



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.
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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

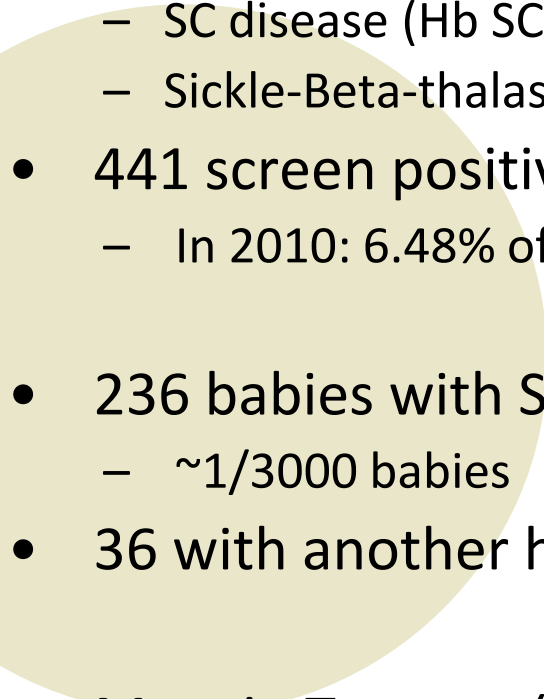

Newborn Screening in Ontario

- 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



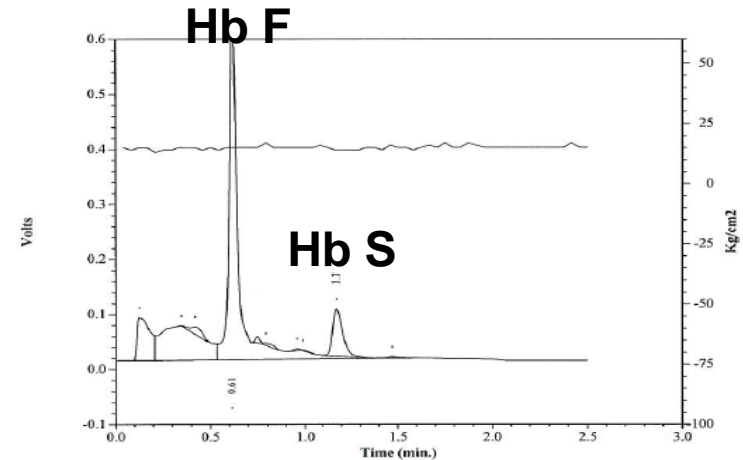
NBS for Hemoglobinopathies in Ontario



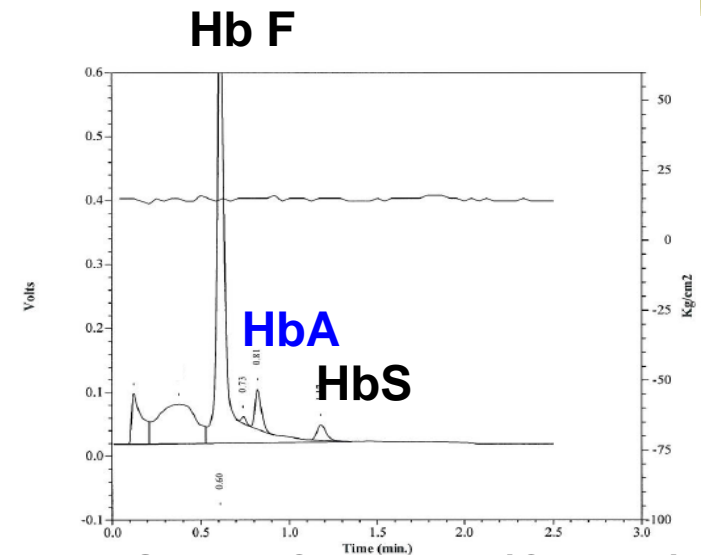
- Since November 24, 2006 all newborns in Ontario are screened for:
 - Sickle Cell Anemia (Hb SS)
 - SC disease (Hb SC)
 - Sickle-Beta-thalassemia (Hb S/B-thal)
 - 441 screen positive for hemoglobinopathies
 - In 2010: 6.48% of all screen positives
 - 236 babies with Sickle Cell Disease
 - ~1/3000 babies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



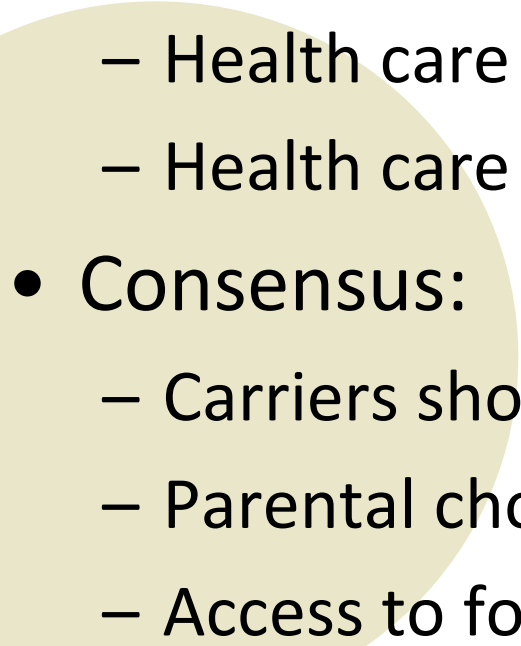

Sickle Cell Disease



Sickle Cell Trait (Carrier)

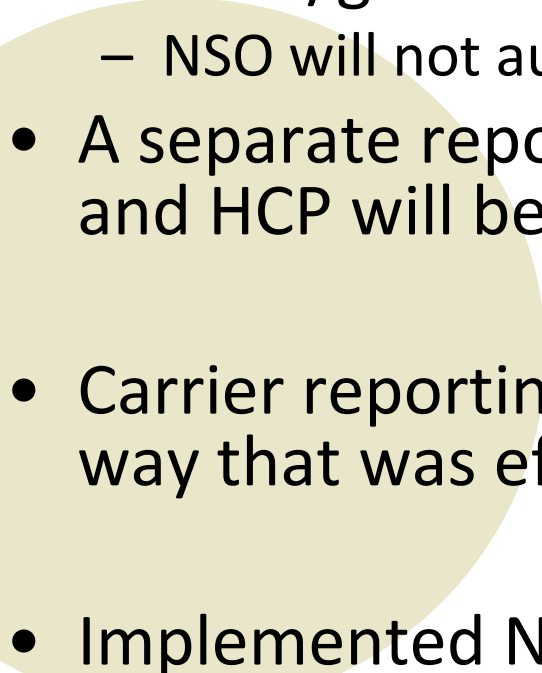

What to do?



- 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
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Policy: Positive Disclosure Model

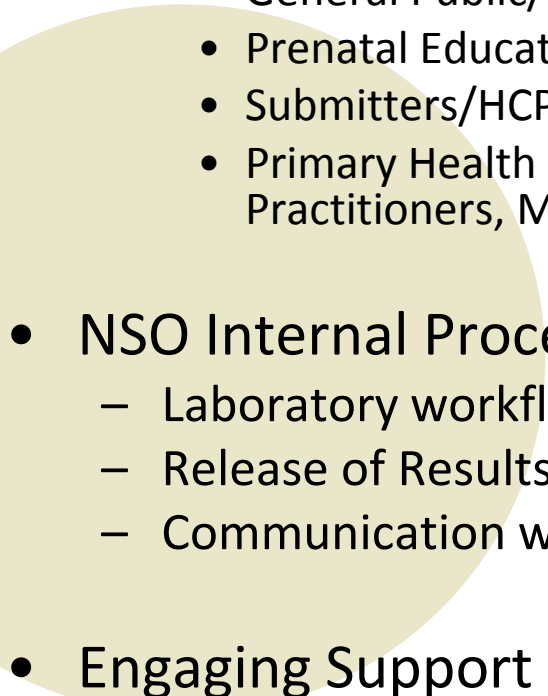



- Parents/guardians have a choice about whether or not to receive the carrier result on their child
 - Parents/guardians must actively request the result
 - NSO will not automatically report carrier results
 - A separate report and information package for parents and HCP will be generated
 - Carrier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

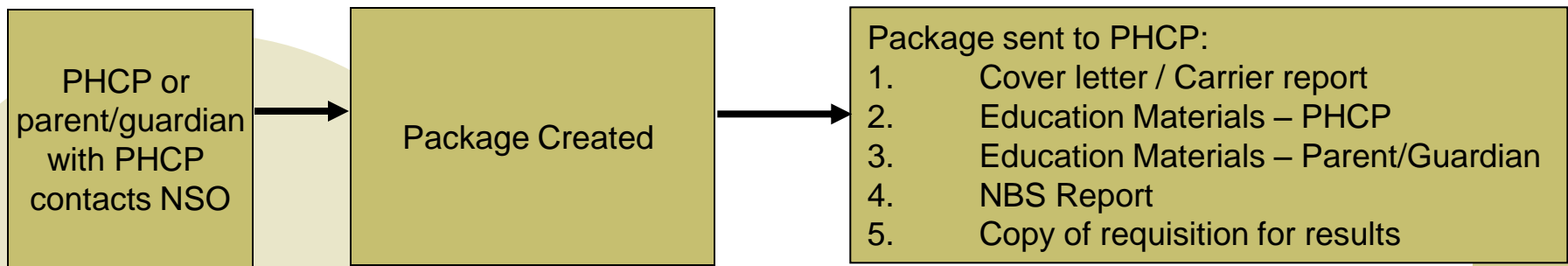


Major Components

- 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
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Carrier Reporting Model

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



Requests received via:

- Online form
- Fax
- Mail
- Phone (Toll-free)
- Email

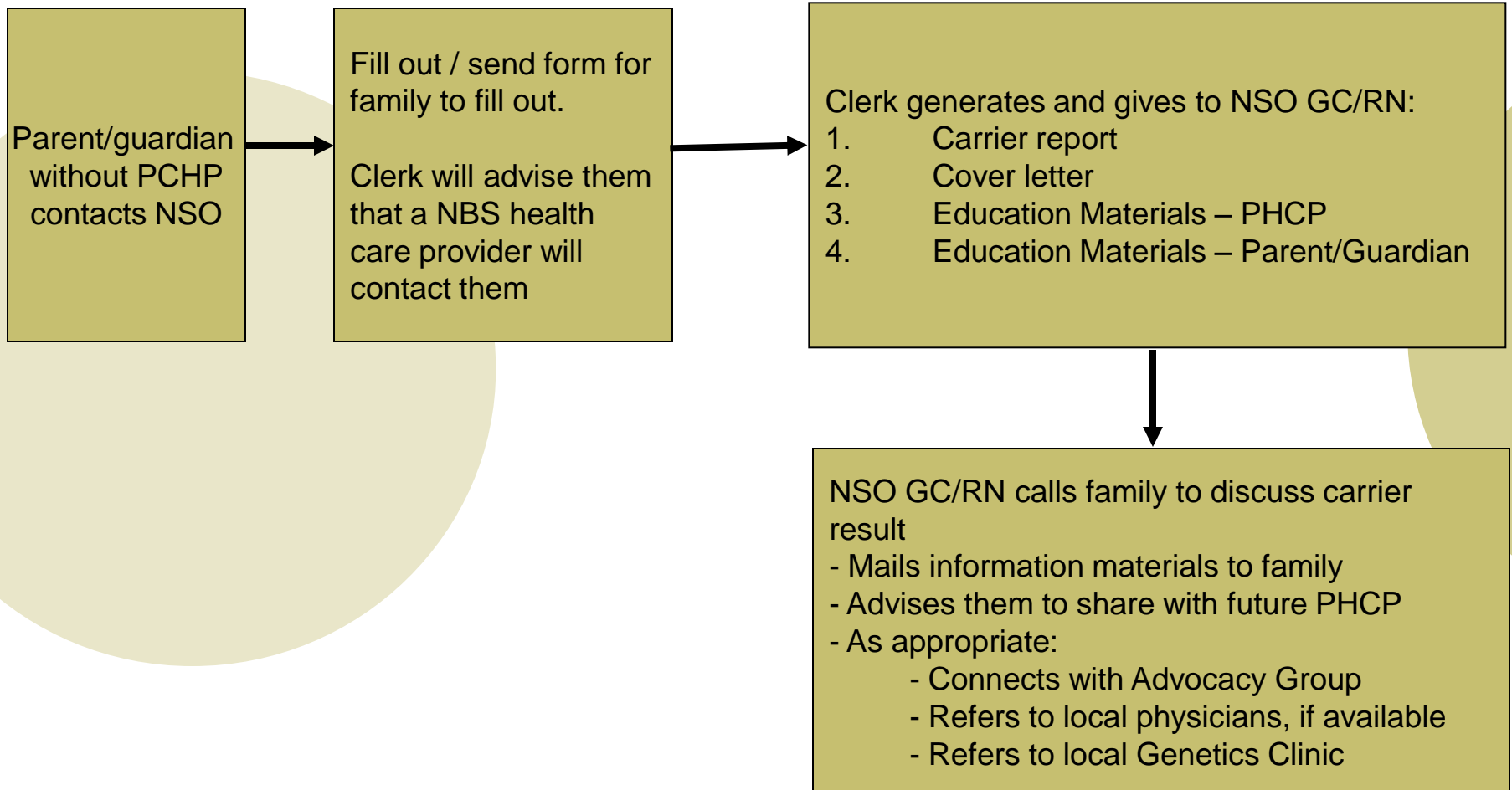
Carrier vs. Non-Carrier packages

- Carrier of Hb S
- Carrier of Hb C
- Carrier of Hb D
- Carrier of Hb E
- Carrier of an unspecified variant
- Not a carrier (of the above)

Carrier Reporting Model

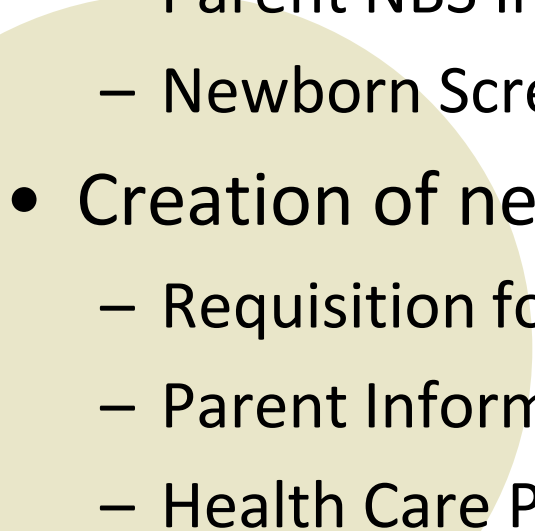



Stream 2: Family without a PHCP



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
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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”

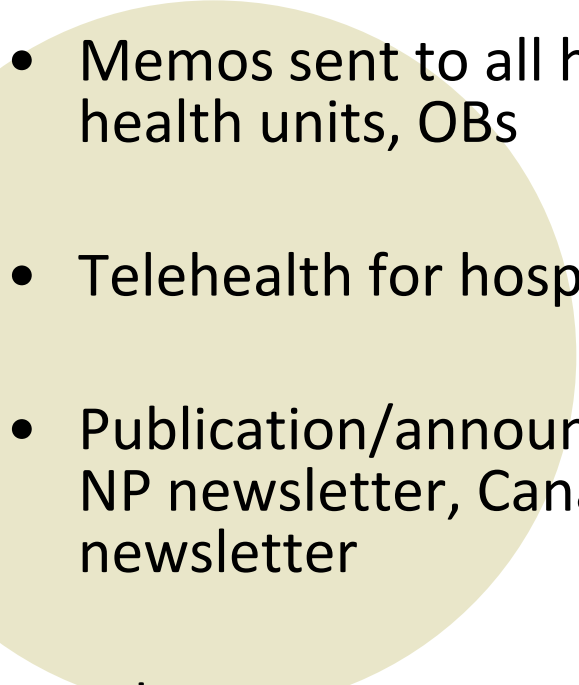

New NSO Materials



- 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”



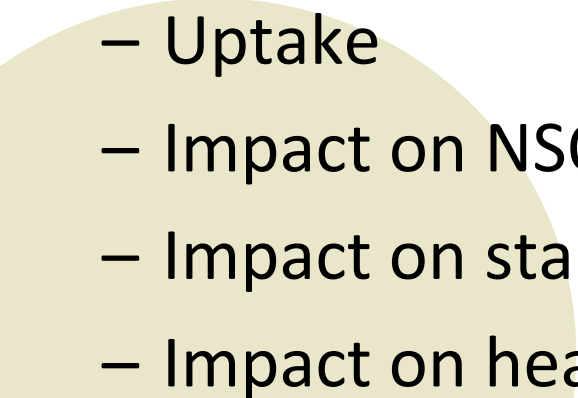

- Information packages sent to all family physicians, pediatricians & midwives in Ontario
 - 13,000 packages!
 - Memos sent to all hematologists, genetics clinics, public health units, OBs
 - Telehealth for hospitals who submit NBS samples
 - Publication/announcements – Ontario Medical Association, NP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

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

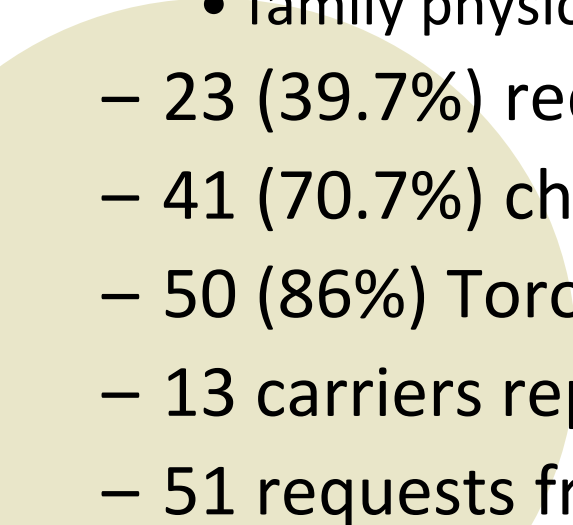

Outcome Measures



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

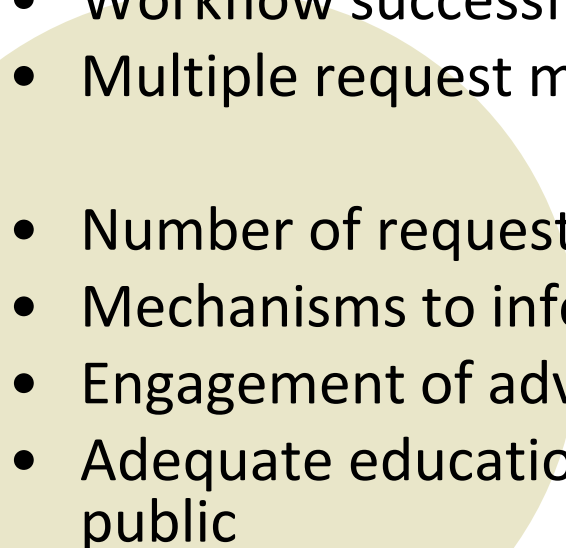



- Requests to-date: 58
 - 35 (60.3%) requests from HCPs
 - family physicians, midwives, pediatricians
 - 23 (39.7%) requests from parents
 - 41 (70.7%) checked off “high-risk” group*
 - 50 (86%) Toronto area, 4 (7%) Ottawa area
 - 13 carriers reported (Hb S and Hb C carriers)
 - 51 requests from children born in 2010 or 2011
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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
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Acknowledgements



- NSO Working Group
 - Dr. Pranesh Chakraborty
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 - Dr. Beth Potter (University of Ottawa)
 - Christine Chretien (CHEO)
 - Denise Labrecque (CHEO)
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Thanks!

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Key Messages

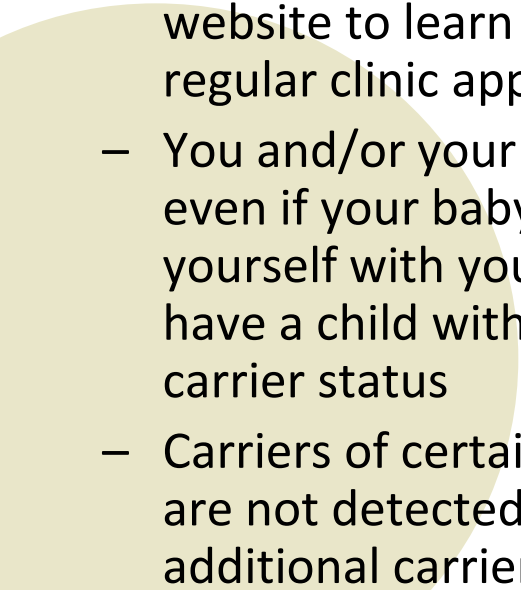

All Audiences

- 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
- Carrier 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

Key Messages



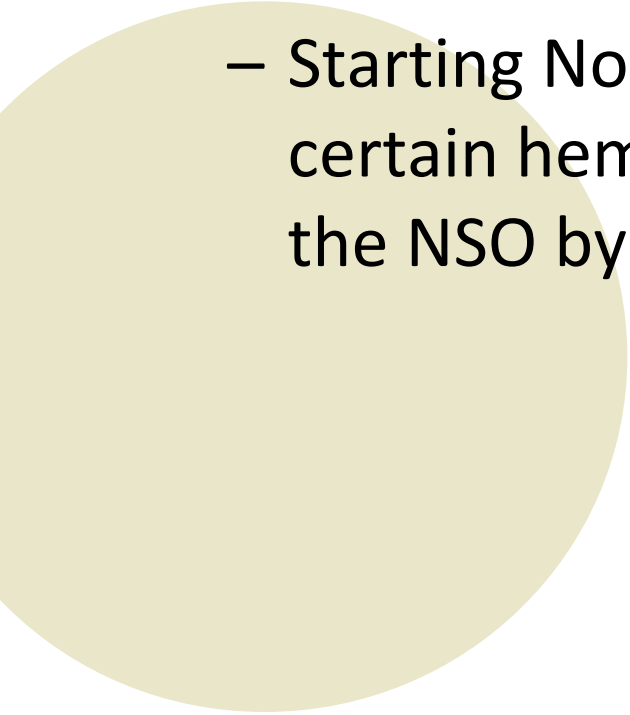

Parents/Guardians

- 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
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Key Messages



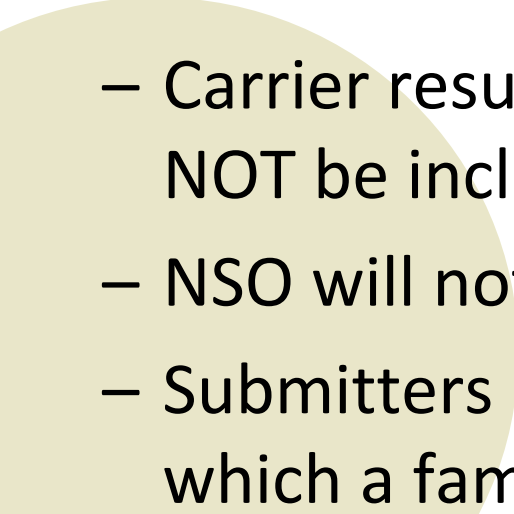

Prenatal Educators

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



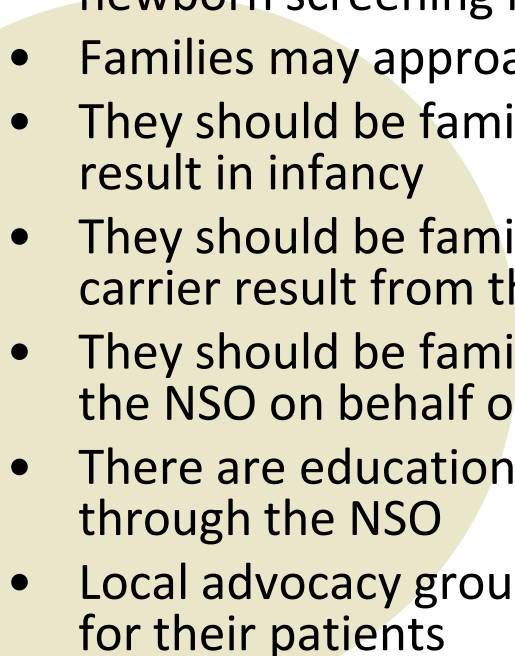
Submitters/HCPs Performing the NBS

- 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
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Key Messages



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
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