Public Health England

NHS

Data Collection and Performance Analysis Report

Newborn blood spot screening in the UK 2014/15

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.

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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 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.

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Executive summary

This is the eleventh 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.

All four UK countries screen for sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT), phenylketonuria (PKU) and medium-chain acyl-CoA dehydrogenase deficiency (MCADD). England and Wales began screening for maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (pyridoxine unresponsive) (HCU) in January 2015 – this report therefore presents partial data for these four conditions.

Data was returned by child health records departments (CHRDs) for 183 clinical commissioning groups (CCGs) (87%) out of the 211 that existed in England in 2014/15. 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) has increased year on year from 81.8% in 2010/11 to 94.6% in 2014/15. Standard 1b introduces an effective timeframe of 21 calendar days for movers in. In England, coverage for movers in with the timeframe applied was 75.6% and without the timeframe was 91.5%.

Over the last three 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. In England, processes for recording declines for movers in vary between regions.

In England, 99.4% of blood spot cards included the baby's NHS number, and 74.5% 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, 95.8% 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.

Laboratories have accepted blood spot cards of varying quality. New consensus guidelines were implemented in England and Wales in April 2015 and avoidable repeat rates will be more comparable from 2015/16.

Laboratory accreditation (standards 8 and 10) is in the process of being published at www.ukas.com.

The acceptable standard for timeliness of first appointment for CF screen positive babies with two mutations was not met in England, Northern Ireland or Scotland. The acceptable standard for one or no mutations was not met in England or Scotland. This data is based on babies with age at first appointment reported. CF diagnostic outcome data is challenging for the laboratories to collect – in England approximately one sixth of CF outcome data remains missing for babies with two mutations, and one third of data remains missing for babies with one or no mutations.

The acceptable standard for timeliness of first appointment for CHT screen positive babies detected on first sample was not met in England or Wales. The acceptable standard for babies detected on second sample was not met in England or Northern Ireland. This data is based on babies with age at first appointment reported. In England, approximately one sixth of data on CHT treatment outcome remains missing. The findings of a CHT British Paediatric Surveillance Unit study are due to be reported in mid-2016.

Following successful implementation of expanded screening in January 2015, full data on MSUD, IVA, GA1 and MSUD will be available in the 2015/16 report. The acceptable standard for timeliness of appointment for PKU and MCADD screen positive babies was not met in England in 2014/15. However, with minimal chasing outcome data was reported for all babies.

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

Introduction

Background

This is the eleventh 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.

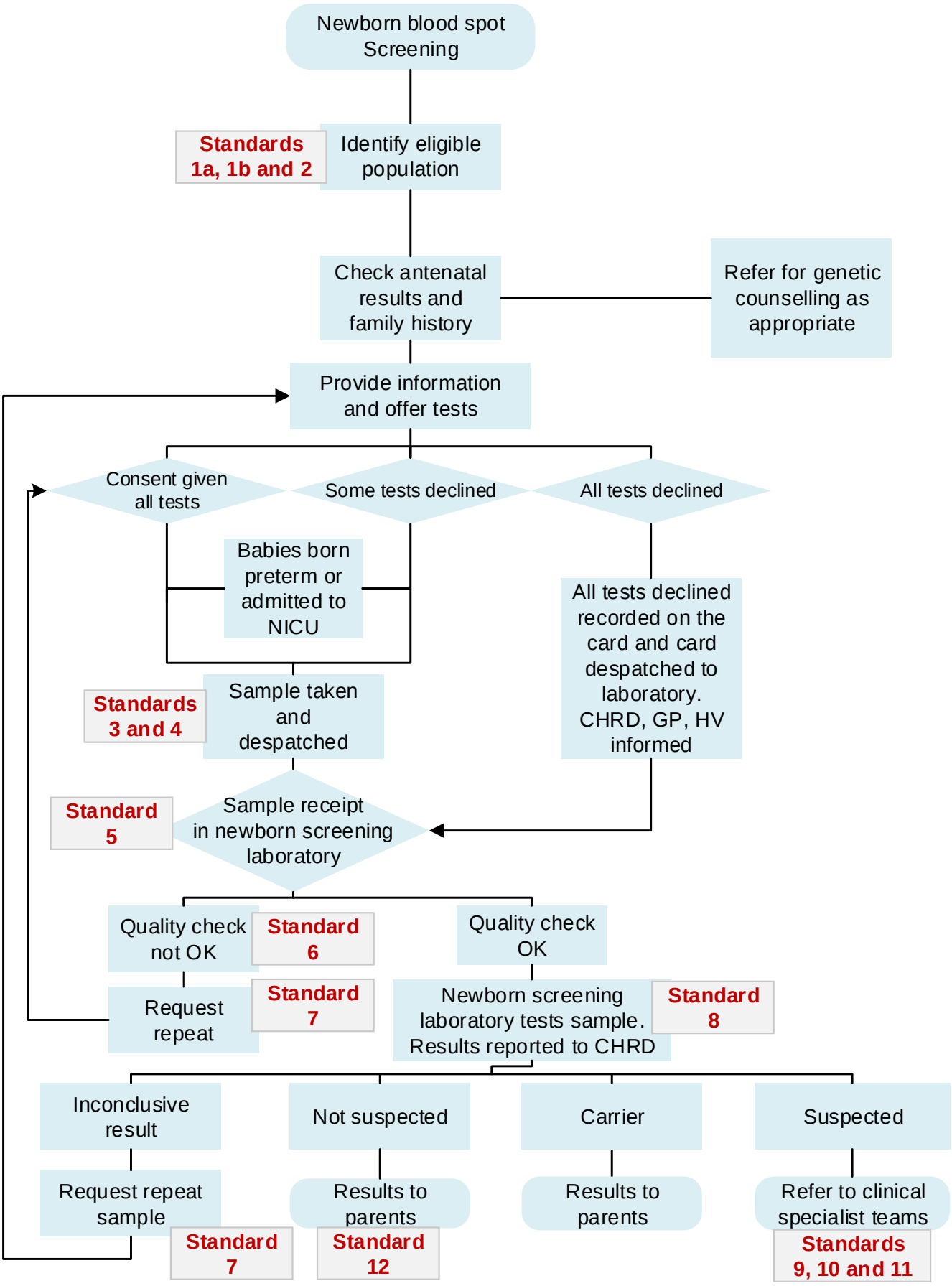
One of the objectives of the NHS NBS Screening Programme is to set national standards (see Table 1 and Figure 1)[1-2]. National standards are important to support the delivery and quality assurance of the screening programme and are used by local commissioners and quality improvement groups. The aim 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

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 Microsoft Excel spreadsheets; these documents 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. Deadlines are given for data collection; however, incomplete data is followed up for a set period prior to analysis.

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.

Aggregate data is collected annually for the previous fiscal year to measure performance against the standards:

- 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

All four UK countries screen for SCD, CF, CHT, PKU and MCADD, and England and Wales began screening for MSUD, IVA, GA1 and HCU in January 2015. This report therefore presents partial data for these four conditions. The report does not include 2014/15 data for Wales on the number of babies tested and screen positive for SCD due to different reporting structures.

Data was returned by CHRDs for 183 CCGs (87%) out of the 211 that existed in 2014/15 in England. Exclusions were made if the data was incomplete (for example the numerator or denominator was missing) – see individual standards for details.

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.

Analysis of screening performance

Overview of UK national screening figures

SCD*	
Babies tested	**742,138**
Screened positive	**287**

PKU	
Babies tested	**780,879**
Screened positive	**71**

CF	
Babies tested	**779,621**
Screened positive	**302**

MCADD	
Babies tested	**780,883**
Screened positive	**58**

CHT	
Babies tested	**780,831**
Screened positive	**622**

MSUD, IVA, GA1, HCU	
Babies tested	**461,445**
Screened positive	**17****

*Does not include data for Wales.

**Includes two screen positive results for one baby. Two further screen positive results were reported but excluded as denominator data was not returned.

***This reflects timeliness of coverage – see Table 5 for completeness of coverage data.

Coverage	
Percentage of babies with a conclusive result for PKU recorded on the CHIS by 17 days of age	**94.6%***** (England) **99.2%***** (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 2014/15

Laboratory	SCD		CF		CHT	
	Number tested	Number of screen positives	Number tested	Number of screen positives	Number tested	Number of screen positives
Bristol	40,670	2	40,702	24	40,702	*25
Cambridge	27,813	2	27,624	8	27,813	25
GOSH	120,284	89	120,676	39	120,676	160
Leeds	40,097	11	42,628	21	42,682	**34
Liverpool	27,292	4	28,521	16	28,521	27
Manchester	55,477	21	55,469	17	55,700	36
Newcastle	33,195	5	33,087	13	33,087	44
Oxford	28,205	13	29,216	9	29,278	19
Portsmouth	36,809	6	36,676	17	37,304	19
SE Thames	56,500	62	56,500	26	56,502	37
Sheffield	72,563	16	72,563	28	72,563	40
SW Thames	52,799	32	51,864	13	51,684	30
West Midlands	69,728	15	70,152	24	70,152	55
England	661,432	278	665,678	255	666,664	551
Northern Ireland	24,363	1	24,290	14	24,398	20
Scotland	56,343	8	56,345	24	56,302	26
Wales	Not reported	Not reported	33,308	9	33,467	25
UK total	742,138	287	779,621	302	780,831	622

Data source: Newborn screening laboratories

*Number of screen positives based on national borderline cut-off (12 further babies were referred based on local borderline cut-off) – presentation of data will be reviewed next year.

**Includes two screen positive babies for whom no clinical data is available.

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 2014/15

	PKU		MCADD		MSUD, IVA, GA1, HCU*	
Laboratory	Number tested	Number of screen positives	Number tested	Number of screen positives	Number tested	Number of screen positives
Bristol	40,702	2	40,702	2	9,413	0
Cambridge	27,813	1	27,813	4	6,517	0
GOSH	120,676	6	120,676	6	120,676	1
Leeds	42,682	5	42,682	4	42,682	1
Liverpool	28,521	3	28,521	1	Not reported	**
Manchester	55,700	5	55,700	6	55,700	3
Newcastle	33,087	2	33,087	6	8,183	0
Oxford	29,278	4	29,278	1	Not reported	**
Portsmouth	37,307	4	37,313	4	Not reported	-
SE Thames	56,500	2	56,500	6	56,500	3
Sheffield	72,563	7	72,563	7	72,563	6
SW Thames	51,684	2	51,684	3	12,171	1
West Midlands	70,152	11	70,152	3	70,152	2
England	666,665	54	666,671	53	454,557	**17
Northern Ireland	24,401	5	24,401	3	-	-
Scotland	56,346	11	56,344	2	-	-
Wales	33,467	1	33,467	0	6,888	0
UK total	780,879	71	780,883	58	461,445	17

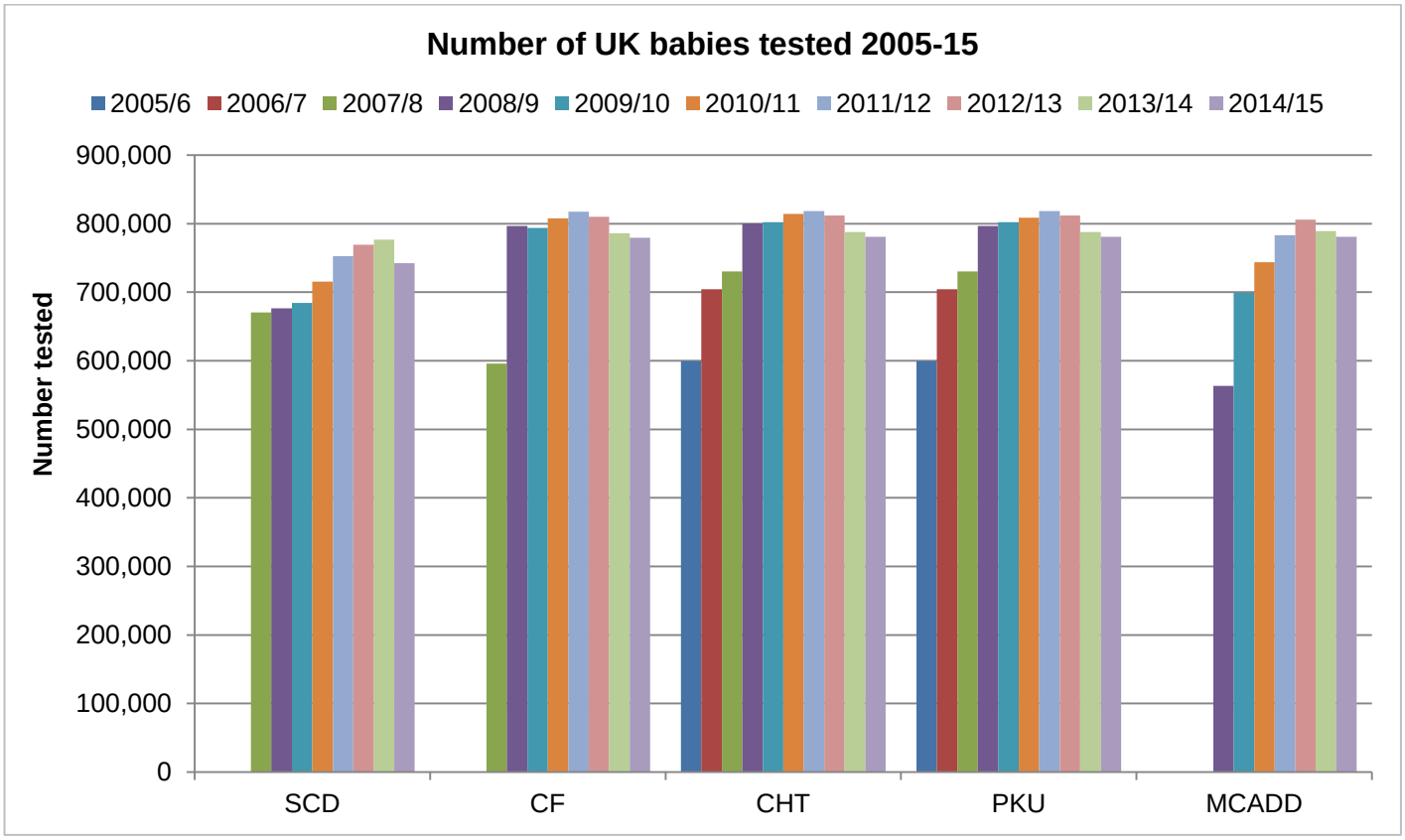
Data source: Newborn screening laboratories

*Screening implemented fully in England and Wales in January 2015. The six laboratories that participated in the expanded screening pilot were asked to provide full data for 2014/15; non-pilot laboratories were asked to provide data for January to March 2015 only.

**Includes two screen positive results for one baby. Two further screen positive results were reported but excluded from the total as denominator data was not returned.

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

Figure 2: Number of UK babies tested for SCD, CF, CHT, PKU and MCADD 2005-15

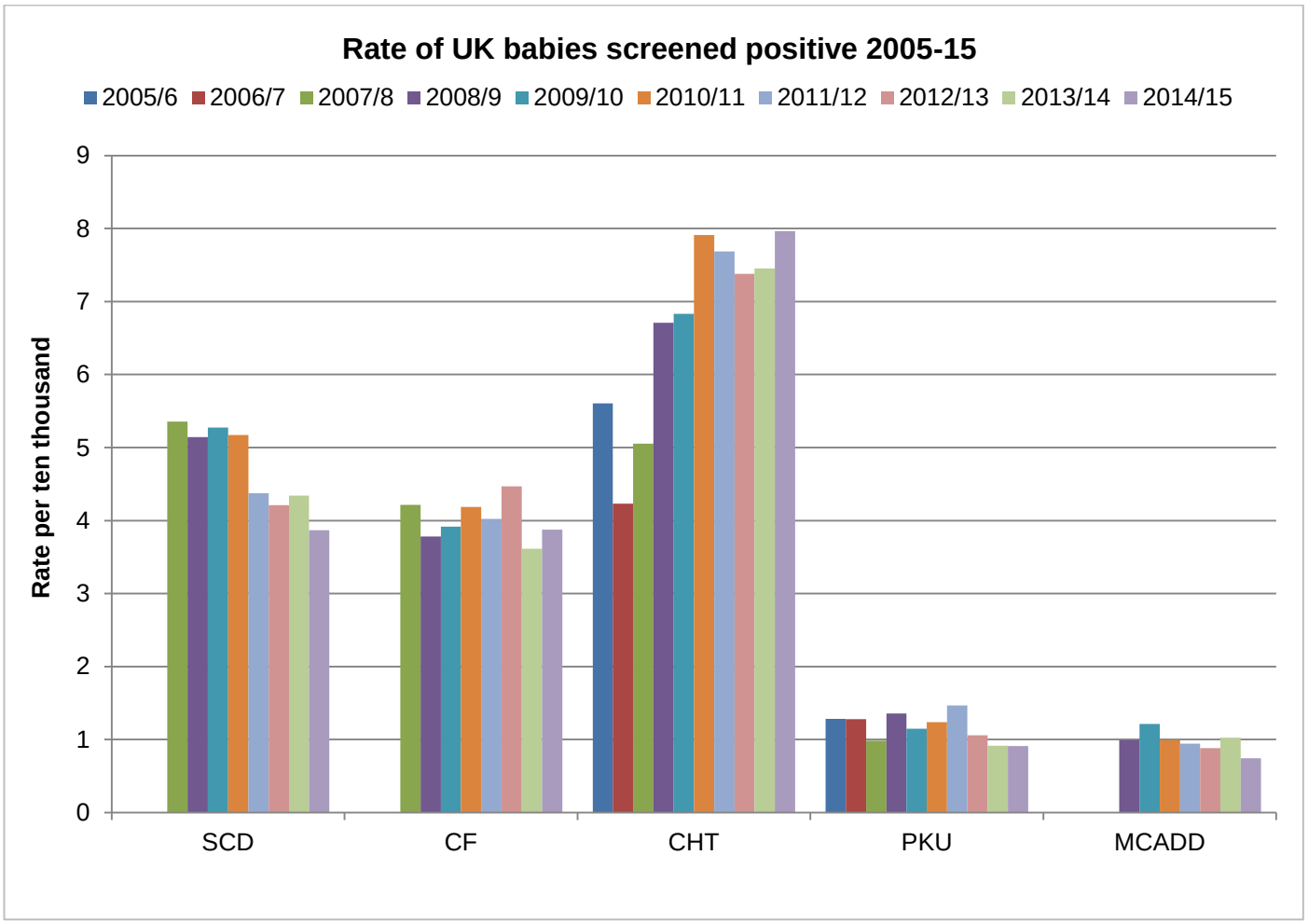


Data source: Newborn screening laboratories

Wales began screening for SCD in June 2013. 2014/15 SCD data for Wales is not included due to different reporting structures.

The Office for National Statistics reported a 4.3% decrease in the number of live births in England and Wales in 2013 compared with 2012, and a 0.5% decrease in 2014 compared with 2013.

Figure 3: Rate per ten thousand of UK babies screened positive for SCD, CF, CHT, PKU and MCADD 2005-15



Data source: Newborn screening laboratories

Wales began screening for SCD in June 2013. 2014/15 SCD data for Wales is not included due to different reporting structures.

Table 4: UK incidence of SCD, CF, CHT, PKU and MCADD

Conditions	Incidence based on research prior to the introduction of the national screening programmes	Incidence	Date range
SCD	1:2,000	1:2,100	2007-15
CF	1:2,500	1:2,500	2007-15
CHT	1:3,000	1:1,500	*2005-15
PKU	1:10,000	1:8,600	2005-15
MCADD	1:10,000	1:10,400	2008-15

Data source: Newborn screening laboratories

*Please note that incidence of CHT for 2008-15 is 1:1,300.

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. The table below shows coverage data with and without the timeframe applied.

Table 5: Completeness of coverage for PKU (CCG responsibility at birth) 2014/15

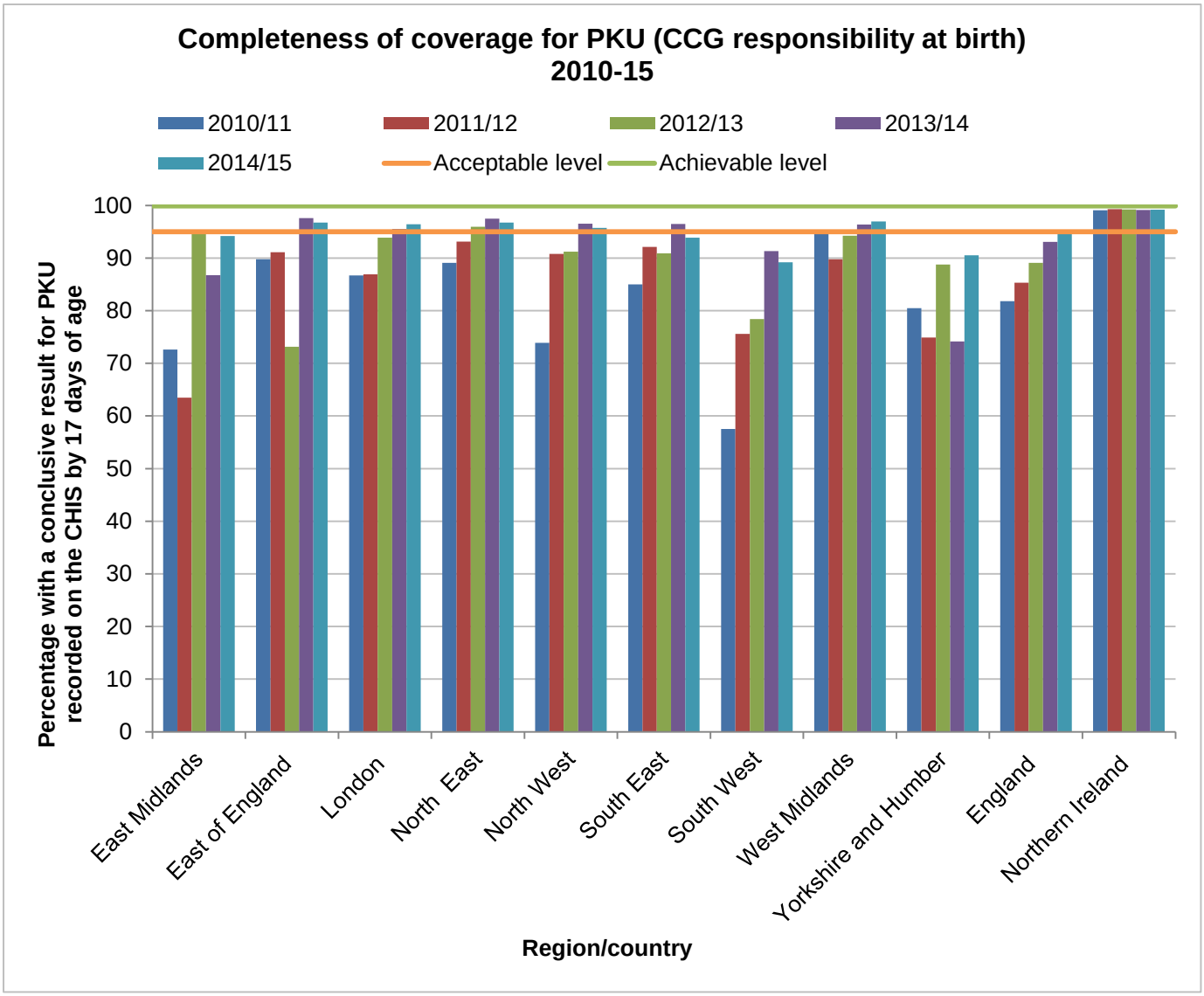
	Babies for whom the CCG/ country is responsible*	Babies tested for PKU		Babies for whom the CCG/ country is responsible	Babies with a conclusive result for PKU** recorded by 17 days of age	
Region/country	n	n	%	n	n	%
East Midlands	47,098	46,955	99.70	47,098	44,360	94.19
East of England	61,514	61,135	99.38	61,514	59,508	96.74
London	84,693	83,408	98.48	88,927	85,762	96.44
North East	21,070	21,064	99.97	21,070	20,385	96.75
North West	69,795	69,512	99.59	73,007	69,898	95.74
South East	88,469	87,799	99.24	88,469	83,024	93.85
South West	49,157	49,062	99.81	49,157	43,843	89.19
West Midlands	56,926	55,805	98.03	56,926	55,190	96.95
Yorkshire and Humber	53,286	53,241	99.92	53,286	48,230	90.51
Unknown region^	43	43	100	43	41	95.34
England	532,051	528,024	99.24	539,497	510,241	94.58
Northern Ireland	23,820	23,812	99.97	23,820	23,626	99.19

Data source: CHRDs

*Three returns were excluded based on missing data. **Status codes 04, 07, 08[3]. ^Not registered with a GP.

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) 2010-15 (measured at 17 days)



Data source: CHRDs

Incomplete returns for 2014/15 were excluded – no exclusions were made in previous years.

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 – 2014/15 data is presented with the timeframe in addition to year-on-year data without the timeframe.

Table 6: Completeness of coverage for PKU (movers in) 2014/15

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 within 21 calendar days	
	n	n	%	n	n	%
East Midlands	1,203	1,108	92.10	1,203	758	63.01
East of England	2,973	2,648	89.07	2,973	2,349	79.01
London	8,958	8,443	94.25	7,607	5,738	75.43
North East	1,126	1,079	95.83	1,126	821	72.91
North West	3,367	2,782	82.63	3,413	2,626	76.94
South East	4,649	4,288	92.23	3,323	2,777	83.57
South West	1,387	1,127	81.25	1,387	654	47.15
West Midlands	1,372	1,256	91.55	931	586	62.94
Yorkshire and Humber	3,374	3,275	97.07	2,979	2,537	85.16
Unknown region^	2	2	100	-	-	-
England	28,411	26,008	91.54	24,942	18,846	75.56
Northern Ireland	343	291	84.84	-	-	-

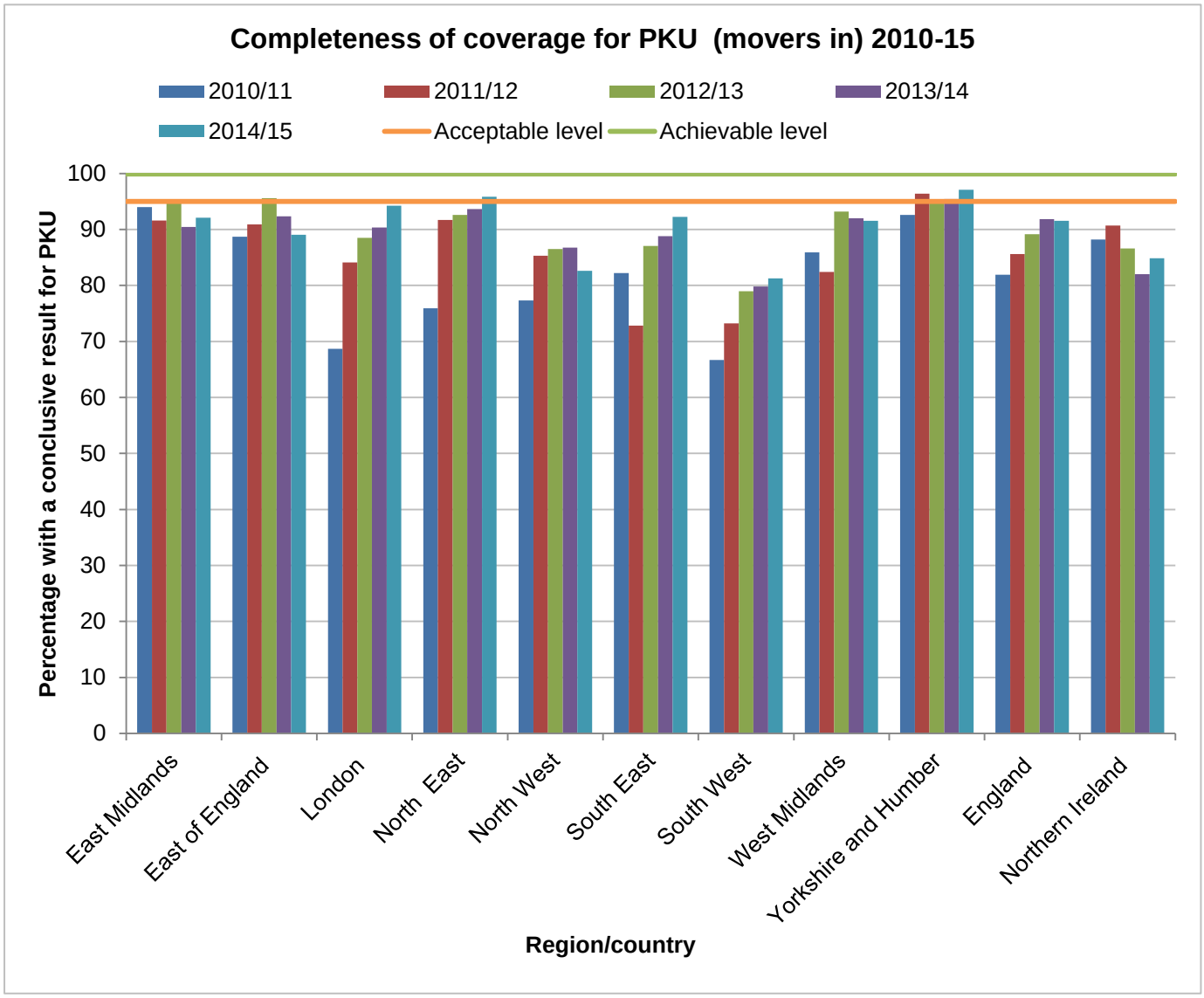
Data source: CHRDs

*12 returns were excluded based on missing data. **33 returns were excluded based on missing data. ***Status codes 04, 07, 08[3]. ^Not registered with a GP.

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

Nine screen positive babies that had their first blood spot sample taken over 28 days of age.

Figure 5: Completeness of coverage for PKU (movers in) 2010-2015 (no timeframe applied)



Data source: CHRDs

Incomplete returns for 2014/15 were excluded – no exclusions were made in previous years.

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 three 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 7: Number and rate per ten thousand babies of declines for PKU (CCG responsibility at birth) 2014/15

Region/country	Babies for whom the CCG/country is responsible*	Declined screening for PKU	
	n	n	Rate per ten thousand
East Midlands	47,098	34	7.22
East of England	61,514	37	6.01
London	84,693	93	10.98
North East	21,070	3	1.42
North West	67,867	40	5.89
South East	88,469	106	11.98
South West	49,157	62	12.61
West Midlands	56,926	28	4.92
Yorkshire and Humber	53,286	30	5.63
Unknown region^	43	0	0
England	530,123	433	8.17
Northern Ireland	23,820	8	3.36

Data source: CHRDs

*Four returns were excluded based on missing data. ^Not registered with a GP.

59 CCGs reported greater than 10 declines per ten thousand and more than five declines in total.

42 CCGs reported zero 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.

Wales reported 19 declines for the sample taken on days 5-8.

Declines: movers in

Table 8: Number and rate per ten thousand babies of declines for PKU (movers in) 2014/15

Region/country	Babies for whom the CCG/country is responsible*	Declined screening for PKU	
	n	n	Rate per ten thousand
East Midlands	1,203	37	307.56
East of England	2,973	247	830.81
London	9,620	12	12.47
North East	1,126	22	195.38
North West	3,497	55	157.28
South East	5,107	71	139.02
South West	1,387	40	288.39
West Midlands	1,998	48	240.24
Yorkshire and Humber	3,374	55	163.01
Unknown region^	2	0	0
England	30,287	587	193.81
Northern Ireland	343	51	1486.88

Data source: CHRDs

*16 returns were excluded based on missing data. ^Not registered with a GP.

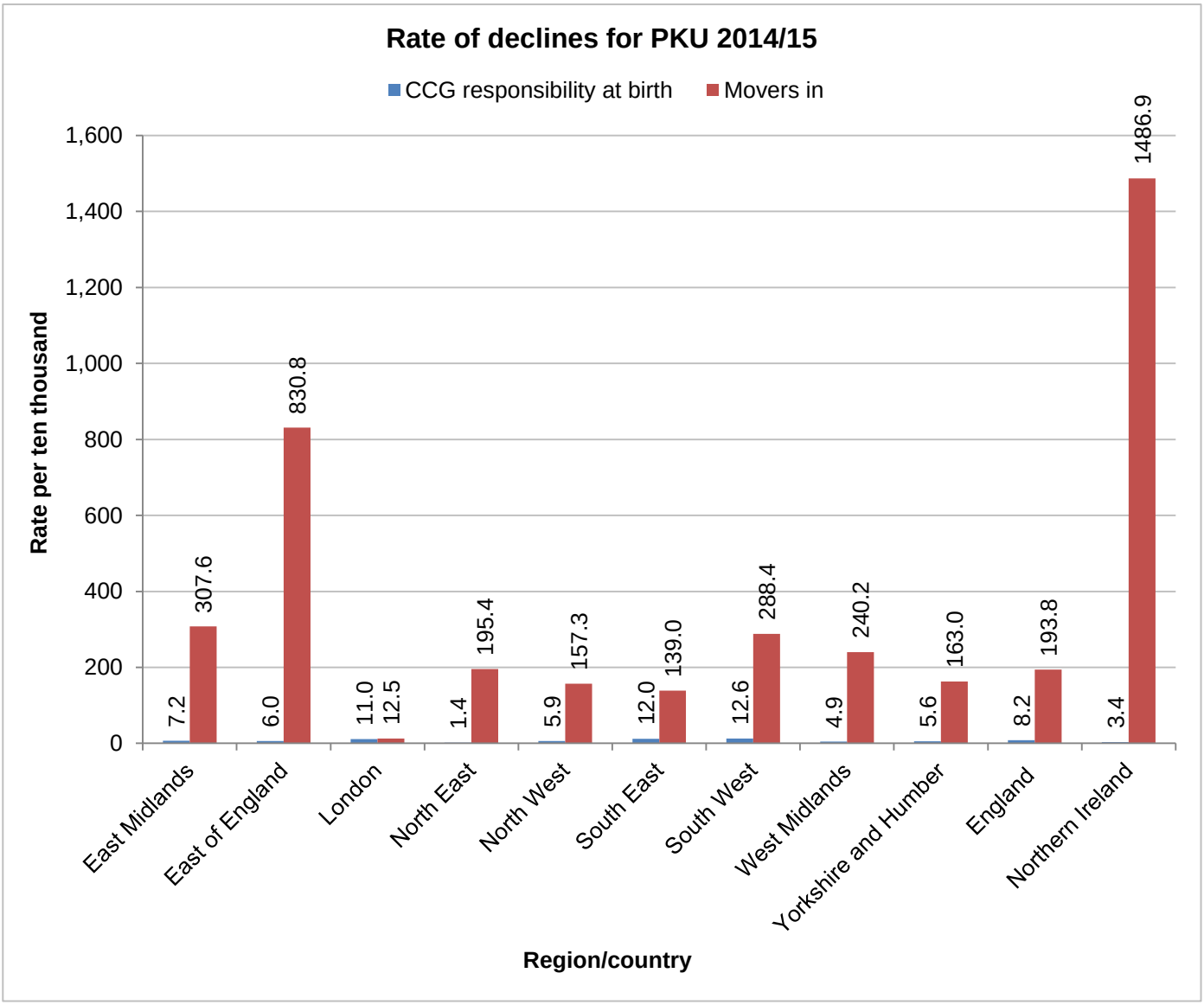
Northern Ireland has a higher rate of declines amongst movers in because all babies under a year of age that move in are offered screening – a proportion of parents will decline if they believe that their child has already been screened. In England these babies are only offered screening if they do not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

In England, processes for recording declines for movers in vary between regions.

Wales reported 89 declines following work with interim failsafe to ensure movers in aged less than one year have been offered screening.

Declines: Comparing CCG responsibility at birth and movers in populations

Figure 6: Rate per ten thousand babies of declines for PKU – CCG responsibility at birth and movers in populations 2014/15



Data source: CHRDs

CHRD process data

Table 9: Receipt, recording and despatch of results by CHRDs 2014/15

Region/ country	Number of CHRDs* that:												
	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
	n	%	n	%	n	%	n	%	n	%	n	%	n
East Midlands	5	26.3	14	73.7	0	0	19	100	19	100	14	73.7	19
East of England	3	13.6	21	95.5	0	0	22	100	22	100	17	77.3	22
London	8	30.8	25	96.2	9	34.6	25	96.2	25	96.2	18	69.2	26
North East	6	66.7	8	88.9	0	0	9	100	9	100	9	100	9
North West	25	86.2	27	93.1	7	24.1	29	100	28	96.6	8	27.6	29
South East	27	79.4	26	76.5	18	52.9	34	100	34	100	18	52.9	34
South West	11	100	9	81.8	1	9.1	11	100	11	100	8	72.7	11
West Midlands	4	19.0	1	4.8	16	76.2	21	100	17	81.0	9	42.9	21
Yorkshire and Humber	11	47.8	10	43.5	18	78.3	23	100	23	100	22	95.7	23
England	100	51.3	141	72.3	69	35.4	194	99.5	188	96.4	123	63.1	195
Northern Ireland	1	100	0	0	0	0	1	100	1	100	0	0	1

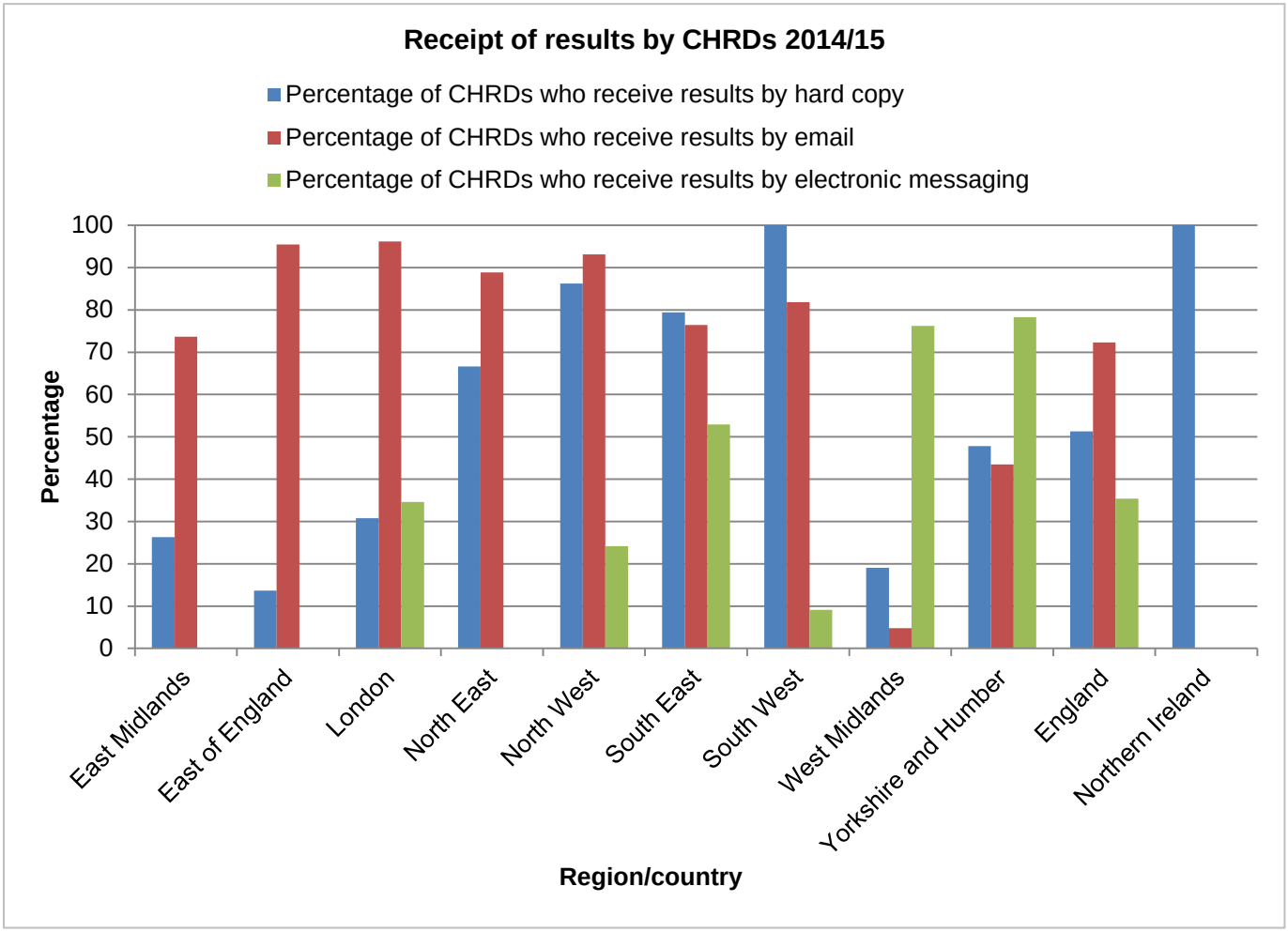
Data source: CHRDs

*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 2014/15



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 10: Number and percentage of CHRDs that search for missing results at 17 days, 14 days and 'other' 2014/15

	Number of CHRDs* that perform:										
	daily checks at 17 days		daily checks at 14 days		other – meeting standard		other – not meeting standard		no data		total
Region/country	n	%	n	%	n	%	n	%	n	%	n
East Midlands	0	0	11	57.9	4	21.1	2	10.5	2	10.5	19
East of England	6	27.3	14	63.6	1	4.5	1	4.5	0	0	22
London	5	19.2	20	76.9	0	0	0	0	1	3.8	26
North East	0	0	6	66.7	1	11.1	2	22.2	0	0	9
North West	6	20.7	22	75.9	0	0	1	3.4	0	0	29
South East	1	2.9	18	52.9	15	44.1	0	0	0	0	34
South West	0	0	9	81.8	2	18.2	0	0	0	0	11
West Midlands	13	61.9	8	38.1	0	0	0	0	0	0	21
Yorkshire and Humber	7	30.4	7	30.4	2	8.7	6	26.1	1	4.3	23
England	39	19.9	115	58.7	26	13.3	12	6.1	4	2.0	195
Northern Ireland	0	0	0	0	1	100	0	0	0	0	1

Data source: CHRDs

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

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 11: Use of the baby's NHS number and bar-coded label 2014/15

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	45,304	45,188	99.74	39,775	87.80
Cambridge	27,787	27,738	99.82	20,822	74.93
GOSH	127,278	126,852	99.67	**Not reported	-
Leeds	45,754	44,020	96.21	33,155	72.46
Liverpool	28,933	28,847	99.70	14,455	49.96
Manchester	58,934	58,632	99.49	***37,835	64.20
Newcastle	34,813	34,635	99.49	31,539	90.60
Oxford	31,558	31,462	99.70	24,570	77.86
Portsmouth	39,049	38,417	98.38	25,802	66.08
SE Thames	59,624	59,362	99.56	***46,083	77.29
Sheffield	75,979	75,645	99.56	55,518	73.07
SW Thames	52,853	52,775	99.85	39,915	75.52
West Midlands	73,630	73,488	99.81	58,282	79.16
England	701,496	697,061	99.37	427,751	74.49
Northern Ireland*	-	-	-	-	-
Scotland	59,742	59,135	98.98	-	-
Wales	33,515	33,304	99.37	-	-
UK	794,753	789,500	99.34	427,751	74.49

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.

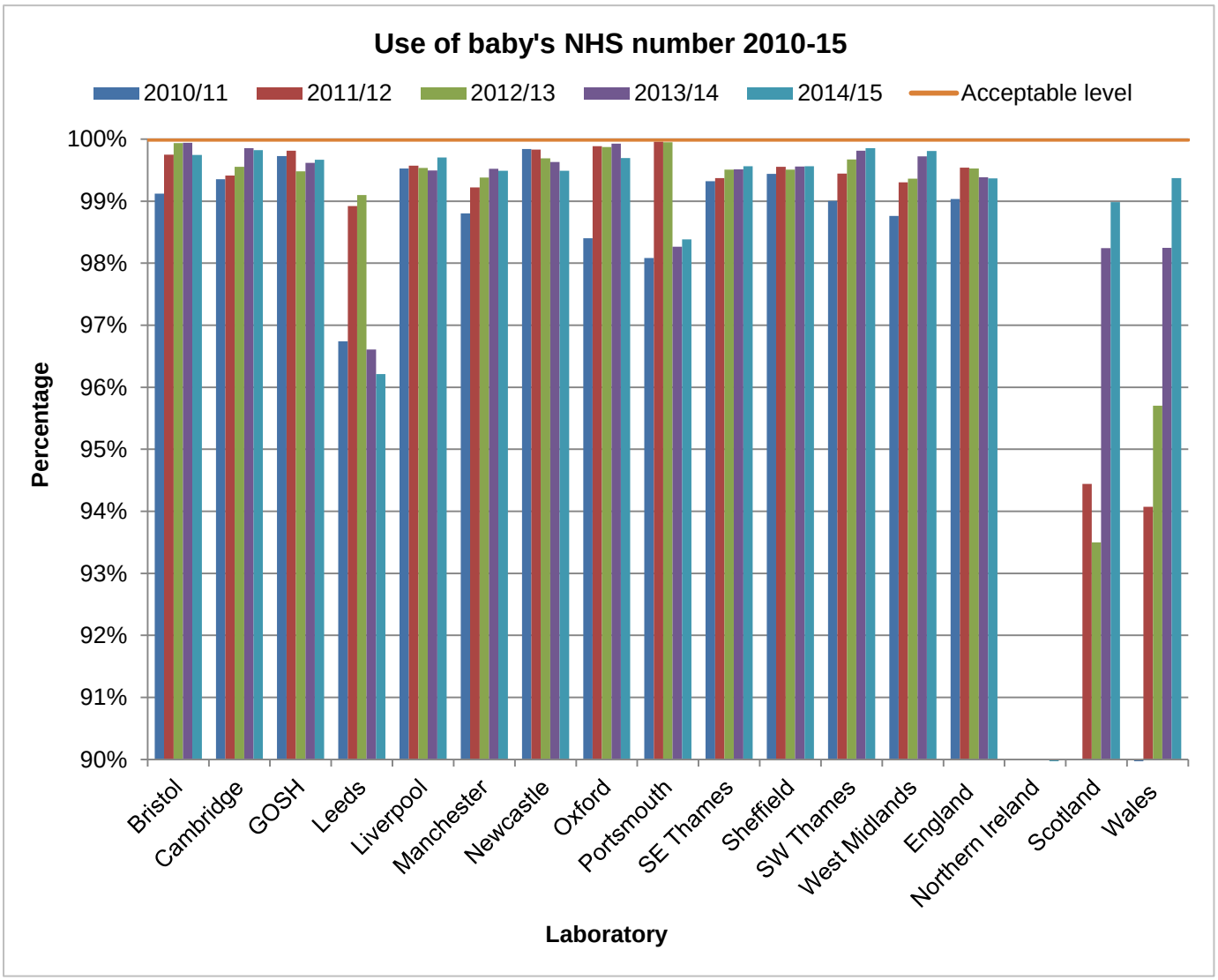
**GOSH unable to report data due to laboratory information management system limitations.

***Manchester unable to provide full data due to an IT error – this figure was extrapolated from an estimated percentage based on Q3 and Q4 data for 2014/15 only. SE Thames unable to provide full data – this figure was extrapolated from a percentage based on March 2015 data only.

Data on areas returning the lowest and highest percentage of samples including the baby's NHS number has not been presented this year. The lowest performing area overall was 83.33% and represents a small number of samples taken.

Figure 8: Percentage of blood spot cards including the baby's NHS number 2010-15

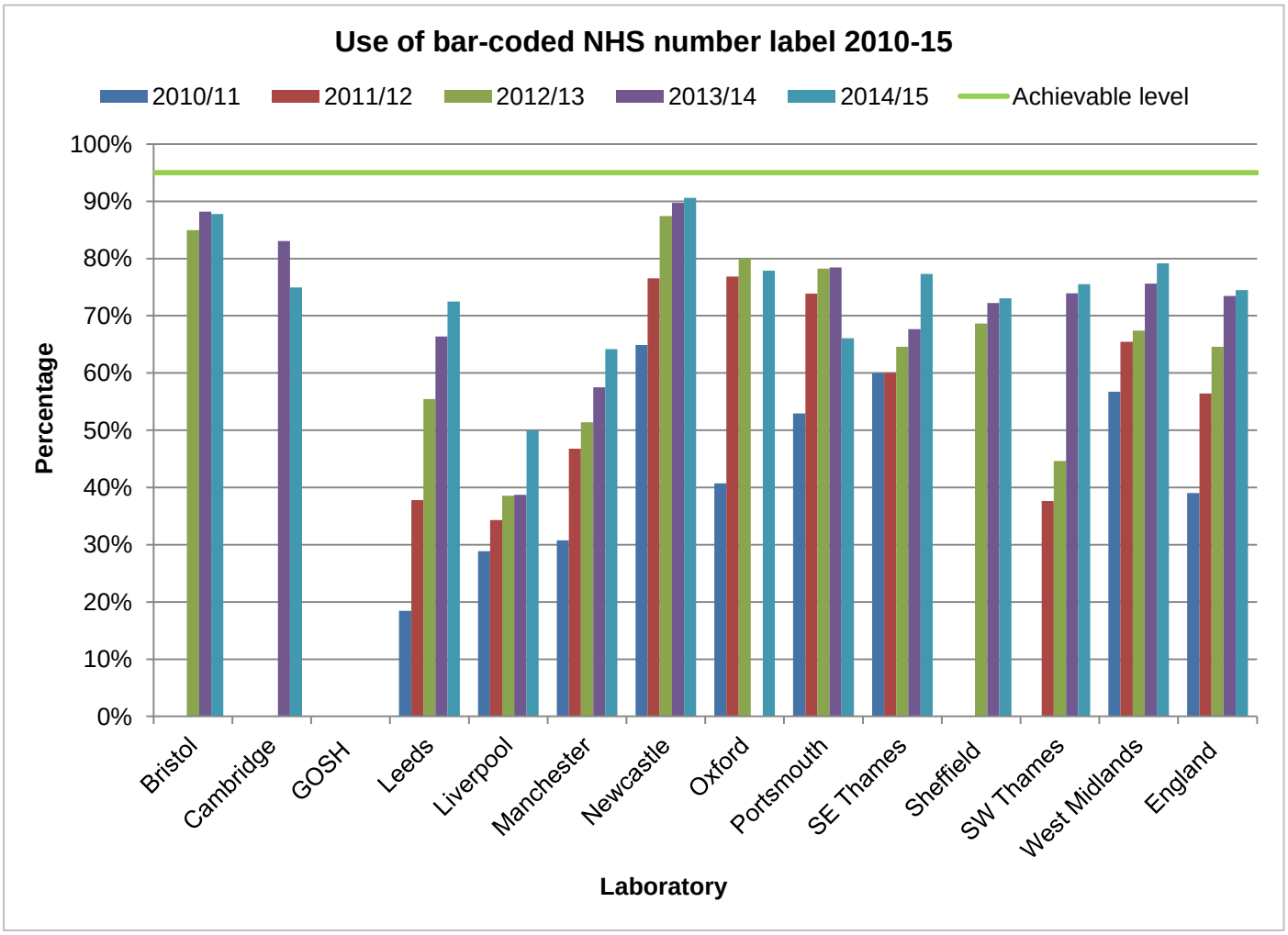
Please note that the Y axis does not begin at zero.



Data source: Newborn screening laboratories

Samples from Jersey and Guernsey (where there is no equivalent NHS number) are processed by Leeds and Portsmouth respectively and included in the denominator for these laboratories.

Figure 9: Percentage of blood spot cards including a bar-coded NHS number label 2010-15



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).

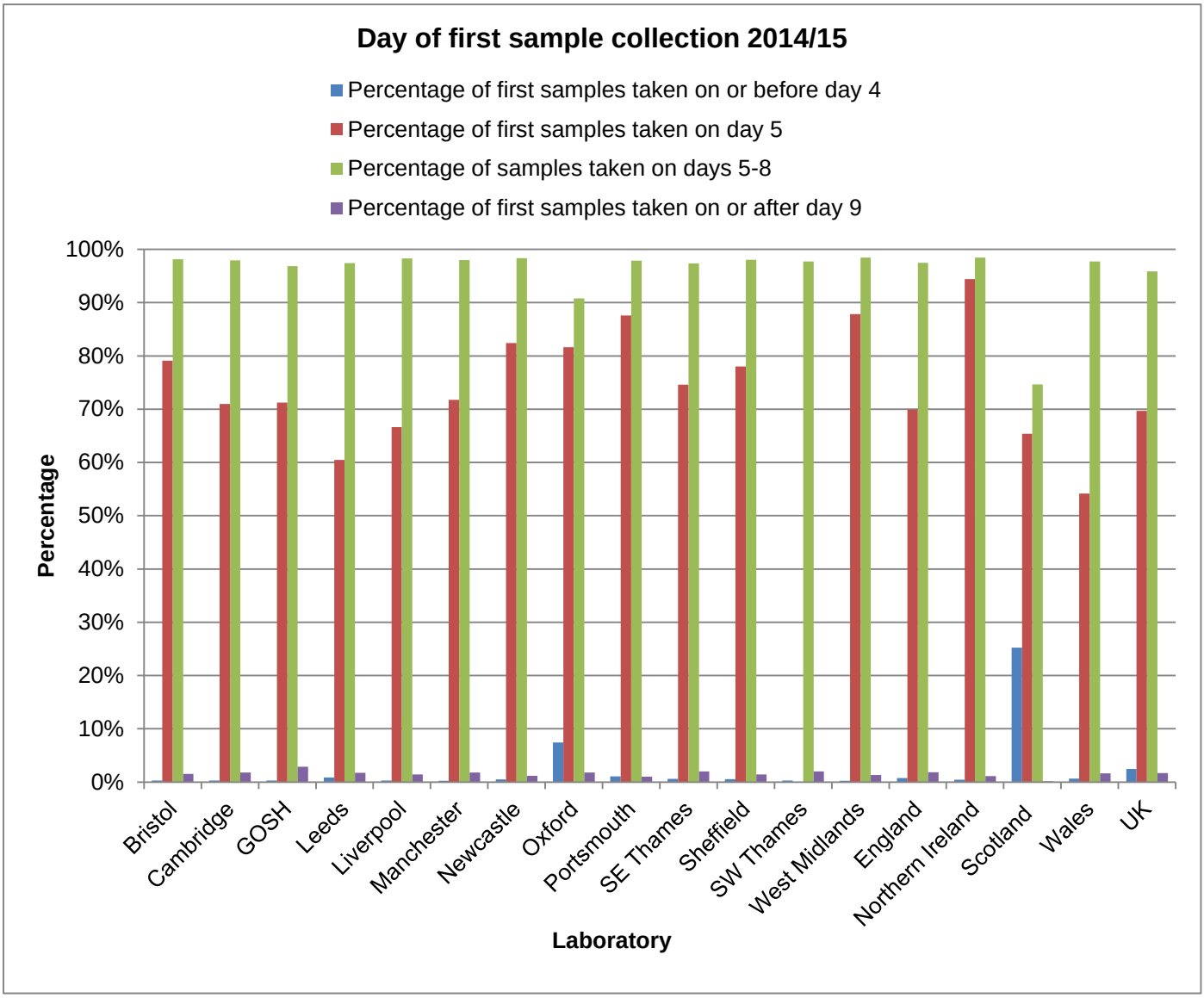
Table 12: Day of first sample collection 2014/15

	First samples taken:							
	on or before day 4		on day 5		on days 5-8		on or after day 9	
Laboratory	n	%	n	%	n	%	n	%
Bristol	121	0.30	31,665	79.09	39,293	98.14	623	1.56
Cambridge	71	0.27	18,839	70.99	25,986	97.93	479	1.81
GOSH	347	0.29	85,448	71.22	116,183	96.84	3,443	2.87
Leeds	381	0.87	26,435	60.45	42,593	97.40	757	1.73
Liverpool	76	0.27	18,555	66.61	27,384	98.31	395	1.42
Manchester	131	0.24	39,672	71.77	54,168	97.99	980	1.77
Newcastle	157	0.48	27,101	82.42	32,342	98.36	382	1.16
Oxford	2,135	7.41	23,519	81.61	26,164	90.79	520	1.80
Portsmouth	403	1.09	32,354	87.57	36,163	97.88	380	1.03
SE Thames	334	0.60	41,862	74.59	54,656	97.38	1,135	2.02
Sheffield	382	0.53	55,778	78.01	70,112	98.06	1,008	1.41
SW Thames	158	0.31	Not reported	Not reported	50,467	97.71	1,027	1.99
West Midlands	175	0.25	60,428	87.82	67,735	98.44	901	1.31
England	4,871	0.74	461,656	69.93	643,246	97.44	12,030	1.82
Northern Ireland	105	0.43	23,038	94.41	24,027	98.47	269	1.10
Scotland*	14,043	25.21	36,428	65.39	41,584	74.65	80	0.14
Wales	210	0.63	18,152	54.16	32,749	97.71	556	1.66
UK	19,229	2.49	539,274	69.69	741,606	95.84	12,935	1.67

Data source: Newborn screening laboratories

*Scotland allows samples to be taken on day 4.

Figure 10: Day of first sample collection 2014/15



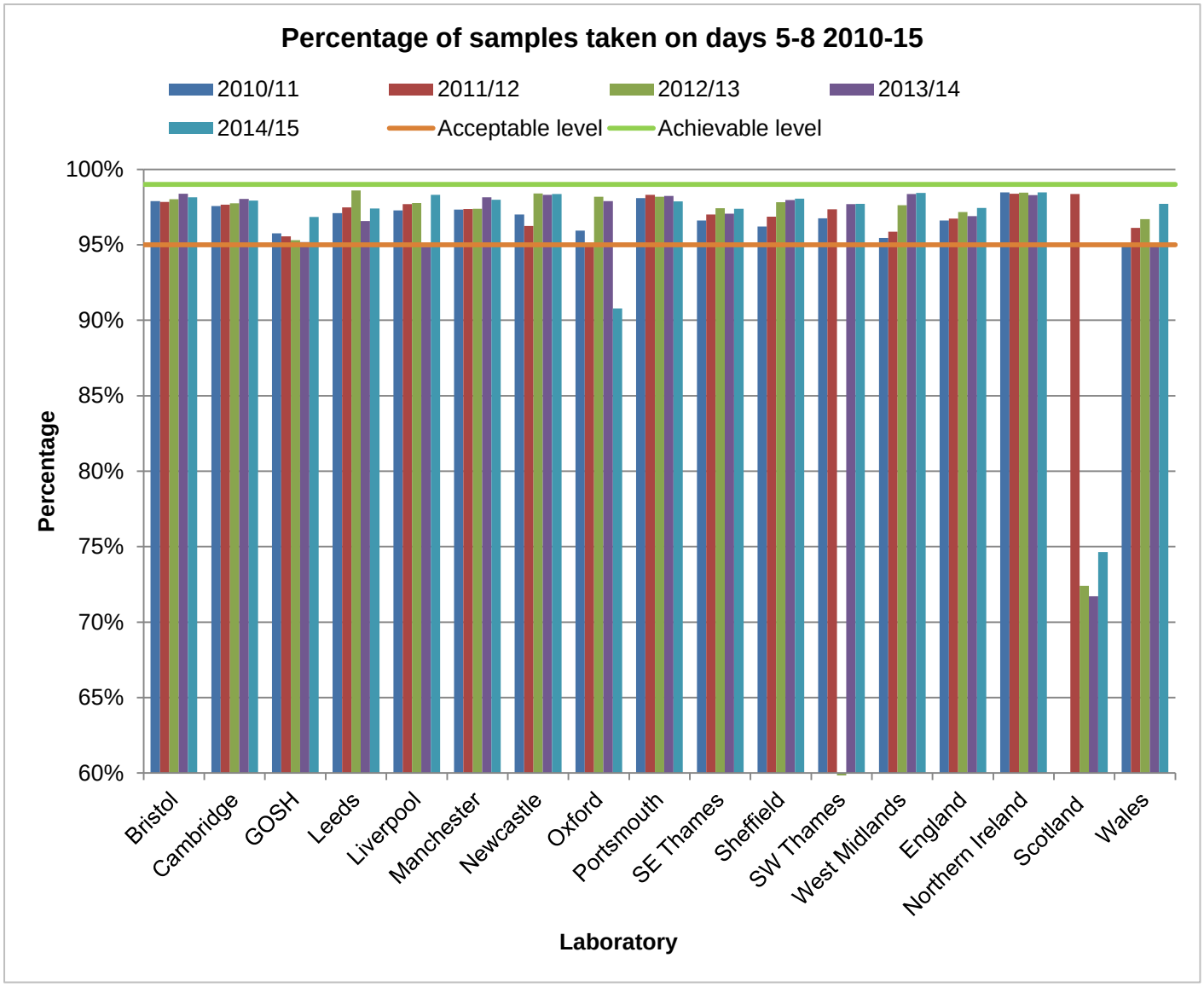
Data source: Newborn screening laboratories

Scotland allows samples to be taken on day 4.

Data on areas returning the lowest and highest percentage of samples taken on days 5-8 has not been presented this year. The lowest performing area overall was 66.67% and represents a small number of samples.

Figure 11: Percentage of samples taken on days 5-8 2010-15

Please note that the Y axis does not begin at zero.



Data source: Newborn screening laboratories

Scotland allows samples to be taken on day 4.

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.

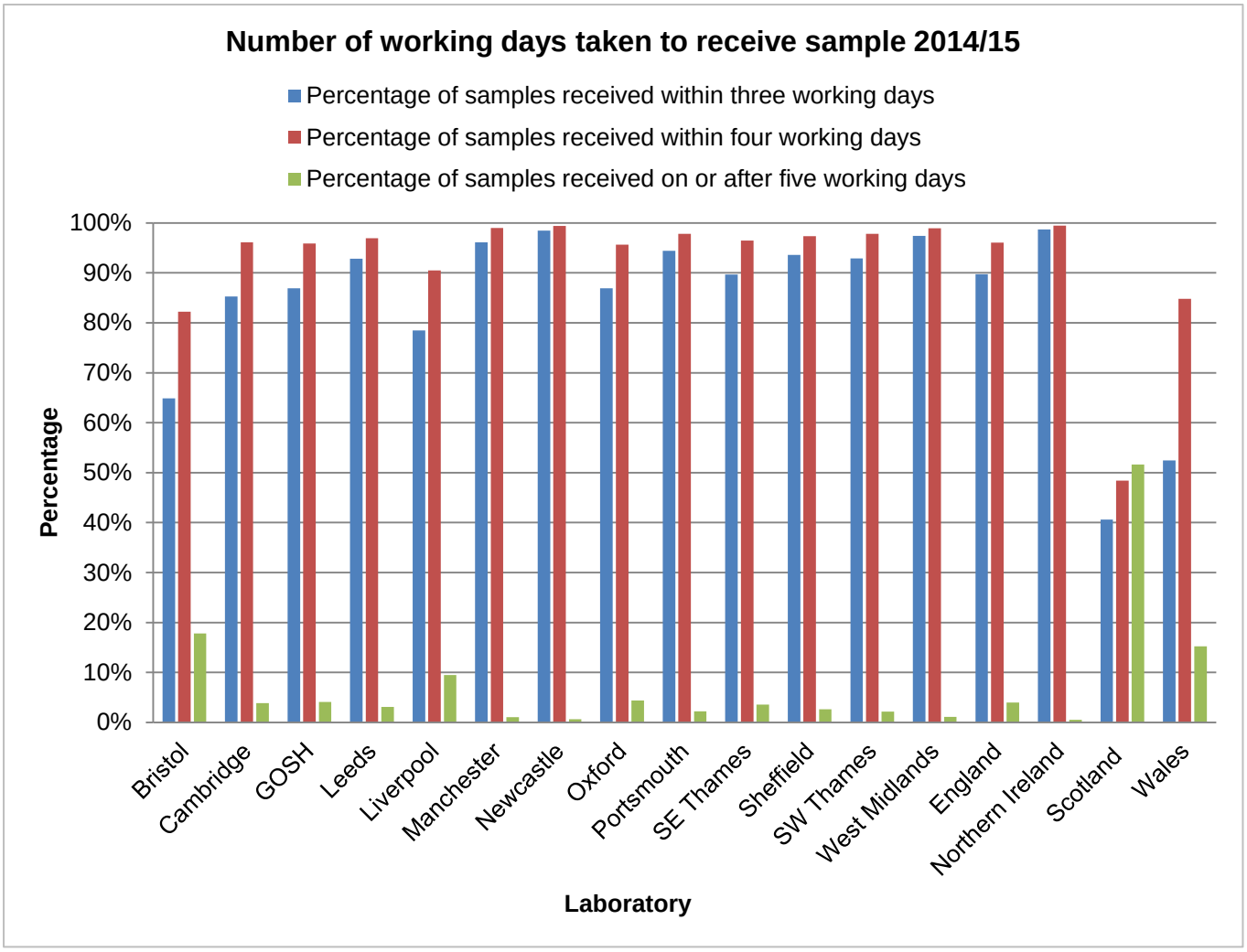
Achievable level
Equal to or greater than 99% of all samples received within three working days of sample collection.

Table 13: Number of working days taken to receive sample 2014/15

	Samples received:					
	within three working days		within four working days		on or after five working days	
Laboratory	n	%	n	%	n	%
Bristol	29,380	64.85	37,240	82.20	8,064	17.80
Cambridge	23,500	85.30	26,486	96.14	1,063	3.86
GOSH	110,115	86.93	121,485	95.91	5,181	4.09
Leeds	42,486	92.86	44,343	96.92	1,411	3.08
Liverpool	22,706	78.48	26,189	90.52	2,744	9.48
Manchester	55,352	96.10	57,011	98.98	588	1.02
Newcastle	32,460	98.48	32,761	99.39	201	0.61
Oxford	27,537	86.91	30,301	95.63	1,385	4.37
Portsmouth	36,860	94.43	38,171	97.79	862	2.21
SE Thames	53,134	89.65	57,166	96.45	2,103	3.55
Sheffield	70,487	93.61	73,309	97.36	1,991	2.64
SW Thames	49,092	92.88	51,708	97.83	1,145	2.17
West Midlands	71,920	97.38	73,047	98.91	806	1.09
England	625,029	89.70	669,217	96.05	27,544	3.95
Northern Ireland	26,196	98.72	26,391	99.45	145	0.55
Scotland	42,697	40.59	50,902	48.39	54,292	51.61
Wales	17,584	52.47	28,415	84.78	5,100	15.22

Data source: Newborn screening laboratories

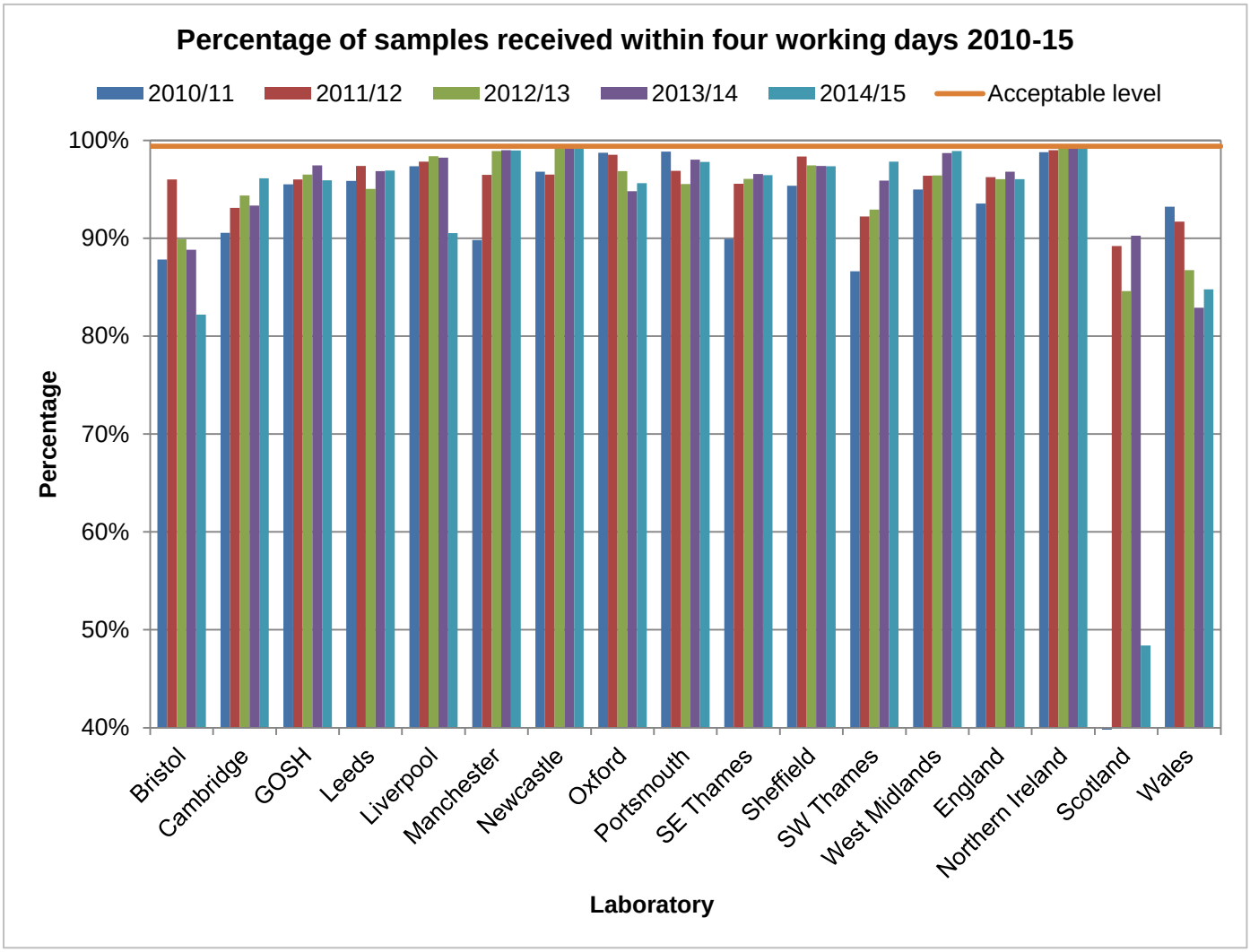
Figure 12: Number of working days taken to receive sample 2014/15



Data source: Newborn screening laboratories

Figure 13: Percentage of samples received within four working days 2010-15

Please note that the Y axis does not begin at zero.



Data source: Newborn screening laboratories

There is anecdotal evidence that sample transport times are continuing to increase in some areas.

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%.

Laboratories have accepted blood spot cards of varying quality. New consensus guidelines were implemented in England and Wales in April 2015 and avoidable repeat rates will be more comparable from 2015/16.

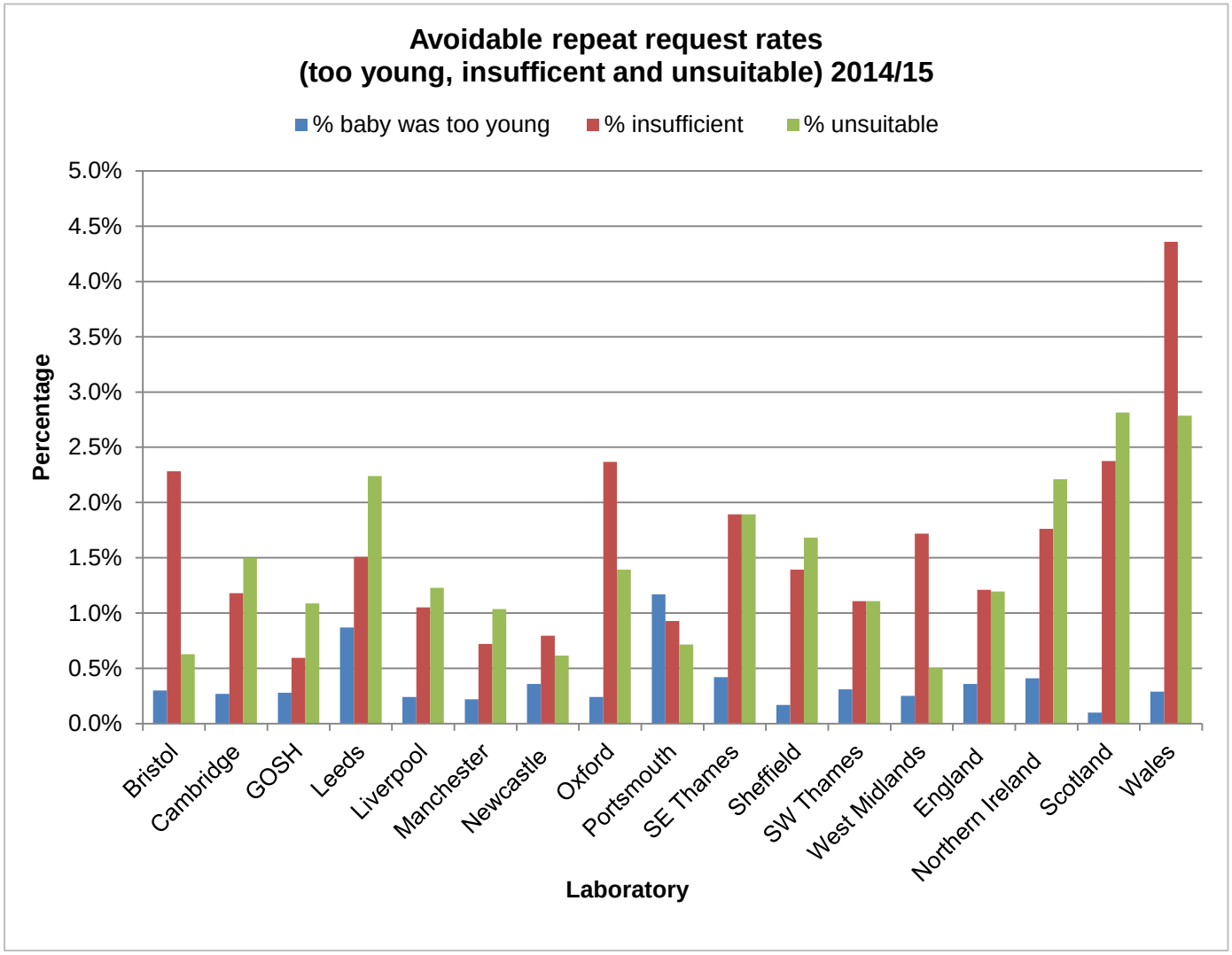
Table 14: Avoidable repeat request rates 2014/15

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	40,037	121	0.30	914	2.28	252	0.63	3.21
Cambridge	26,776	71	0.27	316	1.18	401	1.50	2.94
GOSH	120,552	335	0.28	718	0.60	1,310	1.09	1.96
Leeds	43,767	381	0.87	660	1.51	980	2.24	4.62
Liverpool	27,855	68	0.24	293	1.05	342	1.23	2.52
Manchester	55,351	121	0.22	399	0.72	574	1.04	1.98
Newcastle	33,007	118	0.36	262	0.79	203	0.62	1.77
Oxford	28,819	69	0.24	682	2.37	401	1.39	4.00
Portsmouth	36,890	431	1.17	342	0.93	264	0.72	2.81
SE Thames	56,500	237	0.42	1,070	1.89	1,070	1.89	3.38
Sheffield	72,118	121	0.17	1,004	1.39	1,214	1.68	3.24
SW Thames	51,676	158	0.31	572	1.11	572	1.11	2.65
West Midlands	69,728	175	0.25	1,199	1.72	350	0.50	2.47
England	663,076	2,406	0.36	8,035	1.21	7,933	1.20	2.77
Northern Ireland	24,557	100	0.41	433	1.76	543	2.21	4.38
Scotland	56,746	57	0.10	1,348	2.38	1,597	2.81	5.29
Wales	33,515	96	0.29	1,461	4.36	934	2.79	7.43

Data source: Newborn screening laboratories

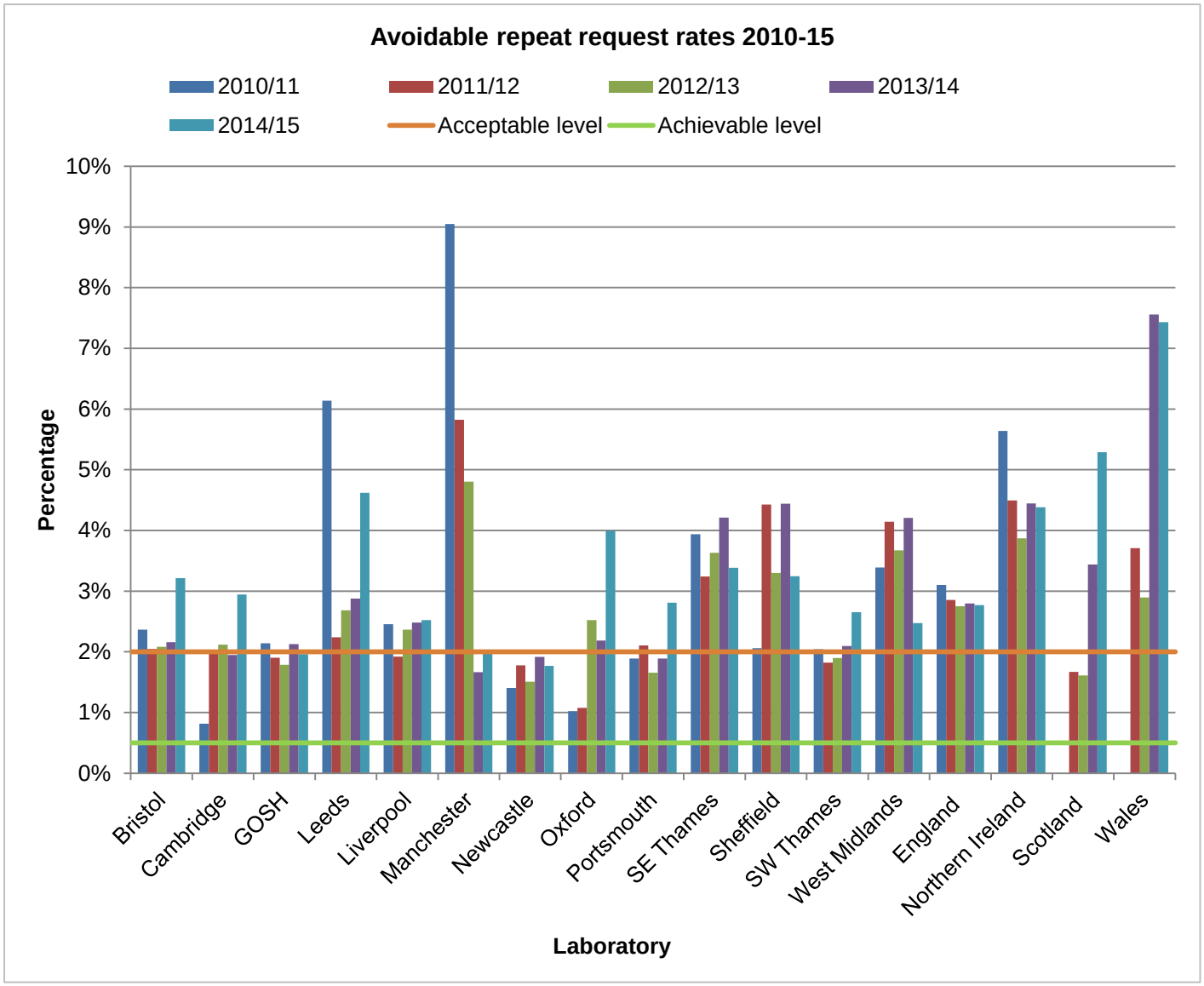
*Not all English laboratories ask for a repeat when the first sample was taken on or before day 4.

Figure 14: Avoidable repeat request rates (too young, insufficient and unsuitable) 2014/15



Data source: Newborn screening laboratories

Figure 15: Avoidable repeat request rates 2010-15



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, 2013-15 data includes repeat requests due to samples taken when the baby was too young, insufficient and unsuitable.

Four samples were reported to be repeated in 2014/15 due to ‘unsatisfactory analysis’.

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 four working days of sample receipt by screening laboratory.

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

Table 15: Numbers of samples processed within the standard in the UK 2014/15

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	71	71	100	71	100
CHT	622 (601*)	573	95.3	570	94.8
MCADD	58	58	100	57	98.3

Data source: Newborn screening laboratories

*Data only available for 601 babies.

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

The following is an extract from the *NHS Sickle Cell and Thalassaemia Screening Programme Data Report 2014/15: Trends and performance analysis* (England only report)[4]:

Data indicates that approximately 99% of screen positive babies have their initial clinical referral by 8 weeks of age (median 16 days), which suggests that programme standard NP4 (effective follow-up of infants with positive screening results) to be both realistic and achievable. Approximately 86% of screen positive babies are reported to have had their first visit to a paediatrician at a specialist health team or local health team by 90 days (median 58 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

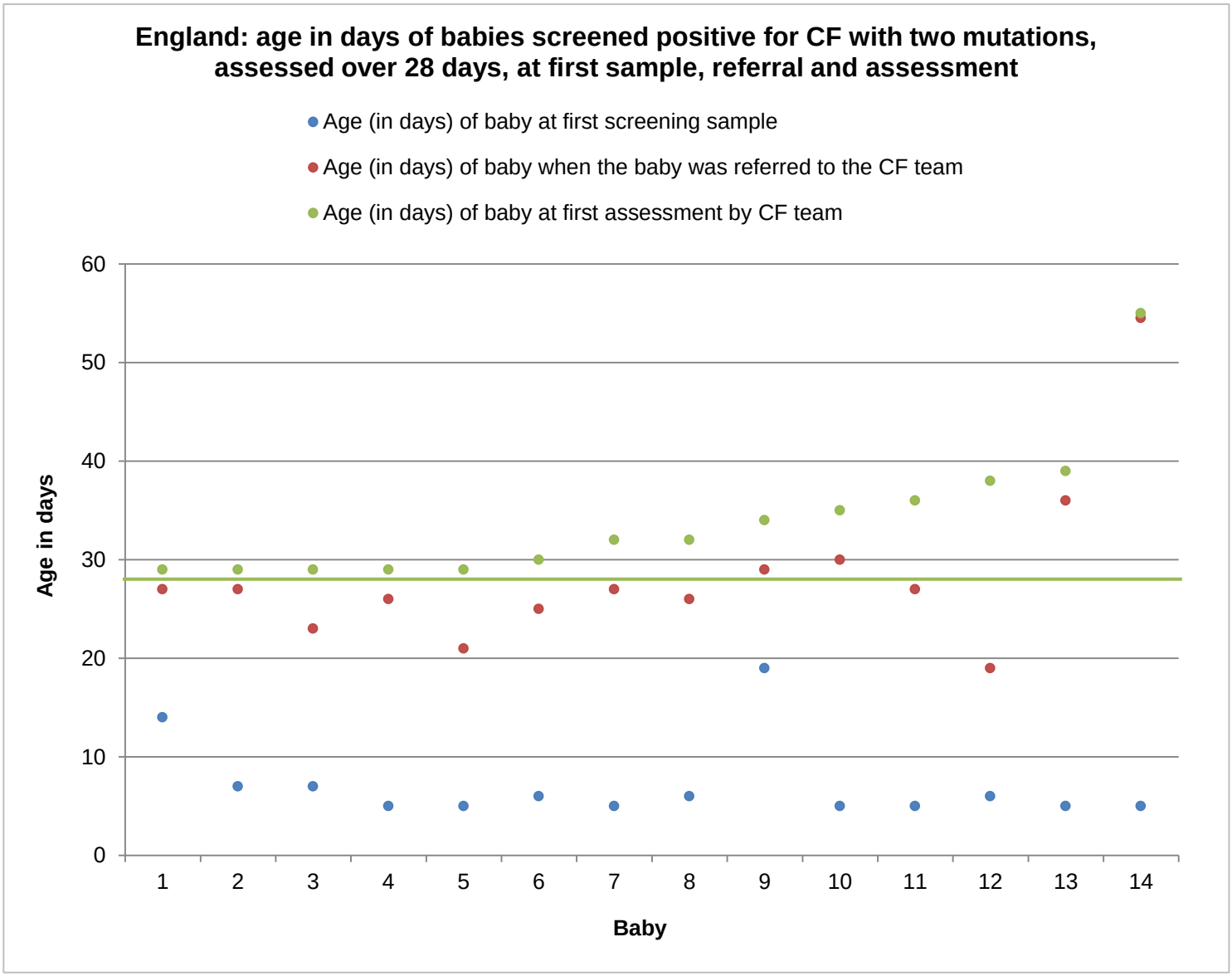
Table 16: Timeliness of appointment and outcome for CF screen positive babies with two mutations 2014/15

	England	Northern Ireland	Scotland	Wales
Number of CF screen positive babies with two mutations	**179**	**9**	**17**	**8**
Number clinically diagnosed before screening (excluded from following age data)	37	1	4	2
Number of babies with age at first appointment reported	104	8	13	6
Number seen ≤ 28 days (% of known data)	90 (87%)	7 (88%)	11 (85%)	6 (100%)
All babies mean age at first appointment	22 days	23 days	24 days	19 days
All babies median age at first appointment	22 days	23 days	22 days	23 days
Age range at first appointment	8-55 days	19-30 days	17-40 days	3-27 days
Number of babies with age at first appointment not reported	38	0	0	0
Inpatient	1	-	-	-
Not reported	37	-	-	-
Outcome				
Confirmed	140	7	17	8
CF SPID	6	2	0	0
Excluded	0	0	0	0
Not reported	33	0	0	0

Data source: Newborn screening laboratories

Note that different screening and diagnostic protocols are followed in the UK – see Figures 19-22.

Figure 16: England: CF screen positive babies with two mutations assessed over 28 days 2014/15



Data source: Newborn screening laboratories

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 17: Timeliness of appointment and outcome for CF screen positive babies with one or no mutations 2014/15

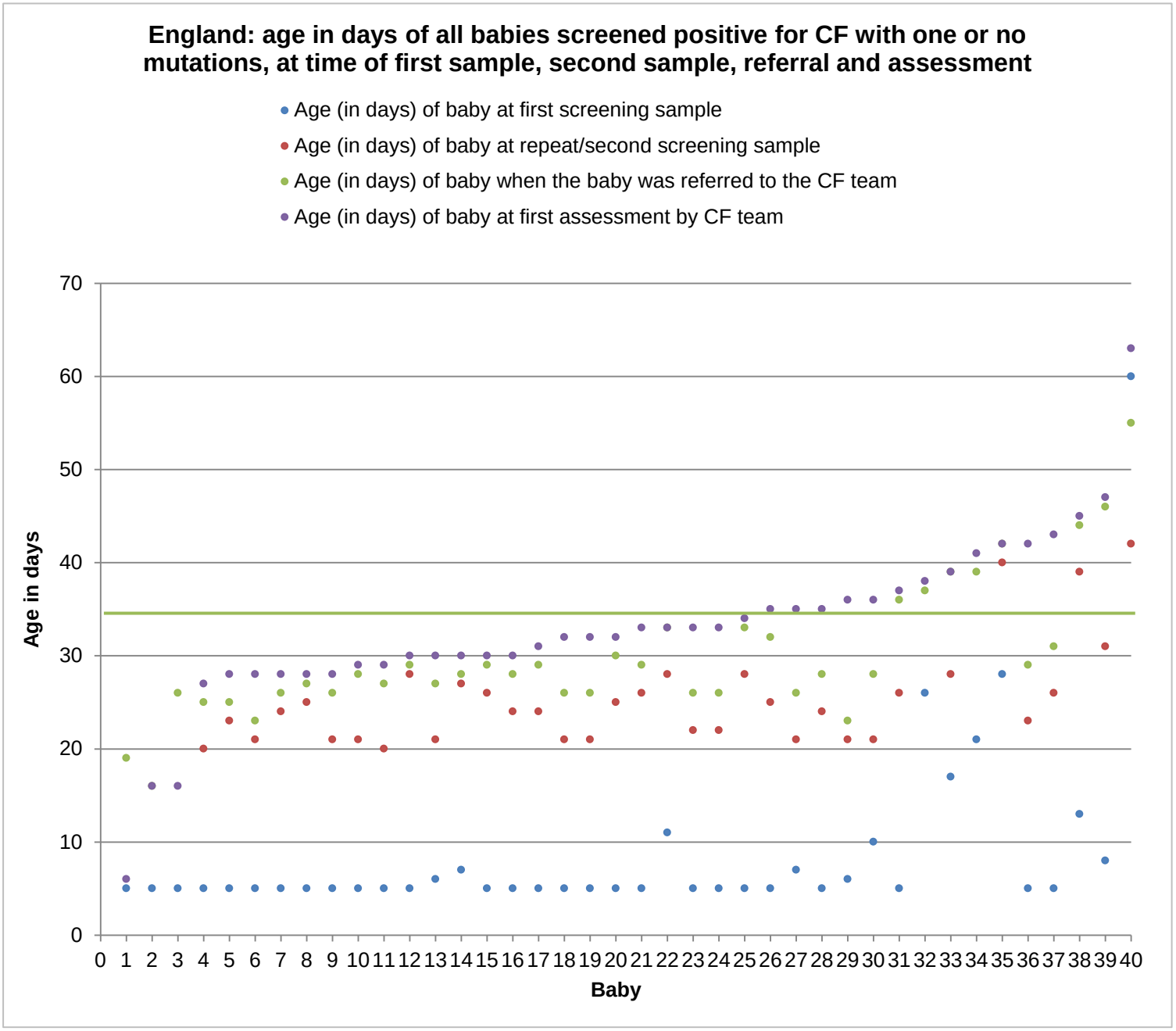
	England	Northern Ireland	Scotland	Wales
Number of CF screen positive babies with one or no mutations	**76**	**5**	**7**	**19**
Number of babies with age at first appointment reported	40	4	4	1
Number seen ≤ 35 days (% of known data)	28 (70%)	4 (100%)	2 (50%)	1 (100%)
All babies mean age at first appointment	33 days	24 days	38 days	5
All babies median age at first appointment	33 days	29 days	40 days	5 days
Age range at first appointment	6-63 days	4-33 days	25-49 days	5 days
Number of babies with age at first appointment not reported	36	1	3	18
Inpatient	5*	0	0	0
Baby died	3	1	0	0
Not reported	31	0	3	18
Outcome				
Confirmed	17	1	1	1
CF SPID	9	1	0	0
Excluded	25	2	5	18**
Baby died	0	1	0	0
Not reported	25	0	1	0

Data source: Newborn screening laboratories

*Of the five inpatients with age at first appointment unknown in England, three died.

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

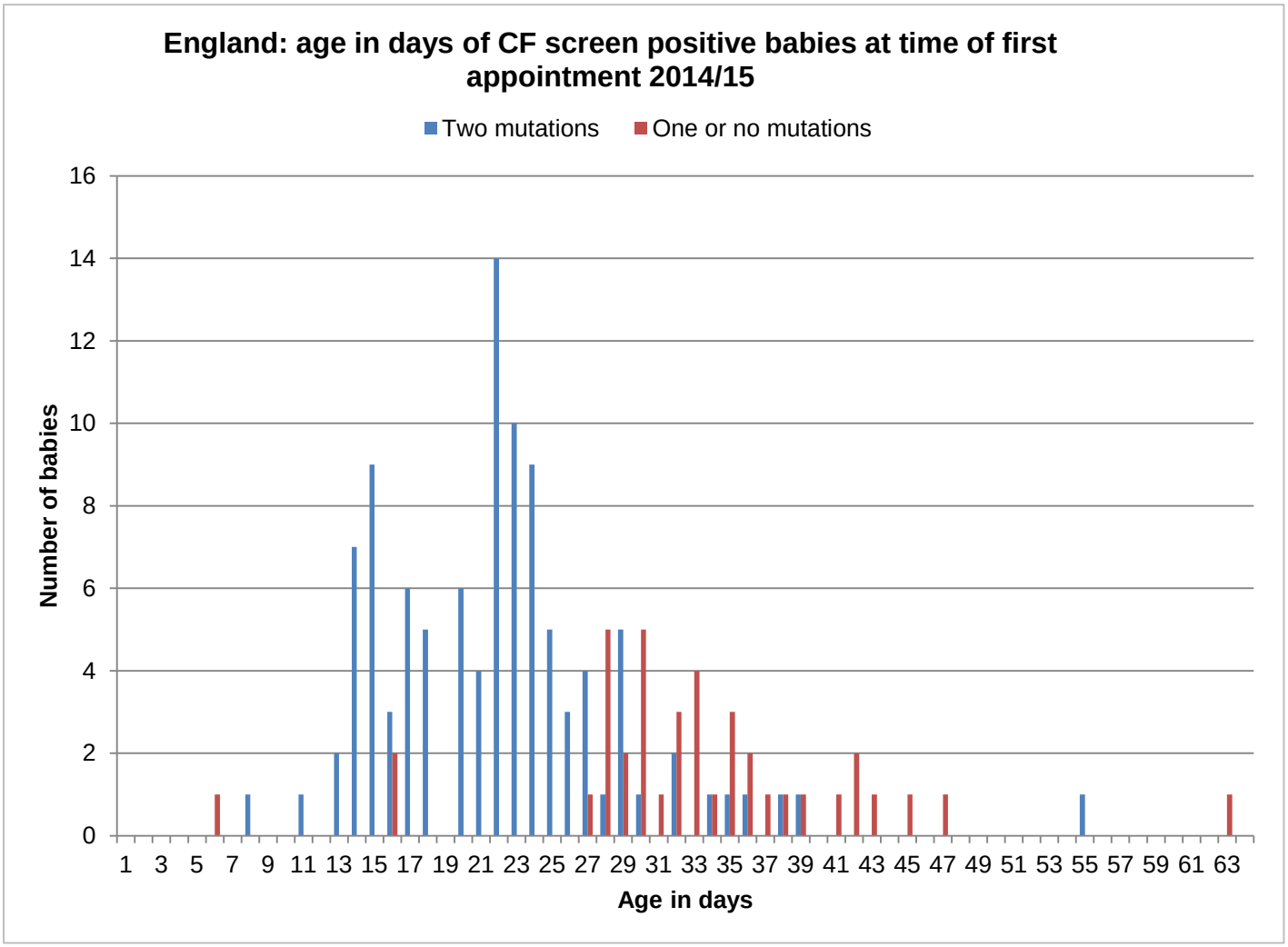
Figure 17: England: CF screen positive babies with one or no mutations 2014/15



Data source: Newborn screening laboratories

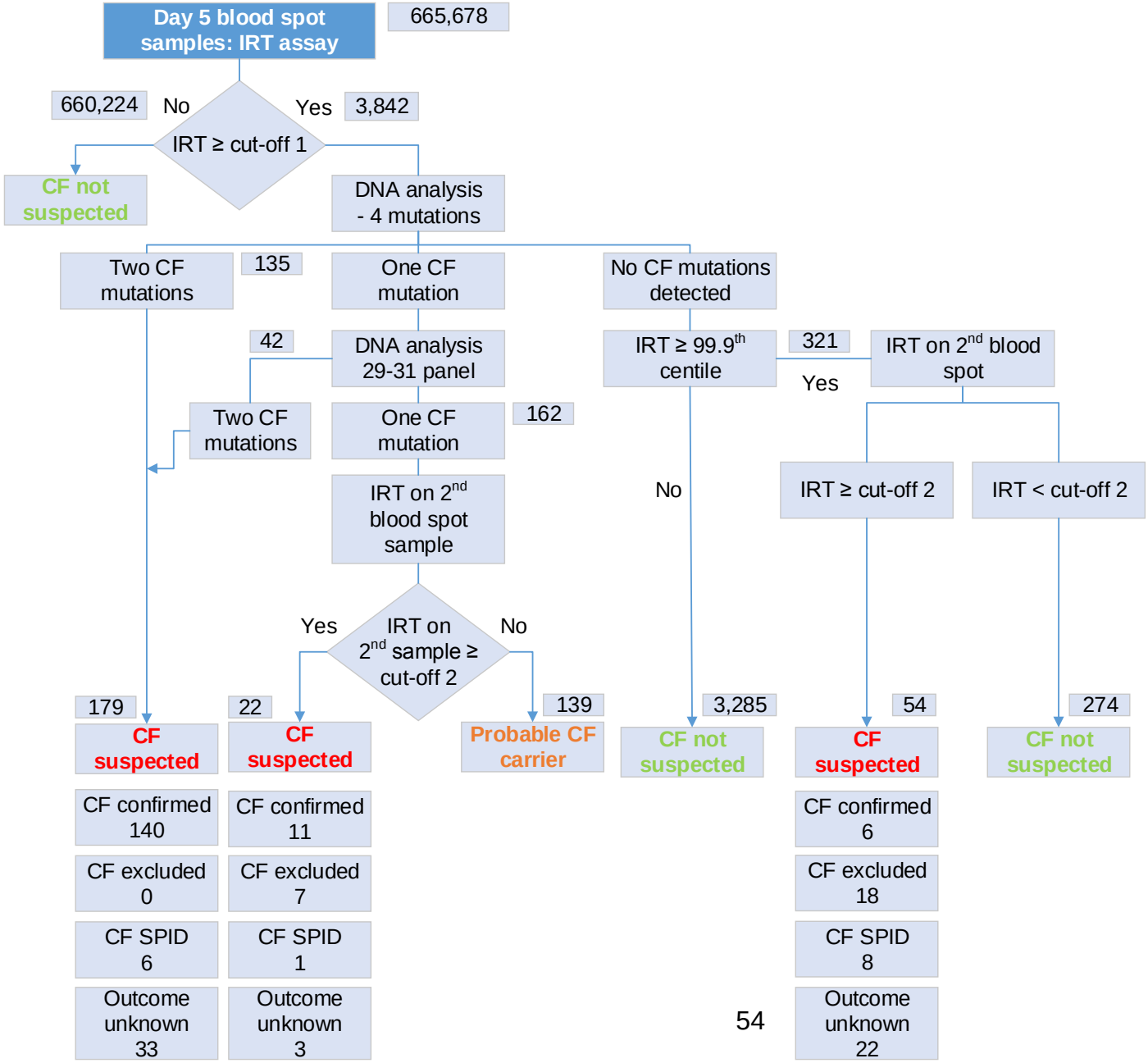
For baby 40, please note that the age in days at first screening sample was given as 60.

Figure 18: England: age in days of CF screen positive babies at time of first appointment 2014/15



Data source: Newborn screening laboratories

Figure 19: England CF screening and diagnostic algorithm 2014/15



Please note that the algorithm is based on data reported by the screening laboratories and that not all discrepancies could be followed up.

Figure 20: Northern Ireland CF screening and diagnostic algorithm 2014/15

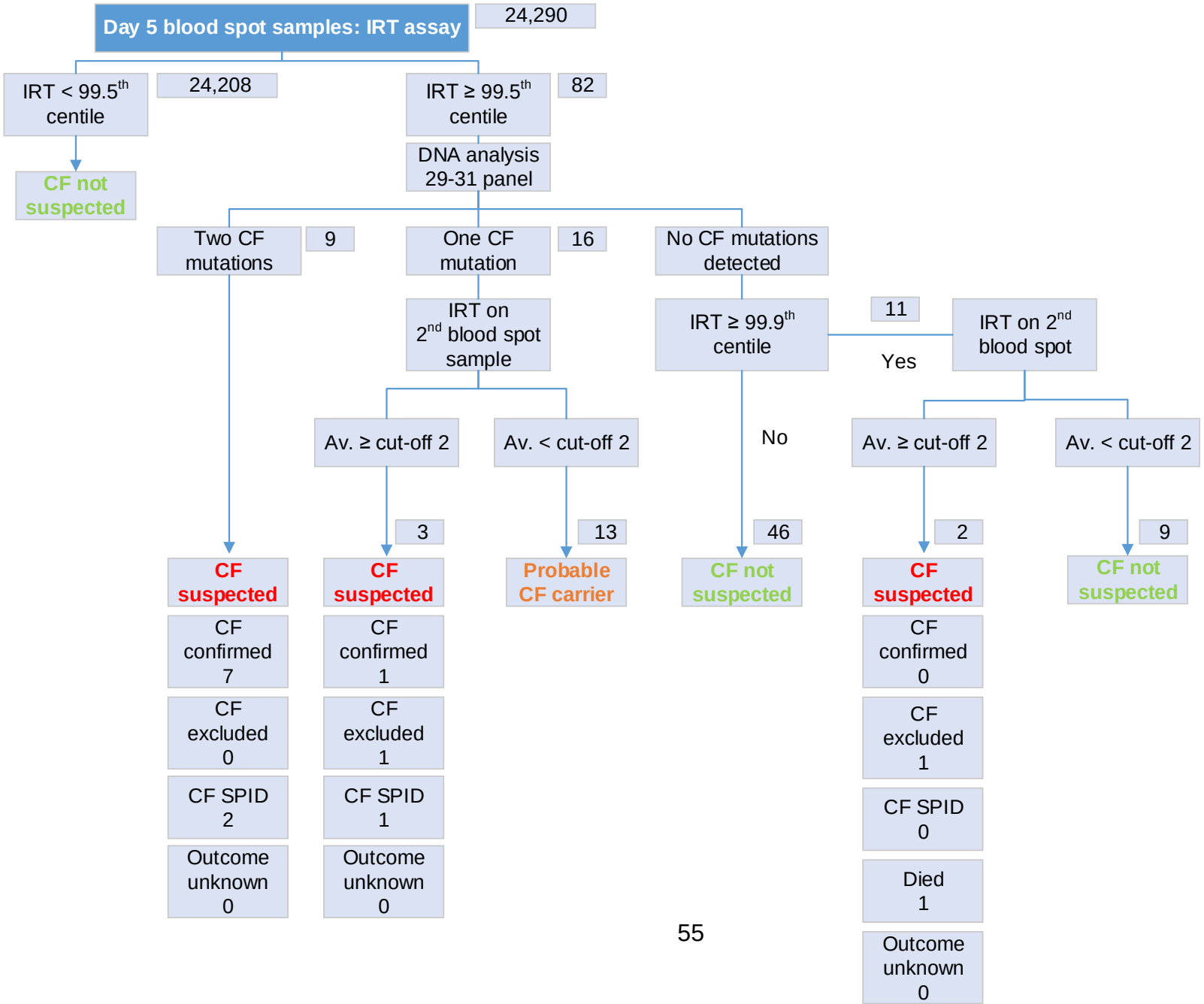


Figure 21: Scotland CF screening and diagnostic algorithm 2014/15

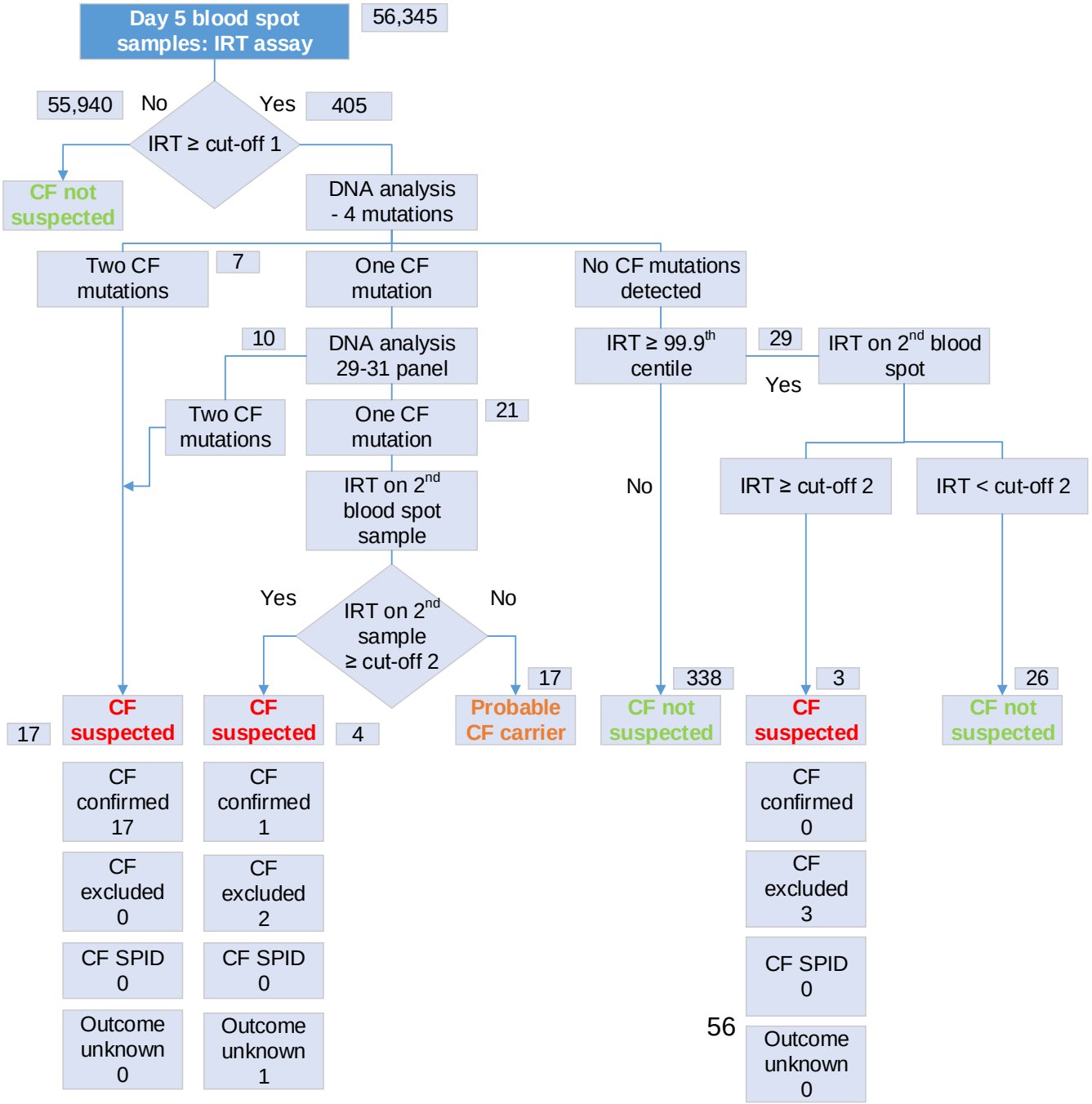
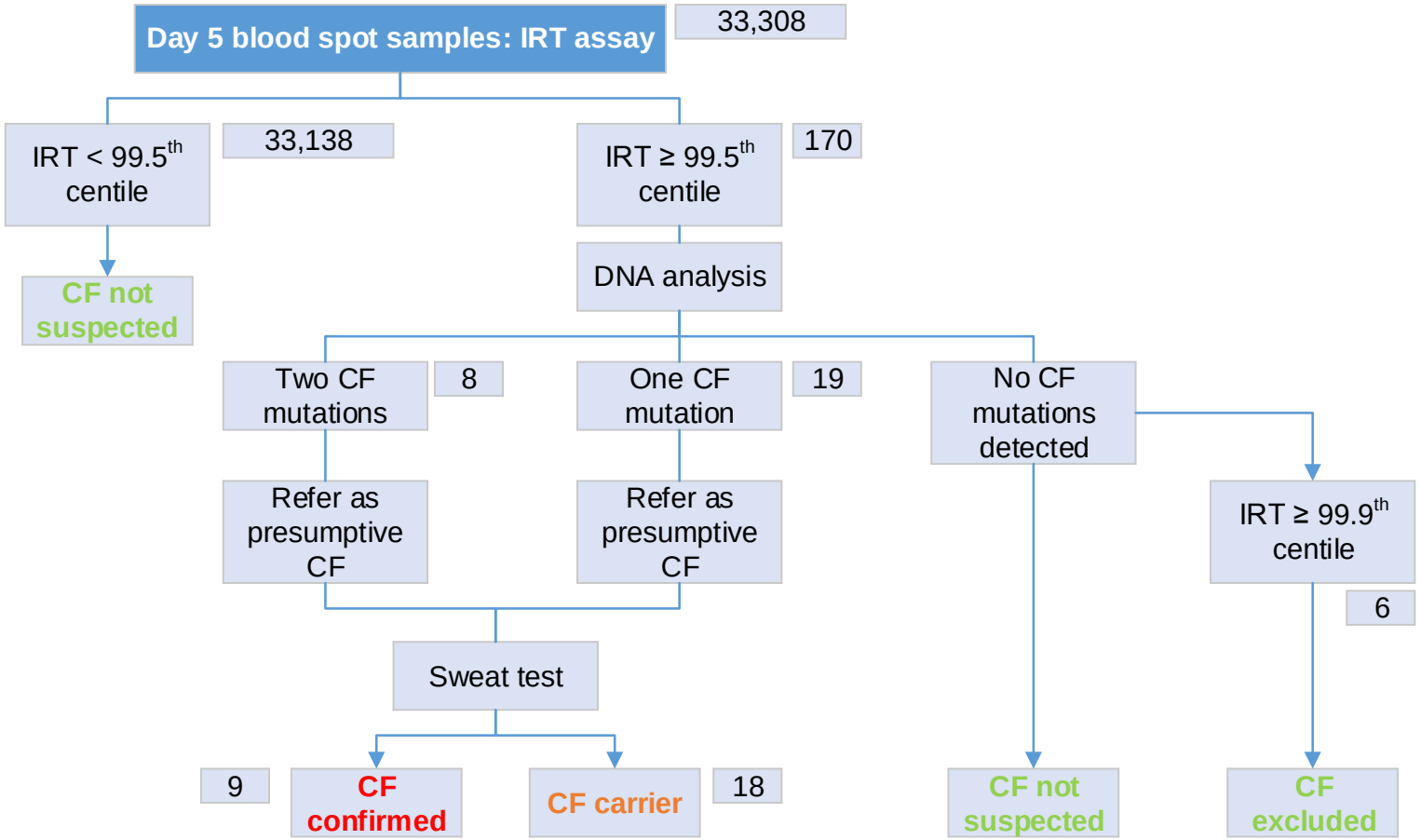


Figure 22: Wales CF screening and diagnostic algorithm 2014/15



CF screen positive data 2007-15

Table 18: CF screen positive data 2007-15

Laboratory	Babies tested for CF	CF screen positives	Rate of CF screen positives
	n	n	Rate per ten thousand
Bristol	331,282	188	5.67
Cambridge	222,369	95	4.27
GOSH	949,969	258	2.72
Leeds	359,478	151	4.20
Liverpool	232,516	132	5.68
Manchester	422,431	155	3.67
Newcastle	275,999	132	4.78
Oxford	238,124	60	2.52
Portsmouth	296,606	108	3.64
SE Thames	408,759	150	3.67
Sheffield	593,697	253	4.26
SW Thames	402,743	126	3.13
West Midlands	573,949	208	3.62
England	5,307,922	2,016	3.80
Northern Ireland	140,819	80	5.68
Scotland	408,796	219	5.36
Wales*	278,682	168	6.03
UK	6,136,219	2,483	4.05

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 19: Timeliness of appointment and treatment outcome for CHT screen positive babies detected on first sample 2014/15

	England	Northern Ireland	Scotland	Wales
Number of CHT screen positive babies detected on first sample	**297**	**12**	**16**	**20**
Number clinically diagnosed before screening (excluded from following age data)	10	0	1	0
Number of babies with age at first appointment reported	265	12	15	20
Number seen ≤ 14 days standard (% of known data)	224 (85%)	11 (92%)	15 (100%)	14 (70%)
Number seen ≤ 17 days standard (% of known data)	256 (97%)	12 (100%)	15 (100%)	18 (90%)
All babies mean age at first appointment	12 days	11 days	10 days	14 days
All babies median age at first appointment	12 days	10.5 days	10 days	13 days
Age range at first appointment	7-25 days	8-15 days	8-13 days	9-27* days
Number of babies with age at first appointment not reported	22	0	0	0
Inpatient	4	-	-	-
Baby died	1	-	-	-
Not reported	17	-	-	-
Has the baby started on thyroxine at the first appointment?				
Yes	216	12	14	16
No	11	-	-	4
Not reported	46	-	-	-
Thyroxine not given but follow up required	12	-	1	-
Thyroxine not given and baby discharged	12	-	1	-

Data source: Newborn screening laboratories

Please note that the data does not include two screen positive babies for whom no clinical data is available.

*Issues contacting family.

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:

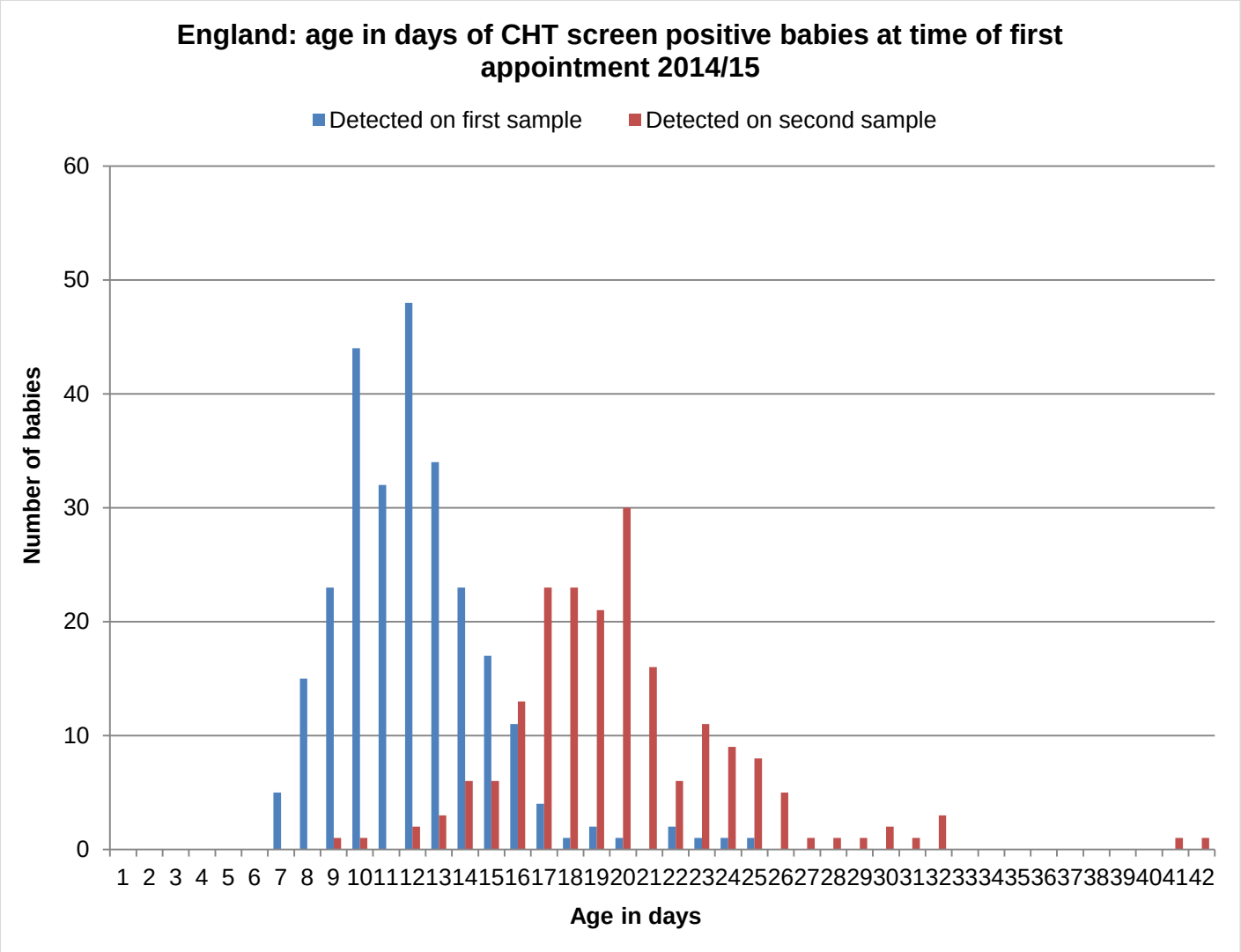
Acceptable level: 100% by 24 days of age
Achievable level: 100% by 21 days of age

Table 20: Timeliness of appointment and treatment outcome for CHT screen positive babies detected on second sample 2014/15

	England	Northern Ireland	Scotland	Wales
Number of CHT screen positive babies detected on second sample	**217**	**6**	**10**	**2**
Number clinically diagnosed before screening (excluded from following age data)	5	0	0	0
Number of babies with age at first appointment reported	195	6	10	2
Number seen ≤ 21 days standard (% of known data)	145 (74%)	5 (83%)	9 (90%)	2 (100%)
Number seen ≤ 24 days standard (% of known data)	171 (88%)	5 (83%)	10 (100%)	2 (100%)
All babies mean age at first appointment	20 days	18 days	17 days	15 days
All babies median age at first appointment	19 days	15 days	18 days	15 days
Age range at first appointment	9-42 days	14-32 days	14-23 days	14-16 days
Number of babies with age at first appointment not reported	17	0	0	0
Inpatient	3	-	-	-
Not reported	14	-	-	-
Has the baby started on thyroxine at the first appointment?				
Yes	113	5	6	2
No	47	0	0	0
Not reported	35	0	0	0
Thyroxine not given but follow up required	8	1	3	0
Thyroxine not given and baby discharged	14	0	1	0

Data source: Newborn screening laboratories

Figure 23: England: age in days of CHT screen positive babies at time of first appointment 2014/15



Data source: Newborn screening laboratories

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

Table 21: Timeliness of sample and appointment for CHT screen positive babies born at less than 32 weeks 2014/15

	England	Northern Ireland	Scotland	Wales
Number of CHT screen positive babies born at less than 32 weeks	**35**	**2**	**0**	**3**
Number clinically diagnosed before screening (excluded from following age data)	1	0	-	0
Age at routine sample				
Babies with age at routine sample reported	33	2		3
Median age at routine sample	7 days	11.5 days		5 days
Age range at routine sample	5-34 days	5-18 days		5-6 days
Age at preterm sample				
Babies with age at preterm sample reported	20	1		0
Median age at preterm sample	28 days	30 days		-
Age range at preterm sample	12-38 days	30 days		-
Age at first appointment				
Babies with age at first appointment reported	32	2		2
Median age at first appointment	33 days	29 days		14 days
Age range at first appointment	9-44 days	27-31 days		13-15 days

Data source: Newborn screening laboratories

Nine babies had a preterm repeat sample taken after 28 days.

Table 22: England: treatment outcome for CHT screen positive babies born at less than 32 weeks 2014/15

	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
35 babies	**10**	**8**	**7**	**5**	**3**	**1**	**1**
Has the baby started on thyroxine at the first appointment?							
Yes		4	4	3	2	1	-
No	9	1	2	1			-
Not reported	1	3	1				-
Thyroxine not given but follow up required							-
Thyroxine not given and baby discharged				1	1		-

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

Table 23: Northern Ireland and Wales: treatment outcome for CHT screen positive babies born at less than 32 weeks 2014/15

	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
Northern Ireland		**1**		**1**			**0**
Wales				**3**			**0**
Has the baby started on thyroxine at the first appointment?							
Yes		1		1			
No				1			
Not reported				1			
Thyroxine not given but follow up required							
Thyroxine not given and baby discharged				1			

Data source: Newborn screening laboratories

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.

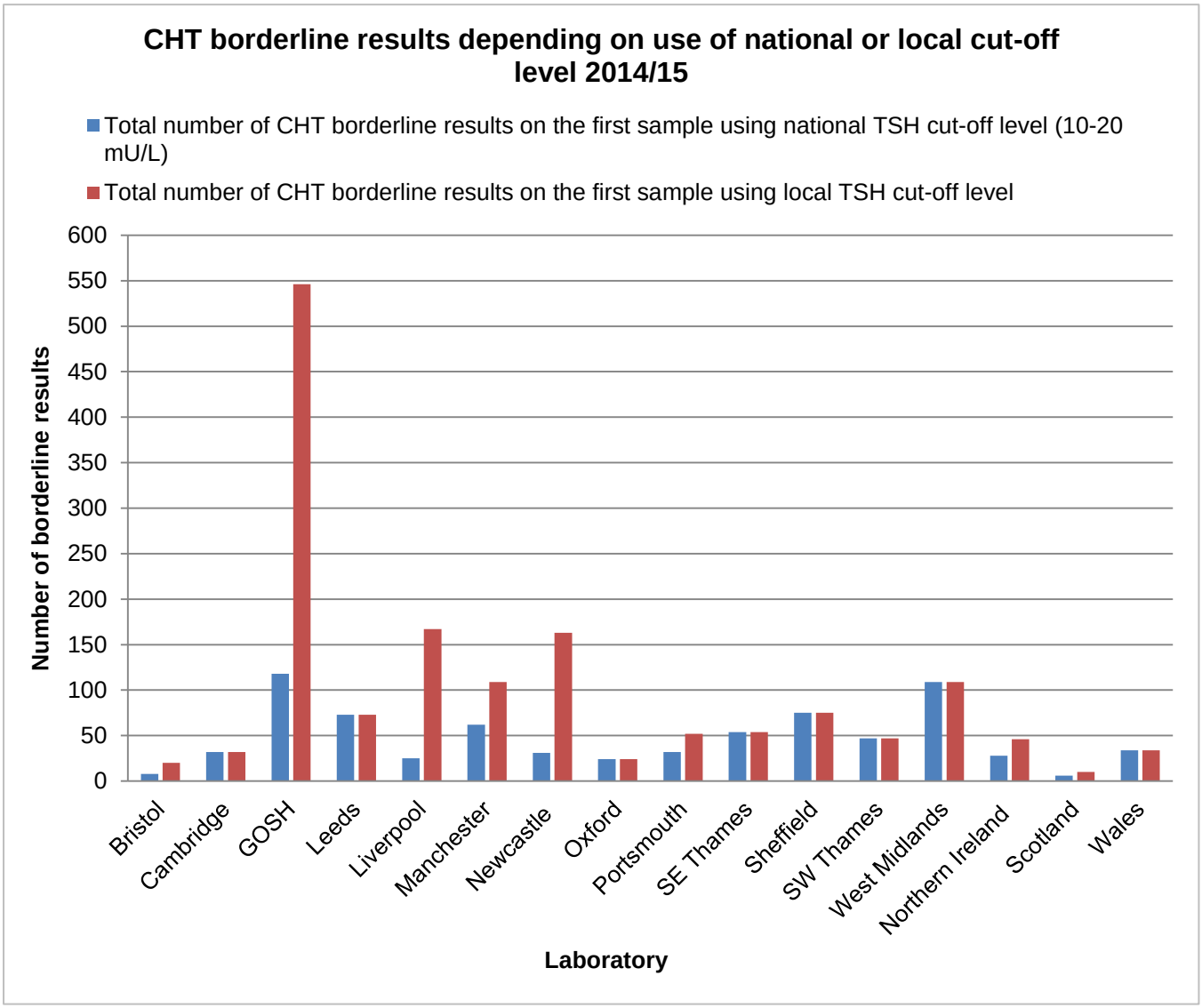
Table 24: CHT borderline results depending on use of national or local cut-off level 2014/15

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	20
Cambridge	18 (GSP)	9 (GSP)	32	32
GOSH	20	6	118	546
Leeds	20	10	73	73
Liverpool	>20	>5	25	167
Manchester	20	8	62	109
Newcastle	20	6	31	163
Oxford	>20	>10	24	24
Portsmouth	20	8	32	52
SE Thames	20	10	54	54
Sheffield	18 (GSP)	9 (GSP)	75	75
SW Thames	20	10	47	47
West Midlands	20	10 to 20	109	109
England			658	1439
Northern Ireland	≥20	≥8	28	46
Scotland	≥25	8-24.9	6	10
Wales	20	10	34	34

Data source: Newborn screening laboratories

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 2014/15



Data source: Newborn screening laboratories

CHT screen positive data 2005-15

Table 25: CHT screen positive data 2005-15

Laboratory	Babies tested for CHT	CHT screen positives	Rate of CHT screen positives
	n	n	Rate per ten thousand
Bristol	406,405	204	5.02
Cambridge	272,909	172	6.30
GOSH	1,220,372	1,180	9.67
Leeds	444,865	277	6.23
Liverpool	290,031	255	8.79
Manchester	507,033	406	8.01
Newcastle	342,754	232	6.77
Oxford	295,118	191	6.47
Portsmouth	354,269	171	4.83
SE Thames	565,858	318	5.62
Sheffield	731,050	384	5.25
SW Thames	511,362	289	5.65
West Midlands	707,840	517	7.30
England	6,649,896	4,596	6.91
Northern Ireland	246,081	164	6.66
Scotland	408,295	190	4.65
Wales	345,355	219	6.34
UK	7,649,627	5,169	6.76

Data source: Newborn screening laboratories

PKU

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

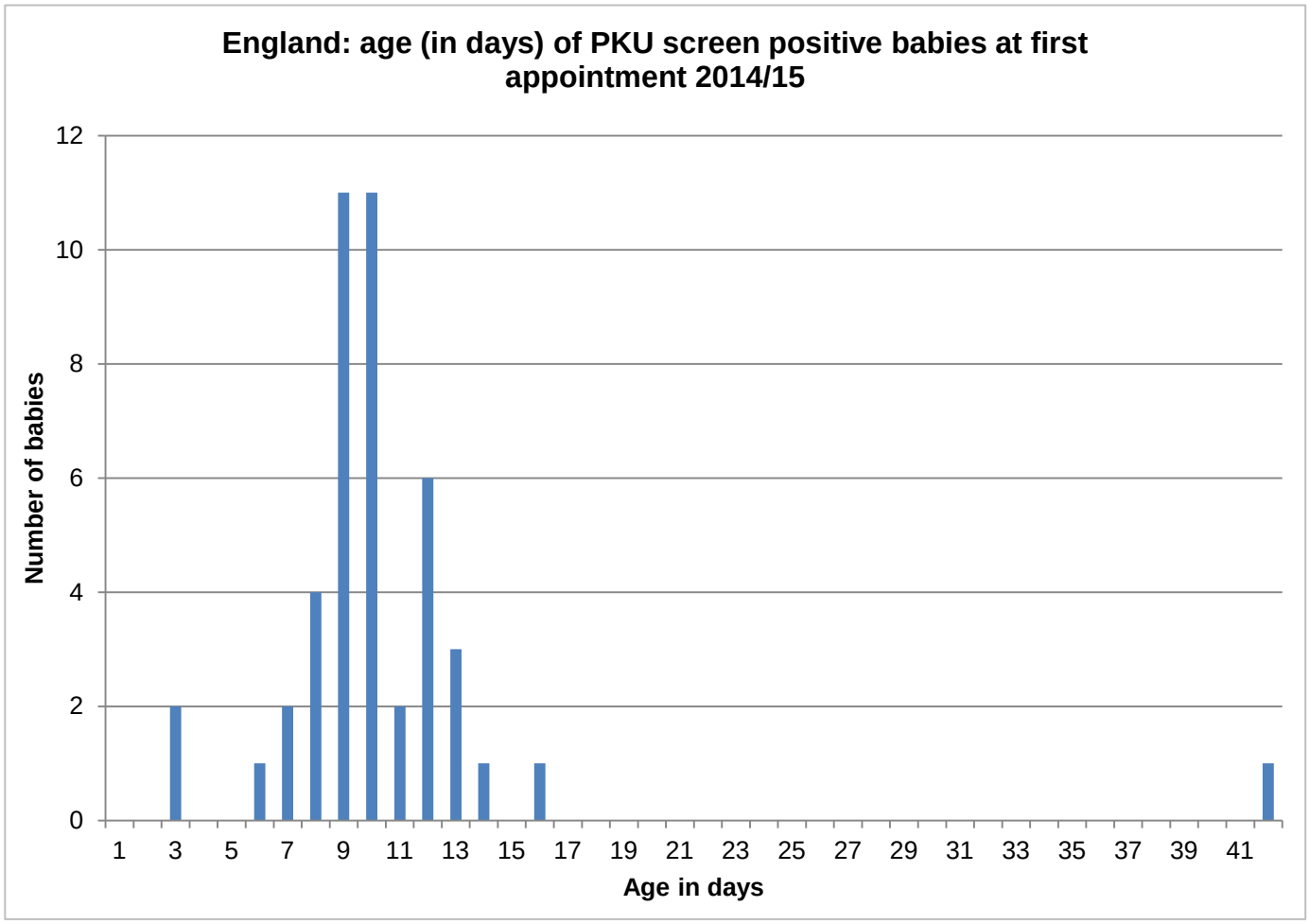
Acceptable level: 100% by 17 days of age
Achievable level: 100% by 14 days of age

Table 26: Timeliness of appointment and outcome for PKU screen positive babies 2014/15

	England	Northern Ireland	Scotland	Wales
Number of PKU screen positive babies	**54**	**5**	**11**	**1**
Number of babies with age at appointment reported	45	5	11	1
Number seen ≤ 14 days (% of known data)	41 (91%)	5 (100%)	11 (100%)	1 (100%)
Number seen ≤ 17 days (% of known data)	42 (93%)	5 (100%)	11 (100%)	1 (100%)
All babies mean age at appointment	10 days	9 days	9 days	11 days
All babies median age at appointment	10 days	9 days	8 days	11 days
Age range at first appointment	3-42 days	7-11 days	4-13 days	11 days
Number of babies with age at appointment not reported	9	0	0	0
Baby died	0	-	-	-
Not reported	9	-	-	-
Outcome				
PKU confirmed, treatment required	36	3	7	1
Non PKU e.g. biopterin disorders	5	1	3	0
No persistent abnormalities - false positive (PKU excluded)	5	0	1	0
PKU monitoring required	8	1	0	0
Not reported	0	0	0	0

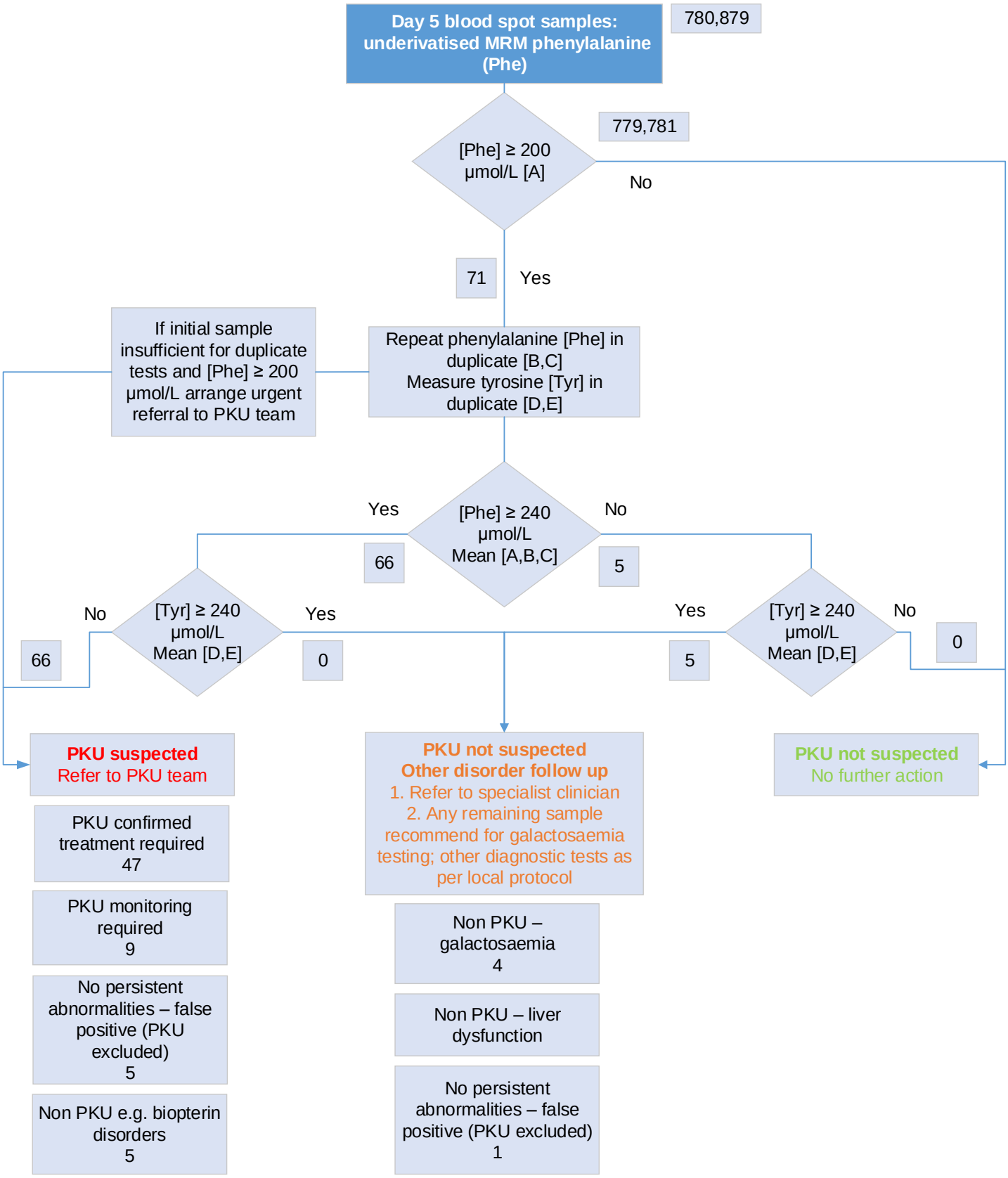
Data source: Newborn screening laboratories

Figure 25: England: age at first appointment for PKU screen positive babies 2014/15



Data source: Newborn screening laboratories

Figure 26: UK PKU screening and diagnostic algorithm 2014/15



Please note that the algorithm is based on data reported by the screening laboratories and that not all discrepancies could be followed up.

PKU screen positive data 2005-15

Table 27: PKU screen positive data 2005-15

Laboratory	Babies tested for PKU	PKU screen positives	Rate of PKU screen positives
	n	n	Rate per ten thousand
Bristol	406,409	30	0.74
Cambridge	272,909	42	1.54
GOSH	1,211,339	113	0.93
Leeds	444,865	55	1.24
Liverpool	290,031	34	1.17
Manchester	507,075	78	1.54
Newcastle	342,754	41	1.20
Oxford	295,121	24	0.81
Portsmouth	354,396	24	0.68
SE Thames	565,856	57	1.01
Sheffield	731,052	99	1.35
SW Thames	511,362	38	0.74
West Midlands	707,840	85	1.20
England	6,641,036	720	1.08
Northern Ireland	246,092	57	2.32
Scotland	408,340	63	1.54
Wales	345,413	48	1.39
UK	7,640,881	888	1.16

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

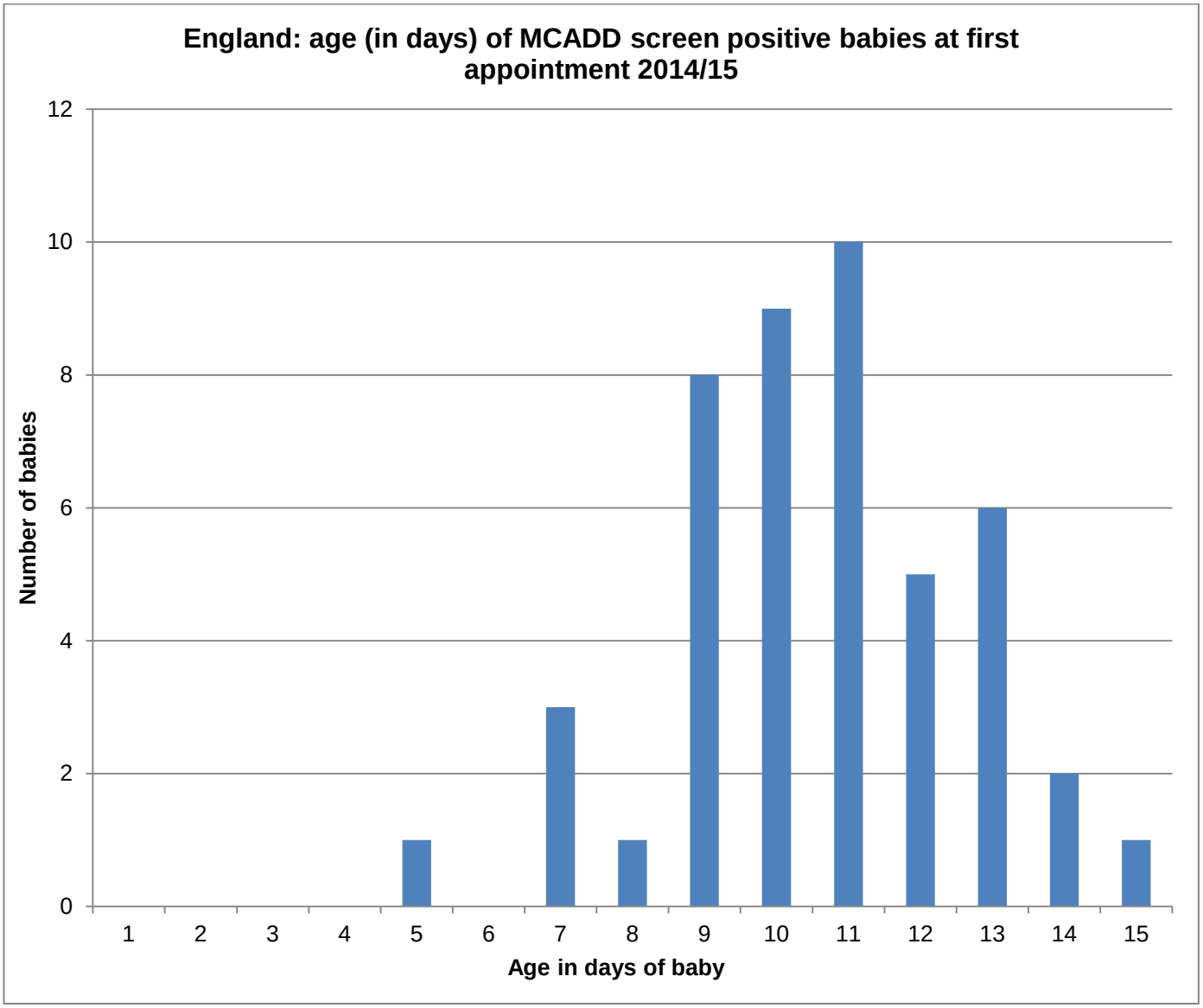
Table 28: Timeliness of appointment and outcome for MCADD screen positive babies 2014/15

	England	Northern Ireland	Scotland	Wales
Number of MCADD screen positive babies	**53**	**3**	**0**	**2**
Number of babies with age at appointment reported	48	3	0	2
Number seen ≤ 14 days (% of known data)	46 (95%)	3 (100%)	0	1 (50%)
Number seen ≤ 17 days (% of known data)	47 (96%)	3 (100%)	0	2 (100%)
All babies mean age at appointment	13 days	7 days	0	13 days
All babies median age at appointment	11 days	8 days	0	13 days
Age range at first appointment	5-142* days	2-11 days	0	8-17 days
Number of babies with age at appointment not reported	5	0	-	0
Family history (early testing)	2	-	-	-
Not reported	3	-	-	-
Outcome				
MCADD	47	3	-	0
Unaffected carrier	1	0	-	0
MCADD unlikely	0	0	-	0
No persistent abnormality, false positive	5	0	-	2
Not reported	0	0	-	0

Data source: Newborn screening laboratories

*Day 142 baby was mover in.

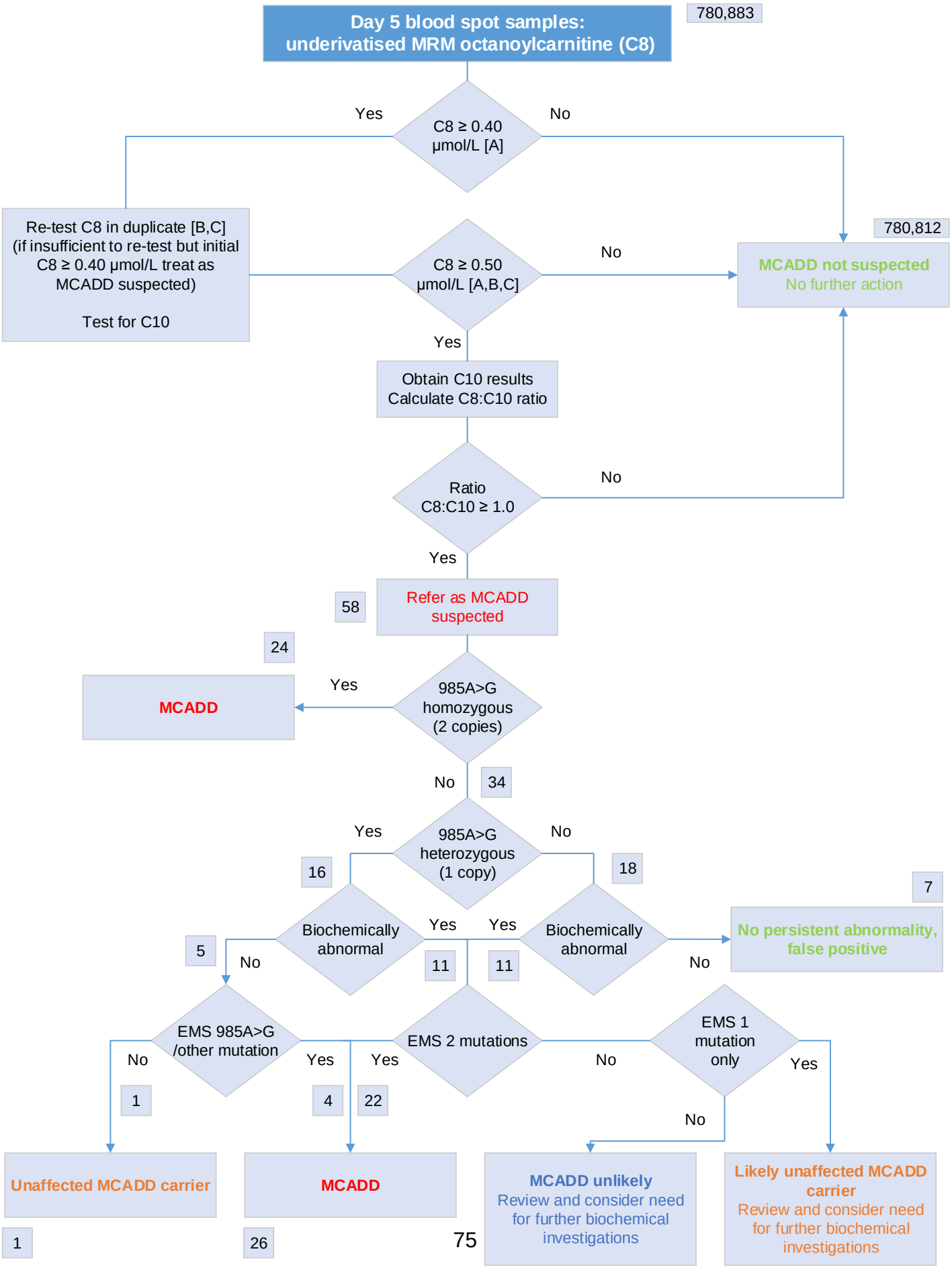
Figure 27: England: age at first appointment for MCADD screen positive babies 2014/15



Data source: Newborn screening laboratories

One further baby has their first appointment at day 142.

Figure 28: UK MCADD screening and diagnostic algorithm 2014/15



MCADD screen positive data 2008-15

Table 29: MCADD screen positive data 2008-15

Laboratory	Babies tested for MCADD	MCADD screen positives	Rate of MCADD screen positives
	n	n	Rate per ten thousand
Bristol	270,157	23	0.85
Cambridge	191,377	26	1.36
GOSH	861,819	62	0.72
Leeds	315,890	44	1.39
Liverpool	196,435	21	1.07
Manchester	396,606	44	1.11
Newcastle	221,581	24	1.08
Oxford	183,417	20	1.09
Portsmouth	256,351	27	1.05
SE Thames	404,448	30	0.74
Sheffield	520,619	76	1.46
SW Thames	342,920	27	0.79
West Midlands	503,332	39	0.77
England	4,664,952	463	0.99
Northern Ireland	141,347	18	1.27
Scotland	262,938	10	0.38
Wales	95,873	8	0.83
UK	5,165,110	499	0.97

Data source: Newborn screening laboratories

MSUD, IVA, GA1 and HCU

Table 30: England and Wales: Timeliness of appointment and outcome for MSUD, IVA, GA1 and HCU screen positive babies 2014/15

	MSUD	IVA	GA1	HCU
Number of screen positive babies	**3**	**7**	**4**	**5**
Number of babies with age at first appointment reported	3	6	4	3
Number seen ≤ 14 days	2	6	3	2
Number seen ≤ 17 days	2	6	3	3
All babies median age at first appointment	5 days	5 days	6.5 days	5 days
Age range at first appointment	8-42 days	7-12 days	11-42 days	14-16 days
Number of babies with age at first appointment not reported	0	1	0	2
Inpatient	-	0	-	1
Not reported	-	1	-	1
Outcome				
Confirmed	2	1	3	3
False positive	1	2	1	2
Mild (IVA only)	0	2	0	0
Other	0	1 (MADD)	0	0
Not reported	0	1	0	0

Data source: Newborn screening laboratories

Screening for MSUD, IVA, GA1 and HCU was implemented fully in England and Wales in January 2015. There were no screen positives for Wales January – March 2015.

The six laboratories that participated in the expanded screening pilot were asked to provide full data for MSUD, IVA, GA1 and HCU for 2014/15; non-pilot laboratories were asked to provide data for January – March 2015 only. Data includes two screen positive results for one baby.

Note that data here is presented on all screen positive babies reported (including two babies with no denominator data).

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 six 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 five conditions available for communication by six weeks of age. The definition of this standard will be reviewed.

Table 31: Timeliness of results to parents 2014/15

	Babies screen negative for all five conditions	Results available for communication by six weeks of age	
Region/country	n	n	%
East Midlands	39,387	39,301	99.8
East of England	53,745	53,321	99.2
London	76,107	75,729	99.5
North East	16,831	16,790	99.8
North West	50,741	50,607	99.7
South East	72,632	72,384	99.7
South West	47,572	47,192	99.2
West Midlands	25,717	25,633	99.7
Yorkshire and Humber	52,010	51,550	99.1
England	434,742	432,507	99.5
Northern Ireland	-	-	-

Data source: CHRDs

37 returns were excluded based on missing data.

Note that standard 1a indicates that 94.6% of results in England are recorded on the CHIS by 17 days of age (CCG responsibility at birth).

Conclusion

Data was returned by CHRDs for 183 CCGs (87%) out of the 211 that existed in England in 2014/15. Exclusions were made if the data was incomplete. In some cases data was reported for a particular CCG by more than one CHRD; this added to the complexity of analysing the data. Changes in networks and reorganisations have meant that the NBS programme does not have an up-to-date contact database for CHRDs – this makes it difficult to request data and follow up any queries.

The CHRD process 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.

Maternity sites now use the NBSFS to ensure all babies born in England are offered screening. Work continues to receive all screening results into the national database. The responsibility for ensuring completeness of coverage remains with the CHRD.

Over the last three 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. In England, processes for recording declines for movers in vary between regions – a programme objective is to ensure that there is consistent and accurate reporting/recording of declines and that parents are supported to make an informed decision about screening.

It is difficult to draw conclusions from the year-on-year data on timeliness of sample receipt, but sample transport remains one of the biggest risks for delayed identification of screen positive babies. There is anecdotal evidence that sample transport times are continuing to increase in some areas – the programme aims to identify best practice and advise lessons learnt.

Laboratories have accepted blood spot cards of varying quality. New consensus guidelines 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. Avoidable repeat rates will be more comparable from 2015/16.

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 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 achievable standard for timely processing of screen positive samples was met for PKU; the acceptable standard was met for MCADD. The acceptable standard was not met for CHT.

Based on data reported, the acceptable standard for timeliness of first appointment for CF screen positive babies with two mutations was not met in England, Northern Ireland or Scotland. The acceptable standard for babies with one or no mutations was not met in England or Scotland.

CF outcome data is challenging for the laboratories to collect. To address this, the NBS programme has supported a Specialist Interest Group to bring together CF clinicians from each referral centre in the UK to facilitate data capture and programme evaluation. Collaborative working has enabled missing outcome data from previous years to be reported – any outstanding data is now considered lost to follow up. Further communication is in progress to complete the 2014/15 data for England as approximately one sixth of CF outcome data remains missing for babies with two mutations (33 out of 179), and one third of data remains missing for babies with one or no mutations (25 out of 76).

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 or Wales. The acceptable standard for babies detected on second sample was not met in England or Northern Ireland.

In England, approximately one sixth of data on CHT treatment at first appointment remains missing for babies detected on first sample (46 out of 297) and second sample (35 out of 217). 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 which is due to report its findings in mid-2016. The NBS programme is also streamlining the data collection template to make the reporting process more robust.

With the implementation of expanded screening in January 2015 in England and Wales, only partial data is presented for the four new conditions. The acceptable standard for timeliness of appointment for PKU and MCADD screen positive babies was not met in England. However, with minimal chasing outcome data was reported for all babies.

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