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Inconsistent lab reporting of possible consanguinity points to need for guidelines

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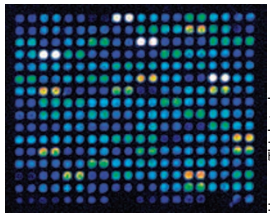
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The recent clinical availability of single-nucleotide polymorphism (SNP) microarray tests that can detect regions of homozygosity indicating that a child may have resulted from a consanguineous or incestuous relationship raises questions about how commercial laboratories handle these findings. Incidental discovery of a potential incestuous relationship raises serious ethical questions with potential legal ramifications. Despite the obvious need, there are no guidelines in place to assist in setting uniform standards for reporting suspected incest. In this issue, a group of investigators from Cincinnati Children's Hospital report that, in the absence of standards, laboratories vary widely in their rationale and approach to reporting. Of the laboratories surveyed, almost half (8 of 18) chose not to report the possibility of parental consanguinity, and 16 did not report potential parental incest even when the findings suggested it. None of the laboratories had ever contacted legal authorities, social-work teams, or an ethics board regarding suspected incest. The study also found variation in the methodology used to determine a cutoff value for suspected uniparental isodisomy, pointing to the need for laboratory standards in these calculations as well. As SNP microarray testing expands, the likelihood of facing incidental findings with associated legal and ethical dilemmas will only increase, hence the importance of addressing these issues now. *—Karyn Hede, News Editor*

Shift from central labs to point-of-care newborn screening prompts dialogue

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The development of rapid and reliable newborn screening technology is expected to increasingly shift the center of gravity from centralized reference laboratories to local, nursery-based settings. However, state public health departments face challenges to implementing new screening recommendations across a variety of birth settings. Using the recent experience of newborn screening for congenital hearing loss as a case study, the US Secretary of Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) conducted a series of meetings to frame the criteria for determining whether to recommend new point-of-care screening for newborns. Kemper et al. report the results of those meetings in this issue. Recognizing the barriers to implementing new screening procedures, the SACHDNC recommends that before clinicians begin new bedside testing, there should be an urgent need for test results, earlier than can be provided by an off-site laboratory, and evidence of better health outcomes using the point-of-care method. In addition, public health authorities must be able to ensure oversight of universal access to follow-up treatment after diagnosis. The authors note that implementing newborn screening protocols will require buy-in from state legislatures and insurance payers, and they expect the bar for such screening to remain high. *—Karyn Hede, News Editor*

NEWS BRIEFS

Data sharing, but with privacy protection, urged for whole-genome sequencing


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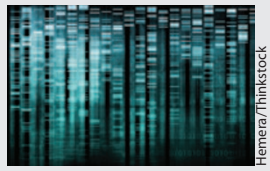
Trying to head off a quagmire of ethical issues expected to be encountered as whole-genome sequencing enters the public consciousness, the Presidential Commission for the Study of Bioethical Issues has issued a new report calling for respect for individuals' genetic privacy. But the report, *Privacy and Progress in Whole Genome Sequencing*, stops short of recommending specific procedures in such gray areas as informed consent as well as for handling the thorny issue of reporting incidental findings. The report acknowledges that whole-genome sequence data gathered almost assuredly will contain information whose importance is currently uncertain but that may raise difficult questions for individuals and their families in years to come. Therefore, the panel cautiously recommends basing consent procedures for clinical whole-genome sequencing on the consent procedures already in place for genetic tests. While stating that "consent processes should ascertain participant or patient preferences at the time the samples are obtained," it stops short of recommending opt-in consent for future studies, which many researchers say is burdensome and could hinder research. The report also notes that "individuals being asked to consent to whole genome sequencing should understand the volume of data and information to be generated, as well as the risks, benefits, and implications." However, given the wide range of public understanding of consent procedures, how to accomplish this remains unclear.
—Karyn Hede, News Editor

…As the 50-hour newborn genome sequence arrives


Hemera/Thinkstock

As if to illustrate the pressing need for genomic privacy protections, two reports published 3 October 2012 describe diagnostic genomic sequencing in gravely ill newborns and persons with severe intellectual disability—two groups that, it could be argued, are among the most in need of privacy protections. Saunders et al. demonstrated a diagnostic approach using whole-genome sequencing technology in a neonatal intensive care setting at Children's Mercy Hospital in Kansas City, Missouri. In their proof-of-principle study, the research team retrospectively diagnosed two

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cases and then prospectively diagnosed four difficult neonatal cases of monogenic disease within two days, using automated bioinformatic analysis that the researchers intend to be a prototype for clinical use in neonatal intensive care units. In a similar study, published in the *New England Journal of Medicine*, exome sequencing identified novel genetic causes of severe intellectual disability in 16 of 100 patients who had already undergone extensive diagnostic workup—including single-nucleotide polymorphism array profiling and targeted gene tests—to no avail. The exome sequencing of affected individuals and both parents identified rare de novo causative point mutations as well as several potential genes of interest for further study. In an accompanying editorial, Heather Mefford of the University of Washington comments on the rapid entry of whole-genome and exome testing into clinical diagnostic laboratories and urges continued collaboration between researchers and clinicians to ensure adequate quality control and continued innovation. *—Karyn Hede, News Editor*