

Screening and Caring for Children with Rare Disorders

By BRUCE K. LIN AND ALAN R. FLEISCHMAN

Newborn screening is a public health program that identifies genetic, metabolic, hormonal, and functional disorders in infants and provides comprehensive follow-up care. Without treatment, these disorders can result in devastating health consequences, and sometimes death. Newborn screening began in the mid-1960s, but has dramatically expanded in recent years. This is primarily due to new technology to reliably identify rare disorders, and to a 2005 report by the American College of Medical Genetics, commissioned by the United States Health Resources and Services Administration, that recommended mandatory newborn screening for a core panel of twenty-nine disorders. This report moves toward a national uniform newborn screening panel for serious disorders that have effective preventive interventions and treatments. It was enthusiastically endorsed by the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, the American Academy of Pediatrics, and the March of Dimes.

Two articles in this issue criticize the process of decision-making and the criteria for inclusion of disorders in the ACMG report and argue that resources spent on newborn screening might be better spent on other child health initiatives. The ACMG process for developing the recommended panel of tests was the creation of a multidisciplinary committee and solicitation of broad expert opinion. Its criteria for including a disorder were: identification of the disorder before symptoms occur through a sensitive, specific test performed shortly after birth; and demonstrated benefit of early detection, timely intervention, and efficacious treatment.

These criteria seem consistent with the ethics of mandatory newborn screening debated over the last forty years. Reasonable people may disagree on the definition of "demonstrated benefit" and "efficacious treatment," but the ACMG group took those concerns quite seriously. Since the report's release, forty-two states—acting on the judgments of advisory committees comprised of health experts and community members—have adopted screening panels that include at least twenty-one of the twenty-nine core disorders.

Bruce K. Lin is manager of public health initiatives and Alan R. Fleischman is senior vice president and medical director at the March of Dimes Foundation.

Critics argue that expert opinion is not enough to determine public health practice, and that only evidence-based data are sufficient to set screening standards. However, the threshold for evidence-based public health practice for screening for rare diseases is intrinsically different from evidence-based clinical practice, and from the approach applied to screening common chronic conditions like cancer or diabetes. The nature of rare disorders generally precludes randomized clinical trials to determine effectiveness of standard approaches to diagnosis and treatment. Case reports of successful interventions in small numbers of affected children may be all that is available to determine efficacy of treatment. Going forward, a national evaluation of newborn screening programs to further assess clinical effectiveness of population-based efforts is certainly warranted.

Newborn screening saves lives and decreases disability. The latest figures show that over four thousand children per year have a treatable metabolic or hormonal disorder, and another twelve thousand have a hearing deficit requiring intervention. Certainly many more children are born prematurely, or develop asthma or obesity, and all deserve adequate resources to prevent and treat these disorders. But it seems wrong to ask groups of children with known serious conditions to compete for resources that will determine their future health. Critics who argue that newborn screening may take resources away from other programs that might benefit more children tacitly accept the reprehensible behavior of state policy-makers and condone injustice when they cite examples like a state's diversion of Medicaid funds from prenatal care or asthma prevention to support newborn screening. Rather than pitting one group of needy children against another, shouldn't we focus our criticism on the injustice caused by inadequate resources allocated to children's health in general?

Newborn screening is an established, effective program to identify children with rare diseases and refer them for needed treatment. Rather than criticize the approach that led to the widely accepted national screening panel, we should assure that other disorders with efficacious treatment are systematically added to it, and that all children who suffer from rare disorders—regardless of where they are born or whether they are insured—receive the comprehensive follow-up they need.