



Human
Genetics
Commission

Choosing the future:

**genetics and reproductive
decision making**

July 2004

Choosing the future:

**genetics and reproductive
decision making**

July 2004

Contents

1.	Introduction	3
2.	Background and history	5
3.	Prenatal testing and clinical genetic services today	8
4.	Potential developments	16
5.	The issues: opportunities or threats?	21
6.	Choosing the future: summary and questions	28
Annex 1:	Terms of Reference and Working Group on Genetics and Reproductive Decision Making Members	33
Annex 2:	Members of the Human Genetics Commission	35





1. Introduction

- 1.1** Progress in molecular biology means we know an increasing amount about our genetic make-up. It also means that we can know more about the genetic make-up of our children. While many people welcome progress in genetic science and what it means for identifying and reducing the risk of having children with genetic disorders, some concerns have been expressed about the impact of this science not only on society generally, but also on our understanding of the meaning and value of human life.
- 1.2** The Human Genetics Commission (HGC) advises the UK Government on the ethical, legal, social and economic aspects of developments in human genetics as well as their effects on health and healthcare. In undertaking this project, HGC established a Working Group to examine the implications of developments in human genetics for the kinds of choices facing people having children and the wider social impact of these choices. The terms of reference and membership of the Working Group are given in Annex 1.
- 1.3** This discussion document summarises information and views that we have so far considered. It includes an examination of the history of genetics and reproduction, prenatal screening, diagnostic and genetic services, potential changes in the near future and some of the key arguments and concerns about where society is heading in this area. We also asked HGC's Consultative Panel for their initial views on some of these issues. The Consultative Panel was established by HGC to act as a sounding board for our reports and recommendations, as well as to give us insight into the types of issues and concerns facing people affected by a genetic disorder. The panel is made up of over 100 people with direct experience of living with a genetic disorder. A number of their comments have been included in this document as direct quotations in order to illustrate some of the issues discussed.
- 1.4** This work needs to be set alongside a number of other projects and reviews in several related areas. These include:
- the House of Commons Science and Technology Committee's inquiry into human reproductive technologies and the law;
 - the Department of Health's review of the Human Fertilisation and Embryology Act 1990;
 - the Human Fertilisation and Embryology Authority's review of the case for using preimplantation genetic diagnosis for tissue typing purposes;
 - a range of initiatives aimed at identifying the type and level of information required to ensure informed consent and decision making in the prenatal screening and diagnostic contexts;
 - a pending judicial review on criteria for abortion for fetal abnormality;
 - and developments of a better understanding of public attitudes to genetic science and its applications.

In this document, we are not addressing any of these issues directly, though outcomes of these reviews and projects will of course inform our thinking.



The consultation process

July – Oct 2004: this consultation on the key issues under consideration

Nov – March 2005: HGC to consider responses and gather additional information

April – July 2005: prepare and draft the report

Late 2005: report to Ministers

- 1.5** This document summarises information and issues HGC has considered to this point. We welcome comments on any aspect of this document, and especially the areas where specific questions are raised – these can be found at the end of the document (page 29). In particular, we are seeking additional viewpoints and evidence to inform our thinking.
- 1.6** This is one stage of our evidence gathering. We will continue to hear from a range of individuals and organisations about their views on particular issues they have brought to HGC's attention. In addition, we will be commissioning a review of research on public attitudes on this topic.



2. Background and History

2.1 Today, genetic screening and diagnostic procedures, and genetic counselling, emphasise the provision of information to facilitate informed decision making. But this has not always been the case. Early last century, some countries had eugenic policies and employed coercive practices aimed at the so-called “improvement” of the human gene pool. Some feel that current practices retain this legacy.

Our eugenic past

2.2 The term eugenics, literally well born, was coined by Francis Galton in the 1883. He defined it as “the science of improving stock, not only by judicious mating, but by whatever tends to give the more suitable races or strains of blood, a better chance of prevailing over the less suitable”.

Eugenics spread throughout the industrialised world as a popular movement, a research programme and, in many countries, a coercive legislative programme. The aim of supporters of eugenics was to limit reproduction of those considered “unfit” and encourage the “fit” to have lots of children.

2.3 The scope of what was done, however, varied widely. In Britain, the movement was driven by a belief that the growth of medical care and social welfare policies were allowing the “unfit” to survive and reproduce, hence causing a national “degeneration”. Falling birth rates amongst the middle and professional classes were thought to further promote this trend. This led to the *Mental Deficiency Act (1913)* which allowed the incarceration of the “feeble minded” to prevent them having children. Other countries (including the USA, Canada and many European countries) went further and had legislation which permitted forced sterilisation of the “unfit”. In the 1920s and 1930s, Sterilisation Acts were adopted with variations in many American states and were targeted at the “feeble minded, insane, criminalistic, epileptic, inebriate, diseased, blind, orphans, tramps, the homeless and paupers”.

2.4 The most well known application of eugenics was by the Nazis in Germany during the 1930s and 1940s. Eugenics was initially used to justify the killing of disabled children, and then disabled adults. This was later extended to a range of populations considered “unfit” including Jews, gypsies and homosexuals, and those from families that carried inherited disorders such as Huntington’s disease.

2.5 It was during the 1940s that many geneticists began to distance themselves from eugenic policies. Nonetheless, in the USA, eugenic laws remained in force in certain states until the 1970s. In Sweden and some other European countries, people with learning difficulties and the inmates of mental institutions continued to be forcibly sterilised until about the same time.

“A doctor once suggested to my mother that I should be sterilised when I was 16 years old... He thought it would be for the best so I would not pass on the disease. Fortunately my parents felt the same way as me and it didn’t happen... When my mother was pregnant with me a Ward Sister told her she should be ashamed of herself and that she would never carry me full term anyway ...” Consultative Panel Member



Prenatal genetic screening and diagnosis

- 2.6** By the 1950s, knowledge about inheritance had developed in such a way that genetic clinics were being set up to offer diagnosis and advice and counselling to couples about passing on genetic disorders that might run in the family. A number of choices could be offered including:
- that people take the chance that their child may be born with a genetic disorder;
 - to conceive children through artificial insemination by sperm donor;
 - to not have children or adopt.
- 2.7** Clinical genetics relied on professionals taking a family history in order to estimate the risk of developing a genetic disorder and passing this to any children. There was no way of identifying who had inherited the genetic disorder except by the appearance of the disease itself. Later, with developments such as the identification of biochemical markers and the identification of genes and the mutations which led to diseases, genetic testing became possible for a growing list of conditions.
- 2.8** One of the earliest developments in testing for a genetic disorder that could be identified, in this case in newborns, was the Guthrie Test for phenylketonuria (PKU). The Guthrie test is a blood test that newborns receive and involves the pinprick of the baby's heel. Blood is then squeezed out and put on a card which is then tested. PKU is one of the commonest inherited metabolic disorders and affects 1 in 10,000 births in the UK. It causes severe learning problems but these problems can largely be avoided by dietary treatment from infancy. It is through this diagnosis that parents learn they are carriers of PKU and will have a one in four chance that any future children will be affected. Screening is now offered for all newborns.
- 2.9** It was during the late 1950s and 1960s that prenatal tests (i.e. on the unborn baby) began to be developed. One of the earliest of these was for testing the sex of a fetus which could be useful where that baby had a high risk of having a sex-linked condition such as Duchenne muscular dystrophy. It later became possible to identify and diagnose some chromosome disorders including Down's syndrome which has an incidence of around 1 in 600 births. Its occurrence – like that for many other chromosome disorders – rises with the age of the woman. Following this was the development of tests for sickle cell disease, a serious blood disorder. Many of these tests, however, were not widely offered or available until the mid 1990s.
- “I was offered no prenatal screening twenty-five years ago ... As a carrier of a genetic disease, I would have liked some screening offered, but twenty-five years ago screening was not available for the disease that I carry. However it is available now, which is a marvellous development.”*
Consultative Panel Member
- 2.10** Today there are genetic tests for a number of the single gene disorders and tests to detect chromosomal disorders. There is also considerable research being done to develop a better understanding of multifactorial diseases that involve the interaction of multiple genes with the environment such as in many cases of heart disease. There have also been advances in techniques for screening and diagnosis of single gene disorders in pregnancy, as well as for pre-implantation genetic diagnosis (PGD) where in vitro fertilisation (IVF) is combined with the testing and selection of preimplantation embryos which allows only unaffected embryos to be transferred to



the woman. There have also been improvements in treatments of the symptoms of some serious single gene disorders such as cystic fibrosis and sickle cell disease, with more people now living into adulthood.

Single gene disorders are inherited disorders caused by a mutation of a single gene. The degree of severity can vary between individuals with the same single gene disorder. Examples include cystic fibrosis and sickle cell disease.

In **chromosomal disorders**, the disorder is caused not by a single gene but by either an excess or deficiency of the genes contained in a chromosome or of whole chromosomes. For example, people with Down's syndrome have an extra copy of chromosome 21. As with single gene disorders, how chromosomal disorders manifest varies widely between individuals.

X-linked or sex linked genetic disorders refer to a form of genetic inheritance where the gene responsible is carried on the X chromosome. Most of them such as Duchenne muscular dystrophy or X-SCID, a serious disorder of the immune system, predominantly affect boys.

Multifactorial diseases are caused by a combination of particular variations in more than one gene combined with environmental factors. Most cancers and coronary heart disease are multifactorial. As yet, only a small number of the gene variation involved in such diseases have been identified. Some of the diseases that are generally multifactorial also have a small number of cases where the disease is associated with mutations in a single gene (in the same way as single gene disorders). For example, only about 5% of breast cancers have been associated with mutations in the BRCA1/2 genes.

- 2.11** Some people see current practices in the screening and diagnosis of genetic conditions as reflecting eugenic beliefs because they feel the overall aim is to prevent the birth of disabled children. In addition, some feel that the provision of the services themselves convey a negative message about the social acceptability of genetic disorders and/or disability. However, others contend that in genetic testing and counselling, the emphasis is on providing information and allowing people to make choices about the tests they have and other courses of action they may wish to follow. On this basis, it is claimed that recent developments are not eugenic but instead reflect changing attitudes about choice and the right of individuals to make decisions they believe are best for them and their families.

“At least 10 years ago a consultant told me that if my wife became pregnant my wife could be tested for the gene. The clear implication being she could seek an abortion.”

Consultative Panel Member

“Now anyone with a history ... in the family can ask for screening by a simple blood test or can have a test taken during pregnancy of the unborn child, to find if the child is affected.”

Consultative Panel Member



3. Prenatal testing and clinical genetic services today

- 3.1** Screening for fetal abnormality has become a significant part of prenatal care for women in the UK today. Nearly every pregnant woman is offered a range of screening and diagnostic tests during the course of her pregnancy. People with known genetic disorders or family related medical histories may face particular issues when considering having children and may use the services of specialist genetic centres.

Population prenatal screening and diagnosis

- 3.2** Prenatal *screening tests* can help identify women who are at increased risk of having a baby with a disorder. Those women identified at increased risk are then offered a *diagnostic test*. Diagnostic tests can tell a woman if the baby has a chromosomal abnormality such as Down's syndrome, or a genetic disorder like cystic fibrosis. Most women will be offered some testing while they are pregnant. The offer of screening and diagnostic tests could potentially be taken up by about 700,000 women per year, the number of births in 2003.

Prenatal screening is a public health service which offers pregnant women a test to see if the baby is at significant risk of having a particular disorder such as Down's syndrome.

Prenatal diagnosis is a type of test which is person and condition specific that aims to provide a diagnosis of the particular condition the baby might have.

- 3.3** A diagnostic test is used to determine the presence or absence of a specific condition and may or may not be invasive. Invasive tests such as amniocentesis and chorionic villus sampling (CVS) involve inserting a needle into the amniotic sac or the chorion to take a sample. Both techniques carry with them a small risk of miscarriage. In the UK, recent figures from the Association of Clinical Cytogeneticists indicate that there were about 30,000 amniocentesis tests and more than 8,000 CVS tests conducted in the year period 2002/03. This number of tests can be expected to increase given the recent NHS commitment to ensure that all women, irrespective of age, are offered a screen for Down's syndrome. If prenatal diagnosis confirms that the baby has an abnormality, this information can be used to prepare for the birth of an affected baby or to decide whether or not to terminate the pregnancy.*

"I think prenatal screening is an important service for parents. This must be backed up by good support if a problem is discovered. Support in termination, or support if the parents want to continue with the pregnancy." Consultative Panel Member

* In 2002, there were 1863 abortions performed for fetal abnormality. Some of these abnormalities would have been genetic in origin. This was out of a total of 175,600 abortions performed overall in that same year. Over the last 5 years, on average about 1% of abortions have been for fetal abnormalities.



“These tests may be useful to prepare for a child with a genetic or medical problem even if the parents would not consider termination under any circumstances ...” Consultative Panel Member

3.4 Current clinical best practice rejects the notion that women will necessarily end a pregnancy after the identification of a fetal abnormality. Health policy is increasingly focused on offering women choices about whether to take up the offer of prenatal screening, to go through diagnostic procedures when there may be an increased risk of having a baby with a genetic disorder, and then for some, whether or not to terminate the pregnancy. For others, the identification of an affected pregnancy means they can plan how best to care for the child after its birth.

“I had my children in the early 1970s when there were no prenatal screening services whatsoever... Services have changed dramatically over time and fortunately women have a choice to take up prenatal screening tests.” Consultative Panel Member

3.5 One area of current development concerns the timing and type of information people receive prior to screening and diagnostic tests, as well as the support they get before and after making a decision. This, however, remains contentious as while many would argue that improving and increasing levels of information, as well as offering a wider range of choices, is a positive move, others have concerns. These concerns are raised on the basis that too much information and choice can become a burden and that screening sends out particular signals about whether or not certain disabilities and/or genetic disorders are acceptable.

“I cannot support screening for any condition unless the purpose is to ensure treatment ... I find it discriminatory and a cover-up for cost-cutting... a further example of how the elimination of disabled foetus is seen as more important than the birth of a child – disabled or not. As usual our right to being regarded as human beings is being denied.”
Consultative Panel Member

3.6 There are a number of organisations that provide information and a number of initiatives currently in place to look at levels of patient and professional information and education needs. These include:

- Contact a Family [www.contactfamily.org.uk], who provide support and advice to parents of children with disabilities. They have also produced an information pack for health professionals with practical suggestions on how best to communicate information and offer appropriate support to parents at significant times from pregnancy to pre-school;
- The Genetic Interest Group [www.gig.org.uk], a national alliance of organisations that support individuals and families with genetic conditions. Its primary goal is to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them;
- Antenatal Screening Web Resources [www.antenataltesting.info], a website with the aim of providing information to enable people to make choices about whether or not to have prenatal tests, and outlining the options available if they are diagnosed as having an affected pregnancy;
- and the UK National Screening Committee [www.nsc.nhs.uk] which is conducting a project on professional training for informed choices in prenatal and newborn screening.



- 3.7** Issues of equity in the provision of prenatal screening services across the UK have been raised with HGC. This is partly because different tests are offered between, as well as within, England, Northern Ireland, Scotland and Wales (Table 3.1 page 15). In 1996, the UK National Screening Committee was established in order to advise on what to screen for, by what methods, and how this may be done in a uniform way across the UK at a population level. A key aim is to address issues about equitable access and standards.

The UK National Screening Committee (www.nsc.nhs.uk)

The UK National Screening Committee (NSC) was established in 1996 and advises Ministers, the devolved national Assemblies and the Scottish Parliament on all aspects of screening policy. In forming its proposals, it draws on the latest research evidence and the skills of specially convened multi-disciplinary expert groups which include patient and service user representatives. The NSC assesses proposed new screening programmes against a set of internationally recognised criteria covering each condition, the test, the treatment options and effectiveness and acceptability of the screening programme.

- 3.8** While the focus in this document is on screening and diagnostic tests offered to pregnant women, population screening and diagnostic tests conducted on newborn babies for particular genetic conditions may also have a significant impact on the decisions women and couples make about having children in the future. Currently, newborn screening is carried out by using a bloodspot taken from the heel of a newborn baby in the first few days of life. Across the UK, babies are routinely tested for PKU, a rare metabolic disorder which can cause severe learning problems but can largely be avoided by dietary treatment from infancy. They may also be tested for a range of other conditions such as cystic fibrosis and/or sickle cell disease. Newborn screening is relevant to questions about reproductive choice because parents' knowledge that their child has an inherited genetic disease has implications for the types of future choices they may make.

Genetic services and reproductive decision making

- 3.9** People who know they have, or are carriers of, genetic disorders can be faced with difficult decisions when considering having children. While some people might discuss their options with their GP or obstetrician, others might be referred to or seek out specialist genetic services. One of the key aims of genetic services is to help people who are at risk of developing, carrying or passing on, a genetic disease to be fully informed about the risks, their implications and their options for the future, including reproductive options.
- 3.10** Specialised genetic services were developed in the UK as local centres of expertise (Regional Genetic Centres), often based on former Regional Health Authority boundaries or, as in Scotland, associated with teaching centres. Each generally serves a population between two and five million, and is multidisciplinary; they might include doctors, specialist nurses, genetic counsellors and laboratory scientists. Related laboratories form the basis of the UK Genetic Testing Network (UK GTN) which was established just over a year ago. UK GTN aims to provide high quality, equitable laboratory services for people who require genetic advice, diagnosis and management. It also seeks to co-ordinate the evaluation, commissioning, funding and prioritisation of services for inherited genetic disorders.



3.11 Testing for inherited blood disorders including sickle cell disease, thalassaemia and haemophilia is generally provided outside regional genetic services through separate specialist services. For example, laboratories within the UK Haemophilia Genetic Laboratory Network provide testing for haemophilia and related inherited bleeding conditions and are usually located in comprehensive care haemophilia centres. Similarly, counselling for these conditions is often provided by specialist counsellors working only with people with these specific genetic disorders.

Genetic Counsellors are specially trained professionals who generally come from medical or nursing backgrounds and have first hand knowledge of genetic disorders and their impact.

Prenatal Genetic Diagnosis (PND) is a type of test which is person and condition specific and aims to provide a diagnosis of the particular **genetic** condition the baby might have.

Preimplantation Genetic Diagnosis (PGD) is a technique where embryos created outside the body by IVF can be tested to see if they have a genetic disorder. One or two cells are removed for testing from the embryo at the 6-10 cell stage (day 3 of development). Implantation into the woman's uterus will generally only be attempted for embryos without the genetic disorder.

3.12 People who have a significant chance of having a baby with an inherited genetic disorder may have discussed options with a genetic counsellor. The aim of genetic counselling is to be non-directive in supporting people to make their own decisions based on their own set of circumstances. Counsellors may provide a list of possible options for people to choose from when deciding about having children. These options might include:

- to have a pregnancy with or without genetic testing;
- to not have children or adopt;
- or to use donor eggs, sperm or embryos so as to avoid the risk altogether of passing on the inherited genetic disorder.

However, for many, using donors or adoption will be a last resort as people tend to prefer have children using their own eggs or sperm.

“Genetic counselling has been a major impact in our lives as it stopped me believing that I was to blame for not looking after myself well during my pregnancies.” Consultative Panel Member

“Services have most definitely changed over time as there is a greater awareness and understanding of genetic issues and approaches... When I first went to the genetic service, the approach was very technical... it has definitely become much more person friendly as well as more informed on genetic conditions.” Consultative Panel Member

3.13 Another option in some circumstances is to proceed with a pregnancy and to have the baby tested using prenatal genetic diagnosis (PND). Figures from the Clinical Molecular Genetics Society indicate that there were about 2,600 prenatal genetic tests carried out for conditions including cystic fibrosis and Duchenne muscular dystrophy for the year period 2002/03. The purpose of these tests is to determine whether the baby has the genetic abnormality being tested for. If the baby is found to be affected, people can then use this information either to prepare for the birth of a child with a genetic disorder or terminate the pregnancy.



*“Screening services have changed over the last 30 years when very little was on offer. Tests should and **must** be offered at the **earliest** possible period during pregnancy and no judgement should be made on parents who opt out of testing for whatever reason.”*
Consultative Panel Member

*“Routine genetic testing should be available prenatally **but** always accompanied by counselling in the event of a genetic illness being discovered.”* Consultative Panel Member

3.14 Preimplantation genetic diagnosis (PGD) developed in the 1980s, provides another option. PGD was developed primarily in response to requests for help from people at risk of passing on a serious genetic disorder to their children. This technique has now been practised for several years and has developed because of the availability of IVF and new genetic testing techniques. In the UK, since 2001, there have been about 45 live births using about 430 PGD cycles. The Human Fertilisation and Embryology Authority regulates PGD as well as a range of other assisted reproductive technologies. There are currently 12 Centres licensed to carry out PGD.

Human Fertilisation and Embryology Authority (www.hfea.gov.uk)

The Human Fertilisation and Embryology Authority (HFEA) was established in August 1991 following the passing of the *Human Fertilisation and Embryology Act 1990* (HFE Act).

The HFEA's principal tasks are to

- License and monitor clinics that carry out in vitro fertilisation (IVF) and donor insemination
- License and monitor research centres undertaking human embryo research
- Regulate the storage of gametes and embryos
- Maintain a Register of licensed assisted conception treatments and donors used in these treatments
- Produce a Code of Practice giving guidance on the proper conduct of licensable activities and the proper discharge of the functions of licensees.

3.15 PGD was first successfully used in 1990 to produce two sets of twin girls where the parents were at a high risk of passing on a serious X-linked disorder were they to have sons. The first autosomal recessive disorder, that is, where both parents are carriers and any child has a 25% of having the condition, for which PGD was used, resulted in the birth of a child unaffected by cystic fibrosis.

3.16 PGD is currently being offered for three major categories of disease including:

- to determine the sex of the embryo with the aim of avoiding sex-linked disorders such as Duchenne muscular dystrophy;
- to identify embryos with single gene disorders such as cystic fibrosis;
- and to identify embryos with chromosomal disorders, where a technique called fluorescence in situ hybridisation (FISH) can be used to identify or confirm abnormal chromosomal rearrangements.



3.17 The use of PGD is not uncontroversial. In particular, concerns have been expressed about the range of conditions for which this technique is currently used and may be used in the future. There is some concern that as the number of identifiable genetic disorders extends, so too will the uses to which PGD is put. One area of controversy is the use of PGD not to merely avoid a genetic disorder, but to identify embryos having a particular tissue type and so select for a “saviour sibling”. These embryos could then be transferred into the woman’s uterus in the hope that the resulting baby will be able to provide matched tissue (blood from the umbilical cord or, potentially, bone marrow, in order to extract stem cells) for the treatment of an existing sibling affected by a serious or life-threatening disorder. The HFEA is in the process of conducting a review of the case for using PGD for tissue typing and we will be taking this into account when writing our final report.

“The advantage of PGD is that it eliminates the need for PND and possible TOP (termination of pregnancy).” Consultative Panel Member

3.18 Techniques such as these are physically and emotionally demanding on the people involved and do not bring with them guarantees of success. PGD is also more expensive than standard IVF treatment owing to the complex technologies needed to test an embryo while still maintaining it in a suitable state for transplantation. In addition, with IVF, on average there is a baby born in 20% of cases after implantation with fresh embryos and 12% with frozen ones. According to research, just over half of women under 34 will have conceived after five attempts at IVF. Thus access to techniques such as PGD are currently limited by the rate of successful outcomes and the ability of patients to meet the costs themselves, or by the willingness of Primary Care Trusts and other relevant funding bodies to fund the treatment.

Current Treatment Options

3.19 Over the past two decades, there have been significant developments in the treatment of the symptoms of some single gene disorders such as sickle cell disease and cystic fibrosis, leading both to increased life expectancies and improvements in quality of life. There are also a limited number of conditions for which treatment is available such as PKU, where treatment consists of dietary intervention and/or giving the individual an enzyme that makes up for what their own body cannot produce. Another example includes a very rare genetic disorder that prevents individuals from making leptin. Leptin is involved in switching off appetite when we eat. These children are constantly hungry and become morbidly obese. Following identification of the gene and its mutations, a handful of children who have this condition have been treated. They develop appetite control and dramatically lose weight. However, treatments of this kind are not usually this straightforward.

3.20 Most single gene disorders are not curable though there have been a couple of exceptions: bone marrow or stem cell transplantation for β thalassaemia, a blood disorder; and gene therapy or transplantation for X-SCID, a severe combined immunodeficiency disorder where boys are vulnerable to infectious disease. It is important to recognise however that such treatments remain at an experimental stage. While bone marrow transplantation offers a possible cure for X-SCID for example, and the chances of success are very good (90%+) when a fully matched sibling donor is available, only a third of patients have a fully matched donor. The chances of success from other donors such as people not related and parental donors are reduced (around 60%). In addition, even where the transplant is successful, there may be ongoing problems with the



person's immune system and side-effects years after the transplant. Gene therapy clinical trials for X-SCID indicate that it might be possible to correct the immune system for this condition in the long term.

- 3.21** The early treatment of symptoms for some genetic disorders in improving health outcomes has been part of the rationale for the introduction of a range of newborn screening programmes. If treatments for such symptoms improve, or if these diseases become curable, there may well be changes in the reproductive decisions made by parents who are at risk of transmitting genetic diseases to their children.
- 3.22** In conclusion, women and couples have a range of decisions to make before and during pregnancy: whether or not to have screening, and/or genetic diagnosis if medically indicated; what to do if a genetic disorder is identified or if there is a known risk in the family; and whether or not to continue with pregnancy. If they continue with the pregnancy, other decisions need to be made about how to use that information to improve health outcomes for the baby. Such decisions sit within a broader framework of services, funding provision, professional norms and social values. Each of these processes is contested and this will be picked up in Chapter 5.

**Table 3.1: Established Antenatal Screening Programmes for Genetic and Chromosomal Conditions (by country)**

Genetic Disorder	England	Northern Ireland	Scotland	Wales
Cystic fibrosis	No, will be offered for newborns	No, offered for newborns	Edinburgh and the Lothians only; newborn screening across Scotland offered	No, offered for newborns
Down's syndrome/ Aneuploidies	Yes, detailed protocol being implemented.	No, not routinely	Yes, offered to all women	Yes, detailed protocol being implemented
Fetal anomalies – ultrasound scan	Yes	Yes	Offered in about 60% of maternity units but under review	Yes, detailed protocol being implemented
Inherited blood disorders – Sickle cell diseases and thalassaemia;	Screening for thalassaemia recommended. Enhanced screening for haemoglobin variants being implemented in all high prevalence Primary Care Trusts (2004-5). In other areas, recommendation is to offer testing to those at high risk passing on the disorder (2005-6).	Not currently offered Screening for thalassaemia recommended.	Not currently offered universally but under review. Screening for thalassaemia recommended.	Screening for thalassaemia recommended.
Neural tube defects	Yes	Not routinely offered other than where a family history	Yes	Yes
Rhesus haemolytic disease	Yes	Yes	Yes	Yes



4. Potential Developments

- 4.1** Throughout HGC discussions on this topic, we have been made aware of concerns about “designer babies”, “playing God” and the issue of genetic determinism. One way to address some of these concerns might be to begin to consider the implications of new technologies and social changes before they arrive. For example, in genetic science, there is a widespread tendency to overestimate what is possible. Sometimes particular techniques that are remote possibilities, for example, are talked about as if they are already a reality. The aim of this chapter is to focus discussion on what is already possible and technologies that we can be reasonably certain will be available in the foreseeable future.

“Over time technology has vastly increased the ability of professionals to understand more. However, the greater the knowledge the bigger and more complex the questions.” Consultative Panel Member

Population Screening

- 4.2** Most women who are pregnant in the UK today will be offered some type of prenatal screening, and diagnosis if medically indicated. While the risks are low with screening, there is a small risk of miscarriage with the use of invasive diagnostic tests such as amniocentesis and CVS. Work is underway to try and develop less invasive and so safer techniques for prenatal diagnosis. For example, it has been known for some time that cells and DNA from a fetus pass into the mother’s blood circulation. If it did become possible to collect and to test these fetal cells, this would allow screening and diagnosis by taking a blood sample from the mother or testing cells from the cervix in early pregnancy.
- 4.3** There are also moves to develop technologies, such as DNA analysis on gene chips, so that a much larger number of gene mutations and variations could be assessed. Traditionally, screening and diagnosis have relied on looking at one or a limited number of genes. Gene chips could potentially be used, in the future, to monitor the entire genome on one chip so that doctors and other health professionals could understand how hundreds or even thousands of genes interact.
- 4.4** Technologies like these have the potential to make screening and diagnosis simpler and faster, and potentially may be able to deliver results in the first trimester (three months) of pregnancy. They should be less invasive and could potentially have much lower operating costs than current methods. They could also make it more feasible to screen the whole population for relatively rare conditions. We have heard concerns that such developments might lead to arguments being made for prenatal screening programmes to be used to screen for an even wider range of genetic disorders than is currently the case.
- 4.5** A range of other potential developments may have implications for choices people make. These include a growing list of conditions for which newborns are screened. The HGC is currently working with the UK National Screening Committee to consider the case for and against genetically profiling newborn babies. They will be reporting to Health and Science Ministers by the end of 2004. At first, this may not seem relevant to this discussion, but if genetic profiling were adopted for the whole population within the next 15-20 years, those profiled and their parents may face a range of complex issues when it comes to making decisions about having



children as these profiles could reveal information that might be significant for future reproductive choices.

Social Developments

- 4.6** Over the past few decades, there have been a number of important social changes relevant to reproductive decision making. First, there is a trend for women to have fewer children and these later in life. This is important for two reasons; because older women are more at risk of having babies with chromosomal and some other abnormalities, and the ability to have children declines as women get older. If this trend of women wanting to have babies at older ages continues, it has been suggested to us that there could be a number of effects such as increased pressure to extend women's fertility beyond menopause. Current knowledge makes it possible for post-menopausal women to be given hormone treatment so that they can carry a pregnancy after IVF.
- 4.7** There are a number of other techniques that could potentially be used to extend fertility beyond menopause. These include:
- egg and embryo storage;
 - the storage of ovarian tissue;
 - and the splitting of an embryo* and the storage of the extra IVF embryos produced for future implantation.

Embryo splitting involves separating cells in early preimplantation embryos. At this stage in development, each cell is able to develop into a complete embryo. The products of such embryo splitting would be genetically identical, like identical twins. Such technologies may also be useful for women who are at high risk of premature ovarian failure. As present, embryo splitting is not permitted in the UK.

- 4.8** There are a number of developments that may affect men's fertility. For example, research into novel methods of male contraception continues. Possibly more significant, however, is that there has been a number of reports signalling a reduction of human sperm counts over the last century. As yet, it is unclear whether this is a general phenomenon or what the extent of such a change might be, or, indeed, its cause. If there were such a decline, there may be an increasing use of donor insemination and pressures to develop new techniques to allow subfertile and infertile men to have children with their own sperm. For example, a technique increasingly used in cases of reduced fertility with low sperm counts or low sperm mobility is intracytoplasmic sperm injection (ICSI). This technique involves injecting a single sperm directly into the egg as part of IVF treatment.
- 4.9** A rather distant possibility here would be the use of stem cell technology to reprogramme cells from the body to produce eggs and sperm. Another would be to create a baby using the unfertilised eggs of two women. Both techniques have been used in animals, though more research would be needed before the practicalities of such developments for humans become clearer. However, it is certainly not just the practical issues that need to be considered. If techniques such as these were ever used in humans, a whole range of ethical issues would need to be addressed.

* The term "cloning" has been applied to embryo splitting but unlike the process used to produce Dolly, the cloned sheep, there is no manipulation or substitution of the nuclear genome so the safety issues are different.



4.10 Given some of these broader trends, we think it likely that there could be increased pressures on services for people with infertility problems. Recently, the National Institute of Clinical Excellence published a Guideline for Fertility which covers England and Wales. This recommends that couples should be offered up to three stimulated cycles of IVF where the woman is aged 23-39 at the time of treatment for an identified cause of their fertility problems or where infertility has been evident for the previous three years. With the implementation of the Guideline, we could expect to see an expansion of fertility services being offered through the NHS, though the expectation is that this will take time. We also note that currently there is much less use of IVF treatments in the UK than in several other European countries.

Treatment of Genetic Disorders

- 4.11** Future developments are likely to include treatments for mitochondrial disease. Mitochondrial disease usually occurs because of mistakes or mutations in the genes inside small “energy factories” called mitochondria, which are inside all cells. This can cause serious metabolic disease. Mitochondria are passed from generation to generation in eggs, not sperm, so it is via the female line. Women who carry a mitochondrially transmitted* disease can avoid having affected children by using egg donation and IVF. But there are other possible future approaches. One involves injection of normal mitochondria from a donor into the egg, another is to use cloning of a newly fertilised egg nucleus into a healthy donor egg from which the nucleus has been removed. These techniques are illegal in the UK.
- 4.12** There are a range of potential treatments in utero which have been considered for genetic disorders. In 1997, the Government’s Gene Therapy Advisory Committee (GTAC), whose key role is to consider and advise on the acceptability of proposals for gene therapy research on human subjects, considered some of the key issues raised by both in utero stem cell transplantation and gene therapy interventions [see GTAC, 1997, *Report on the Potential Use of Gene Therapy in Utero*]. One key issue identified is that in utero gene therapy is currently considered unacceptable due to significant safety and ethical concerns. There may, however, be an increasing role for fetal surgery in correcting some serious genetic disorders. Such techniques would generally only be used where there is clear medical benefit prenatally as opposed to once the baby is born.
- 4.13** There will be important issues about the long-term effects of such interventions and such approaches are only likely to be effective for a very small proportion of the 6000 or so single gene disorders. The fact that most of these are extremely rare severely limits research possibilities. This, together with the complexity of the effects of gene mutations and our limited understanding of these, means that the sad reality is that most genetic disease will remain untreatable for the foreseeable future, despite some successes.
- 4.14** Advances in preimplantation genetic diagnosis (PGD) techniques, such as improved IVF success rates combined with embryo selection, rather than terminating pregnancy, may be increasingly possible for people wanting to have children without a particular inherited genetic disorder. However, PGD and embryo selection have a number of important limitations:
- it is limited by the number of embryos that are available to choose from;
 - hyper-stimulation of the ovaries carries its own hazards;

* Mitochondrial enzymes may be influenced by nuclear genes, so not all mitochondrial disorders imply mitochondrial inheritance.



- the success rates for achieving a pregnancy are limited;
- and technologies have a relatively high cost.

4.15 Potentially, if a dramatic improvement in pregnancy rates using IVF were achieved, there may be an increase in demand for PGD and embryo selection. But without this, it seems likely that PND and possible termination will remain the commoner approach for couples who are at risk of transmitting a serious disease to their children.

Selection, Enhancement and “Designer Babies”

4.16 Public concerns about developments in genetic science become most apparent when it comes to the topic of “designer babies”. While a number of these concerns will be discussed in the following chapter, this section will examine in what ways it might be possible to “design” babies now and in the near future in order to debunk some of the myths in this area.

Designer Babies

Embryo selection refers to the selection of embryos using PGD. PGD is a technique currently used to select embryos without a specific serious genetic disorder or chromosomal abnormality. It has also been used to select for female embryos to avoid an X-linked genetic disorder which is a disorder that will only affect male children. In one case, the HFEA has licensed a clinic to use selection to obtain an embryo that is both free of the relevant genetic disorder (β Thalassaemia) and would be a tissue (HLA) match for an existing child.

Embryo enhancement refers to using techniques to enhance the genetic make-up of a child, and is prohibited. In theory, embryo enhancement might involve either the selection of an embryo with genetic characteristics indicative of desirable traits such as beauty or intelligence, or a process of genetic modification to enhance such traits. Such a process is not yet technically possible because of our lack of knowledge about the complex interaction between many genes and the environment which work together to produce individual characteristics such as beauty or intelligence. Clearly any genetic selection or modification of an embryo would at least in principle be transmitted to the next generation by germline changes. Germline genetic modification is currently banned.

4.17 Embryo selection is currently being used to avoid the birth of children with a serious genetic disorder. As discussed in Chapter 3, what is being selected for is a viable embryo that does not have a particular genetic or chromosomal abnormality. It is also being used to select an embryo that does not have a particular serious genetic disorder and one that could be a potential tissue match for a sibling with the disorder. This is what has been called a “saviour sibling”. HFEA are in the process of reviewing licensing rules for this.

4.18 There is sometimes a belief that in the near future, we will be able to enhance our children genetically and be able to select for certain characteristics such as beauty, intelligence or sporting ability. As HGC has heard, at the moment scientists know almost nothing about which genes might be involved in making up these characteristics and the role of the environment. Even if scientists did know this, an additional problem with selecting for such attributes would be that an impossibly large numbers of embryos would be required to find one with the desired genetic make-up. An even more remote possibility is enhancement through the genetic modification of



embryos or fetuses. As with embryos, we have almost no idea of which genes to target or the means of changing them. Any such approach remains science fiction for the foreseeable future.

4.19 An important point that has been brought to our attention on a number of occasions is that at the moment, technologies like PGD are not being used for what might be considered trivial or social reasons. People who use PGD may see this as their only option for reasons including:

- having already had multiple miscarriages because of chromosomal abnormalities in the fetus;
- having had direct experience of serious genetic disorders either via their families or their own children;
- having had terminations due to abnormalities identified via screening and diagnosis;
- people who may find the termination of pregnancy unacceptable under any circumstances and know they are at high risk of passing on a genetic disorder;
- or may have lost children due to a genetic disorder.

The difficulties associated with getting pregnant using IVF and the limited availability of embryos also need to be taken into account as possible limitations on the future use of these techniques.

Framework and organisation of services

4.20 The developments outlined above have implications for the framework and organisation of not just genetic services, but general medical services as well. As our understanding of the links between genes and environment and disease develops, there may be improvements in the diagnosis and treatment for a range of diseases with some genetic component. It may also give us a better idea of the processes involved in disease and this is necessary for developing improved methods of disease prevention, management and treatment.

4.21 It is difficult to predict the pace at which these changes will occur but in the future a much wider range of healthcare professionals will need an understanding of the role of genetics in disease. There will also be a need for a greater capacity to do genetic testing, but at the same time we can expect that new technologies will allow much larger numbers of tests to be processed more quickly.

“I am not sure if I am ready for “Brave New World” just yet!” Consultative Panel Member

5. The issues: opportunities or threats?

5.1 In late 2000, HGC commissioned a review of public attitudes to human genetic information. One result to emerge was that while most people agreed or strongly agreed that parents should be able to use genetic information to decide if a child with certain disabling conditions is born, a significant minority disagreed. While the survey did not go into the detail of why people felt this way, it does indicate that there is disagreement about prenatal testing and what happens once results are received. For HGC, a key issue is to consider how an individual's right to make reproductive choices is influenced by the social and medical context in which choices are made. We are also concerned with the wider consequences of these choices for both the individual and society.

Should individuals be able to make unlimited choices?

5.2 Questions of choice about children are at the heart of our examination of the implications of developments in human genetics to the types of decisions people might make in having children. This is because we are interested in the kinds of reproductive choices presented to people, the context in which they are presented and what these technical changes might mean for the kind of society we live in. Using the example of IVF, it could be argued that techniques that were once considered socially undesirable are now widely accepted and, therefore, we are not in a position to know the consequences of current choices on future social norms. Others would argue that society should be more concerned about the potential impact of technical changes to reproduction, and an increasing medicalisation of pregnancy, as these could alter our understanding of a whole range of key issues from the concept of family to what it is to be human and how we value human life.

5.3 The United Nations Universal Declaration of Human Rights established the right of men and women to found a family. This, in addition to a right to privacy and to respect for family life, is enshrined in a range of European and international charters and covenants (see the United Nations *Universal Declaration of Human Rights*, Article 16, 1948, the *European Convention on Human Rights*, Article 8 and Article 12, 1950 and the *International Covenant of Civil and Political Rights*, Article 23, 1976). While these and similar statements in themselves are relatively unproblematic, issues arise over their interpretation at both national and individual levels.

5.4 In the UK, it is generally accepted that people should be able to choose their partner for relationships and with whom they have children*. It is also legally and widely socially accepted that women can have abortions subject to provisions of the *Abortion Act* (1967) and its amendment in 1990**, which allowed the possibility of the termination at any stage of a pregnancy when the fetus was affected by a “serious medical handicap”. The definition of “serious” remains contentious as it was never clearly defined within the Act. This discussion document sits alongside a pending judicial review on criteria for abortion for fetal abnormality. We will be taking the outcomes of this review into account when drafting our final report.

* Exceptions to this include prisoners, individuals sterilised subject to court rulings, and minors.

** On a case law basis, “medical” abortions were carried out before this legislation.



- 5.5** While people in the UK have the right to reproduce, there are restrictions on their access to assisted reproductive technologies. For example, there may be limits because of current regulatory restrictions, limited financial resources, and subject to availability of certain technologies and techniques, or clinicians may impose restrictions based on the age of the woman. There is also a range of practices that would mitigate against technologies being used in particular ways. For example, many people would argue that using PGD to select against embryos that are unaffected carriers of genetic disorders would be unethical, as being a carrier would not have a direct impact on that child's health.
- 5.6** Some argue that such limits are unreasonable, suggesting that people should have the right to use developments in genetic science in the way they see fit for their circumstances. Advocates of this position argue that limiting access and the use of developments in genetics in the field of reproduction through regulatory, legislative or other means, is unacceptable unless they can be demonstrated to harm others, or society as a whole. This line of thinking might be extended to say reproductive freedoms need be seen as a fundamental human right. That is, it should be accepted that people should be able to make their own choices even if they are not the choices others would make, or even agree with.

“...all should have the choice, parents have the ultimate choice to not get pregnant, and if science can give people more facts why not.” Consultative Panel Member

“I believe that prenatal screening is an essential part of the preparation for the birth of a new baby, as, whether or not the parents take any action, such as abortion, as a result of the tests, the screening helps them to prepare themselves mentally and perhaps practically for the arrival.” Consultative Panel Member.

“... where there is a genetic problem, and the parents of a possible fetus consider it a great enough problem to wish not to pass it on to their child, help should be on offer. We all want the best for our children and I am sure we would never purposefully choose a lesser option. Even for conditions that usually kick in in mid life such as Huntington's, as none of us would choose half a life over a full life. I certainly want a full life for all of my children.”
Consultative Panel Member

- 5.7** In direct opposition to the above position are arguments for the prohibition of certain genetic technologies based on the belief that all human life is sacred and warrants protection from the moment of conception. However, although various religious objections have been voiced, it must not be thought that, as is often supposed, this debate is one between religion and science. There are many secular and ethical objections to the use of the new techniques. For example, the use of PGD has been seen by some as problematic because it could lead to “designer babies” and the production of children for instrumental reasons. Contention also remains with what might be considered “serious”. There may be competing definitions of this terms. For example, some people might want to have their embryos tested for BRCA1/2 mutations. Inheritance of these mutations means that the person born may have a higher risk of developing breast or ovarian cancer in adulthood. Others, however, would not consider this particular genetic mutation as warranting the use of PGD.



“I am not in favour of the abortion of a fetus with a “disorder”. However, even if we assume that parents have the right to choose, I am concerned that prenatal screening and diagnosis will result in abortion. The prevailing view in the UK is that living with impairment is a tragedy and no right-minded person would want a child with a genetic condition.”

Consultative Panel Member

“A shotgun approach to screening can lead to all sorts of legal, moral and social injustices for the affected individual – assuming that individual has been allowed to come to birth...”

Consultative Panel Member

- 5.8** Others are concerned about the balance between individual choice and the broader social values which shape these choices. Some people argue that reproductive choices are made against a background of inadequate social support for, and widespread discrimination against, disabled people and people with genetic disorders. Furthermore, it is claimed that promoting prenatal selection against potentially affected children stigmatises both those living with that condition and their parents.
- 5.9** Another related issue is the choices people are given. For example, one of our Consultative Panel members told us about her unplanned pregnancy. Both she and her partner each had a genetic disorder that the child might or might not have inherited. The couple made the decision not to have prenatal screening for the condition. They knew though that they would need additional help from Social Services to raise the child. According to this member, Social Services “...saw me as ‘a problem’ from the outset ...”. She felt she had little choice and terminated the pregnancy. This raises the question about the extent to which people have the freedom to choose, particularly for those with knowledge of existing genetic disorders in the family.
- 5.10** While perhaps not entirely accepting any of the arguments above, many people in the UK seem to accept that it is appropriate to have some degree of regulatory and legislative limit on access to reproductive and genetic technologies. There might be a range of reasons for this including:
- the need to regulate standards and practices in IVF and genetics clinics;
 - the need to put safeguards in place for recipients of donor eggs, sperm and embryos such as testing donors HIV status;
 - to ensure that treatments take into account the welfare of any children produced through these techniques (for example, one reason for banning cloning or germline interventions is because little is currently known about the safety of using such technologies);
 - to consider the equity of access to services;
 - and to ensure that embryo research is conducted for acceptable reasons.

Many of these reasons are based on the wish to protect the interests and welfare of individuals.

- 5.11** The arguments briefly outlined above are wide ranging and cover a variety of secular and religious views. In the UK, there are various limits placed on the extent to which people can exercise their choices. These limits are a combination of regulation, legislation and established good practice. There continue to be a range of arguments and concerns about whether these limits are appropriate or necessary. Our concern is not with legislative and regulatory limits – there are a number of reviews currently being undertaken to address these – but with the broader issues: should limits be set, how might lines be drawn and how will decisions influence society?



What's wrong with “designer babies”?

5.12 “Designer babies” is a term often used in the media when talking about current or potential future uses of genetic technologies. It has been used as a catchall phrase for a number of different things that involve changing or creating the kinds of children we might have. Examples have included:

- the use of sex selection techniques to avoid the birth of a child with an X-linked condition;
- the use of PGD to select for embryos free from having a particular inherited serious genetic disorder;
- the selection of donors of sperm, egg or embryos for particular characteristics such as deafness;
- and the enhancement or selection of features such as intelligence, sporting prowess and attractiveness.

“If we allow access to reproduction related genetic technologies, where and when are we going to draw the line? Between fair skins and dark skins? Between boys and girls?... What is the difference in saying no to these differences and saying yes to the differences between impairments?” Consultative Panel Member

5.12 It is important to stress that the last of these, selecting for or enhancing genetic features such as intelligence is neither technically possible nor legal. This was discussed in the previous chapter and will not be discussed here. There are, though, very real concerns about what is currently possible and what may be possible in the near future.

5.13 There are clear regulatory and legislative limits on the extent to which we can “design” babies in the UK. For example, while there are techniques such as PGD that can be used to ensure the birth of a girl rather than a boy so as to avoid a serious X-linked genetic disorder, people are not allowed to select for “family balancing” reasons such as when a woman or couple have a boy and would like a girl. Changing the genetic structure of embryos and reproductive cloning is illegal.

5.14 There are, however, ongoing concerns about how current techniques are being used. For example, when PGD has been licensed to screen for embryos free of a serious genetic condition, at the same time in a few cases tissue typing has been permitted to ensure a genetic match for an ill sibling. In these cases, when the “saviour sibling” is born, its umbilical cord blood which contains stem cells has been used to treat the older child. Applications for licenses to conduct such procedures are considered by HFEA on a case by case basis. However, there has been an application to use PGD for tissue typing which does not also have the aim of avoiding a serious genetic disorder and so would not be of direct benefit to the prospective donor child. This was not licensed and so not carried out in the UK. For many, this raises issues about social values. Should people be able to select an embryo on the grounds that the child born may be the source of life saving therapies for a sibling? Should people be able to screen for carrier status, either prenatally or at an embryonic stage using PGD? Should they be able to use PGD to avoid the implantation of an embryo with a late onset genetic disorder such as Huntington’s disease or an embryo with a predisposition to develop conditions such as diabetes, high blood pressure or breast cancer?

5.15 One type of “designer baby” in the US which received significant media attention a couple of years ago was born to a lesbian couple who were both deaf. The mothers sought out and selected a sperm donor with an inheritable form of deafness so that any child would have a high chance of



being deaf. They went on to have two children, both of whom are deaf. The rationale for the couple's choice of donor is that deafness should not be considered a disability but a cultural identity, and they wanted their children to share their identity. This issue sparked controversy on a number of grounds. While some argued against such developments on the basis that “designing babies” for any reason is wrong, many others argued that such developments are wrong because it is the parents, and indeed health professionals', duty to maximise a child's advantages in life. For many, there are clear differences between parents with inheritable genetic conditions such as deafness having children, and choosing donors on the basis of increasing the chance to have a deaf child or potentially selecting embryos for having particular genetic disorders.

- 5.16** The term “designer babies” has also been used to describe the selection of donors for specific attributes such as intelligence. For example, in the US in 1982, Robert K. Graham set up a company called Repository for Germinal Choice that he hoped to stock with the sperm of Nobel Prize Winners. This was in order to provide services to women who hoped to have an intelligent child. Most fertility clinics in the US offer a wide range of donors who can be selected for such characteristics as education level, appearance and ethnic origin. In the UK, the HFEA closely regulates the use of donor eggs, sperm and embryos and clinics are expected to strive as far as possible to match the ethnic background and physical characteristics of donors and ethnic background to those of the affected or infertile partner (or both partners in the case of embryo donation) so that the opportunity for recipients to choose donors with particular characteristics is limited. However, regulation only applies to situations in which treatment is being provided by a clinic. It does not extend to arrangements between private individuals and companies offering to mediate, for example, between would-be recipients and donors whose fresh gametes are supplied anonymously for self-insemination. These examples are perhaps peripheral to our review, but raises questions about the extent to which people should be able to select donor characteristics, as well as issues about the commercialisation of reproductive technologies there are also concerns about the emergence of a “backstreet” industry and reproduction tourism where people may be encouraged to treat themselves without proper screening or the legal protection that can only be provided by licensed clinics.
- 5.17** Some would argue that people should generally be given a choice to use artificial reproductive technologies as they feel appropriate as long as it does not harm that child. Others argue that using these technologies undervalues the life of the child born. For example, if a couple is using PGD for a condition like cystic fibrosis and there may be a choice between embryos with cystic fibrosis, healthy carriers, and embryos which neither have the genetic disorder nor are carriers, then parents should have the option to implant the embryo without the condition or carrier status. On the other hand, some argue that in some situations, carriers of cystic fibrosis may have a genetic advantage because of resistance to certain diseases. It could certainly be suggested that we should not exclude such genetic variation from human populations. It does mean that there is a risk for the next generation that if the person who is a carrier has a child with another carrier, they will run the risk of having a child with cystic fibrosis.
- 5.18** Some feel that in “designing” babies, society is no longer valuing children for who they are, but rather for what they are, making them into consumer items, with embryos or fetuses being discarded when they are not suitable. Others welcome such developments but with some clear reservations. While many would appreciate the value of such technologies in allowing people to have healthy children, they would also have concerns about where such technologies are leading us, or the purposes that such technologies may be put such as selecting for specific impairments such as deafness.



“I believe that all cases need to be dealt with on an individual basis. However I do not believe in people being able to choose the sex, personality traits or physical attributes of their child. This is totally different to having a test for serious medical conditions.” Consultative Panel Member

What obligations do we have to future generations?

5.19 A question that has been raised in a number of HGC meetings discussing this topic is: what, if any, obligations do we have to our children and future generations? While many people will make significant lifestyle changes both before and during pregnancy to ensure the health of their child, different sorts of questions arise with developments in genetics.

“I believe that parents should be allowed to give their children the best possible chance to live a healthy and successful life. On the other hand I would not have been born...” Consultative Panel Member

5.20 Some argue that our obligations only extend so far as ensuring children are provided with an “open future”. This means that while parents have a responsibility to help their children develop the range of skills and abilities to become mature adults, they should not attempt to enhance aspects of their child’s life to such an extent that it closes off other possibilities. While this does not foreclose on the right of people to utilise genetic technologies for the health of their children, it does raise issues about the extent to which such technologies could be used to enhance or shape that child’s life in specific ways. An argument often used against allowing parents to select the sex of their child for non-medical reasons is that parents will be doing so with a range of assumptions about what that child’s life will be. Similar arguments have been put forward about human cloning which is neither legal in the UK nor technically feasible. Some concern has been raised however that if it were possible to clone a child, then it would be done with the intention of shaping that child’s life in predetermined ways.

5.21 Others argue that people should, as far as possible, avoid passing on genetic disorders or even carrier status to their children as they have an obligation to that child and to society to have the best possible child. The obligation to the child is that it has the best possible life in terms of health; and obligation to society is that the child should not burden future generations in terms of health and social care.

5.22 In conclusion, as we have seen, there are a diverse range of views about the social and ethical implications of developments in genetic science. Recognising that we are simplifying complex arguments, we might sum up the debate by saying that for some, developments in human genetics raise questions about who should inhabit the world. For others, the key issue is about the rights of people to make autonomous decisions for themselves and their families. Our concern is with how a person’s right to make reproductive choices are influenced by the social and medical context in which they are made, and the wider consequences of these choices both for people and society.



“ALL parents wish the best for the health and wellbeing of their successive generations but that does not mean that children born with congenital disorders/genetic conditions are valued any the less by their parents/families or even society in general. Quality of life issues are very subjective ... individuals and society can gain a great deal from diversity. Diversity not only includes races, cultures and ethnicity but also the diversity and richness that contact with individuals living with certain disorders can bring, providing that the services and support structures are in place to provide fully for the needs of such individuals and their carers...”

Consultative Panel Member



6. Choosing the future

This section summarises information and views the Human Genetics Commission has considered to this point. We welcome comments on any aspect of this document, and especially the areas where specific questions are raised. In particular, we are seeking additional viewpoints and evidence to inform our thinking.

How to respond

The deadline for responses is **Friday 15 October 2004**. A response can be submitted by letter, fax or email or on our website at www.hgc.gov.uk/choosingthefuture. If by letter or fax, it would be appreciated if you could send us an email version in addition if at all possible.

Responses can be sent to:

Baroness Kennedy
Chair, Human Genetics Commission
Freepost LON15696
Maidenhead
SL6 2BZ

Email: HGC@doh.gsi.gov.uk
Fax: 020 7972 1717

Additional Copies

Additional copies of this document may be made without seeking permission. Further copies, including in the alternative formats described below, can be obtained from the above address.

An electronic version can be found at www.hgc.gov.uk/choosingthefuture

Other versions of the document in Braille, large print, other languages, for people with learning difficulties or on audio cassette are available on request.

Confidentiality

Your response may be made public by the Human Genetics Commission. If you do not want all or part of your response or name made public, please state this clearly in the response.

We would be grateful if you could provide us with your contact details to aid in our analysis of responses. Please also let us know if you would be interested in future consultations and/or you would like a summary of the outcomes from this consultation.



Questions HGC is seeking your views on

A. Population screening in pregnancy

(More information is given in sections 3 and 5 of the main document)

- Offering screening to pregnant women to identify possible fetal abnormalities has become a significant part of prenatal care in the UK.
- Women are given choices about whether or not to take up the offer of prenatal screening and/or diagnosis. They are then free to use that information to prepare for the birth of a child with a genetic disorder or to terminate the pregnancy.
- There are a range of views about why prenatal screening has been offered. Some say it is about promoting health and extending reproductive choice. Others have argued that it is about reducing the number of children with genetic disorders in a socially acceptable manner.

Questions

1. Various forms of prenatal screening have now become a routine part of medical practice in the UK today. An increasing number of genetic conditions may be included in screening programmes in the future. How do you feel about these developments?
2. We are interested in the extent to which you have confidence in the current provision of prenatal screening and diagnostic services. For example:
 - Is adequate counselling provided?
 - Is sufficient and appropriate information offered at all stages of the process?
 - Is the information provided fully accessible to all groups in the community?
 - Is counselling non-directive?
3. It has been claimed that prenatal screening and diagnosis presupposes that most women and couples will opt for termination if a genetic disorder is identified, some feel this reflects a wider negative assessment in society of the value of the lives of disabled people and/or people with genetic disorders. Do you agree or disagree with this view? And why?



B. Genetic services

(More information is given in sections 3 and 5 of the main document)

- People who have a history of genetic disorders in the family are faced with difficult decisions when having children. Some will speak to their GPs about this, others will go to specialist genetic clinics.
- Some women and couples may have prenatal genetic diagnosis to find out whether or not their baby has a particular genetic disorder. Some will decide to terminate the pregnancy if the disorder is present.
- Other women and couples may seek to use preimplantation genetic diagnosis (PGD) where IVF embryos are tested for a specific genetic disorder. Those without the disorder will be implanted in the uterus.

4. There are a number of genetic disorders for which embryos and fetuses can be tested. Should the use of PGD to test and select an embryo be governed by the same principles as the use of prenatal genetic testing (PND)? And to what extent should people have the right to request the testing of an embryo or fetus for particular genetic conditions?

- Some people with a known family history of a genetic disorder may use sperm, egg or donor embryos to avoid the risk of having the disorder passed on. In the UK, it is the role of the Human Fertilisation and Embryology Authority to regulate their use. In many countries overseas, however, it may be possible to select some of the characteristics of donors such as height, educational attainment, ethnicity or an impairment such as deafness.

5. Whilst treatment using donor sperm, eggs and embryos is regulated in the UK, there exist companies outside the regulatory framework who can match potential donors with recipients. To what extent should people be able to choose the characteristics of a donor in the hope that they will conceive a child who inherits these characteristics?

C. Developments in genetics

(More information is given in sections 4 and 5 of the main document)

- There are a number of potential technical, social and structural developments that may have an impact on genetics and reproductive choice. For example, developments in screening technologies means that we might be able to screen for genetic disorders earlier in pregnancy; improved treatments (though very few cures as yet) for a limited number of single gene disorders; developments with techniques such as preimplantation genetic diagnosis and so on.
- Such developments might influence decisions about having a child with a specific genetic disorder.

6. What, if any, are the potential future developments in this field that give you hope and/or concern? How might your hopes or concerns be addressed most effectively?
7. Genetics is a rapidly changing field, particularly in relation to reproduction. Are there any issues you would like to raise about the framework and organisation of services in light of potential developments over the next decade?
8. Are there any additional issues or concerns you would like to bring to the attention of the Human Genetics Commission that have not been addressed in this document?



Annex 1.

Terms of Reference

1. To collate information, take evidence and consider past, current and future developments in genetic services related to reproduction within the current legal framework and in terms of the technology and public attitudes towards its use.
2. To examine, in particular, advances as they relate to prenatal genetic screening services, prenatal genetic diagnosis and preimplantation genetic diagnosis.
3. To work with existing bodies responsible for regulating and/or advising Government on genetics and reproduction including the National Screening Committee and the Human Fertilisation and Embryology Authority.
4. To work with HGC groups as appropriate to develop strategies for public consultation and discussion, to develop the working group's knowledge about genetic services and horizon scan in the area of genetics and reproduction.
5. To contribute to and/or respond, where appropriate, to emergent national debates about genetics services and their implications for reproductive decision making.
6. To prepare and publish a consultation document and to consider other methods for obtaining the views of stakeholders and others.
7. To identify from consultation and deliberation, sound ethical principles appropriate to genetic advances and services related to reproduction.
8. To publish a report identifying our conclusions and recommendations pertaining to the ethical principles on genetic advances and services related to reproduction and to communicate these to Health & Science Ministers.

Working Group on Genetics and Reproductive Decision Making Members

Baroness Helena Kennedy (Co-chair, HGC Member)
Barrister and broadcaster

Professor Martin Richards (Co-chair, HGC Member)
Professor of Family Research, Centre for Family Research, University of Cambridge

Dr Bill Albert (HGC Member)
Chair of the Norfolk Coalition of Disabled People

Professor Brenda Almond (HGC Member)
Emeritus Professor of Moral and Social Philosophy, Hull University



Professor Elizabeth Anionwu (HGC Member)
Professor of Nursing, Head of the Mary Seacole Centre for Nursing Practice,
Thames Valley University

Professor John Burn (HGC Member)
Professor of Clinical Genetics, University of Newcastle upon Tyne and Director, Northern
Genetics Service

Dr Heather Draper (Co-opted HGC Member)
Senior Lecturer, Centre for Biomedical Ethics, University of Birmingham

Dr Frances Flinter (Co-opted HGC Member)
Clinical Director and Consultant Clinical Geneticist, Genetics Centre, Guy's and St Thomas'
Hospital Trust.

Dr Hilary Harris (HGC Member)
General Practitioner, Manchester

Professor John Harris (HGC Member)
Sir David Alliance Professor of Bioethics, University of Manchester

Mr Alastair Kent (HGC Member)
Director, Genetic Interest Group (GIG)

Ms Suzi Leather (ex officio Member)
Chair of Human Fertilisation and Embryology Authority

Dr Christine Patch (HGC Member)
Senior Research Fellow and Genetic Nurse Counsellor, University of Southampton

Mr Peter Sayers (HGC Member)
Chair, Telecommunications Advisory Panel

Professor Veronica van Heyningen (HGC Member)
Head of Cell Genetics Section, MRC Human Genetics Unit, Edinburgh

Dr Martin Whittle (Co-opted – UK NSC Observer)
Professor of Fetal Medicine, Birmingham Women's Hospital



Annex 2: Members of the Human Genetics Commission

Chair

Baroness Helena Kennedy QC
Barrister and broadcaster

Vice-Chair

Sir John Sulston
Former Director of the Sanger Center, part of the Wellcome Trust Genome Campus, Cambridge

Members

Dr Bill Albert
Chair of the Norfolk Coalition of Disabled People

Professor Brenda Almond
Emeritus Professor of Moral and Social Philosophy, Hull University

Professor Elizabeth Anionwu
Professor of Nursing, Head of the Mary Seacole Centre for Nursing Practice, Thames Valley University

Dr Stephen Bain
Reader in Diabetic Medicine at Birmingham University and Consultant Physician at Birmingham Heartlands Hospital NHS Trust

Dr Celia Brazell
Director, Genetics Science and Technology, GlaxoSmithKline Research and Development

Professor John Burn
Professor of Clinical Genetics, University of Newcastle upon Tyne and Director, Northern Genetics Service

Dr Hilary Harris
General Practitioner, Manchester

Professor John Harris
Sir David Alliance Professor of Bioethics, University of Manchester

Mr Alastair Kent
Director, Genetic Interest Group (GIG)



Ms Suzi Leather (ex-officio)
Chair of Human Fertilisation and Embryology Authority

Ms Hilary Newiss
Solicitor

Dr Christine Patch
Senior Research Fellow and Genetic Nurse Counsellor, University of Southampton

Professor Martin Richards
Professor of Family Research, Centre for Family Research, University of Cambridge

Mr Peter Sayers
Chair, Telecommunications Advisory Panel

Professor Veronica van Heyningen
Head of Cell Genetics Section, MRC Human Genetics Unit, Edinburgh

Mr Geoff Watts
Journalist and presenter of BBC Radio 4's Leading Edge

Mr Philip Webb
Member of the Board of Trustees of Genetic Interest Group

Representatives of the Chief Medical Officers

Each of the four UK Chief Medical Officers will be able to participate in HGC or nominate a representative with observer status.

Professor Peter Harper (Wales)
Professor and consultant in medical genetics, University of Wales College of Medicine

Professor Patrick Morrison (Northern Ireland)
Consultant clinical geneticist, Belfast City Hospital

Dr Stephen Singleton (England)
Medical Director, Northumberland, Tyne and Wear Strategic Health Authority

Dr Rosalind Skinner (Scotland)
Principal Medical Officer of Public Health Medical Division, Scottish Executive Health Department

© Crown copyright 2004

Produced by the Department of Health

40293 1p 3.5k Jul 04 (STE)