Analysis and comment

Ethical debate

Refusing to provide a prenatal test: can it ever be ethical?

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A couple in which the man carries the mutation for Huntington's disease request prenatal testing during their first pregnancy. Though they would not terminate an affected pregnancy, they would like the information. There is no treatment available that can change the course of the disease so the diagnosis will not result in medical benefit for the child

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Introduction

Huntington's disease is an autosomal dominant lethal neurodegenerative disorder, with a mean age of onset of 37 years.¹ There is no treatment that modifies the natural course of the disease. Prenatal genetic testing can raise difficult ethical issues for clinicians.

The International Huntington Association and the World Federation of Neurology have published guidelines on the provision of predictive genetic testing for Huntington's disease.² Two aspects of these guidelines are relevant. Firstly, recommendation 2.1 states that "the test is available only to individuals who have reached the age of majority." This stance is echoed in guidelines published by human genetics societies internationally.³⁻⁴ Secondly, recommendation 7.2 states that "the couple requesting antenatal testing must be clearly informed that if they intend to complete the pregnancy if the fetus is a carrier of the gene defect, there is no valid reason for performing the test."

Here we present two opposing professional views about the ethics of the courses of action available in this case.

Not to test (M B Delatycki)

In about 95% of cases the onset of Huntington's disease is in adulthood.¹ All major human genetic societies recommend against testing minors for genetic disorders that have their onset in adulthood and for which no preventive treatment exists.³⁻⁴ The recommendation is based on two principles. Firstly, individuals should be able to make an autonomous decision about whether or not to have predictive testing at a time when they are intellectually and emotionally capable of making this decision. Secondly, the child's confidentiality is breached in that the genetic result is made known to third parties (the parents) without the tested individual's consent.

The same societies' policies take a more permissive stance from the perspective of prenatal diagnosis. For example, the guidelines of the Human Genetics Society of Australasia state that "... prenatal diagnosis

Immunofluorescent light micrograph of a nerve cell from the striatum of the brain. An immunofluorescent antibody specific to the mutant protein, huntingtin, that causes Huntington's disease has been added to show where it is expressed.

should be available for women who have a high risk fetus with a DNA identifiable condition" and that "the action to be taken on the basis of the abnormal result is a decision for the couple concerned based on the information given with full counselling support."⁵

Testing of a fetus for Huntington's disease without termination of the pregnancy is no different from testing a minor. The parents will be aware of their child's disease status and their child will not have a choice about whether or not to be tested if the parents choose to disclose the result of the test. This is different from prenatal diagnosis for conditions that are diagnosed soon after birth such as Down's syndrome and cystic fibrosis. In such cases, there is no time before the onset of symptoms for the affected individual to reach an age at which they can make an informed choice about whether or not to have predictive testing.

The decision about whether to provide a prenatal test in this scenario therefore must be based on who is

deemed to have the greater right. Is the future individual's right to decide about testing for paramount, or is the right of the parents to have prenatal diagnosis irrespective of the condition being tested for, or the action taken in response, more important?

In this scenario I consider that the future individual's rights outweigh the parents' rights and that prenatal testing for Huntington's disease should not be offered. By avoiding prenatal testing for adult onset disorders when no preventive treatments exist, the autonomy of the future individual is preserved. Individuals can decide for themselves if and when to have predictive testing. By these arguments, not offering prenatal testing is no different from not offering predictive testing for a child too young to make an informed decision about such testing. The rights of a pregnant woman are of great importance and are paramount when no third party has an equal or greater right. If the ethical consequences dictate that it is preferable not to offer such a test, however, the fact that the test is prenatal rather than being a test on an individual outside the womb does not make it any more justifiable.

The outcome of such testing might be harmful to the parents. Three cases have been reported in which parents experienced considerable and long term anxiety related to learning that their young child was gene positive for a late onset genetic disease.⁶ In two of the three cases this followed prenatal diagnosis when a decision was made to not terminate the pregnancy. The parents were greatly troubled by when and how to tell their children.

How should such requests be handled? In my view, issues regarding termination of pregnancy need to be explored in depth, and if it is clear that a couple will not terminate a pregnancy then the test should not be offered. The specific reasons for this decision should be communicated to the parents. Clearly some couples embark on prenatal testing with the genuine intention of terminating an affected pregnancy but decide against it when they receive the result.⁷ This outcome will occur from time to time but can be minimised by in-depth counselling before the test as well as ongoing discussions between the time of the test and the results.—MBD

To test (R E Duncan and B Foddy)

We think that this couple should be offered a prenatal test for Huntington's disease. Refusal to offer prenatal testing to women who are not willing to consider termination of pregnancy suggests that termination of pregnancy is the only appropriate reason to undergo such testing. This is incorrect. There are several other equally valid motivations for wanting a prenatal test. For example, the possibility of a negative diagnosis represents a chance to be spared a long period of uncertainty about a child's health prospects and to spare the child from the same uncertainty in future years. Uncertainty about genetic status can be associated with anxiety and frustration.⁸ Additionally, parents may want this information to assist them in making a decision about whether to have more children.

Even if it were true that termination of pregnancy was the only appropriate reason to undergo testing,

this would not justify withholding testing from parents who doubted that they would consider termination. There is no way of ensuring that those who say they will terminate their pregnancy after a positive test result will do so. Similarly, there is no guarantee that those who say they will not terminate their pregnancy will indeed continue the pregnancy. It may be that gaining information about the unborn child is part of the process of decision making regarding termination. Requiring individuals to be sure of what they will do in a hypothetical situation is unreasonable. Such demands are never placed on patients before other diagnoses.

If tests are provided only to parents who say they would terminate the pregnancy in the event of a positive result, those not seeking a termination of pregnancy may simply lie. Because of this, the clinician's stance is not only completely ineffective as a deterrent, but it breaks down the important relationship of trust between patient and clinician. Such a policy will prevent people from having honest and supportive relationships with their clinicians at a time when this is crucial.

It is an abuse of power for a clinician to threaten to withhold a medical diagnosis unless the patient conforms to a particular behaviour. It is a means of coercing the patient into making what the clinician deems to be the best decision. Clinicians should not demand that their patients will react in the "right" way to a diagnosis; that a lung transplant recipient will stop smoking; that an athlete receiving a knee reconstruction will cease competing. These decisions should remain in the control of the patient.

Not everyone would agree that when parents have a prenatal test in this setting their child's autonomy is harmed. But even if we accept that the child's autonomy is harmed, a mother's right to information about her unborn child outweighs this harm. During pregnancy the fetus is part of the mother's body, and the mother has a right to know facts about the fetus as these constitute facts about her body and her future. For (at least) the first trimester, it is entirely up to the mother or couple whether the fetus will even exist as a person. If people have the right to decide if their child will exist, surely they have the right to learn of genetic information that can help them to make this decision.

This couple have already decided that their child will exist. If they had not articulated this decision, the prenatal test would be provided to them—that is, they would have a right to genetic information about their unborn child. They cannot lose this right to information simply because they decided not to terminate the pregnancy.

Finding out that one's unborn child will develop a severe genetic condition later in life is difficult. It is, however, paternalistic to assume that testing should not proceed because people will find this burdensome. Any adverse medical diagnosis can be distressing, but few would claim that we should withhold diagnoses to spare patients' feelings, even in cases when there is no available treatment. Clinicians and counsellors should aim to help parents to understand the difficulties that may arise in such situations and to support them through the process of making an informed

Summary points

Huntington's disease is an autosomal dominant, lethal neurodegenerative disorder with an average age of onset of 37 years. There is no cure and or current preventive treatment

Predictive genetic testing and prenatal diagnosis are available for Huntington's disease

Guidelines recommend that prenatal tests should be provided only when termination of pregnancy is planned in the event of a positive test result

Offering prenatal diagnosis to parents who have no desire to terminate their pregnancy is controversial

decision. The ultimate decisions should be those of the parents, not their clinicians.—RED, BF

Discussion

Requests for prenatal tests are likely to become more common as genetic knowledge continues to increase. The case we present highlights the ethical challenges and the contradictory views that professionals may hold.

There are four key ethical issues for clinical practice. Firstly, and in our view most importantly, how should the autonomy of parents be balanced with the future autonomy of their unborn children? Secondly, is it paternalistic for a clinician to refuse to administer a prenatal test requested by a woman (or a couple) and, if so, is such paternalism ever acceptable? Thirdly, is it appropriate for clinical guidelines to make recommen-

dations that may encourage patients to lie to achieve a desired test or treatment? Finally, does it constitute a breach of confidentiality when parents are informed about genetic characteristics of their unborn child? These ethical issues provide a basis for consideration by clinicians faced with similar requests for prenatal testing in the future.

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Evidence based medicine

Analogies between reading of medical and religious texts

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Whether you take a fundamental or liberal view of scientific evidence will affect how you interpret it. But just as with religion, we need to admit there will be legitimate differences in our views

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Conventional medicine can be seen as a belief system characterised by a profession of faith in evidence based medicine.¹ Faith in evidence based medicine follows from the benefits it has delivered in the past and continues to deliver. It is the best method we have for navigating our way through potential new treatments.¹

Evidence based medicine, analogous to many religious traditions, is a system for the interpretation of a canon of sacred texts (medical literature). Differences in interpretation can often be traced to different assumptions underlying our reading of the literature. Just as there are fundamentalist, conservative, and liberal views of religious texts there are similar views of

evidence based medicine. An examination of these views and assumptions can tell us much about differences in medical opinion.

Religious fundamentalism

The Oxford dictionary defines fundamentalism as the strict maintenance of the ancient or fundamental doctrines of any religion or ideology.² Fundamentalism sees truth as unified, revealed, absolute, and inerrant. Supporters have a black and white view, seeing themselves as the true keepers of the faith with good reason for an absolute belief that they are right. Critics