



Human  
Genetics  
Commission

**Increasing options, informing  
choice: A report on preconception  
genetic testing and screening.**

by the Human Genetics Commission. April 2011



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# Executive Summary and Recommendations

## Executive Summary

The Human Genetics Commission (HGC) was asked by the UK National Screening Committee (UK NSC) to provide advice on the social, ethical and legal implications relevant to whole-population preconception genetic screening. In order to respond to this request, a collaborative expert working party was set up, including members of both the HGC and the UK NSC. While conscious that the request for advice by the UK NSC specifically related to national screening programmes, the working party held the view that it should address preconception genetic testing more broadly than in the context of NHS-led preconception genetic screening programmes, to ensure that all the relevant general issues were considered. This report has been designed to supplement the HGC report *Making Babies: reproductive decisions and genetic technologies* (January 2006), which made recommendations about the use of genetic testing to inform the choices people make about having children, but did not cover preconception genetic testing and screening in any depth.

In this report we have developed a number of principles that relate to preconception genetic testing and screening. If followed, they would ensure that safe and equitable preconception genetic screening services are available to all those who may benefit from them, increasing the reproductive options available to these individuals. We have framed the issues that need to be considered prior to the implementation of specific preconception screening programmes, but have not gone further to make any direct recommendations about how individual screening programmes should be established – a task which is the responsibility of the UK NSC.

Our thoughts and recommendations have been guided by two principles in particular: namely, (i) the principle that respect for reproductive autonomy implies that a range of reproductive options should be available (all other things being equal). This in turn implies that preconception genetic testing should be available as one such option and that it should be equally available to all those who may benefit from it; and (ii) the principle that, where there are reproductive options available, individuals should be supported in making informed choices.

**Having considered the issues associated with preconception genetic testing, in our view, there are no specific ethical, legal or social principles that would make preconception genetic testing within the framework of a population screening programme unacceptable.** Indeed, there are good reasons why earlier testing is favoured over later testing, as it facilitates wider patient choice and improved access to information supporting reproductive decision-making.

We conclude that, in general, the ethical issues relating to preconception genetic testing within the framework of a population screening programme are similar to the ethical issues associated with other types of population screening, although appropriate limits for preconception genetic screening need to be considered.

Having come to this view, we have made recommendations about processes that we believe should support preconception genetic testing within the framework of population screening programmes.



## Recommendations

### Increased reproductive autonomy and choice

To maximise choice in terms of the various reproductive options available and to increase reproductive autonomy, it is desirable that, wherever possible, individuals are offered genetic screening prior to conception rather than after a pregnancy is established. **We therefore recommend that, if antenatal carrier screening is offered for a genetic condition then, where technically feasible, preconception screening should also be offered for that same condition; however, the availability of a preconception screening programme does not remove the need to offer antenatal screening to those who have not had screening prior to conception.**

### Education about genetic risks

Education is the key to providing individuals with the ability to engage with the concepts and utility of preconception testing and screening. **We recommend that children and young people should learn about antenatal and preconception screening in the final years of compulsory schooling. In addition, high-quality information about preconception tests (and, in the future, relevant screening programmes) should be made available from a range of other sources such as primary care, family planning centres, well man/woman clinics, IVF clinics, pharmacies and community organisations in order to reinforce knowledge, and also to make it accessible to people who have missed out on education or not been educated in the UK.**

### Counselling

Those who are considering the offer of screening should be given an opportunity to have their questions answered and to discuss the possible helpful or unhelpful consequences of screening with a knowledgeable and insightful professional, who could be, but need not necessarily be, a genetic counsellor. **We recommend that appropriate information about preconception genetic testing should be provided by a competent person to people considering a test. Couples who discover that they are at an increased risk of conceiving a pregnancy with a genetic condition should be referred to a health professional who has knowledge of the condition and its management, and who is also able to discuss all the reproductive options that are available to them. In the UK this service is offered by clinical geneticists and genetic counsellors based in regional genetics centres, and sometimes also by specialist counsellors working elsewhere, for example haemoglobinopathy counsellors.**

### Timing of the offer of preconception testing and screening

Voluntary screening of older school children, following mandatory on-site education, has been shown to be successful.<sup>1</sup> A genetic screening programme in Australia for Tay-Sachs disease and cystic fibrosis led to an increase in the number of students with a positive attitude towards genetic testing and an increased uptake of testing, with no stigma experienced by genetic carriers identified during the programme. **We recommend that, if screening for older school children and young people is introduced, it will be important to ensure that individuals are not pressured or induced to take tests, they are provided with proper information (in an age-appropriate form), and there is a suitable interval between receiving the information and taking the test to allow time for reflection and discussion with parents, counsellors or others if they wish.** Young people are not normally offered carrier testing below the age of 15–16 years, by which stage most will be competent to give consent for testing themselves and will not need parental consent.

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1 Barlow-Stewart *et al* (2003).



## Sharing information relevant to reproductive decision-making

A moral obligation may fall both on people who are tested and on clinicians to share personal genetic information that may be relevant to family members; however, we recognise that, for some, sharing carrier status information about a genetic condition would raise grave social difficulties.

**We recommend that professionals who counsel those considering genetic tests make explicit to the individual concerned the importance of sharing test results with family members, in order to facilitate the offer of cascade testing for relatives who may also be at increased risk of carrying certain genetic conditions.**

## Duties and responsibilities

The UK NSC will determine the framework and principles for preconception screening for each specified condition considered as part of its remit. It is responsible for ensuring that programmes are developed equitably and based on sound evidence. **We recommend that others setting up screening programmes outside the NHS, for example in the voluntary sector, should take cognisance of these principles.**

## Support for those living with conditions

Preconception genetic screening programmes will increase the choices available to an individual or couples; one such choice is whether or not to use available medical technologies to avoid having a child affected by a serious genetic condition. Individuals and couples can only have a true choice, however, if there are adequate services available to support them were they to have an affected child. **We recommend that the development of preconception genetic screening programmes should not detract from the provision of treatment and support for affected individuals. Prior to testing, individuals should be made aware of the services and support available to them if they were to have an affected child.**

## Key measure of preconception genetic screening programmes

In line with the principle of reproductive autonomy, individuals and couples should only ever be offered preconception genetic testing (within a framework of non-directive counselling) and never be made to feel obliged, expected or forced to take a genetic test. Therefore, it follows that unlike other screening programmes, whose success can be measured by the uptake of a test, **the key measure of a preconception genetic screening programme should be the proportion of people of reproductive age who are made aware of the availability of testing, or offered testing (as appropriate), and not the proportion who have a test.**

## Management and disclosure of information

**We recommend that carrier status information obtained incidentally through tests or investigations carried out for other purposes (for example, after prenatal diagnosis or through newborn screening) should be provided to GPs and stored in a secure and accessible format.**

**We recommend that the UK NSC reviews and formalises policy to allow the routine communication of carrier status results identified through newborn screening to GPs.**



# An outline of the preconception genetic testing and screening report

## Background to the work

1. The Human Genetics Commission (HGC) is the UK Government's independent advisory body on developments in human genetics and their implications for individuals and society. In 2006, the HGC published a report entitled *Making Babies: reproductive decisions and genetic technologies* which contained recommendations about the use of genetic testing to inform the choices people make about having children. These include choices about how to prepare for the birth and upbringing of a child affected by an inherited genetic condition, the possible use of assisted reproductive technologies such as preimplantation genetic diagnosis (PGD) and about the option of prenatal diagnosis, which can generate information facilitating a choice as to whether to continue or terminate a pregnancy. The report also made recommendations about the conditions under which testing should be made available. For example, it was recommended that couples undergoing testing should be offered supportive genetic counselling in order to uphold important principles such as individuals' reproductive freedom, social responsibility and the protection of the interests of future children.
2. The UK National Screening Committee (UK NSC) was established in 1996 and advises ministers and the Devolved Administrations 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 UK NSC assesses proposed new screening programmes against a set of internationally recognised criteria covering each condition, the test, the treatment options and the effectiveness and acceptability of the screening programme. It also examines the cost benefit and/or effectiveness of screening programmes, and it ensures there is a plan for managing and monitoring screening programmes. Currently, the UK NSC does not support any national preconception genetic screening programmes.
3. Improvements in genetic testing technology have facilitated the development of tests for significantly more markers, at greater speed and more cheaply than ever before. These technologies are becoming available both within the national healthcare system and, without significant limitation, to those who may wish to buy them privately.<sup>2</sup> While screening fetuses for inherited conditions during pregnancy is widely available for some conditions, preconception genetic testing – that is, genetic testing performed before a child is conceived in order to identify whether an individual tested carries a genetic condition which could be passed on to, and might affect, any offspring – has until now been practised on a much more restricted scale. Some communities, in which there is known to be a high risk of a genetic condition occurring,

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<sup>2</sup> The HGC has recently published a *Common Framework of Principles for direct-to-consumer genetic tests* in order to promote good practice in this sector and protect the interests of consumers (Human Genetics Commission 2010).



have organised very effective screening programmes.<sup>3</sup> In other cases, testing has been offered systematically for certain conditions, in certain areas, for a certain period.<sup>4</sup>

4. Preconception genetic testing, which includes testing for carrier status for a range of genetic conditions, can be beneficial if offered either as a screening test to those who belong to an identified population, or on an individual basis because of a personal or family history of a genetic condition. It is clear, however, that not all those who might be eligible access preconception genetic testing for a variety of reasons, including a reflective decision not to do so or being unaware that such testing is both possible and available. As technologies become more powerful and less expensive, the use of preconception genetic testing is likely to increase and it is important that policy anticipates this and ensures appropriate use of such technology.
5. Recently, the UK NSC has been asked to consider the introduction of preconception genetic screening programmes.<sup>5</sup> As genetic diagnostic technologies improve in technical efficiency, clinical utility and cost-effectiveness, alongside the anticipated discovery of an increasing number of genetic markers for hereditary conditions, testing strategies that aim to identify genetic variations, or their surrogates, are likely to have an increased role in informing healthcare decisions. In line with the Coalition Government's commitment<sup>6</sup> to putting patients at the heart of the NHS, and empowering them to make decisions about their healthcare through better information and more choice among providers, the importance of knowledge gained from genetic testing is likely also to grow.
6. As genetic carrier screening potentially raises some distinctive issues that do not pertain to non-genetic screening programmes, the UK NSC approached the HGC in late 2009 to develop some general advice on the social and ethical considerations raised by preconception genetic screening, with a view to applying and, where appropriate, supplementing the relevant principles identified in the earlier *Making Babies* report. These include ensuring reproductive autonomy and protection of the rights of children when information may have been obtained collaterally at a time when reproductive options were not being considered, for example following prenatal diagnosis or neonatal screening. Different groups within the population are at increased risk of a variety of different genetic conditions and people may seek information and testing at different points in their lives. The report highlights the challenge of enabling people to have access to up-to-date information and testing relevant to their situation in a rapidly changing technological environment.

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3 See, for example, the Tay-Sachs screening service organised by Jewish Care for the Ashkenazi Jewish community: although the prevalence of the recessive Tay-Sachs trait in the Ashkenazi Jewish community is 1 in 25 and there is a high level of marriage within the community (62% of married Jewish people reported that they had a Jewish spouse according to the 2001 census), there are fewer births of affected babies than would be expected in that community. However, reliable evidence remains elusive, as the laboratories that make the diagnoses do not collect ethnicity and other relevant data that would help to separate those born to Jewish parents from those born to non-Jewish parents. Current screening for Tay-Sachs occurs both preconceptually and antenatally on an ad hoc basis in the Jewish population. It may be that a natural tendency towards informal cascade screening (whereby individuals are more likely to present for screening if they are already aware of a carrier in their family) has helped to keep the occurrence of this condition low. See Burton *et al* (2009).

4 See, for example, Bekker H *et al* (1994).

5 "The Advisory Group recommends that the Tay-Sachs disease programme should be formally constituted as with any other screening programme" (Burton *et al* 2009).

6 See *Equity and Excellence: Liberating the NHS* (Department of Health 2010).



## Purpose of the work

7. The present report responds to the need for a clearer policy context within which to consider preconception genetic testing and screening by addressing specifically the conditions that should be met for preconception genetic screening to be socially and ethically acceptable, beneficial to individuals or groups and appropriate for the NHS to provide.
8. To do this we have brought together a working party that comprises members of the HGC and UK NSC, together with experts from other bodies and organisations, in order to provide a broad range of expertise and perspectives.<sup>7</sup>

## Scope of the work

9. Our working definition of preconception genetic testing is as follows:  
**Preconception genetic testing identifies carriers of genetic mutations responsible for a range of genetic conditions, knowledge of which can inform – and may qualitatively affect – the reproductive choices of individuals tested or those of their close biological relatives.**
10. According to our definition, it is not important whether the analyte (the substance tested) is DNA itself or some surrogate, such as the presence of certain proteins or metabolites, which identifies the presence or absence of a specific genetic variant.
11. From the outset the working party was persuaded that it would be necessary to consider preconception genetic testing more broadly than just in the carefully controlled context of organised and systematic NHS-led preconception screening programmes (of the sort that UK NSC exists to consider) to avoid inconsistencies and ensure that relevant general issues were considered.
12. In our report we will therefore distinguish between different types of genetic testing by the context in which it is provided (for example, commissioned privately by the consumer or a medical intermediary, or as part of an organised screening programme):
  - ‘**genetic testing**’ refers to testing of individuals to determine the presence or absence of a genetic biomarker, regardless of the context in which the test is commissioned or performed, for example, testing in the context of a positive family history (also called ‘cascade testing’). The test may or may not be taken as part of a genetic screening programme.
  - ‘**genetic screening**’ is the offer of testing to whole population groups, often when the population is known to be at increased risk of a genetic condition; however, individual members of the population are at no higher risk than other members of the group. It aims to identify which individuals are carriers (for example school-aged  $\beta$ -thalassaemia testing in Cyprus, antenatal sickle cell and thalassaemia screening in England and Tay-Sachs population screening in Israel) and therefore which individuals are more likely to be helped than harmed by either further tests or treatment or information about reproductive options. If offered, preconception genetic screening should be delivered as a whole programme with follow-up counselling and support and not just the provision of testing.<sup>8</sup>

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7 Information about the membership of the working party and the terms of reference is provided at Appendix 1 to this report.

8 The UK NSC defines a screening programme as “the whole system of activities needed to deliver high-quality screening. This includes identifying and informing those to be offered screening through to the treatment and follow-up of those found to have an abnormality, as well as support for those who develop disease despite screening”.



13. Using our definition of preconception genetic testing, we will consider genetic testing that is relevant to reproductive decisions, regardless of the context in which it is performed; at the same time, we also recognise that information relevant to reproductive decision-making does not become available only as the result of a deliberate test for that purpose. Tests carried out on other family members and collateral information from tests for other purposes may also be relevant; for example, a full blood count may suggest  $\beta$ -thalassaemia. As well as testing for the purpose of informing reproductive decisions, we will, therefore, also consider the management of information about individuals held in health records that could help to inform their reproductive decisions.
14. The scope of our report therefore includes:
  - testing for carrier status of specific single gene conditions
  - testing offered to those known to be at significant risk because of their ethnic or family origin following a screening questionnaire or screening whole populations where there is a significant risk of particular harmful genetic variants
  - testing for a range of genetic conditions that may occur with low and variable frequency within the general population
  - cascade testing offered to relatives of individuals who are affected by, or carry, a single gene condition
  - management of genetic information relevant to reproductive decisions obtained from genetic tests carried out for another purpose (for example, collateral information from antenatal screening tests carried out on other biological relatives and genetic profiling or genome sequencing).
15. However, there is a specific exception: instead of considering this in the context of our report **we recommend that the UK NSC reviews and formalises policy to allow the routine communication of carrier status results identified through newborn screening to GPs.**
16. The work should consider how incidental carrier status information that is obtained in this way can be systematically stored, how the information should be recorded on the primary care record and whether this information should be relayed and discussed with the individual and at what age. We note that some screening programmes will identify a large proportion of carriers, such as screening programmes for sickle cell anaemia, while others will identify only a subset, for example cystic fibrosis. (Neonatal cystic fibrosis screening programmes only test for the more common DNA mutations in the subgroup of neonates whose positive immunoreactive trypsinogen (IRT) screening test suggests a clinical diagnosis of cystic fibrosis.)
17. There are certain genetic tests that, while their outcomes are relevant to reproductive decisions (and many other decisions about the future besides), are principally concerned with the diagnosis and care of affected individuals. Therefore, our report does not consider:
  - presymptomatic testing for autosomal dominant conditions where there is a positive family history
  - testing for predispositions to complex conditions (either singly or multiply).



## Legal context

18. There is a variety of legal instruments, both binding and non-binding, ranging from universal declarations, through international conventions, European treaties and national statutes to domestic common law, that assert the fundamental rights of men and women to found a family and to protect family life from intrusion by the state or by other people. These include:
- United Nations, Universal Declaration of Human Rights (1948), Article 16 (right to marry and found a family)
  - Council of Europe, Convention for the Protection of Human Rights and Fundamental Freedoms (1950), Article 8 (right to respect for private and family life) and Article 12 (right to marry and found a family)
  - United Nations, International Covenant on Civil and Political Rights (1966), Article 23 (right to marry and found a family)
  - European Parliament, Charter of Fundamental Rights of the European Union (2000), especially Article 1 (human dignity); Article 3 (right to integrity of the person), including Article 3(2)(i) (respect for free and informed consent) and Article 3(2)(ii) (prohibition of eugenic practices); Article 7 (respect for private and family life); Article 9 (right to marry and found a family); Articles 20–26 (equality); and Article 35 (right of access to preventive health care)
  - Council of Europe, Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine (Oviedo Convention, 1996) and Additional Protocol concerning Genetic Testing for Health Purposes (2008).
19. The interpretation of these statements can raise issues at international, national and individual levels. Many of these instruments, while advocating individual autonomy and the interests of the individual above the sole interests of science or society, also advocate a principle of solidarity grounded in the recognition of the shared human genome. This may be summarised by saying that the human genome is a common feature that importantly (although not uniquely) both distinguishes us from each other and relates us to each other, as individuals, families, populations and as a species. A number of these high-level declarations and conventions also include the protection of future generations, the prevention of eugenic practices and a recognition of the importance of human genetic diversity, for example:
- UNESCO, Universal Declaration on Bioethics and Human Rights (2005), Article 16 (protecting future generations)
  - UNESCO, Declaration on the Responsibilities of the Present Generations Towards Future Generations (1997), Article 6 (preservation of the human genome in its diversity).
20. In the UK, it is generally accepted that people should be able to choose their intimate partners and those with whom they have children, excepting certain restrictions relating to incest and marriage between people within defined degrees of relatedness. It is also accepted widely within society, and provided for in law, that women may have abortions subject to the provisions of the Abortion Act 1967 (as amended by the Human Fertilisation and Embryology Act 1990). This amending legislation allowed the possibility, among other things, of the termination of pregnancy at any stage when the fetus was affected by a “serious mental or physical handicap”. The term “serious” is not addressed in the abortion legislation and is a matter of clinical



judgement. The law permitting PGD (the Human Fertilisation and Embryology Act 1990 as amended by the Human Fertilisation and Embryology Act 2008) makes the rationale for using PGD to exclude affected embryos consistent with that for termination of pregnancy, through a judgement about the seriousness of “a physical or mental disability, illness or other medical condition” of which the pregnancy is “at significant risk”.

21. Medical examination and treatment are subject to the law requiring patients’ informed consent. This comprises the protection of their autonomy or self-determination (*Re T* (1992) 9 BMLR 46) and the requirement that they are provided with appropriate information to allow them to exercise this (*Pearce v United Bristol Healthcare NHS Trust* [1999] 48 BMLR 118 and *Chester v Afshar* [2004] UKHL 41). Consent for the testing of biological samples from living persons (for example, to examine them for DNA variations) is further subject to statutory consent requirements imposed by the Human Tissue Act 2004.
22. The Coalition Government’s health White Paper *Equity and Excellence: Liberating the NHS* (July 2010) promises opportunities for the more active involvement of individuals in choosing their healthcare options (“no decision about me without me”). The Government emphasises the importance of patient choice and equity of access to healthcare.



## Current guidance and relevant publications

23. We have taken as our starting point for this report the HGC's own 2006 report, *Making Babies: reproductive decisions and genetic technologies*. We found comparatively little extant guidance on preconception genetic testing. A list of sources is included at Appendix 2; we found the following, in particular, to be useful to us during our deliberations:
- Association of Biomedical Andrologists, Association of Clinical Embryologists, British Andrology Society, British Fertility Society and Royal College of Obstetricians and Gynaecologists (2008) UK guidelines for the medical and laboratory screening of sperm, egg and embryo donors. *Human Fertility* 11(4): 201-10.
  - Burton H, Levene S, Alberg C and Stewart A (2009) *Tay-Sachs Disease carrier screening in the Ashkenazi Jewish population: A needs assessment and review of current services*, available at: [www.phgfoundation.org/reports/4987/](http://www.phgfoundation.org/reports/4987/)
  - Christianson AL, Howson CP and Modell B (2006) *March of Dimes Global Report on Birth Defects: the hidden toll of dying and disabled children*. White Plains, New York: March of Dimes Birth Defects Foundation.
  - Dormandy E, Bryan S, Gulliford MC *et al* (2010) Antenatal screening for haemoglobinopathies in primary care: a cohort study and cluster randomised trial to inform a simulation model. The Screening for Haemoglobinopathies in First Trimester (SHIFT) trial. *Health Technol Assess* 14(20).
  - Health Council of the Netherlands (2007) *Preconception care: a good beginning*, available at: [www.gezondheidsraad.nl/en/publications/preconception-care-good-beginning](http://www.gezondheidsraad.nl/en/publications/preconception-care-good-beginning)
  - Smith A, Shakespeare J and Dixon A (2010) *The role of GPs in maternity care: what does the future hold?* Research paper. London: The King's Fund.
  - World Health Organization (2010) *Birth defects: Report by the Secretariat*. Sixty-third World Health Assembly (provisional agenda item 11.7).
  - World Health Organization (2006) *Sickle-cell anaemia: Report by the Secretariat*. Fifty-ninth World Health Assembly (provisional agenda item 11.4).
24. The 2010 World Health Assembly report on birth defects<sup>9</sup> identifies carrier screening as one of the important measures for the prevention of birth defects (paragraph 9 notes the “timely identification of a family risk of inherited disease, and carrier screening with genetic counselling, enable couples to limit family size where there is a known risk”). As a potential action it recommends “initiation and monitoring population-screening programmes such as screening of newborn infants, premarital/pre-pregnancy screening, and screening during pregnancy”.
25. A resolution was adopted to help redress the limited focus to date on preventing and managing birth defects, especially in low- and middle-income countries. The resolution calls on Member States to prevent birth defects wherever possible, to implement screening programmes, and to provide ongoing support and care to children with birth defects and their families.

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9 World Health Organization (2010).



## Reproductive options

26. The prevalence of clinically relevant genetic conditions in the general population is approximately 2–3%.<sup>10</sup> These range in severity from the relatively mild to the severely disabling and life-limiting. While no effective treatment has been found for Tay-Sachs disease, for example, so that affected individuals often do not survive beyond their second birthday, other conditions such as cystic fibrosis and  $\beta$ -thalassaemia have seen significant improvements in life expectancy over the last decade, with many affected individuals now surviving into their 30s and 40s. People with a family history of genetic conditions, or from a population group in which the condition(s) are prevalent, have a significantly higher risk of being carriers of those genetic conditions and of having an affected child.
27. **Reproductive options for individuals or couples who know before conception that they carry a genetic mutation/s.** There are a number of reproductive options available to individuals or couples who know before conception that they carry a genetic mutation/s that causes a specific clinical condition. These may include:
- to remain childless through choice
  - to adopt a child rather than having a child who is genetically related to them
  - to accept the chance of having an affected child, perhaps using the information to prepare for the possibility of this eventuality; for example, by accessing social or clinical networks supporting affected families or making plans to optimise immediate treatment for the child after birth
  - to conceive naturally (through sexual intercourse) and to undergo prenatal diagnosis to establish whether the fetus is affected; if the test result is positive (affected) this information can then be used to decide whether to terminate or continue the pregnancy
  - to conceive a child using donated sperm or eggs
  - to undergo *in vitro* fertilisation (IVF) to allow the PGD of embryos fertilised in the laboratory to determine which may be affected before transferring unaffected embryos to the woman to begin a pregnancy.
28. **Reproductive options for individuals or couples who only know about their carrier status after pregnancy starts.** The options available to couples who discover their carrier status only after they have embarked upon a pregnancy are substantially fewer than those for couples who are aware of their risk in advance of a pregnancy. The additional options available to those who know their carrier status in advance of a pregnancy are also available in future pregnancies to those who know they are carriers because they already have an affected child.
29. In 2009, 2,085 abortions were carried out under statutory ground E of the Abortion Act (“there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped”). Of these, 144 were coded as being related to a family history of a heritable condition.

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10 “The birth frequency of single gene and chromosomal disorders is around 2%, while 2–3% of couples have a high risk of a recurring condition in their children. In adult life, genetic factors are prominent in around one-third of serious chronic disorders, while specific genes are increasingly being identified in many common disorders with a familial component” (Royal College of Physicians of London 1998).



30. While individuals or couples usually choose to have genetic carrier testing in order to identify whether or not any current/future pregnancy will be at increased risk of inheriting a genetic condition, there may be broader consequences in addition to the reproductive choices they empower. For example, there may be consequences for personal relationships within the family and for the sense of identity and family belonging of those unaffected individuals who are shown to carry or not to carry the condition in question.



## Ethical issues

31. In our view, the key, ethical principles are: (i) reproductive autonomy, (ii) the protection of the interests of children who will be born, and (iii) genetic solidarity and the moral duty on individuals to share relevant results with family members. These principles were elaborated in the HGC's earlier *Making Babies* report.
32. The principle of **reproductive autonomy**, which is a corollary of respect for persons, entails a requirement to respect a person's right to control their own body and their choice to try to have (or not to have) children. It stops short, however, of an entitlement to assistance from others to try to have children, and in no way amounts to entitlement to have children (or children of a certain sort).
33. The **protection of the interests of children who will be born** potentially constrains the scope of the principle of reproductive autonomy, at least insofar as giving effect to the autonomous decision necessitates the involvement of a professional. An extreme example is the Human Fertilisation and Embryology Authority's regulation that does not permit the preferential selection of an embryo affected by a genetic condition following PGD, even if this is what the potential parents request.
34. The principle of **genetic solidarity** is articulated in a number of the high-level declarations that we cited earlier in this report. It amounts to an observation of the fact that we all share the same basic human genome and provides the conditions for certain basic sympathies and for common interests. In *Inside Information* (2002), the HGC described the principle as follows: "We all share the same basic human genome, although there are individual variations which distinguish us from other people. Most of our genetic characteristics will be present in others. This sharing of our genetic constitution not only gives rise to opportunities to help others but it also highlights our common interest in the fruits of medically-based genetic research."<sup>11</sup> In the context of preconception genetic testing, the principle also has a narrower and more specific reading: it enjoins us to help those with whom we share close biological links, for example by offering to share information about our own diagnoses or known carrier status of particular genetic conditions with family members who may also be at risk, in order to make them aware of the possible benefits of taking a test themselves. It is in this sense that it is frequently encountered in clinical genetics, where in some cases it can be taken to limit the principle of confidentiality (for example, where a serious harm may be avoided by disclosing personal genetic information without consent).
35. Preconception genetic testing is currently available in an inconsistent and inequitable fashion across the UK, with access to tests being dependent on individuals knowing whom to ask, which may, in turn, depend on which school they attend or their own knowledge about the increased carrier risks for certain genetic conditions, such as Tay-Sachs disease or sickle cell disease, that are associated with specific ethnic groups.
36. In certain circumstances, however, preconception genetic screening entails the potential for harm. The next section identifies possible harms and relevant predisposing conditions.

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11 Human Genetics Commission (2002)



## Negative eugenics

37. Negative eugenics is the organised, deliberate discouragement or prevention of natural reproduction between certain groups of people (whereas positive eugenics is the deliberate encouragement of breeding between certain classes of people) in order to produce offspring who have or lack specific heritable characteristics. An example would be a programme of preventing uncontrolled reproduction between people with a known or supposed risk of passing on a negatively valued, heritable condition in a way that was designed to eradicate that condition from society over the course of a few generations. The disturbing history of eugenic policies, and its relation to contemporary public health policies, was described in Chapter 2 of *Making Babies*.
38. While the choices of individuals to use available medical technologies to avoid having a child affected by a serious genetic condition do not entail the judgement that it is undesirable to have affected individuals with that condition in society, the cumulative effect of all individual choices could, depending on the nature of those choices, result in fewer affected individuals being born. In other words, universalising the maxim of individual choice may lead towards a conclusion that it would be morally unacceptable to reach by imposed or co-ordinated design (although it would be necessary to examine carefully what the maxim of such a judgement may be).
39. Some have pointed to the possible impact of such choices in increasing the stigmatisation of people living with conditions that may be genetic, or may appear to many in society as likely to be genetic, and the practical impact it may have on the services available to them. The individual choice to avoid having a child affected by a given genetic condition, or the collective decision to permit the use of technologies to help people avoid having affected children, may be interpreted as devaluing affected children by supporting the view that their birth should be prevented. Parents may also come to be expected to bear more personal responsibility for having a child with the condition if they are seen, in effect, as having chosen not to avoid it. With respect to service provision, if genetic conditions are avoidable and, for the most part, avoided, then the decreasing number of people with those conditions may see the availability of services diminish as expertise is lost and the marginal cost of providing those services increases. Alternatively, facilities for treatment could improve if there were less demand. All these possible consequences may exert a pressure on reproductive decision-making by altering the circumstances under which these decisions will be made. Those who see genetic conditions as something more complex than a simple disadvantage, particularly those who view disability, its construction and effects, as social rather than medical issues, may find this position undermined by a spreading acceptance of genetic testing to avoid disease. **We recommend that the development of preconception genetic screening programmes should not detract from the provision of treatment and support for affected individuals. Prior to testing, individuals should be made aware of the services and support available to them if they were to have an affected child.**
40. Arguing from a different set of convictions, those who have a principled opposition to the use of assisted conception technologies and to the artificial termination of pregnancy, may feel their beliefs are compromised by genetic testing to avoid disease. The tension between respect for autonomy and the duty to intervene for the putative good of others underlies almost all problems in medical ethics and is always only provisionally resolved at any given point in our history. There is an interval that remains to be crossed between the defence of individual autonomy and the accepted principle that people may be offered testing (within a framework of non-directive counselling) so that they have the opportunity to avoid the birth of children with genetic conditions. Awareness of the social context within which such decisions are made and the influence of societal factors (such as availability of healthcare, constructions of disability and norms of health) may help with understanding and bridging different positions.



41. Unlike many other screening programmes, whose success can be measured by the uptake of a test (for example, cervical screening), **the key measure of a preconception genetic screening programme should be the proportion of people of reproductive age who are made aware of the availability of testing, or offered testing (as appropriate), and not the proportion who have a test.**
42. One concern that may be raised about private purchasing of preconception genetic tests is that such tests may be supplied by a company that has an ideological commitment to, for example, the eradication of avoidable genetic conditions. It is a question of ethics, therefore, to what extent the knowing patronage of these services entails a culpable complicity with the provider's ideological commitments. At the very least, it may support the company's capacity to promote that ethos by providing revenue.

### The context of reproductive choice: support for those living with genetic conditions

43. For individuals, couples and families where there is an identified risk of a known genetic condition, there are more reproductive opportunities available than there are for those who may be at a similar biological risk but who are unaware of the fact. Knowledge creates opportunities and brings with it consequences that can create pressures not experienced by those unaware of the potential challenges that a genetic risk may pose for the health and well-being of a future pregnancy. Lack of awareness does not eliminate this risk; it simply removes opportunities to intervene to avoid it manifesting itself.
44. The options available to individuals and couples with a known genetic risk are described in paragraph 27. When choosing between these options, individuals and couples need to feel able to make decisions that they judge to be right for them in the circumstances in which they find themselves at the time. They should not feel pressured into making choices that have been unnecessarily constrained by the expectations of others as to what the "correct" decision might be (subject, of course, to the constraints imposed by legislation and current clinical and scientific possibilities). It is the individual (plus their child and partner, if any, as well as their wider family) who will live with the consequences of any decision, and the weighting given to different elements in reaching this decision will vary between individuals and couples and over time. The options preferred or chosen may be influenced by knowledge of the condition, awareness of the impact of affected children on themselves and other members of the family, access to services and support, personal religious and moral codes or community norms. They may differ between pregnancies; an individual or couple with one affected child may choose to proceed with a second affected pregnancy, while another individual or couple in an apparently similar situation may choose to terminate and avoid the birth of a second affected child. Perception will also vary between individuals as to the severity of the effect on a child with a given condition, and of their capacity to raise it.
45. Whatever the internal algorithms that individuals and couples use to establish their genetic status and to exercise their reproductive choices within the law, healthcare professionals and others in positions of authority should beware of exerting undue influence and pushing individuals in directions which they find uncomfortable.
46. Of course, context-based decisions are often constrained by, for example, limits on access to restricted services. Supporting informed choices by individuals and couples in no way precludes campaigning for better services and support for those affected. Nevertheless, it remains the case that, even where support is readily available and provided in a timely, user-friendly manner that minimises the disabling aspects associated with a specific condition, some individuals and couples will still seek to avoid the birth of an affected child. The right of individuals and couples to make a choice should be respected.



47. We are conscious that other societies hold values that may differ from the ethical values prevailing in the UK. In some countries there are highly organised screening programmes and, in some cases, prohibitions on marriage between carriers of certain conditions, or requirements for couples to show that they have undergone carrier testing before marriage ceremonies can take place. In Cyprus, for example, where one in seven adults carries a mutation that causes  $\beta$ -thalassaemia,<sup>12</sup> individuals who wish to marry must present documentation of thalassaemia screening to obtain a marriage licence (although carriers continue to marry, with most carrier couples opting for prenatal diagnosis and the termination of affected pregnancies). This screening programme was introduced with the support of the World Health Organization, and was accompanied by a softening of the Orthodox Church's attitude to abortion, in the expectation that, without intervention, treatment for affected people would have come to consume the entire health budget within 40 years.<sup>13</sup>

### Consent/capacity/autonomy – young people and testing

48. Possession of information that is relevant to one's reproductive choices is a benefit as it provides individuals and couples with more options; however, autonomy also entails the freedom of each individual to determine when and in what manner to seek and receive that information according to their own values. This must include the freedom to decide whether or not to be given information, although care must be taken to ensure that this does not lead to an unhelpful and paralysing paradox. Individuals must be offered sufficient information for them to appreciate that there is more they could find out if they wished.
49. Therefore, those being offered information about preconception screening must be able to decline further and more detailed information once they have appreciated that screening could help them avoid the (usually small) risk of having a child affected by a serious genetic condition.
50. In addition, as in any other case when samples for testing are obtained, informed consent should of course be sought and obtained before testing is carried out. The aim of pre-test counselling should be to ensure that people have (i) sufficient information, presented in a non-judgemental way, to enable them to make an informed decision about whether or not to have a test, including an opportunity to consider the possible social, psychological and cultural impact of testing and, if they choose to have testing, (ii) adequate knowledge to understand the possible test results and the likely implications for their future reproductive choices.
51. It may be difficult to identify the best time to offer information about genetic testing to young people. Ideally, they should learn about the risks of having a child with a genetic condition alongside education about sex, reproduction and the benefits of adopting a healthy lifestyle, and before they become sexually active; but young people mature at different ages and have varying capacities to understand and remember information that may be complex, confusing and not immediately relevant at the time of testing. Individual counselling may help to determine their educational needs and understanding, although in the context of a population screening programme there will inevitably be limits to the degree to which individualised counselling can be provided.
52. Sensitivity is always required when raising and discussing reproductive choices with young people because their personal views, their religious faith or the traditions of their communities may constrain or direct their thoughts, feelings and decisions. This may be especially relevant for those who come from a religious or ethnic minority community within which there are strong expectations that a particular practice will be followed that is unusual within the mainstream of family life in the UK (for example, the practice of arranged marriage).

12 Ashiotis *et al* (1973).

13 Zeuner *et al* (1999).



53. Establishing robust population-based preconception genetic screening programmes will be a major undertaking for the UK NSC; however, the current situation in which access to tests for conditions such as Tay-Sachs, for example, is patchy and inequitable is unacceptable, particularly as some couples do not discover their pregnancy is at increased risk of a genetic disease until they are already pregnant, leading to considerable stress and distress. We regret the fact that this situation is sometimes an avoidable consequence of the failure to offer genetic carrier testing prior to conception.

### Responsibility for sharing information relevant to reproduction

54. The moral obligation to share personal genetic information that may be relevant in order to avoid harm may fall both on the people tested and on clinicians (or others who are aware of a relevant test result).
55. It is recognised that it can raise grave social difficulties, within the family or with strangers, for an individual to reveal their carrier status for a genetic condition. This may lead to anger, blame, denial or rejection, especially if there are likely to be adverse consequences such as stigmatisation or an impact on the prospects for marriage of the individual or of others in the family. In addition, even without adverse social consequences, individuals may differ in the extent to which they are comfortable sharing personal information with relatives and/or with others. Education of the public is required in order to normalise the issues surrounding the inheritance of recessive conditions and the sharing of carrier status information, and to create a better understanding that we all carry genes for a few recessive conditions, some of which may be identified by genetic testing.
56. **We recommend that professionals who counsel those considering genetic tests make explicit to the individual concerned the importance of sharing test results with family members, in order to facilitate the offer of cascade testing for relatives who may also be at increased risk of carrying certain genetic conditions.** This is supported by the guidance on confidentiality and information sharing in clinical genetics from the Joint Committee on Medical Genetics of the Royal College of Physicians, the Royal College of Pathologists and the British Society for Human Genetics.<sup>14</sup> Both this and the General Medical Council's guidance on confidentiality recognise that genetic information is a class of "shared information".<sup>15</sup> Indeed, they acknowledge that in some cases, where consent to share medically relevant information for the benefit of others is not obtained or even if it is explicitly refused, there can be a duty to disclose the information, "to help protect the other person from serious harm".<sup>16</sup>

14 "...we support the good practice of encouraging individuals to agree to the appropriate (and where possible anonymised) use of their genetic information to benefit the clinical management of family members" (Joint Committee on Medical Genetics of the Royal College of Pathologists *et al* 2010).

15 "Genetic and some other information about your patient might at the same time also be information about others the patient shares genetic or other links with" (General Medical Council 2009: paragraph 67).

16 *Ibid.*, paragraph 69.



## Stigmatisation and discrimination

57. Stigmatisation and discrimination are potential harmful effects of preconception screening (alongside other suggested harms that include inducing stress, anxiety, excessive worrying and misunderstanding the test results).<sup>17</sup> This may include stigmatisation of the individual, or an ancestry-based group on which the carrier screening may be based, for example the stigmatisation that occurred after implementation of sickle cell testing and screening in the US in the early 1970s.<sup>18</sup> Stigmatisation in this case arose due to a lack of effective communication and education about the difference between the carrier state and the disease state, which resulted in, for example, insurance companies erroneously rejecting applicants who were identified as carriers.
58. Studies from around the world on carrier screening have, on the whole, shown positive support and satisfaction about carrier status; although some identified carriers experienced initial anxiety and maybe a reduction in self-esteem, later attitudes were similar in carriers and non-carriers, in terms of retention of knowledge about their status and an intention to use the results when considering child-bearing.
59. A major follow-up of both Western and non-Western participants in a study of preconception ancestry-based carrier couple screening for cystic fibrosis and haemoglobinopathies in the Netherlands showed no major adverse psychological effects in either group, with low anxiety levels, improvement in knowledge and intention to utilise test results for reproductive decision-making. Patients did not regret participating, nor did they report major feelings of discrimination or stigmatisation.<sup>19</sup> However, it has been observed that parental knowledge of a child's carrier status can make parents try to guide the child's future reproductive decision-making.<sup>20</sup>
60. The potential for stigmatisation and discrimination should not prevent the introduction of preconception genetic screening programmes; however, when setting up such programmes, consideration should be given to the potential for stigmatisation and discrimination to occur and measures should be taken to minimise the likelihood of their occurrence, for example by appropriate education.

## Equity and equality

61. Unlike many other countries, the UK has a National Health Service that provides health services free at the point of use to every member of the population according to their need. Within this system, however, practice in the commissioning of diagnostic tests for particular conditions may vary from place to place, depending on the judgement of the clinicians involved and the resources available. Although there are programmes approved by the UK NSC to screen women and fetuses during pregnancy and infants shortly after birth for a number of conditions, there is currently no national preconception genetic screening programme for any condition.

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17 Lakeman *et al* (2008).

18 Markel (1992).

19 Lakeman *et al* (2008).

20 Parsons and Atkinson (1992).



62. Equity in access to preconception genetic testing arises at the three main levels identified within the scope of this document:
- a. **Genetic testing of the individual** where there is known to be an increased risk, for example because of a positive family history. This should normally happen already as part of a planned and systematic follow-up of the extended family and will usually be organised by the regional genetics centre, although for some large volume services, such as haemoglobinopathies, there will be specific counsellors working in specialist units or centres. Although equity of access across the country may be expected as a result of agreed professional practice standards, in reality this may be limited by differences in budgets for genetic tests and different priorities determined by commissioners. In populations with high levels of consanguinity, this might entail follow-up of at-risk family members through extensive and complex pedigrees.
  - b. **Genetic testing offered to groups known to be at increased and significant risk as a result of ethnic background.** Examples in the UK include Ashkenazi Jewish groups, known to have a high carrier risk for a number of conditions including Tay-Sachs disease, familial dysautonomia, Canavan disease and cystic fibrosis. The report by Burton *et al* on carrier testing for Tay-Sachs disease<sup>21</sup> identified the important equity issue whereby carrier screening may be more easily offered in areas of higher population density for these ancestral groups. For example, about half of all Jewish children could be reached by targeting Jewish schools, and in general the majority of Tay-Sachs carrier screening could be achieved by concentrating efforts on a small number of districts in the London region and Greater Manchester. In order to achieve an equitable service for Jewish people who do not live in these areas, however, it would be necessary to introduce a screening programme that included the whole population, with a question to identify the relevant group; Tay-Sachs carrier testing would then be offered to anyone of Jewish ancestry, regardless of where they live.
  - c. **Carrier screening for a range of genetic conditions that may occur with low and variable frequency within the general population.** The issue of whether or not to offer such screening to the general population is beginning to arise in the context of the more common conditions, such as cystic fibrosis, and will need to be debated by the public, NHS and genetics experts. In principle, it would be expected that equity would be ensured by these tests being offered within an agreed NHS screening programme context; however, tests such as the Counsyl test,<sup>22</sup> which purports to offer a universal carrier test for around 100 Mendelian conditions, and is currently available privately, will provide a challenge to equity. Policy-makers in the NHS will need to take a view on whether they will recommend these tests to those who can afford to pay and/or whether (and how) they will provide services for those who need advice about the results of tests that they purchased privately.
63. The emergence of companies offering genetic tests direct-to-consumer (DTC) has been the subject of a recent set of principles by the HGC.<sup>23</sup> Individuals or couples who choose to purchase DTC tests may subsequently request help from the NHS in order to understand the results and their reproductive implications. This could cause additional work for the NHS, both in order to reassure people who are unnecessarily worried by their results, but occasionally also to counsel people about any increased reproductive risks that have been identified. Currently, the clinical value of many DTC tests is limited, and individuals who have a valid clinical reason

21 Burton *et al* (2009).

22 [www.counsyl.com](http://www.counsyl.com)

23 *A Common Framework of Principles for direct-to-consumer genetic testing services* (Human Genetics Commission 2010). These principles recommend that carrier tests should be provided only to consumers who are given suitable opportunities to receive individualised pre- and post-test counselling.



for requesting carrier tests should be able to obtain them via the NHS. In future, however, the fact that access to DTC tests is restricted to those who can afford to pay for them may contribute to inequalities of healthcare provision, particularly if DTC testing companies offer carrier testing for a wider range of conditions than the NHS.

- 64.** Having considered the issues associated with preconception genetic testing, in our view, there are no specific ethical, legal or social principles that would make preconception genetic testing within the framework of a population screening programme unacceptable. Indeed, there are reasons to favour the earlier offer of testing over a later offer of testing in order to achieve equality of access to information and wider patient choice when it comes to reproductive decision-making. In general, the ethical issues relating to preconception genetic testing within the framework of a population screening programme are similar to the ethical issues associated with other types of medical screening, although appropriate parameters for preconception genetic screening need to be considered.

### Duties and responsibilities

- 65.** The UK NSC, as part of its consideration of individual proposals for the case for preconception screening, will set the framework and principles for preconception screening that come within its remit. It will define the boundaries of responsibility for the development of services and ensure that services develop equitably on a sound evidence base. **We recommend that others setting up screening programmes outside the NHS, for example in the voluntary sector, should take cognisance of these principles.**



## Practical issues

### Education about genetic risk

66. Antenatal screening programmes focus attention on conditions where the maternal contribution is greater than the paternal (especially Down's syndrome) and pay less attention to conditions where the converse is true (for example, new dominant mutations). They might therefore inadvertently help to perpetuate the belief that having an affected child is attributable solely or primarily to the mother. Men, as well as women, could be offered more education in order to understand how the future health of a child can be affected by genes inherited from both the father and mother.
67. The ability of an individual to engage with the concepts and utility of preconception testing and screening, both on an individual basis and for society as a whole, must be developed through programmes of education and, in particular, scientific and genetic literacy, that begin at school and continue through engagement of the media.
68. Health professionals, particularly those who may interact with individuals at salient times such as in family planning centres, well man/woman clinics, pharmacies and those involved with adolescent health, must also be competent to provide the necessary education and professional support individuals need to inform decision-making, or signpost them to colleagues who can help.
69. Similarly, for those involved with particular high-risk groups, community organisations such as religious organisations and other support groups should also be encouraged and enabled to provide high-quality education and support.
70. In the development of the necessary educational programmes, both for school children and the community, the active involvement of health professionals will be vital for any planned (or unplanned) expansion in preconception screening activity, which should be based on the best available evidence of benefits and possible harm.
71. **We recommend that children and young people should learn about antenatal and preconception screening in the final years of compulsory schooling. In addition, high-quality information about preconception tests (and, in the future, relevant screening programmes) should be made available from a range of other sources such as primary care, family planning centres, well man/woman clinics, IVF clinics, pharmacies and community organisations in order to reinforce knowledge, and also to make it accessible to people who have missed out on education or not been educated in the UK.**
72. There are ethical, social and political issues connected with the way in which any group of individuals is defined, and religious, political and cultural sensitivities must be understood and taken into account. Laboratories may need knowledge of an individual's ethnic background and family history in order to analyse samples and interpret test results accurately.

### People involved in consanguineous marriage

73. Gene mutations can be "concentrated" in large families with a high degree of consanguinity or in isolated endogamous communities, giving rise to a concentration of individuals at high risk of being carriers. In spite of this, the majority of couples will still have unaffected children. The excessive



focus of health professionals' attention on consanguinity, rather than recognising that everyone is a carrier for a few recessive conditions, can lead to these families having difficulty understanding the situation and feeling stigmatised. Education of health professionals about the reasons for families practising consanguinity, and education of families about the option and availability of carrier testing, should reduce the risks of stigmatisation. Educational initiatives among relevant communities, but also aimed at the wider society, pitched in a neutral manner that does not make judgements about choice of marriage partner, will also be important in raising awareness.<sup>24</sup>

### People undergoing assisted conception

74. Individuals and couples having assisted reproductive treatments present at clinics prior to pregnancy, providing an ideal opportunity for discussion about preconception screening and the offer of testing if appropriate. People undergoing assisted conception should have a detailed family and medical history reviewed in order to identify whether any preconception genetic tests are indicated: for example, a history of recurrent unexplained miscarriages is an indication to test both partners for a chromosome rearrangement; and a history of congenital bilateral absence of the vas deferens should lead the male partner to be tested for cystic fibrosis mutations (with his partner having a carrier test too if his result is positive).
75. One further group for whom preconception genetic testing would be appropriate includes couples where both partners are known carriers of an autosomal recessive condition and who wish to use donor gametes to avoid having an affected child, and individuals who are using donor gametes and know that they are a carrier of a condition. In these cases, it would be appropriate to test the carrier status of candidate donors, providing appropriate consent is obtained.

### Management and disclosure of information

76. **We recommend that carrier status information obtained incidentally through tests or investigations carried out for other purposes (for example, after prenatal diagnosis or through newborn screening) should be provided to GPs and stored in a secure and accessible format.**
77. GPs should have an efficient “bring forward” system to ensure that young people are made aware that carrier status information may be available in their health records and could be communicated to them as they mature and become adult. Otherwise, they may never be given the opportunity to make use of this information at the most appropriate time, i.e. before they conceive.
78. Generally, clinicians should give carrier status information that they obtain about small children to their parents; carrier status information about teenagers should be given to the teenagers themselves.
79. The point at which information is provided to an individual may be: (i) if it is sought by the individual, (ii) when an appropriate opportunity arises for that information to be provided, (iii) if additional information becomes available in the future due to technological developments, or (iv) if it is made available as an integral part of pre-pregnancy care. We would expect that people are less likely to consider fully the implications of carrier status information, or even to remember it, if they have not actively and deliberately sought it.

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24 For further information, see Pembrey (2010) and [www.borninbradford.nhs.uk/](http://www.borninbradford.nhs.uk/)



## Timing of the offer of preconception testing and screening

80. Offering preconception genetic testing within the framework of a population screening programme offers many advantages over later offers, primarily because it provides people with information at a time when they have a greater range of choices available to them.
81. In principle, for an individual, testing should be offered at an earlier stage in an individual's reproductive history rather than a later one (i.e. preconception rather than preimplantation; preimplantation rather than antenatal; antenatal rather than neonatal) in order to increase the range of reproductive options available. This principle must, however, be weighed against possible harms for the individual or population, which might include the cost of providing information and offering genetic testing to a greater number of people, some of whom might not go on to have children; bearing the burden of knowledge of carrier status for a longer time or forgetting or losing the information. Thus, for example, carrier testing undertaken early must be accompanied by the systematic use of opportunities to remind people of their status at relevant stages (for example, in GP surgeries and family planning clinics).
82. When might it be best to make preconception testing available as part of a population screening programme? Carrier screening at birth or in the neonatal period has some advantages (it may be logistically simple) but it has many disadvantages, such as the chance of parents forgetting the results or failing to pass them on to the child at an appropriate time. Screening during childhood, but before reproductive age, may give children information at a stage of life when they can accept it readily, but their understanding of the test and its implications may be limited and it may then be difficult for them to access information when it becomes more personally relevant. Furthermore, the question of consent may be most difficult at this stage as it may be unclear whether the child is able to consent and therefore whether the consent of their parents may also be necessary. Guidance published by the British Society for Human Genetics on the genetic testing of children recommends that genetic testing for future reproductive risks should normally be delayed until the young person can decide for him/herself when, or whether, to be tested. The rationale for this recommendation is that testing in childhood removes the opportunity of the future young person to make their own choices about such decisions, and that this opportunity should not be denied to them without good reason.<sup>25</sup> Screening offered once young people reach reproductive age, but before they plan to start a family (for example, 15–16 years of age), may be practical if they are still at school and at that stage they may welcome the information as relevant. There will then be a trade-off between the relevance of screening to the young person (growing in relevance as they become older, nearer the time that they may wish to start a family) and the ease of population coverage (becoming progressively less complete – or more costly – as the age of the target group increases).
83. Voluntary screening of older school children, following mandatory on-site education, has been shown to be successful.<sup>26</sup> A genetic screening programme in Australia for Tay-Sachs disease and cystic fibrosis led to an increase in the number of students with a positive attitude towards genetic testing and an increased uptake of testing, with no stigma experienced by genetic carriers identified during the programme. **We recommend that, if screening for older school children and young people is introduced, it will be important to ensure that individuals are not pressured or induced to take tests, they are provided with proper information (in an age-appropriate form), and there is a suitable interval between receiving the information and taking the test to allow time for reflection and discussion with parents, counsellors or others if they wish.** Young people are

25 British Society for Human Genetics (2010: recommendation 2).

26 Barlow-Stewart *et al* (2003).



not normally offered carrier testing below the age of 15–16 years, by which stage most will be competent to give consent for testing themselves and will not need parental consent.

84. When preconception screening is introduced in an area it will require discussion with schools, GPs and other primary healthcare professionals. In addition, parents of school children and other members of the local community may wish to be involved in discussions about how screening could be introduced and operated most appropriately in the local circumstances. For the programme to operate successfully, it will be important for such local sensitivities to be treated with respect.
85. Where preconception genetic testing and antenatal testing are both technically feasible for the same condition, a decision will have to be made as to whether an antenatal or a preconception screening programme should be made available, or both. It is desirable that, wherever possible, individuals are offered genetic screening prior to conception rather than after a pregnancy is established, otherwise the options available to them will be limited, reducing the scope of their reproductive autonomy. **We therefore recommend that, if antenatal carrier screening is offered for a genetic condition then, where technically feasible, preconception screening should also be offered for that same condition; however, the availability of a preconception screening programme does not remove the need to offer antenatal screening to those who have not had screening prior to conception.**

## Counselling

86. In addition to the information provided with the invitation to consider screening, there should be an opportunity for those considering the offer of screening to have their questions answered and to discuss the possible helpful or unhelpful consequences of screening with a knowledgeable and insightful professional, who could be, but need not necessarily be, a genetic counsellor. Those who are found to be carriers of a genetic condition may be offered referral to clinical genetic services both for genetic counselling for themselves and to enable the offer of cascade carrier testing (if appropriate) within their extended family. With some conditions, such as haemoglobin conditions, individuals are more likely to be referred to dedicated health centres outside regional genetics centres for counselling and testing, if such centres exist locally. **We recommend that appropriate information about preconception genetic testing should be provided by a competent person to people considering a test. Couples who discover that they are at an increased risk of conceiving a pregnancy with a genetic condition should be referred to a health professional who has knowledge of the condition and its management, and who is also able to discuss all the reproductive options that are available to them. In the UK this service is offered by clinical geneticists and genetic counsellors based in regional genetics centres, and sometimes also by specialist counsellors working elsewhere, for example haemoglobinopathy counsellors.**



## Appendix 1: Terms of reference and membership of the working party

### Terms of reference

To define appropriate approaches to preconception genetic testing in the UK, in particular:

- the conditions for the ethical and social acceptability of preconception genetic testing
- arrangements for providing information to people about heritable genetic conditions, genetic testing and genetic test results that are relevant to their reproductive decisions
- the proper role of the state and the NHS vis-à-vis other bodies, groups or individuals in permitting, promoting and/or providing preconception genetic testing.

### Membership of the working party

#### Chair

- Dr Frances Flinter (HGC)

#### Members

- Professor Roger Brownsword (UK NSC)
- Dr Hilary Burton (PHG Foundation)
- Professor Angus Clarke (HGC)
- Mr Alastair Kent (HGC)
- Dr Carole McKeown (UK NSC)
- Dr Heather Mellows (Professional Adviser in Obstetrics and Gynaecology)
- Dr Lola Oni (HGC)
- Dr Ros Skinner (ex-HGC and UK NSC)
- Dr Allison Streetly (UK NSC/NHS Sickle Cell and Thalassaemia Screening Programme)



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