By MELISSA KLEIN
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The drop of blood taken from a needle stick to newborn Matthew Fisch's heel showed no sign of PKU, a disorder that can cause mental retardation, or a biotinidase deficiency that may have prompted seizures and hearing loss.

Matthew, like all babies born in New York state, was tested for congenital disorders that most parents have never worried about, let alone heard of.

Although the test results were negative, Matthew's mother knew immediately her son had a problem, because he could not suck his bottle.

"I thought there was something wrong with the nipple. We were buying nipples. We were buying different bottles," said Jill Fisch of Edgemont. "We just had no idea."

When Matthew was 2 1/2 years old, and after he had failed to gain any weight for 18 months, doctors finally pinpointed the cause of his symptoms. He had a rare metabolic disorder called SCAD. Had he been born in Alaska, Mississippi or a few miles away in New Jersey, that newborn heel stick would have yielded a diagnosis.

"There are many states that do screen for this disorder," Fisch said. "Unfortunately, Matthew was not born in one of them."

Intense lobbying by parents such as Fisch may have helped change that. New York is planning to expand the number of disorders for which infants are screened, although the state Health Department will not release details of the program.

Such expansion could take place nationally. A federal advisory committee recommended in September that all states test for a panel of what are being called 30 core disorders and an additional 25 conditions related to the first group. A final recommendation is to go to Health and Human Services Secretary Tommy Thompson early next year and could work toward establishing a screening standard. Screening now varies so much that some states test for three disorders and some for more than 30.

"It's just mind-boggling to see the discrepancies from state to state," said Dr. Piero Rinaldo, a biochemical geneticist at the Mayo Clinic in Minnesota who served on the advisory panel and supports a national standard.

Although the disorders are rare, the goal of early identification is to

avoid complications that can be prevented, often with mere dietary changes.

"These are kids that don't need to be sick," said Dr. Louis Elsas, professor and director of the Dr. John T. MacDonald Foundation Center for Medical Genetics at the University of Miami, who has been working on newborn screening for four decades.

Elsas said that without screening, parents go from doctor to doctor until someone figures out what's wrong with their child.

"By that time, many times there's irreversible damage done, which is what we're trying to prevent by having newborn screening," he said.

The idea of screening babies started in the 1960s when Dr. Robert Guthrie, a Buffalo pediatrician whose son was mentally retarded, developed a method of taking a drop of an infant's blood and drying it on filter paper to be analyzed for phenylketonuria, or PKU. By the early 1970s, most states screened infants for PKU, and now every state does.

As technology improved, more disorders were added to the panel of those that could be screened.

New York now screens for 11, including HIV and cystic fibrosis. The screening still is done the way Guthrie developed. A tiny bit of blood from an infant's heel, optimally taken a day or two after birth, is placed on filter paper and sent to the state's Wadsworth Laboratory in Albany.

New York is among some 30 states that use tandem mass spectometry, a method of quickly detecting many disorders by looking for chemicals that are present in abnormal amounts or not at all.

"The efficiency of this is that you can do one test and get the results and screen for many diseases at once," said Dr. David Kronn, co-director of the Inherited Metabolic Disease Center at Westchester Medical Center in Valhalla. "It's possible, using the technology, to screen over 30 diseases at once, depending how you set the parameters of the machines."

Kronn, a member of the state's Newborn Screening Task Force, said looking for additional disorders should not raise costs significantly. The price of the screening is included in a hospital's charge for delivering a baby.

"The testing machine is already in place," he said. "It's just a question of modifying the software."

A baby identified with a metabolic disorder such as PKU - in which a missing enzyme cannot break down a protein in food - is referred to one of many

centers throughout the state for further analysis. At Westchester Medical Center, which handles the referrals for the lower Hudson Valley, testing is done to confirm the presumptive diagnosis.

In 2003, the state laboratory analyzed 267,848 blood samples from newborns. Excluding those with HIV, there were 502 confirmed cases of metabolic or endocrine disorders and sickle-cell diseases.

Jill Fisch said she began to search for answers when Matthew, the youngest of her three children, did not smile or turn over on time and had not gained weight. She took Matthew to the Children's Hospital of Philadelphia, where he was evaluated by genetic specialists. A feeding tube was placed in his abdomen, and a urine test showed he could have SCAD.

SCAD is an acronym for "short chain acyl-coA dehydrogenase deficiency." People with SCAD cannot process fatty acids because they either lack an enzyme to do so or the enzyme is not working properly.

Matthew's diet was changed to limit his fat intake. He is now given a formula through his feeding tube three times a day, the content varying with how much fat he has eaten. He also takes supplements to improve his muscles and build energy.

After the dietary changes, Matthew, who is now 3 1/2, began to grow and get stronger, although he still has deficits. He gets physical and occupational therapy for muscle weakness, and it was only recently that he has been able to walk up or down the stairs upright instead of crawling. A speech therapist and special education teacher visit the family's home weekly, as does a therapist to strengthen the muscles in his mouth that were too weak to allow him to suck his bottle as an infant.

Fisch said the family will never know what Matthew's full potential could have been.

"So would he be developmentally delayed had we known from the beginning? I don't know," she said. "Would he have needed a feeding tube? I don't know. We were never given the chance to know."

Fisch said that as she researched SCAD, she learned it could have been detected by newborn screening. Test kits are available that parents can take to the hospital and have sent to a private laboratory, although Fisch never knew she had that option. The cost for the testing ranges from about $60 to $90.

Fisch, who worked as a paralegal supervisor before having children, became national director of education and awareness for the advocacy group Save

Babies Through Screening. She testified before the federal advisory committee and met with Health Department officials in Albany to encourage expanded newborn screening.

"Hopefully, parents will not have a child who suffers a crisis or dies," she said. "And they won't have a journey like we had that took us over two years, when the answer really was right at our fingertips."