Case Analysis: Ethical Issues in Predictive Genetic Testing of Minors for Adult-Onset Hereditary Conditions

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Abstract

Introduction: Predictive genetic testing for adult-onset conditions in children is an issue that has initiated ethical discussions and polarized views.
Case Presentation: A genetic counselor is contacted by a mother requesting predictive genetic testing of her young son for Hereditary Nonpolyposis Colorectal Cancer (HNPCC) due to an extensive family history of the condition. The ethics team subsequently worked to balance professional guidelines and ethical considerations to evaluate whether to comply with the parental request. It was necessary to consider the importance preserving the child's future right to exercise his autonomy in participating in such testing, preserving the confidentiality of the test result, and considering the possibility of potential psychological harm.
Conclusion: Determining whether to perform predictive genetic testing in a minor for conditions of adult-onset should be guided by the potential outcomes that indicate that medical benefits obtained will outweigh the risks of testing.

Introduction

Pre-symptomatic and predictive genetic testing are rapidly developing technologies which allow for identification of genetic defects before manifestation of symptoms and inform individuals of their future health risks.1,2 Nevertheless, genetic testing in pediatrics remains controversial and has initiated ethical discussions and polarized views. Many parents seek genetic testing for their children when hereditary conditions are prevalent in the family in order to implement early interventions or to obtain the reassurance that accompanies a negative test result.3 These requests create a dilemma for the clinician, whose primary concern is with the child's best interest. This case analysis concerns a parent's request for predictive genetic testing of her young child for a hereditary disease of adult-onset.

Case Report

A woman with a family history of Hereditary Nonpolyposis Colorectal Cancer (HNPCC) contacted a genetic counselor to request genetic testing for her 4-year-old son. The mother reported multiple bloodlines in the family with a history of HNPCC, the youngest affected individual being 18 years of age at initial diagnosis with colorectal cancer (CRC). She requested testing of her son for HNPCC mutations and regular colonoscopy screenings. The cancer genetic counselor contacted the ethics team with concerns regarding predictive genetic testing of this young child.

There are two clinical questions identified in this case, both of which have ethical implications. First, is it appropriate to perform genetic testing for HNPCC in a child with a known family history of early-onset CRC? Second, is it appropriate to initiate colonoscopy screenings at this child's current age?

Discussion

HNPCC is an autosomal dominant cancer syndrome of adults. The mean age at initial CRC diagnosis is 45 years.4,5 Guidelines established by the American Gastroenterological Association (AGA) advocate screening for CRC in at-risk persons every 1-2 years beginning ages 20-25 or every 2-5 years before the youngest age of diagnosis of CRC in the family if diagnosed before age 25.4 Thus, the recommendation is to offer colonoscopy screenings to children in this family beginning as young as age 13.

Nevertheless, the American Academy of Pediatrics advises against genetic testing of children for conditions with adult-onset, recommending deferral of testing until adolescence or adulthood, after the development of mature decision-making capacities to request testing as an autonomous individual.3 This correlates with American College of Medical Genetics (ACMG) guidelines, which advocate delaying testing until adulthood unless there exists a risk of malignancy in childhood or adolescence.4,5,6 In this case, even if genetic testing were postponed until the age indicated by AGA or ACMG guidelines, the patient will have not yet reached adolescence and appeals to postpone testing until adulthood are not applicable.

The first consideration in genetic testing involves disclosure to the child about the hereditary disease and its implications.7 Discussing this information throughout the child's stages of maturity enables him to make informed decisions about predictive genetic testing in the future

and incorporate such knowledge into his self-identity.7 Thereafter, the clinician should ensure that screening and testing recommendations are discussed in an age-appropriate manner with the child, as they are capable.3,8 The clinician must consider a plan for disclosure about the test and results, confidentiality of genetic information, and the psychological impact on the child.

Testing the immature child for a hereditary condition of adult-onset is ethically problematic as it disregards his future right to make informed, autonomous decisions.9,10 Young children are generally considered incapable of making complex medical decisions, therefore the authority usually falls to the parents. When the child has developed the capacity to participate in the decision-making process, he should be informed about pertinent medical issues. However, the repercussions of genetic testing and implications for future health suggest that discussions be deferred until the child is able represent his autonomy during the decision-making process .9,10

Medical benefit to the child must be the primary justification for genetic testing.9 The availability of preventative or therapeutic measures is supportive of testing and may serve as a requirement to perform a genetic test. In this case, preventative measures (regular colonoscopy screenings) are not indicated until the child reaches the age of 13-16, based on AGA guidelines.4 Therefore, the lack of medical benefit in testing the child as a minor supports the ethical indication to defer testing until adolescence. There are ethical concerns related to potential harm, as testing a young child may lead to a phenomenon known as “vulnerable-child syndrome,” in which parents become overprotective of their child in the absence of symptoms.9,11 Further, the child who tests gene-positive may exhibit psychosocial harm to themselves, as knowledge of their health risks alters their self-concept and can lead to feelings of unworthiness and guilt, subsequently affecting relationships with family and peers.9,11

Nevertheless, there are several ethical arguments in support of predictive genetic testing. In response to concerns about the child’s autonomy, it is asserted that parents are considered effective surrogates and thus should be provided with the right to make informed decisions about their child.9 Having early knowledge of gene status allows parents to help psychologically prepare their child for future medical interventions.9,10

Despite the potential psychosocial benefits for this 4-year-old child, the lack of indication for performing preventative screenings at his current age demonstrates that the risks outweigh likely medical benefits. It is recommended to defer discussions regarding HNPCC testing until the child has reached adolescence and can provide an autonomous decision regarding testing. This approach adheres to AGA guidelines and most fairly balances risks and benefits of predictive genetic testing.

Conclusion

As genetics become increasingly integrated into patient care, one must consider ethical indications for performing genetic testing. Decision-making regarding compliance with parental demands for predictive genetic testing of a minor should be guided by potential clinical benefits

versus risks. This involves disclosing health risks to the child, preserving their future right to exercise autonomy, and considering potential psychological harm. In the case of testing a minor for HNPCC, the ultimate decision to defer genetic testing and screening until adolescence is supported by ethical considerations as well as professional guidelines which ultimately function to preserve the child's best interest.

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