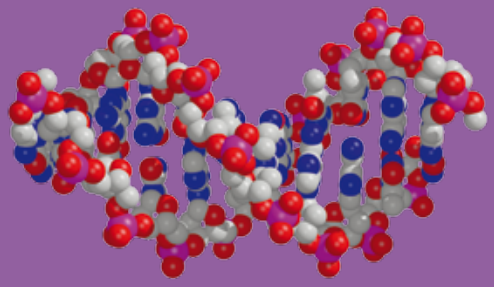
Portfolio Review Summary

Human Genetics
1990–2009

June 2010



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This summary describes a portfolio review of human genetics over the past 20 years, focusing on the Wellcome Trust's role within this landscape.

Historically, we have provided substantial funding for genetics – and, specifically, human genetics – research and have been an active player in the development of international research policy and strategy in the field. Support for genetics continues to be a cornerstone of our funding strategy today. Although we recognise that we are not the only funder in this rapidly evolving field, it is important to take time to reflect on how our input is making a difference and to learn from this.

Introduction and background

This portfolio review is part of a new approach we have developed to help us evaluate the impact of our funding on the landscape and inform potential future directions, taking a macro and long-term view. The analysis combines retrospective analysis (looking back) with prospective analysis (looking forward). Our aims in doing this were three-fold:

- to identify the key landmarks in and influences on the human genetics research landscape over the past two decades (1990–2009)
- to consider the key features of our impact on this landscape
- to speculate on the future direction of human genetics and where there may be opportunities for future strategy funding for us and other research funders.

We undertook three complementary streams of work (see the full report for further detail: **www.wellcome.ac.uk/humangeneticsreview**):

- landscape analysis, comprising bibliometric analysis (which describes patterns in scientific research publication outputs over time) and funding analysis
- narrative case studies describing the impact of a range of significant funding investments in people and science
- an expert group, convened to discuss and assess the key landmarks in the development of human genetics to date and to speculate on future directions.

We hope that this portfolio review will highlight areas where new and continued research is required and help to inform how we and other funders of human genetics research can provide support in the best ways.

Overview and key findings

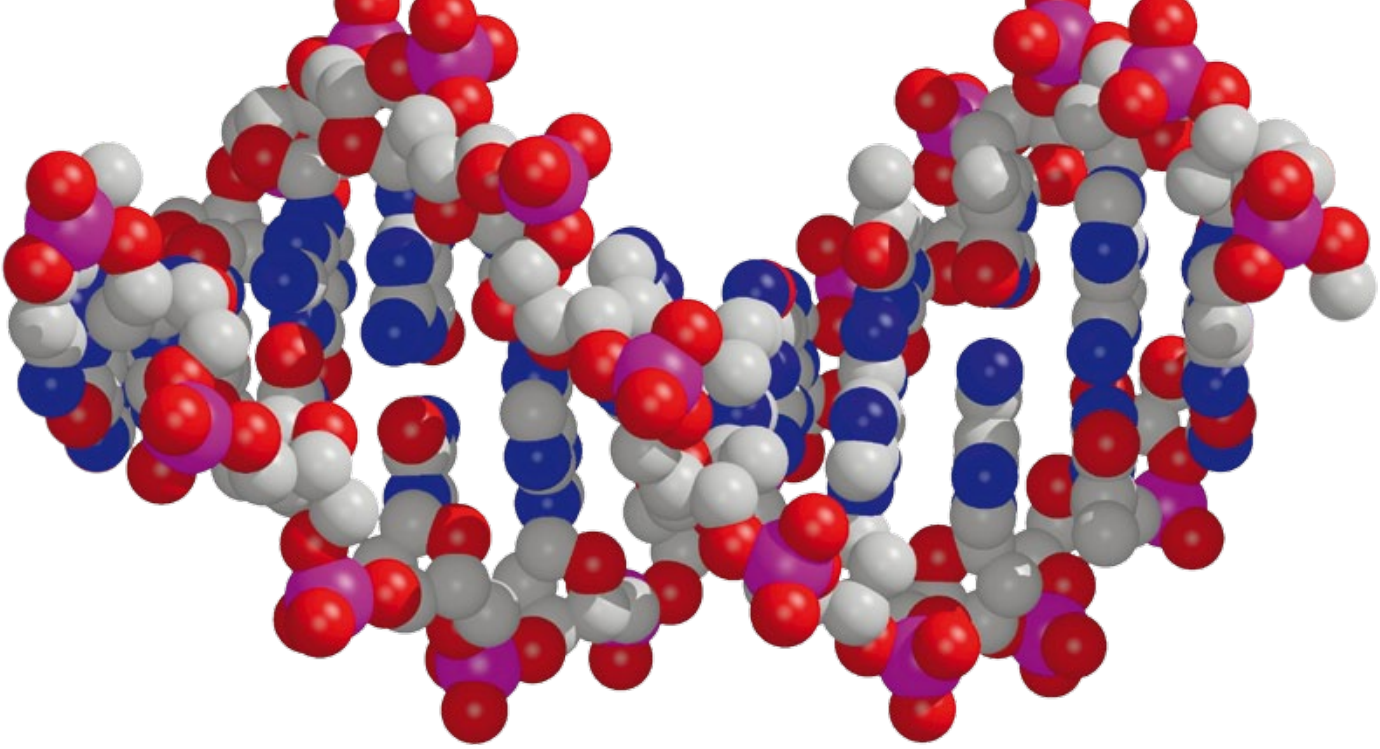
Between 1990 and 2009, we committed £740 million to research focused on human genetics (including funding to the Wellcome Trust Sanger Institute), accounting for approximately 10 per cent of our total funding commitment during this time. The drive to maximise the health benefits of human genome research remains a core component of our funding strategy today.

We have invested substantially in the field of human genetics over the past 20 years, from our contribution to the Human Genome Project at the Sanger Institute to our role in major research consortia and collaborations, such as the Wellcome Trust Case Control Consortium.

Our most notable impacts are thought to have been through:

- providing stable and sustained funding to build **research capacity and infrastructure** to support human genetics and genomics
- providing generous and sustained support for excellent researchers, who have made several key **advances in knowledge** and discoveries in human genetics that are likely to underpin future research and have broad impacts
- **forging partnerships and developing research policies** that have helped to shape the direction and openness of human genetics research and its findings.

Achieving progress in science is not always down to how much money is invested; it is about being bold, flexible and agile enough to recognise and act upon the requirements of a particular scientific area at the most opportune moments and giving support in the most appropriate ways. It is about working with excellent researchers and, where value can be added, with other partners to explore new areas and build critical mass.



Over the past 20 years, much of the research leading to perhaps the most significant breakthroughs in human genetics has resulted from 'big', technology-based, collaborative efforts. Now that the human genome has been deciphered, the paradigms within human genetics are shifting. The challenge is to harness and act upon this new wealth of knowledge and understanding to bring about real changes to the health and wellbeing of populations – and to support research in ways best suited to deliver these changes.

Challenges ahead

Looking to the future of human genetics with the help of our experts, we identified several needs and challenges to be addressed in further research and development, including:

- The need for **further underpinning research** ('wet lab') into basic biological mechanisms and genetics, including research into monogenic disease and the genetics of infectious disease.
- The need for **improved phenotypic definition and understanding among populations**, as well as a requirement to undertake more human genetic studies on non-medical traits. Our experts felt that it would be valuable to include quantitative phenotyping elements in new and existing cohort studies, such as the 2012 Birth Cohort Study. In addition, support for epigenetic research is thought to be key to enable further insight into phenotype and genotype studies.
- The need for funders to support **high-quality epidemiology**; well-powered cohorts and genome-wide association studies are required to generate robust associations between genetic factors and health outcomes and to secure adequate statistical power on rare variants. There may also be opportunities to **develop genetic components to existing cohorts and longitudinal studies**. Funding for longitudinal-based research needs to be sustained over the long term, however, and stability of funding is essential.
- It is timely to consider how funders and researchers might **engage with private genomics-based companies**, including those not focusing on primary research. Many private companies offering services directly to the public have a wealth of genomic data at their disposal; if consent and ethical considerations can be accommodated, such companies could offer much to research.
- Ensuring and facilitating the **involvement of clinicians** in human genetics-related research is an important strategic goal to help assure its benefits. This could also include pathologists; our expert group felt that the establishment of a **central pathology laboratory** in the UK would offer many benefits to human genetics research.
- The need to **guarantee global data sharing and open access** to genetic and genomic data remains an important principle. To enable this, it is crucial that key funders work together to develop sound governance frameworks to ensure that public and researcher confidence is maintained.
- The need to harness the **opportunities to develop strategic partnerships** with new and emerging science 'markets' across the world, both to develop fruitful research collaborations and to support the transfer of simple DNA-based technologies – for example, to assist in the management of communicable and non-communicable disease.

In all of this, it remains crucial that we, as funders, work with researchers to ensure that considerations of the ethical and social implications of human genetics research are integral to all that we do.

For further detail on the findings and methodology of this portfolio review, see the full report at **www.wellcome.ac.uk/humangeneticsreview**.

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