



Annual ReportCalendar Year 2014

Contents

1. SA	MPLE VOLUMES IN 2014	4
1.1 1.1.1 1.1.2 1.1.3 1.2	SCREENING SAMPLES INFANTS SCREENED DECLINED/DEFERRED TESTING MISSED SCREENS NON-SCREENING SAMPLES	5 6
	EMOGRAPHICS OF SCREENING SAMPLES	
2.1 2.2 2.3 3. UN	AGE AT COLLECTION	8
3.1 3.2 3.2.1 3.2.2 3.2.3 3.3	Sample Quality — Laboratory Unsats Data Quality and Process Related Unsats Insufficient Information Expired Cards Transportation Repeat Rates for Unsatisfactory Specimens	11 11 11 12
4. TU	JRN AROUND TIMES	13
4.1 4.2 5. SC	TRANSPORTATION TIME	14
5.1	REFERRALS BY TREATMENT CENTRE	
5.1	SCREEN POSITIVES BY DISORDER	16
5.3	DIAGNOSTIC FEEDBACK	16
5.4	CLASSIFICATION OF TRUE/FALSE POSITIVES	
5.5	DEFINITIVE DIAGNOSIS DATA AND POSITIVE PREDICTIVE VALUES	18

Introduction

In 2014, Newborn Screening Ontario (NSO) focused its efforts towards renewing the information systems that support the program and ensuring readiness to adapt to growth and change to the screening system in the coming years.

Project Lancet, with a mandate to develop a flexible IS ecosystem that NSO can use to better manage its current and future workflows and communication needs, began its project inception phase by reviewing the current IS systems in NSO and identifying user requirement to direct the build of the new system. As an early initiative in Project Lancet, a team from Algonquin College built a proof of concept Data Warehouse, a large store of NSO data accumulated from a wide range of sources, as a tool to support quality assurance and guide management decisions. This project was successful, and continued work has been done to extend the data and reports available in the NSO data warehouse. For the first time, the data warehouse has been used to pull the data for the creation of this annual report, which necessitated additional data validation and cleansing steps, and therefore an initial delay in the release of this report. Standard reports and processes have been created, which will make future annual reports more consistent and less time consuming to produce.

A number of other initiatives took place in 2014 to support all areas of the NSO mandate and ensure quality services. The laboratory underwent Ontario Laboratory Accreditation (OLA) assessment in June, 2014 with over a 99% compliance and only three identified minor non-conformances. The program has achieved ISO 15189 Plus designation.

Following direction from the Health Ministers meeting in 2013, a pan-Canadian working group has been formed to explore areas of cooperation in newborn screening with a focus on developing a national core panel of diseases. NSO is participating in the development of all recommendations and is leading work on the evaluation of new technologies and the development of NBS-related research sharing protocols.

While the volume of Ontario screening samples has remained constant in 2014, in June NSO entered into a contract with Dynacare to provide newborn screening services for Nunavut territory, resulting in a higher volume overall. This initiative has also encouraged the work on a CPT1 assay development and integration to our screening logic, due to the high incidence in this population.

Another significant change to laboratory testing in 2014 was the change to a new instrument and enzymatic method for Galactosemia and Biotinidase deficiency. This new method had a very big impact on lab workflow, due to the improved instrumentation, but some increases in referrals were noted due to the conservative cutoffs chosen for implementation. This is further described in section 5.2.

A number of educational initiatives took place in 2014 for all NSO stakeholders, including a Joint Garrod & Canadian Newborn and Child Screening Symposium; three NSO submitter workshops held in Toronto, Peterborough, and Chatham; and the launch of the NSO Submitter Manual on the NSO website. NSO has also been working with MOHLTC on the distribution of new NSO information pamphlets and an updated video for parents about newborn screening, both of which are available in 20 languages.

In December 2014, NSO began a new strategic planning process with a staff survey based on a SWOT analysis, and leadership retreat. The strategic goals and objectives resulting from the process will set the direction for NSO over the next 3-5 years.

1. Sample Volumes in 2014

Table 1. Sample volumes between 2014-2011 by sample type.

Indication	Sample Type	2014	2013	2012	2011
Double concenies	Satisfactory	144,099	144,402	143,979	142,993
Routine screening	Unsatisfactory	4,349	3,191	3,826	2,912
Routine Screening – Summary		148,448	147,593	147,805	145,905
Referred in sample: full panel	Satisfactory	192	8	17	49
Referred in sample, full parier	Unsatisfactory	5	0	0	0
Referred in screening: Full pan	el – Summary	197	8	17	49
Cord Blood		469	160	0	0
Post Mortem		333	276	195	179
Diagnostic/Monitoring		481	464	326	115
Non-screening sample – Summ	nary	1,283	900	521	294
Total	149,928	148,501	148,343	146,248	

1.1 Screening samples

In 2014, NSO saw increased sample volumes for routine screening, primarily due to an increased number of unsatisfactory samples.

NSO entered into an agreement with Dynacare in June 2014 to accept referred in screening samples for all Nunavut babies. Thus, the 197 referred in samples is largely composed of Nunavut samples.

1.1.1 Infants Screened

This is the total number of samples received for newborn screening purposes only. The retrospective data is based on current linking algorithms, not on numbers from previous reports, therefore numbers may have changed slight if the system has identified older samples that "match" (ie. two samples received from the same infant). The number of infants tested is an estimate which may be impacted by the efficiency of the linking algorithm as well as data quality.

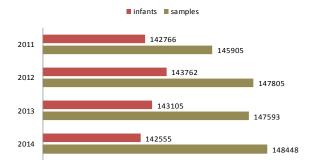


Figure 1. The total number of infants and samples screened between 2011-2014.

The number of infants tested is always lower than the number of samples received due to repeats required for transfusion, prematurity/low birth weight and laboratory and data unsatisfactory samples.

The overall number of infants tested is relatively constant each year with only ~1000 infants difference between the highest and lowest years. The unsatisfactory rate in 2013 was 2.16%. The rate in 2014 increased to 2.93%. The may partially account for the increase in number of samples received.

The NSO Babies in Ontario is the total number of infants NSO was made aware of through defers/ declines, missed screen alerts and newborn screening samples. 99.7% of all infants born in ON were screened by NSO.

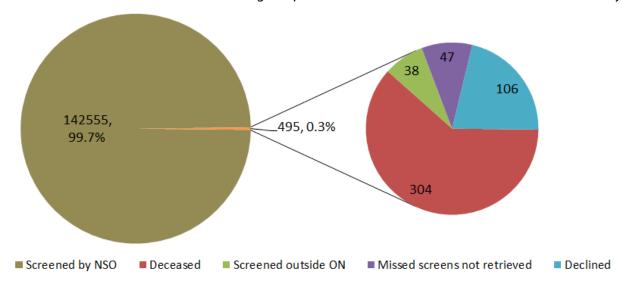


Figure 2. Coverage of screening in Ontario births.

1.1.2 Declined/Deferred Testing

In 2013 NSO developed and implemented a decline/defer form with the NBS blood dot cards. These cards went into circulation in January 2014. If parents wished to decline or defer newborn screening, health care providers could have the parents sign the form and submit the card with demographic information completed. In the case of a decline, it would avoid unnecessary follow up when a missed screen alert was received and it allowed formal documentation on the part of the health care provider that they offered NBS. Upon receipt of the decline form, NSO entered the information into their system and generated a letter to the

submitter documenting the receipt of the decline. In the case of a deferral, the family would once again sign the NBS card and the submitter would send it in to NSO. Similar to the decline process, the information would be entered and a letter generated to the submitter. If a NBS sample was not received by 14 days from the receipt of the deferral notice, NSO would generate a letter that would be sent to the family directly.

In 2014, there were 23 declines from this process. There were 32 deferrals in this time period with only one case where the deferred sample was never received despite a letter to submitter and family.

Table 2. Declined, deferred samples and potential missed screens between 2014 – 2011.

Case Type	2014	2013	2012	2011
Declined/deferred form received	54	<5	N/A	N/A
Potential missed newborn screen	454	558	212	0

1.1.3 Missed Screens

In 2014, there were 454 potential missed newborn screen alerts from BORN that required follow up by NSO. This is down by approximately 100 cases from 2013. Hospitals were the responsible facility in 56% of cases and midwives were involved in roughly 42% of the missed screen alerts. Other (representing 2% of alerts) include centres or nursing stations involved in follow up care post discharge from hospital. There were 65 different midwifery practices involved in the alerts and 62 different hospitals. Action on the part of NSO resulted in 116 of the 163 (71%) missed screens being completed.

Table 3. Potential missed screen alerts requiring follow-up in 2014, by reason and responsible submitter, and samples received post follow-up.

	TOTAL	Samples Received	% Received
Incomplete or incorrect information	291		
True missed screens	163	116	71%

Missed Screens and BORN entry

NSO is dependent upon timely data entry into BORN on the part of responsible health care providers for missed screen alerts. Of the missed screen alerts, 179 (54 true misses) were received >14 days of age and the age at which true missed screen alerts were received ranged from 15 to 317 days of age. In addition, there were 150 cases with no alerts were triggered because of late data entry into the BORN system, but samples received ≥14 days of age. However in examining the age at collection and time from collection to receipt, many of these samples were collected an appropriate age and were delayed in transit. See Section 4 for further description of NSO's initiatives to decrease transit times.

Missed Screens and Declines

In 2014 there were 83 declines identified in the missed screen alerts, compared to 87 declines in 2013. Including the 23 declines from the decline process outlined above, the total number of declines actually rose in 2014. Midwives were the health care provider in 83% (n=69) of declined cases.

1.2 Non-Screening Samples

In addition to routine screening samples, and screening samples referred from other jurisdictions, Newborn Screening Ontario accepts non-screening samples of various types.

In 2013, NSO began accepting cord blood samples for hemoglobin screening to support the national cord blood registry. Cord blood samples have been submitted from Ontario since 2013, and Alberta and British Columbia were included in 2014.

Since 2010, NSO has had an agreement with the Ontario Forensic Pathology Service to provide postmortem dried blood spot and dried bile spot sample analysis for all unexplained deaths of children under two years of age. These sample volumes have been steadily increasing each year, likely due to coroner awareness and compliance. Although a blood and bile sample is requested for each case, both sample types may not always be retrievable. NSO received 164 blood samples and 169 bile samples in 2014.

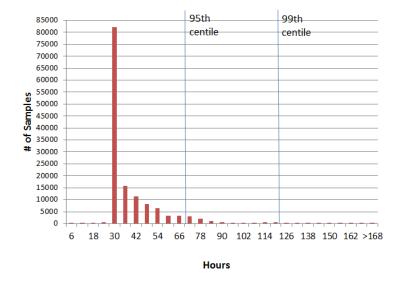
NSO offers diagnostic and monitoring testing for targets of newborn screening, and volumes are relatively steady from 2013, with the biggest volume being from PKU home monitoring.

2. Demographics of Screening Samples

2.1 Age at Collection

Table 4. Age at collection for 2014 initial samples only.

Age at Collection	Number of Initial Samples	% of Initial Samples
Less than 24 hours	818	0.55%
24-47 hours	116,537	78.50%
48-72 hours	15,897	10.71%
3-7 days	8,651	5.83%
Greater than 7 days	6,476	4.36%
Not specified	69	0.05%



The majority of newborn screening samples are collected between 24-48 hours of age. Approximately 90% of samples are collected by 72 hours of age. NSO will be looking into possibly changing the recommended age of collection to 24-48 hours of age from 48-72 hours in the upcoming year.

There were 628 samples that were collected <24 hours of age and considered unsatisfactory. NSO has a 10 min grace period for samples obtained between 23:50 and 24:00. There were 190 samples that fell into the grace period of <24 hours but considered satisfactory for testing.

Figure 3. The number of samples collected by the age in hours of the infant.

Of the 628 samples collected <24 hours that were considered unsatisfactory, 74 were reported to have had early hospital discharge. A further 39 were collected early due to a pending transfusion. The majority of <24 hour samples that were unsatisfactory were taken early for an unknown reason.

2.2 Transfusion Status

NSO recommends that a repeat sample is taken 4-6 months after the most recent transfusion, therefore some infants who have had multiple transfusions will be greater than six months old when they are eligible for a repeat newborn screening sample. 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). If NSO is informed by the submitter that the infant is deceased, the case is closed as no repeat sample will be received.

Table 5. Transfusion cases in 2014.

Repeat Requirement	# of cases
Repeat Not Required	192 (58.5%)
Repeat Received	61 (18.5%)
Repeat Not Received (e.g.	
deceased, family moved, etc.)	75 (23%)
Total	328

Table 6. Age at which transfusion repeats were received in 2014.

Age	# of samples
4-6 months	12
6-12 months	44
>12 months	5
Total	61

There were 328 transfusion cases created in 2014. 77% of cases either have a repeat received or a repeat was not required as a satisfactory pre-transfusion sample was already received. Only 13 cases from 2014 remain open with no repeat received as of yet. There were 32 cases where letters were sent to submitters from NSO advising of the need for a repeat sample. There were 61 cases where a repeat transfusion sample was received, the majority of which were received between 6-12 months of age.

2.3 Gestational Age and Birth Weight

NSO introduced an extreme premature infant policy in January 2013 where any infant <1500 g or <33 weeks gestation would be recommended to have a repeat sample obtained around 21 days of age or sooner if the infant was to be discharged. In 2014, there were 2139 infants that fit the premature infant policy. Of these, 1402 (66%) had a 3 week (or equivalent) sample obtained.

3. Unsatisfactory Samples

Table 7. Unsatisfactory samples by reason between 2014-2011.

	orisatisfactory samples by reason between	2014	2013	2012	2011
	Satisfactory Samples	144,099	144,402	143,979	142,993
	Unsatisfactory Samples	4,349	3,191	3,826	2,912
	Unsatisfactory Rate	2.93%	2.16%	2.59%	2.00%
	Samples collected at <24 hours	628	718	648	693
	Unsatisfactory Samples excluding <24 h samples	3,721	2,473	3,178	2,219
	Unsatisfactory Rate excluding <24 h samples	2.51%	1.68%	2.15%	1.52%
	Quantity of blood insufficient	1,707	1,168	1,251	863
	Blood spots appear scratched or abraded	1,353	758	1,131	595
sats	Blood spots are supersaturated	1,140	718	1,220	810
Lab Unsats	Blood spots appear clotted or layered	958	248	154	174
Lab	Blood spots appear diluted	65	9	7	14
	Blood spots exhibits serum rings	65	28	24	23
	Blood spots are wet and/or discolored	16	15	35	41
ts	Blood dot collection paper is expired	120	68	123	62
<u>U</u> nsats	Insufficient data provided	32	36	43	46
Data <u>U</u>	Damaged or delayed in transit	23	<5		
Da	Delivered to lab > 14 days after collection	30	120	37	117
	Other	23	41	32	19

The number of unsatisfactory samples rose in 2014. There were a number of samples that had more than one unsatisfactory reason (the discrepancy between the total number of unsatisfactory reasons and number of unsatisfactory samples).

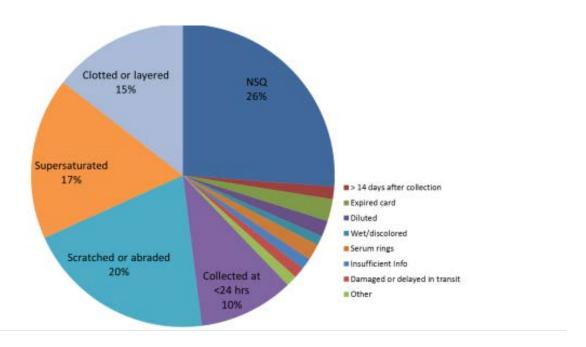


Figure 4. Distribution of unsatisfactory reasons in 2014.

3.1 Sample Quality – Laboratory Unsats

The majority of unsatisfactory samples are related to the quality of the blood sample collection directly, including too little or too much blood, or improper application of the blood on the card. There has been a significant increase in these types of unsatisfactory samples in 2014, resulting in an overall unsat rate (excluding <24 h samples) of 2.58%.

In June 2014, NSO increased the internal criteria for the quantity of blood required to perform newborn screening. The number of dried blood spot punches taken from the bloodspot cards increased from 8 to 9 due to the addition of Severe Combined Immunodeficiency (SCID) and changes in NSO's screen methods for galactosemia and biotinidase deficiency. While this change would have contributed to the higher number of unsats in 2014, much of the increase was the result of a change to the pre-analytical review process for samples, causing a higher number of samples to be rejected. The procedure has been reviewed and the number of samples being rejected has reduced in 2015.

3.2 Data Quality and Process Related Unsats

3.2.1 Insufficient Information

The number of samples ultimately deemed unsatisfactory related to insufficient information remains consistently low, due to the efforts made by NSO to contact submitting providers for missing data fields.

3.2.2 Expired Cards

Expired cards can fluctuate year to year, depending on when the lots of cards expire. There were three lots of cards that expired in 2014, in January, July and December, likely resulting in higher numbers of unsatisfactory samples for this reason. Typically, NSO sends out bulletin reminders to submitters when an expiry date is

approaching, asking them to check their stock and circulate their stock. In November, NSO implemented an enhanced reminder system to reduce expired stock.

3.2.3 Transportation

NSO began to examine ways to improve transit time of newborn screening samples in 2014. NSO introduced batchograms which were visualizations of transit times across the province for submitters as a whole as well as individual report cards. The majority of samples collected for newborn screening should be received within 24-48 hours of shipping (exceptions in more rural areas of the province). Further information on transit time is outlined in section 4.

In the fall of 2014, NSO transitioned all submitters from Canpar to Purolator for courier services, as Purolator is the Government of Ontario Vendor of Record and provides significantly lower rates for comparable service.

3.3 Repeat Rates for Unsatisfactory Specimens

The majority (80.52%) of repeat samples required due to unsatisfactory initial samples are received within 3 weeks of the initial sample. By 6 weeks, 90.43% of unsatisfactory samples have had screening completed via a repeat sample. A further \sim 5% (total of 95.36%) repeats are received \geq 6 weeks. Repeat samples have not been received for 4.64% of unsatisfactory samples in 2014.

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 **85**th **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 **94**th **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.

Table 8. Days in transit (date of collection to receipt of sample in laboratory).

Statistic	2014	2013	2012	2011	2010
Average	3.0	3.2	3.2	3.3	3.4
Median	3	3	3	3	3
Mode	2	2	2	2	2
85th Percentile	4	5	5	5	5
94th Percentile	5	6	6	6	6

NSO began using Canpar as their courier in 2010. In July 2014, NSO switched to Purolator. With a consistent mode of 2 it is clear that overnight delivery is the norm for the majority of samples.

4.2 Reporting Times

Table 9. Time from receipt of sample to report by date of collection and date of birth.

Statistics	DOC to received (transport)	Received to report	DOB to report	DOC to report
Average	2.9	4	8	7
Median	3	3	8	6
Mode	2	2	7	6
85th Percentile	4	6	11	9
94th Percentile	5	7	13	11

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.

Table 10. Date of sample collection to report.

Statistic	2014	2013	2012	2011	2010
Average	6.6	7.2	6.8	6.5	6.3
Median	6	7	6	6	6
Mode	6	6	6	6	6
85th Percentile	9	10	9	9	8
94th Percentile	10	12	11	11	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.

Table 11. Date of sample receipt to report.

Statistic	2014	2013	2012	2011	2010
Average	3.6	4.1	3.3	3.3	2.9
Median	3	3	3	2	2
Mode	2	2	3	2	2
85th Percentile	6	6	5	5	5
94th Percentile	7	8	5	7	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, cystic fibrosis, and SCID (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.

5. Screen Positives

In 2014, there were 1674 screen positive referrals, from a total of 1614 infants. This represents 1.13% of the total number of infants screened by NSO. There were 1710 total screen positives but 29 had an elevated TSH in samples taken at <24 hours and 7 were premature infants who screened positive for SCID. All 36 infants were screen negative on repeat sample testing and were not referred.

The number of screen positive infants referred in 2014 rose from 2013 by just under 200 referrals. This is discussed further in Section 5.2.

5.1 Referrals by Treatment Centre

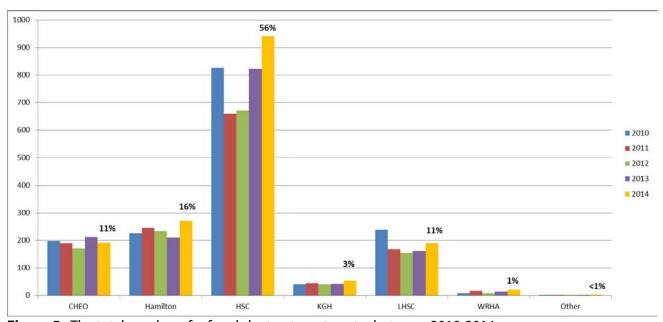


Figure 5. The total number of referrals by treatment centre between 2010-2014.

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 above. 'Other' represents infants 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 increased in 2014, with the exception of CHEO.

5.2 Screen Positives by Disorder

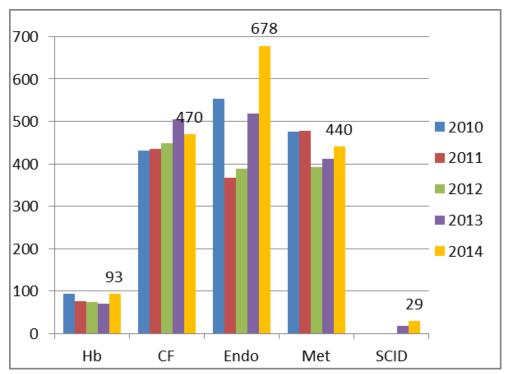


Figure 6. The total number of screen positives by disease grouping between 2010-2014. Cystic fibrosis, endocrinopathies and metabolics represent approximately 28%, 40%, and 25% of screen positives respectively. Hemoglobinopathies represent approximately 5% of screen positives.

5.3 Diagnostic Feedback

Approximately 12.3% (210 cases) of feedback information (DERFs = diagnostic evaluation report forms) was not received or not entered into our electronic record for the referrals made in 2014 as of October 2015.

Of the 1500 referrals on which feedback was received, 185 were classified as true positive. This represents 12.3% of all returned information and provides a true positive rate of 0.13% (~1:789) 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:

Table 12. The definitions of the classification of true positive.

True	Definition	Example		
Positive?				
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.

Table 13. The true positive categories.

True Positive Categories Not Affected	→ No	
Primary Target Primary Target – Variant or Indeterminate	→ Yes → Variant	
Secondary Target Secondary Target – Variant or Indeterminate Untargeted Disease Persistent laboratory anomalies	→ Incidental	
Carrier Maternal Disease Maternal persistent laboratory abnormalities	2 meiaemai	
Lost to follow up Deceased Other	→ Other	
Twin (screen negative)	→ Twin	

5.5 Definitive Diagnosis Data and Positive Predictive Values

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 = 120-359), 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.

There 7 conditions in which there have been disorder logic updates since NSO began operations in 2006: CAH, Hemoglobinopathies, PA/MMA, Tyrosinemias, MSUD, Biotinidase Deficiency and Galactosemia. Disorder logics that were updated in 2014 include Galactosemia and Biotinidase Deficiency. The current PPVs are for current disorder logics.

The data below includes all follow up information received prior to March 31, 2015.

Table 14. The current PPVs for each disorder screened by NSO and the current PPVs if the extremely premature infants were removed from the calculations.

	Current PPV			Current PPV (Prems removed)		
Disease		PPV (yes)	PPV (yes + variant)		PPV (yes)	PPV (yes + variant)
Congenital Hypothyroidism						
Referred		41.7%	46.0%		41.5%	45.6%
< 24 hrs		0.0%	0.0%		0.0%	0.0%
Total		36.6%	40.4%		36.5%	40.0%
Congenital Adrenal Hyperplasia		2.7%	2.7%		5.1%	5.1%
Hemoglobinopathies		68.9%	69.6%		71.2%	72.0%
Cystic Fibrosis						
Category A		99.2%	100.0%		99.3%	100.0%
Category B		1.8%	5.3%		1.8%	5.3%
Category C		0.6%	1.1%		0.6%	1.1%
Total		6.4%	9.2%		6.5%	9.3%
SCID		3.8%	6.7%		4.3%	10.0%
Citrullinemia		17.5%	18.6%		17.2%	18.4%
PA/MMA		5.0%	5.0%		6.1%	6.1%
Isovaleric Acidemia		2.2%	3.3%		4.0%	6.0%
Glutaric Aciduria type 1		6.8%	6.8%		7.2%	7.2%
С5ОН		5.5%	5.5%		5.6%	5.6%
CUD		5.4%	5.4%		6.8%	6.8%
СРТІ		6.5%	54.8%		8.3%	47.9%
CPTII		3.6%	3.6%		4.0%	4.0%
Homocystinuria		0.0%	0.0%		0.0%	0.0%
LCHAD		71.4%	71.4%		66.7%	66.7%
MCAD		29.5%	35.9%		32.8%	39.9%
Phenylketonuria		17.9%	33.6%		24.0%	45.2%
Tyrosinemia		20.0%	20.0%		25.0%	25.0%
MSUD		1.9%	1.9%		2.2%	2.2%
Galactosemia		5.3%	15.8%		7.1%	21.4%
Biotinidase Deficiency		0.0%	26.1%		0.0%	26.1%
VLCAD		8.5%	14.1%		8.5%	14.1%