Ministry of Health and Long-Term Care

McGuinty government increasing protection for newborns

Expanding Screening Program For Inherited Metabolic Disorders From Two To 21 Tests

TORONTO, Sept. 7 /CNW/ - The McGuinty government will expand its newborn screening program by adding 19 tests for inherited metabolic disorders (IMDs) for all children born in Ontario, Health and Long-Term Care Minister George Smitherman announced today. It is the first expansion of the screening program in 27 years.

"There is nothing more important than keeping our children healthy and providing them with the best possible care," Smitherman said. "We are expanding our newborn screening program so that we can identify infants with these problems quickly and get them the treatment they need when they need it."

Currently, Ontario tests for phenylketonuria (PKU) and congenital hypothyroidism (CH). Beginning in 2006, the government will phase in 19 new tests for Inherited Metabolic Disorders. These IMDs are very rare, affecting only approximately one in 2000 newborns, but they can cause serious health difficulties and in some rare cases even death. Three new Tandem Mass Spectrometry machines will be purchased to allow Ontario's enhanced newborn screening program to test for these additional IMDs, ensuring immediate treatment.

The expanded screening panel is based on a preliminary recommendation of the Newborn Screening Subcommittee of the Ontario Advisory Committee on Genetics.

"I want to thank the subcommittee for its excellent work," Smitherman said. "I am looking forward to additional recommendations later this year on the further expansion of Ontario's newborn screening program."

"This expansion in newborn screening will ensure a number of conditions are caught and treated early," said Dr. Joe Clarke, chair of the Advisory Committee on Newborn Genetics and head of clinical metabolic genetics at The Hospital for Sick Children. "Many children will be saved from potentially disastrous consequences."

This initiative is part of the McGuinty government's commitment to build a health care system that delivers on three priorities - keeping Ontarians

healthy, reducing wait times and improving access to doctors and nurses.

This news release, along with other media materials, such as matte stories and audio clips, on other subjects, are available on our website at: http://www.health.gov.on.ca under the News Media section.

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Backgrounder

MCGUINTY GOVERNMENT EXPANDS NEWBORN SCREENING PROGRAM

The McGuinty government is expanding its newborn screening program by adding 19 tests for Inherited Metabolic Disorders (IMDs). These tests will detect IMDs and allow for quick treatment, preventing serious medical problems.

The 19 new tests for IMDs fall under three categories: Organic Acid Disorders (OAs), Fatty Acid Oxidation Disorders (FAODs) and Amino Acid Disorders (AAs). These are groups of rare inherited conditions caused by enzymes that do not work properly.

In May 2005, the Ministry appointed an Advisory Committee on Newborn Screening to undertake a comprehensive review of the Newborn Screening program and to provide expert advice and report back with recommendations by the end of the year. This advisory committee is a Subcommittee of the Ontario Advisory Committee on Genetics (OACG).

Organic Acid Disorders (OAs)

Ontario is adding nine new tests for organic acid disorders. Enzymes are needed to process protein from the food we eat for use by the body. Problems with one or more of these enzymes can cause an OA.

People with OAs cannot break down protein properly causing harmful substances to build up in their blood and urine, which can affect health, growth, and learning.

These tests include:

- Isovaleric acidemia (IVA)

- Glutaric Acidemia type 1 (GA I)
- 3-OH 3-CH3 glutaric aciduria (HMG)
- Multiple carboxylase deficiency (MCD)
- Methylmalonic acidemia (mutase deficiency) (MUT)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- Methlymalonic acidemia (Cbl A,B)
- Propionic acidemia (PROP)
- Beta-Ketothiolase deficiency

Fatty Acid Oxidation Disorders (FAODs)

Ontario is adding five new tests for fatty acid oxidation disorders.

Enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder.

People with FAODs cannot properly break down fat from either the food they eat or from fat stored in their bodies.

These tests include:

- Medium-chain acyl-CoA dehydrogenase (MCAD)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)
- Trifunctional protein deficiency (TFP)
- Carnitine uptake defect

Amino Acid Disorders (AAs)

Ontario is adding five new tests for amino acid disorders.

Protein is made up of smaller building blocks called amino acids. A number of different enzymes are needed to process these amino acids for use by the body. Because of missing or non-working enzymes, people with AAs cannot process certain amino acids. These amino acids, along with other toxic substances, then build up in the body, and cause problems.

These tests include:

- Maple syrup disease (MSUD)
- Homocystinuria (due to CBS deficiency) (HCY)
- Citrullinemia (CIT)
- Argininosuccinic acidemia (ASA)

- Tyrosinemia type I (TYR I)

The members of the subcommittee, who are all experts in the fields of newborn screening and inherited metabolic diseases (IMD), are:

Dr. Joe Clarke (Chair)
Head, Clinical Metabolic Genetics
Division of Metabolic Genetics
Hospital for Sick Children

Dr. Ron Carter
Pathology & Molecular Medicine
McMaster University
Hamilton

Dr. Pranesh Chakraborty
Department of Genetics
Children's Hospital of Eastern Ontario

Dr. Natalie Lepage
Head, Biochemistry
Children's Hospital of Eastern Ontario

Dr. Jennifer Mackenzie
Department of Genetics
Queen's University

Dr. Kenneth D. Onuska
Clinical Biochemist, Laboratory
St. Joseph's Health Centre, Sudbury

Dr. Murray Potter
Head, Biochemical Genetics
Hamilton Health Sciences
McMaster University Medical Centre

Dr. Ken Pritzker

Pathology and Laboratory Medicine
Mt. Sinai Hospital

Dr. Tony Rupar
Director, Biochemical Genetics Laboratory

The screening program will be conducted by three new tandem mass spectometry machines. Tandem mass spectrometry allows for the screening of many more IMDs than the technology currently used in Ontario, which can only screen for phenylketonuria (PKU). The ministry issued a letter of invitation to seven Ontario hospitals on August 2, 2005, inviting them to submit a proposal to host the new machines. The deadline for submission of proposals was Friday, September 2, 2005. Review of responses is underway and a decision on the successful site will take place in late September 2005.

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