Summary: Ontario will begin screening newborns for cystic fibrosis (CF) in April, 2008. CF is the latest addition to the panel of diseases screened in the blood sample obtained routinely from newborns via the heel prick test. Other diseases screened include metabolic, endocrine, and blood diseases.

Bottom line: New or expecting parents should be informed about expanded newborn screening, which now includes cystic fibrosis. The vast majority of babies screened will receive a screen negative result. A positive screen does not mean that the baby has a disease; rather that follow-up testing is required.

Newborn screening in Ontario
- Ontario’s Newborn Screening Program expanded in April 2006 and is based at the Children’s Hospital of Eastern Ontario (CHEO) in Ottawa.
- All newborns should receive a heel prick test between 24 hrs and 1 week of age.
- Newborn screening reports are currently issued to the institution or health care provider who submitted the heel prick blood specimen.
- Newborn Screening Follow-up Centres in London, Hamilton, Toronto, Kingston & Ottawa are responsible for retrieving babies with screen positive results, arranging and interpreting appropriate diagnostic investigations, initiating treatment, and providing feedback to the provincial program. There are regional differences in the way the Centres are structured, so procedures vary by region.

The Disease: Cystic Fibrosis
- A disease involving primarily the respiratory and digestive systems that affects an estimated 1 in 3600 children born in Ontario.
- Thick mucus clogs the lungs and heightens susceptibility to infection.
- Mucus also obstructs the pancreatic ducts, preventing release of digestive enzymes.
- Most babies with CF are initially asymptomatic.
- Symptoms tend to be non-specific, including recurrent cough, abdominal pain, loose stools and failure to thrive.
- Treatment for CF includes chest physiotherapy, inhaled medications, and artificial digestive enzymes taken orally.
- Predicted median life expectancy for individuals with CF is in the late 30s.

The Gene
- CFTR (cystic fibrosis transmembrane conductance regulator gene) encodes a protein that regulates the transport of salts across cell membranes.
- Autosomal recessive inheritance. Disease results from two non-working copies of the gene.
- Delta F508 is the most common CF gene mutation, but upwards of 1000 mutations have been described.
- Approximately 1 in 30 Ontarians “carries” a defective copy of CFTR, meaning one of the pair is changed while one is normal. Carriers do not have symptoms of CF.
Benefits of newborn screening for CF
- Early identification, permitting immediate and optimum treatment
- Improved nutrition, lung function, and growth; decreased hospitalization
- Improved cognitive outcome and psychosocial advantages to the family
- Genetic counselling opportunities: parents of a child with CF are likely carriers of the disease and have a 25% chance of another child with CF. The option of prenatal testing is available and referral for genetic counselling is recommended.

Screening and confirmatory testing for CF
- Newborn screening for CF starts with analysis of immunoreactive trypsinogen (IRT), a pancreatic enzyme, in the blood spot. Babies with CF usually have pancreatic insufficiency, resulting in abnormally high levels of IRT.
- IRT levels above the screening cutoffs will trigger genetic testing using a panel of 39 common CF mutations. There are 3 types of screen positive results:
  - **Category A**: Elevated IRT and 2 CFTR mutations
  - **Category B**: Elevated IRT and 1 CFTR mutation
  - **Category C**: Markedly elevated IRT (>99.9th centile) and NO CFTR mutations
- Diagnostic confirmation involves sweat chloride testing and repeat genetic testing.
- As a result of follow-up testing, some babies will be identified as CF carriers.

Harms/Limitations of newborn screening for CF
- Parental anxiety awaiting confirmatory testing following screen positive results.
- As with any screening program, false negative results will occur infrequently. If symptoms of CF are suspected in an infant or child, a diagnostic work-up for CF is indicated, regardless of the newborn screening results.

Role of Family Physician
- Discuss newborn screening via the heel prick test with new or expecting parents.
- Help expecting parents understand that a positive screening test does not mean that their baby has a disease, rather that follow-up is required.
- Well baby care.

ONSP Educational Resources:
- Downloadable PDFs are available in the provider section at [www.newbornscreening.on.ca](http://www.newbornscreening.on.ca)
- Parent Brochures: “My Baby Has a Positive Cystic Fibrosis Newborn Screening Result” & “My Baby is a Cystic Fibrosis (CF) Carrier”
- Healthcare provider information: Ontario Ministry of Health CF fact sheet & “A discussion guide to counsel patients about a positive newborn screen for cystic fibrosis”

Web Resources:
- Ontario Newborn Screening Program website [www.newbornscreening.on.ca](http://www.newbornscreening.on.ca)
- Ontario Ministry of Health website: [www.health.gov.on.ca/newbornscreening](http://www.health.gov.on.ca/newbornscreening)
- The Canadian CF Foundation: [www.cysticfibrosis.ca](http://www.cysticfibrosis.ca)

- More references available at [www.newbornscreening.on.ca](http://www.newbornscreening.on.ca)