

**Human Genetics Commission Consultation,  
Choosing the future: Genetics and reproductive decision making**

*A response from the Genetic Interest Group, November 2004*

The Genetic Interest Group (GIG) is a national alliance representing individuals and families affected by or at risk of genetic disorders. We have 130 groups and many individuals in membership. We welcome the opportunity afforded by this consultation for debate and discussion around current practice and broader background issues.

This response was circulated to our members for discussion. It was also discussed and approved by GIG's Trustee Board.

**Introductory comments**

For parents at risk of having a child with a genetic disorder, testing of embryos prior to implantation or during pregnancy is an important option. Indeed, for those who believe that embryos do not have rights or interests that could interfere with selection prior to birth, the currently available reproductive and genetic technologies represent a very positive development. They have allowed many infertile couples to have children who are related to them. They have allowed women at risk of having a child with a genetic condition to achieve what others almost now take for granted—a child with every chance of a healthy and full life. They have made possible a higher chance of a successful and healthy pregnancy for older women. More recently they have made it possible to have a child who will very likely be a tissue match for a seriously ill sibling, an event that is generally hailed as a wonderful thing when it occurs by chance following conception in the traditional manner.

In an important sense, those fundamentally opposed to these practices have lost the argument. Just as it can be confidently stated that abortion is broadly accepted as a legal practice in Britain, so we can say that genetic testing during pregnancy and 'conventional' applications of Pre-implantation Genetic Diagnosis (PGD) are now widely accepted, even if some or even many people have some unease about aspects of this. Indeed, there is slow drift towards NHS funding for a part of IVF and also PGD<sup>1</sup>. GIG welcomes this, and would like to see it implemented more speedily.

However, some media and public discussion focuses on a different set of issues. What are almost always tough but sensible decisions taken by reasonable people in difficult circumstances are either discussed as problematic in themselves or else situated in a wider context that makes them so. It is not uncommon for example to see commentators

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<sup>1</sup>[http://www.dh.gov.uk/PolicyAndGuidance/HealthAndSocialCareTopics/Genetics/GeneticsGeneralInformation/GeneticsGeneralArticle/fs/en?CONTENT\\_ID=4072467&chk=hX18v9](http://www.dh.gov.uk/PolicyAndGuidance/HealthAndSocialCareTopics/Genetics/GeneticsGeneralInformation/GeneticsGeneralArticle/fs/en?CONTENT_ID=4072467&chk=hX18v9)

refer to a ‘market’ in operation which is encouraging parents to see children as ‘commodities’ whose properties they can choose or shape.

Internationally, influential writers such as Francis Fukuyama<sup>2</sup> and Jurgen Habermas<sup>3</sup> have taken up this theme. In the UK feminist academic Hilary Rose and (opportunistically?) some of the anti-abortion campaigners are obvious proponents. Roger Brownsword, Professor of Law at King’s College, who is sympathetic to these concerns, labels this new alignment the ‘dignitarian alliance’<sup>4</sup>. Unlike previous absolutist arguments based on ‘Pro-Life’ grounds, the arguments put forward under this heading do not support outright rejection of embryo selection and research. But they do suggest that we should move forward with caution or prevent particular applications.

In the final chapters of the consultation, the HGC sets out to ‘debunk some of the myths’ in this area. This is an important task we should all address in the coming period. Outlining the real choices people make, and the circumstances in which they make them, is an important part of that. People’s lives and the decisions they make are being stigmatised by a portentous public discussion that often bears little relationship to the facts.

We do not wish to deny that there are, and will always be, new applications of reproductive and genetic technologies that cause people to pause for thought and which raise new issues. The example of ‘Saviour Siblings’ as they have been called is a recent example of this. Another is sex selection, perhaps for reasons of family balancing. These issues have troubled and continue to trouble regulators and advisors to government. The HGC raises concerns, or at least suggests that some people might have concerns, about ‘commodification’ in the former example, while both might be examples of the concern outlined in the consultation 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.’

However, the difficulty is that beyond opinions and fears, what evidence is there that existing and short to middle term possibilities will have the consequences suggested or are so influenced by (presumably troubling) social and medical forces? One of the difficulties regulators and advisors have run into in recent times is that in attempting to draw lines and raise issues, they have relied upon speculative arguments and opinion surveys. People whose choices might be restricted will rightly expect better than this.

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<sup>2</sup> F. Fukuyama, *Our Posthuman Future* (Profile Books 2002).

<sup>3</sup> J. Habermas, *The Future of Human Nature* (Polity Press 2003).

<sup>4</sup> He develops his argument, in the context of a critical survey of regulation and recent court cases, in two recent articles: Regulating Human Genetics: New Dilemmas For a New Millennium, *Medical Law Review*, **12**, Spring 2004, pp. 14-39; and Reproductive Opportunities and Regulatory Challenges, *Modern Law Review*, 2004, pp. 304-321.

*Questions HGC is seeking your views on*

We have answered those questions that are or appear important to us.

*A. Population screening in pregnancy*

- 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?*

The Genetic Interest Group broadly supports genetic screening programmes. They offer a reproductive choice to people, many of whom would have been unaware of the risks they faced. Evidence suggests that they are consistent with the values women and couples hold.

Informed decision-making is critical within antenatal screening. Research continues on how better to provide antenatal screening in ways that ensure that women can make informed choices. Such programmes inevitably cause some anxiety, but most feel this is worthwhile for the reassurance gained in the case of a negative result and the choices available in the unfortunate event of a positive genetic test result on the embryo. To not develop such programmes when it was possible to do so would be to deny people what we know to be an important choice.

The decision to undergo genetic testing during pregnancy rests with the woman, usually after consultation with her partner. In the case of a known family history of a genetic condition, the initiative, the initial suggestion that testing could or should be used, will come from the woman as well in many instances. In the case of population screening it is health professionals who take the initiative. For example couples might be offered

screening with the aim of determining whether they are carriers for a condition; if they are and a pregnancy is established, antenatal testing will be offered.

The involvement of the health service and in this sense government causes some to raise questions about the purposes of antenatal screening. For Modell, Harris and colleagues, the purpose of such carrier screening is to ‘permit couples who are at risk an informed choice among available options, including prenatal diagnosis in every pregnancy.’<sup>5</sup> A reduced birth incidence of the condition in the population as a whole might be an outcome, but it is not the aim. Others present things rather differently. Writing about cystic fibrosis, Murray, Cuckle and colleagues boldly state that: ‘the aim of genetic screening for CF is to reduce the birth prevalence of the disorder. This is primarily achieved by identifying carrier couples who can have prenatal diagnosis and selective termination of pregnancy.’<sup>6</sup>

The truth of the matter is probably that both aims inform screening and testing programmes: however the issue is posed, the primary choice, in screening and also in patient-initiated testing, is the choice to avoid the birth of a child with a genetic condition. Certainly, many if not all the background papers drawn up to inform the thinking of the National Screening Committee in this area consider the issue from a classic public health perspective, including financial aspects. Knowing the proportion of women who choose to terminate in given circumstances for a particular condition allows such calculations to be made.

Is this a problem, is this perhaps even ‘eugenic’? There are a number of aspects to the argument that it is, which is put with varying degrees of vigour by different critics of antenatal screening and selective termination; and clearly, whether or not these procedures are thought to be ‘eugenic’ will depend on what that emotive term is taken to mean.

It is GIG’s view that the values of modern medical genetics are fundamentally different from those of the main strands in historical eugenics, and that approval for genetic testing prior to birth is compatible with equal treatment for people living with disabilities.

Francis Galton defined eugenics as ‘the scientific study of the biological and social factors which improve or impair the inborn qualities of human beings and of future generations.’ Such ‘study’ suggests a practice of eugenics. A modern definition might be any policy that alters the composition of the human gene pool. The philosopher Philip Kitcher develops this interpretation in his thoughtful book *The Lives to Come: the genetic revolution and human possibilities*. He then subdivides the notion into different types.

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<sup>5</sup> Modell, B. et al (2000), ‘Informed choice in genetic screening for thalassaemia during pregnancy: audit from a national confidential inquiry’, *BMJ*, **320**: 337-41.

<sup>6</sup> Murray, J. et al (1999), ‘Screening for cystic fibrosis; executive summary’, *Health Technology Assessment*, **3**(8): 3.

Interestingly, he also characterises doing nothing when we have the ability to do something as eugenic. At this point the critics of genetic testing part company with him. For them, eugenics is about humanity changing the gene pool, specifically reducing the incidence of genetic disorders, whether government policy or the aggregate of individual decisions brings this about.

Both Kitcher's and the critics' notions have their merits. But posing the issue in such a general way also tends to obscure crucial differences between historical eugenics and modern genetics. At the turn of the century there was a widespread belief that genetics influenced morals and personality traits. The preoccupation was with controlling the spread of these traits, rather than medical conditions. The dominant strand in eugenics of old was a programme for a coercive, state-led drive to alter the gene pool. It was used to justify the sterilisation, and even murder, of people classed as mentally insane and genetically inferior.

At the time, not enough was known about genetics and disease / behaviour to highlight the scientifically irrational character of many of the eugenic proposals. However, enough was known about population genetics by 1920 to invalidate, on scientific, never mind humane, grounds eugenic arguments for sterilisation. That such programmes continued regardless highlights perhaps the most important point to understand about the dominant strand of old eugenics: it was driven neither by science nor by humanitarian concern but by a strong political belief and fear—of national, racial, and social decline. As the historian Daniel Kevles puts it, using the example of Britain at the turn of the century: 'To many British, the general fiber of the nation—its overall moral character, intelligence, energy, ambition, and capacity to compete in the world—was declining.'<sup>7</sup>

After the Second World War, eugenic practices did continue for some time, up until the 1970s in the case of Sweden. Eugenicists did seek to pursue their goals through the new field of reproductive and genetic counselling. And some people do still believe that the moral worth and future of nations depend upon genetics. But in our view the predominant ethos of all work in human genetics today, and in medical genetics in particular, has little or nothing in common with historical eugenics.

The new genetics is concerned more with identifiable medical diseases than with personality traits and behaviours. It represents a biological approach to biological problems, not a reductionist approach to the whole human being. This is not to say that modern behaviour genetics and the genetics of mental health are marginal fields of inquiry; they are not. But leading researchers in the field understand the limited contribution of many different genes. Their study is primarily individual variation, not purported race or social-group differences, and very few working in the field link genetics to ideas of racial or national success and failure. Finally, these fields do not

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<sup>7</sup> Kevles, D., *In the Name of Eugenics*, University of California Press, Berkeley and Los Angeles, 1985: 73.

impinge on services offered prior to implantation or birth, and are unlikely to do so for the foreseeable future.

Some within the disability rights movement might accept the distinction we have drawn, but continue to object to antenatal testing and screening because they believe it necessarily devalues those living with the condition. Focussing on the motivations of parents in the first instance, we believe that this is wrong, and that the critics are guilty of conflating impairment and the moral status of people—something they often accuse supporters of testing and screening programmes of doing.

We have located modern genetics within the traditions of humanistic medicine. Clearly, selective termination, a possible outcome of one aspect of genetic science, is not a ‘cure’ or treatment. However, in our view parental attitudes towards fetal abnormality are framed by attitudes towards illness and not unreasonable expectations about the impact of genetic disorders on their own and their children’s lives. If they choose to terminate an affected pregnancy they are making a judgement about impairment, which is the level at which antenatal selection operates, and a guess about the life they and a child with the particular condition would have given existing levels of medical knowledge and social support. That judgement is a *relative* one; that life without the condition is better than life with it. Parents are not, as the caricature sometimes has it, saying that life with a genetic disorder is not worth living or is too terrible to contemplate. And certainly, they do not see themselves as making a moral judgement about the worth or rights of people living with the same condition.

We would argue that not only does the service not have the faults attributed to it, but that the arguments of the critics could add to the faults in the service as it exists, and may hinder its future development. In short, the danger is one of poor guidance and restrictive regulations.

It is our experience that some parents are left to make their decisions in a vacuum because health professionals fear being seen as directive if they fully discuss the available options. Genetic counselling is concerned with facilitating informed reproductive decisions. Following the eugenic experience prior to World War II, the emphasis has always been placed firmly on the ‘non-directive’ part of ‘non-directive genetic counselling’. This is as it should be. But we would introduce some caveats:

- (1) There is a danger of making anything appear directive: some say presenting testing as part of antenatal care is directive, or that clarifying likely implications of a condition is directive; or that ensuring that risks are properly understood is directive. All these procedures should rather be seen as perfectly reasonable features of patient care, and quite consistent with the goal of informed choice. We note that in a study of

this patients expressed satisfaction even when they thought aspects of their care were ‘directive’.<sup>8</sup>

(2) We cannot avoid the fact that the primary choice offered by these services is the choice to avoid having a child with a genetic condition—and that this is the choice made by most people. If this is directive then non-directive genetic counselling is impossible (a majority of delegates to the Third European Meeting on the Psychological Aspects of Genetics, held in 1992, took this point of view).

(3) A rigidly applied policy of non-directiveness may not meet patients’ needs in all circumstances. Certainly, in a screening context, if a family is unaware of the nature and implications of the condition that may affect a future child and if the goal is indeed informed choice, then it is the duty of the health professionals to present the family with the facts; to inform them about the reality of the condition.

Currently, the principal national antenatal genetic screening programme is for the haemoglobinopathies—thalassaemia and sickle cell disorder, conditions that mainly affect people who have originated from Africa, the Caribbean, the Middle East, Asia and the Mediterranean. The policy for antenatal screening is for the phased implementation of a programme that will eventually offer screening to all women as a part of early antenatal care. Debate continues on whether other programmes should be developed. In 1999, the Health Technology Assessment programme recommended universal antenatal screening for cystic fibrosis. The National Screening Committee has not agreed this, although debate continues. Similarly, debate continues on whether and how to implement screening for fragile X syndrome, a condition which causes learning difficulties largely, but not exclusively, in boys.

A key practical issue is the ability of health services to meet the information needs of the couples involved in the existing programmes. On the whole, specialist genetic staff do not oversee or take part in the existing programmes. Other staff may have less training or simply time to fully inform women and then couples at each stage of the process. Furthermore, some people approached in the course of screening programmes will require materials to be accurately translated. These issues are being addressed,<sup>9</sup> but they do raise important resource and practice questions that will need to be addressed if and when more programmes are considered.

*Questions HGC is seeking your views on*  
*B. Genetic Services*

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<sup>8</sup> Michie, S. et al (1997), ‘Nondirectiveness in Genetic Counselling: An Empirical Study’, *Am. J. Hum. Genet.*, **60**: 40-47.

<sup>9</sup> <http://www-phm.umds.ac.uk/haemscreening/Documents/ServicesReport.pdf>

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 testing (PND)? And to what extent should people have the right to request the testing of an embryo or fetus for particular genetic conditions?*

It is GIG's view that individual's and families, after consultation with their clinical team in some instances, are the best judges of what is appropriate for them. Accordingly, we believe that the ultimate decision should rest with the parents of the child to be. It is clear that they should be well informed and that is the role of the genetic services and other professionals. However, in principle GIG is opposed to rules and regulations restricting or preventing the intentions of the parents to be.

In the context of PGD it is hard to see why, if a test is reliable, there should be any restriction on the genetic conditions that could be selected against. To take but one example that has been debated recently, we can well understand that a woman might want to have a child free from a serious adult onset condition or the substantial risk of one. After all, many if not all people would view removing the certainty or significant risk of developing a disorder in later life for an individual already born as an unequivocal good. If PGD is not in itself wrong, it is hard to see why its use to achieve the same ends should not be greeted with the same positive endorsement.

In practice, taking account of all that is involved, there are many disincentives to using PGD. Most if not nearly all women would not seek out PGD services in the absence of a known family history or unless they were receiving IVF already and it was thought that chromosomal analysis could improve the success rate. However, looking to the future, if a woman was already undergoing IVF and was to make a request for a broader genetic screen, we can see no objection in principle. Indeed, it may become possible in time to effectively screen pre-implantation embryos for a range of conditions, much as presently the technology has been developed to screen such embryos to improve the chances of a successful pregnancy. As the HFEA recently acknowledged,<sup>10</sup> there is no evidence of harm to the future child as a result of biopsy, and much evidence of safety. Additionally, there is no 'slippery slope' because the only outcome is a child free of a particular condition. Of course, there is a question of whether there should be public funding in all circumstances, but that is a separate issue.

GIG takes a similar approach to Prenatal Genetic Testing. It should be up to the woman concerned whether and for what to conduct tests during pregnancy. It may be possible at some point in the future, as the consultation document suggests, to isolate fetal cells from the mother's bloodstream, thus facilitating risk-free diagnosis. But this is still quite futuristic. The reality for now and for some time to come is that invasive tests will continue to be necessary and entail a small but definite risk of miscarriage. Accordingly,

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<sup>10</sup> <http://www.hfea.gov.uk/AboutHFEA/HFEAPolicy/Preimplantationtissueotyping>

as with PGD, there is an important reality check: women support tests which offer them information on risks for serious conditions, but are unlikely to be interested in tests for mild and late onset predispositions without good reason. However, against a background in which social abortion is legal and widely available, we would insist that it should be up to the woman to decide for herself what is serious. We would also want to highlight that while guidelines are needed in a screening context, to guide decisions on what to offer and when, we shouldn't prejudge how an individual would trade off the risk of miscarriage against the risk of having a child with a particular disorder.

*Questions HGC is seeking your views on*

*C. Developments in genetics*

- 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?*

We have raised some points already. The range of applications of PGD is likely to continue to grow, if slowly. PGD is widely thought to be safe. It offers an important choice to some women and couples. And it undoubtedly has a significant impact on outcomes. As such we believe the NHS will need to keep under review its policy and guidance on the funding of this treatment. In general, GIG would like to see commissioners look more favourably on requests for funding.

Genetic screening raises a number of questions. Resource issues may once again figure in future debates, in terms of the training of qualified staff and the allocation of appropriate time. Currently, programmes target specific groups and / or are focused on a specific condition. It is possible that in the future it may make sense to offer a more generic form of genetic testing as a part of a screening programme. For example, a single procedure to identify carriers of many recessive conditions that are individually very rare. In principle GIG believes that such an approach would be a reasonable one. However, in this case as in other possible developments of screening programmes, there will likely be quite a degree of individual variation in the assessments people make of the risks and benefits of different kinds of testing, necessitating a degree of flexibility in any future programmes.