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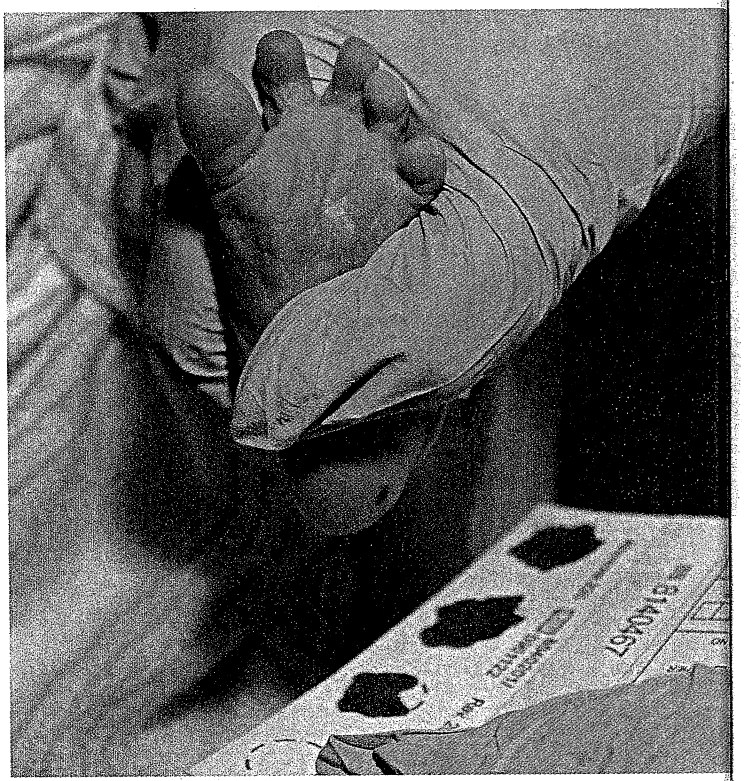
Perils of Newborn Screening

Doctors may be testing infants for too many diseases

The first symptoms often appear a month or two after birth. The babies' muscles stiffen. They lose their hearing and vision, stop sleeping and scream in pain. Some develop seizures. By the time many parents learn that their children have Krabbe disease—a rare genetic disorder that degrades nerve cells—it is too late for the only viable treatment, a transfusion of umbilical cord blood stem cells from healthy donors. Children with full-blown Krabbe who do not receive medical treatment, as well as many who do get treated, usually die by age two.

In some cases, doctors can prevent this grim outcome by screening infants at birth for genetic harbingers of disease. Right now such tests are mandatory in only a few states—something that many parents want to change. "If we don't screen for this disease at birth, those children will never have a chance at life," says Jacque Waggoner, CEO of Hunter's Hope Foundation, one of several advocacy groups lobbying state politicians to add mandatory tests for Krabbe and other rare diseases. The politicians are starting to listen. In the past year four states have passed legislation that requires hospitals to check newborns for abnormal enzyme levels linked to as many as seven new diseases.

Within the medical community, however, doctors are debating the rapid expansion of screening programs. As a whole, the programs have saved many lives. But some experts worry that states may be aggressively demanding tests for diseases that do not always develop in those who show signs of risk or cannot be safely or effectively treated even when they are caught. Doctors who have recently started screening for Krabbe and similar rare diseases are swiftly realizing that, in many cases, the results of such mandatory tests unnecessarily frighten parents and fail to help the children the tests were designed to save.



HARD TO HEAL: Screening infants' blood for signs of disease may not make sense if effective treatment does not exist.

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THE BIRTH OF NEWBORN SCREENING

THE CURRENT DEBATE has origins in the earliest forms of newborn screening. By the early 1960s microbiologist Robert Guthrie had perfected a test for phenylketonuria (PKU) that simply required a drop of blood from a baby's heel. Children with PKU suffer brain damage and seizures because they cannot break down the amino acid phenylalanine, which is found in high-protein foods.

Although most states adopted the procedure, a few doctors worried that some babies who did not have PKU would test positive and suffer malnourishment as a consequence of a low-protein diet. Ultimately the doctors' fears proved unfounded. (In a 2006 review of the medical literature on PKU, Jeffrey Brosco and his colleagues at the University of Miami found "no published cases of children who suffered permanent harm after an erroneous [newborn screening] test and treatment for a condition they did not have.") States soon began using similar tests to screen for the likelihood of developing other easily treatable diseases, including congenital hypothyroidism and sickle cell disease.

Today all states require newborn screening for between 28 and 57 medical disorders. Overall, these mandatory programs mark "one of the most significant advances ever in public health," says Stuart Shapira, a medical geneticist at the Centers for Disease Control and Prevention. Of the four million babies born in the U.S. every year, newborn screening identifies 12,500 with medical disorders. Catching and treating many of these disorders early, Shapira says, can prevent intellectual and developmental disabilities, organ damage and death.

Recently, however, doctors have raised new concerns, this time about the repercussions of widespread newborn screening. By the 1990s a tool known as tandem mass spectrometry had drastically expanded the number of disorders laboratory technicians could detect with a single drop of blood—from one to as