

Choices and Boundaries

Should people be able to select embryos free from an inherited susceptibility to cancer?

The responses of the Genetic Interest Group (GIG) to the consultation by the Human Fertilisation and Embryology Authority (HFEA), January 2006

Introduction

As the UK alliance of charities, support groups and voluntary bodies for those affected by or at risk from all forms of genetic disorder GIG welcomes the decision by the HFEA to consult on the issue of selecting embryos free from an inherited susceptibility to cancer. Our response to the consultation is based on our experience of and contact with our member organisations (currently approximately 140 in number) and with the families and individuals they support. This response has also been approved by GIG's Trustee Board, which comprises members nominated and elected by our members.

In response to the questions raised we would offer the following observations:

Question 1

Pre-implementation Genetic Diagnosis (PGD) offers an important option for couples at risk from serious genetic disorders for whom other options (abstaining from having children, trusting to chance or ante-natal testing and termination when an affected fetus is detected) are unacceptable. We have consulted our members about aspects of PGD on a regular basis, and as a result of this, GIG has supported the development of PGD services, and endorses the current range of uses made of it fully – including the use to select embryos that will not only be free of a given disease themselves, but who may also be able to provide compatible tissues for the benefit of affected siblings.

Given the substantial impact of IVF on women undertaking this procedure, the psychological impact of the procedure on the couple, and the relatively low chance of the process producing the desired result (i.e. a healthy baby free from the condition in question), seeking PGD is not a decision taken lightly by those for whom it might be an option. Indeed a proportion of those who initially ask for PGD subsequently drop out when they discover the likely impact of the process on them. We believe that those who do wish to follow this route should be supported and helped in their quest for a child free of serious; possibly life threatening disease.

Question 2

Notwithstanding the fact that some inherited cancers may be treatable to some extent (and treatment options are improving), and notwithstanding the incomplete penetrance of the genes associated with these conditions there can be no doubt that these are serious conditions. The treatments currently available do not prevent or cure the condition in all cases and some conditions remain effectively untreatable. Many of the options are invasive, disfiguring and carry substantial risks themselves. Some are lifelong (from the point of symptom onset at least) and access to them is dependent on the availability of resources from either public or private funds, making it far from certain that speedy access to state of the art intervention will be possible for all who need it now or in the future. Even if safe, effective affordable interventions were guaranteed to be available, the psychological impact of late onset genetic disease on those at risk and on their families, partners or carers is significant. This in itself can have a damaging effect. In families where there is a known genetic risk and PGD is seen as a way of removing the threat to the immediate children (and also to future generations) of inherited cancer – and of the sequelae of its treatment (possible colostomy, mastectomy, chemo and/or radio-therapy and other unpleasant procedures) then this is a choice which we say they should be permitted to make, and for which they should expect support.

Question 3

GIG does not believe that it is appropriate to pick a figure and use it to establish a boundary above which PGD is permissible and below which it is not. There are a number of reasons for this:

Firstly, the concept of a “significant risk” necessarily and unavoidably contains a subjective element – both for the person applying the risk to themselves, and also for the person imparting the information. The literature relating to genetic counselling contains numerous examples of ways of presenting risk information both numerically and by analogy. Each has associated pros and cons, and none is guaranteed to give a consistent message to the recipient. (A 1 in 4 chance of an event happening may be the same as a 3 in 4 chance that it won't, but people “read” the information differently). Then, even if the information is communicated accurately, the salience of the event to the person(s) affected will impact on the importance of reducing or accepting the risk associated with the chance offered. Thus for a woman who has lost a mother and a sister to a genetic cancer, the chance of freeing her daughter from the same fate may make any option (including PGD) worth exploring.

Secondly, figures about the penetrance of genes are, even with good quality research inexact. Estimates will vary substantially, and will often change over time as new data comes to light. The penetrance of the same genetic cancer may vary between different populations.

For those who actually develop a familial condition such as genetic breast cancer the penetrance of the gene in the population is of little relevance. They have it,

and they experience the impact of the condition for themselves. This will be highly significant in determining their perception of the risk, and of their desire to use PGD to avoid passing it on to their children. If a line were to be drawn then it would create inequity between those above the line able to seek PGD, and those below – with the same condition – for whom it is ruled out.

Thirdly, because PGD is a process that few if any women embark upon lightly, we believe that the decision about the appropriateness of the procedure is best left to the woman and her partner with the support of expert clinicians in a counselling environment. This is so that as fully informed a decision as possible can be made in the light of all available relevant information.

Question 4

GIG believes that the views of people seeking treatment should take precedence of those who may have other opinions about whether or not such procedures are ethical. The legitimacy of using genetic techniques to avoid the birth of a person who will be affected by serious, often life limiting, diseases has been debated extensively since the passage of the Abortion Act, the Human Fertilisation and Embryology Act, in connection with the use of embryonic stem cells and on countless other occasions over the years. In each case the majority opinion has endorsed the legitimacy of taking steps to avoid or reduce the impact of genetic disease through measures such as ante-natal testing and termination of affected pregnancies. The fact that, notwithstanding the increasing number of genetic conditions that can be tested for; the number of terminations each year has remained more or less constant indicates that the decision is not taken lightly by those affected. This and the number of women who drop out of PGD prior to commencing treatment supports the idea that those seeking to avoid a given condition do so only after considerable thought. This seriousness of intent should be respected. Those for whom PGD poses ethical problems are under no compulsion to avail themselves of this option, but should respect the views of those whose ethical values do not reflect their own, but are no less deeply held, and who find PGD a humane and acceptable option in order to avoid passing on inheritable conditions from one generation to the next.

Question 5

Whilst current guidance from the HFEA states that the use of PGD should be consistent with current practice in pre-natal diagnosis (PND) in practice this has already been superseded by developments licensed by the Authority – particularly in the case of selecting for the purposes not only of avoiding disease. In a future child but also to benefit an affected sibling, where PND would not generally be seen as appropriate or acceptable. Indeed the explicit use of PND for the selection of a fetus that was not at risk of a genetic disease itself but to provide an existing sibling with a tissue match would not be permitted by the Abortion Act.

As has been noted above the decision to terminate a wanted pregnancy is not one taken lightly by those who find themselves facing it. Experience of the requests that come forward for funding PGD by the NHS show that many of those who have experienced pre-natal diagnosis and termination find PGD a more acceptable, less traumatising way to try for a healthy baby. Whilst few would want to use PND to avoid the risk of passing on a lower-penetrance condition it is not an option that is ruled out absolutely by law or in current clinical practice. The risks to the mother and the possible psychological impact of PGD are also not trivial, emphasizing the importance of skilled support for those contemplating this option.

Given that the link between PND and PGD has in practice been at least stretched if not actually broken, it would be inappropriate to seek to re-instate it, and conflate the two procedures, in the case of lower penetrance inherited disorders where PGD provides an acceptable route for some to avoid putting their descendants at risk.

Question 6

The drawing up of lists and the setting of arbitrary boundaries is inappropriate in our view. Conditions vary in their natural history, their age of onset, their severity and in the impact they have on those affected or at risk. Setting boundaries is likely to create suffering and injustice for those who fall on the “wrong” side of the line but who turn out to have a particularly unfortunate version of the presenting condition. For example, cleft lip and palate is sometimes cited as a condition for which termination of pregnancy should not be allowed as it is repairable by surgery. Where as it can be repaired in most cases, in some there is nothing with which to re-construct the face, resulting in very severe deformity and in all probability severe psychological distress. Similarly the experience of a parent will colour their view of the seriousness of a disease for their children, and decisions taken at one point may not be made in the same way should similar circumstances arise later in life. Different people, faced with similar circumstances will respond very differently – sometimes for reasons unrelated to any “quasi-objective” assessment of the seriousness of the condition or of the risk of it manifesting itself in their offspring. For these reasons we feel that the appropriate place for decisions to be made as to whether PGD could be an option to be explored is in the clinic, where multi-disciplinary expertise is on hand to work with the woman (and her partner where appropriate) to reach a conclusion that is most likely to be liveable with by the woman/couple in the long term. There is no evidence that we are aware of that indicates that women/couples are seeking to use PGD/IVF for reasons that might be characterised by some as “trivial”. Indeed the reverse seems to be more likely to be the case, with women who are highly motivated to avoid the birth of a/another child with a serious, perhaps even a lethal genetic condition being put off by the physical and psychological stresses that this process creates.

Should the HFEA feel, at some point, that the use of PGD is being “trivialised” by a particular centre or clinician then its existing powers of Inspection would surely give it the power to “call in” cases for discussion/further examination. However we do not believe that this will become a widespread problem, and to impose arbitrary limits runs the risk of stigmatising women who might otherwise come forward for help, even if they subsequently decide that following the provision of information, counselling, care and support PGD is not for them.

We would be happy to expand on any or all of the above points if it would be helpful for us to do so.

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