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Patients diagnosed with a genetic disorder also want secondary findings

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In a report that may add fuel to the ethical debate over whether and how to return secondary findings to patients, Shahmirzadi et al. found that, among 200 families seeking diagnostic exome sequencing in a commercial clinical setting, an overwhelming majority wanted a report on secondary findings as well. In the first published study of its kind, the authors analyzed preferences for receiving secondary findings among those seeking a genetic diagnosis after referral by a genetic counselor. The patients—161 severely affected children and 39 adults with less serious illnesses—and their families received genetic counseling prior to testing and signed consent forms that asked specifically about four categories of potential secondary findings: carrier of recessive disorders, predisposition to later-onset disease, predisposition to higher cancer risk, and early-onset disease. All but one adult chose to receive secondary findings in at least one category, and six adults abstained from hearing results in at least one category. Parents and guardians were given the option to receive secondary findings only about early-onset disease; seven chose not to receive that information. The authors point out that these results may not be generalizable because all the patients were dealing with chronic and/or life-threatening illnesses, which may color outlook on future risk of illness. They also suggest that the variation between the guardians' and adults' choices may reflect a difference between information parents would like to learn about their children and information that adults would like to learn about themselves. *—Karyn Hede, News Editor*

Two enzyme treatments for Gaucher disease type 1 have similar safety profiles

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A real-world evaluation of two commercial forms of enzyme therapy for treatment of Gaucher disease type 1 (GD1) revealed nearly identical safety profiles, according to results reported in this issue. Pastores et al. evaluated patients who transitioned from one form of glucocerebrosidase, imiglucerase, to another, velaglucerase alfa, during a worldwide shortage of imiglucerase caused by temporary shutdown of a manufacturing facility in 2009. The observational study involved 211 GD1 patients with varying exposure to velaglucerase alfa, which was an investigational drug at the time. GD1, an autosomal recessive lysosomal storage disease due to mutations in the encoding gene (*GBA1*), requires lifetime infusion of glucocerebrosidase. At the time of the study, imiglucerase was the only treatment approved by the US Food and Drug Administration. The study addressed such safety concerns as generation of neutralizing antibodies to the medication and adverse events related to infusion and other treatments. Almost all the patients (189) completed the protocol. Inhibition was very low, and essentially identical for anti-imiglucerase and anti–velaglucerase alfa neutralizing antibodies. Only 3 of the patients who discontinued treatment with velaglucerase alfa did so because of an adverse event suspected of being related to the drug. The authors of the study reported payments from the company sponsoring the study, the maker of velaglucerase alfa. *—Karyn Hede, News Editor*

NEWS BRIEFS

Keeping medical genetics patient-centric

Thanks to the recent Supreme Court ruling in Association for Molecular Pathology v. Myriad Genetics, laboratories are now free to engage in a full range of diagnostic genetic testing, largely unencumbered by concerns of patent infringement. The resulting competition, which looks to be good for our patients, is already allowing *BRCA1/2* testing for substantially reduced costs and spurring the availability of tests that sequence broad panels of genes. However, with this welcome new progress comes a potential downside. There are

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increasing concerns about laboratories using manipulative tactics to capture market share and persuade clinicians to use a particular lab.

In response to these concerns, the Cancer Genetic Counseling Program at the Yale School of Medicine has issued a laboratory position statement, emphasizing that decisions about which laboratory to use for genetic testing should focus on test quality, turnaround time, and cost. Moreover, the statement includes a pledge not to accept gifts (such as speaking fees and trips) or funding from testing laboratories. Finally, the statement advocates that, whenever possible, laboratories that make their data publicly available should be favored, so as to advance our communal knowledge (e.g., regarding the interpretation of variants of uncertain significance). This is also relevant to payers (e.g., Medicare), which could make such sharing a requirement for coverage.