Predictive genetic testing of minors: evidence and experience with families

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In this issue of *Genetics in Medicine*, Wakefield et al.¹ report data from a systematic review of 13 studies involving 966 minors who received risk information through either genetic testing or family history, most frequently for hereditary cancer and cardiovascular syndrome. In the majority of cases, the minors and their parents reported no adverse psychological outcomes.

A small number of participants reported the transient anxiety and/or depressive symptoms that are expected upon receipt of threatening health information. The authors contrast their results to 10 practice guidelines that advocate limiting the genetic testing of minors to conditions with clinical interventions in childhood, citing the guidance as unsupported by the evidence. Yet they caution that their review involved a limited number of studies that assessed short-term rather than long-term outcomes.¹

Published guidance advocates for minors to reach the age of majority before undergoing predictive genetic testing, recognizing exceptions in cases where providers endorse minors as being well equipped to make informed decisions.² Outcomes from the review by Wakefield et al.¹ reveal limited evidence to support such caution toward predictive testing. Evidence suggests that there are few adverse psychological outcomes among the majority of those tested, probably resulting from the self-selection of psychologically sound clients making well-considered decisions. Those who seek out predictive testing of minors are often members of families who are experts on living with the condition that affects their relative(s). One source of concern is that parents may pursue testing of their children to relieve their worry but inadvertently confirm their fears. Genetic counselors and other health-care providers can help facilitate decision making by partnering with parents and adolescents to deliberate the potential consequences of results, recognizing and respecting their capabilities and experiences.

The justification for the history of limiting predictive genetic testing of minors is reviewed in a 2015 American Society of Human Genetics position statement.² This update to the 1995 Points to Consider paper published in collaboration with the American College of Medical Genetics reviews ethical and social arguments to defer testing.³ Consequences of the testing of minors have been hypothesized to lead parents to raise their

children differently from how they would raise them if their risk status is uncertain.³ Further, preserving a child's right to an open future⁴ defends adult preferences to live with uncertain risk. If this option is removed during childhood by parents opting for testing, the adult's autonomy is compromised. Yet whether a child raised by parents who knew his or her risk status would come to regret having that knowledge as an adult is unknown but unlikely. The long-term outcomes of testing minors have not been reported and would be difficult to capture given the human tendency to adapt to our experiences, having never known an alternative.

Cautionary guidance is warranted in the absence of evidence about a vulnerable population. Parents can be naive about the consequences of testing and may place their own desire to know ahead of their children's needs. Yet rarely do we witness such narcissism among parents. Families affected by genetic conditions often have spent years considering the risk status of their child(ren). Many have rehearsed testing options and considered the outcomes before seeking care from a genetics provider. These parents take testing decisions seriously and have a history of making decisions on behalf of their children and family. Taken together with a lack of evidence of adverse outcomes from the systematic review,¹ the case of a cautionary position toward predictive testing is unsupported. Furthermore, the 2015 American Society of Human Genetics position statement acknowledges a shift toward greater parental discretion in the face of uncertainty about the best interests of a child, yet the shift is not reflected in the guidance.² Patient-centered care and patient empowerment have gained favor in health-care delivery reflecting respect for patient preferences and parental capabilities to make well-considered decisions. The option of predictive genetic testing of minors should join this trend.

Families should be encouraged to discuss the predictive testing of minors in genetic counseling and, after collaborative deliberation, be offered testing if that is the preferred outcome.⁶ Similarly, adolescents should be supported in their pursuit of predictive testing following shared decision making within genetic counseling. Decisions to undergo predictive testing are complex, nuanced, and influenced by family values and beliefs, interpersonal dynamics, and personality traits. Optimists, for example, have more

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resources for managing threatening health information and are more likely to pursue testing.⁷ Similarly, those who are averse to ambiguity seek to reduce uncertainty by learning their status.⁸ As such, it is unclear whether further evidence would be used to reverse guidance to forgo predictive testing of minors in the absence of clinical intervention. Robertson and Kerruish⁵ suggest that further evidence may not resolve discrepant views on the testing of minors. They call for “acknowledging that choosing to do “nothing” (or not test) is not necessarily better than choosing to intervene (or test) under conditions of uncertainty.” Their proposed desirable outcome is to advocate for a thorough and informed comparative assessment of quality of life.

Wakefield et al.¹ provide valuable data that may reassure genetics providers of the small chances of adverse psychological outcomes among minors undergoing predictive testing. They regard their evidence as promising but insufficient to change current guidance to defer testing to the age of majority. However, more data may never adequately address the personal and complex nature of predictive testing of minors. Although few parents and adolescents may pursue predictive testing, those interested should be empowered to make a carefully considered informed choice in this era of patient-centered care.

DISCLOSURE

The author declares no conflict of interest.

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