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Genetic testing in children and young people

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Abstract Guidelines on childhood genetic testing are in broad agreement that where there are no 'urgent medical reasons', presymptomatic and predictive testing for adult-onset disorders, and carrier testing should be postponed until a child is able to give his or her own consent, either as a competent young person, or as an adult. This paper explores situations in which this requirement can be in tension with genetics professionals' and others' judgement of what is in the child's best interests. It concludes that whilst the guidelines do reflect a broad agreement that in most cases testing children for adult onset conditions or carrier status is inappropriate, there are at least some situations in which testing may be thought by genetics professionals to be appropriate. Many of the morally relevant features of such cases will often be context specific, i.e. to do with the child's family and other relationships or other features of the local context and this suggests that any revision of the guidelines on genetics testing in childhood will need to take into account the need to allow space for the utilisation of judgement by genetics professionals about whether genetic testing is in the child's best interests. In making such judgements the genetics professional will need to pay close attention to the views of the child's parents and do all they can to facilitate input from the child him or herself.

Keywords Children · Genetic testing · Ethics

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Introduction

The genetic testing of children and young people presents important ethical challenges for genetics professionals [5, 8, 12]. National guidelines have been developed in many countries on various aspects of childhood testing [2–4]. There is broad agreement in these guidelines that whilst genetic testing in childhood may be appropriate in the context of conditions presenting or treatable in childhood, the testing of particularly very young children for adult onset disorders or for carrier status is more problematic. In a systematic review of guidelines and position papers on presymptomatic and predictive testing, Borry et al. found that all the guidelines surveyed recommended that where there were 'no urgent medical reasons [testing should be postponed] until the child could consent to testing as a competent adolescent or as an adult' [2, p. 374]. In their review of guidelines on carrier testing they again found that all guidelines agreed that 'carrier testing should not be performed in childhood and, testing should be deferred until the child can give proper consent to be tested' [2, p. 133].[1] These formal positions allow little room for the use of judgement by genetics professionals about testing for adult-onset conditions or carrier status in the light of the implications of the facts of particular cases for the child's best interests. In the United Kingdom there are currently moves afoot to revise the guidelines on childhood testing. For any revised guidelines to be effective, it is vital that they reflect and address the complexity of the ethical issues that arise in practice. This paper explores some of the ways in which the current guidelines may be in tension with the

[1] There was rather less agreement about testing for *childhood* onset disorders where no preventative or therapeutic measures are available.

ethical challenges confronting genetics professionals in their day-to-day practice.

The UK genethics club

The discussion in this paper is grounded in the experience of the UK Genethics Club. The Genethics Club (www.genethicsclub.org) was established in 2001 as a national forum for genetics professionals to discuss ethical issues in their day-to-day work and to enable those working in different regional genetics centres to develop and share models of good practice. Those attending the 22 meetings which have thus far taken place, include: geneticists, counsellors, nurses, laboratory staff, general practitioners, clinicians in other specialties, researchers, lawyers, and members of patient-groups. The Genethics Club meets three times a year. Discussion is facilitated by an ethicist (the author) and is primarily concerned with the discussion of cases identified and presented by those who attend. Genetics professionals from each of the 23 genetics centres in the United Kingdom have attended. Whilst the cases presented—approximately 200 at the time of writing—are not strictly representative they are a valuable resource because they offer a rich picture of the range and diversity of ethical issues which arise for genetics professionals in their day-to-day practice. Requests for the genetic testing of children can be experienced as morally problematic by genetics professionals. Given this, it is unsurprising that a number of cases concerning the genetic testing of children and young people have been discussed at the Genethics Club. While the Genethics Club has inevitably not reached a consensus about solutions to the problems presented by requests for childhood testing, the issues discussed in this paper do to some extent reflect a degree of agreement regarding the nature of the problems arising in the day-to-day practice of genetics professionals.

The genetic testing of children: a complex landscape

Testing for adult-onset conditions

Current guidelines are largely agreed that the genetic testing of young children for adult onset disorders is something to be avoided until the child is competent to consent. Despite this, requests for testing do occur on a reasonably regular basis. Parents will generally request a genetic test in their child because they feel that if the result is negative this will provide reassurance both for themselves and for their child and remove the need for many years of worry, potentially to be endured throughout childhood and beyond. When challenged to consider the implications of a positive test result, they will sometimes argue that as the child's parents they are going to have a long-term caring relationship with the child as he or she grows up, and as the people responsible for his or her care, they are in the best position to choose the moment and the method by which the topic of the child's genetic risk might be sensitively introduced and discussed in a supportive way. There is broad agreement in the national guidelines surveyed by Borry et al. that these tests should generally not be undertaken. The main reasons for this are: that child should be able to make their own decision about testing, based on their own values, when they are ready (many adults choose not to be tested, the child should not have this choice taken away from them); respect for the child's confidentiality (if the test goes ahead the parents will know about the child's genetic status and it will appear in his or her medical record); and concerns about the possibility of harmful consequences for the child of testing (such as increased anxiety; changes in the parent-child relationship; stigmatisation, or changes in the child's self-image) [7].

In addition to its effects on parents, the position taken by the guidelines can present practical ethical difficulties for genetics professionals who find themselves in situations where they believe the harms to a child in not testing outweigh those of testing. Such harms are likely to be very context specific, arising perhaps out of the nature of the child's family and other relationships. An example might be where, despite the best efforts of the genetics professionals, a child who is at 50% risk of an autosomal dominant condition such as inherited breast cancer is treated by their family or others as if he or she were in fact already known to be affected by the condition in ways which are having serious detrimental effects on his or her development and well-being. Where the situation is unlikely to improve, genetics professionals can sometimes come to the judgement that on balance a test would be in the child's interests because the fact that there is a possibility that the result will be positive is outweighed by other considerations of importance to the child's well-being. In such cases there is a tension between the guidelines and good practice. The fact that such cases are likely to be relatively infrequent does not obviate the need for guidelines to take into account the fact of their possibility.

An increasingly common situation in which there can also be a tension between the genetics professional's judgement of the child's best interests and the position taken by the guidelines is where a request for childhood testing for an adult onset condition comes not from the child's parent but from a social worker [9]. This kind of situation typically involves a child who is living in residential social care who has information in his or her record about an inherited disorder in one or more relatives and where the presence of this information has been enough to

deter a series of potential parents from adoption with the implication that the child is likely to remain in residential care indefinitely. In this kind of situation social workers may have the strong belief, grounded in experience, that were a test to be carried out and the child found to be free of the disease-causing mutation this would dramatically increase the likelihood that he or she would be found an adoptive family. The main argument against testing in such cases, over and above those against childhood testing generally, is that a positive test result may itself have very negative consequences for the child. This means in the majority of such cases that it will probably be right to refuse a test. However, this position is weaker in situations where social workers are strongly of the view that the only possibility of adoption for the child is if a test is carried out and the result is negative, and the implications of growing up in residential care for the child are sufficiently against his or her interests as to warrant testing despite the other possible implications of a positive result.

Further examples of situations in which a child might have a strong interest in being tested include those in which testing the child for an inherited condition might provide information to benefit other family members and those in which providing a test to the child is considered to be vital to maintaining an effective relationship between the genetics professionals and the family or within the family itself where the continuation and development of these relationships is considered to be in the child's best interest.

Testing for adult on-set conditions, where early screening is available

A related set of cases is those in which a request is made by parents for a test in their child for a condition which although not presenting until adulthood, is one for which there is a well-established screening programme available fairly early on in adulthood. A good example of this is HNPCC for which screening is available from the early twenties. As this is a case involving an adult-onset condition, it is one in which the guidelines would be currently against childhood testing. But to many genetics professionals this kind of case seems very different to those involving adult onset conditions such as HD for which no intervention is available. In the case of HNPCC a well-established screening programme is in place which might be missed out on by people who were not tested as children and were not informed by their family members about the need for screening or their risk status. In such cases there are good reasons for wanting to ensure that the child is aware as a young adult that screening is available. Similar issues arise in the context of conditions where screening is available rather later in adulthood such as inherited breast cancer [6]. Moreover, in cases such as HNPCC it is much less likely than it is in HD, that an at-risk adult would choose not to be tested and followed up with screening and this puts pressure on arguments against testing which rest on a claim that children should be free to make decisions for themselves on achieving adulthood. These arguments seem much less convincing in the case of adult onset conditions for which screening is available and seem weaker still in cases where the genetics professional feels that information will not be passed on, or where it is very likely that the child will be lost to follow up. It might reasonably be argued that greater attention should be focused on ensuring effective and comprehensive follow up rather than on providing testing during childhood particularly given the evidence suggesting that accurate information may not be passed on even where childhood testing has occurred [10]. But whilst it is certainly true that effective follow up is important this does not obviate the need for decisions to be made in the best interests of the child where this is not available or where there are serious concerns about its effectiveness.

Carrier testing

Borry et al. also found broad agreement between the guidelines they surveyed that carrier testing should not be performed in childhood [2]. The main reason for this is that in the vast majority of cases, being a carrier of an autosomal recessive condition or a female carrier of an x-linked recessive mutation has no implications for the child's health, other than in its implications for the child's future reproductive choices. Despite this, requests for carrier testing in childhood are relatively common and these requests are not necessarily based on a misunderstanding about the health implications of carrier status. In some cases, parents request testing because they hope for a negative result and believe that this will relieve their anxiety and that of their child about the implications of carrier status for future reproductive decision-making. This can sometimes be based on the parents' own personal experience and a resulting strong wish to find a way to avoid their child experiencing what they went through. In other cases, parents want testing so that if the result is positive they will in a good position to inform and support their child as they grow up and to prepare them for what they themselves experienced as a traumatic reproductive choice. Many genetics professionals tend to take the view that these factors do not outweigh the importance of allowing the child to make his or her own choice as an adult and, given the absence of implications for the child's health, tend to feel that the right thing to do in such cases is to refuse testing. The problem with this position and with the guidelines as they stand is that such refusals take place against the background of national newborn screening

programmes for conditions such as Cystic Fibrosis and for haemaglobinopathies which routinely reveal carrier status and pass this information on to parents. Given this, parents quite reasonably ask, how can a refusal to carry out carrier testing during childhood be fair when such information is routinely available to other parents [11]?

Testing for childhood onset conditions

The guidelines surveyed by Borry et al. were less clear cut in relation to childhood testing for childhood onset conditions where there is no intervention available. An important sub-set of cases which can present ethical challenges to health professionals comprises situations in which parents request a test on an infant for a childhood onset condition for which screening or interventions of other kinds are available but not until the child is much older. An example would be a request for testing for FAP, for which screening is only offered once an at-risk child is a young teenager, in an infant. There are three main sets of reasons why genetics professionals might on balance consider the carrying out a test in this situation to be a child's best interests. The first situation is where the genetics professional is concerned that contact with the family might be lost and that the child might miss out on screening as a teenager. A second, and related type of reason arises in situations where a genetics professional feels that providing a test may be an important way to maintain an effective relationship with the parents and the family. The third reason for childhood testing in such cases is that it has the potential to avoid the need for unnecessary screening and monitoring in those children who are considered to be 'at-risk' but do not in fact have the mutation. Given that all at-risk children are going to go through the screening process, testing is not against the best interests of those who turn out to be positive, and it is in the interests of those who are found to be free of the mutation. In such situations, genetics professionals sometimes come to the view that it is in the child's best interests to be tested for conditions such as FAP well before screening is available.

Conclusion

Guidelines on childhood genetic testing are in broad agreement that where there are no 'urgent medical reasons', presymptomatic and predictive testing for adult-onset disorders, and carrier testing should generally be postponed until a child is able to give his or her own consent, either as a competent young person, or as an adult. This paper has explored situations in which this requirement can be in tension with genetics professionals' and others' judgement of what is in the child's best interests. Whilst the guidelines do reflect a broad agreement that in most cases testing children for adult onset conditions or carrier status is inappropriate, the cases discussed above suggest that there are at least some situations in which testing may be thought to be appropriate by genetics professionals. This suggests that any guidelines in the area of childhood testing will need to make it clear that good practice depends upon the use of judgement in individual cases. Many of the morally relevant features of such cases will often be context specific, i.e. to do with the child's family and other relationships or other features of the local context and this suggests that any revision of the guidelines on genetics testing in childhood will need to take into account the need to allow space for the utilisation of judgement by genetics professionals about whether genetic testing is in the child's best interests. In making such judgements the genetics professional will need to pay close attention to the views of the child's parents and do all they can to facilitate input from the child him or herself [1].

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