequent skull growth lags.

Microcephaly is also used, in a much narrower sense, to refer to a particular variety of genetically determined mental defect associated with a characteristic appearance of the head. At birth, the skull is noticeably small, because brain growth has been retarded in utero, and the anterior fontanelle is abnormally small or closed. The forehead is low and receding, and the skin of the scalp may be wrinkled like a puppy's neck. This condition, which is apparently due to a rare recessive gene, is sometimes distinguished by the name of true microcephaly (microcephalia vera) and is rare in comparison with that affecting the large, heterogeneous group of small-headed mental defectives.

If the size of the head is abnormally small at birth it is not necessarily a genetically determined microcephaly. Maternal infection with rubella, or toxoplasmosis, or heavy doses of ionizing radiation, in the first trimester of pregnancy, may interfere with the growth of the brain. If the head is small, it is therefore always worth trying to determine the cause of the abnormality by taking a careful history and carrying out serological tests for toxoplasmosis on mother and infant. If these steps are not taken, pessimistic genetic advice may be given unjustifiably.

Recording head circumference

It is appropriate at this point to stress the importance of recording the head circumference at birth in all babies, whether it appears to be abnormal or not. This record serves as a baseline for comparison should subsequent growth of the skull appear to be abnormal. In addition to its obvious diagnostic importance, such a record may have medico-legal value. It has been known for the

parents of a microcephalic idiot to threaten proceedings against the obstetrician, the implication being that the child suffered brain injury at the time of birth. In such circumstances, if it can be shown that the infant's head circumference was abnormal at birth, it is clearly established that the brain was abnormal before the end of pregnancy.

Craniostenosis

In craniostenosis, or craniosynostosis, one or more cranial sutures closes prematurely. Although the abnormal sutures must fuse before or very soon after birth, the consequent abnormal shape of the skull is not usually noticeable for a few weeks or months. The seriousness of the disorder depends upon whether the skull can grow fast enough to contain the expanding brain. Generally speaking, if only one suture is involved the calvarium will be abnormal in shape but adequate in size; closure of the sagittal suture results in a long, thin head (scaphocephaly), and of the coronal suture in a broad, flat-backed head (brachycephaly). If all the major sutures close prematurely, adequate skull growth is impossible. The frontal region is expanded so that the head is turret-like (oxycephaly or acrocephaly), and the orbital plates of the sphenoid bone push the eyeballs forward. The optic nerves are stretched and, without treatment, this will lead to blindness. At the same time, increasing intracranial pressure interferes with normal growth of the brain. In this group of infants, early diagnosis is of the utmost importance; operative treatment, repeated if necessary, will permit expansion of the skull and hence normal brain growth, thus preventing the tragic late *sequelae* that are otherwise inevitable.

A number of infants with turret skull also have fusion of fingers or toes, the combination being called acrocephalosyndactyly. This is a rarity, but when an infant is noted to have syndactyly (by no means rare), the cranial sutures should be examined by palpation, and radiographs taken in cases of doubt.

Craniosynostosis can be demonstrated radiologically but is shown better by special positioning than by the standard views of the skull. It is, therefore, always wise to discuss this possibility with the radiologist or at least to indicate clearly to him on the request form that craniostenosis is being considered, stating which suture or sutures are thought to be involved.

If uncertainty regarding the diagnosis remains, the infant should be reviewed fairly frequently. In addition to serial measurements of the head circumference, measurement of the anteroposterior and biparietal diameters with calipers may be informative.

An anterior fontanelle bone occupying the anterior fontanelle is a great rarity and may lead to a false diagnosis of craniostenosis.

Soft tissue swellings on the head rarely give rise to diagnostic difficulty. The large swellings, including caput succedaneum, cephalhaematoma, subaponeurotic haematoma, occipital meningocele, and meningo-encephalocele, are all sufficiently characteristic to be recognized on sight. However, small swellings may cause diagnostic difficulty. Cranial meningocele and meningo-encephalocele (to which the collective name cranium bifidum cysticum is sometimes given) are usually large and occipital, although they may be small, and occur anywhere in the midline of the calvarium. Being in the midline they may be mistaken for dermoid cysts, but the reducible nature of the swelling distinguishes them. Very rarely they are not midline.

There is an interesting, small soft-tissue swelling, sometimes seen near the posterior fontanelle, which may be mistaken for an encephalocele or meningocele. This is an irreducible, fluctuant swelling in the line of the lambdoid suture, often close to the midline but never exactly median. It either disappears rapidly or hardens and then resolves more slowly. If the swelling is aspirated, a procedure that has nothing to commend it, a small amount of blood is removed, and the swelling disappears. The evidence suggests that these swellings are minute cephalhaematomata,

presumably related to Wormian bones. They need to be distinguished from encephaloceles because no treatment is required and the prognosis is excellent.

Congenital skin defect

The only congenital abnormality of the skin of the scalp that may, when first seen, puzzle the clinician is the so-called congenital skin defect. This takes the form of a round or slightly oval area of granulation tissue, usually in the midline over the vertex and about half an inch across (Fig. 17). The cause of the lesion is unknown, although the naked-eye appearance suggests pressure necrosis. It heals spontaneously leaving a small, bald area.

The white hair of albinism may be closely mimicked in babies born with very fair hair, but examination of the eyes immediately settles the diagnosis (*see* p. 60).

EXAMINATION OF THE FACE

Apart from revealing obvious abnormalities, such as cleft lip, inspection of the faces of newborn babies gives the lie to the belief that all infants look alike.

Mothers may require, and should be given, reassurance about the dilated blood vessels so often seen on the upper eyelids and the centre of the forehead. This type of naevus (naevus flammeus), like the naevi almost universally present on the nape of the neck of newborn babies, clears up spontaneously and in this way differs from the port-wine stain most commonly seen on the cheek or neck.

It is perhaps surprising that so many conditions that are later recognizable by the characteristic facies are scarcely ever diagnosed in the neonatal period. Amongst such conditions are craniofacial dysostosis (Crouzon) and mandibulofacial dysostosis (Treacher-Collins). Similarly, in gargoylism (Hunter-Hurler syndrome) the facial appear-

ance in an affected infant 6 months old is typical, but the condition is rarely diagnosed with certainty in the neonatal period. Two important facies are, however, always apparent at the moment of delivery: Potter facies and the facial appearance in mongolism.

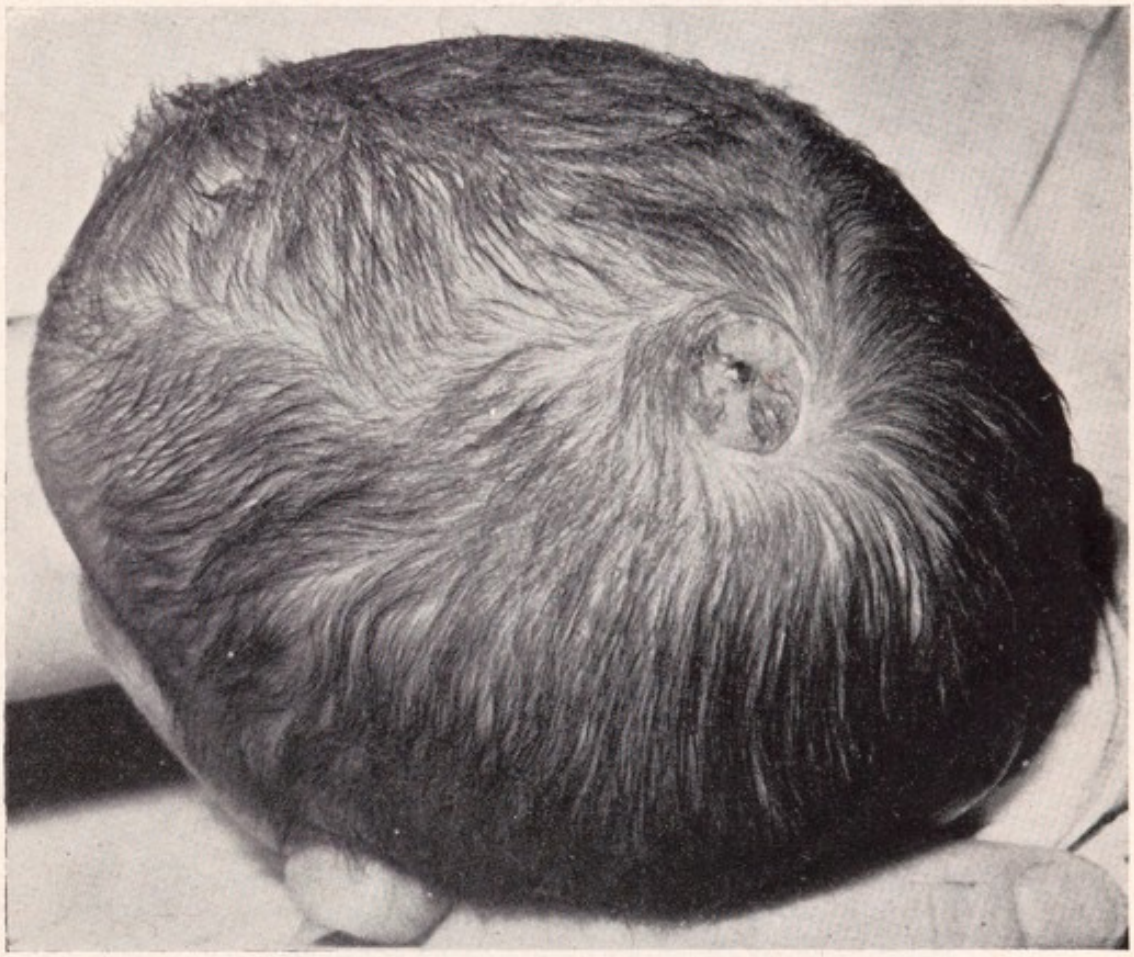


FIG. 17. Congenital skin defect.

Potter facies

Potter facies (Figs. 18 (*a*) and (*b*)), usually associated with renal agenesis, was described by Potter (1961) thus: 'The most constant facial characteristic is a very prominent epicanthic fold that forms a semicircle arising on the forehead, swinging down to cover the medial palpebral commissures and ending on the cheek. It is different from the fold in mongolism, which ordinarily ends at the level of the commissure. The nose is usually flattened, the

depression below the lower lip is unusually prominent and the ears, which ordinarily contain little cartilage, are flat without a formed helix and often are set at an angle exceptionally low on the sides of the head.' The flat nose

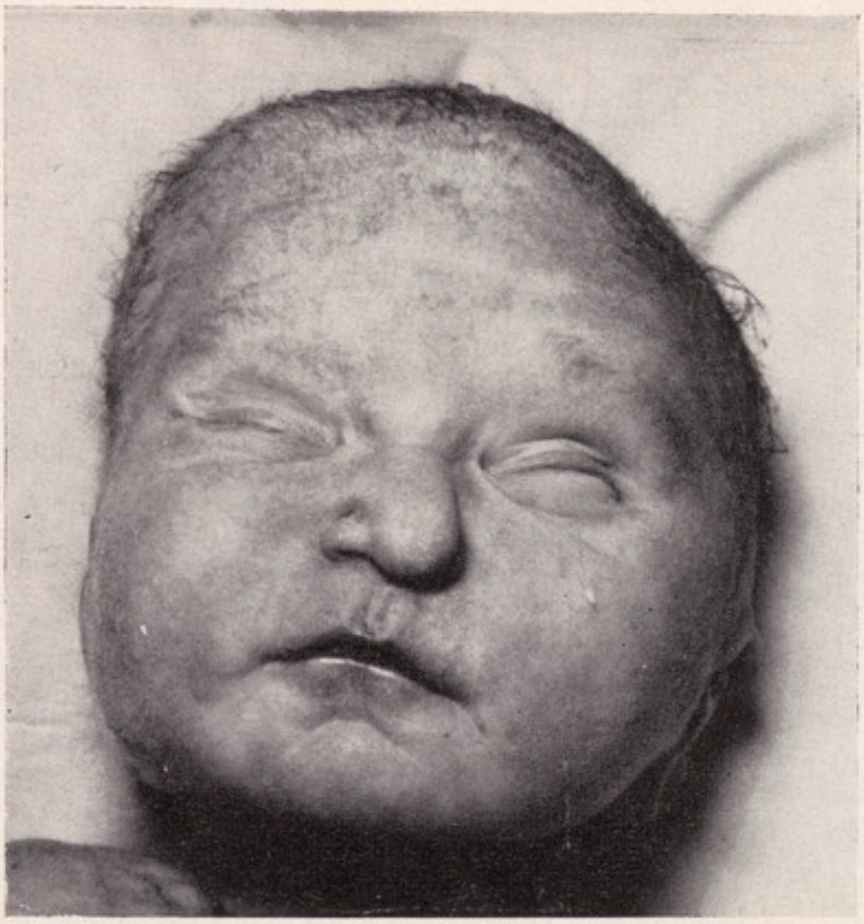


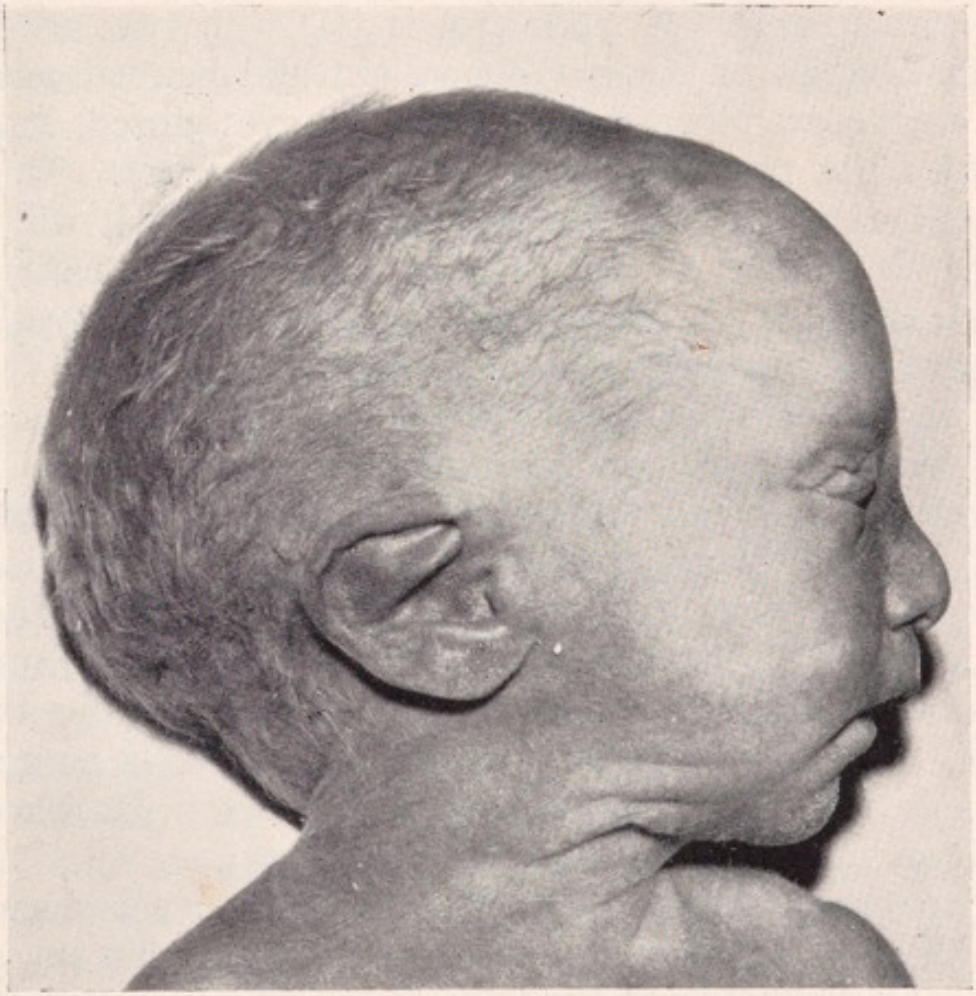
FIG. 18 (*a*). Potter Facies. Note flattened nose and epicanthic folds.

has been attributed to pressure consequent upon oligohydramnios, but the author has seen typical Potter facies, including the flat nose, in a baby with polycystic kidneys whose mother had polyhydramnios.

Mongolism

The diagnosis of mongolism (Down's syndrome) is not, as a rule, in doubt. Only two matters will be discussed here: the diagnosis of the doubtful case, and the significance of chromosome studies in relation to genetic prognosis. There is no single physical characteristic that is diagnostic

of mongolism. Apart from the trisomic state of chromosome 21, almost without exception each feature that has been described in mongols may be found in healthy individuals. Clinical diagnosis usually rests on the co-existence in one infant of several of these features. The most characteristic, apart from the facial appearance, are



(*b*). Potter Facies. Note large, low-set ears.

the flat occiput, the squared-off helix of the ear, the loose skin at the back of the neck and the squat palms. It sometimes happens that a newborn baby shows some of these characteristics, yet there is doubt or a difference of opinion about the diagnosis. The appearance of the parents has to be taken into consideration; the normal child of oriental or mixed parentage does not really look like a mongol, but the inexperienced eye may be deceived. Not uncommonly, when there is suspicion or doubt at

birth, re-examination three or four days later clears up all indecision.

If at re-examination doubt still remains, it can sometimes be resolved by a simple clinical test. Although any single one of the visible features of mongolism may be found in normal babies, muscular hypotonia is usually very striking in the mongol baby from birth and is not found in healthy full-term infants. If a mongol is put with two or three normal babies of comparable weight and handed one by one to the examiner, he can, with his eyes closed, readily identify the mongol by its extreme floppiness. This is sometimes so striking that the baby feels as if it will slip from his hands. If the suspect baby cannot easily be distinguished from the normal controls by this test, the diagnosis of mongolism should remain in doubt.

If facilities for chromosome studies are available, it is not necessary to rely upon clinical judgement. The majority of mongols (almost all mongols born to mothers over 35 years of age) possess an extra small chromosome (number 21), and it is a relatively simple matter to demonstrate that the nuclei of somatic cells (for example, leucocytes) contain 47 chromosomes. If, on the other hand, the diploid number is shown to be 46, a complete analysis of the karyotype is necessary to determine whether the individual chromosomes appear normal.

Cytogenetic studies will not only help to establish the diagnosis in doubtful cases of mongolism, but by distinguishing between two groups of parents, they also serve as a basis for counselling about the prognosis for future children. One group of parents has only a random chance of having another mongol; the other, much smaller, group has a very much greater chance. Thus the early diagnosis of mongolism alone is not always sufficient. For genetic counselling it may be equally important to diagnose early the *type* of mongolism, that is to say the chromosomal constitution of the mongol. This has the same practical value as establishing the aetiology in a case of microcephaly.

In order to clarify the difference between these two groups of mongols in relation to prognosis, the genetic situation is explained in a little more detail on page 174. Apart from the diagnosis of mongolism itself, its association with congenital heart disease and duodenal stenosis should be constantly borne in mind. These conditions are discussed elsewhere; and it is sufficient to say here that the significance of cyanosis or bilious vomiting should be appreciated even more rapidly in a mongol than in other infants.

EXAMINATION OF THE EYES

Examination of the eyes of the newborn baby is important and rather apt to be neglected. Retinoscopy cannot adequately be carried out without dilating the pupils, and opinions differ as to whether it is advisable as a routine procedure. In most parts of the world the doctors are so hopelessly outnumbered by the babies that routine retinoscopy is out of the question. Inspection of the anterior structures of the eye, on the other hand, is extremely simple and should never be omitted. A good source of light, preferably natural daylight, is all that is required. The eyes can be seen most satisfactorily when the baby is awake and content. Any attempt to force the eyelids apart, whether the infant be crying or peacefully asleep, is likely to stimulate strong contraction of the orbicularis oculi muscles so that examination becomes impossible.

Two points about congenital ocular abnormalities in general are worth noting. First, they are often hereditary, and a history of congenital eye disease in either parent should prompt a particularly careful scrutiny of the infant's eyes. Frequently, blind people (and deaf people, too) find their marriage partners amongst members of a club or society composed of people with similar disabilities. So it is that two blind people may marry, and yet the causes of their respective blindness may, in fact, be quite different. Unfortunately the paediatrician is not always as well

informed about the parents as might be desired; a cataract or defect of the iris in the mother may only come to light after the same abnormality has been discovered in the baby.

The second general principle is that an eye defect is often only part of a more widespread syndrome. Its discovery should therefore lead to a particularly careful examination of the rest of the baby, and especially of the central nervous system, the heart, and structures derived from the first branchial arch (the ear and mandible). Any or all of these may be involved in infection of the embryo by rubella virus or *Toxoplasma gondii*, in some autosomal trisomic syndromes, and in the varied manifestations of the first arch syndrome (McKenzie, 1958, *see* p. 65).

When examining the eye, the following points should be noted:

(1) The shape, size, and position of the eyeball.
(2) Abnormalities of the conjunctiva, cornea, iris, and lens.
(3) Abnormalities of the retina (if an ophthalmoscope is used).

An abnormally small eye (microphthalmos) can be recognized immediately; this may be the result of toxoplasmosis. In some instances not only the eyeball but one entire side of the face appears smaller then the other.

Of much greater clinical importance is an abnormally large eye, because the cause may be *congenital glaucoma*. This condition, also called buphthalmos, is usually determined by an autosomal recesssive gene. The parents are unaffected, but previous siblings may have suffered from the same condition. In about half the affected children diagnosis can be made at birth (Barkan and Ferguson, 1958), the characteristic signs being both cloudiness and enlargement of the cornea. As long as the condition is constantly borne in mind and the diameter of the cornea is noted, it is unlikely that a case will be overlooked. Actual measurement is only necessary to confirm a clinical impression. The normal cornea in a full-term newborn

infant is not more than 11 mm., or 12 mm. at most, in diameter. In borderline cases, and when the clinical diagnosis seems certain, an ophthalmologist should be consulted so that the diagnosis may be confirmed by measurement of the intra-ocular tension (tonometry). The importance of early diagnosis in congenital glaucoma lies in the fact that the prognosis for vision depends upon the stage at which treatment is started in the disease. Unfortunately, those infants in whom the condition is recognizable at birth have a worse outlook than those in whom signs are first detectable at a few months of age. Nevertheless, their best hope lies in detection in the neonatal period.

In approximately half the infants affected, congenital glaucoma is unilateral; bilateral cases may be confused with conditions that cause prominence of the eyes. *Exophthalmos* is sometimes seen in infants born to thyrotoxic mothers, and to those whose thyrotoxicosis has been treated by antithyroid drugs or thyroidectomy. This is presumably the result of placentally transferred maternal thyrotropic hormone. Some of these infants also show evidence of thyrotoxicosis, but in all but a very few instances the condition is temporary and the eyes soon become normal. Again, prominence of the eyes is sometimes seen in the offspring of parents with high degrees of myopia, and these infants may well prove to be myopic too. In craniostenosis involving all the sutures, proptosis develops but is not usually apparent at birth (*see* p. 50).

Abnormalities of conjunctiva, cornea, iris, and lens

The sclera in the newborn baby is often slightly blue or grey. Rarely, it is strikingly blue and brings to mind the possibility that the infant is suffering from osteogenesis imperfecta.

Conjunctival dermoids are white areas, usually small and very slightly raised; they may occur anywhere on the scleral conjunctiva and may overlap the edge of the cornea.

Unless they overlap a large part of the cornea, they are not of great significance. There may, however, be associated anomalies. In the case illustrated by Potter (1961), and in another the author has seen recently, there were several skin tags along a line between the ear and the angle of the mouth on the side of the dermoids. In the case the author saw, there was also a congenital heart lesion from which the baby died when a few days old. This illustrates well the association between eyes, heart, and first arch to which reference has already been made (*see* p. 58).

The cornea should be clear. Haziness in congenital glaucoma has already been mentioned. Cloudiness and corneal opacities are also seen in the Hunter-Hurler syndrome (gargoylism) but these are not as a rule observed at birth.

Lens opacities (Fig. 19) may be overlooked at first glance if the light is not shining obliquely into the eye. Although expert opinions are not united about the best age at which to operate upon congenital cataracts, early diagnosis will at least permit early treatment to be considered. In some cases the cataracts are hereditary; in others they follow maternal rubella or infection by *Toxoplasma gondii*. They have been described as sequelae of maternal tetany during pregnancy and following neonatal tetany. Cataracts are a feature of untreated galactosaemia (*see* p. 111), but in this disorder they are not present at birth and should therefore be preventable.

The iris of newborn Caucasian babies usually contains little pigment and is commonly blue. In the negro infant the irides are very dark from birth, and this is occasionally true of infants of other races. The absence of pigment in the albino gives the iris an unmistakable pink shimmer. The examiner feels he cannot focus his own eyes properly for he is endeavouring to look at an iris which is in effect invisible.

The most common and most striking congenital abnormality of the iris is *coloboma* (Fig. 20). This is a segmental deficiency of the iris, usually of the inferior part,

which results in a pear-shaped pupil. The condition may be unilateral or bilateral. Often it is associated with coloboma of the corresponding segment of the

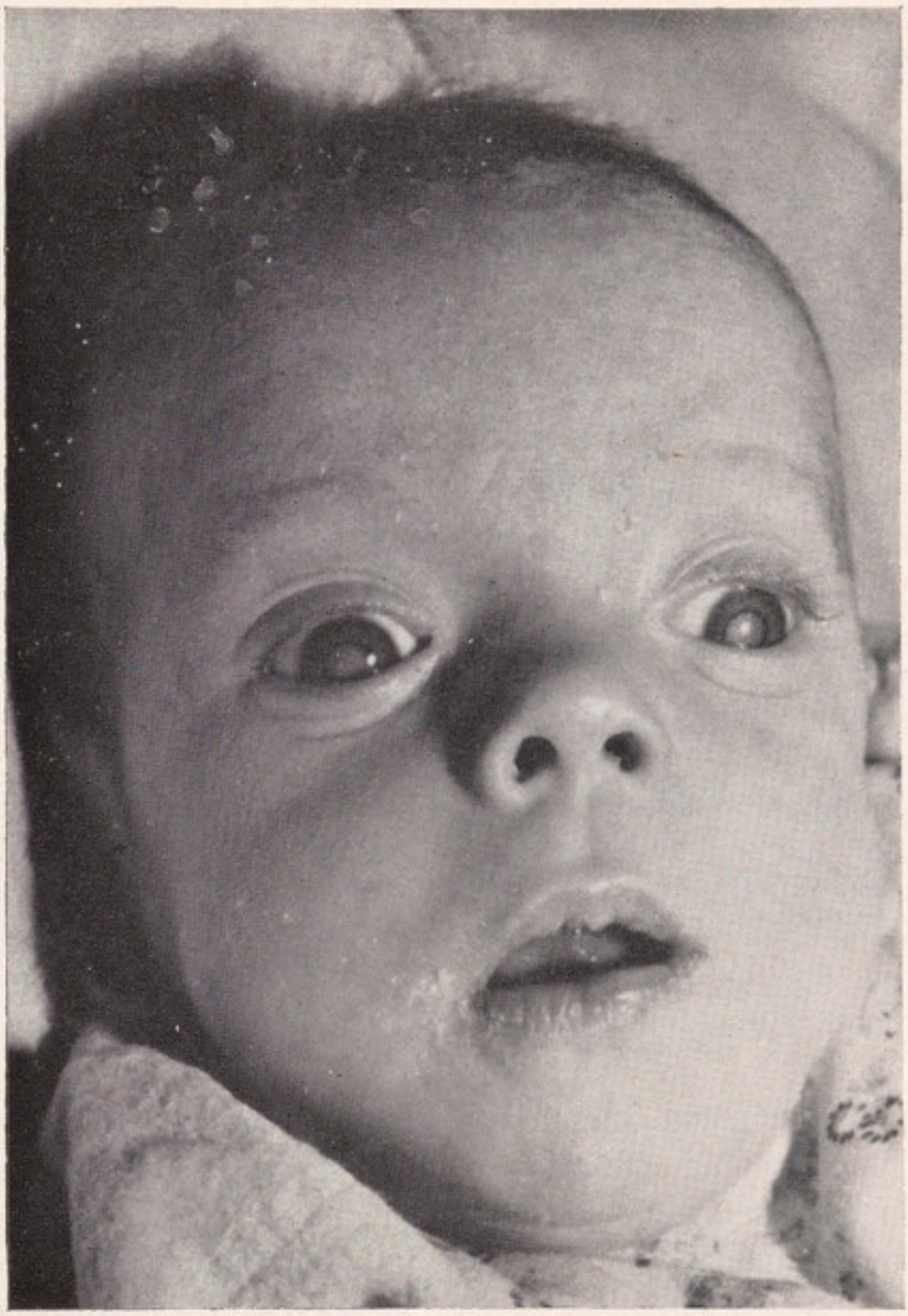


FIG. 19. Congenital cataract. This baby also had bilateral choanal atresia and is breathing through the mouth.

choroid and retina. This can be recognized only with an ophthalmoscope and appears as a pale wedge in the red retina, narrowing towards the optic disc. A corresponding defect in the lens may also be present but is less common.

Coloboma, although giving a curious appearance to the eye, has little or no effect on vision provided the lens is normal. But once again there is a tendency for other and more serious malformations to occur in the same infant, the heart and central nervous system being most often involved. Angelman (1961) described colobomata in four children, of

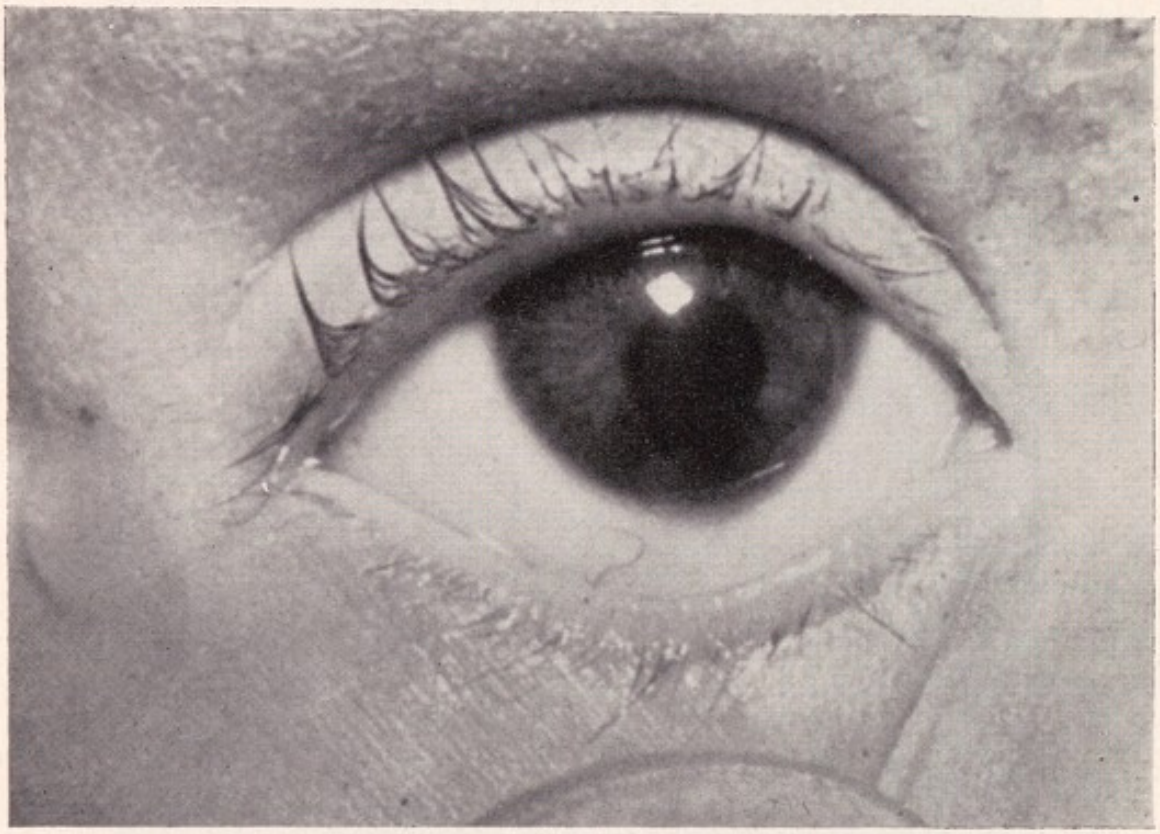


FIG. 20. Coloboma of the iris. There was an associated coloboma of the retina. The cause in this infant was thalidomide.

whom three were mentally defective and had congenital heart disease; in addition, one child had hydronephrosis, one had abnormal vertebrae, while the third had a cleft palate and the fourth a meningo-encephalocele. Nevertheless, coloboma does occur as an isolated abnormality and, if an affected infant appears otherwise healthy, it is improbable that any further disorder will become apparent later.

Examination of the irides of a mongol infant often reveals a ring of whitish dots (Brushfield spots) or a continuous white circle surrounding the pupil, rather nearer

the sclera than the pupil. These spots become more obvious when the iris darkens. They are sometimes seen in normal infants and children and are not present in every mongol. Too great a diagnostic importance should not be attached to them.

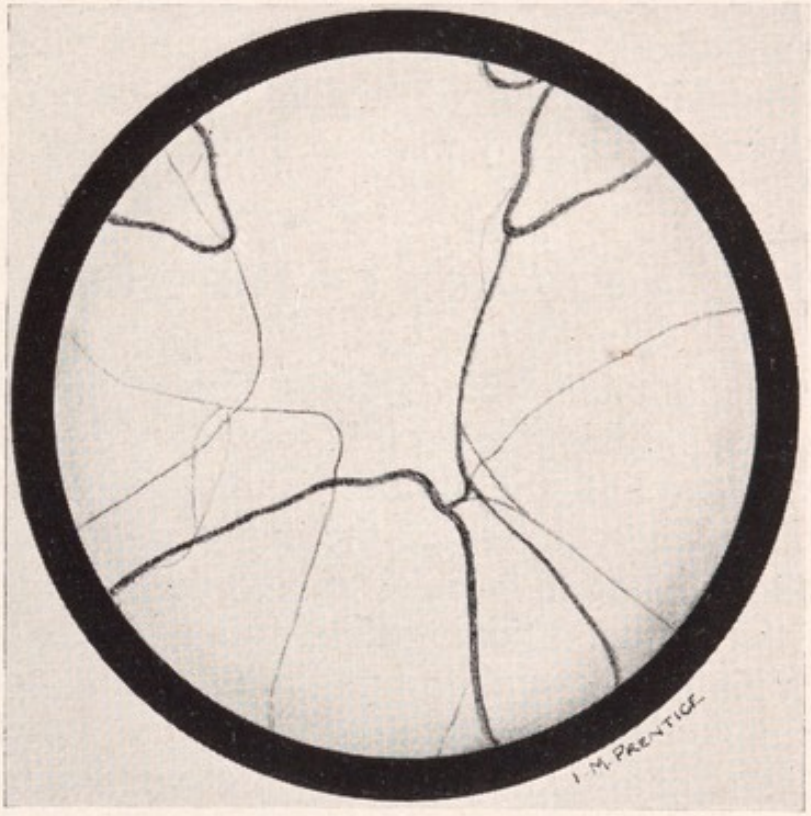


FIG. 21. Pupillary membrane in a premature infant, 10 days old, as seen with a +20 lens. (*By courtesy of Dr. Bruno Gans and the Editors of Archives of Disease in Childhood.*)

Abnormalities of the retina

Before the eyes are examined with an ophthalmoscope, the pupils should be dilated with homatropine. In premature babies, it is normal to see remains of the pupillary membrane as fine filaments festooned around the periphery of the pupil (Fig. 21). The appearance of coloboma of the retina and choroid has already been described. *Chorioretinitis* appears as irregular patchy white areas with pigmentation around them. It may be seen in congenital

toxoplasmosis, congenital syphilis and congenital tuberculosis and after transplacental virus infections of the foetus, such as herpes simplex and cytomegalic inclusion disease. In any of these conditions the uveal tract may be normal at birth, but the lesions of chorioretinitis may appear later, if the infant survives. In the same way, the characteristic cherry-red spot at the macular region in Tay-Sachs disease (familial amaurotic idiocy) is not present at birth, and it is not possible with presently available techniques to exclude this condition in the early weeks of life.

EXAMINATION OF THE EARS

Abnormalities of the pinna: Inspection of the ear drums of apparently healthy newborn babies is not necessary, but careful scrutiny of the pinnae is always worth while. Variations in the patterns of the convolutions are almost infinite and are said to be as characteristic of the individual as are finger-prints. Nevertheless, it is a simple matter to identify a pinna that is abnormal in shape, size, position or configuration. The presence of accessory auricles (pre-auricular skin tags) is common, especially in negro infants, and they are without special significance.

Abnormalities of the pinna are often associated with internal malformations, especially of the renal tract. The classic example of this association is the Potter facies of renal agenesis. In this instance, the large, low-set, poorly cartilaginized ears are but a single feature of a characteristic facial appearance (Fig. 18 (*b*)) that may occur in association with other renal abnormalities, such as horseshoe kidney. In some examples of unilateral renal agenesis, the ipsilateral pinna is deformed in this way, while the opposite ear appears absolutely normal.

A more general association between auricular and renal abnormalities (including double ureters, cystic kidneys, and congenital hydronephrosis) was pointed out by Hilson (1957). He found that in many of these cases relatives were

known to have similar malformations of the ears and the renal tract. As in renal agenesis, unilateral deformity of an ear is often associated with abnormality of the kidney or ureter on the same side. The link between the ears and the kidneys lies in the fact that they reach a critical stage of development at the same time (the seventh week of embryonic life). The occurrence of unilateral lesions leaves room for speculation, especially as families with several affected members may 'specialize' in one side or the other.

Malformations of the pinna are a common feature of several syndromes grouped together by McKenzie (1958) under the name of the *first arch syndrome*. This group includes the Treacher-Collins syndrome (mandibulofacial dysostosis), the Pierre-Robin syndrome (micrognathia, cleft palate, and glossoptosis), isolated micrognathia, and congenital deaf-mutism. The pinnae of mongols often show an excessive down-turning of the upper border, giving a squared-off appearance. Deformed pinnae have also been described in association with other autosomal trisomies.

Microtia is gross hypoplasia or aplasia of the pinna associated with a blind or absent external auditory meatus. The lesion may be unilateral or bilateral, and there may be a facial palsy on the affected side. This lesion became more common in some parts of the world from 1959 through 1961, and is attributed in many instances to mothers having taken the hypnotic drug, thalidomide, early in pregnancy. In spite of the complete absence of an external auditory meatus, the middle ear in some affected infants is normally formed. An otologist will advise on the nature and timing of surgical measures to improve hearing.

The discovery of malformation of the ear or ears in an otherwise healthy newborn infant raises two practical problems: Should the renal tract be investigated? Should the baby be followed up for hearing tests? To answer the last first, if there is no family history of deafness, hearing tests are probably not necessary. The majority of infants with malformed ears hear normally, and most of those born

deaf have normal auricles. As to the first question, renal-tract investigation means estimation of the blood urea and contrast pyelography, at the very least. The level of the blood urea will be elevated only if there is serious bilateral renal disease. Pyelography is far more informative, but there are technical difficulties in doing this in the newborn infant; furthermore, the radiographs are not always of the quality one would desire. In general, it is probably wise to postpone renal tract investigation beyond the neonatal period, unless a medical crisis arises, and to consider it later in the following circumstances:

(1) If there are other malformations, particularly if they involve the genitalia.

(2) If there is a family history of renal disorder.

(3) If the malformation of the pinna is gross.

(4) If urinary infection supervenes. In this connexion, should an older infant or child present with infection of the urinary tract, the finding of any auricular malformation should be regarded as an added indication for pyelography.

Congenital deafness is only exceptionally associated with deformed pinnae, and early suspicion that a child may prove to be deaf normally rests on the history. In approximately 25 per cent. of cases the deafness is of genetic origin, one parent, or sometimes both, being deaf. Perinatal damage to the nervous system, principally by asphyxia and jaundice, accounts for a further 20 per cent., while maternal rubella may account for another 10 per cent. Occasionally, cases are attributable to drugs given to the mother during pregnancy (quinine, or possibly streptomycin) or are associated with familial goitrous cretinism. However, in almost half the cases of congenital deafness the cause is poorly understood.

Hearing tests in the neonatal period are at present only reliable in the hands of a few experts and for general purposes testing must usually be deferred until the age of 4–6 months. It is therefore essential to ensure that infants known to be at risk in this respect should be subjected to

simple screening tests at this age and referred for more detailed examination if they do not respond normally. Detection of deafness before the age of 9 months permits the fitting of a suitable hearing-aid before speech is established, but no deaf infant is too young for an aid and the sooner the diagnosis is made, the better. The sooner the child receives auditory communication the more he will learn. Suitable tests for screening have been described by Sheridan (1958) and Ewing and Ewing (1954) and are described in Appendix 2.

Infants in any of the following categories should be considered at risk and must be tested for deafness:

(1) Those with an immediate family history of congenital deafness (that is, with an affected parent or sibling).

(2) Those with a history of severe perinatal asphyxia or intracranial birth injury.

(3) Those with a history of severe neonatal jaundice, whether haemolytic or 'physiological' (that is, with a maximum unconjugated bilirubin level of 20–25 mg. per cent. or more).

(4) Premature infants weighing 2000G. (4½ lb.) or less at birth.

(5) Infants whose mothers had rubella in the first three months of pregnancy.

It is worth repeating that almost half the children born deaf will not fall into any of these categories, and any sign of imperfect hearing in an older infant (whether first recognized by parents, friends, or neighbours, or detected at a medical examination) should never be ignored on the grounds of a negative history.

It is evident that infants in all these categories, except the first, are also at risk in respect of cerebral palsy, mental defect, or both, and require full neurological and developmental assessment periodically as well as hearing tests. If frequent review is impracticable, the best age for follow-up examination is around 6 months for full-term infants and proportionately later for prematures.

EXAMINATION OF THE BACK

Congenital abnormalities of the spine are usually apparent at birth and the purpose of this section is only to discuss differential diagnosis and to call attention to associated abnormalities which might be overlooked.

Spina bifida cystica is a useful term including both meningocele and myelomeningocele. When it occurs in the cervical region it is usually a simple meningocele, but unfortunately the anomaly is far more common in the lumbar region, and in this situation the lesion is usually a myelomeningocele. A meningocele is always covered by normal skin (McNab, 1958) whereas a myelomeningocele is usually covered by granulation tissue. Transillumination of a skin-covered swelling may show whether or not it contains nerve tissue, but more is to be learned from a functional examination of the lower limbs and the anal and vesical sphincters.

Motor function in legs

Motor function in the legs is best assessed by observing voluntary movements when the child is awake. Sometimes stimulation of the raw area on the back, by accident or design, results in brief contraction of groups of muscles in the legs and these must not be mistaken for voluntary movements. When paralysis is severe there is muscle wasting, especially below the knee; bilateral club-foot is common. The lower abdominal muscles may also bulge if they are paralysed. Sensation can be tested crudely by pin-prick. If it is normal, the pricked limb will be withdrawn unless it is paralysed, in which case the infant's cries may give a clear indication that sensation is intact.

Anal and vesical sphincters

The function of the anal and vesical sphincters can also be assessed in spite of the fact that the normal infant is

doubly incontinent. If the anal sphincter is paralysed, the normal natal cleft between the gluteal folds is absent, the anus lying at the apex of a smooth eminence. Small amounts of meconium or faeces escape whenever the intra-abdominal pressure is raised and the rectal mucous membrane may prolapse. Similarly the uterus may prolapse if the muscles of the pelvic floor are very weak. Stimulation of the perianal skin normally causes contraction of the anal sphincter. Absence of this reflex indicates a defect of sensation or muscle power or both.

Paralysis of the bladder sphincter is evidenced by continual dribbling of small quantities of urine. It is often possible to express urine by suprapubic pressure on the bladder.

Myelomeningocele

The association of hydrocephalus with myelomeningocele is well known. Even if the head appears normal at birth, a record of the head circumference will be of the utmost value subsequently in assessing the rate of growth of the skull. Less widely appreciated is the frequent occurrence of hip dislocation when paralysis of the muscles around the joint is severe. Unlike the isolated 'congenital dislocation', which is a subluxation or joint instability (*see* p. 97), the femoral head is completely displaced on one or both sides. If the pelvis is fixed with one hand and the femur grasped with the other, the two can be moved independently. This is so striking that radiological confirmation is scarcely necessary.

The importance of assessing the extent of the neurological deficit and the presence of associated lesions in a newborn infant with myelomeningocele depends very much upon the policy adopted by the surgeon concerned. There is good evidence that the extent of the muscle paralysis may spread within literally hours of birth, and paediatric neurosurgeons in Great Britain are more and more in favour of immediate repair of the back. If the results of this approach

are to be judged satisfactorily, a very careful appraisal of the infant will be required at birth.

Spina bifida occulta

Spina bifida occulta may be suggested by the presence over the lumbar region of a tuft of dark hair, a pad of fat or a vascular naevus. Sometimes there is a very small scar of thin tissue crossed by dilated blood vessels. In some infants with these lesions, the absence of spinous processes on the lower lumbar vertebrae may be noted clinically, but only a radiograph will show the true condition of the underlying spine. In some infants with lumbar hair tufts, the spine is perfectly normal. Yet others have diastematomyelia, a rare condition in which a bony spur from a vertebral body passes through the spinal cord, dividing it into two halves. There are no symptoms at birth, but as the spine and the spinal cord grow at different rates as the years pass, weakness of the legs or sphincters may develop later.

Skin anomalies over the lower spine

Two anomalies of the skin over the lower spine need to be distinguished. The more common and less important condition is post-anal dimple or pilonidal sinus. This, as the name suggests, is situated behind the anus in the sacrococcygeal region and is a midline dimple. Some are so deep that the bottom of the pit cannot definitely be seen at birth, but review after a few weeks shows that it is indeed only a dimple.

A dermal sinus is a rare anomaly but is important because it communicates with the meninges and, if unrecognized and untreated, may lead to recurrent meningitis. It is a much smaller hole than a post-anal dimple and is therefore easily overlooked unless, as sometimes happens, there is a lumbar hair-tuft, lipoma, or other landmark, to attract attention. It is also situated higher up, usually in

the lumbar region. Communication with the spinal canal may be demonstrated by injection of radio-opaque dye, but if this is done it is important to choose a contrast medium which will normally be used for myelography. The condition requires urgent surgical treatment if meningitis is to be averted.

The neck

Sometimes an infant is born with a neck that appears short and wide. It may look wide because it *is* short or it may be wide and therefore *look* short. A wide (webbed) neck is a feature of the Bonnevie-Ullrich syndrome and is often associated with gonadal dysgenesis which is discussed on page 89. If the neck is short there is usually a malformation of the cervical spine. This may simply be an absent vertebra or hemivertebra, but commonly there is a more diffuse deformity and fusion of the cervical vertebrae with high scapulae and low occipital hair-line. This association of abnormalities is the Klippel-Feil syndrome. In some of these cases there is a history of polyhydramnios, and many of them have congenital heart lesions, especially ventricular septal defect (Nova, Cohen, and Maxwell, 1961). Nothing in the way of treatment can be offered for the neck, but the diagnosis of Klippel-Feil syndrome may lead to the recognition of a treatable cardiac defect.

EXAMINATION OF THE HEART

Important congenital abnormalities of the respiratory system will give rise to symptoms which are apparent at birth or become so within a few hours. This is equally true of many kinds of congenital heart lesion. These are discussed in the section on respiratory difficulties (p. 117). The importance of early diagnosis of cardiovascular abnormalities can scarcely be over-emphasized. As open heart surgery is developed, many complex heart lesions,

which are now fatal, will become amenable to operative treatment. There seems little doubt that, before long, the majority of congenital heart lesions will be treatable.

The differential diagnosis of congenital heart lesions is often a complex business necessitating cardiac catheterization, angiocardiography and other specialized techniques. In this section only the very broad principles are discussed and for further details the reader should consult a textbook of paediatric cardiology, such as that by Nadas (1957) or Keith, Rowe and Vlad (1958).

Congenital heart disease is usually manifest by murmurs, cyanosis, cardiac failure, tachypnoea, or a combination of these. If none of these signs is present at birth, diagnosis is likely to be delayed until a murmur develops; this may be in the neonatal period or later. If the delayed phenomenon of a murmur is overlooked, later cyanosis or cardiac failure may direct attention to a heart lesion.

Auscultation of the heart

Auscultation of the heart of a small infant can only be carried out satisfactorily when he is quiet and fairly still. If he is crying or very restless, it is quite wrong to suppose that a murmur will be heard 'if it is loud enough to mean anything'. It is true that if the heart is examined under ideal circumstances, many murmurs will be heard which mean nothing in terms of organic lesions, but the murmurs of congenital heart disease are not necessarily loud, especially in the newborn baby. The clinician's task is therefore to detect every murmur he possibly can, and then to decide whether further investigation or observation is necessary.

Because auscultation of the heart, like estimation of the fontanelle tension, needs to be done when the infant is quiet, it is often wise to do it very early in the examination. If the baby is asleep in a cot or incubator, this may be done before he is awakened and lifted out. If respiratory noises create difficulty in the sleeping baby, gentle and momentary

compression of the nose will interrupt breathing without waking the baby. The end-piece of the stethoscope should be warm; a sleeping baby is quickly roused by the application of cold metal to the chest. If the baby is crying, and examination of the heart cannot be postponed, a dummy (pacifier) will usually quieten him for a few moments, particularly in the first day or two of life. If an older infant is crying because he is due for a feed, peace can often be purchased only with food. The way to a hungry infant's heart is through his stomach.

Other evidence of heart disease

The discovery of a heart murmur in an apparently healthy newborn will prompt a search for other evidence of heart disease. Cardiac enlargement, cardiac failure, cyanosis, or the presence of other congenital abnormalities will all suggest that a murmur has an organic basis, but their absence in no way refutes a diagnosis of congenital heart disease. Not uncommonly, a systolic murmur is heard in the pulmonary area. Phonocardiography shows that this murmur often has the characteristics of the systolic murmur of patent ductus arteriosus (Burnard, 1958). The sooner after birth the examination is made, the more likely is such a murmur to be heard; it is more common in babies that have been asphyxiated during birth. It usually disappears within a few hours or days. Because of the changing haemodynamic situation in the neonatal period, murmurs may appear and disappear in a rather disconcerting fashion. Occasionally, the murmur of an organic lesion may disappear within a few days of birth and reappear later. Alternatively, no murmur may be audible at all until after the neonatal period. This is one of the more valuable purposes of the periodic routine examination of infants and school-children, because a heart malformation that is not recognized until cyanosis or failure supervenes may have passed the optimum time for repair or even have become inoperable.

Another phenomenon, unexplained until recently, is the permanent disappearance of a murmur which had been confidently attributed to a ventricular septal defect. It has been shown by cardiac cathererization before and after disappearance of the murmur that ventricular septal defects do rarely close spontaneously, probably as a result of muscle hypertrophy (Evans, Rowe, and Keith, 1960).

Palpation of femoral pulses

Palpation of the femoral pulses in the newborn is as important a part of routine examination as auscultation of the heart, because the absence of a murmur does not exclude the possibility of coarctation of the aorta. It may be less disturbing to feel first for the dorsalis pedis or posterior tibial pulses, but if these are not certainly palpable the femorals must be examined. The femoral pulses are not always easy to feel, particularly in plump or restless babies, but with patient persistence (and the help of a 'dummy' for restive infants) they are always palpable normally. They are best felt immediately below the inguinal skin crease, approximately at the junction of the inner and middle thirds of the crease. The hips should be semi-flexed. If they are fully extended the baby will often struggle and make the examination more difficult. In the presence of coarctation of the aorta the femoral pulses are, almost without exception, impalpable or very weak.

Familiarity with the normal strength of the femoral pulses will enable an abnormally forceful one to be recognized in some cases of patent ductus arteriosus. In these infants the femoral pulses seem to come up and hit the examiner's finger without waiting to be felt. In most of these babies, the posterior tibial and dorsalis pedis arterial pulses are also very easily palpable. Similarly, large volume pulses are found in the less common anomaly of persistent truncus arteriosus.

Chest radiograph and ECG

In assessing the significance of a heart murmur in an otherwise healthy infant useful information may be obtained from a chest radiograph and an electrocardiogram (ECG). These are commonly normal in newborn babies in whom a heart lesion has not yet caused any symptoms. The value of a radiograph depends very much upon the skill and experience of the radiologist and radiographer, for positioning and exposure need to be carefully controlled and interpretation is not easy. Even with a satisfactory film, considerable caution should be felt before declaring that the cardiac outline or lung vascularity is abnormal. The size and shape of the heart vary with systole and diastole, with inspiration and expiration, and with even minor degrees of rotation of the infant in relation to the X-ray plate.

The ECG is often normal in infants with congenital heart disease, but it is well worth doing because it may serve as a useful baseline for comparison with later records. Furthermore, in a few cases (for example, babies with primary myocardial disease or tricuspid atresia) the ECG may be of great diagnostic help.

Although the ECG only rarely makes a major contribution to the anatomical diagnosis of a heart lesion in the neonatal period, it may be extremely valuable in the diagnosis of cardiac arrhythmias. In congenital heart block the ventricular rate is below 70 per minute. Reference has been made earlier to the fact that prenatal recognition of this bradycardia may lead to an erroneous diagnosis of foetal distress (*see* p. 24). Congenital heart block is usually associated with a systolic murmur and sometimes with a diastolic murmur (Smithells and Outon, 1959), but it is exceptional for a structural lesion of the heart to be present. Supraventricular tachycardia may occur in the neonatal period and, if prolonged, is likely to lead to cardiac failure. The ECG will indicate the site of the ectopic pacemaker. Extrasystoles are sometimes heard in newborn babies and have the same significance as at any other age.

EXAMINATION OF THE MOUTH

The most important abnormalities of the alimentary tract are the obstructive lesions. The majority of these, however, cannot be diagnosed until symptoms develop, and are therefore discussed in Chapter 4.

Examination of the back of the throat in the newborn baby is extremely difficult and is not necessary in the symptomless infant. In contrast, the inside of the mouth should always be inspected because minor anomalies are common. This part of the routine examination can be left with advantage until the end, because if the infant starts crying at any stage the procedure is much simpler.

Teeth present at birth are almost invariably the lower central incisors. They present no diagnostic difficulty unless, as sometimes happens, they are covered by mucous membrane. Because of their extreme mobility they may then be difficult to distinguish from retention cysts. However, within a few days the teeth break through the mucous membrane and the diagnosis becomes apparent. Because of their mobility, these teeth rarely interfere with breast feeding.

Retention cysts of salivary and mucous glands in the floor of the mouth are quite common. A *ranula* is a sublingual salivary gland retention cyst. These cysts are usually small but are occasionally so large that early surgery is required. They tend to regress spontaneously.

Epithelial pearls are very small yellowish-white, calcareous nodules most commonly seen on the outer surface of the upper gum and on the hard palate near the midline. They are said to be retention cysts of mucous glands but are often surprisingly hard. They are of no pathological significance and should not be regarded as congenital abnormalities. They disappear spontaneously. Occasionally they are sufficiently large and conspicuous to be mistaken for teeth in the upper gum, but they are always well above the alveolar margin. Epithelial pearls on the palate should not be mistaken for thrush.

Congenital abnormalities of the tongue are rare. There is no doubt that most babies said to be tongue-tied are perfectly normal, but rarely the lingual fraenum is so short that the tip of the tongue cannot be protruded over the lower gum. This may lead to feeding difficulties and require early treatment, but it must be stressed that this is a rarity. Difficulty is more likely to arise with a breast-fed baby, who feeds with his mouth wide open, than with a bottle-fed infant.

Ankyloglossia is an uncommon anomaly in which there are bands of adhesions between the dorsum of the tongue and the roof of the mouth. The only case the author has seen also had ankyloblepharon (adhesions of the eyelids) and other abnormalities. *Macroglossia* may be caused by lymphangiectasis or glycogen-storage disease. It may also be the presenting symptom of congenital hypothyroidism.

EXAMINATION OF THE ABDOMEN

Some clues to the early diagnosis of congenital abnormalities of the gastrointestinal and urinary tracts have already been mentioned. Polyhydramnios may be associated with obstructive lesions of the upper bowel, and oligohydramnios with obstructive lesions of the urinary tract or renal agenesis. The significance of a single umbilical artery, of malformed ears, and of the Potter facies has also been discussed. There is also a close association between renal-tract abnormalities and developmental deficiency of the abdominal musculature. This is a rare condition, but the bulging abdomen and corrugated skin are unmistakable. The most usual internal anomaly to be found with abdominal-muscle hypoplasia is bilateral hydronephrosis, grossly dilated and tortuous ureters, and an enlarged bladder.

A further clue, which may be apparent on a radiograph of the abdomen, is defective development of the sacrum.

Sacral hypoplasia or agenesis, as an isolated defect or in association with more extensive spinal deformities, is often accompanied by developmental defects of the pelvic organs.

The importance of the early exclusion of oesophageal atresia has already been stressed and this condition is further discussed on p. 136.

Enlarged abdomen

Enlargement of the abdomen before birth may give rise to difficulties during the second stage of labour. It is not always due to developmental disorders but may be caused by hydrops foetalis, by foetal ascites, or by hepato-splenomegaly of infective or neoplastic origin. It is never the consequence of simple intestinal obstruction, in which the distension appears after birth. The most common developmental abnormality, causing abdominal swelling which is apparent at birth, is bilateral renal enlargement due to polycystic disease or hydronephrosis. In either case, enormous kidneys fill the whole of both loins and often approach each other in the midline, compressing the bowel into the centre of the abdomen. The X-ray appearances may simulate those of intestinal obstruction, the greater part of the abdomen being opaque (Fig. 22). Infants with polycystic disease of the kidneys may pass urine, the amount depending upon how much functional renal tissue remains. If there is an obstructive lesion causing hydronephrosis, and in some cases of polycystic disease, no urine will be passed. There may be a history of oligohydramnios, and the foetal membranes may show amnion nodosum (p. 31). If no urine is passing down the ureters because of bilateral obstruction or non-functioning kidneys, a catheter can readily be passed into the bladder but no urine is obtained. If the failure to void urine is due to urethral valves, catheterization will release urine from a distended bladder; if it is due to bladder-neck obstruction or atresia of some part of the urethra, catheterization will not be possible.

Abdominal distension is also a conspicuous feature of

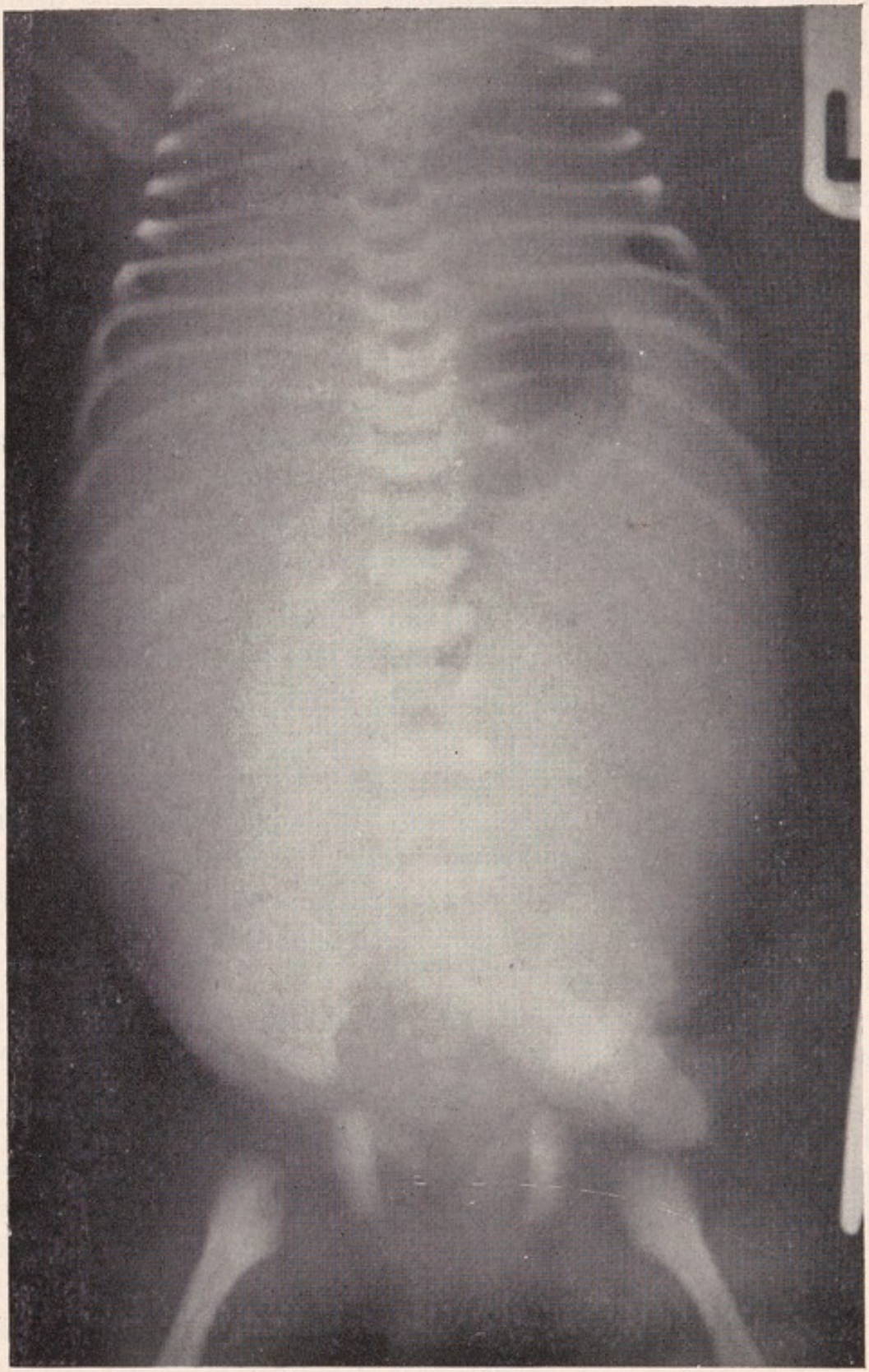


FIG. 22. Congenital polycystic kidneys. The huge kidneys occupy most of the abdomen, compressing the air-containing bowel into the centre of the abdomen. (Same case as Fig. 9.)

cloacal atresia. In this condition the rectum and ureters enter a common sac from which there is no exit. Affected infants are stillborn or die immediately after birth.

Polycystic disease and hydronephrosis may affect one kidney only, but the other kidney will function normally and normal amounts of urine will be passed. The diagnosis can only be made by the discovery of a mass in one flank or if infection supervenes. It is not possible to distinguish between these two conditions clinically, and it is not possible to distinguish them from a Wilms' tumour of the kidney which may rarely be present at birth. Any renal enlargement discovered in a newborn infant requires urgent and thorough investigation. An enlarged kidney may prove to be perfectly normal, except in size; in that case the opposite kidney may be small or absent.

Renal agenesis may be unilateral or bilateral. Unilateral cases will be overlooked unless there are associated malformations (which are quite often lethal) or the one kidney becomes diseased. It is, of course, impossible to estimate the number of healthy people who only have one kidney. Bilateral renal agenesis is usually associated with oligohydramnios and Potter facies. Early diagnosis is of little consequence because no treatment can be offered. These infants die soon after birth, probably as a result of pulmonary hypoplasia.

There is a very large number of congenital abnormalities of the urinary tract of which no mention is made in this book. Many remain undetected throughout life and are found unexpectedly at necropsy; others are brought to light by the investigation of individuals who are subject to urinary-tract infections.

EXAMINATION OF THE PERINEUM

Abnormalities in the perineal region are surprisingly easy to overlook. This is not because they are difficult to recognize but because of human nature. If a newborn

infant at the time of routine examination is found to be in a napkin full of meconium, there is a temptation to take this (rightly, as a rule) as evidence of patency of the anus and to assume (wrongly, perhaps) that there is no anatomical abnormality lurking beneath the meconium. This temptation must be resisted, the baby cleaned up and a careful inspection of the anus and external genitalia made.

Anal anomalies

The subject of anal anomalies has been made somewhat more difficult than it need be by the variety of nomenclature used in publications and by disagreement as to what abnormalities actually exist. An outstanding attempt to reconcile these views has been made by Partridge and Gough (1961). Fortunately, the clinician will never see the anomalies which do not exist and he need not worry about nomenclature. He need only satisfy himself that the anus is normal in position and calibre and that the rectum communicates with it and with nothing else.

An ectopic anus may be situated anywhere in the midline between its normal position and the vagina. This must be distinguished from a covered anus, a condition in which a normally situated anal canal leads into a subcutaneous tunnel which runs forward in the midline to open somewhere on the under surface of the perineum, scrotum, or penis. An ectopic or a covered anus is often narrower than a normal one. A normally situated anus may also be stenosed, the constriction being close to the muco-cutaneous junction. Extreme examples of anal stenosis, in which the orifice is microscopic, may be mistaken for an imperforate anus. Another anomaly, illustrated in Fig. 23, is anal bar; the anus is divided into two separate orifices by a median antero-posterior bridge of soft tissue.

If there is no anus, or merely a blind dimple, the extent to which the rectum has developed can only be determined radiologically. In some infants, the rectum terminates

immediately beneath the anal dimple; in more severe cases, there is agenesis of the greater part of the rectum, and in these infants there is commonly a rectovaginal, rectovesical,

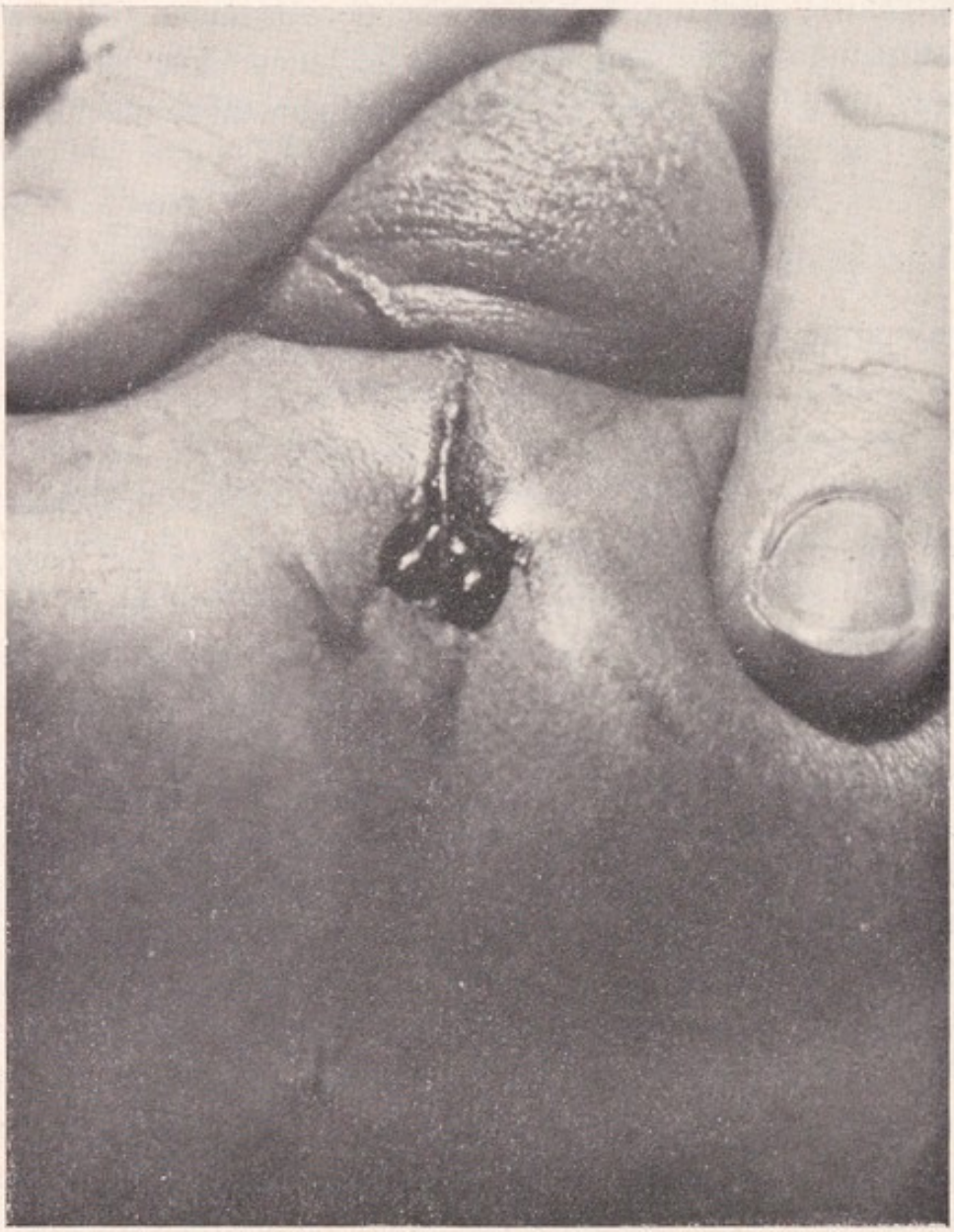


FIG. 23. Anal bar. Meconium is emerging on both sides of the bar. (*By courtesy of Mr. P. P. Rickham.*)

or rectourethral fistula. Sacral agenesis may also be present in these cases. In all infants without an anal orifice, a lateral radiograph should be taken with the baby upside down and a metal marker in the position of the anus (Fig. 24). The distance between the terminal air bubble

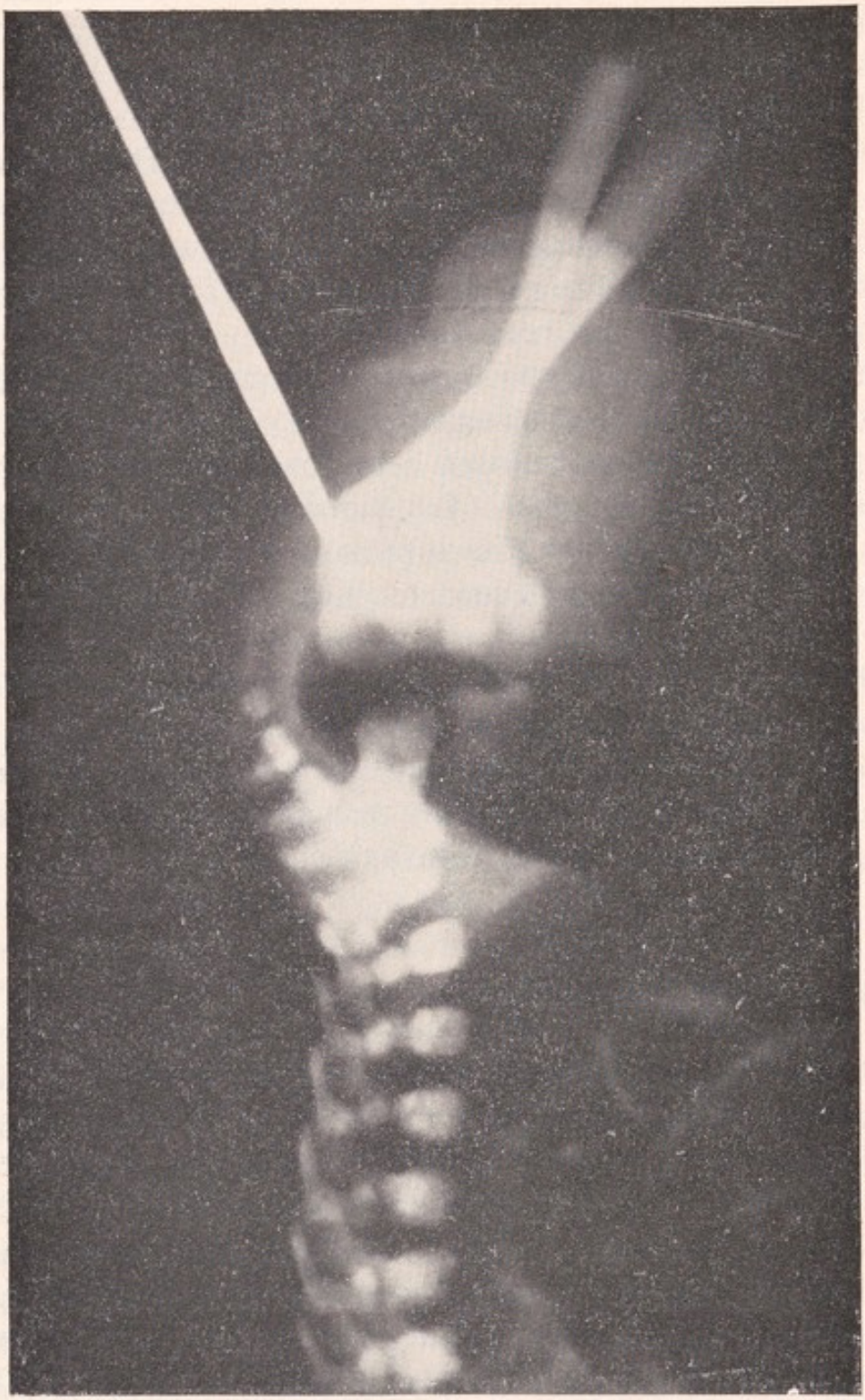


FIG. 24. Rectal atresia. The radiograph is taken with the infant inverted. The tip of the forceps indicates the site of the anal dimple. (*By courtesy of Mr. P. P. Rickham.*)

and the metal marker can then be estimated and the appropriate surgical approach made.

In high rectal atresia, the anal canal and the lower part of the rectum are normal and the obstruction is too high to be discovered by digital examination. The condition will therefore not be suspected until symptoms develop (*see* p. 14). A particular watch for symptoms should be kept on infants with oesophageal atresia, for in some cases there is an associated rectal atresia.

In all these abnormalities, the urgency of treatment depends upon the functional integrity of the anus. The ectopic, covered, or stenosed anus requires urgent surgery only if the free passage of meconium and stools is impossible. In contrast, the infant with no anal orifice or only a microscopic anus requires surgical treatment urgently.

The passage of meconium *per urethram* is clear evidence of a fistulous connection between the rectum and the bladder or urethra. The passage of meconium *per vaginam* indicates that there is either a rectovaginal fistula, or an ectopic anus opening into the vagina.

Genitalia

Abnormalities of the genitalia are common and a few of them are of great importance. In boys, hydrocele, hypospadias, and undescended testicles are all extremely common. The great majority of hydroceles resolve spontaneously and require no treatment. Hypospadias is usually glandular or coronal. In the most minor degrees of glandular hypospadias, the diagnosis is usually evident from the hooded prepuce rather than from the position of the urethral orifice, which may be only fractionally removed from its normal situation. Not infrequently, the ectopic meatus is a very small, slit-like opening which is scarcely visible except when the infant micturates. Meatal atresia is extremely rare, and if an orifice cannot be identified, the infant should

be observed for at least twenty-four hours. The passage of time usually leads to the reassuring discovery of a wet napkin.

The testes are quite commonly undescended at birth, but in the majority of cases they descend soon afterwards. If one testis is in the scrotum and the other cannot be felt, there is more reason for anxiety. A careful search should be made for the absent testicle in ectopic sites, of which the perineum is the most common.

Abnormalities of the genitalia of girls are less common than those of boys. Small polyps of the vaginal mucosa are seen frequently but are of no serious significance. Vaginal ectopic anus and rectovaginal fistula have already been mentioned. Imperforate hymen and vaginal atresia are rare but may lead to hydrometrocolpos. In this condition copious mucoid secretions accumulate in and greatly distend the uterus and vagina. These may reach sufficient size to press upon the bladder and ureters and cause retention of urine. If the obstruction is due to an imperforate hymen, examination of the vulva reveals a bulging membrane. If a hollow needle is inserted through this, large amounts of milky fluid can be withdrawn, the swelling becomes smaller and the urinary retention is relieved.

Of far greater importance, generally speaking, than the genital abnormalities in babies that are obviously boys or girls, are the anomalies which leave room for doubt whether a baby is male or female. These range over a broad spectrum of anatomical variations resulting from a number of different underlying disorders. At one end is the girl with a somewhat enlarged clitoris; at the other, the boy with perineal or penile hypospadias; in the middle is a group of infants whose external genitalia seem to be midway between those of a normal male and those of a normal female. The term intersex, meaning 'between the sexes', can be used for any individual whose external genitalia are not clearly those of one sex or the other, as long as the word is not regarded as carrying any aetiological implications.

The importance of determining the sex of a child

accurately at birth is twofold: it is clearly of great importance to the parents; it is also vitally important to the child, because sexual orientation is determined very early in life and normally corresponds to the baptismal sex and sex of upbringing. That is to say, if an infant is named and brought up as a boy, but is later found to be a masculinized girl, any attempt to 'change the sex' after the age of three years is likely to fail psychologically. Masculine interests and attitudes will already be firmly ingrained. It is perhaps surprising that sexual orientation depends so greatly upon upbringing and so little on the histology of the gonads or the number of X chromosomes, but it is so.

The most common type of intersex is the female pseudohermaphrodite with congenital adrenal hyperplasia (the adrenogenital syndrome). This condition is determined by an autosomal recessive gene which causes defective production of the enzyme responsible for the synthesis of hydrocortisone. The deficiency of glucocorticoids and mineralocorticoids leads to an increased production of ACTH by the foetal pituitary gland, and this in turn stimulates the adrenal cortex to secrete excessive amounts of androgens. This process begins early in gestation and the degree of masculization of a female foetus, although variable, is usually sufficiently marked to be obvious at birth. In the male, who is less often affected, the effects of the androgen excess are not usually apparent for a few years, but electrolyte disturbances may develop early in life (*see* p. 110).

The female foetus may also be masculinized by androgenic drugs taken by the mother during pregnancy. A few cases have been attributed to testosterone, but the majority in this group have been caused by progestational drugs taken to prevent abortion. A few examples have been described of masculinization of babies born to mothers with androgen-secreting tumours, principally arrhenoblastomata. The majority of infants exposed to exogenous androgens have shown enlargement of the clitoris with or without labial fusion. Occasionally, when progestogens have been taken

very early in pregnancy, development of the internal genitalia has also been disturbed.

True hermaphrodites are very rare indeed. By definition, they have gonads which contain both ovarian and testicular tissue. The external genitalia may be male or female, but are usually intermediate between the two.

In the testicular feminization syndrome, an enzyme block results in the failure of the foetal adrenal cortex to produce masculinizing hormones. The external genitalia therefore appear female, but the gonads are testes and the sex chromosomes are those of a normal male.

When inspection of the newborn infant arouses doubts in the doctor's mind as to its sex, the diagnosis is approached along the following lines.

(1) Maternal history: Have any drugs with known androgenic action been taken, particularly progestogens?

(2) Family history: Have any previous children in the family suffered from the adrenogenital syndrome, or died unexpectedly in the early weeks of life?

(3) A 24-hour specimen of urine is examined for 17-ketosteroids. Infants with adrenal hyperplasia show a raised level. The result cannot be interpreted with confidence in the first two weeks of life, but after this time 17-ketosteroid excretion in excess of 1 mg. in 24 hours may be regarded as abnormal.

(4) Buccal smears may be examined for nuclear chromatin. This appears as a small, darkly-staining body immediately beneath the nuclear membrane. It is present in a high proportion of cells from normal females but not in those of males. It is thought to represent the second X chromosome. The interpretation of buccal smears requires experience and is not an investigation to be undertaken on an amateur basis. Female infants with masculinized genitalia, whether caused by congenital adrenal hyperplasia or maternal drugs, will have normal amounts of nuclear chromatin for females; that is, they are chromatin positive. Infants with testes and hypospadias, or with undescended testes, will have no nuclear chromatin;

that is, they are chromatin negative. If the nuclear chromatin is at variance with the apparent phenotypic sex, there may well be an abnormality of the sex chromosomes, and chromosome studies should be undertaken. These are discussed further below.

(5) If the results of clinical examination and the above investigations should fail to settle the question of sex, it may be necessary to perform a laparotomy in order to inspect the internal genitalia and to take biopsy specimens from both gonads. Laparotomy is, of course, entirely unnecessary if the diagnosis of adrenal hyperplasia has been established biochemically, or if the history suggests that drugs given to the mother have been responsible for masculinization.

The urgency of sex assignment bears repetition. The adrenogenital syndrome needs early treatment with cortisone; without this, death from adrenal cortical insufficiency may occur early, suddenly, and unexpectedly. A protracted period of uncertainty about the infant's sex is intolerable for the parents. Finally, as has already been stressed, sex orientation is settled early in life. There is therefore a very limited time within which it is possible to change one's mind about a baby's sex without impairing the child's future welfare.

Abnormalities of the sex chromosomes are not, as a rule, associated with abnormalities of the external genitalia at birth and are therefore not usually diagnosed until later. A number of surveys have been done in which buccal smears of several thousand newborn babies have been examined for nuclear chromatin. A surprisingly high proportion of phenotypic males are chromatin positive (about 1 in 400, according to Moore, 1959) but chromatin negative females are found less frequently. This procedure has no place in the normal routine examination of the newborn infant.

The three most common sex-chromosome abnormalities are *gonadal dysgenesis* (often called Turner's syndrome), in which the somatic cells contain 45 chromosomes instead of the normal 46, there being only one X chromosome;

Klinefelter's syndrome with 47 chromosomes, XXY; and the *triplo-X syndrome* with 47 chromosomes, XXX. The second and third of these will not be recognized at birth unless buccal smears are examined. The baby with Klinefelter's syndrome appears to be a normal boy, except that the testicles are small or undescended, but he is chromatin positive. The triplo-X individual appears to be a normal girl but the buccal smear shows double nuclear-chromatin bodies in many cells. In true hermaphrodites, the sex chromosomes may be male (XY), female (XX), or a mosaic mixture of the two (XX/XY).

Gonadal dysgenesis (Turner's syndrome) may sometimes be diagnosed at birth. The external genitalia are those of a normal girl. The principal signs which suggest the diagnosis are oedema of the feet or legs and webbing of the neck. The cause of the leg swelling is unknown, but it may be very striking; sometimes it lasts for years. It may be confined to the feet, more commonly spreads up to the knees, and rarely involves the hands also. It pits on pressure. The discovery of such oedema in a newborn girl should prompt the examination of buccal smears which, in gonadal dysgenesis, are chromatin negative. The neck webbing must be distinguished from the short neck associated with the Klippel-Feil syndrome and other developmental defects of the cervical spine. If gonadal dysgenesis is diagnosed, a careful search should be made for evidence of coarctation of the aorta, which is present in a number of cases. However, the majority of affected individuals have normal hearts, and most children with coarctation of the aorta have normal sex chromosomes.

EXAMINATION OF MECONIUM AND URINE

It is a long-standing and valuable tradition that the midwife records the first passing of urine and meconium by the newborn baby. The importance of this custom cannot be over-emphasized, for the failure of either of

these events to take place within a reasonable time may be the first indication of a serious but remediable condition.

Failure to pass urine

The majority of infants pass urine within twenty-four hours of birth (93 per cent. of them, according to Sherry and Kramer, 1955). Failure to do so should prompt the midwife or nurse to report the fact to the clinician. The majority of babies that have not passed urine within this period do so during the next twelve hours. Failure to pass urine within thirty-six hours of birth is strongly suggestive of an underlying abnormality, and after forty-eight hours, the suspicion almost amounts to a certainty. Reference has already been made (p. 6) to the fact that oligohydramnios and amnion nodosum are strong evidence that there has been no foetal micturition. The association of Potter facies with renal agenesis has also been mentioned, but the infant with no kidneys does not live long enough for his failure to pass urine to cause anxiety. The lesions which give rise to this symptom are therefore those which obstruct the urinary tract. These include not only intrinsic lesions (polycystic kidneys, ureteric obstructions, bladder neck and urethral obstructions), but extrinsic lesions which may cause pressure from without. These include hydrometrocolpos and a variety of cysts and tumours.

Whatever the precise site and cause of the urinary-tract obstruction, both kidneys, if present, are likely to be enlarged. The bladder will also be distended if the obstruction is below this level. The discovery of renal enlargement in an infant that has not passed urine (and, indeed, in one that has) is an indication for urgent investigation and treatment. Estimation of the blood urea is a simple and informative test. Descending pyelography has drawbacks in the newborn period. Abdominal gas shadows often mask the important areas; intravenous pyelography requires a continuous intravenous infusion to be set up, while intramuscular pyelograms are often

unsatisfactory. Finally, if the blood urea is appreciably raised, the kidneys are probably incapable of concentrating the dye sufficiently to show up on a radiograph. For these

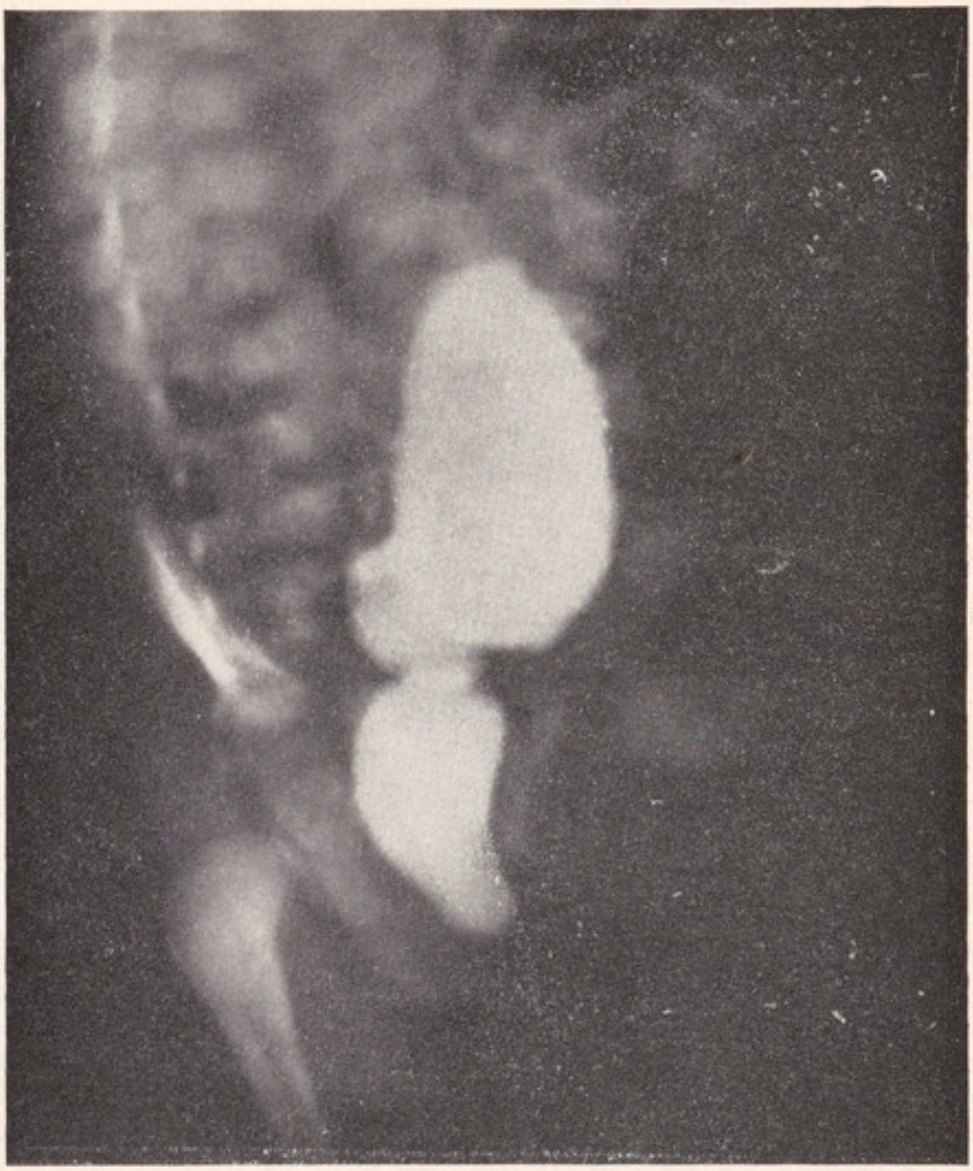


FIG. 25. Urethral valves. The bladder has been filled with radio-opaque dye. It is enlarged, and below, like an inverted pear, is the distended proximal urethra. A thin stream of dye can just be seen passing along the penile urethra. (*By courtesy of Mr. P. P. Rickham.*)

reasons retrograde pyelography is often preferable. The information required can frequently be obtained very simply by introducing radio-opaque fluid into the bladder by way of a urethral catheter. If the ureters are much dilated, they will be filled by reflux (unless they are obstructed).

Fig. 25 shows a cystogram carried out on an infant with urethral valves.

Failure to pass meconium

Meconium is usually passed within twenty-four hours of birth and delay beyond this time is scarcely ever seen. However, it is worth remembering that an infant that passes meconium before birth may not do so again for more than twenty-four hours. The customary watch for the first bowel movement sometimes fails to take this into account, and a nurse may express anxiety about an infant who 'has not passed meconium' but whose skin is stained with it.

True failure to pass meconium within twenty-four to thirty-six hours of birth suggests the likelihood of intestinal obstruction. However, obstructive lesions, especially those in the small intestine or above, can usually be recognized before this time because vomiting and abdominal distension develop (*see* Chapter 4). In lower obstructions, such as Hirschsprung's disease, rectal atresia, and meconium plugs, the failure to pass meconium may be the first sign of trouble, although distension is usually obvious by this time.

It is important to appreciate that meconium may be passed by an infant with complete intestinal obstruction. At birth, meconium fills the greater part of the large intestine, but there is very little in the small bowel. This situation is found not only in normal infants but in many with high intestinal obstruction. Fig. 26, for example, shows the colon full of meconium in an infant with duodenal atresia. There can only be two possible explanations for this. Either meconium is formed largely *in situ* and is not derived to any great extent from swallowed material, or these obstructions arise by closure of a previously patent bowel. Such 'acquired' atresias can be produced experimentally by ligation of the mesenteric vessels in foetal animals (Louw, 1959). Swallowed elements also contribute towards meconium, the squamous cells normally

demonstrable in it being derived from foetal skin and swallowed with amniotic fluid. This forms the basis of Farber's test (1933), in which the presence or absence of squames in meconium is interpreted as showing whether

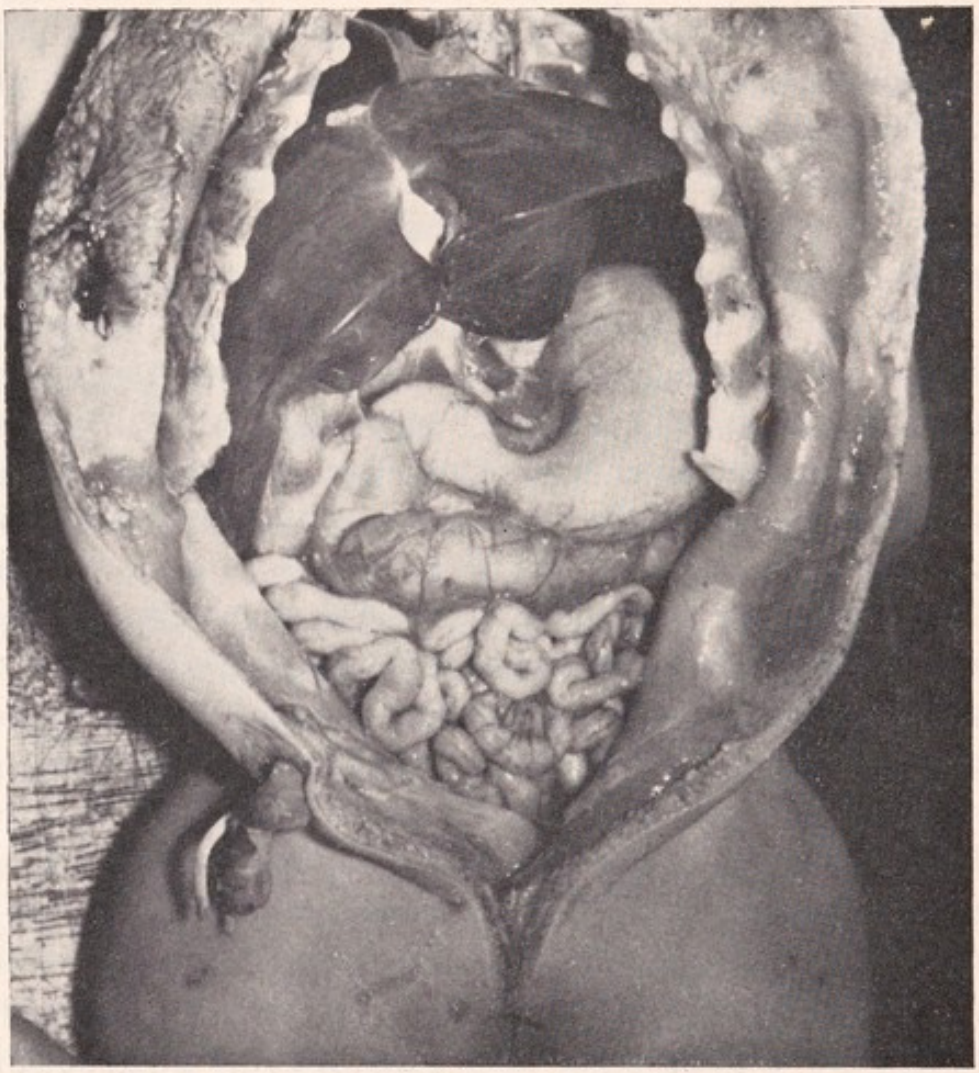


FIG. 26. Duodenal atresia. The small bowel is empty. The transverse colon is full of meconium. The stomach and the first part of the duodenum are only moderately distended. The baby died soon after birth from associated malformations.

or not there is intestinal obstruction. Unfortunately this test may give false positives and false negatives (Emery, 1952) and is no longer used.

The practical consequence of these observations is that the passage of meconium is compatible with complete intestinal obstruction, although in these circumstances the

meconium is often small in amount and has rather a dry, tenuous appearance. The fact that an infant has had a bowel action must never be allowed to detract from the significance of vomiting and abdominal distension.

When an infant is reported not to have passed meconium, it should first be confirmed that meconium was not passed before birth, then inquiry should be made about fibrocystic disease (mucoviscidosis) or Hirschsprung's disease in siblings, and whether the infant has vomited. Examination of the abdomen for distension, visible peristalsis, and palpable masses, must be supplemented by inspection of the anal region to see if the anus is normal in size and position. The principal anal anomalies have already been described (*see* p. 81). Rectal examination will determine whether the anal canal is of adequate calibre and the lower two inches or so of the rectum can be felt. Meconium plugs (p. 149) may be found in the rectum and can be removed digitally or washed out. Finally, rectal examination is the finest stimulus to bowel evacuation known, and removal of the finger is often followed by a gush of meconium which affords more relief to the doctor than to the patient.

If the rectum is found to be empty, a firm rectal tube may be passed gently up and will go considerably further than a finger. This technique is also valuable in premature infants in whom the small anus will not admit a finger without considerable trauma. If any evidence of obstruction is found, or if doubt remains, a radiograph of the abdomen should be taken with the infant upright. The size and distribution of gas shadows and the presence or absence of fluid levels will indicate whether there is obstruction. The diagnosis of obstructive lesions is considered in Chapter 4.

The stools

Examination of the stools sometimes provides the earliest clue to congenital abnormality. Blood may be mixed with meconium from the first stool, or appear on the second or

third day of life; more rarely, an infant may pass a typical melaena stool after this time. Hiatus hernia and duplication cysts of the bowel may be associated with melaena, but only exceptionally does this sign prove to be due to a malformation. Melaena within twenty-four hours of birth is usually the the result of the infant having swallowed maternal blood during delivery. In most instances a history of antepartum or intrapartum haemorrhage is forthcoming. Melaena on the second or third day, with or without haematemesis, is conveniently labelled *haemorrhagic disease of the newborn*, a descriptive term which evades the vexed question of aetiology.

These remarks apply only to macroscopic melaena obvious to the naked eye. Occult blood detected chemically is present in the stools of most infants in the first few days of life and is without special significance in a sick infant.

Biliary atresia may first be diagnosed from the pallor of the stools, although this is often overlooked until the infant becomes jaundiced. The anatomical lesion cannot be diagnosed before operation, and the term 'atresia' should be construed as meaning any congenital abnormality causing obstruction of the biliary tract. All will lead to a deficiency of bile in the stools. The meconium may be noted to be paler than normal although it is by no means colourless. From the fourth day onwards, when milk stools are passed, the absence of pigment is very striking. This usually antedates jaundice, which generally develops in the second week of life but may not appear until the third or fourth week. Enlargement of the liver may also take some time to develop. As jaundice progresses, there may from time to time be some bile in the stools. This in no way negates the diagnosis of complete biliary obstruction; the bile is thought to be excreted from the intestinal wall. Biliary atresia is further discussed on p. 153.

Examination of the stools is helpful in fibrocystic disease of the pancreas (mucoviscidosis). Only a minority of affected infants present the clinical picture of meconium

ileus in the newborn period (p. 145), the majority appearing normal until they develop respiratory or digestive disturbances later in infancy. In the presence of a family history of the disease, diagnosis may be made before symptoms develop by testing the stools for the presence of trypsin. This is normally present both in meconium (Emery, 1952) and in milk stools from birth. According to Emery, trypsin can be detected in meconium at a dilution of 1 in 8 or more. He suggested that deficiency of trypsin provided useful confirmatory evidence of intestinal obstruction below the ampulla of Vater. However, in the absence of other signs of obstruction, and with a family history of fibrocystic disease, deficiency of trypsin would have a very different significance. If trypsin is consistently absent from the stools, the enzyme content of duodenal juice should be estimated for confirmation before such a serious diagnosis is made. An additional sign sometimes seen is pronounced jaundice, presumably caused by increased viscosity of the bile. Examination of the electrolyte content of the sweat is of limited diagnostic value in the early days of life.

EXAMINATION OF THE MUSCULO-SKELETAL SYSTEM

Skeletal malformations are very common and, for the most part, they are impossible to overlook. Talipes, the early treatment of which is so important, is obvious at a glance. Extra digits are usually conspicuous by the abnormality of their position, shape, or size. Normally formed supernumerary digits are astonishingly easy to overlook, especially on the feet.

Long, thin fingers and toes associated with dolichocephaly are suggestive of Marfan's syndrome (arachnodactyly), but the important complications of this disorder—dissecting aortic aneurysm and dislocation of the lens—are not apparent in the neonatal period.

Defective ossification of the skull

Three generalized skeletal disorders are characterized by defective ossification of the skull and may therefore be confused with one another or with hydrocephalus.

The first is *achondroplasia*, in which the vault of the skull is large and the limbs are short in proportion to the trunk (Fig. 27).

The second is *fragilitas ossium* (*osteogenesis imperfecta*) in which the skull is poorly ossified and multiple fractures are usually present at birth. Sometimes intrauterine fractures of the limb bones have united in such a way as to cause shortening, and the infant may then have a superficial resemblance to an achondroplastic. However, the facial appearance is usually characteristic in achondroplasia, and the X-ray appearances are different in the two conditions.

The third disorder in which the skull is soft at birth is *cleidocranial dysostosis*. The condition is commonly present in one parent, being caused by a dominant gene; the limbs are perfectly formed, and in most cases it is possible to feel that the clavicles are absent or rudimentary. The clavicular abnormality can easily be confirmed radiologically (Fig. 16, p. 47).

To these three might be added a fourth, *osteochondrodystrophy*, of which the best-known variety is the Hunter-Hurler syndrome (gargoylism). However, unless there has been a previously affected child, it is unusual to diagnose this condition with confidence in the neonatal period. The characteristic physical features and mental handicap become apparent later in the first year of life.

The hip-joint

In company with talipes, the skeletal malformation most in need of early diagnosis is congenital 'dislocation' of the hip. A true dislocation is virtually never seen in the newborn baby except in those with muscle paralysis due to

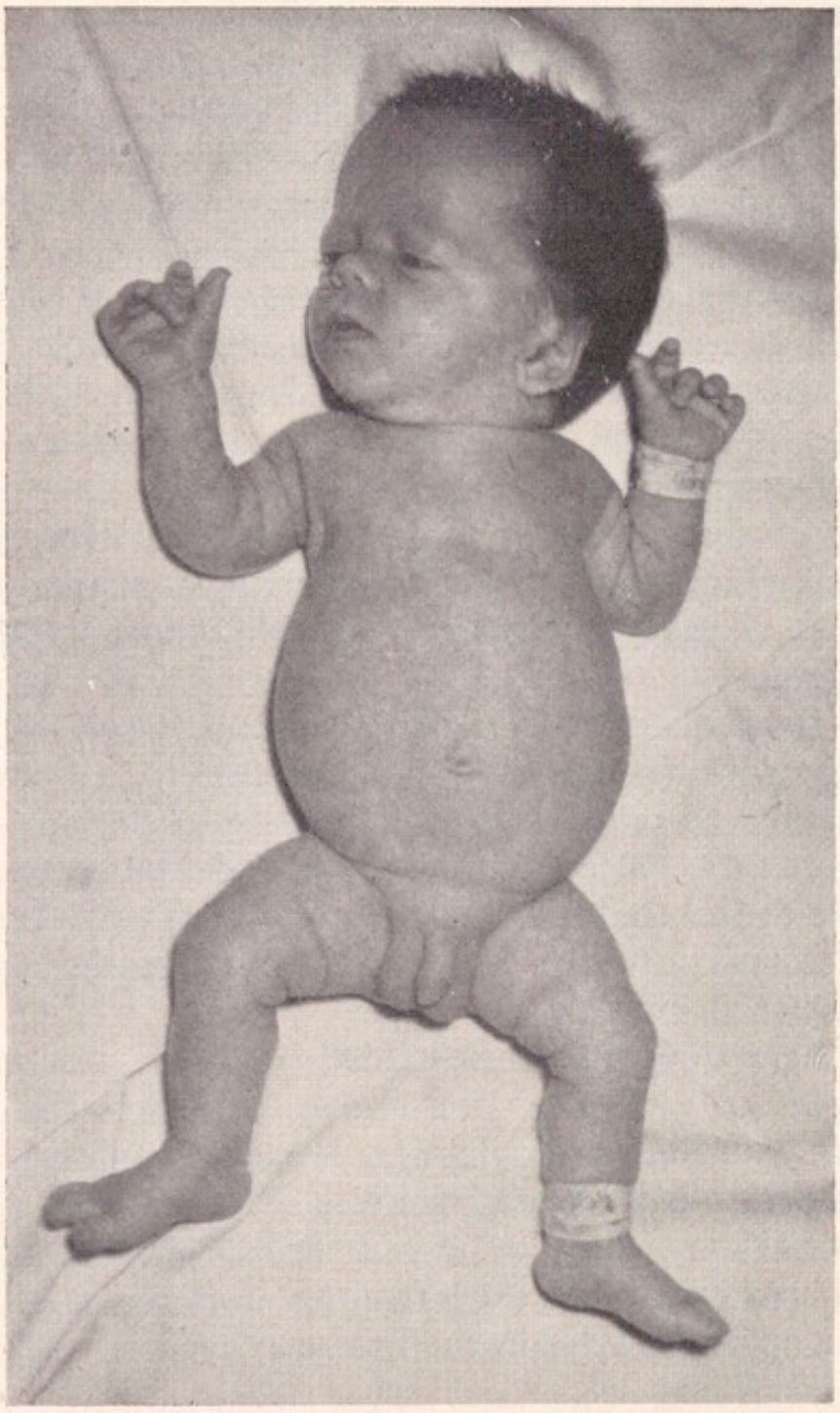


FIG. 27. Achondroplasia. Note the broad forehead, snub nose, short limbs and large abdomen. (*By courtesy of Dr. F. P. Hudson.*)

myelomeningocele. The aim must be to detect hips which, if left untreated, are likely to dislocate when the baby starts bearing weight on the legs. In such an unstable hip-joint the head of the femur may be displaced laterally, but not to the extent of being dislocated. This is subluxation. Alternatively, it may be possible to produce a subluxation deliberately; the condition of the hip is then described as preluxation. An unstable hip-joint is due partly to acetabular dysplasia and partly to joint laxity. Both are to some extent genetically determined, and a family history of congenital dislocation is therefore not uncommon. Girls are far more often affected than boys; unilateral cases nearly always involve the left side; and an abnormally high proportion of affected infants are delivered by the breech.

If the hip joint is completely dislocated, as it is in some infants with myelomeningocele, the head of the femur rides up on to the ilium above the acetabulum. The leg is therefore shortened in comparison with the opposite side (in unilateral cases), and it is possible to move the femur independently of the pelvis (telescoping). In addition, there will be limitation of abduction of the flexed hip. The skin creases on the internal aspect of the thigh may be asymmetrical, but this is so often seen in normal infants that it is of no diagnostic importance.

Diagnostic tests for subluxation and preluxation

Three tests are used in the diagnosis of subluxation and preluxation of the hip. They are illustrated in Figs. 28 (*a*), (*b*), (*c*), and (*d*). In all these tests the baby and the examiner's hands should be warm and the manoeuvres should be gentle. If the infant begins to struggle, the examination is unsatisfactory.

(1) *Hart's test* (1950) is a test of the degree of abduction possible in the flexed hip. The infant's knees and hips are flexed, the legs being together, and the hips are then gently abducted. In the normal newborn, abduction

to 90° or slightly less is possible. After a few weeks or months the increase in muscle tone limits abduction to a rather smaller arc and it may be more difficult to decide

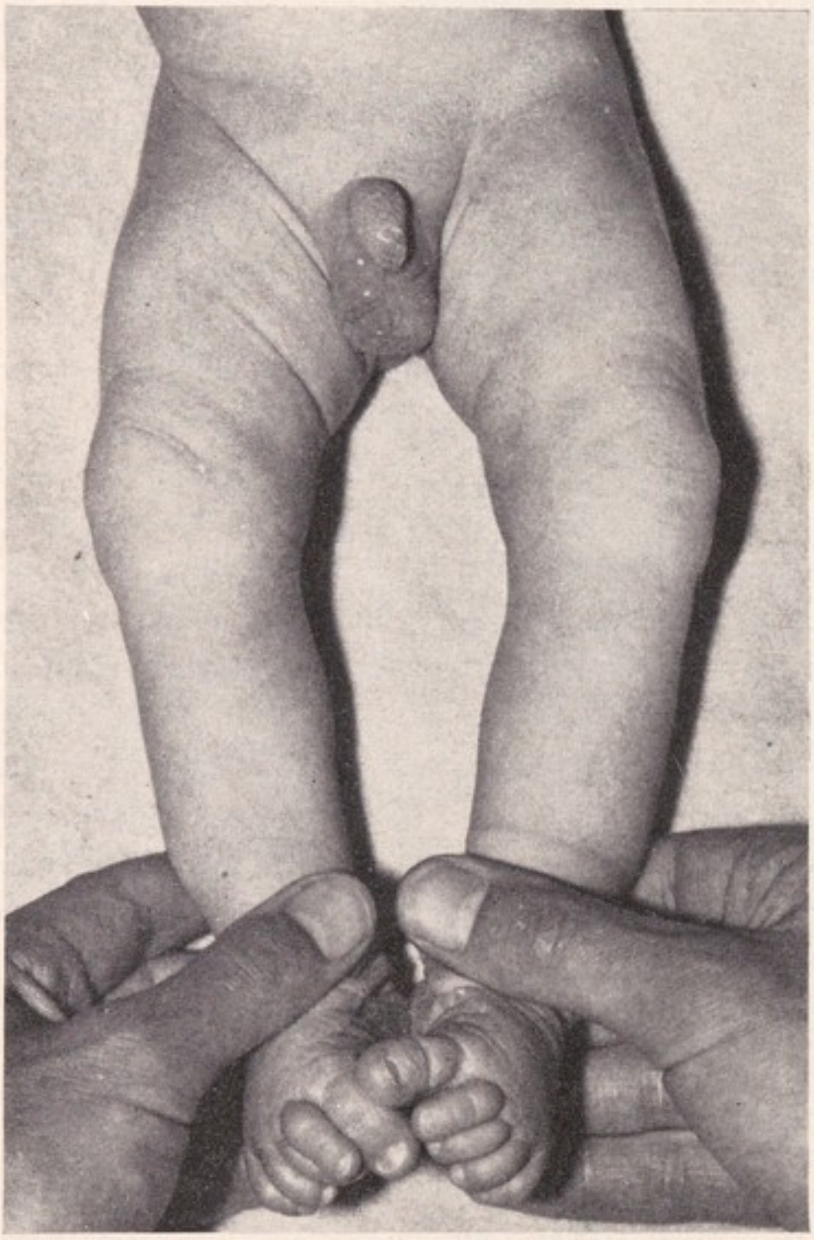
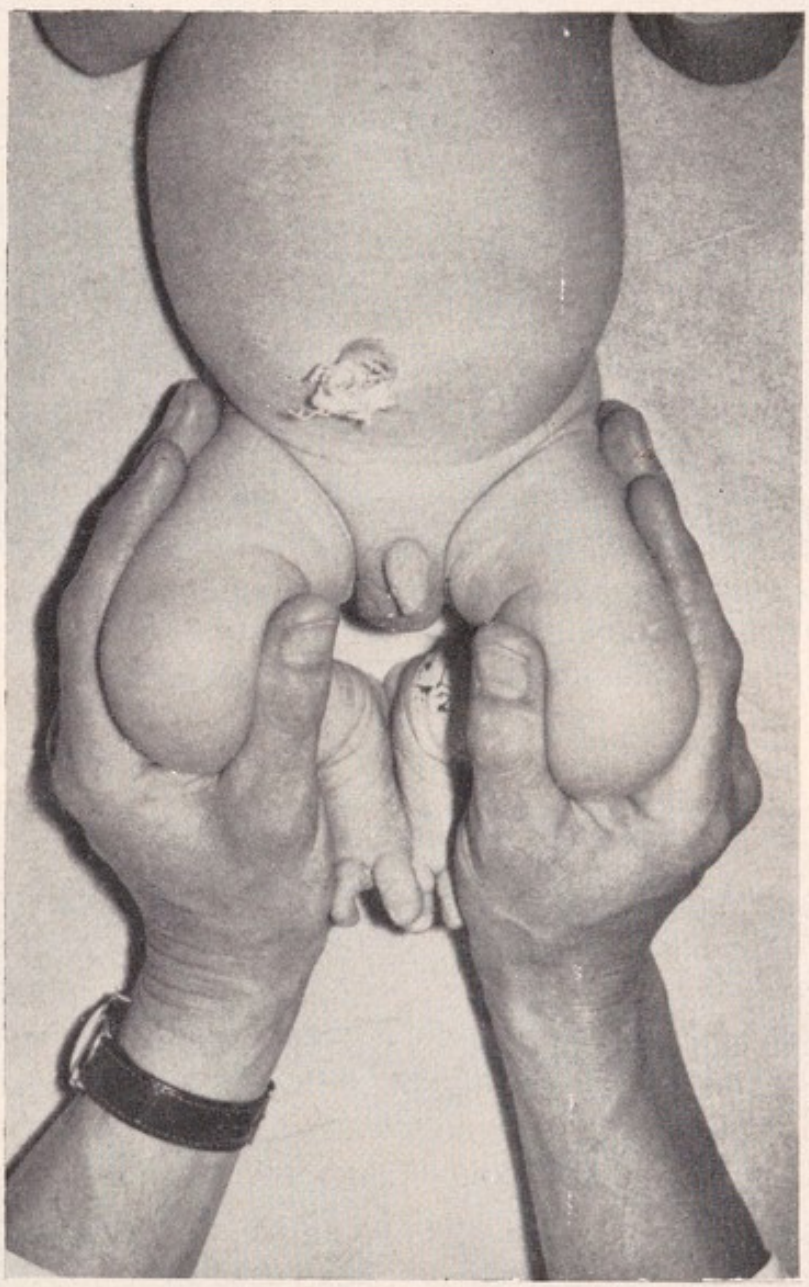


FIG. 28. Examination of the hips.
(*a*) An unstable hip joint is most likely to subluxate when the legs are extended.

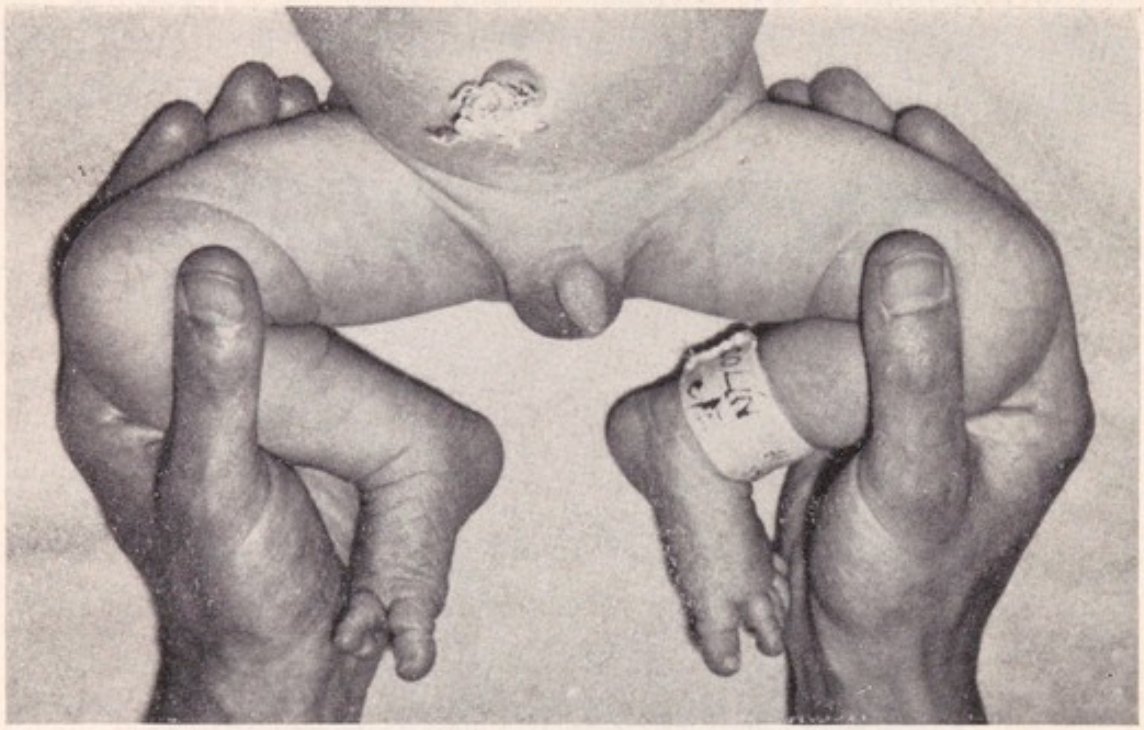
what is normal and what is abnormal. If there is dislocation or subluxation of the hip, abduction will be limited to about 60° or less. Hart's test is a very useful and simple screening test and will detect most cases, but it will not detect preluxation of the hip.

(2) *Ortolani's test* (1948). This may be regarded as an extension of Hart's test, although it was described earlier. The manoeuvre is designed to reduce a subluxation

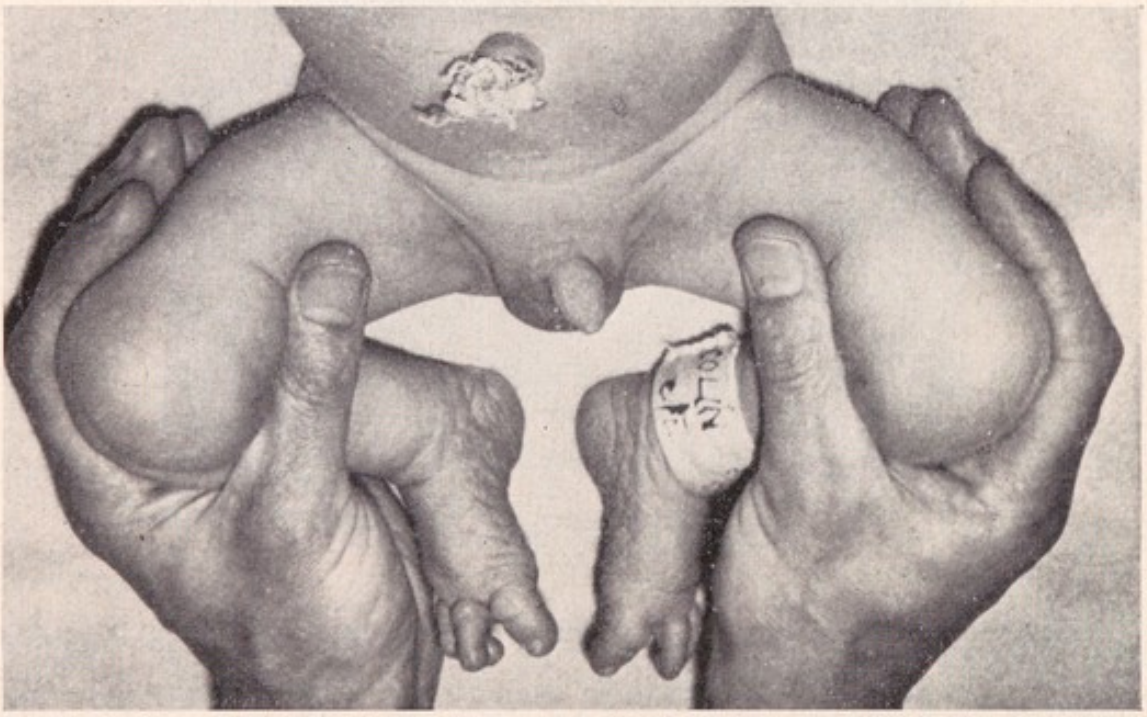


(*b*) The hips and knees are flexed prior to abduction.

of the hip. As the flexed hips are abducted, the fingers over the greater trochanter push the upper end of the femur forward. At the same time, slight pressure is applied along the long axis of the thighs. If there is a subluxation, this manoeuvre forces the femoral head back



(*c*) The flexed hips are abducted. The baby illustrated had bilateral congenital subluxation of the hips, but when the subluxation was reduced full abduction was possible.



(*d*) Subluxation provocation test (*see* text).

into the acetabulum with a click which can be clearly felt and sometimes heard. After the click, full abduction becomes possible. If the thighs are then adducted with a view to repeating the test, the femoral heads may be felt to click out again. Ortolani's test is positive in subluxation of the hip, but not always in complete dislocation nor in preluxation.

(3) *Palmén's test* (1961). If the femoral head is in the acetabulum at the time of the examination, neither Hart's test nor Ortolani's test will detect any abnormality. Palmén, therefore, devised a test which he calls subluxation provocation. With the hips flexed and semi-abducted, an attempt is made to displace the femoral head laterally out of the acetabulum. If the joint is unstable it will be possible to do this, sometimes with a palpable click. This test is, in effect, the reverse of the Ortolani manoeuvre. The one tries to produce a subluxation, the other tries to reduce it.

Radiological confirmation

When dislocation, subluxation, or preluxation of the hip is suspected, radiological confirmation is usually attempted, but this may be very difficult (*see* Fig. 29.) In the newborn baby, the epiphysis for the head of the femur is entirely cartilaginous and therefore does not show on an X-ray film. The position of the head and its relationship to the acetabulum must be deduced from the position of the shaft of the femur, and this in turn is affected by the position of the infant's legs. The acetabulum is also to a considerable extent cartilaginous at this age and dysplasia is more difficult to be certain about than it is in older babies. (An additional clue present in the older baby, but not at birth, is that the upper femoral epiphyses often develop unequally in unilateral subluxation or dislocation.)

It is clear from these considerations that, whatever technique is used for X-ray examination of the hips in the

newborn infant, the position of the baby, the pelvis, the legs, and the X-ray tube must be carefully standardized. Multiple films are to be avoided because in girls (which most of these infants are) the gonads will inevitably be exposed to irradiation.

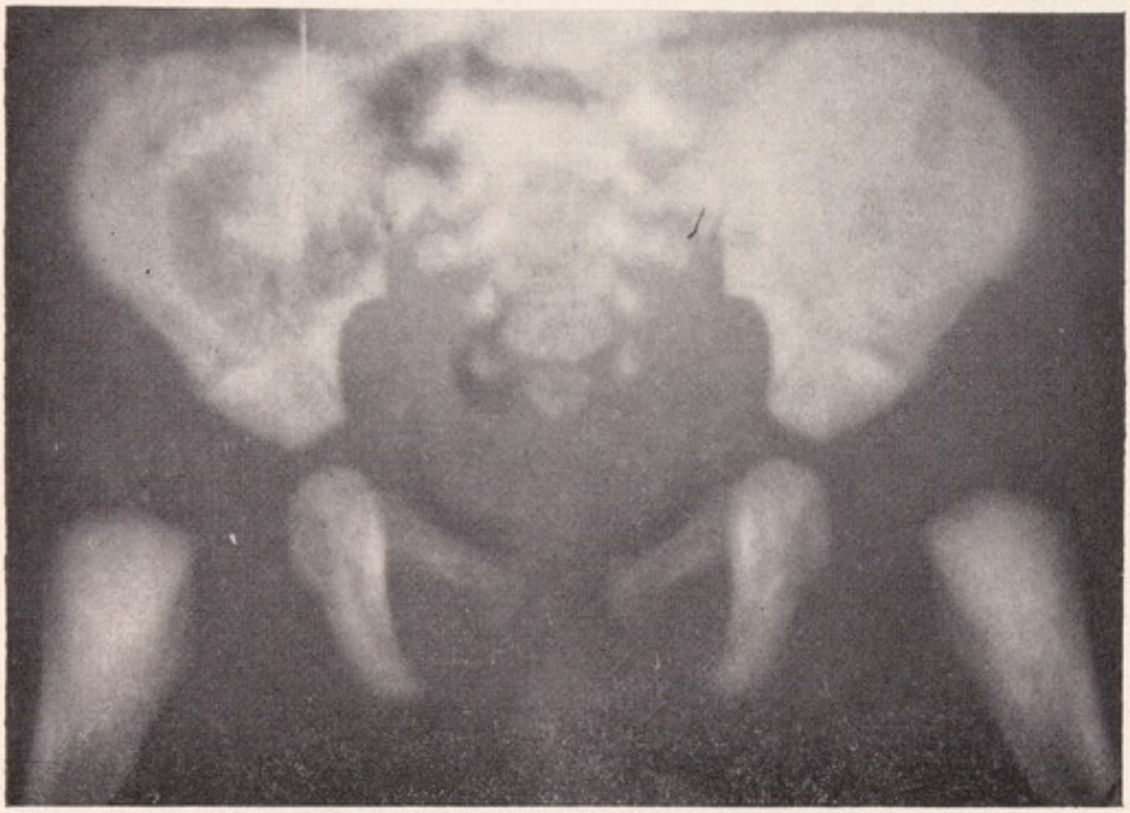


FIG. 29. X-ray of a newborn infant whose hip joints were both grossly unstable. Radiological diagnosis is difficult and unnecessary.

Muscles

Little need be said of congenital abnormalities of muscle. Generalized muscular hypotonia is discussed in Chapter 4. Congenital absence of the abdominal muscles is immediately obvious from the lax, redundant appearance of the abdominal wall which bulges when the infant cries. Its importance lies in the frequent association with internal malformations, especially hydronephrosis and hydroureter, to which reference has already been made (p. 77).

The muscle most commonly congenitally absent is pectoralis major. Usually the sternal head is absent and

the clavicular head present. The ipsilateral nipple may be hypoplastic or absent. The absence of this muscle does not cause any significant functional disability and often passes unnoticed for years.

EXAMINATION OF THE SKIN

There is little difficulty in recognizing disorders of the skin in the newborn baby, although their identification sometimes calls for the assistance of a dermatologist. Reference has already been made to post-anal dimples and dermal sinuses (p. 70). Deep skin dimples may also be seen elsewhere than the back, particularly on the proximal parts of the limbs. These appear to arise from the adhesion of the dermis to the deep fascia at one small point. The later development of subcutaneous fat between these layers leaves a dimple at the site of the adhesion. The dimples are deepest where the fat is thickest, that is, on the thighs.

Although the common skin diseases seen in the neonatal period, whether infective, allergic, or toxic, may reasonably be regarded as acquired after birth, a few are prenatally determined. Apart from diffuse disorders of pigmentation, brief mention will be made of three skin diseases which have characteristic appearances. In *congenital ichthyosis* the skin is hard, dry, and thickened. For severe examples, the term 'harlequin foetus' is sometimes used but needs to be distinguished clearly from the unrelated harlequin phenomenon seen in some premature infants, in which one half of the body is flushed and the other pale, with a sharp line of demarcation down the midline. Congenital ichthyosis is determined by a recessive gene. The severe form is fatal.

Epidermolysis bullosa is also caused by a recessive gene and is usually fatal. The suffix 'letalis' is sometimes used to emphasize the bad prognosis, but there have been a few cases recorded of recovery. The lesions are bullae, often

large and multiple. Many of the lesions rupture before birth leaving red, raw areas of denuded skin (Fig. 30). A few bullae may still be intact and contain fluid.

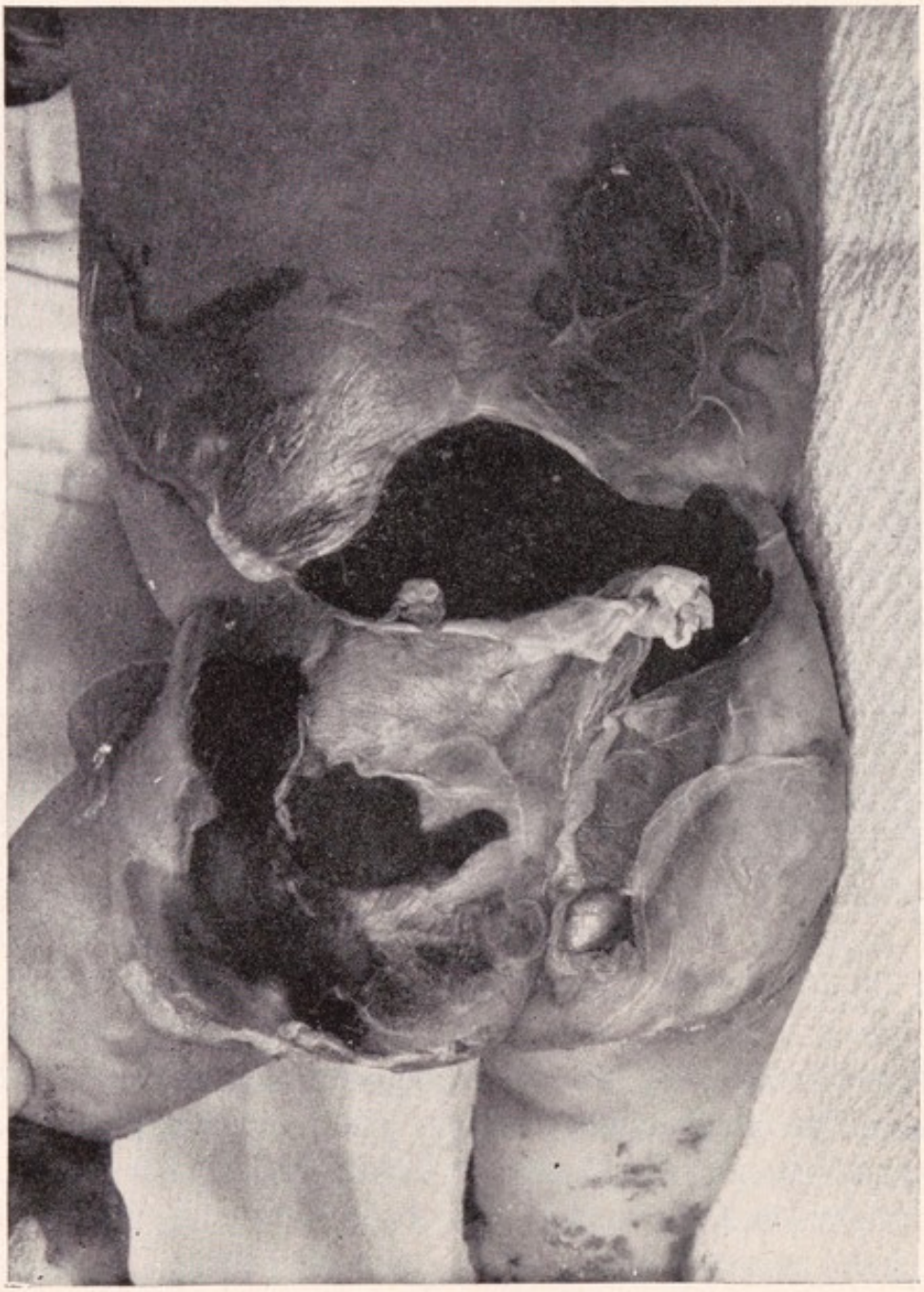


FIG. 30. Epidermolysis bullosa.

Incontinentia pigmenti is a curious skin disorder. The pigmentation which gives the disease its name is not present early in life, but the lesions are, nevertheless, characteristic. They are papules and vesicles distributed

over a variable amount of the body surface in a linear pattern. On the limbs the lines are longitudinal; on the trunk they are transverse.

Congenital defects of the skin occur most commonly over the vertex of the head and appear as round or oval lesions about half an inch across (Fig. 17, p. 53). The cause is unknown. The defect heals but the scar does not bear hair.

Complete *albinism*, which is determined by a recessive gene, is obvious at birth not so much from the appearance of the skin as from the white hair and the pink eyes. Partial albinism in negroes is usually due to a dominant gene and is therefore seen in successive generations. The size, number, and position of the non-pigmented areas are very variable, but the centre of the forehead is commonly involved.

Pigmentation in white infants is usually limited to one or two small pigmented naevi present at birth. Occasionally, however, pigmentation is very extensive (Fig. 31). Flat lesions, however extensive, tend to be benign and often fade considerably with time, but raised, warty lesions may be malignant.

Congenital *malformations of the dermal blood vessels* are common. The flat, red lesions seen on the nape of the neck, the upper eyelids and the centre of the forehead are so common as scarcely to justify the term 'abnormality'. They regress spontaneously. Capillary naevi on the face may be associated with intracranial vascular malformations, particularly if they cover the territory supplied by the first or second divisions of the trigeminal nerve (Sturge-Weber syndrome). The intracranial naevi later calcify, resulting in a characteristic radiological appearance, but they are not calcified in early infancy.

Extensive capillary naevoid formation may be associated with subcutaneous lymphangiectasis and sometimes with hypertrophy of the underlying tissues. This occurs most commonly in an arm or leg, or may involve the whole of one side of the body (hemihypertrophy). A similar effect

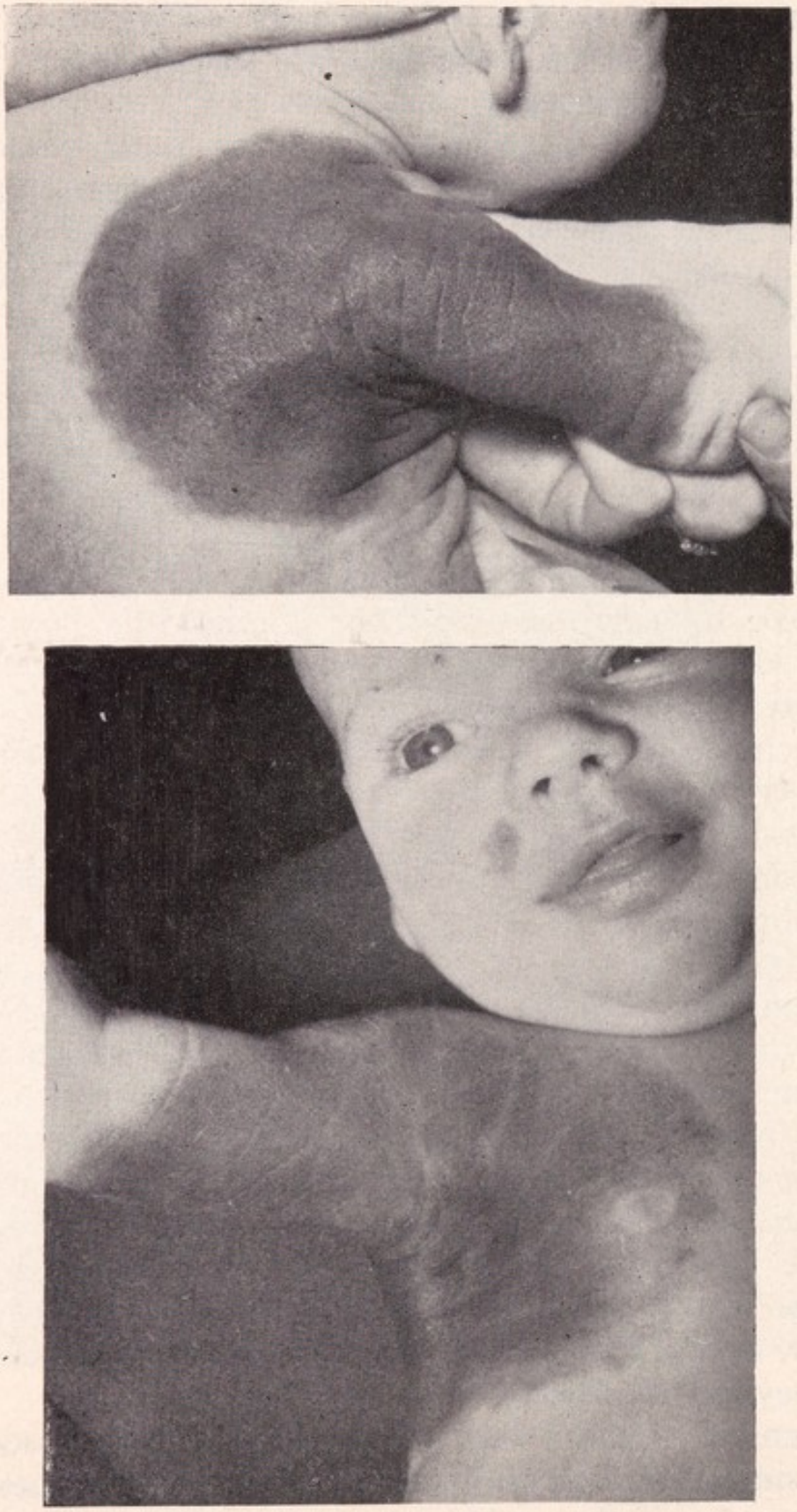


FIG. 31. Naevus pigmentosus. In addition to one large pigmented naevus, this child had multiple smaller naevi, of which two are visible on the face.

may be seen with lymphangiectasis in the absence of any skin naevus.

It is interesting to note that strawberry naevi, the most common birthmarks of all, are not apparent at birth but develop within the first two weeks of life. They first appear as flat, pink, sharply demarcated areas which darken in colour and then become raised above the skin surface.

Cavernous naevi, on the other hand, are present at birth. They involve deeper structures than do strawberry naevi and contain venous blood, although the most superficial part is often bright red. A few infants have been reported in whom very large cavernous naevi were associated with profound thrombocytopaenia. The naevi appear to act as sponges (or perhaps graves) for platelets. The bone marrow, rather unexpectedly, shows the features of idiopathic thrombocytopenic purpura, with plentiful megakaryocytes showing maturation arrest. However, neither cortisone nor splenectomy affect the course of the disorder which is progressively downhill unless the naevi are treated promptly and effectively. In any infant with extensive or multiple cavernous naevi, therefore, the platelets should be counted.

EXAMINATION OF METABOLIC FUNCTIONS

Metabolic and endocrine disorders can only rarely be recognized in the neonatal period without recourse to biochemical tests. Albinism, for example, can be diagnosed clinically, but with most conditions in this group it is not practicable to exclude them unless there is some special reason for doing so. The congenital metabolic disorders are mostly caused by recessive genes, and a common indication to do special tests is a history of a previously affected sibling. In other cases investigation is prompted by evidence of ill-health in the baby. The number of genetically-determined disorders of metabolism now runs

into hundreds and even the most common are rare. Fibrocystic disease of the pancreas, which is the most common of these disorders amongst Caucasian peoples, has already been discussed (p. 95).

Congenital adrenal hyperplasia

In a girl this presents at birth as female pseudo-hermaphroditism (p. 86), but the male infant and his genitalia appear normal at birth. If there is a history of a previously affected child, a 24-hour specimen of urine should be examined for 17-ketosteroids and pregnanetriol as soon as possible after the second week of life. In the absence of a family history, diagnosis may be delayed until it becomes apparent that the boy's growth and development are precocious. However, many affected infants have profound electrolyte disturbances which cause serious symptoms as early as the second or third week of life. Vomiting, dehydration, and collapse may suggest a diagnosis of pyloric stenosis, but no pyloric tumour is palpable and adrenal cortical insufficiency is suggested by the high levels of serum potassium and urinary chloride. The diagnosis must be confirmed by steroid excretion studies, but it is unwise to delay giving extra salt while the urine is being collected. This condition presents a medical emergency. Delay may be as lethal as it would be in a case of intestinal obstruction.

Phenylketonuria

Phenylketonuria needs early diagnosis because early treatment can prevent mental deficiency. Unfortunately the urine test with ferric chloride, which is so simple to do (especially with the test strip, Phenistix), does not become positive in affected children until the age of 3 to 6 weeks, and very rarely later. For routine purposes the urine should be tested as soon as possible after the age of 4 weeks. This is adequate because effective treatment,

started within two months of birth and maintained subsequently, will lead to normal growth and development.

Many premature babies remain in hospital for more than four weeks and their urine can be tested before they go home. The test should also be done on children with eczema, convulsions, or retarded development.

Diagnosis can be established at birth by estimating the level of phenylalanine in the serum, and this should be done whenever there has been an older sibling affected by phenylketonuria. This should also be done on any infant found to have a positive urine test because there are other substances which give an identical colour reaction with ferric chloride. The normal serum phenylalanine concentration is less than 2 mg. per 100 ml.

Details of the urine tests are as follows (Hudson, 1960):

(1) Using 5 per cent. ferric chloride solution:

Add a few drops of 5 per cent. ferric chloride solution to a fresh sample of urine. In the presence of phenylpyruvic acid, a deep, olive-green colour appears within a minute and fades within half an hour.

(2) Using Phenistix test-strips (made by Ames Co.):

The test-strip can be moistened with a drop of urine or a wet napkin. In either case the urine must be fresh. Care must be taken not to soak or rub the colour out of the test-strip. The colour fades after half an hour, and for that reason the strip must not be left in the napkin with a view to reading the result later. If filter paper is soaked in urine and dried in the air, it can be remoistened and tested with Phenistix up to a week later.

Galactosaemia

Galactosaemia is another condition in which prompt diagnosis and treatment are vital. Like phenylketonuria, it is determined by a recessive gene, and an older child in the family may have been affected. Otherwise the condition may be suggested by early vomiting, failure to

thrive, jaundice, hepatomegaly, or any combination of these. A reducing sugar is demonstrable in the urine, but enzyme tests for glucose are usually negative. However, since reducing sugars (including galactose) are sometimes found in the urines of normal infants in the first few days of life, complete laboratory investigation, including chromatography, is essential. It must be remembered that infants who have not taken, or who have not retained, milk feeds will not have galactosuria.

Cretinism

The importance of diagnosing cretinism early cannot be over-emphasized. The earlier treatment is started, the better is the chance of a satisfactory response. Congenital hypothyroidism is nearly always a sporadic condition, and the clinician will only be particularly alerted in three circumstances, all of which are rare:

(1) In an area of endemic cretinism.

(2) If the mother has taken antithyroid drugs or other goitrogens (for example, iodides) during pregnancy.

(3) If there is a family history of cretinism in older siblings. Familial goitrous cretinism is a rarity. Several different biochemical varieties have been described but all appear to be controlled by recessive genes. The goitres are not necessarily present at birth. Deafness is also present in some families.

In any other circumstance cretinism will only be diagnosed early by the clinician who is looking for it round every corner. Sluggish feeding, constipation, umbilical hernia, enlargement of the tongue, a hoarse cry, or the facial appearance may first attract attention. One of the most constant features of the facies is the transversely furrowed brow. Only exceptionally do the features arouse suspicion in the neonatal period, but the photograph shown in Fig. 32 was taken on the twenty-eighth day of life.

There is an important sign which may enable the diagnosis of cretinism to be made before any of the classical symptoms

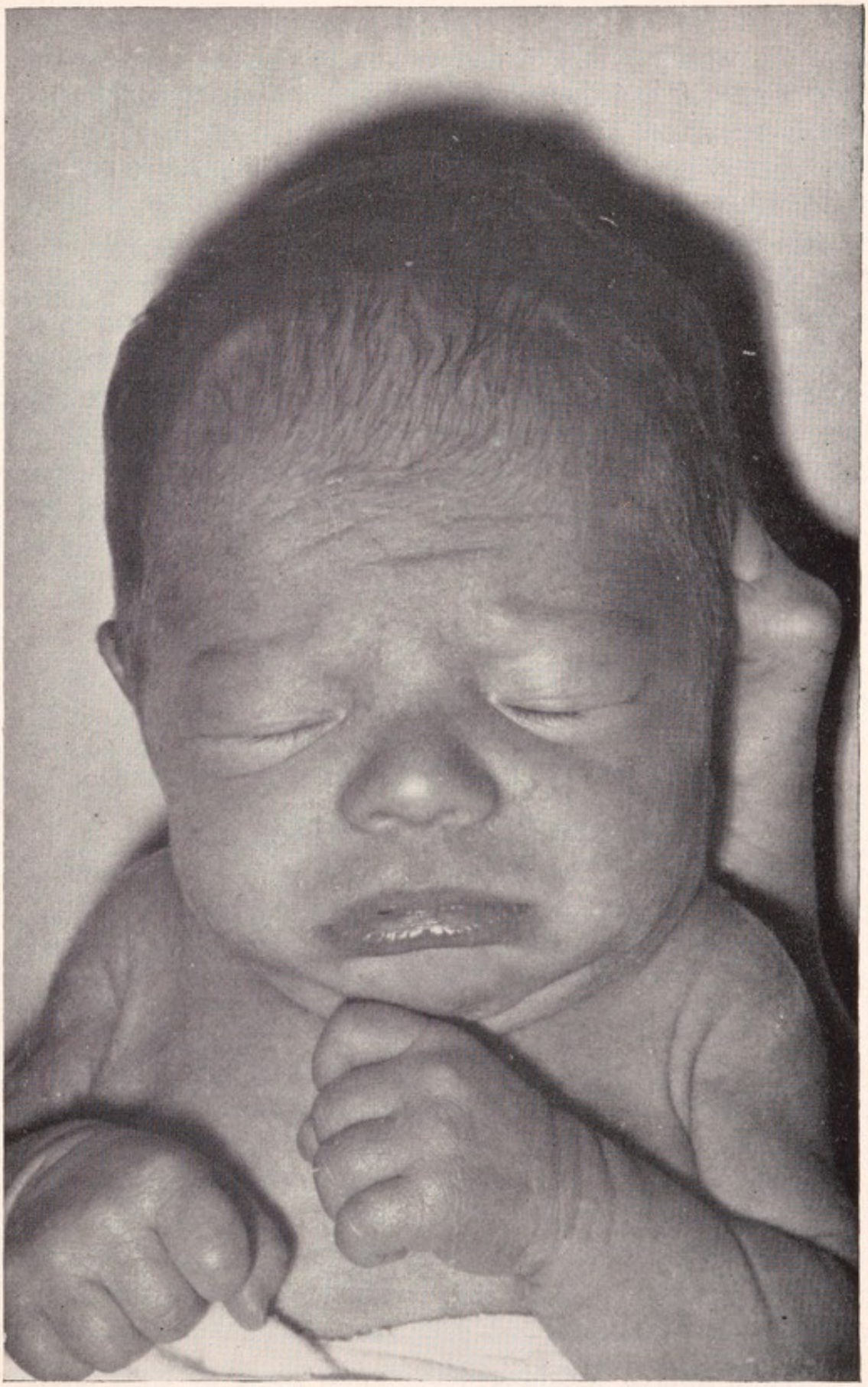


FIG. 32. Cretinism. Note the furrowed brow.

or signs have appeared. This is jaundice of a severity or duration which is out of proportion to the maturity of the baby ('icterus prolongatus'). It is an exaggerated physiological jaundice, appearing on the third or fourth day of life, and with the serum bilirubin almost entirely of the unconjugated variety. The differential diagnosis of jaundice is considered on page 152, and hypothyroidism is a rare but important cause. The clinical suspicion may receive radiological support. For this purpose, the knee and foot should be X-rayed. The great majority of normal full-term infants have two epiphyseal centres at the knee and two or three tarsal centres (*see* Table 2). The diagnosis of hypothyroidism is confirmed by estimation of the serum protein-bound iodine, which is normally 7–12 μg.% in the first week of life and gradually falls to half this value at the age of one year.

TABLE 2

OSSIFICATION CENTRES PRESENT AT BIRTH

(Data from Christie, 1949)

Ossification Centre	*Proportion in whom present*	
	Boys *	*Girls* *
Calcaneus	100%	100%
Talus	100%	100%
Distal femur	100%	100%
Proximal tibia	97%	90%
Cuboid	60%	76%

* White infants weighing 4000 G. or more at birth and X-rayed within seventy-two hours of birth.

Radio-active isotopes in diagnosis

A word may be said about the use of radio-active isotopes in the diagnosis of cretinism. Formal radio-active iodine uptake studies are of limited value. The relatively long half-life of I^{131} means that, even with tracer doses, the newborn baby may be exposed to undesirable amounts of ionizing radiation. The short half-life of I^{132} means that uptake and excretion can only be studied over a short

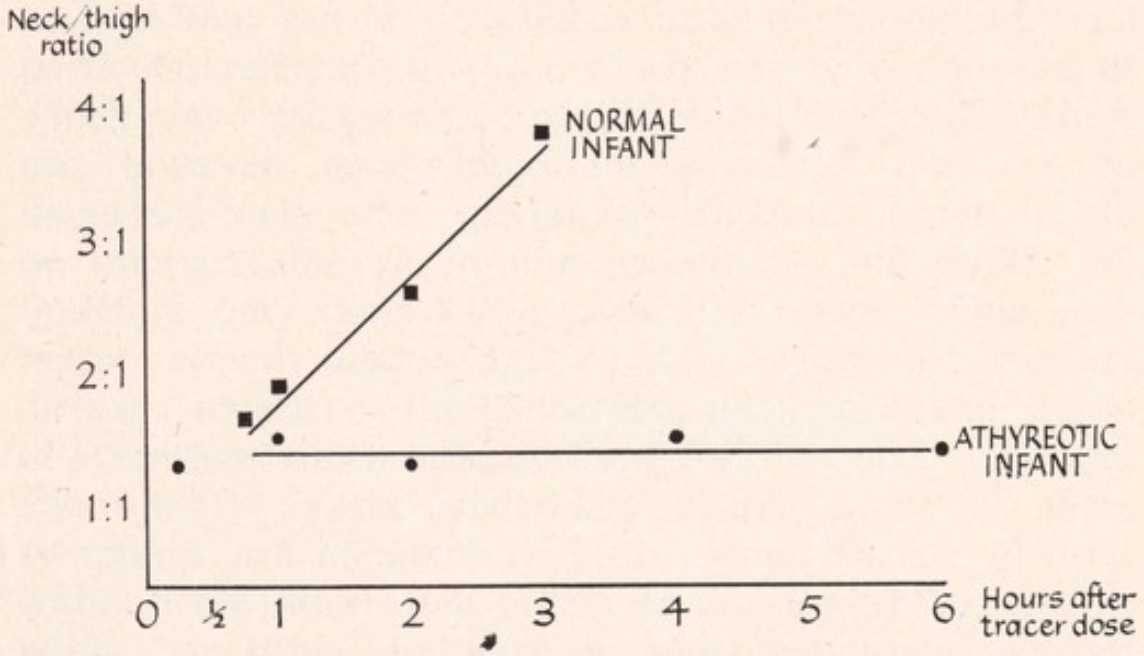


FIG. 33. Uptake of I^{132} in normal and athyreotic newborn infants. (5 microcuries of I^{132} were given by gastric tube and radiation counts measured over neck and thigh.)

period. Interpretation of the results may also be difficult because the normal newborn has iodine uptake levels which would be interpreted in an adult as indicating hyperthyroidism.

It is, however, a relatively safe and simple matter to establish, by the use of isotopes, whether or not there is any functioning thyroid tissue in the neck. A tracer dose of I^{132} is given by mouth and readings are subsequently taken with a radiation counter over the front of the neck and, for comparison, the thigh. In the absence of active thyroid

tissue in the neck, serial readings will show comparable rates of decay in the neck and the thigh. The test shown in Fig. 33 was done because the mother had previously given birth to two cretins. It enabled the diagnosis to be established very early in life.

CHAPTER 4

Early Symptoms of Congenital Abnormalities

IN THE last chapter the diagnosis of congenital abnormalities that can be detected at birth was discussed, with particular reference to those which may be overlooked or which may raise problems of differential diagnosis. There are, however, many important anomalies that can be diagnosed only after special investigations which would be impracticable as routine procedures and which are therefore only done for a particular reason. One such reason, already discussed, is a family history of hereditary disease; another is the appearance in the neonatal period of symptoms which indicate the possibility of a congenital abnormality as the underlying cause. Some of these symptoms are apparent at birth, some become obvious within a few hours, and others do not appear for days or weeks. In some hereditary disorders symptoms may not appear for many years, an extreme example being Huntington's chorea. Only those symptoms which are likely to appear in the neonatal period are considered here.

RESPIRATORY DIFFICULTIES

There are three main groups of newborn infants with respiratory symptoms. In the first, there is no satisfactory attempt to breathe; in the second, there is an attempt which meets with mechanical opposition; in the third, respiration is satisfactory, but oxygenation of the blood is imperfect.

The earliest symptom of these three is failure to establish

normal respirations. This is almost invariably the result of foetal asphyxia and only rarely a consequence of congenital abnormality. It is precisely because of this rarity that a malformation may be overlooked whilst the doctor's attention becomes fixed more and more closely upon resuscitative measures.

The asphyxiated infant is either cyanosed or pallid. If the asphyxia has been severe enough to make him pallid, he is limp, often covered with meconium, and makes no attempt to breathe. No congenital abnormality will simulate this condition. Confusion may be caused if an infant's pallor is the result of acute blood loss. The bleeding may have occurred into the maternal circulation, into the circulation of an identical twin, from the placenta (by premature separation in abruptio placentae or placenta praevia, or by accidental incision at Caesarean section), or from the end of an avulsed umbilical cord. The history will usually suggest the true explanation for the infant's pallor except in the case of foeto-maternal transfusion. This diagnosis can only be proved by the demonstration of large quantities of foetal red cells and foetal haemoglobin in the mother's blood. Profound anaemia from any of these causes will also lead to anoxia, which makes the diagnostic difficulty even greater. Haemoglobin and haematocrit readings will be normal if the haemorrhage was recent. The heart rate may provide a clue, being rapid after haemorrhage but slow in severe asphyxia from other causes.

The blue asphyxiated infant usually has shallow, irregular, or infrequent respirations. This is an important point of distinction from the baby whose cyanosis has some basis other than asphyxia. Respiratory difficulty and cyanosis may be caused by choanal atresia, micrognathia, laryngeal and tracheal obstruction, gross abnormalities of the lungs, and diaphragmatic hernia. In each of these conditions the cyanosed infant tries to breathe but only meets with partial success. This contrasts with the blue asphyxiated infant who appears not to be trying very hard.

Upper respiratory-tract obstruction

Obstruction of the upper respiratory tract is most commonly due to choanal atresia or micrognathia. In *choanal atresia*, there is a membranous or bony obstruction of the posterior nares. Since the inborn pattern of breathing of the newborn infant is through the nose, obstruction of the nasal airway causes respiratory difficulty at birth or within a very short time afterwards. Some infants manage to take occasional breaths through the mouth; others attempt to breathe through the nose until they lose consciousness, the jaw drops, and air is able to enter the lungs. In either case dyspnoea may be intermittent. The head is sometimes held tilted backwards. The infant's distress is aggravated by attempts to feed him and by covering the mouth. The condition may be suspected if long strings of clear mucus can be pulled from the nares.

The simplest way to determine whether an infant is breathing through the mouth or the nose is to hold a cold, smooth object (such as a pen-torch, metal spatula, or pocket mirror) close to the mouth and nares and to observe where water vapour condenses. In choanal atresia, catheters passed down the nose are held up before they reach the pharynx. The atresia may be demonstrated radiologically by instilling Lipiodol into the anterior nares (Fig. 34).

Choanal stenosis is a condition of partial obstruction of the posterior nares. An affected infant may breathe through the nose with obvious difficulty and indrawing of the chest wall. There is a noticeable stertor, reminiscent of the noise made by a child with enlarged adenoids. Choanal atresia and stenosis may be unilateral. Here, diagnosis is much more difficult, but is also much less urgent. Bilateral atresia demands early treatment to relieve the obstruction.

Obstruction of the pharynx is most commonly due to *micrognathia* (hypomandibulosis), the lower jaw being too small to hold the tongue away from the posterior pharyngeal

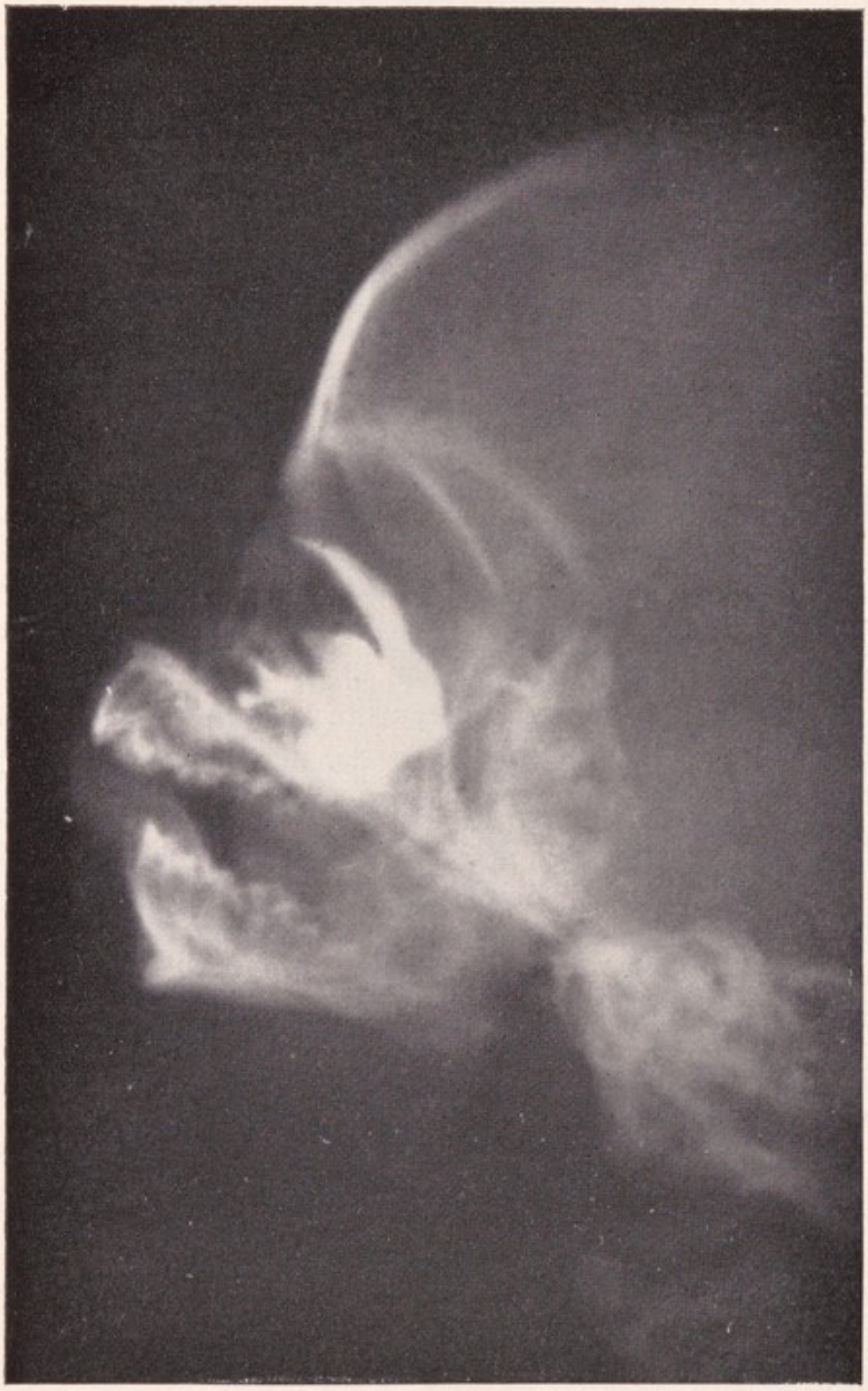


FIG. 34. Choanal atresia. Lipiodol has been instilled into the anterior nares. It cannot flow into the nasopharynx because the posterior choanae are obstructed. (*By courtesy of Mr. P. P. Rickham.*)

wall. Minor degrees of micrognathia are usually symptomless and the jaw soon develops to a normal size, but if the condition is severe the airway may be almost completely obstructed. This obstruction may be relieved by putting the baby in the prone position, and severe cases require to be nursed prone on a special frame for several months.

Less common lesions which may obstruct the pharynx include macroglossia (p. 177), large lingual cysts, haemangiomata and lymphangiomata of the tongue and pharynx. Very rarely a baby is born with adhesions between the tongue and the palate (ankyloglossia) which may cause pharyngeal obstruction.

Congenital stridor

Stridor may be of laryngeal or tracheal origin and is rarely severe. It is most commonly due to a congenital softness of the cartilage of the larynx which falls inwards on inspiration. This condition gradually improves with the passage of time, so that the stridor has disappeared before the infant is a year old. This condition is commonly called *congenital laryngeal stridor*. Much more rarely the larynx may be obstructed by webs, papillomata, lymphangiectasis, or laryngeal palsy. These conditions can only be distinguished by laryngoscopy which should be carried out if the stridor is severe or if it does not improve in the expected way with time. A few infants have been reported in whom a solid, cartilaginous bar obstructed the trachea immediately below the larynx. In this condition, and the equally rare laryngeal atresia, respiration is impossible and death almost immediate. If the tremendous respiratory effort and absent breath sounds are noted and their significance appreciated, a tracheostomy would be life-saving.

Narrowing of the trachea may result from deficiency of the cartilage in the walls, but is more often caused by extrinsic pressure. This may be due to a congenital goitre or some other mass in the neck, which will be

obvious, but is more commonly due to *abnormalities of the great vessels* at the thoracic inlet. A right-sided aorta, double aorta, or aberrant left subclavian artery (arising from the first part of the aorta and passing behind the oesophagus) may press upon the trachea a little above the bifurcation. Less commonly, an aberrant pulmonary artery may compress the trachea immediately above the bifurcation. Most of the aortic abnormalities result in vascular rings which also compress the oesophagus, a fact which can be readily demonstrated by means of a barium swallow. Operation is necessary in severe cases.

Abnormalities involving the lungs

Dyspnoea and cyanosis will be evident if a substantial proportion of the lung tissue is non-functional, or functioning inadequately. The congenital abnormalities which may cause this clinical picture include pulmonary agenesis, congenital emphysema, extensive cystic disease of the lung, and diaphragmatic hernia. Respiratory difficulty is likely to be evident from birth in all these conditions. This is important, because, in the respiratory distress syndrome (hyaline membrane disease), dyspnoea is usually only evident after a few hours, although the experienced eye may detect evidence of respiratory difficulty earlier. This is also true of most cases of neonatal pneumonia. Even when the infection appears to have been acquired *in utero* it is exceptional for respiratory symptoms to be apparent at birth, although they may appear within a matter of hours.

In the newborn infant respiratory disease may, in a disconcerting way, be simulated by intracranial haemorrhage. Dyspnoea and cyanosis (or cyanotic attacks) may be the dominant symptoms, there may be no twitchings or convulsions and the fontanelle tension may be normal. This confusing picture is particularly common in premature babies.

With the congenital anomalies of the lung it is con-

venient to include *pneumothorax*, although it should perhaps be regarded as an acquired lesion.

These intrathoracic abnormalities are nearly always unilateral, if only because bilateral lesions of comparable severity would be incompatible with existence for more than a few minutes. Because the lesions are unilateral, the physical signs in the chest are different on the two sides. If the abnormality is a diaphragmatic hernia, congenital emphysema, pulmonary cysts, or a pneumothorax, the mediastinum is shifted away from the lesion. Only in pulmonary agenesis or extensive unilateral collapse or atelectasis does the mediastinum move towards the abnormal side. In practice, it is rarely possible to make an accurate diagnosis without recourse to radiography.

The combination of early dyspnoea or cyanosis with asymmetrical signs in the chest or displacement of the heart will therefore prompt an urgent request for radiographs of the chest. These should be taken with the infant upright; they should include lateral views, which are particularly helpful in diaphragmatic hernia. They are also valuable when the antero-posterior film appears to show an intrathoracic mass. Such masses are rare and may prove to be bronchogenic cysts, duplication cysts of the alimentary tract, anterior meningoceles, or congenital neoplasms.

Radiographs are often invaluable in the diagnosis of neonatal disorders, not only of pulmonary disease but also of cardiac malformations and abdominal disease, especially intestinal obstruction. They need to be done well; much depends upon the radiologist, the radiographer, and the standardization of radiological techniques. A film taken with the baby erect is generally more helpful than one taken with the baby horizontal. Films taken with portable X-ray machines are usually disappointing.

Although the modern infant incubator is fitted with a slide to hold an X-ray plate, enabling the baby to be X-rayed without disturbance, this gadget is best ignored. Its use combines the disadvantages of using a portable

X-ray machine with the drawbacks of X-raying the baby in the horizontal position. The more seriously ill the infant, the more important it is to have a good film at the first attempt. If the infant is in an incubator he should be taken to the X-ray department in it and, when the radiographer is ready, removed, held in position whilst the exposure is made, and immediately returned to the incubator. There is scarcely any circumstance in which this policy cannot be adopted and it pays handsome dividends in the form of minimizing the number of exposures necessary. This not only saves time and tempers but also reduces the amount of radiation to which the infant is exposed.

Diaphragmatic hernia and pneumothorax

Two particular intrathoracic disorders—diaphragmatic hernia and pneumothorax—warrant further discussion. *Diaphragmatic hernia* is the accepted term used to indicate a major defect in one or both domes of the diaphragm in a newborn baby and is not to be taken to include hiatus hernia (p. 140) or herniae through the foramina of Bochdalek or Morgagni. Developmentally, it represents persistence of the pleuro-peritoneal canal, allowing free communication between the pleural and peritoneal cavities. It is usually unilateral and occurs most commonly on the left side. The stomach, most of the small bowel, and a variable amount of the liver lie within the chest. The lung on the abnormal side is usually extremely hypoplastic and the other lung is compressed by the displaced heart.

It is therefore understandable that affected infants are dyspnoeic and cyanosed from birth. The apex beat of the heart may be detected on the right side of the chest if the hernia is on the left, and bowel sounds may be heard over the hernia. The location of the apex beat is of great diagnostic importance in this condition. If it is found to be on the right side it should not be attributed to dextrocardia; if the heart beat is felt on the right side in a dyspnoeic newborn infant, it has most probably been pushed or

pulled across from the left. In diaphragmatic hernia the chest X-ray picture is characteristic (Fig. 35) with gross mediastinal displacement and bowel shadows in the chest. It must be admitted that the outlook for these infants is

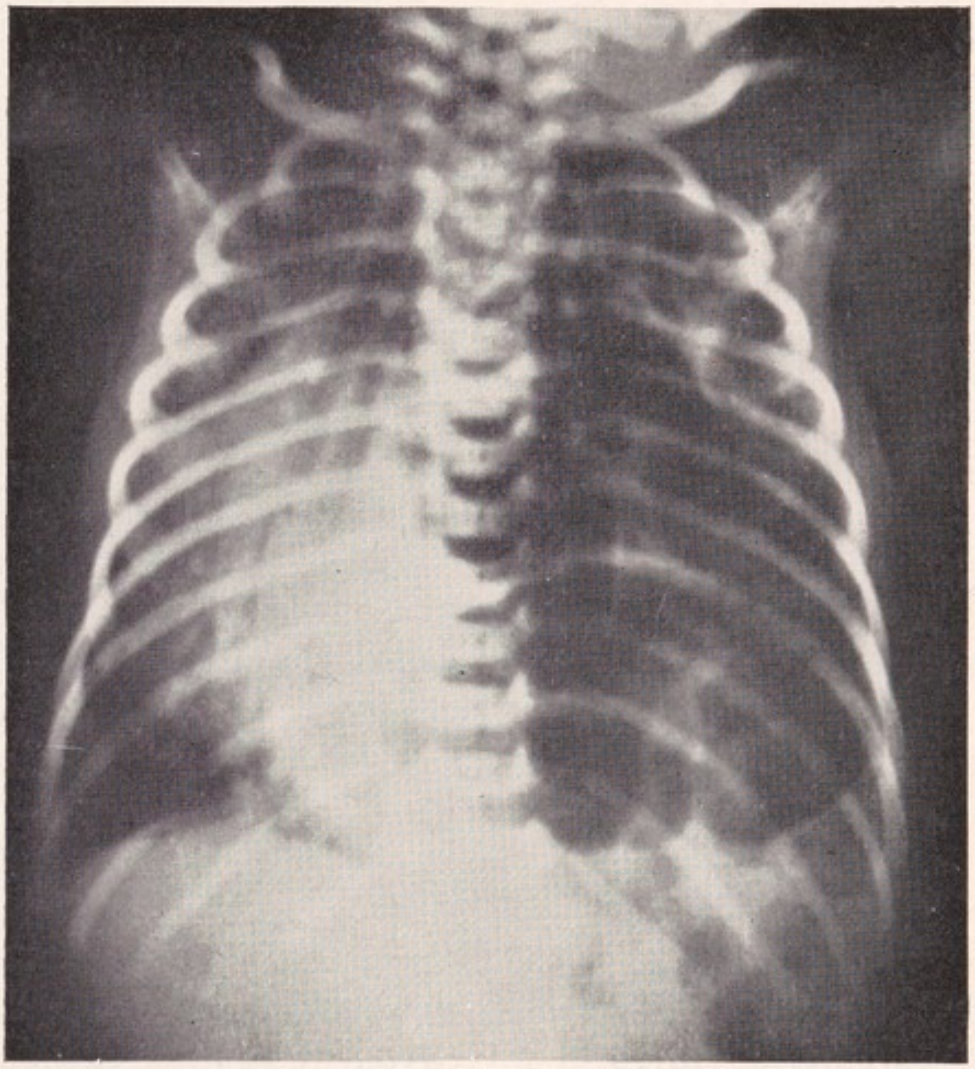


FIG. 35. Diaphragmatic hernia. The heart is displaced to the right. There are bowel shadows in the left hemithorax. The normal line of the diaphragm can be seen on the right, but not on the left.

poor, but prompt diagnosis and immediate operation gives them their only chance of survival. Apart from the technical difficulties of repairing such a large defect, there remains a hypoplastic lung, and in many cases there are other malformations.

Pneumothorax, although not strictly a congenital abnormality, may complicate such anomalies as cystic lung and congenital emphysema. It may also occur spontaneously, as a complication of endotracheal insufflation or in association with the respiratory distress syndrome (hyaline membrane disease). A simple pneumothorax may cause little trouble, and indeed a small one may be discovered unexpectedly on an X-ray film. A tension pneumothorax, however, is an emergency requiring urgent recognition and treatment. It occurs most commonly as a complication of endotracheal insufflation. Respiratory difficulty and cyanosis increase progressively, the mediastinum is displaced away from the affected side and the abdominal viscera are displaced downwards. On the right side, this may give the mistaken impression of hepatic enlargement, leading to a diagnosis of cardiac failure. The percussion note over the affected side is hyper-resonant, becoming almost dull as the tension increases. Breath sounds are absent or very distant. The condition may be confirmed radiologically (Fig. 36), but if a tension pneumothorax is suspected and the infant's condition is deteriorating, a hollow needle should be inserted through an intercostal space without delay.

CYANOSIS

Cyanosis in a newborn baby may be the result of respiratory, cardiac, or cerebral disorders. Cyanosis caused by respiratory disease is associated with dyspnoea. A cyanosed infant that is breathing without apparent difficulty is far more likely to have heart disease than lung disease. Cyanosis of cerebral origin is often associated with shallow or irregular breathing, although dyspnoea is occasionally seen; there is usually a history of difficult delivery or of foetal distress during labour. Cerebral damage may be shown by reduced activity, pallor, and a frown, or by wakefulness, restlessness, twitching, and convulsions. In irritable babies, cyanosis becomes more pronounced during

phases of hyperactivity. The administration of oxygen improves or abolishes cyanosis of pulmonary or cerebral origin. Rather surprisingly, it may also improve cyanosis resulting from a veno-arterial shunt.

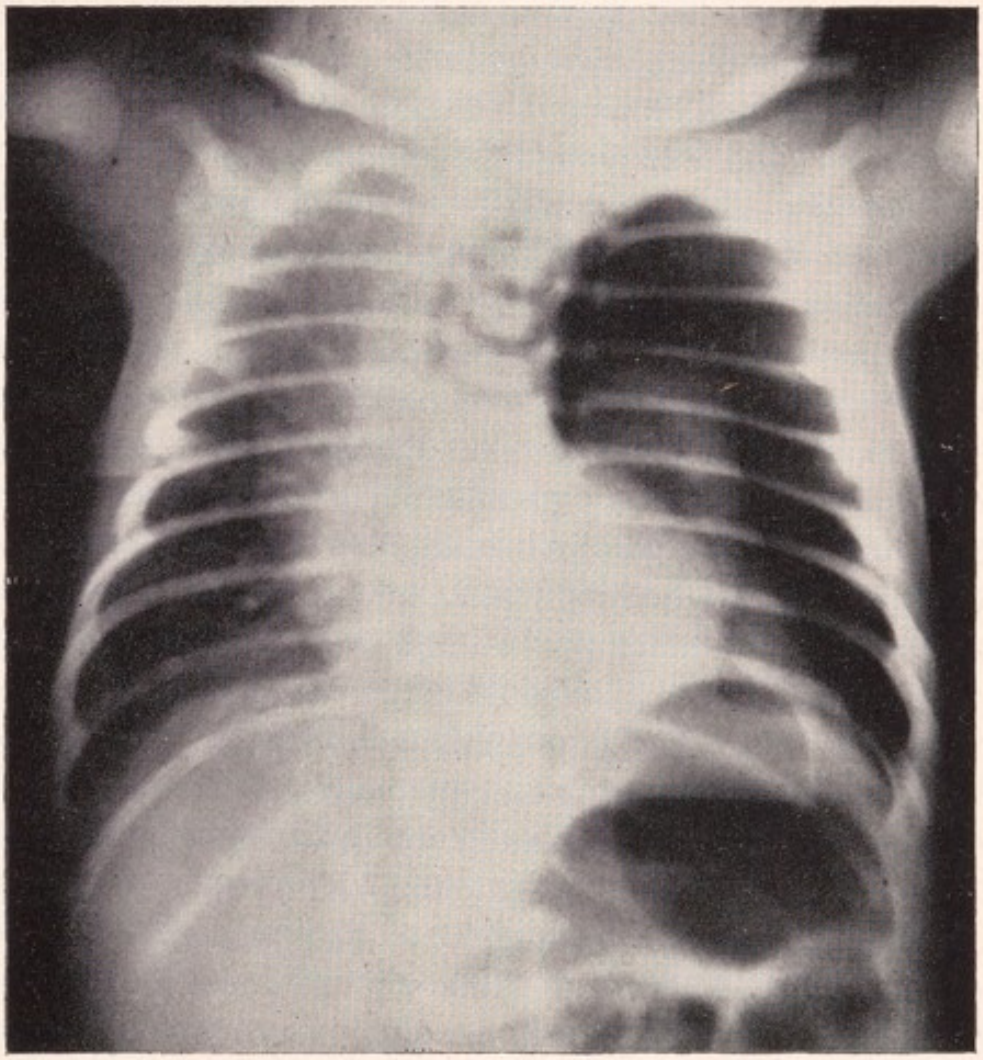


FIG. 36. Tension pneumothorax. The left lung is completely collapsed. The left pleural sac extends across the mediastinum. The heart is displaced to the right.

Continued cyanosis in an otherwise normal-looking baby usually indicates congenital heart disease. Unfortunately, when cyanosis of cardiac origin is apparent within a few hours of birth (or at birth) the underlying lesion is usually serious. *Bilocular* and *trilocular hearts*, *persistent truncus arteriosus*, *transposition of the great vessels*, and gross *anomalies of the pulmonary veins* may

present in this way. It is exceptional for infants with Fallot's tetralogy to be cyanosed in the first few days of life, and even some babies with pulmonary atresia remain pink for a little while. Cyanosis may also be due to severe cardiac failure which may complicate any lesion.

Examination of the heart in these infants may be disappointing. Cardiac enlargement is not as a rule sufficiently marked to be clinically detectable, although palpation in the xiphisternal region may reveal overactivity of the right ventricle. The absence of clinical enlargement is hardly surprising because most of these lesions cause no intrauterine difficulties. Murmurs are often absent when there is early cyanosis, whilst many lesions which characteristically cause murmurs are not associated with cyanosis. A small ventricular septal defect, for example, may produce a loud murmur but no disturbance of health. Absence of the interventricular septum may disturb health to the extent of killing the baby but may cause no murmur. It thus comes about that when a heart murmur is expected it is often absent, whereas most murmurs are heard in symptomless babies.

If cardiac enlargement is conspicuous in a newborn baby, whether demonstrated clinically or radiologically, there is likely to be a severe cardiac malformation or primary myocardial disease. Cyanosis is common in the first group and cardiac failure often complicates both, while heart murmurs are frequently absent. Myocardial diseases which may cause symptoms in the neonatal period include *endomyocardial fibroelastosis*, *glycogen-storage disease of the heart*, *myocarditis*, *rhabdomyoma of the heart*, and myocardial ischaemia caused by an *aberrant coronary artery* arising from the pulmonary artery. The differential diagnosis of these conditions is not always possible during life, but the electrocardiogram is sometimes helpful. The baby should always have the benefit of an expert opinion in case early operation offers hope of help.

Anomalous pulmonary venous drainage cannot always be diagnosed clinically as it often presents with the same

triad of cyanosis, cardiac enlargement, and cardiac failure, often without heart murmurs. Two clues occasionally point in the right diagnostic direction: if the veins from only one lung drain abnormally, a chest radiograph may show a difference in the degree of vascularization of the two lungs; if the anomalous veins drain below the diaphragm into the hepatic veins or the inferior vena cava, there is likely to be jaundice and enlargement of the liver. The combination of cyanosis and jaundice produces a very striking colour. It is not peculiar to anomalous pulmonary venous drainage, but in combination with cardiac and hepatic enlargement it makes this a likely diagnosis.

A clinical suspicion of congenital heart disease, however normal the heart feels and sounds, will prompt a chest radiograph and an electrocardiogram. The latter is often normal, even when there is a severe heart lesion, but it is nevertheless worth doing. It serves as a base-line with which to compare future tracings and it is sometimes of diagnostic help. The chest film is more rewarding but great caution must be felt in its interpretation, and the general precautions discussed on page 75 must be observed. The apparent size and shape of the infant's heart on a radiograph vary with his posture, with the phases of respiration and the cardiac cycle, and with the height of the diaphragm, which is influenced by the fullness of the stomach. There are therefore wide variations of cardiac contour within the range of normal. The vascularity of the lung fields also gives useful information, provided, again, that the limits of normal variation are appreciated.

Cardiac failure in the neonatal period is usually a clear indication of a cardiac lesion except in the presence of severe respiratory distress (including that due to pneumonia) or of profound anaemia. The clinical picture of cardiac failure at this age bears little relation to that seen in the adult: the heart is not necessarily enlarged; tachycardia is usual but not invariable; oedema is a late sign. The most reliable signs are dyspnoea (which may lead to feeding difficulties), enlargement of the liver, cough and crepitations

at the lung bases. The size of the liver should be assessed at least once a day in all sick infants with cardiac disease.

Cyanotic attacks. It may at first appear paradoxical that the principal causes of continued cyanosis are only exceptionally responsible for cyanotic attacks. On reflection, however, it is clear that the gross cardiac and pulmonary lesions that so often underlie continued cyanosis can never be associated with normal tissue oxygenation. Cyanotic attacks are almost always due either to temporary obstruction of the upper respiratory tract or to defective respiration consequent upon cerebral disorders. Cyanotic attacks are particularly common in premature infants, because they are especially prone to difficulties in both categories. Immature coughing and swallowing reflexes allow secretions to pool in the pharynx and obstruct the airway. Immaturity of the respiratory centre results in periods of apnoea which, if sufficiently prolonged, lead to cyanosis. Similar apnoeic attacks may be seen in any infant with cerebral disturbance, and the soft brain of the premature infant is far more likely to be damaged than that of the full-term infant.

Reference has already been made to choanal atresia and micrognathia (p. 119), both of which may be responsible for cyanotic attacks. Rarely, enlargement of the tongue or large cysts within the mouth may cause similar trouble.

Cyanotic attacks with feeds are of special significance. They are likely to be seen in association with the following conditions:

(1) Nasal obstruction (choanal atresia or stenosis; rhinitis).
(2) Weakness of the swallowing muscles (prematurity; 'bulbar palsy').
(3) Reversible intracardiac shunts (for example, atrial septal defect).
(4) Oesophageal atresia and oesophagotracheal fistula.

Reversible veno-arterial shunts include many cases of

atrial septal defect and some examples of *patent ductus arteriosus*. Provided that the pressure difference between the two communicating chambers is small, a shunt that is from the arterial side to the venous side (and therefore not associated with cyanosis) when the infant is at rest may be reversed by a respiratory effort which raises the pulmonary arterial pressure. Feeding, crying, and intercurrent respiratory infections may all produce temporary cyanosis in infants with this type of lesion.

Oesophageal atresia can usually be diagnosed before the infant is fed (*see* p. 136). He may have bubbly breathing and cyanotic attacks from the overspill of pharyngeal secretions into the trachea. If the diagnosis has not been made earlier, the first few mouthfuls of the first feed will precipitate such dramatic symptoms that the diagnosis becomes apparent. Milk rapidly fills the blind oesophageal pouch and then runs over into the lungs causing instant distress, cyanosis, and choking.

The rare condition of oesophagotracheal fistula without oesophageal atresia is responsible for some cyanotic spells of obscure origin. Because the oesophagus is patent, a tube can be passed into the stomach without difficulty and cyanotic attacks may occur with some feeds but not with others. Attempts to demonstrate a fistula radiographically may not succeed, and even at oesophagoscopy it may not be seen. Cyanotic attacks which occur with oral feeds but not with feeds given by gastric tube suggest a fistula, but may also occur if there is some defect of the swallowing mechanism (*see* p. 132).

If an otherwise healthy baby has apparently inexplicable cyanosis, it is worth bearing in mind the possibility of methaemoglobinaemia, rare though this condition is. The hereditary variety may present in the newborn period. Acquired methaemoglobinaemia has been described in newborn infants, chiefly as a result of poisoning by nitrates or aniline. Nitrate poisoning, now very rare, arises from the ingestion of well-water with a high nitrate content. Aniline poisoning has been reported when napkins marked

with Indian ink and used before being washed have been in contact with excoriated buttocks, the dye being absorbed through the skin. The suspicion of methaemoglobinaemia may be aroused by the sight of a deeply cyanosed infant that is otherwise well, without dyspnoea and without signs of cardiac, respiratory, or cerebral disease. The diagnosis is established by spectrographic examination of the blood.

DIFFICULTY WITH SWALLOWING

Difficulty with feeding may give the first clue to the presence of a congenital abnormality. It may be difficult to distinguish between the infant that has a mechanical obstruction to swallowing and the infant that has a defective swallowing mechanism. However, mechanical obstruction to respiration by choanal atresia or micrognathia, which will cause feeding difficulty, should be readily recognizable, and oesophageal atresia can be excluded (*see* p. 136).

If the swallowing mechanism is defective, the fault may be on the sensory side, but more usually there is muscle weakness. Such weakness is normal in small premature babies and may be seen in infants who have suffered perinatal brain damage. Bulbar palsy may occur as an isolated defect or it may be a feature of Möbius's syndrome, being sometimes associated with weakness of muscles supplied by the facial or other cranial nerves and, often, with skeletal deformities, particularly of the hands and feet. The dysphagia of bulbar palsy and Möbius's syndrome tends to improve very slowly and ultimately to disappear, but tube-feeding may be necessary for many months.

Defective swallowing may also be a feature in any infant with generalized muscular hypotonia. This is an important symptom and is discussed in the next section.

THE FLOPPY INFANT

Generalized hypotonia is physiological in small premature babies, but may be the result of systemic disease (especially some metabolic disorders), of muscular disease, or of disorders of the nervous system. Many of the metabolic and muscular disorders, and some of the neurological diseases, are genetically determined, although they are not necessarily apparent at birth. Amongst those which present after the neonatal period are infantile spinal muscular atrophy (Werdnig-Hoffman disease) and muscular dystrophy, both of which are genetically determined. Polymyositis, polyneuritis, and poliomyelitis may also occur in infancy.

Muscular hypotonia is found in many infants that have sustained cerebral birth injury and is an integral part of the clinical picture of white asphyxia. Hypotonia is a constant finding in mongolism and reference has already been made to this as a diagnostic test in cases of doubt (p. 56). There is often diffuse muscle flaccidity in infants with Möbius's syndrome, and in the Ehlers-Danlos syndrome (cutis laxa) hypotonia is combined with excessive mobility of the joints and laxity of the skin. Glycogen-storage disease of the skeletal muscles may present in the neonatal period; if the tongue is involved it is enlarged and provides a useful diagnostic pointer.

If hypotonia results from perinatal brain injury, it begins to improve within a few days, except in the most severe cases. In Möbius's syndrome the improvement is very much more gradual, and in glycogen-storage disease the floppiness tends to increase as time passes.

There remains one other important cause of hypotonia in the newborn baby, namely, *myasthenia gravis*. At this age it may occur in two forms: the first is comparable with the disease seen in adults and is extremely rare; the second is transient neonatal myasthenia, which occurs as a temporary phenomenon in some infants born to mothers with myasthenia gravis. Only a minority of babies born to

myasthenic mothers, about 20 per cent., are thus affected. Infants with transient myasthenia are limp at birth or, more usually, become limp within a few hours. Sucking and swallowing become sluggish or absent and breathing is shallow. Inhalation of feeds occurs readily and may be fatal.

In both varieties of myasthenia gravis the diagnosis is established by the dramatic response to edrophonium chloride (Tensilon), of which 0·1 ml. should be injected intramuscularly. This drug is far better than neostigmine for a diagnostic test, because it acts more quickly and a response is obvious within ten minutes of the injection. Muscle tone improves, respirations become stronger, and sucking and swallowing may be restored to normal. This response is seen in no other condition. Early diagnosis is important because myasthenic infants with symptoms of bulbar palsy are greatly helped by treatment with neostigmine (Prostigmin) or pyridostigmine (Mestinon).

VOMITING AND REGURGITATION

Vomiting and regurgitation in the newborn infant will immediately bring to mind the possibility of intestinal obstruction. This is as it should be, although the suspicion will only be confirmed in a minority of instances. Vomiting is a symptom that can never be neglected and should prompt a thorough examination of the whole infant—not just the abdomen.

In some of these infants the vomiting or regurgitation is associated with swallowing of much mucus or amniotic fluid. This is particularly common in babies delivered by Caesarean section. Aspiration and washing out the stomach will often relieve the vomiting in these cases. Frequent regurgitation of small amounts of fluid is not uncommon in infants with cerebral disturbance. It is maximal in the first day or two and improves spontaneously.

Vomiting may be associated with infection, enteral or parenteral, at this as at any other age. It is this possibility in particular that prompts examination of the whole infant. The diagnosis of internal infection may be very difficult in the neonatal period, especially in prematures. Fever and leucocytosis are often conspicuous by their absence. Microscopy of the urine and radiography of the chest are indicated if the cause of the vomiting is not obvious.

Vomiting resulting from feeding difficulties is rarely noted in the first two or three days of life, and is therefore unlikely to be confused with that due to intestinal obstructions. Regurgitation of feeds is especially common in premature infants.

In considering the possibility of intestinal obstruction, attention will be paid to the following points in the history:

(1) At what age did the vomiting start? The higher the obstruction, the earlier the vomiting. With oesophageal atresia there is trouble with the second swallow, with duodenal atresia at the second feed, and with rectal atresia on the second day.

(2) What does the vomitus look like? With hiatus hernia the vomitus is milk, sometimes containing small amounts of altered blood. In duodenal obstruction, whatever the underlying lesion, the vomitus is usually bile-stained, because the obstruction is nearly always below the ampulla of Vater. With obstruction below this level, the vomitus may be faeculent.

(3) Has the infant passed meconium? The significance of this has already been discussed (p. 91), but it is worth repeating that the passage of meconium does not exclude complete intestinal obstruction.

(4) Is there any relevant family history? This may be helpful in meconium ileus (with or without meconium peritonitis) and in some examples of Hirschsprung's disease.

In the examination of the abdomen, a particular search is made for distension, visible peristalsis, and palpable masses. Distension is usual with obstruction but is by no

means invariable. Furthermore, it may be quite marked in normal infants that are not vomiting, especially just after a feed. It tends to be confined to the upper part of the abdomen in duodenal obstruction, although if diagnosis is delayed the stomach may become enormously distended so that the whole abdomen is swollen. With lower obstructions, distension appears somewhat later but involves the whole abdomen. It is progressive if the obstruction is not relieved.

Visible peristalsis is commonly seen in healthy premature infants, because the abdominal wall is thin, and it may be seen in some full-term infants that have little subcutaneous fat. Gastric peristalsis runs across the epigastrium from left to right. Small bowel peristalsis forms a 'ladder' pattern, the coils lying more or less horizontally across the centre of the abdomen.

Palpable masses are rarely found. In meconium ileus, faecal masses may be felt; in other infants some non-obstructive lesion, such as polycystic kidneys, may be found to explain the distension. If there is obstruction, palpation may reveal gurgling.

Examination of the abdomen is completed by inspection of the anus and digital examination of the rectum. The latter may be of therapeutic benefit if obstruction is caused by meconium plugs (p. 149).

Atresia of the oesophagus

Atresia of the oesophagus can often be suspected before birth because of its common association with hydramnios (p. 6). If a woman with an excess of amniotic fluid gives birth to an apparently normal infant, a tube should be passed into the stomach without delay. The tube should not be very fine or very soft, otherwise it may coil up in the upper oesophageal pouch (Fig. 37) and the examiner will mislead himself into thinking that the end must have reached the stomach. The tube can be introduced through the nose or over the tongue. The procedure

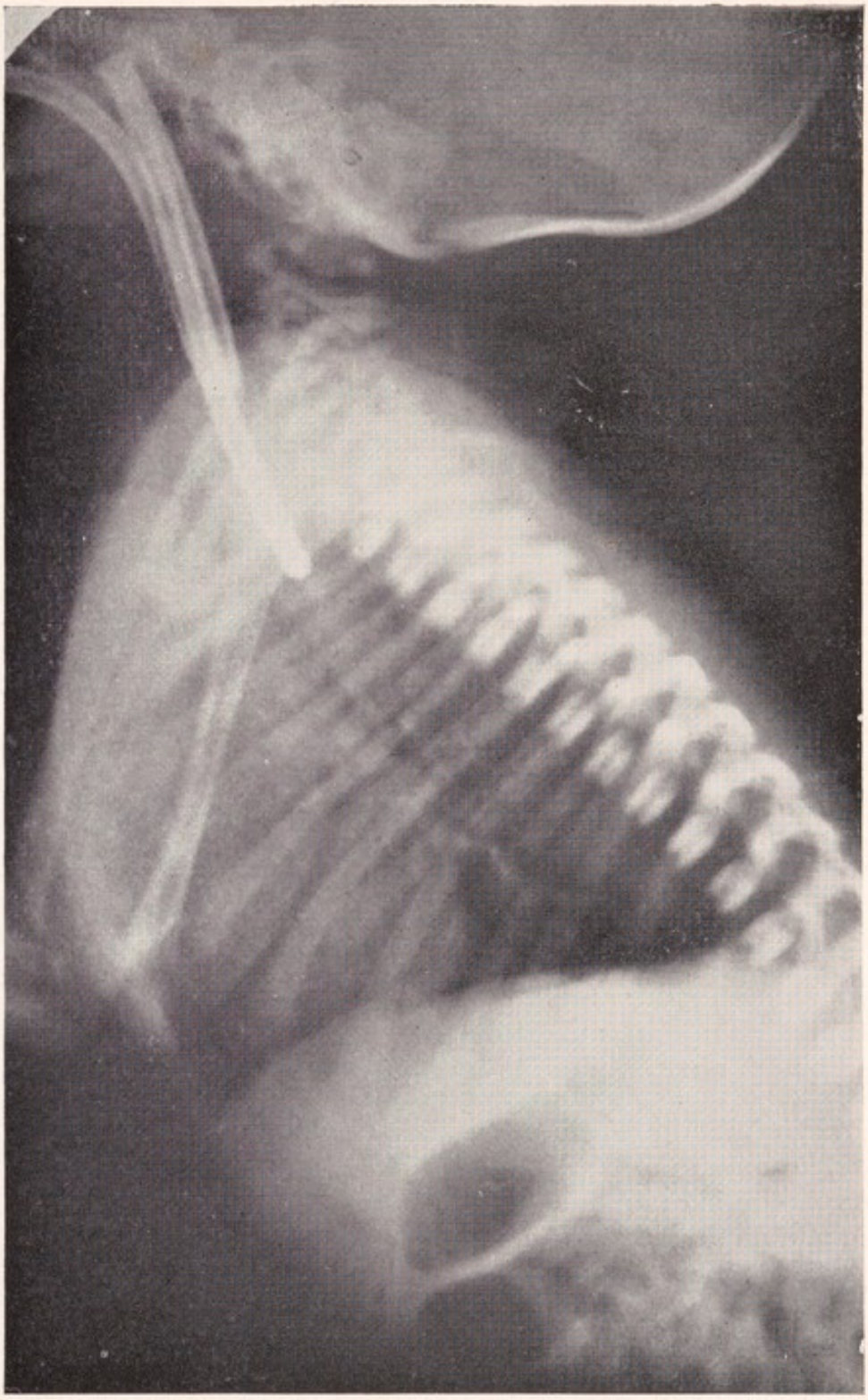


FIG. 37. Oesophageal atresia. A soft catheter has been used and has doubled up in the oesophageal pouch. The presence of air in the bowel shows that there is an oesophago-tracheal fistula.

should be gentle, and the tube will slip down easily if the oesophagus is patent. A dry tube slips down less easily than a wet one, but oils such as liquid paraffin should not be used as lubricants lest some should be aspirated into the lungs.

If there is any doubt as to whether the tube has entered the stomach, air should be blown gently down the tube with a syringe whilst the examiner listens with a stethoscope over the epigastrium. The air can readily be heard bubbling in. This simple test will sometimes obviate the necessity for an X-ray.

In the absence of hydramnios, the first clue to oesophageal atresia is the excessive amount of mucus which accumulates in the infant's pharynx, necessitating repeated sucking out. This symptom is also seen in infants with defective swallowing mechanisms, but such conditions are far more rare than oesophageal atresia. Spilling over of pharyngeal secretions into the trachea may cause cyanotic attacks or choking.

If these symptoms are not reported, or if their significance is overlooked, the truth should become apparent the first time the baby is fed. The first mouthful or two of milk fills the upper oesophageal pouch and thereafter it spills over into the trachea causing choking, cyanosis, and great distress. If this situation is reached, there is a grave danger of pneumonia developing; every endeavour should be made to establish the diagnosis before the baby is ever fed.

Apart from the findings when a stomach tube is passed, clinical examination is often negative. In something like 85 per cent. of cases there is a fistulous connection between the lower part of the oesophagus and the trachea, allowing air to enter the gastrointestinal tract. In most of the remainder, however, there is atresia without a fistula: in these cases there is no gas in the bowel (Fig. 38) and the scaphoid appearance of the abdomen, which resembles that of a stillborn baby, immediately suggests the diagnosis.

It should be emphasized that about half of all infants with

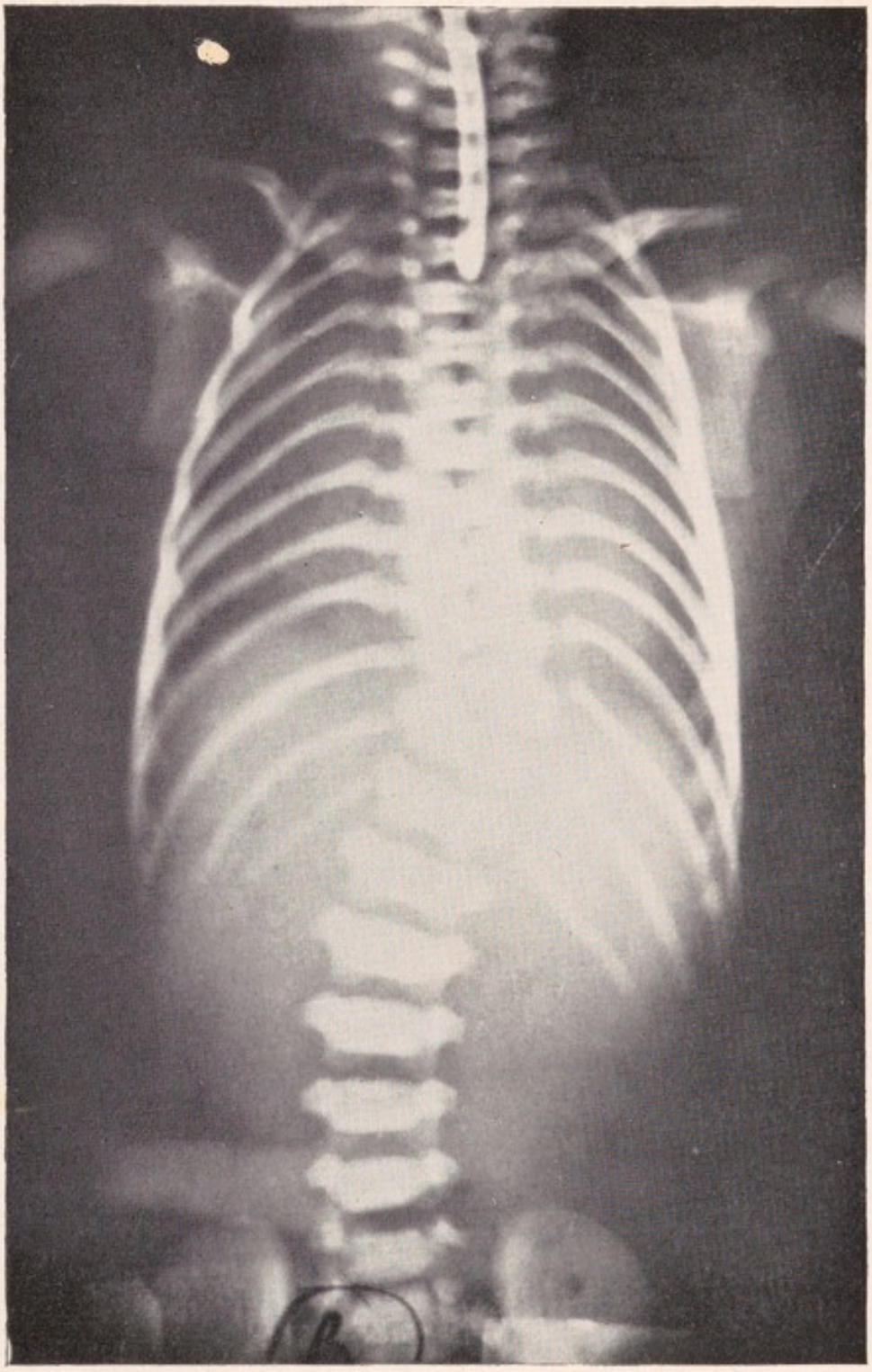


FIG. 38. Oesophageal atresia without fistula. The catheter is held up in the upper oesophageal pouch. The absence of air in the abdomen shows that there is no oesophago-tracheal fistula. There is an associated malformation of the spine.

oesophageal atresia have other serious internal malformations. Some of these are incompatible with life, which accounts for the apparently disappointing results of surgery even in the most competent hands, but others are curable. There is a particular association between atresias of the oesophagus and rectum, and the diagnosis of one should prompt a search for the other.

Radiography may be used to confirm the diagnosis and to demonstrate for the surgeon the size and position of the upper pouch. A radio-opaque tube may be inserted as far as it will go with gentle pressure, and a lateral or oblique film taken to show its position. Alternatively, a contrast medium may be introduced into the upper oesophageal pouch to delineate it more exactly (Fig. 39). Only a few ml. of medium are necessary and it should be aspirated after the films have been taken. Only contrast media suitable for bronchography should be used, because, if atresia is present, some contrast medium is almost bound to spill over into the bronchial tree. Under no circumstances whatever should barium be given to an infant, if there is the least doubt about the patency of the oesophagus, because it is injurious to the lungs.

Hiatus hernia

Hiatus hernia often causes no conspicuous symptoms in the neonatal period but in some instances vomiting or regurgitation is noted from birth. The vomitus may contain a little altered blood. Abdominal distension is not seen, and constipation occurs only if the vomiting is exceptionally severe. The diagnosis is made by barium meal, the baby being screened in the Trendelenberg position. Early diagnosis is important, because early treatment by nursing the infant upright may prevent a troublesome oesophagitis. If reflux oesophagitis develops, it may lead to the formation of a stricture (Fig. 40). Hiatus hernia is sometimes associated with pyloric stenosis.

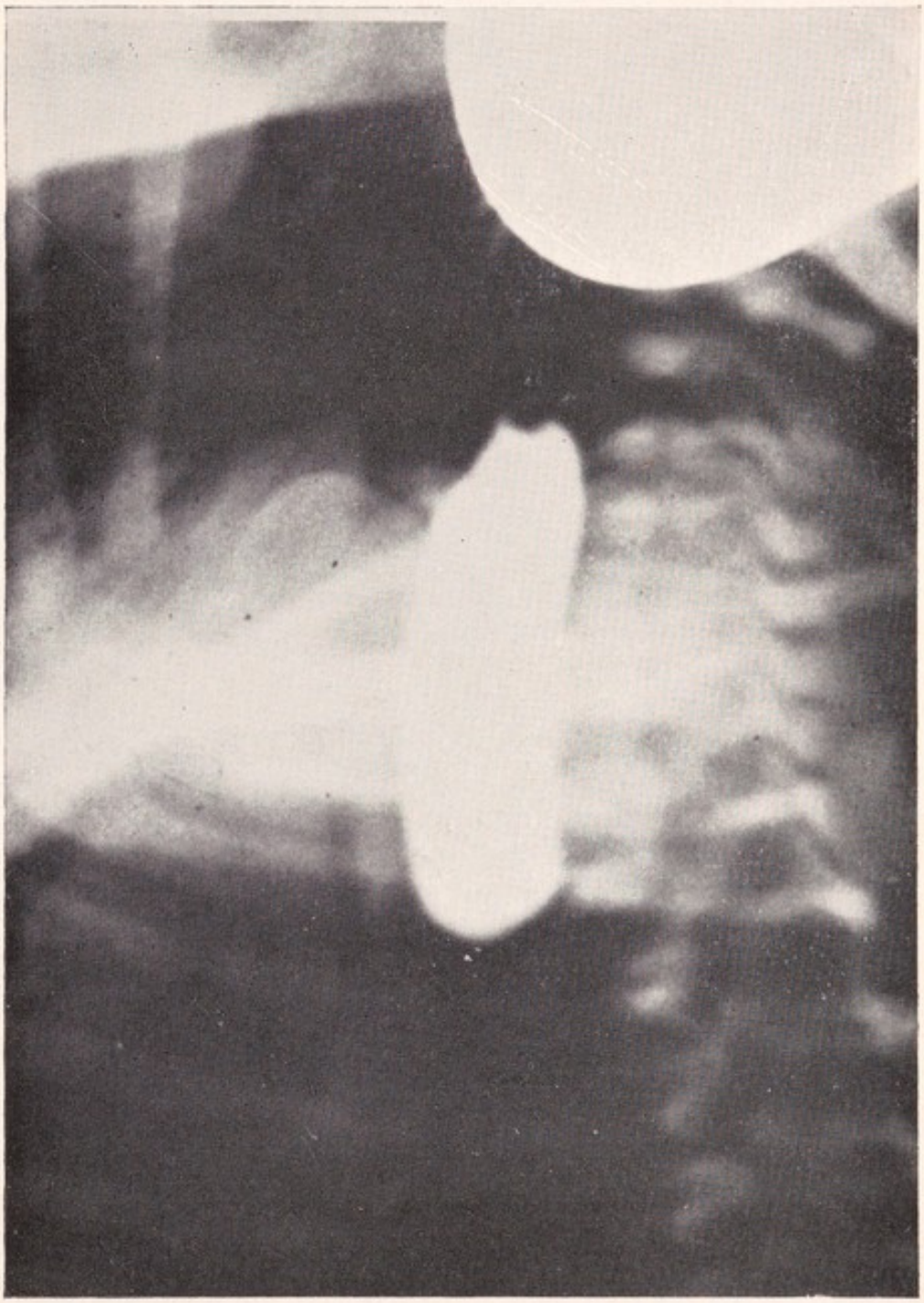


FIG. 39. Oesophageal atresia. The pouch has been filled with Lipiodol.

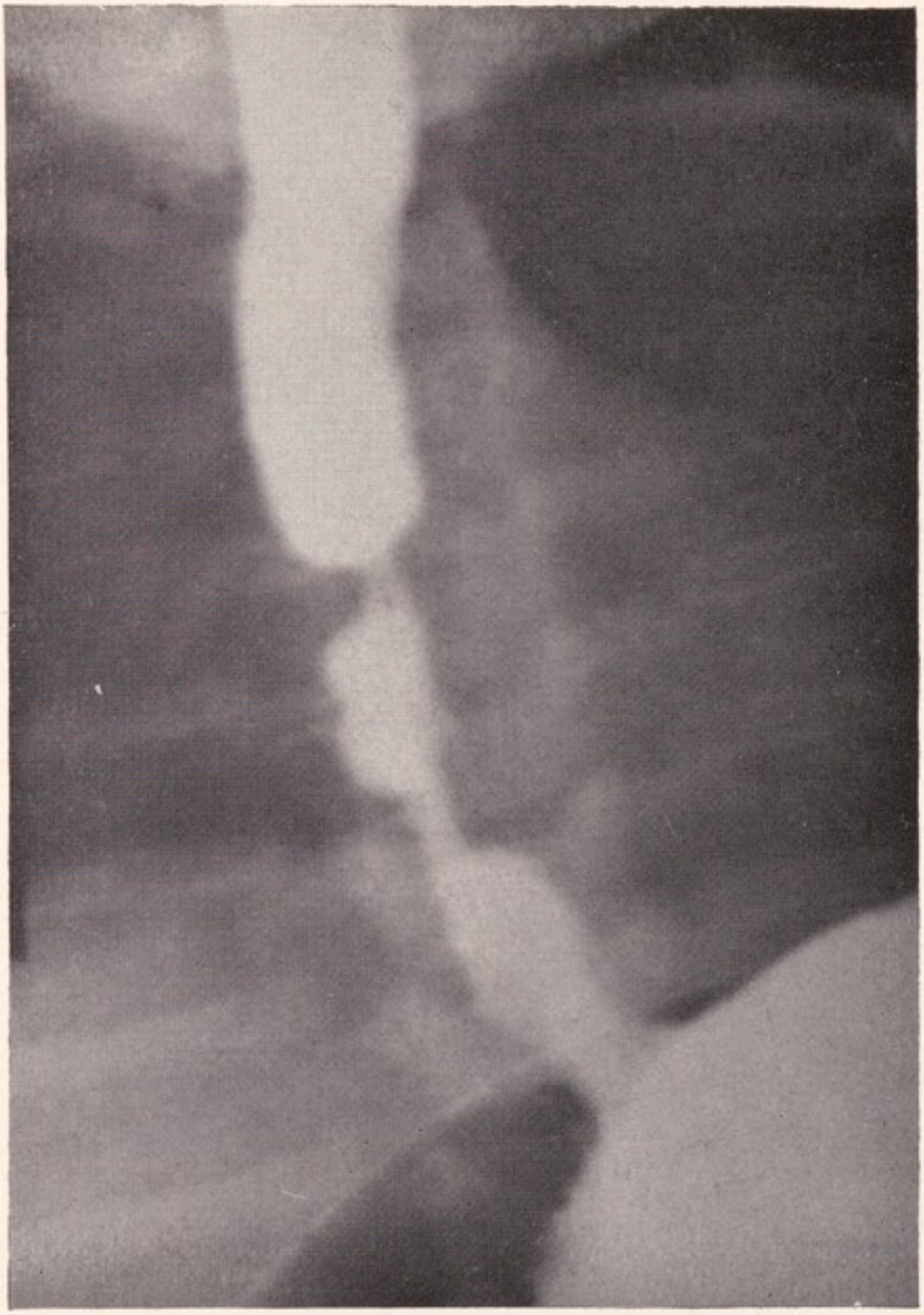


FIG. 40. Hiatus hernia with oesophageal stricture. The upper constriction in the oesophagus is an inflammatory stricture. The lower constriction is the oesophagogastric junction.

Duodenal obstruction

Vomiting may be due to duodenal obstruction which may arise from one of several causes. The clinical picture is the same in all, namely, bilious vomiting, upper abdominal distension, and constipation. Duodenal obstructions are

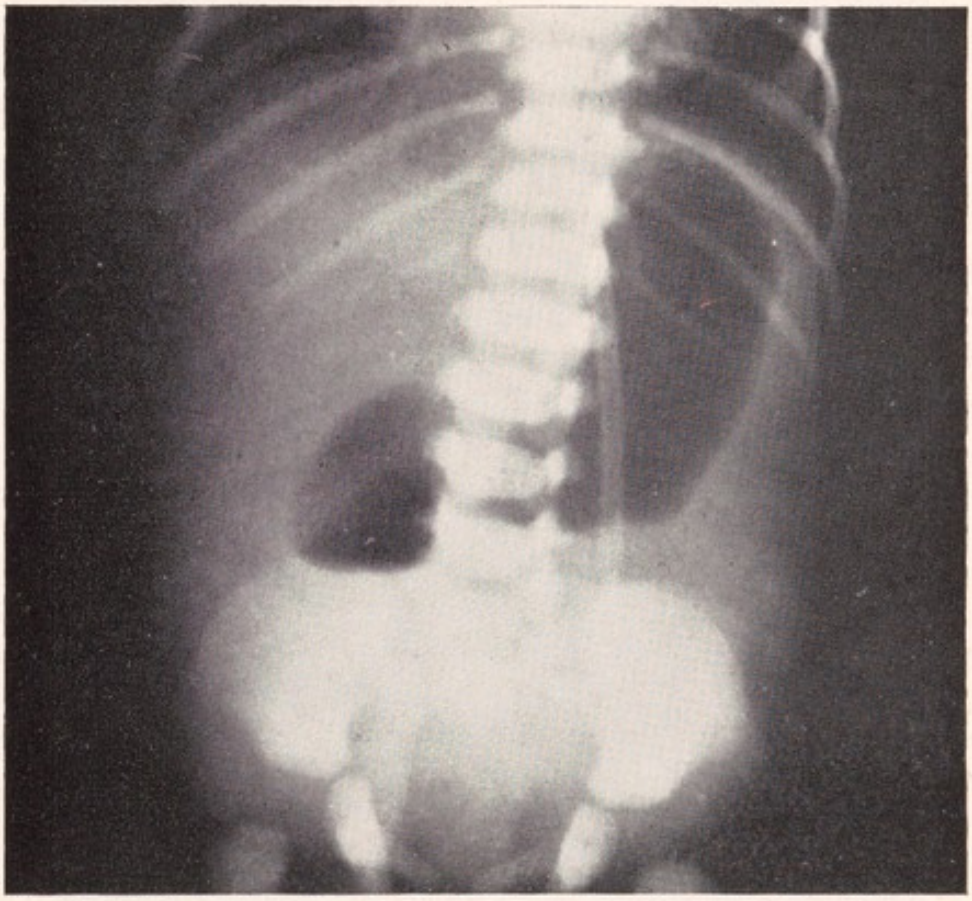


FIG. 41. Complete duodenal obstruction. The stomach and proximal duodenum are distended, forming two large gas shadows with fluid levels. There is no gas beyond. A tube has been passed into the stomach.

almost always distal to the ampulla of Vater and the vomitus, therefore, usually contains bile. The vomiting usually starts after the second or third feed and is thereafter continuous. The abdomen swells and the swelling is principally above the umbilicus. A straight X-ray film taken with the infant upright shows two gas shadows with fluid levels, one in the stomach and the other in the first part of the duodenum. This is the 'double bubble' sign (Fig. 41)

If the obstruction is incomplete, a few small gas shadows may be seen beyond the duodenum (Fig. 42).

It is scarcely ever possible for the physician to decide the

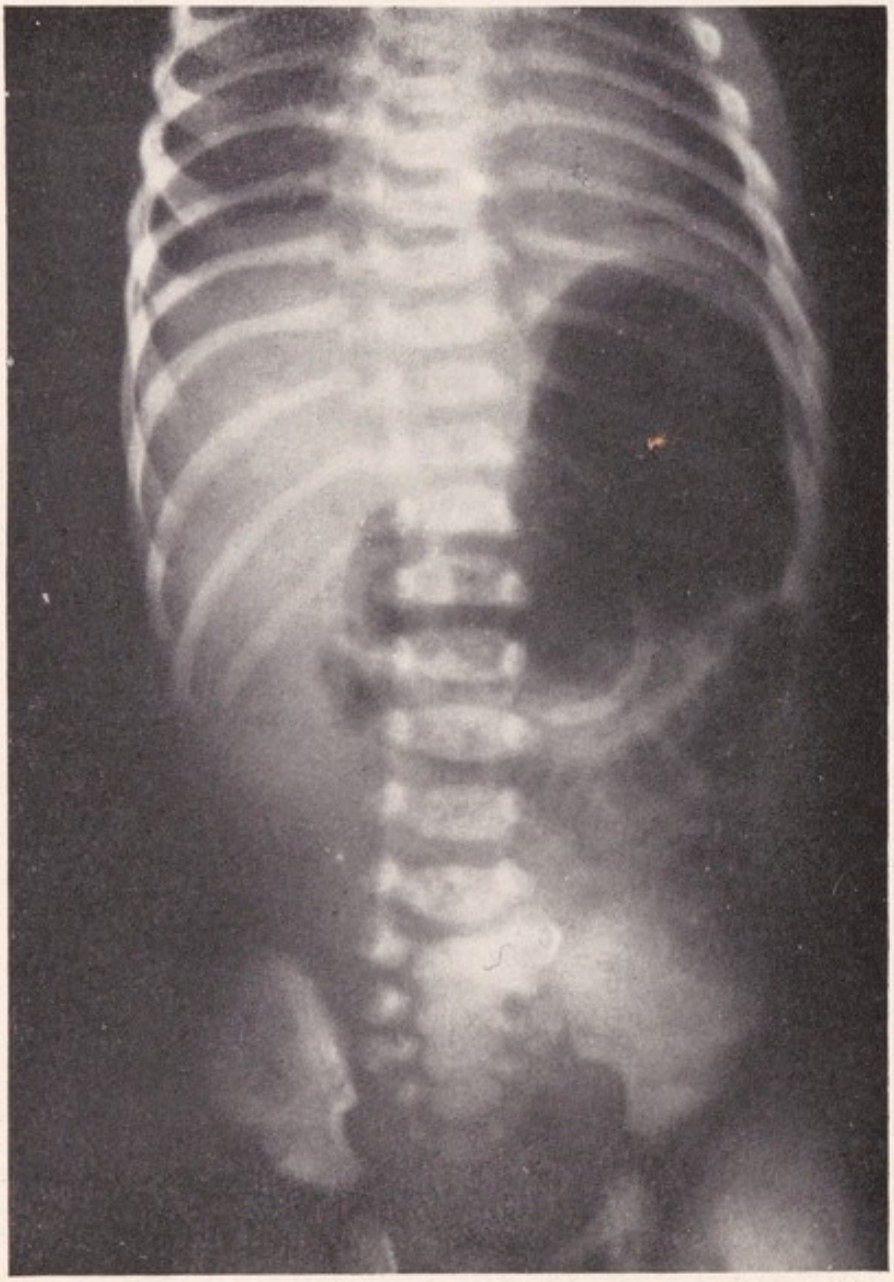


FIG. 42. Incomplete duodenal obstruction. The stomach is distended but there is some gas in the small bowel.

nature of the lesion in any particular case. This does not matter, because operative treatment will inevitably be needed. The probabilities favour duodenal atresia in

babies that are otherwise healthy, and stenosis in mongols. Some prove at operation to be due to malrotation of the intestine, and others to annular pancreas.

Obstruction of the small bowel

Vomiting due to obstruction of the small bowel is usually associated with generalized abdominal distension. The obstruction is most commonly due to meconium ileus. This condition is an early manifestation of fibrocystic disease of the pancreas (mucoviscidosis), the meconium being so abnormally viscid that the peristaltic action of the bowel is incapable of pushing it onwards. The obstruction is most often in the terminal ileum and is usually incomplete, so that multiple, small bowel fluid levels are seen on the X-ray (Fig. 43). The radiological appearances and a history of fibrocystic disease in older siblings will strongly suggest the diagnosis, but it can only be established with certainty by laparotomy.

Less commonly, small bowel obstruction may be due to ileal or jejunal atresia (Fig. 44). In this case the obstruction is complete, there is no air in the lower bowel, and the family history is usually negative. However, the author has seen an infant with ileal stenosis associated with protrusions of a Meckel's diverticulum into the umbilical cord, the previous sibling having had identical lesions.

Meconium ileus is sometimes associated with the even more grave condition of meconium peritonitis. This is a chemical peritonitis resulting from prenatal perforation of the foetal bowel, and leads to dense, fibrotic adhesions within the abdomen. These adhesions undergo patchy calcification, usually before birth, thus simplifying diagnosis (Fig. 45). When the abdomen is opened, the immediate impression is that the intestines are congenitally absent, for they are quite invisible, buried behind a wall of fibrous tissue. Meconium peritonitis is not always the result of mucoviscidosis, but this is the most common condition underlying the perforation. The bowel of the newborn

baby becomes colonized with bacteria within a few hours of birth; this will immediately convert a chemical into a bacterial peritonitis. It is, therefore, imperative that

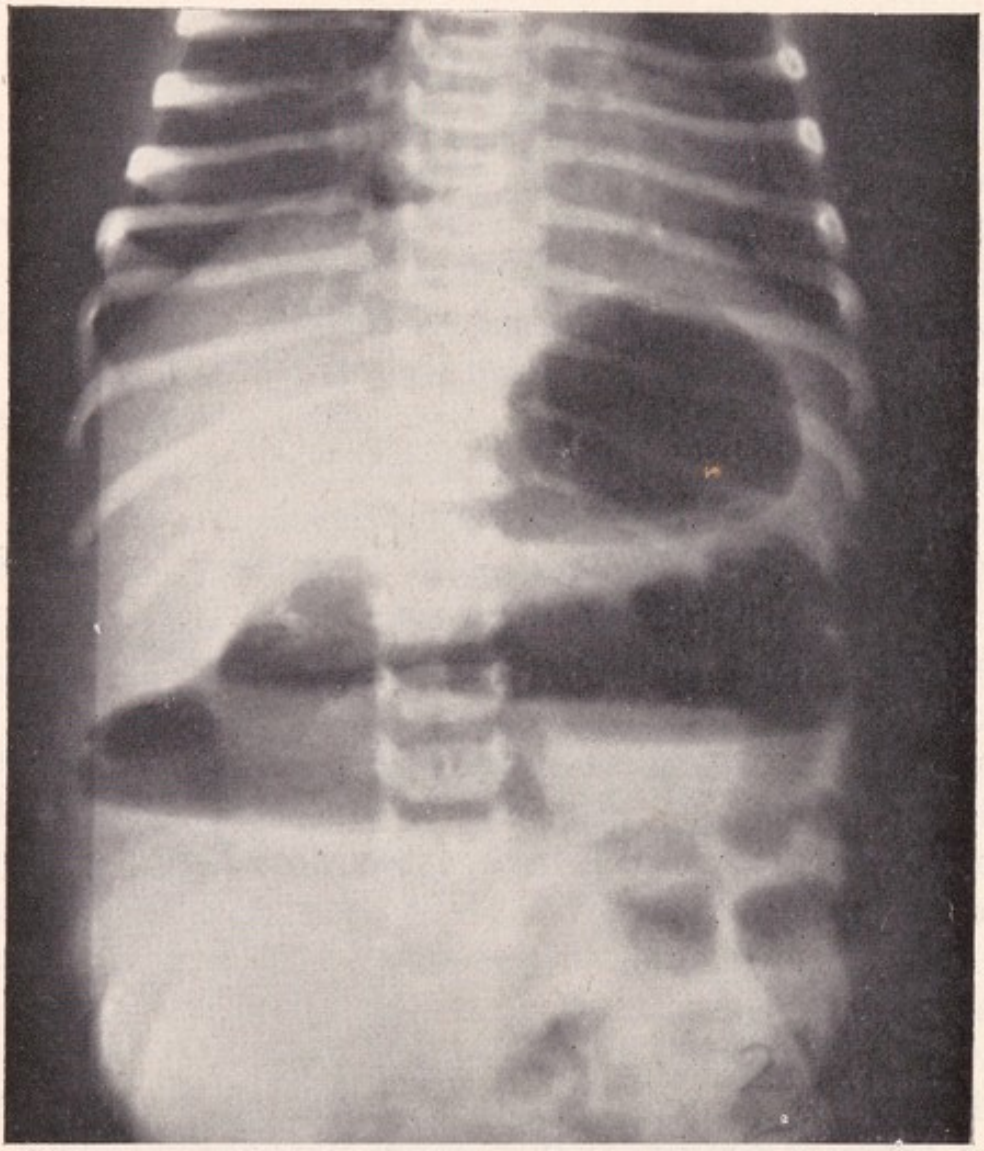


FIG. 43. Meconium ileus. There are multiple fluid levels in the small bowel. There is a little calcification to the right of the first lumbar vertebra, suggestive of meconium peritonitis (*see* Fig. 45).

diagnosis be made really early, if the surgeon is to have any chance at all of dealing successfully with this very difficult condition.

Amongst the other causes of small bowel obstruction are internal hernias and duplication cysts, but these can only be diagnosed at operation. The physician's task is to

recognize at the earliest possible moment that the bowel is obstructed; the radiologist can usually determine the level of the obstruction; and the surgeon will establish the nature of the underlying lesion.

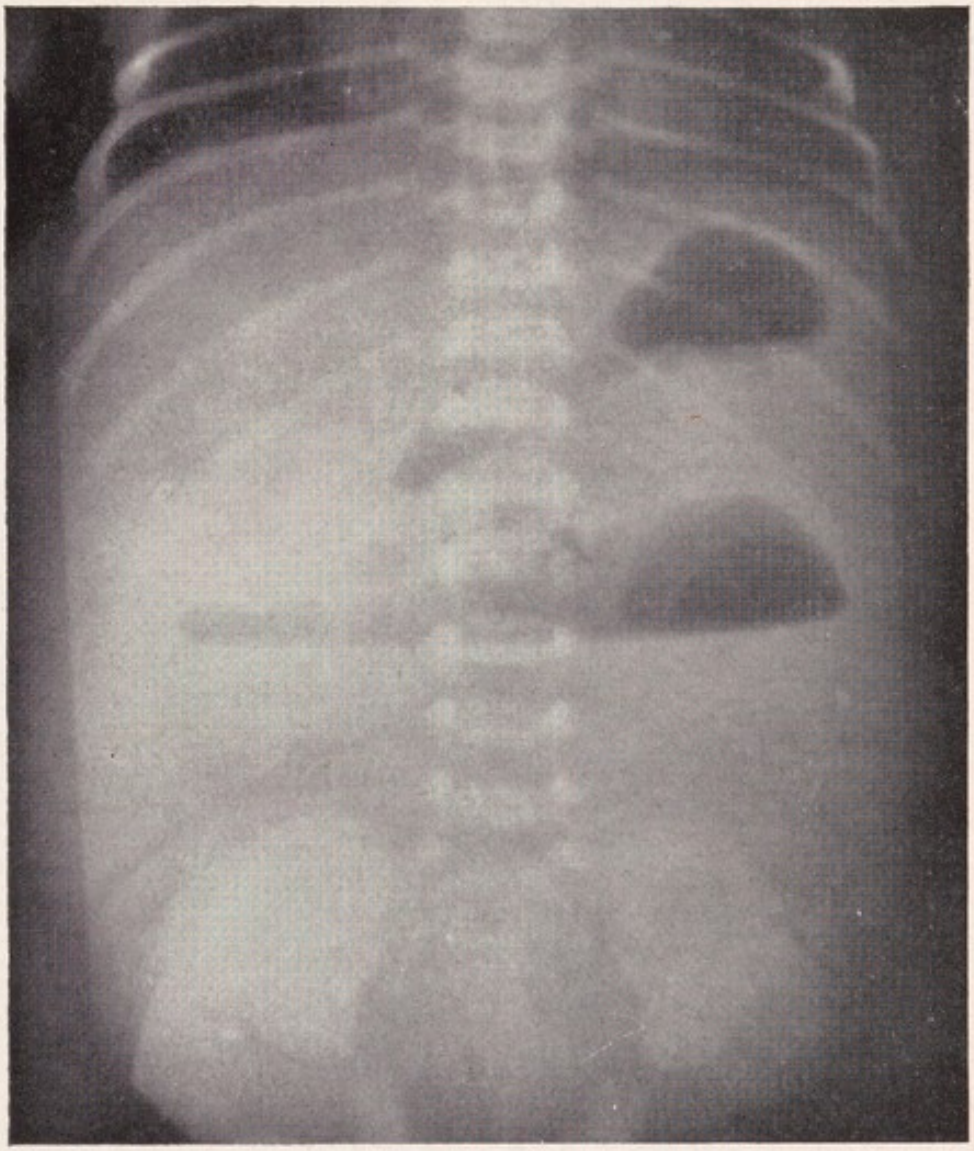


FIG. 44. Small bowel obstruction. In this example the cause was ileal atresia.

Obstruction of the large bowel

Obstruction of the large bowel also gives rise to vomiting and generalized abdominal distension. These symptoms tend to appear rather later than with obstruction of the small bowel. Atresias of the colon are rare. If there is complete obstruction of the rectum or anus, meconium is

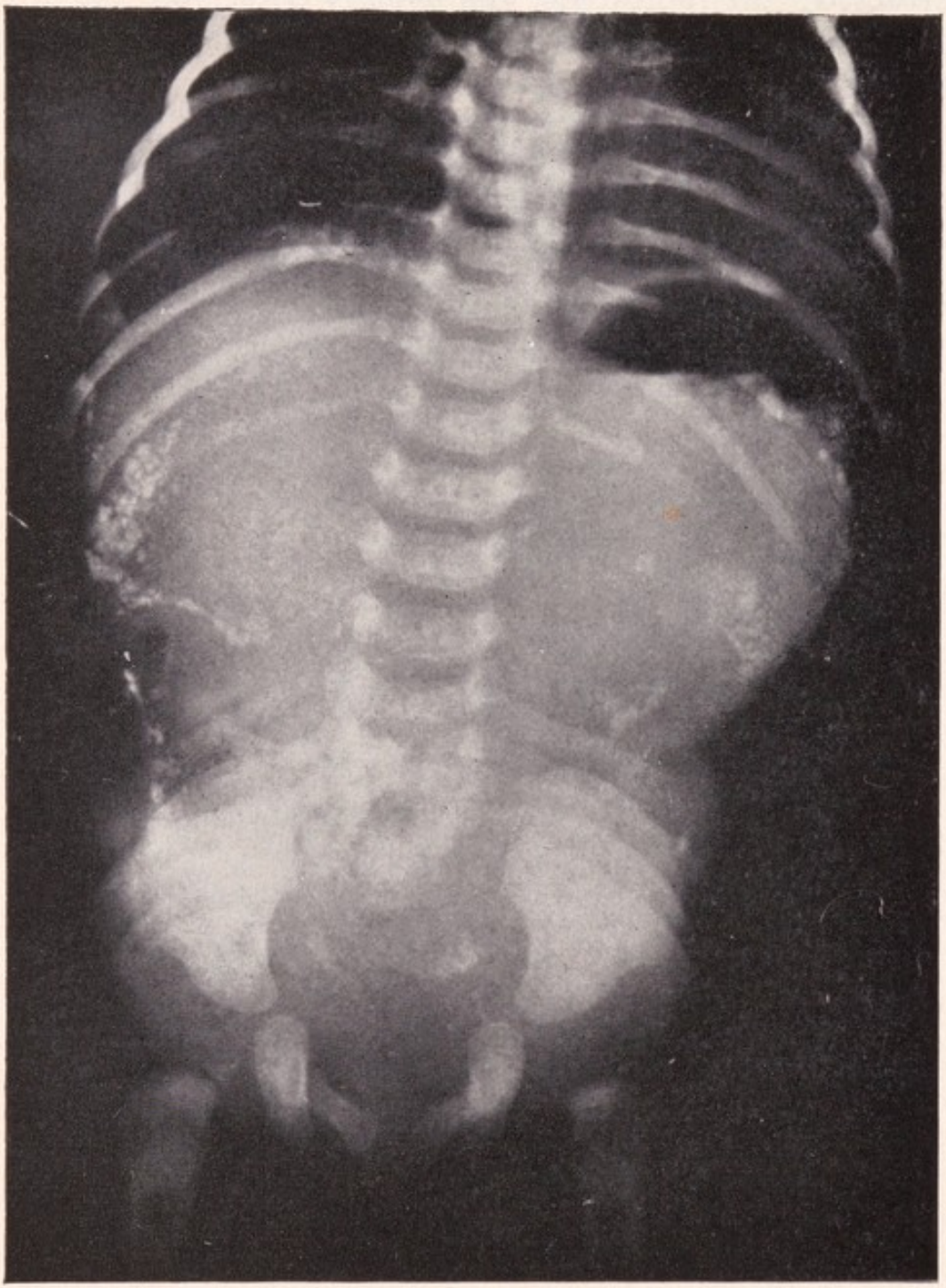


FIG. 45. Meconium peritonitis. The bowel is almost airless except for the stomach bubble. There is diffuse speckled calcification within the abdomen.

not passed. In anorectal agenesis the anus is represented by no more than a dimple and the cause of the obstruction is obvious. In rectal atresia, however, there is an anal pouch that is blind, and the diagnosis will not be apparent until the infant's failure to pass meconium, or the development of other symptoms, leads to appropriate investigation. Incomplete lower bowel obstruction may be due to Hirschsprung's disease, to a variety of anal anomalies (p. 81), or, rarely, to abnormalities of mesenteric attachment or duplication cysts.

Obstruction of the lower bowel is occasionally caused by meconium plugs. These are plugs of firm, inspissated meconium which the normal bowel peristalsis is unable to move on. Although the mechanism is analogous to that in meconium ileus, the condition is in no way related to mucoviscidosis and the plugs are most commonly in the rectum or colon, not in the ileum.

Rectal examination helps in the diagnosis of two of these conditions. Meconium plugs may be palpable in the rectum and can sometimes be removed digitally. The leading end of the plug often consists of white, mucoid jelly, the remainder being dark meconium formed like a stool and not in the semi-liquid state of normal meconium. Rectal examination may also reveal the lower end of a narrowed rectosigmoid junction in Hirschsprung's disease. The distal rectum is usually empty, and the tip of the finger encounters a sudden narrowing about two inches above the anus. It feels not unlike the external os of the cervix uteri.

Whenever a tentative diagnosis of intestinal obstruction is made, a straight X-ray of the abdomen is essential. If the anus is not patent, the infant should be X-rayed in the inverted position (Fig. 24, p. 83), the rectal or anal air bubble indicating the distance between patent bowel and the surface. In Hirschsprung's disease the colon above the aganglionic segment is seen on a straight X-ray to be grossly dilated (Fig. 46). The cautious introduction of a small amount of Dionosil will confirm this and will also demonstrate the length of the narrowed segment (Fig. 47).

Later vomiting

Two important causes of later vomiting deserve special mention. Pyloric stenosis is not a congenital abnormality

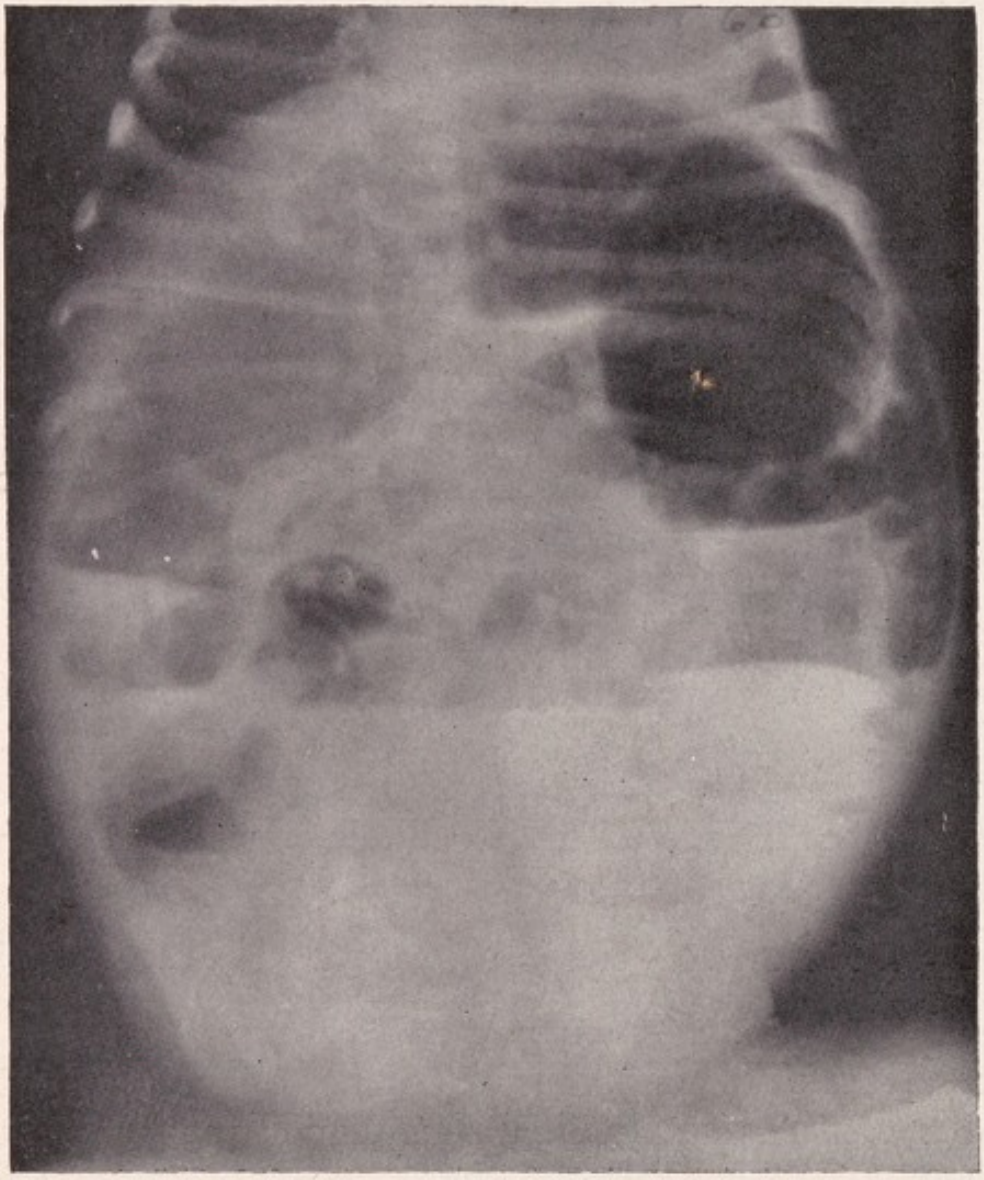


FIG. 46. Hirschsprung's disease. The abdomen is greatly enlarged. The colon is distended. There are multiple fluid levels.

and it is not a diagnosis that readily comes to mind in a maternity unit. This is because the onset of symptoms usually dates from the second or third week of life, or later still, after the infant has returned home from hospital. It is, therefore. easy to overlook this condition in a vomiting

premature baby. It is said that pyloric stenosis develops in prematures at the same time as it would have done had they been born at term. That is to say, a baby born after

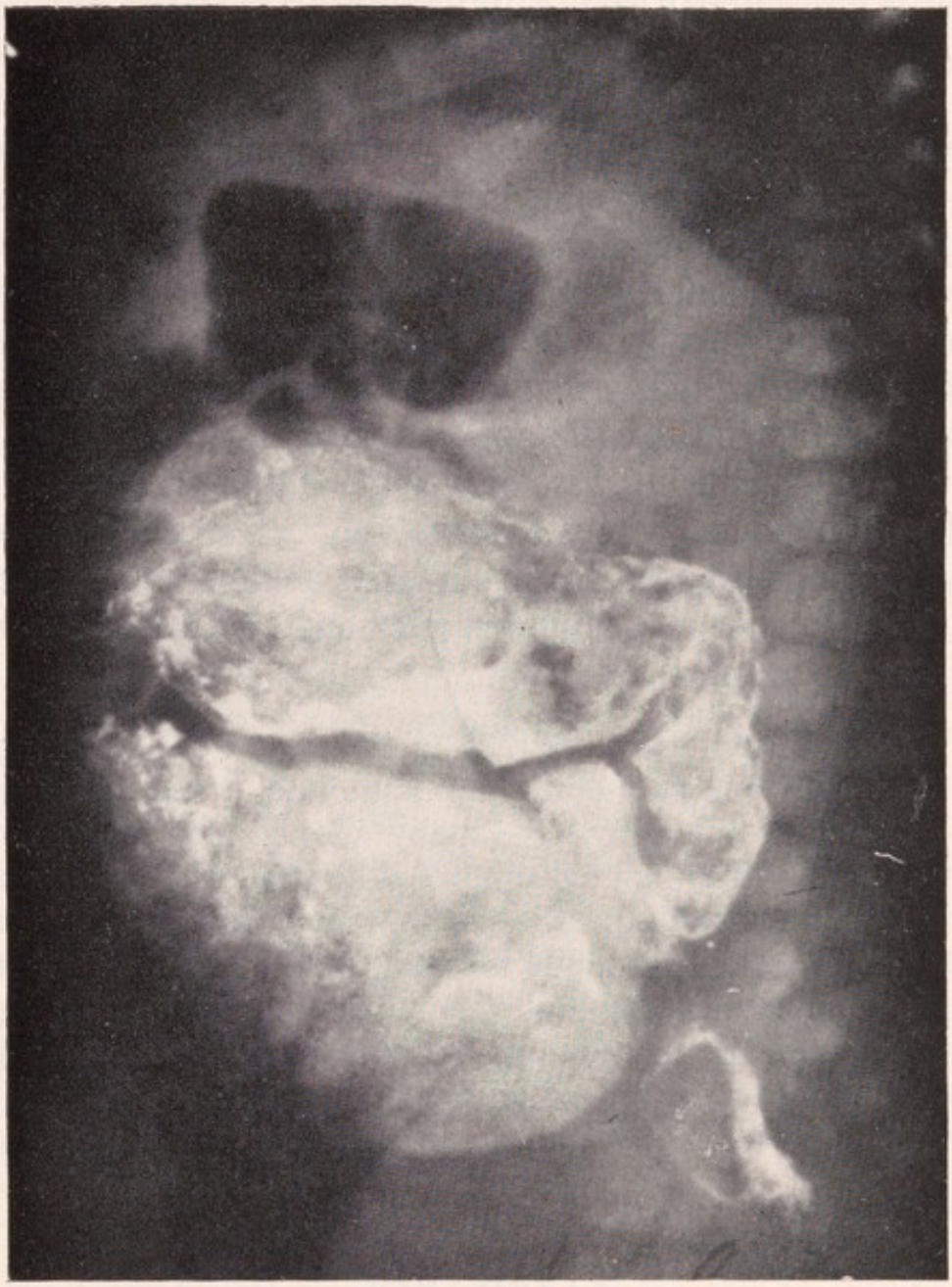


FIG. 47. Hirschsprung's. disease. A Dionosil enema demonstrates a grossly distended sigmoid colon and a narrowed segment at the recto-sigmoid junction.

32 weeks' gestation would not begin to vomit until it was about three months old. Be that as it may, premature babies often have to remain in hospital longer than the time

of their unfulfilled gestation, and symptoms of pyloric stenosis may develop whilst they are still in the maternity unit. There is nothing difficult about the diagnosis; indeed, the thin abdominal wall makes the palpation of the pylorus particularly easy, but the tumour (and the diagnosis) may be missed if the physician does not think of feeling for it.

Another cause of vomiting worth bearing in mind is congenital adrenal hyperplasia. In girls, this condition presents at birth as female pseudohermaphroditism. In boys, no abnormality may be apparent until it becomes clear that they are growing and developing with abnormal rapidity; but in many instances disturbances of electrolyte balance result in the early appearance of symptoms. These symptoms—vomiting, loss of weight, and later, dehydration and collapse—are rarely conspicuous before the second or third week of life, but if the clinician is alerted by a history of the disorder in previous siblings, evidence may be recognized sooner.

> In a recent illustrative case, a boy was born to a mother whose previous two sons had both had congenital adrenal hyperplasia. At birth, the baby looked perfectly normal. The penis was of normal size, and the urinary 17-ketosteroids measured a few days after birth were within normal limits. On the seventh day of life he was noted to have lost 9 oz. in weight since the previous day, although apparently taking feeds well. The next day he had one small vomit and rapidly developed the picture of adrenal cortical insufficiency. His response to treatment was equally dramatic.

JAUNDICE

Jaundice is an important symptom at any age, but never more important than in the neonatal period. If it develops within twelve hours of birth, it is likely to be due to rhesus or ABO blood-group incompatibility between the mother and child, although systemic infections, including congenital

syphilis, and blood dyscrasias are occasionally responsible. Appearing on the second or third day of life, jaundice is usually 'physiological', but mild cases of haemolytic disease may declare themselves at this time. Hepatitis, from infection with viruses, bacteria, *Treponema pallidum* or *Toxoplasma gondii*, may cause jaundice which is apparent at birth, though more usually it develops during the first few days of life.

Congenital abnormalities are only responsible for a minority of instances of neonatal jaundice. Of these, obstructive lesions of the biliary system are the most familiar, but jaundice may be an important clue in the diagnosis of galactosaemia (p. 111), fibrocystic disease of the pancreas (p. 95), and congenital hypothyroidism (p. 112).

The characteristics of obstructive jaundice in the neonatal period are its relatively late onset, its fluctuating intensity and the fact that the bilirubin in the blood is principally of the conjugated or direct-acting type. Pallor of the stools is often conspicuous before jaundice has developed. It is also variable in intensity, but usually the stools are almost white. Although the meconium is sometimes paler than normal, it is not as pale as the subsequent milk stools and rarely gives rise to comment. The jaundice may be noted towards the end of the first week of life, commonly appears in the second week, and may be delayed until the third or fourth week. At this stage the liver is likely to be enlarged and the urine is abnormally dark and contains bile pigments.

The anatomical lesion varies from a localized narrowing of the common bile duct to complete agenesis of the intrahepatic biliary system. The obstruction may also be associated with a choledochal cyst; this is a huge dilatation of the terminal part of the bile duct, the reason for which is not always apparent. Clinically, it is impossible to determine the site or extent of the lesion. Furthermore, it is impossible on clinical grounds to distinguish between this group of surgical lesions on the one hand and neonatal

hepatitis on the other. Very early onset of the jaundice—during the first three or four days of life—and early hepatomegaly are more in keeping with hepatitis than with biliary tract anomalies. In both conditions, however,

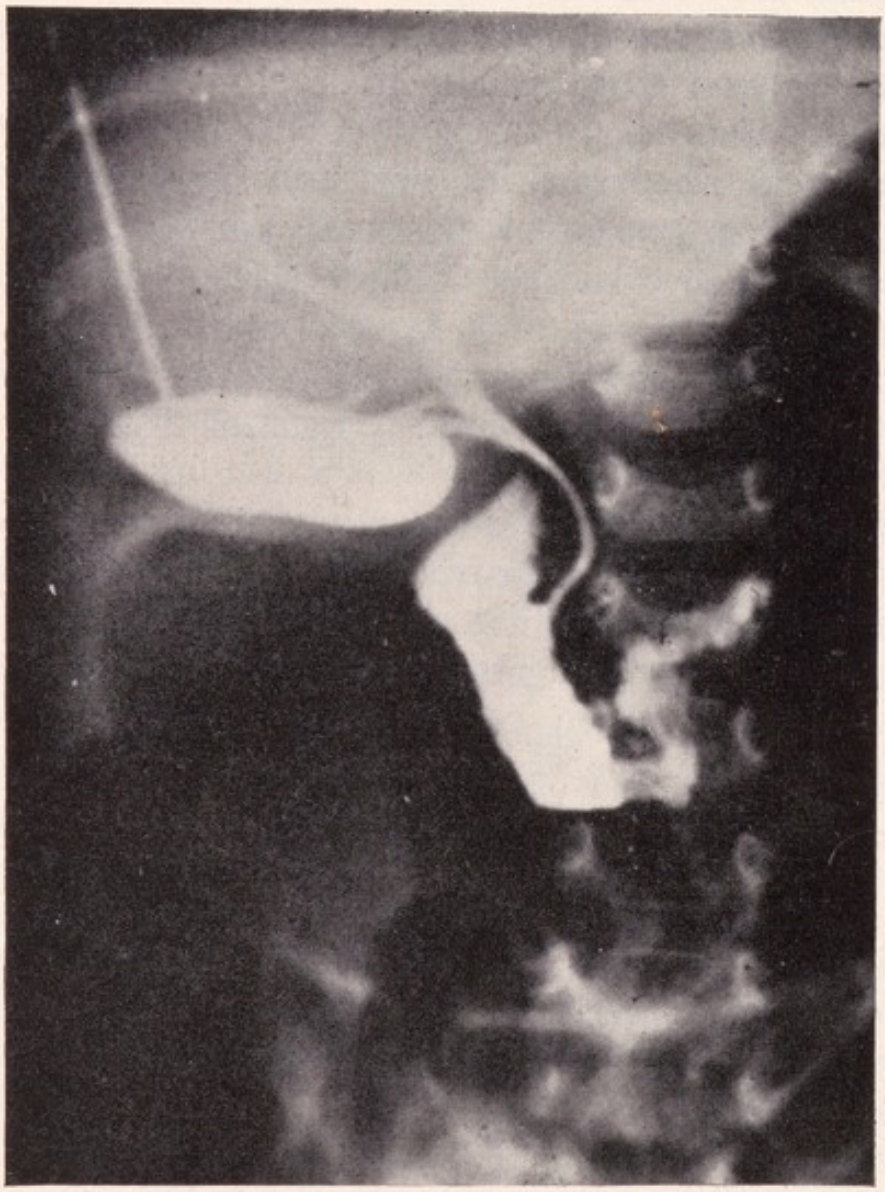


FIG. 48. Normal cholangiogram. Dye is being injected into the gall-bladder and is filling the intrahepatic and extrahepatic bile ducts. It has entered the duodenum. This infant proved to have hepatitis.

these signs commonly become apparent towards the end of the first week of life, and pallor of the stools with dark urine is common to both. Repeated attempts have been made to find a liver-function test which would distinguish between the two, but so far these have not been successful.

At the time of writing, great interest is being shown in the radio-active rose bengal test.

Until some reliable liver-function tests have been established, it is better to rely upon microscopic examination of the liver to distinguish between hepatitis and biliary atresia. Needle biopsy of the liver has been recommended, but there are good reasons for preferring open biopsy. On the whole it is probably a rather safer procedure in the newborn infant; it permits removal of a wedge of liver tissue, which simplifies the task of the pathologist; the extrahepatic part of the biliary tree can be examined directly; the intrahepatic part can be examined indirectly by cholangiography; finally, if an obstruction is found which can be relieved or by-passed, it can be dealt with on the spot.

The cholangiogram is performed by injecting radio-opaque dye into the gall-bladder. If there is no obstruction, the dye passes up into the intrahepatic bile ducts and down the common bile duct into the duodenum. Fig. 48 shows a normal cholangiogram.

CHAPTER 5

Interviewing Parents of Malformed Babies

EVERY clinician in his working life experiences occasional moments which, by their brightness or their darkness, stand out from the routine of hospital or general practice. The bright spots may be the unexpected recovery of a patient from a serious illness, gratitude expressed by patients or relatives (usually those for whom very little has been done), or, on a smaller scale, a diagnostic triumph or a technical *tour de force*. The black moments include the corresponding diagnostic, technical, and therapeutic failures, but above all those occasions when it falls to his lot to break bad news to patients or their relatives. The darkness of such moments always seems even greater when a child is involved. The news of a death, or the diagnosis of incurable disease, is difficult to convey; to make known that a new baby is malformed is certainly no easier.

Every person who by the nature of his work is involved in this situation gradually learns ways of handling it, although in this context familiarity never breeds contempt and the task never becomes any easier. Those who have already had to face this problem may leave this chapter unread, but others to whom it is a newer trial may find some help.

In brief, the parents of an abnormal baby need to be told the truth, nothing but the truth, and eventually the whole truth. When the deformity is something like a cleft lip, the whole truth is obvious at a glance. With myelomeningocele, the condition of the back is obvious but the neurological implications are not. A mongol

infant usually appears perfectly normal to the parents, and they may at first flatly refuse to accept any suggestion to the contrary.

The whole truth may therefore have to be spread out over a period of time; it does not have to be delivered in a single, shattering broadside. There is a story of a dying man whose relatives called in one specialist after another for further opinions about his illness. After the patient had died, the family doctor asked the relatives which specialist they had regarded most highly. They replied; 'The last one. He was the only one who held out any hope.' Nobody has the right to deprive the parents of hope. This is not to say that they must be encouraged to hope for anything that is known to be impossible, for that would be dishonest and, in the long run, unkind; but the situation often seems to the parents far more disastrous than it is.

There is always something which can be justifiably hoped for, and there is therefore no place in this context for the word 'hopeless' or for the phrase 'nothing can be done'. Congenital abnormalities are often incurable, but rarely untreatable, and never hopeless. After all, the hope is not in the malformation, nor in the malformed child, but in the hearts of his parents. It is often helpful to the parents if they can be told what they themselves will be able to do to help their child from the earliest days. The frustration of feeling that they are useless often makes the situation seem worse.

Every parent knows that congenital malformations occur, but it is part of the nature of humanity to believe that such things happen only to other people's children. It is exceptional for an expectant mother to believe that her baby will be other than perfectly formed. (Nevertheless, most mothers will express relief if, soon after the baby is born, the doctor examines him and pronounces him healthy.) A few mothers are seriously concerned that their babies may not be normal. Sometimes the fear is the consequence of an attempt to terminate the pregnancy. It

seems possible that recent public discussion of the effects of ionizing radiation, and of drugs taken, and illness suffered, by the mother in pregnancy will make such fears more common. Even so, these half-felt fears are only for physical imperfections; the idea of mental handicap seems but rarely to be contemplated.

The first reaction of parents on learning that their child has a serious malformation is therefore often something approaching incredulity, a rejection of the news, a refusal to believe it. This is especially so if the news is of mental handicap. When doubts (and often hopes) on this point have dissolved, there follows an agonized phase in which logic and reason have no place, only the knowledge that a situation has arisen which is unbearable and at the same time inescapable. In this situation, fathers (not least when they are themselves doctors) are every bit as vulnerable as mothers, perhaps more so. They feel strongly their failure to procreate a perfect child and they cannot escape so easily into maternal feelings of caring for the child. Fathers tend to look forward to the child becoming a companion and partner in business and games, especially if it is a boy and above all if it is a first boy.

The news that one's child is handicapped strikes deeply. It strikes at a parent's feeling of adequacy at reproducing (especially if it is the first child); it strikes at the feeling of being adequate at the task of caring for the child; it may arouse feelings of guilt that this is a deserved punishment.

It is perhaps helpful to consider a comparable event that also seems at the time inescapable and unbearable, that of bereavement. It is better realized today that anger and resentment, as well as deep sorrow, depression, and the aching void, are feelings normal to this situation. It is often helpful to let parents know that to feel anger and resentment at their child's malformation is neither unusual nor deplorable.

Obviously the parents' reaction to the first news will depend to some extent on the seriousness of the infant's disability, but the doctor does well to remember that nearly

always this seems far more serious, far more hopeless, than it does to him. Parents will fairly easily accept the news that a child has a malformation such as a cleft palate. But to learn that their child is a mongol is such a knock-out blow that of this first interview almost nothing is likely to be remembered: none of the words of mitigation, the expressions of sympathy, the explanations of long-term measures; only the bare, disastrous fact that the child will always be mentally handicapped. If the doctor imagines that, with the first telling, he has dealt and finished with an awkward situation, he is almost certainly in error.

If a handicapped person is to be accepted into the community, it is necessary for him first to be accepted into his family. The greatest tragedy that can happen to a child is for him to be rejected by his parents from birth. The abnormal arouses a natural biological revulsion, against which maternal, paternal, and humanitarian feelings will act. If the child's malformation is a grave one, and especially if it involves mental defect, there is real danger of the parents rejecting the child. The doctor has to weigh everything he does in the early days from the point of view of whether the mother and father are helped to accept the child. Serious news clumsily conveyed can militate against this; keeping the mother and child apart unnecessarily is even more potent. In the early days and weeks after her child is born, the mother is emotionally ready to fall in love with her child. To hold and feed the baby will help her to do this. Not to see the infant will hinder it. Even if the child must be in an incubator, he can travel to the mother's room and she may let him hold her finger.

When the infant is so severely handicapped that later in life he will need to go away from his home, he will do better for having had experience of a mother's continuing care, and the parents will have had pleasure and satisfaction from his presence and from having given him the best start. The parents of a newborn mongol can be helped to give him what he needs at the present, a place in his family;

he may have to go into residential care later, but that bridge can be crossed when the family comes to it.

It may be argued that if the malformation is such that the child is likely to die within a few days or weeks it is better for the mother not to see the child; but this argument is nearly always fallacious. It seems that it is easier for a mother to get over the loss of a child if she has loved and mourned it than if she has not really known it and feels she perhaps could have done more.

Before breaking news of a malformation to parents, it is as well to run through a simple catechism: When shall I tell? whom shall I tell? what shall I say? how shall I say it? There are no stock answers that are always right, and the decisions must be made afresh every time this situation arises.

When to tell

The choice of time will depend upon the nature and severity of the malformation. With minor anomalies the news can be broken at the first convenient moment and the parents reassured that the baby is otherwise perfectly normal. If urgent surgical treatment is required, the situation must perforce be explained to them immediately. If the child's life is in danger it may be necessary to give some preliminary warning. If the deformity is obvious, delay is not possible for more than a day or two. If, however, there is no necessity for the news to be broken at once, there is much to be said for waiting a few days. Many mothers in the first few days of the puerperium are physically tired and emotionally fragile; bad news, at this time, is likely to appear even worse. Premature disclosure may lead to profound depression, even to the point of attempted suicide. This is particularly likely if the infant is mentally handicapped but may also occur with severe malformations.

Whom to tell

There are, in general, objections to asking one partner to keep information a secret from the other, but the doctor may well consult one partner for guidance on how the other is to be informed. It is usually desirable that the doctor should find an early opportunity to discuss the problem with both parents together.

What to say

Parents want to know four things about their abnormal infant, although they will not always ask the questions directly. The answers must nevertheless be provided at the appropriate time.

(1) What exactly is the matter? What are the implications? This may be obvious, or it may need explaining in language suited to the individual.

(2) What can be done about it? What can the doctors do? What can we do ourselves?

(3) Why did it happen? with the corollary, is it in any way our fault? This is the question least often put into words, but it is one of the most important to discuss with parents. This is amplified below.

(4) Will this happen again if we have more children? This question also may not be asked but the answer is wanted just the same.

How to say it

What the doctor says when he first tells the parents depends largely on the 'quantity' of bad news the parents will have to take in. Parents can, in one interview, easily absorb the news that the child has a cleft palate and that an operation will be necessary during the second year, for they will also be learning that all will be well in the end. But the parents of a child who is gravely handicapped deserve to be allotted three interviews for explanation.

They will want information, they will need support, they will need hope; they will usually need to go over the ground again and again.

I am not sure that any one person can help another except in the most general terms. The doctor has to remember that his understanding of the situation derives from long years of training; the parents are novices and even if they understand the words, they draw very different inferences from those the doctor draws. The choice of words is important. They must be intelligible to the person spoken to and they must be devoid of ambiguity.

Take as examples the three conditions already mentioned, namely, cleft lip, myelomeningocele, and mongolism. Words are not necessary to explain a cleft lip, but it will come as a shock to the mother to see it for the first time, especially if it is a bilateral cleft. It is tremendously helpful to her to be shown photographs of a similarly affected child before and after surgical repair. This demonstrates far more clearly than words what can, and will, be done. She should also be given some idea of the age at which operation will be performed so that the inconvenience and embarrassment of taking a deformed child home with her will be to some extent mitigated by the knowledge that it is only for a matter of weeks.

Although the parents may think that only the back is abnormal, myelomeningocele is, as a rule, a much more disabling anomaly. They must be given some account of any neurological complications, and it is better to speak of muscle weakness and bladder or bowel weakness rather than to use the word paralysis. At present, the answer to 'What can be done?' will vary from centre to centre, and the timing of surgical procedures leaves room for individual judgement. If early surgery is not contemplated and the infant thrives in the first week or two of life, it is reasonable to put a horizon at the age of three months and arrange full appraisal then. By this age, infants with myelomeningocele who are thriving will, with very few exceptions, continue to thrive.

One of the most unpalatable tasks which can fall to the lot of a general practitioner, paediatrician, or obstetrician is informing parents that their child is a mongol, or, indeed, that it has any kind of mental handicap. It is especially in this context that the problems of whom to tell and when to tell arise. Most parents have no idea that there is anything the matter with the infant at all. Occasionally, if the mother knows a mongol child, she may make the diagnosis herself as soon as she sees the baby, but this is exceptional.

As regards the time at which to break the news, there is a wide diversity of opinion. If the mother is told as soon as the diagnosis is made, she will still be in the early days of the puerperium, when the emotions are known to be labile. The opposite policy is to say nothing until the parents spontaneously express anxiety about the baby's progress. There are at least two disadvantages in this scheme. It will be a long time, usually a matter of months, before the parents are convinced that something may be wrong. When it comes to explaining away the shortcomings of children, the parents' minds have almost unlimited resources; even when the initial doubts have approached certainty, parents will be fearful of seeking confirmation of their suspicions. This means that if the first move is left to the parents, they will go through a protracted period of uncertainty before finding the courage to discuss their anxieties with the doctor. Furthermore, at any time during this phase somebody may say: 'Of course, you know he's a mongol!' This may happen if the infant is admitted to hospital because of an intercurrent chest infection, for example. This is the worst possible way in which the information can be imparted. The parents may feel that their normal child has been snatched from them. They may lose trust in their doctor.

On the whole it seems better to convey the news within a week or two of birth, but not in the first few days. If it is postponed for months, parents will often say how much they wish they had known sooner. On the other hand, if

the information is given too soon, before the normal mother-child bond has been forged, the infant may be completely rejected. When such serious news has to be conveyed, the doctor should try to set aside time for several interviews, on successive days. It may be helpful if a parent of a mongol from a similar social background can meet them and answer their questions.

There is much to be said for interviewing the father first without the mother. When the situation has been explained to him, he may be invited to decide whether he would prefer to break the news to his wife himself or prefer the doctor to do this. If he expresses a wish to do it himself, he will, in almost every case, postpone this unpalatable task week after week, or express himself so vaguely as virtually to have said nothing. It is therefore absolutely essential, if the father takes this responsibility, to check that he has carried it out. This can best be done by arranging to see both parents with the baby a few weeks after birth.

If the father asks the doctor to inform the mother, this should be done whenever possible in the father's presence. They will need each other's support in the times ahead, and the beginning is a good place to start.

Why did it happen?

In the great majority of instances the cause of a malformation is not known. Genetic disorders, maternal rubella, toxoplasmosis, and teratogenic drugs account for only a small part of the total. It is therefore exceptional for the clinician to be able to give a direct answer to the question: 'What was the cause of this?' It is, however, usually possible to enumerate a number of things which are so unlikely to have been responsible that they can be ruled out. This can be of the greatest help to the parents. There is a very deep-seated and primitive belief, still embodied in many religions, that malformations, together with illnesses, deaths, disappointments, and all manner of

setbacks, are punishments for sins committed. When a baby is born abnormal there is a strong tendency for the mother in particular to search her memory for something she did that she should not have done, or something she did not do that she ought to have done. This feeling of guilt is common and may be strong; it is part of the doctor's job to try to help to dispel it. A point must always be made of explaining to the parents that it is not their fault, that it could have happened to anybody.

In the case of genetic disorders, the statement that it is 'not their fault' needs some modification. Disorders caused by dominant genes will be present in one parent and the doctor will not have to explain the cause. With recessive genes it must be emphasized that *both* parents are carriers. It is worth explaining that every individual is a carrier of about half a dozen serious disorders, but most of them never find out.

With the rare disorders caused by sex-linked genes, the affected boy has received the abnormal gene from his mother. This should *not* be explained to the parents if it can be avoided, although the mode of inheritance of haemophilia is fairly widely known. If there have been previous cases in the family, it will be obvious from which side the disorder comes. If the family history is negative, however, there is no necessity to blame one partner and exonerate the other. The prognosis for further children can be given empirically without explaining the genetic background.

Will it happen again?

The parents of a malformed child always want to know whether subsequent children are likely to be similarly affected. Some parents ask this question outright, some, because they dread the answer, are afraid to, and others do not think of it until the opportunity is past. The clinician must, therefore, make it his personal responsibility to provide this information at a suitable time.

The time is not always easy to judge. If the congenital

abnormality is a serious one, the parents will not absorb any additional information in the early days. Even then, the shock may have been so great that they cannot possibly visualize themselves deliberately having another child and thereby risking another disappointment. These considerations, together with the emotional instability so common in mothers in the puerperium, makes it advisable to postpone this discussion for a few weeks, unless the parents raise the question themselves.

On the other hand, when the recurrence risk is high, that is, in genetically determined disorders, it is unwise to postpone the interview for too long. This is particularly true of abnormalities that cannot be recognized until some months after birth. For example, Tay-Sachs disease (familial amaurotic idiocy) and Werdnig-Hoffmann disease (progressive spinal muscular atrophy) are both controlled by recessive genes and the risk of recurrence in future children is therefore 1 in 4. Some parents of infants affected with these disorders, knowing that their children cannot live long, decide to plan another baby forthwith. If genetic advice is to be of any help to them it must be given promptly. The terms 'genetic advice' and 'genetic counselling' are apt to suggest that the doctor advises the parents what they should or should not do; this is not the case. Advice is here used in its older sense of presenting facts. The temptation to make recommendations, and even the direct invitation to do so, should be studiously resisted.

If the abnormality is not evident until the infant is more than three months old, it is as well to interview the father first and to inquire whether his wife is again pregnant. In Great Britain at present the existence of hereditary disease in a family is not accepted in law as sufficient grounds to justify the termination of pregnancy. If, therefore, another baby is already expected it is pointless to spread gloom and despondency about its prospects. Even if the whole truth has to be told at this stage, it is surprising how the emphasis can be changed without altering the facts.

Somehow a 1 in 4 risk of abnormality sounds much more depressing than odds of 3 to 1 in favour of normality.

The risk of future children being affected should always be expressed as odds or chances or probabilities. Although most people are familiar with these terms, two important points should be explained to them. The first is that there is an overall risk of about 1 in 50 that any baby will have a serious malformation. This is the base-line with which all other risks must be compared. The second point to be made concerns a popular misunderstanding about probabilities. If a penny is tossed nine times and comes down 'heads' every time, many people feel that at the tenth toss it is more likely to be 'tails' than 'heads'. Although they may accept that the penny cannot know which way it fell last time, it may still seem 'common sense' that the more often it comes down heads, the more likely it is to be tails next time. On the same reasoning, the parents of a child with fibrocystic disease, being told of the 1 in 4 incidence, may feel that the next three children should be normal. (This is reminiscent of the man who was told that the operation he was about to undergo carried a 90 per cent. mortality rate, but as the last 9 patients had died he should be all right!)

Before genetic advice is given, a full family history is taken together with a history of events early in the pregnancy (virus infections, drugs taken, threatened abortion) which may have relevance. Contrary to popular opinion, only a small minority of malformations are of genetic origin. Of the others, however, parents will always welcome suggestions as to what might have been the cause, even though they appreciate that it is only an intelligent guess. To be able to attribute it to something, preferably something which is unlikely to happen again and for which they were not responsible, will give them confidence in the outcome of future pregnancies and will help the mother in particular to banish any feelings of guilt.

GENETIC PROGNOSIS

The following notes on genetic prognosis are not intended to be comprehensive, nor is it assumed that every clinician concerned with newborn babies wants to be a geneticist. However, the doctor who diagnoses a congenital abnormality and speaks to the parents about it must be prepared to meet some questions.

Achondroplasia

This is usually the result of a mutation, the parents being normal. The risk of recurrence within a sibship is therefore no higher than random. The mutant gene is dominant, and the children of an achondroplastic dwarf will therefore be, on average, normal and achondroplastic in equal numbers.

Adrenal hyperplasia, congenital

This may present with virilization or with electrolyte disturbances. In some cases there is hypertension. It therefore seems likely that more than one enzyme deficiency may be involved. The defect is determined by an autosomal recessive gene or genes. The chances of recurrence in a sibship are 1 in 4.

Albinism

This is determined by an autosomal recessive gene. The incidence of the disorder is about 1 in 20,000 of the population. The gene is therefore carried by about 1 person in 70. The recurrence risk is 1 in 4.

Amaurotic idiocy, familial (Tay-Sachs disease)

This condition is not, as was once thought, confined to the Jewish race. It is determined by an autosomal recessive gene and the recurrence risk is 1 in 4.

Anencephaly

From the genetic viewpoint, there is a close relationship between anencephaly, spina bifida, and hydrocephalus. The occurrence of any of these in a sibship increases the likelihood that one of them will recur. The random incidence of anencephaly varies very widely from one part of the world to another. The figure most widely quoted is 1 per 1000 births; in Liverpool it is 3·5 per 1000 births. The recurrence risk of a major nervous system malformation is about 1 in 25. After two affected children, the chances of a third are in the region of 1 in 4. This suggests that there may be genetic and non-genetic forms of anencephaly, the former carrying a 1 in 4 risk from the start, but at present one cannot distinguish between them until the birth of a second affected child suggests a genetic basis. The well-known preponderance of female over male anencephalics appears to be due to the selective loss of male anencephalics by early abortion.

Cataract

The causation is variable, being in some instances infective, in some metabolic and in others genetic. The latter are usually caused by autosomal dominant genes.

Cleft lip, with or without cleft palate

This is genetically distinct from cleft palate alone. It is more common in males. If the parents are normal the recurrence risk is about 1 in 25. If one parent has a similar defect, the recurrence risk in the sibship is about 1 in 7.

Cleft palate alone

This is genetically distinct from cleft palate with cleft lip. It is more common in females. The recurrence risk is about 1 in 50 if the parents are normal and about 1 in 15 if one parent is similarly affected.

Cleidocranial dysostosis

This is usually due to an autosomal dominant gene; one parent and about half the children are therefore affected. However, the manifestations of the disorder are very variable and in mild cases may only be revealed by radiographs. The recurrence risk in a sibship is 1 in 2.

Craniofacial dysostosis (*Crouzon*)

Some cases are sporadic while others appear to be due to an autosomal dominant gene. If both parents appear normal and there are no affected relatives, the recurrence risk is small.

Deafness, congenital

The causation is variable. About 25 per cent. of cases are of genetic origin, the responsible gene being dominant in some cases and recessive in others. Deafness may be associated with congenital goitrous cretinism (recessive) and is a feature of Waardenburg's syndrome (deafness, lateral displacement of the inner canthus, white forelock, and eyes of different colours) which is dominant. In each case of congenital deafness a full history of the family, the pregnancy, and the neonatal period must be taken. Conversely, any infant born to a congenitally deaf parent must have his hearing tested as early as possible and, if necessary, repeatedly (*see* Appendix 2).

Dislocation of the hip, congenital

This is about ten times more common in females than in males. The recurrence risk in a sibship is about 1 in 20 if both parents are normal, and 1 in 10 if one parent is affected.

Epidermolysis bullosa

This 'disease' is probably a group of diseases, some due to dominant genes and others to recessive genes. The severe form which commonly leads to death in the neonatal period is, at least in some families, due to a recessive gene and the recurrence risk is 1 in 4.

Fibrocystic disease of the pancreas (*mucoviscidosis*)

This is caused by an autosomal recessive gene and therefore carries a 1 in 4 recurrence risk. Heterozygotes can sometimes be identified by the finding of sweat sodium and chloride levels intermediate between those of normals and affected individuals. The finding of abnormal sweat electrolytes in some adults with bronchiectasis suggests that it may be necessary to revise the earlier concept of this disorder as something incompatible with survival beyond childhood.

Fragilitas ossium (see *Osteogenesis imperfecta*)

Galactosaemia

Like most inborn errors of metabolism, galactosaemia is caused by an autosomal recessive gene. The recurrence risk is 1 in 4, but when there has been an affected child in the family any subsequent cases will be diagnosed very early and treated appropriately. This largely mitigates the high recurrence risk.

Gargoylism (*Hunter-Hurler syndrome: chondrodystrophy*)

This is caused by a recessive gene, usually an autosomal one. In the less common sex-linked variety, corneal clouding is absent. The recurrence risk is 1 in 4 for the autosomal type, 1 in 2 males in the sex-linked variety.

Gaucher's disease

The acute, infantile form of the disease is caused by an autosomal recessive gene (recurrence risk, 1 in 4). The adult form of the disease appears to be due, at least in some families, to a dominant gene. The family history should be studied in detail.

Gonadal dysgenesis (*ovarian agenesis: Turner's syndrome*)

This is usually associated with an XO sex-chromosome constitution resulting from non-disjunction during gametogenesis. Although a few families have been described in which more than one member had a chromosomal abnormality, the empirical chance of recurrence within a sibship is very small indeed.

Harelip (see *Cleftlip*)

Haemophilia

This is the classic example of sex-linked inheritance, the abnormal gene being attached to an X chromosome. The disease is manifest in males and transmitted by symptomless females. If such a carrier has children, on average half her sons will be affected and half her daughters will be carriers. If a haemophiliac male should have children, all his sons will be normal and all his daughters will be carriers. Christmas disease is inherited in the same way.

Hirschsprung's disease

This disorder is very much more common in boys than in girls. The risk of recurrence in a brother has been estimated at about 1 in 5.

Hunter-Hurler syndrome (see *Gargoylism*)

Hydrocephalus

Isolated hydrocephalus is the least likely to recur of the three major malformations of the central nervous system (*see* Anencephaly). The empirical risk of recurrence within a sibship is of the order of 1 in 100. Hydrocephalus with spina bifida is more likely to recur (*see* Spina bifida), and the birth of a hydrocephalic infant increases somewhat the chances of anencephaly or spina bifida occurring in later children. A few cases of hydrocephalus are caused by a sex-linked recessive gene; at present these can only be distinguished by a close study of the family history.

Ichthyosis

The variety of ichthyosis which is apparent at birth or soon afterwards is caused by an autosomal recessive gene. The recurrence risk is 1 in 4. The more common variety of ichthyosis which becomes apparent later in life is usually inherited in dominant fashion.

Klinefelter's syndrome

This is associated with an XXY sex-chromosome constitution resulting from non-disjunction, probably during gametogenesis. Exactly the same recurrence risks apply as for Gonadal dysgenesis (q.v.).

Mandibulofacial dysostosis (*Treacher-Collins syndrome*)

A dominant gene is usually responsible although those bearing the gene do not necessarily show all the features of the syndrome. About half the siblings of an affected child may be expected to show some features of the disorder.

Microcephaly

This condition is discussed on page 48. The cases of genetic origin are usually due to a recessive gene, the recurrence risk being 1 in 4.

Mongolism

Mongolism is the consequence of possessing the genetic material of an additional chromosome 21. In regular mongols this is present as an extra, separate chromosome (trisomy), making the total number of chromosomes in somatic-cell nuclei 47 instead of 46. Less commonly, and principally in mongols born to young mothers, the additional genetic material may be attached to another chromosome (translocation) so that the total number of chromosomes remains normal. If a translocation is identified in the chromosomes of a mongol, it may have arisen during gametogenesis or the abnormal chromosome may have been inherited from a normal parent. It is therefore necessary in these cases to study the chromosomes of both parents. If their chromosomal constitutions are normal, the mongol's translocation must be assumed to have arisen during gametogenesis. In this case, the parents have no special risk of giving birth to another mongol. If, however, the translocated chromosome is identified in a parent (a translocation carrier with 45 chromosomes), the probability that any future child will be a mongol is 1 in 3. Furthermore, half the 'normal' children of these parents will be translocation carriers (*see* Fig. 49).

If the mongol is shown to be trisomic, it can be assumed for ordinary purposes that non-disjunction occurred during gametogenesis and that the parents have no greater risk of having another mongol than any parents of the same age. There is, however, a rare exception to this rule. One or other parent may be a chromosomal mosaic, their somatic cells being a mixture of some with 46 normal chromosomes and others trisomic for chromosome 21. Such an individual may have all, some or none of the physical and mental stigmata of mongolism. The recurrence risk would depend upon the type of cells from which the ova develop. If a mongol has already been born, it is reasonable to assume that the gonadal tissue consists wholly or predominantly of

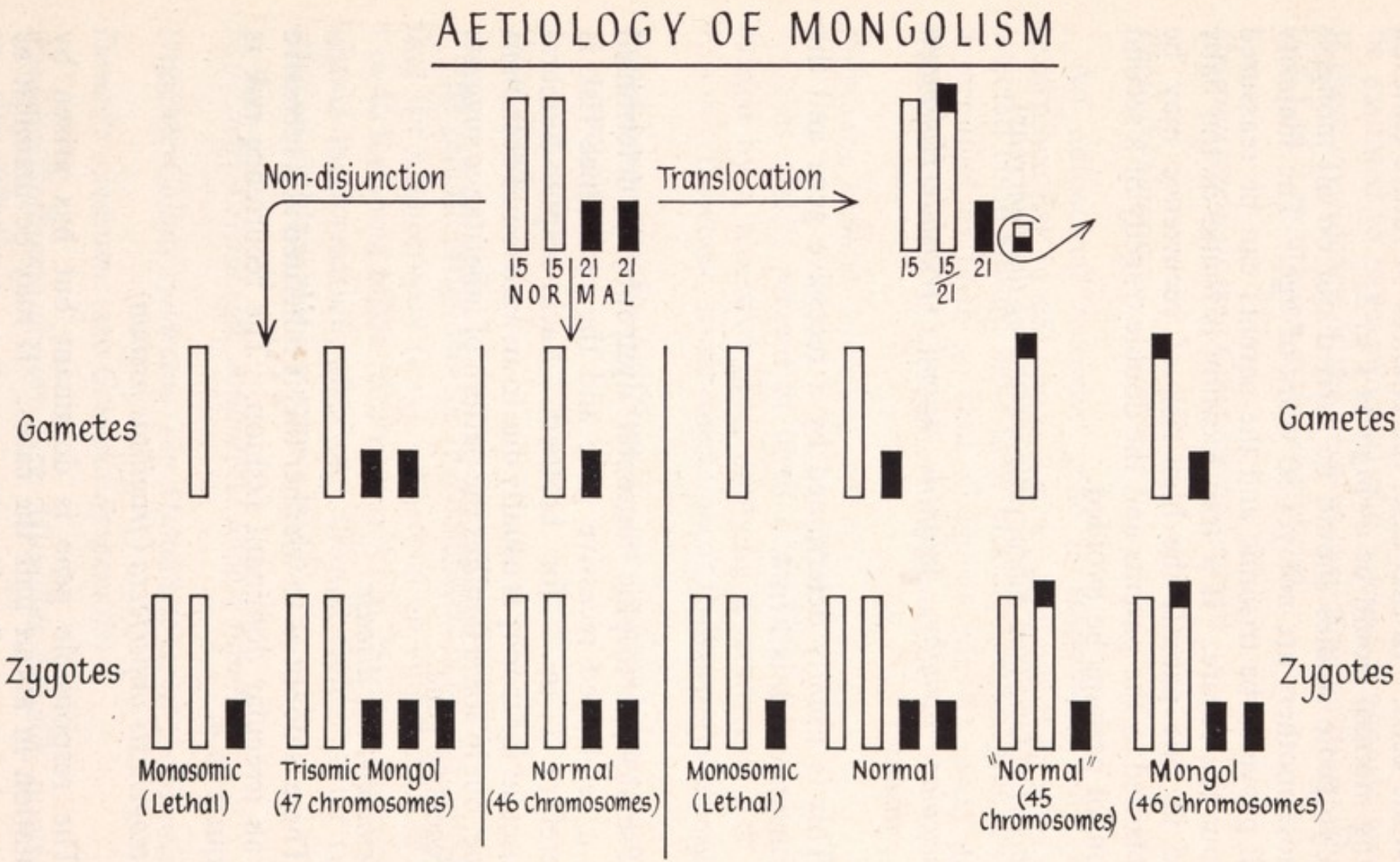


FIG. 49. The aetiology of mongolism. For explanation, *see* text.

trisomic cells. In this case, the chances of future children being mongol would be as high as 1 in 2.

Cytogenic studies should be carried out on all mongols whose mothers are not yet 30 years of age. The majority will prove to be trisomic and the parents can be reassured about the future. If a translocation is found in the baby and in one parent, the high risk of recurrence may be explained to the parents and the double tragedy of a second mongol perhaps be avoided.

Mucoviscidosis (see *Fibrocystic disease of the pancreas*)

Muscular atrophy, infantile spinal (*Werdnig-Hoffmann disease*)

This is usually determined by a recessive gene and the recurrence risk is 1 in 4.

Muscular dystrophy

Pseudohypertrophic muscular dystrophy is determined by a sex-linked recessive gene and the recurrence risk is therefore 1 in 2 for brothers. Facio-scapulo-humeral muscular dystrophy is usually due to an autosomal dominant gene, but in some families the pattern of inheritance suggests a recessive gene.

Niemann-Pick disease

There is doubt as to whether this is inherited in a recessive or an irregular dominant fashion. The recurrence risk is about 1 in 4.

Osteogenesis imperfecta (*fragilitas ossium*)

The responsible gene is dominant but has arisen by mutation in about half the cases. It may be manifest as blue sclerae or otosclerosis as well as brittle bones. If

none of these feature in the family history, a mutation may be assumed to have occurred and a good prognosis given. If there is evidence of the gene in earlier generations, there is a 1 in 2 recurrence risk for siblings.

Ovarian agenesis (see *Gonadal dysgenesis*)

Phenylketonuria

An autosomal recessive gene is responsible and the recurrence risk in siblings is 1 in 4. Siblings born after an affected child should have their blood phenylalanine levels determined at birth so that diagnosis can be made and treatment started at the earliest moment.

Polycystic kidneys

Cases which present at birth or in infancy are usually caused by a recessive gene, implying a recurrence risk of 1 in 4. However, some cases appear to be sporadic.

Pseudohermaphroditism, female (see *Adrenal hyperplasia, congenital*)

Spina bifida

The genetic prognosis for this condition is bound up with that for anencephaly (q.v.). There is an empirical risk of 1 in 40 for spina bifida recurring and 1 in 25 for any nervous system malformation, after one affected child. After two affected children the recurrence risk is 1 in 4.

Tay-Sachs disease (see *Amaurotic idiocy, familial*)

Treacher-Collins syndrome (see *Mandibulofacial dysostosis*)

Turner's syndrome (see *Gonadal dysgenesis*)

Werdnig-Hoffmann disease (see *Muscular atrophy, progressive spinal*)

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APPENDIX 1

Notes on the Transfer of Newborn Infants for Emergency Surgery

THERE are still relatively few centres in Great Britain which are adequately staffed and equipped to deal with surgical emergencies in the first few days of life. This implies that the majority of infants requiring such surgery will need to be transported from their place of birth to a special unit. The pre-operative care of these babies begins at the moment a surgical emergency is suspected. The importance of early diagnosis has been stressed throughout this book; of equal importance to infants requiring surgery is their safe and speedy transport.

Before the baby leaves the maternity unit or home, the doctor responsible for the case should:

(*a*) Telephone the hospital to which the infant is being transferred. He should speak to a doctor, or senior nurse, giving the name and age of the infant and the provisional diagnosis. He should *not* leave messages with junior staff or telephonists.

(*b*) In cases of suspected intestinal obstruction, ensure that a nasogastric tube has been passed and that it remains in place. In cases of myelomeningocele, encephalocele, and exomphalos, cover the exposed tissues with sterile saline packs.

(*c*) Remember to explain to the mother what is happening to her baby.

(*d*) Ensure that the following are ready to go with the infant:

(i) A trained person who is competent to deal with any emergency that may arise on the journey.
(ii) The case notes or a full summary.
(iii) Any radiographs.
(iv) An operation consent form signed by a parent.

(v) A note to say whether or not the infant has been baptized; if not, give the parents' religion and the infant's baptismal name.

(vi) A sample of venous blood from the mother (5–10 ml. in a plain tube) is desirable. This may be used for cross-matching blood for transfusion if a sample of the infant's blood is not readily available. It is also valuable if the infant later develops signs of haemolytic disease.

During the journey, ensure that the infant remains (*a*) well oxygenated and (*b*) warm:

(*a*) The airway must be kept clear. If secretions accumulate in the pharynx they must be aspirated with a mucus extractor. If a nasogastric tube has been passed, the stomach should be aspirated every 10 minutes. If the lungs cannot function properly (for example, with a large diaphragmatic hernia), endotracheal intubation and positive pressure insufflation may be necessary. A supply of oxygen and efficient means of resuscitation must be available at all times in case the infant collapses in transit. The infant should be kept horizontal and lying on one side to prevent the inhalation of vomitus.

(*b*) Newborn infants, especially if premature, may cool remarkably rapidly even on relatively short journeys. Although some degree of hypothermia may have theoretical advantages, it appears in practice to be dangerous to infants undergoing emergency surgery. The rectal temperature, recorded with a low-reading thermometer, should not be allowed to fall below 95° F (35° C). The infant should be transported *either* in a portable incubator *or* in a portable cot. If in a cot, he should be well wrapped in blankets and hot-water bottles placed outside them. The main advantages of a portable incubator are that temperature, humidity, and oxygen concentration can be accurately controlled, and the infant can be easily observed. If one is not readily available, however, these refinements should be sacrificed for the sake of speedy transfer.

APPENDIX 2

Developmental, Vision, and Hearing Tests in Young Infants

(Reproduced from Ministry of Health Report No. 102 (London: H.M.S.O., 1960) by kind permission of the Controller of H.M. Stationery Office.)

Age: One month

POSTURE AND LARGE MOVEMENTS

Lies on back with head to one side; arm on same side outstretched, or both arms flexed: legs flexed, knees apart, soles of feet turned inwards.

Large jerky movements of limbs, arms more active than legs.

At rest, hands closed and thumb turned in. Fingers and toes fan out during extensor movements of limbs.

When cheek touched, turns to same side; ear touched, turns away.

When lifted head falls loosely.

Held sitting, head falls forward, with back in one complete curve.

Placed downwards on face, head immediately turns to side; arms and legs flexed under body, buttocks humped up. Held standing on hard surface, presses down feet and often makes reflex 'stepping' movements.

VISION AND FINE MOVEMENTS

Stares expressionlessly at brightness of window or blank wall.

Shuts eyes tightly when pencil light shone directly into them at 2·5–5·0 cm. (1–2 in.)

Follows pencil flash-lamp briefly with eyes at one foot.

Notices dangling toy or rattle shaken in line of vision at 10–15 cm. (4–6 in.) and follows its slow movement with eyes from side towards mid-line on level with face through approximately quarter circle, before head falls back to side.

Beginning to watch mother's nearby face when she feeds or talks to him.

Age: One month

HEARING AND SPEECH

Startled by sudden loud noises, stiffens, quivers, blinks, screws eyes up, extends limbs, fans out fingers and toes, and may cry.

Movements momentarily 'frozen', when small bell rung gently 7·5–12·5 cm. (3–5 in.) from ear for 3–5 seconds with 5-second pauses: may move eyes towards sound.

Stops whimpering to sound of near-by soothing human voice, but not when screaming or feeding.

Cries lustily when hungry or uncomfortable.

Utters little guttural noises when content.

(*Note.*—Deaf babies also cry and vocalise in this reflex way, but if very deaf will not show startle reflex to sudden noise.)

SOCIAL BEHAVIOUR AND PLAY

Sucks well.

Sleeps most of the time when not being fed or handled.

Expression vague, but tending to become more alert, progressing to smiling at about 5–6 weeks.

Hands normally closed, but if opened, grasps examiner's finger when palm is touched.

Stops crying when picked up.

Mother supports head when carrying, dressing and bathing.

Age: Three months

POSTURE AND LARGE MOVEMENTS

Now prefers to lie on back with head in mid-line.

Limbs more pliable, movements smoother and more continuous.

Waves arms symmetrically. Hands now loosely open.

Brings hands from side into mid-line over chest or chin.

Kicks vigorously, legs alternating or occasionally together.

Held sitting, holds back straight, except in lumbar region, with head held erect and steady for several seconds before bobbing forwards.

Placed downwards on face lifts head and upper chest well up in mid-line, using forearms as support, and often scratching at table surface; legs straight, buttocks flat.

Held standing with feet on hard surface, sags at knees.

VISION AND FINE MOVEMENTS

Visually very alert, particularly preoccupied by nearby human face.

Moves head deliberately to look around him.

Follows adult's movements near cot.

Follows rattle or dangling toy at 6–10 inches above face through half circle from side to side, and usually also vertically from chest to brow.

Watches movements of own hands before face and beginning to clasp and unclasp hands together.

Recognises feeding bottle and makes eager welcoming movements as it approaches his face.

Regards still objects within 15–25 cm. (6–10 in.) for more than a second or two, but seldom able to fixate continuously.

Age: Three months

HEARING AND SPEECH

Sudden loud noises still distress, provoking blinking, screwing up of eyes, cry and turning away.

Definite quietening or smiling to sound of mother's voice before she touches him, but not when screaming.

Vocalises when spoken to or pleased.

Cries when uncomfortable or annoyed.

Quietens to rattle of spoon in cup or to bell rung gently out of sight for 3–5 seconds at 15–30 cm. (6–12 in.) from ear.

May turn eyes towards sound; brows may wrinkle and eyes dilate, may move head from side to side as if searching vaguely for sound.

Often licks lips in response to sounds of preparation for feeding.

Shows excitement at sound of approaching footsteps, running bath water, etc.

(*Note*.—Deaf baby, instead, may be obviously startled by mother's sudden appearance beside cot.)

SOCIAL BEHAVIOUR AND PLAY

Fixes eyes unblinkingly on mother's face when feeding.

Beginning to react to familiar situations —showing by smiles, coos, and excited movement that he recognises preparations for feeds, baths, etc.

Responds with obvious pleasure to friendly handling, especially when accompanied by playful tickling and vocal sounds.

Holds rattle for few moments when placed in hand, but seldom capable of regarding it at same time.

Mother supports at shoulders when dressing and bathing.

Age: Six months

POSTURE AND LARGE MOVEMENTS

Lying on back, lifts up head from pillow.

Sits with support in cot or pram and turns head from side to side to look around him.

Moves arms in brisk purposeful fashion and holds them up to be lifted.

When hands grasped, pulls himself up.

Kicks strongly, legs alternating.

Can roll over.

Held sitting, head is firmly erect, and back straight.

Placed downwards on face lifts head and chest well up supporting himself on extended arms.

Held standing with feet touching hard surface bears weight on feet and bounces up and down actively.

VISION AND FINE MOVEMENTS

Visually insatiable: moves head and eyes eagerly in every direction.

Eyes move in unison: squint now abnormal.

Follows adult's movements across room.

Immediately fixates interesting small objects within 15–30 cm. (6–12 in.) (for example, toy, bell, wooden cube, spoon, sweet) and stretches out both hands to grasp them.

Uses whole hand in palmar grasp.

When toys fall from hand forgets them or searches only vaguely round cot with eyes and patting hands.

Age: Six months

HEARING AND SPEECH

Turns immediately to mother's voice across room.

Vocalises tunefully, using single syllables, for example, ka, muh, goo, der.

Laughs, chuckles and squeals aloud in play.

Screams with annoyance.

Shows evidence of response to different emotional tones of mother's voice.

Responds to baby hearing tests at 45 cm. (1½ ft.) from each ear by correct visual localization, but may show slightly delayed response.

[Tests employed—voice, rattle, cup and spoons, paper, bell; 2 seconds with 2-second pause.]—(*See* Sheridan, M. D. (1958). *Brit. med. J.*, ii, 999; and (1960). *ibid.*, ii, 453.)

SOCIAL BEHAVIOUR AND PLAY

Hands competent to reach for and grasp small toys. Most often uses a two-handed, scooping-in approach, but occasionally a single hand.

Takes everything to mouth.

Beginning to find feet interesting and even useful in grasping.

Puts hands to bottle and pats it when feeding.

Shakes rattle deliberately to make it sound, often regarding it closely at same time.

Still friendly with strangers but occasionally shows some shyness or even slight anxiety.

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