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Too few genetic tests: Marin

Ontario behind in screening infants, ombudsman says
Small investment would prevent some deaths

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Dozens of Ontario children are condemned to lives of "misery and despair" or death from rare but preventable diseases because the province doesn't do enough genetic testing of newborns, Ontario's ombudsman charges.

André Marin said last night he's launching a special investigation into the problem, which his office discovered during a probe into why the province won't fund life-saving drugs for a Barrie teenager with Batten's disease.

"Ontario only tests for two such conditions for newborns while international experts recommend testing for 29," Marin told the *Toronto Star*.

"We rank like a Third World country."

Marin said early checks with the Ontario coroner's office suggest "there have been about half a dozen questionable deaths we'll be following up."

Wider screening, done through blood tests, would cost an extra \$2 million a year according to preliminary estimates, but save the health system and families untold anguish and expense, Marin said.

"You're talking about a minimal investment with exponential dividends ... these conditions, to some extent, are preventable if they're caught early enough."

Some can be fixed with diet, such as MCAD, which leaves children unable to digest fats to make energy. They can die or quickly suffer severe mental disabilities if they don't eat frequently.

The treatment for medium-chain acyl-coA dehydrogenase deficiency is a diet low in fat and high in carbohydrates.

Ontario doesn't test for MCAD, only for phenylketonuria (PKU), which can also cause mental disabilities and hypothyroidism.

PKU is an inherited body chemistry disorder and affects about 1 in 14,000 children in the United States, according to the March of Dimes. Because of a missing enzyme, children with PKU cannot process a common food protein and this causes a chemical to build up in the brain. Children born with the condition appear normal for the first few months but eventually lose interest in their surroundings.

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"We're extremely concerned about this issue," said Marin, who is sending two investigators today to interview Tammy Clark, an Ottawa-area woman whose 9-month-old daughter Jenna died from MCAD two years ago.

Marin's concerns echo a study published this May in the *Journal of Paediatrics and Child Health*, which found Ontario is behind four other provinces that test for MCAD and more than two dozen other disorders that can be treated if caught early through blood tests.

The problems are known as inherited metabolic diseases and Ontario does not test for them now because it does not have the right technology in place, said David Spencer, spokesperson for Health Minister George Smitherman.

Smitherman ordered civil servants in June to investigate options and make recommendations by early September, Spencer added. "It's something we want to move forward on."

Saskatchewan, British Columbia and Nova Scotia already have the proper technology, known as tandem mass spectrometry, to do testing for all 29 conditions as recommended by international experts. One U.S. study estimated the conditions affect one in 800 births, which translates to as many as 75 babies a year in Ontario.

The Barrie teen whose case prompted this latest probe is suffering from Batten's disease, a potentially fatal neurodegenerative illness that has left him blind and prone to psychotic hallucinations. His father pays \$1,500 a month for a drug called Cystagon from the U.S., which is not covered here because Health Canada has not cleared it for general sale.

The investigation into newborn screening marks the third time since his appointment earlier this year that Marin has taken the government to task. His first effort was on behalf of severely disabled children whose parents could not afford to provide the round-the-clock care they often needed, forcing them to relinquish custody to local children's aid societies as the only way to get treatment.

The province has since pledged an extra \$10 million to help the children and restored parental custody to many families.

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