NEWBORN SCREENING **ONTARIO**

DÉPISTAGE NÉONATAL **ONTARIO**

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Newborn Screening Ontario Annual Public Report

Calendar Year 2012

Contents

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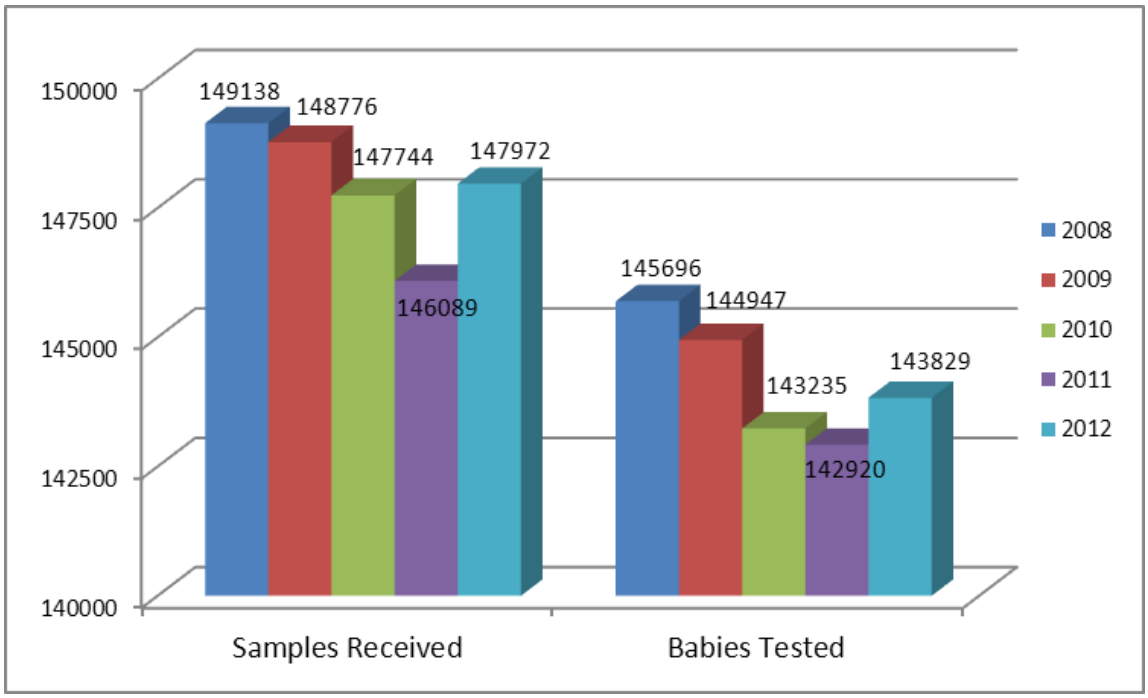
1. Total Samples and Babies Screened

Total Samples Received: 147,972
Estimated Babies Screened: 143,829



The total number of samples received includes newborn screening samples, monitoring and diagnostic samples, and coroner requests. The number of infants tested assumes that the program's linking and matching algorithms are correct, therefore, this number is an estimate.

The number of infants tested is always lower than the number of samples received due to repeats required for transfusion, and laboratory and data unsatisfactory samples. The unsatisfactory rate improved from 3% to 2%, resulting in a decrease in the number of samples received in 2011 as compared to 2010 with a relatively small change in the number of infants tested. However it rose to 2.5% in 2012. In 2012 NSO saw the highest rate in the number of repeat samples received which may be part of the explanation as to why the total number of samples received has increased.

The overall number of infants tested appears to have been going down over time. However, there was no way to know if this was truly the case or if it was due to changes in NSO's linking

algorithm for babies tested for repeat samples, an overall decrease in the birth rate for Ontario, or to missed/declined screens. This is one area where BORN has and will continue to be of great value providing accurate birth rates, and helping NSO to track missed and declined cases. The increase in number of babies tested and samples received in 2012 cannot be fully explained but it is speculated that it could be due to the introduction of BORN – entering all birth encounters and capturing whether or not a baby had a NBS may have increased tracking within hospitals prior to a missed screen notification.

2. Demographics of Screening Samples

2.1 Age at Collection

This table represents the age at collection for 2012 initial samples only.

Age at collection	Number of Samples	% of total samples
Less than 24 hours	690	0.48%
24-47 hours	109,891	76.56%
48-72 hours	20,450	14.25%
1-7 days	142,029	98.95%
Greater than 7 days	813	0.57%

Distribution of Age for initial samples	Hours (Days)				
	2012	2011	2010	2009	2008
Mode	25 (1)	25 (1)	25 (1)	25 (1)	25 (1)
95th percentile	93 (3.9)	93 (3.9)	94 (3.9)	93 (3.9)	89 (3.7)
99th percentile	143 (6.0)	143 (6.0)	148 (6.2)	158 (6.6)	161 (6.7)

2.2 Transfusion Status

Transfused babies should have a repeat sample collected at 4-6 months post transfusion. In the **calendar year** of 2012, 235 (0.16%) initial samples indicated that the baby was transfused prior to the sample being taken. This is lower than in 2011 where 286 samples indicated that the infant was transfused. Overall, of the samples where a repeat sample could be expected, 45% of these have completed screening with a repeat sample. While the percentage of repeats received is lower than last year, a number of cases from 2012 are still open.

Samples received between 4-6 months are sent to NSO without a reminder having been sent to the submitter (ie the submitter has their own tracking system in place). At 6 months submitters receive a reminder by fax that a repeat NBS is required. If the submitter responds to the fax that a health care provider (HCP) has been notified, NSO also sends a letter to the HCP. At 12 months, the case is closed with a close case letter to the submitter (and HCP if indicated).

Year	Samples without a repeat				Cases still open	Samples with a repeat			Total	% repeat received (of total required)
	No repeat	Deceased	Repeat not required	Not transfused		4-6 mo	6-12 mo	>12 mo		
2011	92	37	31	19	0	20	74	13	286	54%
2012	57	21	20	12	43	13	68	1	235	45%

Repeat not required = cases where an initial sample was not transfused but a second sample was received indicating transfused. As screening had already been adequately completed, a third sample in 4-6 months was not required.

Not transfused = transfusion status was written as 'yes' on the NBS requisition but the program was subsequently notified that the infant either did not have a transfusion or had a transfusion other than a packed red blood cell transfusion.

Year	Quarter	Repeat not received	Repeat not expected	Repeat Received			Case still open	Total	% repeat received (of total required)
				4-6 mo	6-12 mo	>12 mo			
2011	Jan - Mar	22	11	3	21	2	0	59	54%
	Apr - Jun	29	35	7	18	3	0	92	49%
	Jul - Sep	20	21	4	21	6	0	72	61%
	Oct - Dec	21	20	6	14	2	0	63	51%
2012	Jan - Mar	26	32	5	19	0	0	82	48%
	Apr - Jun	22	16	4	21	1	0	64	54%
	Jul - Sep	8	0	0	18	0	16	42	43%
	Oct - Dec	1	5	4	10	0	27	47	33%

Note: Repeat not expected encompasses infants who are deceased, where the transfusion status was incorrectly identified as 'yes', or where an initial sample was normal negating the need for a repeat NBS.

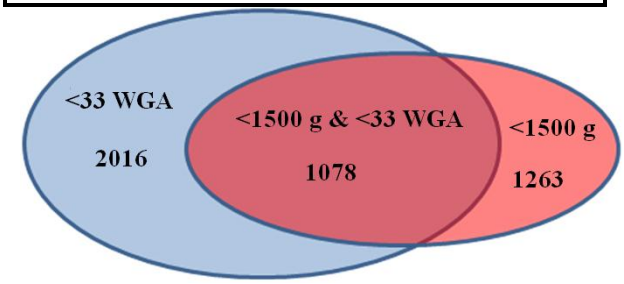
2.3 Gestational Age and Birth Weight

In calendar year 2012, **10,950 babies (7.6%)** were identified as being premature (<37 WGA).

Gestational Age (ww.d)	Number of babies	% of population
<25	86	0.06
25-27.6	363	0.25
28-32.6	1567	1.09
33-36.6	8934	6.24
Total	**10950**	**7.63**

Birth Weight (g)	Number of babies	% of population
<1000	439	0.31
1000-1499	824	0.58
1500-2499	7686	5.37
>2500	132403	92.42
Unknown	1962	1.37

NSO has developed a policy for repeat testing in premature babies which recommends repeat testing for babies <33 weeks gestation or very low birth weight (<1500g) at time of screening. Therefore an increase of approximately 2200 samples (the number of babies <1500g OR <33 weeks) may be expected with this policy.



3. Unsatisfactory Samples

There were a total of **3681** unsatisfactory samples in calendar year 2012, and some samples were unsatisfactory for more than one reason. On page 5 is a table summarizing the most frequent unsatisfactory reasons, as well as the distribution throughout the year.

Overall the unsatisfactory rate in 2012 was 2.11% (excluding samples that are collected at <24 hours as this is recommended in the case of early discharge). This is higher than the rate for calendar year 2011 of 1.51%. This increase results in 754 more infants than last year requiring follow up and repeated samples.

It is speculated that the unsatisfactory rate inversely correlates with the number of submitter workshops in the preceding year. In 2012 there were 3 submitter workshops (Toronto area and Ottawa). Workshops are planned for Toronto area, Thunder Bay, Sudbury, and Woodstock

in 2013. NSO is also in the process of developing a submitter handbook which will be published in 2013.

3.1 Quantity of Blood

The two main reasons that samples are deemed unsuitable for testing both involve having the right quantity of blood on the filter paper; supersaturated or insufficient quantity of blood. The newborn screening assays are validated under the assumption of an equal distribution of blood through the filter paper. It is estimated that 75 uL - 100 uL of blood is required to fill one circle on the filter paper. If there is too little blood applied to the card, the unsatisfactory sample will be coded as "Quantity of blood insufficient". If too much blood is applied to the card, the sample will be coded as "Blood spots are supersaturated".

Quantity of Blood Insufficient

There was an increase in the number of samples deemed unsatisfactory due to insufficient quantity of blood (up from 0.61% in 2011 to 0.85% in 2012). However these rates are both lower than the 2010 rate of 1.14%. Educational materials target this issue as it is the most common cause or contributing factor for unsatisfactory samples.

Blood Spots are Supersaturated

Supersaturated samples have also increased over the last year from 0.56% in 2011 to 0.82% in 2012, and account for over a quarter of unsatisfactory samples received. Increased education for submitters about this reason is warranted as it has been the second most common reason for sample rejection for the last two years.

3.2 Scratched or Abraded Samples

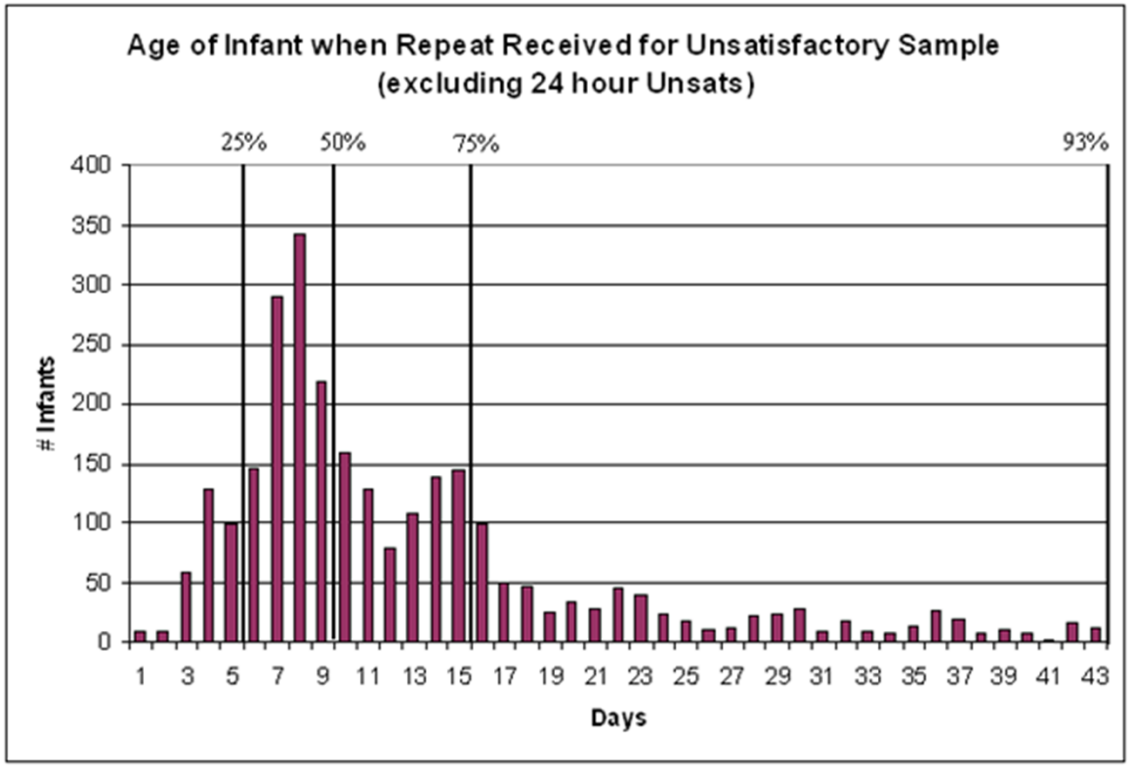
Although this had improved greatly in 2011, there was an increase in the number of samples that appear scratched or abraded. In 2010 it was noticed that a large number of samples were scratched or abraded so investigations were made into the cause. When the samples were applied to paper with capillary tubes there appeared to be an increase in this category of unsatisfactory samples. NSO training documents indicate that capillary tubes should not be used for the application of blood, and if used should not make contact with the filter paper.

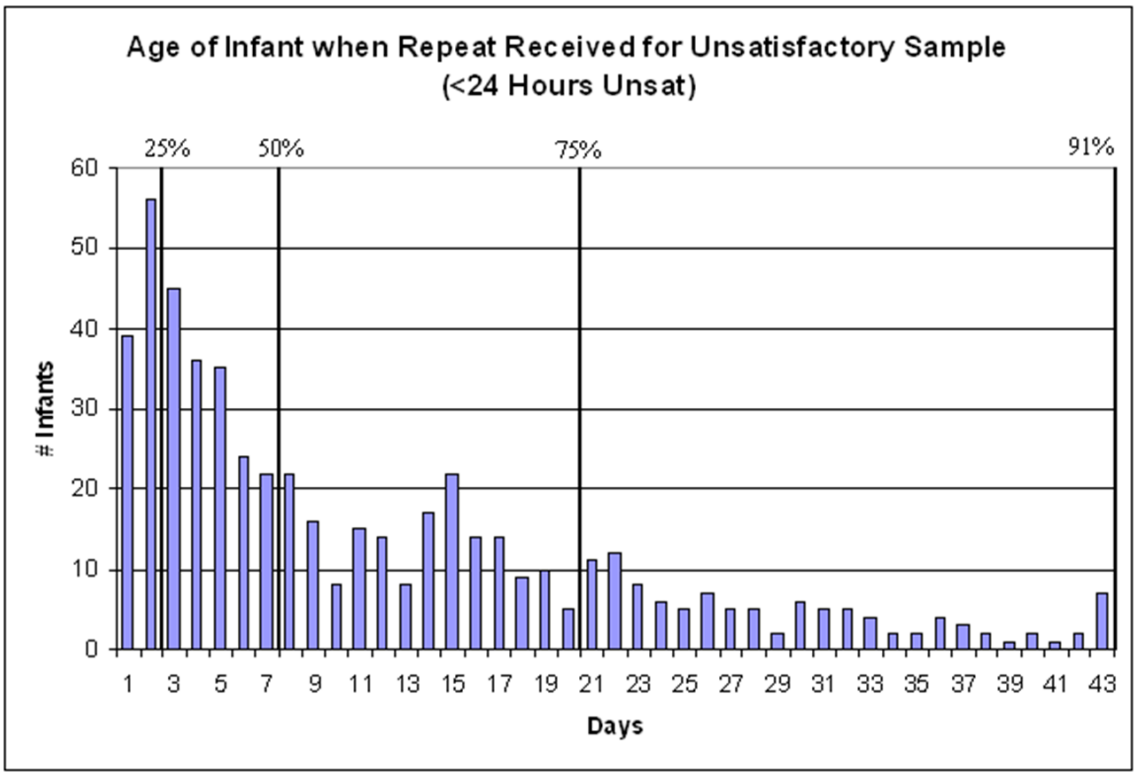
Quarter	Unsat Samples	Samples collected at <24 hrs of age	% unsats excluding <24 hrs	Lab unsats (most common)				Data unsats			Other *
				Quantity of blood insufficient	Blood spots are super-saturated	Blood spots appear scratched or abraded	Blood spots appear clotted or layered	Expired blood spot card	Insufficient data provided	Transport related (delayed, batching)	
Q1 2012	734 (2.13%)	143	**1.71%**	281 (0.81%)	186 (0.54%)	194 (0.56%)	30 (0.09%)	16 (0.06%)	5 (0.01%)	7 (0.02%)	19 (0.06%)
Q2 2012	991 (2.77%)	170	**2.30%**	350 (0.98%)	311 (0.87%)	286 (0.80%)	40 (0.11%)	24 (0.06%)	11 (0.03%)	13 (0.04%)	44 (0.12%)
Q3 2012	950 (2.52%)	181	**2.04%**	265 (0.70%)	347 (0.92%)	279 (0.74%)	29 (0.08%)	16 (0.06%)	5 (0.01%)	12 (0.03%)	18 (0.05%)
Q4 2012	1006 (2.82%)	157	**2.38%**	319 (0.90%)	330 (0.93%)	328 (0.92%)	57 (0.16%)	31 (0.06%)	19 (0.05%)	18 (0.05%)	48 (0.13%)
2012	3681 (2.56%) ↑	651 ↓	**2.11%** ↑	1215 (0.85%) ↑	1174 (0.82%) ↑	1087 (0.76%) ↑	156 (0.11%) ↓	87 (0.06%) ↑	40 (0.03%) ↓	50 (0.03%) ↓	129 (0.09%) ↓
2011	2927 (2.00%) ↓	711 ↓	**1.51%** ↓	888 (0.61%) ↓	819 (0.56%) ↓	619 (0.42%) ↓	184 (0.13%) ↓	70 (0.03%) ↓	60 (0.04%) ↓	157 (0.10%) ↓	173 (0.12%) ↓
2010	4396 (2.97%)	807	**2.42%**	1682 (1.14%)	1092 (0.74%)	1275 (0.86%)	604 (0.41%)	200 (0.12%)	164 (0.11%)	272 (0.18%)	461 (0.31%)

↓ Lower than previous year ↑ Higher than previous year * Other includes: wet and/or discoloured, serum rings, diluted, and Other

3.3 Repeat Rates for Unsatisfactory Specimens

The majority (88.24%) of repeat samples required due to unsatisfactory initial samples are received within 6 weeks of the initial sample. By 3 months, more than 93.97% of unsatisfactory samples have had screening completed via a repeat sample. Repeat samples have not been received for 4.89% of unsatisfactory samples in 2012.





4. Turn Around Times

A number of turnaround times and other quality indicators are monitored to ensure timely and good quality service.

The reasons for using mode, 85th centile, 94th centile are outlined below:

1) The **mode** will primarily reflect samples where at most one weekend interrupts transportation or analysis, and the time at which all tests are completed such that an initial screening determination can be made. For example, a sample which has a screen positive result will have initial results available one working day before the report due to the practice of reanalyzing for confirmation. The mode will reflect better the time at which that initial result is examined for an alert result.

2) The **85th centile** will reflect primarily the turnaround times for samples where at least one weekend interrupts either transportation or analysis, an initial screening result is positive and where analytical QC issues cause a delay in reporting.

3) the **94th centile** will primarily reflect the turnaround times for samples where transportation or analysis is interrupted by a long weekend or by two weekends, while still excluding those initially positive for Cystic Fibrosis where NSO is aiming to introduce a delay in reporting.

Both centiles and the mode will be sensitive to issue such as reporting or data entry delays.

4.1 Transportation Time

Currently the best measurement of transportation time at the sample level is the difference between the date of collection (DOC) and the date the sample is received in the laboratory. Submitting institutions are asked to dry samples for three hours prior to sending via courier to NSO. Most submitters have a scheduled pick up once daily, therefore any samples that are not yet dry and/or packaged for shipment will be delayed by at least 24 hours.

Days in transit (Date of Collection to Receipt of Sample in Laboratory)

Statistic	2012	2011	2010	2009	2008
Average	3.2	3.3	3.4	4.3	4.4
Median	3	3	3	4	4
Mode	2	2	2	4	4
85th Percentile	5	5	5	6	6
94th Percentile	6	6	6	7	7

The reduction in transportation time achieved during 2010 switching to a courier model has been maintained in 2011 and 2012 with Canpar service. With a consistent mode of 2 it is clear that overnight delivery is the norm for the majority of samples.

4.2 Reporting Times

The turn around times from various points to the printing of a full report are described in the tables below. Screen positive infants may be referred prior to the full report being available, due to ongoing testing.

Date of Birth to Report

	2012	2011	2010	2009	2008
Average	8.4	9.1	8.8	10.9	9.0
Median	8	8	7	10	8
Mode	7	7	7	9	7
85th percentile	11	11	11	14	11
94th percentile	13	14	14	17	14

Date of Collection to Report

	2012	2011	2010	2009	2008
Average	6.8	6.5	6.3	8.6	6.9
Median	6	6	6	8	6
Mode	6	6	6	7	6
85th percentile	9	9	8	12	9
94th percentile	11	11	11	14	11

For most infants results are available by the time they are a week old. Logically, the time from collection to report is 1-2 days less than the time from birth to report, since most infants are sampled at 24-48 hours of age. These periods include the time for sampling, transportation, and analysis of the sample, and may be impacted by later sampling, batching of samples at the hospital/midwifery practice, delays in transport, or delays in reporting due to further testing or quality issues.

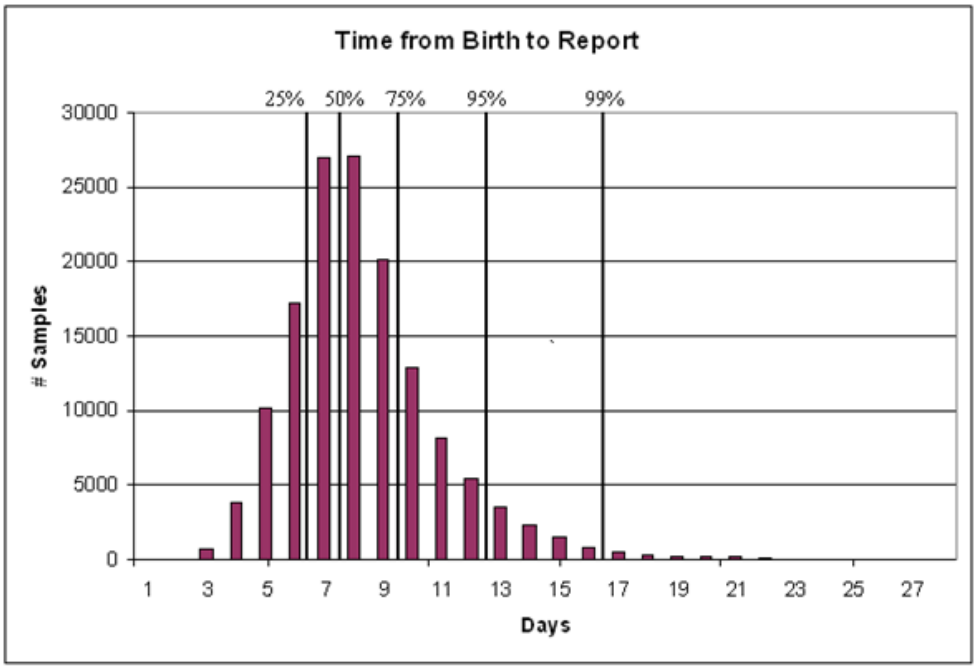
Turn around times have remained generally constant over the last two years, still showing a decrease from 2009 levels which reflects the change in sample transportation services in 2010.

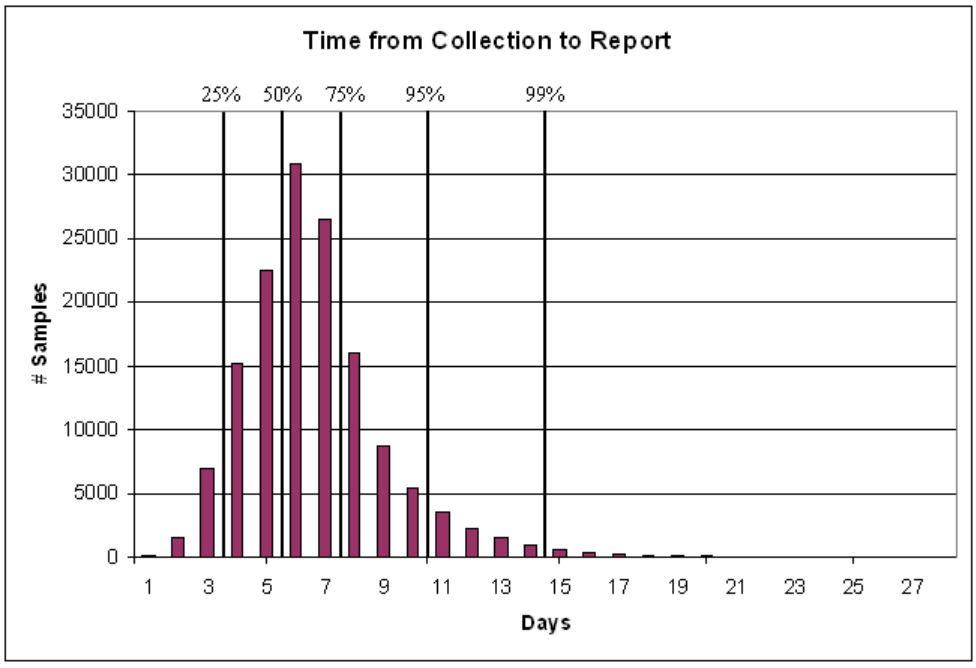
Sample Receipt to Report

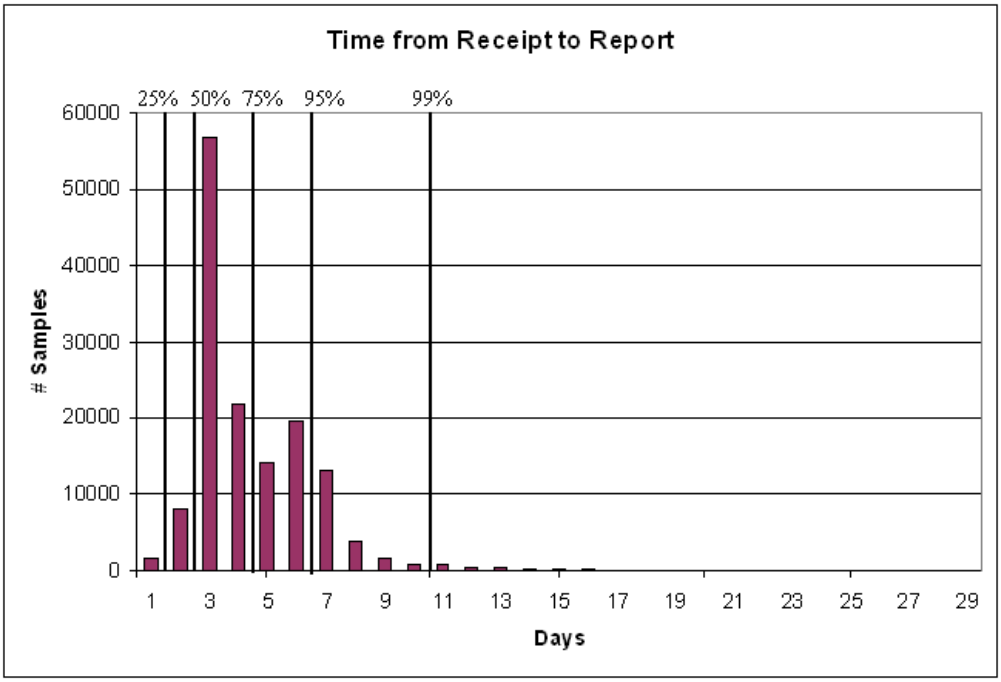
	2012	2011	2010	2009	2008
Average	3.3	3.3	2.9	4.25	2.5
Median	3	2	2	4	2
Mode	3	2	2	2	1
85th percentile	5	5	5	6	4
94th percentile	5	7	6	9	6

Once a sample is received in the laboratory, the demographic entry must be complete and all test results accepted before a report is available for printing. Reports are generated once daily in time for the mail run. Due to the batching of reporting for hemoglobinopathies and cystic fibrosis (these test results are not accepted on a daily basis) there are some delays in printing the reports. The turn around times for reporting have remained constant over the last four years, with the majority of reports available within 2 days of receipt.

Some outliers in turn-around-time reports are due to older children being screened, so for a homogeneous population where a sample was taken within a week of birth, the turn around times are displayed graphically below.


Time from Birth to Report
25%
50%
75%
95%
99%
30000
25000
20000
15000
10000
5000
0
Samples
1
3
5
7
9
11
13
15
17
19
21
23
25
27
Days

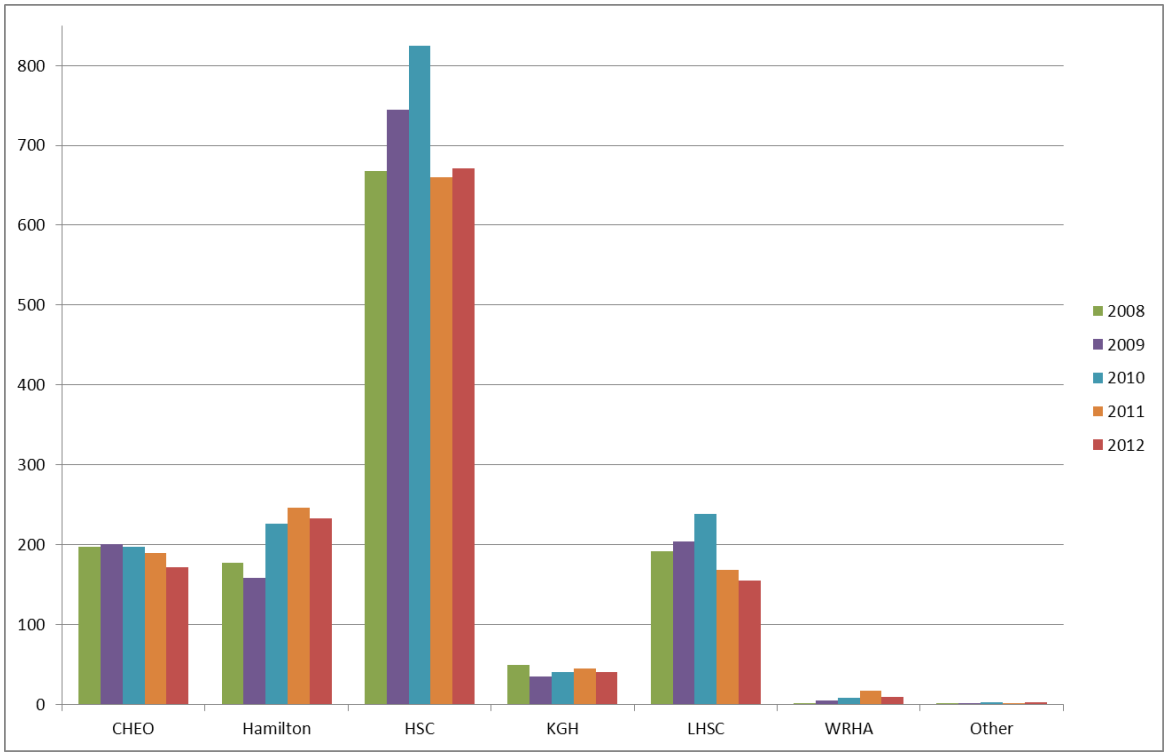

Time from Collection to Report
25%
50%
75%
95%
99%
35000
30000
25000
20000
15000
10000
5000
0
Samples
1
3
5
7
9
11
13
15
17
19
21
23
25
27
Days



5. Screen Positives

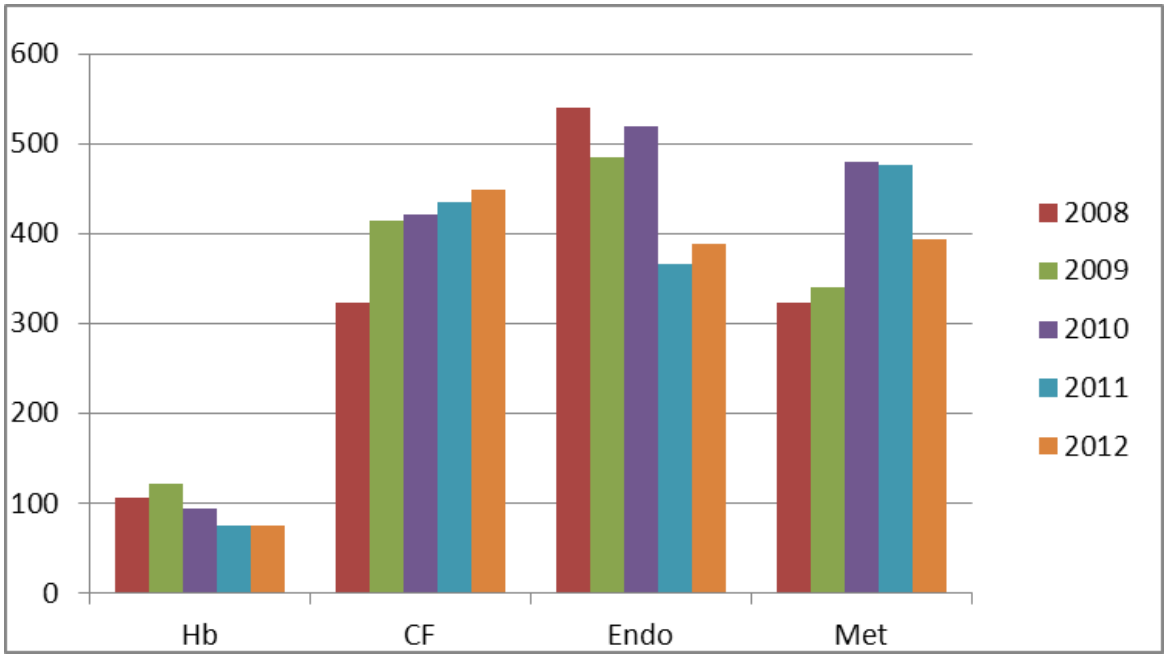
In 2012, there were 1284 screen positive referrals, from a total of 1249 infants. This represents 0.87% of the total number of infants screened by NSO. There were 1307 total screen positives but 23 had an elevated TSH in samples taken at <24 hours which were screen negative on repeat sample testing.

5.1 Referrals by Treatment Centre



The number of referrals over the last 5 calendar years to the five Ontario treatment centres and the Winnipeg treatment centre are depicted in the graph. 'Other' represents babies referred to treatment centres outside of Ontario/ Winnipeg, such as Quebec or USA, or a centre in Ontario that is outside of the standard treatment centres. The Hospital for Sick Children in Toronto receives approximately half of the screen positive referrals. The total number of referrals per treatment centre has mostly decreased in 2012.

5.2 Referrals by Disorder



Hemoglobinopathies
The algorithm for reporting changed in October 2010 where FAX hemoglobin patterns (patterns of unknown significance) were no longer reported. Therefore, 2011 is the first complete year of data with this reporting algorithm. As anticipated, the 2012 data was very similar.

Cystic Fibrosis
The number of referrals in 2012 was relatively consistent with previous years, with just over 400 referrals per year.

Congenital Adrenal Hyperplasia
The algorithm was modified in December 2010 with the addition of steroid profiling. With this addition, there was a significant reduction in the referrals from 2010 to 2011. In August 2012 the algorithm changed again when NSO stopped reporting CAH screen positives which were critically positive on automated immunoassay but negative on steroid profiling. The number of referrals for 2013 is expected to decrease as a result.

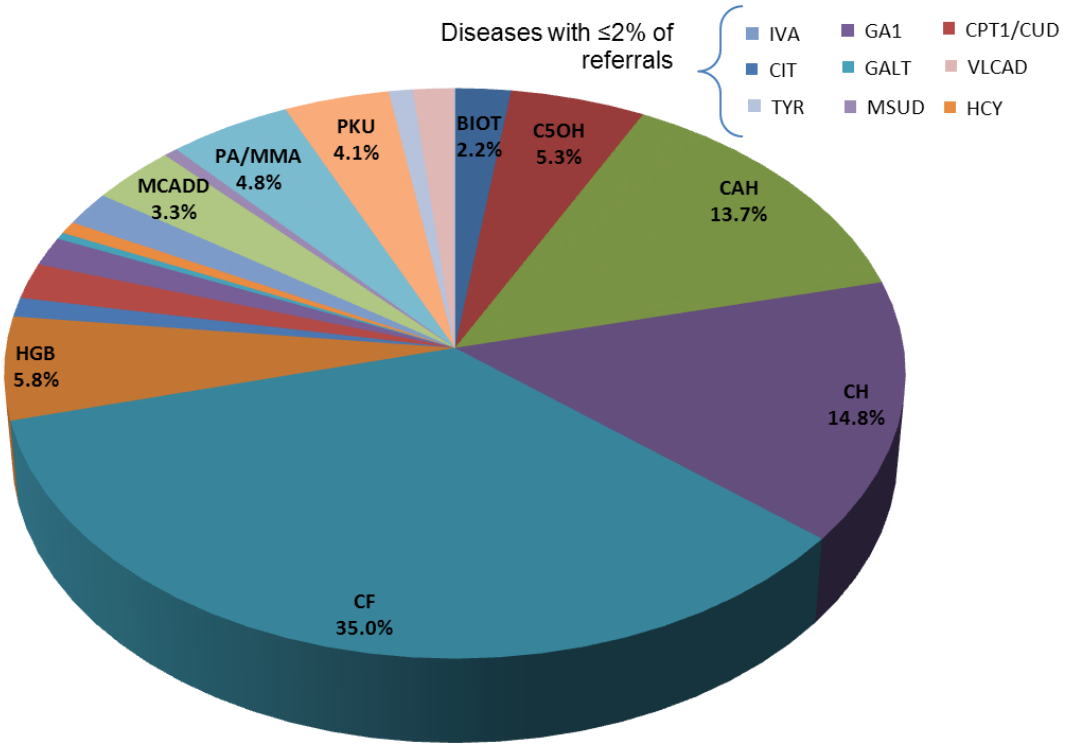
Congenital hypothyroidism
The referral rate for this condition decreased in 2012.

Metabolics
The majority of metabolic conditions had a decrease in number of referrals in 2012.

MSUD and Tyrosinemia had disorder logic changes toward the end of 2011. This resulted in a significant reduction in referrals in 2012.

The number of IVA referrals has gone up in 2012 but unlike previous years, ~50% of referrals were sent to Toronto (which would be expected based on their referral rate). Hamilton's referral rate for IVA has dropped significantly. In 2010 and 2011 it was thought that the use of pivalic acid at McMaster caused a spike in their referrals as it interfered with the IVA assay.

Percentage of Referrals by Disorder in 2012



Cystic fibrosis, endocrinopathies and metabolics each represent approximately 30% of screen positive referrals. Hemoglobinopathies represent approximately 6% of referrals.

5.3 Diagnostic Feedback

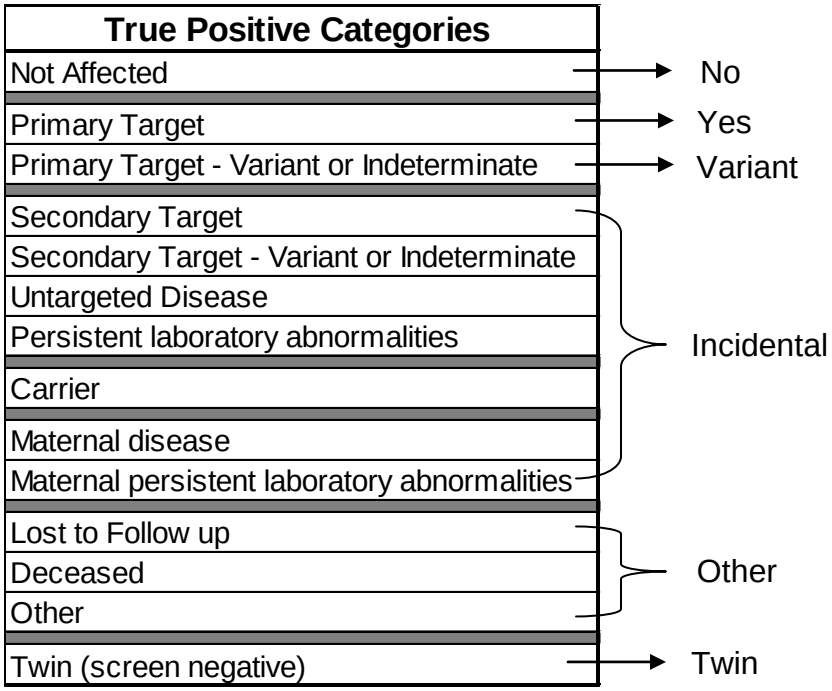
Approximately 15.3% of feedback information (DERFs = diagnostic evaluation report forms) was not received or not entered into our electronic record for the referrals made in 2012. Of the 1107 referrals on which feedback was received, 158 were classified as true positive. This represents 14% of all returned information and provides a true positive rate of 0.1% (~1:910) of all infants screened by NSO. Based on the information available, the positive predictive values are estimated in the table in Section 5.5.

5.4 Classification of True/False Positives

NSO has developed a classification system for true positives to take into account the variability of definitive diagnoses and the impact of variant conditions and incidental findings. The definitions are as follows:

True Positive?	Definition	Example
Yes	confirmed diagnosis of a targeted condition	Classical PKU
No	confirmed to be NOT affected by a target or related disease	Not Affected
Other	lost to follow up; family refused follow up; infant deceased prior to completion of diagnostic evaluation	Deceased
Variant	confirmed diagnosis of a variant of the targeted condition	CF indeterminate or gray zone
Incidental	not affected by target or variant disease but not unaffected; affected with secondary target or other condition; carriers; reason intrinsic to baby or mother that caused the baby to screen positive	Vitamin B12 deficient (PA/MMA screen positive), maternal Grave's disease (CH screen positive)

The category of incidental is a large group – consisting of reasons due to mom and baby. Now that the DERF information is captured in BORN, we have added additional classifications to allow for more useful data extractions in the future.



5.5 Positive Predictive Values

The current PPVs are for current disorders logics.

The PPV for yes is calculated using our classification of 'yes' in the numerator and the sum of 'yes', 'variant', 'incidental' and 'no' in the denominator. 'Other' is excluded as follow up data is not known. The PPV including yes plus variant is calculated with the addition of 'variant' in the numerator.

Variant is particularly important in CF = indeterminate or gray zone where there are borderline sweat results and 1 or more CFTR mutations identified, biotinidase deficiency (partial biot def), PKU variant = mild hyperphe (Phe = 120-600) and mild PKU (Phe = 600-1200), and CPT1 deficiency with the Inuit common mutation (which is questionable as to whether or not it is associated with disease). Therefore, our PPVs are higher when these primary disease variants are taken into account.

Current and Previous Method PPV Data

Disease	Current PPV			Previous PPV		
	Total No. Positive	PPV (yes)	PPV (yes + variant)	Total No. Positive	PPV (yes)	PPV (yes + variant)
Congenital Hypothyroidism						
Referred	1190	44.5%	44.5%			
< 24 hrs	151	0.7%	0.7%			
Total	1341	39.4%	39.4%			
Congenital Adrenal Hyperplasia	67	0.0%	0.0%	244	4.5%	4.5%
Hemoglobinopathies	190	71.6%	71.6%	353	59.9%	59.9%
Cystic Fibrosis						
Category A	108	99.0%	100.0%			
Category B	1584	1.5%	5.2%			
Category C	345	0.6%	1.0%			
Total	2037	6.3%	9.3%			
Citrullinemia	83	19.1%	20.6%			
PA/MMA	202	3.3%	3.3%	231	2.6%	2.6%
Isovaleric Acidemia	163	2.0%	2.7%			
Glutaric Aciduria type 1	90	8.6%	8.6%			
C5OH	355	7.3%	7.3%			
CUD/CPT1	306	5.6%	13.0%			
CPTII	26	3.8%	3.8%			
Homocystinuria	67	0.0%	0.0%			
LCHAD	9	77.8%	77.8%			
MCAD	187	27.4%	37.2%			
Phenylketonuria	329	13.4%	36.3%			
Tyrosinemia	17	7.7%	7.7%	70	1.5%	1.5%
MSUD	7	14.3%	14.3%	86	2.7%	2.7%
Galactosemia	68	37.5%	40.6%			
Biotinidase Deficiency	208	6.3%	27.6%			
VLCAD	138	10.3%	17.9%			

There were only 5 conditions in which there have been disorder logic updates: CAH, Hemoglobinopathies, PA/MMA, Tyrosinemias, and MSUD.

With the change in disorder logic, the PPV for Hemoglobinopathies has increased form 59.9% to 71.6%. Of note, the PPV calculation is performed with incidental in the denominator. Incidentals are non-targeted conditions, such as beta thalassemia, EE disease, etc. So while they are diseases, they are not the targeted conditions.

For tyrosinemia, the previous cutoff was too close to the detection limits. With the new cutoffs, the PPV has risen to 17% (which includes type 1 as well as other types of tyrosinemia). The initial positive rate has also decreased which is reflected in the current PPV.

For CAH, the PPV has decreased, reflecting that there have been no true positives identified as of yet with the current logic disorder. However 1/3 of the DERFs during this time period are still pending. There has been a slight reduction in the total number of referrals (109 over a 7 month period of time vs. 67 over a 5 month period which works out to a reduction of 2-3 referrals per month).

6. New and Ongoing Initiatives

6.1 Monitoring and Diagnostic Testing

NSO had received feedback requesting an option for home monitoring or diagnostic testing by dried blood spot. This test would be easier for families as it would reduce the number of visits to a treatment centre, thereby reducing time away from work and expenses. This is also a cost effective and timely way to obtain results as NSO performs daily runs of screening samples. The offering is limited to tests that are currently used in the screening program, but have other uses in the diagnosis and monitoring of patients. Monitoring results are reported to the ordering physician, other identified HCPs, plus the patient/family. Diagnostic testing results are reported to the ordering physician only. In the first year of offering (2011), NSO received 158 diagnostic or monitoring samples from 50 different individuals. In 2012, 327 diagnostic or monitoring samples from 67 individuals were received.

NSO also receives coroner samples, in the form of dried blood spots and bile samples. In 2012, NSO received 195 samples from 106 patients.

Criteria	Samples	Patients
Coroner samples	195	106
Monitoring/Diagnostic samples	327	67

6.2 Reporting of Screen Positives in Multiples

NSO implemented a multiple birth policy for screen positive infants in 2011. This involved referring out all babies in a birth set (twins, triplets, or even higher multiples) even when only one baby in a birth set was screen positive. The reason for this was two-fold:

1) Chance of sample mix-up
2) If one infant is at increased risk for a condition on the NBS, most of which are purely genetic, then the other infant(s) in the set would also be at increased risk above the general population.

Referral paperwork is generated for all babies in the birth set, complete with DERFs. In 2012, NSO referred 93 screen positive infants who were a twin or triplet.

Of the 93 screen positive referrals, this represents 82 sets of twins/triplets. There were 11 sets where both twins would have already been referred as they were either screen positive for the same condition or for different diseases. There were 76 twins/triplets referred as a result of this

policy where one was positive and one was negative. Since this policy was implemented in 2011, 1 screen negative twin was found to be affected on follow up investigations.

6.3 Updated Disorder Logic for Congenital Adrenal Hyperplasia

Implemented: August 2012

Newborn Screening Ontario (NSO) updated the disorder logic for congenital adrenal hyperplasia by no longer reporting fluoroimmunoassay (FIA) 17-hydroxyprogesterone (17OHP) exceeding the critical cutoff regardless of the Steroid Profiling (SP) results by LC-MS/MS. Therefore, a baby would only be reported as screen positive for CAH if the SP result was also positive. It is anticipated that this will decrease the false positive rate without changing the screening sensitivity for CAH.

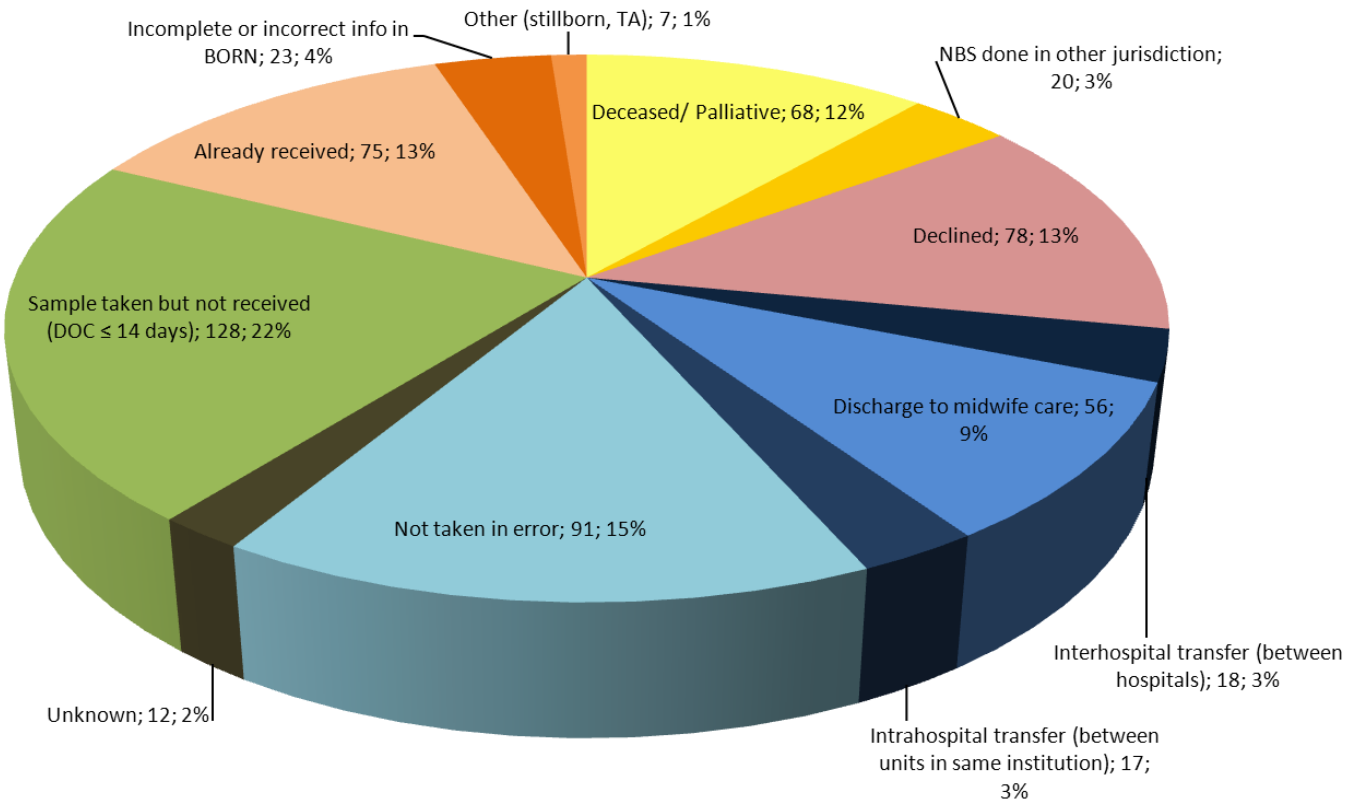
6.4 BORN Data Sharing

Implemented: January 26, 2012

Beginning in January 2012, NSO began sending HL7 messages containing real time demographic information and laboratory results for NSO samples to BORN for inclusion in the registry data.

There have been challenges related to data quality, both on our requisitions and in the data entry being done directly or by batch upload into BORN at hospital sites. This has led to a great deal of manual linking and matching of NSO records to mothers and babies in the BORN system. We continue to work through these challenges with the software vendors and BORN coordinators to achieve more accurate and automated linking and matching.

One of the primary benefits of BORN integration for NSO was the identification of missed screens (ie. babies born in Ontario but no NSO sample was received by 14 days of age). On a daily basis, BORN notifies NSO of infants who are ≥14 days of age and who have no matching newborn screening record in BORN. The NSO team then follows up with the birth hospital or midwifery practice to determine why an infant has not had a newborn screen. In the first twelve months, NSO followed up on 593 potential missed screen alerts. The data regarding these infants is attached, and indicates that 194 (33%) infants truly had missed newborn screens. Following the notification from NSO, the birthing hospital or midwifery practice involved with the family was able to ensure that the newborn screen was completed for 165 of the 194 infants.



In 78 (13%) missed screen alerts a sample will never be obtained because the parents have declined the screening test. While a minority (31%) of babies are in midwifery care, 68 (87%), of the declines were from babies in midwifery care (see graph page 25). The reasons for this are unclear and require further study. For example, education and collection processes in hospital may leave less opportunity for parents to decline screening, or parents using midwifery services may be more likely to decline newborn screening in general.

Of the alerts, 181 (31%) had midwifery (MW) involvement, while only 10% of births in Ontario are under midwifery care. Of the 83 midwifery practices in the province which submitted samples over this time period, 68 (82%) were involved in missed screen alerts versus 74 (47%) of the 158 Ontario submitting hospitals. MWs were involved in 60 (31%) of the true missed screens and obtained samples following notification from NSO for 53 (88%) cases. Between the true misses and the samples taken but not yet received, 80 samples from MWs arrived ≥14 days of age.

6.5 Severe Combined Immune Deficiency

Screening for Severe Combined Immune Deficiency (SCID) was approved by the Ministry of Health and Long Term Care in November 2012. With this announcement came a 3 year funding commitment, beginning in April 2013. NSO has begun acquiring additional equipment and new lab space, hiring new staff, developing a SCID screening method, liaising with the treatment centres and immunologists across the province, and developing a workflow and educational material. SCID screening is expected to commence in the summer of 2013.

6.6 Infant Hearing Program

A collaboration between the Infant Hearing Program (IHP) and NSO was established to develop a framework to screen newborn dried blood spots for risk of congenital / non-congenital early hearing impairment. The risks for non-congenital hearing impairment include congenital CMV and well-defined mutations in genes associated with hearing impairment. Analysis of historical blood spots will allow for the determination of Ontario prevalence of congenital CMV DNA in blood spots and the allele frequencies of the common mutations associated with hearing impairment

6.7 Annual Symposium

NSO hosted the 2013 Canadian Newborn and Child Screening Symposium on April 11-12, 2013, in Ottawa, Ontario. NSO has hosted annual symposia, since 2008 to gather together specialists from the different treatment centres in Ontario and Winnipeg for advice and development of practice guidelines. For the first time, in response to participant feedback and a growing interest from other provincial programs, NSO reinvented the 2013 symposium for a national audience.

The first day of the conference focused on disease specific workshops where attendees could share challenges and successes. These workshops were led by disease specialists from across Canada, who moderated discussion about disease specific issues and practices. Themes also emerged about challenges that span the different specialties, such as the disclosure of carrier information, which were highlighted in a combined specialty panel discussion. The first day ended with dinner and a key note presentation by Dr. Guy Van Vliet.

The second day of the two day conference attracted a broader audience for poster sessions, abstract presentations, and invited speakers. The main themes of the second day were screening for Severe Combined Immune Deficiency (SCID) and developmental delay. Dr. Alex Kemper from Duke University School of Medicine in North Carolina opened the day with a summary of the newborn screening review process in the United States. He was followed by Dr. John Cairney from McMaster University in Hamilton, who spoke about early infancy/ childhood screening for developmental delay and the application of tools in Ontario. These talks were complimented by six abstract presentations of diverse work related to novel screening techniques, educational initiatives, and research. Dr. Mei Baker from the Wisconsin Newborn Screening Program and Dr. Francis Lee from the Center for Disease Control in Atlanta spoke in the afternoon about their pioneering work on SCID screening in the US.

The success of the conference was due in part to the diversity of the participants, with attendees from across Canada and the United States, from various specialties and training backgrounds. The feedback from the attendees was positive and NSO is planning for the 2014 Canadian Newborn and Child Screening Symposium which will occur along with the 2014 Garrod Symposium in Ottawa in the spring. The tentative theme for next year is ‘Personalized medicine through screening – rare diseases and beyond’.

6.8 Public/ Parent Education Activities

NSO has been working on updating and harmonizing the NSO and Ministry of Health websites:

www.newbornscreening.on.ca

www.health.gov.on.ca/newbornscreening

Part of the updating has included the fact sheets for the public (12 languages), the bilingual parent's section on NSO website, and materials for screen positive families: "My baby is screen positive for…".

NSO is incorporating social media into its strategies for marketing and education. In December, 2012, NSO launched a Twitter account (@NBS_Ontario). Twitter is an online social networking and micro blogging service that allows its users to share and read text-based messages of up to 140 characters, known as "tweets". The NSO Twitter account has a broad audience which includes parents, healthcare providers, patient advocacy groups, and healthcare organizations and was created as a forum for education about newborn screening in Ontario. Depending on the success if of this initiative, NSO will consider increasing its social media presence in the future by joining other social media platforms (i.e. Facebook, LinkedIn).

6.9 Exploration

NSO has also been involved in research projects highlighting the impact of educational initiatives and messages.

- One of the projects is based out of the University of Toronto and examines health care providers' and moms' experience with a Cystic Fibrosis (CF) screen positive result through a self-administered survey. NSO is involved with the control population (CF screen negative moms) and health care providers. This project is still ongoing.
- Another project is examining whether and how NSO screening information can help with the care of families known to be at risk of having a child with a target condition. The findings are that there is a potential benefit to having a systematic approach for the infant and HCPs involved with postnatal care. Such a system could enhance NBS test performance, ensure congruency in prenatal and NBS results and identify discordant or missed cases. One of the concerns raised was regarding privacy and the need for consent to share prenatal information with postnatal HCPs.
- Another project based out of the University of Ottawa, is intended to measure and compare expecting mothers' responses to specific NBS educational messages: How does the message impact the decision making process? The study population consists of 500 women with low-risk pregnancies attending routine 2nd trimester ultrasound clinics in Ottawa. The participants are randomized to receive different combinations of messages about NBS. This project is still ongoing.

6.10 Ontario Laboratory Information System

Planning project: Completed in January 2013

The Ontario Laboratory Information System (OLIS) is a system to electronically share all laboratory information between practitioners and laboratories. OLIS allows practitioners and laboratory staff to review results, referral workflow and order submissions. NSO plans to have

newborn screening results for all infants available through OLIS in the Fall of 2013, and electronic test requests in early 2014

6.11 Vision and Mission

In 2012, the NSO leadership team with feedback from the entire staff, has developed a vision and mission statement for the program:

Vision

- The Best Possible Health through Screening

Mission

- Ensure every newborn receives the highest quality screening and care for serious diseases.
- Continuously develop and improve the integrated system of screening for the population we serve.
- Be global leaders in screening through research, development, education, and innovation.