# SHOULD THE LAW LIMIT GENETIC TESTS ON EMBRYOS AND FOETUSES?

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As genetic technology improves, there will be an increasing number of tests available to detect foetal abnormalities. Many couples will wish to take advantage of these tests to improve their chances of having a 'healthy' baby and avoiding the transmission of genetic conditions to their children and grandchildren. An issue arises whether there should be limits on the tests that are offered and, if so, whether those limits should be imposed by the law or in some other way. These questions are not merely academic. Already, some European countries have legislated to restrict the genetic tests that may be conducted on embryos created in infertility treatment programs (so-called pre-implantation diagnosis ('PGD')). In Germany, PGD tests are prohibited by law and Italy has recently introduced similar restrictions. This paper argues that Australia should not take this path, even if some people are concerned about the proliferation of new genetic tests and believe that testing should only be permitted for 'serious' genetic disorders.

It should be recognised at the outset that there are two types of tests for foetal abnormality. The first, which has been mentioned already, is PGD, which can be conducted by removing a cell from an embryo created in an infertility program in order to detect a mutation that is known to exist in a family. If an embryo is found to be affected, that embryo can be discarded and an unaffected embryo can be chosen in its place. Conditions that can be detected in this way include chromosomal abnormalities that cause early death (like trisomy 13) or serious intellectual impairment but prolonged survival. They also include other conditions that affect the child from birth, like Down syndrome, cystic fibrosis, genetic deafness, haemophilia and phenylketonurea ('PKU'). Down syndrome involves a varied prognosis but will inevitably involve mild to moderate intellectual disability. There are treatments for other conditions mentioned: cochlear implant for deafness; blood transfusions for haemophilia; dietary restrictions for PKU. However, the conditions have a significant impact on day to day life, the interventions are burdensome and they must be undertaken throughout life.

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Other conditions that can be detected by PGD have an effect later in life, like the colorectal cancer familial adenomatous polyposis ('FAP'), which is fatal if not detected and treated with surgery; and Huntington disease, a chronic and life-shortening condition that develops later in life. The mutations that cause these conditions are of high penetrance and people who inherit them will certainly develop the condition at some stage. Other tests are designed to detect conditions that may not affect the child at all, such as increased susceptibility to develop breast cancer later in life; or carrier status, where the child is normal but has a risk of passing on a genetic condition to his or her offspring. However, even if a child has only a susceptibility to develop a condition, and not a certain prospect of developing it, the parents may choose to avoid that risk if they can do so easily by choosing an unaffected embryo for implantation. Similarly, if they are able to choose an embryo that will not make the child a carrier for a family mutation, then the condition can be eradicated in the family.

Finally, PGD tests may reveal that a child will be healthy but have particular characteristics, such as gender, being a matched tissue donor for a sick sibling, or having a particular hair and eye colour.

Thus, important factors in considering the PGD tests that may be undertaken include the reason for the test, especially the severity of the condition being tested; the degree of risk that the child will be affected; the time of onset of the condition; and the availability of treatment for the condition.[1] On the other hand, it should be noted that the outcome of a positive test is the choice of an unaffected embryo, not the termination of a pregnancy.

Other genetic tests can be done only after a woman has become pregnant and some of the conditions revealed by these tests cannot be detected until late in the pregnancy. Conditions that can be detected by these *prenatal* tests (in contrast to PGD) include anencephaly, where the brain fails to develop and the baby will die soon after birth; other severe brain damage causing profound and irremediable mental retardation; spina bifida; severe physical malformation; cleft foot; cleft palate and dislocated hips, which are all correctable by surgery; and achondroplasia (dwarfism). If a prenatal test reveals a genetic condition, the woman may choose to terminate her pregnancy and, inevitably, this may occur late in the pregnancy.

As noted earlier, some European countries have recently legislated to prevent PGD apparently in order to protect early embryos from potential harm; or, especially in the case of Germany, due to concerns about 'eugenics'. However these countries do not prevent prenatal tests and termination of pregnancy if a foetus is affected. If a foetal abnormality could be detected by PGD and that is not permitted by the new laws, then the provisions have the anomalous result that, instead of an early embryo being discarded, a foetus may be aborted at a much later stage of development. On the other hand, women may be reluctant to terminate a pregnancy for relatively minor reasons, so the availability of prenatal tests, but not PGD, may in practice limit choices that are seen as 'eugenic'.

1 Being a genetic condition, it cannot be 'cured'.

However one views the desirability of limiting genetic tests on embryos or foetuses, the question remains whether the law is the most appropriate means of regulation. It is clear from the discussion above that it would be necessary to prohibit or restrict both PGD and prenatal tests if one were determined to ensure that all embryos and foetuses are fully protected from tests and possible destruction. Or, one might adopt the German and Italian option of prohibiting PGD and allowing prenatal tests on the assumption that only severely impaired foetuses are likely to be aborted.

The most effective way to prohibit or limit genetic tests would be to legislate to ban or restrict all genetic tests on an embryo that will be implanted into a woman or on a foetus in utero; or all tests except those prescribed in the legislation; or all but those prescribed in regulations, or by a body authorised by the legislation to make such decisions. The latter options would enable greater flexibility in the regulations as new tests could be added later as they become available.

However, assuming that legislation of that kind was proposed, problems arise in drafting it. An initial issue is where to draw the line in deciding which tests (if any) should be allowed and which prohibited. It would be clearer to list the particular tests that are allowed or prohibited, rather than use a generic expression like 'severe conditions'. What seems 'severe' to one person is 'within the range of human difference' to another. However, if the genetic tests are listed, it may be difficult to describe them. One might refer to the test by name, such as X, but that would not prevent a test by other names for a particular condition. Similarly, if one referred to 'a genetic test for condition X', the meaning of 'genetic' would be problematic. Does it mean, as in common parlance, a DNA test? If so, other tests would still be possible, such as a test for a gene 'product', which is a biological test that does not involve testing the DNA itself.[2] Women could also avoid the ban by seeking tests in another country.

An alternative regulatory scheme might focus on the disposal of particular embryos on genetic grounds; or the termination of pregnancy because of foetal abnormality. However, the former is inconsistent with accepted best clinical practice in choosing the 'best' embryos for implantation; and the latter is inconsistent with the law on abortion, which in practice is available on request even for 'social' reasons at least in the early stage of pregnancy.

In summary, legislation could be passed to prohibit or restrict the genetic tests that may be undertaken before an embryo is implanted or after a woman has become pregnant.[3] It would be necessary to have both types of provision in order

2 See Australian Health Ethics Committee of the National Health and Medical Research Council, *Ethical Aspects of Human Genetic Testing: an Information Paper* (2000) 9 <http://www.nhmrc.gov.au/publications/_files/e39.pdf> at 16 July 2006: 'A genetic test is one that reveals genetic information. It may be performed on DNA, RNA or protein (the 'gene product'), or involve measurement of a substance that indirectly reflects gene function. Examples of the latter two groups are haemoglobin electrophoresis to diagnose carriers of beta-thalassaemia and measurement of blood cholesterol to diagnose familial hypercholesterolaemia in a child whose parent has the disorder'.

3 One might limit the grounds for termination of pregnancy to 'serious' conditions, or require all embryos formed in infertility treatment to be implanted in a woman but, if legislation were proposed, it would be better to limit the tests that may be conducted.

to prevent entirely what some people regard as a 'eugenic' approach to human reproduction. However, at present we allow women to undertake PGD and prenatal tests of various types, both physical (like ultrasound) and biological (like genetic tests), and to discard an embryo or to terminate a pregnancy if there is an adverse result.[4] If it is possible to test an embryo before implantation and to choose an unaffected embryo, then the potential 'harm' is discarding an affected embryo, not terminating a pregnancy. We accept that embryos that are not needed in infertility treatment programs may be discarded (left to succumb), even if they are healthy. We also accept the medical practice of choosing for implantation those embryos that have the best prospect of development into a healthy baby. PGD seems to fit in with these principles and, in my view, it should not be prevented or restricted by law. There is no reason, on the basis of clinical experience to date, to believe that women will use PGD for frivolous reasons such as determining minor characteristics of appearance and it is not necessary to legislate against such tests. Although views may differ about where to draw the line on the tests that are desirable, if an occasional couple feels so strongly about the immuno-compatibility status of their child, or even its sex, that is not sufficient reason to enact legislation to prevent, or even restrict, PGD. The European countries that have done so appear not to have considered the risk that women will terminate pregnancies instead of choosing an unaffected embryo after PGD, yet the latter seems to me a better option. However, it is also necessary to allow prenatal genetic testing to continue in order to detect conditions that cannot be detected by PGD. Women do not lightly discard affected embryos after PGD and they are even less likely to terminate pregnancies for frivolous reasons. Indeed, experience indicates to the contrary. Couples are prepared to accept even substantial disability in their child. For these reasons, neither PGD nor prenatal genetic tests should be limited by law.

4 In most Australian jurisdictions, foetal abnormality is not a ground in itself for terminating a pregnancy but may be regarded as constituting a threat to the mother's physical or mental health, which is a ground for lawfully terminating a pregnancy.