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HEADLINE: Parents look to save 'a lot of heartache';
State weighs adding 30 diseases to infant screening program

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BODY:

Every baby born in Maryland is tested for eight diseases that can cause mental retardation, deafness or even death. Now, the state is considering adding up to 30 more diseases so parents can take steps to stave off medical crises and boost their babies' chances for a healthy life.

"It would save a lot of parents and kids a lot of heartache," says Shantel Matthews of Easton, whose 4-year-old daughter, Heather, has a rare metabolic disorder. "It's extremely important, more important than anything else."

If she had known her daughter's condition at birth, says Matthews, she could have prevented seizures that left Heather with a type of brain damage that causes her to fall about 20 times a day. The seizures occurred when she was 15 months old, a seemingly healthy child.

Although the issue seems obvious on its face - early diagnosis to prevent lifelong disability - it has raised perplexing questions about the degree to which American medicine can apply its exploding knowledge base to prevent or cure disease. While many of the 30 diseases are clearly treatable, others are not.

The technology used to find these diseases looks for abnormal chemicals, not genes. But the debate over its use foreshadows a much larger issue that could face society as the genetics revolution provides tools to identify a vast array of illnesses.

"This is just the beginning," said Dr. Maimon Cohen, a geneticist at the Greater Baltimore Medical Center in Towson.

Most of the diseases under discussion are rare and unknown to the vast majority of people. They go by such alphabet-soup names as MCAD, VLCAD, GA-1 and GA-2. The most common, MCAD, is sufficiently rare that screening 80,000 newborns in Maryland each year would turn up about five cases. Others occur no more frequently than once in 100,000 births.

Expanded screening has become an issue with the introduction of tandem mass spectroscopy, which scans blood for the presence of unusual compounds. Many of the compounds are signs of metabolic diseases, defects in the way the body converts food to energy that can cause toxic chemicals to accumulate.

Adopting the new technology would require an initial investment of $1 million, according to state officials - the cost of purchasing two machines and hiring technicians needed to run them. But it could cost an additional $500,000 to $1 million a year to provide counseling and other services to parents whose children are identified by the new tests.

Pressure to adopt the technology is coming from parents who say their children could have been spared untold suffering if their illnesses had been identified at birth and treated before symptoms arose. Now, they say, Maryland has the opportunity to protect other children.

Since its introduction in the 1960s, Maryland's program of infant screening has prevented hundreds of children from becoming severely disabled from congenital diseases that include sickle cell anemia, hypothyroidism and PKU, a metabolic disorder. The disorders are identified through a simple blood test.

Infants diagnosed with sickle-cell disease receive antibiotics that prevent fatal infections. Children with hypothyroidism get thyroid hormone, a simple treatment that prevents mental retardation. Babies with PKU are placed on strict diets that prevent medical crises that can leave a child with brain damage.

While doctors generally agree that the new technology could be used to keep many children healthy, they say that might not be true for all of the diseases.

"Some are more treatable than others," said **Dr. Susan Panny,** who runs the genetics program at the state Department of Health and Mental Hygiene. "I do not believe there is good long-term evidence in the medical literature that shows what the outcome is from treating those disorders."

Panny said she can imagine that some parents who end up losing a child to an untreatable disease might, in hindsight, conclude that they would have been happier not learning the diagnosis until the child got sick. This would have preserved for them a few months or years of peace - free from the dread of a fatal medical crisis.

So far, only Massachusetts, North Carolina and Wisconsin have purchased the new machines to conduct universal screening of infants. Pennsylvania runs a voluntary program and contracts with a private laboratory.

The decision in Maryland will ultimately rest with Dr. Georges Benjamin, the state's health secretary. He will receive the recommendation of a state advisory committee.

To parents such as Shantel Matthews, the issue is clear.

Her daughter, Heather, seemed perfectly normal until she came down with a routine stomach disorder at 15 months. One morning, her parents could barely wake her. She became listless and started falling to one side.

In the ambulance, she developed seizures that affected the right side of her body. Later, while doctors were taking a CT scan, she developed a violent "grand mal" seizure.

Eventually, she was diagnosed with GA-1 and stabilized with medication. But the episode had the effect of a near-drowning, damaging part of her brain that controls movement. Today, Heather has normal intelligence but suffers from poor balance.

Matthews says the episode could have been prevented had she realized - before the first crisis - that the girl needed intravenous glucose infusions whenever she got sick.

One disease, MCAD, has attracted particular attention because its devastating effects can be almost uniformly prevented if parents take steps early. Children with this disorder can lead healthy lives as long as they do not go without food for more than about 12 hours. The situation often comes into play when a child gets the flu or some other illness and feels too sick to eat.

"If the kid is not eating for any length of time, come to the emergency room, get an IV and he's perfect," said Dr. Ada Hamosh, a Johns Hopkins geneticist who heads a state subcommittee that is considering the issue. Waiting too long, she said, could leave a child who has the disorder severely retarded.

She said many parents argue that detecting any of the disorders under study is worthwhile, even if there were no clear remedies. Most of the disorders are genetic, and parents who have passed on the disease to one child might want the information before they conceive a second.

Dr. Eric Wulfsberg, a University of Maryland geneticist who sits on the subcommittee, suggested a phased approach: The state could first screen all children for the handful of diseases that are without question treatable. At the same time, it could enlist a group of parents in a pilot study to measure the benefits of screening more widely.

Shantel Matthews and Dawn Dougherty, who has a child with VLCAD, are among those who have testified at public hearings held by the state's advisory council on hereditary and genetic disorders.

The next hearing is scheduled for 6 p.m. March 8 at the O'Conor Building, 201 W. Preston St., Baltimore.

Dougherty, of Ellicott City, said her daughter, Jordan, was lucky to escape severe brain damage. She, too, had gone

too long without food and developed seizures.

But Dougherty says she knows of other children who weren't so lucky.

"It's like a time bomb waiting to go off," she said.