Clinical Reports

A Narrative Experience with Adolescent Genetic Testing

Katherine P. Wu, Amanda Ruth, and Daniel P. Mahoney

ABSTRACT

The genetic testing of adolescents for delayed onset illnesses has raised a number of ethical debates and policy discussions, including issues about pediatric assent, open future, and genetic privacy. Specifically, what role, if any, should an adolescent play in the decision-making process, and whether irreversible decisions made by caregivers on behalf of an adolescent breach their right to a future otherwise filled with more possibilities. Despite abundant theoretical discourse in the literature, there are few first-person accounts from young adolescents that describe their own experiences with genetic testing. In this article, I describe my experience getting a BRCA2 genetic test; advocate for the preservation of an open future; suggest deferring the age of predictive genetic testing until the age of 21; and encourage transparency with regards to who can access genetic information.

Katherine P. Wu, BA, is Student in the Department of Neuroscience at Rice University in Houston, Texas. *katherinewu@alumni.rice.edu*

Amanda Ruth, MD, is an Assistant Professor in the Department of Pediatrics at Baylor College of Medicine, Texas Children's Hospital, in Houston. *axruth@texaschildrens.org*

Daniel P. Mahoney, MD, is **an Associate Professor** in the Department of Pediatrics, Baylor College of Medicine, Texas Children's Hospital. *dx.mahone@texaschildrens.org*

INTRODUCTION

Adolescent genetic testing for disorders with delayed onset is an ongoing debate in ethical and policy discussions because it strikes a careful balance between the growing importance of genetic privacy, pediatric agreement, and open-ended future possibilities. The story told in this article sheds light on the ethical challenges surrounding pediatric genetic testing by giving a firsthand account of my experience getting a BRCA2 genetic test as a late adolescent. Although the majority of policy recommendations oppose predictive genetic testing for teenagers, my narrative encourages a closer look at the complex ethical issues that involve parental consent for children, unresolved concerns for the future, and the changing field of genetic privacy.

In this article I argue that choices should be left to individuals. I believe the more mature you are, the more equipped you are to handle situations. In my case, age 21 was a good time to undergo a genetic test. Due to the gravity and maturity required for genetic testing, I suggest that adolescents delay testing until they are mature enough to handle the information testing provides. Based on my experience, my policy

recommendation is to encourage children and parents to wait until they are ready, especially if there is no current medical benefit, such as medical interventions or preventative actions.

AN OPEN FUTURE

Current policy statements generally argue against predictive genetic testing for adolescents. In 2013 the American Academy of Pediatrics (AAP) and American College of Medical Genetics and Genomics issued a joint policy statement on genetic testing in children that recommends against predictive genetic testing. Their exceptions to the recommendation were when early disclosure allows a medical intervention that decreases mortality, or if diagnostic uncertainty poses a psychosocial burden.[1] In 2015 a similar statement was issued by the American Society of Human Genetics, which encouraged adolescents to defer predictive testing for adult-onset conditions until adulthood, due to the potential impact of this information at formative life stages, as long as there is no substantial psychosocial distress over risk status or specific life-planning decisions.[2]

To help guide policy about adolescent genetic testing, philosophers have explored various ways to think about the complex issues involved. One prominent issue is the open future argument. In 1971, American philosopher Joel Feinberg coined the phrase "the child's right to an open future" to refer to the idea that children should have the freedom to make choices about their own lives and therefore shape their own destinies.[3] While the concept of an open future was originally made in the context of education, the same principles can be applied to healthcare. In the context of healthcare, violating a child's right to an open future may include disclosing predictive genetic information during childhood, which violates the autonomy of the future adult. Doing so is a strong claim against testing, as it may take away a child's right to an open future.

Accordingly, a 2017 article written for an audience of genetic counselors summarized the status quo as "unless testing has current medical benefit, it should be deferred until a child is old enough to make her/his own decision protecting what Feinberg called the child's right to an open future."[4] The consensus appears to preserve the right to an open future by delaying decisions until an adolescent is capable of making autonomous decisions on their own.

However, the current literature has shifted slightly to regard an open future not as a right, but as one factor to be weighed among many.[5] Regarding an open future as one factor allows greater flexibility in determining if knowing one's genetic status may cause more benefit than harm. Other factors to consider include alleviating anxiety and uncertainty, informing planning for a future health condition, and enabling children to begin to incorporate information into their developing identity.[6]

When I consider the open future concept in the context of my own care, I agree that an open future is not a right in itself, but instead one factor among many to consider. I first learned that genetic testing for BRCA2 was a possibility for me when I was 16 years old. At the time, my sister was 18 years old and had a genetic test. Thankfully, the result for my sister was negative. Despite this, I waited until I was 21 years old to finally take a genetic test for myself. My parents kept the possibility open for me to choose when to undergo a genetic test. My mom, an emergency room physician, recommended that I test before I was 25 years old because I might have to have medical screenings, but the choice of when to test was ultimately up to me. In an investigation of BRCA testing one year after a breast cancer diagnosis, 13 percent of women did not report taking a BRCA genetic test.[7] Like me, I think part of my mom did not want to know whether her positive BRCA2 status was passed on to me. In the years prior to my genetic test, I felt a bit of anxiety, wondering at times if I could get breast cancer before I was 25 years old. When I tested, I found out that I was positive for the same BRCA2 gene my mom had. Oddly enough, having a definitive answer of whether I was positive had an almost calming effect. Following the diagnosis, I felt less anxiety about wondering if I was positive.

But there are cons as well. On receiving the diagnosis, I felt doors closed in terms of what

futures lay ahead of me. If I wanted children, it would have to be before a certain age so that I could have my ovaries removed before ovarian cancer became a likely outcome. In turn, the timing of children informed the occupation I would see myself in. Do I continue on my current path to become a medical doctor, going through four years of medical school and another three years of residency, before I can finally start my job at the age of 31? If I knew that I had a higher chance of dying younger, would I choose to live my life differently? I had chosen Rice University for its opportunities at the Texas Medical Center and studied neuroscience for four years as a premed. On the flip side, while certain doors closed for me, other doors opened—career options outside of medicine felt like more of a possibility.

In terms of parent involvement, the literature states that parents remained a primary source of emotional and financial support, slowing age-appropriate independence and complicating patients' privacy.[8] I believe my parents facilitated my growth, however, by giving me the freedom to choose without imposing their own will. I appreciate what my parents did for me, which was to give me the freedom of when to test, thereby preserving my options for an open future. In doing so, they gave me the autonomy to decide when to take a genetic test, which allowed me to take charge of my own healthcare.

PEDIATRIC ASSENT

Pediatric assent centers around the question: How do we involve children in their own care? Research indicates that involving children in decision making about their healthcare may lead to better health outcomes because it encourages better compliance[9] and verbal or nonverbal contributions about their care,[10] and develops a child's capacity for future autonomy.[11] Furthermore, including child patients in their treatment options helps to empower them.[12] By making my own decisions on when to test, I felt empowered to take charge of my own health, especially as I was transitioning to becoming an adult and determining my career and where I might want to live in the future.

While informed assent generally leads to a better connection between the patient and doctor,[13] I did not feel this was the case. I wasn't given much information on the pros and cons of early predictive genetic testing, and what to expect in my healthcare future should I test positive. It was only until after I was positive that my ob-gyn referred me to a breast specialist, gynecologic oncologist, dermatologist, and gastroenterologist. I was then given ages and frequency at which I would have to undergo mammogram screening and visit each of these specialties. I would have liked this information prior to a genetic test, so that I could be prepared for the different outcomes. While I did not feel closer to my doctor, I did feel closer to my family. I learned to live life with just a bit more gratitude, cherishing each moment with loved ones, living in the present, and having a mindset that allows me to let go of situations that I cannot change.

The AAP notes that one of the benefits of pediatric assent is that the patient may disclose relevant information that could help in the diagnosis, whether verbal or nonverbal, which could contribute to their care.[14] But I did not believe I had relevant information to disclose. The genetic counselor had my family tree that showed my cousins had tested positive, as well as my mom's BRCA2 status.

In terms of whether I felt more compliant, I do feel this is true. I felt more of a responsibility and obligation to take charge in my own healthcare, to have yearly mammogram screenings in the future. Now that I know I'm at a greater risk, I feel I am more likely to follow up with regular scans. I consider myself lucky that I had no time-sensitive medical treatments, as breast and ovarian cancer isn't something that typically happens until later in life. Resolving uncertainty early on helped me be forward thinking about my career, family plans, and so on.

Requirements for pediatric assent should be informed by what is developmentally appropriate for the individual child patient.[15] There are disagreements on what age a child can provide informed consent. For example, David Wendler and Seema Shah equate pediatric assent and informed consent, arguing that the grounding for

the moral value of assent rests on autonomous decision making, and therefore, at least in the context of research, assent should be available only to children age 14 or older.[16]

I consider myself an early bloomer in mental development. By age 14, I knew that going out past 9 PM was too risky and holding my phone loosely in a public setting was an invitation to be robbed. Even at that age, though, I was not prepared to understand or interpret what a positive genetic test could mean for my future. At age 14, I would have been mature enough to be able to understand what was happening, but not mature enough to be able to make a fully informed decision about my own healthcare. It wasn't until I was 21 years old that I could fully grasp the magnitude of the visits to specialists I would have in the future. Therefore, I suggest the age of pediatric assent should not be age 14, but instead 21 and older. To give a young person information too soon poses a risk on the spectrum between informed consent and acquiescence.[17] At that age of 21, I felt mature enough to weigh the options between the anxiety of uncertainty and the burden of unwanted information. I'm glad that I waited until I was mature enough to handle the information without being overwhelmed.

By making my own decisions on when to test, I felt empowered to take charge of my own health, especially as I was transitioning to becoming an adult and determining my career.

GENETIC PRIVACY

Whole-genome sequencing is now more affordable and accessible thanks to developments in sequencing technologies. While genome sequencing allows researchers to test hypotheses, it is also possible to identify the people whose deoxyribonucleic acid (DNA) sequences they include. Accordingly, individual rights in health law and medical ethics have brought attention to the necessity to safeguard the privacy of health information. In response, federal statutes and regulations created the Health Insurance Portability and Accountability Act (HIPAA) to protect human research subjects. HIPAA restricts certain unauthorized uses of patients' identities, but does not specifically mandate institutional review board (IRB) oversight or subject consent for the public release of sequenced data.[18]

I remember visiting the Exploratorium in San Francisco recently with my father and sister. I was unnerved by the exhibit, "Probably Chelsea: Twenty different sculptural portraits, all based on the same person's DNA information," made by the Exploratorium artist-in-residence Heather Dewey-Hagborg.[19] On reading the description, I realized that these seemingly different faces were all based on the same person's DNA information. The exhibit shows how the same DNA data can be interpreted in many different ways. Despite this, advancements in bioinformatics and medical informatics indicate that researchers in the fields of medicine and science can no longer guarantee complete anonymity and confidentiality.[20]

In the context of pediatric consent, parents are often asked to provide consent in place of their child. In a study that explored differences in data-sharing preferences between parents of pediatric patients and adult patients, most parents (73.5 percent) and adult participants (90.3 percent) ultimately consented to broad public release of personal data.[21] But parents were much more controlling when it came to decisions about the release of data—not because they understood or thought involvement would be beneficial, but rather because they valued their child's future autonomy and control.

Compared to adult research patients, parents who decided for their child expressed greater concern about the unknown future hazards and wanted to be more involved in the decision-making process about data sharing.[22]

Sometimes genetic information from one individual reveals information for the rest of their family, whether they like it or not. When my mother was encouraged by her genetic counselor to take a genetic test for BRCA2 following her breast cancer, it had implications for my extended family. Her positive test result led to my cousins taking a BRCA genetic test. Even though my aunt did not have a genetic test, we knew she was positive for the BRCA2 gene based on her daughters' positive test results.

Thinking back on my experience with taking a genetic test, I wasn't told with whom the data would be shared. This aligns with most cases, in which subjects were simply told that genetic analysis will be performed, without any explanation of what that means for their privacy.[23] While a lot of information can be shared via the internet without a user's knowledge or consent, my genetic code inherently feels a bit more personal than my browsing history. Personally, I would have appreciated transparency about where my genetic data might be stored and who might have access. This consent might even include a further layer: my consent to be recontacted and to reconsent, for example, when new information becomes available that is relevant to me, or if further research is being considered.[24]

CONCLUSION

Examining the narrative experience of teenage DNA testing uncovers a complex web of ethical issues that go beyond traditional frameworks. This complex web of decisions is exemplified by the idea of an open future, and the interaction of pediatric assent with a growing understanding of genetic privacy. A sophisticated approach is needed, as policy frameworks and philosophical viewpoints keep changing. This approach should acknowledge individual autonomy, grapple with the intricacies of an open future, and navigate the challenges associated with safeguarding genetic privacy.

While information about the future specialists I might visit was given to me after my positive test result, I would have liked this information before testing so that I could be prepared for the outcome. However, I would not have liked to receive this information before the age of 21. Given that I consider myself mature for my age, I believe knowing too much before I was ready to handle the information would have posed a burden. Therefore, I advocate for age-appropriate information to be provided to pediatric patients, so that they can be capable of assent. I agree that an open future should be preserved, because I appreciate the autonomy that it gave me to choose my own future. Choosing at what age to test, and when I was ready to handle the information, was a step towards taking charge of my own health. I believe this gives me an obligation to monitor my own future health. I believe genetic data should be accessible to researchers to be able to advance research. However, this genetic information should still be closed to the public. When an individual undergoes a genetic test, they should be told where their genetic data might be stored and who might have access to it.

Choosing at what age to test, and when I was ready to handle the information, was a step towards taking charge of my own health.

As we continue to navigate this complicated terrain, there are a few important areas that need more research and academic study. The first areas for future research must include improving age-based recommendations and investigating

the mental and emotional maturation required for teenagers to properly understand the consequences of genetic testing. By examining the equilibrium between protection and autonomy, we may more clearly define the point at which pediatric assent should change to informed consent. Second should be research that analyzes the complex effects of genetic testing on teenagers. Individuals who were genetically tested as children should be followed over time through longitudinal study to examine the effects of this information on their life decisions, professional paths, and general well-being. A more comprehensive understanding of the idea of an open future results from an awareness of the complex interactions that exist between genetic information and life choices. Third, we can examine how well the current regulatory frameworks protect individuals' sensitive genetic information as genomic technologies progress.

The complexity of predictive genetic testing underscores the importance of age-appropriate decision making, transparent communication, and a nuanced understanding of the open future principle. As the history of genetic testing moves us to continued research and refinement of regulatory frameworks, it also stands as a testament to the need for a sophisticated approach that embraces the delicate balance between the empowering adolescents in their healthcare choices and safeguarding their genetic privacy. In navigating this intricate terrain, in telling my story, I seek not only to contribute a personal perspective to the ethical discourse, but also to inspire a collective commitment to shaping policies that prioritize informed decision making, respect autonomy, and adapt to the ever-evolving dynamics of genetic testing.

NOTES

1. Committee on Bioethics, Committee on Genetics, the American College of Medical Genetics, et al., "Ethical and Policy Issues in Genetic Testing and Screening of Children," *Pediatrics* 131, no. 3 (2013): 620-2, doi:10.1542/peds.2012-3680.

2. J.R. Botkin et al., "Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents," *American Journal of Human Genetics* 97, no. 1 (2015): 6-21, doi: 10.1016/j.ajhg.2015.05.0223.

3. *Wisconsin v. Yoder,* 406 U.S. 205 (1972). Justia Law, *https://supreme.justia.com/cases/federal/us/406/205/.*

4. A. Fenwick, M. Planting, S. Dheensa, and A. Lucassen, "Predictive Genetic Testing of Children for Adult-Onset Conditions: Negotiating Requests with Parents," *Journal of Genetic Counselling* 26, no. 2 (2017): 244-50, doi:10.1007/s10897-016-0018-y.

5. L.B. McCullough et al., "Professionally Responsible Disclosure of Genomic Sequencing Results in Pediatric Practice," *Pediatrics* 136, no. 4 (2015): e974-e982, doi:10.1542/peds.2015-0624; J.R. Garrett et al., "Rethinking the 'open future' argument against predictive genetic testing of children," *Genetics in Medicine* 21, no. 10 (2019): 2190-8, doi:10.1038/s41436-019-0483-4.

6. B. Wilfond and L.F. Ross, "From Genetics to Genomics: Ethics, Policy, and Parental Decision-making," *Journal of Pediatric Psychology* 34, no. 6 (2009): 639-7, doi:10.1093/jpepsy/jsn075; J. Harris and K. Keywood, "Ignorance, Information and Autonomy," *Theoretical Medicine and Bioethics* 22, no. 5 (2001): 415-36, doi:10.1023/A:1013058801622; P. Malpas, "Predictive Genetic Testing in Children and Respect for Autonomy," in *Children's Health and Children's Rights,* ed. M. Freeman (Leiden, the Netherlands: Brill Nijhoff, 2006), 297-309.

7. S.M. Rosenberg et al., "BRCA1 and BRCA2 Mutation Testing in Young Women With Breast Cancer," *JAMA Oncology* 2, no. 6 (2016): 730-76, doi:10.1001/jamaoncol.2015. 5941.

8. A. Werner-Lin, L.M. Hoskins, M.H. Doyle, and M.H. Greene, " 'Cancer doesn't have an age': Genetic testing and cancer risk management in BRCA1/2 mutation-positive women aged 18-24," *Health* 16, no. 6 (2012): 636-54, doi:10.1177/136345931 2442420.

9. A.L. Bredenoord, W. Dondorp, G. Pennings, and G.D. Wert, "Ethics of modifying the mitochondrial genome," *Journal of Medical Ethics* 37, no. 2 (February 2011): 97-100, doi:10.1136/jme.2010.037481.

10. R.J.L. Darby, "The child's right to an open future: Is the principle applicable to non-therapeutic circumcision?" *Journal of Medical Ethics* 39, no. 7 (2013): 463-8, doi:10.1136/medethics-2012-101182.

11. T. Hainz, "The Enhancement of Children versus Circumcision: A Case of Double Moral Standards?" *Bioethics* 29, no. 7 (2015): 507-15, doi:10.1111/bioe.12141.

12. B.N. Waller, "Patient Autonomy Naturalized," P*erspectives in Biology and Medicine* 44, no. 4 (2001): 584-93.

13. B.S. Wilfond et al., "Navigating Growth At-

tenuation in Children with Profound Disabilities," *Hastings Center Report* 40, no. 6 (2010): 27-40, doi:10.1002/j.1552-146X.2010.tb00075.x.

14. Darby, "The child's right to an open future," see note 10 above.

15. R.E. Duncan et al., "An international survey of predictive genetic testing in children for adult onset conditions," *Genetics in Medicine* 7, no. 6 (2005): 390-6, doi:10.1097/01.GIM.0000170775. 39092.44.

16. D. Wendler and S. Shah, "Should Children Decide Whether They Are Enrolled in Nonbeneficial Research?" *American Journal of Bioethics* 3 (2003): 1-7, doi:10.1162/152651603322614382.

17. Duncan et al., "An international survey," see note 15 above.

18. A.L. McGuire and R.A. Gibbs, "No Longer De-Identified," *Science* 312, no. 5772 (2006): 370-1, doi:10.1126/science.1125339.

19. H. Dewey-Hagborg, "Probably Chelsea: Twenty different sculptural portraits, all based on the same person's DNA information," Exploratorium Museum Exhibit, San Francisco, Ca., January 2024, *https://www.exploratorium.edu/exhibits/probably-chelsea.*

20. W.W. Lowrance and F.S. Collins, "Identifiability in Genomic Research," *Science* 317, no. 5838 (2007): 600-2, doi:10.1126/science.1147699.

21. M.D. Burstein et al., "Pediatric Data Sharing in Genomic Research: Attitudes and Preferences of Parents," *Pediatrics* 133, no. 4 (2014): 690-7, doi:10.1542/peds.2013-1592.

22. Ibid.

23. Dewey-Hagborg, "Probably Chelsea," see note 19 above.

24. R. Dal-Ré et al., "When Should Re-consent of Subjects Participating in a Clinical Trial Be Requested? A Case-Oriented Algorithm to Assist in the Decision-Making Process," *Clinical Pharmacology and Therapeutics* 83, no. 5 (May 2008): 788-93, https://doi.org/10.1038/sj.clpt.6100357.