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Assisted reproduction and Prader–Willi syndrome

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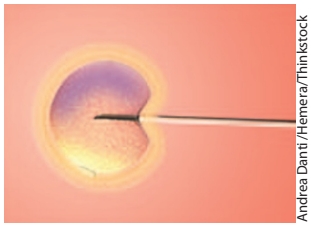

Andrea Danti /Hemera/Thinkstock

Assisted reproductive technology (ART), which enables couples having difficulty conceiving to bear children, is not without risk. The technology has been associated with an increased frequency of certain imprinting disorders. Gold et al. studied a cohort of children with Prader–Willi syndrome (PWS) and found that the use of ART by their parents did not seem to be higher than that for parents of other children. In an analysis of surveys completed by members of the Prader–Willi Syndrome Association, along with medical records, the research team determined that 1.1% of individuals with PWS were conceived using ART, compared with 1% of the general population, an insignificant difference. However, there was a significant difference in the cause of PWS among ART users. In this group, more cases were caused by maternal uniparental disomy (UPD) and imprinting defects than in the general population. The study is the first to report cases of an imprinting-center defect in PWS patients conceived via ART. The investigators suggest that the increase may be explained by the fact that the parents of those in the ART-conceived group were significantly older (mean age 36.3 and 39.7 years for mothers and fathers, respectively) than those of the naturally conceived group (31.5 and 31.6 years, respectively), and maternal UPD is associated with maternal age at conception. The age of parents using reproductive technologies must be taken into account when evaluating the risks associated with the technology. *—Karyn Hede, News Editor*

Next-gen sequencing company reports clinical carrier screening technique

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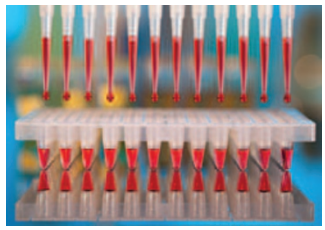

YAY Media AS /Alamy

Next-generation DNA sequencing is poised to supplant traditional genotyping methods for many genetic screening applications. But before it can do so, the technique must jump the hurdles of speed, accuracy, and cost per test. Researchers from the clinical diagnostic company Good Start Genetics, based in Cambridge, Massachusetts, report that they have developed a proprietary method that can provide cost-efficient genotyping that is as accurate as traditional methods while providing more detailed information. Noting that accuracy and reproducibility have been an issue with previously described next-gen methods, the authors explain that the new method uses commercial sequencing technology combined with probes designed to capture regions of interest in 15 well-known recessive Mendelian disorders. The sample set of 194 yielded a single-nucleotide-variant false-positive rate of 1.1 per million base pairs and one false negative. The authors do not provide precise measurements of insertion/deletion detection accuracy but report a sensitivity of 95.3% in the reference set. The company has developed an automated workflow that processes the steps from isolating DNA from blood or cell lines to delivery of a clinical report within 6 days. Of course, next-gen methods are not yet appropriate for some types of mutations, including certain large deletions and chromosomal rearrangements, but for routine testing of common Mendelian disease traits, they are rapidly gaining ground. *—Karyn Hede, News Editor*

NEWS BRIEFS

Don't scrap your '80s-era DNA sequencer

Scientists tend not to be a sentimental lot, especially when it comes to antiquated equipment. If a device or technique becomes outmoded, it gets tossed onto the scrap heap. In a fast-moving field like genomics, 10-year-old equipment may as well be a century old, given what it's worth today. But the curators of 12 science museums are now saying, "Not so fast"—the out-with-the-old mentality could mean losing a piece of genomics history. So was born the Museum Genomics Initiative, in which museums are working together to acquire selected genomics instruments that have historic value. In a recent report on the initiative published in *Nature* News (http://www.nature.com/news/museums-hunt-for-relics-from-genomics-early-days-1.14287), museum curators describe some of their recent efforts, from salvaging a '90s-era colony-picking machine from MIT to creating an art display from old microarray slides. The goal of the initiative is to develop a prioritized list of pieces recommended for acquisition. Just don't ask what's on the list. According to the *Nature* report, Robert Bud, chief curator of medicine at the Science Museum, London, isn't saying—at least not publicly—for fear of driving up the prices of items he'd like to acquire. So save those old PCR machines; your local science museum may want them.

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—Karyn Hede, News Editor