Newborn Blood Spot Screening in Northern Ireland

Annual Report 2016 - 17

HSC Public Health Agency

Newborn Blood Spot Screening Programme

CONTENTS:

Summary

This report, the first annual report of the Northern Ireland Newborn Blood Spot Screening Programme (NBSP), summarises the performance of the programme against key standards for the financial year 2016-17.

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

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

Headline Results

The most recently published national report (2016-17)[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 2016-17:

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

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

- NI was the best performing UK region in relation to timing of sample collection and processing, with 98.3 % of samples collected between 5-8 days of age, and 99.5% of samples received in the newborn screening laboratory within 4 working days of collection.

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

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

- Over 23,000 babies had newborn blood spot screening testing. In total, across all of the five conditions tested for, 44 babies were identified as screen positive and 32 of these babies were confirmed as having one of the conditions.

At a national level, meeting the standard (acceptable = ≤2%; achievable = ≤0.5%) in relation to 'avoidable repeats' has proved challenging since the introduction of the programmes, and variation exists across the UK. An avoidable repeat refers to a sample that has not met the required quality standard to be accepted by the laboratory for analysis, e.g. an insufficient quantity of blood may have been collected and the laboratory will request a repeat sample.

In Northern Ireland the avoidable repeat rate was 4.39% in 2016-17. The regional NBSP Quality Improvement (QI) group continues to work to understand and reduce avoidable repeats.

SECTION A: INTRODUCTION AND HEADLINE RESULTS FOR 2016-17

Background

The Northern Ireland Newborn Blood Spot Programme (NBSP) offers all newborn babies a blood spot screening test to identify if they are at increased risk of five rare, but serious, inherited conditions (these are described on pages 7-9). 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 2016 - 31st March 2017 (hereafter referred to as 2016-17) against national standards.

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

- **Phenylketonuria (PKU)**

About 1 in 6,000 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, mental disability. Screening means babies with PKU can be treated early through a

[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

special diet, which will prevent severe disability and allow them to lead a normal life.

• **Congenital Hypothyroidism (CHT**)

About 1 in 3,000 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 mental disability. Screening means babies with CHT can be treated early with thyroxine medication, which will prevent serious disability and allow them to develop normally.

• **Cystic Fibrosis (CF)**

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

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

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

• **Sickle Cell Disorders (SCD)**

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

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

Screening pathway

Who is eligible for NBSP screening?

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

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

What does screening involve?

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

The blood spot test involves taking a small sample of blood from the baby's heel; this is often referred to as the 'heel prick' test. The sample is sent to the Regional Newborn Screening Laboratory in 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.

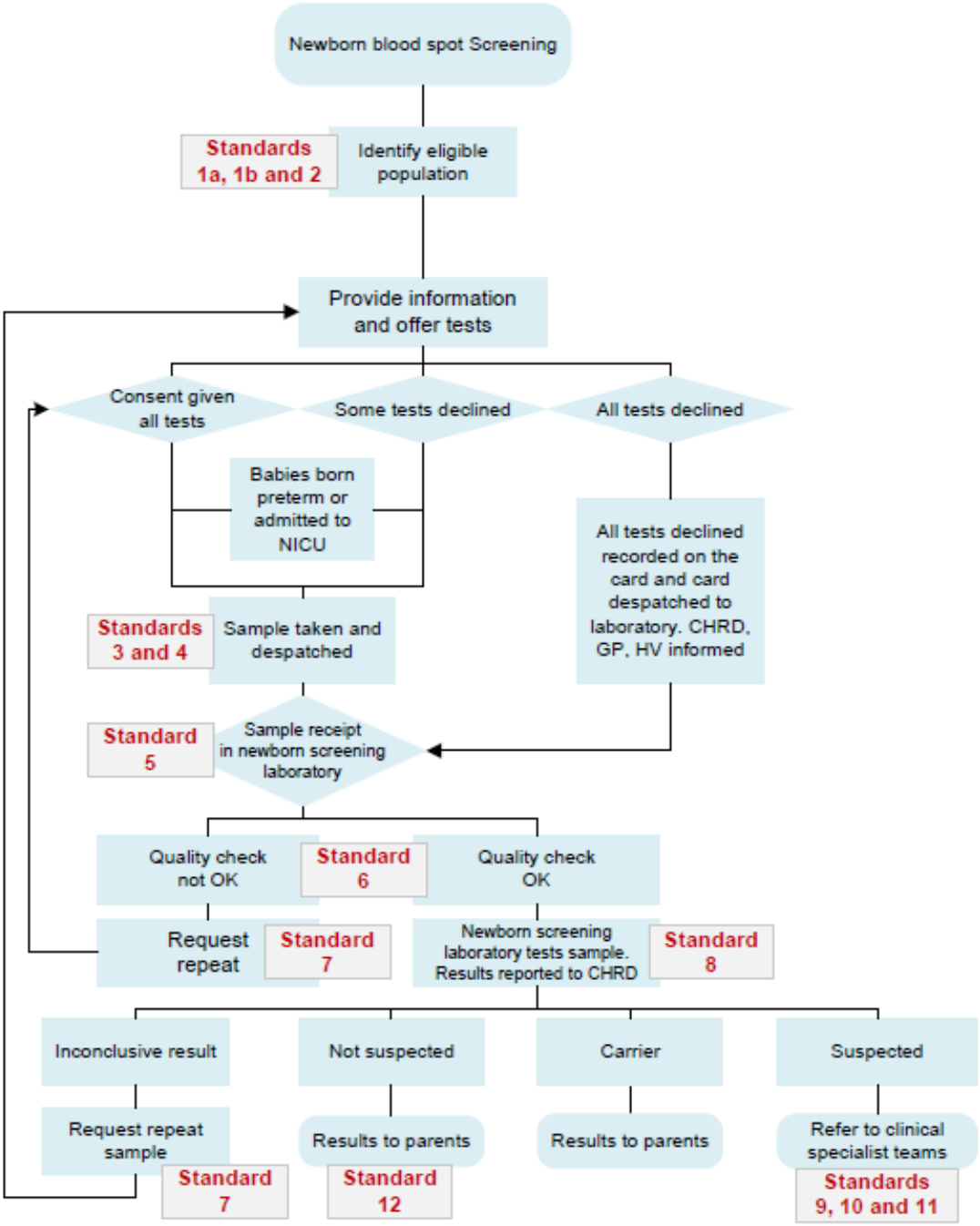
The purpose of screening is to identify babies more likely to have these conditions. If the screening test is positive, a baby will be offered further tests or investigations to confirm the diagnosis. Where one of these conditions is confirmed as present, effective interventions are available to prevent 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 involved. However, there are also built in 'failsafes' to improve safety and quality. 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 2015 https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/463734/AAA_screening_2015-09-23_Failsafe_v2.1.pdf

Figure 1: Screening pathway mapped to programme standards[4]



NICU = neonatal intensive care unit
CHRD = child health records department

[4] © Crown Copyright 2018. This information was originally developed by Public Health England Screening (https://www.gov.uk/topic/population-screening-programmes) and is used under the Open Government Licence v3.0

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 number of multi-disciplinary teams support and deliver the programme within each of the five Health and Social Care Trusts (HSCTs) in Northern Ireland. These include: midwifery, neonatal, paediatric and health visiting staff who deliver informed choice and collection of a high quality blood spot sample. 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 also conduct 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 multidisciplinary professionals in Trust teams delivering the programme to promote compliance with national standards and continuous improvement. A regional NBSP Quality Improvement (QI) group chaired by PHA, with representatives from each of the professional groups and Trusts involved, also meets biannually**.**

Key developments 2016-17

Key improvements in the NBSP in Northern Ireland (NI) during 2016-17 included the development of an electronic interface for the transfer and receipt of newborn blood spot results that are sent from the Laboratory Information System to the Child Health System. An electronic interface reduces the need to manually enter

results, increases efficiency and improves quality and patient safety by automatically matching results using a baby's unique demographic details (gender, date of birth, surname and Health and Care Number).

In July 2016 the Department of Health approved recommendations to expand the Newborn Blood Spot Screening Programme in Northern Ireland to include a further four inherited metabolic diseases. Preparatory work has included developing business processes to support procurement of new laboratory equipment and the enhancement of laboratory services to support expanded screening.

The expanded screening programme will test for the following additional inherited metabolic diseases:

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

Implementation of expanded screening is anticipated to commence during 2019-20.

In 2016-17, to reflect developments in the regional CF service, work also commenced on updating the standardised operational protocol and practitioner guidance for CF.

Programme performance 2016-17

In 2016-17, NBS programmes across the UK monitored performance against 12 national standards published in 2013 (these are outlined in detail in Appendix A).

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

Headline results

The most recently published national report (2016-17), which describes performance against national standards in each region of the UK[5], 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 2016-17:

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

- Northern Ireland was the best performing UK region in relation to timing of sample collection and processing, with 98.3 % of samples collected between 5-8 days of age, and 99.5% of samples received in the newborn screening laboratory within 4 working days of collection.

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

- 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 Northern Ireland exceeded acceptable national standards. Northern Ireland was also the only region of the UK to meet the achievable standard for timeliness of first appointment for CF screen positive babies with 2 mutations, with 100% of babies seen by 28 days.

- Over 23,000 babies had newborn blood spot screening testing. In total, across all of the five conditions tested for, 44 babies were identified as screen positive, and 32 of these babies were confirmed as having one of the conditions.

Areas for further improvement

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

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

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

At a national level, meeting the standard (acceptable = ≤2%; achievable = ≤0.5%) 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 2016-17 was 4.39% which did not meet the acceptable or achievable standard. The regional NBSP QI group continues to work to understand and reduce the avoidable repeat rate, including scoping potential variance across Northern Ireland. Within HSCTs, additional training was provided to midwifery staff in the use of a new incision device and understanding of the new laboratory quality acceptance criteria. A 'Once is enough' information leaflet that had previously been developed by the group, was recirculated as a refresher guide to all test takers within HSCTs. This concise publication provides guidance to support accurate first-time completion of blood spot sampling. Photographic evidence of what constitutes acceptable and unacceptable blood spot samples was also shared with test takers to provide a visual aid to help improve the quality of the first sample.

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

APPENDIX A: DATA SOURCES, CORE DEFINITIONS AND UK NEWBORN BLOOD SPOT SCREENING PROGRAMME STANDARDS, 2013

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 2016 and 31st March 2017 and is reported by the Trust that took the sample.

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 2016/17 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 2016-17.

Key definitions

'Born and Resident'

Babies who were born to Northern Ireland residents (at time of birth) in 2016-17 and were still resident in Northern Ireland at 31st March 2017.

'Movers in'

Babies who were born in 2016-17, who moved to a CHS area of Northern Ireland between 1st April 2016 and 31st March 2017, who were not born to residents of that CHS area and were still resident at 31st March 2017.

'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.[6,7]

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

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

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 2016-17 is based upon standards that were introduced across the UK in 2013. The 12 standards are summarised in Table 1:

Table 1: UK Newborn Blood Spot Screening Programme Standards, 2013[8]

Standard		Description	Acceptable	Achievable
1a	Completeness of coverage (CCG[9] responsibility at birth)	The proportion of *eligible babies for whom a conclusive screening result for each of the five conditions is recorded on the child health information system (CHS) by 17 days of age. **Eligible babies (denominator) are 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[9] was responsible at birth and remains responsible on the last day of the reporting period*.	≥ 95.0%	≥ 99.9% for PKU,MCADD and SCD ≥ 98% for CHT and CF
1b	Completeness of coverage (movers in)	The proportion of **eligible babies for whom a conclusive screening result for PKU is recorded on CHS by 21 calendar days of movement in being recorded on the CHS. ***Eligible babies (denominator) are 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[9] during the reporting period and for*	≥ 95%	≥ 99.9%

[8]UK Newborn Screening Programme Centre - Standards and guidelines for newborn blood spot screening, 2013. http://newbornbloodspot.screening.nhs.uk/standards

[9]CCG – 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)*	*whom the CCG[9] remains responsible on the last day of the reporting period.*		
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 for 5 conditions for babies aged 17-364 days	100% perform regular checks for 5 conditions for babies aged 14-364 days
3	Baby's NHS number (or UK equivalent) is included on the blood spot card	The NHS number (or UK equivalent) is a unique identifier that aids the identification and tracking of babies as they progress through the screening pathway.	100% of cards received by a laboratory include the baby's NHS number	95% of cards received by a laboratory have the baby's NHS number on a bar-coded label
4	Timely sample collection	The proportion of first samples taken between 5 and 8 days after birth	≥ 95%	≥ 99%
5	Timely receipt of a sample in the laboratory	Proportion of all samples received within 3 or 4 working days of sample collection	≥ 99% within 4 working days	≥ 99% within 3 working days

Standard		Description	Acceptable	Achievable
6	Quality of the blood spot sample	Proportion of avoidable repeat samples, of the total number of first blood spot samples received, requested by the laboratory because the previous sample was: • Taken when the baby was too young (on or before day 4) • Insufficient blood • Unsuitable sample/card	≤ 2%	≤ 0.5%
7	Timely taking of a repeat blood spot sample	The proportion of repeat samples taken as defined for individual tests: • An avoidable repeat sample must be taken within three calendar days of receipt of request • A 2nd sample for raised IRT[10] should be taken ideally on day 21 (between day 21 and 28) • A 2nd sample for borderline TSH[11] should be taken between seven and 10 days after the initial borderline sample • A 2nd sample for TSH should be taken from babies born at less than 32 weeks gestation (less than or equal to 31 weeks + six days) when they reach 28 days of age, or	≥ 95% of repeat samples taken as defined ≥ 95% of repeat samples taken as defined	≥ 99% of repeat samples taken as defined ≥ 99% of repeat samples taken as defined

[10] Immunoreactive trypsinogen
[11] Thyroxine stimulating hormone

Standard		Description	Acceptable	Achievable
7	Timely taking of a repeat blood spot sample (*cont'd)*	day of discharge home whichever is the sooner		
8	Clinical Pathology Accreditation (screening)	CPA 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 CPA looks at both the ISO standards and the UK screening specific laboratory standards, as an integrated process	Laboratory is CPA accredited with specialist assessment of NBS screening by the next full visit	
9	Timely processing of all PKU, CHT and MCADD screen positive samples	The proportion of positive screening results available and clinical referral initiated within 3 or 4 working days of sample receipt by screening laboratory.	100% within 4 working days	100% within 3 working days
10	Clinical Pathology Accreditation (diagnosis)	CPA accredits pathology laboratories against a set of defined standards which are allied to international standards for competence in medical laboratories – ISO 15189	Laboratory is CPA accredited	
11	Timely receipt into clinical care	A baby in whom PKU, CHT (on first sample) or MCADD is suspected should attend their first clinical appointment by:	100% by 17 days of age	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 24 days of age	100% by 21 days of age

Standard		Description	Acceptable	Achievable
11	Timely receipt into clinical care *(cont'd)*	A baby in whom CF is suspected (2 CFTR mutations detected) 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 be referred and registered with designated healthcare professional by:	90% by 8 weeks of age	95% by 8 weeks of age
		A baby in whom SCD is suspected should attend local clinic by 3 months of age:	90%	95%
		SCD screen positive babies should be offered and prescribed penicillin V (or alternative) by 3 months of age:	90%	99%
12	Timeliness of results to parents	Proportion of screen negative results letters despatched direct to parents from the child health records department by six weeks of age	100%	

Application of 2013 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 Health and Care Number (equivalent to NHS number) on blood spot cards is currently not mandatory in Northern Ireland. There are plans to make the use of Health and Care Number on blood spot cards compulsory in the future.

Standard 7

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

Standard 8 and 10

Compliance with Standards 8 and 10 is monitored as part of the UKAS accreditation (previously known as Clinical Pathology Accreditation (CPA) of the Regional Newborn Screening Laboratory. The Northern Ireland Regional Newborn Screening Laboratory has achieved this accreditation.

Standard 12

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

APPENDIX B: PERFORMANCE OF NBSP IN NI 2016-17

The performance of the NBSP in Northern Ireland 2016-17 against each of the UK standards[1] is outlined in Table 2.

Table 2: Performance of the Northern Ireland Newborn Blood Spot Screening Programme 2016-17 *(data collected by NI Child Health System)*

Standard		Performance 2016-17			Acceptable	Achievable
1a	Total number of 'born and resident' babies = 23,584				≥ 95.0%	≥ 99.9% for PKU,MCADD and SCD ≥ 98% for CHT and CF
	Completeness of coverage (CCG[12] responsibility at birth) by Day 17 (Numbers and %)	PKU[13] CHT CF SCD	23,315 23,142 23,233 23,323	98.86% 98.13% 98.51% 98.89%		
	Declines to screening (Numbers and %)	PKU[10] CHT CF SCD	16 16 16 16	0.07% 0.07% 0.07% 0.07%		
1b	Total number of 'movers in' babies = 400				≥ 95%	≥ 99.9%
	Completeness of coverage (movers in)	PKU[10] CHT CF SCD	311 310 267 310	77.75% 77.50% 66.75% 77.50%		
	Declines to screening (Numbers and %)	PKU[10] CHT CF SCD	79 80 80 80	19.75% 20.00% 20.00% 20.00%		
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 5 conditions for babies aged 11-364 days		100%	100% perform regular checks for 5 conditions for babies aged 17-364 days	100% perform regular checks for 5 conditions for babies aged 14-364 days

[12]CCG – Clinical Commissioning Group (Northern Ireland data are provided by Child Health System (CHS) area)
[13]PKU data is reported as proxy to MCADD data in line with UK reporting.

Standard		Performance 2016-17		Acceptable	Achievable
3	Baby's NHS number (or UK equivalent) is included on the blood spot card	This is currently not mandatory in NI Total number of samples = 27,007	79.11% (21,366)	100% of cards received by a laboratory include the baby's NHS number	95% of cards received by a laboratory have the baby's NHS number on a bar-coded label
4	Timely sample collection	The proportion of first samples taken between 5 and 8 days after birth Total number of samples taken = 24,171[14]	98.27% (23,752)	≥ 95%	≥ 99%
5	Timely receipt of a sample in the laboratory	% of ALL samples received by the lab in 3 or fewer working days of sample being taken % of ALL samples received by the lab in 4 or fewer working days of sample being taken Total number of all samples = 26,807[15]	98.52% (26,410) 99.45% (26,660)	≥ 99% within 4 working days	≥ 99% within 3 working days

[14] 168 samples had no date of sample recorded and therefore were not included

[15] 200 samples had no date of sample recorded and therefore were not included

Standard		Performance 2016-17		Acceptable	Achievable
6	Quality of the blood spot sample (avoidable repeat rate)	Total number of first samples = 24,339 Total number of avoidable repeats = 1068	4.39%	≤ 2%	≤ 0.5%
9	Timely processing of all PKU, CHT and MCADD screen positive samples	PKU 4/4 MCADD 2/2 CHT 20/20	100% within 3 working days for each condition	100% within 4 working days	100% within 3 working days
11	Timely receipt into clinical care	Age range at first appointment (in days) PKU 4/4 MCADD 2/2 CHT* 9/9 *suspected on first sample	100% by 14 days of age	100% by 17 days of age	100% by 14 days of age
		CHT** 10/10 ***suspected on a repeat sample*	100% by 21 days of age	100% by 24 days of age	100% by 21 days of age
		CF 9/9 (*2 mutations*)	100% by 28 days of age	95% by 28 days of age	100% by 28 days of age
		CF 8/9 (*1 or 0 mutations*)	88.89% by 35 days of age	80% by 35 days of age	100% by 35 days of age

Standard		Performance 2016-17		Acceptable	Achievable
11	Timely receipt into clinical care (*cont'd*)	SCD Age when baby was referred and registered with designated healthcare professional[16]	N/A	90% by 8 weeks of age	95% by 8 weeks of age
		Baby in whom SCD is suspected should attend local clinic by 3 months of age	N/A	90% by 3 months of age	95% by 3 months of age

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 3 shows consistently high performance in relation to coverage in babies 'born and resident' in NI. Similar to 2015-16 and 2014-15, during 2016-17 the programme exceeded the acceptable standard for completeness of coverage, with more than 98% of 'born and resident' babies with conclusive results for all conditions recorded on CHS by 17 days of age and 99.9% of all born and resident babies having conclusive results by the end of the reporting period.

[16] See further details on page 38

Table 3 Coverage - Born and Resident (B&R) - Standard 1a[17] *(data collected by NI Child Health System)*

	Year		
	2016-17	**2015-16**	**2014-15**
Total number **born and resident** (**B&R**) at 31st March	23,584	23,858	23,820
Number of **B&R** with decline to screening (02)			
PKU[18]	16(0.07%)	16(0.07%)	8(0.03%)
CHT	16(0.07%)	16(0.07%)	8(0.03%)
CF	16(0.07%)	18(0.08%)	8(0.03%)
SCD	16(0.07%)	16(0.07%)	10(0.04%)
Number (%) of **B&R** with conclusive results by the end of the reporting period (codes 04, 05, 06, 07, 08, 10)			
PKU[17]	23,568 99.93%	23,840 99.92%	23,812 99.97%
CHT	23,565 99.92%	23,840 99.92%	23,812 99.97%
CF	23,563 99.91%	23,838 99.92%	23,810 99.96%
SCD	23,568 99.93%	23,840 99.92%	23,810 99.96%
Number (%) of **B&R** with conclusive results available by Day 17 (by 17 days of age) codes 04, 05, 06, 07, 08,10)			
PKU[17]	23,315 98.86%	23,644 99.10%	23,626 99.19%
CHT	23,142 98.13%	23,448 98.28%	23,406 98.26%
CF	23,233 98.51%	23,620 99.00%	23,616 99.14%
SCD	23,323 98.89%	23,642 99.09%	23,628 99.19%

The NBSP in NI is currently unable to report on the completeness of coverage for 'mover in' babies by 21 calendar days of 'movement in' being recorded on the CHS (Standard 1b); this would require software development. However, by the end of the reporting period, compared with 2015-16, there was a slight decrease

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

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

in the proportion of ‘mover in’ babies with a conclusive result for PKU (77.8% compared to 81.5% in 2015-16).

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 2016-17 there were 45 ‘mover in’ babies who had no conclusive result for CF, as they were over 8 weeks old and therefore too old (code 09/4) for testing for CF at this point. This is similar to 2015-16 and 2014-15 when 44 and 43 babies (respectively) were over 8 weeks old.

There were 4 ‘mover in’ babies, resident at the end of the reporting period, with outstanding screening results for a variety of reasons including moving out of the area prior to offer of screening test, prior to test being carried out or not completing screening by the age of 1 year.

Table 4 – Completeness of coverage – Movers in - Standard 1b[19] *(data collected by NI Child Health System)*

	Year		
	2016-17	**2015-16**	**2014-15**
Total number 'Movers In' (**MI**) resident at 31st March	400	356	343
Number of **MI** with decline to screening (02)			
PKU[20]	79 (19.75%)	61 (17.13%)	51 (14.87%)
CHT	80 (20.00%)	61 (17.13%)	53 (15.45%)
CF	80 (20.00%)	58 (16.29%)	50 (14.58%)
SCD	80 (20.00%)	62 (17.42%)	53 (15.45%)
Number (%) of **MI** with reason for not starting/incomplete (code 09-)			
PKU[19]	2 (0.50%)	0 (0)	0 (0)
CHT	2 (0.50%)	0 (0)	0 (0)
CF	45 (11.25%)	44 (12.36%)	43 (12.54%)
SCD	2 (0.50%)	0 (0)	0 (0)
Number (%) of **MI** with conclusive results(codes 04, 05, 06, 07, 08,10)			
PKU[19]	311 77.75%	290 81.46%	291 84.84%
CHT	310 77.50%	290 81.46%	289 84.26%
CF	267 66.75%	250 70.22%	249 72.59%
SCD	310 77.50%	290 81.46%	289 84.26%

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 18 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.

[19] excludes data on some inconclusive results (codes 01 = received and not tested, 03 = repeat/further sample required

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

Sample identification (Standard 3)

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

In 2016-17, just under 80% of all samples received by the Laboratory had the HCN included[21]. It is recognised that including the unique HCN is an important additional safety and quality mechanism for identifying and matching baby records in the NBSP; work to improve recording on blood spot cards continued during 2016-17.

Timely sample collection and processing (Standards 4 and 5)

The NBSP exceeded the acceptable standard for timely sample collection, with more than 98% of first samples taken between 5 to 8 days after birth; this performance is similar to 2015-16 and 2014-15.

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

	Year		
	2016-17	**2015-16**	**2014-15**
Total number of first samples	24,171	24,430	24,401
Number of samples not included in audit (because DOB or the date that sample was taken was not recorded on the cards)	168	145	156
Number of first samples taken on or before day 4 (%)	111 (0.46%)	108 (0.44%)	105 (0.43%)

[21] Data source: Regional Newborn Screening Laboratory

	Year		
	2016-17	**2015-16**	**2014-15**
Number of first samples taken on day 5 (%)	22,818 (94.40%)	23,066 (94.42%)	23,038 (94.41%)
Number of first samples taken on or after day 9 (%)	308 (1.27%)	273 (1.12%)	269 (1.10%)
Number of first samples taken between day 5 and day 8 (%)	23,752 (98.27%)	24,049 (98.44%)	24,027 (98.47%)

In 2016-17 the programme also exceeded the acceptable standard for timely receipt of a sample in the laboratory (≥ 99.0% of all samples received in the laboratory **within 4 working days**).

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

	Year		
	2016-17	**2015-16**	**2014-15**
Total number of ALL samples (first, repeat and second samples) included	26,807	26,517	26,536
Number of samples EXCLUDED (because date of specimen was not recorded on the cards)	200	182	205
Number of ALL samples received by the lab in 3 or fewer working days of sample being taken (%)	26,410 (98.52%)	26,187 (98.76%)	26,196 (98.72%)
Number of ALL samples received by the lab in 4 or fewer working days of sample being taken (%)	26,660 (99.45%)	26,407 (99.59%)	26,391 (99.45%)

Sample quality (Standard 6)

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

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

The avoidable repeat rate in 2016-17 was 4.39% and therefore the NBSP did not meet the acceptable standard (avoidable repeat rate ≤2% - see Table 7). This was similar to performance in 2014-15 (4.38%) and comparable to other UK countries (Scotland 5.72% Wales 5.56%) except England (2.89%).[22]

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

	Year		
	2016-17	**2015-16**	**2014-15**
Total number of first samples received	24,339	24,575	24,557
Total number of repeat samples requested	1,378	1,302	1,396
REASON FOR REPEAT			
Avoidable Repeats			
Too young for reliable screening (≤ 4 days) (%)	105 (0.43%)	108 (0.44%)	100 (0.41%)
Insufficient sample	724 (2.97%)	460 (1.87%)	433 (1.76%)
Unsuitable sample[23]	239 (0.98%)	441 (1.79%)	543 (2.21%)
Total Avoidable Repeats	**1068 (4.39%)**	**1009 (4.11%)**	**1076 (4.38%)**

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

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

In 2016-17, the majority of avoidable repeats (2.97%) were due to insufficient sampling (Table 7). Potential contributory factors may have included the introduction of more stringent laboratory acceptance criteria following implementation of new UK standards for blood spot quality in February 2016. The introduction of a new incision device for sample collection in NI on 1st March 2017 may also have had an impact (see below). However, national trend data highlights that the issue of avoidable repeats has been a long term problem across all regions of the UK.

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

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

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

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

PKU and MCADD

In 2016-17, 4 babies (including 1 who was tested early due to a family history) were identified with positive screening tests for PKU, and 2 babies were identified as having positive screening tests for MCADD. As in previous years, all (100%) of these babies were referred within 3 working days of sample receipt in the

laboratory and seen by clinical teams by 17 days of age, therefore meeting both the acceptable and achievable standards (by 14 days of age - see Tables 8a and b).

Outcomes

A diagnosis of PKU was confirmed in all 4 babies who were PKU screen positive and a diagnosis of MCADD was confirmed in 1 of the 2 babies who were MCADD screen positive.

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

	Year		
	2016-17	**2015-16**	**2014-15**
Number of babies with screen positive result (status code = 07 - not suspected other disorder follow up or 08 - suspected)	4	14	5
Number of screen positive babies with clinical referral initiated within 3 working days of sample receipt in lab (%)	4 (100%)	14 (100%)	5 (100%)
Number of screen positive babies who were seen by 17 days of age (%)	4 (100%)	14 (100%)	5 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	4 (100%)	12 (85.7%)	5 (100%)

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

	Year		
	2016-17	**2015-16**	**2014-15**
Number of babies with screen positive result (status code = 08 - suspected)	2	2	3
Number of screen positive babies with clinical referral initiated within three working days of sample receipt in lab (%)	2 (100%)	2 (100%)	3 (100%)
Number of screen positive babies who were seen by 17 days of age (%)	2 (100%)	2 (100%)	3 (100%)
Number of screen positive babies who were seen by 14 days of age (%)	2 (100%)	2 (100%)	3 (100%)

CHT

In 2016-17, 9 babies were identified as CHT screen positive on their first blood spot sample; all 9 (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, therefore meeting the achievable standard.

A further 11 babies were also identified as screen positive. Ten of these babies were positive on a repeat sample following a borderline result (Table 8c). As in previous years, all 11 (100%) were referred within 3 days of sample receipt in the laboratory and 10 of the 11 (90.91%) babies were seen by the clinical team by 24 days of age.

<u>Outcomes</u>

A diagnosis of CHT was confirmed in 17 of the 20 (85%) 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					
	2016-17		**2015-16**		**2014-15**	
	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)	9	10	11	7	12	6
Number of screen positive babies with clinical referral initiated within 3 working days of sample receipt in lab (%)	9 (100%)	10 (100%)	11 (100%)	7 (100%)	12 (100%)	6 (100%)
Number of screen positive babies who were seen by 17 days of age (suspected on first sample)	9 (100%)	N/A	11 (100%)	N/A	12 (100%)	N/A
Number of screen positive babies who were seen by 24 days of age (suspected on a repeat blood spot sample following a borderline result)	N/A	10 (100%)	N/A	7 (100%)	N/A	5 (83%)

SCD

In 2016-17, no babies were identified as screen positive for SCD; however, the screening results for 43 babies required further testing/assessment: 28 were identified as sickle cell 'carriers' and 15 as a carrier of another unusual haemoglobin gene. 97.7% (42/43) of these babies were referred and registered with a designated healthcare professional by 8 weeks of age and 93.0% (40/43) were seen by 3 months of age.

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

Condition/Disorder	Year		
	2016-17	**2015-16**	**2014-15**
Sickle Cell Disease or Other Potentially Clinically Significant Condition[24]	0	4	7
Condition/Disorder	**Year**		
	2016-17	**2015-16**	**2014-15**
Sickle Cell Carrier or Carrier of Other Unusual Haemoglobin Gene[25] or Possible Benign Disorder[26]	43	49	38

CF

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

A further 9 babies were identified as suspected CF with 1 or 0 mutations and an IRT ≥ cut-off 2 on screening. Eight out of 9 babies (88.9%) were seen by the clinical team by 35 days of age. CF was confirmed in 2 of these children.

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

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

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

Outcomes

In 2016-17, a total of 18 babies were referred into the clinical service for further testing following an initial positive screen for suspected CF. A diagnosis of CF was confirmed in 10 of these babies. In addition, 18 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		
	2016-17	**2015-16**	**2014-15**
No. of CF suspected babies with 2 mutations	9	9	9
Number of screen positive babies who were seen by 28 days of age (%)	9 (100%)	9 (100%)	8 (89%)

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

	Year		
	2016-17	**2015-16**	**2014-15**
No. of CF suspected babies with 1 or 0 mutations and IRT ≥ cut-off 2	9	5	5
Number of screen positive babies who were seen by 35 days of age	8 (89%)	5 (100%)	4 (80%)

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

	Year		
	2016-17	**2015-16**	**2014-15**
No. of babies with 1 CF mutation detected and second IRT < cut-off 2	18	6	13

ACKNOWLEDGEMENTS

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)

Data analysed and report written by:

Dr Rachel Doherty, Leanne McMullan and Nicola Cunningham (Public Health Agency)

HSC 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:

06/19