

NEWBORN SCREENING **ONTARIO** DÉPISTAGE DES NOUVEAU-NÉS **DE L'ONTARIO** 

### Optional Reporting of Sickle Cell Disease Carrier Results from Newborn Screening the Ontario model

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IDENTIFICATION. PREVENTION. TREATMENT. IDENTIFICATION. PRÉVENTION. TRAITEMENT.

# Overview

- Background on NBS in Ontario
- Background on NBS for hemoglobinopathies in Ontario
- What to do about carrier detection for hemoglobinopathies?
- Formation of Ontario policy
- Implementation of policy
- Current statistics
- Program evaluation
- Successes and challenges to-date

### Newborn Screening in Ontario

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- Large geographic area
- Laboratory and referral activities are centralized in Ottawa
- Screen positives are referred to 1 of 6 treatment centres
- Provincially determined
- Not mandated by law
- Implied consent or informed refusal
- Incidental findings



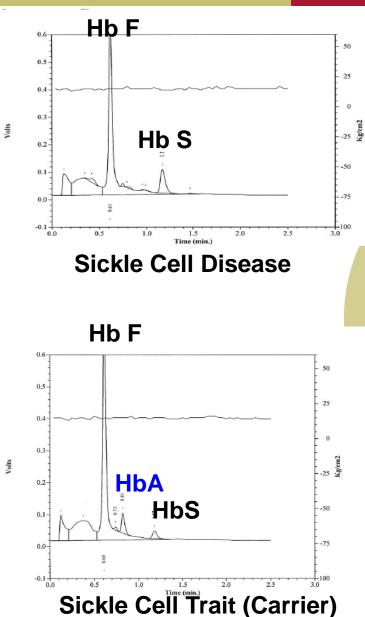
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#### NBS for Hemoglobinopathies in Ontario

- Since November 24, 2006 all newborns in Ontario are screened for:
  - Sickle Cell Anemia (Hb SS)
  - SC disease (Hb SC)
  - Sickle-Beta-thalassemia (Hb S/B-thal)
- 441 screen positive for hemoglobinopathies
  - In 2010: 6.48% of all screen positives
- 236 babies with Sickle Cell Disease
  - ~1/3000 babies
- 36 with another hemoglobinopathy
- Most in Toronto (~80%) or Ottawa (~12%)

### **Carrier Detection**

- HPLC also detects carriers of:
  - Sickle Cell (Hb S)
  - Hemoglobin C (Hb C)
  - Hemoglobin D (Hb D)
  - Hemoglobin E (Hb E)
- Carriers of other hemoglobin variants (including thalassemia) are NOT detected.
- Occasionally carriers of unspecified (and rare) hemoglobin variants are also detected
  - Further testing is needed to confirm which variant



# What to do?

- A study was performed to solicit input from stakeholders in Ontario:
  - Health care providers
  - Health care consumers
- Consensus:
  - Carriers should be reported
  - Parental choice to receive results
  - Access to follow-up if desired

*J Med Ethics* 2009 35: 626-634 *Eur J Hum Genet* 2010 18: 303-308

### Policy: Positive Disclosure Model

- Parents/guardians have a choice about whether or not to receive the carrier result on their child
- Parents/guardians must actively request the result
   NSO will not automatically report carrier results
- A separate report and information package for parents and HCP will be generated
- Carrier reporting program had to be implemented in a way that was effective, efficient and affordable
- Implemented November 1, 2010

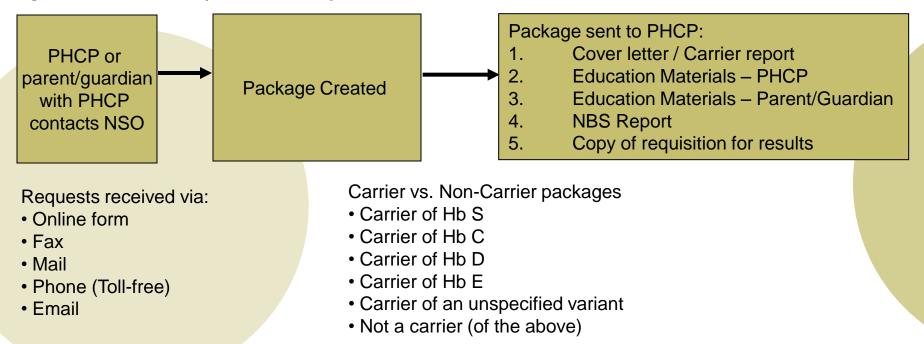
### **Hb** Carrier Reporting Initiative

#### **Major Components**

- Educational Initiatives
  - To raise awareness of availability of carrier results amongst:
    - General Public/Parents/Guardians
    - Prenatal Educators
    - Submitters/HCPs performing NBS
    - Primary Health Care Providers: Family Physicians, Pediatricians, Nurse Practitioners, Midwives, Public Health Nurses
- NSO Internal Procedures
  - Laboratory workflow and workflow in Patient Care
  - Release of Results
  - Communication with families and HCPs
- Engaging Support of Advocacy Groups

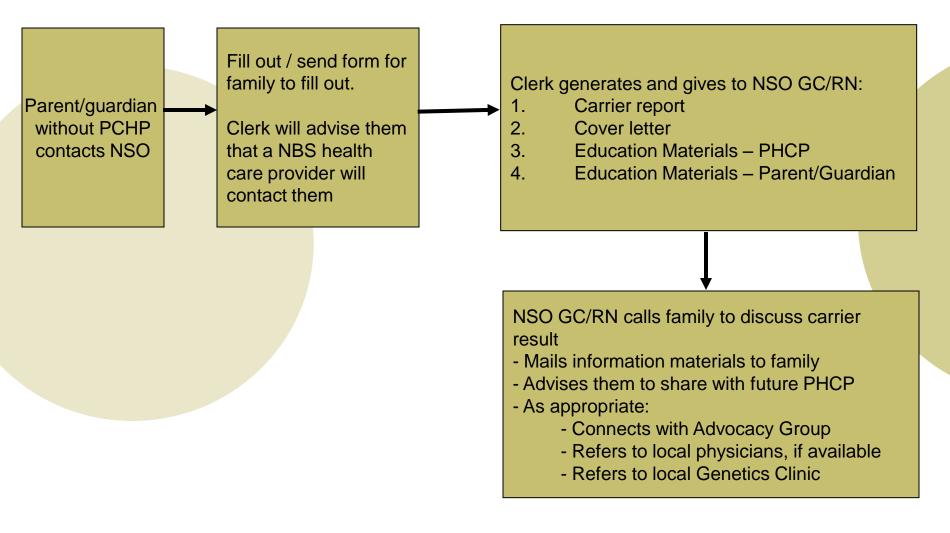
### **Carrier Reporting Model**

# Stream 1: Family with a primary health care provider (PHCP)



### **Carrier Reporting Model**

#### Stream 2: Family without a PHCP



#### **Educational Materials**

- Development of "key messages"
- Modification to existing materials:
  - Parent NBS Information Sheet
  - Newborn Screening Report
- Creation of new materials
  - Requisition form
  - Parent Information Sheets
  - Health Care Provider Information Sheets

### Parent NBS Information Sheet

NEWBORN SCREENING ONTARIO DÉPISTAGE DES NOUVEAU-NÉS DE L'ONTARIO A123456

NEWBORN SCREENING ONTARIO DÉPISTAGE DES NOUVEAU-NÉ A123456

Children's Hospital of Eastern Ontario Centre hospitalier pour les enfants de l'est de l'Ontario 401 Smyth Road, Ottawa, ON K1H 8L1

#### PARENT INFORMAT Is anything else detected through the screening test?

A blood sample has been can cause mental retardat Ontario (NSO) will report s sent the sample.

A "screen positive" result and NSO physicians will r up testing.

A "screen negative" resul currently screened for is v If your baby's sample is ta practitioner or your baby's a repeat sample is needed who submitted the sample If you have any questions doctor, nurse practitioner, Screening: A healthy start www.newbornscreening.o

Turn page over

Screening also detects babies who are carriers of sickle cell and some other red blood cell diseases. Babies who are carriers of these diseases are not more likely to get sick than any other baby. As of November 1<sup>st</sup>, 2010 carrier results will be available by request. More information about sickle cell carriers and how to obtain your baby's carrier result is available on the NSO website and/or talk to your baby's health care provider.

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#### FEUILLET D'INFORMATION POUR LES

Un échantillon sanguin a été prélevé chez votre bé graves qui peuvent causer l'arriération mentale, la elle ne sont pas traitées. Dépistane des pouveau

as possible. In some cases this can be life-saving while in others, serious effects of the diseases like mental retardation or growth problems can be avoided.

#### Is anything else detected through the screening test?

Screening also detects babies who are carriers of sickle cell and some other red blood cell diseases. Babies who are carriers of these diseases are not more likely to get sick than any other baby. As of November 1st, 2010 carrier results will be available by request. More information about sickle cell carriers and how to obtain your baby's carrier result is available on the NSO website and/or talk to your baby's health care provider.

#### How do I find out the results?

All results are mailed to the hospital or health care provider that did the test. The NSO does not release results directly to parents or guardians. If you would like a copy of your baby's results, please ask your health care provider or the hospital where your baby was born.

#### Can I decline newborn screening for my baby?

Newborn screening is not mandatory in Ontario. However, it is considered standard of care for babies and is strongly recommended. Newborn screening is designed to identify serious diseases in babies at a young age so that treatment can be started early in life. Early diagnosis and treatment helps to provide the best possible outcomes for babies with these diseases. Any decision to decline screening should first be discussed with your baby's health care provider.

#### Protection de vos renseignements personnels et votre confidentialité

Dépistage des nouveau-nés de l'Ontario (NSO) est administré par le Centre Hospitalier pour Enfants de l'Est de l'Ontario (CHEO) sous la direction du ministère de la Santé et des Soins de longue durée. NSO s'est engagé à fournir le meilleur dépistage possible

#### Newborn Screening Report

- Concern that the "Negative" or "Positive" may be incorrectly thought to include carrier status
- Additional footnote:
- "This report does NOT contain information about this newborn's carrier status for the hemoglobinopathies. Families wishing to learn their child's carrier result should ask their primary health care provider to contact the NSO or visit www.newbornscreening.on.ca"

#### **New NSO Materials**

- Parent Guardian Materials
  - General information section of NSO website
  - Do I Want to Know My Child's Carrier Status
  - Sickle Cell Carrier Pamphlet
  - Hemoglobin C Carrier Pamphlet
  - Hemoglobin D Carrier Pamphlet
  - Hemoglobin E Carrier Pamphlet
  - Not a HbS, C, D, E Carrier Pamphlet
- Health Care Provider Materials
  - General information section of NSO website
  - Pros and Cons of Learning Carrier Results in Childhood
  - Carrier pamphlets for each variant

#### "Advertising"

- Information packages sent to all family physicians, pediatricians & midwives in Ontario
  - 13,000 packages!
- Memos sent to all hematologists, genetics clinics, public health units, OBs
- Telehealth for hospitals who submit NBS samples
- Publication/announcements Ontario Medical Association, NP newsletter, Canadian Association of Genetic Counsellors newsletter
- Advocacy group engagement visit to CHEO Oct 15, 2010

#### **Program Evaluation**

- Is the program working well?
  - For families?
  - For primary care providers?
  - For NSO?

#### **Outcome Measures**

- Five categories of outcome measures:
  - Awareness and understanding
  - Uptake
  - Impact on NSO
  - Impact on stakeholders
  - Impact on health care system

#### **Outcome Measure: Uptake**

- Requests to-date: 58
  - 35 (60.3%) requests from HCPs
    - family physicians, midwives, pediatricians
  - 23 (39.7%) requests from parents
  - 41 (70.7%) checked off "high-risk" group\*
  - <mark>– 50 (86%) Toro</mark>nto area, 4 (7%) Ottawa area
  - 13 carriers reported (Hb S and Hb C carriers)
  - 51 requests from children born in 2010 or 2011

# **Successes and Challenges**

- The "worried-well" have not submitted a lot of request for results
- The majority of requests are from those "at-risk"
- Workflow successfully streamlined
- Multiple request mechanisms
- Number of requests much less than anticipated
- Mechanisms to inform at-risk groups
- Engagement of advocacy groups
- Adequate education of parents, health care providers and public

#### Acknowledgements

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# Thanks!

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#### All Audiences

- Carrier results for sickle cell and some less common red blood cell diseases can be requested from the NSO via a parent/guardian or PHCP starting November 1, 2010
- Requesting carrier results is optional
- Certain ethnic groups have a higher chance to be carriers than others
- Carriers results will be communicated to a parent/guardian by their PHCP or a NSO health care professional if they do not have a PHCP
- Babies who are carriers of a red blood cell disease are not more likely to get sick than any other baby
- Carrier results may have implications for reproductive decision making
  - For the child in the future
  - For the parents in a future pregnancy

#### **Parents/Guardians**

- Additional Messaging:
  - Obtaining your child's carrier result is NOT urgent visit the NSO website to learn more information and discuss with your PHCP at a regular clinic appointment
  - You and/or your partner may be a carrier of a Hemoglobin Disease even if your baby is not – discuss the option of carrier testing for yourself with your PHCP. There are ways to learn if you are at risk to have a child with a Hemoglobin Disease without learning your baby's carrier status
  - Carriers of certain Hemoglobin Diseases (for example: Thalassemia) are not detected on the newborn screen - discuss the option of additional carrier testing for your child with your PHCP

#### **Prenatal Educators**

- Additional Messaging:
  - Starting November 1, 2010 carrier results for certain hemoglobinopathies will be available from the NSO by request

#### Submitters/HCPs Performing the NBS

- Additional Messaging:
  - Carrier results for the hemoglobinopathies will
    NOT be included on the newborn screening report
  - NSO will not automatically report carrier results
  - Submitters should be familiar with the process by which a family/PHCP can obtain a carrier result

#### **Primary Health Care Providers**

- Additional Messaging:
- Carrier results for the hemoglobinopathies will NOT be included on the newborn screening report
- Families may approach them to learn their child's carrier result
- They should be familiar with the pros and cons of learning a child's carrier result in infancy
- They should be familiar with how a parent/guardian can request a child's carrier result from the NSO
- They should be familiar with how to request a child's carrier result from the NSO on behalf of a family
- There are educational resources available for them and their patients through the NSO
- Local advocacy groups are a source of additional support and information for their patients