Newborn Blood Spot Screening in Northern Ireland

Annual Report 2020 - 22

HSC Public Health Agency

Newborn Blood Spot Screening

CONTENTS:

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 2020-22.

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 released national annual KPI data, which outlines performance against key performance indicators in the NBSP in England, shows that in terms of these indicators the programme in NI is performing well. [1]

Regional data relating to the Northern Ireland NBSP highlight that in 2020-21:

- In terms of coverage, >98% 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, 93.3% of samples were collected on day 5 and 98.5% of samples were received in the newborn screening laboratory within 3 working days of collection.

[1] NHS screening programmes: KPI reports 2021 to 2022 - GOV.UK (www.gov.uk)

- 100% of positive screening results (for PKU, IVA, GA1 and CHT) 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. This included achieving:

    - the acceptable standard for timeliness of first appointment for PKU screen positive babies with 100% of babies seen by 14 days of age.
    - 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 21,000 babies had newborn blood spot screening testing. In total, across all of the conditions tested for, 55 babies were identified as screen positive and 45 of these babies were confirmed as having one of the conditions.

Regional data relating to the Northern Ireland NBSP highlight that in 2021-22:

- 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, 93.1% of samples were collected on day 5 and 98.1% of samples were received in the newborn screening laboratory within 3 working days of collection.

- 100% of positive screening results (for PKU and CHT) 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. This included achieving:

    - the acceptable standard for timeliness of first appointment for PKU screen positive babies with 100% of babies seen by 14 days of age.
    - 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 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
        - 100% of babies with 0 or 1 mutations seen by 35 days of age.

- Over 21,000 babies had newborn blood spot screening testing. In total, across all the conditions tested for, 36 babies were identified as screen positive, and 30 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 in 2020-21 was 4.4% and in 2021-22 was 4.7%. The regional NBSP Quality Improvement (QI) group continues to work to understand and reduce avoidable repeats.

SECTION A: INTRODUCTION AND HEADLINE RESULTS FOR 2020-22

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 7 - 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 2020 - 31st March 2022 (hereafter referred to as 2020-22) against national standards.

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

[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

- **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.

- **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.

And 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)

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.

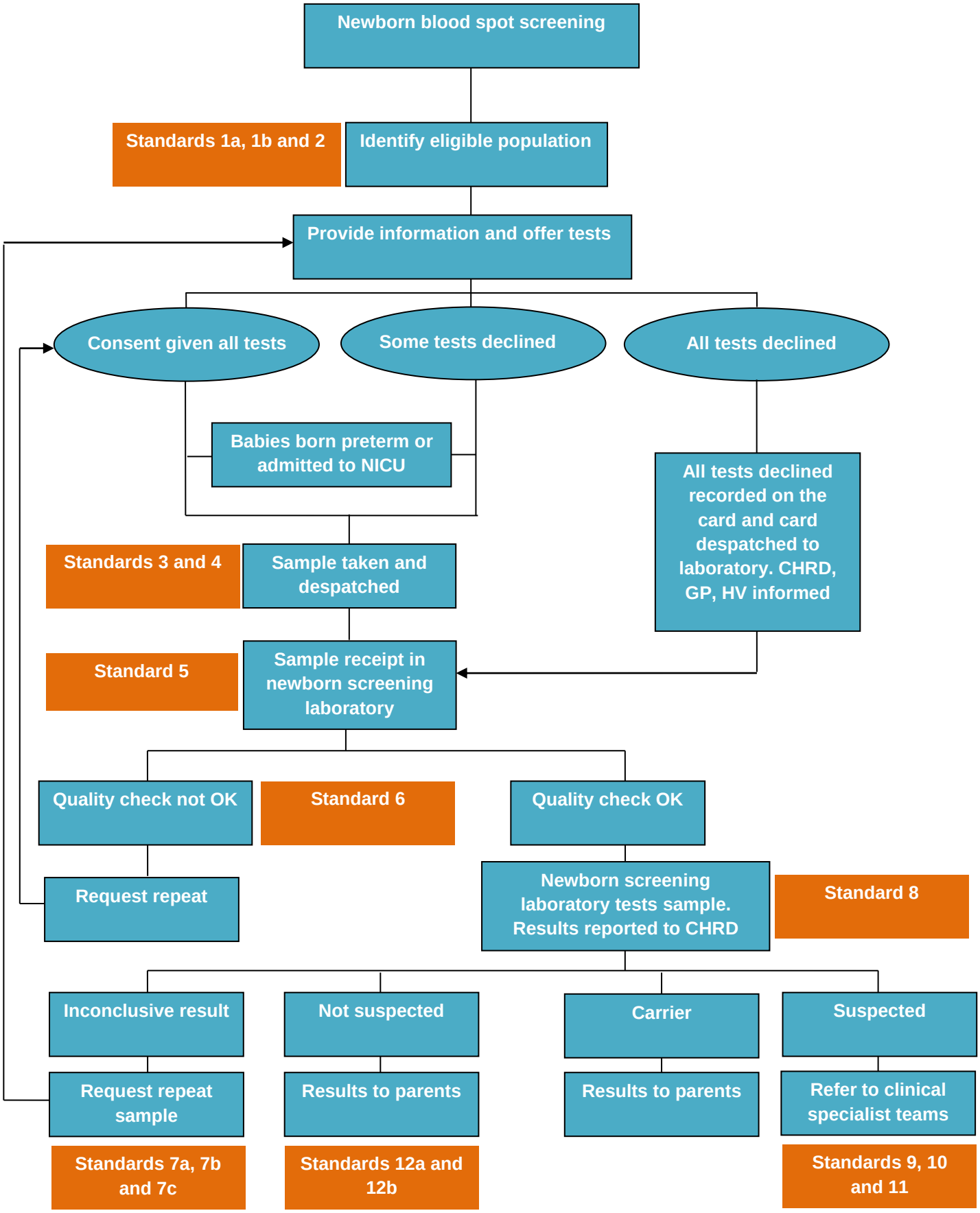
If the screening tests are negative, a copy of the results report is usually given to the child's parent/guardian by their health visitor at their 6 – 8-week review visit. Occasionally, a midwife or health visitor will need to take a second blood spot sample because there was not enough blood collected 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 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 2020-22

Key improvements in the NBSP in Northern Ireland (NI) during 2020-22 have included:

- *implementation of an expanded screening programme on 2nd March 2020 to include screening for the additional inherited metabolic conditions: glutaric aciduria (GA1), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA) and homocystinuria (HCU);*

- *further work on enhancing the failsafe within the NBSP including*
    - *facilitating the implementation of the expanded screening programme.*
    - *reviewing, enhancing and standardising the format of the weekly failsafe report which identifies babies with outstanding conclusive blood spot screening results recorded on the Child Health System and work to develop and implement 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; enhancing the 'failsafe' and providing information for ongoing performance management including the development of failsafe trend reports;*

    - *Development of an additional failsafe for 'older babies' to facilitate a change in national policy regarding timeframes for sampling within the NBSP in England. In line with this policy change, it was agreed that within the NBSP in NI from September 2020 there would also be an extension to the sampling window, of up to 14 calendar days after the first birthday, for babies who are consented and accept screening whilst they are eligible to be offered (i.e. under age of 1 year). Whilst screening and taking of the screening sample is recommended as early as possible, this updated policy allows further flexibility particularly for 'mover in' babies who are close to their first birthday.*

- *the avoidable repeats/blood spot quality subgroup continued to provide regular monitoring, education resources and additional support for test takers to address avoidable repeats rate.*

- *ongoing work with regard to enhancing Quality Assurance structures and processes within the NBSP. This has included the development of a new dedicated part time 'Quality Assurance Facilitator' role to enhance internal QA within each Trust in 2021. Following successful recruitment these posts were operational in all Trusts by December 2022.*

- *the establishment of a subgroup working to prepare for the introduction of an updated regional Laboratory Information Management System across HSCNI. This group has been reviewing and planning for the impact of this change on the NBSP including:*
    - *the electronic interface between the Child health and Regional Newborn Screening Laboratory information systems,*
    - *the programme failsafe,*
    - *and coding structures related to NBSP within these systems.*

- *the Public Health Agency also continues to represent the Northern Ireland NBSP at the national Blood Spot Advisory Group which is hosted by NHS England.*

Programme performance 2020-22

In 2020-22, the Northern Ireland NBS programme 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 NHS England (nationally).

Headline results

Regional data relating to the Northern Ireland NBSP highlight that in 2020-21:

- In terms of coverage, >98% 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, 93.3% of samples were collected on day 5, and 98.5% of samples were received in the newborn screening laboratory within 3 working days of collection.

- 100% of positive screening results (for PKU, IVA, GA1 and CHT) 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 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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- Over 21,000 babies had newborn blood spot screening testing. In total, across all the conditions tested for, 36 babies were identified as screen positive, and 30 of these babies were confirmed as having one of the conditions.

  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 achievable standard for CF

screen positive babies with 0 or 1 mutations with 100% 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 = ≤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 2020-21 and 2021-22 was 4.4% and 4.7% 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 NHS 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 2020 and 31st March 2022 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 2020-21 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 2020-21.

Key definitions

'Born and Resident'

Babies who were born to Northern Ireland residents (at time of birth) in 2020-21 or 2021-22 and were still resident in Northern Ireland at 31st March.

'Movers in'

Babies who were born in 2020-21 or 2021-22, 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]

[4]Patient info https://patient.info/treatment-medication/genetic-testing
[5]NHS 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) 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 2020-22 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. **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[6] was responsible at birth and 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
1b	Completeness of coverage (movers in)	The proportion of **eligible babies for whom a conclusive	≥ 95.0% (for all conditions)	≥ 99.0% for IMDs

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

Standard		Description	Acceptable	Achievable
1b (cont'd)	Completeness of coverage (movers in)	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[6] during the reporting period and for whom the CCG[6] remains responsible on the last day of the reporting period.*		≥ 98.0% for CF, CHT and SCD

[6]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 (cont'd)	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	100%	N/A

Standard		Description	Acceptable	Achievable
12a (cont'd)	Timeliness of results to parents (CCG responsibility at birth)	weeks of birth.		
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 IMD 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 requires a software development and work to develop, agree and finalise. The specification for this development has commenced

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)

Collection of data on Standard 7 commenced in 2021-22.

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. Work to scope out how this could be reported has commenced and has included the development and finalisation of a specification for a software development within CHS to facilitate this.

The performance of the NBSP in Northern Ireland 2020-22 against each of the UK standards is outlined in Table 2.

APPENDIX B: PERFORMANCE OF NBSP IN NI 2020-22

Table 2: Performance of the Northern Ireland Newborn Blood Spot Screening Programme 2020-22 *(data collected by NI Child Health System)*

Standard	Performance 2020-22						Acceptable	Achievable
1a	Total number of 'born and resident' babies =			**2021-22** 21,256		**2020-21** 21,027	≥ 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[7] CHT CF SCD	21,005 20,807 20,970 21,019	98.82% 97.89% 98.65% 98.89%	20,814 20,634 20,776 20,826	98.99% 98.13% 98.81% 99.04%		
	Declines to screening (Numbers and %)	PKU[7] CHT CF SCD	12 12 14 13	0.06% 0.06% 0.07% 0.06%	16 16 16 16	0.08% 0.08% 0.08% 0.08%		
1b	Total number of 'movers in' babies =			**2021-22** 367		**2020-21** 354	≥ 95.0%	≥ 99.0% for IMDs ≥ 98.0% for CF, CHT and SCD
	Completeness of coverage (movers in)	PKU[7] CHT CF SCD	294 292 267 294	80.11% 79.56% 72.75% 80.11%	278 277 245 278	78.53% 78.25% 69.21% 78.53%		
	Declines to screening (Numbers and %)	PKU[7] CHT CF SCD	66 68 63 66	17.98% 18.53% 17.17% 17.98%	73 74 73 73	20.62% 20.90% 20.62% 20.62%		

[7]PKU is reported as proxy to IMD data in line with UK reporting

Standard	Performance 2020-22				Acceptable	Achievable
			2021-22	2020-21		
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 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 =	93.12% (20,336) 21,838	93.34% (20,155) 21,592	≥ 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 =	98.12% (23,441) 23,889	98.50% (23,068) 23,420	≥ 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 2020-22				Acceptable	Achievable
			2021-22	2020-21		
6	Quality of the blood spot sample (avoidable repeat rate)	Avoidable repeat rate	4.7%	4.4%	≤ 2%	≤ 1%
		Total number of first samples =	21,901	21,662		
		Total number of avoidable repeats =	1,029	961		
7a[8]	Timely taking of a second blood spot sample following a CF inconclusive result	% second blood spot samples taken on 21 to 24 days of age following a CF inconclusive result	81.3% (13/16)		≥ 95.0% of second samples taken on day 21 to day 24	≥ 70.0% of second blood spot samples taken on day 21
		% second blood spot samples taken on 21 days of age following a CF inconclusive result				
7b[8]	Timely taking of a second blood spot sample following a borderline CHT result	% second blood spot samples taken between 7 and 10 calendar days after the initial borderline TSH result for CHT	94.1% (32/34)		≥ 95.0%	≥ 99.0%
7c[8]	Timely taking of a second blood spot sample for CHT screening for preterm infants	% second blood spot samples taken for CHT screening for preterm infants taken on 28 days of age or discharge home	79.2% (152/192)		≥ 95.0%	≥ 99.0%
9	Timely processing of all CHT and IMD (excluding HCU) screen positive samples	IMDs	100% (3/3)	100% (15/15)	100% clinical referral initiated within 3 working days of sample receipt in the laboratory	N/A
		CHT	100% (22/22)	100% (18/18)		

[8]Collection of data on Standards 7a, 7b and 7c commenced in 2021-22. NI data relates to repeat samples received by the laboratory in 2021-22. Grey shaded cells indicate where data is not available

Standard	Performance 2020-22				Acceptable	Achievable
			2021-22	2020-21		
11	Timely entry into clinical care	*By 14 days of age* IMDs CHT* **suspected on first sample*	100% (3/3) 100% (13/13[9])	100% (13/13[10]) 100% (9/9[11])	100% by 14 days of age	N/A
		By 21 days of age CHT** ***suspected on a repeat sample*	100% (7/7)	100% (7/7)	100% by 21 days of age	N/A
		By 28 days of age CF (2 mutations)	100% (2/2[12])	100% (14/14[13])	≥ 95% by 28 days of age	100% by 28 days of age
		By 35 days of age CF (1 or 0 mutations)	100% (4/4)	100% (5/5)	≥ 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:	100% (2/2)	100% (1/1)	≥ 90% by 90 days of age	≥ 95% by 90 days of age
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%	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.	NI is currently unable to report on this standard		100%	N/A

[9]One additional baby was diagnosed before screening and is excluded from age data
[10]Two additional babies were diagnosed before screening and are excluded from age data
[11]One additional baby was diagnosed before screening and is excluded from age data
[12]Three additional babies were diagnosed before screening and is excluded from age data
[13]Two 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 2020-21 and 2021-22 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 1a[14] *(data collected by NI Child Health System)* ~ 2019-22

	Year					
	2021-22		2020-21		2019-20	
Total number '**born and resident**' (**B&R**) at 31st March	21,256		21,027		21,929	
Number of **B&R** with decline to screening (02)						
PKU[7]	12	0.06%	16	0.08%	11	0.05%
CHT	12	0.06%	16	0.08%	11	0.05%
CF	14	0.07%	16	0.08%	11	0.05%
SCD	13	0.06%	16	0.08%	12	0.05%
Number (%) of **B&R** with conclusive results by the end of the reporting period (codes 04, 05, 06, 07, 08, 10)						
PKU[7]	21,244	99.94%	21,010	99.92%	21,915	99.94%
CHT	21,244	99.94%	21,010	99.92%	21,913	99.93%
CF	21,241	99.93%	21,006	99.90%	21,915	99.94%
SCD	21,243	99.94%	21,010	99.92%	21,916	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[7]	21,005	98.82%	20,814	98.99%	21,648	98.72%
CHT	20,807	97.89%	20,634	98.13%	21,457	97.85%
CF	20,970	98.65%	20,776	98.81%	21,603	98.51%
SCD	21,019	98.89%	20,826	99.04%	21,658	98.76%

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

[14]excludes 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 requires a software development. Work has commenced to develop, agree and finalise the specification for this.

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 2020-21 and 2021-22 there were 35 and 30 ‘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 - Standard 1a[14,15] *(data collected by NI Child Health System)* ~ 2020-21

	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 31/03/21	7,597		5,065		4,802		3,563		21,027	
Number (%) of **B&R** with decline to screening (02):										
PKU[7]	3		2		7		4		16	0.08%
CHT	3		2		7		4		16	0.08%
CF	3		2		7		4		16	0.08%
SCD	3		2		7		4		16	0.08%
Number (%) of **B&R** with reason for not starting/ incomplete screening (09):										
PKU[7]	0		0		1		0		1	0.00%
CHT	0		0		1		0		1	0.00%
CF	0		1		2		2		5	0.02%
SCD	0		0		1		0		1	0.00%
Number (%) of **B&R** with conclusive results (04, 05, 06, 07, 08, 10):										
PKU[7]	7,594	99.96%	5,063	99.96%	4,794	99.83%	3,559	99.89%	21,010	99.92%
CHT	7,594	99.96%	5,063	99.96%	4,794	99.83%	3,559	99.89%	21,010	99.92%
CF	7,594	99.96%	5,062	99.94%	4,793	99.81%	3,557	99.83%	21,006	99.90%
SCD	7,594	99.96%	5,063	99.96%	4,794	99.83%	3,559	99.89%	21,010	99.92%
Number (%) of **B&R** with conclusive results available by Day 17 (04, 05, 06, 07, 08, 10):										
PKU[7]	7,529	99.10%	5,021	99.13%	4,750	98.92%	3,514	98.62%	20,814	98.99%
CHT	7,463	98.24%	4,978	98.28%	4,717	98.23%	3,476	97.56%	20,634	98.13%
CF	7,519	98.97%	5,017	99.05%	4,741	98.73%	3,499	98.20%	20,776	98.81%
SCD	7,532	99.14%	5,023	99.17%	4,755	99.02%	3,516	98.68%	20,826	99.04%

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

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

[15]unlike 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 - Standard 1a[14,15] *(data collected by NI Child Health System)* ~ 2021-22

	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 31/03/22	7,686		5,039		5,002		3,529		21,256	
Number (%) of **B&R** with decline to screening (02):										
PKU[7]	4		0		3		5		12	0.06%
CHT	4		0		3		5		12	0.06%
CF	4		1		3		6		14	0.07%
SCD	4		1		3		5		13	0.06%
Number (%) of **B&R** with reason for not starting/ incomplete screening (09):										
PKU[7]	0		0		0		0		0	0.00%
CHT	0		0		0		0		0	0.00%
CF	1		0		0		0		1	0.00%
SCD	0		0		0		0		0	0.00%
Number (%) of **B&R** with conclusive results (04, 05, 06, 07, 08, 10):										
PKU[7]	7,682	99.95%	5,039	100.00%	4,999	99.94%	3,524	99.86%	21,244	99.94%
CHT	7,682	99.95%	5,039	100.00%	4,999	99.94%	3,524	99.86%	21,244	99.94%
CF	7,681	99.93%	5,038	99.98%	4,999	99.94%	3,523	99.83%	21,241	99.93%
SCD	7,682	99.95%	5,038	99.98%	4,999	99.94%	3,524	99.86%	21,243	99.94%
Number (%) of **B&R** with conclusive results available by Day 17 (04, 05, 06, 07, 08, 10):										
PKU[7]	7,592	98.78%	4,985	98.93%	4,956	99.08%	3,472	98.38%	21,005	98.82%
CHT	7,519	97.83%	4,941	98.06%	4,918	98.32%	3,429	97.17%	20,807	97.89%
CF	7,581	98.63%	4,977	98.77%	4,950	98.96%	3,462	98.10%	20,970	98.65%
SCD	7,602	98.91%	4,988	98.99%	4,956	99.08%	3,473	98.41%	21,019	98.89%

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

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

[15]unlike 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 1b[14] *(data collected by NI Child Health System)* ~ 2019-22

	Year					
	2021-22		2020-21		2019-20	
Total number 'Movers In' (**MI**) resident at 31st March	367		354		410	
Number of **MI** with decline to screening (02)						
PKU[7]	66	17.98%	73	20.62%	66	16.10%
CHT	68	18.53%	74	20.90%	68	16.59%
CF	63	17.17%	73	20.62%	63	15.37%
SCD	66	17.98%	73	20.62%	66	16.10%
Number (%) of **MI** with reason for not starting/incomplete (code 09-)						
PKU[7]	0	0.00%	1	0.28%	0	0.00%
CHT	0	0.00%	1	0.28%	0	0.00%
CF	30	8.17%	35	9.89%	30	7.32%
SCD	0	0.00%	1	0.28%	0	0.00%
Number (%) of **MI** with conclusive results(codes 04, 05, 06, 07, 08,10)						
PKU[7]	294	80.11%	278	78.53%	294	71.71%
CHT	292	79.56%	277	78.25%	292	71.22%
CF	267	72.75%	245	69.21%	267	65.12%
SCD	294	80.11%	278	78.53%	294	71.71%

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

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

Table 4b Completeness of coverage – Movers in by Child Health Area - Standard 1b[14,15] *(data collected by NI Child Health System)* ~ 2020-21

	Eastern CHS Area		Northern CHS Area		Southern CHS Area		Western CHS Area		N Ireland	
Total number of '**Movers In**' (**MI**) resident at 31/03/21	113		92		101		48		354	
Number of **MI** with decline to screening (02):										
PKU[7]	29		8		28		8		73	20.62%
CHT	29		8		29		8		74	20.90%
CF	30		7		28		8		73	20.62%
SCD	29		8		28		8		73	20.62%
Number of **MI** with reason for not starting/incomplete screening (09):										
PKU[7]	0		1		0		0		1	0.28%
CHT	0		1		0		0		1	0.28%
CF	10		6		10		9		35	9.89%
SCD	0		1		0		0		1	0.28%
Number (%) of **MI** with conclusive results (04, 05, 06, 07, 08, 10):										
PKU[7]	84	74.34%	83	90.22%	71	70.30%	40	83.33%	278	78.53%
CHT	84	74.34%	83	90.22%	70	69.31%	40	83.33%	277	78.25%
CF	73	64.60%	79	85.87%	62	61.39%	31	64.58%	245	69.21%
SCD	84	74.34%	83	90.22%	71	70.30%	40	83.33%	278	78.53%

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

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

[15]unlike 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 by Child Health Area - Standard 1b[14,15] *(data collected by NI Child Health System)* ~ 2021-22

	Eastern CHS Area		Northern CHS Area		Southern CHS Area		Western CHS Area		N Ireland	
Total number of '**Movers In**' (**MI**) resident at 31/03/22	112		113		80		62		367	
Number of **MI** with decline to screening (02):										
PKU[7]	33		10		16		7		66	17.98%
CHT	33		10		18		7		68	18.53%
CF	30		10		16		7		63	17.17%
SCD	33		10		16		7		66	17.98%
Number of **MI** with reason for not starting/incomplete screening (09):										
PKU[7]	0		0		0		0		0	0.00%
CHT	0		0		0		0		0	0.00%
CF	10		7		5		8		30	8.17%
SCD	0		0		0		0		0	0.00%
Number (%) of **MI** with conclusive results (04, 05, 06, 07, 08, 10):										
PKU[7]	74	66.07%	101	89.38%	64	80.00%	55	88.71%	294	80.11%
CHT	74	66.07%	101	89.38%	62	77.50%	55	88.71%	292	79.56%
CF	67	59.82%	94	83.19%	59	73.75%	47	75.81%	267	72.75%
SCD	74	66.07%	101	89.38%	64	80.00%	55	88.71%	294	80.11%

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

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

[15]unlike 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 page 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 2020-21 and in 2021-22, 98.7% and 98.9% respectively of all samples received by the Laboratory had the HCN included[16]. 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 2020-21 and 2021-22, with more than 98% of first samples taken between 5 to 8 days after birth; this performance is similar to 2019-20.

[16]Data Source: Regional Newborn Screening Laboratory

Table 5a – Sample collection – Standard 4 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

	Year		
	2021-22	**2020-21**	**2019-20**
Total number of first samples	21,838	21,592	22,639
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	63	70	79
	Year		
	2021-22	**2020-21**	**2019-20**
Number of first samples taken on or before day 4 (%)	41 (0.19%)	49 (0.23%)	79 (0.35%)
Number of first samples taken on day 5 (%)	20,336 (93.12%)	20,155 (93.34%)	21,447 (94.73%)
Number of first samples taken on or after day 9 (%)	327 (1.50%)	343 (1.59%)	367 (1.62%)
Number of first samples taken between day 5 and day 8 (%)	21,470 (98.31%)	21,200 (98.18%)	22,193 (98.03%)

Table 5b – Sample collection – Standard 4 by Trust[17] *(data collected by the Regional Newborn Screening Laboratory)* ~ 2020-21

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[18]	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	1,742	4,088	1,899	5,142	5,040	3,681	21,592
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	9	12	7	16	18	8	70
Number of first samples taken on or before day 4	7	9	4	9	11	9	49
Number of first samples taken on day 5	1,618	3,863	1,810	4,594	4,743	3,527	20,155
Number of first samples taken on or after day 9	44	64	12	49	116	58	343
Number of first samples taken between day 5 and day 8	1,691	4,015	1,883	5,084	4,913	3,614	21,200
% of first samples taken on or before day 4	0.40%	0.22%	0.21%	0.18%	0.22%	0.24%	0.23%
% of first samples taken on day 5	92.88%	94.50%	95.31%	89.34%	94.11%	95.82%	93.34%
% of first samples taken on or after day 9	2.53%	1.57%	0.63%	0.95%	2.30%	1.58%	1.59%
% of first samples taken between day 5 and day 8	97.07%	98.21%	99.16%	98.87%	97.48%	98.18%	98.18%

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

[18]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

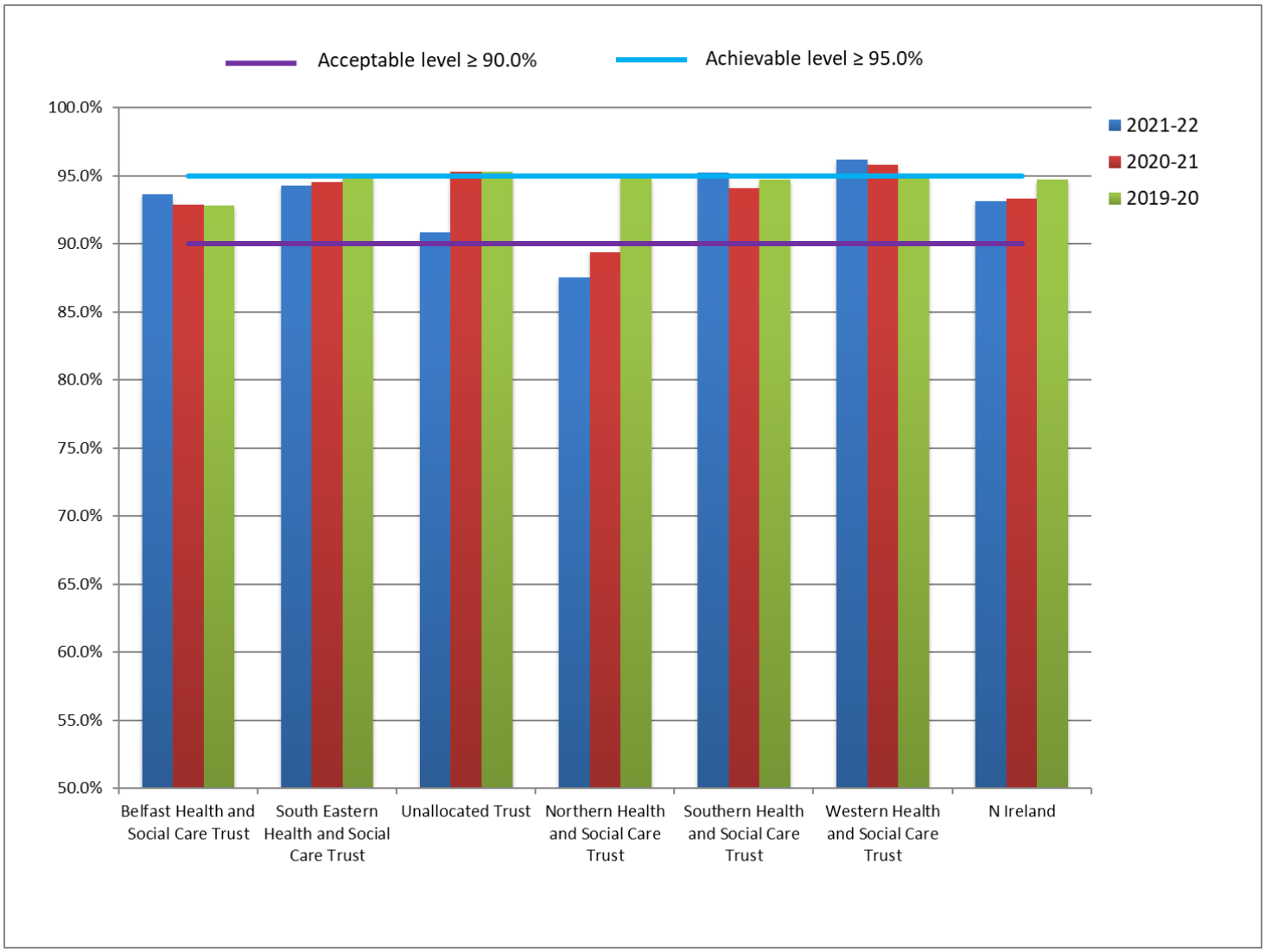
Table 5c – Sample collection – Standard 4 by Trust[17] *(data collected by the Regional Newborn Screening Laboratory)* ~ 2021-22

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[18]	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	2,249	5,488	153	5,172	5,184	3,592	21,838
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	11	11	2	12	14	13	63
Number of first samples taken on or before day 4	6	8	1	7	10	9	41
Number of first samples taken on day 5	2,106	5,172	139	4,528	4,936	3,455	20,336
Number of first samples taken on or after day 9	50	78	4	55	87	53	327
Number of first samples taken between day 5 and day 8	2,193	5,402	148	5,110	5,087	3,530	21,470
% of first samples taken on or before day 4	0.27%	0.15%	0.65%	0.14%	0.19%	0.25%	0.19%
% of first samples taken on day 5	93.64%	94.24%	90.85%	87.55%	95.22%	96.19%	93.12%
% of first samples taken on or after day 9	2.22%	1.42%	2.61%	1.06%	1.68%	1.48%	1.50%
% of first samples taken between day 5 and day 8	97.51%	98.43%	96.73%	98.80%	98.13%	98.27%	98.31%

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

[18]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 ~ 2019-22



In 2020-21 and 2021-22 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).

In general, the proportion of samples taken on Day 5 has been increasing over the 2019-22 period. In NHSCT the proportion of samples taken on Day 5 has declined year on year from 2019-22, and the Trust advise that due to staffing pressures and weekend workload Day 5 samples may be deferred until Day 6.

Table 6a – Timely receipt of a sample in the laboratory - Standard 5 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

	Year		
	2021-22	**2020-21**	**2019-20**
Total number of ALL samples (first, repeat and second samples) included	23,889	23,420	25,046
Number of samples EXCLUDED (because date of specimen was not recorded on the cards)	70	74	93
Number of ALL samples received by the lab in 3 or fewer working days of sample being taken (%)	23,441 (98.12%)	23,068 (98.50%)	24,630 (98.34%)
Number of ALL samples received by the lab in 4 or fewer working days of sample being taken (%)	23,727 (99.32%)	23,303 (99.50%)	24,909 (99.45%)

Table 6b Timely receipt of a sample in the laboratory by Trust[17]– Standard 5 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2020-21

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[18]	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	1,971	4,418	2,043	5,477	5,446	4,065	23,420
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	9	12	8	19	18	8	74
Number of all samples received by the lab in 3 or fewer working days of sample being taken	1,953	4,388	2,029	5,449	5,381	3,868	23,068
Number of all samples received by the lab in 4 or fewer working days of sample being taken	1,967	4,411	2,043	5,467	5,418	3,997	23,303
Number of all samples received by the lab on or after 5 working days of sample being taken	4	7	0	10	28	68	117

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[18]	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
% of all samples received by lab in 3 or fewer working days of sample being taken	99.09%	99.32%	99.31%	99.49%	98.81%	95.15%	98.50%
% of all samples received by lab in 4 or fewer working days of sample being taken	99.80%	99.84%	100.00%	99.82%	99.49%	98.33%	99.50%
% of all samples received by lab in 5 or more working days of sample being taken	0.20%	0.16%	0.00%	0.18%	0.51%	1.67%	0.50%

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

[18]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

Table 6c Timely receipt of a sample in the laboratory by Trust[17] – Standard 5 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2021-22

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[18]	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	2,537	5,948	175	5,665	5,648	3,916	23,889
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	12	13	2	13	17	13	70
Number of all samples received by the lab in 3 or fewer working days of sample being taken	2,483	5,918	167	5,624	5,597	3,652	23,441
Number of all samples received by the lab in 4 or fewer working days of sample being taken	2,513	5,937	169	5,650	5,628	3,830	23,727
Number of all samples received by the lab on or after 5 working days of sample being taken	24	11	6	15	20	86	162

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[18]	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
% of all samples received by lab in 3 or fewer working days of sample being taken	97.87%	99.50%	95.43%	99.28%	99.10%	93.26%	98.12%
% of all samples received by lab in 4 or fewer working days of sample being taken	99.05%	99.82%	96.57%	99.74%	99.65%	97.80%	99.32%
% of all samples received by lab in 5 or more working days of sample being taken	0.95%	0.18%	3.43%	0.26%	0.35%	2.20%	0.68%

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

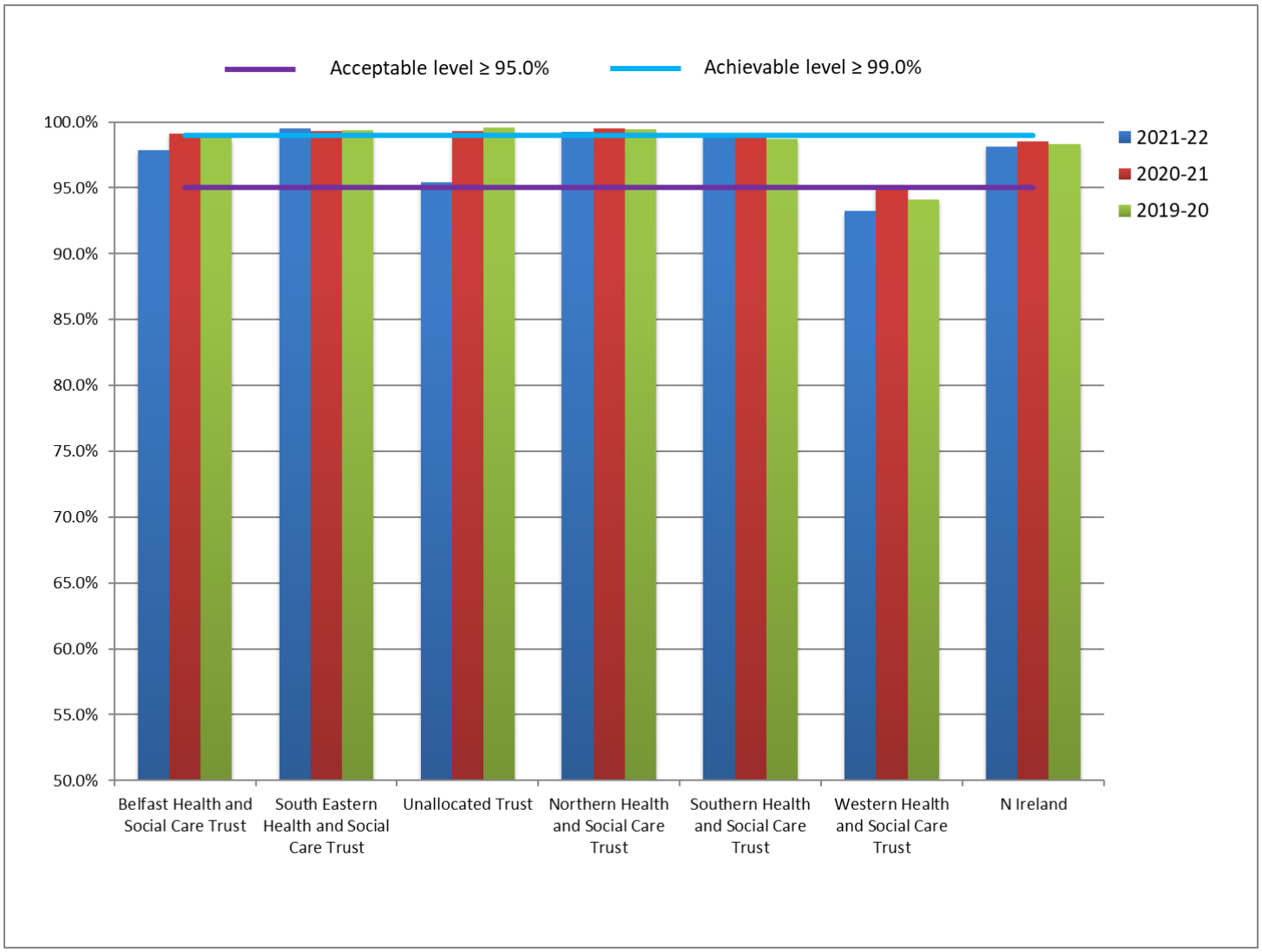
[18]Unallocated Trust is assigned where it is not possible to determine from the data entered by the test taker on the card whether the responsible Trust is Belfast Health and Social Care Trust or South Eastern Health and Social Care Trust

Historically the Western Trust, has a lower percentage of samples being received in the laboratory in 3 or fewer days of the sample being taken, and this could be due to its wider geographical area.

During this reporting period WHSCT identified issues with the postage system as a result of Covid with major delays to postage services in a local sorting office due to staffing shortages from October 2020 – June 2021. This is a contributing factor in the time it took for samples to get to the Regional Newborn Screening Laboratory. Also, the community midwives relocated to a community hub in January 2021 and commenced postnatal clinics. Newborn bloodspot samples were then posted in

batches the next working day and this combined with Royal Mail staffing issues are factors that potentially contributed to delays.

Figure 2: Percentage of samples received by the laboratory in less than or equal to 3 working days of sample collection ~ 2019-22



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 2021-22 and 2020-21 was 4.70% and 4.44% respectively and therefore the NBSP did not meet the acceptable standard (avoidable repeat rate ≤2%) - see Tables 7a and 7b.

Table 7a - Quality of the blood spot sample - Standard 6 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

	Year		
	2021-22	**2020-21**	**2019-20**
Total number of first samples received	21,901	21,662	22,718
Total number of avoidable repeat samples requested	1,029	961	1,371
REASON FOR REPEAT			
Avoidable Repeats			
Too young for reliable screening (≤ 4 days) (%)	41 (0.19%)	47 (0.22%)	78 (0.34%)
Insufficient sample	682 (3.11%)	632 (2.92%)	919 (4.05%)
Unsuitable sample[19]	306 (1.40%)	282 (1.30%)	374 (1.65%)
Total Avoidable Repeats	**1,029 (4.70%)**	**961 (4.44%)**	**1,371 (6.03%)**

[19]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)* ~ 2020-21

	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[18]	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	1,751	4,100	1,906	5,158	5,058	3,689	21,662
Total number of repeat samples requested	**137**	**212**	**115**	**232**	**257**	**243**	**1,196**
REASON FOR REPEAT							
Avoidable Repeats							
Too young for reliable screening (≤4 days)	7	8	4	8	11	9	47
Insufficient sample	79	112	61	103	129	148	632
Unsuitable sample[19]	29	54	25	67	61	46	282
Total Avoidable Repeats	**115**	**174**	**90**	**178**	**201**	**203**	**961**
Avoidable Repeats Request Rates							
% Too young for reliable screening (≤4 days)	0.40%	0.20%	0.21%	0.16%	0.22%	0.24%	0.22%
% Insufficient sample	4.51%	2.73%	3.20%	2.00%	2.55%	4.01%	2.92%
% Unsuitable sample[19]	1.66%	1.32%	1.31%	1.30%	1.21%	1.25%	1.30%
% Avoidable Repeats	**6.57%**	**4.24%**	**4.72%**	**3.45%**	**3.97%**	**5.50%**	**4.44%**
Unavoidable Repeats[20]							
Total Unavoidable Repeats	**22**	**38**	**25**	**54**	**56**	**40**	**235**
Unavoidable Repeats Request Rates							
% Unavoidable Repeats	**1.26%**	**0.93%**	**1.31%**	**1.05%**	**1.11%**	**1.08%**	**1.08%**

[18]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

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

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

[21]Two samples that were taken on day 4 were accepted under COVID-19 UK revised sample acceptance criteria and are therefore not included in the 'Too young for reliable screening (≤4 days)' figures above

Table 7c - Quality of the blood spot sample by Trust - Standard 6 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2021-22

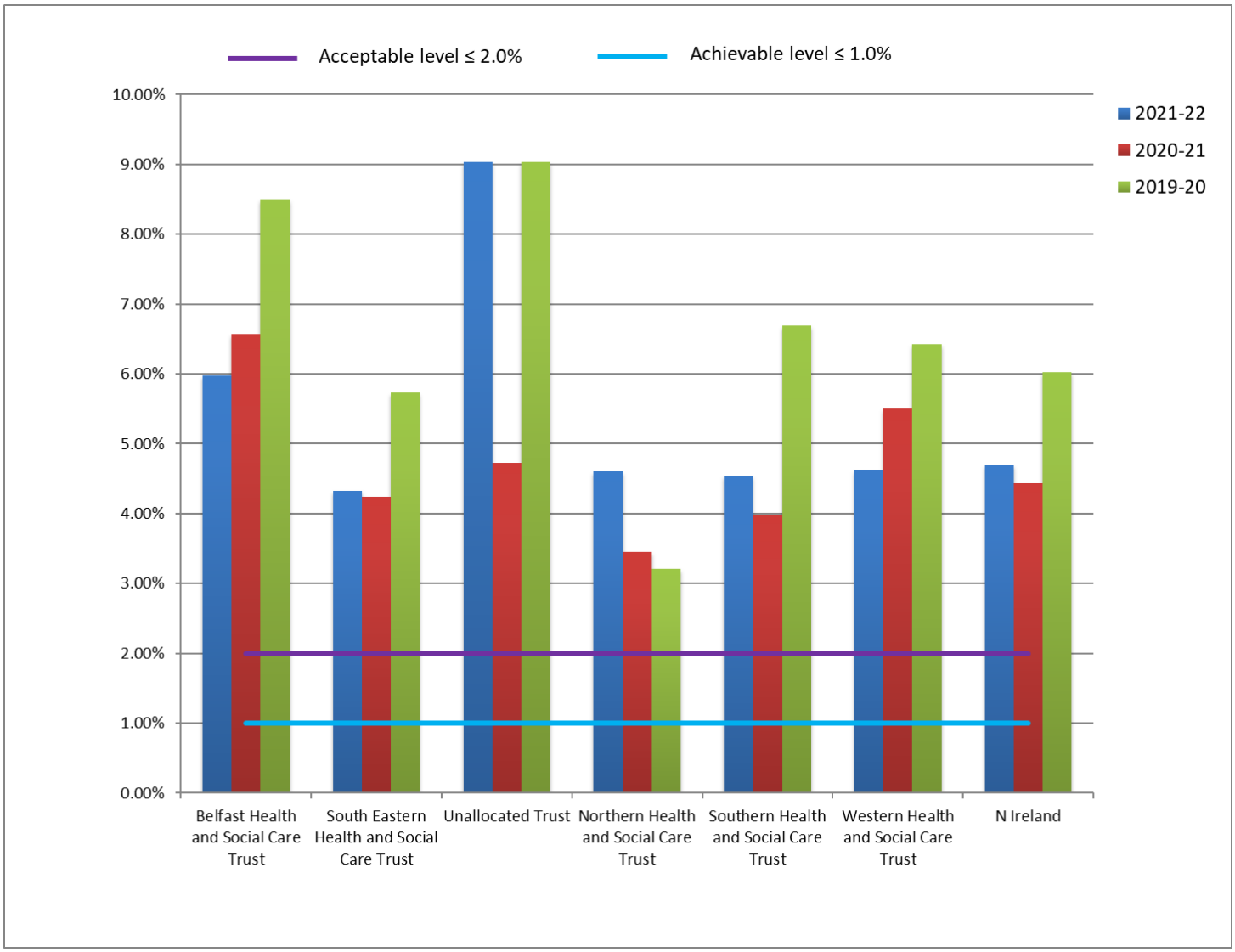
	Belfast Health and Social Care Trust	South Eastern Health and Social Care Trust	Unallocated Trust[18]	Northern Health and Social Care Trust	Southern Health and Social Care Trust	Western Health and Social Care Trust	N Ireland
Total number of first samples received	2,260	5,499	155	5,184	5,198	3,605	21,901
Total number of repeat samples requested	**179**	**285**	**18**	**294**	**295**	**209**	**1,280**
REASON FOR REPEAT							
Avoidable Repeats							
Too young for reliable screening (≤4 days)	6	8	1	7	10	9	41
Insufficient sample	87	164	7	173	149	102	682
Unsuitable sample[19]	42	66	6	59	77	56	306
Total Avoidable Repeats	**135**	**238**	**14**	**239**	**236**	**167**	**1,029**
Avoidable Repeats Request Rates							
% Too young for reliable screening (≤4 days)	0.27%	0.15%	0.65%	0.14%	0.19%	0.25%	0.19%
% Insufficient sample	3.85%	2.98%	4.52%	3.34%	2.87%	2.83%	3.11%
% Unsuitable sample[19]	1.86%	1.20%	3.87%	1.14%	1.48%	1.55%	1.40%
% Avoidable Repeats	**5.97%**	**4.33%**	**9.03%**	**4.61%**	**4.54%**	**4.63%**	**4.70%**
Unavoidable Repeats[20]							
Total Unavoidable Repeats	**44**	**47**	**4**	**55**	**59**	**42**	**251**
Unavoidable Repeats Request Rates							
% Unavoidable Repeats	**1.95%**	**0.85%**	**2.58%**	**1.06%**	**1.14%**	**1.17%**	**1.15%**

[18]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

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

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

Figure 3: Avoidable Repeat Request Rates ~ 2019-22



In 2020-21 and 2021-22, the majority of avoidable repeats (2.92% and 3.11% (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 general trend in Trust avoidable repeat rates has been a steady decline over the 2019-22 period. In 2019 the NHSCT had the lowest avoidable repeat rate of (3.1%), however this has increased year on year since then. The Trust has advised that there has been an increase in the number of new staff involved in

NBSP over this period in response to staff retirements, and this may have attributed to the increase in the avoidable repeat rate.

The NHSCT in 2020-21 and the SEHSCT in 2021-22 had the lowest rates of avoidable repeats (3.45% and 4.33% respectively) and the BHSCT had the highest rates in both 2020-21 (6.57%) and 2021-22 (5.97%).

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, MCADD, IVA, GA1, HCU, MSUD

In 2020-21, 12 babies (including 2 who were tested early due to a family history) were identified with positive screening tests for PKU. In 2021-22 3 babies were identified with positive screening tests for PKU. In 2020-21 and in 2021-22, there were no babies identified as having positive screening tests for MCADD, HCU or MSUD. As in previous years, all babies who had a positive screening result for PKU (2020-22) (100%) of these babies were referred within 3 working days of sample receipt in the laboratory and seen by the clinical team by 14 days of age, therefore meeting the acceptable standard (by 14 days of age - see Table 8a).

In 2020-21, 2 babies were identified with positive screening tests for IVA. Both babies were referred within 3 working days of sample receipt in the laboratory (100%) and seen by the clinical team by 14 days of age (100%), therefore meeting the acceptable standard. In 2021-22 there were no babies identified with a positive screening test for IVA (see Table 8b).

In 2020-21, 1 baby was identified with a positive screening test for GA1 and was referred within 3 working days of sample receipt in the laboratory (100%) and seen by the clinical team by 14 days of age (100%), therefore meeting the acceptable standard. In 2021-22 there were no babies identified with a positive screening test for GA1 (see Table 8c).

Outcomes

A diagnosis of PKU was confirmed in 10 babies who were PKU screen positive in 2020-21 and in 3 babies who were PKU screen positive in 2021-22. A diagnosis of GA1 was confirmed in 1 baby who was GA1 screen positive in 2020-21.

Table 8a – PKU Clinical Data – Standards 9 and 11 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

	Year		
	2021-22	**2020-21**	**2019-20**
Number of babies with screen positive result (status code = 07 - not suspected other disorder follow up or 08 - suspected)	3	12[22]	6[23,24]
Number of screen positive babies with clinical referral initiated within 3 working days of sample receipt in lab (%)	3 (100%)	12 (100%)	6 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	3 (100%)	10 (100%)	4 (100%)

[22]Two babies were diagnosed before screening and are excluded from age data
[23]One baby did not require follow up with clinical services and is therefore excluded from age data
[24]One baby was diagnosed before screening and is excluded from age data

Table 8b IVA Clinical data – Standards 9 and 11 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2020-22

	Year	
	2021-22	**2020-21**
Number of babies with screen positive result (status code = 08 - suspected)	0	2
Number of screen positive babies with clinical referral initiated within three working days of sample receipt in lab (%)	N/A	2 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	N/A	2 (100%)

Table 8c GA1 Clinical data – Standards 9 and 11 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2020-22

	Year	
	2021-22	**2020-21**
Number of babies with screen positive result (status code = 08 - suspected)	0	1
Number of screen positive babies with clinical referral initiated within three working days of sample receipt in lab (%)	N/A	1 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	N/A	1 (100%)

CHT

In 2020-21, 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 9 were seen by the clinical team by 14 days of age (one baby who was diagnosed before screening has been excluded from the age data).

A further 8 babies were also identified as screen positive. 7 of these babies were positive on a repeat sample following a borderline result (Table 8d) and were seen by the clinical team by 21 days of age.

In 2021-22, 14 babies were identified as CHT screen positive on their first blood spot sample; all 14 babies (100%) were referred to specialist clinical teams within 3 days of sample receipt in the laboratory and 13 were seen by the clinical team by 14 days of age (one baby who was diagnosed before screening has been excluded from the age data).

A further 8 babies were also identified as screen positive. 7 of these babies were positive on a repeat sample following a borderline result (Table 8d) and were seen by the clinical team by 21 days of age.

<u>Outcomes</u>

In 2020-21 a diagnosis of CHT was confirmed in 17 (94.4%) of the 18 babies who were identified as screen positive. In 2021-22 a diagnosis of CHT was confirmed in 20 (90.9%) of the 22 babies who were identified as screen positive.

Table 8d - CHT Clinical Data - Standards 9 and 11 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

	Year					
	2021-22[25]		**2020-21[27]**		**2019-20**	
	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)	14[26]	7	10[28]	7	10	6[29]
Number of screen positive babies with clinical referral initiated within 3 working days of sample receipt in lab (%)	14 (100%)	7 (100%)	10 (100%)	7 (100%)	10 (100%)	6 (100%)
Number of screen positive babies who were seen by 14 days of age (suspected on first sample)	13 (100%)	N/A	9 (100%)	N/A	10 (100%)	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	7 (100%)	N/A	7 (100%)	N/A	5 (100%)

[25]There was an additional pre-term baby who was screen positive for CHT
[26]One baby was diagnosed before screening and has been excluded from age data
[27]There were an additional pre-term baby who was screen positive for CHT
[28]One baby was diagnosed before screening and has been excluded from age data
[29]One baby was diagnosed before screening and has been excluded from age data

SCD

In 2020-21 1 baby who moved in to Northern Ireland after 90 days of age was identified as screen positive for SCD and was seen by specialist clinical services within 8 days of clinical referral. In 2020-21, the screening results for an additional 35 babies required further testing/assessment: 1 was identified with other potentially clinically significant condition, 21 were identified as sickle cell 'carriers' and 13 as a carrier of another unusual haemoglobin gene.

In 2021-22, 2 babies were identified as screen positive for SCD and were seen by specialist clinical services within 90 days of age, meeting the achievable standard. In 2021-22, the screening results for an additional 54 babies required further testing/assessment: 5 were identified with other potentially clinically significant condition, 34 were identified as sickle cell 'carriers' and 15 as a carrier of another unusual haemoglobin gene.

<u>Outcomes</u>

In 2020-21 a diagnosis of SCD was confirmed in the baby who was identified as screen positive and in 2021-22, in 1 (50.0%) of the 2 babies who were identified as screen positive.

Table 8e - SCD Clinical Data - Standard 11 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

Condition/Disorder	Year		
	2021-22	**2020-21**	**2019-20**
Sickle Cell Disease or Other Potentially Clinically Significant Condition[30]	2	1	1
Condition/Disorder			
	2021-22	**2020-21**	**2019-20**
Sickle Cell Carrier or Carrier of Other Unusual Haemoglobin Gene[31] or Possible Benign Disorder[32]	54	35	53

[30] Other potentially clinically significant condition comprise F, FE, FEA or HbAF
[31] Carrier of other unusual haemoglobin gene comprise FAC, FAD, ?FAD, FAE or FAO^{Arab}
[32] Other possible benign disorder comprise FC, FD, FO^{Arab}, FCA, FDA or FO^{Arab}A

CF

2 mutations

In 2020-21, 16 babies were identified on screening as suspected CF with 2 genetic mutations; 14 (100%) were seen by the clinical team by 28 days of age, meeting the achievable standard (2 babies diagnosed before screening have been excluded from the age data). Fourteen of the 16 babies identified on screening as suspected CF with 2 genetic mutations were confirmed as having CF.

In 2021-22, 5 babies were identified on screening as suspected CF with 2 genetic mutations; 2 (100%) were seen by the clinical team by 28 days of age, meeting the achievable standard (3 babies were diagnosed before screening and have been excluded from the age data) (Table 8f). CF was confirmed in 4 babies identified on screening as suspected CF with 2 genetic mutations.

1 or 0 mutations

In 2020-21, a further 5 babies were identified as suspected CF with 1 or 0 mutations and an IRT ≥ 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 2 of these children.

In 2021-22, a further 4 babies were identified as suspected CF with 1 or 0 mutations and an IRT ≥ cut-off 2 on screening and required follow-up. All 4 babies (100%) were seen by the clinical team by 35 days of age. CF was confirmed in 2 of these babies.

Outcomes

In 2020-21, a total of 21 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 16 of these babies. In addition, 5 babies were identified as probable carriers of a gene for CF (Table 8h).

In 2021-22, 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 6 of these babies. In addition, 7 babies were identified as probable carriers of a gene for CF (Table 8h).

Table 8f - CF Clinical Data - Standard 11 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

	Year		
	2021-22	**2020-21**	**2019-20**
No. of CF suspected babies with 2 mutations	5[33]	16[34]	4[35]
Number of screen positive babies who were seen by 28 days of age (%)	2 (100%)	14 (100%)	3 (100%)

[33]Three babies were diagnosed before screening and have been excluded from age data
[34]Two babies were diagnosed before screening and have been excluded from age data
[35]One baby was diagnosed before screening and has been excluded from age data

Table 8g - CF Clinical Data - Standard 11 *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

	Year		
	2021-22	**2020-21**	**2019-20**
No. of CF suspected babies with 1 or 0 mutations and IRT ≥ cut-off 2	4	5	5
Number of screen positive babies who were seen by 35 days of age	4 (100%)	5 (100%)	4 (80%)

Table 8h – CF Clinical Data – CF gene carriers *(data collected by the Regional Newborn Screening Laboratory)* ~ 2019-22

	Year		
	2021-22	**2020-21**	**2019-20**
No. of babies with 1 CF mutation detected and second IRT < cut-off 2	7	5	10

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