



2009

An Introduction to the
**Human Genetics
Commission**

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The Human Genetics Commission is the UK Government's advisory body on new developments in human genetics and how they impact on society and individuals' lives.

The Commission advises the Government on human genetics with a particular focus on social, ethical, legal and economic issues. One of its key roles is to promote debate and to listen to what the public and other stakeholders have to say.

HOW DOES THE COMMISSION WORK?

Since the unravelling of the human genome, our understanding of human genetics has continued to develop rapidly. The issues are complex, whether we are thinking, for example, about the possibilities raised by testing three-day-old embryos for particular genetic traits or using genetic information to identify criminals through DNA profiling. New advances are being made all the time.

It is the role of the Human Genetics Commission (HGC) to consider the ethical, social and legal implications of these and other genetic advances, to listen to the hopes and concerns of the public about these developments and to advise Ministers about what, if anything, the Government needs to do.

The HGC is a special body – funded by Government but firmly independent. It is made up of 21 members and includes experts in science, healthcare, ethics, law, and patient and consumer affairs. It is chaired by Jonathan Montgomery, Professor of Health Care Law at the University of Southampton.

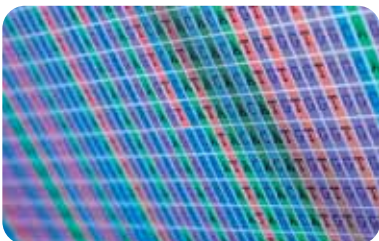
The HGC takes an inclusive and deliberative approach to its work and sees promoting debate as one of its key roles. It is committed to conducting its business openly and sharing its thinking with the public and stakeholders as much as possible. It holds all its plenary meetings in public (usually one in London and three in other cities across the UK each year). The audience is always given

The Consultative Panel is made up of people with direct experience of living with genetic conditions

the opportunity to ask questions or comment on any of the issues raised during these public meetings. For all of its major investigations it has held evidence gathering sessions and had in-depth public consultations. In addition, the Commission maintains regular communication with its Consultative Panel. The Panel, which is made up of people with direct experience of living with genetic conditions, acts as a sounding board for HGC reports and recommendations, and enriches the Commission's understanding of the impact of genetic conditions, and of policies that affect research, treatment or support, on individual lives.

At the same time the Commission has five monitoring groups, which keep a close eye on genetic discrimination, intellectual property, genetic databases, genetic services and the use of DNA for identity testing (including forensic) purposes.

The HGC's major reports, annual reports, interim documents and working papers are all published on its website: www.hgc.gov.uk.



WHAT ADVICE HAS THE COMMISSION GIVEN?

The Commission exists to keep Ministers, Parliament and the public properly informed about the benefits, limitations and implications of developments in human genetics and to think seriously and sensibly about any social, legal and ethical issues involved.

While the HGC is enthusiastic about the benefits of new developments in genetics, it looks carefully at their broader implications and has clear reservations about some of them. From its beginnings in 1999 a key consideration for the HGC has been the rights of individuals and, in particular, an individual's right to privacy.

All the HGC's deliberations start from the position that a person's genetic make-up is a personal matter and the results of their genetic tests should normally remain private. However, the Commission believes that society needs to achieve the right balance between an individual's right to privacy and the interests of others who might benefit from the use of personal genetic information, perhaps for research or medical treatment.

One of the Commission's major concerns has been about the predictive value of some genetic information. This creates the possibility of 'genetic discrimination': discrimination against people because of



their genetic inheritance. Concerns have included that employers might insist on their employees taking genetic tests or that companies providing life insurance might demand to know the results of genetic tests or insist on people having them.

The HGC campaigned for the present genetics and insurance moratorium – the voluntary agreement with the insurance industry that ensures insurers' use of predictive genetic tests is transparent, fair, and subject to independent oversight. The existing genetics and insurance Moratorium has recently been extended until 2014.

From early on the HGC has also monitored the use of DNA in criminal investigations. It has broadly supported the use of DNA analysis to identify and convict criminals, but has reservations about gradual inclusion, in the National DNA Database, of large numbers of people who may never have committed any crime. It is also concerned about possible police access to bio-medical databases and the potential for genetic samples to be used for further purposes beyond simply identifying suspects.

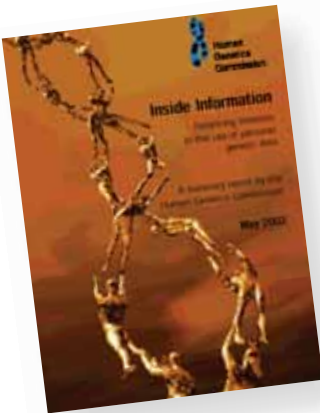
The HGC has been concerned for some time about the marketing and sale of genetic tests directly to the public

In its reports and its discussions the HGC has often taken a sceptical view of the more extreme claims surrounding some genetic applications. In the report “Making Babies: reproductive decisions and genetic technologies” the Commission was clear that “designer babies” – babies genetically engineered so that they display certain physiological traits such as eye colour – are not on the horizon. It has been equally sceptical about organisations claiming extraordinary results in tracing remote ancestors through DNA tests. Commission members have argued that while DNA can be a useful tool for genealogical research, it is inappropriate to rely on it alone.

The HGC has been concerned for some time about the marketing and sale of genetic tests that provide health related information directly to the public, often over the internet. While not arguing for a complete ban, the Commission believes that the public could be misled about the usefulness of genetic tests by aggressive marketing campaigns. There are fears that consumers might suffer unnecessary anxiety or make radical changes to their lifestyles as a result of valueless test results or seek further, unnecessary tests through the NHS.

OUR REPORTS

Inside Information (2002)



The Commission's first report, *Inside Information* (2002), looked at how people's personal genetic information was obtained, what it was used for and how it was stored.

It established important principles, such as respect for persons and genetic solidarity and altruism that have guided much of the Commission's subsequent work. The report made clear that:

“Society needs to achieve the right balance between an individual's interest in privacy and the interests of others in benefiting from the use of personal genetic information for medicine or research.”

A key recommendation contained in the report was that in most circumstances it should be a criminal offence to take and analyse someone's DNA without their permission. The Government agreed and a new offence was created in the Human Tissue Act 2004, which came into force in September 2006.

The report also recommended a series of safeguards to protect people's privacy and the ways their genetic information could be used. Particularly concerns were expressed about the need to protect children and families from the possible consequences of the misuse of their DNA. The report recommended that

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data for medical and research purposes should be routinely anonymised – i.e. separated from information that could link it directly to an individual.

'*Inside Information*' came out strongly against improper use of genetic information by employers. The Commission argued that there were other, better ways of detecting a person's risk of future ill health and genetic tests should not be demanded as a condition of employment.

There was also considerable concern about the National DNA Database, used by the police to identify suspects in criminal investigations. The Commission recommended that the Home Office and the Association of Chief Police Officers establish an independent body with lay members to have oversight of the work of the National DNA Database custodian and the laboratories that supply the profiles for the Database. Following publication of the report, the National DNA Database Strategy Board invited the HGC to put forward one, then two, of their Commissioners to sit on the Board. In 2007 the Home Office agreed and set up the independent DNA Database Ethics Group – another recommendation of the report.

Genes Direct (2003)



Genes Direct (2003) was concerned with the growing market in private DNA testing and identified the danger that some private testing companies might exploit people's lack of understanding or their fears about their health to sell genetic tests.

The Commission recommended that most genetic tests offering predictive health information should not be sold directly to the public. The Commission also had concerns about 'do-it-yourself' home DNA testing, such as tests to establish paternity or tests which claimed to show how to improve health. The Commission likened genetic tests to medicines: these are often only available with a doctor's prescription, although some are provided by pharmacists and others can even be bought in any shop or over the Internet. The Commission recommended that a similar arrangement should apply to genetic tests. The report also urged consumers to think twice before undergoing tests that they might not need or that could cause them unnecessary concern.

The HGC's advice to Government was that the existing regulatory framework could be developed to safeguard against bad and exploitative practice. Agencies such as the Medicines and Healthcare products Regulatory Agency, the Human Tissue Authority, the Office of Fair Trading and the Advertising Standards Authority all had an important role to play in protecting consumers.

Profiling the Newborn (2005)



In 2004, the Government asked the Commission to consider the arguments for and against setting up a scheme to take DNA profiles of every baby born in Britain. After considering the evidence, *Profiling the Newborn* (2005) came out clearly against this idea at the present time. The report – which was produced by a joint working party of the HGC and the UK National Screening Committee – said that there were strong ethical, legal and social barriers to mass genetic profiling of this kind and, in any case, the cost would be prohibitive at this stage. What is more, whilst genetic profiling may in the future have clinical potential, its effectiveness could not yet be judged.

The report also said that scientific knowledge was not sufficiently advanced to make use of the mass of genetic information that such an exercise would produce. However, the Commission agreed that mass profiling of babies in this way was technically feasible, and that there might be medical advantages in the future. A number of these advantages were identified: following analysis of the baby's DNA, doctors might be able to plan medical treatment in advance and people at heightened risk of a particular condition could be advised to adapt their lifestyles to minimise the environmental contribution to this risk. The Commission therefore recommended that the area should be kept under review and revisited in 2010.

Making Babies: reproductive decisions and genetic technologies (2006)



The report, *Making Babies* (2006) looked at what science can, and might in future, offer to enhance reproductive choice. The options open to couples having children – particularly those who know they might have children with an inherited genetic condition – has increased dramatically in recent years but these advances have brought with them new concerns. The report recognised that there is a need to balance reproductive autonomy – the right of people to make their own reproductive decisions – with the welfare of the child that may be born as a result and the wider interests of society.

The HGC felt it was important to recognise that there were limits to what such technologies could offer. It was not inevitable that pre-implantation genetic diagnosis (PGD) would lead to ‘designer babies’: children genetically engineered so that they have certain physical or behavioural characteristics such as beauty, intelligence or an outgoing personality. The complex nature of our genetic make-up and the interaction between genes and the environment in which we live makes being able to control outcomes simply by genetic selection highly unlikely.

While generally endorsing the benefits of new reproductive technologies, the Commission was concerned that there should be more

The report looked at what science can, and might in the future, offer to enhance reproductive choice



research and follow-up studies looking at their long-term consequences, for example, there was a need for studies into the health and development of children conceived by PGD. Similarly, research was needed into the experience of ‘saviour siblings’ – children resulting from embryos specially selected to help a brother or sister affected by a serious disease – to see what impact this had on them and their families.

The Commission accepted that many parents, particularly those who know that any children they have may be affected by an inherited genetic condition, might want to have access to prenatal screening. However, it felt it was important that genetic testing should not be presented as purely routine: there needed to be clear advice so that women did not feel they were on a ‘conveyor belt’ but felt empowered to ‘opt out’ of screening and testing if they so wished.

More Genes Direct (2007)



In 2007, recognising the rapid increase in the number of companies offering genetic tests directly to the public, the Commission published the follow-up report *More Genes Direct – A report on the developments in the availability, marketing and regulation of genetic tests supplied directly to the public.*

They found that there were significant variations in the effectiveness of consumer genetic tests, from tests that can accurately diagnose single-gene disorders such as cystic fibrosis to those that might provide only an indication that, all other things being equal, an individual had a marginally increased susceptibility to developing a multifactorial disease, such as cancer, cardiovascular disease or diabetes.

Because genetic test results have the ability to significantly alter the lifestyle choices an individual makes, the report recognised that there should be some way of distinguishing tests that provided well-founded and medically relevant information. However, despite the HGC's recommendations in the 2003 Genes Direct report, there continued to be no significant regulatory oversight of these tests and so claims about the efficacy and utility of the tests remain substantially up to the companies who supply them.

Working with experts, consumers' representatives and suppliers in the UK and Europe the Commission made a number of practical recommendations to supplement the findings of the earlier Genes Direct report. Firstly, genetic tests giving health-relevant information should be made subject to independent review before they are sold to the public. Secondly, a Code of Practice for tests not covered by the Directive should be developed, which would help to ensure high standards in the provision services. Finally, the marketing of genetic tests should be subject to greater oversight, and tests that provide health-relevant information should not be advertised directly to the public.



Genetic test results have the ability to significantly alter the lifestyle choices an individual makes

RECENT WORK & NEW WORK

Recent work (2007-09)

In 2007-08 the Commission underwent an independent review of its role, remit and effectiveness. The outcome of the review was very positive for the HGC, describing the Commission as ‘exceptional value for money’ and confirming that it has consistently ‘punched above its weight in terms of influencing public policy’. It also contained some helpful recommendations on how HGC can develop its ways of working to become an even more effective voice.

The publication of the Government’s proposals for a Single Equality Bill for Great Britain in July 2007 provided a stimulus for the HGC to draw together its advice on genetics and discrimination. In responding to the proposals, the Commission made specific recommendations on how the threat of unfair treatment on grounds of genetic difference might be addressed practically through legislation. In setting out the way forward, the Government subsequently drew attention to the importance of the monitoring role that the HGC plays in relation to genetic discrimination.

Concerned about the way in which growth in the consumer genetic testing sector was outstripping developments in regulation, the HGC has continued to bring together stakeholders and shape a consensus on the need for agreement about the principles of good practice in this area. Following on from the publication of *More Genes Direct*, in December 2007, in June 2008 the HGC held a meeting at which service providers, professional bodies, government officials, regulators, consumers' representatives and other experts from around the world came together to agree on the need for new guidance for this maturing market. HGC has now taken the lead in developing an international 'Common Framework of Principles' for consumer genetic testing services.

News of a new technique that can give information about a fetus from a sample of the pregnant woman's blood led the HGC to respond by holding a public discussion, supported by evidence from expert speakers in February 2008. Our conclusion – that the technique had significant potential advantages but should be subject to careful evaluation before being implemented as a clinical service – aimed to strike a balance between the benefits of non-invasive testing and the risk that the test will be misused. The HGC has continued to work with the UK National Screening Committee and the Human



Fertilisation and Embryology Authority to ensure consistency and the appropriate introduction of new antenatal screening techniques, following consideration of their potential social and ethical implications.

The HGC's Citizens' Inquiry into the forensic use of genetic information took place in the first few months of 2008 and the Citizens' conclusions were published – in their own words – by the HGC in July. These included recommendations for the database to be controlled by an independent statutory authority and the need for a campaign to inform the public about the database. In initiating this exercise, the HGC brought together collaborators with complementary expertise from the ESRC Genomics Forum and the Policy, Ethics and Life Sciences Research Centre (PEALS) in Newcastle to commission an innovative, citizen-led approach to public dialogue. Publication of the Citizens' findings was followed by a wider public consultation which, with a number of other sources of evidence helped the Commission's to develop its final advice and recommendations.

New work (2009 and beyond)

HGC has taken the lead in developing an international 'Common Framework of Principles' for consumer genetic testing services

Consumer genetic tests

Following on from 'More Genes Direct' and in recognition of the growing market for private genetic tests the HGC is leading the production of a common framework of principles for the responsible provision of genetic tests directly to the consumer. As the market for these tests is an international one, with some tests readily available from other countries over the internet, the HGC feels that the principles should have a similarly international scope and establish a consistency of approach across different jurisdictions.

To develop the principles the HGC is working with an international group of experts in law and regulation, clinical and molecular genetics and representatives of companies that supply tests and consumers who may buy them. The draft principles will be subject to extensive consultation before publication in their final form.

Forensic use of genetic information and the National DNA database

The Commission has often expressed concern that whilst the expansion of the National DNA Database raises significant concerns for the public there has been little debate about it either amongst the public or in Parliament.

Following the Citizens' Inquiry and the HGC's consultation in 2008, the Commission has established a working group to produce a report on the forensic use of genetic information and the National DNA Database. The report will draw together the Commission's deliberations informed by all the evidence collected through the Citizens' Inquiry, the consultation and from a variety of other sources, and will contain advice and recommendations on what the information on the National DNA Database should be used for and how the database should be managed.

Horizon scanning

Building on recommendations in the 2008 independent review report the Commission will be implementing improvements in its horizon scanning and business planning procedures. In February 2009 the Commission held a 'forward look' to identify what are likely to be significant trends organised under four themes: democracy and the citizen, biomedicine and society, technological possibilities in genomics and bioinformatics, and globalisation. In the light of this, the Commission will produce a separate business plan containing an assessment of the challenges ahead and proposals for new work to address these.

THE HGC CONSULTATIVE PANEL

The HGC Consultative Panel consists of over 80 people with direct experience of living with genetic conditions. Much of the Panel's work is by correspondence, with Panel members being sent summaries of reports that the HGC is writing or being alerted to issues that the Commission is discussing. In addition, there are annual meetings with the Panel to allow them to meet with Commissioners to discuss genetics issues in depth.

The Panel includes people who have experience of single gene, chromosomal or multifactorial conditions, which may have become apparent in either childhood or adulthood. Some people are affected themselves or are carriers, some have experience as a parent of a child affected by a genetic difference and some are carers for someone in their family who is affected. The Panel membership has a wide age range and includes people who live in England, Scotland, Wales and Northern Ireland.

HGC COMMISSIONERS

The Commission is made up of the following members:

- Chair:** Professor Jonathan Montgomery
- Vice-Chair:** Professor Sir John Sulston
- Members:** Professor Timothy Aitman
Professor Thomas Baldwin
Professor Sarah Cunningham-Burley
Dr Paul Debenham
Mrs Nicola Drury
Dr Frances Flinter
Mrs Ros Gardner
Professor John Harris
Mr Michael Harrison
Professor Lisa Jardine (ex officio)
Mr Alastair Kent
Professor Anneke Lucassen
Dr Duncan McHale
Dr Alice Maynard
Dr Lola Oni OBE

Representatives of the Chief Medical Officers for England, Wales, Scotland and Northern Ireland:

- Professor Angus Clarke (Wales)
Dr Paul Darragh (Northern Ireland)
Dr Rosalind Skinner (Scotland)
Dr Anita Thomas (England)

CONTACT US

Your views matter to us and contribute to our advice to Government.

We want to know what you think. For example: What genetic issues do you consider will be important in the next twenty years? Do you have experience of genetic services in this country you would like to tell us about? Have you bought a genetic test over the Internet?

You can write to the HGC Secretariat at:
The Human Genetics Commission
Area 605
Wellington House
133-155 Waterloo Road
London SE1 8UG

Alternatively, you can call, fax or email us:
Public enquiries: 020 7972 4351
Fax: 020 7972 4300
E-mail: hgc@dh.gsi.gov.uk

To receive HGC news and publications you can register your details with us by contacting the Secretariat.

We can also provide this leaflet in formats suitable for people with physical or sensory impairments and versions for those whose first language is not English.

Please contact the HGC Secretariat on telephone number **020 7972 4325** or by email at **hgc@dh.gsi.gov.uk**



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