



Newborn Blood Spot Screening in Northern Ireland

Annual Report 2018 - 20

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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 2018-20.

The NBSP in Northern Ireland offers all newborn babies a blood spot screening test to identify if they are at increased risk of nine 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 (2018-19)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 2018-19:

- In terms of coverage, >97% of 'born and resident' babies in Northern Ireland had a conclusive screening result for each of the conditions recorded on the child health system by 17 days of age.

¹ PHE 2018 - 2019 annual report accessed at:

[Newborn blood spot screening data collection and performance analysis report 1 April 2018 to 31 March 2019 - GOV.UK \(www.gov.uk\)](https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/684441/newborn-blood-spot-screening-data-collection-and-performance-analysis-report-1-april-2018-to-31-march-2019.pdf)

- In relation to timing of sample collection and processing, 94.3% of samples were collected on day 5 and 98.8% of samples were 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 met and exceeded acceptable national standards. NI met the acceptable standard for timeliness of first appointment for PKU screen positive babies with 100% of babies seen by 14 days of age. The NI programme met the achievable standard for timeliness of first appointment for CF screen positive babies with 100% of babies with 2 mutations seen by 28 days of age and 100% of babies with 0 or 1 mutations seen by 35 days of age.
- Over 23,000 babies had newborn blood spot screening testing. In total, across all of the conditions tested for, 30 babies were identified as screen positive and 18 of these babies were confirmed as having one of the conditions.

Regional data relating to the Northern Ireland NBSP highlight that in 2019-20:

- In terms of coverage, >97% of 'born and resident' babies in Northern Ireland had a conclusive screening result for each of the conditions recorded on the child health system by 17 days of age.
- In relation to timing of sample collection and processing, 94.7% of samples were collected on day 5 and 98.3% of samples were 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 met and exceeded the acceptable national standards. NI met the acceptable standard for timeliness of first appointment for MCADD screen positive babies with 100% of babies seen by 14 days of age. The NI programme met the acceptable standard for timeliness of first appointment for CHT screen positive babies, suspected on first sample, with 100% of babies seen by 14 days of age and 100% of babies, suspected on a repeat sample, seen by 21 days of age.

The NI programme met the achievable standard for timeliness of first appointment for CF screen positive babies with 100% of babies with 2 mutations seen by 28 days of age and met the acceptable standard for CF screen positive babies with 0 or 1 mutations with 80% of babies seen by 35 days of age.

At a national level, meeting the standard (acceptable = $\leq 2\%$; achievable = $\leq 1\%$) 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 in 2018-19 was 5.44% and in 2019-20 was 6.03%. The regional NBSP Quality Improvement (QI) group continues to work to understand and reduce avoidable repeats.

SECTION A: INTRODUCTION AND HEADLINE RESULTS FOR 2018-20

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 a number of rare, but serious, inherited conditions (these are described on pages 6 - 8). 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 2018 - 31st March 2020 (hereafter referred to as 2018-20) against national standards.

In Northern Ireland the NBSP currently offers screening for nine 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 disability.

²Department 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>

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.

- **6 Inherited Metabolic Disorders (IMDs):**

1. Phenylketonuria (PKU)
2. Medium Chain acyl-CoA Dehydrogenase Deficiency (MCADD)
3. Glutaric aciduria (GA1)*
4. Isovaleric acidaemia (IVA) *
5. Maple syrup urine disease (MSUD)*
6. Homocystinuria (HCU)*

**In line with recommendations from the UK National Screening Committee newborn screening for MSUD, GA1, IVA and HCU was introduced in Northern Ireland on 2nd March 2020.*

Approximately 1 in 5,000 babies born in Northern Ireland will have PKU and 1 in 10,000 will have MCADD. The other conditions are rarer, occurring in 1 in 100,000–300,000 babies. Babies with these disorders cannot process certain substances in their food. Without treatment, babies with some of these conditions can become suddenly and seriously ill. The symptoms of the conditions are different; some may be life threatening or lead to severe developmental problems. They can all be treated by a carefully managed diet, which is different for each condition and may include additional medicines. If babies are not screened, but are later found to have an IMD, it may be too late for the special diet to make a real difference.

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.

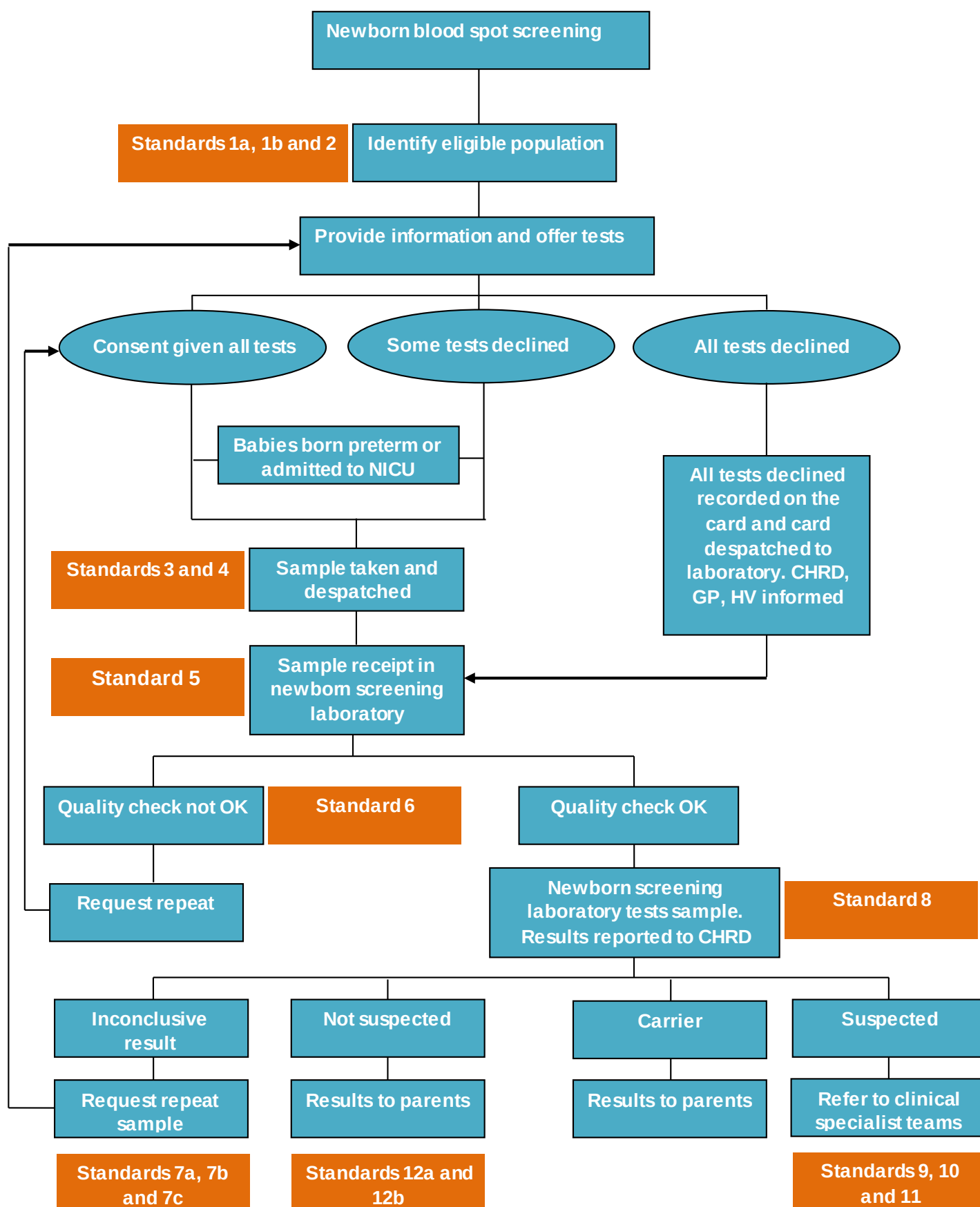
The purpose of screening is to identify babies more likely to have these conditions. If the screening test is positive, a baby will be offered further tests or investigations to confirm the diagnosis. Where one of these conditions is confirmed as present, effective interventions are available to prevent 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 taken³.

³PHE 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 2018-20

Key improvements in the NBSP in Northern Ireland (NI) during 2018-20 included the

- introduction of failsafe/information workshops with key programme stakeholders (2018-). This group:
 - Has facilitated the implementation of an expanded screening programme
 - Has reviewed, enhanced and standardised the format of the weekly failsafe report which identifies babies with outstanding conclusive blood spot screening results recorded on the Child Health System and
 - Is working to develop Quality Assurance Information and Safety Requirements which will assess the feasibility of software changes to the Child Health System to enable reporting on specific programme standards against which performance cannot currently be measured; enhance the 'failsafe' and provide information for ongoing performance management;
- introduction of the mandatory use of the health and care number on the blood spot cards (2018) which are sent to the Laboratory to improve quality, timeliness and ultimately the safety of babies within the screening programme;
- implementation of an expanded screening programme (2020) to include screening for the additional inherited metabolic conditions: glutaric aciduria (GA1), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA) and homocystinuria (HCU);
- setting up of an avoidable repeats subgroup which provides monitoring, education resources and additional support for test takers to address avoidable repeats rates (2019-20) and
- ongoing work with regard to Quality Assurance structures and processes within the NBSP.

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.

Programme performance 2018-20

In 2018-20, 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 (2018-19), which describes performance against national standards in each region of the UK¹, 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 2018-19:

- In terms of coverage, >97% of born and resident babies in Northern Ireland had a conclusive screening result for each of the conditions recorded on the child health system by 17 days of age.
- In relation to timing of sample collection and processing, 94.3% of samples were collected on day 5, and 98.8% of samples were 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.

¹PHE 2018-19 annual report can be accessed at:

Newborn blood spot screening data collection and performance analysis report 1 April 2018 to 31 March 2019 - GOV.UK (www.gov.uk)

- In relation to timeliness of receipt into clinical care, the programme in Northern Ireland met the acceptable standard for timeliness of first appointment for PKU screen positive babies with 100% of babies seen by 14 days of age. The NI programme met the achievable standard for timeliness of first appointment for CF screen positive babies with 100% of babies with 2 mutations seen by 28 days of age and 100% of babies with 0 or 1 mutations seen by 35 days of age.
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suspected on first sample, with 100% of babies seen by 14 days of age and 100% of babies, suspected on a repeat sample, seen by 21 days of age.

The NI programme met the achievable standard for timeliness of first appointment for CF screen positive babies with 100% of babies with 2 mutations seen by 28 days of age and met the acceptable standard for CF screen positive babies with 0 or 1 mutations with 80% of babies seen by 35 days of age.

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 = $\leq 2\%$; achievable = $\leq 1\%$) 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 2018-19 and 2019-20 was 5.44% and 6.03% respectively, neither of which met 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. In 2019-20 a regional Avoidable Repeats working group was established to monitor avoidable repeat rates and to provide education resources and additional support for test takers.

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 2018 and 31st March 2020 and is

reported by the Trust work location of the test taker and not the baby's 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 2018-19 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 2018-19.

Key definitions

'Born and Resident'

Babies who were born to Northern Ireland residents (at time of birth) in 2018-19 or 2019 – 20 and were still resident in Northern Ireland at 31st March.

'Movers in'

Babies who were born in 2018-19 or 2019-20, who moved to a CHS area of Northern Ireland between 1st April and 31st March, who were not born to residents of that CHS area and were still resident at 31st March.

'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^{4,5}

⁴Patient info <https://patient.info/treatment-medication/genetic-testing>

⁵NHS choices 2018 www.nhs.uk/conditions/genetics/inheritance

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*)
- 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) Sickie – 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 2018-20 is based upon standards that were introduced across the UK in 2017.

The 12 standards are summarised below in Table 1.

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

Standard		Description	Acceptable	Achievable
1a	Completeness of coverage (CCG ⁶ 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. <i>*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 CCG⁶ was responsible at birth and remains responsible on the last day of the reporting period.</i>	≥ 95.0% (for all conditions)	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD
1b	Completeness of coverage (movers in)	The proportion of **eligible babies for whom a conclusive screening result for each of the nine	≥ 95.0% (for all conditions)	≥ 99.0% for IMDs

⁶CCG – Clinical Commissioning Group (Northern Ireland data are provided by Child Health System (CHS) area)

Standard		Description	Acceptable	Achievable
1b	Completeness of coverage (movers in) (cont'd)	conditions is recorded on CHS by 21 calendar days of movement in being recorded on the CHS. <i>**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 CCG⁶ during the reporting period and for whom the CCG⁶ remains responsible on the last day of the reporting period.</i>		≥ 98.0% for CF, CHT and SCD
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.

⁶CCG— Clinical Commissioning Group (Northern Ireland data are provided by Child Health System (CHS) area)

Standard		Description	Acceptable	Achievable
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. <i>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.</i>	≥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%
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	≤2%	≤1%

Standard		Description	Acceptable	Achievable
6	Quality of the blood spot sample (<i>cont'd</i>)	young (on or before day 4) • Insufficient blood • Unsuitable sample/card		
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 sample for CHT screening for preterm infant	The proportion of second blood spot samples taken on or before 28 days of age. Only taken earlier if baby discharged home.	≥95%	≥99%
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	Laboratory is UKAS accredited with specialist assessment of NBS screening by the next full visit	

Standard		Description	Acceptable	Achievable
8	UKAS (screening) (cont'd)	ISO standards and the UK screening specific laboratory standards, as an integrated process		
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	
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	

Standard		Description	Acceptable	Achievable
11	Timely entry into clinical care (<i>cont'd</i>)	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
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. However, the mandatory use of the health and care number on the blood spot cards was introduced in 2018.

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.

Standards 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 2018-20 against each of the UK standards is outlined in Table 2.

APPENDIX B: PERFORMANCE OF NBSP IN NI 2018-20

Table 2: Performance of the Northern Ireland Newborn Blood Spot Screening Programme 2018-20 (data collected by NI Child Health System)

Standard	Performance 2018-20 ⁷						Acceptable	Achievable
1a	2019-20			2018-19			≥ 95.0%	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD
	Total number of 'born and resident' babies =			21,929		22,574		
	Completeness of coverage (CCG responsibility at birth) by Day 17 (Numbers and %)	PKU ⁸	21,648	98.72%	22,331	98.92%		
		CHT	21,457	97.85%	22,115	97.97%		
		CF	21,603	98.51%	22,279	98.69%		
		SCD	21,658	98.76%	22,330	98.92%		
1b	Declines to screening (Numbers and %)	PKU ⁸	11	0.05%	12	0.05%	≥ 95.0%	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD
		CHT	11	0.05%	13	0.06%		
		CF	11	0.05%	13	0.06%		
		SCD	12	0.05%	12	0.05%		
	2019-20			2018-19				
1b	Total number of 'movers in' babies =			410		366	≥ 95.0%	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD
	Completeness of coverage (movers in)	PKU ⁸	318	77.56%	297	81.15%		
		CHT	316	77.07%	297	81.15%		
		CF	276	67.32%	279	76.23%		
		SCD	316	77.07%	297	81.15%		
	Declines to screening (Numbers and %)	PKU ⁸	89	21.71%	66	18.03%		
		CHT	91	22.20%	66	18.03%		
		CF	87	21.22%	65	17.76%		
		SCD	91	22.20%	66	18.03%		

⁷In 2018-19, 5 conditions only were screened for in Northern Ireland. In March 2020 screening for isovaleric acidemia (IVA), glutaric acidemia (GA1), homocystinuria (HCU) and maple syrup urine disease (MSUD) was introduced and therefore performance data regarding these conditions is not included in this report.

⁸PKU is reported as proxy to MCADD data in line with UK reporting

Standard	Performance 2018-20 ⁷				Acceptable	Achievable
			2019-20	2018-19		
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 ² conditions for babies aged 11-364 days			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 (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 =	94.73% (21,447) 22,639	94.34% (21,728) 23,032	≥ 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 =	98.34% (24,630) 25,046	98.82% (25,132) 25,431	≥ 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 2018-20 ⁷				Acceptable	Achievable
			2019-20	2018-19		
6	Quality of the blood spot sample (avoidable repeat rate)	Avoidable repeat rate	6.03%	5.44%	≤ 2%	≤ 1%
		Total number of first samples =	22,718	23,078		
		Total number of avoidable repeats =	1,371	1,256		
9	Timely processing of all CHT and IMD (excluding HCU) screen positive samples	PKU	100% (6/6)	100% (8/8)	100% clinical referral initiated within 3 working days of sample receipt in the laboratory for each condition	N/A
		MCADD	100% (4/4)	N/A		
		CHT	100% (16/16)	100% (14/14)		
11	Timely entry into clinical care	By 14 days of age			100% by 14 days of age	N/A
		PKU	100% (4/4 ^{9,10})	100% (6/6 ¹¹)		
		MCADD	100% (4/4)	N/A		
		CHT*	100% (10/10)	83.33% (5/6)	100% by 21 days of age	
		*suspected on first sample				
		By 21 days of age				
		CHT**	100% (5/5 ¹²)	80% (4/5)	≥ 95% by 28 days of age	100% by 28 days of age
		**suspected on a repeat sample				
		By 28 days of age			≥ 80% by 35 days of age	100% by 35 days of age
		CF (2 mutations)	100% (3/3 ¹³)	100% (4/4 ¹⁴)		
		By 35 days of age			≥ 90%	≥ 95%
		CF (1 or 0 mutations)	80% (4/5)	100% (3/3)		
		SCD A baby in whom SCD is suspected should attend first clinical appointment by 90 days of age:	N/A	N/A		

Standard	Performance 2018-20 ⁷			Acceptable	Achievable
			2019-20	2018-19	
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%
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%

⁹One baby did not require follow up with clinical services and is therefore excluded from age data

¹⁰One additional baby was diagnosed before screening and is excluded from age data

¹¹Two additional babies were diagnosed before screening and are excluded from age data

¹²One additional baby was diagnosed before screening and is excluded from age data

¹³One additional baby was diagnosed before screening and is excluded from age data

¹⁴One additional baby was diagnosed before screening and is excluded from age data

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. During 2018-19 and 2019-20 the programme exceeded the acceptable standard for completeness of coverage, with more than 97% of 'born and resident' babies with conclusive results for all conditions recorded on CHS by 17 days of age and 99% 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 1a¹⁵ (data collected by NI Child Health System)

	Year					
	2019-20		2018-19		2017-18	
Total number 'born and resident' (B&R) at 31st March	21,929		22,574		22,582	
Number of B&R with decline to screening (02)						
PKU ⁸	11	0.05%	12	0.05%	9	0.04%
CHT	11	0.05%	13	0.06%	9	0.04%
CF	11	0.05%	13	0.06%	10	0.04%
SCD	12	0.05%	12	0.05%	9	0.04%
Number (%) of B&R with conclusive results by the end of the reporting period (codes 04, 05, 06, 07, 08, 10)						
PKU ⁸	21,915	99.94%	22,553	99.91%	22,568	99.94%
CHT	21,913	99.93%	22,551	99.90%	22,567	99.93%
CF	21,915	99.94%	22,549	99.89%	22,567	99.93%
SCD	21,916	99.94%	22,555	99.92%	22,568	99.94%
Number (%) of B&R with conclusive results available by Day 17 (by 17 days of age) codes 04, 05, 06, 07,08,10)						
PKU ⁸	21,648	98.72%	22,331	98.92%	22,362	99.03%
CHT	21,457	97.85%	22,115	97.97%	22,155	98.11%
CF	21,603	98.51%	22,279	98.69%	22,302	98.76%
SCD	21,658	98.76%	22,330	98.92%	22,356	99.00%

⁸PKU is reported as proxy to MCADD data in line with UK reporting

¹⁵excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required, 09 = incomplete/not screened)

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.

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 2018-19 and 2019-20 there were 22 and 45 'mover in' babies respectively 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.

Table 3b Coverage - Born and Resident (B&R) by Child Health Area 2018-19 - Standard 1a^{15,16} (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 31/03/19	8,386		5,293		5,180		3,715		22,574	
Number (%) of B&R with decline to screening (02):										
PKU ⁸	4		2		5		1		12	0.05%
CHT	4		2		5		2		13	0.06%
CF	4		2		5		2		13	0.06%
SCD	4		2		5		1		12	0.05%
Number (%) of B&R with reason for not starting/ incomplete screening (09):										
PKU ⁸	7		0		0		0		7	0.03%
CHT	7		0		0		0		7	0.03%
CF	9		0		1		0		10	0.04%
SCD	7		0		0		0		7	0.03%
Number (%) of B&R with conclusive results (04, 05, 06, 07, 08, 10):										
PKU ⁸	8,373	99.84%	5,291	99.96%	5,175	99.90%	3,714	99.97%	22,553	99.91%
CHT	8,372	99.83%	5,291	99.96%	5,175	99.90%	3,713	99.95%	22,551	99.90%
CF	8,371	99.82%	5,291	99.96%	5,174	99.88%	3,713	99.95%	22,549	99.89%
SCD	8,375	99.87%	5,291	99.96%	5,175	99.90%	3,714	99.97%	22,555	99.92%
Number (%) of B&R with conclusive results available by Day 17 (04, 05, 06, 07, 08, 10):										
PKU ⁸	8,293	98.89%	5,239	98.98%	5,130	99.03%	3,669	98.76%	22,331	98.92%
CHT	8,214	97.95%	5,187	98.00%	5,082	98.11%	3,632	97.77%	22,115	97.97%
CF	8,262	98.52%	5,233	98.87%	5,126	98.96%	3,658	98.47%	22,279	98.69%
SCD	8,285	98.80%	5,245	99.09%	5,132	99.07%	3,668	98.73%	22,330	98.92%

⁸PKU is reported as proxy to MCADD data in line with UK reporting

¹⁵excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required)

¹⁶unlike data produced by the lab, data provided by CHS relates to the baby's residence area at time of reporting

Table 3c Coverage - Born and Resident (B&R) by Child Health Area 2019-20 - Standard 1a^{15,16} (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 31/03/20	8,024		5,213		5,112		3,580		21,929		
Number (%) of B&R with decline to screening (02):											
PKU ⁸	4		2		4		1		11	0.05%	
CHT	4		2		4		1		11	0.05%	
CF	4		2		4		1		11	0.05%	
SCD	4		2		5		1		12	0.05%	
Number (%) of B&R with reason for not starting/ incomplete screening (09):											
PKU ⁸	0		0		0		0		0	0.00%	
CHT	0		0		0		0		0	0.00%	
CF	0		0		0		0		0	0.00%	
SCD	0		0		0		0		0	0.00%	
Number (%) of B&R with conclusive results (04, 05, 06, 07, 08, 10):											
PKU ⁸	8,018	99.93%	5,211	99.96%	5,107	99.90%	3,579	99.97%	21,915	99.94%	
CHT	8,017	99.91%	5,211	99.96%	5,106	99.88%	3,579	99.97%	21,913	99.93%	
CF	8,018	99.93%	5,211	99.96%	5,107	99.90%	3,579	99.97%	21,915	99.94%	
SCD	8,020	99.95%	5,211	99.96%	5,106	99.88%	3,579	99.97%	21,916	99.94%	
Number (%) of B&R with conclusive results available by Day 17 (04, 05, 06, 07, 08, 10):											
PKU ⁸	7,900	98.45%	5,157	98.93%	5,056	98.90%	3,535	98.74%	21,648	98.72%	
CHT	7,820	97.46%	5,120	98.22%	5,013	98.06%	3,504	97.88%	21,457	97.85%	
CF	7,882	98.23%	5,153	98.85%	5,044	98.67%	3,524	98.44%	21,603	98.51%	
SCD	7,908	98.55%	5,158	98.94%	5,059	98.96%	3,533	98.69%	21,658	98.76%	

⁸PKU is reported as proxy to MCADD data in line with UK reporting

¹⁵excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required)

¹⁶unlike data produced by the lab, data provided by CHS relates to the baby's residence area at time of reporting

Table 4a Completeness of coverage – Movers in - Standard 1b¹⁵ (data collected by NI Child Health System)

	Year					
	2019-20		2018-19		2017-18	
Total number 'Movers In' (MI) resident at 31/03/19	410		366		356	
Number of MI with decline to screening (02)						
PKU ⁸	89	21.71%	66	18.03%	52	14.61%
CHT	91	22.20%	66	18.03%	50	14.04%
CF	87	21.22%	65	17.76%	48	13.48%
SCD	91	22.20%	66	18.03%	49	13.76%
Number (%) of MI with reason for not starting/incomplete (code 09-)						
PKU ⁸	1	0.24%	2	0.55%	0	0.00%
CHT	1	0.24%	2	0.55%	0	0.00%
CF	45	10.98%	22	6.01%	29	8.15%
SCD	1	0.24%	2	0.55%	0	0.00%
Number (%) of MI with conclusive results(codes 04, 05, 06, 07, 08,10)						
PKU ⁸	318	77.56%	297	81.15%	303	85.11%
CHT	316	77.07%	297	81.15%	305	85.67%
CF	276	67.32%	279	76.23%	279	78.37%
SCD	316	77.07%	297	81.15%	306	85.96%

⁸PKU is reported as proxy to MCADD data in line with UK reporting

¹⁵excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required)

Table 4b – Completeness of coverage – Movers in 2018-19 by Child Health Area - Standard 1b^{15,16} (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 31/03/19	112		107		82		65		366	
Number of MI with decline to screening (02):										
PKU ⁸	35		3		20		8		66	18.03%
CHT	35		3		20		8		66	18.03%
CF	35		3		19		8		65	17.76%
SCD	35		3		20		8		66	18.03%
Number of MI with reason for not starting/incomplete screening (09):										
PKU ⁸	2		0		0		0		2	0.55%
CHT	2		0		0		0		2	0.55%
CF	10		1		11		0		22	6.01%
SCD	2		0		0		0		2	0.55%
Number (%) of MI with conclusive results (04, 05, 06, 07, 08, 10):										
PKU ⁸	75	66.96%	103	96.26%	62	75.61%	57	87.69%	297	81.15%
CHT	75	66.96%	103	96.26%	62	75.61%	57	87.69%	297	81.15%
CF	67	59.82%	103	96.26%	52	63.41%	57	87.69%	279	76.23%
SCD	75	66.96%	103	96.26%	62	75.61%	57	87.69%	297	81.15%

⁸PKU is reported as proxy to MCADD data in line with UK reporting

¹⁵excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required)

¹⁶unlike data produced by the lab, data provided by CHS relates to the baby's residence area at time of reporting

Table 4c – Completeness of coverage – Movers in 2019-20 by Child Health Area - Standard 1b^{15,16} (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 31/03/20	136	102	101	71	410
Number of MI with decline to screening (02):					
PKU ⁸	48	5	23	13	89 21.71%
CHT	49	5	24	13	91 22.20%
CF	47	5	22	13	87 21.22%
SCD	49	5	24	13	91 22.20%
Number of MI with reason for not starting/incomplete screening (09):					
PKU ⁸	1	0	0	0	1 0.24%
CHT	1	0	0	0	1 0.24%
CF	17	5	16	7	45 10.98%
SCD	1	0	0	0	1 0.24%
Number (%) of MI with conclusive results (04, 05, 06, 07, 08, 10):					
PKU ⁸	86 63.24%	97 95.10%	77 76.24%	58 81.69%	318 77.56%
CHT	85 62.50%	97 95.10%	76 75.25%	58 81.69%	316 77.07%
CF	71 52.21%	92 90.20%	62 61.39%	51 71.83%	276 67.32%
SCD	85 62.50%	97 95.10%	76 75.25%	58 81.69%	316 77.07%

⁸ PKU is reported as proxy to MCADD data in line with UK reporting

¹⁵excludes data relating to inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required)

¹⁶unlike data produced by the lab, data provided by CHS relates to the baby's residence area at time of reporting

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 19 - 20 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 has been mandatory in Northern Ireland since July 2018.

In 2018-19 and in 2019-20, 97.5% and 99.3% respectively of all samples received by the Laboratory had the HCN included¹⁷. 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. 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 in 2018-19 and 2019-20, with more than 98% of first samples taken between 5 to 8 days after birth; this performance is similar to 2017-18 and 2016-17.

¹⁷ Data Source: Regional Newborn Screening Laboratory

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

	Year		
	2019-20	2018-19	2017-18
Total number of first samples	22,639	23,032	22,953
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	79	46	119
	Year		
	2019-20	2018-19	2017-18
Number of first samples taken on or before day 4 (%)	79 (0.35%)	91 (0.40%)	103 (0.45%)
Number of first samples taken on day 5 (%)	21,447 (94.73%)	21,728 (94.34%)	21,780 (94.89%)
Number of first samples taken on or after day 9 (%)	367 (1.62%)	365 (1.58%)	256 (1.12%)
Number of first samples taken between day 5 and day 8 (%)	22,193 (98.03%)	22,576 (98.02%)	22,594 (98.44%)

Table 5b – Sample collection 2018-19– Standard 4 by Trust¹⁸ (data collected by the Regional Newborn Screening Laboratory)

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust¹⁹	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	2,775	5,286	460	5,176	5,428	3,907	23,032
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	5	11	2	8	13	7	46
Number of first samples taken on or before day 4	6	22	2	21	20	20	91
Number of first samples taken on day 5	2,629	5,027	423	4,834	5,094	3,721	21,728
Number of first samples taken on or after day 9	50	77	16	49	103	70	365
Number of first samples taken between day 5 and day 8	2,719	5,187	442	5,106	5,305	3,817	22,576
% of first samples taken on or before day 4	0.22%	0.42%	0.43%	0.41%	0.37%	0.51%	0.40%
% of first samples taken on day 5	94.74%	95.10%	91.96%	93.39%	93.85%	95.24%	94.34%
% of first samples taken on or after day 9	1.80%	1.46%	3.48%	0.95%	1.90%	1.79%	1.58%
% of first samples taken between day 5 and day 8	97.98%	98.13%	96.09%	98.65%	97.73%	97.70%	98.02%

¹⁸Trust, reported by the Regional Newborn Screening Laboratory, relates to the work location of the test taker and not the baby's residence area

¹⁹Unallocated 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

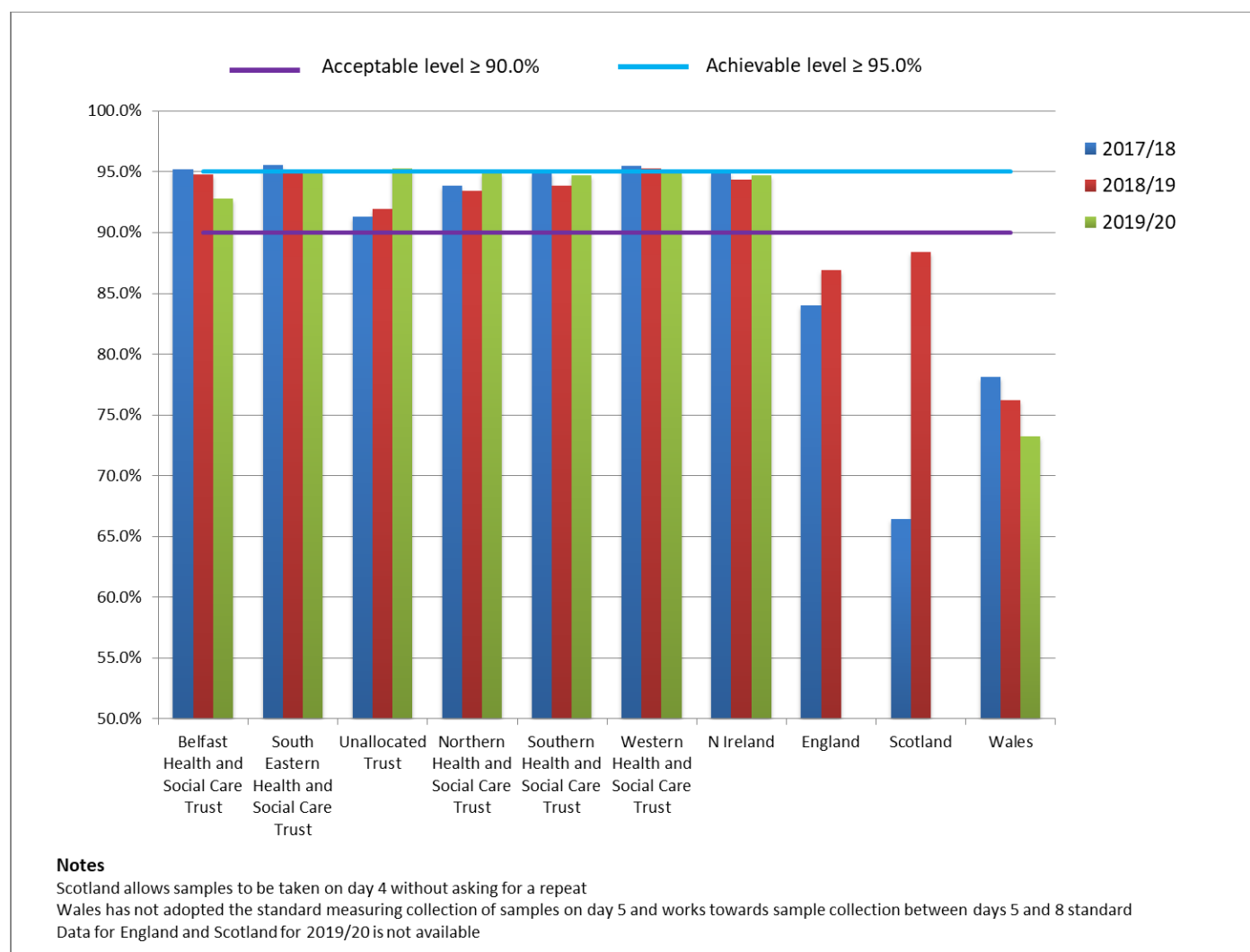
Table 5c – Sample collection 2019-20– Standard 4 by Trust¹⁸ (data collected by the Regional Newborn Screening Laboratory)

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust ¹⁹	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	2,018	3,995	2,154	5,375	5,413	3,684	22,639
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	6	17	14	19	17	6	79
Number of first samples taken on or before day 4	12	18	13	6	20	10	79
Number of first samples taken on day 5	1,873	3,796	2,052	5,101	5,128	3,497	21,447
Number of first samples taken on or after day 9	49	68	28	51	103	68	367
Number of first samples taken between day 5 and day 8	1,957	3,909	2,113	5,318	5,290	3,606	22,193
% of first samples taken on or before day 4	0.59%	0.45%	0.60%	0.11%	0.37%	0.27%	0.35%
% of first samples taken on day 5	92.81%	95.02%	95.26%	94.90%	94.73%	94.92%	94.73%
% of first samples taken on or after day 9	2.43%	1.70%	1.30%	0.95%	1.90%	1.85%	1.62%
% of first samples taken between day 5 and day 8	96.98%	97.85%	98.10%	98.94%	97.73%	97.88%	98.03%

¹⁸Trust, reported by the Regional Newborn Screening Laboratory, relates to the work location of the test taker and not the baby's residence area

¹⁹Unallocated 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 2017-18 to 2019-20



In 2018-19 and 2019-20 the programme also exceeded the acceptable standard for timely receipt of a sample in the laboratory ($\geq 95.0\%$ 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		
	2019-20	2018-19	2017-18
Total number of ALL samples (first, repeat and second samples) included	25,046	25,431	25,222
Number of samples EXCLUDED (because date of specimen was not recorded on the cards)	93	54	146
Number of ALL samples received by the lab in 3 or fewer working days of sample being taken (%)	24,630 (98.34%)	25,132 (98.82%)	24,712 (97.98%)
Number of ALL samples received by the lab in 4 or fewer working days of sample being taken (%)	24,909 (99.45%)	25,316 (99.55%)	25,073 (99.41%)

Table 6b Timely receipt of a sample in the laboratory by Trust¹⁸ 2018-19 – Standard 5 (data collected by the Regional Newborn Screening Laboratory)

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust¹⁹	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	3,105	5,829	523	5,561	6,040	4,373	25,431
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	6	13	2	8	14	11	54
Number of all samples received by the lab in 3 or fewer working days of sample being taken	3,086	5,801	521	5,532	6,004	4,188	25,132
Number of all samples received by the lab in 4 or fewer working days of sample being taken	3,092	5,818	521	5,547	6,025	4,313	25,316

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust¹⁹	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
Number of all samples received by the lab on or after 5 working days of sample being taken	13	11	2	14	15	60	115
% of all samples received by lab in 3 or fewer working days of sample being taken	99.39%	99.52%	99.62%	99.48%	99.40%	95.77%	98.82%
% of all samples received by lab in 4 or fewer working days of sample being taken	99.58%	99.81%	99.62%	99.75%	99.75%	98.63%	99.55%
% of all samples received by lab in 5 or more working days of sample being taken	0.42%	0.19%	0.38%	0.25%	0.25%	1.37%	0.45%

¹⁸Trust, reported by the Regional Newborn Screening Laboratory, relates to the work location of the test taker and not the baby's residence area

¹⁹Unallocated 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

Table 6c Timely receipt of a sample in the laboratory by Trust¹⁸ 2019-20 – Standard 5 (data collected by the Regional Newborn Screening Laboratory)

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust¹⁹	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	2,367	4,450	2,439	5,667	6,037	4,086	25,046
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	13	19	14	21	19	7	93
Number of all samples received by the lab in 3 or fewer working days of sample being taken	2,338	4,421	2,429	5,635	5,961	3,846	24,630
Number of all samples received by the lab in 4 or fewer working days of sample being taken	2,351	4,440	2,437	5,655	6,004	4,022	24,909

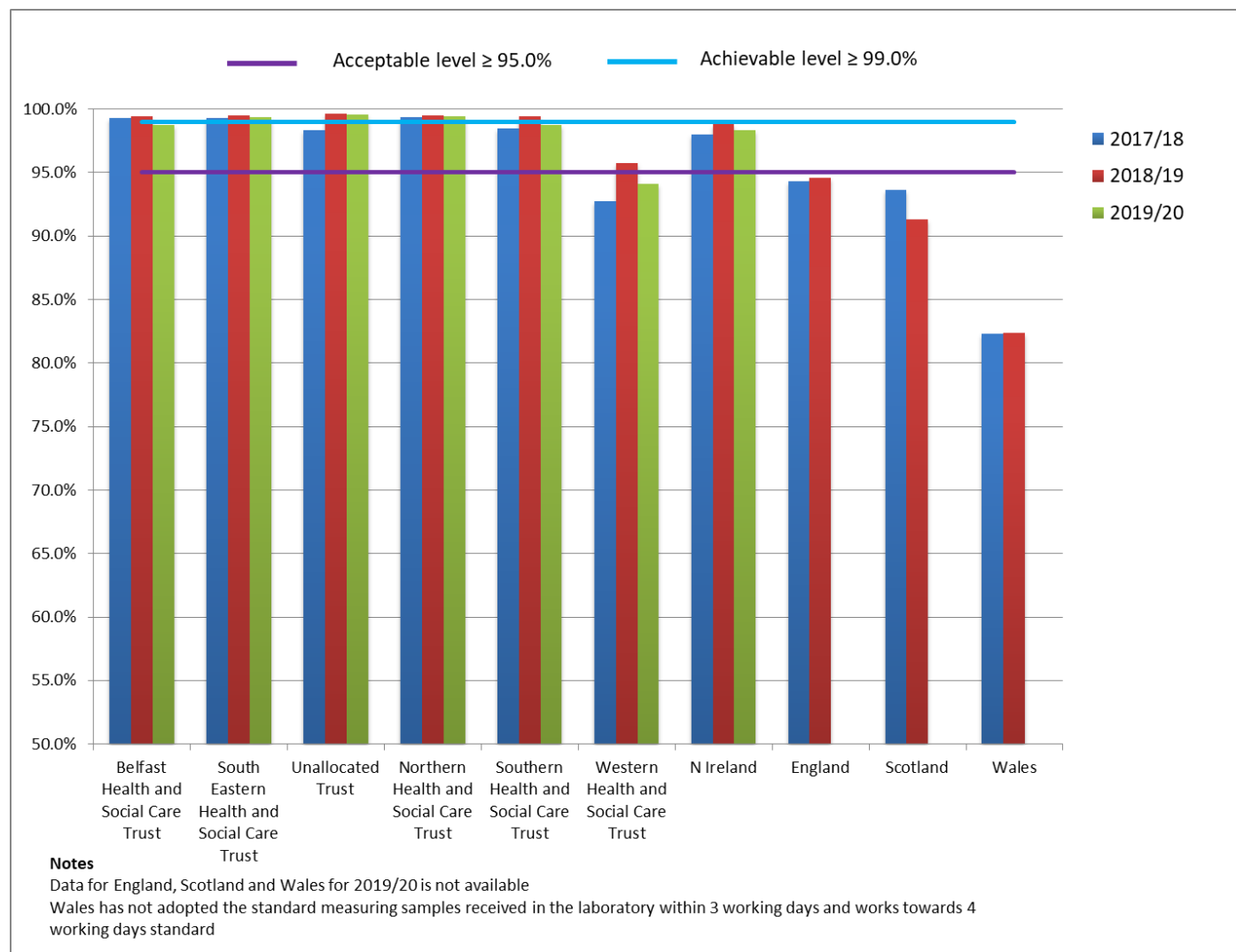
	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust¹⁹	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
Number of all samples received by the lab on or after 5 working days of sample being taken	16	10	2	12	33	64	137
% of all samples received by lab in 3 or fewer working days of sample being taken	98.77%	99.35%	99.59%	99.44%	98.74%	94.13%	98.34%
% of all samples received by lab in 4 or fewer working days of sample being taken	99.32%	99.78%	99.92%	99.79%	99.45%	98.43%	99.45%
% of all samples received by lab in 5 or more working days of sample being taken	0.68%	0.22%	0.08%	0.21%	0.55%	1.57%	0.55%

¹⁸ Trust, reported by the Regional Newborn Screening Laboratory, relates to the work location of the test taker and not the baby's residence area

¹⁹Unallocated 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 2017-18 to 2019-20



Sample quality (Standard 6)

Avoidable repeat requests are 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 2019-20 and 2018-19 was 6.03% and 5.44% respectively and therefore the NBSP did not meet the acceptable standard (avoidable repeat rate $\leq 2\%$) - see Tables 7a and 7b. Performance in 2018-19 (5.44%) is comparable to other UK countries (Scotland 4.69% Wales 8.30%) except England (2.16%)^{1,20}.

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

	Year		
	2019-20	2018-19	2017-18
Total number of first samples received	22,718	23,078	23,072
Total number of avoidable repeat samples requested	1,371	1,256	1,143
REASON FOR REPEAT			
Avoidable Repeats			
Too young for reliable screening (≤ 4 days) (%)	78 (0.34%)	91 (0.39%)	104 (0.45%)
Insufficient sample	919 (4.05%)	861 (3.73%)	857 (3.71%)
Unsuitable sample ²¹	374 (1.65%)	304 (1.32%)	182 (0.79%)
Total Avoidable Repeats	1,371 (6.03%)	1,256 (5.44%)	1,143 (4.95%)

¹<https://www.gov.uk/government/publications/newborn-blood-spot-screening-data-collection-and-performance-analysis-report/newborn-blood-spot-screening-data-collection-and-performance-analysis-report-1-april-2018-to-31-march-2019>

²⁰Not all English laboratories ask for a repeat when the first sample was taken on or before day 4

²¹Unsuitable 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 2018-19 - 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 ¹⁹	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	2,780	5,297	462	5,184	5,441	3,914	23,078
Total number of repeat samples requested	205	339	54	242	393	280	1,513
REASON FOR REPEAT							
Avoidable Repeats							
Too young for reliable screening (≤4 days)	6	22	2	21	20	20	91
Insufficient sample	133	208	21	108	231	160	861
Unsuitable sample ²¹	33	65	16	60	74	56	304
Total Avoidable Repeats	172	295	39	189	325	236	1,256
Avoidable Repeats Request Rates							
% Too young for reliable screening (≤4 days)	0.22%	0.42%	0.43%	0.41%	0.37%	0.51%	0.39%
% Insufficient sample	4.78%	3.93%	4.55%	2.08%	4.25%	4.09%	3.73%
% Unsuitable sample ²¹	1.19%	1.23%	3.46%	1.16%	1.36%	1.43%	1.32%
% Avoidable Repeats	6.19%	5.57%	8.44%	3.65%	5.97%	6.03%	5.44%
Unavoidable Repeats²²							
Total Unavoidable Repeats	33	44	15	53	68	44	257
Unavoidable Repeats Request Rates							
% Unavoidable Repeats	1.19%	0.83%	3.25%	1.02%	1.25%	1.12%	1.11%

¹⁹Unallocated 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

²¹Unsuitable sample = missing data, card past expiry, sample anti-coagulated, over-layered or contaminated, >14 days transit

²²Unavoidable repeat = too premature for testing (SCD), preterm CHT, borderline CHT or inconclusive CF

**Table 7c - Quality of the blood spot sample by Trust 2019-20 - Standard 6
(data collected by the Regional Newborn Screening Laboratory)**

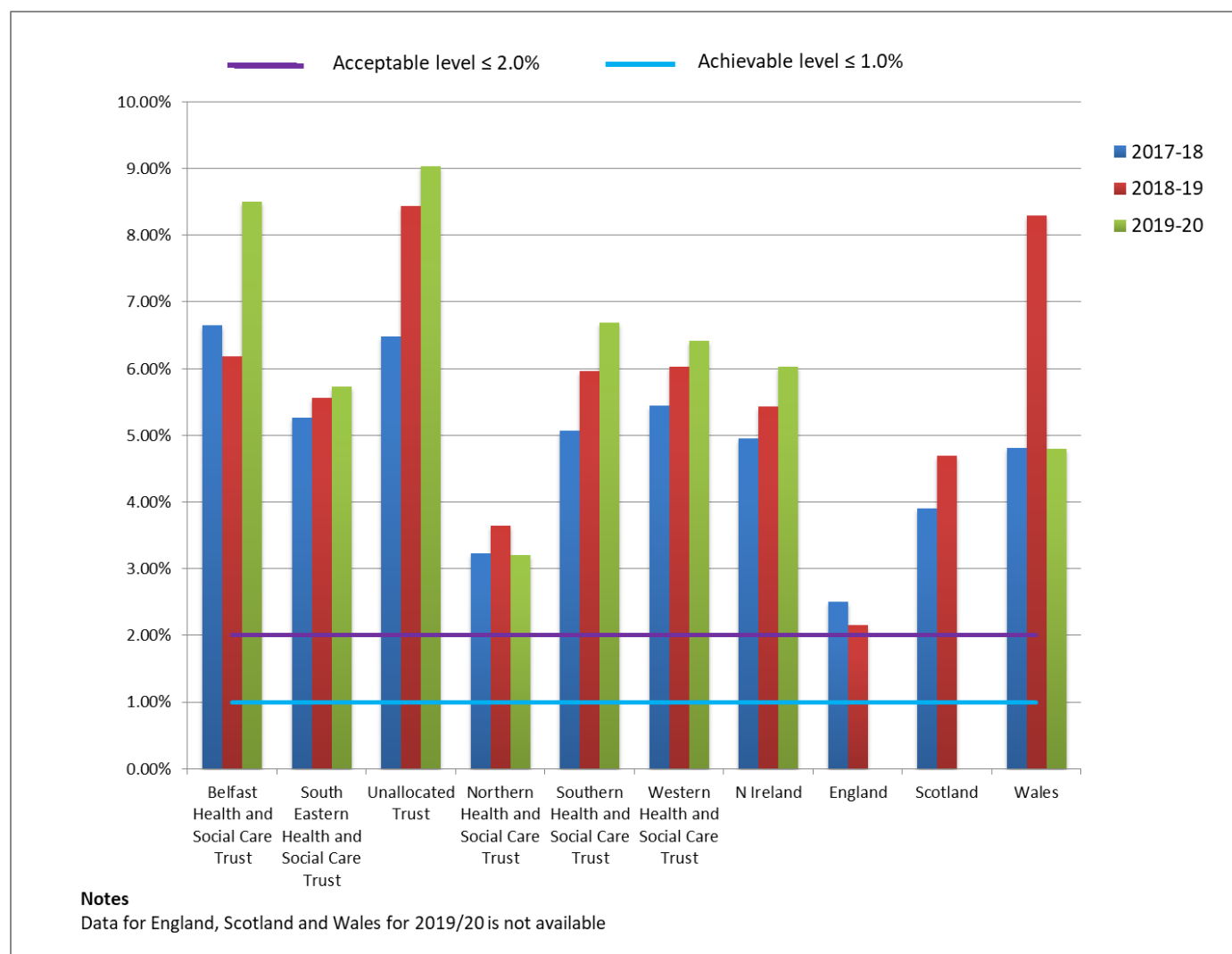
	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust ¹⁹	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
Total number of first samples received	2,024	4,012	2,168	5,394	5,430	3,690	22,718
Total number of repeat samples requested	199	272	228	217	429	267	1,612
REASON FOR REPEAT							
Avoidable Repeats							
Too young for reliable screening (≤4 days)	12	18	12	6	20	10	78
Insufficient sample	118	150	139	94	256	162	919
Unsuitable sample ²¹	42	62	45	73	87	65	374
Total Avoidable Repeats	172	230	196	173	363	237	1,371
Avoidable Repeats Request Rates							
% Too young for reliable screening (≤4 days)	0.59%	0.45%	0.55%	0.11%	0.37%	0.27%	0.34%
% Insufficient sample	5.83%	3.74%	6.41%	1.74%	4.71%	4.39%	4.05%
% Unsuitable sample ²¹	2.08%	1.55%	2.08%	1.35%	1.60%	1.76%	1.65%
% Avoidable Repeats	8.50%	5.73%	9.04%	3.21%	6.69%	6.42%	6.03%
Unavoidable Repeats²²							
Total Unavoidable Repeats	27	42	32	44	66	30	241
Unavoidable Repeats Request Rates							
% Unavoidable Repeats	1.33%	1.05%	1.48%	0.82%	1.22%	0.81%	1.06%

¹⁹ 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

²¹ Unsuitable sample = missing data, card past expiry, sample anti-coagulated, over-layered or contaminated, >14 days transit

²² Unavoidable repeat = too premature for testing (SCD), preterm CHT, borderline CHT or inconclusive CF

Figure 3: Avoidable Repeat Request Rates from 2017-18 to 2019-20



In 2018-19 and 2019-20, the majority of avoidable repeats (3.7% and 4.1% (respectively) of first samples) were due to insufficient sampling (Table 7a). 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 in 2018-19 and 2019-20 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.

In 2019-20 a regional avoidable repeats subgroup was established to monitor avoidable repeats rates and provide educational resources and additional support for test takers.

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

PKU and MCADD

In 2018-19, 8 babies (including 2 who were tested early due to a family history) were identified with positive screening tests for PKU. In 2019-20 6 babies (including 1 baby who was diagnosed before screening) were identified with positive screening tests for PKU. In 2018-19, there were no babies identified as having positive screening tests for MCADD and in 2019-20 there were 4 babies identified with positive screening tests for MCADD. As in previous years, all babies who had a positive screening result for PKU and MCADD (2018-20) (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).

Outcomes

A diagnosis of PKU was confirmed in 6 babies who were PKU screen positive in 2018 -19 and in 5 babies who were PKU screen positive in 2019-20. A diagnosis of MCADD was confirmed in all 4 babies who were MCADD screen positive in 2019 - 20.

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

	Year		
	2019-20	2018-19	2017-18
<i>Number of babies with screen positive result (status code = 07 - not suspected other disorder follow up or 08 - suspected)</i>	6 ^{23,24}	8 ²⁵	7
Number of screen positive babies with clinical referral initiated within 3 working days of sample receipt in lab (%)	6 (100%)	8 (100%)	7 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	4 (100%)	6 (100%)	7 (100%)

²³One baby did not require follow up with clinical services and is therefore excluded from age data

²⁴One baby was diagnosed before screening and is excluded from age data

²⁵Two babies were diagnosed before screening and are excluded from age data

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

	Year		
	2019-20	2018-19	2017-18
Number of babies with screen positive result (status code = 08 - suspected)	4	0	3
Number of screen positive babies with clinical referral initiated within three working days of sample receipt in lab (%)	4 (100%)	0 (100%)	3 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	4 (100%)	0 (100%)	3 (100%)

CHT

In 2018-19, 6 babies were identified as CHT screen positive on their first blood spot sample; all 6 (100%) were referred to specialist clinical teams within 3 days of sample receipt in the laboratory and 5 (83.33%) were seen by the clinical team by 14 days of age.

A further 8 babies were also identified as screen positive. Five of these babies were positive on a repeat sample following a borderline result (Table 8c) and 4 (80%) of these babies were seen by the clinical team by 21 days of age.

In 2019-20, 10 babies were identified as CHT screen positive on their first blood spot sample; all 10 (100%) were referred to specialist clinical teams within 3 days of sample receipt in the laboratory and seen by the clinical team by 14 days of age.

A further 6 babies were also identified as screen positive. Five of these babies were positive on a repeat sample following a borderline result (Table 8c) and were seen by the clinical team by 21 days of age (one baby was diagnosed before screening and has been excluded from the age data).

Outcomes

In 2018-19 a diagnosis of CHT was confirmed in 7 (50.0%) of the 14 babies who were identified as screen positive. In 2019-20 a diagnosis of CHT was confirmed in 8 (50.0%) of the 16 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					
	2019-20		2018-19 ²⁶		2017-18	
	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)	10	6 ²⁷	6	5	13 ²⁸	6
Number of screen positive babies with clinical referral initiated within 3 working days of sample receipt in lab (%)	10 (100%)	6 (100%)	6 (100%)	5 (100%)	12 (100%)	6 (100%)
Number of screen positive babies who were seen by 14 days of age (suspected on first sample)	10 (100%)	N/A	5 (83.33%)	N/A	11 (91.67%)	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	5 (100%)	N/A	4 (80%)	N/A	6 (100%)

²⁶ In 2018/19 there were 3 additional babies who were CHT screen positive following a preterm repeat at 28+ days of age

²⁷ One baby was diagnosed before screening and has been excluded from age data

²⁸ One baby was diagnosed before screening and has been excluded from age data

SCD

In 2018-19 and in 2019-20, no babies were identified as screen positive for SCD; however, the screening results for 39 babies in 2018-19 and 54 babies in 2019-20 required further testing/assessment: in 2018-19, 2 were identified with other potentially clinically significant condition, 21 were identified as sickle cell 'carriers'

and 16 as a carrier of another unusual haemoglobin gene. In 2019-20, one baby was identified with other potentially clinically significant condition, 34 were identified as sickle cell 'carriers' and 19 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		
	2019-20	2018-19	2017-18
Sickle Cell Disease or Other Potentially Clinically Significant Condition ²⁹	1	2	2
Condition/Disorder	Year		
	2019-20	2018-19	2017-18
Sickle Cell Carrier or Carrier of Other Unusual Haemoglobin Gene ³⁰ or Possible Benign Disorder ³¹	53	37	38

²⁹ Other potentially clinically significant condition comprise F, FE, FEA or HbAF

³⁰ Carrier of other unusual haemoglobin gene comprise FAC, FAD, ?FAD, FAE or FAO^{Arab}

³¹ Other possible benign disorder comprise FC, FD, FO^{Arab}, FCA, FDA or FO^{Arab}A

CF

2 mutations

In 2018-19, 5 babies were identified on screening as suspected CF with 2 genetic mutations; 4 (100%) were seen by the clinical team by 28 days of age (one baby was diagnosed before screening and has been excluded from the age data). Four of the 5 babies identified on screening as suspected CF with 2 genetic mutations were confirmed as having CF.

In 2019-20, 4 babies were identified on screening as suspected CF with 2 genetic mutations; 3 (100%) were seen by the clinical team by 28 days of age (one baby was diagnosed before screening and has been excluded from the age data) (Table 8e).

CF was confirmed in all 4 babies identified on screening as suspected CF with 2 genetic mutations.

1 or 0 mutations

In 2018-19, a further 3 babies were identified as suspected CF with 1 or 0 mutations and an IRT $\geq$ cut-off 2 on screening and required follow-up. All babies (100%) were seen by the clinical team by 35 days of age. CF was confirmed in one of these children.

In 2019-20, a further 5 babies were identified as suspected CF with 1 or 0 mutations and an IRT $\geq$ cut-off 2 on screening and required follow-up. Four babies (80%) were seen by the clinical team by 35 days of age. CF was confirmed in one of these children.

Outcomes

In 2018-19, a total of 8 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 5 of these babies. In addition, 6 babies were identified as probable carriers of a gene for CF (Table 8g).

In 2019-20, a total of 9 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 5 of these babies. In addition, 10 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		
	2019-20	2018-19	2017-18
No. of CF suspected babies with 2 mutations	4 ³²	5 ³³	8
Number of screen positive babies who were seen by 28 days of age (%)	3 (100%)	4 (100%)	8 (100%)

³²One baby was diagnosed before screening and has been excluded from age data

³³One baby was diagnosed before screening and has been excluded from age data

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

	Year		
	2019-20	2018-19	2017-18
No. of CF suspected babies with 1 or 0 mutations and IRT ≥ cut-off 2	5	3	10
Number of screen positive babies who were seen by 35 days of age	4 (80%)	3 (100%)	7 (77.8%)

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

	Year		
	2019-20	2018-19	2017-18
No. of babies with 1 CF mutation detected and second IRT < cut-off 2	10	6	11

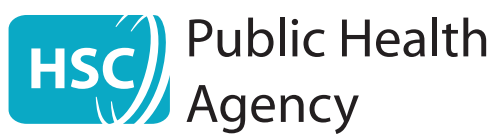
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Data for this report was produced and collated by:

Dr Jennifer Cundick (Regional Newborn Screening Laboratory)
Nicola Cunningham (Public Health Agency)
Ciara Daly (Regional Newborn Blood Spot Screening Service, BHSCT)
Valerie Doyle (CHS – SHSCT)
Gwen Gillespie (CHS – NHSCT)
Deborah Kelly (Performance and Service Improvement, WHSCT)
Claire McIlroy (Regional Newborn Blood Spot Screening Nurse, BHSCT)
Maria Monaghan (CHS – BHSCT and SEHSCT)
Kathleen Slavin (CHS – WHSCT)
Adam Steele (CHS – NHSCT)
Tracy Thompson (CHS – SHSCT)

Data analysed and report written by:

Dr Rachel Doherty, Dr Jenny Gingles, Dr Adrian Mairs, Annie McGeown, Ursula Gaffney and Nicola Cunningham (Public Health Agency)



Public Health Agency

12-22 Linenhall Street, Belfast BT2 8BS.
Tel: 0300 555 0114 (local rate).
www.publichealth.hscni.net

Find us on:

