Public Health England

NHS

Newborn Blood Spot Screening Programme in the UK

Data collection and performance analysis report 2016 to 2017

Public Health England leads the NHS Screening Programmes

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About PHE Screening

Screening identifies apparently healthy people who may be at increased risk of a disease or condition, enabling earlier treatment or better informed decisions. National population screening programmes are implemented in the NHS on the advice of the UK National Screening Committee (UK NSC), which makes independent, evidence-based recommendations to ministers in the 4 UK countries. The Screening Quality Assurance Service ensures programmes are safe and effective by checking that national standards are met. PHE leads the NHS Screening Programmes and hosts the UK NSC secretariat.

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Executive summary

This report presents annual screening data for the NHS Newborn Blood Spot (NBS) screening programme for the financial year 1 April 2016 to 31 March 2017. The aim of the report is to feedback performance against the national standards.

Coverage of newborn screening continues to improve and more Clinical Commissioning Groups (CCGs) were at or above the acceptable level (greater than or equal to 95%) for key performance indicator (KPI) NB1 (Standard 1a). This year we also report data on coverage for movers in through KPI NB4 (Standard 1b).

Data were returned by Child Health Records Departments (CHRDs) for all 209 (100%) CCGs in England, in 2016 to 2017 for coverage measured at 17 days (CCG responsibility at birth) and was reported to be 96.5%. This is an increase from2015 to 2016 when manually submitted CHRD returns (from 191 CCGs out of 211 in total) revealed coverage at 17 days reach 94.5%. For movers in, Standard 1b introduces an effective timeframe of 21 calendar days. Data were returned for 207 (99%) out of 209 CCGs, reporting coverage at 87.1% for 2016 to 2017 in England, compared with 78.3% in 2015 to 2016.

Sub-regions are a sub-set of regions, allowing us to report data at a more granular level. Coverage (CCG responsibility at birth) was at or above the acceptable level of 95% for all sub-regions within England except the South West. No region within England attained the achievable level of 99%. Coverage for movers in showed marked improvement in the proportion of babies tested within 21 days of movement in when compared to 2015 to 2016, however all regions in England continued to perform below the acceptable threshold of 95%.

In England, 99.8% of blood spot cards included the baby's NHS number, and 73.9% included the NHS number on a bar-coded label. Although use of bar-coded labels continues to increase, no region is yet meeting the standard (achievable level at 95%) despite the investment made in funding trusts to purchase printers and scanners.

In the UK, 97.0% of samples were taken on days 5 to 8, an increase on 2015 to 2016 at 95.7%. Year-on-year data on timeliness of sample receipt shows no clear trends, but sample transport remains one of the biggest risks for delayed identification of screen positive babies.

Newborn screening laboratories in England and Wales adopted consensus quality guidelines in April 2015. After an initial anticipated increase, quarterly (KPI) collection of avoidable repeat data show avoidable repeat rates are decreasing as sample quality improves. Whilst the number of avoidable repeats due to insufficient samples has been

decreasing, it still remains the largest contributor of avoidable repeats for most laboratories.

Laboratory accreditation (standards 8 and 10) will be published by the United Kingdom Accreditation Service (UKAS).

The acceptable standard for timeliness of first appointment for CF screen positive babies with 2 mutations, when reported, was met in Northern Ireland, whilst for England and Scotland, it accounted for 85% and 92% of babies respectively. For Wales, three of four babies (75%) for whom age at first appointment was reported, met the standard. The acceptable standard for one or no mutations was met in Wales, whilst for England, Northern Ireland and Scotland, the rates were 74%, 89% and 55% respectively. These data are based on babies with age at first appointment reported.

Northern Ireland and Wales met the acceptable standard for CHT screen positive babies detected on first or second sample. These data are based on babies with age at first appointment reported.

Acknowledgements

The NHS Newborn Blood Spot (NBS) screening programme would like to thank all those who provided data to the annual collection in particular the UK newborn screening laboratories and child health records departments (CHRDs) that submitted the 2016 to 2017 data.

Introduction

Background

This report presents screening data and performance analysis for the UK's Newborn blood spot (NBS) screening programmes for the financial year 1 April 2016 to 31 March 2017. The UK National Screening Committee (UK NSC) recommends that all babies in the UK are offered NBS screening for sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT) and 6 inherited metabolic diseases (IMDs): phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (pyridoxine unresponsive) (HCU). The overall goal is to prevent ill health, disability and death through early diagnosis and effective intervention.

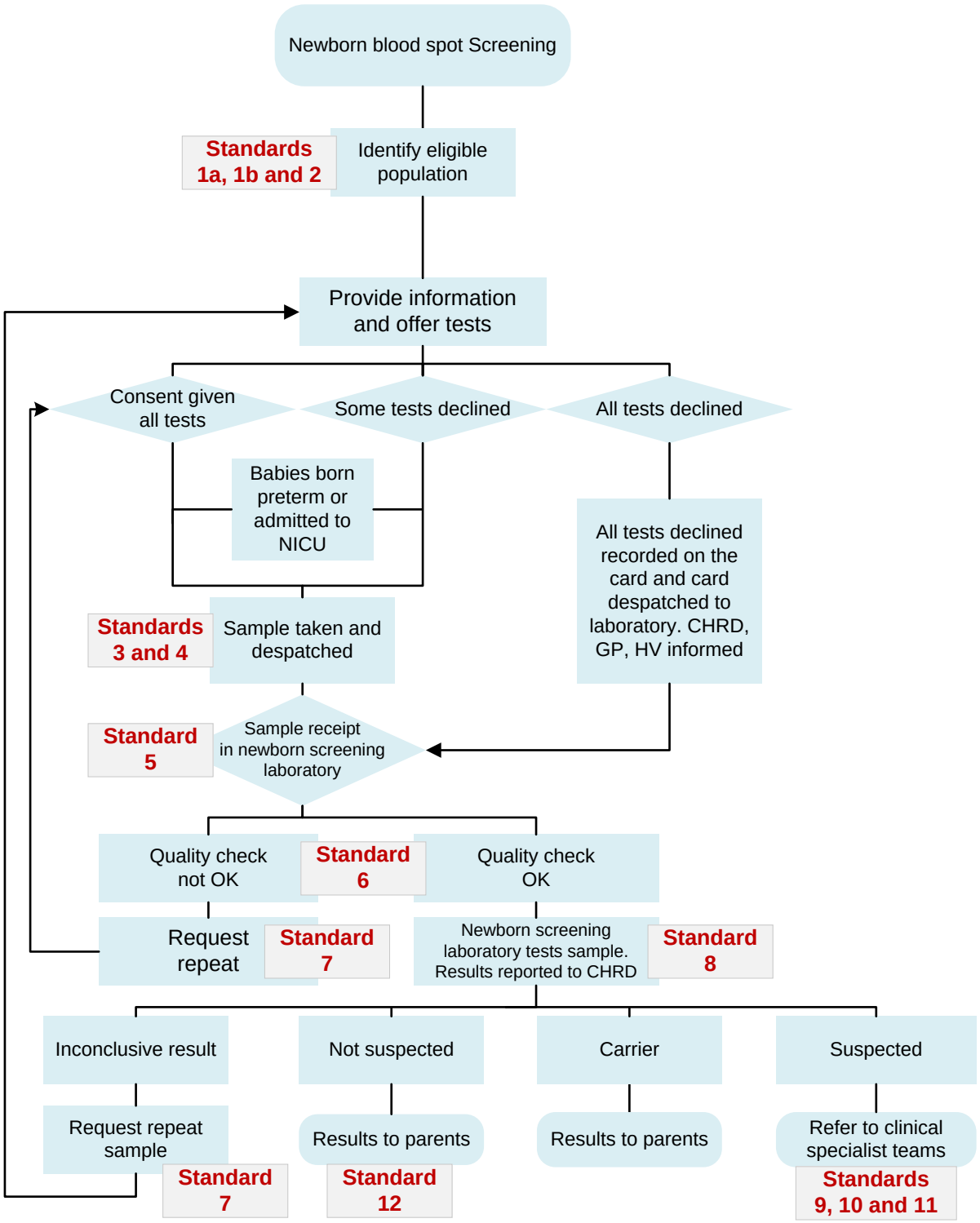
One of the objectives of the NHS NBS screening programme is to set national standards (see Table 1 and Figure 1)[1-2]. National standards are important to support the delivery and quality assurance of the screening programme and are used by local commissioners and quality improvement groups. The aim of this report is to feedback performance against the national standards. Providers, commissioners and the Screening Quality Assurance Service (SQAS) are encouraged to review this report to identify areas for improvement locally.

Table 1: NBS standards (2013)

Standard	Reporting responsibility
Standard 1a: Completeness of coverage (CCG responsibility at birth). Collected as KPI NB1	CHRD
Standard 1b: Completeness of coverage (movers in). Collected as KPI NB4	CHRD
Standard 2: Timely identification of babies with a null or incomplete result recorded on the child health information system	CHRD
Standard 3: Baby's NHS number (or UK equivalent) is included on the blood spot card	Newborn screening laboratory
Standard 4: Timely sample collection	Newborn screening laboratory
Standard 5: Timely receipt of a sample in the newborn screening laboratory	Newborn screening laboratory
Standard 6: Quality of the blood spot sample	Newborn screening laboratory
Standard 7: Timely taking of a repeat blood spot sample	Not currently collected
Standard 8: CPA (screening)	Part of UKAS accreditation
Standard 9: Timely processing of all PKU, CHT and MCADD screen positive samples	Newborn screening laboratory
Standard 10: CPA (diagnosis)	Part of UKAS accreditation
Standard 11: Timely receipt into clinical care	Newborn screening laboratory
Standard 12: Timeliness of results to parents	CHRD

For more information on the NBS standards please see: **Error! Hyperlink reference not valid.** www.gov.uk/government/collections/newborn-blood-spot-screening-programme-standards-and-data.

Figure 1: NBS standards mapped to a generic screening pathway



Methodology

Data are collected using spreadsheet based templates; these templates are accessible from www.gov.uk/government/collections/newborn-blood-spot-screening-programme-standards-and-data. The spreadsheets must be downloaded, completed and returned to the NHS NBS screening programme by email. The data are checked on receipt and if required, the relevant laboratory is contacted for any clarifications that are required.

With the intention of improving clarity of definitions, completeness and accuracy of data, and to keep up to date with changes in the programme, the definitions, methods and tools are reviewed annually and amended if required. Data are presented by financial year (1 April to 31 March) unless stated otherwise. The year '2016 to 2017' for example, refers to the financial year 1 April 2016 to 31 March 2017.

Data on standards 1a and1b are collected as key performance indicators (KPIs); compiled from 4 quarterly returns. In the annual KPI data, providers are excluded where data has not been submitted for all 4 quarters in that year.

Data on standards 2 and 12 are returned by child health records departments (CHRDs) per clinical commissioning group (CCG) and presented by region or country (England) or returned and presented by country (Northern Ireland); please note that one CHRD is not always coterminous to a single CCG.

Data on standards 3, 4, 5 and 6 are returned by newborn screening laboratories per CHRD/CCG/Maternity unit (England), child health service (Northern Ireland), health board (Wales) or laboratory catchment area (Scotland), and presented by laboratory catchement area.

Data on standard 7 are not currently collected.

Data on standard 9 are returned by newborn screening laboratories and presented by condition.

Data on standard 11 (including diagnostic outcome data) are returned by newborn screening laboratories per individual baby (anonymous) and presented by country/condition *(SCD data for England is presented in the NHS Sickle Cell and Thalassaemia Screening Programme's annual report).*

Laboratory accreditation (standards 8 and 10) will be published by the United Kingdom Accreditation Service (UKAS).

Completeness of data

CHRD data

Data was returned by CHRDs for all 209 (100%) CCGs for coverage for standard 1a and for 207 (99%) out of 209 CCGs for coverage for movers in; standard 1b, in 2016 to 2017 in England. This data are collected as a KPI (NB1 and NB4) on a quarterly basis. Annual data are derived from the quarterly returns, but exclusions are made for any provider that did not provide data in one or more quarter in that year.

For standards 2 and 12 returns are reported at CCG level collated by CHRDs, who then submit these returns directly to the annual data collection. Approximately 75% (160) of CCGs in England returned data out of the total number (209) in existence; a percentage decrease of 17% in 2016 to 2017 compared with 2015 to 2016. Regional variations in performance were more pronounced than others in 2016 to 2017 with London and the South East observing noticeable falls in the number of returns received compared with numbers returned in 2015 to 2016.

Laboratory data

The programme received data from the 13 newborn screening laboratories in England, and from the laboratories in Northern Ireland, Scotland, and Wales (100% response rate). Data are compared for consistency and clarifications are sought if required. Not all laboratories were able to submit data for all fields that were requested, in particular, outcome data which clinicians should supply to the laboratory.

Newborn screening laboratories inform the designated paediatrician directly when a baby is suspected of having one of the conditions screened for and request diagnostic outcome data on each baby. The laboratories hold the information on screen positive babies within their catchment area. Laboratories can experience difficulties in collecting this data, and as a result information is not always complete. These gaps in the data mean that diagnostic outcomes of the NHS NBS screening programme cannot be evaluated fully.

For 2016 to 2017 data, the number of babies tested for PKU and MCADD will also have been tested for MSUD, IVA, GA1 and HCU as the test for these conditions cannot be declined individually as these are tested together.

Data returns were excluded where providers were unable to submit data so as to not bias reported rates which depend on aggregating these figures. Where exclusions were made, these are highlighted in appropriate footnotes below the relevant charts and tables.

Analysis of screening performance

Overview of UK national screening figures

SCD	
Babies screened*	**778,973**
Screen positive	**280**
Screen positive rate**	**3.59**

PKU	
Babies tested	**779,688**
Screen positive	**107**
Screen positive rate**	**1.37**

CF†	
Babies tested	**776,086**
Screen positive	**346**
Screen positive rate**	**4.46**

MCADD	
Babies tested	**779,688**
Screen positive	**80**
Screen positive rate**	**1.03**

CHT	
Babies tested	**779,501**
Screen positive	**664**
Screen positive rate**	**8.52**

MSUD, IVA, GA1, HCU††	
Babies tested	**668,668**
Screen positive	**31**
Screen positive rate	
MSUD & GA1	0.2
IVA	0.1
HCU	0.1

*For SCD babies screened includes those tested (normal+abnormal) and decline

**Screen positive rates per 10,000 babies for 2016 to 2017

†Fewer babies are screened for CF as screening is not undertaken in babies over 8 weeks of age for this condition.
††Fewer babies are screened for MSUD, IVA, GA1 and HCU as screening for these conditions has not been implemented in Scotland or Northern Ireland.

Coverage	
Percentage of babies with a conclusive result for PKU recorded on the CHIS by 17 days of age	**96.5%** (England) **98.9%** (Northern Ireland)

Number of babies tested and number of screen positive results

Table 2: Number of UK babies tested and number of screen positive results for SCD, CF and CHT 2016 to 2017

Laboratory	SCD		CF		CHT	
	Number tested	Number of screen positives	Number tested	Number of screen positives	Number tested	Number of screen positives
Bristol	40,981	*	40,870	24	40,870	38
Cambridge	27,673	*	27,429	13	27,601	15
GOSH	122,726	98	121,668	41	122,436	149
Leeds	43,035	18	42,404	15	42,404	31
Liverpool	27,640	*	28,715	16	28,715	41
Manchester	55,599	9	55,361	14	55,661	52
Newcastle	33,357	*	32,687	12	32,687	28
Oxford	28,542	9	29,093	10	29,093	16
Portsmouth	32,138	*	36,907	19	37,988	20
SE Thames	60,555	56	55,699	21	56,043	29
Sheffield	71,188	9	70,610	31	71,129	51
SW Thames	54,281	29	52,857	22	52,854	34
West Midlands	69,806	32	71,000	35	71,000	56
England	**667,521**	**274**	**665,300**	**273**	**668,481**	**560**
Northern Ireland	24,311	0	23,996	18	24,115	20
Scotland	54,365	*	54,313	34	54,313	32
Wales	32,776	*	32,477	18	32,592	52
UK total	**778,973**	**280**	**776,086**	**343**	**779,501**	**664**

Data source: Newborn screening laboratories

*Numbers are suppressed to mask small numbers less than 5. This explains why the total is sometimes greater than the sum of the individual numbers recorded in the table.

We would normally expect to see a lower number of babies tested for CF, than for the other conditions, as the screening test is not reliable, and therefore not undertaken, in babies over 8 weeks of age. This will apply to some movers in.

Note that a significant proportion of screen positive results will not be confirmed cases.

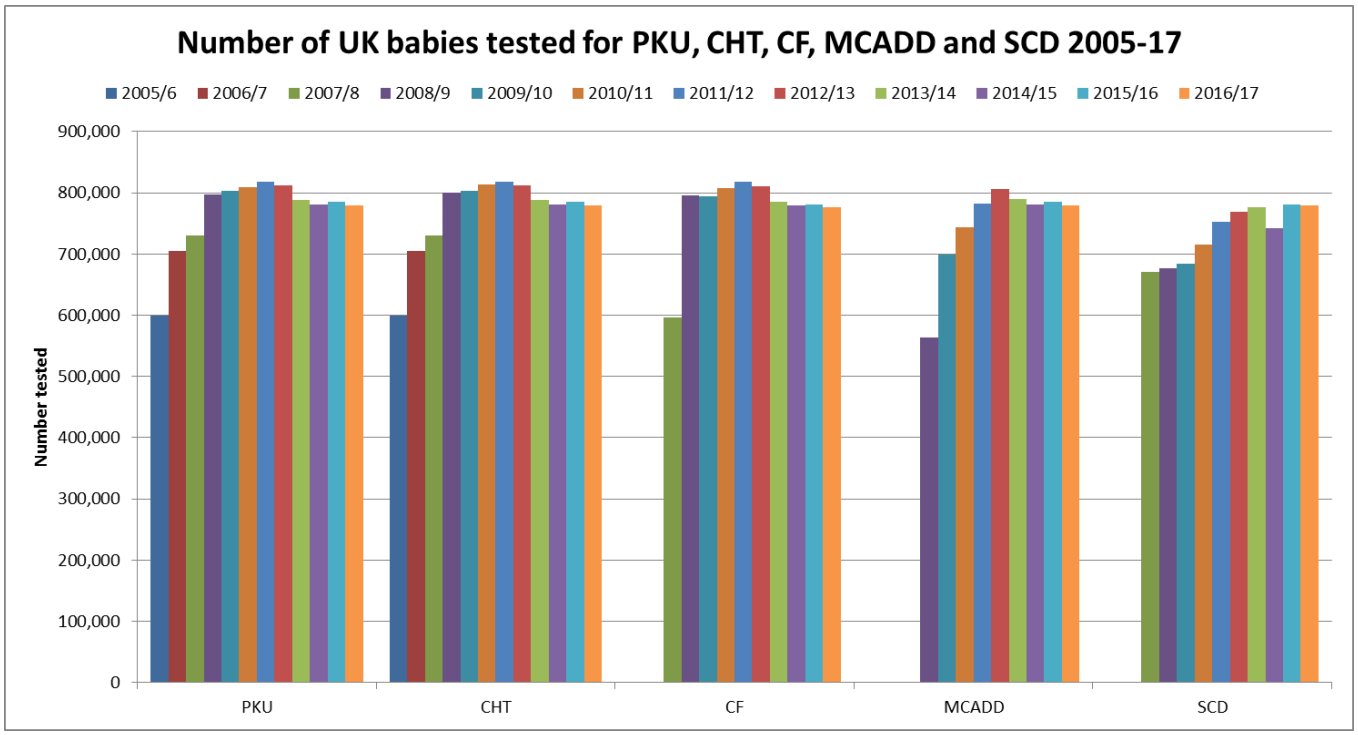
Table 3: Number of UK babies tested and number of screen positive results for IMDs 2016 to 2017

Laboratory	PKU		MCADD		MSUD, IVA, GA1, HCU	
	Number tested	Number of screen positives	Number tested	Number of screen positives	Number tested	Number of screen positives
Bristol	40,870	5	40,870	*	40,870	*
Cambridge	27,601	5	27,601	*	27,601	*
GOSH	122,436	18	122,436	8	122,436	8
Leeds	42,404	5	42,404	8	42,404	*
Liverpool	28,715	6	28,715	*	28,715	*
Manchester	55,661	9	55,661	5	55,661	*
Newcastle	32,687	7	32,687	*	32,687	*
Oxford	29,093	*	29,093	6	29,093	*
Portsmouth	38,177	*	38,177	5	38,177	0
SE Thames	56,043	7	56,043	*	56,043	0
Sheffield	71,129	14	71,129	14	71,129	7
SW Thames	52,852	*	52,852	*	52,852	*
West Midlands	71,000	4	71,000	8	71,000	*
England	**668,668**	**88**	**668,668**	**73**	**668,668**	**31**
Northern Ireland	24,115	*	24,115	*	0	0
Scotland	54,313	6	54,313	*	0	0
Wales	32,592	9	32,592	*	32,592	0
UK total	**779,688**	**107**	**779,688**	**80**	**701,260**	**31**

Data source: Newborn screening laboratories

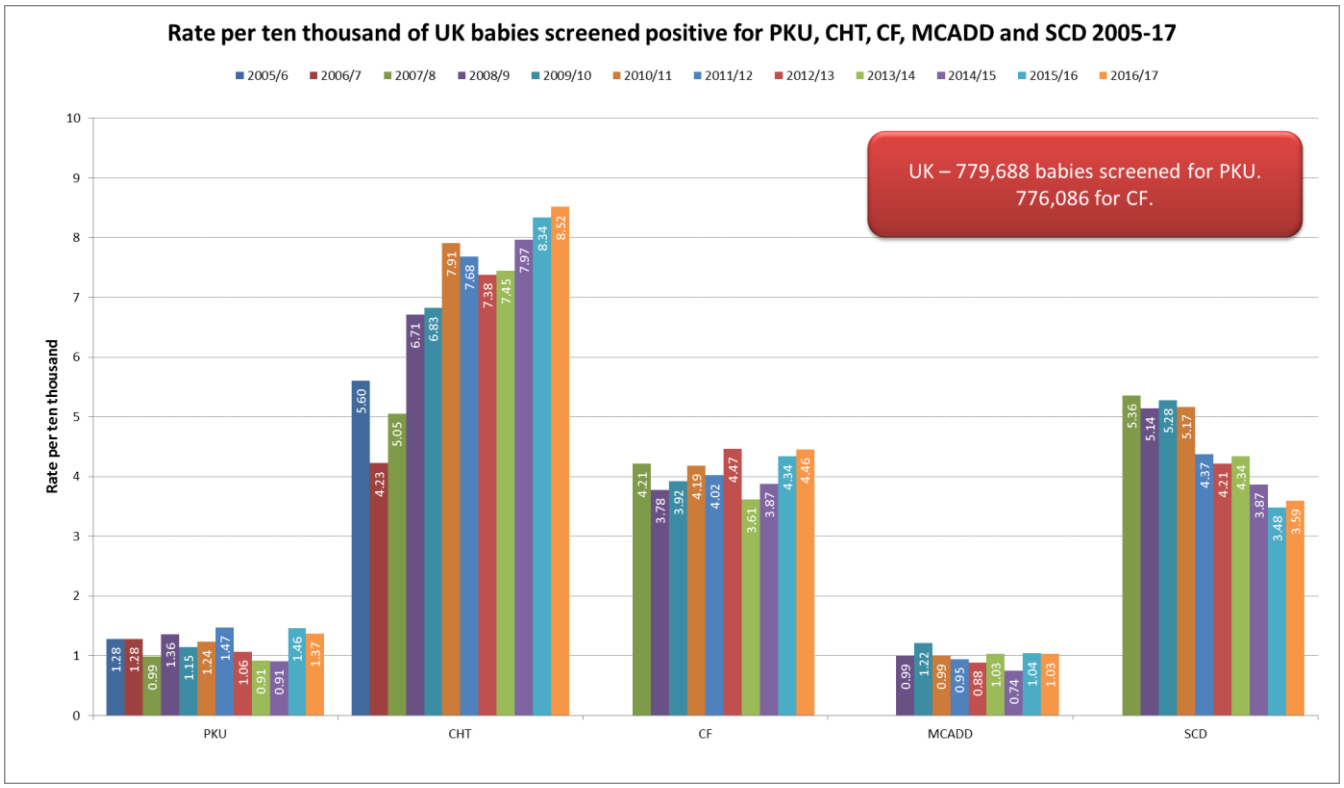
*Numbers are suppressed to mask small numbers less than 5. This explains why the total is sometimes greater than the sum of the individual numbers recorded in the table.

Figure 2: Number of UK babies tested for PKU, CHT, CF, MCADD and SCD 2005 to 2017



Data source: Newborn screening laboratories

Figure 3: Screen positive rate (per ten thousand) for babies screened for PKU, CHT, CF, MCADD and SCD 2005 to 2017



Data source: Newborn screening laboratories

Standard 1a: Completeness of coverage (CCG responsibility at birth)

Description

The proportion of babies registered within the CCG both at birth and on the last day of the reporting period who are eligible for NBS screening and have a conclusive result recorded on the Child Health Information System (CHIS) by 17 days of age.

Acceptable level: greater than or equal to 95.0% all tests
Achievable level: greater than or equal to 99.9% PKU, MCADD, SCD
Achievable level: greater than or equal to 98% CF, CHT

Newborn screening coverage data is collected as part of KPI NB1 on a quarterly basis. Performance against this KPI is calculated as the proportion of eligible babies for whom a conclusive screening result was available by 17 days. For this indicator, PKU is used as a proxy for all conditions screened for, through newborn blood spot screening. More information on KPI definitions can be found on gov.uk. Annual data are derived from the quarterly data submissions, but exclusions are made for any trust that did not provide data in one or more quarters in that year.

It should be noted that the coverage figures from KPI NB1 only include those born and registered in the sub-region and will not include movers in. Coverage of movers in presented in this report is collected separately as KPI NB4 which uses an effective timeframe of 21 calendar days from the CHRD being notified of movement in.

Table KPI NB-1. Coverage of newborn screening measured against PKU: CCG responsibility at birth (born and registered population), 2016 to 2017: England

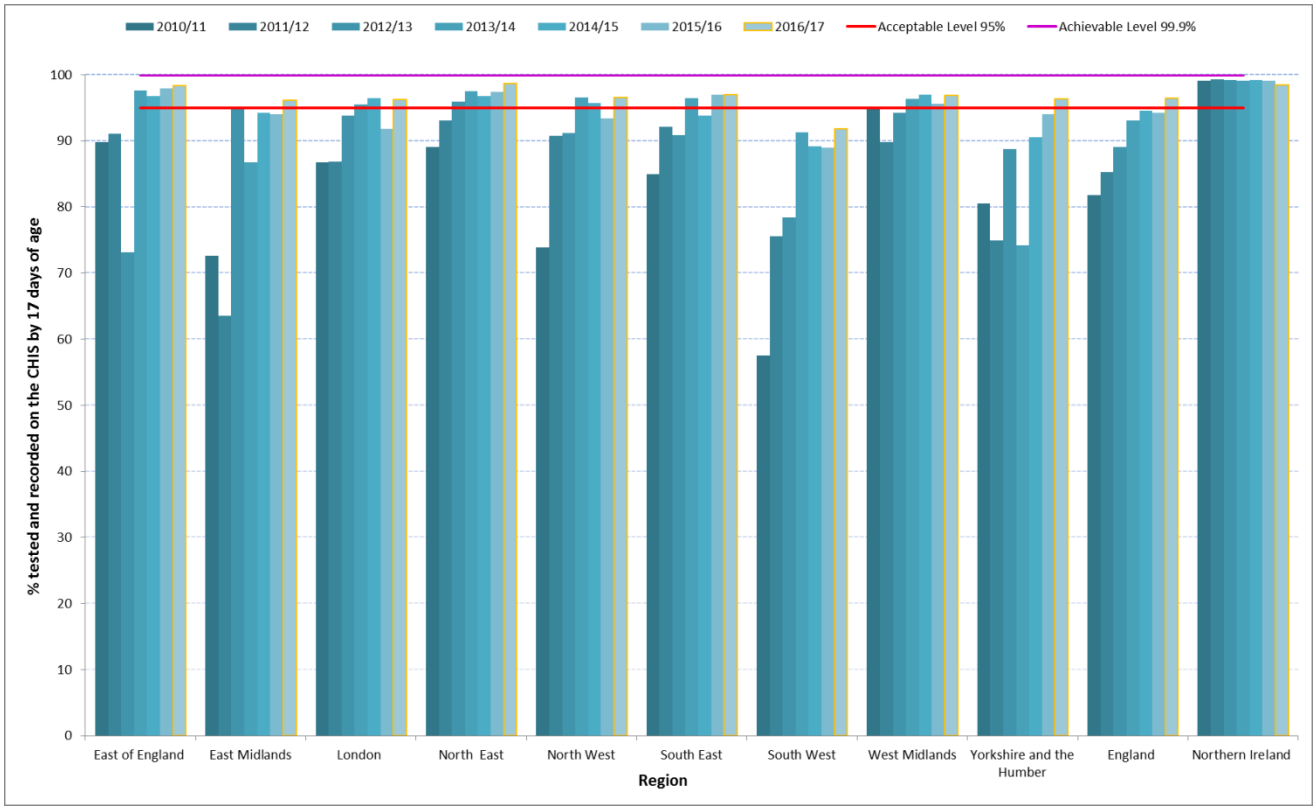
Sub-region	Completeness: Submitted all 4 quarters/ no.of providers	Tested babies	Eligible babies	Coverage (%)
East Midlands	20 / 20	47,188	49,084	96.1
East of England	19 / 19	63,840	64,946	98.3
London	32 / 32	109,784	114,069	96.2
North East	10 / 10	26,553	26,912	98.7
North West	33 / 33	76,515	79,202	96.6
South East	40 / 40	93,918	96,864	97.0
South West	11 / 11	44,432	48,384	91.8
West Midlands	22 / 22	65,258	67,331	96.9
Yorkshire & The Humber	22 / 22	57,207	59,368	96.4
England total	209 / 209	584,695	606,160	96.5

Data source: national quarterly [annual aggregate] KPI data collection

Maternity sites now use the Newborn Blood Spot Failsafe Solution (NBSFS) to ensure all babies born in England are offered screening. The percentage increase shown for England in 2016 to 2017 in the chart below is likely due, in large part, to NBSFS identifying babies missed at the day 5 screen. The responsibility for ensuring completeness of coverage continues to remain with the CHRD.

For Northern Ireland, coverage data for standard 1a is provided direct from a submission to the routine annual data collection rather than the quarterly KPI collection. Out of 23,584 eligible babies for whom the CCG was responsible at birth, 23,315 babies were tested for PKU (98.9%).

Figure 4: Coverage of newborn screening measured using PKU: CCG responsibility at birth (born and registered population measured at 17 days), 2016 to 2017



Data source: national quarterly [annual aggregate] KPI data collection

Standard 1b: Completeness of coverage (movers in)

Description
The proportion of babies who:

- are born within the reporting period, and
- change responsible CCG since birth or move in from abroad under a year of age and become the responsibility of the CCG during the reporting period, and
- for whom the CCG remains responsible on the last day of the reporting period, and
- are eligible for NBS screening and have a conclusive test result for PKU recorded on the CHIS equal to or less than 21 calendar days of movement in being recorded on the CHIS

Acceptable level: greater than or equal to 95% of eligible babies are tested for PKU

Achievable level: greater than or equal to 99.9% of eligible babies are tested for PKU

Table KPI NB-4. Coverage of newborn screening measured using PKU: movers in, 2016 to 2017: England

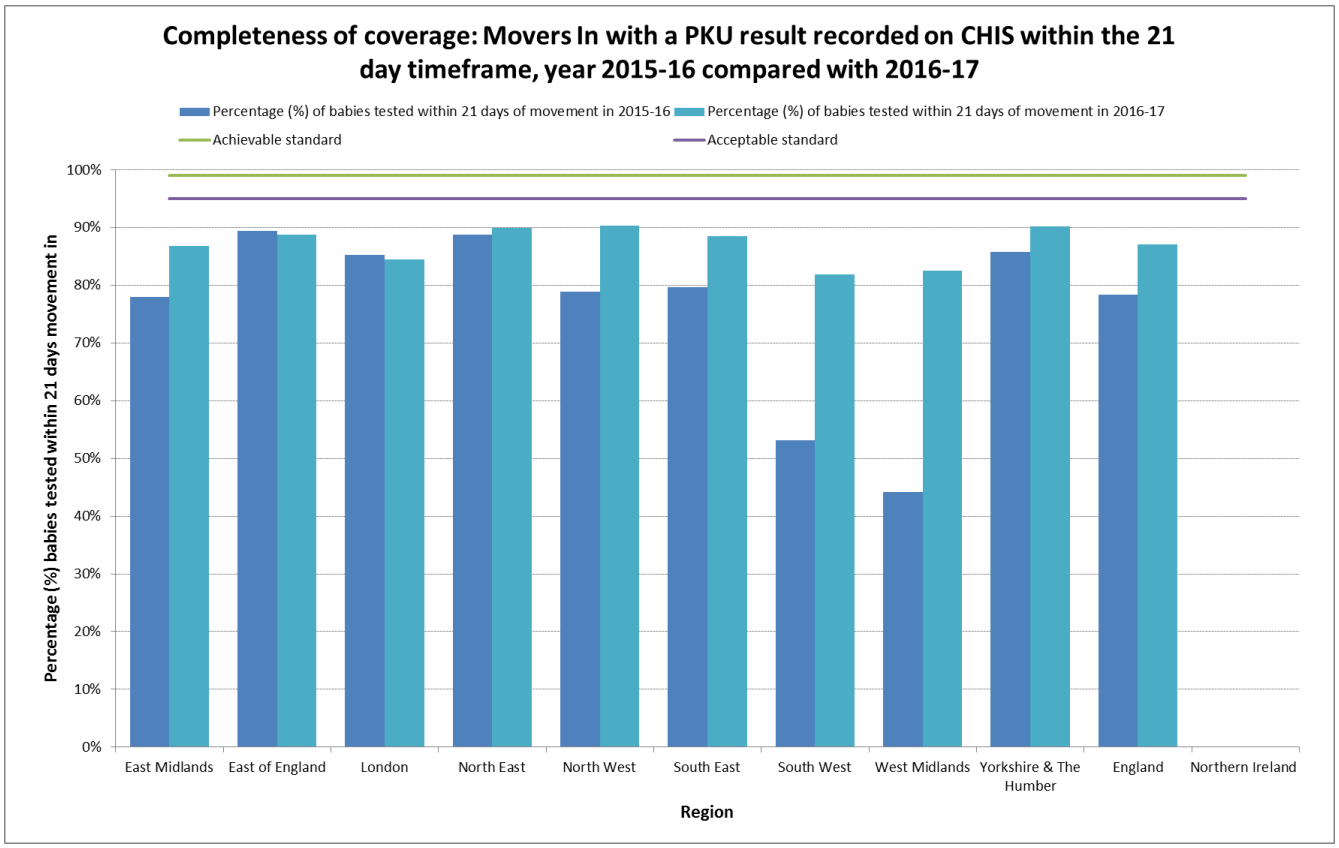
Sub-region	Completeness: Submitted all 4 quarters/ no.of providers	Tested babies	Eligible babies	Coverage (%)
East Midlands	20 / 20	1,620	1,866	86.8
East of England	19 / 19	5,280	5,948	88.8
London	30 / 32	4,740	5,608	84.5
North East	10 / 10	1,760	1,956	90.0
North West	33 / 33	4,060	4,493	90.4
South East	40 / 40	4,245	4,797	88.5
South West	11 / 11	1,900	2,322	81.8
West Midlands	22 / 22	3,598	4,360	82.5
Yorkshire & The Humber	22 / 22	3,155	3,495	90.3
England total	207 / 209	30,358	34,845	87.1

*2 providers excluded where data was not returned in all four quarters.

Data source: national quarterly [annual aggregate] KPI data collection

From 2010 to 2014, data was collected to measure coverage for movers in without applying an effective timeframe. Standard 1b introduces an effective timeframe of 21 calendar days; 2014 to 2017 data are presented with the timeframe in addition to year-on-year data without the timeframe.

Figure 5: Completeness of coverage for PKU (movers in) 2016 to 2017 within the 21 day timeframe



Data source: national quarterly [annual aggregate] KPI data collection

In England, processes for identifying and offering screening for movers in vary between regions. Northern Ireland is currently unable to report on the number of babies tested and recorded on CHIS within 21 days.

CHRD process data

Table 4: Receipt, recording and despatch of results by CHRDs 2016 to 2017 (reported per CCG)

Number of CHRDs† that:													
Region / Country	receive results by hard copy		receive results by email		receive results by electronic messaging		receive results with status codes		record results using status codes		send letters directly to parents when 04* is reported on all conditions		total number of CCGs**
	n	%	n	%	n	%	n	%	n	%	n	%	n
East Midlands	3	13.04	23	100.00	0	0.00	23	100.00	23	100.00	22	95.65	23
East of England	0	0.00	16	100.00	0	0.00	16	100.00	16	100.00	16	100.00	16
London	0	0.00	10	90.91	1	9.09	11	100.00	11	100.00	11	100.00	11
North East	0	0.00	11	100.00	0	0.00	11	100.00	11	100.00	7	63.64	11
North West	22	88.00	22	88.00	7	28.00	25	100.00	24	96.00	11	44.00	25
South East	10	55.56	16	88.89	10	55.56	18	100.00	18	100.00	15	83.33	18
South West	10	76.92	6	46.15	4	30.77	10	76.92	10	76.92	9	69.23	13
West Midlands	0	0.00	0	0.00	15	100.00	15	100.00	15	100.00	15	100.00	15
Yorkshire and Humber	8	30.77	12	46.15	19	73.08	25	96.15	25	96.15	26	100.00	26
England	53	33.54	116	73.42	56	35.44	154	97.47	153	96.84	132	83.54	158
Northern†† Ireland	0	0.00	0	0.00	0	0.00	1	100.00	1	100.00	1	100.00	1

Data source: CHRDs

*Status code 04; condition screened for not suspected

**For which a return was received. For some CCGs more than one return was received

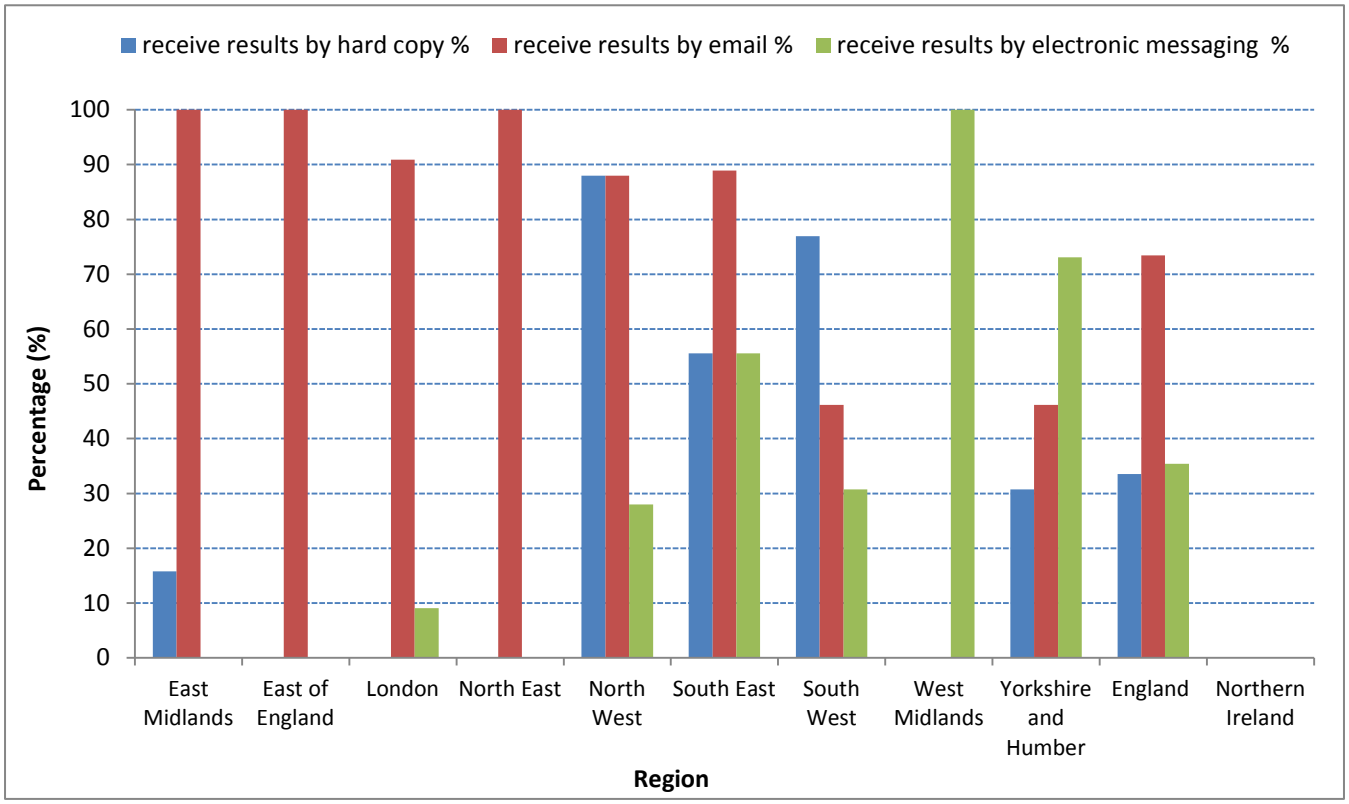
†Note some CHRDs might be double counted as data is returned per CCG and not CHRD

††Northern Ireland does not issue letters regarding results to parents. Results go to parents via the health visitor and a second set of results are inserted in the PCHR

The data highlights the multiplicity of methods used by CHRDs to receive results.

Full use of electronic messaging will enable greater efficiency.

Figure 6: Percentage of CHRDs who receive results by hard copy, email and electronic messaging 2016 to 2017



Data source: CHRDs

Region / Country	total number of CCGs 2016/17*		total number of CCGs 2015/16*
	n		n
East Midlands	23	↑	18
East of England	16	↓	19
London	11	↓	32
North East	11	↑	6
North West	25	↓	28
South East	18	↓	38
South West	13	↑	11
West Midlands	15	↓	16
Yorkshire and Humber	26	↑	23
England	158	↓	191
Northern Ireland	1		1

*For which there is a return. NB for some CCGs more than one return was received

The number of CCGs returning data decreased in some regions compared to the number of returns received in 2015 to 2016.

The most significant decreases can be seen in London with returns received down by over 65%; with regional percentage performance at 34% in 2016 to 2017.

In the South East the decrease is just over 50% in the number of returns received; with a regional percentage performance at 45% in 2016 to 2017.

*there were 2 returns in 2016 to 2017 with no sub-region indicated

Standard 2: Timely identification of babies with a null or incomplete result on the CHIS

Description

CHRDs perform regular checks for a null or incomplete result. If screening is found to be incomplete it is their responsibility to initiate follow-up arrangements to ensure parents are offered the screening test and babies are tested and have a conclusive result as soon as possible. CHRDs were asked if they performed daily checks for missing results at 17 days, 14 days or used a different search strategy.

Acceptable level

100% of CHRDs perform regular checks (ideally daily, minimum weekly) to identify babies with null values or status codes 01 specimen received in laboratory or 03 repeat/further sample required, for any of the 5 conditions, for all babies equal to or more than 17 days and equal to or less than 364 days.

Achievable level

100% of CHRDs perform regular checks (ideally daily, minimum weekly) to identify babies with null values or status codes 01 specimen received in laboratory or 03 repeat/further sample required, for any of the 5 conditions, for all babies equal to or more than 14 days and equal to or less than 364 days.

Table 5: Number and percentage of CHRDs that search for missing results at 17 days, 14 days and 'other' 2016 to 2017

Region/country	Number of CCGs†	Total number of CCG returns*	Number reaching the acceptable standard (100% at ≥17 days)**		Number reaching the achievable standard (100% at ≥14 days)**		Number reporting 'other' search strategy (non-compliant)	
	n	n	n	%	n	%	n	%
East Midlands	20	23	5	21.74	9	39.13	9	39.13
East of England	19	16	1	6.25	8	50.00	7	43.75
London	32	11	0	0.00	10	90.91	1	9.09
North East	10	11	0	0.00	8	72.73	3	27.27
North West	33	25	3	12.00	22	88.00	0	0
South East	40	18	2	11.11	6	33.33	10	55.56
South West	11	13	0	0.00	11	84.62	2	15.38
West Midlands	22	15	6	40.00	9	60.00	0	0
Yorkshire & The Humber	22	26	5	19.23	21	80.77	0	0
England	209	158	22	13.92	104	65.82	32	20.25
Northern Ireland	1	1	1	100.00	1	100.00	0	0

Data source: CHRDs

†Number of CCGs in existence as recorded in the KPI annual dataset 2016 to 2017

*For some CCGs more than one return was received.

**Where this information was available from submitted returns.

Standard 3: Baby's NHS number (or UK equivalent) is included on the blood spot card

Description

This standard is intended to ensure use of the baby's NHS number throughout the newborn screening process. The NHS number is a unique identifier that will aid the identification and tracking of babies as they progress through the screening pathway. Since April 2010 it has been mandatory for the NHS number to be used in England, ideally in a bar-coded label with an eye-readable NHS number.

Acceptable level

100% of blood spot cards received by a laboratory include the baby's NHS number.

Achievable level

95% of blood spot cards received by a laboratory have the baby's NHS number included on a bar-coded label.

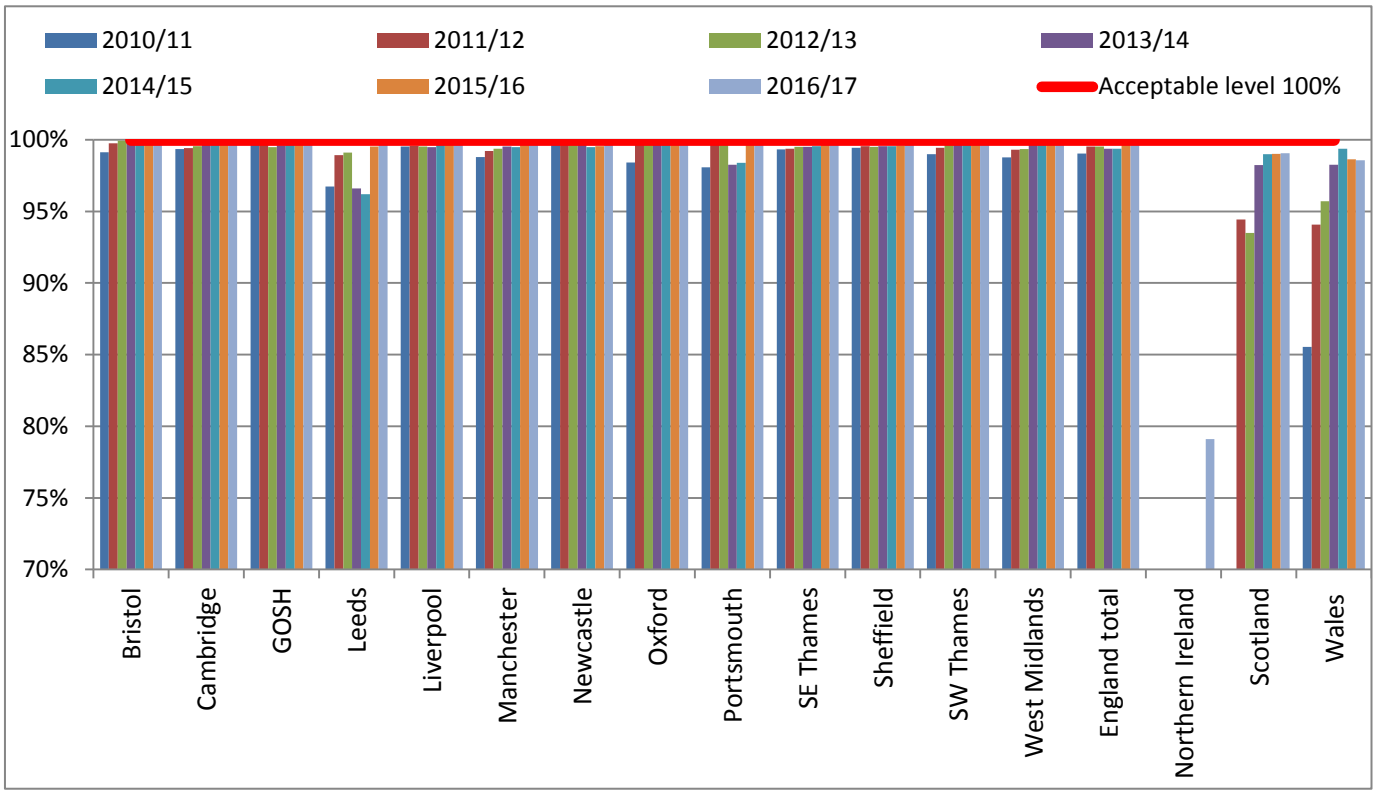
Table 6: Use of the baby's NHS number and bar-coded label 2016 to 2017

Laboratory	Number of all samples (including repeats)	Blood spot cards including baby's NHS number (or UK country equivalent)		Number of blood spot cards including ISB label bar-coded babies' NHS number (or UK country equivalent)	
	n	n	%	n	%
Bristol	40,743	40,611	99.7%	36,316	89.1%
Cambridge	28,693	28,632	99.8%	22,836	79.6%
GOSH	127,803	127,554	99.8%	56,599	44.3%
Leeds	44,926	44,773	99.7%	32,663	72.7%
Liverpool	30,325	30,293	99.9%	22,227	73.3%
Manchester	59,541	59,375	99.7%	47,578	79.9%
Newcastle	34,551	34,444	99.7%	22,897	66.3%
Oxford	30,684	30,618	99.8%	24,637	80.3%
Portsmouth	33,389	33,341	99.9%	25,366	76.0%
SE Thames	61,944	61,873	99.9%	51,050	82.4%
Sheffield	74,841	74,609	99.7%	63,438	84.8%
SW Thames	54,821	54,770	99.9%	45,542	83.1%
West Midlands	74,053	73,934	99.8%	63,045	85.1%
England total	696,314	694,827	99.8%	514,194	73.8%
Northern Ireland*	27,007	21,366	79.1%		0.0%
Scotland	57,457	56,913	99.1%	0	0.0%
Wales	35,388	34,885	98.6%	0	0.0%
UK total	816,166	807,991	99.0%	514,194	63.0%

Data source: Newborn screening laboratories

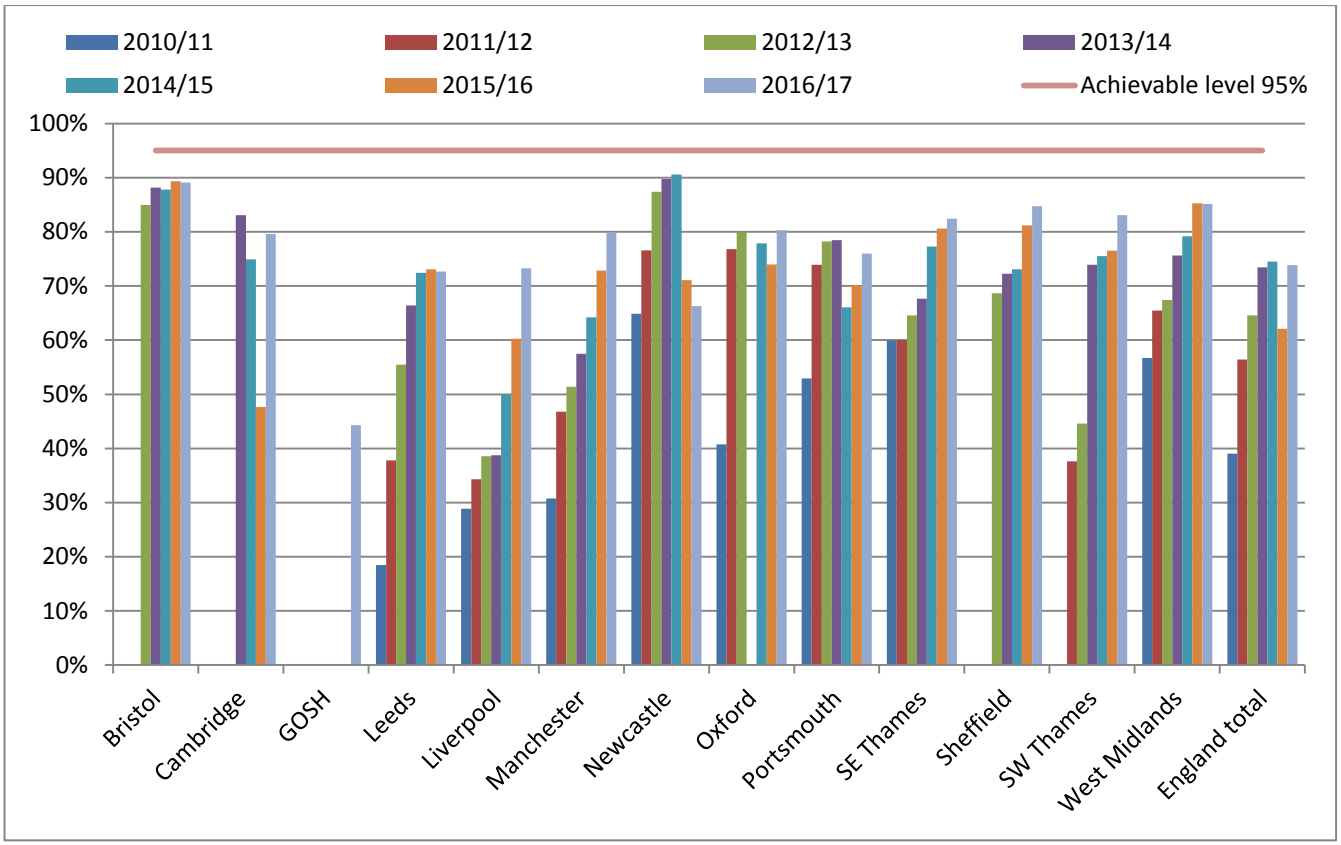
*Use of Health and Care number (equivalent to NHS number) in Northern Ireland is not mandatory

Figure 7: Percentage of blood spot cards including the baby's NHS number (or UK equivalent) 2010 to 2017



Data source: Newborn screening laboratories
Please note that the Y axis does not begin at zero
Use of Health and Care number (equivalent to NHS number) in Northern Ireland is not mandatory

Figure 8: Percentage of blood spot cards including a bar-coded NHS number (or UK equivalent) 2010 to 2017



Data source: Newborn screening laboratories
Great Ormond Street Hospital (GOSH) was unable to report data in previous years due to laboratory information management system limitations. GOSH reported data in 2016 to 2017

The data indicates that the investment made in funding trusts to purchase printers and scanners to produce bar-coded labels is not being fully realised.

Standard 4: Timely sample collection

Description

It is essential to take the blood spot sample promptly (ideally on day 5 and in exceptional circumstances between days 5 and 8) to give each screen positive baby the best possible chance of receiving early treatment. The health professional responsible for taking the blood sample should adhere to the guidelines for newborn blood spot sampling to ensure a valid sample is taken.

Acceptable level

Equal to or greater than 95% of first samples taken on days 5 to 8 (ideally on day 5).

Achievable level

Equal to or greater than 99% of first samples taken on days 5 to 8 (ideally on day 5).

Table 7: Day of first sample collection 2016 to 2017

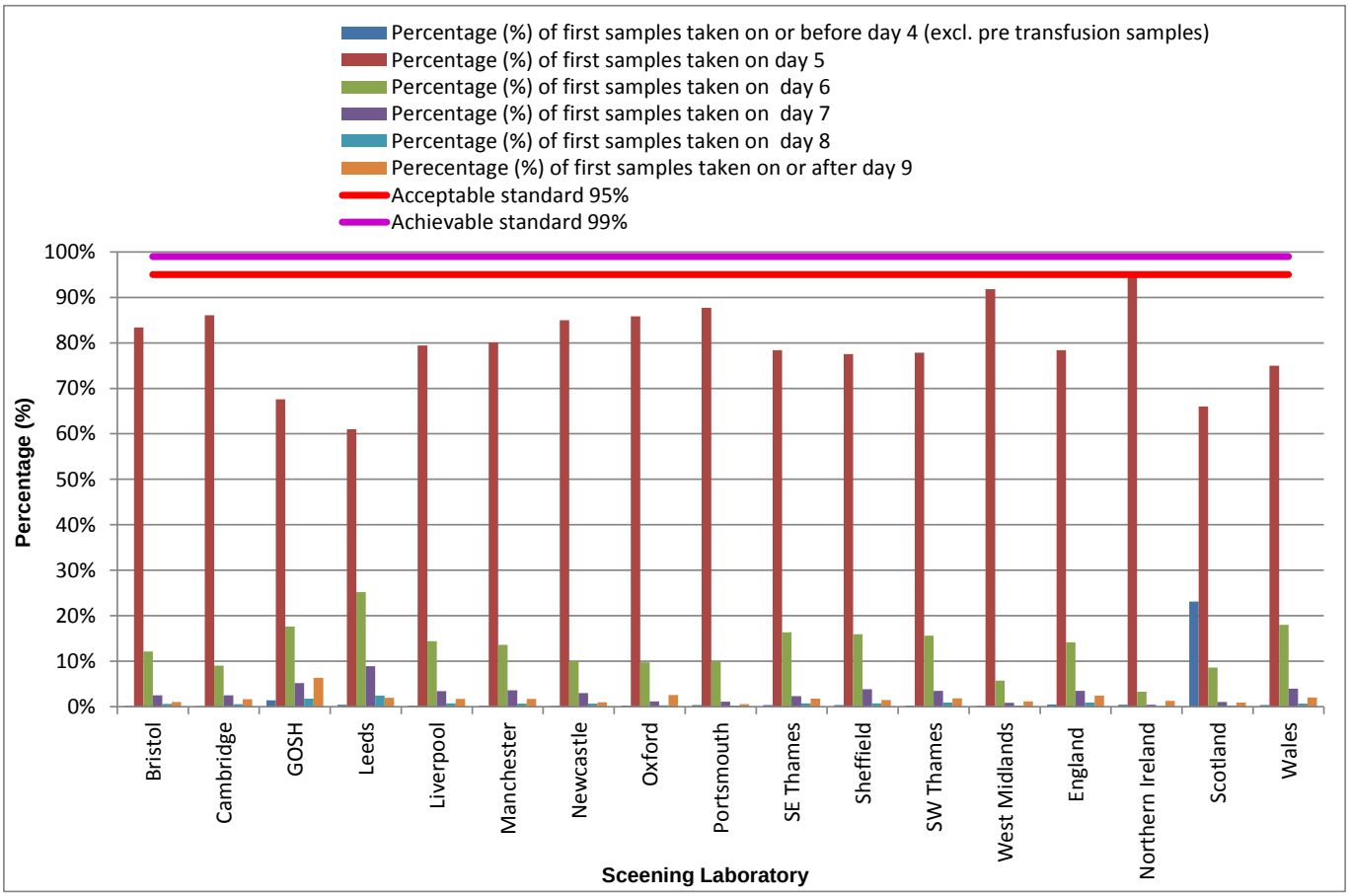
Laboratory	First samples taken							
	on or before day 4*		on day 5		on days 5 to 8		on or after day 9	
	n	%	n	%	n	%	n	%
Bristol	83	0.2	30,877	83.4	36,553	98.7	395	1.1
Cambridge	38	0.1	23,620	86.1	26,944	98.2	452	1.6
GOSH	1,799	1.4	85,859	67.6	117,144	92.2	8,090	6.4
Leeds	178	0.4	25,864	61.0	41,364	97.6	842	2.0
Liverpool	73	0.3	22,957	79.4	28,323	98.0	499	1.7
Manchester	131	0.2	44,379	80.1	54,295	98.0	962	1.7
Newcastle	71	0.2	27,766	84.9	32,283	98.8	333	1.0
Oxford	70	0.2	24,486	85.8	27,719	97.2	741	2.6
Portsmouth	121	0.4	28,052	87.7	31,670	99.1	180	0.6
SE Thames	202	0.4	43,883	78.4	54,760	97.8	1,007	1.8
Sheffield	269	0.4	54,404	77.6	68,858	98.2	1,018	1.5
SW Thames	146	0.3	41,113	77.8	51,700	97.8	990	1.9
West Midlands	118	0.2	63,887	91.8	68,681	98.7	806	1.2
England total	3,299	0.5	517,147	78.4	640,294	97.0	16,315	2.5
Northern Ireland	111	0.5	22,818	94.4	23,752	98.3	308	1.3
Scotland†	12,542	23.1	35,774	66.0	41,171	75.9	503	0.9
Wales	123	0.4	24,417	75.0	31,792	97.6	655	2.0
UK total	16,075	2.1	600,156	77.9	737,009	95.6	17,781	2.3

Data source: Newborn screening laboratories

*For the purposes of this standard, day of birth is taken as day 0. Pre-transfusion samples are excluded from the denominator and numerator

†Scotland accept samples to be taken on or before day 4 without asking for repeat. In 2016 to 2017 23.1% of samples were received on or before day 4. If these are included, the percentage (%) will be higher.

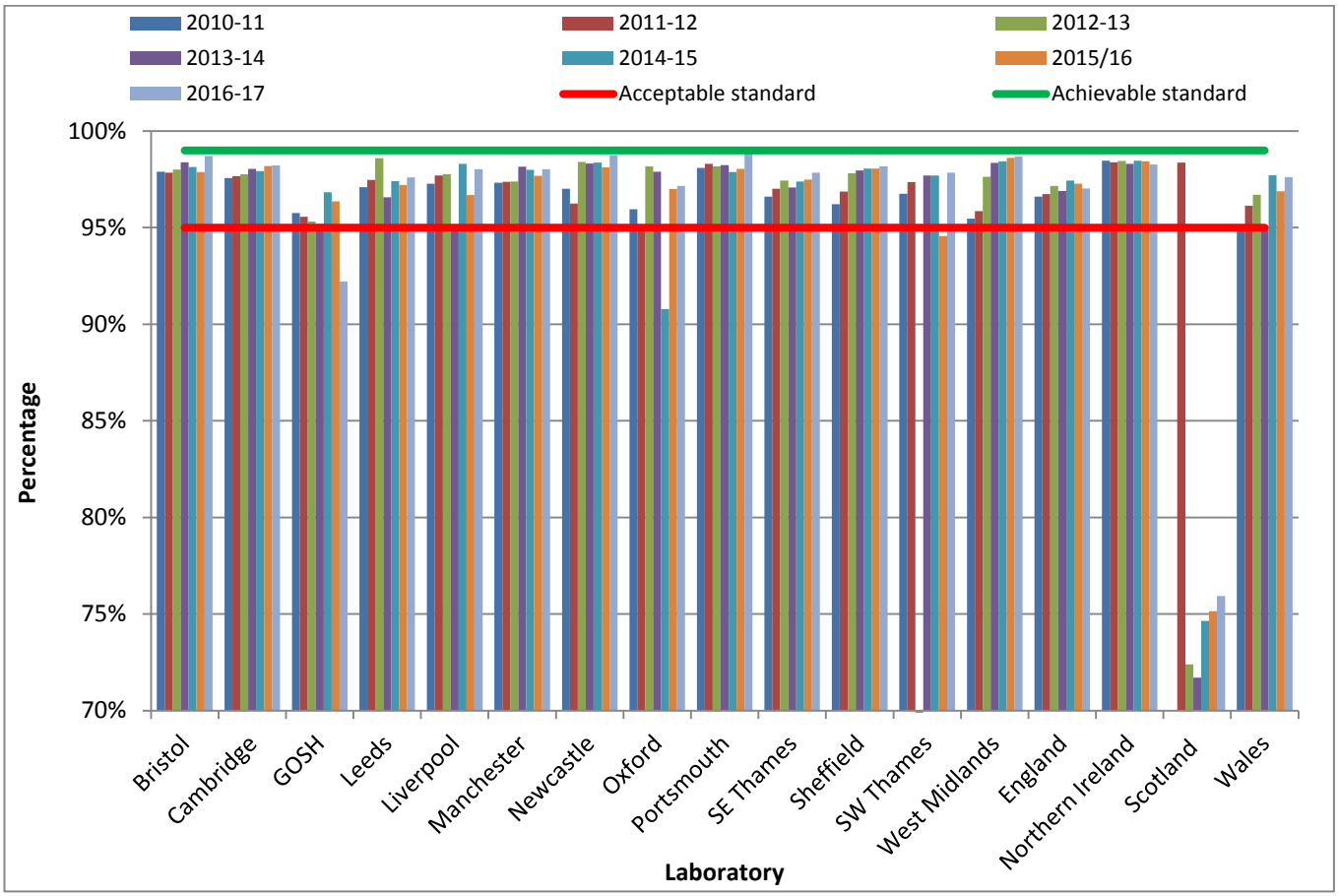
Figure 9: Day of first sample collection 2016 to 2017



Data source: Newborn screening laboratories

*Scotland accept samples to be taken on or before day 4 without asking for repeat. In 2016 to 2017 23.1% of samples were received on or before day 4. If these are included, the percentage (%) will be higher.

Figure 10: Percentage of samples taken on days 5 to 8 from 2010 to 2017



Data source: Newborn screening laboratories
Please note that the Y axis does not begin at zero

Standard 5: Timely receipt of a sample in the newborn screening laboratory

Description

To maximise accuracy of the screening test. All samples must arrive within the screening laboratory as soon as possible after the sample has been taken. This enables the laboratory to analyse the sample at the earliest opportunity and also reduces the risk of sample deterioration due to prolonged despatch.

Acceptable level

Equal to or greater than 99% of all samples received within 4 working days of sample collection.

Achievable level

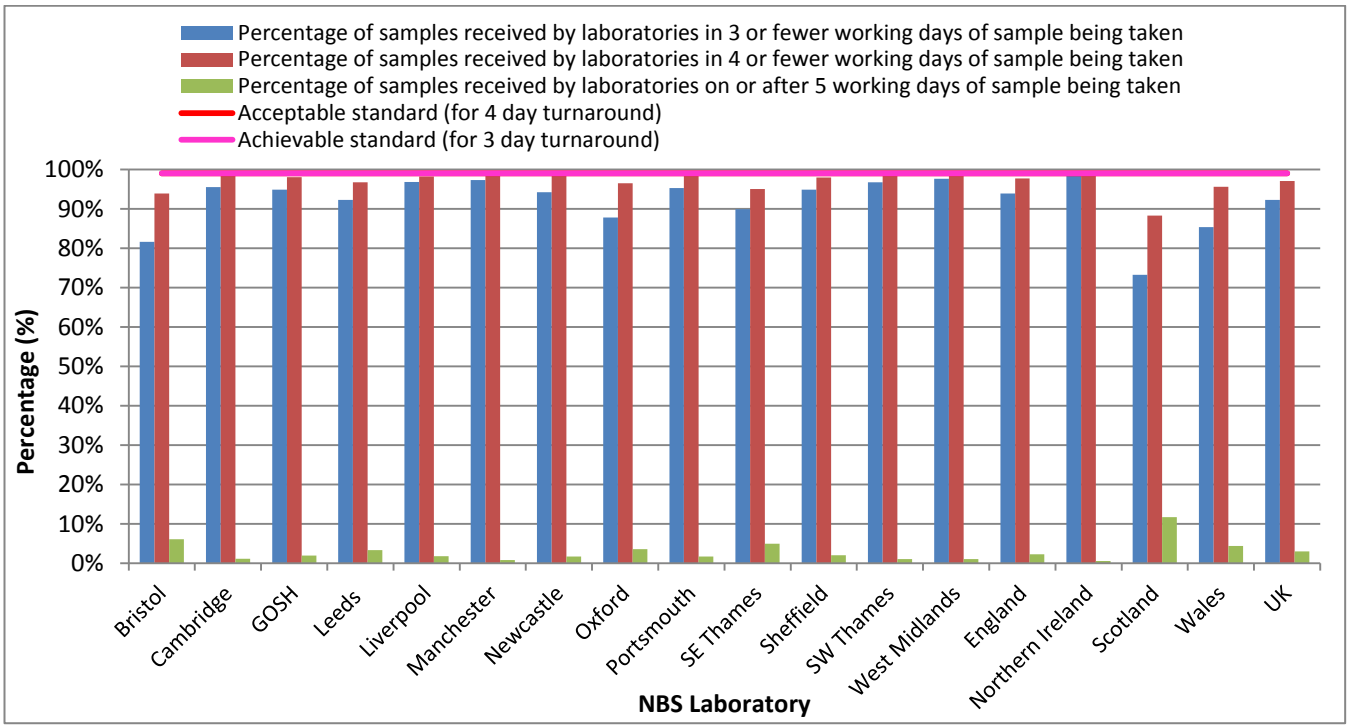
Equal to or greater than 99% of all samples received within 3 working days of sample collection.

Table 8: Number of working days taken to receive sample 2016 to 2017

Laboratory	Samples received					
	within 3 working days		within 4 working days		on or after 5 working days	
	n	%	n	%	n	%
Bristol	33,258	81.6	38,264	93.9	2,479	6.1
Cambridge	27,163	95.5	28,132	98.9	312	1.1
GOSH	121,283	94.9	125,349	98.1	2,464	1.9
Leeds	41,464	92.3	43,456	96.7	1,470	3.3
Liverpool	29,354	96.8	29,792	98.2	533	1.8
Manchester	56,256	97.3	57,382	99.2	437	0.8
Newcastle	32,486	94.2	33,894	98.3	575	1.7
Oxford	27,770	87.8	30,500	96.5	1,116	3.5
Portsmouth	31,628	95.2	32,655	98.3	554	1.7
SE Thames	55,417	89.9	58,577	95.0	3,056	5.0
Sheffield	70,491	94.9	72,796	98.0	1,492	2.0
SW Thames	53,024	96.7	54,270	99.0	551	1.0
West Midlands	72,560	97.6	73,585	99.0	756	1.0
England total	652,154	93.9	678,652	97.7	15,795	2.3
Northern Ireland	26,410	98.5	26,660	99.5	147	0.5
Scotland	40,528	73.2	48,858	88.3	6,478	11.7
Wales	30,201	85.4	33,814	95.6	1,557	4.4
UK total	749,293	92.3	787,984	97.0	23,977	3.0

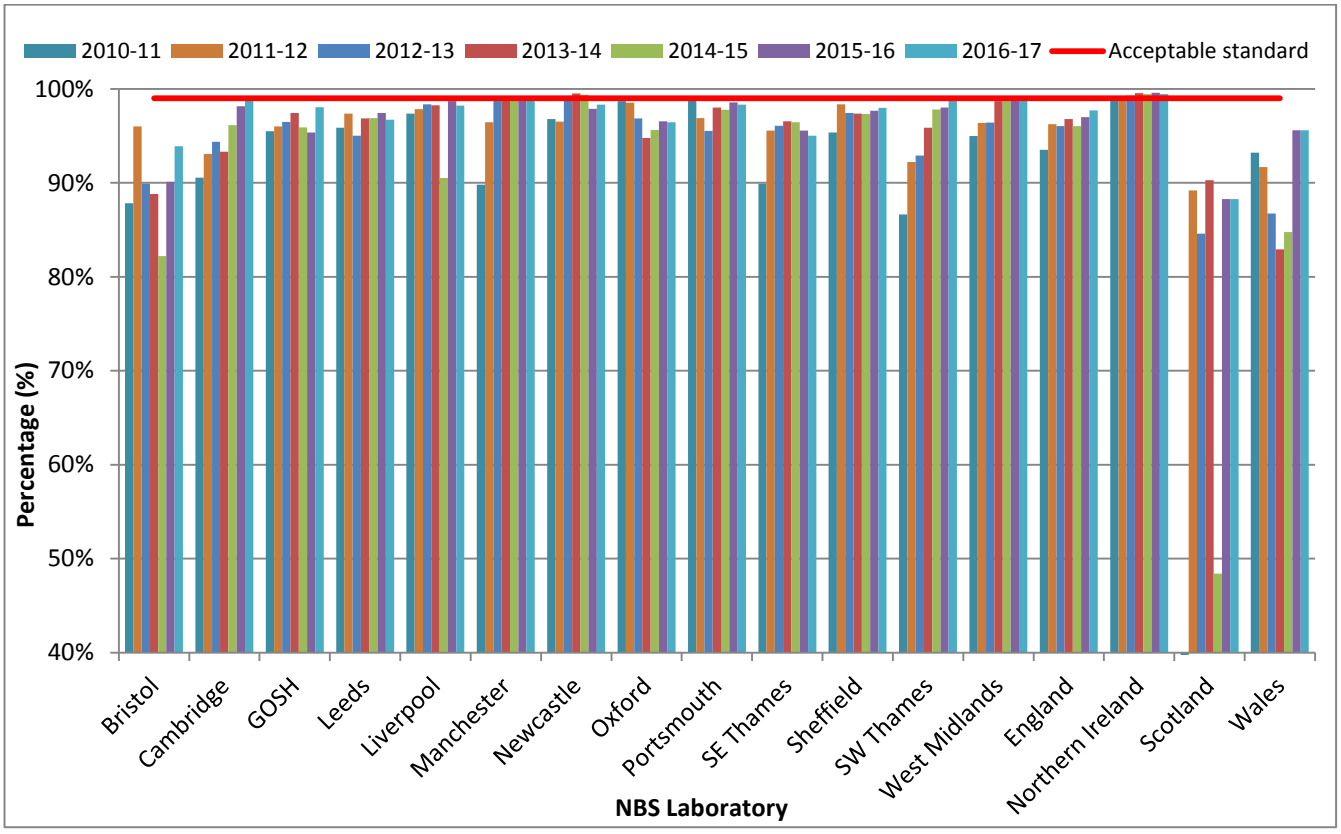
Data source: Newborn screening laboratories

Figure 11: Number of working days taken to receive sample 2016 to 2017



Data source: Newborn screening laboratories

Figure 12: Percentage of samples received within 4 working days 2010 to 2017



Data source: Newborn screening laboratories
Please note that the Y axis does not begin at zero

Standard 6: Quality of the blood spot sample

Description

A good quality blood spot sample is one that is taken at the right time, has all data fields completed on the blood spot card, contains sufficient blood to perform all tests, has not been contaminated, and arrives in the laboratory in a timely manner.

Avoidable repeat requests (numerator) 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)
- insufficient blood
- unsuitable sample/card (eg on an expired blood spot card, contaminated, in transit for more than 14 days, anti-coagulated sample, baby's NHS number and/or other details not accurately recorded on the blood spot card)

Acceptable level: the avoidable rate is less than or equal to 2%.
Achievable level: the avoidable rate is less than or equal to 0.5%.

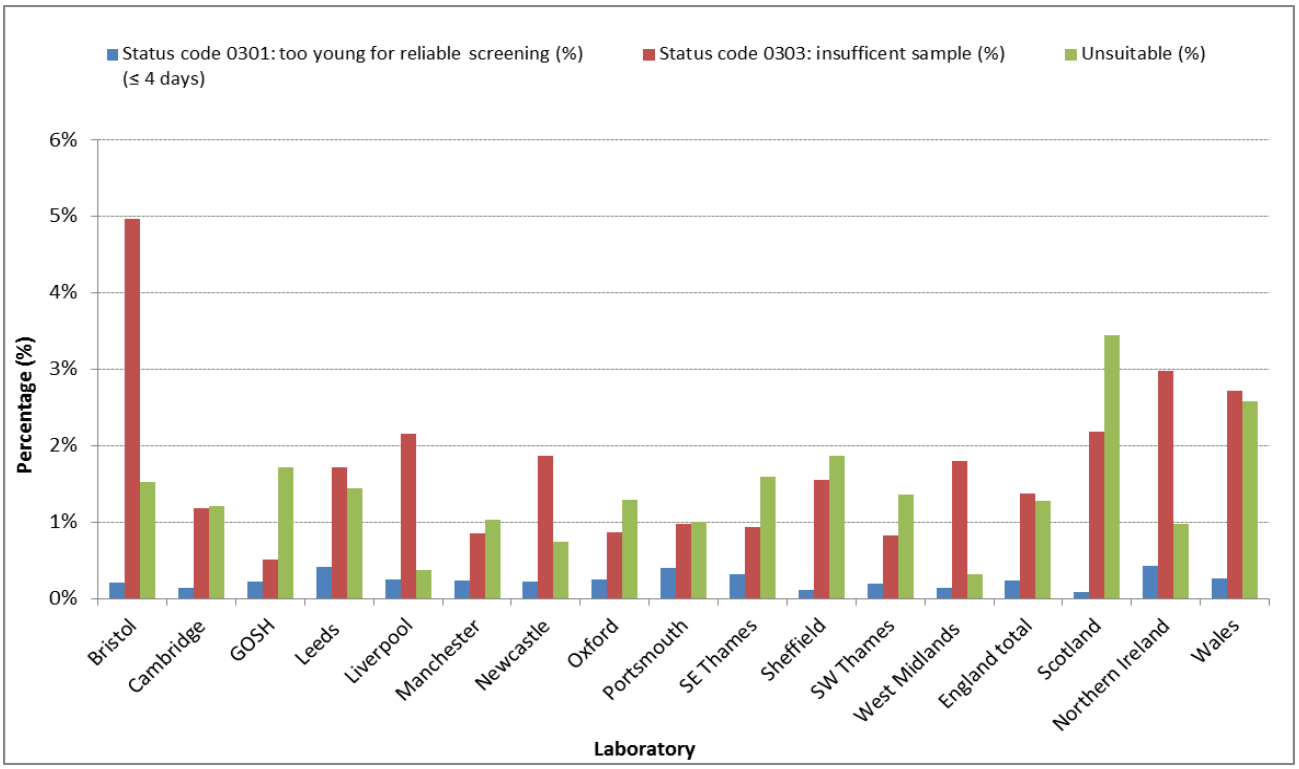
Table 9: Avoidable repeat request rates 2016 to 2017

Laboratory	First samples received/babies tested	Repeat (second or subsequent) samples requested by the laboratory because the previous sample was:						Avoidable repeat request rate
		taken when the baby was too young*		insufficient		unsuitable		
	n	n	%	n	%	n	%	%
Bristol	37,032	79	0.21	1,839	4.97	565	1.53	6.71
Cambridge	27,585	38	0.14	326	1.18	333	1.21	2.53
GOSH	127,803	282	0.22	648	0.51	2,190	1.71	2.44
Leeds	42,819	178	0.42	733	1.71	617	1.44	3.57
Liverpool	28,924	73	0.25	625	2.16	108	0.37	2.79
Manchester	55,661	128	0.23	478	0.86	572	1.03	2.12
Newcastle	32,687	71	0.22	611	1.87	245	0.75	2.84
Oxford	28,530	70	0.25	249	0.87	370	1.30	2.42
Portsmouth	32,217	131	0.41	315	0.98	324	1.01	2.39
SE Thames	56,213	182	0.32	529	0.94	899	1.60	2.86
Sheffield	70,658	80	0.11	1,095	1.55	1,323	1.87	3.54
SW Thames	52,842	106	0.20	438	0.83	717	1.36	2.39
West Midlands	69,655	98	0.14	1,256	1.80	224	0.32	2.27
England total	662,626	1,516	0.23	9,142	1.38	8,487	1.28	2.89
Northern Ireland	24,339	105	0.43	724	2.97	239	0.98	4.39
Scotland	54,313	49	0.09	1,185	2.18	1,870	3.44	5.72
Wales	32,570	85	0.26	886	2.72	841	2.58	5.56
UK total	773,848	1,755	0.23	11,937	1.54	11,437	1.48	3.22

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

Data source: Newborn screening laboratories

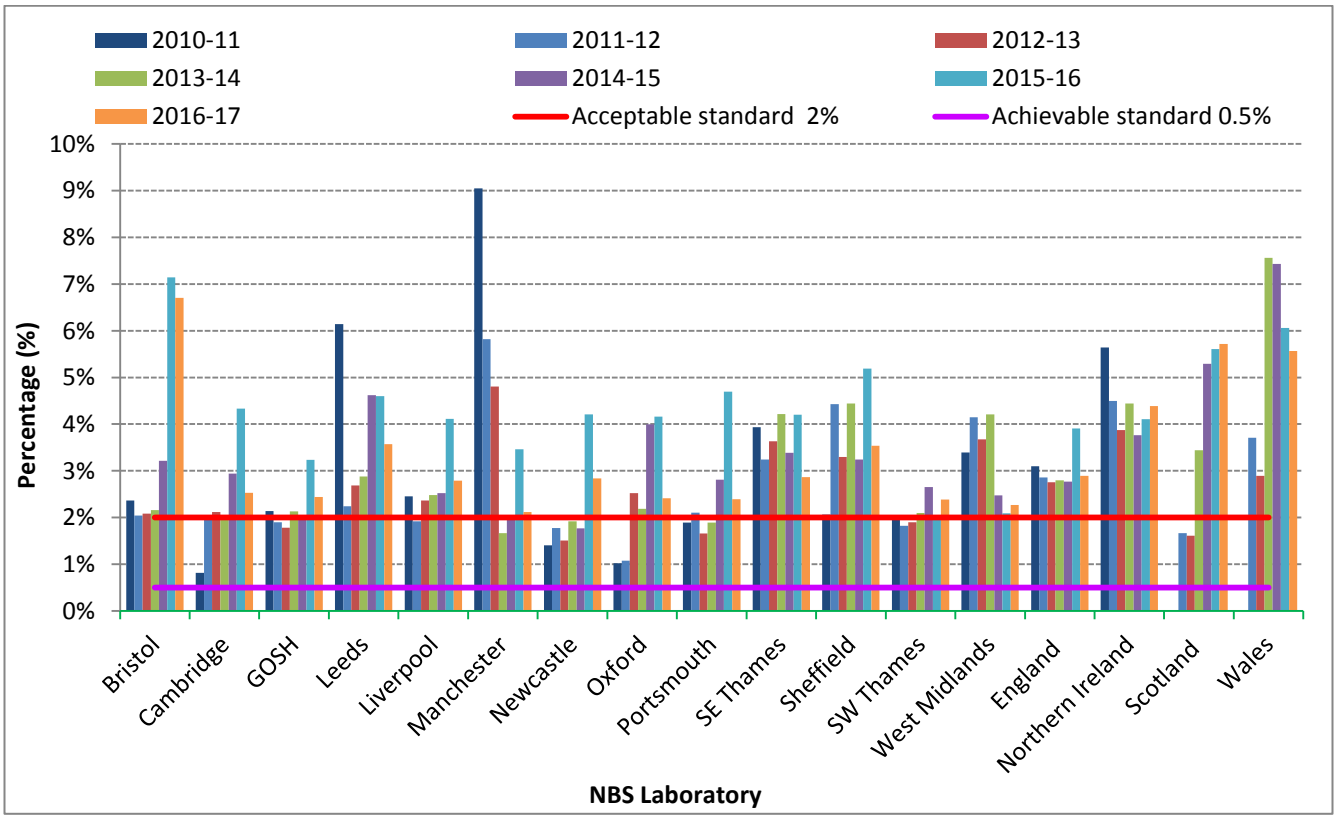
Figure 13: Avoidable repeat request rates (too young, insufficient and unsuitable) 2016 to 2017



Data source: Newborn screening laboratories

† 'Unsuitable total' includes status codes: 0304: unsuitable sample (blood quality): incorrect blood application, 0305: unsuitable sample (blood quality): compressed/damaged, 0306: unsuitable sample: day 0 and day 5 on same card, 0307: unsuitable sample for CF: discrepant IRT replicates, possible faecal contamination, 0308: unsuitable sample: NHS number missing/not accurately recorded, 0309: unsuitable sample: date of sample missing/not accurately recorded, 0310: unsuitable sample: date of birth not accurately matched, 0311: unsuitable sample: expired card used, 0312: unsuitable sample: >14 days in transit, too old for analysis and 0313: unsuitable sample: damaged in transit.

Figure 14: Avoidable repeat request rates 2010 to 2017



Data source: Newborn screening laboratories

Please note that 2010 to 2013 data include avoidable repeat requests due to insufficient and unsuitable samples only. In line with standard 6, 2013 to 2016 data include repeat requests due to samples taken when the baby was too young, insufficient and unsuitable.

New consensus guidelines on quality blood spot were introduced in April 2015 following which the percentage of avoidable repeats predictably rose. Gratifyingly they fell for this year.

Standard 7: Timely taking of a repeat blood spot sample

Description

This standard covers repeat/second samples requested by the laboratory because the first sample was of poor quality, not valid for testing or required by the UK protocol for the specific condition. In order that treatment and clinical referral targets are met, the timely receipt of a repeat/second blood spot sample is imperative.

Acceptable level

Equal to or greater than 95% of repeat samples taken as defined.

Achievable level

Equal to or greater than 99% of repeat samples taken as defined.

Laboratory information management systems do not currently support collection of data for this standard.

Standard 8: CPA (screening)

Description

Laboratories undertaking newborn blood spot screening shall be accredited by Clinical Pathology Accreditation (UK) Ltd (CPA), now formally part of the United Kingdom Accreditation Service (UKAS). This shall include the NBS specialist assessment. DNA laboratories shall be a member of the UK Genetic Testing Network (UK GTN) and comply with the quality criteria laid down by the UK GTN Steering Group.

Acceptable level

The laboratory is CPA accredited (with the specialist assessment of NBS screening by the next full visit).

Laboratory accreditation (standards 8 and 10) will be published by the United Kingdom Accreditation Service (UKAS).

Standard 9: Timely processing of all PKU, CHT and MCADD screen positive samples

Description

This standard relates to PKU, CHT and MCADD and subsequent action on positive screening results. It is intended to measure the timeliness of screening laboratory processes and clinical referral. The purpose is to facilitate high quality and timely intervention for those who wish to participate.

Acceptable level

100% of babies with a positive screening result have a clinical referral initiated within 4 working days of sample receipt by screening laboratory.

Achievable level

100% of babies with a positive screening result have a clinical referral initiated within 3 working days of sample receipt by screening laboratory.

Table 10: Numbers of samples processed measured against the standard in the UK 2016 to 2017

Condition*	Screen positive samples	Screen positive babies with clinical referral initiated within 4 working days of sample receipt		Screen positive babies with clinical referral initiated within 3 working days of sample receipt	
	n	n	%	n	%
PKU	87	86	98.9	85	97.7
CHT	561	560	99.8	545	97.1
MCADD	73	73	100.0	73	100.0
England total	721	719	99.7	703	97.5

Data source: Newborn screening laboratories

*Data is reported against the 2013 standards therefore this does not reflect all the conditions

Standard 10: CPA (diagnosis)

Description

Follow up screening and diagnostic tests shall be undertaken in line with the diagnostic protocols.

Acceptable level

The laboratory is CPA accredited.

Laboratory accreditation (standards 8 and 10) will be published by the United Kingdom Accreditation Service (UKAS).

Standard 11: Timely receipt into clinical care

Sickle cell disease

Newborn screening for sickle cell disease (SCD) saw approximately 667,500 newborn babies screened for SCD in England in 2016 to 2017 (includes both tested and declines) and approximately 778,900 for the whole of UK.* Of those screened in England, 274 babies were identified with significant conditions** (0.41 per 1000 screened) and approximately 8,530 babies were identified as carriers (12.78 per 1000 screened).

There was a decline in the number of screen positives in London in newborn screening but rates remain steady in the rest of England. The uptake of testing of the baby's biological father continues to improve slightly nationally, and uptake in low prevalence areas appears to have improved following a decline since 2013 to 2014. This may possibly reflect the change to the programme guidance to recommend testing fathers in every pregnancy.

The programme requests data on laboratory processes and timeliness of entry into care for screen positive babies. There were, however, some gaps in the numbers for age at receipt of sample in the laboratory and for age at first visit to a paediatrician at a specialist health team or local health team. Just under 40% of newborn screen positives had no information provided for their age at first visit to a paediatrician, but of those that did have information approximately 90% were seen by 90 days of age (where data was provided).

While beta thalassaemia is not currently screened for in newborn screening, F-only cases are picked up as a by-product of screening for sickle cell disease. These are likely to be beta thalassaemia major cases and require follow-up. In 2016 to 2017 there were 25 F-only cases in England, and across the whole of the UK.

Rates of declined screening continue to rise and are now at approximately 2.3 per 1,000 babies screened which is similar to the rate of declined antenatal screening. It is not possible to say why there is this increase, but some possible explanations include mover in babies who have been tested elsewhere and re-testing is declined, better reporting of declines now that there is a sub-code for this, or it may be that the figures include declined repeat samples rather than having declined screening entirely.

*Newborn laboratories report on samples rather than babies tested
**Significant conditions comprise FS, FSC, FS-other and FE results. Carrier results comprise FAS, FAC, FAD, FAE and other haemoglobin variants.

Data source: NHS Sickle Cell and Thalassaemia Screening Programme Data Report 2016 to 2017: trends and performance analysis (England only report)

CF – screen positive babies with 2 cystic fibrosis transmembrane conductance regulator (CFTR) mutations

Description
A baby in whom CF is suspected should have their first clinical appointment by 28 days of age:

Acceptable level: 95% of babies seen by 28 days of age

Achievable level: 100% of babies seen by 28 days of age

Table 11: Timeliness of appointment and outcome for CF screen positive babies with 2 mutations 2016 to 2017

	England	Northern Ireland	Scotland	Wales
Number of CF screen positive babies with two mutations	177	9	17	8
Number diagnosed before screening (excluded from following age data)	29	2	4	0
Number of babies with age at first appointment reported	126 (85%)	7 (100%)	12 (92%)	4 (50%)
Number seen ≤ 28 days (% of known data)	113 (90%)	7 (100%)	11 (92%)	3 (75%)
All babies mean age at first appointment	22	21	23	24
All babies median age at first appointment	21	21	23	23
Age range at first appointment	8-33	14-26	17-37	20-30
Number of babies with age at first appointment not reported	22 (15%)	0	1 (8%)	4 (50%)
Outcome (out of ALL babies screened positive with 2 mutations. Includes number diagnosed before screening)				
Confirmed	152 (86%)	8 (89%)	13 (76%)	8 (100%)
CF SPID	10 (6%)	1 (11%)	0	0
Excluded	1	0	0	0
Not reported	14 (8%)	0	4 (24%)	0

Data source: Newborn screening laboratories
Note that different screening and diagnostic protocols are followed in the UK – see Figures 17-20.

CF – screen positive babies with one or no mutations

Description
A baby in whom CF is suspected should have their first clinical appointment by 35 days of age:

Acceptable level: 80% of babies seen by 35 days of age

Achievable level: 100% of babies seen by 35 days of age

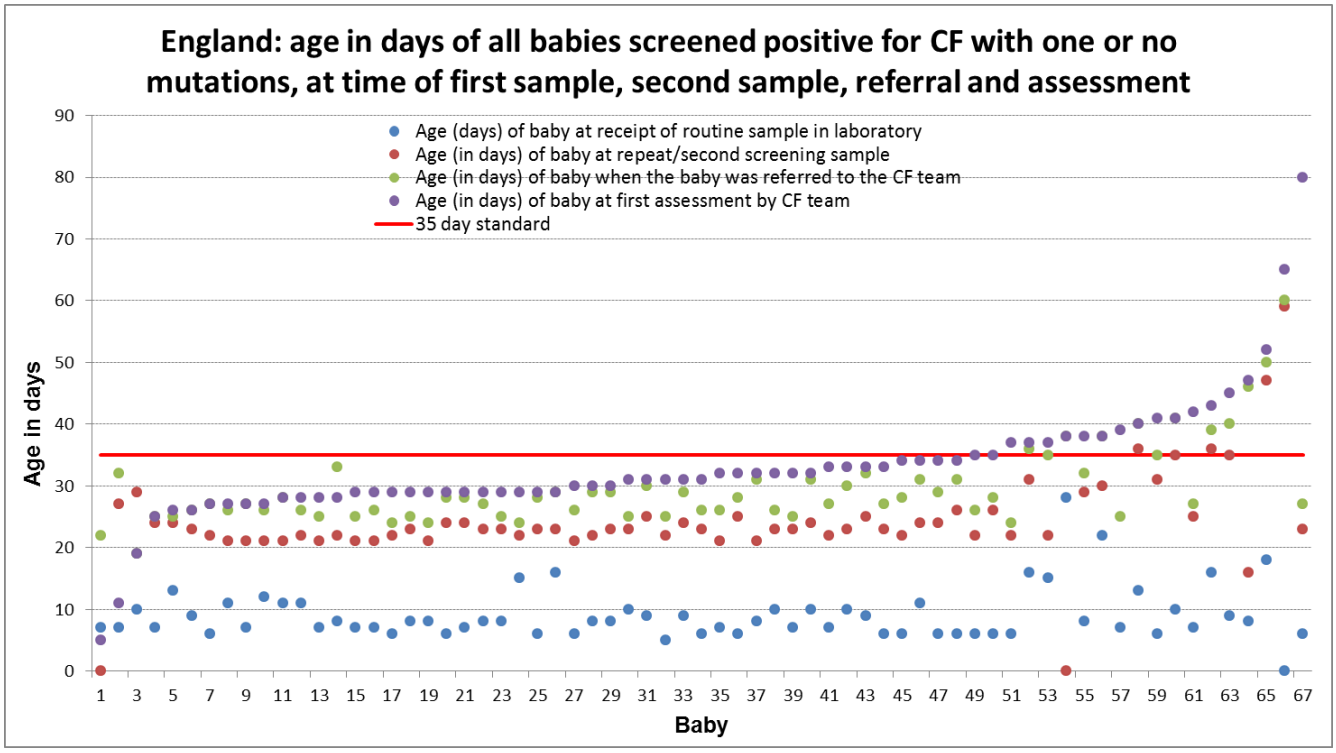
Table 12: Timeliness of appointment and outcome for CF screen positive babies with one or no mutations 2016 to 2017

	England	Northern Ireland	Scotland	Wales*
Number of CF screen positive babies with one or no mutations	**99**	**9**	**14**	**10**
Number diagnosed before screening (excluded from following age data)	2	0	0	0
Number of babies with age at first appointment reported	68 (70%)	9 (100%)	11 (79%)	4 (40%)
Number seen ≤ 35 days (% of known data)	49 (72%)	8 (89%)	6 (55%)	4 (100%)
All babies mean age at first appointment	34	30	36	24
All babies median age at first appointment	32	30	35	23
Age range at first appointment	19-80	24-39	18-57	20-29
Number of babies with age at first appointment not reported	29 (30%)	0	3 (21%)	6 (60%)
Outcome (out of ALL babies screened positive with 1 or 0 mutations. Includes number diagnosed before screening)				
Confirmed	24 (24%)	2 (22%)	3 (21%)	0
CF SPID	2 (2%)	1 (11%)	0	1 (10%)
Excluded	44 (44%)	4 (44%)	1 (7%)	7 (70%)
Baby died	1 (1%)	0	0	0
Not reported	26 (26%)	0	5 (36%)	2 (20%)
Carrier	2 (2%)	0	5 (36%)	0
Other	0	2** (22%)	0	0

Data source: Newborn screening laboratories
*Wales data does not include 0 mutations
**For Northern Ireland there were 2 babies who did not have further testing

Figure 15a: CF screen positive babies with one or no mutations 2016 to 2017, England only

Age in days of all babies screened positive for CF with one or no mutations, at time of first sample, second sample, referral and assessment



Data source: Newborn screening laboratories

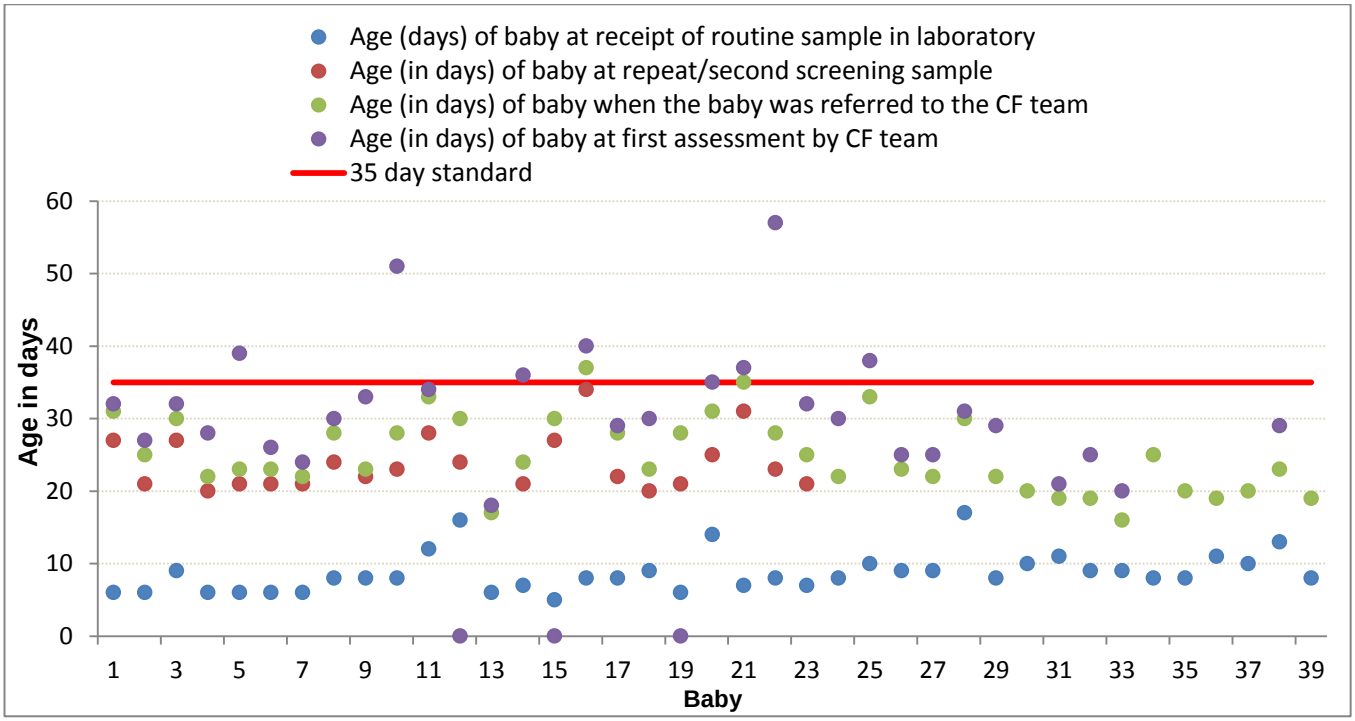
Baby 1: Age (in days) of baby at repeat/second screening sample; reported as N/A
Outcome CF confirmed. Additional comments include "already diagnosed, same mutations as mother who has CF"

Baby 54: Age (in days) of baby at repeat/second screening sample; reported as N/A. Outcome CF excluded.

Baby 66: Age (in days) of baby at receipt of routine sample in laboratory; reported as N/A. Outcome CF excluded.

Figure 15b: CF screen positive babies with one or no mutations 2016 to 2017, Northern Ireland, Scotland and Wales

Age in days of all babies screened positive for CF with one or no mutations, at time of first sample, second sample, referral and assessment



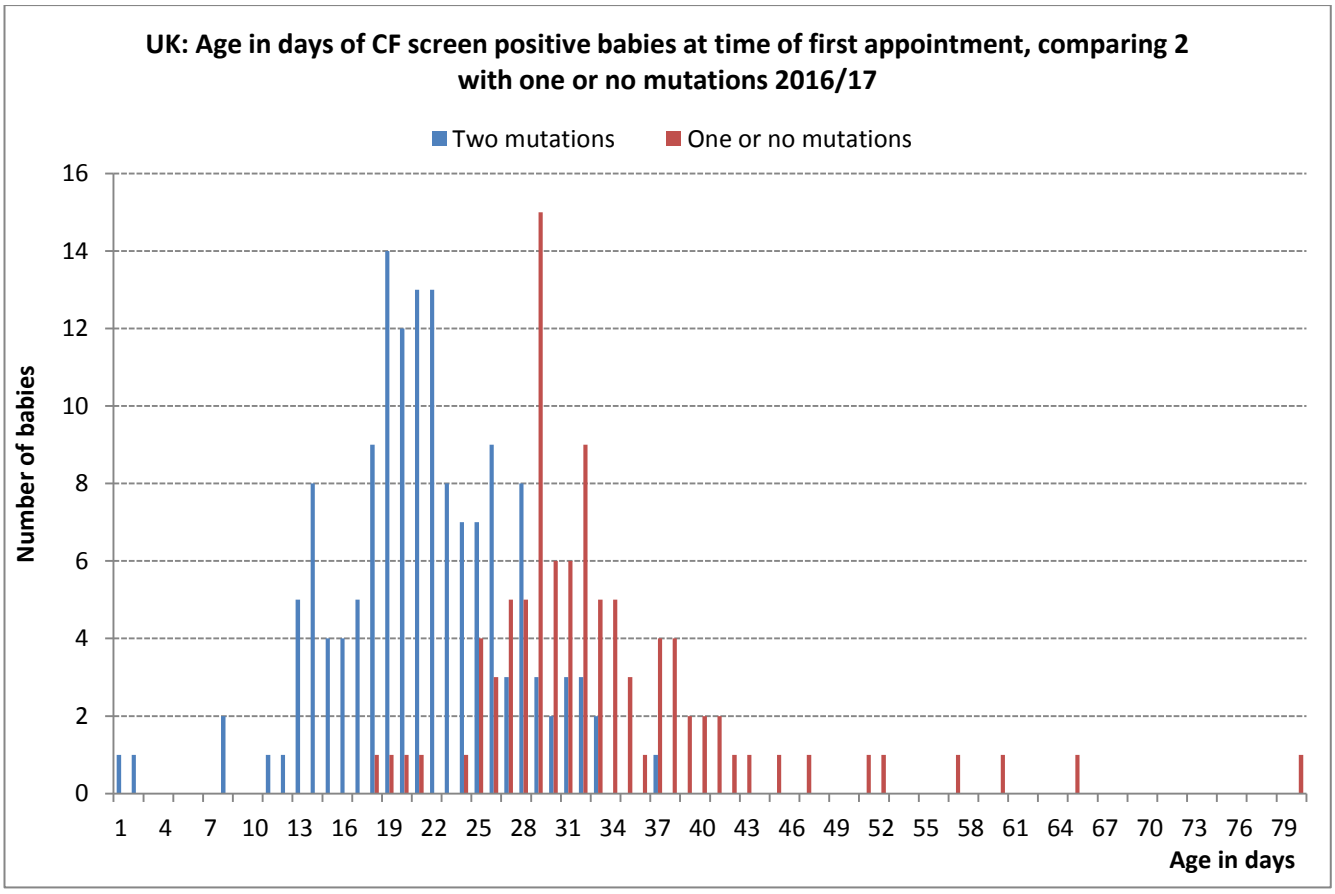
Data source: Newborn screening laboratories

Baby 12: Age (in days) of baby at first assessment by CF team; reported as N/A
No outcome data reported

Baby 15: Age (in days) of baby at first assessment by CF team; reported as N/A. No outcome data reported.

Baby 19: Age (in days) of baby at first assessment by CF team; reported as N/A. No outcome data reported .

Figure 16: CF screen positive babies comparing 2 with one or no mutations 2016 to 2017, UK: Age in days at time of first appointment



Data source: Newborn screening laboratories

Figure 17: England CF screening and diagnostic algorithm 2016 to 2017

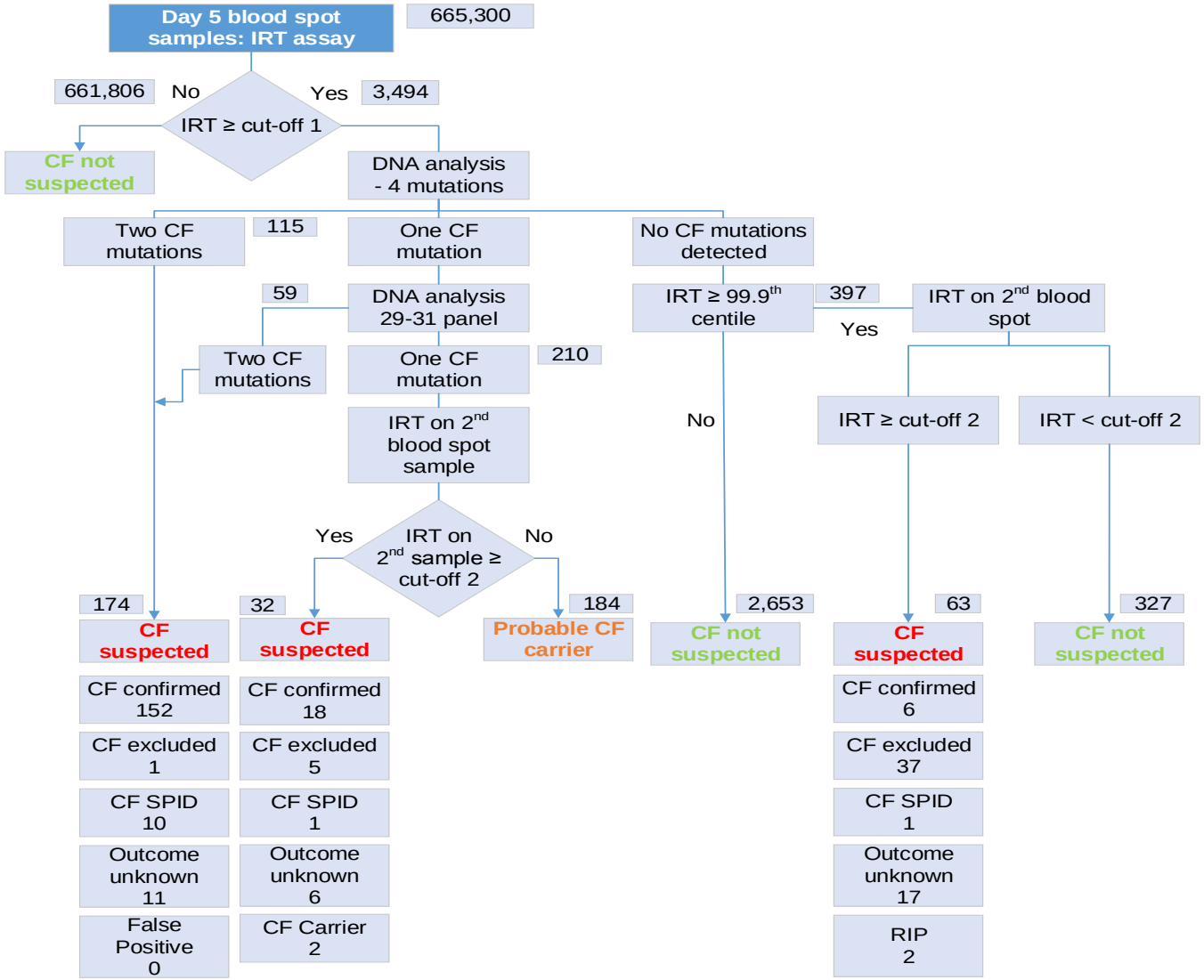


Figure 18: Northern Ireland CF screening and diagnostic algorithm 2016 to 2017

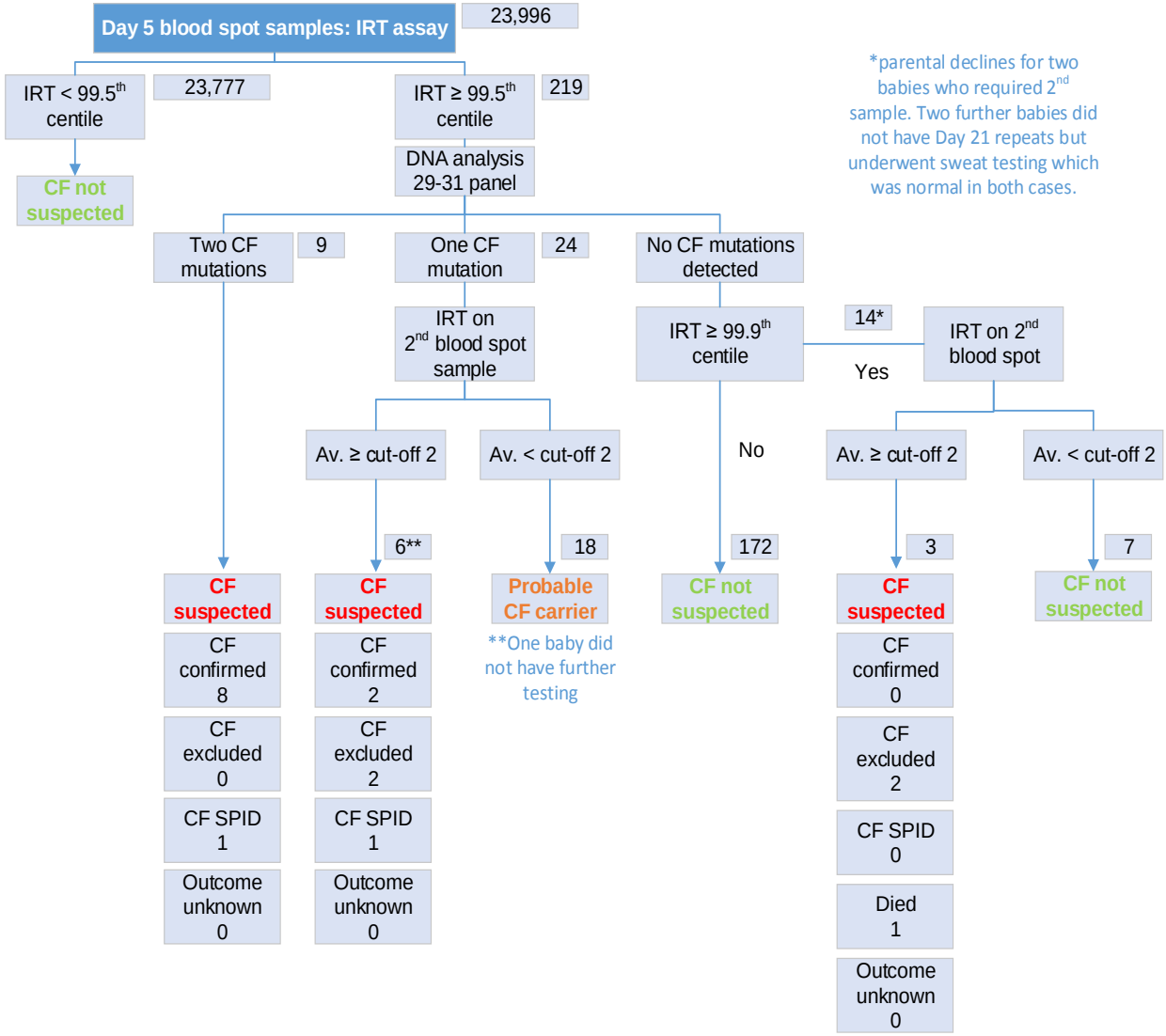


Figure 19 Scotland CF screening and diagnostic algorithm 2016 to 2017

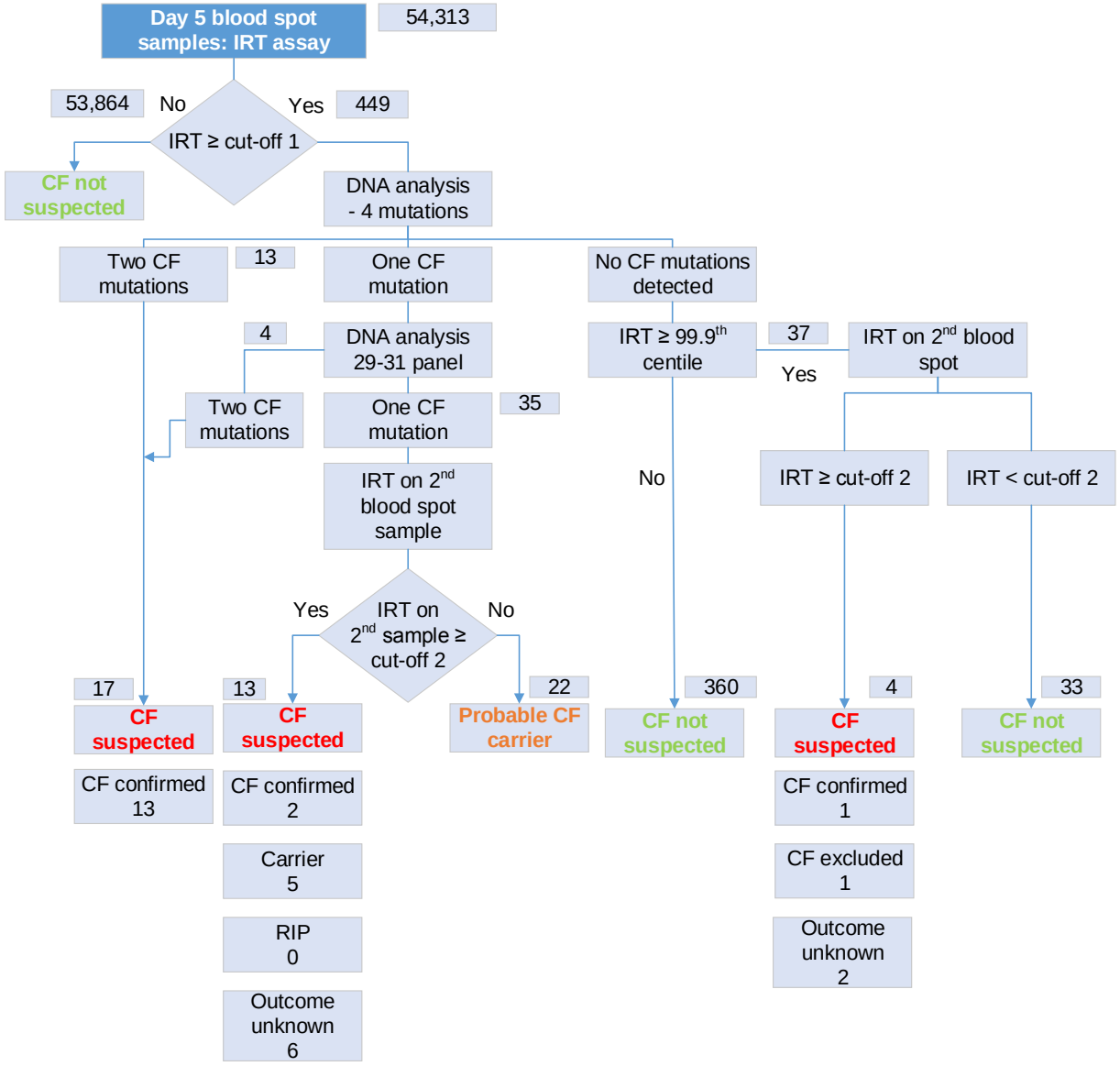
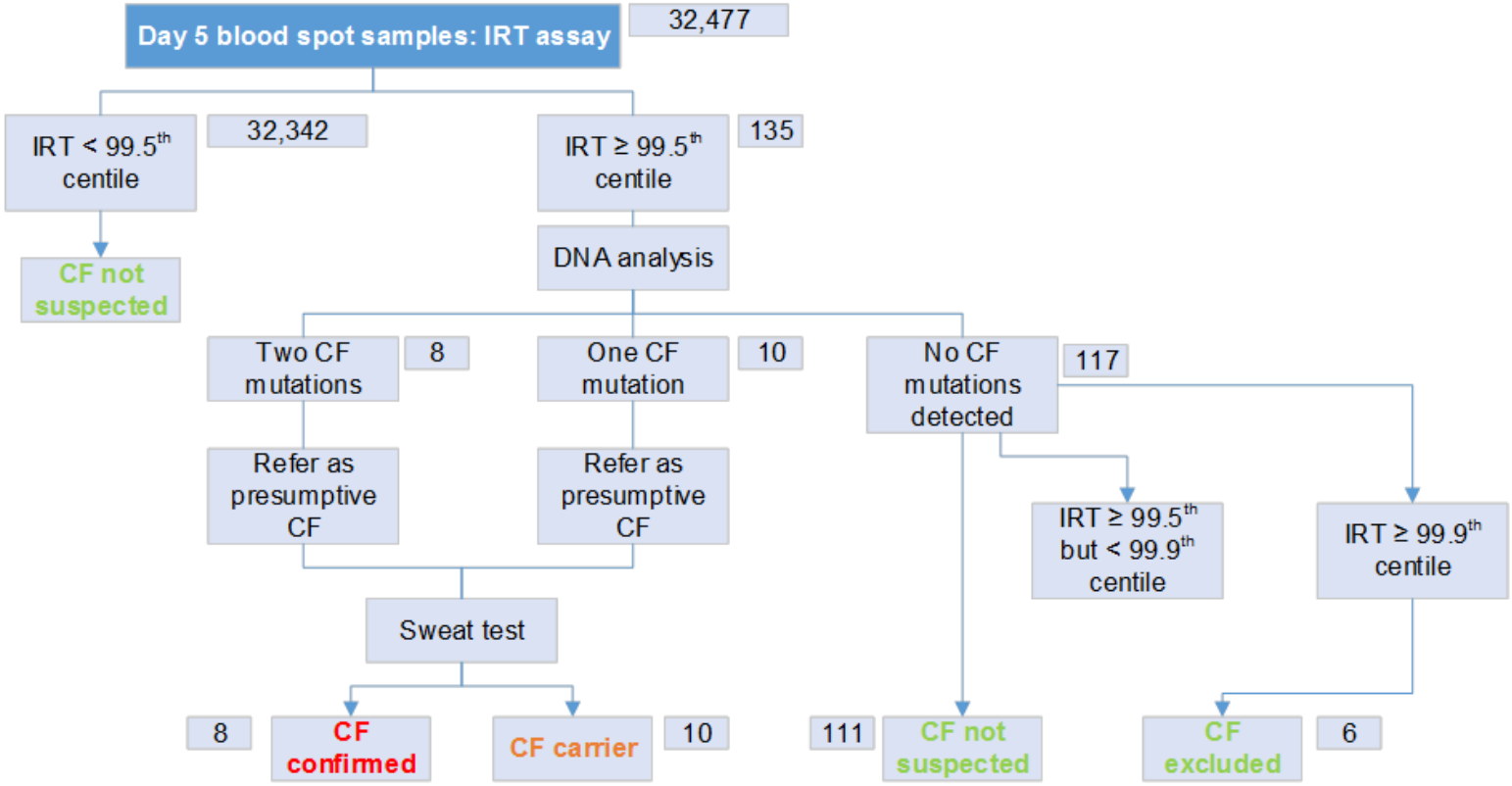


Figure 20: Wales CF screening and diagnostic algorithm 2016 to 2017



CF screen positive data 2007 to 2017

Table 13: CF screen positive data 2007 to 2017

Laboratory	Babies tested for CF 2007 to 2017	CF screen positives 2007 to 2017	Rate of CF screen positives 2007 to 2017
	n	n	Rate per 10,000
Bristol	412,401	243	5.89
Cambridge	277,793	117	4.21
GOSH	1,191,734	327	2.74
Leeds	445,064	181	4.07
Liverpool	290,132	164	5.65
Manchester	533,199	194	3.64
Newcastle	341,863	156	4.56
Oxford	296,819	80	2.70
Portsmouth	370,931	147	3.96
SE Thames	521,190	193	3.70
Sheffield	736,422	329	4.47
SW Thames	507,211	165	3.25
West Midlands	715,906	271	3.79
England total	6,640,665	2568	3.87
Northern Ireland	189,106	114	6.03
Scotland	518,554	285	5.50
Wales	344,201	200	5.81
UK total	7,692,526	3167	4.12

Data source: Newborn screening laboratories

CHT – screen positive babies detected on first sample (not including preterm babies)

Description
A baby in whom CHT is suspected on the first sample should attend their first clinical appointment by:

Acceptable level: 100% by 17 days of age
Achievable level: 100% by 14 days of age

Table 14: Timeliness of appointment and treatment outcome for CHT screen positive babies detected on first sample 2016 to 2017

	England	Northern Ireland	Scotland	Wales
Number of CHT screen positive babies detected on first sample	283	9	18	6
Number diagnosed before screening (excluded from following age data)	7	0	0	0
Number of babies with age at first appointment reported	221 (80% of 275)	9 (100%)	18 (100%)	6 (100%)
Number seen ≤ 14 days standard (% of known data)	207 (94% of 221)	9 (100%)	15 (83%)	6 (100%)
Number seen ≤ 17 days standard (% of known data)	217 (98% of 221)	9 (100%)	16 (89%)	6 (100%)
All babies mean age at first appointment	11	10	12	12
All babies median age at first appointment	11	10	10	12
Age range at first appointment	3 to 22	8 to 11	6 to 27	10 to 14
Number of babies with age at first appointment not reported	49 (18% of 275)	0	0	0
Inpatient	5	0	0	0
Baby died	0	0	0	0
Other*	1	0	0	0
Has the baby started on thyroxine at the first appointment?				
Yes	197	9	17	5
No	4	0	0	0
Not reported	63	0	0	0

Thyroxine not given but follow up required	15	0	1	1
Thyroxine not given and baby discharged	3	0	0	0

*One baby indicated as “DECLINE;mother refused to bring child, GP had to treat at home”
Data source: Newborn screening laboratories

CHT – screen positive babies detected on second sample (not including preterm babies)

Description

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:

Acceptable level: 100% by 24 days of age
Achievable level: 100% by 21 days of age

Table 15: Timeliness of appointment and treatment outcome for CHT screen positive babies detected on second sample 2016 to 2017

	England	Northern Ireland	Scotland	Wales
Number of CHT screen positive babies detected on second sample	240	10	13	0*
Number diagnosed before screening (excluded from following age data)	5	0	0	0
Number of babies with age at first appointment reported	171 (73% of 235)	10 (100%)	13 (100%)	
Number seen ≤ 21 days standard (% of known data)	135 (79% of 171)	10 (100%)	8 (62%)	
Number seen ≤ 24 days standard (% of known data)	154 (90% of 171)	10 (100%)	8 (62%)	
All babies mean age at first appointment	22	17	24	
All babies median age at first appointment	19	17	20	
Age range at first appointment	4 to 374	15 to 19	15 to 46	

Number of babies with age at first appointment not reported	61 (26% of 235)	0	0	
Inpatient	2	0	0	
Other**	1	0	0	
Has the baby started on thyroxine at the first appointment?				
Yes	111	9	5	
No	7	0	0	
Not reported	68	0	0	
Thyroxine not given but follow up required	37	1	6	
Thyroxine not given and baby discharged	17	0	2	

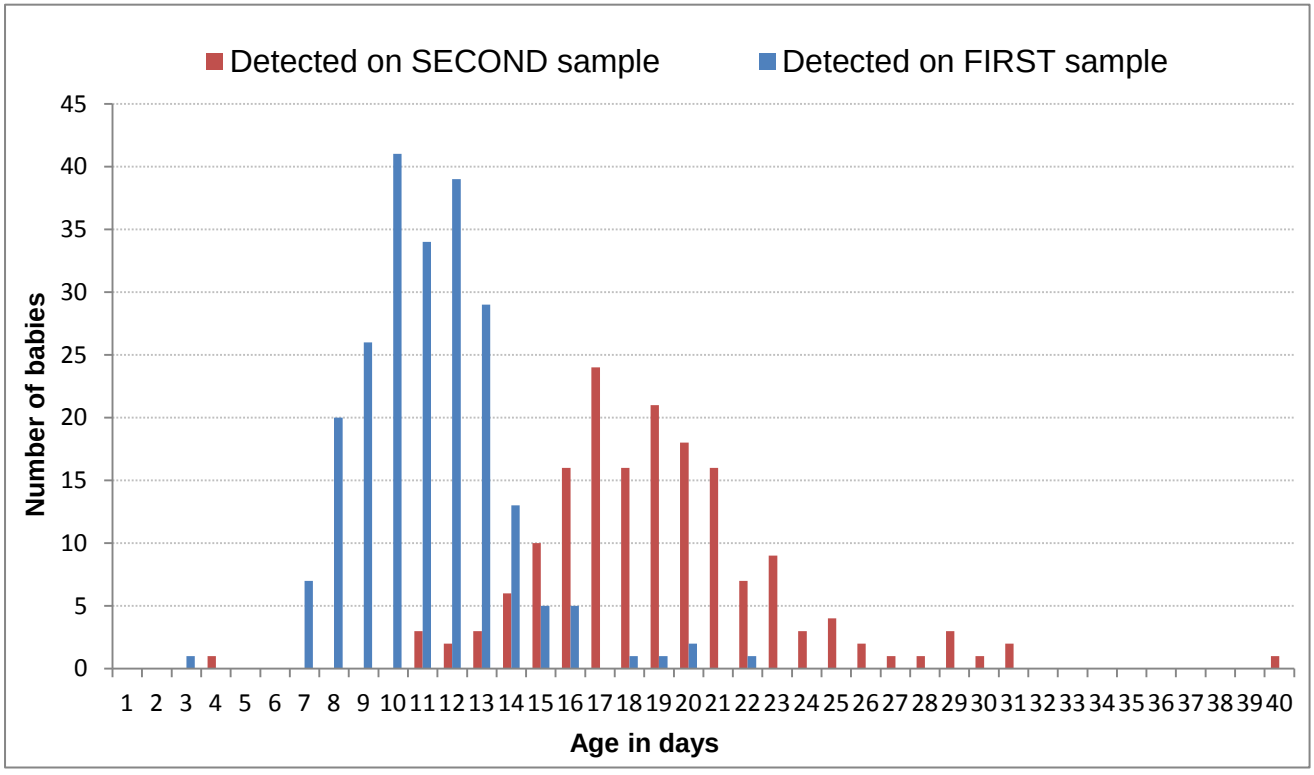
Data source: Newborn screening laboratories

*For Wales there were no full-term babies categorised as detected on second sample. However there were 8 full-term babies categorised as detected CHT suspected on preterm repeat (double borderline TSH result)

**No appointment, CHT suspected on double borderline TSH result. Age at referral 21 days, outcome CHT excluded,

Figure 21: Age in days of CHT screen positive babies (not including preterm babies)

Detected on first and second sample at time of first appointment 2016 to 2017, England only



Data source: Newborn screening laboratories

CHT – screen positive preterm babies (born at less than 32 weeks)

Data on CHT preterm babies will be further analysed in detail separately

CHT results depending on use of national or local borderline cut-off level

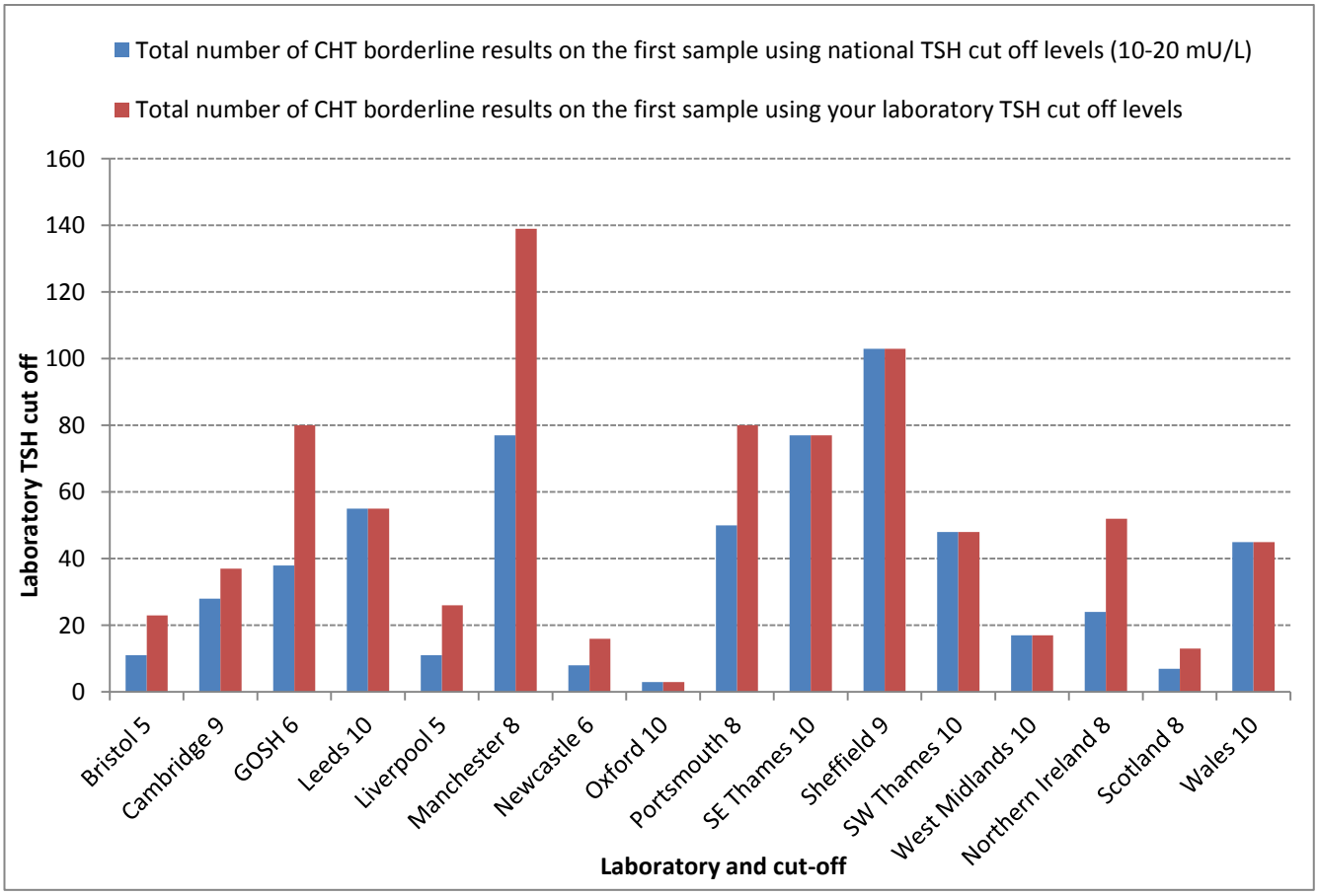
CHT is the only screening protocol in which a borderline result necessitates a second sample before a conclusive result can be achieved. The national borderline cut-off level is 10 mU/L. Some laboratories use a local cut-off level.

Table 16: CHT borderline results depending on use of national or local cut-off level 2016 to 2017

Laboratory	What TSH* cut-off levels do you use to determine a positive screen for CHT (mU/L)?	What TSH cut-off levels do you use to determine a borderline screen for CHT (mU/L)?	Total number of CHT borderline results on the first sample using national TSH cut-off level (10-20 mU/L)	Total number of CHT borderline results on the first sample using local TSH cut-off level
Bristol	18	6	11	23
Cambridge	18 (GSP)	9 (GSP)	28	37
GOSH**	18 (GSP)	6 (GSP)	38	80
Leeds	20	10	55	55
Liverpool	20	5	11	26
Manchester	≥20	≥8	77	139
Newcastle	20	6	8	16
Oxford	20	10	3	3
Portsmouth	20	8	50	80
SE Thames	20	10	77	77
Sheffield	18 (GSP)	9 (GSP)	103	103
SW Thames	20	10	48	48
West Midlands	20	10-20	17	17
Northern Ireland	20	8	24	52
Scotland	20	8	7	13
Wales	20	10	45	45

*Thyroid stimulating hormone (TSH)
**Great Ormond Street Hospital (GOSH) laboratory changed to Genetic Screening Processor (GSP) December 2015.
Note that GSP cut-offs are equivalent to national cut-offs.
Data source: Newborn screening laboratories

Figure 22: CHT borderline results depending on use of national or local cut-off level 2016 to 2017



Data source: Newborn screening laboratories

CHT screen positive data 2007 to 2017

Table 17: CHT screen positive data 2007 to 2017

Laboratory	Babies tested for CHT 2007 to 2017	CHT screen positives 2007 to 2017	Rate of CHT screen positives 2007 to 2017
	n	n	Rate per 10,000
Bristol	412,526	256	6.21
Cambridge	278,527	191	6.86
GOSH	1,237,896	1359	10.98
Leeds	445,208	307	6.90
Liverpool	291,524	305	10.46
Manchester	563,599	434	7.70
Newcastle	341,875	276	8.07
Oxford	297,920	201	6.75
Portsmouth	382,195	199	5.21
SE Thames	573,716	329	5.73
Sheffield	737,869	428	5.80
SW Thames	521,227	320	6.14
West Midlands	715,905	547	7.64
England total	6,799,987	5152	7.58
Northern Ireland	248,355	176	7.09
Scotland	518,053	253	4.88
Wales	344,671	273	7.92
UK total	7,911,066	5854	7.40

Data source: Newborn screening laboratories

PKU

Description

A baby in whom PKU is suspected should attend their first clinical appointment by:

Acceptable level: 100% by 17 days of age

Achievable level: 100% by 14 days of age

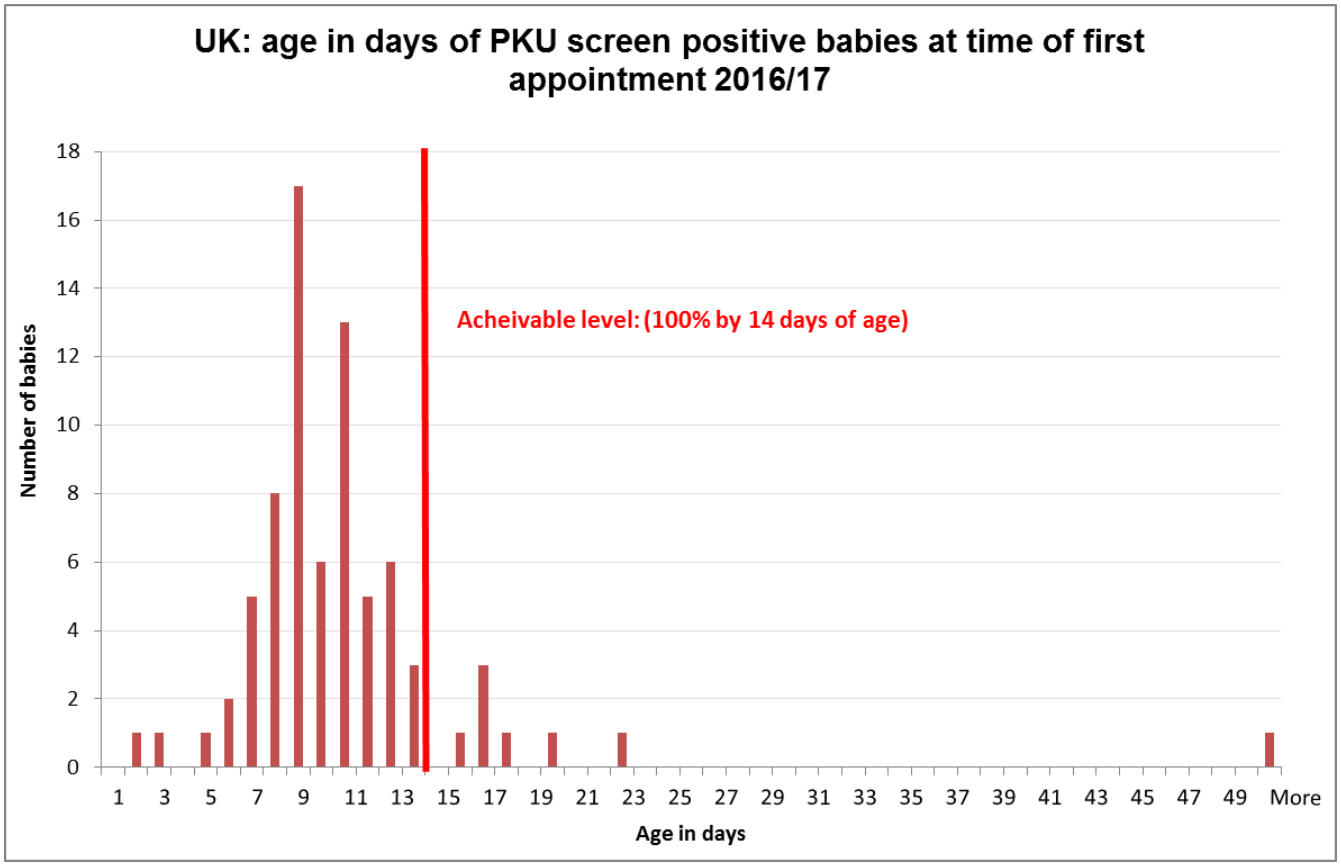
Table 18: Timeliness of appointment and outcome for PKU screen positive babies 2016 to 2017

	England	Northern Ireland	Scotland	Wales
Number of PKU screen positive babies	88	4	6	9
Number diagnosed before screening e.g family history of PKU *(excluded from following age data)*	10	1	1	0
Number of babies with age at appointment reported	50	3	5	9
Number seen ≤ 14 days (% of known data)	46 (92%)	3 (100%)	5 (100%)	7 (78%)
Number seen ≤ 17 days (% of known data)	48 (96%)	3 (100%)	5 (100%)	8 (89%)
All babies mean age at appointment	15 days	9 days	9 days	13 days
All babies median age at appointment	10 days	9 days	9 days	11 days
Age range at first appointment	6-262* days	7-11 days	7-11 days	8-23 days
Number of babies with age at appointment not reported	22 (28% of 78)	0	0	0
N/A/Other**	6	0	0	0
Outcome (includes number diagnosed before screening)				
PKU confirmed, treatment required	47	4	3	6
Non PKU eg biopterin disorders and liver dysfunction	7	0	1	0
Non PKU eg galactosaemia	7	0	0	1
PKU monitoring required	19	0	0	2

No persistent abnormalities - false positive (PKU excluded)	8	0	2	0

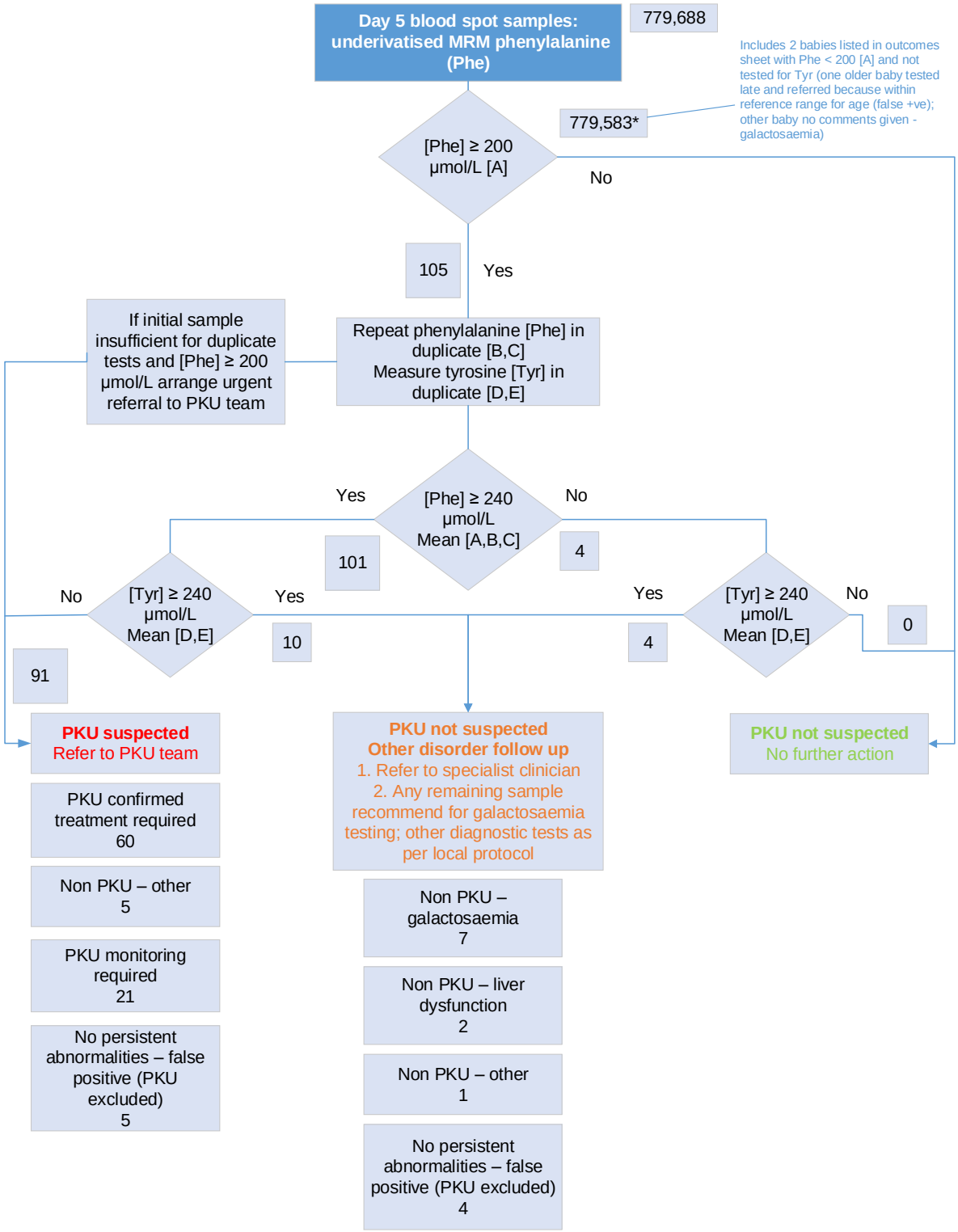
Data source: Newborn screening laboratories
*Age at 262 days was an older baby referred for further investigation as Phe>RR for age
**These babies were all non PKU (e.g biopterin disorders), other conditions include galactosaemia, liver dysfunction. One baby died at 9 days “before sample arrived in the laboratory”

Figure 23: UK: age at first appointment for PKU screen positive babies 2016 to 2017



Data source: Newborn screening laboratories

Figure 24: UK PKU screening and diagnostic algorithm 2016 to 2017



PKU screen positive data 2007 to 2017

Table 19: PKU screen positive data 2007 to 2017

Laboratory	Babies tested for PKU 2007 to 2017	PKU screen positives 2007 to 2017	Rate of PKU screen positives 2007 to 2017
	n	n	Rate per 10,000
Bristol	412,530	34	0.82
Cambridge	278,527	50	1.80
GOSH	1,228,863	113	0.92
Leeds	445,208	53	1.19
Liverpool	291,524	34	1.17
Manchester	563,641	87	1.54
Newcastle	341,875	45	1.32
Oxford	297,920	26	0.87
Portsmouth	382,405	26	0.68
SE Thames	573,714	57	0.99
Sheffield	737,871	113	1.53
SW Thames	521,225	37	0.71
West Midlands	715,905	67	0.94
England total	6,791,208	742	1.09
Northern Ireland	248,368	66	2.66
Scotland	518,098	75	1.45
Wales	344,726	60	1.74
UK total	7,902,400	943	1.19

Data source: Newborn screening laboratories

MCADD

Description

A baby in whom MCADD is suspected should attend their first clinical appointment by:

Acceptable level: 100% by 17 days of age

Achievable level: 100% by 14 days of age

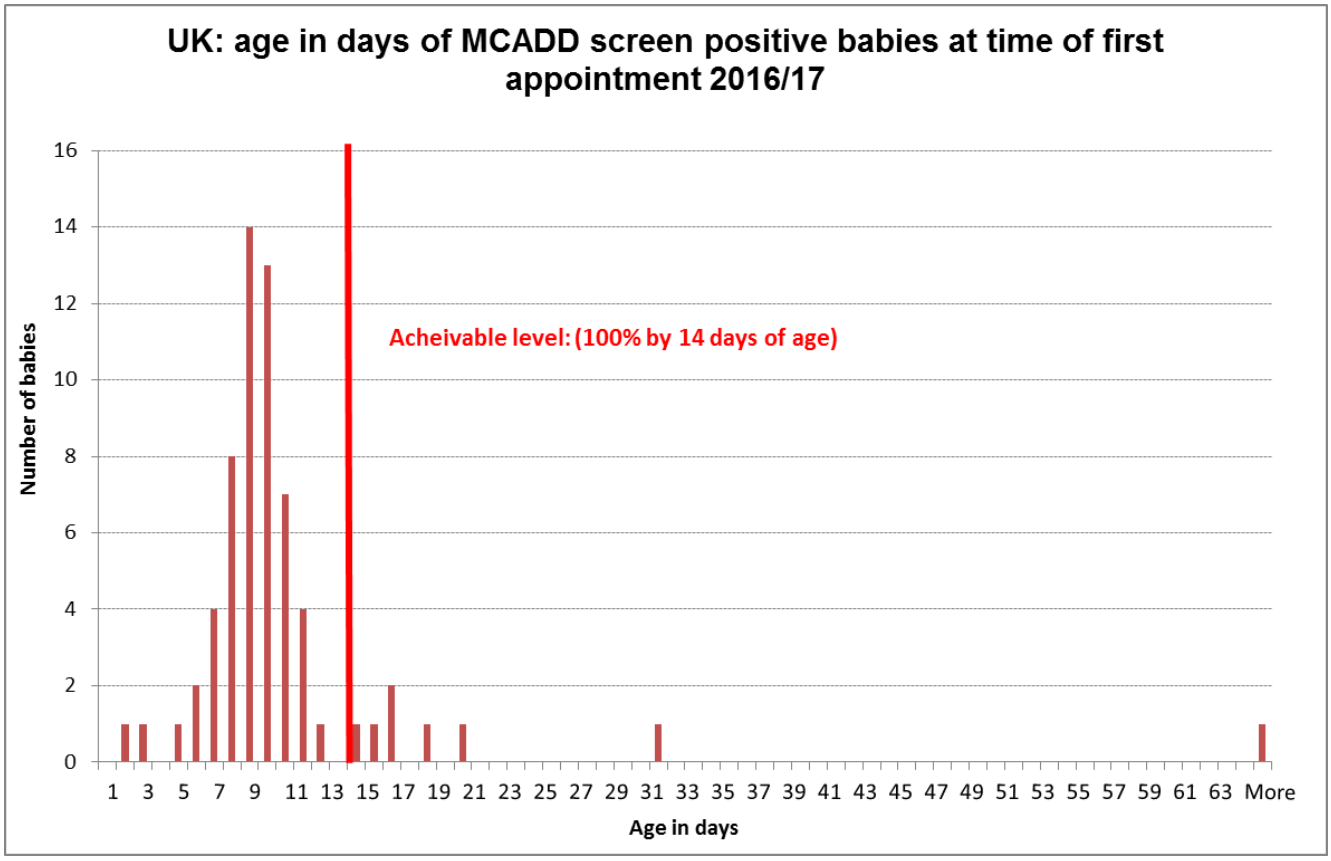
Table 20: Timeliness of appointment and outcome for MCADD screen positive babies 2016 to 2017

	England	Northern Ireland	Scotland	Wales
Number of MCADD screen positive babies	73	2	1	4
Diagnosis before routine screening? (eg affected sibling, family history)	10	0	0	0
Number of babies with age at appointment reported	50	2	1	3
Number seen ≤ 14 days (% of known data)	42 (84%)	2 (100%)	1 (100%)	3 (100%)
Number seen ≤ 17 days (% of known data)	46 (92%)	2 (100%)	1 (100%)	3 (100%)
All babies mean age at appointment	13 days	8 days	6 days	12 days
All babies median age at appointment	10 days	8 days	6 days	12 days
Age range at first appointment	2-209* days	7-8 days	n/a	10-12 days
Number of babies with age at appointment not reported	8 (13% of 63)	0	0	1 (25% of 4)
N/A/Inpatient	5	0	0	0
Outcome (includes number diagnosed before screening)				
MCADD	55	1	1	1
Unaffected carrier	1	0	0	0
No persistent abnormality, false positive	9	1	0	0
Other disorder	4	0	0	0
Not reported	4	0	0	3

Data source: Newborn screening laboratories

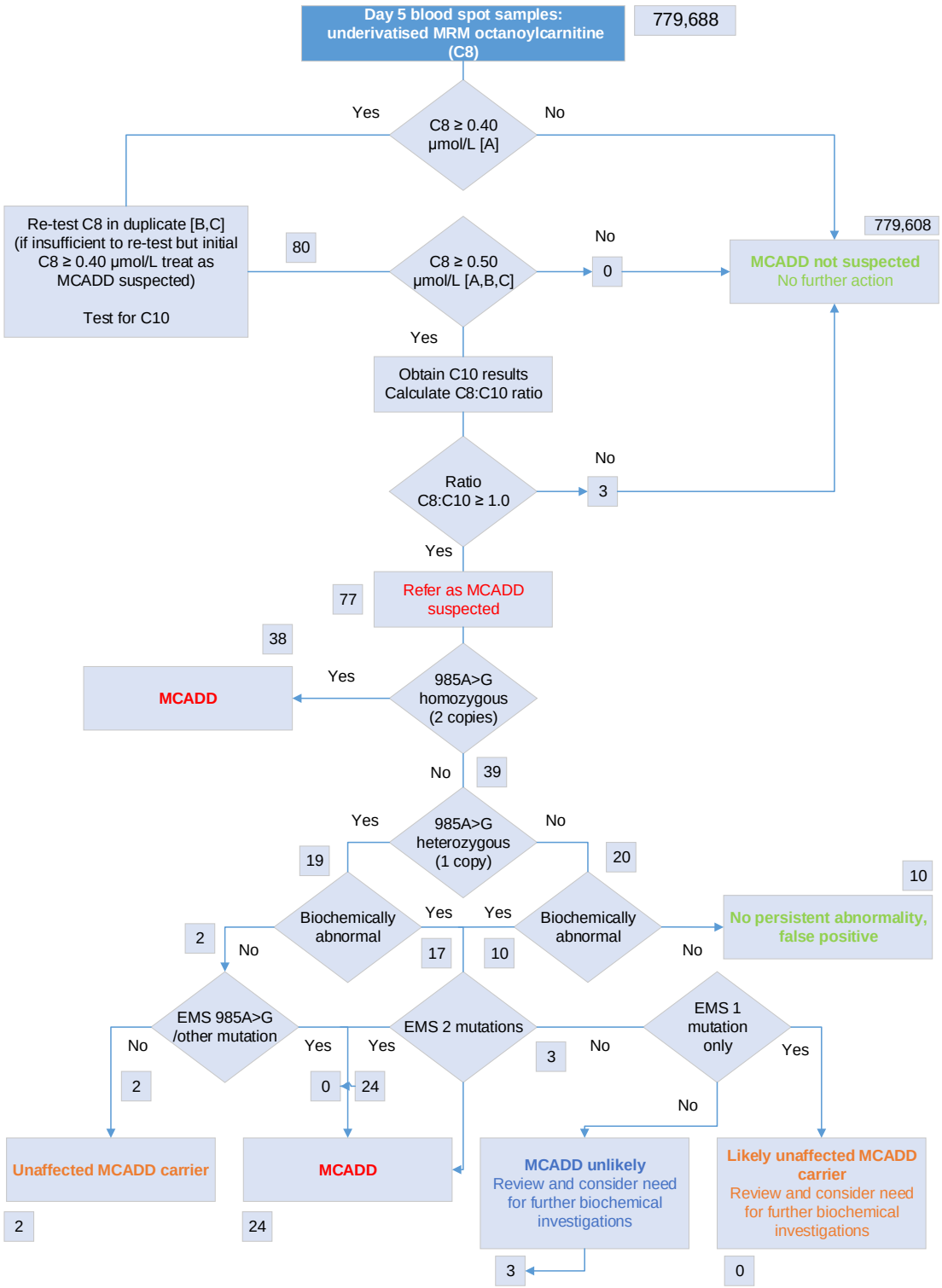
*Age at 209 days is a baby born abroad and a mover in, so screening was performed late

Figure 25 UK: age at first appointment for MCADD screen positive babies 2016 to 2017



Data source: Newborn screening laboratories

Figure 26: UK MCADD screening and diagnostic algorithm 2016 to 2017



MCADD screen positive data 2008 to 2017

Table 21: MCADD screen positive data 2008 to 2017

Laboratory	Babies tested for MCADD 2008 to 2017	MCADD screen positives 2008 to 2017	Rate of MCADD screen positives 2008 to 2017
	n	n	Rate per 10,000
Bristol	351,276	29	0.83
Cambridge	247,168	32	1.29
GOSH	1,105,361	78	0.71
Leeds	401,476	58	1.44
Liverpool	254,101	26	1.02
Manchester	507,988	58	1.14
Newcastle	287,513	30	1.04
Oxford	242,112	31	1.28
Portsmouth	332,899	36	1.08
SE Thames	517,521	39	0.75
Sheffield	664,451	103	1.55
SW Thames	449,231	37	0.82
West Midlands	645,289	55	0.85
England total	6,006,386	612	1.02
Northern Ireland	189,863	22	1.16
Scotland	372,696	12	0.32
Wales	161,507	15	0.93
UK total	6,730,452	661	0.98

Data source: Newborn screening laboratories

MSUD, IVA, GA1 and HCU

Table 22: England and Wales: Timeliness of appointment and outcome for MSUD, IVA, GA1 and HCU screen positive babies 2014 to 2017

	MSUD	IVA	GA1	HCU
Number of screen positive babies	18	23	21	18
Family history (early testing)	3	4	0	1
Number of babies with age at first appointment reported	7	13	11	4
Number seen ≤ 14 days	5	12	9	2
Number seen ≤ 17 days2	5	13	10	2
All babies median age at first appointment	16 days	9 days	11 days	17 days
Age range at first appointment	10-42 days	8-16 days	8-249 days	9-30 days
Number of babies with age at first appointment not reported	7	6	10	10
Inpatient	1	0	0	3
Outcome (includes number diagnosed before screening)				
Confirmed	11	3	6	5
Mild (IVA only)	0	5	0	0
Other	0	1	2	2
False positive	5	12	9	5
Not reported	2	2	4	6

Data source: Newborn screening laboratories

Due to small numbers, the data for IMDs are shown for a 3 year collection period.

Standard 12: Timeliness of results to parents

Description

CHRDs issue normal results for all 9 conditions to parents in a timely manner.

Acceptable level

100% of screen negative results letters are despatched direct to parents from the CHRD by 6 weeks of age.

Table 23: Timeliness of results to parents, England 2016 to 2017

Region/country	Number of babies with screen negative results for all nine conditions	Number of Babies with results despatched within six weeks of birth	
	n	n	%
England	**429,512**	**417,110**	**97.11**
North	144,511	135,601	93.83
North East	26,787	26,725	99.77
North West	58,126	49,593	85.32
Yorkshire & The Humber	59,598	59,283	99.47
South	97,157	96,613	99.44
South East	46,707	46,679	99.94
South West	50,450	49,934	98.98
Midlands & East	153,639	151,046	98.31
East Midlands	56,495	56,246	99.56
East of England	49,630	49,527	99.79
West Midlands	47,514	45,273	95.28
London	34,205	33,850	98.96

Data source: CHRDs
Data is shown for England only. Data is based on 158 CCG returns (out of a total of 209) received, submitted by CHRDs to the annual data collection

CHRDs were asked to report the number of babies with screen negative results for all 9 conditions available for communication by 6 weeks of age. In comparison to 2015 to 2016, the low number of CCG returns made in 2016 to 2017 (158 vis-à-vis 191), saw marked decreases in both the numbers of babies with screen negative results for all 9 conditions and numbers of babies with results despatched within 6 weeks of age. With the exception of the North which saw just over 20% increases in both categories, London by contrast, saw significant percentage decrease in both babies with screen negative results and babies with results despatched by 6 weeks of age; down by 64% and 51% respectively compared to 2015 to 2016.
Note that standard 1a indicates that 96.5% of results in England in 2016 to 2017 are recorded on the CHIS by 17 days of age (CCG responsibility at birth).

Conclusion

CHRD data

Data was returned by CHRDs to the annual data collection for 160 CCGs (77%) out of 209 in existence in England in 2016 to 2017. This represented a much lower response rate compared with the number of CCGs returning data last year (91%) which saw 191 CCGs out of 211 report in 2015 to 2016. In some cases data was reported for a particular CCG by more than one CHRD which added to the complexity of analysing the data. By comparison, all 209 CCGs (100%) submitted data to the national quarterly KPI data collection in 2016 to 2017. Exclusions were made if the data was incomplete.

Gaps in reporting reflects an ongoing issue with manual CHRD data returns; where non-submission of data and/or incomplete or partial data being returned, continues to cause difficulties in analysis and verification of data with providers. It reaffirms however that the KPI data is much more complete and robust as evidenced in the higher rates of coverage for standards 1a and 1b.

There also continues to be a large variety of methods used by CHRDs to receive results and a discrepancy between the number receiving and recording results using screening status codes.

Screening laboratory data

All 16 UK newborn screening laboratories returned data and incomplete data was followed up where possible. Collection of timeliness of appointment and diagnostic outcome data is an issue every year. The laboratory is reliant on the clinician that received the screen positive referral reporting the age at first appointment and the conclusive result to the screening laboratory.

All but 1 English screening laboratory reported their maternity sites are meeting the acceptable level of first samples taken on days 5 to 8 (greater than or equal to 95%). Scotland accepts day 4 samples. Samples received within 4 working days has increased overall, with 2 screening laboratories in England reaching the achievable level of 99%.

New consensus guidelines for the acceptance of the quality of blood spot cards were implemented in England and Wales in April 2015 following which the percentage of avoidable repeat rates predictably rose. Gratifyingly they fell in 2016 to 2017. As the avoidable repeat rate is a key performance indicator, maternity service providers are working hard to reduce their rates.

The current standard for timely processing of screen positive samples only applies to PKU, CHT and MCADD (England only data). The acceptable level (100%) was met for MCADD for referral initiated within 4 working days of sample receipt. Likewise the achievable level (100%) was met for MCADD for referral initiated within 3 working days of sample receipt. For PKU and CHT neither the acceptable nor the achievable levels were met in England.

Based on data reported, the acceptable standard for timeliness of first appointment for CF screen positive babies with 2 mutations (95% of babies seen by 28 days of age) was only met in Northern Ireland. England, Wales and Scotland did not meet this standard. The acceptable standard for babies with one or no mutations (80% of babies seen by 35 days of age) was not met in England or Scotland which is the same as 2015 to 2016. CF outcome data remains challenging for the laboratories to collect, however establishing closer links to CF regional centres is helping to bridge some of the inconsistencies in collating this information.

Based on data reported, the acceptable standard for timeliness of first appointment for CHT screen positive babies detected on first sample (100% by 17 days of age) was not met in England or Scotland. The acceptable standard for babies detected on second sample (100% by 24 days of age) was only met in Northern Ireland.

In England, 22% of data on CHT treatment at first appointment remains missing for babies detected on first sample (63 out of 282) and 28% of data were not reported on second sample (68 out of 240). CHT outcome data is reported by laboratories but is very incomplete and therefore not presented in this report. It is acknowledged that long-term outcome data is necessary to fully evaluate the screening programme; this is being addressed through a British Paediatric Surveillance Unit study. It reported that the performance of the NHS NBS screening programme for detecting permanent CHT was good (sensitivity 97.84%, specificity 99.98%, positive predictive value 67.36%). Out of the 16 UK newborn screening laboratories, only 6 were using the national cut-off (thyroid stimulating hormone [TSH] ≥10mU/L), the rest continue to use lower thresholds. Further analysis of the impact of the use of lower TSH cut-offs on referral rates for diagnostic investigation and diagnosis of permanent CHT is underway.

Based on data reported, the acceptable standard for timeliness of first appointment (100% by 17 days of age) was met in Northern Ireland and Scotland for PKU. For MCADD the acceptable standard was met in all 3 countries except for England which was 92%.

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