

Ont. newborn screening program expanding to include more tests

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Up to 20 additional screens to include sickle cell anemia and galactosemia

OTTAWA | Ontario's newborn screening program, now housed at the Children's Hospital of Eastern Ontario (CHEO), is gearing up to become the most comprehensive program of its kind in Canada.

Ontario has screened newborns for decades, but the program in place prior to April, when the province's screening program moved from the public health laboratory in Toronto to CHEO in Ottawa, only tested for phenylketonuria (PKU) and hypothyroidism. A third test, for medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, has already been added, and the lab expects to be screening for another two dozen conditions by the end of the year.

"Over the coming months we're going to add an additional number of inborn errors of metabolism, up to about 20 in total," said Dr. Michael Geraghty, co-director of the newborn screening lab at CHEO.

"In late summer we will start screening for sickle cell disease. Toward the end of the year we will add biotinidase and galactosemia, and then at the very end of the year we will start screening for congenital adrenal hyperplasia."

These changes will bring the program in line with guidelines set by the American College of Medical Geneticists, he added.

"Many of these inborn errors of metabolism are treatable. MCAD deficiency is the best example. Unrecognized, one-quarter of babies with this condition die during their first episode," Dr. Geraghty said. "And of the babies who survive, one-third of them have major neurological problems and only one-third are totally normal. And if we screen and diagnose these kids early, the outcome is excellent."

In its first four weeks of operation, the CHEO lab screened 17,400 babies.

"We have found five babies with proven hypothyroidism, two babies with PKU and two babies with MCAD deficiency. I think that's really what it's all about," Dr. Geraghty said.

"We weren't screening for that before," he said of the MCAD deficiency findings. "They wouldn't have been picked up, and they had a high likelihood of sudden death."

The lab will receive blood samples from infants born all over Ontario, and will use tandem mass spectrometry to test a single sample for many compounds simultaneously. It will also establish a computer system capable of tracking samples and hospital compliance, and will also enable followup with infants' primary care providers.

"Most of this testing is done in the birthing units, and that's who we report to," Dr. Geraghty said. "If this child has a GP or a pediatrician, then a copy of the report would go there, because a newborn screening report is just as important as the height, weight and head circumference and the vaccination record of the baby."