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Outcomes of children diagnosed via newborn screening

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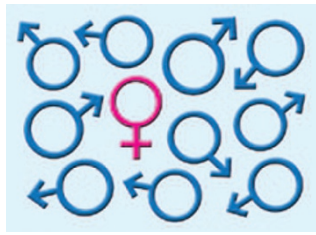
Cathy Yeulet/Hemera/Thinstock

The nationwide expansion of newborn screening programs provides an opportunity to better understand how early identification affects health outcomes of children. But lack of standardized data collection and of collaborative long-term follow-up has hindered efforts to determine whether children diagnosed with metabolic disorders receive appropriate health-care services. To address this issue, the US Centers for Disease Control and Prevention funded a three-year pilot project to expand and enhance public health data-collection programs in four states: California, Iowa, New York, and Utah. The authors report that the project not only improved state-level data but also, importantly, provided pooled data that sped the process of acquiring sufficient information to better understand how these rare disorders affect health outcomes. A total of 261 infants diagnosed with 1 of 19 metabolic disorders were followed through age 3. The research revealed that 88% received care at a metabolic clinic at least once in their first year of life. Those figures fell slightly to 77% in their second year and 74% in their third year. In the first year of follow-up, almost all the children (94%) saw a metabolic geneticist at least once; most (78%) also saw a metabolic dietitian, and half received services from a genetic counselor. The authors argue that continuing to track the long-term outcome of children diagnosed with metabolic disorders through newborn screening is necessary to maintain quality of care, identify areas for improvement, and observe the epidemiology of disorders recently added to the screening panel.
—Karyn Hede, News Editor

Connecting the dots between prenatal testing and a violent society

see pages 425 and 433



Noninvasive prenatal testing undoubtedly serves a vital role in early detection of genetic defects, but the authors of a review and an accompanying commentary in this issue bring critical focus to the technology's darker uses. Widespread use of prenatal diagnostic technologies to discover a fetus' sex early in pregnancy has led to highly skewed female-to-male ratios in China and (the focus of the review) in India. The review's authors draw troubling correlations between low female birth rates in several Indian provinces and higher levels of violence against women. They point out that, despite Indians' ingrained cultural preference for a boy, the problem had previously been contained. Recently, however, it has resurfaced owing to the availability of increasingly common and inexpensive methods for prenatal sex selection as well as financially burdensome demands for brides' dowries. The ability to identify a male fetus via noninvasive prenatal testing, available since the mid-2000s, is further encouraging prenatal sex determination, the authors state. They report that India's current female/male sex ratio of 0.914 among children under 6 is historically low and that in some areas, it is well below 0.9.

In his related commentary, Peter Benn, Director of Diagnostic Human Genetics Laboratories at the University of Connecticut Health Center, argues that clinical geneticists must confront these difficult moral quandaries: "The example of prenatal gender identification illustrates that even the most basic genetic information can fundamentally affect societies in worrisome ways." He notes that the recent public outcry about gruesome crimes against women in India could be used to put pressure on authorities to enforce laws banning prenatal sex selection.
—Karyn Hede, News Editor

NEWS BRIEFS

Oncologists unsure about use of complex genomic tests in cancer

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The coming genomic-testing revolution has practicing oncologists uneasy about how such tests should be used and how to return results to patients, according to a physician survey published recently in the *Journal of Clinical Oncology*. Cancer researcher Stacy Gray and her colleagues at Dana Farber Cancer Institute surveyed 160 of their colleagues ahead of a planned research project to implement predictive multigene genomic testing at their institution. They found a high degree of variability in how physicians planned to incorporate the predictive genomic tests into practice. Physicians who were confident and comfortable in their knowledge of genomics planned to test more patients than did those with less confidence. Alarmingly, about a quarter of those surveyed reported having little or no confidence in their knowledge of genomics or in their ability to base treatment recommendations on genomic data (22% and 26%, respectively). In addition,

NEWS BRIEFS *(continued)*

23% said that they would only rarely or sometimes explain clinically relevant findings to patients. Survey participants also varied widely in the language and terminology they planned to use in explaining results. The authors suggest that without standardized language, physicians run the risk of confusing patients and making treatment decisions more difficult, indicating an urgent need for innovative education to help cancer physicians integrate genomic data into their clinical practices.
—Karyn Hede, News Editor

Proactive intervention urged as NIPT goes global

In the face of escalating concern about how noninvasive prenatal testing (NIPT) is being applied in developing countries, Subhashini Chandrasekharan, an assistant professor at Duke University's Institute

Jake Lyell / Alamy

for Genome Sciences and Policy, and colleagues are calling for research into how commercial NIPT is being disseminated and its impact on prenatal care. In a commentary published in *Science Translational Medicine*, they outline several areas of concern pertaining to the dissemination of these tests and the general lack of regulatory control in many countries. For example, although both China and India forbid returning information to parents about fetal sex to help prevent sex-based abortion, those laws are weakly enforced (see the review by Madan and Breuning and the commentary by Benn in this issue). The authors note that many prenatal tests are offered in other countries through licensed purveyors of United States–produced tests, but that US companies rely on local regulatory agencies to enforce local laws. In addition, patients may be able to send samples to jurisdictions where such laws do not apply. Decisions by companies, local and national governments, nongovernmental organizations, professional societies, regulatory agencies, and international agencies will determine how effectively and ethically NIPT is implemented. The authors argue that promotion of these tests without societal safeguards in place may negate the benefit the tests are designed to provide.
—Karyn Hede, News Editor