A bird’s eye view of the debate about predictive genetic testing in minors

Genetic testing now allows us to reliably predict whether people will develop certain late-onset genetic conditions such as Huntington disease. If you were at risk for a genetic disease which will only take effect in the distant future, would you want to know now whether you will later develop this disease? This can be a profoundly difficult question for many. But from a clinical point of view, the question is often relatively simple: we should let patients decide whether to undergo such genetic testing. It’s their difficult decision to make. Things get more complicated when we consider the use of predictive genetic testing in minors, for conditions that will only manifest themselves in adulthood. Many oppose this kind of testing. They think we should wait until the child grows older, and can decide for herself. Others object that this ignores the interests of the child, as well as those of the parents, who could, for example, make appropriate future plans.

This ethical debate has been going on for several decades. The important feature article by Cara Mand and colleagues (See page 519, Editor’s choice) that opens this issue doesn’t directly enter into this debate. Instead, it takes a bird-eye view of how the debate has evolved since the seminal first articles of the 90s. On the basis of a comprehensive review of the relevant literature, Mand and her colleagues have identified 53 articles focusing on this ethical issue, which they then proceed to analyse using interpretative content analysis. In this way, they were able to chart the evolution of arguments for and against predictive genetic testing in minors for conditions for which such testing provides no immediate medical benefit. The picture that emerges is interesting, and perhaps disturbing: the very same arguments appear to be repeated by either side of the debate, with little change over time. Worse, some of the key arguments on both sides are based on empirical claims for which at the moment there is little hard evidence. Mand and her colleagues conclude that if we want to make genuine progress on this important ethical issue, we need to begin to rigorously assess these empirical claims—claims, for example, about the relative benefits and harms both of testing and of the decision not to test.

Mand et al’s article is followed by several vigorous commentaries. The commentary by Stephen Robertson and Nicola Kerruish (See page 525) raises a number of worries. They are sceptical about the attempt to reduce a heterogeneous and complex moral landscape to quantifiable date. Ethical questions about predictive genetic testing in minors can only be addressed from close up, by paying attention to the distinctive psychosocial features of particular situations. Thus although more empirical date can be useful, it will not resolve the ethical debate.

A similar line is taken by Anneke Lucassen and Angela Fenwich (See page 531). They argue that the group of cases falling under the label of predictive testing in minors is far too heterogeneous. It is thus unhelpful to seek a single overarching resolution to the debate—the ethical solutions should be sought at a more fine-grained level. Lucassen and Fenwich thus resist Mand et al’s unflattering representation of the current debate as stuck in an empirical uninformed stalemate. They think it’s unlikely that further empirical data will really remove disagreement, pointing out the longstanding impasse about issues such as abortion, where empirical data is not lacking. They further suggest that the appearance of stalemate is illusory, since advances are nevertheless being made through the development of guidelines, and the growing experience of clinicians.

Angus Clarke’s critical commentary (See page 527) goes further. Clarke raises doubts about the methodology of counting the frequency in which certain arguments are cited. He doubts, in particular, whether the main arguments against these forms of predictive genetic testing in minors are really dependent on empirical claims. They main argument here, he thinks, is the principled worry about autonomy, a worry that cannot be addressed by further empirical research. But even if we thought that such empirical research is worthwhile, Clarke points out that it is far more difficult to pursue than assumed by Mand et al. Such studies, for example, would need to follow children and their perhaps reluctant parents over the course of decades. They raise formidable ethical and methodological challenges. In line with the previous commentators, Clarke suggests that it would be better to proceed by letting clinicians use their judgment in particular cases. When it is agreed that it’s appropriate to allow predictive testing in some minor, doctors could keep contact with families and later report their long term experience.

The commentary by Bernice Elger (See page 529) is more supportive. Elger agrees that opposition to predictive genetic testing in minors is often based on little empirical evidence. But she thinks that the literature review by Mand and her colleagues actually excludes some key articles, and thus their survey overlooks a number of important arguments in favour of allowing such predictive testing.

In their reply (See page 533), Mand and her colleagues reaffirm the importance of rigorous empirical research. They think that the kind of opportunistic, anecdotal data drawn from particular cases, recommended by several of the commentators, cannot support any serious substantive conclusions. The research needs to be more systematic, conducted through a formal international effort. They also clarify some of their suggestions: we need to start, they think, with mature minors, a form of research which is not that different from similar research already done on adults, and which requires a shorter follow up. And they point out that although such longitudinal studies raise many difficulties, they have been successfully carried out in other domains. The difficulties are genuine, but not insurmountable. This seems to me right.

The debate about predictive testing in minors is extremely important, but this discussion is also of general interest. When we engage in some heated ethical debate, it is natural to take sides, and to focus on the current (or next) step in the dialectic. But we sometime also need to...
step back, and consider the debate from a greater distance. How did it get to where it is now? Is progress actually being made? What might change the playing field, or even lead to a satisfactory resolution? In the context of the debate they are discussing, Mand and her colleagues suggest that what is missing is further empirical evidence. Some of their commentators doubt this. This is in part because such evidence would be very hard to obtain, but also because of doubts about the role of empirical evidence in the ethical debate. It will be interesting to see if this is true: if the relevant empirical evidence would really change the debate about such testing in minors, or whether the two sides will remain entrenched, shifting their focus to other, less empirical concerns.

This issue of the Journal also contains interesting discussions of a range of other topics. Here we only have space to highlight a number of the other articles.

Ethics of Personally Controlled Electronic Health Records (PCEHR)
Merle Spriggs and her colleagues examine ethical issues arising in connection with the government developed PCEHR that is to be launched in Australia, a system whereby patients will be able to exercise some personal control over the stored data. Spriggs and her colleagues worry that debate about, and development of, this system has so far focused on technical issues and functionality, without proper acknowledgment of some subtle ethical issues. These include questions about equity, in particular in relation with those who do not opt to join the system, questions about standards of consent and privacy, and questions about the legitimate use of the data, and about who genuinely benefits from it, and might thus be expected to pay for it. Although these questions are largely discussed here in the Australian context, they relate to general ethical issues about the ethics of national electronic health record initiatives.

German perspectives on futility
Ralf Jox and his colleagues report the results of a qualitative study of perspectives on medical futility of intensive and palliative care clinicians in Germany. The first stage of the study involved an analysis of the protocols of ethics consultations in a centre over a period of 12 months. The next stage of the study involved interviews with physicians and nurses who were present in these consultations; these included clinicians from both intensive care and palliative care units. The interviews revealed the clinicians regularly provide patients with futile treatment, such as ineffective chemotherapy for end-stage cancer. The key reason given for this wasn’t pressure from patients or family, but fear about communicating the full situation. The interviews also revealed intriguing differences in the way in which news about futility were delivered by intensive as opposed to palliative care clinicians. Intensive care clinicians tended to be more cautious, indirect and matter of fact, whereas palliative care clinicians were more direct and personal. Most importantly, clinicians did not share a accepted definition of futility. The authors end by offering an interesting algorithm for end of life decision-making which tries to integrate the perspectives on futility emerging from this admittedly exploratory study.

An argument against abortion, revisited
Finally, let me draw your attention to the latest step in the exchange between Don Marquis and Carson Strong. Some years ago, Marquis put forward an extremely influential argument against the morality of abortion. He famously argued that abortion is wrong because it deprives an embryo or foetus of a ‘future like ours’. In a recent paper in this Journal, Strong criticised Marquis’s argument, forcing Marquis to offer several clarifications to his argument. Strong now replies to this reply. He argues that Marquis has in effect made some significant concessions to his critics, and that Marquis’s revised version of the argument begs important questions. This, needless to say, is a debate that is unlikely to be resolved any time soon, but with each round of the exchange one senses that critical issues are being clarified, and significant progress is nevertheless being made.