Genetic testing of children

By Margaret Otlowski

As a result of the joint Australian Law Reform Commission and the Australian Health Ethics Committee Inquiry into the protection of human genetic information which culminated in a major report released in May 2003, increasing attention is being given in Australia to legal, social and ethical issues associated with predictive genetic testing and the use of an individual's genetic information. The predictive genetic testing of minors raises particularly difficult problems and poses some dilemmas for law and policy makers.

This article addresses only the issue of predictive genetic testing of children as opposed to genetic testing for diagnostic purposes, or other DNA testing such as parentage testing.[1]

Predictive genetic testing of children is undertaken on children who are asymptomatic but who—because of family history—may be at risk of developing a genetic disease or disorder in the future. The defining feature of such testing is that it is relevant to the person's future health status, identifying a condition or disorder which the person may present with at some later time. In the case of some rare, single gene, late onset conditions, inherited on an autosomal dominant basis, such as Huntington's Disease, testing may reveal that the person has the mutation that will lead to the development of that genetic disease virtually as a matter of certainty. More commonly, however, predictive genetic testing identifies predisposition or susceptibility to a genetic condition or disorder rather than pinpointing pre-symptomatic status. Whether or not the condition or disorder ultimately manifests will depend on the complex interplay between the individual's genes and his or her environment. In some circumstances, lifestyle or other prophylactic interventions may be possible, however for many conditions and disorders no preventive or therapeutic treatments are presently available. Particularly in such situations, the impact of genetic test information can be significant, with potentially harmful psychosocial consequences for individuals resulting from this information. This is recognised by key organisations and health care professionals in the field who seek to promote the importance of informed and free choice with regard to genetic testing, facilitated by the availability of appropriate pre-testing counselling by qualified professionals.[2] Underpinning this view is the recognition of the significance of the right to know as well as its corollary, namely the right not to know as encapsulated in major international instruments.[3]

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The regulatory framework for the predictive genetic testing of children

Few would dispute that decisions about predictive genetic testing can have profound consequences for an individual. Ideally, these decisions should be made by the individual concerned at a time that they are in a position to weigh up all the issues. This may not always be feasible and there are, undoubtedly, some situations where predictive genetic testing of minors is justified, and ultimately in that person's best interests because it allows appropriate interventions to be undertaken. There is, however, considerable debate about the circumstances in which parents should be able to initiate predictive genetic testing of their children, and growing awareness and concern about the potentially harmful effects of predictive genetic testing of minors if inappropriately carried out.

Currently, there are no laws in Australia which directly regulate the predictive genetic testing of children. Further, there is nothing to restrict laboratories performing predictive genetic tests where parents have consented on behalf of the child, and the availability of direct-to-consumer genetic testing via the internet has expanded the opportunity for parents to access genetic testing. Although there is no direct regulation of this area various professional bodies and health organisations have developed guidelines regarding the acceptability of predictive genetic testing of children, including the World Health Organisation,[4] the Nuffield Council on Bioethics,[5] the American Society of Human Genetics[6] and the Human Genetics Society of Australasia.[7]

These organisations have overwhelmingly concluded that predictive testing for adult onset diseases—for which there is no known treatment or preventive strategy has no immediate benefits and should not be performed on children but deferred until adulthood—or at least until the person is able to appreciate the relevant genetic facts as well as the emotional and social consequences of what predictive genetic testing entails. Where, however, direct benefit to the child can be demonstrated through medical surveillance or intervention, predictive genetic testing of children is generally regarded as acceptable. For example, children in danger of contracting familial adenomatous polyposis (leading to colon cancer) can establish whether they carry the genetic mutation responsible for the disease. They can, thereby, decide whether they need to undertake ongoing surveillance which is usually offered to at risk individuals between the ages of 10-15 years.

Underpinning these guidelines on predictive genetic testing on children are ethical concerns about the effects of such testing, in particular the potential of psychological damage to the child. Testing may result in diminished self-esteem, difficulties in interpersonal relationships and altered parental perception of, and behaviour towards, the child.

Testing children—who cannot give their informed consent—breaches their autonomy and interest in genetic privacy and their right to choose not to know about their long-term health prognosis. This may create difficulties in coping with the knowledge of the likelihood of disease in later life. It can also potentially lead to detriments such as discrimination in insurance and employment. It is recognised, however, that testing may bring some psychosocial benefits such as relieving anxiety about possible early signs of the disorder, reducing uncertainty about the future, providing the possibility of appropriate forward planning of matters such as education, housing and family finances, including estate planning, and, in the context of later reproductive choices, identifying children who might benefit from predictive genetic testing in the future.[8] Currently, fears about the possible harms that could be caused by testing in childhood are believed to outweigh any potential advantages,[9] particularly at the present time when there is a lack of evidence-based research demonstrating the psychosocial consequences of predictive genetic testing of minors.

Notwithstanding the strong support for the prevailing approach against predictive genetic testing of minors, there does appear to be recognition that opinions on this issue do differ, and that the circumstances in which these matters arise are variable. This has resulted in general support for the position that there should not be a categorical prohibition on predictive genetic testing of children which offers no immediate therapeutic benefit. Rather, it is regarded as preferable to allow health care professionals and genetic counsellors to work through the problems with the families, and to be permitted to make exceptions in individual cases, in situations where it is believed to advance the welfare of a particular child. Most of the guidelines on this subject are couched in strong but not absolute terms, thus permitting flexibility in appropriate cases. There is, however, a consensus that predictive genetic testing of children must be undertaken with care.

Role of parents in decision-making about genetic testing of their children

Parents have parental responsibility in respect of their children (unless a court orders otherwise),[10] so potentially they may have the legal authority to authorise the predictive genetic testing of their child or children. Parental decisions about predictive testing for genetic disorders should be made according to whether, objectively assessed, the child will benefit from such testing, not in order to relieve the anxieties of the parents. In other contexts (for example, cases of non-therapeutic sterilisation to be performed on mentally retarded girls), the courts have indicated that children should be given the chance to make choices about medical care for themselves if it

is possible to wait until they are able to do so without risking their health.[11]

Circumstances in which parental decision-making may come under legal scrutiny

Whilst one might hope that sensitive counselling could resolve most potential disagreements amongst family members, or between family members and health care professionals, about the desirability of predictive genetic testing for individual children, there may be situations where it is necessary to resort to the courts in an effort to safeguard the best interests of a child. A number of possible scenarios can be put forward. One possibility is that parents want to have the child tested but the child objects. In these circumstances, other persons (for example, a social worker or health care professional) may become involved as advocate for the child, with a view to assisting the child to withstand inappropriate parental pressure to be tested.[12] Even in the absence of objection from the child, a third party may intervene because they believe that the course proposed by the parents in relation to predictive genetic testing is not in the child's interests.

Another possibility is that the child may wish to undergo predictive genetic testing but the parents object. It is also conceivable that parents are in conflict about whether or not predictive genetic testing should be carried out on their child. In each of these scenarios, the jurisdiction of the Family Court could conceivably be invoked to determine whether a particular child should be tested. There is specific provision in the *Family Law Act 1975* (Cth) to deal with applications for an order in relation to the welfare of a child[13] which clearly would encompass the issue of predictive genetic testing of children. In deciding whether to make such an order, a court must regard the best interests of the child as the paramount consideration.[14] The process of assessing the child's best interests would need to take account of a whole range of matters, going well beyond the question of health benefits of predictive genetic testing. Amongst other things, the Court would be required to take into consideration any wishes expressed by the child and any factors (such as the child's maturity or level of understanding) that the Court thinks are relevant to the weight it should give to the child's wishes.[15] In cases of conflict between parents and the child, it is likely that an Independent Children's Lawyer (ICL) would be appointed on behalf of the child.[16] The role of the ICL would *inter alia* be to present information to the Court about the child's wishes, but also to submit his or her own assessment of what is ultimately in the child's best interests. It is difficult to predict the outcome of such cases—the child's level of decision-making competence would obviously be a significant factor—but even where it could be shown that the child is of an age and maturity to express well founded wishes and views, this would not necessarily be decisive. The Family Court would need to assess whether these wishes are consistent with the child's best interests which are the paramount consideration.[17] This would inevitably involve a process of weighing up the harms and benefits for that particular child of proceeding with testing. In view of the complexity of this assessment of pros and cons of testing, caution would be required in acting on the decision of a child.

Whilst the availability of this jurisdiction is useful as a possible check on inappropriate decision-making on the part of either parents or children, it does have inherent limitations. Invoking this jurisdiction is time consuming and expensive and in practice, it will often depend on health care professionals, social workers or others becoming drawn in to bring the matters in conflict before the court. There are also constitutional constraints due to the fact that the State referral of power in respect of ex-nuptial children extended only to matters of custody, guardianship and access. Accordingly, this welfare jurisdiction only applies to children of married parties.[18]

What form of regulation is appropriate?

Questions remain about the most appropriate course of action for the regulation of this area—whether to rely on the 'soft' approach of professional guidelines underpinned by the potential intervention of the Family Court in circumstances where the testing may be regarded as contrary to the child's best interests—or whether a more structured, interventionist strategy is required providing some form of independent vetting of decisions for predictive genetic testing to be undertaken, at least in circumstances where there are no immediate or clear health benefits for the child.

It must be acknowledged that the current arrangements are by no means foolproof, and will not necessarily protect a child from inappropriate predictive genetic testing which

may be perceived objectively to be against the child's interests (eg a doctor may be inappropriately influenced by wishes of the parents, testing may be contrary to a child's interests, possibly even going against the child's expressed wishes, no third party to raise objection etc.) However, for the majority of cases, the combined effect of professional guidelines, professional practice, and the safety net of the family law legislation which can be invoked to protect a child's interests will suffice to ensure that predictive genetic testing is not undertaken inappropriately. From a practical point of view, it would in any event be impossible to effectively prohibit or regulate access to genetic testing for children, due to the availability of direct-to-consumer genetic testing which transcends national boundaries.[18]

Introducing a more interventionist approach, requiring oversight of family decision making, would probably be anathema to believers in family autonomy, implying as it does, that families working together with their genetic counsellor and doctor/geneticist cannot be trusted to make wise choices in the best interests of their children. Whilst objectively, this course of action would be more protective against the risk of inappropriate predictive genetic testing, the costs of such intervention must also be weighed in the balance—in particular, the negative aspects of disempowering parents as decision makers, and the bureaucracy and inevitable financial burden associated with such a system of oversight. In these circumstances, it seems far more appropriate to acknowledge the need for some flexibility in decision-making, to try and educate and support families as much as possible, and to encourage responsible use of predictive genetic testing of children.

Conclusion

Where predictive genetic testing is performed on a child, that person's freedom of choice, in particular, the right not to know, is taken away, as is any claim to confidentiality about their genetic status. The position taken in this article is that there are compelling reasons why the use of predictive genetic testing on children should be confined to situations where it is clearly justifiable in the child's best interests. The existing regulatory framework, although somewhat open textured and non-mandatory, is probably, on balance, preferable to a more interventionist model. This is not to say, however, that there are not areas where improvements can be made, for example, stepping up requirements for pre-testing genetic counselling for the families involved, so that the full range of issues, including potentially negative consequences for the child, can be addressed.

Endnotes

1. On the subject of parentage testing, see the Australian Law Reform Commission/ Australian Health Ethics Committee. Report, *Essentially Yours: Protection of Human Genetic Information in Australia*, ALRC 96, (2003).
2. See, NHMRC, *Ethical Aspects of Human Genetic Testing: An Information Paper* (2003).
3. See, the Council of Europe, *Convention on Human Rights and Biomedicine*, Strasbourg, November (1996), Article 10; United Nations Educational, Scientific and Cultural Organization (UNESCO), *Universal Declaration on the Human Genome and Human Rights* (1997) Article 5.
4. World Health Organisation, *Proposed International Guidelines on Ethical Issues in Medical Genetics and Genetics Services* (1997).
5. Nuffield Council on Bioethics. *Mental Disorders and Genetics* (1998).
6. ASHG/ACMG Report, 'Points to Consider: Ethical, Legal and Psychosocial Implications of Genetic Testing in Children and Adolescents' (1995) 57 *American Journal of Human Genetics* 1233.
7. Human Genetics Society of Australasia, *Predictive Testing in Children and Adolescents* (1999).
8. Holland, J., 'Should Parents be Permitted to Authorise Genetic Testing for Their Children?' (1997) 31 *Family Law Quarterly* 321.
9. Clarke, A. and Flinter F., 'The Genetic Testing of Children: A Clinical Perspective' in Marteau, T. and Richards, M., (eds) *The Troubled Helix: Social and Psychological Implications of the New Human Genetics* (1995) 164.
10. See s 61C and s 61D *Family Law Act 1975* (Cth) dealing with parental responsibility and the effect of parenting orders.
11. *Secretary, Department of Health and Community Services v JWB and SMB* (1992) FLC 92-293 ('Re Marion'); *Re D* 1976 Fam 185.
12. Under the *Family Law Act 1975* (Cth), any person concerned with the care, welfare or development of the child can bring proceedings in respect of that child: s 69C(d).
13. Section 67ZC *Family Law Act 1975* (Cth) .
14. Section 67ZC(2) *Family Law Act 1975* (Cth).
15. Section 60CC *Family Law Act 1975* (Cth).
16. Sections 68L and 68LA *Family Law Act 1975* (Cth). Note also the Family Court's Guidelines for Child Representatives: Practice Directions and Guidelines.
17. *H and W* (1995) FLC 92-598, R and R: *Children's Wishes* (2000) FLC 93-000.
18. But see the case *In the Marriage of Schorel and Elms* (2000) 26 FamLR 88 which is authority for the proposition that the reference of power in respect of ex-nuptial children by the state of Victoria encompassed welfare matters arising under the wardship jurisdiction of the Supreme Court of Victoria, such that children who are wards of the Supreme Court would be covered by the state referral of power. Given the similar terms of the referral of powers legislation, this reasoning could apply more generally to referring states.