



# The Ontario Newborn Screening Program: An Introduction

**February 19, 2007**

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# Newborn Screening

- ~135,000 babies/yr born in Ontario
- most appear to be (and are!) completely healthy
- a few have rare disorders that are not apparent at birth
- If untreated, these babies may develop serious health problems, mental retardation, or even die



# Why NBS?

- ***Intervention***
  - Early diagnosis/treatment improves prognosis
- Enumeration and surveillance
- Reproductive counselling

# A Brief History

- Ontario began screening for PKU (phenylketonuria) in 1965 and congenital hypothyroidism in 1978
- Nov 2005 – MOHLTC announced that the NBS Program would be expanding and would be moving to CHEO
- At least 27 rare disorders to be screened
  - 22+ metabolic disorders
  - 2 endocrine disorders
  - 3 blood disorders



# GO LIVE – April 4, 2006

- In less than 6 months, the Ontario Newborn Screening Laboratory at CHEO was up and running and began reporting

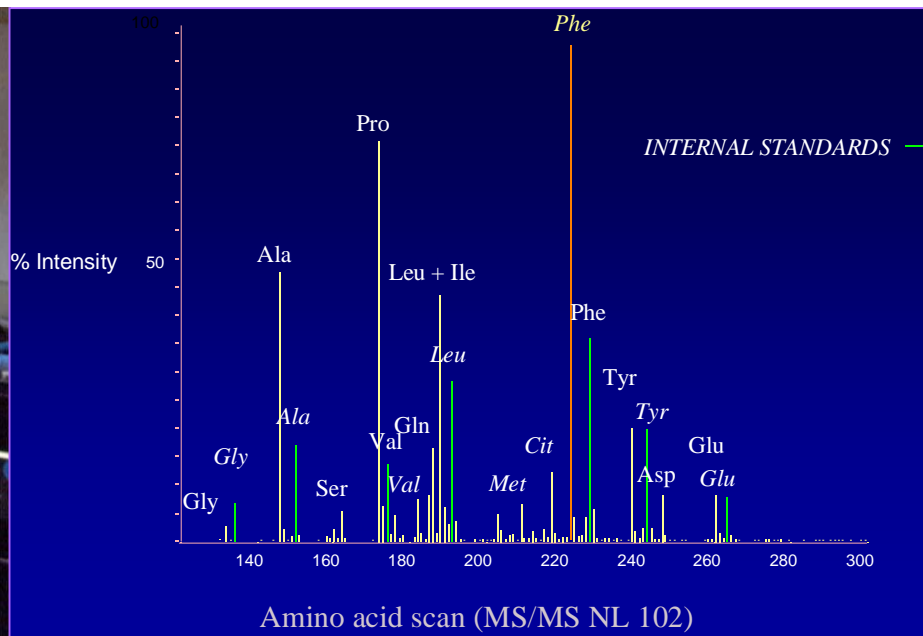
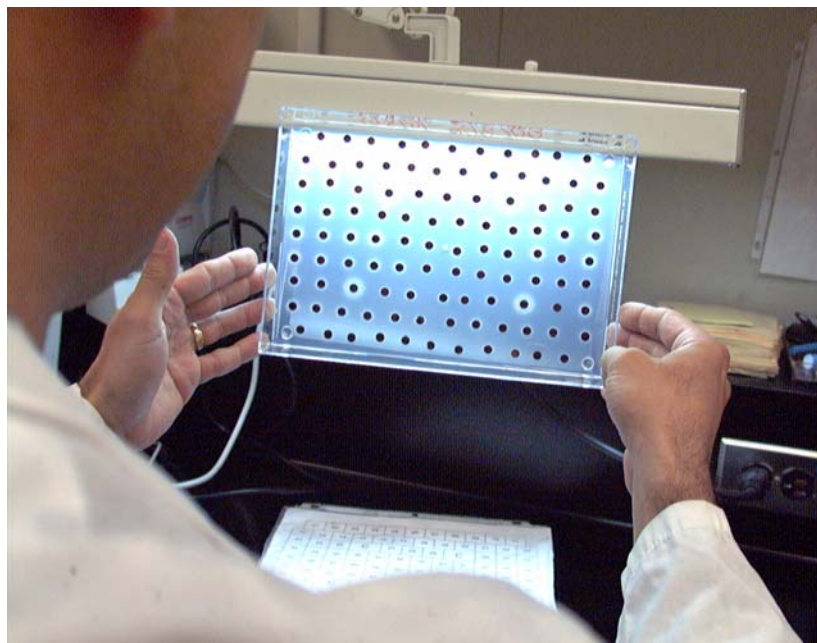


Ontario Newborn Screening Laboratory  
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# Expanded NBS

- April 4/06- PKU, CH, MCADD
- August 8/06- 18 additional metabolic conditions (AAs, OAs, FAODs)
- November 24/06- 3 hemoglobinopathies (sickle cell & variants)
- February 19/07- Galactosemia and biotinidase deficiency

# New Technology



# New Blood Collection Cards



**Ontario Newborn Screening Program**  
Children's Hospital of Eastern Ontario  
401 Smyth Road, Ottawa, Ontario  
K1H 8L1 Tel: 613-738-3222

XXXXXXXXXX

Dear Parent,

A blood sample has been taken from your baby's heel to test for very rare, but serious disorders which can cause mental retardation, poor growth, or death if not treated. The pamphlet "Newborn Screening: A healthy start leads to a healthier life" describes the newborn screening tests. If you have not received that pamphlet, ask your nurse, baby's physician or your midwife for a copy.

If your baby's test is done before he or she is 24 hours old, ask your midwife or your child's physician to repeat the newborn screening test within 5 days. If your baby seems sick, bring him or her to a physician as soon as possible.

The Newborn Screening Program office will report test results to the hospital, doctor, midwife or individual who sent the test, and to your baby's health care provider if his/her name is on the newborn screening requisition form. If the results indicate that a repeat test is needed, the Newborn Screening Program office will notify that healthcare provider. If the test is positive, the Program office will also notify a treatment centre that may contact you directly. If you have any questions, speak to your baby's doctor, your midwife or contact the Newborn Screening Program at (613) 738-3222.

**INSTRUCTIONS TO HOSPITAL:**  
Remove this top copy and give to parent.  
Then fill out multi-copy form for Lab.

**PARENT'S COPY**

USE BALL POINT PEN. PRESS HARD. INSTRUCTIONS ON BACK. PRINT LEGIBLY. IF A STAMP IS USED, STAMP ALL COPIES. COMPLETE ALL FIELDS.

COMPLETELY FILL ALL CIRCLES WITH BLOOD      Whatman 903® Lot # X-XXX Exp. XXXXXX

NBS Barcode  
FOR OFFICE USE ONLY

1234567

NBS Barcode  
FOR OFFICE USE ONLY

1234567

**Ontario Newborn Screening Program**  
Children's Hospital of Eastern Ontario  
401 Smyth Road, Ottawa, Ontario  
K1H 8L1 Tel: 613-738-3222

INFANT	Last Name		Sex: <input type="checkbox"/> M <input type="checkbox"/> F	Date of Birth
	First Name		Multiple Birth: <input type="checkbox"/> A <input type="checkbox"/> B <input type="checkbox"/> C	Time of Birth
	Health Card Number		Birth Weight: _____ g	Date of Collection
<input type="checkbox"/> 1st Test      Feeding: <input type="checkbox"/> Breast <input type="checkbox"/> Formula <input type="checkbox"/> Retest Prior Unsatisfactory <input type="checkbox"/> TPN <input type="checkbox"/> NPO		Transfusion: <input type="checkbox"/> Y <input type="checkbox"/> N, if yes: _____ <input type="checkbox"/> Retest Prior Screen Positive      Premature: <input type="checkbox"/> Y <input type="checkbox"/> N, if yes: gestational age _____ wks		Time of Collection
MOTHER	Last Name		Date of Birth	
	First Name		Phone Number	
	Address		Alternate Phone Number	
City		Province		Postal Code
SUBMITTING HEALTH CARE PROVIDER	Last Name		First Name	
	Address		City	
	Address		Province	
Postal Code		Phone Number		x
Birth Hospital (if different from above)		FOR SUBMITTER USE:		
Last Name		Submitter Unique Number		
First Name				
City				
Phone Number				
Ext.				

Report:  Screen Positive  Screen Negative

USE BALL POINT PEN. PRESS HARD. INSTRUCTIONS ON BACK. PRINT LEGIBLY. IF A STAMP IS USED, STAMP ALL COPIES. COMPLETE ALL FIELDS.

1. Please see instructions on back and complete all fields.  
2. Please ensure this flap covers blood spots when sending in the specimen.

**General Instructions:**

- ALL information on this form must be complete for identification and reporting purposes. Print identifying information on this form with pressure using a ball-point pen. If a stamp or label is used, stamp or label all copies.
- Do not use this kit after expiry date (shown on filter paper).
- Blood samples are to be taken on every infant before discharge from the hospital regardless of age, milk intake, or age at discharge.
- Optimum time for screening is 48 to 72 hours after birth. Any infant discharged before the age of 24 hours must have a repeat test within 5 days.

**Instructions for Blood Collection:**

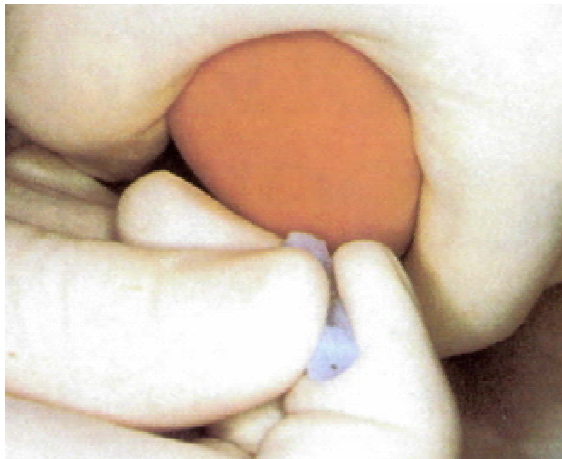
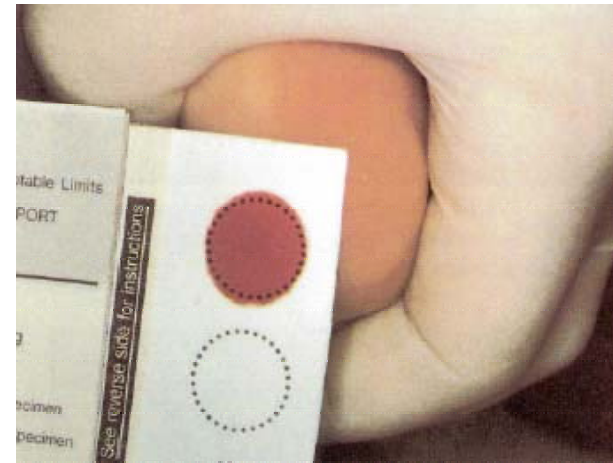
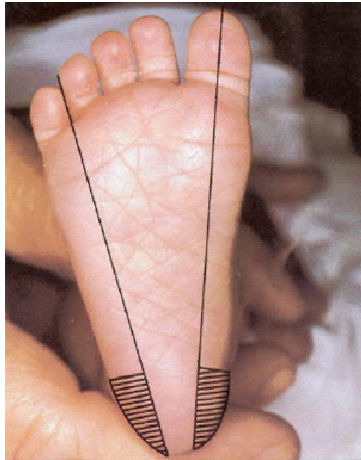
- Warm infant's heel with soft cloth moistened with warm water for 3 to 5 minutes.
- Cleanse skin with alcohol only, let dry and puncture heel with 2.0 mm disposable sterile lancet (see diagram). Wipe away first blood drop with sterile gauze and allow another large blood drop to form.
- Lightly touch the filter paper to the large blood drop while viewing from the other side to ensure complete penetration and saturation of the entire circle with a single application. Apply blood to one side of the filter paper only. Do not use capillary tube to transfer blood as this may damage filter paper.
- Fill the remaining circles in the same manner. If blood flow is diminished, repeat steps 1 and 2. To enhance blood flow, very gentle intermittent pressure may be applied to area surrounding puncture site.
- Allow blood on card to dry thoroughly for a minimum of four hours at room temperature in a horizontal position. Do not allow card to touch anything else while drying. Never superimpose one wet specimen collection paper on another before thoroughly drying. Do not place collection form in plastic bag. Keep away from direct sunlight and heat.
- Submit completed form on day of sampling, but not later than 48 hours after blood collection.

# New Transport System



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Children's Hospital of Eastern Ontario

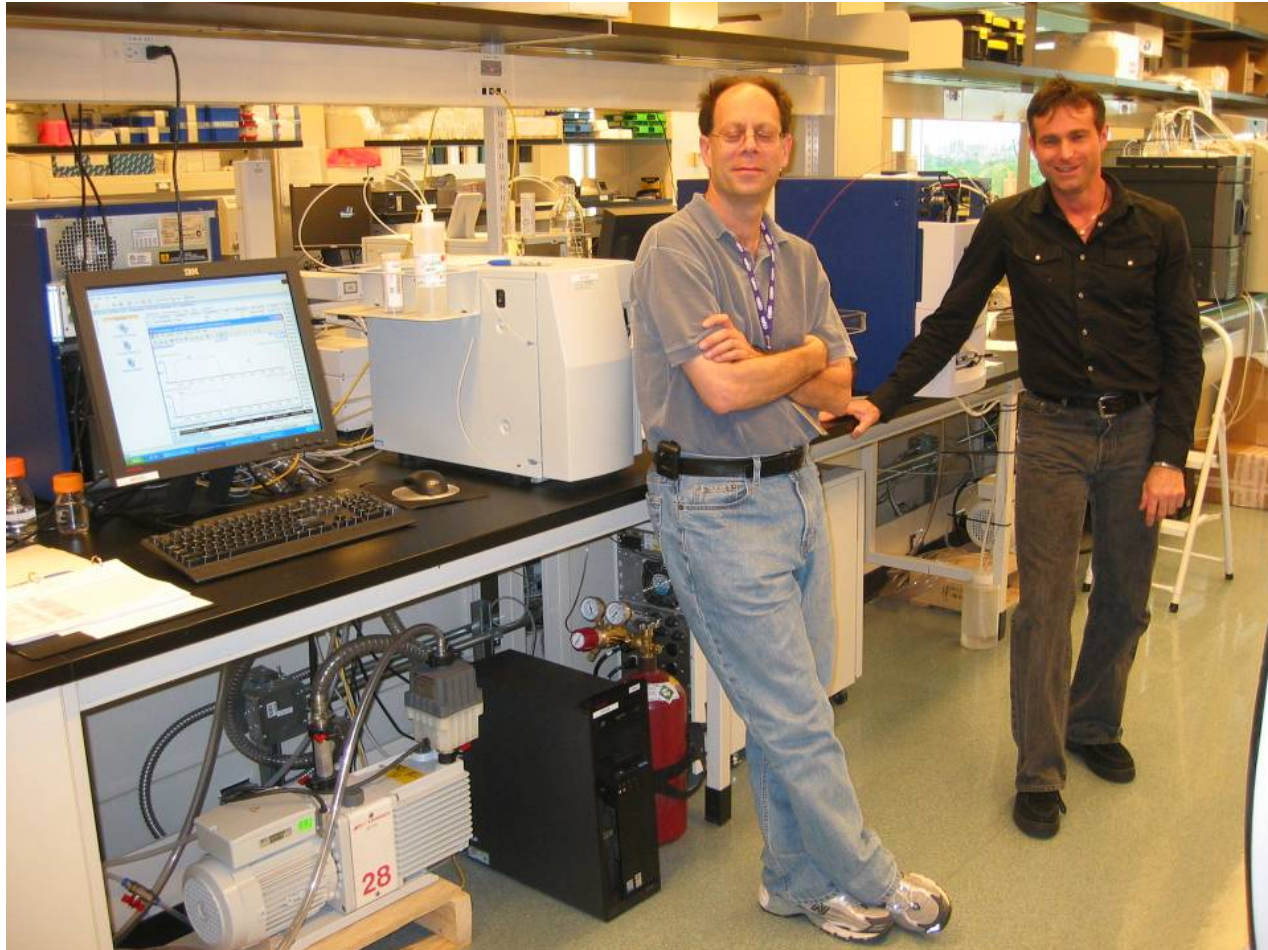
# Same Heelprick Test





# The Process

~2800 samples/week are received and tested in the lab



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Children's Hospital of Eastern Ontario

# Samples

- When samples are received by the laboratory, they are accessioned (barcoded) and inspected to see if they are suitable for testing

We are unable to analyze the sample from your patient for the following reason(s):	Reason:
Quantity of blood insufficient	X
Blood spots appear scratched or abraded	
Blood spots are wet and/or discolored	
Blood spots are supersaturated	
Blood spots appear diluted	
Blood spots exhibit "serum rings"	X
Blood spots appear clotted or layered	
Specimen delivered to lab more than 14 days after collection	
Sample collected at < 24 hours	
Insufficient data provided	

# A small sample of the blood spot is punched for testing



Ontario Newborn Screening Laboratory  
Children's Hospital of Eastern Ontario

All the info from the requisitions is  
entered into a database



Ontario Newborn Screening Laboratory  
Children's Hospital of Eastern Ontario

# Reporting

- Reports on most samples are issued the day after they are received
- Reports can say that the NBS was
  - Negative (majority)
  - Positive
  - Sample unsatisfactory for testing

# Reports

Department of Genetics, CHED  
Room 3127, 401 Smyth Road  
Ottawa, ON, K1H 8L1

Ontario Newborn Screening Laboratory

Tel: (613) 738-3222  
Fax: (613) 738-0853

Health Card Number: UNINSURED  
Mother's Name: SMITH, JANE  
Infant's Health Care Provider: JONES, DR.

D.O.B.: 2008/08/09 10:00 am  
D.O.C.: 2008/08/10 10:01 am  
Date Received: 2008/08/11  
Date Reported: 2008/08/15

SMITH, BABY BOY

ONSL Accession Number: 200808110538

Submitter Unique Number:

Test	Screening Result
<b>Amino Acidurias:</b>	
Phenylketonuria and Variants / Biopren Defects	Negative
Maple Syrup Urine Disease	Negative
Homocystinuria (Hypermethioninemia)	Negative
Citrullinemia / Argininosuccinic Aciduria	Negative
Tyrosinemia	Negative
Amino Acidopathies, other	Negative
<b>Organic Acidurias:</b>	
Propionic / Methylmalonic Acidemias	Negative
Isovaleric Acidemia / 2 Methylbutyric Acidemia	Negative
Glycine Acidemia Type I	Negative
3 Methylcrotonic / Hydroxymethylglutanic / Methylglutaconic / 2-Methyl, 3-Hydroxybutyric Acidemias, or (j) Ketothiolase Deficiency	Negative
Organic Acidurias, other	*
<b>Fatty Acid Oxidation Defects:</b>	
Medium Chain Acyl Dehydrogenase Deficiency / Glutaric Acidemia Type 2	Negative
Very Long Chain Acyl Dehydrogenase Deficiency	Negative
Long Chain Hydroxy Acyl Dehydrogenase / Trifunctional	Negative
Protein Deficiencies	
Carnitine Uptake Defect / CPT I Deficiency	Negative
Fatty Acid Oxidation Disorders, other	Negative
<b>Galactosemia:</b>	
	*
<b>Biotinidase Deficiency:</b>	
	*
<b>Endocrine Disorders:</b>	
Congenital Hypothyroidism	Negative
Congenital Adrenal Hyperplasia	*
<b>Hemoglobinopathies:</b>	
Hb SS, Hb S/C, Hb S/I, Thal	*
Hemoglobinopathies, other	*

1. Screen negative means that this infant is at decreased risk for the disorder(s).
2. Screen positive means that this infant is at increased risk for the disorder(s). It does not mean that a disease is present, but further testing is indicated. If a test is positive and you and / or your patient have not already been contacted, please call the NBS laboratory at (613) 738-3222.
3. \* indicates that the laboratory is not yet testing for the disorder(s).

# Screen Positive Results

- One of five Ontario treatment centres (Metabolics, Hematology, Endocrinology) is notified immediately to arrange for diagnostic testing for the baby
- If baby is truly affected, treatment begins immediately
  - Better outcomes
  - Healthier babies



# To date...

April 3, 2006 to January 31, 2007- Samples received: 121,479

	<b>True Pos.</b>	<b>False Pos.</b>	<b>Results Pending</b>	<b>Total</b>
<b>PKU</b> <sup>1</sup>	10	35	3	48
<b>MCADD</b> <sup>2</sup>	10	6	5	21
<b>CH</b> <sup>3</sup>	45	42 (+22)*	31	140
<b>Amino Acidemias</b>	1	13	9	23
<b>Organic Acidemias</b>	5	38	34	77
<b>FAODs</b> <sup>4</sup>	2 (VLCAD)	14	11	27
<b>Hb-pathies</b> <sup>5</sup>	4	0	6	10

\*22 samples received from babies <24h old. Repeat samples were all negative for CH.

1 Phenylketonuria

2 Medium Chain Acyl-CoA Dehydrogenase Deficiency

3 Congenital Hypothyroidism

4 Fatty Acid Oxidation Defects

5 Hemoglobinopathies: HbSS, HbSC, and HbS/b-thal


-Screening for AAs, OAs, and FAODs began August 8, 2006

-Screening for HbSS, HbSC, and HbS/b-thal began on November 24, 2006

# The Future

- February 19, 2007 (Today!)-  
Galactosemia and Biotinidase deficiency
- Mid 2007- Congenital Adrenal  
Hyperplasia
- Late 2007- Cystic Fibrosis

# Resources

- 
- A vertical decorative strip on the left side of the slide, featuring a series of alternating purple and orange footprints of varying sizes, arranged in a slightly curved path.
- [http://www.health.gov.on.ca/english/public/program/child/screening/fact\\_sheets.html](http://www.health.gov.on.ca/english/public/program/child/screening/fact_sheets.html)
  - [http://www.health.gov.on.ca/english/providers/program/child/screening/screen\\_sum.html](http://www.health.gov.on.ca/english/providers/program/child/screening/screen_sum.html)
  - 613-738-3222 Newborn Screening Office
  - [NewbornScreening@cheo.on.ca](mailto:NewbornScreening@cheo.on.ca)
  - [www.nbs.on.ca](http://www.nbs.on.ca) (Coming Soon!)



# Questions?