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Are guidelines for genetic testing of children necessary?

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Abstract There is now a plethora of guidance on the genetic testing of children. This paper explores the recommendation that childhood testing for adult on-set conditions should be delayed until the child can make up their own mind. It analyses the underpinning arguments used to support this position and asks whether, given some of the problems with these, the guidelines are really necessary.

Keywords Autonomy · Best interests · Children · Genetic testing · Guidelines

There is now a plethora of guidance on genetic testing of children: Borry et al. [1] found 27 different guidelines/position papers worldwide from 1991–2005. There appears to be a consensus that genetic testing in children is appropriate where medical treatment/surveillance can be offered but that it is inappropriate to test children for adult onset conditions where no interventions are available should the predictive test be positive. This paper sets out to critically explore the latter recommendation and to ask whether guidelines on this are really necessary.

The delay for testing children for adult onset conditions is seen as desirable because it respects the child as a future adult [2], who will then be able to make their own decisions/choices about when, and indeed if, they want to undergo testing and because it protects the child from potential harm. Despite the apparent agreement this is not without its detractors for a number of reasons.

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The first concern is that drawing on the child's future autonomy as an underpinning reason for the guidance is problematic. Harris and Keywood [3] point out that there are different sides to the equation. On the one hand an autonomy argument is used to support the right of a child to wait for test results until they can make an autonomous decision; however, a converse argument can be made that not testing takes away the rights of a child to live in and grow up in an environment which recognises their situation and where actions and decisions are made which take account of this context.

Another perspective is that if a child is not able to make a decision about whether to undergo genetic tests then they are not autonomous. In such circumstances, we would expect others, such as their parents, to make decisions on their behalf until such times as they are able to do this for themselves. Pelias [4] writing within the context of private healthcare in the US, asserts that, whilst it may be appropriate to advise and discuss the recommendation about delaying testing with parents, to override their request for testing would be to override a parent's right to decide what is in their children's best interest. She views this as ethically problematic because it would restrict parents' autonomous decision making abilities. It can, of course, be argued that where genetic testing for adult onset conditions is an issue, families will have lived with and will know about the condition and its effects more deeply and profoundly than the healthcare professionals advising them [4–6]. However, her point is more fundamental: parents' autonomy is the primary ethical concern: parents have both a right and duty, as carers, to make decisions on behalf of their family and these should be respected.

These arguments do not necessarily undermine a belief that waiting until the individual who may be affected can make up their own mind up about testing is a good thing.

However, they do illustrate how drawing on 'autonomy' as a key underpinning principle in the guidelines is problematic because there are different sides to the same principle which all have competing claims; for example: the autonomy of the future adult, the rights of the child and the autonomy of the present parents. Harris and Keywood [3] usefully allude to this as: "autonomy walks on both sides of the street" (p. 424).

A further issue with autonomy arguments is that they tend towards an individualistic notion of the self—whether they are referring to the individual child as later adult or the autonomy of the parent/s—which positions self-determination as a right of primary importance. An alternative approach would be to see a person and the decision making process as being located within a social group/family. Considering such an approach would mean thinking through the ethical dilemmas in a different way and giving the family a 'voice' [7]. This more family-orientated perspective has been raised in genetic medicine more generally in relation to genetic data and attempts have been made to encourage a view of such data as 'belonging' to a family/group rather than the individual [8]. Adopting an individualistic approach which privileges the rights of individuals to have their confidentiality protected will necessarily conflict with a view that harms and benefits need to be more widely considered.

The second underpinning reason given to support a guideline that testing should be delayed is that its delay protects children from harm that might accrue from testing: for example, because knowledge of having a condition may cause psychological harm to the child or to minimise the potentially harmful effect on the relationship between the child and their parents/family should the test be positive [9]. Rhodes [5] has questioned this position as being too heavily focussed on the potential harms associated with knowing test results without taking into account the potential benefits; for example: that there is some evidence to show that for both parents and the child, living with uncertainty can be worse than knowing the 'bad news' and that children can cope with disclosure of such information. There has been a widespread acknowledgement that there is little research in the area of either harms or benefits and there have been calls for research to be undertaken yet, to-date, few convincing data have been generated. However, it is surely likely that there will be harms and benefits associated with both positive and negative predictive test results and these may be influenced by a range of factors such as the nature of the condition, what is currently known about it, the family context, and the individual child themselves. This would indicate that viewing each case on its own merits might be the most appropriate approach to adopt.

Although parents make healthcare decisions for children all the time, this does not mean that they can decide on anything they want: children do not belong to their parents and others have an interest, and should have an interest, in their well being. The current practice in the UK is that we expect and require parents to make healthcare decisions for their children with the proviso that in doing so they should act in each child's 'best interest'. We mostly leave the parent/s to make decisions on their children's behalf because they know their children, their situation etc. [10, 11] and legal pathways are in place where serious disagreements exist as to the best course of action/inaction to take. This situation applies to genetic testing as with other healthcare contexts. Why then does there need to be guidelines, which outline a consensus view, in this area? Why not just rely on parents and healthcare professionals—and children when they are able—to discuss the medical and wider benefits and harms of testing to determine what is best in *each* case and, where there is disagreement about what is best, to adopt the usual process/pathways for resolution? We might question the notion that consensus is actually a desirable goal as is assumed [1, 9] and, moreover, whether guidelines are actually necessary given some of the already highlighted problems associated with them.

One further question to consider in trying to answer these issues is what the purpose of guidelines is and how are they used: are they 'guidance' to help inform different parties involved in decisions about whether to test or are they more 'policy', aiming to direct action? Do clinicians use them as their 'default' position? Duncan et al. [12] in a survey of geneticists in the UK, US and Australia found that, whilst they did test for adult onset conditions (for wider best interest reasons), their test numbers were low and they indicated that they supported the guidelines. Rhodes [5] interprets this as the guidelines becoming the 'standard of care' for testing.

Compare the recommendations from the UK Genetics Society:

> We would generally advise against such testing unless there are clear cut and unusual arguments in favour… formal genetic testing should generally wait until "children" request such tests for themselves as autonomous adults [9, p. 785].

With those of the European Society of Human Genetics:

> Presymptomatic and predictive testing of minors for conditions with adult-onset is only recommended if preventative actions… can be initiated before adulthood. Otherwise… testing should be deferred until the person has the maturity and competence to understand the nature of the decision and its implications [13, p. 3]

The former presents itself as *advice* whilst the latter, written over a decade later, is more prescriptive indicating that this is, or *should* be, the default position. This

difference may also indicate that earlier advisory-type guidance is increasingly being seen as the default position and may reflect a more general shift in healthcare for 'guidance' to be developed which is based on evidence and which, therefore, should be 'complied' with. There are a number of problems with adopting such a compliance approach to genetic testing guidelines: one, the guidelines are primarily based on ethical considerations—whatever the framework/approach is adopted—with little underpinning 'evidence' that you might expect to be available or uncovered through research for medical treatments; and two, if the testing guidance is seen as something that needs to be complied with, how they are used in practice will reflect this. It will be more difficult for parents to openly discuss the best interests of their child with the clinician/s concerned; and if they do believe that testing their child is the best option, what will be required of them is to convince the healthcare professions about their case which may set up adversarial relationships from the outset. In a context where families experience extremely challenging health problems, and are likely to need all the support possible, this seems to undermine the rhetoric that healthcare professionals and patients make 'decisions together' [14].

Not withstanding all of these issues, do guidelines still have a purpose? My answer to this question is: yes, if it is to inform all parties (parents, children, clinicians across specialities, stakeholders) what the issues are and what sorts of things need to be thought through and grappled with when testing is being discussed and considered. A general consensus about what is important to consider in this area can be viewed as helpful for everyone to sharpen their thinking about the ethical issues involved. I think the answer should be no, if guidance sets out to be, or is seen to be, a set of rules which need to be followed or complied with, and which discourage critical and open engagement.

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