

November 23, 2006

## CYSTIC FIBROSIS AND NEWBORN SCREENING

### Cystic Fibrosis

Cystic Fibrosis (CF) is the most common fatal genetic disease affecting young Canadians. It is estimated that one in every 3,600 children born in Ontario has CF. CF affects mainly the lungs and the digestive system. CF causes severe breathing problems in the lungs. A build-up of thick mucus makes it difficult to clear bacteria and leads to cycles of infection and inflammation, which damage the delicate lung tissues. In the digestive tract, CF makes it extremely difficult to digest and absorb adequate nutrients from food.

The Advisory Committee on Newborn and Childhood Screening (ACNBCS), which is chaired by Dr. Joe Clarke of the Hospital for Sick Children, recommended the implementation of screening for CF at its September 1, 2006 meeting. The ACNBCS is a permanent and independent advisory body which has been established to provide the Ministry of Health and Long-Term Care with ongoing advice on its policies and programs related to newborn screening. The committee was established in February 2006.

### Newborn Screening

Ensuring that Ontario's newborns get the best start in life is a key priority for the McGuinty government. That's why it is expanding the province's newborn screening program to screen for 28 rare disorders.

Although most babies with these disorders look healthy at birth, they may be at risk of having serious health problems – including developmental disabilities, recurrent sickness and even death – if their disorder is not detected in the first days of life and treated. Early identification is the key to effective treatment.

The government has established a state-of-the-art screening program, based at the Children's Hospital of Eastern Ontario (CHEO), in Ottawa. Blood samples from every child born in Ontario are sent to the lab where they are tested for 20 inherited metabolic disorders (IMDs), three blood disorders, two endocrine disorders and two classified as "others". The lab is now screening for 24 of the 27 conditions in the expanded panel that was announced in 2005, with three more on track to begin by the end of the year. Screening for CF, which will bring the total number of tests to 28, will begin by December 2007.

Although these disorders are very rare – as a group, they will affect about 110 out of approximately 130,000 babies born each year in Ontario – testing newborns within the first days of life means they can be treated early, reducing the chance of serious health problems later in life.

## *Screening Process*

To perform the screening test, a small sample of blood is taken from the child by pricking the baby's heel. The blood sample is collected on a special paper card and sent to the Newborn Screening Program Laboratory at CHEO.

The screening test shows whether there is a high or low risk of the child having a disorder and identifies babies who need further testing.

More than 99 per cent of babies screened will receive a "screen negative" result. This means there is a very low risk the newborn has one of the rare disorders. A positive test does not necessarily mean that the baby has a disorder, only that further testing is required.

## *Screened Disorder Categories*

- **Metabolic disorders :**  
When the body is not able to break down (metabolise) certain substances in food like fats, proteins or sugars, they can accumulate in the body and cause serious health problems.
- **Endocrine disorders :**  
The endocrine system, which is responsible for producing hormones, can sometimes produce too much or too little of some hormones, leading to sickness or developmental disabilities.
- **Blood disorders :**  
Oxygen is carried through the body in the hemoglobin found in red blood cells. If hemoglobin is not formed or not working properly, red blood cells break down, leading to health problems including severe anemia and serious infections.
- **Other disorders :**  
Biotinidase and Galactosemia are disorders related to defective enzymes. CF is a disorder that impedes the normal functioning of the respiratory and digestive systems.

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