The importance of newborn screening is vividly and tragically demonstrated by the story of Zachary Black and Zachary Wyvill—known as the tale of two Zacharys in California.

The two boys were born one month and 60 miles apart in northern California. Zachary Black was tested for the rare genetic disorder glutaric acidemia type 1 (GA1) in a pilot program run by the California state public health laboratory’s Genetic Disease Screening Program (GDSP). Zachary Wyvill, born seven weeks premature, was not tested; his blood arrived at the laboratory but without the required parental consent form.

Zachary Black tested positive for GA1, which renders the body unable to digest two amino acids found in most food proteins. He was immediately placed on a special, low-protein diet and is a happy, healthy child.

Zachary Wyvill also has GA1. Absent a newborn screening result, however, it took physicians months (at considerable expense) to diagnose the disorder; long enough for toxic levels of undigested protein to accumulate in his body causing severe neurological damage. Zachary Wyvill may never be able to walk, talk or even feed himself. His family’s health insurance coverage is nearly maxed out.

While the tragedy is obvious, the outcome could have been worse: without the work of GDSP scientists, there would have been no pilot program and neither boy would have been screened.

More than 4 million babies are born in the United States each year and virtually all—97%—undergo screening for genetic and metabolic conditions in programs run by state public health laboratories. In addition to assuring the quality of newborn screening, these laboratories conduct research to improve existing tests and to demonstrate the efficacy of new tests, store newborn screening blood spots for future research and link families with specialists who can make final diagnoses and provide medically necessary follow-up services.

The California newborn screening program receives technical assistance from the CDC Newborn Screening Quality Assurance Program. It receives no direct federal funding.
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With data from the public health laboratory showing the utility of the GA1 test, in 2005 the state legislature added the disorder to the standard newborn screening panel used for the 560,000 babies born in California each year. Subsequent pilot programs have led to the addition of cystic fibrosis and biotinidase deficiency to the screening panel.

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Altogether, the GDSP now supervises screening for 76 disorders in a public-private partnership involving several contract laboratories.

Fred Lorey, PhD, acting GDSP chief, said, “We oversee those contract laboratories. We provide the equipment and screening methodology with quality control measures built in. They follow our protocols exactly.”

Since time is such a critical factor in newborn screening, test results are transmitted electronically back to the GDSP as soon as they are available. Public health laboratory scientists conduct a second level of quality control and then release the results to the family physician and birth hospital.

But the role of the public health laboratory does not stop there. GDSP scientists are constantly evaluating new technologies and working to make current tests better.

One example is the test for congenital adrenal hyperplasia (CAH), which is subject to a high rate of false positives, especially among sick or low birth-weight babies. Said Lorey, “False positives cause quite a bit of consternation to doctors and families, plus extra expense. Many babies with presumptive positives for CAH are already ill and in the neonatal intensive care unit, and it might be some time before the test resolves to a negative result.”

GDSP scientists supplemented the initial biochemical test for CAH with a second tier test that looks for five additional disease markers. The testing process now picks up more cases of true disease and yields fewer false positive results.

By helping to diagnose serious and potentially deadly disorders quickly and accurately, the public health laboratory benefits babies, families and the entire healthcare system.

Contact

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