IN THIS ISSUE

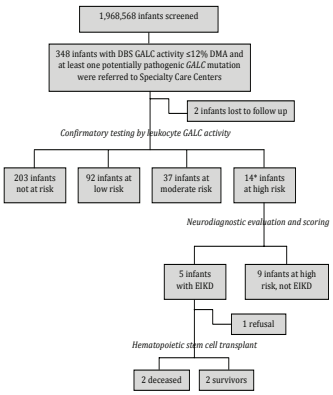
NGS superior to genotyping for carrier screening in an Ashkenai Jewish population

see page 1214

Next-generation sequencing (NGS) is poised to take over from traditional genotype-based screening as economical NGS tests become available. As reported in this issue, a recent carrier screening study of 506 Ashkenazi Jews living in Chicago suggests that commercially available NGS in this population performs better than traditional screening methodologies. Researchers at the Center for Jewish Genetics identified 288 carriers and two carrier couples at risk for recessive disorders that would have been missed through traditional screening methods. Genetic testing in this heavily screened population typically employs targeted mutation analysis (genotyping) for founder mutations in Tay–Sachs disease and a handful of other disorders, typically a total of 19. In contrast, the commercial NGS panel reported here encompassed 85 genetic disorders. The research team reports that 288 (57%) people were carriers of at least one genetic condition, with a smaller number carrying two or more mutations, for a total of 434 pathogenic variants revealed. The findings showed that only 312 (72.5%) would have been detected using a targeted mutation analysis panel. The researchers point out that this is a narrowly defined, high-risk, and self-selecting population. Nonetheless, the results raise questions about best practices for patient education and how best to describe the nature and limitations of various forms of genetic testing. *—Karyn Hede, News Editor*

Newborn screeing for Krabbe disease raises ethical considerations

see page 1235

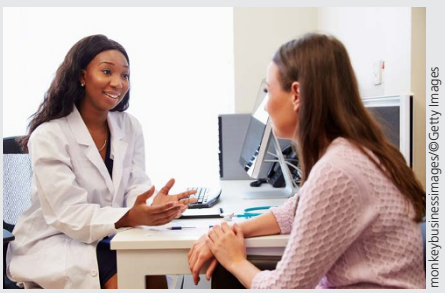


Newborn screening programs that include testing for lysosomal storage diseases such as Pompe disease have been essential in identifying infants who could benefit from available early treatment, for example, enzyme replacement therapy . But for some disorders, treatments are not as well established. In this issue, Wasserstein *et al.* and the New York State Krabbe Consortium discuss the outcomes for New York State's newborn screening for Krabbe disease, a rare lysosomal storage disorder affecting the central and peripheral nervous systems. There is no enzyme replacement for galactocerebrosidase (GALC), the missing enzyme, which is responsible for myelin turnover. Early hematopoietic stem cell transplantation (HSCT) is thought to improve outcomes if performed soon after birth. However, in New York State's experience, the risks may outweigh the benefits of screening, according to the authors. Of the nearly 2 million infants screened during the study, five were diagnosed with early infantile Krabbe disease. Three of them died—two from HSCT-related complications and one from untreated disease. The two remaining children, who both received HSCT, have moderate to severe developmental delays. The authors suggest that in the case of Krabbe disease, the risk-versus-outcome analysis of newborn screening may need to be reassessed from an ethics standpoint. *—Karyn Hede, News Editor*

NEWS BRIEFS

A simmering turf war: who can provide genetic counseling?

A September 2016 report by Kaiser Health News (KHN) is raising questions about a relatively new requirement by some insurers that women receive counseling by a certified genetic counselor before getting tested for cancer-predisposing mutations. Two insurers, United Healthcare and Cigna, have insisted for some time now that genetic counseling should take place prior to insurance approval for *BRCA1/2* testing. Earlier this year, Cigna's Genetic Testing and Counseling Program went further, making genetic counseling



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mandatory prior to testing for all forms of hereditary cancers. But that stance is not sitting well with some physician groups. In a statement released a year ago, the American College of Obstetricians and Gynecologists and the American Congress of Obstetricians and Gynecologists (collectively ACOG) released a formal statement opposing such restrictions and suggesting that the requirement would limit access to care. The statement implies that such restrictions only make it harder for patients to obtain care at a time "when delays may lead to worse outcomes." The group states, "ACOG opposes such attempts to restrict the scope of practice of obstetrician-gynecologists, who are fully qualified to provide pre-test counseling to their patients." Requiring women to receive genetic counseling from certified genetic counselors could particularly impact patients in underserved areas where fewer certified genetic counselors are available. In a conciliatory gesture, Cigna