Public Health England

NHS

Data Collection and Performance Analysis Report

Newborn blood spot screening in the UK 2015/16

Public Health England leads the NHS Screening Programmes

About Public Health England

Public Health England exists to protect and improve the nation's health and wellbeing, and reduce health inequalities. It does this through world-class science, knowledge and intelligence, advocacy, partnerships and the delivery of specialist public health services. PHE is an operationally autonomous executive agency of the Department of Health.

Public Health England, Wellington House, 133-155 Waterloo Road, London SE1 8UG
Tel: 020 7654 8000 www.gov.uk/phe
Twitter: @PHE_uk Facebook: www.facebook.com/PublicHealthEngland

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 four 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.

PHE Screening, Floor 2, Zone B, Skipton House, 80 London Road, London SE1 6LH
www.gov.uk/topic/population-screening-programmes
Twitter: @PHE_Screening Blog: phescreening.blog.gov.uk

Prepared by: Tessa Morgan, Screening and Information Manager
For queries relating to this document, please contact: phe.screeninghelpdesk@nhs.net

Published March 2017
PHE publications gateway number: 2016679

Contents

Executive summary

This is the twelth annual data report for the UK's newborn blood spot screening programmes. The aim of the report is to feedback performance against the national standards.

Data was returned by child health records departments (CHRDs) for 196 (93%) clinical commissioning groups (CCGs) out of the 209 that existed in England in 2015/16. Exclusions were made if the data was incomplete. All 16 UK newborn screening laboratories returned data and incomplete data was followed up where possible.

In England, coverage measured at 17 days (CCG responsibility at birth) was reported at 94.25%. Standard 1b introduces an effective timeframe of 21 calendar days for movers in. In England, coverage for movers in with the timeframe applied was 78.33% and without the timeframe was 89.78%.

Declines reported by the CHRDs were 9 per 10,000 (CCG responsibility at birth) and 246 per 10,000 (movers in). In England, 99.73% of blood spot cards included the baby's NHS number, and 75.79% 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 despite the investment made in funding trusts to purchase printers and scanners.

In the UK, 95.73% of samples were taken on days 5-8. 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 concensus quality guidelines in April 2015. Since implementation, monthly and quarterly (KPI) collection of avoidable repeat data shows 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. The Newborn Blood Spot (NBS) Screening Programme will continue to collect monthly avoidable repeat data until all screening laboratories have implemented status codes and subcodes into the Newborn Blood Spot Failsafe Solution (NBSFS). The NBSFS must also be configured to accurately report this data for babies in the location where they are screened. This will enable avoidable repeat data to be collected via failsafe.

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

The acceptable standard for timeliness of first appointment for CF screen positive babies with two mutations was met in England, Northern Ireland and Wales. The acceptable standard for one or no mutations was met in Northern Ireland and Wales. This data is based on babies with age at first appointment reported. It is apparent from data supplied by the laboratories that the

requirement for a second blood spot test to be taken for babies with one or no mutations causes delay in time to appointment and a smaller proportion of these babies meet the standard.

Northern Ireland was the only country to meet the acceptable standard for CHT screen positive babies detected on first or second sample. This data is based on babies with age at first appointment reported.

A national surveillance study has been undertaken through the British Paediatric Surveillance Unit (BPSU). This study has now collected three years of follow-up data on all UK infants identified with a presumed-positive newborn blood spot screening test result for congenital hypothyroidism (CHT), or who were aged under 5 years and referred to a paediatrician for investigation after a clinical diagnosis of CHT between 1 June 2011 and 30 June 2012. During 13 months of surveillance, 676 infants (62% girls) were identified after a presumptive-positive newborn screening result and 22 further children (50% girls) presented clinically. Of these 462 children had permanent CHT, which was defined as a requirement for treatment with thyroxine replacement therapy after three years of follow-up. One-third of children in whom CHT was excluded by three years were commenced on treatment at diagnosis but this was later discontinued. Based on this study, the incidence of permanent CHT was approximately 5 per 10,000 live births.

Acknowledgements

The NHS Newborn Blood Spot Screening Programme would to thank the UK newborn screening laboratories and child health records departments (CHRDs) that submitted 2015/16 data. The programme would also like to thank the UK screening programme colleagues for their contributions.

Introduction

Background

This is the twelth annual data report for the UK's newborn blood spot (NBS) screening programmes. 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 six 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.

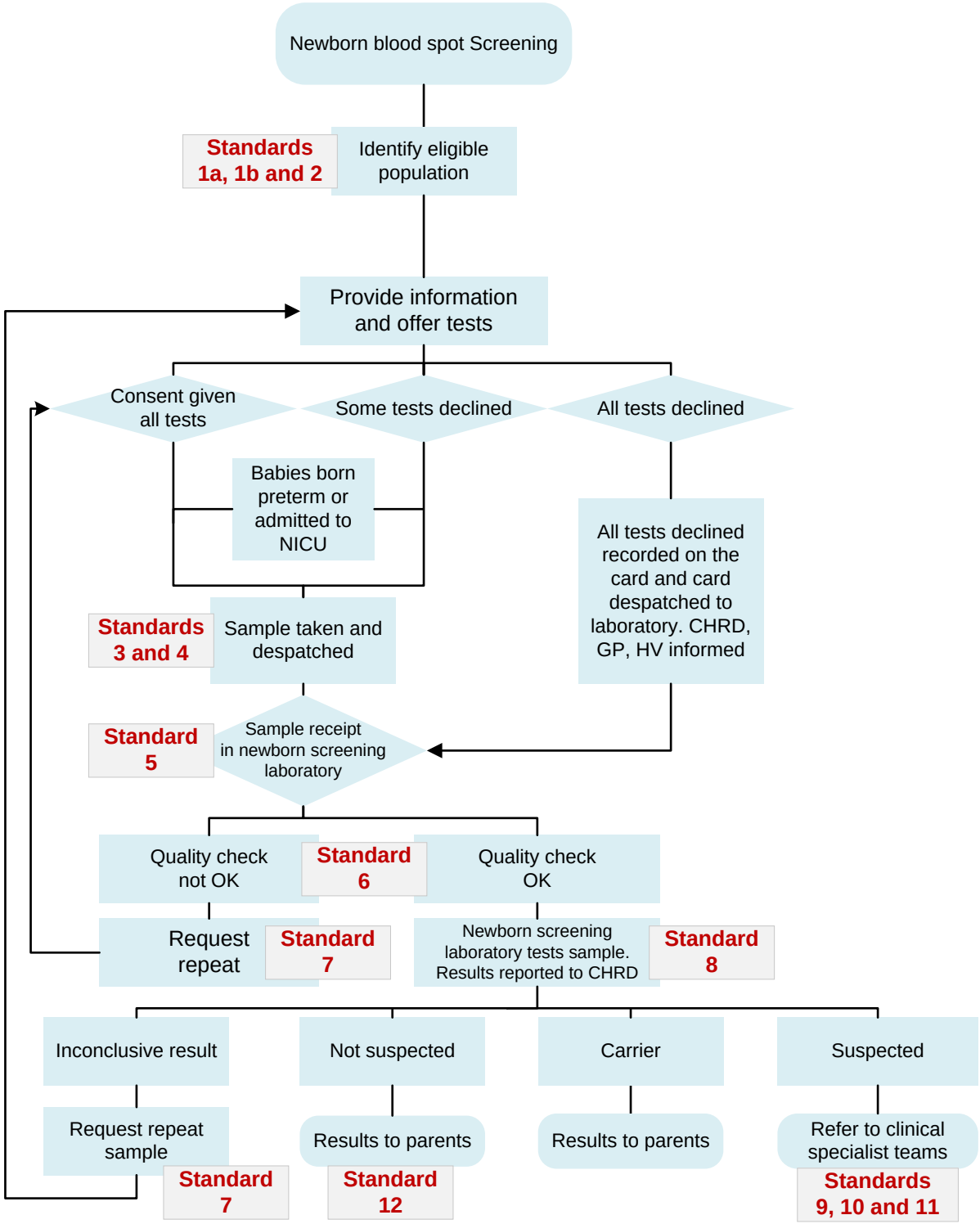
One of the objectives of the NHS NBS Screening Programme is to set national standards (see Table 1 and Figure 1)[1-2]. National standards are important to support the delivery and quality assurance of the screening programme and are used by local commissioners and quality improvement groups. The aim 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)	CHRD
Standard 1b: Completeness of coverage (movers in)	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: www.gov.uk/government/collections/newborn-blood-spot-screening-programme-standards-and-data.

Figure 1: NBS standards mapped to screening pathway



Methodology

Data is 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.

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 is presented by fiscal year (1 April – 31 March) unless stated otherwise. The year '2014/15', for example, refers to the financial year 1 April 2014 to 31 March 2015.

- data on standards 1a, 1b, 2 and 12 is 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 is 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 catchment area

- data on standard 7 is not currently collected

- data on standard 9 is returned by newborn screening laboratories per laboratory catchment area and presented by condition

- data on standard 11 (including diagnostic outcome data) is 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) is in the process of being published at www.ukas.com

Completeness of data

Data was returned by CHRDs for 196 CCGs (93%) out of the 209 that existed in 2015/16 in England. There were 27 exclusions made as the data was incomplete (for example the numerator or denominator was missing) or the quality of the data was poor. Some CHRDs reported that the data provided by their informations system was inaccurate and therefore agreed to withdraw their data from publication. Work is ongoing to rectify these IT issues.

All 16 UK newborn screening laboratories returned data and incomplete data was followed up where possible. 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 and are the logical place to capture follow-up and outcome data. 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 2015/16 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.

Analysis of screening performance

Overview of UK national screening figures

SCD	
Babies tested	**779,448**
Screened positive	**272**
Incidence*	**2,200**

PKU	
Babies tested	**785,653**
Screened positive	**115**
Incidence*	**8,400**

CF	
Babies tested	**780,221**
Screened positive	**339**
Incidence*	**2,500**

MCADD	
Babies tested	**785,654**
Screened positive	**82**
Incidence*	**10,200**

CHT	
Babies tested	**785,757**
Screened positive	**655**
Incidence*	**1,500**

MSUD, IVA, GA1, HCU	
Babies tested	**705,808**
Screened positive	**26**
Incidence	
MSUD & GA1	172,000
IVA	120,000
HCU	300,000

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.

*Incidence per 10,000 babies base upon screening data collected from 2005 onwards.

Coverage	
Percentage of babies with a conclusive result for PKU recorded on the CHIS by 17 days of age	**94.3%** (England) **99.1%** (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 2015/16

Laboratory	SCD		CF		CHT	
	Number tested	Number of screen positives	Number tested	Number of screen positives	Number tested	Number of screen positives
Bristol	40,249	2	40,249	30	40,249	44
Cambridge	28,190	6	27,995	9	28,190	24
GOSH	120,958	105	120,097	28	121,106	158
Leeds	43,573	11	43,182	15	43,182	36
Liverpool	28,300	0	28,901	16	28,951	36
Manchester	55,569	13	55,407	25	55,721	34
Newcastle	33,035	1	33,177	12	33,245	41
Oxford	29,002	12	29,602	10	29,602	18
Portsmouth	37,142	6	37,418	20	38,477	26
SE Thames	55,493	49	56,732	22	57,030	27
Sheffield	72,703	18	72,115	45	72,703	47
SW Thames	51,845	20	51,611	17	53,459	42
West Midlands	70,382	22	70,957	28	70,957	47
England	**666,441**	**265**	**667,443**	**277**	**672,872**	**580**
Northern Ireland	24,394	0	24,291	16	24,398	18
Scotland	55,571	3	55,445	32	55,445	31
Wales	33,042	4	33,042	14	33,042	26
UK total	**779,448**	**272**	**780,221**	**339**	**785,757**	**655**

Data source: Newborn screening laboratories

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

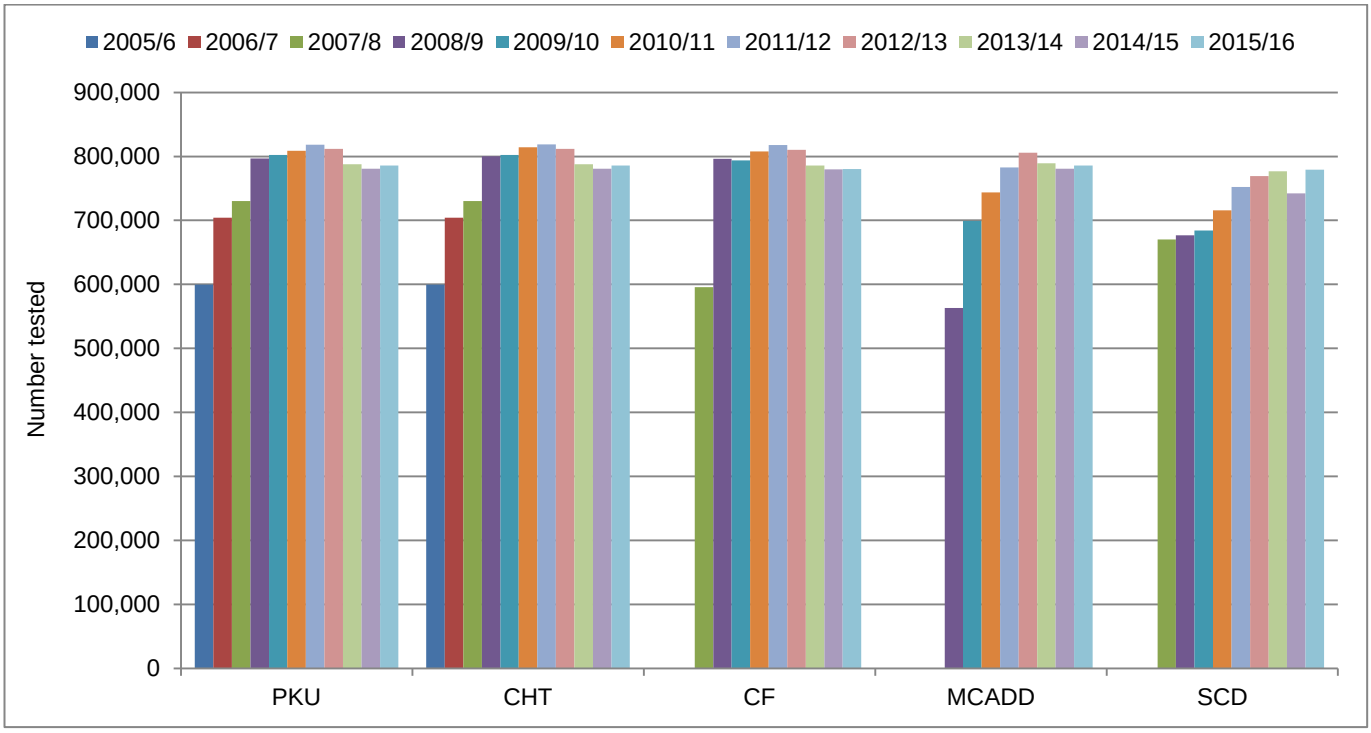
Note that a significant proportion of screen positive results will not be confirmed cases.

Table 3: Number of UK babies tested and number of screen positive results for IMDs 2015/16

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	40,249	3	40,249	2	40,249	2
Cambridge	28,190	10	28,190	3	28,190	0
GOSH	121,106	11	121,106	8	121,106	3
Leeds	43,182	8	43,182	6	43,182	0
Liverpool	28,951	1	28,951	4	28,951	0
Manchester	55,721	12	55,721	9	55,721	3
Newcastle	33,245	6	33,245	3	33,245	2
Oxford	29,602	4	29,602	5	29,602	1
Portsmouth	38,371	3	38,371	4	38,371	3
SE Thames	57,030	6	57,030	5	57,030	2
Sheffield	72,703	14	72,703	13	72,703	5
SW Thames	53,459	5	53,459	6	53,459	2
West Midlands	70,957	4	70,957	8	70,957	2
England	**672,766**	**87**	**672,766**	**76**	**672,766**	**25**
Northern Ireland	24,400	14	24,401	2	0	0
Scotland	55,445	6	55,445	1	0	0
Wales	33,042	8	33,042	3	33,042	1
UK total	**785,653**	**115**	**785,654**	**82**	**705,808**	**26**

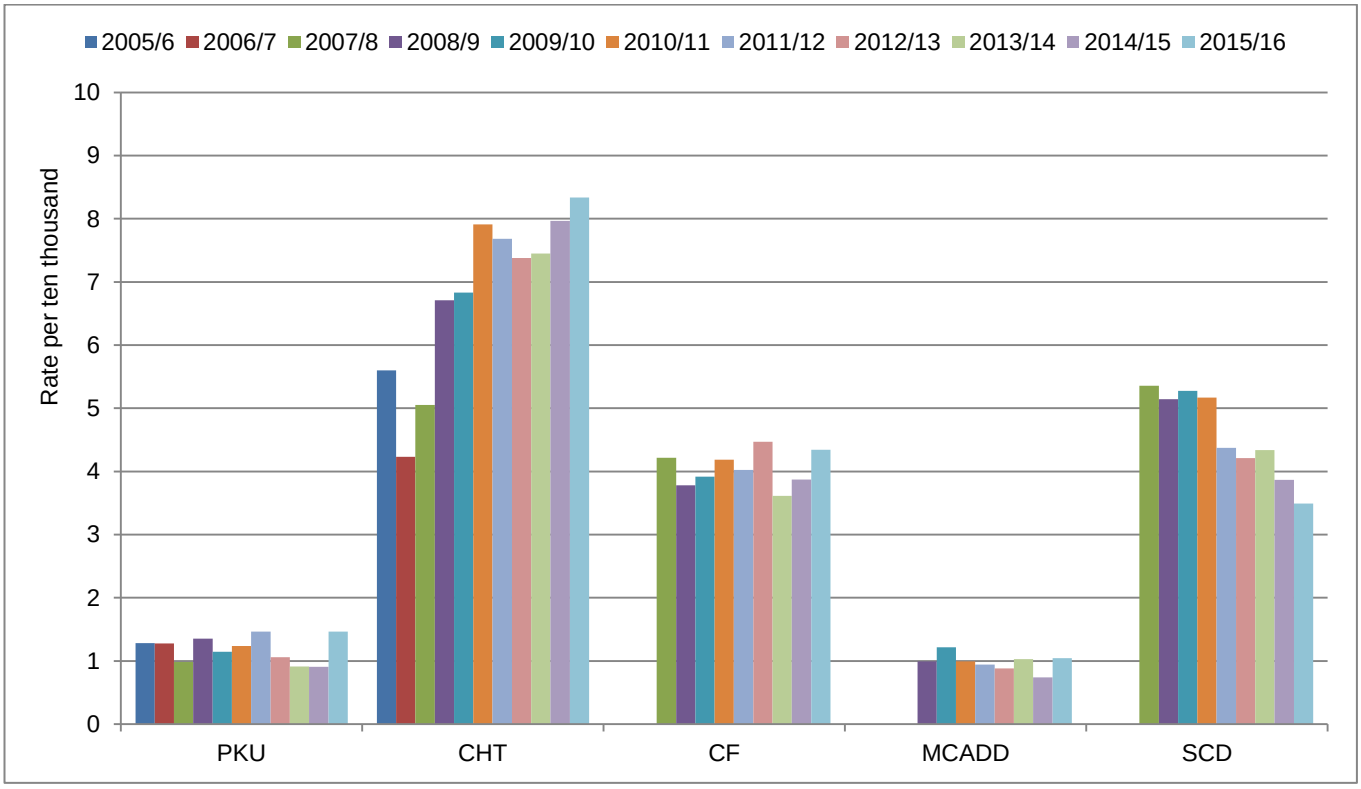
Data source: Newborn screening laboratories

Figure 2: Number of UK babies tested for SCD, CF, CHT, PKU and MCADD 2005/16



Data source: Newborn screening laboratories

Figure 3: Rate per ten thousand of UK babies screened positive for SCD, CF, CHT, PKU and MCADD 2005-16



Data source: Newborn screening laboratories

The antenatal sickle cell and thalassaemia programme affects birth prevalence. Data collected over an 8 year period shows that in PND tests with a sickle cell affected diagnosis approximately 66% opted to terminate, compared to 82% with a beta thalassaemia diagnosis and 88% with an alpha thalassaemia diagnosis[4]. Increased awareness of risks before conception may be increasing demand for pre-implantation genetic diagnosis.

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: ≥ 95.0% all tests
Achievable level: ≥ 99.9% PKU, MCADD, SCD
Achievable level: ≥ 98% CF, CHT

Coverage is measured at the time of the report (there is a two month period allowed for data return) and at 17 days of age. PKU is used as a proxy for all conditions. There were196 CHRD returns submitted for 2015/16. Of these 24 were omitted for Standard 1a and 28 were omitted for Standard 1b due to innacurate or incomplete submissions.

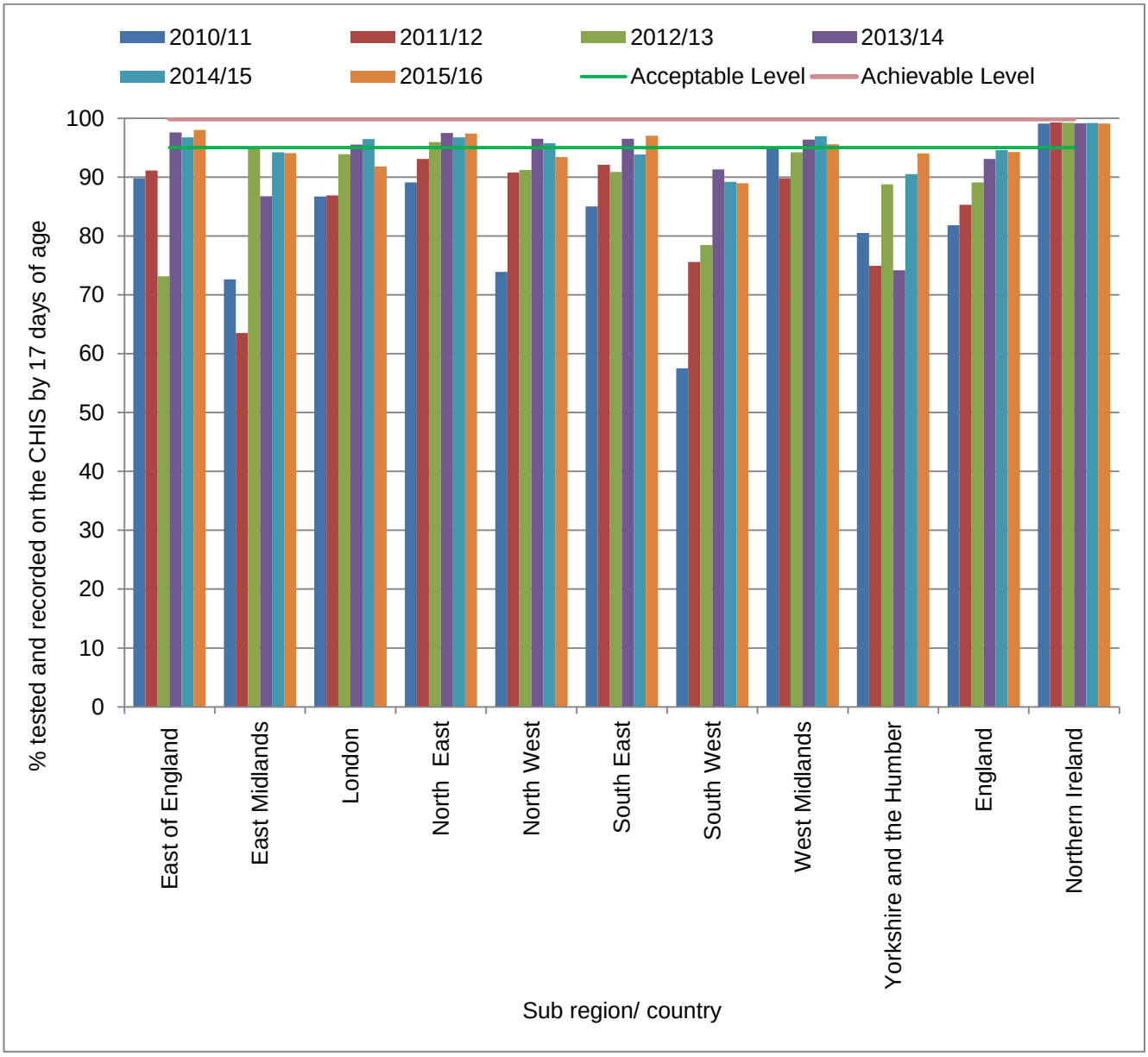
Table 4: Completeness of coverage for PKU (CCG responsibility at birth) 2015/16

Region/country	Babies for whom the CCG/ country is responsible	Babies tested for PKU		Babies for whom the CCG/ country is responsible	Babies with a conclusive result for PKU recorded by 17 days of age	
	n	n	%	n	n	%
England	**504,203**	**499,443**	**99.06**	**476,037**	**448,681**	**94.25**
North Region	**139,549**	**139,433**	**99.92**	**139,549**	**131,343**	**94.12**
North East	15,226	15,215	**99.94**	15,226	14,828	97.39
North West	63,495	63,446	**99.92**	63,495	59,325	93.43
Yorkshire & The Humber	60,828	60,772	**99.91**	60,828	57,190	94.02
South Region	**101,930**	**101,576**	**99.65**	**101,930**	**95,477**	**93.67**
South Central	40,701	40,623	**99.81**	40,701	40,024	98.34
South East	12,371	12,279	**99.26**	12,371	12,001	97.01
South West	48,858	48,674	**99.62**	48,858	43,452	88.94
Midlands & East Region	**158,248**	**157,089**	**99.27**	**158,248**	**151,812**	**95.93**
East Midlands	54,035	53,978	**99.89**	54,035	50,829	94.07
East of England	57,452	57,388	**99.89**	57,452	56,298	97.99
West Midlands	46,761	45,723	**97.78**	46,761	44,685	95.56
London Region	**104,476**	**101,345**	**97.00**	**76,310**	**70,049**	**91.80**
Northern Ireland	**23,858**	**23,840**	**99.92**	**23,858**	**23,644**	**99.10**

Data source: CHRDs

Maternity sites now use the Newborn Blood Spot Failsafe Solution (NBSFS) to ensure all babies born in England are offered screening. The responsibility for ensuring completeness of coverage remains with the CHRD.

Figure 4: Completeness of coverage for PKU (CCG responsibility at birth) 2010/16 (measured at 17 days)



Data source: CHRDs

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: ≥ 95% of eligible babies are tested for PKU
Achievable level: ≥ 99.9% of eligible babies are tested for PKU

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 – 2014-16 data is presented with the timeframe in addition to year-on-year data without the timeframe.

Table 5: Completeness of coverage for PKU (movers in) 2015/16

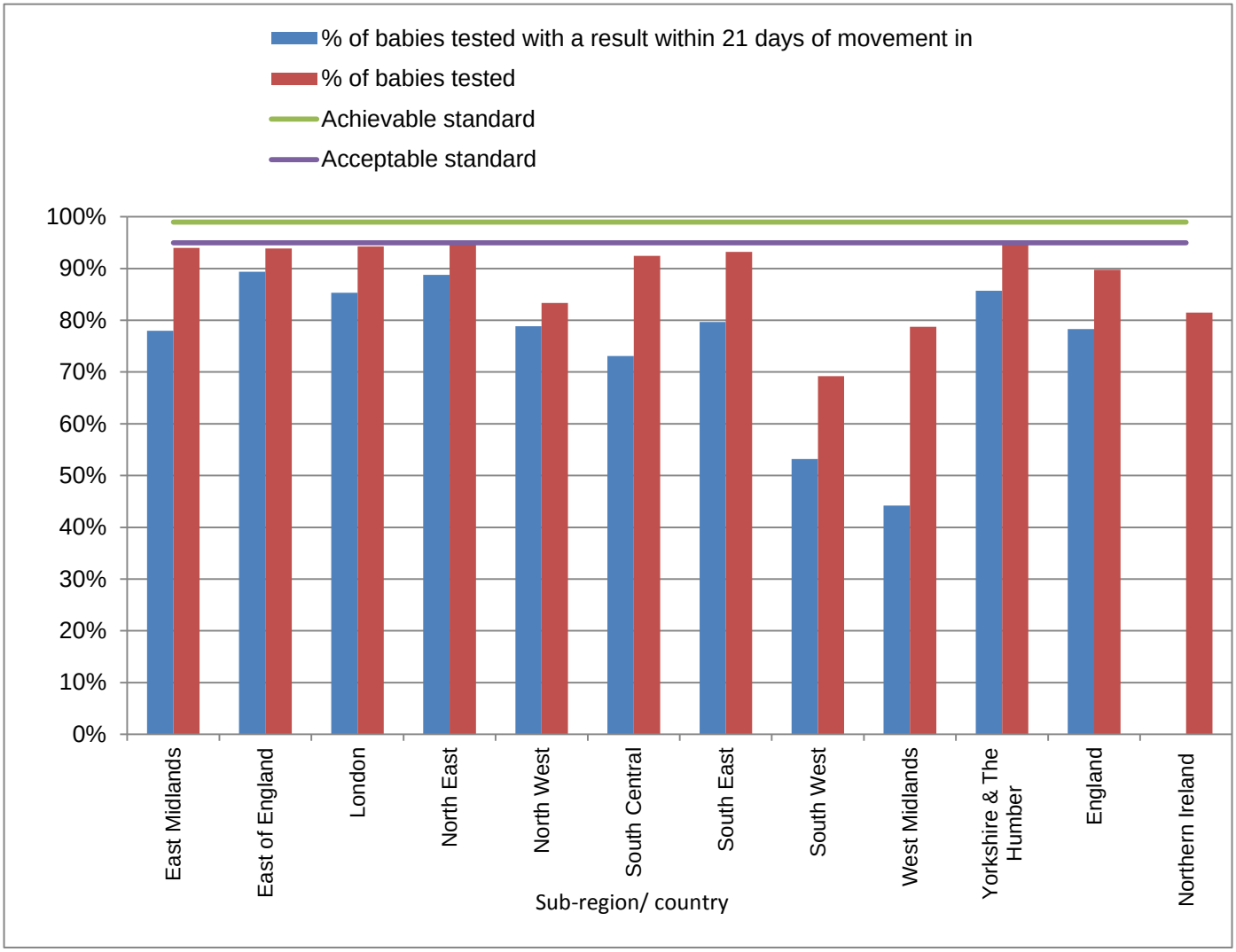
Region/country	Babies for whom the CCG/ country is responsible*	Babies tested for PKU		Babies for whom the CCG/ country is responsible*	Babies with a conclusive result for PKU** recorded by 21 calendar days	
	n	n	%	n	n	%
England	**25,482**	**22,877**	**89.78**	**25,367**	**19,869**	**78.33**
North Region	**8,808**	**7,862**	**89.26**	**8,808**	**7,273**	**82.57**
North East	819	775	**94.63**	819	727	88.77
North West	4,400	3,667	**83.34**	4,400	3,469	78.84
Yorkshire & The Humber	3,589	3,420	**95.29**	3,589	3,077	85.73
South Region	**4,212**	**3,590**	**85.23**	**4,212**	**2,843**	**67.50**
South Central	2,462	2,276	**92.45**	2,462	1,799	73.07
South East	428	399	**93.22**	428	341	79.67
South West	1,322	915	**69.21**	1,322	703	53.18
Midlands & East Region	**7,044**	**6,319**	**89.71**	**7,044**	**5,229**	**74.23**
East Midlands	1,640	1,541	**93.96**	1,640	1,279	77.99
East of England	3,454	3,243	**93.89**	3,454	3,088	89.40
West Midlands	1,950	1,535	**78.72**	1,950	862	44.21
London Region	**5,418**	**5,106**	**94.24**	**5,303**	**4,524**	**85.31**
Northern Ireland	**356**	**290**	**81.46**	**Not reported**		

Data source: CHRDs

*27 returns were excluded based on missing or inaccurate data. **Status codes 04, 07, 08[3].

In England, processes for identifying and offering screening for movers in vary between regions.

Figure 5: Completeness of coverage for PKU (movers in) 2015-16 and babies tested (no timeframe applied)



Data source: CHRDs

Declined screening

Parents can choose not to have their baby screened. Data on declined screening is collected and reported alongside coverage data to aid interpretation. PKU is used as a proxy for all conditions. It is difficult to draw any conclusions about the data due to the small numbers of declines reported and differences in local processes for recording declines. Over the last 4 years there has been no significant change in the overall rate of declines in England and no clear patterns have emerged within regions. Year-on-year data is therefore not presented.

Declines: CCG responsibility at birth

Table 6: Number and rate per ten thousand babies of declines for PKU (CCG responsibility at birth) 2015/16

Region/country	Babies for whom the CCG/country is responsible	Declined screening for PKU	
	n	n	Rate per ten thousand
England	**476,037**	**405**	**8.51**
North Region	**139,549**	**77**	**5.52**
North East	15,226	8	**5.25**
North West	63,495	38	**5.98**
Yorkshire & The Humber	60,828	31	**5.10**
South Region	**101,930**	**106**	**10.40**
South Central	40,701	30	**7.37**
South East	12,371	7	**5.66**
South West	48,858	69	**14.12**
Midlands & East Region	**158,248**	**113**	**7.14**
East Midlands	54,035	50	**9.25**
East of England	57,452	33	**5.74**
West Midlands	46,761	30	**6.42**
London Region	**76,310**	**109**	**14.28**
Northern Ireland	**23,858**	**16**	**6.71**

Data source: CHRDs

19 CCGs reported greater than 10 declines per ten thousand and more than 5 declines in total.

49 CCGs reported no declines. We have not been able to determine if this data is accurate (i.e. there were no true declines) or if it reflects local recording/reporting processes.

Declines: movers in

Table 7: Number and rate per ten thousand babies of declines for PKU (movers in) 2015/16

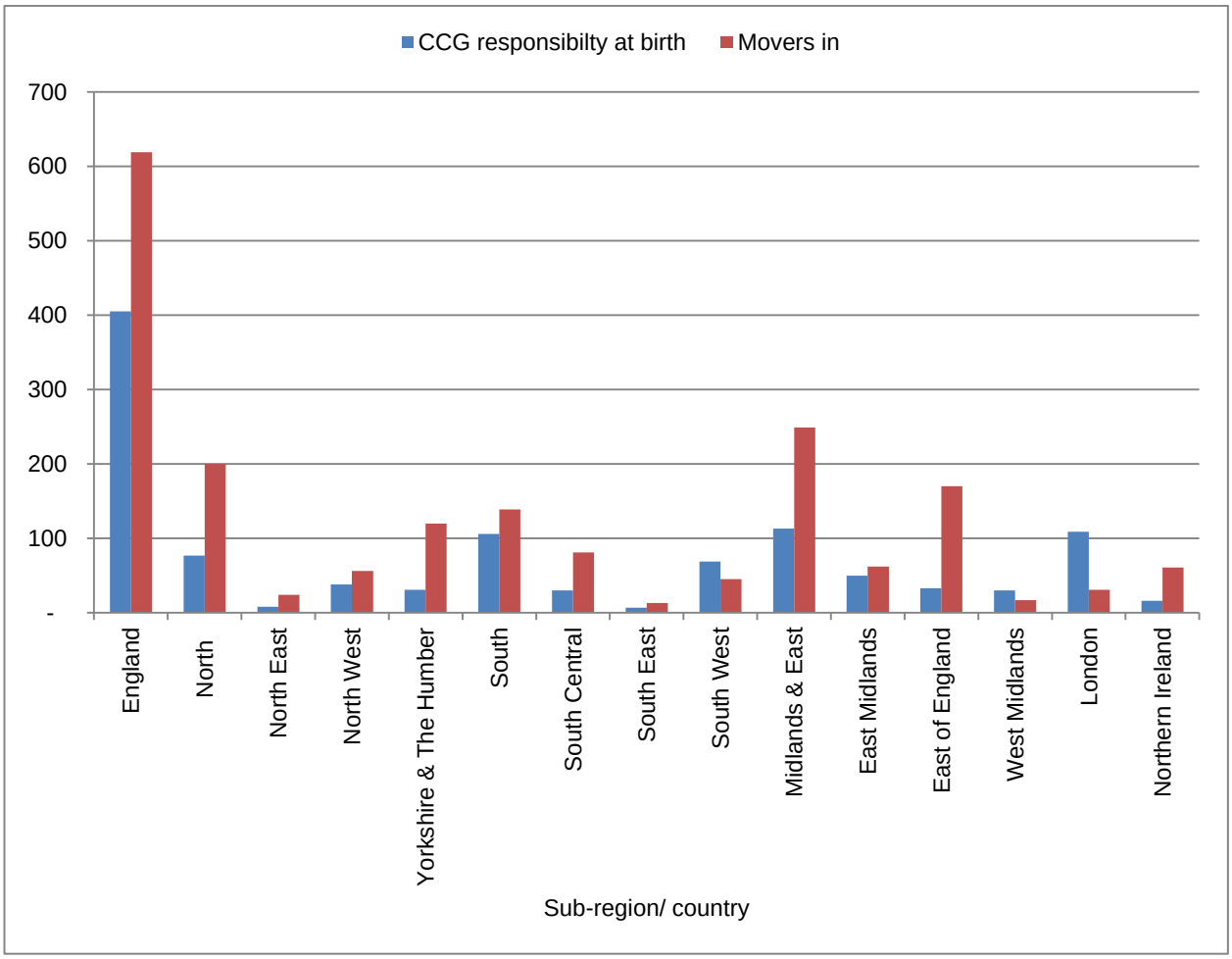
Region/country	Babies for whom the CCG/country is responsible	Declined screening for PKU	
	n	n	Rate per ten thousand
England	**25,162**	**619**	**246**
North	**8,728**	**200**	**229**
North East	739	24	**325**
North West	4,400	56	**127**
Yorkshire & The Humber	3,589	120	**334**
South	**4,212**	**139**	**330**
South Central	2,462	81	**329**
South East	428	13	**304**
South West	1,322	45	**340**
Midlands & East	**6,919**	**249**	**360**
East Midlands	1,640	62	**378**
East of England	3,454	170	**492**
West Midlands	1,825	17	**93**
London	**5,303**	**31**	**58**
Northern Ireland	356	61	1713

Data source: CHRDs

Northern Ireland has a higher rate of declines amongst movers in because all babies under a year of age that move in are offered screening – a proportion of parents will decline if they believe that their child has already been screened. In England these babies are only offered screening if they do not have documented results.

Declines: Comparing CCG responsibility at birth and movers in populations

Figure 6: Number of declines for PKU – CCG responsibility at birth and movers in populations 2015/16



Data source: CHRDs

CHRD process data

Table 8: Receipt, recording and despatch of results by CHRDs 2015/16 (reported per CCG)

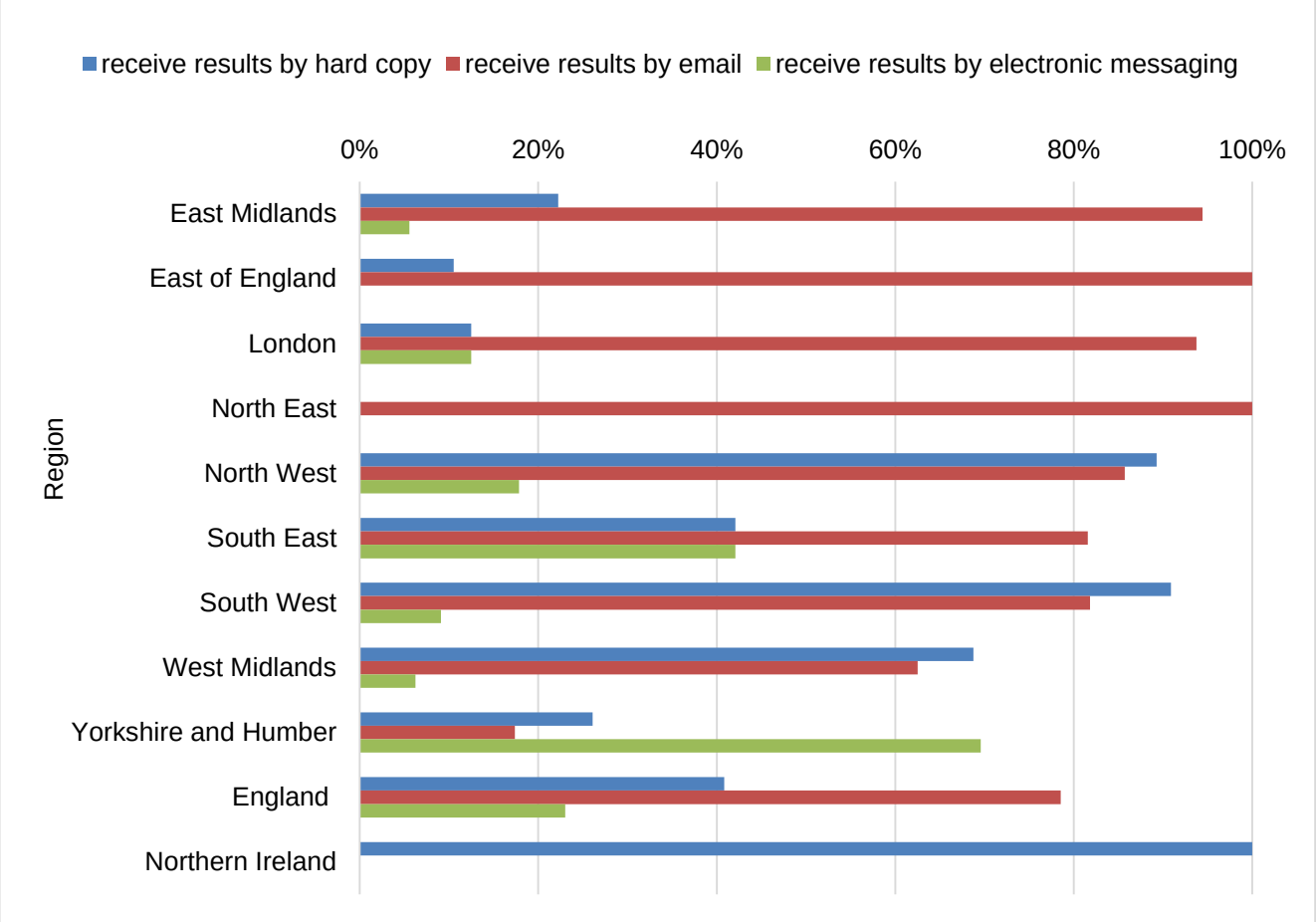
Region/ country	Number of CHRDs* that:												total number of returns
	receive results by hard copy		receive results by email		receive results by electronic messaging		receive results with status codes		record results using status codes		send letters directly to parents when 04** is reported on all conditions		
	n	%	n	%	n	%	n	%	n	%	n	%	n
East Midlands	4	22.22	17	94.44	1	5.56	18	100	18	100	18	100	18
East of England	2	10.53	19	100	0	0.00	19	100	19	100	19	100	19
London	4	12.50	30	93.75	4	12.50	31	96.88	32	100	24	75	32
North East	0	0.00	6	100	0	0.00	6	100	6	100	6	100	6
North West	25	89.29	24	85.71	5	17.86	28	100	27	96.43	16	57.14	28
South East	16	42.11	31	81.58	16	42.11	38	100	38	100	21	55.26	38
South West	10	90.91	9	82	1	9	11	100	11	100	8	72.73	11
West Midlands	11	68.75	10	62.50	1	6.25	12	75.00	12	75.00	15	93.75	16
Yorkshire and Humber	6	26.09	4	17.39	16	69.57	16	69.57	16	69.57	23	100	23
England	**78**	**40.84**	**150**	**78.53**	**44**	**23.04**	**179**	**94**	**179**	**93.72**	**150**	**78.53**	**191**
Northern Ireland	1	100	0	0.00	0	0.00	1	100	1	100	0	0	1

Data source: CHRDs

*Note that some CHRDs might be double counted as data is returned per CCG not CHRD.

**Status code 04 – condition screened for not suspected[3].
The data highlights the multiplicity of methods used by CHRDs to receive results and a discrepancy between the number receiving and recording results using status codes – full use of electronic messaging will enable greater efficiency.

Figure 7: Percentage of CHRDs who receive results by hard copy, email and electronic messaging 2015/16



Data source: CHRDs

Note that some CHRDs might be double counted as data is returned per CCG not CHRD.

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.

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 five 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 five conditions, for all babies equal to or more than 14 days and equal to or less than 364 days.

CHRDs were asked if they performed daily checks for missing results at 17 days, 14 days or used a different search strategy.

Table 9: Number and percentage of CHRDs that search for missing results at 17 days, 14 days and 'other' 2015/16

Region/country	Number reaching the acceptable standard		Number reaching the achievable standard		Number of non compliant		Number not reported	
	n	%	n	%	n	%	n	
East Midlands	16	80.00	11	55.00	2	10.00	1	5.00
East of England	19	100.00	16	84.21	0	0.00	0	0.00
London	32	100.00	27	84.38	0	0.00	0	0.00
North East	6	60.00	4	40.00	0	0.00	4	40.00
North West	22	66.67	20	60.61	3	9.09	3	9.09
South East	32	84.21	28	73.68	4	10.53	2	5.26
South West	10	83.33	10	83.33	1	8.33	1	8.33
West Midlands	15	68.18	9	40.91	1	4.55	6	27.27
Yorkshire & The Humber	15	65.22	9	39.13	1	4.35	0	0.00
England	167	80.38	134	64.11	12	5.74	17	8.13
Northern Ireland **	1	100.00	1	100.00	0	0.00	0	0.00

Data source: CHRDs
*Note that some CHRDs might be double counted as data is returned per CCG not CHRD.
** Northern Ireland carry out weekly checks for babies aged 11-364 days.

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 10: Use of the baby's NHS number and bar-coded label 2015/16

Laboratory	Number of all samples (including repeats)	Blood spot cards including baby's NHS number		Blood spot cards including baby's NHS number on a bar-coded label	
	n	n	%	n	%
Bristol	44635	44475	99.64%	39872	89.33%
Cambridge	29841	29733	99.64%	14221	47.66%
GOSH	129616	129297	99.75%	**	0.00%
Leeds	45584	45371	99.53%	33321	73.10%
Liverpool	30704	30652	99.83%	18509	60.28%
Manchester	60158	59951	99.66%	43815	72.83%
Newcastle	35566	35410	99.56%	25276	71.07%
Oxford	32441	32372	99.79%	24006	74.00%
Portsmouth	38723	38627	99.75%	27155	70.13%
SE Thames	61591	61512	99.87%	49649	80.61%
Sheffield	78163	77853	99.60%	63464	81.19%
SW Thames	55530	55472	99.90%	42481	76.50%
West Midlands	73875	73759	99.84%	62986	85.26%
England	716,427	714,484	99.73%	433,188	75.79%
Northern Ireland*					
Scotland	58765	58187	99.02%	0	0.00%
Wales	36828	36324	98.63%	0	0.00%
UK	812,020	795,353	99.63%	433,188	75.79%

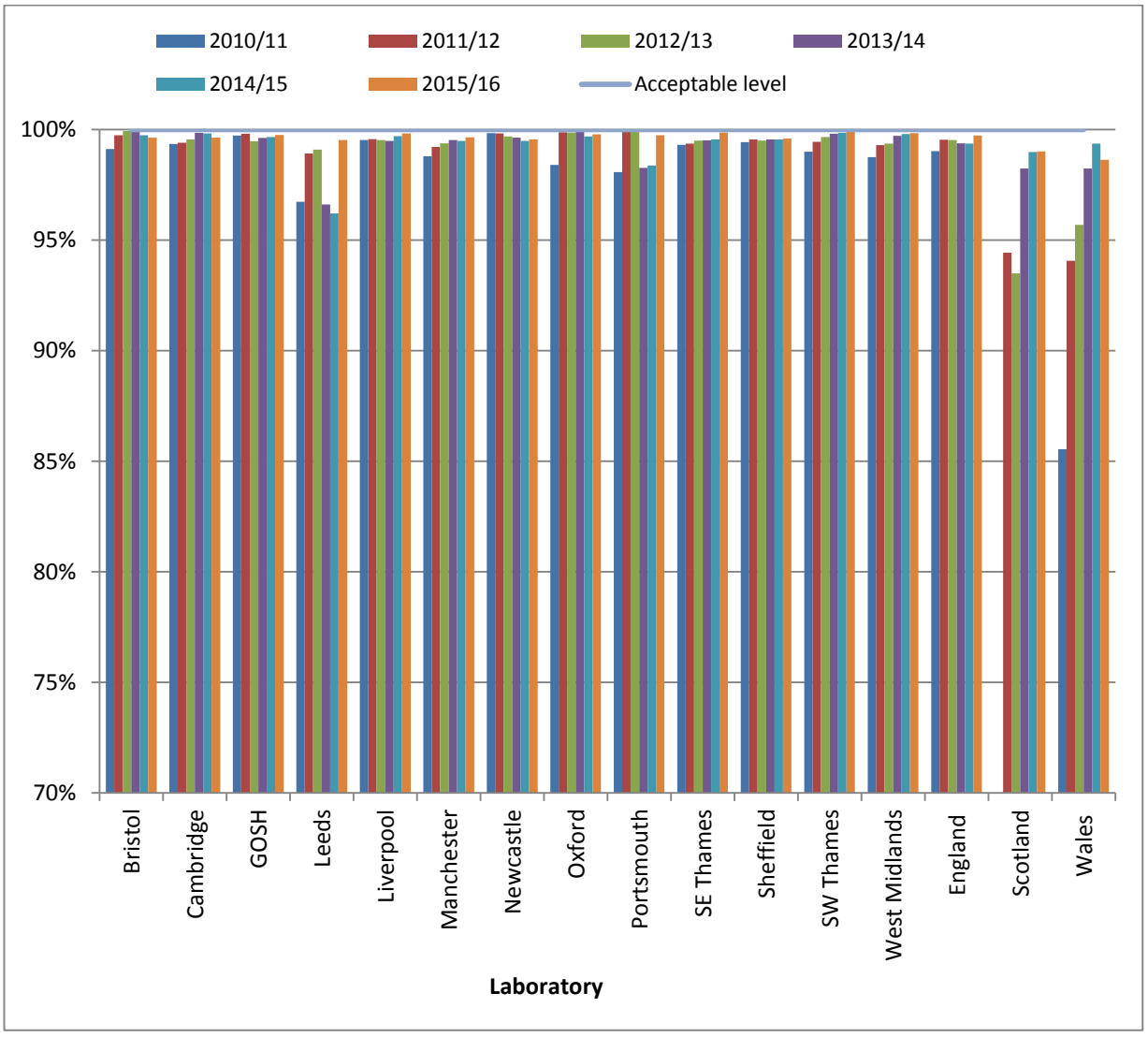
Data source: Newborn screening laboratories

*The Health + Care number (Northern Ireland equivalent to NHS number) is currently recorded on blood spot cards and plans are underway for the regional screening laboratory to routinely capture and report on use of the number.

**GOSH unable to report data due to laboratory information management system limitations.

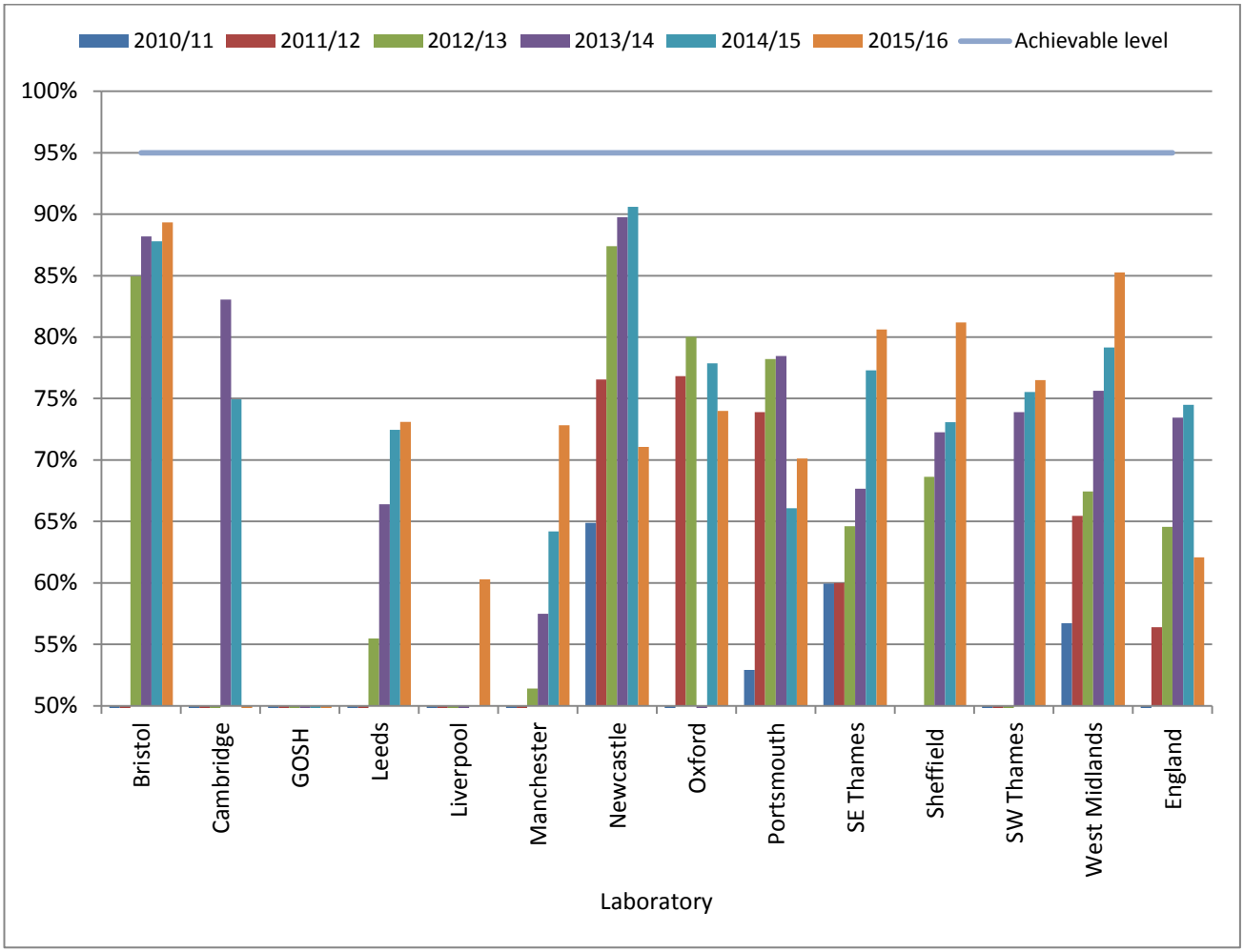
It has been reported that some maternity units are using bar coded labels which are not compliant with the national specification and therefore not counted in the data.

Figure 8: Percentage of blood spot cards including the baby's NHS number (or UK equivalent) 2010/16



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

Figure 9: Percentage of blood spot cards including a bar-coded NHS number (or UK equivalent) 2010/16



Data source: Newborn screening laboratories

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 5-8 (ideally on day 5).

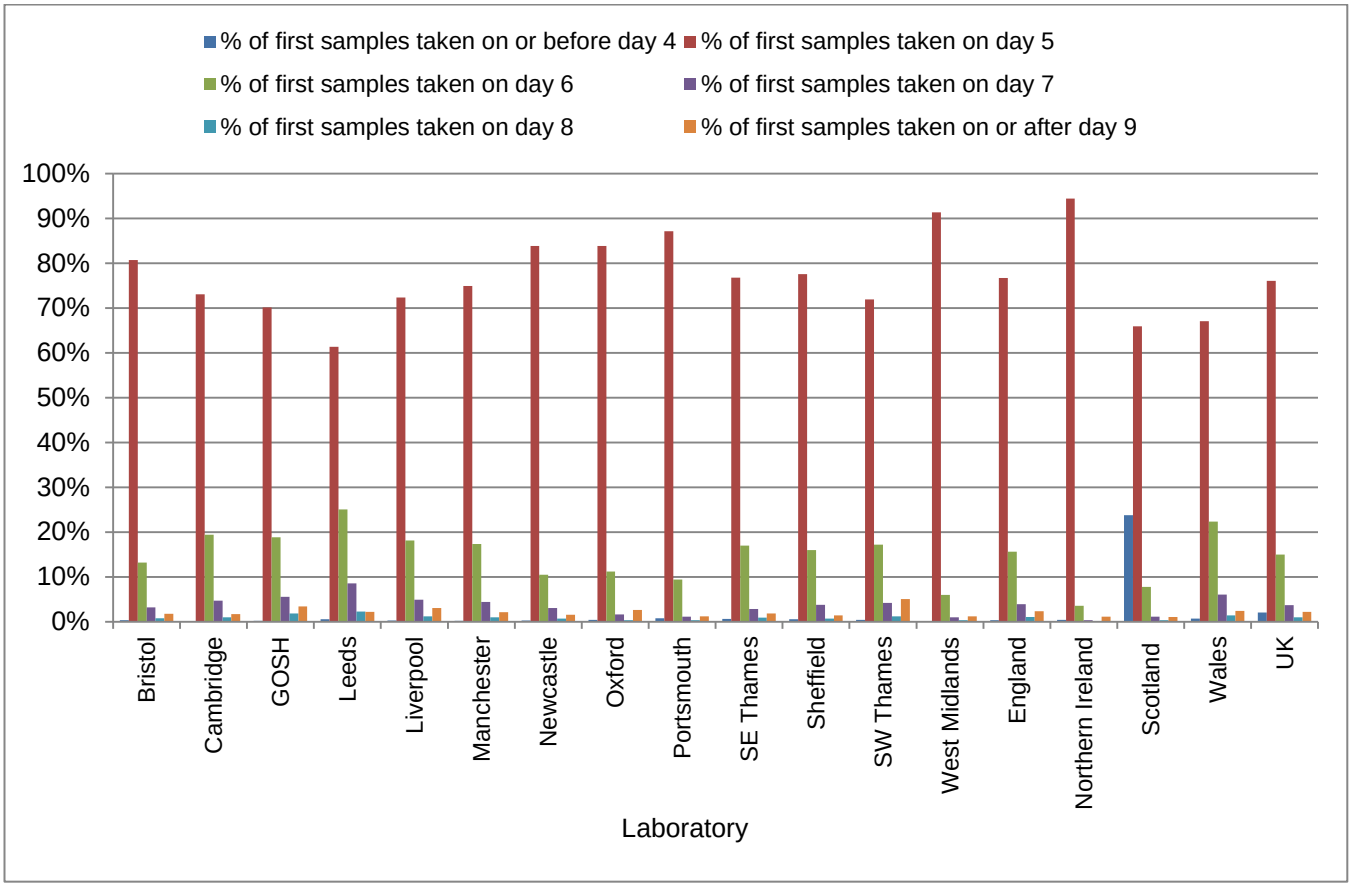
Achievable level
Equal to or greater than 99% of first samples taken on days 5-8 (ideally on day 5).

Table 11: Day of first sample collection 2015/16

	First samples taken:							
	on or before day 4		on day 5		on days 5 to 8		on or after day 9	
Laboratory	n	%	n	%	n	%	n	%
Bristol	141	0.35	32,564	80.73	39,481	97.88	716	1.78
Cambridge	33	0.12	20,437	73.08	27,460	98.19	473	1.69
GOSH	285	0.23	85,355	70.12	117,302	96.37	4,138	3.40
Leeds	249	0.57	26,698	61.31	42,331	97.22	963	2.21
Liverpool	82	0.28	20,988	72.37	28,037	96.68	880	3.03
Manchester	107	0.19	41,679	74.90	54,352	97.68	1,184	2.13
Newcastle	101	0.30	27,883	83.87	32,622	98.13	522	1.57
Oxford	110	0.40	23,267	83.82	26,923	96.99	725	2.61
Portsmouth	275	0.75	32,103	87.16	36,110	98.04	448	1.22
SE Thames	364	0.64	43,778	76.76	55,596	97.49	1,070	1.88
Sheffield	384	0.53	55,911	77.54	70,703	98.06	1,015	1.41
SW Thames	220	0.40	39,711	71.95	52,198	94.57	2,775	5.03
West Midlands	109	0.16	63,504	91.33	68,567	98.62	853	1.23
England	**2,460**	**0.37**	**513,878**	**76.71**	**651,682**	**97.28**	**15,762**	**2.35**
Northern Ireland	108	0.44	23,066	94.42	24,049	98.44	273	1.12
Scotland*	13,178	23.80	36,490	65.91	41,609	75.15	580	1.05
Wales	227	0.68	22,246	67.04	32,149	96.89	805	2.43
UK	**15,973**	**2.04**	**595,680**	**76.09**	**749,489**	**95.73**	**17,420**	**2.23**

Data source: Newborn screening laboratories
*Scotland allows samples to be taken on day 4.

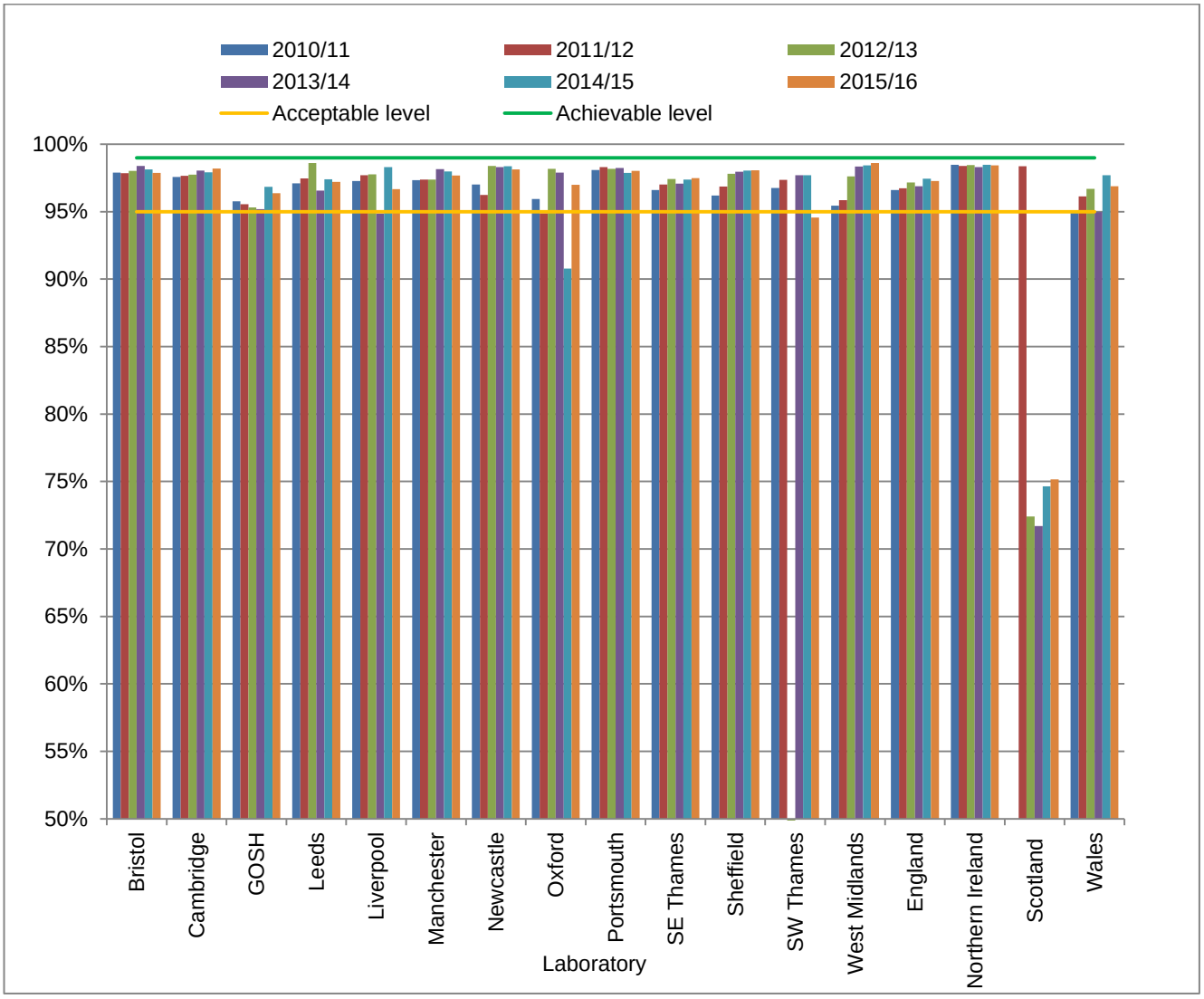
Figure 10: Day of first sample collection 2015/16



Data source: Newborn screening laboratories

Data on areas returning the lowest and highest percentage of samples taken on days 5-8 has not been presented this year.

Figure 11: Percentage of samples taken on days 5-8 2010/16



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 four working days of sample collection.

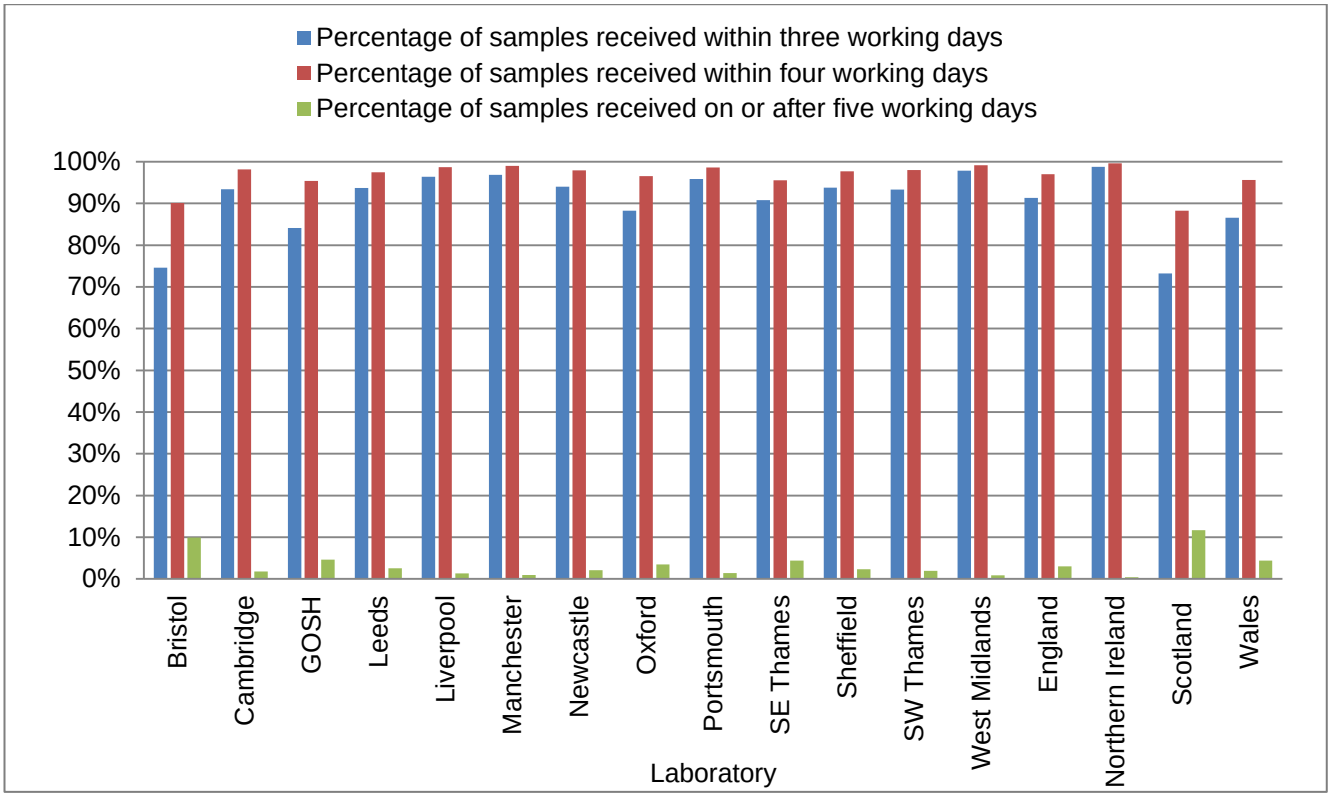
Achievable level
Equal to or greater than 99% of all samples received within three working days of sample collection.

Table 12: Number of working days taken to receive sample 2015/16

Laboratory	Samples received:					
	within 3 working days		within 4 working days		on or after 5 working days	
	n	%	n	%	n	%
Bristol	28,615	74.57	34,577	90.11	3,797	9.89
Cambridge	27,537	93.44	28,931	98.17	540	1.83
GOSH	108,843	84.15	123,351	95.37	5,994	4.63
Leeds	42,704	93.68	44,418	97.44	1,166	2.56
Liverpool	29,591	96.38	30,304	98.70	400	1.30
Manchester	56,845	96.83	58,126	99.02	577	0.98
Newcastle	33,424	94.01	34,806	97.90	748	2.10
Oxford	29,292	88.26	32,043	96.54	1,147	3.46
Portsmouth	36,983	95.88	38,028	98.59	545	1.41
SE Thames	55,621	90.82	58,538	95.59	2,702	4.41
Sheffield	72,678	93.81	75,676	97.68	1,796	2.32
SW Thames	51,820	93.32	54,440	98.04	1,090	1.96
West Midlands	72,535	97.82	73,529	99.16	623	0.84
England	**646,488**	**91.33**	**686,767**	**97.02**	**21,125**	**2.98**
Northern Ireland	26,187	98.76	26,407	99.59	110	0.41
Scotland	40,554	73.25	48,885	88.29	6,482	11.71
Wales	31,872	86.60	35,188	95.61	1,616	4.39

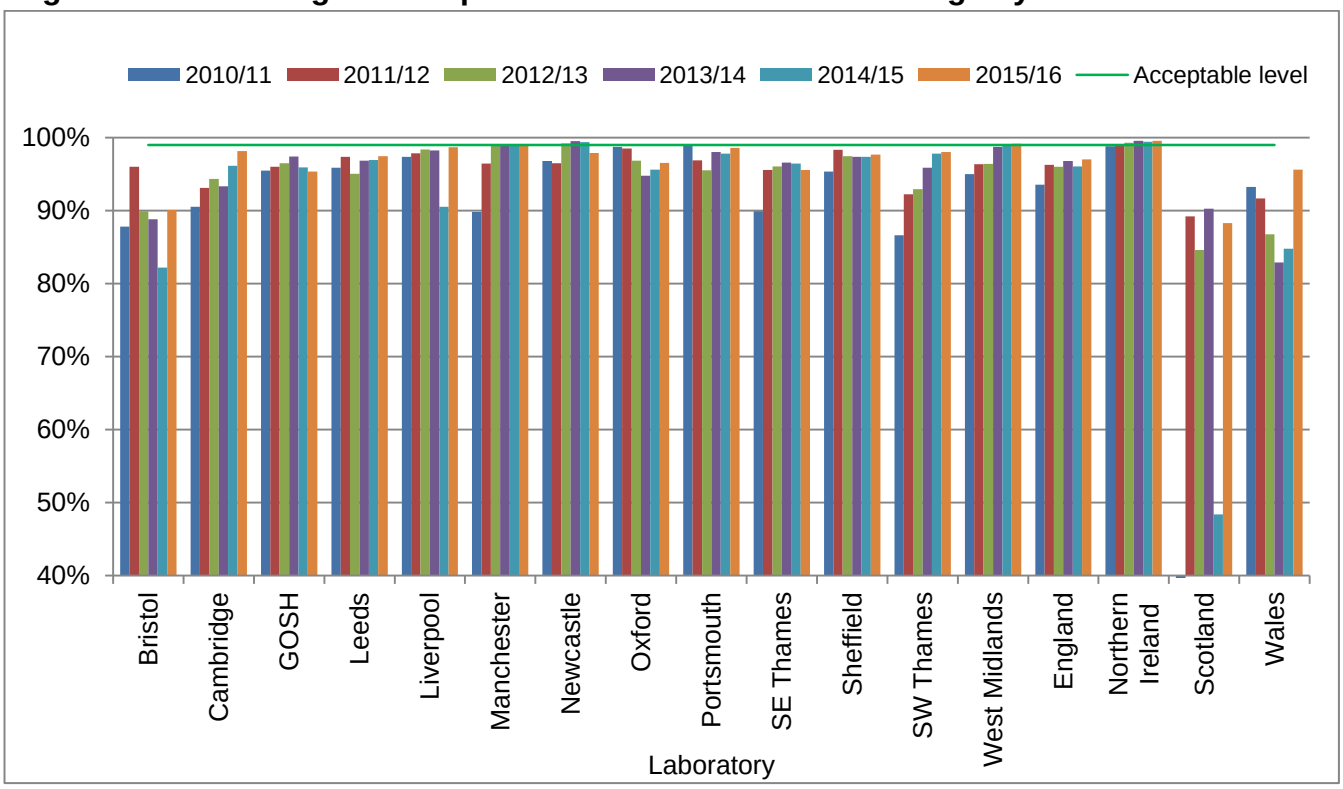
Data source: Newborn screening laboratories

Figure 12: Number of working days taken to receive sample 2015/16



Data source: Newborn screening laboratories

Figure 13: Percentage of samples received within four working days 2010/16



Data source: Newborn screening laboratories

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%.

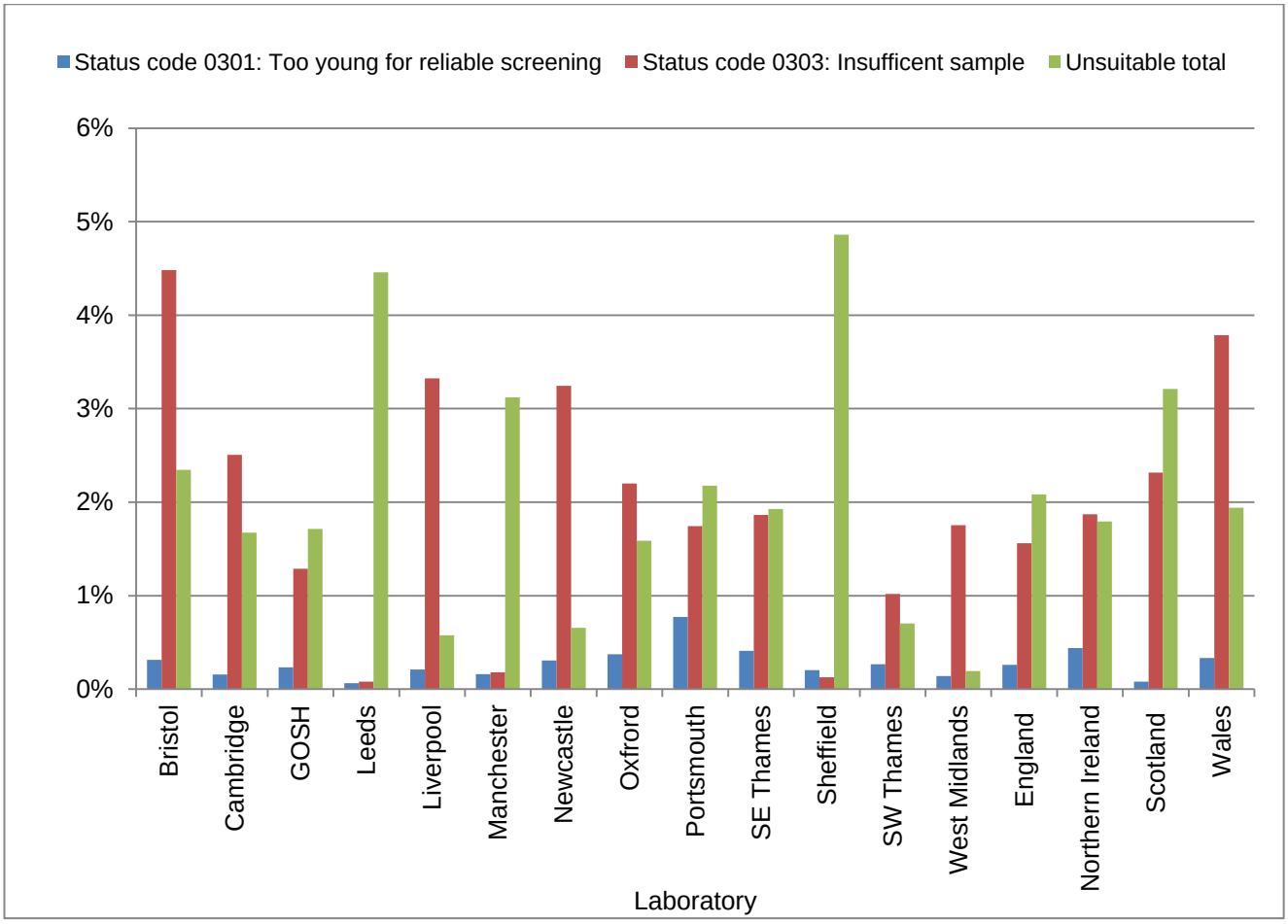
Table 13: Avoidable repeat request rates 2015/16

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	40,348	127	0.31	1,808	4.48	946	2.34	7.14
Cambridge	28,190	44	0.16	706	2.50	472	1.67	4.33
GOSH	121,730	285	0.23	1,569	1.29	2,087	1.71	3.24
Leeds	43,182	28	0.06	34	0.08	1,925	4.46	4.60
Liverpool	28,999	61	0.21	964	3.32	167	0.58	4.11
Manchester	55,721	89	0.16	100	0.18	1,739	3.12	3.46
Newcastle	33,245	102	0.31	1,079	3.25	218	0.66	4.21
Oxford	29,602	110	0.37	651	2.20	470	1.59	4.16
Portsmouth	36,873	285	0.77	643	1.74	802	2.18	4.69
SE Thames	57,339	235	0.41	1,069	1.86	1,104	1.93	4.20
Sheffield	73,144	149	0.20	92	0.13	3,555	4.86	5.19
SW Thames	53,457	142	0.27	544	1.02	376	0.70	1.99
West Midlands	70,382	98	0.14	1,235	1.75	136	0.19	2.09
England	**672,212**	**1,755**	**0.26**	**10,494**	**1.56**	**13,997**	**2.08**	**3.90**
Northern Ireland	24,575	108	0.44	460	1.87	441	1.79	4.11
Scotland	55,477	45	0.08	1,285	2.32	1,782	3.21	5.61
Wales	33,181	111	0.33	1,256	3.79	644	1.94	6.06

Data source: Newborn screening laboratories

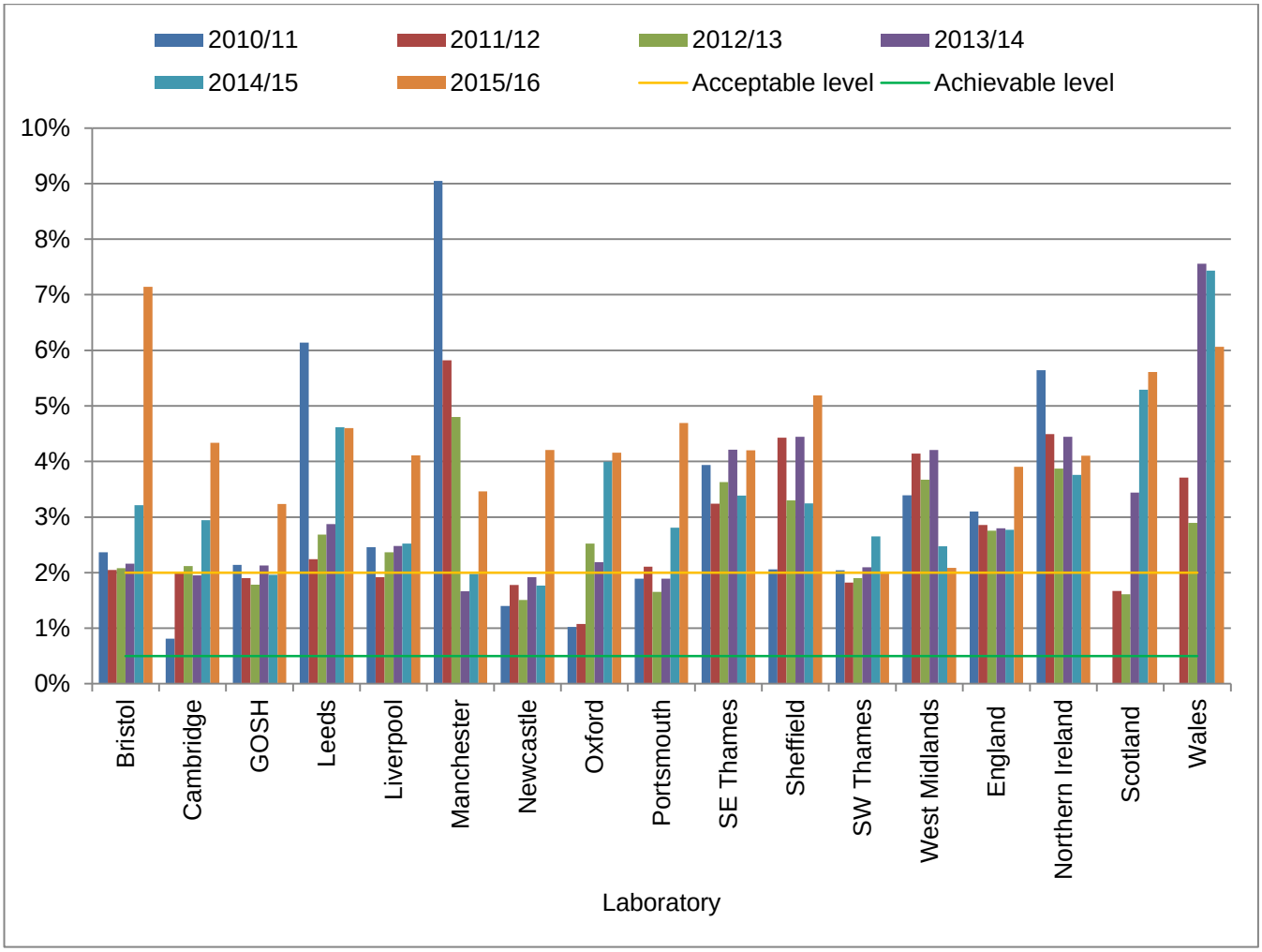
*Not all English laboratories ask for a repeat when the first sample was taken on or before day 4.

Figure 14: Avoidable repeat request rates (too young, insufficient and unsuitable) 2015/16



Data source: Newborn screening laboratories

Figure 15: Avoidable repeat request rates 20010/16



Data source: Newborn screening laboratories

Please note that 2010/13 data includes avoidable repeat requests due to insufficient and unsuitable samples only. In line with standard 6, 2013/16 data includes repeat requests due to samples taken when the baby was too young, insufficient and unsuitable.

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 is in the process of being published at www.ukas.com.

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 14: Numbers of samples processed within the standard in the UK 2015/16

Condition	Screen positive samples	Screen positive babies with clinical referral initiated within four working days		Screen positive babies with clinical referral initiated within three working days	
	n	n	%	n	%
PKU	115	115	100	115	100
CHT	655	632	96.5	619	94.5
MCADD	82	80	97.6	79	96.3

Data source: Newborn screening laboratories

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 is in the process of being published at www.ukas.com.

Standard 11: Timely receipt into clinical care

SCD

This year marks the 10th anniversary of full roll-out of newborn screening for sickle cell disease in England in July 2006. Since data collection began in 2005/06 7.3 million babies were screened, of which approximately 3,600 babies were identified with significant conditions (1 in 2,000 babies screened) and approximately 101,000 babies were identified as carriers (1 in 72 babies screened).

In newborn screening in 2015/16 there were 667,800 babies screened in England, and 779,448 for the whole of the UK (not including declines). Coverage for newborn screening in 2015/16 was 95.6%, which is similar to the figure for 2014/15. This figure uses PKU as a proxy for all conditions tested in newborn blood spot screening.

In 2015/16 there were 265 babies (1 in 2,520) with significant conditions identified in England, and approximately 8,600 (1 in 78) identified as carriers. The rate per 1,000 babies screened with significant conditions and carrier results appears to be falling in London, but these rates remain higher than those in the rest of England.

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 2015/16 there were 27 F-only cases in England, and 30 across the whole of the UK.

Rates of declined screening have continued to rise and are now at approximately 2 per 1,000, 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. Of the newborn screening declines, 40% did not have ethnicity recorded and 11% did not have region recorded in the data. This could indicate that this information is not being recorded where testing is declined and may be a reflection on the quality of the conversation between midwife and parents, and could indicate a training need.

Breakdown data on screen positive babies from the newborn laboratories shows a median age at time of initial clinical referral in 2015/16 of 16 days, and a median age at first visit to a paediatrician at a specialist haemoglobinopathy centre or local haemoglobinopathy centre of 51 days.

CF – screen positive babies with two 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 15: Timeliness of appointment and outcome for CF screen positive babies with two mutations 2015/16

	England	Northern Ireland	Scotland	Wales
Number of CF screen positive babies with two mutations	177	9*	21	11
Number clinically diagnosed before screening (excluded from following age data)	35	3	4	2
Number of babies with age at first appointment reported	93	6	16	9
Number seen ≤ 28 days (% of known data)	90 (96.7%)	6 (100%)	13 (81.3%)	9 (100%)
All babies mean age at first appointment	21	23	25	23
All babies median age at first appointment	22	24	24	23
Age range at first appointment	7-32	21-25	19-38	19-27
Number of babies with age at first appointment not reported	49	0	1	0
Outcome				
Confirmed	158	8	21	11
CF SPID	13	1	0	0
Excluded	0	0	0	0
Not reported	4	0	0	0
False negative (Meconium ileus)	2	0	0	0

Data source: Newborn screening laboratories
*for a further two babies it was not possible to determine if one or two CF mutations were present at the time of the screening mutation analysis.
Note that different screening and diagnostic protocols are followed in the UK – see Figures 19-22.

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:

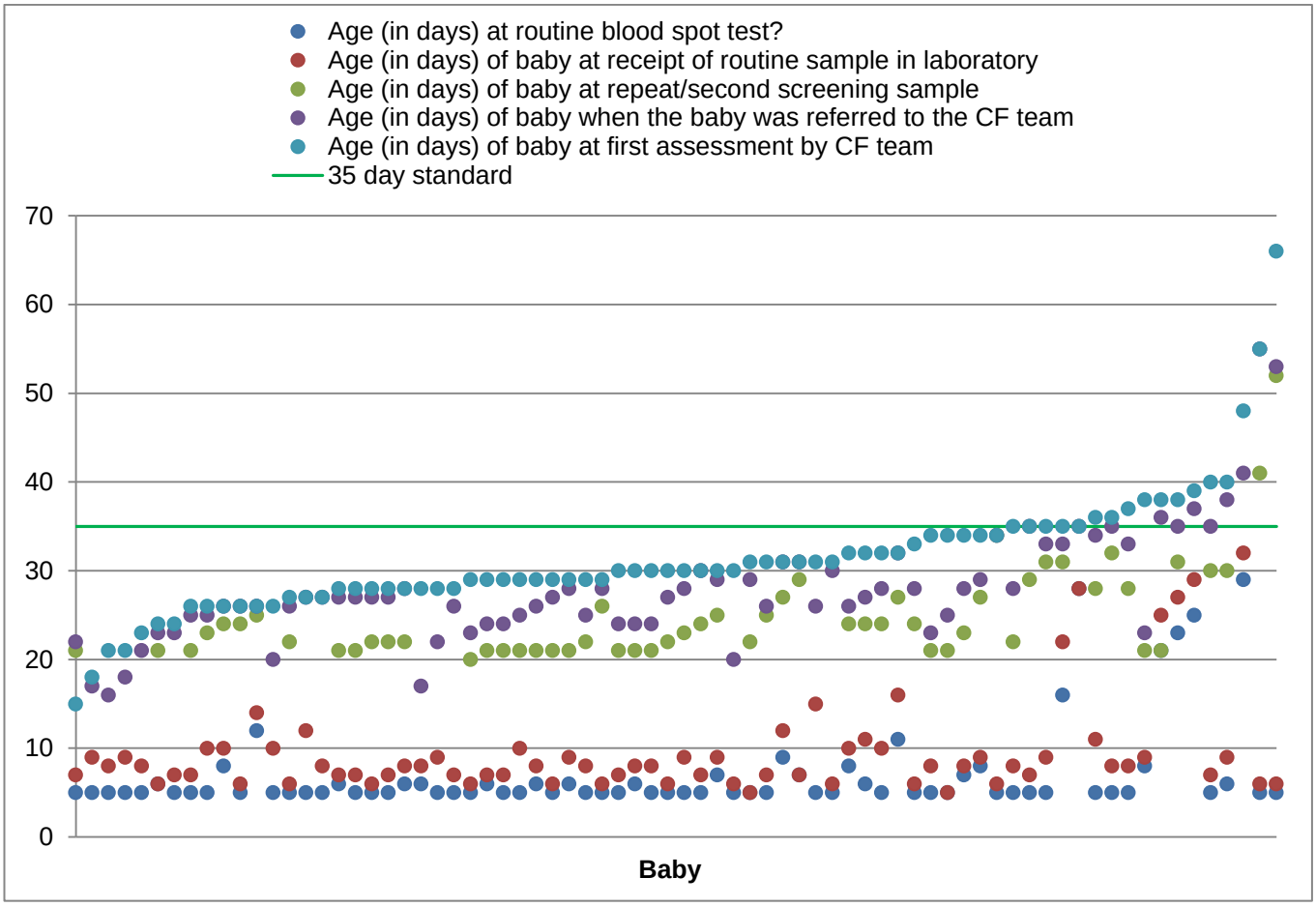
Acceptable level: 80% of babies seen by 35 days of age
Achievable level: 100% of babies seen by 35 days of age

Table 16: Timeliness of appointment and outcome for CF screen positive babies with one or no mutations 2015/16

	England	Northern Ireland	Scotland	Wales
Number of CF screen positive babies with one or no mutations	**100**	**5**	**11**	**12**
Number clinically diagnosed before screening (excluded from following age data)	1	1	0	0
Number of babies with age at first appointment reported	53	4	8	7
Number seen ≤ 35 days (% of known data)	42 (79.2%)	4 (100%)	6 (75%)	7 (100%)
All babies mean age at first appointment	30	31	37	24
All babies median age at first appointment	30	32		23
Age range at first appointment	24-279	24-34	28-66	18-31
Number of babies with age at first appointment not reported	46	0		
Outcome				
Confirmed	21	2	2	3
CF SPID	8	2		
Excluded	37		4	8
Baby died	4		1	
Not reported	19	1	2	1
Carrier	9		2	
Screening incomplete – lost to follow up	2			

Data source: Newborn screening laboratories

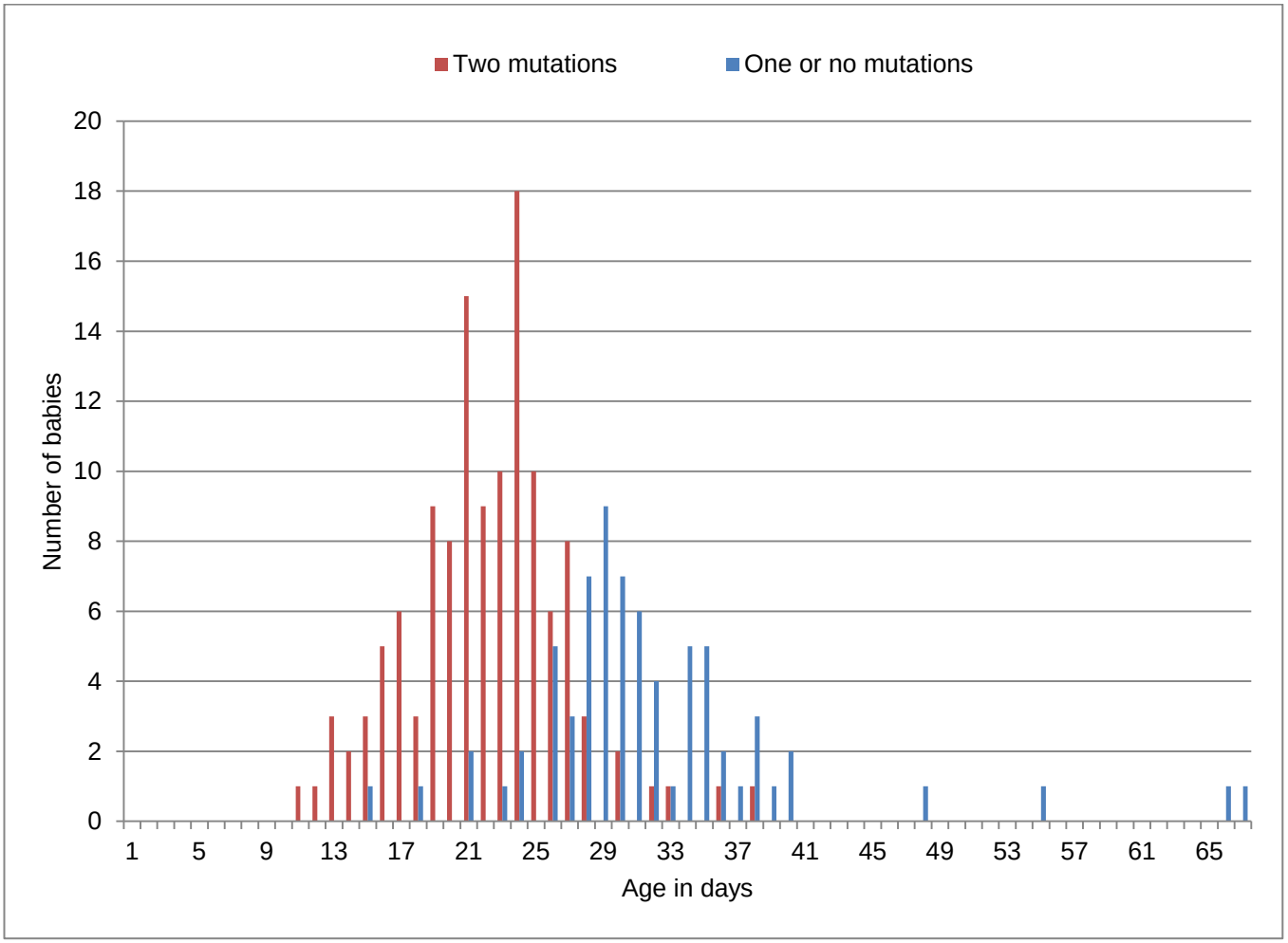
Figure 17: UK: CF screen positive babies with one or no mutations 2015/16
Age in days of all babies screened positive for CF with one or no mutations, at time of first sample, second sample, referral and assessment



Data source: Newborn screening laboratories

Exclusions have been made form the chart above for two babies that moved into the country late and were therefore tested late.

Figure 18: UK: age in days of CF screen positive babies at time of first appointment 2015/16



Data source: Newborn screening laboratories

Figure 19: England CFscreening and diagnostic algorithm 2015/16

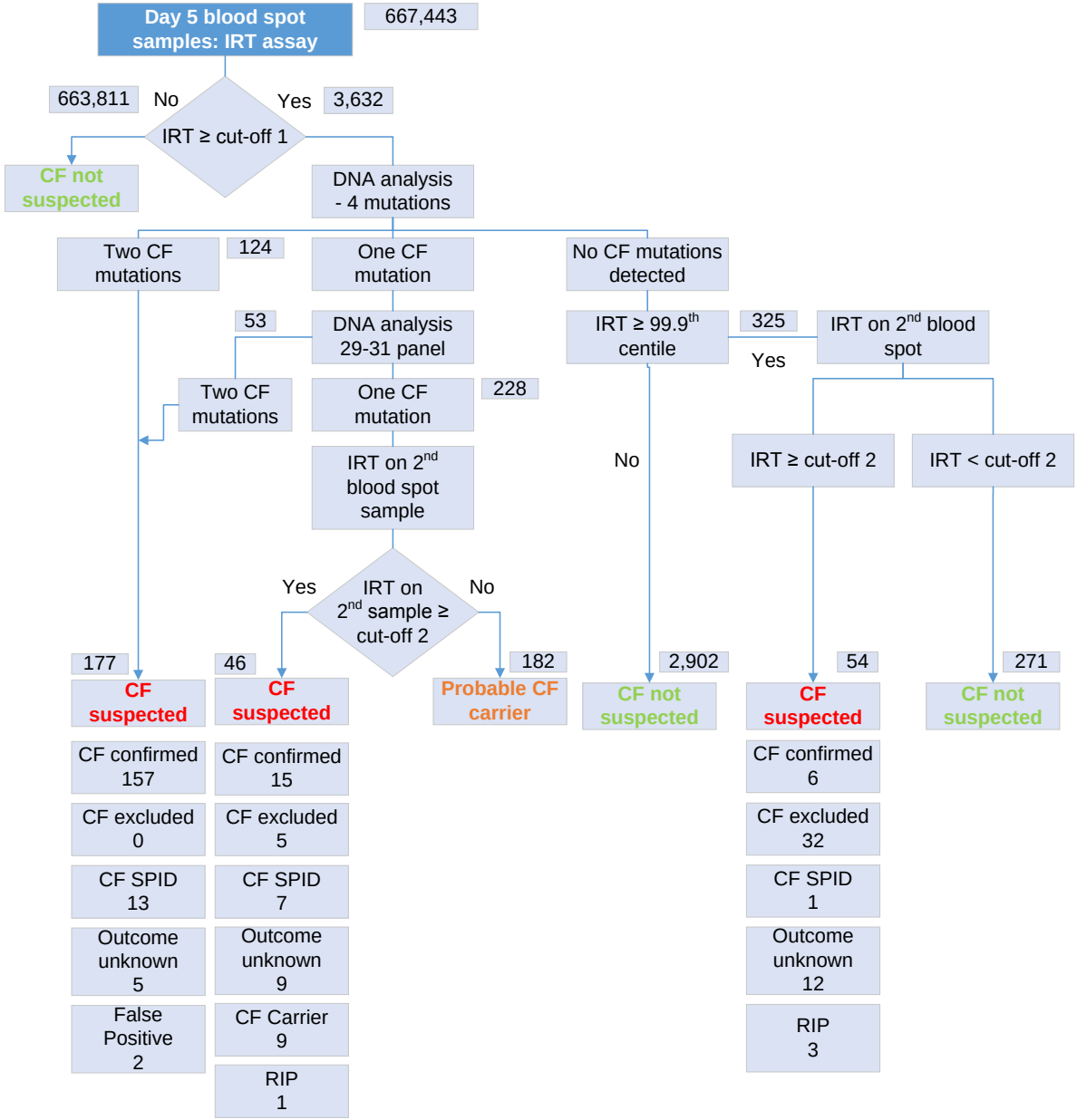
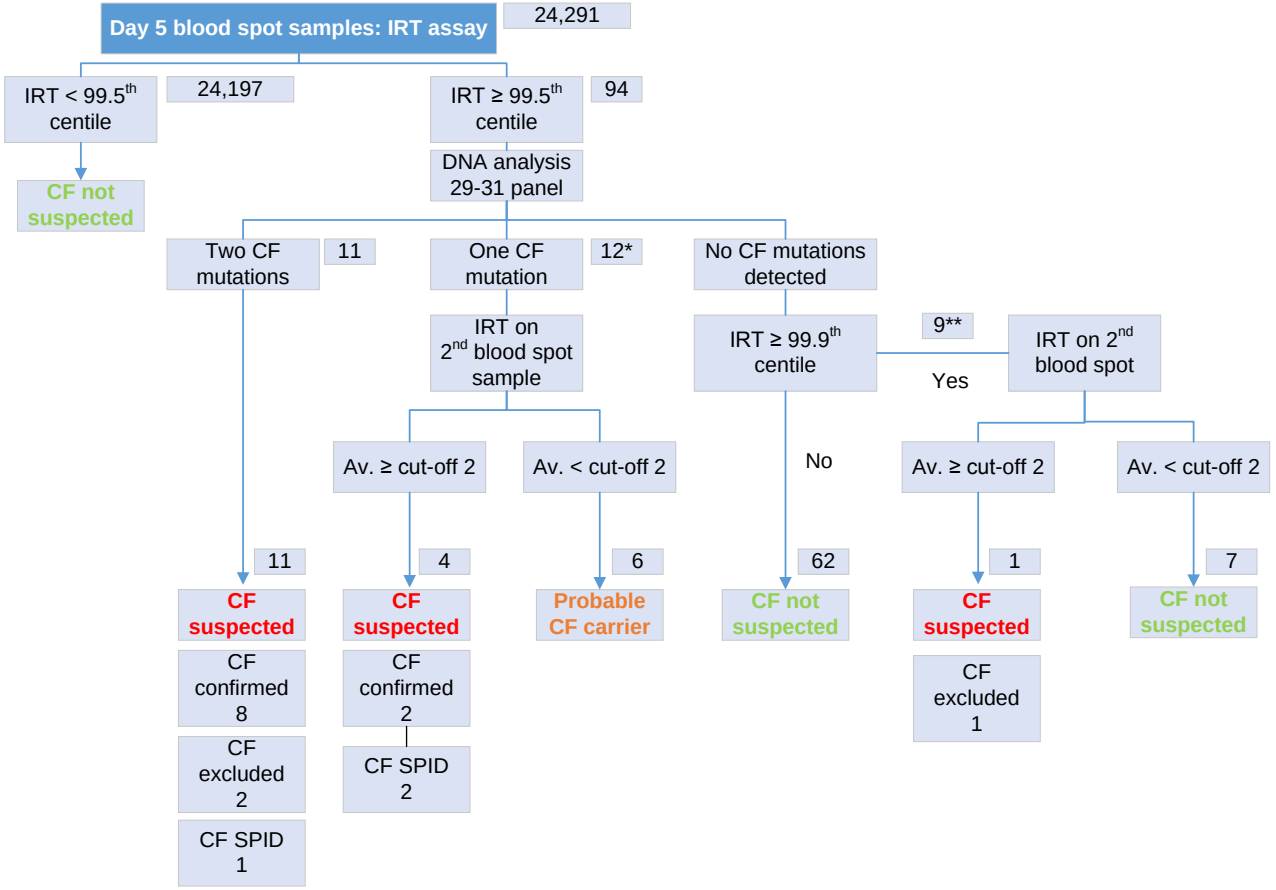


Figure 20: Northern Ireland CF screening and diagnostic algorithm 2015/16



*2 declined second sample as had already had CF Genetic testing outside screening pathway
**1 died before day 21 sample

Figure 21: Scotland CF screening and diagnostic algorithm 2015/16

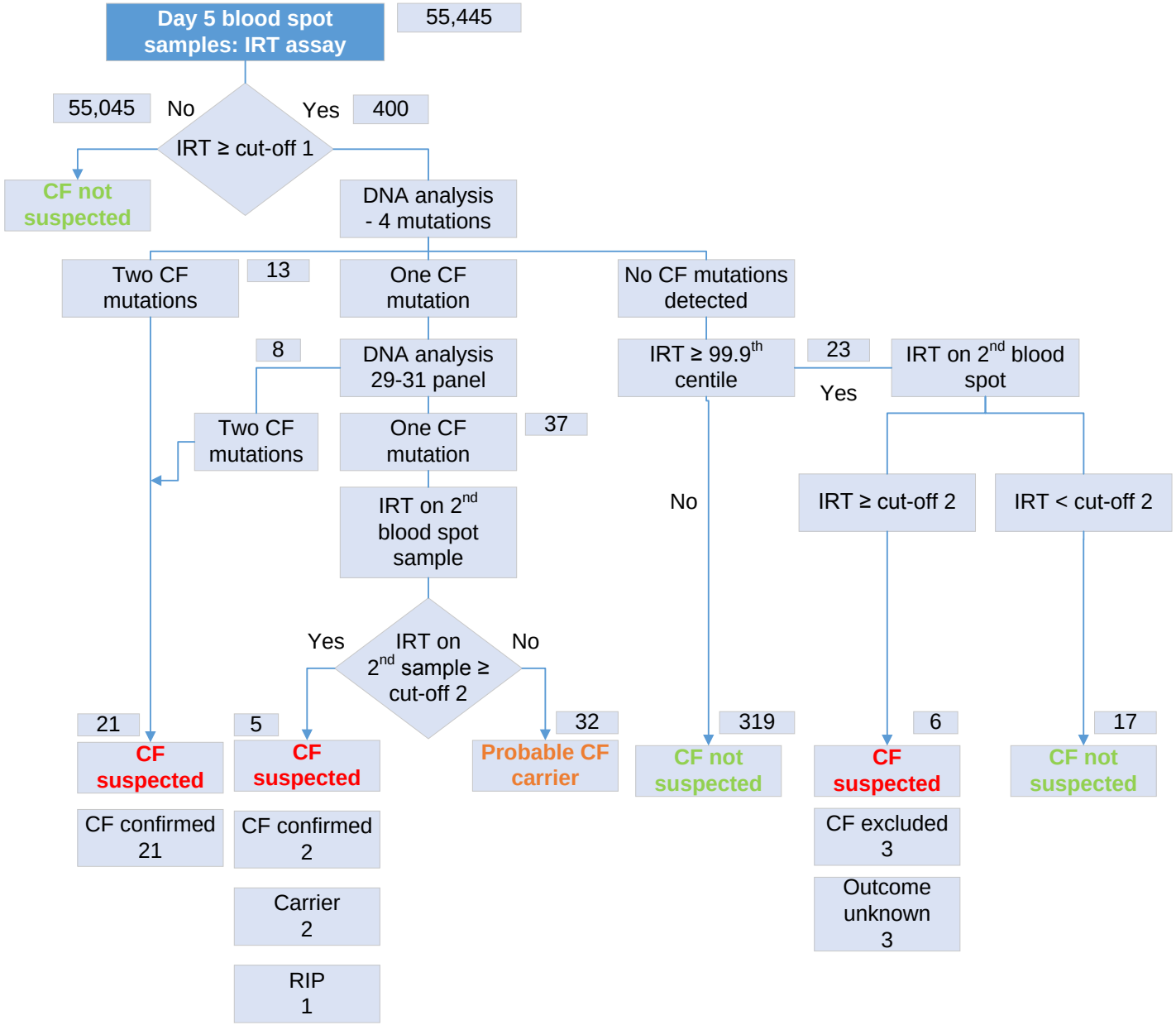
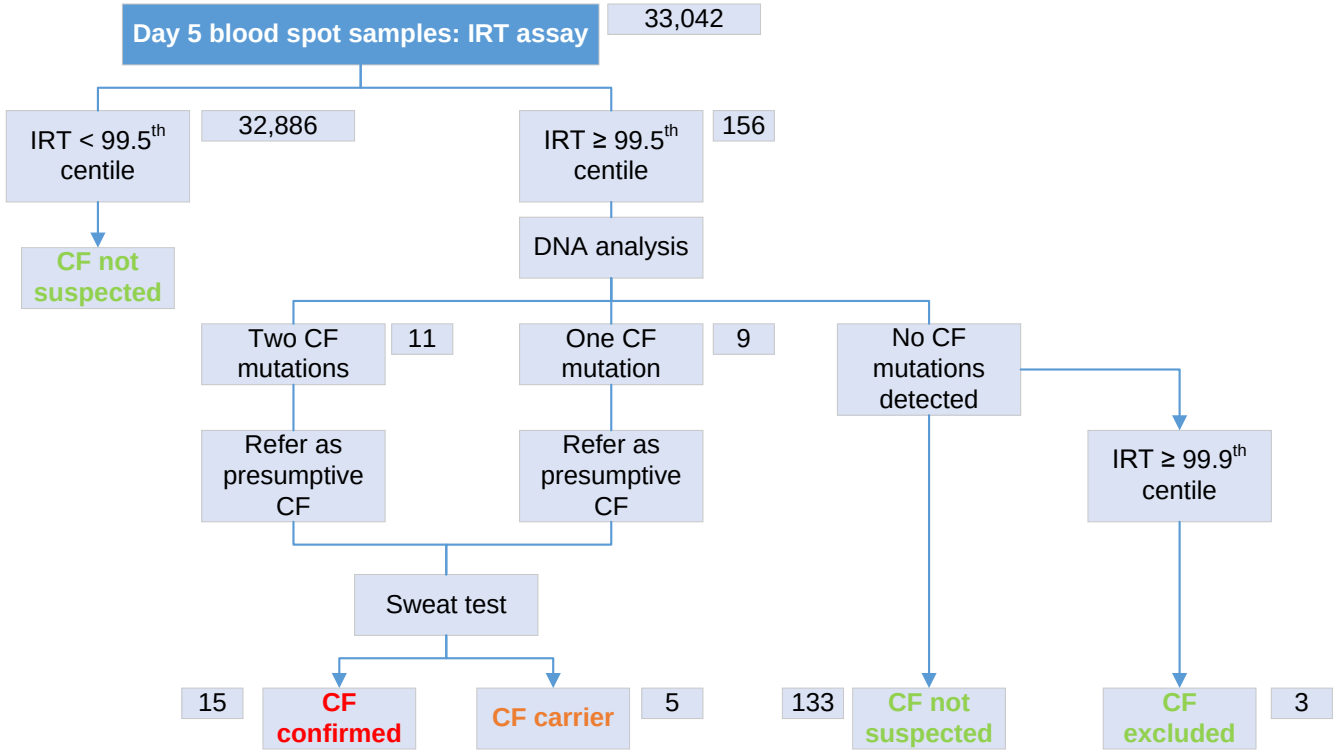


Figure 22: Wales CF screening and diagnostic algorithm 2015/16



CF screen positive data 2007/16

Table 17: CF screen positive data 2007/16

Laboratory	Babies tested for CF	CF screen positives	Rate of CF screen positives
	n	n	Rate per ten thousand
Bristol	371,531	218	5.87
Cambridge	250,364	104	4.15
GOSH	1,070,066	286	2.67
Leeds	402,660	166	4.12
Liverpool	261,417	148	5.66
Manchester	477,838	180	3.77
Newcastle	309,176	144	4.66
Oxford	267,726	70	2.61
Portsmouth	334,024	128	3.83
SE Thames	465,491	172	3.70
Sheffield	665,812	298	4.48
SW Thames	454,354	143	3.15
West Midlands	644,906	236	3.66
England	5,975,365	2,293	3.84
Northern Ireland	165,110	96	5.81
Scotland	464,241	251	5.41
Wales*	311,724	182	5.84
UK	6,916,440	2,822	4.08

Data source: Newborn screening laboratories

*Wales data does not include 'no mutations' – a different algorithm is followed.

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 18: Timeliness of appointment and treatment outcome for CHT screen positive babies detected on first sample 2015/16

	England	Northern Ireland	Scotland	Wales
Number of CHT screen positive babies detected on first sample	**281**	**11**	**23**	**18**
Number clinically diagnosed before screening (excluded from following age data)	11	0	3	0
Number of babies with age at first appointment reported	217	11	19	18
Number seen ≤ 14 days standard (% of known data)	175 (91%)	10 (91%)	18 (95%)	11 (61%)
Number seen ≤ 17 days standard (% of known data)	189 (98%)	11 (100%)	18 (95%)	16 (89%)
All babies mean age at first appointment	11	11	10	13
All babies median age at first appointment	11	11	10	13
Age range at first appointment	7-21	8-15	7-18	9-35
Number of babies with age at first appointment not reported	53	0	1	0
Has the baby started on thyroxine at the first appointment?				
Yes	188	9	16	15
No	0			
Not reported	64		5	3
Thyroxine not given but follow up required	21	2	2	
Thyroxine not given and baby discharged	8			

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:

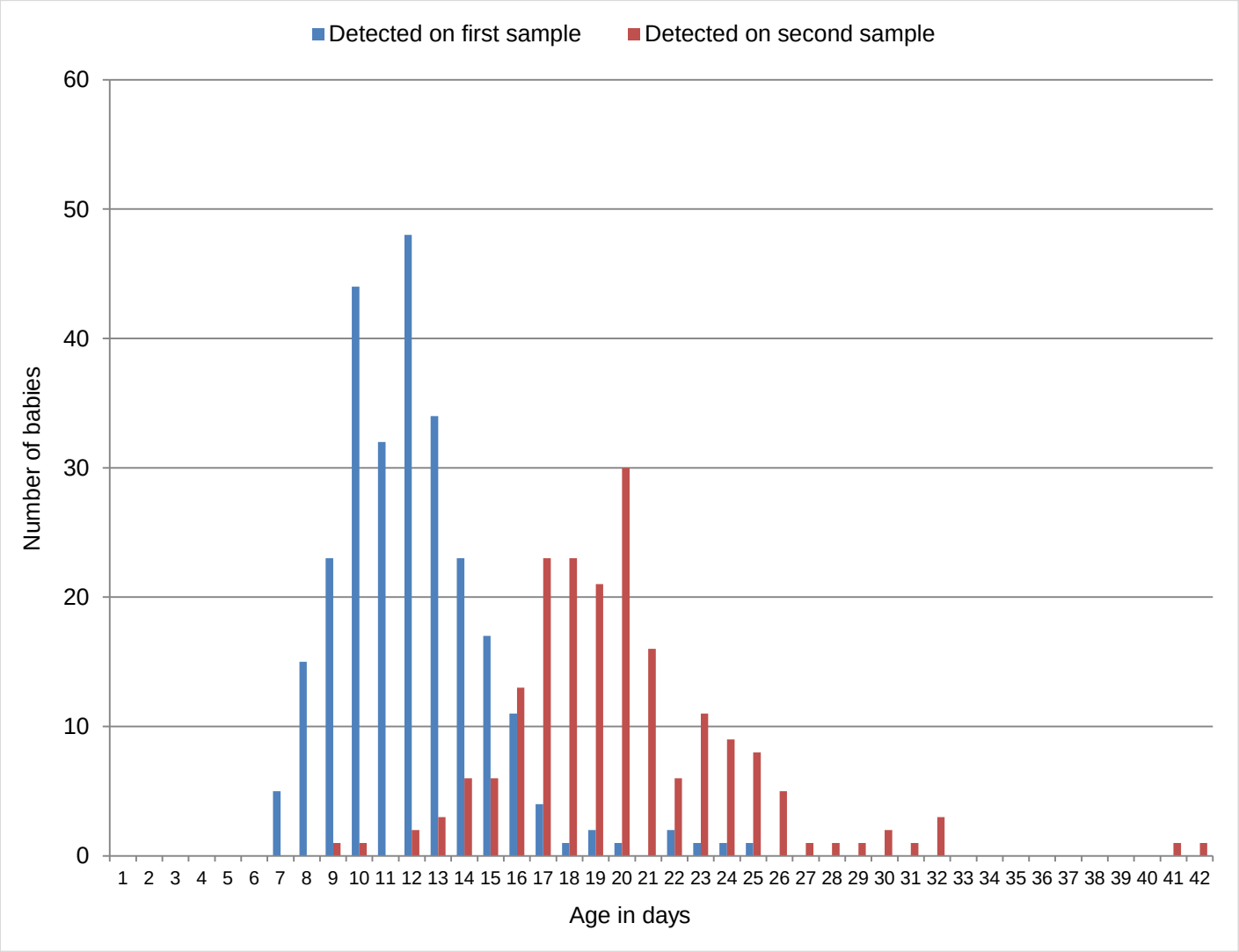
Acceptable level: 100% by 24 days of age
Achievable level: 100% by 21 days of age

Table 19: Timeliness of appointment and treatment outcome for CHT screen positive babies detected on second sample 2015/16

	England	Northern Ireland	Scotland	Wales
Number of CHT screen positive babies detected on second sample	**227**	**7**	**8**	**8**
Number clinically diagnosed before screening (excluded from following age data)	5	0	1	0
Number of babies with age at first appointment reported	151	7	7	8
Number seen ≤ 21 days standard (% of known data)	116 (75%)	6 (86%)	3 (43%)	4 (50%)
Number seen ≤ 24 days standard (% of known data)	140 (93%)	7 (100%)	5 (71%)	7 (88%)
All babies mean age at first appointment	19	16	23	19
All babies median age at first appointment	20	16	22	20
Age range at first appointment	10-48	12-23	12-42	10-26
Number of babies with age at first appointment not reported	62	0	0	0
Has the baby started on thyroxine at the first appointment?				
Yes	92	5	3	6
No	0	0	0	0
Not reported	63	0	0	0
Thyroxine not given but follow up required	54	2	2	1
Thyroxine not given and baby discharged	18	0	3	1

Data source: Newborn screening laboratories

Figure 23: England: age in days of CHT screen positive babies at time of first appointment 2015/16



Data source: Newborn screening laboratories

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

Table 20: Timeliness of sample and appointment for CHT screen positive babies born at less than 32 weeks 2015/16

	England	Northern Ireland	Scotland	Wales
Number of CHT screen positive babies born at less than 32 weeks	**72**	**0**	**0**	**0**
Number clinically diagnosed before screening (excluded from following age data)	2			
Age at routine sample				
Babies with age at routine sample reported	69			
Median age at routine sample	5			
Age range at routine sample	5-18			
Age at preterm sample				
Babies with age at preterm sample reported	56			
Median age at preterm sample	28			
Age range at preterm sample	21-52			
Age at first appointment				
Babies with age at first appointment reported	23			
Median age at first appointment	33			
Age range at first appointment	20-61			

Data source: Newborn screening laboratories

Table 21: England: treatment outcome for CHT screen positive babies born at less than 32 weeks 2015/16

	CHT suspected from other blood spot sample*	CHT suspected on preterm repeat @ 28 days/ discharge	CHT suspected on routine sample	CHT suspected on double borderline TSH result	CHT suspected on preterm repeat (double borderline TSH result)	CHT suspected on repeat TSH > 20 following borderline initial result	Clinically diagnosed before screening
72 babies	15	22	6	16	**9**	**2**	**2**
Has the baby started on thyroxine at the first appointment?							
Yes	1	10	3	2	2		1
No							
Not reported	6	7	3	6	5	2	
Thyroxine not given but follow up required	7	2		5	2		1
Thyroxine not given and baby discharged	1	3		3			

Data source: Newborn screening laboratories

*For example:

- single borderline result after initial unsuitable sample
- single borderline result on preterm repeat, first sample borderline, second sample normal
- preterm repeat borderline

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

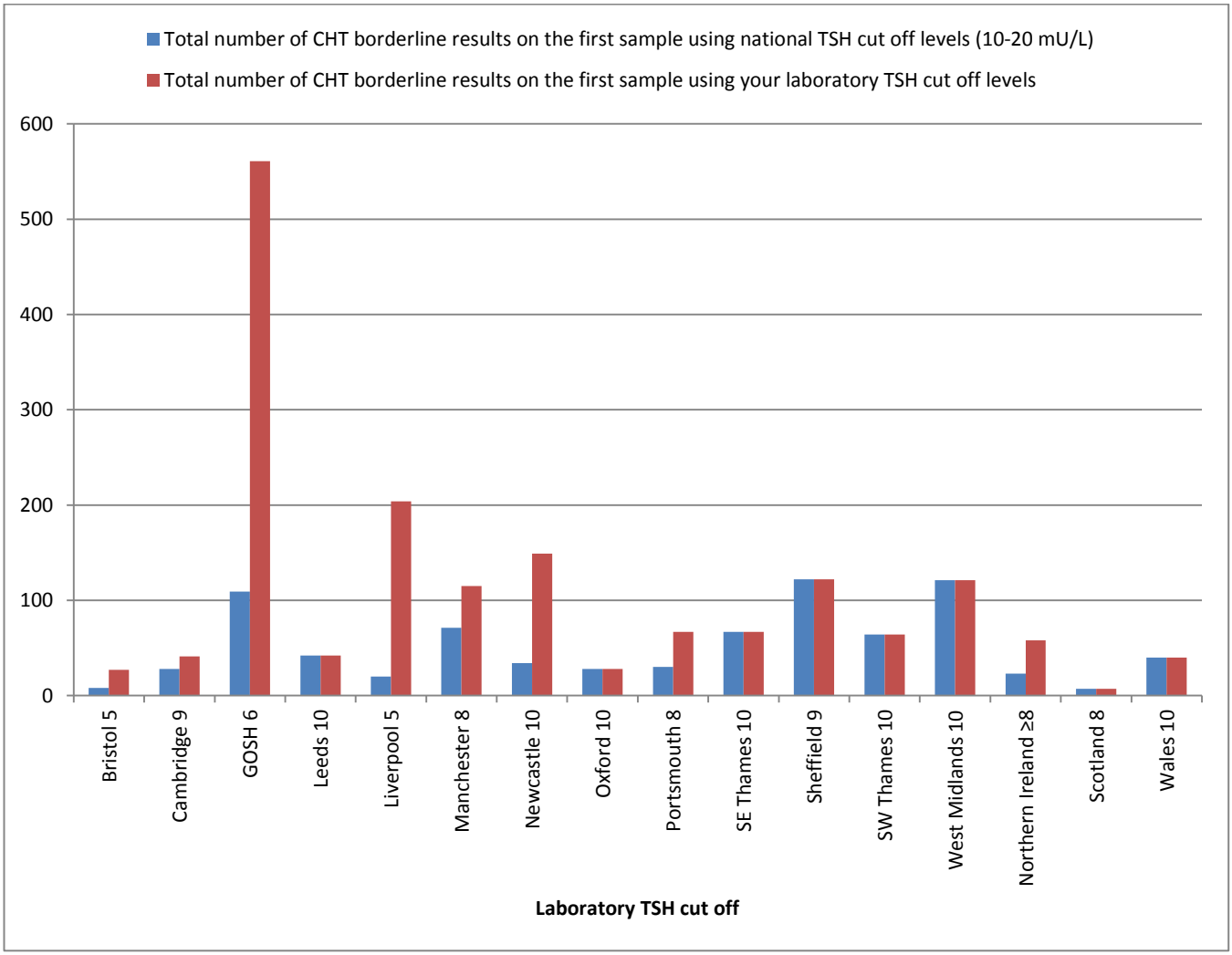
CHT is the only screening protocol in which a borderline result necessitates a second sample before a conclusive result can be achieved. The national borderline cut-off level is 10 mU/L. Some laboratories use a local cut-off level.

Table 22: CHT borderline results depending on use of national or local cut-off level 2015/16

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	20	6	8	27
Cambridge	18 (GSP)	9 (GSP)	28	41
GOSH	18 (GSP)*	6 (GSP)*	109	561
Leeds	20	10	42	42
Liverpool	20	5	20	204
Manchester	20	8	71	115
Newcastle	20	6	34	149
Oxford	20	10	5	5
Portsmouth	20	8	30	67
SE Thames	20	10	67	67
Sheffield	18 (GSP)	9 (GSP)	122	122
SW Thames	20	10	64	64
West Midlands	20	10	121	121
England			629	903
Northern Ireland	20	8	23	58
Scotland	20	8	7	7
Wales	20	10	40	40

Data source: Newborn screening laboratories
*GOSH laboratory changed to GSP December 2015.
Note that GSP cut-offs are equivalent to national cut-offs.

Figure 24: CHT borderline results depending on use of national or local cut-off level 2015/16



Data source: Newborn screening laboratories

CHT screen positive data 2005/16

Table 23: CHT screen positive data 2005/16

Laboratory	Babies tested for CHT	CHT screen positives	Rate of CHT screen positives
	n	n	Rate per ten thousand
Bristol	446,654	248	5.55
Cambridge	301,099	196	6.51
GOSH	1,341,478	1,338	9.97
Leeds	488,047	313	6.41
Liverpool	318,982	291	9.12
Manchester	562,754	440	7.82
Newcastle	375,999	273	7.26
Oxford	324,720	209	6.44
Portsmouth	392,746	197	5.02
SE Thames	622,888	345	5.54
Sheffield	803,753	431	5.36
SW Thames	564,821	331	5.86
West Midlands	778,797	564	7.24
England	7,322,768	5,176	7.07
Northern Ireland	270,479	182	6.73
Scotland	463,740	221	4.77
Wales	378,397	245	6.47
UK	8,435,384	5,824	6.90

Data source: Newborn screening laboratories

PKU

Description

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

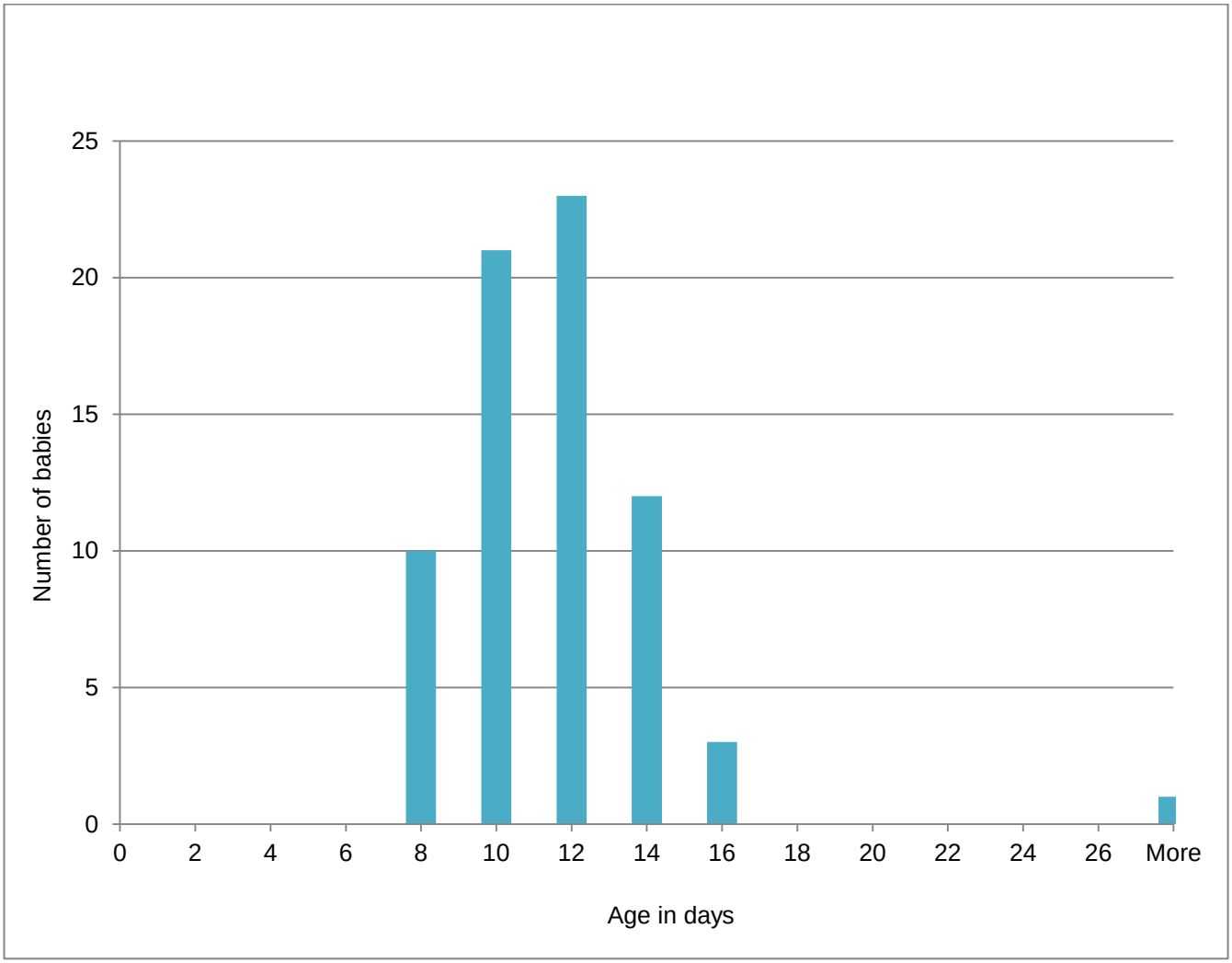
Acceptable level: 100% by 17 days of age
Achievable level: 100% by 14 days of age

Table 24: Timeliness of appointment and outcome for PKU screen positive babies 2015/16

	England	Northern Ireland	Scotland	Wales
Number of PKU screen positive babies	87	14	6	8
Number clinically diagnosed before screening (excluded from following age data)	10	4	1	0
Number of babies with age at appointment reported	73	10	4	8
Number seen ≤ 14 days (% of known data)	68 (93%)	8 (80%)	4 (100%)	8 (100%)
Number seen ≤ 17 days (% of known data)	71 (97%)	10 (100%)	4 (100%)	8 (100%)
All babies mean age at appointment	11	10	9	12
All babies median age at appointment	11	10	10	12
Age range at first appointment	7-350	6-16	7-10	11-14
Number of babies with age at appointment not reported	3	0	1	0
Outcome				
PKU confirmed, treatment required	53	10	4	6
Non PKU e.g. biopterin disorders	11	1	1	
No persistent abnormalities - false positive (PKU excluded)	6	1	1	1
PKU monitoring required	17	2		1
Not reported	0	0		

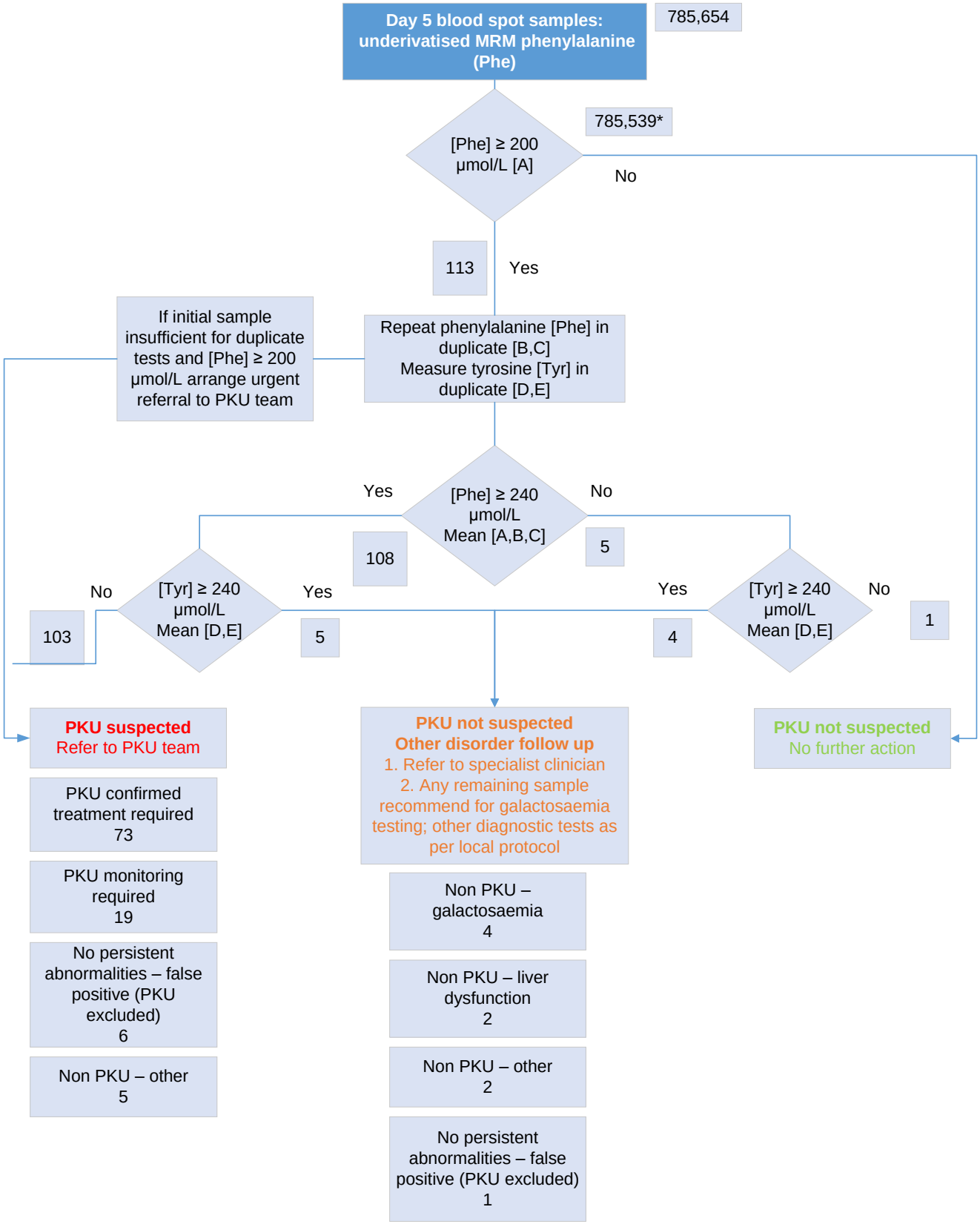
Data source: Newborn screening laboratories
Two older babies that moved in from abroad were tested late. Both were referred with elevated tyrosinaemia and phenylanine levels compared to reference ranges for their age.

Figure 25: UK: age at first appointment for PKU screen positive babies 2015/16



Data source: Newborn screening laboratories
One baby was seen late and was a late entry to the country.

Figure 26: UK PKU screening and diagnostic algorithm 2015/16



*Two babies are not mapped to the algorithm as they were late entrants and referred with elevated tyrosine and phenylaninine levels compared to reference ranges for their age.

PKU screen positive data 2005/16

Table 25: PKU screen positive data 2005/16

Laboratory	Babies tested for PKU	PKU screen positives	Rate of PKU screen positives
	n	n	Rate per ten thousand
Bristol	446,658	33	0.74
Cambridge	301,099	52	1.73
GOSH	1,332,445	124	0.93
Leeds	488,047	63	1.29
Liverpool	318,982	35	1.10
Manchester	562,796	90	1.60
Newcastle	375,999	47	1.25
Oxford	324,723	28	0.86
Portsmouth	392,767	27	0.69
SE Thames	622,886	63	1.01
Sheffield	803,755	113	1.41
SW Thames	564,821	43	0.76
West Midlands	778,797	89	1.14
England	7,313,802	807	1.10
Northern Ireland	270,492	71	2.62
Scotland	463,785	69	1.49
Wales	378,455	56	1.48
UK	8,426,534	1,003	1.19

Data source: Newborn screening laboratories

MCADD

Description

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

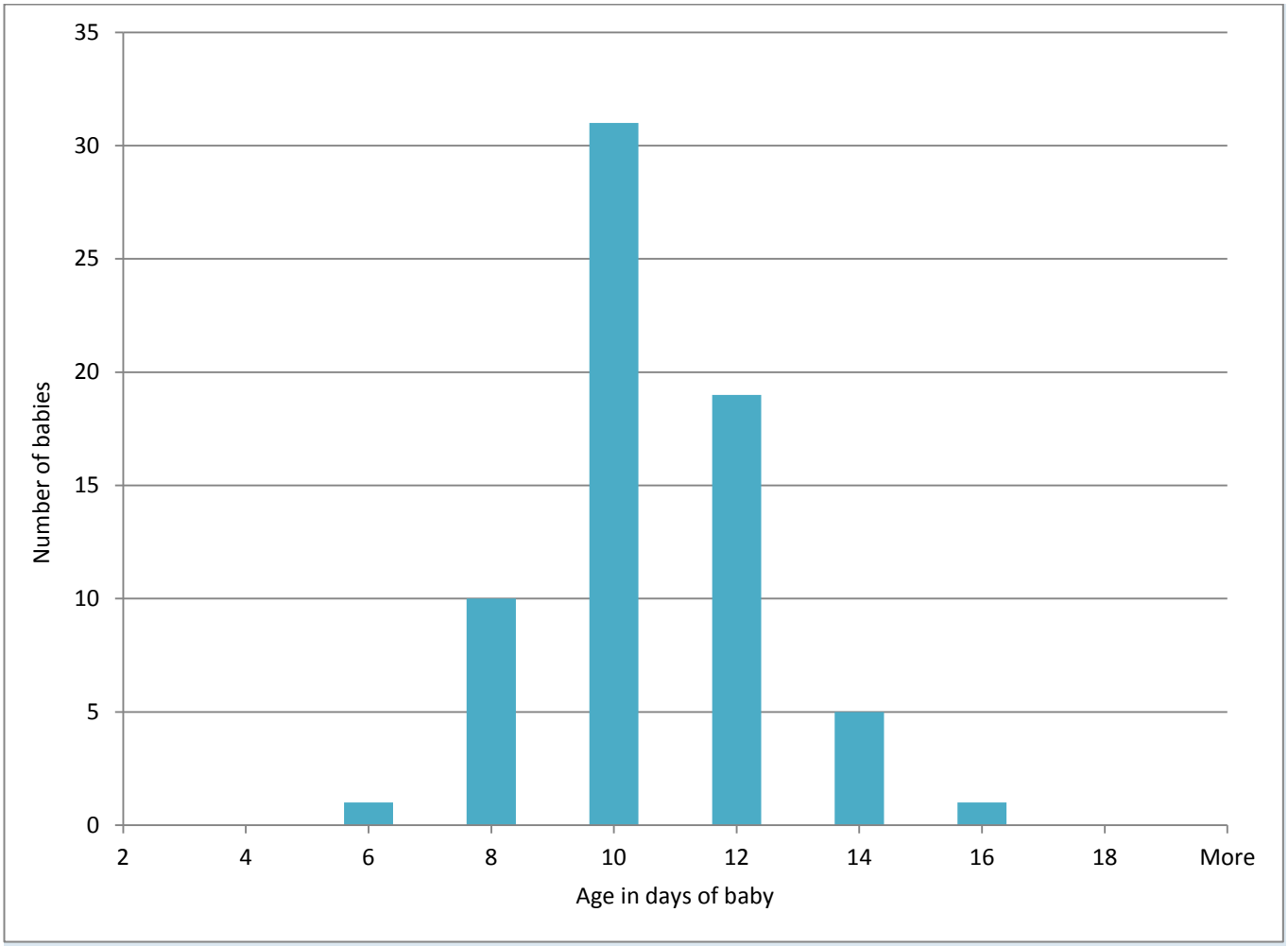
Acceptable level: 100% by 17 days of age
Achievable level: 100% by 14 days of age

Table 26: Timeliness of appointment and outcome for MCADD screen positive babies 2015/16

	England	Northern Ireland	Scotland	Wales
Number of MCADD screen positive babies	**77**	**2**	**1**	**3**
Number clinically diagnosed before screening (excluded from following age data)	**6**	**1**	**0**	**0**
Number of babies with age at appointment reported	67	1	1	3
Number seen ≤ 14 days (% of known data)	66 (99%)	1 (100%)	1 (100%)	3 (100%)
Number seen ≤ 17 days (% of known data)	67 (100%)	1 (100%)	1 (100%)	3 (100%)
All babies mean age at appointment	10	8	10	11
All babies median age at appointment	10	8	10	11
Age range at first appointment	5-15	n/a	n/a	10-12
Number of babies with age at appointment not reported	4	0	0	0
Outcome				
MCADD	59	2	1	3
Unaffected carrier	3			
MCADD unlikely				
No persistent abnormality, false positive	6			
Not reported	9			

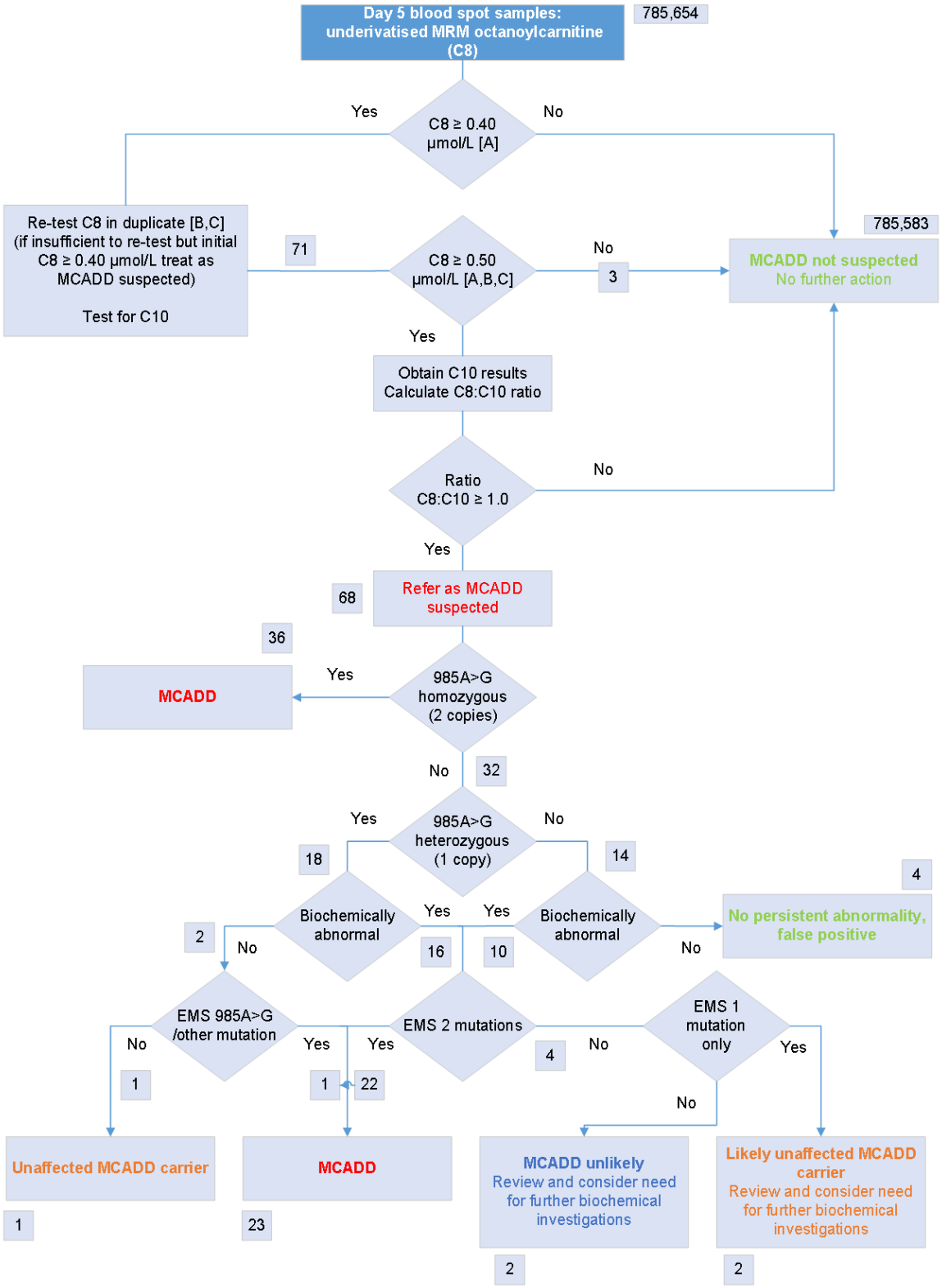
Data source: Newborn screening laboratories

Figure 27: UK: age at first appointment for MCADD screen positive babies 2015/16



Data source: Newborn screening laboratories

Figure 28: UK MCADD screening and diagnostic algorithm 2015/16



MCADD screen positive data 2008/16

Table 27: MCADD screen positive data 2008/16

Laboratory	Babies tested for MCADD	MCADD screen positives	Rate of MCADD screen positives
	n	n	Rate per ten thousand
Bristol	310,406	25	0.81
Cambridge	219,567	29	1.32
GOSH	982,925	70	0.71
Leeds	359,072	50	1.39
Liverpool	225,386	25	1.11
Manchester	452,327	53	1.17
Newcastle	254,826	27	1.06
Oxford	213,019	25	1.17
Portsmouth	294,722	31	1.05
SE Thames	461,478	35	0.76
Sheffield	593,322	89	1.50
SW Thames	396,379	33	0.83
West Midlands	574,289	47	0.82
England	5,337,718	539	1.01
Northern Ireland	165,748	20	1.21
Scotland	318,383	11	0.35
Wales	128,915	11	0.85
UK	5,950,764	581	0.98

Data source: Newborn screening laboratories

MSUD, IVA, GA1 and HCU

Table 28: England and Wales: Timeliness of appointment and outcome for MSUD, IVA, GA1 and HCU screen positive babies 2014/16

	MSUD	IVA	GA1	HCU
Number of screen positive babies	**11**	**14**	**12**	**11**
Family history (early testing)	**2**	**1**	**0**	**1**
Number of babies with age at first appointment reported	5	12	10	4
Number seen ≤ 14 days	4 (80%)	12 (100%)	7 (70%)	3 (75%)
Number seen ≤ 17 days	4 (80%)	12 (100%)	10 (100%)	3(75%)
All babies median age at first appointment	10	11	13	15
Age range at first appointment	8-42	7-16	8-249	14-30
Number of babies with age at first appointment not reported	4	1	2	6
Outcome				
Confirmed	7	3	4	4
False positive	4	6	6	5
Mild (IVA only)	0	3	0	0
Other	0	1	2	1
Not reported	0	1	0	1

Data source: Newborn screening laboratories

Due to small numbers the data for IMDs are shown for a two year period.

Standard 12: Timeliness of results to parents

Description
CHRDs issue normal results for all five 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.

Data against this standard was previously collected quarterly as KPI NB3 – this is the first time that data has been presented in the annual report.

CHRDs were asked to report the number of babies with screen negative results for all 5 conditions available for communication by 6 weeks of age. The definition of this standard will be reviewed.

Table 29: Timeliness of results to parents 2015/16

Region/country	Babies screen negative for all five conditions	Results available for communication by six weeks of age	
	n	n	%
England	**472,576**	**433,432**	**91.7%**
North	**120,312**	**112,019**	**93.1%**
North East	17,810	17,787	99.9%
North West	43,944	35,800	81.5%
Yorkshire & The Humber	58,558	58,432	99.8%
South	**102,658**	**98,343**	**95.8%**
South Central	40,421	40,414	100.0%
South East	13,841	13,775	99.5%
South West	48,396	44,154	91.2%
Midlands & East	**155,280**	**154,105**	**99.2%**
East Midlands	53,335	53,130	99.6%
East of England	56,713	55,800	98.4%
West Midlands	45,232	45,175	99.9%
London	**94,326**	**68,965**	**73.1%**

Data source: CHRDs

Note that standard 1a indicates that 94.3% of results in England are recorded on the CHIS by 17 days of age (CCG responsibility at birth).

Conclusion

The NBS programme streamlined the data collection template to make the reporting process more robust. Data was returned by CHRDs for 196 CCGs (93%) out of the 211 that existed in England in 2015/16. Exclusions were made if the data was incomplete. Some CHRDs reported that the data their informations system had provided was inaccurate and therefore agreed to withdraw their data from publication. Work is ongoing to rectify these IT issues. In some cases data was reported for a particular CCG by more than one CHRD; this added to the complexity of analysing the data.

There 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.

In February 2015 all 167 maternity sites were utilising the NBSFS to ensure all babies born in England are offered screening. In November 2016 the last laboratory began to send all screening results into the national database but it is not sending a complete set of screening subcodes. The responsibility for ensuring completeness of coverage remains with the CHRD.

There continues to be no significant change in the overall rate of declines for the CCG responsibility at birth cohort in England and no clear patterns have emerged within regions. However, only 19 CCGs reported greater than 10 declines per ten thousand and more than 5 declines in total, this is 30 fewer CCGs than in 2014/15. The NBSFS can capture declines and work is continuing to develop accurate reporting.

All but 1 English screening laboratory reported their maternity sites are meeting the acceptable level of first samples taken on days 5-8 (≥95%). Scotland accepts day 4 samples. Transport of samples to the laboratories has been a focus for this year as it has been one of the biggest risks for delayed identification of screen positive babies. Samples received within four working days has increased in 12 out of 16 UK laboratories, with one other laboratory maintaining the achievable level of 99%. This can be accounted for by maternity services changing their mode of transport for the samples. See a recent blog ‘Are your blood spot samples taking too long to arrive at the screening lab?’ https://phescreening.blog.gov.uk/2016/12/16/are-your-blood-spot-samples-taking-too-long-to-arrive-at-the-screening-lab/

New consensus guidelines for the acceptance of the quality of blood spot cards were implemented in England and Wales in April 2015. However, prior to this some laboratories that accepted poor quality samples began to apply stricter rejection criteria. As anticipated, overall avoidable repeat rates have increased as a result although these rates are now more comparable in 2015/16. As the avoidable repeat rate is a key performance indicator, maternity service providers are working hard to reduce their rates.

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.

The current standard for timely processing of screen positive samples only applies to PKU, CHT and MCADD. The achievable level was met for PKU. The acceptable standard was not met for CHT or MCADD.

Based on data reported, the acceptable standard for timeliness of first appointment for CF screen positive babies with 2 mutations was met in England, Wales and Northern Ireland but not Scotland. This is an improvement for England and Northern Ireland from 2014/15. The acceptable standard for babies with one or no mutations was not met in England or Scotland which is the same as 2014/15. CF outcome data remains challenging for the laboratories to collect though closer links to CF regional centres is helping.

Based on data reported, the acceptable standard for timeliness of first appointment for CHT screen positive babies detected on first sample was not met in England, Scotland or Wales. The acceptable standard for babies detected on second sample was not met in England, Scotland or Wales.

In England, just over a quarter of data on CHT treatment at first appointment remains missing for babies detected on first sample (65 out of 281) and second sample (63 out of 227). This is worse than 2014/15 when approximately one sixth of CHT treatment data was missing. 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 Newborn Blood Spot Screening Programme for detecting permanent CHT was good (sensitivity 97.84%, specificity 99.98%, positive predictive value 67.36%). However only 4 of the UK newborn screening laboratories were using the national cut-off (thyroid stimulating hormone [TSH] ≥10mU/L) and many were using 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 for PKU was met in Northern Ireland, Scotland and Wales. For MCADD the acceptable standard was met in all 4 countries.

Abbreviations

CF	cystic fibrosis
CCG	clinical commissioning group
CFTR	cystic fibrosis transmembrane conductance regulator
CHIS	child health information system
CHRD	child health records department
CHT	congenital hypothyroidism
CPA	Clinical Pathology Accreditation
GA1	glutaric aciduria type 1
GOSH	Great Ormond Street Hospital
GSP	Genetic Screening Processor
HCU	homocystinuria
HV	health visitor
IMD	inherited metabolic disease
IVA	isovaleric acidaemia
KPI	key performance indicator
MCADD	medium-chain acyl-CoA dehydrogenase deficiency
MSUD	maple syrup urine disease
NBS	newborn blood spot
NBSFS	Newborn Blood Spot Failsafe Solution
NICU	neonatal intensive care unit
PHE	Public Health England
PKU	phenylketonuria
SCD	sickle cell disease
SE Thames	South East Thames
SW Thames	South West Thames
SQAS	Screening Quality Assurance Service
TSH	thyroid stimulating hormone
UKAS	United Kingdom Accreditation Service
UK GTN	UK Genetic Testing Network
UK NSC	UK National Screening Committee

References

1. NHS Newborn Blood Spot Screening Programme (2013) *Standards for Newborn Blood Spot Screening* [Online] Available at: www.gov.uk/government/publications/standards-for-nhs-newborn-blood-spot-screening (accessed: 22nd February 2017).

2. NHS England (2015) *NHS public health Functions agreement 2016-17 Service specification no.19, NHS Newborn Blood Spot Screening Programme* [Online] Available at: www.england.nhs.uk/commissioning/pub-hlth-res/ (accessed: 22 February 2017).

3. NHS Newborn Blood Spot Screening Programme (2014) *Status codes: version 4.2* [Online] Available at: www.gov.uk/government/publications/status-codes-for-the-newborn-blood-spot-nbs-screening-programme (accessed: 22 February 2017).

4. NHS Sickle Cell and Thalassaemia Screening Programme (2016) *NHS Sickle Cell and Thalassaemia Screening Programme Data Report 2015/16: Trends and performance analysis* [Online] Available at: www.gov.uk/government/publications/sickle-cell-and-thalassaemia-screening-data-trends-and-performance-analysis (accessed: 22 February 2017).