

State is first to test newborns for T-cell deficiency diseases

By [Mark Johnson](#) of the Journal Sentinel

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Wisconsin, the first state to screen newborns for "bubble boy disease," has now become the first to test for the broad family of diseases known as T-cell lymphopenia, a development that could change detection and treatment of these illnesses around the world.

In January 2008, the state began testing drops of blood taken from the heels of babies in their first 24 to 36 hours in order to screen for "bubble boy disease," a rare immune system disorder marked by the presence of virtually no T-cells, which defend the body against invaders.

"Bubble boy disease," more formally known as severe combined immune deficiency, is thought to occur in between 1 in 50,000 and 1 in 100,000 infants, and the screen has yet to pick up a case in Wisconsin.

However, doctors from the Medical College of Wisconsin write in this week's Journal of the American Medical Association that the screen has a wider application in detecting diseases marked by a deficiency in T-cells. In the course of a year, Wisconsin tested 71,000 babies and found that eight had such a deficiency. Doctors were able to treat the one infant who had a severe T-cell deficiency, using a bone marrow transplant to essentially replace a faulty immune system.

"In 2008, the first year, we picked up a baby that had a severe immunodeficiency with T-cell lymphopenia. If the baby didn't get a bone marrow transplant, it would have died," said John M. Routes, section chief in the division of allergy and immunology at the Medical College and lead author of the paper. "The baby was given a bone marrow transplant here at Children's (Hospital of Wisconsin), and he's doing fine."

Wisconsin screens all newborns for 48 different disorders, well above the federal recommendation of 29 disorders.

Long-term effect

Although cases of "bubble boy disease" and T-cell lymphopenia are not frequent, doctors said the new screening could have a profound effect. The long-term survival rate is significantly higher when babies with the disease receive a bone marrow transplant in the first month of life than it is when they receive the transplant after six months.

But up to now, T-cell deficiencies in infants have often gone undetected. The babies get very sick from infections, fail to gain weight and often bounce from one hospital to another as doctors struggle to find an explanation for their health problems. The diseases are difficult to diagnose

because the children come to the hospital with seemingly common problems - ear infections, for example.

Doctors have had to be trained to look for signs that the common ailment is actually being caused by something far more sinister and severe. Even then, T-cell lymphopenias may not be picked up until a child is very sick.

With early screenings, "there's the potential for people to never again see a very sick patient (from these diseases)," said Kate Sullivan, an associate professor of pediatrics at the University of Pennsylvania. "It could be a completely different landscape for immunology."

"There have been papers on the technique, but this is the first paper to do it live - to run the screening and look at the results," said Ramsay Fuleihan, an associate professor of pediatrics at Northwestern University's Feinberg School of Medicine.

Fuleihan said it would be interesting to look at the long-term outcomes of the patients identified with T-cell lymphopenia "to see what their needs are."

Other states follow suit

In February, the state of Massachusetts followed Wisconsin and began performing the new T-cell lymphopenia screening. Other states, including Louisiana, Illinois, Minnesota and Iowa, are considering adding the test. Brazil, Sweden and Saudi Arabia also have expressed interest in adopting the screening.

"We are committed to helping any state get this up and going," Routes said. "We have a manual of how to do this, A to Z."

Routes said the cost of the screening is about \$5 to \$5.50 per infant. In Wisconsin, that cost has been picked up through grants from the Jeffrey Model Foundation, Children's Hospital of Wisconsin, the Wisconsin State Laboratory of Hygiene and the Centers for Disease Control and Prevention.

T-cell deficiencies can be treated when they're detected early but become deadly and more costly later.

"Not only can you save lives with the tests," Routes said, "but it would be cost effective and eventually save money."