IN THIS ISSUE

An effective decision aid for secondary findings selection from genomic sequencing

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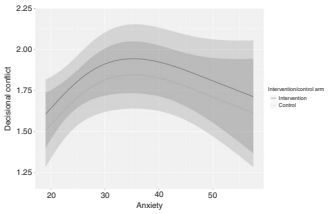

When faced with the return of results from genetic/genomic sequencing, there's a lot of information patients may, or may not, want to sift through. There are thousands of possible secondary findings—things like common disease risk and late-onset untreatable brain diseases that are outside of results currently recommended for routine reporting by ACMG—and only some are medically actionable. Helping patients understand every secondary finding is complicated and can take up a significant chunk of a clinician's time. That's why Bombard and colleagues created Genomics ADvISER (http://www.genomicsadviser.com), an online decision aid that educates patients about selecting secondary findings for reporting. Genomics ADvISER takes patients through five categories of secondary findings using videos and written explanations in plain language. Patients then fill out a questionnaire to gauge their interest in each category, are quizzed on the information presented, and ultimately make their decision. In this issue, the team presents the first clinical trial of this decision aid, evaluating the effectiveness of Genomics ADvISER by comparing participants who were randomly assigned to use the decision aid before genetic counseling with those who received genetic counseling alone. The participants were recruited from patients at cancer genetics clinics who had had a negative result in their first-tier genetic testing and thus were eligible for genomic sequencing. While those who used the decision aid said it improved their knowledge and provided enough information to make a decision based on their personal values, it did not reduce their decisional conflict. There were also no differences between the two groups in knowledge of sequencing limitations, preparation for decision-making, or satisfaction with the decision. However, the decision aid group did have better knowledge of secondary findings and sequencing benefits and spent significantly less time (24 minutes less) with their genetic counselor. The authors conclude that Genomics ADvISER could therefore reduce in-clinic time and costs while effectively educating patients. *—A. N. Grennell, News Editor*

Genetic testing as a clinical tool for infants and children with inherited eye disorders

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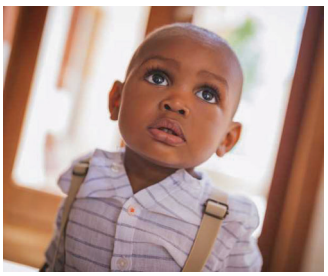

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More than 400 genes are associated with inherited eye disorders. While genetic testing has been recommended for several pediatric eye conditions by the American Academy of Ophthalmology since 2012, it is not uniformly applied. Previous work demonstrated that genetic testing can accurately identify variants of interest as well as aid in diagnosis of clinically defined disorders. But how genetic tests affect patient management and health outcomes is not well known. In this issue, Lenassi and colleagues examine the outcome of genetic testing for 201 children with a diverse range of inherited eye disorders. Their team focused on a broad definition of clinical utility—the ability of a test to measurably improve patient outcomes, management, and decision-making. Patients 5 years of age and younger with clinically diagnosed inherited eye disorders were retrospectively identified from the North West Genomic Laboratory Hub. Their disorders included pediatric cataracts, ectopia lentis, anterior segment dysgenesis, albinism, and inherited retinal disease. The team reviewed health-care records and clinical notes on each patient in conjunction with the genetic test results. In a majority of cases (64%), a molecular diagnosis was identified, although the diagnostic yield varied significantly depending on the disorder. Ectopia lentis, albinism, and inherited retinal disease all had a diagnostic yield above 75%. In one-third of cases, the diagnosis led to a change in disease management via personalized surveillance measures. With a diagnosis in hand, one-fifth of patients also avoided unnecessary tests. For those with inherited retinal disease, testing reduced uncertainty in their prognosis and determined eligibility for gene-based therapeutic trials. Apart from management of the disease, a precise diagnosis from genetic testing offers a better understanding of the condition, resolves uncertainty sooner, and helps inform life planning. The authors conclude that genetic testing could be used as a frontline diagnostic tool for the disorders studied, significantly impacting downstream clinical management. *—A. N. Grennell, News Editor*

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