Newborn Blood Spot Screening in Northern Ireland

Annual Report 2017 - 18

HSC Public Health Agency

Newborn Blood Spot Screening Programme

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APPENDIX B: PERFORMANCE OF NBSP IN NI 2017-18 *(cont'd)*

Summary

This Northern Ireland Newborn Blood Spot Screening Programme (NBSP) annual report will summarise the performance of the programme against key standards for the financial year 2017-18.

The NBSP in Northern Ireland offers all newborn babies a blood spot screening test to identify if they are at increased risk of five rare, but serious, inherited conditions. The aim of the programme is to improve the outcomes for babies born with one of these conditions, which can cause critical illness, severe disability and death, by achieving early diagnosis and treatment.

Throughout the United Kingdom, NBSP performance is monitored against national standards, which promote safety and quality within the programme.

Headline Results

The most recently published national report (2017-18)[1], which describes performance against national standards in each region of the UK, shows that the NBSP in Northern Ireland is of high quality and performing well.

Regional and national data relating to the Northern Ireland NBSP highlight that in 2017-18:

- In terms of coverage, >98% of 'born and resident' babies in Northern Ireland had a conclusive screening result for each of the five conditions recorded on the child health system by 17 days of age.

[1] PHE 2017 - 2018 annual report accessed at:

https://www.gov.uk/government/publications/newborn-blood-spot-screening-data-collection-report-2017-to-2018

- In relation to timing of sample collection and processing, 94.9% of samples collected on day 5 and 98.0% of samples received in the newborn screening laboratory within 3 working days of collection.

- 100% of positive screening results (for PKU, CHT and MCADD) were available, and clinical referral had been initiated, within 3 days of the sample being received by the screening laboratory.

- In relation to timeliness of receipt into clinical care, the programme in NI exceeded acceptable national standards. NI met the achievable standard for timeliness of first appointment for CF screen positive babies with 2 mutations, with 100% of babies seen by 28 days.

- Over 23,000 babies had newborn blood spot screening testing. In total, across all of the five conditions tested for, 49 babies were identified as screen positive and 24 of these babies were confirmed as having one of the conditions.

At a national level, meeting the standard (acceptable = ≤2%; achievable = ≤1%) in relation to 'avoidable repeats' has proved challenging since the introduction of the programmes, and variation exists across the UK. An avoidable repeat refers to a sample that has not met the required quality standard to be accepted by the laboratory for analysis, e.g. an insufficient quantity of blood may have been collected and the laboratory will request a repeat sample.

In Northern Ireland the avoidable repeat rate was 4.95% in 2017-18. The regional NBSP Quality Improvement (QI) group continues to work to understand and reduce avoidable repeats.

SECTION A: INTRODUCTION AND HEADLINE RESULTS FOR 2017-18

Background

The Northern Ireland Newborn Blood Spot Programme (NBSP) offers all newborn babies a blood spot screening test to identify if they are at increased risk of five rare, but serious, inherited conditions (these are described on pages 5 - 7). The aim of the programme is to improve the outcomes for babies born with one of these conditions, which can cause critical illness, severe disability and death, by achieving early diagnosis and treatment.

Most babies who are screened do not have any of these conditions, but for the small numbers who do, the benefits of screening are substantial. The programme supports 'giving every child the best start in life', a key objective of the 'Making Life Better' strategy (2013-2023)[2], and offers early diagnosis and intervention to reduce ill health.

Throughout the United Kingdom NBSP performance is monitored against national standards, which promote safety and quality within the programme. This report summarises the performance of the NBSP in Northern Ireland from 1st April 2017 - 31st March 2018 (hereafter referred to as 2017-18) against national standards.

In Northern Ireland the NBSP currently offers screening for five conditions:

- **Phenylketonuria (PKU)**

About 1 in 5,000 babies born in Northern Ireland has phenylketonuria (PKU). Babies with this inherited condition are unable to process a substance in their food called phenylalanine. If untreated, they will develop serious, irreversible, learning

[2] Department of Health and Social Services Making Life Better Whole System Strategic Framework Belfast 2014 available at: : https://www.health-ni.gov.uk/topics/public-health-policy-and-advice/making-life-better-whole-system-strategic-framework-public

disability. Screening means babies with PKU can be treated early through a special diet, which will prevent severe disability and allow them to lead a normal life.

• **Congenital Hypothyroidism (CHT**)

About 1 in 2,000 babies born in Northern Ireland has congenital hypothyroidism (CHT). Babies with CHT do not have enough of the hormone thyroxine. Without this hormone, they do not grow properly and can develop serious, permanent physical and learning disability. Screening means babies with CHT can be treated early with thyroxine medication, which will prevent serious disability and allow them to develop normally.

• **Cystic Fibrosis (CF)**

About 1 in 2,500 babies born in Northern Ireland has cystic fibrosis (CF). This inherited condition can affect digestion and the lungs. Babies with CF may not gain weight and may have frequent chest infections. Screening means babies with CF can be treated early with a high-energy diet, medication and physiotherapy.

• **Medium Chain acyl-CoA Dehydrogenase Deficiency (MCADD)**

About 1 in 10,000 babies born in Northern Ireland has medium chain acyl-CoA dehydrogenase deficiency (MCADD). Babies with this inherited condition have problems breaking down fats to make energy for the body. This can lead to serious illness or even death. Screening means most babies with MCADD can be recognised early, allowing special attention to be given to their diet including making sure they feed regularly.

• **Sickle Cell Disorders (SCD)**

Less than 1 in 10,000 babies born in Northern Ireland has a sickle cell disorder (SCD). These inherited conditions affect the red blood cells. Babies with a SCD have red blood cells that can change to a sickle shape and become stuck in the small blood vessels. This can cause pain and damage to the baby’s body, serious

infection, or even death. Screening means babies with an SCD can receive early treatment, including immunisations and antibiotics.

Screening pathway

Who is eligible for NBSP screening?

Screening commences by identifying all those who are eligible for the test. All babies up to the age of one year (i.e. those from birth, (defined as day 0 of life) up to and inclusive of 364 days of age, or up until 8 weeks old for cystic fibrosis) are eligible for, and offered, Newborn Blood Spot Screening.

This includes babies who are born and resident in Northern Ireland and those who move into Northern Ireland.

What does screening involve?

As part of the programme, in the first week after birth, ideally on day 5 of life, all babies are offered blood spot screening by a health professional, usually their midwife or nurse. It is important that those who participate in screening make an informed choice to do so. Screening tests are not 100% accurate. The screening 'test taker' will communicate clearly with baby's parent/guardian to ensure that they understand why blood spot screening is recommended and how the blood sample is used to test for a number of health conditions. A regional consent policy has been developed in Northern Ireland for use by test takers and this supports parents/guardians in making an informed choice regarding participation in the screening programme.

The blood spot test involves taking a small sample of blood from the baby's heel; this is often referred to as the 'heel prick' test. The sample is sent to the Regional Newborn Screening Laboratory in the Belfast Trust for analysis. Results are forwarded from the Laboratory to the local Child Health System (CHS) offices for recording

and issuing of hard copy result reports.

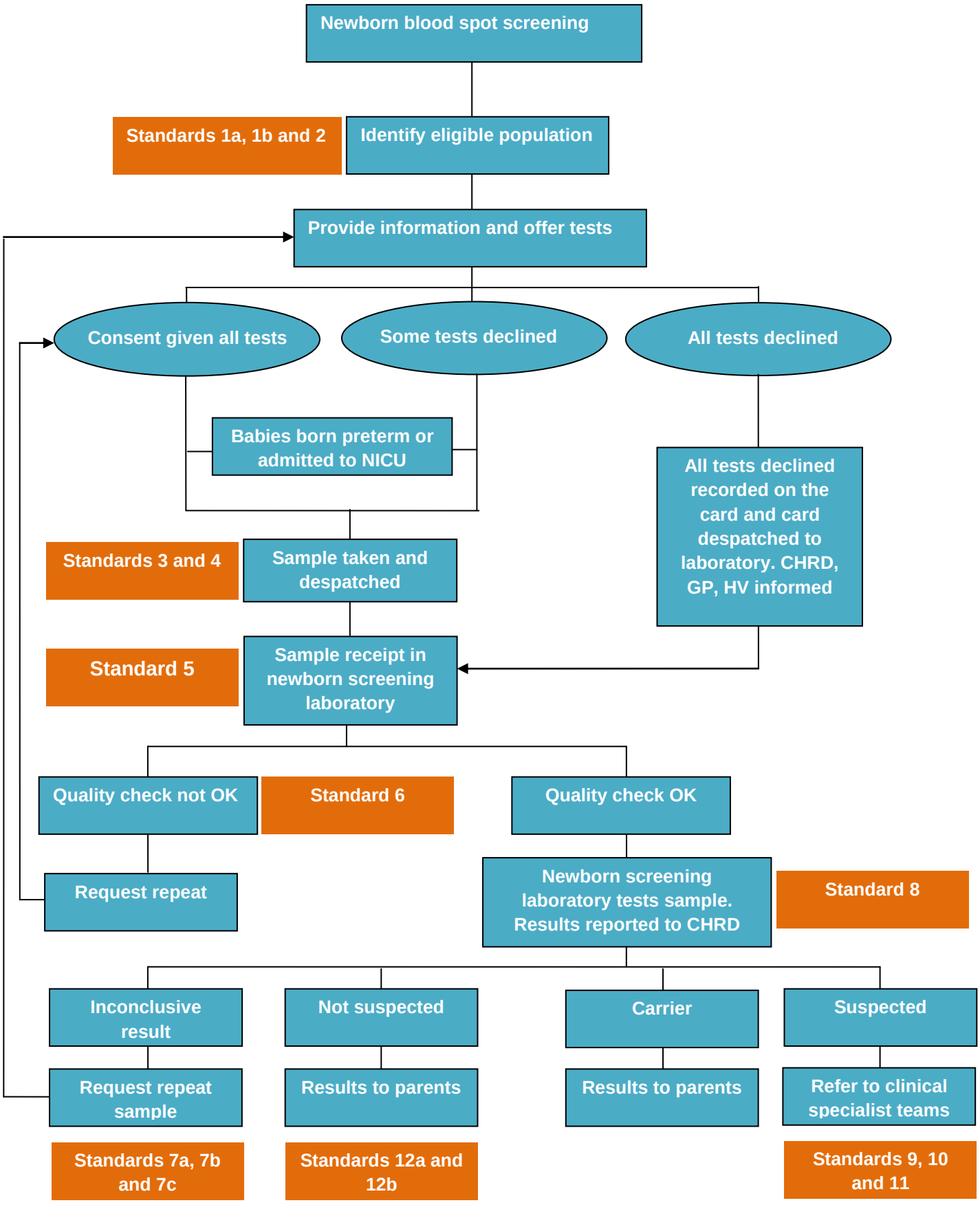
The purpose of screening is to identify babies more likely to have these conditions. If the screening test is positive, a baby will be offered further tests or investigations to confirm the diagnosis. Where one of these conditions is confirmed as present, effective interventions are available to prevent subsequent illness and/or disability arising. There are also specific national standards relating to timely referral and entry into clinical care for each of the conditions.

If the screening tests are negative, a copy of the results report is usually given to the child's parent/guardian by their health visitor at their 6 – 8 week review visit. Occasionally, a midwife or health visitor will need to take a second blood spot sample because there was not enough blood collected from the first sample or the result was unclear – this is referred to as a repeat sample. A second sample may also be required if, for example, the first result was 'borderline', in the case of congenital hypothyroidism or if the baby was premature (i.e. born before 32 weeks of age), or had a blood transfusion prior to the first sample being taken.

Figure 1 outlines the newborn blood spot screening pathway and how each element of the pathway complements the national standards for the programme. It illustrates that screening is a complex process with several stages involving multiple stakeholders. However, there are built in 'failsafes' to improve safety and quality of the programme. A failsafe is a back-up mechanism, in addition to usual care, which ensures that if something does not go to plan in the screening pathway, processes are in place to identify what has happened and that action is taken[3].

[3] PHE 2019 https://www.gov.uk/government/publications/abdominal-aortic-aneurysm-screening-programme-failsafe-procedures/abdominal-aortic-aneurysm-screening-failsafe-processes#failsafe-strategy

Figure 1: Screening pathway mapped to programme standards



NICU = neonatal intensive care unit
CHRD = child health records

Programme delivery

Newborn blood spot screening is a complex programme, involving a wide range of staff and services, from highly specialised laboratories through to individual staff in the community and in hospitals, working closely together.

A wide range of professionals and a number of multi-disciplinary teams support and deliver the programme within each of the five Health and Social Care Trusts (HSCTs) in Northern Ireland. Midwives, nurses and health visitors are responsible for providing families with relevant information to enable informed choice and consent to blood spot screening. They are also responsible for the collection and transport of blood spot samples. Laboratory staff offer timely sample analysis and results reporting and Child Health System (CHS) staff process and distribute results for onward communication and action.

CHS staff are also responsible for conducting weekly 'failsafe' reports, which aim to identify any baby that does not have a conclusive result within a designated timeframe. For each of the conditions tested, there are also agreed pathways for referral to specialist clinical teams who provide further diagnostic testing, assessment and treatment of babies with positive screening results.

At a regional level, the Public Health Agency (PHA) is responsible for commissioning and quality assuring the programme. The PHA works collaboratively with the wide range of professionals responsible for delivering the programme within each Trust to promote compliance with national standards and continuous improvement. A regional NBSP Quality Improvement (QI) group chaired by the PHA, with representatives from each of the professional groups and Trusts involved, also meets biannually**.**

Key developments 2017-18

Key improvements in the NBSP in Northern Ireland (NI) during 2017-18 included the implementation of an electronic interface for the transfer and receipt of newborn

blood spot results that are sent from the Laboratory Information System to the Child Health System. An electronic interface reduces the need to manually enter results, increases efficiency and improves quality and patient safety by automatically matching results using a baby's unique demographic details (gender, date of birth, surname and Health and Care Number).

In 2017, new programme standards were issued by Public Health England and these were implemented in Northern Ireland, with effect from 1st April 2017.

The blood spot screening programme in Northern Ireland provided feedback to a consultation that was launched by Public Health England in 2017 regarding a revised blood spot card. A regional response to this consultation was coordinated by the Public Health Agency.

Laboratory guidance for inherited metabolic diseases (IMDs), published by Public Health England in 2015, was implemented by the Regional Newborn Screening Laboratory in 2017 for the inherited metabolic diseases that were being screened for as an interim measure, prior to the implementation of the guidance in full, with the introduction of the expanded screening programme.

The Public Health Agency continues to represent the Northern Ireland NBSP at the national Blood Spot Advisory Group which is hosted by Public Health England.
Initial planning commenced in order to expand the current NBSP, following direction from the Department of Health. The expanded screening programme will test for the following additional inherited metabolic diseases:

- Glutaric aciduria type 1 (GA1)
- Isovaleric acidaemia (IVA)
- Maple syrup urine disease (MSUD)
- Homocystinuria (pyridoxine unresponsive) (HCU).

Implementation of expanded screening will commence in 2019.

Programme performance 2017-18

In 2017-18, NBS programmes across the UK monitored performance against 12 national standards published in 2017 (these are outlined in detail in Appendix A).

In Northern Ireland performance data on the NBSP is obtained from two main sources - the Child Health System and the Regional Newborn Screening Laboratory. Using agreed templates, data is reported annually to the PHA (regionally) and Public Health England (nationally).

Headline results

The most recently published national report (2016-17), which describes performance against national standards in each region of the UK[4], shows that the NBSP in Northern Ireland is of high quality and performing well.

Regional data relating to the Northern Ireland NBSP highlight that in 2017-18:

- In terms of coverage, >98% of born and resident babies in Northern Ireland had a conclusive screening result for each of the five conditions recorded on the child health system by 17 days of age.

- In relation to timing of sample collection and processing, 94.9% of samples collected on day 5, and 98.0% of samples received in the newborn screening laboratory within 3 working days of collection.

- 100% of positive screening results (for PKU, CHT and MCADD) were available, and clinical referral had been initiated within 3 days of the sample being received by the screening laboratory.

[4] PHE 2018 accessed at: https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/709367/Newborn_blood_spot_screening_data_collection_and_performance_analysis_report_2016_to_2017.pdf

- In relation to timeliness of receipt into clinical care, the programme in Northern Ireland exceeded acceptable national standards. Northern Ireland met the achievable standard for timeliness of first appointment for CF screen positive babies with 2 mutations, with 100% of babies seen by 28 days.

- Over 23,000 babies had newborn blood spot screening testing. In total, across all of the five conditions tested for, 49 babies were identified as screen positive, and 24 of these babies were confirmed as having one of the conditions.

Areas for further improvement

Whilst the NBSP in Northern Ireland consistently performs well against national standards, work continues to promote quality improvement, safety, efficiency and innovation in the programme.

Key Performance Indicators (KPIs) focus specifically on identifying areas for improvement which will allow for targeted quality improvement initiatives to be developed where necessary. One such area that has been identified for improvement across the UK is the avoidable repeat rate.

An avoidable repeat refers to a sample that has not met the required quality standard to be accepted by the laboratory for analysis. For example, an insufficient quantity of blood may have been collected and in this instance the laboratory will request a repeat sample.

At a national level, meeting the standard (acceptable = ≤2%; achievable = ≤1%) in relation to avoidable repeats has proved challenging since the introduction of the programmes and variation exists across the UK.

In Northern Ireland the avoidable repeat rate for 2017-18 was 4.95% which did not meet the acceptable or achievable standard. The regional NBSP QI group

continues to work to understand and reduce the avoidable repeat rate, including scoping potential variance across Northern Ireland. Within HSCTs, additional training was provided to midwifery staff in the use of a new incision device and understanding of the new laboratory quality acceptance criteria. A ‘Once is enough’ information leaflet that had previously been developed by the group, was recirculated as a refresher guide to all test takers within HSCTs. This concise publication provides guidance to support accurate first-time completion of blood spot sampling. Photographic evidence of what constitutes acceptable and unacceptable blood spot samples was also shared with test takers to provide a visual aid to help improve the quality of the first sample.

The Regional NBSP QI group is committed to continual improvement. This includes seeking ways to resolve issues that are identified within the programme and encouraging shared learning, in order to ensure consistency of service delivery across the region.

APPENDIX A: DATA SOURCES, CORE DEFINITIONS AND UK NEWBORN BLOOD SPOT SCREENING PROGRAMME STANDARDS, 2017

In Northern Ireland, data related to the NBSP is obtained from two main sources - the Child Health System (CHS) and the Regional Newborn Screening Laboratory. Using agreed templates, data is reported annually to the PHA (regionally) and Public Health England (nationally).

Child Health System (CHS) data

There are four CHS areas in Northern Ireland and these collectively cover the five HSCT geographies, i.e. Eastern CHS (BHSCT and SEHSCT) Northern (NHSCT) Southern (SHSCT) and Western (WHSCT).

Data provided by the CHS relates to the baby's area of residence at time of reporting.

Laboratory data

The Northern Ireland Newborn Screening Laboratory is provided by Belfast HSCT and located on the Royal Victoria Hospital site. The laboratory processes all newborn blood spot samples in Northern Ireland, and is UKAS (UK Accreditation Service) ISO 15189 accredited for NBS screening and diagnosis.

Laboratory data provided in this report refers to samples received in the Regional Newborn Screening Laboratory between 1st April 2017 and 31st March 2018 and is reported by the Trust work location of the test taker and not the baby' residence area.

As CHS and laboratory data systems define cohorts of interest differently, (outlined above), corresponding totals may vary. For example, the total number of resident babies screened in CHS is calculated based on babies who are screened during 2017-18 and who remain resident at year end. However, the number of first samples received in the laboratory in a given year will also include babies who

move out before year end as well as babies who were born in the previous year, but the sample was received in 2017-18.

Key definitions

'Born and Resident'

Babies who were born to Northern Ireland residents (at time of birth) in 2017-18 and were still resident in Northern Ireland at 31st March 2018.

'Movers in'

Babies who were born in 2017-18, who moved to a CHS area of Northern Ireland between 1st April 2017 and 31st March 2018, who were not born to residents of that CHS area and were still resident at 31st March 2018.

'Carrier' status

To have certain genetic conditions, such as cystic fibrosis or sickle cell disease, an individual must possess two copies of an altered gene (a gene mutation) inherited from parents, both of whom are carriers of that altered gene (gene mutation). A carrier only has one copy of the altered gene (gene mutation) and so does not have the condition, but may pass the gene mutation to their children[5,6]

NI Result Status Codes

There are a number of potential result outcomes for blood spot samples. A standard set of result status codes (see below) are used for reporting which ensures uniformity of result reporting.

Status

01 – Specimen received in laboratory
02 – Declined
03 – Repeat / Further sample required (*see Reason for Repeat Test*)

[5] Patient info 2018 www.patient.info/health/genetic-testing
[6] NHS choices 2018 www.nhs.uk/conditions/genetics/inheritance

04 – Not suspected
05 – Carrier (CF / SCD)
06 – Carrier of other haemoglobin (SCD)
07 – Not suspected – other disorder follow-up
08 – Suspected
09 – Not screened / screening incomplete *(see Reason Not Screened / Screening Incomplete)*
10 – Not suspected – no other Hb/thal excluded (SCD)

Reason For Repeat Test

(A) Raised tyrosine (PKU)
(B) Too young for reliable screening
(C) Too soon after blood transfusion (<72 hours)
(D) Unsuitable sample
(E) Insufficient sample
(F) Unsatisfactory analysis
(G) Borderline result (PKU-tyrosine/CHT)
(H) Inconclusive (CF)/SCD
(I) Sickle – Transfusion
(J) Too premature for testing SCD
(K) Moved In – Reason Unknown
(L) Preterm CHT

Reason Not Screened / Screening Incomplete

(1) Died
(2) Unreliable result
(3) Moved out of area
(4) CF too old > 8 week

National Standards

The performance of the NI NBSP in 2017-18 is based upon standards that were introduced across the UK in 2017. Changes to the standards from 2013 are summarised in the table below.

Standard	Changes
Standard 1a: Coverage (CCG responsibility at birth)	• PKU reported as proxy for all IMDs • Clarified definition • Change to achievable thresholds
Standard 1b: Coverage (movers in)	• PKU reported as proxy for all IMDs • Clarified definition • Change to achievable thresholds
Standard 2: Timely identification of babies with a null or incomplete result recorded on the CHIS	• No change
Standard 3: Barcoded NHS number label is included on the blood spot card	• Change to standard to drive improvement in the use of barcoded NHS number labels as NHS number is mandatory • Acceptable threshold reflects data; achievable threshold remains the same • Denominator excludes samples received from places with no NHS number
Standard 4: Timely sample collection	• Change to standard to measure taking the sample on day 5 only • In mitigating circumstances samples can be taken between day 6 and day 8 inclusive • Numerator and denominator exclude pre-transfusion samples • Change to thresholds to reflect data
Standard 5: Timely receipt of a sample in the newborn screening laboratory	• Change to standard to drive improvement in timely receipt of samples • Numerator and denominator exclude pre-transfusion samples • Change to thresholds to reflect data • Mitigation added

Standard	Changes
Standard 6: Quality of the blood spot sample	• Clarified definition • Change to achievable threshold
Standard 7a: Timely taking of a second blood spot sample for CF screening	• Only includes second samples taken for raised immunoreactive trypsinogen (IRT) – reporting mechanism under development for second samples • Change to standard to measure taking the second sample for raised IRT on day 21 to day 24 • Change to thresholds to reflect data • In mitigating circumstances the second sample for raised IRT can be taken between day 25 and day 28 inclusive
Standard 7b: Timely taking of a second blood spot sample following a borderline CHT screening	• Only includes second samples taken for borderline thyroid stimulating hormone (TSH) – reporting mechanism under development for second samples
Standard 7c: Timely taking of a second blood spot sample for CHT screening for preterm infants	• Only includes second samples taken for thyroid stimulating hormone (TSH) in preterm infants – reporting mechanism under development for second samples
Standard 8: UKAS (screening)	• Laboratories undertaking screening must be accredited by the United Kingdom Accreditation Service (UKAS)
Standard 9: Timely processing of CHT and IMD screen positive samples	• Standard includes IMDs excluding HCU • Single threshold of 100% referrals within 3 working days • Updated CHT sample definition
Standard 10: UKAS (diagnosis)	• Laboratories undertaking screening must be accredited by the United Kingdom Accreditation Service (UKAS)
Standard 11: Timely entry into clinical care	• Standard includes IMDs

Standard	Changes
Standard 12a: Timeliness of results to parents (CCG responsibility at birth)	• Standard retained • Audit tool to be developed to measure standard • Updated definitions section
Standard 12b: Timeliness of results to parents (movers in)	• New standard 12b • Audit tool to be developed to measure standard

The 12 standards are summarised below in Table 1.

Table 1: UK Newborn Blood Spot Screening Programme Standards, 2017[7]

Standard		Description	Acceptable	Achievable
1a	Completeness of coverage (CCG[8] responsibility at birth)	The proportion of *eligible babies for whom a conclusive screening result for each of the nine conditions is recorded on the child health information system (CHS) by 17 days of age. **Eligible babies (denominator) is the total number of babies born within the reporting period, excluding any baby who died before the age of eight days. For the purposes of this standard, the cohort includes babies for whom the CCG[8] was*	≥ 95.0% (for all conditions)	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD

[7]NHS Newborn Blood Spot Screening Programme Standards, 2017.
http://newbornbloodspot.screening.nhs.uk/standards
[8]CCG – Clinical Commissioning Group (Northern Ireland data are provided by Child Health System (CHS) area)

Standard		Description	Acceptable	Achievable
1a	Completeness of coverage (CCG[8] responsibility at birth) *(cont'd)*	*responsible at birth and remains responsible on the last day of the reporting period*.		
1b	Completeness of coverage (movers in)	The proportion of **eligible babies for whom a conclusive screening result for each of the nine conditions is recorded on CHS by 21 calendar days of movement in being recorded on the CHS. ***Eligible babies (denominator) is the total number of babies born within the reporting period and equal to or less than 364 days old. For the purposes of this standard, the cohort includes only babies who have moved in and become the responsibility of the CCG[8] during the reporting period and for whom the CCG[8] remains responsible on the last day of the reporting period.*	≥ 95.0% (for all conditions)	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD

[8]CCG– Clinical Commissioning Group (Northern Ireland data are provided by Child Health System (CHS) area)

Standard		Description	Acceptable	Achievable
2	Timely identification of babies with a null or incomplete result recorded on CHS	Child health records departments perform regular checks for null or incomplete results. Failsafe reports are produced and action taken to follow-up, according to local protocols. There can be flexibility in frequency and age range of reports providing the method complies with the acceptable performance threshold.	100% perform regular checks to identify babies ≥17 days and ≤364 days with a null or incomplete result.	100% perform regular checks to identify babies ≥14 days and ≤364 days with a null or incomplete result.
3	Barcoded NHS number label is included on the blood spot card	The proportion of blood spot cards received by the laboratory with the baby's NHS number (or UK equivalent) on a barcoded label. *Use of a barcoded NHS number (or UK equivalent) label will reduce the risk of an inaccurate NHS number (or UK equivalent) on the blood spot card which would require a repeat sample to be taken.*	≥90.0% of cards received by a laboratory with the baby's NHS (or UK equivalent) number on a barcoded label	≥95.0% of cards received by a laboratory with the baby's NHS (or UK equivalent) number on a barcoded label
4	Timely sample collection	The proportion of first samples taken on day 5 (excludes pre-transfusion samples)	≥90.0%	≥95.0%

Standard		Description	Acceptable	Achievable
5	Timely receipt of a sample in the laboratory	Proportion of all samples received less than or equal to 3 working days of sample collection (excludes pre-transfusion samples)	≥95.0% of all samples received less than or equal to 3 working days	≥99.0% of all samples received less than or equal to 3 working days
6	Quality of the blood spot sample	Proportion of first blood spot samples received that required repeating due to an avoidable failure in the sampling process because the sample was: • Taken when the baby was too young (on or before day 4) • Insufficient blood • Unsuitable sample/card	≤2%	≤1%
7a	Timely taking of a second blood spot sample for CF screening	The proportion of second blood spot samples for raised IRT taken on day 21 to day 24 (day of birth is day 0)	≥95% of second samples taken on day 21 to day 24	≥70% of second samples taken on day 21
7b	Timely taking of a second blood spot sample following a borderline CHT screening	The proportion of second blood spot samples for borderline TSH taken between 7 and 10 calendar days after the initial borderline sample.	≥95%	≥99%
7c	Timely taking of a second blood spot	The proportion of second blood spot samples taken on or	≥95%	≥99%

Standard		Description	Acceptable	Achievable
7c	sample for CHT screening for preterm infant	before 28 days of age. Only taken earlier if baby discharged home.		
8	UKAS (screening)	UKAS accredits pathology laboratories against a set of defined standards which are allied to international standards for competence in medical laboratories – ISO 15189. During the newborn screening specialist assessment UKAS looks at both the ISO standards and the UK screening specific laboratory standards, as an integrated process	Laboratory is UKAS accredited with specialist assessment of NBS screening by the next full visit	
9	Timely processing of CHT and IMD (excluding HCU) screen positive samples	The proportion of CHT and IMD (excluding HCU) screen positive screening results available and clinical referral initiated within 3 working days of sample receipt by screening laboratory.	100% within 3 working days	N/A
10	UKAS (diagnosis)	UKAS accredits pathology laboratories against a set of defined standards which are allied to international standards for competence in medical laboratories – ISO 15189	Laboratory is UKAS accredited	

Standard		Description	Acceptable	Achievable
11	Timely entry into clinical care	A baby in whom an IMD (excluding HCU) and CHT (on first sample) is suspected should attend their first clinical appointment by:	100% by 14 days of age	
		A baby in whom CHT is suspected on a repeat blood spot sample that follows a borderline TSH should have their first clinical appointment by:	100% by 21 days of age	
		A baby in whom CF is suspected (2 CFTR mutations detected) and HCU should have their first clinical appointment by:	≥95% by 28 days of age	100% by 28 days of age
		A baby in whom CF is suspected (none or one CFTR mutation detected) should have their first clinical appointment by:	≥80% by 35 days of age	100% by 35 days of age
		A baby in whom SCD is suspected should attend first clinical appointment by 90 days of age:	≥90%	≥95%
12a	Timeliness of results to parents (CCG responsibility at birth)	The proportion of babies with a not suspected result for each of the conditions for whom a not suspected results letter was despatched directly to parents by the CHRD within 6 weeks of birth.	100%	N/A

Standard		Description	Acceptable	Achievable
12b	Timeliness of results to parents (movers in)	The proportion of babies with a not suspected result for each of the conditions screened for whom a not suspected results letter was despatched directly to parents by the CHRD within 6 weeks of notification of movement in.	100%	N/A

Application of 2017 standards to NBSP in NI - Key information

Standards 1a and b

PKU data is reported as a proxy to MCADD data in line with UK reporting.

Standard 1b

Northern Ireland is currently unable to report on the number of babies tested and recorded on CHS within 21 days of 'movement in' being recorded on the Child Health System; this would require a software development.

Standard 3

The use of barcoded labels with Health and Care Number (equivalent to NHS number) on blood spot cards is currently not mandatory in Northern Ireland. There are plans to make the use of Health and Care Number on blood spot cards compulsory in the future.

Standard 7 (a - c)

Data on Standard 7 is currently not collected in the UK as information management systems do not currently support collection of data for this standard.

Standard 8 and 10

Compliance with Standards 8 and 10 is monitored as part of the UKAS accreditation (previously known as Clinical Pathology Accreditation (CPA)) of the

Regional Newborn Screening Laboratory. The Northern Ireland Regional Newborn Screening Laboratory has achieved this accreditation.

Standard 12

In Northern Ireland, instead of issuing negative result letters to parents, negative results are given directly to parents by the health visitor at the 6 – 8 week health review visit for insertion into the PCHR (Personal Child Health Record – 'red book'). Performance against this specific standard is therefore currently not reported.

The performance of the NBSP in Northern Ireland 2017-18 against each of the UK standards is outlined in Table 2.

APPENDIX B: PERFORMANCE OF NBSP IN NI 2017-18

Table 2: Performance of the Northern Ireland Newborn Blood Spot Screening Programme 2017-18 *(data collected by NI Child Health System)*

	Standard	Performance 2017-18			Acceptable	Achievable
1a	Total number of 'born and resident' babies = 22,582				≥ 95.0%	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD
	Completeness of coverage (CCG responsibility at birth) by Day 17 (Numbers and %)	PKU[9]	22,362	99.03%		
		CHT	22,155	98.11%		
		CF	22,302	98.76%		
		SCD	22,356	99.00%		
	Declines to screening (Numbers and %)	PKU[9]	9	0.04%		
		CHT	9	0.04%		
		CF	10	0.04%		
		SCD	9	0.04%		
1b	Total number of 'movers in' babies = 356				≥ 95.0%	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD
	Completeness of coverage (movers in)	PKU[9]	303	85.11%		
		CHT	305	85.67%		
		CF	279	78.37%		
		SCD	306	85.96%		
	Declines to screening (Numbers and %)	PKU[9]	52	14.61%		
		CHT	50	14.04%		
		CF	48	13.48%		
		SCD	49	13.76%		
2	Timely identification of babies with a null or incomplete result recorded on CHS	The child health records departments (four) perform regular weekly checks for 9[10]conditions for babies aged 11-364 days		100%	100% perform regular checks to identify babies ≥17 days and ≤364 days with a null or incomplete result	100% perform regular checks to identify babies ≥14 days and ≤364 days with a null or incomplete result

[9] PKU is reported as proxy to MCADD data in line with UK reporting

[10] 5 conditions only screened for in Northern Ireland at this time

Standard		Performance 2017-18		Acceptable	Achievable
3	Barcoded NHS number (or UK equivalent) is included on the blood spot card	This is currently not mandatory in NI	-	≥ 90.0% of cards received by a laboratory with the baby's NHS number (or UK equivalent) on a barcoded label	≥ 95.0% of cards received by a laboratory with the baby's NHS number (or UK equivalent) on a barcoded label
4	Timely sample collection	The proportion of first samples taken on day 5 (excludes pre-transfusion samples) Total number of first samples taken = 22,953	94.89% (21,780)	≥ 90.0%	≥ 95.0%
5	Timely receipt of a sample in the laboratory	The proportion of all samples received less than or equal to 3 working days of sample collection (excludes pre-transfusion samples) Total number of all samples = 25,222	97.98% (24,712)	≥ 95.0% of all samples received less than or equal to 3 working days	≥ 99.0% of all samples received less than or equal to 3 working days

Standard		Performance 2017-18		Acceptable	Achievable
6	Quality of the blood spot sample (avoidable repeat rate)	Total number of first samples = 23,072 Total number of avoidable repeats = 1,143	4.95%	≤ 2%	≤ 1%
9	Timely processing of all CHT and IMD (excluding HCU) screen positive samples	PKU 7/7 MCADD 3/3 CHT 21/21	100% within 3 working days for each condition	100% within 3 working days	N/A
11	Timely entry into clinical care	Age range at first appointment (in days) PKU 7/7 MCADD 3/3 CHT* 12/13 **suspected on first sample*	 100% by 14 days of age 100% by 14 days of age 92.31% by 14 days of age	100% by 14 days of age	N/A
		CHT** 6/6 ***suspected on a repeat sample*	100% by 21 days of age	100% by 21 days of age	
		CF 8/8 (*2 mutations*)	100% by 28 days of age	≥ 95% by 28 days of age	100% by 28 days of age

Standard		Performance 2017-18		Acceptable	Achievable
11	Timely entry into clinical care *(cont'd)*	CF 7/10 (*1 or 0 mutations*)	70% by 35 days of age	≥80% by 35 days of age	100% by 35 days of age
		SCD A baby in whom SCD is suspected should attend first clinical appointment by 90 days of age:	N/A	≥90%	≥95%
12a	Timeliness of results to parents (CCG responsibility at birth)	The proportion of babies with a not suspected result for each of the conditions for whom a not suspected results letter was despatched directly to parents by the CHRD within 6 weeks of birth.	NI is currently unable to report on this standard	100%	

Standard		Performance 2017-18		Acceptable	Achievable
12b	Timeliness of results to parents (movers in)	The proportion of babies with a not suspected result for each of the conditions screened for whom a not suspected results letter was despatched directly to parents by the CHRD within 6 weeks of notification of movement in.	NI is currently unable to report on this standard	100%	

Trends in performance

Completeness of Coverage (Standards 1a and 1b)

Monitoring coverage allows us to examine whether all babies who are eligible for the newborn blood spot test, including (a) those born and resident in NI and (b) those who move into a CHS area from another area of NI or from outside NI, are offered the screening test.

Table 3a shows consistently high performance in relation to coverage in babies 'born and resident' in NI. Similar to 2016-17 and 2015-16, during 2017-18 the programme exceeded the acceptable standard for completeness of coverage, with more than 98% of 'born and resident' babies with conclusive results for all conditions recorded on CHS by 17 days of age and 99.9% of all born and resident babies having conclusive results by the end of the reporting period.

Table 3a Coverage - Born and Resident (B&R) - Standard 1a[11] *(data collected by NI Child Health System)*

	Year		
	2017-18	**2016-17**	**2015-16**
Total number '**born and resident'** (**B&R**) at 31st March	22,582	23,584	23,858
Number of **B&R** with decline to screening (02)			
PKU[9]	9 0.04%	16 0.07%	16 0.07%
CHT	9 0.04%	16 0.07%	16 0.07%
CF	10 0.04%	16 0.07%	18 0.08%
SCD	9 0.04%	16 0.07%	16 0.07%
Number (%) of **B&R** with conclusive results by the end of the reporting period (codes 04, 05, 06, 07, 08, 10)			
PKU[9]	22,568 99.94%	23,568 99.93%	23,840 99.92%
CHT	22,567 99.93%	23,565 99.92%	23,840 99.92%
CF	22,567 99.93%	23,563 99.91%	23,838 99.92%
SCD	22,568 99.94%	23,568 99.93%	23,840 99.92%
Number (%) of **B&R** with conclusive results available by Day 17 (by 17 days of age) codes 04, 05, 06, 07,08,10)			
PKU[9]	22,362 99.03%	23,315 98.86%	23,644 99.10%
CHT	22,155 98.11%	23,142 98.13%	23,448 98.28%
CF	22,302 98.76%	23,233 98.51%	23,620 99.00%
SCD	22,356 99.00%	23,323 98.89%	23,642 99.09%

The NBSP in NI is currently unable to report on the completeness of coverage for 'mover in' babies by 21 calendar days of 'movement in' being recorded on the CHS (Standard 1b); this would require software development. However, by the end of the reporting period, compared with 2016-17, there was an increase in the proportion of 'mover in' babies with a conclusive result for PKU (85.11% compared to 77.75% in 2016-17).

[9] PKU is reported as proxy to MCADD data in line with UK reporting

[11] excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required, 09 = incomplete/not screened

It is usual to expect to see a lower number of babies tested for CF than for the other conditions, given that the screening test is not reliable as an indicator of CF over 8 weeks of age and therefore is not undertaken beyond this age. This will apply to some babies who move into NI after birth. In 2017-18 there were 29 ‘mover in’ babies who had no conclusive result for CF, as they were over 8 weeks old and therefore too old (code 09/4) for testing for CF at this point. This is a decrease from 2016-17 and 2015-16 when 45 and 44 babies (respectively) were over 8 weeks old.

Table 3b Coverage - Born and Resident (B&R) by Child Health Area - Standard 1a[11] *(data collected by NI Child Health System)*

	Eastern CHS Area		Northern CHS Area		Southern CHS Area		Western CHS Area		N Ireland	
Daily Search for Untested Babies performed?										
- Day 14	No		No		No		No		-	
- Day 17	No		No		No		No		-	
If different, please describe	Weekly CHS 'failsafe' reports are run for babies (births to N Ireland residents and movers in) with incomplete results (result codes not equal to 02, 04, 05, 07, 08, 09 or 10) and aged between 11 and 364 days. These reports are validated by laboratory staff to identify babies for whom samples have been received, prior to follow up by clinical staff.									
Total number **born and resident** (**B&R**) at 31/3/18	8139		5357		5293		3793		22582	
Number (%) of **B&R** with decline to screening (02):										
PKU[9]	3		3		2		1		9	0.04%
CHT	3		3		2		1		9	0.04%
CF	4		3		2		1		10	0.04%
SCD	3		3		2		1		9	0.04%
Number (%) of **B&R** with reason for not starting/ incomplete screening (09):										
PKU[9]	4		0		0		0		4	0.02%
CHT	5		0		0		0		5	0.02%
CF	4		0		0		0		4	0.02%
SCD	4		0		0		0		4	0.02%
Number (%) of **B&R** with conclusive results (04, 05, 06, 07, 08, 10):										
PKU[9]	8132	99.91%	5354	99.94%	5290	99.94%	3792	99.97%	22568	99.94%
CHT	8131	99.90%	5354	99.94%	5290	99.94%	3792	99.97%	22567	99.93%
CF	8131	99.90%	5354	99.94%	5290	99.94%	3792	99.97%	22567	99.93%
SCD	8132	99.91%	5354	99.94%	5290	99.94%	3792	99.97%	22568	99.94%
Number (%) of **B&R** with conclusive results available by Day 17 (04, 05, 06, 07, 08, 10):										
PKU[9]	8071	99.16%	5317	99.25%	5243	99.06%	3731	98.37%	22362	99.03%
CHT	7984	98.10%	5278	98.53%	5191	98.07%	3702	97.60%	22155	98.11%
CF	8052	98.93%	5308	99.09%	5224	98.70%	3718	98.02%	22302	98.76%
SCD	8071	99.16%	5317	99.25%	5239	98.98%	3729	98.31%	22356	99.00%

[9] PKU is reported as proxy to MCADD data in line with UK reporting

[11]excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required, 09 = incomplete/not screened

Table 4a – Completeness of coverage – Movers in - Standard 1b[11] *(data collected by NI Child Health System)*

	Year		
	2017-18	**2016-17**	**2015-16**
Total number 'Movers In' (**MI**) resident at 31st March	356	400	356
Number of **MI** with decline to screening (02)			
PKU[9]	52 (14.61%)	79 (19.75%)	61 (17.13%)
CHT	50 (14.04%)	80 (20.00%)	61 (17.13%)
CF	48 (13.48%)	80 (20.00%)	58 (16.29%)
SCD	49 (13.76%)	80 (20.00%)	62 (17.42%)
Number (%) of **MI** with reason for not starting/incomplete (code 09-)			
PKU[9]	0 (0%)	2 (0.50%)	0 (0%)
CHT	0 (0%)	2 (0.50%)	0 (0%)
CF	29 (8.15%)	45 (11.25%)	44 (12.36%)
SCD	0 (0%)	2 (0.50%)	0 (0%)
Number (%) of **MI** with conclusive results(codes 04, 05, 06, 07, 08,10)			
PKU[9]	303 (85.11%)	311 (77.75%)	290 (81.46%)
CHT	305 (85.67%)	310 (77.50%)	290 (81.46%)
CF	279 (78.37%)	267 (66.75%)	250 (70.22%)
SCD	306 (85.96%)	310 (77.50%)	290 (81.46%)

[9] PKU is reported as proxy to MCADD data in line with UK reporting

[11] excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required, 09 = incomplete/not screened

Table 4b – Completeness of coverage – Movers in by Child Health Area - Standard 1b[11] *(data collected by NI Child Health System)*

	Eastern CHS Area		Northern CHS Area		Southern CHS Area		Western CHS Area		N Ireland	
Total number of '**Movers In'** (**MI**) resident at 31/3/18	100		108		68		80		356	
Number of **MI** with decline to screening (02):										
PKU[9]	21		4		10		17		52	
CHT	17		4		11		18		50	
CF	17		4		10		17		48	
SCD	17		4		11		17		49	
Number of **MI** with reason for not starting/incomplete screening (09):										
PKU[9]	0		0		0		0		0	
CHT	0		0		0		0		0	
CF	11		1		6		11		29	
SCD	0		0		0		0		0	
Number (%) of **MI** with conclusive results (04, 05, 06, 07, 08, 10):										
PKU[9]	79	79.00%	103	95.37%	58	85.29%	63	78.75%	303	85.11%
CHT	83	83.00%	103	95.37%	57	83.82%	62	77.50%	305	85.67%
CF	72	72.00%	103	95.37%	52	76.47%	52	65.00%	279	78.37%
SCD	83	83.00%	103	95.37%	57	83.82%	63	78.75%	306	85.96%

[9] PKU is reported as proxy to MCADD data in line with UK reporting

[11] excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required, 09 = incomplete/not screened

Timely identification of babies with a null or incomplete result recorded on CHS (Standard 2)

CHS 'failsafe' reports are produced weekly in each CHS Bureau/Central Office in Northern Ireland. These search for and flag up babies with incomplete results (result status codes not equal to 02, 04, 05, 06, 07, 08, 09 or 10 – see pages 16 - 17 for result status codes definitions) and aged between 11 and 364 days, meeting the UK standard. The reports relate to both babies born to Northern Ireland residents at time of birth and to 'mover in' babies.

Sample identification (Standard 3)

Every person that is born or resident in Northern Ireland should be assigned a unique Health and Care Number (HCN). This number can be used to link health and social care records. Completion of a baby's health and care number on the blood spot card by test takers is recommended good practice but not mandatory in Northern Ireland.

In 2017-18, 87.73% of all samples received by the Laboratory had the HCN included[12]. It is recognised that including the unique HCN is an important additional safety and quality mechanism for identifying and matching baby records in the NBSP; work to improve recording on blood spot cards continued during 2017-18. Northern Ireland, however, does not use barcoded labels with health and care number and therefore is currently unable to report on this standard.

Timely sample collection and processing (Standards 4 and 5)

The NBSP exceeded the acceptable standard for timely sample collection, with more than 98% of first samples taken between 5 to 8 days after birth; this performance is similar to 2016-17 and 2015-16.

[12] Data source: Regional Newborn Screening Laboratory

Table 5a – Sample collection – Standard 4 *(data collected by the Regional Newborn Screening Laboratory)*

	Year		
	2017-18	**2016-17**	**2015-16**
Total number of first samples	22,953	24,171	24,430
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	119	168	145
	Year		
	2017-18	**2016-17**	**2015-16**
Number of first samples taken on or before day 4 (%)	103 (0.45%)	111 (0.46%)	108 (0.44%)
Number of first samples taken on day 5 (%)	21,780 (94.89%)	22,818 (94.40%)	23,066 (94.42%)
Number of first samples taken on or after day 9 (%)	256 (1.12%)	308 (1.27%)	273 (1.12%)
Number of first samples taken between day 5 and day 8 (%)	22,594 (98.44%)	23,752 (98.27%)	24,049 (98.44%)

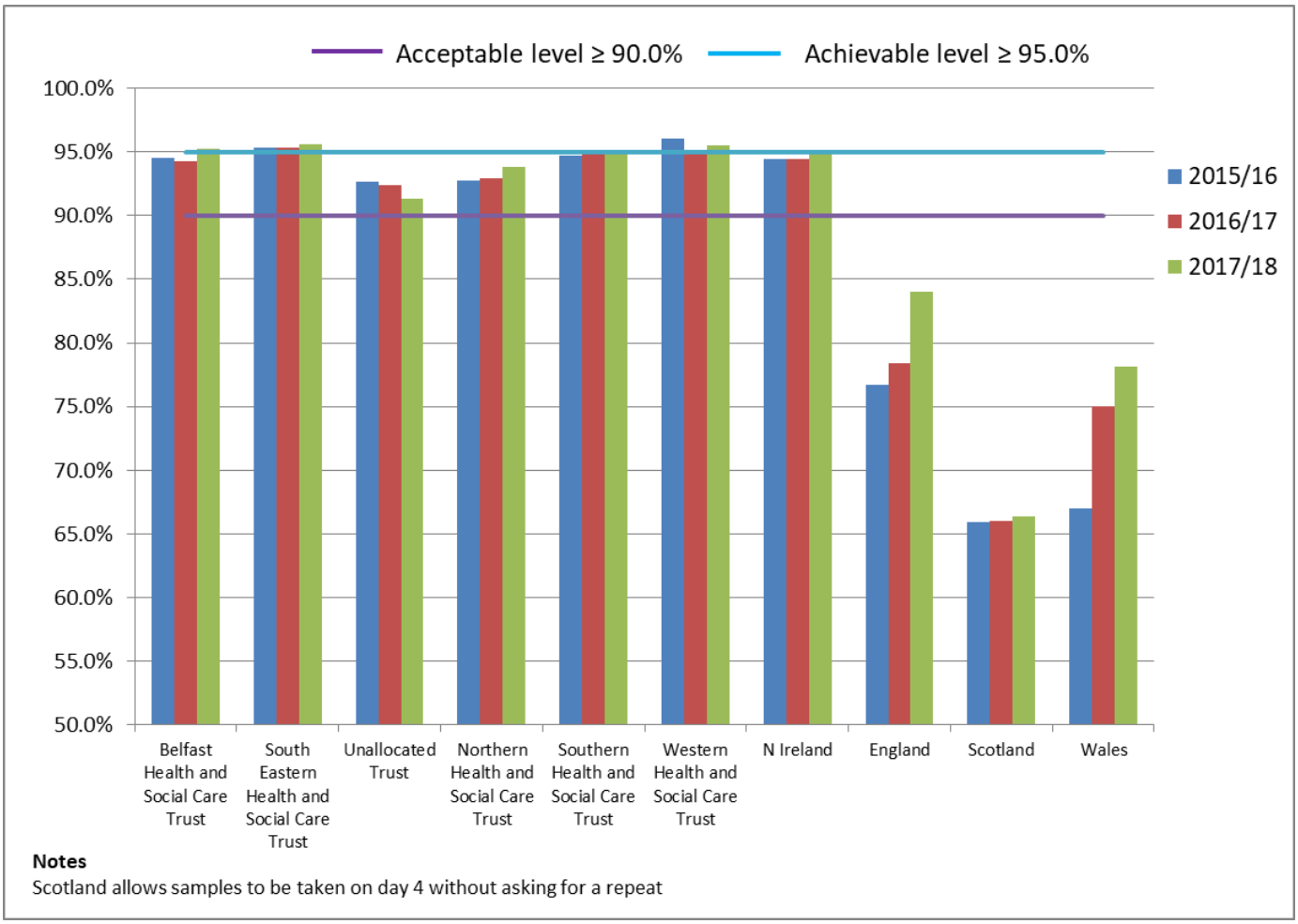
Table 5b – Sample collection – Standard 4 by Trust[13] *(data collected by the Regional Newborn Screening Laboratory)*

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[14]	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
Total number of first samples included in audit	2692	5210	322	5385	5473	3871	22953
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	13	28	2	26	26	24	119
Number of first samples taken on or before day 4	15	26	5	17	28	12	103
Number of first samples taken on day 5	2563	4979	294	5054	5194	3696	21780
Number of first samples taken on or after day 9	34	40	11	47	70	54	256
Number of first samples taken between day 5 and day 8	2643	5144	306	5321	5375	3805	22594
% of first samples taken on or before day 4	0.56%	0.50%	1.55%	0.32%	0.51%	0.31%	0.45%
% of first samples taken on day 5	95.21%	95.57%	91.30%	93.85%	94.90%	95.48%	94.89%
% of first samples taken on or after day 9	1.26%	0.77%	3.42%	0.87%	1.28%	1.39%	1.12%
% of first samples taken between day 5 and day 8	98.18%	98.73%	95.03%	98.81%	98.21%	98.30%	98.44%

[13] Trust, reported by the Regional Newborn Screening Laboratory, relates to the work location of the test taker and not the baby's residence area

[14] Unallocated Trust is assigned where it is not possible to determine from the data entered by the test taker on the card whether the responsible Trust is Belfast Health and Social Care Trust or South Eastern Health and Social Care Trust

Figure 1: Percentage of samples collected on day 5 from 2015-16 to 2017-18



In 2017-18 the programme also exceeded the acceptable standard for timely receipt of a sample in the laboratory (≥ 95.0% of all samples received in the Laboratory within 3 working days).

Table 6a – Timely receipt of a sample in the laboratory - Standard 5 *(data collected by the Regional Newborn Screening Laboratory)*

	Year		
	2017-18	**2016-17**	**2015-16**
Total number of ALL samples (first, repeat and second samples) included	25,222	26,807	26,517
Number of samples EXCLUDED (because date of specimen was not recorded on the cards)	146	200	182
Number of ALL samples received by the lab in 3 or fewer working days of sample being taken (%)	24,712 (97.98%)	26,410 (98.52%)	26,187 (98.76%)
Number of ALL samples received by the lab in 4 or fewer working days of sample being taken (%)	25,073 (99.41%)	26,660 (99.45%)	26,407 (99.59%)

Table 6b Timely receipt of a sample in the laboratory by Trust[13] – Standard 5 *(data collected by the Regional Newborn Screening Laboratory)*

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[14]	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
Total number of ALL samples (first, repeat and second samples) included in the audit	3067	5717	355	5785	6024	4274	25222
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	18	42	2	27	30	27	146
Number of all samples received by the lab in 3 or fewer working days of sample being taken	3045	5675	349	5747	5931	3965	24712
Number of all samples received by the lab in 4 or fewer working days of sample being taken	3057	5709	352	5769	5990	4196	25073
Number of all samples received by the lab on or after 5 working days	10	8	3	16	34	78	149

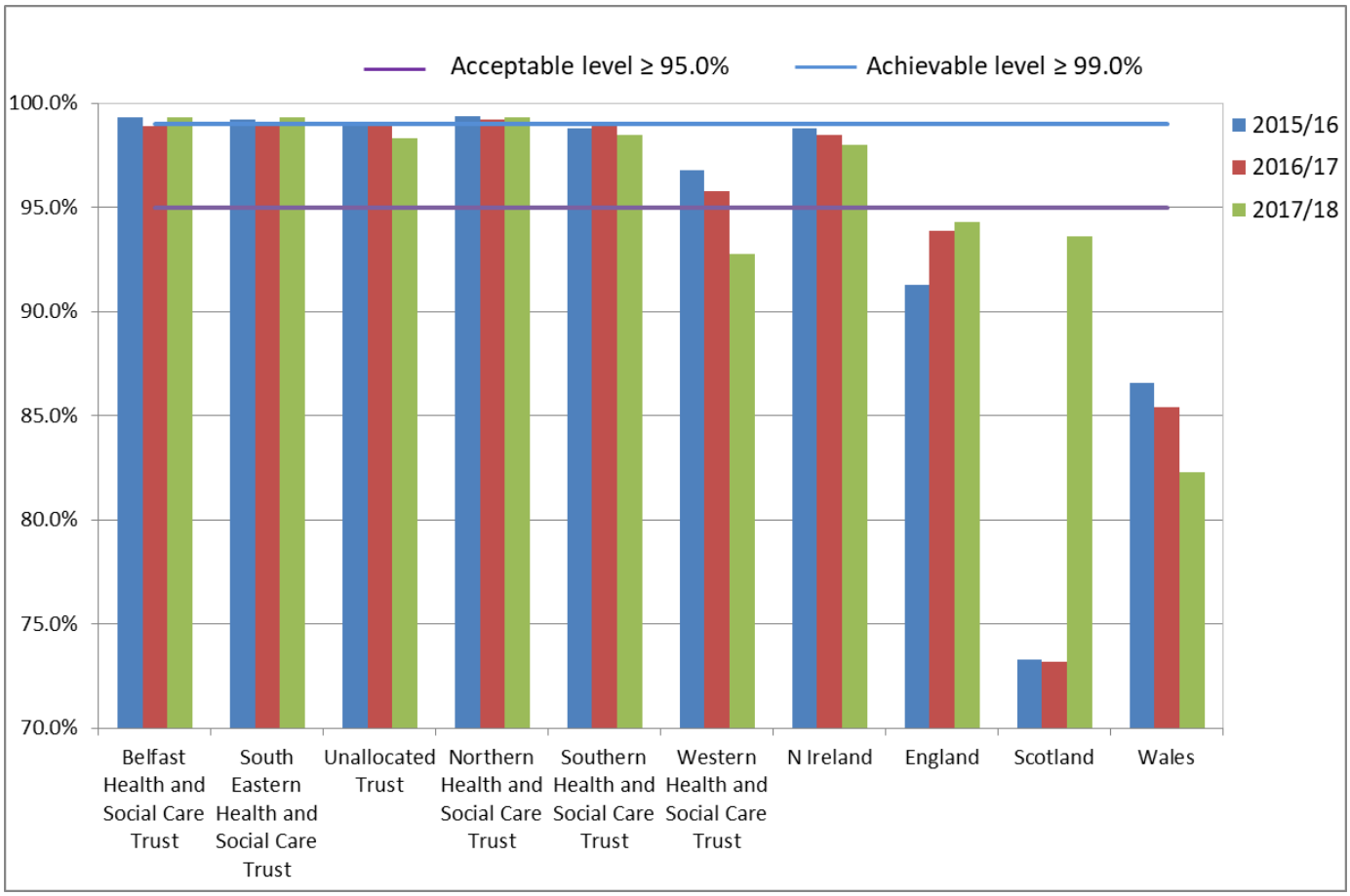
	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[14]	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
of sample being taken							
% of all samples received by lab in 3 or fewer working days of sample being taken	99.28%	99.27%	98.31%	99.34%	98.46%	92.77%	97.98%
% of all samples received by lab in 4 or fewer working days of sample being taken	99.67%	99.86%	99.15%	99.72%	99.44%	98.18%	99.41%
% of all samples received by lab in 5 or more working days of sample being taken	0.33%	0.14%	0.85%	0.28%	0.56%	1.82%	0.59%

[13] Trust, reported by the Regional Newborn Screening Laboratory, relates to the work location of the test taker and not the baby's residence area

[14]Unallocated Trust is assigned where it is not possible to determine from the data entered by the test taker on the card whether the responsible Trust is Belfast Health and Social Care Trust or South Eastern Health and Social Care Trust

The Western Trust, with its' wider geographical area, has a lower percentage of samples being received in the laboratory in 3 or fewer days of the sample being taken.

Figure 2: Percentage of samples received by the laboratory in less than or equal to 3 working days of sample collection from 2015-16 to 2017-18



Sample quality (Standard 6)

Avoidable repeat requests is the total number of repeat (second or subsequent) samples requested by the laboratory during the reporting period because the previous sample:

- was taken when the baby was too young (on or before day 4, where day of birth is day 0) (excluding pre-transfusion admission samples);
- had insufficient blood;
- was an unsuitable sample/card (e.g. on an expired blood spot card, contaminated, in transit for more than 14 days, anti-coagulated sample, or baby's details not accurately recorded on the blood spot card).

The avoidable repeat rate in 2017-18 was 4.95% and therefore the NBSP did not meet the acceptable standard (avoidable repeat rate ≤2%) - see Tables 7a and 7b. This was similar to performance in 2016-17 (4.39%) and comparable to other UK countries (Scotland 5.72% Wales 5.56%) except England (2.89%).[15]

Table 7a - Quality of the blood spot sample - Standard 6 *(data collected by the Regional Newborn Screening Laboratory)*

	Year		
	2017-18	**2016-17**	**2015-16**
Total number of first samples received	23,072	24,339	24,575
Total number of avoidable repeat samples requested	1,143	1,378	1,302
REASON FOR REPEAT			
Avoidable Repeats			
Too young for reliable screening (≤ 4 days) (%)	104 (0.45%)	105 (0.43%)	108 (0.44%)
Insufficient sample	857 (3.71%)	724 (2.97%)	460 (1.87%)
Unsuitable sample[16]	182 (0.79%)	239 (0.98%)	441 (1.79%)
Total Avoidable Repeats	**1,143 (4.95%)**	**1,068 (4.39%)**	**1,009 (4.11%)**

[15] PHE 2018 https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/709367/Newborn_blood_spot_screening_data_collection_and_performance_analysis_report_2016_to_2017.pdf

[16] Unsuitable sample = missing data, card past expiry, sample anti-coagulated, over-layered or contaminated, >14 days transit

Table 7b - Quality of the blood spot sample by Trust - Standard 6 *(data collected by the Regional Newborn Screening Laboratory)*

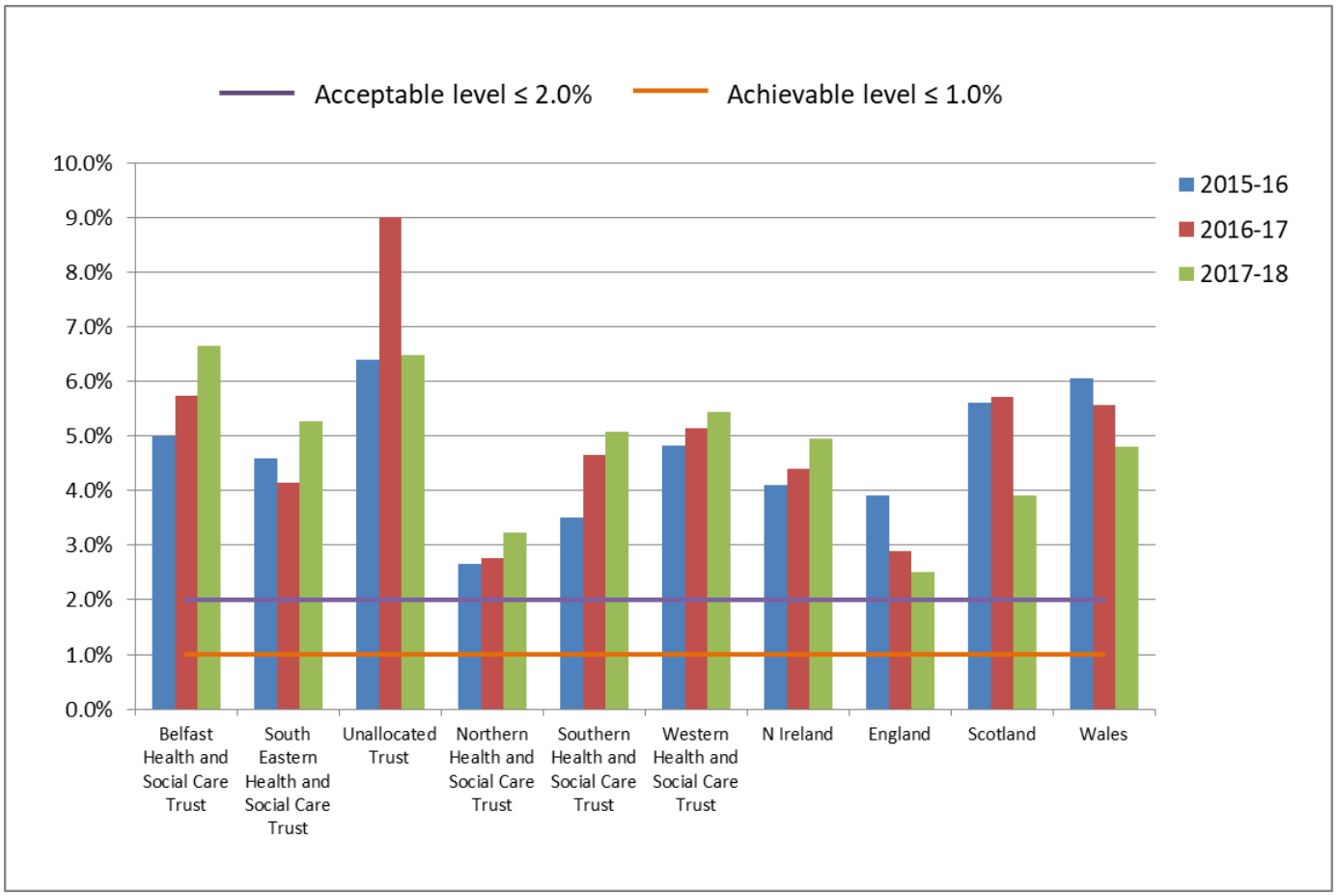
	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[14]	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
Total number of first samples received	2,705	5,238	324	5,411	5,499	3,895	23,072
Total number of repeat samples requested	227	330	24	244	336	244	1405
REASON FOR REPEAT							
Avoidable Repeats							
Too young for reliable screening (≤4 days)	15	26	5	18	28	12	104
Insufficient sample	147	200	13	113	223	161	857
Unsuitable sample[16]	18	50	3	44	28	39	182
Total Avoidable Repeats	**180**	**276**	**21**	**175**	**279**	**212**	**1,143**
Avoidable Repeats Request Rates							
% Too young for reliable screening (≤4 days)	0.55%	0.50%	1.54%	0.33%	0.51%	0.31%	0.45%
% Insufficient sample	5.43%	3.82%	4.01%	2.09%	4.06%	4.13%	3.71%
% Unsuitable sample[16]	0.67%	0.95%	0.93%	0.81%	0.51%	1.00%	0.79%
% Avoidable Repeats	**6.65%**	**5.27%**	**6.48%**	**3.23%**	**5.07%**	**5.44%**	**4.95%**
Unavoidable Repeats[17]							
Total Unavoidable Repeats	**47**	**54**	**3**	**69**	**57**	**32**	**262**
Unavoidable Repeats Request Rates							
% Unavoidable Repeats	**1.74%**	**1.03%**	**0.93%**	**1.28%**	**1.04%**	**0.82%**	**1.14%**

[14] Unallocated Trust is assigned where it is not possible to determine from the data entered by the test taker on the card whether the responsible Trust is Belfast Health and Social Care Trust or South Eastern Health and Social Care Trust

[16] Unsuitable sample = missing data, card past expiry, sample anti-coagulated, over-layered or contaminated, >14 days transit

[17] Unavoidable repeat = too premature for testing (SCD), preterm CHT, borderline CHT or inconclusive CF

Figure 3: Avoidable Repeat Request Rates from 2015-16 to 2017-18



In 2017-18, the majority of avoidable repeats (3.7% of first samples) were due to insufficient sampling (Table 7a). Potential contributory factors may have included the introduction of more stringent laboratory acceptance criteria following implementation of new UK standards for blood spot quality in February 2016. The introduction of a new incision device for sample collection in NI on 1st March 2017 may also have had an impact (see below). However, national trend data highlights that the issue of avoidable repeats has been a long term problem across all regions of the UK.

The NHSCT had the lowest rates of avoidable repeats and the BHSCT the highest rates.

The regional NBSP QI group continues to work to understand and reduce the avoidable repeat rate, including scoping potential variance across NI.

Within HSCTs additional training was provided to midwifery staff in the use of a new incision device and to understand the new quality criteria from the Laboratory.

In 2016-17, an information leaflet entitled ‘Once is enough’, which had been previously developed by the QI group, was reissued as a refresher guide to all test takers within HSCTs. This concise publication provides guidance to support accurate first-time completion of blood sample extraction. Photographic evidence of what constitutes acceptable and unacceptable blood spot samples was also shared regionally to provide a visual aid to improve quality of the first sample. In addition, a training video on blood spot sampling was shared with staff in all Trusts.

Screen positive results- Timely referral and clinical assessment (Standards 9 and 11)

PKU and MCADD

In 2017-18, 7 babies (including 1 who was tested early due to a family history) were identified with positive screening tests for PKU, and 3 babies were identified as having positive screening tests for MCADD. As in previous years, all (100%) of these babies were referred within 3 working days of sample receipt in the laboratory and seen by clinical teams by 14 days of age, therefore meeting the acceptable standard (by 14 days of age - see Tables 8a and b).

<u>Outcomes</u>

A diagnosis of PKU was confirmed in 5 babies who were PKU screen positive and a diagnosis of MCADD was confirmed in all 3 babies who were MCADD screen positive.

Table 8a – PKU Clinical Data – Standards 9 and 11 *(data collected by the Regional Newborn Screening Laboratory)*

	Year		
	2017-18	**2016-17**	**2015-16**
Number of babies with screen positive result (status code = 07 - not suspected other disorder follow up or 08 - suspected)	7	4	14
Number of screen positive babies with clinical referral initiated within 3 working days of sample receipt in lab (%)	7 (100%)	4 (100%)	14 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	7 (100%)	4 (100%)	12 (85.7%)

Table 8b MCADD Clinical data – Standards 9 and 11 *(data collected by the Regional Newborn Screening Laboratory)*

	Year		
	2017-18	**2016-17**	**2015-16**
Number of babies with screen positive result (status code = 08 - suspected)	3	2	2
Number of screen positive babies with clinical referral initiated within three working days of sample receipt in lab (%)	3 (100%)	2 (100%)	2 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	3 (100%)	2 (100%)	2 (100%)

CHT

In 2017-18, 13 babies were identified as CHT screen positive on their first blood spot sample; all 13 (100%) were referred to specialist clinical teams within 3 days of sample receipt in the laboratory and 12 (92.31%) were seen by the clinical team by 14 days of age.

A further 8 babies were also identified as screen positive. As in previous years, all 8 (100%) were referred within 3 days of sample receipt in the laboratory. Six of

these babies were positive on a repeat sample following a borderline result (Table 8c) and all 6 (100%) of these babies were seen by the clinical team by 21 days of age.

Outcomes

A diagnosis of CHT was confirmed in 5 (23.81%) of the 21 babies who were identified as screen positive.

Table 8c - CHT Clinical Data - Standards 9 and 11(data collected by the Regional Newborn Screening Laboratory)

	Year					
	2017-18		**2016-17**		**2015-16**	
	Screen positive on first sample	Screen positive on second sample	Screen positive on first sample	Screen positive on second sample	Screen positive on first sample	Screen positive on second sample
Number of babies with screen positive result (status code = 08 - suspected)	13[18]	6	9	10	11	7
Number of screen positive babies with clinical referral initiated within 3 working days of sample receipt in lab (%)	12 (100%)	6 (100%)	9 (100%)	10 (100%)	11 (100%)	7 (100%)
Number of screen positive babies who were seen by 14 days of age (suspected on first sample)	11 (91.67%)	N/A	9 (100%)	N/A	10 (90.91%)	N/A
Number of screen positive babies who were seen by 21 days of age (suspected on a repeat blood spot sample following a borderline result)	N/A	6 (100%)	N/A	10 (100%)	N/A	6 (85.71%)

[18] One baby was diagnosed before screening and has been excluded from the age data

SCD

In 2017-18, no babies were identified as screen positive for SCD; however, the screening results for 40 babies required further testing/assessment: 2 were identified with other potentially clinically significant condition, 22 were identified as sickle cell 'carriers' and 16 as a carrier of another unusual haemoglobin gene.

Table 8d - SCD Clinical Data - Standard 11 *(data collected by the Regional Newborn Screening Laboratory)*

Condition/Disorder	Year		
	2017-18	**2016-17**	**2015-16**
Sickle Cell Disease or Other Potentially Clinically Significant Condition[19]	2	0	4
Condition/Disorder	**Year**		
	2017-18	**2016-17**	**2015-16**
Sickle Cell Carrier or Carrier of Other Unusual Haemoglobin Gene[20] or Possible Benign Disorder[21]	38	43	49

CF

Eight babies were identified on screening as suspected CF with 2 genetic mutations; all (100%) were seen by the clinical team by 28 days of age. All eight babies were confirmed as having CF.

A further 10 babies were identified as suspected CF with 1 or 0 mutations and an IRT ≥ cut-off 2 on screening: nine of these required follow-up. Seven out of 9 babies (77.8%) were seen by the clinical team by 35 days of age. CF was confirmed in 3 of these children.

[19] Other potentially clinically significant condition comprise F, FE, FEA or HbAF

[20] Carrier of other unusual haemoglobin gene comprise FAC, FAD, ?FAD, FAE or FAO^{Arab}

[21] Other possible benign disorder comprise FC, FD, FO^{Arab}, FCA, FDA or $FO^{Arab}A$

Outcomes

In 2017-18, a total of 18 babies were referred into the clinical service for further testing following an initial positive screen for suspected CF. A diagnosis of CF was confirmed in 11 of these babies. In addition, 11 babies were identified as probable carriers of a gene for CF (Table 8g).

Table 8e - CF Clinical Data - Standard 11 *(data collected by the Regional Newborn Screening Laboratory)*

	Year		
	2017-18	**2016-17**	**2015-16**
No. of CF suspected babies with 2 mutations	8	9	9
Number of screen positive babies who were seen by 28 days of age (%)	8 (100%)	9 (100%)	9 (100%)

Table 8f - CF Clinical Data - Standard 11 *(data collected by the Regional Newborn Screening Laboratory)*

	Year		
	2017-18	**2016-17**	**2015-16**
No. of CF suspected babies with 1 or 0 mutations and IRT ≥ cut-off 2	10	9	5
Number of screen positive babies who were seen by 35 days of age	7 (77.8%)	8 (88.9%)	5 (100%)

Table 8g – CF Clinical Data – CF gene carriers *(data collected by the Regional Newborn Screening Laboratory)*

	Year		
	2017-18	**2016-17**	**2015-16**
No. of babies with 1 CF mutation detected and second IRT < cut-off 2	11	18	6

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