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HEADLINE: Debate Surrounds Screening Newborns for Genetic Disorders

BODY:

TUESDAY, Nov. 18 [HealthDayNews] — A new blood-testing technology is stirring controversy about screening newborns for inherited disorders.

The technology, tandem mass spectrometry, can detect 20 or more genetic disorders by analyzing a single drop of blood, an improvement on the tedious series of tests for a more limited number of conditions that has been used until now.

Because most of the disorders are rare and many are virtually untreatable, health experts are debating not only whether the technology should be used for all newborns in the United States, but also how many conditions should be included in the screening.

The decision to use tandem mass spectrometry is being made state by state. Right now, it is mandated by law in 24 states. Four others will soon begin mandated tandem mass spectrometry screening and another four states allow its use on a voluntary basis, according to a study in the Nov. 19 issue of the Journal of the American Medical Association.

The push for universal use of the technology, despite some health experts' concerns about its safety and effectiveness, comes primarily from parents of children with genetic conditions, most notably deficiency of an enzyme whose complicated name is abbreviated as MCAD. Lack of the enzyme can cause a number of major developmental problems — from mental retardation to chronic muscle weakness — and can be fatal. Its estimated incidence is about one in every 10,000 births.

Commercial interests are also involved. Pediatrix Inc., a company that manufactures tandem mass spectrometry devices, is offering the testing in Maryland at a price lower than that of the state health department.

The latest entry in the tandem mass spectrometry debate compares children whose condition was detected at birth by the technique, to those who were diagnosed later in childhood.

The study, by Susan E. Waisbren and colleagues at Children's Hospital in Boston, also looked at the impact on a family whose child was falsely identified by tandem mass spectrometry as having a genetic disorder.

The children diagnosed at birth have had fewer hospitalizations than those whose condition was detected later. And their parents have suffered less stress than those whose children were diagnosed later.

But this is a preliminary report, and "questions remain," the researchers say.

"Do the benefits of expanded newborn screening outweigh its long-term costs in terms of quality-of-life considerations and financial burden?" the researchers ask. "Hopefully, continued study will permit detailed analyses of these questions so that rational decision-making will occur."

The March of Dimes Birth Defects Foundation takes a cautious approach toward tandem mass spectrometry, says Dr. Nancy Green, the foundation's medical director.

"We have a specific panel of disorders for which every baby should be screened, and MCAD deficiency is on that list," Green says. "So in essence, we are indirectly advocating tandem mass spectrometry, since that is the only way to detect the condition."

But the March of Dimes has not taken a position on use of the technique to detect many other rare disorders "because

the medical consensus on the value of screening for those disorders is not known," Green says.

It's easy enough to screen for a large number of disorders because after the initial setup cost of tandem mass spectrometry — estimated at $400,000 — "the cost of adding another five or 10 additional tests is minimal," Green says. "But we don't have the data to assess the value of those tests."

The Maryland Department of Health is using tandem mass spectrometry to screen for 32 disorders, up from nine before the technique was introduced, says **Dr. Susan Panny,** director of the department's Office for Genetics and Children with Special Health Care Needs.

Panny, like Green, says she is "waiting for more evidence" about the ultimate value of early detection of many rare conditions.

"Many disorders do not respond well, even when children are given the best available treatment," she says. The value of earlier detection can be determined only by comparing children diagnosed at birth with those whose problem is detected later, she says.

"It remains to be seen how much difference there will be between these two groups when the children are a little older," Panny says.

More information

To learn more about newborn screening, visit the National Newborn Screening & Genetics Resource Center. The center also has a list of screenings on a state-by-state basis.