# GENETIC TESTING, SCREENING AND 'EUGENICS'

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Advances in genetic diagnosis—specifically, the ability to test for a growing number of genetic disorders—combined with new reproductive technologies, have made possible an expansion of both ante-natal genetic testing and pre-implantation genetic diagnosis (PGD). Whereas women in families affected by a genetic disorder previously had to either rely on fate or forego having children, they can now choose to have their fetus or fertilised egg tested for the disorder. As a consequence, the reproductive confidence of those affected by genetic disease has increased.

These procedures fall far short of the therapies and cures that GIG looks forward to. Nevertheless, in our view they do represent an important advance. We also view favourably the enhanced choice experienced by women as a result of the development of screening services offered to those without a family history of a disorder.

However others—from the established churches and traditional conservatives to some feminists, academics and disability rights activists—see a negative side to testing and screening. They believe that women are being coerced into testing, and that pre-implantation and pre-natal testing encourages discrimination against disabled people. A parallel between the new genetics and the eugenics of old is often drawn. For example, a report by the National Childbirth Trust published in 1997 claimed that the 'right of parents not to have ante-natal testing is being undermined by health professionals.' This message was reinforced by a study by four leading UK authorities on genetics in the same year. They claimed that professionals in the field were not following the official policy of non-directiveness in genetic counselling.[1] In the press comment on both reports an equation was made between directive counselling and eugenics.

Critics of the new genetics argue that the explicit form of pressure exerted by directive counselling is reinforced by more subtle economic and cultural processes. Indeed, whereas in the past eugenics took the form of outright, state-led, coercion, today, they argue, it results in the main from the marriage of market forces and genetics.

In his book *Brave New Worlds*, the journalist Bryan Appleyard puts it like this: 'so what we face here is, in fact, the opposite of state-sponsored eugenics. This is the eugenics that happens when the state is specifically excluded from reproductive decisions. It is the eugenics of the free market, and it results inevitably from a combination of the current quasi-religious faith in the absolute virtues of unfettered free markets and the

rapid growth of the new genetic knowledge. The whole point is that we are about to be deluged with offers of choices… The free market takes over where Nazism left off.'[2] German sociologist Ulrich Beck is of a similar opinion. Contemporary barbarism, he writes, 'does not appear on the political stage, clothed in the familiar garb of brutality. It gains access through the clinics, laboratories and factories of the new biochemical industries. Its victory parade does not begin with street brawls, the persecution of minorities or people's assemblies, the dissolution of parliaments and the abolition of constitutions. This time it steps onto the stage of world history dressed in white coats, of self-confident research, the good intentions of doctors and the desire of parents to do their "best" for their children.'[3]

For some, free markets mean free choices. But for the feminist writer Hilary Rose, 'when medical interventions are particularly heroic, as with infertility treatments, foetal surgery and embryo genetic screening, women can find themselves having "chosen", yet feeling that it was not "this" that they wanted.' When this happens, she continues, 'both the rights of disabled people and the complexity of decision are undervalued.' In her view, in the age of genetics 'the old and powerful mechanism of devaluing the victims and blaming mothers has been activated.'[4]

The aim of this paper is to present a different, more positive, picture of the origins, purposes and character of modern genetic testing and screening. We begin with a brief overview of the services offered, the patient needs they meet and the legal position regarding the services in the United Kingdom.

Ante-natal testing and screening are now available for an increasing number of genetic conditions. The tests are becoming more accurate and can be carried out at earlier stages of pregnancy than previously. Indeed it may soon be possible to isolate a few fetal cells from the mother's blood, thus removing the small risk of miscarriage that accompanies current procedures. Broadly, three classes of women use the service at present:

(1) Those who have already had an affected child or who have a known family history of a condition and therefore know that they are at high risk for producing another child with the same condition.

(2) Those with no family history but who are at higher than average risk of having a child with a specific condition for a particular reason. Examples include maternal age and exposure to noxious agents.

(3) Those undergoing ante-natal screening, during which a problem with the fetus is detected.

Pre-implantation genetic diagnosis is a more recent development, and is less widely available. In her new pamphlet for the Progress Educational Trust—*Designer myths: the science, law and ethics of preimplantation diagnosis*—Kay Chung explains the science upon which PGD is based. Crucially, 'the introduction of gene amplification techniques, which enable diagnosis using tiny amounts of DNA, means that genetic tests can now be performed on a single cell taken from the early embryo.'[5] This does not destroy the embryo, as cells are pluripotent at this stage of development, which

enables the loss to be made up. Approximately three embryos free from the condition are then implanted. The process was pioneered at the Hammersmith Hospital in London in the period 1990-2. It is still a highly specialised service; in the UK it is only performed at four centres. The first conditions to which it was applied were X-linked disorders such as Duchenne muscular dystrophy. Since then it has been used to test for around ten single-gene disorders. Most recently PGD has been applied to familial breast and ovarian cancer as well as some other late-onset cancers.

Primarily, families use the service because they know they have an increased risk of having a child with a genetic condition, want to have a child free of the condition, but also wish to avoid abortion. Technical difficulties notwithstanding, the invasive and expensive character of the procedure (it is not provided on the NHS) limits demand for the service. Unlike ante-natal testing, health services do not offer a screening service using PGD.

**The Legal Position**

The law on termination for fetal abnormality is set down in The Abortion Act 1967 as amended by the Human Fertilisation and Embryology Act 1990. Prior to 1990, the maximum gestation at which abortion for fetal abnormality could be provided was limited by the Infant Life (Preservation) Act 1929, but this restriction was lifted by the Human Fertilisation and Embryology Act, and such abortions are now possible at any gestation if the specified criteria are met. The Royal College of Obstetricians and Gynaecologists spells out what those conditions are and what they are taken to mean:

> The amended Abortion Act allows two medical practitioners, acting in good faith, to certify that a pregnancy can be terminated at any gestation if '… *there is a substantial risk that if the child were born it would suffer such physical and mental abnormalities as to be seriously handicapped*'. If these criteria are not met, termination after 24 weeks is permissible only if necessary to prevent grave permanent injury to the physical or mental health of the pregnant woman or if the continuation of pregnancy would involve risk to the '*life*' of the pregnant woman, greater than if the pregnancy were terminated. By contrast, up to 24 weeks, the Act allows a pregnancy to be terminated if '*the continuation of the pregnancy would involve risk, greater than if the pregnancy were terminated, of injury to the physical or mental health of the pregnant woman or any existing children of her family.*'[6]

PGD is legal in the UK. It is subject to regulation by the HFEA, although the exact terms of regulation are currently evolving. There is no specified list of conditions for which PGD is or is not allowed. In a joint report on the subject published recently, the HFEA and the Advisory Committee on Genetic Testing outlined a number of issues on which they want to hear the views of the public.[7]

There have been attempts both to tighten the law on selective termination and to define it more sharply: attempts have been made to define 'serious' as 'whether or not the 'child to be' would have a life worth saving through medical intervention', and as 'whether or not it would be legitimate to withhold treatment from a child in the same state as the embryo, even though this might result in death'. But if conditions were

drawn up for which ante-natal testing and termination on the grounds of fetal abnormality were or were not allowed, people would be denied the opportunity to make the choices appropriate to their family. Such a denial of choice would be discriminatory. Moreover, if, in particular, women were denied the opportunity to be tested ante-natally for certain conditions, it might put pressure on some to have abortions who otherwise would not. Accordingly, GIG opposes any attempt to set down conditions for testing and termination.

In relation to pre-implantation diagnosis, the issues are more straightforward: if it is a matter of implanting three fertilised eggs out of a greater number, it seems to GIG sensible to implant those free of known genetic conditions. Most people would want to make that choice, and when it is possible to maximise the chances of a healthy child it appears to GIG to be perverse not to do so. There is no 'slippery slope' because there are no difficult cases: it is good to avoid any and all disease, and that includes predisposition to disease in later life. Accordingly, GIG opposes any and all attempts to restrict the range of medical conditions for which pre-implantation diagnosis can be performed. Of course, PGD is not an easy procedure, therefore it is highly unlikely that women will start requesting PGD for what the critics of the procedure call 'trivial' reasons.

**Under Pressure to Test and Terminate?**

The critics of genetic testing would regard the picture painted above as unduly rosy. Part of their argument is that women are pressurised first into genetic testing and then into termination if the test is positive. One result of this, they argue, is a high level of anxiety and distress for the woman and her family.

Meg Stacey presents evidence of direct pressure to terminate in her article ‘The new genetics: a feminist view’. She draws on Jo Green’s 1993 survey of the attitudes of obstetricians to access to prenatal diagnosis. Obstetricians were asked, ‘Do you generally require that a patient should agree to the termination of an affected pregnancy before proceeding with amniocentesis/chorionic villus sampling (CVS)?’ A third (34%) of 375 consultants said ‘yes’.[8] Ruth Hubbard makes a more general argument along the same lines. It is her belief that while society does not uphold the collective against the individual, ‘we come mighty close when we once again let scientists and physicians make judgements about who should and who should not inhabit the world and applaud them when they develop the techniques that let them implement such judgements.’[9]

Disability rights activist Tom Shakespeare highlights the anxiety he believes is caused by such testing and pressure: ‘I support a woman’s right to choose, but believe that abortion is a traumatic route of last resort. Now that pregnancy is increasingly “provisional”, and thousands of couples are offered information leading to termination, the area of reproduction is becoming filled with anxiety and ethical complicity.’ Prospective parents, he adds, ‘join a conveyor belt without having the opportunity to consider their situation. Prejudice becomes the basis for policy. Genetics becomes the pretext for discrimination.’[10] In Hubbard’s view, taking the example of Huntington’s disease to illustrate a more general point, ‘the existence of this test puts people with a family history of Huntington’s disease in an outrageous position.’[11]

Meg Stacey is undoubtedly right: some obstetricians do put such pressure on women, and GIG is firmly opposed to this. But two corrections are needed to the impression that might be given by the 1993 survey. Firstly, other medical professionals working in the field are more sensitive to the needs of women undergoing ante-natal testing, geneticists in particular. Secondly, as Stacey herself points out, the proportion of obstetricians who operate in such an explicitly directive way is falling quite rapidly—in 1993 the proportion was 34%, but in 1980 it was 75%.

Another way to examine the issue of direct pressure is to take notice of the experiences of the women concerned. Helen Statham and Wendy Solomou at the Centre for Family Research in Cambridge are engaged in a project which is doing just that. Writing in *The Lancet* they report that so far their study has shown that 'most parents do not report pressure from doctors to terminate affected pregnancies.'[12] The National Childbirth Trust argued the opposite when they presented their research on the subject in 1997. However, their own evidence does not seem to support their stronger claims: only 10% of women said they felt pressurised into taking a test, and only 1% refused.

It is GIG's view that in general women are not pressurised either into testing or termination. Women are glad that the service is available because they genuinely want to have the choice whether or not to take the tests that are on offer. In GIG's experience this is particularly true for those with a family history of a disorder. Of course Hubbard is right that deciding whether or not to test for a terrible disorder such as Huntington's disease is very stressful, but it is not the existence of the test which places the family in an 'outrageous position'. Rather, it is the fact that the disease runs in the family that is the problem and the primary cause of anxiety. The availability of the test, and for some the taking of the test, is a means to resolve some of the dilemmas created by this fact. For women without a family history of a disorder, the high uptake of screening tests for conditions such as Down's syndrome by those who face a higher than normal risk simply because of their age suggests to GIG that most women would prefer these services to be available.

None of the above should be taken to imply that GIG believes that the service is free from problems, but GIG does believe that the problems should not be exaggerated. There also needs to be a recognition that however much women want the service, the process is bound to cause them some anxiety: professionals see their job as being on the lookout for problems; women go into the process assuming that everything is fine in the case of a routine process or hoping that everything is fine if they are at high risk. These different expectations inevitably colour attitudes towards the service.

But what about more indirect forms of pressure? The point is often made that women are pressurised into being tested in more subtle ways; more specifically that social and cultural norms, or simply the lack of resources afforded to people with disabilities, inevitably create a pressure on women to test and then terminate affected pregnancies. The suggestion is that screening in particular does not extend choice but rather restricts it by demanding conformity.

So for Ruth Hubbard, 'to the extent that prenatal interventions implement social prejudices against people with disabilities they do not expand our reproductive rights.

They constrict them.'[13] Similarly, Abby Lippman suggests that if there were greater support from society for people with disabilities, women might be more likely to choose not to terminate affected pregnancies, but that at present technological advance combined with insufficient social support implicitly force women to undergo testing and termination, which they find a burden.[14] Accordingly, argues Tom Shakespeare, 'although antenatal testing supposedly offers choice and information to pregnant women, there is implicit pressure to eliminate affected fetuses: decisions are not taken freely or on the basis of balanced information about disability.'[15]

It is undoubtedly the case that for many women the decision to terminate an affected pregnancy is a difficult one. In this sense it is a burden for them. But it would be wrong to conclude that it is a decision they feel they have been coerced into. As Dorothy Wertz and John Fletcher, two American specialists on the subject, have pointed out, pressure from women helped to create many of these services in the first place. Interestingly, Wertz and Fletcher make the further point that women exercise more choice and control in this area than they do in other areas of medicine.[16]

The notion that women are implementing social prejudice if they decide to terminate, acting in ignorance of the realities of disability, makes little sense in the context of the families represented by GIG, many of whom already have a child with a genetic condition and decide to terminate a later pregnancy so that they can offer the necessary care to the first child. They may find it a difficult decision to make, but it is a decision that is made based on a combination of concerns for their own future and first hand experience of the quality of life a child with the disorder would have.

If society were to devote more resources to helping people with disabilities then some women might make different decisions, but it is reasonable to assume that many women would simply want a healthy child regardless of social attitudes towards and resources allocated to disabled people. In any case, it would be wrong to restrict testing in order to make families at risk of a genetic disorder play the role of 'trail-blazers' for better resources.

In GIG's view, the danger may lie in the opposite direction to the one highlighted by the critics of ante-natal genetic testing and screening. It is our experience that many 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 ante-natal 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 recent study patients expressed satisfaction even when they thought aspects of their care were 'directive'.[17]

(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. 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.

**A New Eugenics?**

For GIG, selective termination and pre-implantation genetic diagnosis do not imply a negative judgement about the worth of people living with disabilities. Nor do they imply a neglect of the needs of disabled people. But for the critics of genetic testing, unfair discrimination and a broader 'eugenic' mind-set is an integral part of modern genetics. There are a number of aspects to the argument, which is put with varying degrees of vigour by different critics; and clearly, whether or not modern genetics is thought to be 'eugenic' will depend on what that emotive term is taken to mean. In this section we cannot hope to do justice to the full range of opinion and writings on this subject, but we do hope to establish 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. 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. 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.'[18]

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 GIG's 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 impinge on services offered prior to implantation or birth.

Of course, and to repeat the point, some people do *interpret* some areas of genetics in ways akin to historical eugenics—although cultural theories of group difference and degeneration are more popular today than genetic ones—but that is purely an interpretation of the science and not the science itself. To conflate the science with historical eugenics does a disservice both to modern genetics and to history.

In GIG's view, both the ethos and the science of medical genetics are quite different from historical eugenics. Medical genetics is neither 'reductionist' nor morally discriminatory. Rather, the underlying spirit of the field is to consider people equal as human beings, while recognising that some have, or are at risk of producing children with, a medical condition. Clearly, selective implantation and termination are not 'cures'. However, the spirit of the new genetics is a search for the alleviation of disease and suffering, which often takes the path of pre-implantation diagnosis or selective termination of pregnancy precisely because cures are not available. The attitudes of parents and, in large part society as a whole, to fetal abnormality reflect attitudes towards illness—they feel sympathy for the ill and they want to cure them. They do not want to marginalise still less eradicate them.

The individual choices parents make may alter the gene pool in a very limited way. But the spirit driving those choices has nothing in common with that which drove the old

eugenics. Governments provide genetic services with expectation of health gain from the avoidance of disability. There is nothing wrong with this, so long as, unlike under the eugenics of old, parents remain free to make their own choices.

**Choices and Discrimination**

Diagnosable genetic disorders are always likely to remain a small proportion of the causes of disability. Even in principle therefore, pre-birth genetic diagnosis could never be seen as an alternative to providing resources for disabled people. But more to the point, given that the motivation underlying attitudes towards testing before and during pregnancy is not to marginalize or ignore disabled people, there is no reason that genetic testing should promote such an attitude. Indeed in GIG's view genetic testing and screening are not encouraging neglect of the needs of disabled people. On the level of attitudes, society today is more supportive towards people with disabilities. On the level of resources, the problems seem to GIG to be more to do with government's desire to control public spending than anything to do with genetics and overt prejudice. That is why, through our work, we seek both to promote advances in genetics and to secure equal treatment and better resources for those living with genetic conditions.

## NOTES

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