Disclosure of Genome Sequencing Results: Are Pediatricians Ready?

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In this issue of Pediatrics, McCullough et al1 use the tools of ethical analysis and argument to address the issues surrounding the disclosure of genome sequencing results, specifically whole exome sequencing (WES) and whole genome sequencing (WGS). From this analysis, they develop a framework for pediatricians to use in their practice to address the unique issues surrounding the disclosure of WES/WGS results. This review is particularly helpful because it addresses issues regarding genetic testing that were not readily apparent when the 1995 American Academy of Pediatrics’ Committee on Bioethics report2 was written. In this world of rapidly changing genetic testing, revisiting our previously well-thought-out guidelines is an important step.

In addition to understanding the ethical framework for the disclosure of genomic testing results, pediatricians must have sufficient knowledge of the testing procedures themselves. In the midst of the ethical decisions that must be made in terms of genome sequencing in children is the question regarding the comfort level of the pediatricians in discussing complex genetic test results and testing procedures. In 2012, the Genetics in Primary Care Institute (through the American Academy of Pediatrics Quality Improvement Innovations Network) performed a needs assessment survey of pediatricians in the area of genetic education.3 Of the pediatricians surveyed, 50% stated that they disagreed with the statement “I feel competent in providing health care to my patients that is related to genetics and genomics.” At the time the survey was conducted, WES and WGS were just being developed as clinical tests and were not addressed specifically by the survey.

As noted in the article by McCullough et al,1 “The results of genome sequencing are distinctive for their degree of complexity and subsequent challenges of interpretation for pediatricians, parent, and patients alike.” At the time of the survey, most of the variants of unknown significance were those discovered in doing whole genome chromosomal microarrays (comparative genomic hybridization). The number of variants of unknown significance found through WES or WGS is exponentially higher. With the increasing number of unclear results in these 2 tests, it is easy to imagine that the number of pediatricians who did not feel competent in addressing the issues would be even higher if the survey were repeated today.

The genetics and genomics workforce has not increased significantly since 2012, and at the time of that needs assessment, there were an estimated 1400 geneticists and 3000 genetic counselors. Should WES/WGS testing become more widespread, it is clear that the responsibility for pretest counseling, ordering of tests, and explaining the results obtained will fall more and more on the primary care providers, including pediatricians.

I applaud the authors1 for addressing the changing world of genetics through the lens of ethical principles, but I think the article highlights even more clearly the importance and urgency to increase the genetic and genomic
literacy of pediatricians and other primary care providers. There is clearly a need for a national effort to educate primary care providers and nongenetic specialists about genomic testing so that they can have an informed discussion with the families they serve. The decision to perform genomic testing in a child is complicated, and it requires an in-depth conversation before and after the testing.\(^4\)

Caring for children and families who have genetic differences requires a partnership between the primary care pediatrician and the appropriate specialists. Undertaking WES/WGS testing also requires a partnership between pediatricians and genetic specialists until the nuances of genomic testing become better understood by the majority of pediatricians.

**ABBREVIATIONS**

WES: whole exome sequencing  
WGS: whole genome sequencing

**REFERENCES**

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