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Just a simple blood test

Screening newborns for metabolic disorder saves lives

Many cases of MCAD are only detected in post-mortems

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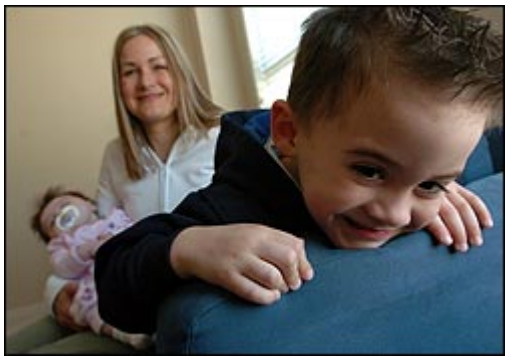

TANNIS TOOHEY / TORONTO STAR
Connor Kahler and his 9-week-old sister Rylie both have MCAD. Their mother Jacey and medical experts want newborns to be routinely screened for the condition.

Connor Kahler was 18 months old and miserable on Christmas Day, 2002. He woke up vomiting and spent the day lying listlessly on the couch instead of happily opening presents at his Brampton home.

When his mother Jacey started getting him ready for bed that night, his eyes rolled back in his head. By the time he was rushed to the Hospital for Sick Children, he was having seizures. After a battery of tests, Connor was diagnosed with MCAD, a disorder in which the body is unable to break down simple fats to make energy. Children with the disorder can die or suffer severe mental retardation very quickly if they go without eating for even a short period of time. "If I had put him to bed that night, he wouldn't be here today," Kahler says. "It was traumatizing."

A simple test at birth can detect MCAD, or medium-chain acyl-coA dehydrogenase deficiency, from a few drops of blood that are already taken from every newborn in Ontario to test for two other conditions, PKU and genetic hypothyroidism.
Once detected, the treatment for MCAD is simple — a low-fat, high-carbohydrate diet and if the child gets sick with the flu or an infection, he needs a simple glucose IV to prevent his blood sugar from dropping.

But Ontario, unlike four other provinces, does not test for MCAD or 26 other disorders that can be treated if caught early. Along with New Brunswick, it is at the bottom of the list in newborn screening in Canada, behind every country in the developed world and even places like Costa Rica, says a study published this spring in the *Journal of Paediatrics and Child Health.*

"Surely we can do better than Costa Rica," says Dr. William Hanley, a pediatrician in clinical and biochemical genetics at the Hospital for Sick Children, who conducted the study and calls the provincial government's inaction "negligence."

"I am getting tired of seeing these damaged children arrive here when something could be done to prevent it when they're newborns," he says in an interview.

So is Dr. Jim Cairns, Ontario's deputy chief coroner. Two years ago, he began testing for the metabolic disorders in babies who die under the age of 2 and "bingo, we started to get positives, especially for MCAD," he says in an interview.

He wrote to the Ministry of Health asking if it had plans to begin screening but hasn't received a definitive reply, he says. "We're now at the stage where we feel unless something happens, we will be calling an inquest," into one of the deaths.

Paradoxically, the coroner's office is paying to have blood samples sent to the U.S. to test for the diseases after a child has died, yet no one is testing newborns to prevent the deaths.

"That is right out of Franz Kafka," says Dr. Joe Clarke, director of the genetic metabolic diseases program at Sick Kids. The screening is possible because of new technology called tandem mass spectrometry (known as MS/MS), now used in Saskatchewan — which tests for all 29 conditions — as well as in Nova Scotia and British Columbia. Hanley estimates it would cost $2 million in the first year to buy the equipment and set up the program in Ontario, about the same amount it costs to treat one severely retarded person for life.

A U.S. study estimates that when combined, the 29 conditions affect one in every 800 births, which in Ontario means between 50 and 75 babies a year are not being diagnosed "until they end up on the post-mortem table," Hanley says. He was a member for six years of a Ministry of Health advisory committee that in 2002 called for expanded screening but the issue has got "lost in the bureaucracy at Queen's Park," he says. "They don't see it as important or vital" and there aren't enough specialists treating these children to "make a noise."

Clarke chaired that committee and resigned in protest when the ministry failed to act on its recommendations. The rest of the committee also later resigned. "There was simply no point in continuing," he says, although he has now agreed to take part in a new review.

The ministry is currently reviewing the whole issue of newborn screening internally but no decision has been made, says David Spencer, press secretary to Health Minister George Smitherman. While the ministry is aware that other jurisdictions test for more disorders, they don't all test for the same ones, Spencer says. "You need to look at what makes the most sense for your population."

Clarke is incensed by that view. The ministry has been reviewing its program for more than five years, he says, and "to use that as an excuse not to go forward is in my opinion irresponsible." Clarke doesn't see the dead children "but I see those unlucky enough to survive — they're severely brain damaged," he says.

In the U.S., the American College of Medical Genetics has just recommended that every state test for the 29 disorders that can be treated once detected.

Connor Kahler was one of the lucky ones who was caught in time and has shown no symptoms of brain damage. Because he was diagnosed, his sister Rylie was tested the day after she was born nine weeks ago and she too has the disease. "But it's so simple to treat as long as you know. I don't understand why they're not doing it," mom Jacey Kahler says.

Yolanda Sampson of Oshawa had never heard of MCAD until her 8-year-old son Nathan suddenly went into a coma in January and started having seizures that lasted up to 15 minutes at a time. After he was rushed by ambulance to Oshawa General Hospital, he was airlifted to Sick Kids.

It wasn't until a month of worry and sleepless nights on a hospital couch that he was diagnosed with the disorder. He's been having problems ever since, with headaches, fatigue and nausea, and his mother worries the damage is permanent. Nathan is so tired he can only go to school half-days and has to stop at least three times to catch his breath on the 15-minute walk to school.

She is still waiting for test results to find out if he has permanent brain damage but "he's just not the same child any more," she says. "I'm angry that I didn't know anything about this or that he could have been tested as a newborn," she says. "He went eight years without us knowing and this whole situation could have been avoided if we had known."

Parents can order a newborn testing kit from several U.S. medical centres for $30 U.S. and send it to a U.S. laboratory for testing. But most parents don't even know about the disorders or the test.

Pediatricians' offices are full of posters and pamphlets outlining the benefits of vaccines for everything from chickenpox to meningitis but there's no mention of screening for these disorders, says Tammy Clark whose 9-month-old daughter Jenna died of MCAD two years ago. "Why are there no posters about this issue so parents can decide?" she asks. "I truly believe they have a duty to inform."

June 1 will mark the 40th anniversary of the introduction of newborn screening to Ontario after then-NDP leader Stephen Lewis introduced a private member's bill to have all babies tested for PKU or phenylketonuria, which also causes severe retardation and has no symptoms until it's too late. Because of that test, former Toronto councillor John Adams' son was found to have the condition in 1986, put on an expensive special diet to treat it and is heading off to university this fall.

Adams has become a passionate advocate for expanded screening in the province. "My son is going to be a taxpayer instead of not being able to walk, talk or even be toilet trained," he says. "It tears my heart out that Tammy Clark's baby died in her arms."

★For information on where to obtain newborn screening test kits, go to: http://www.savebabies.org/NBS/snbs.php.