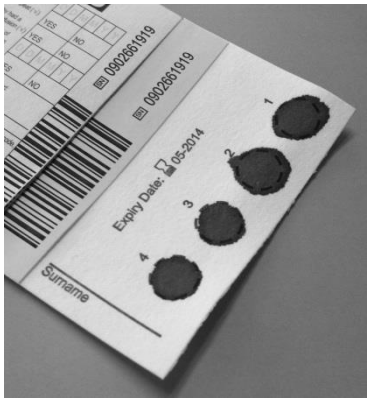
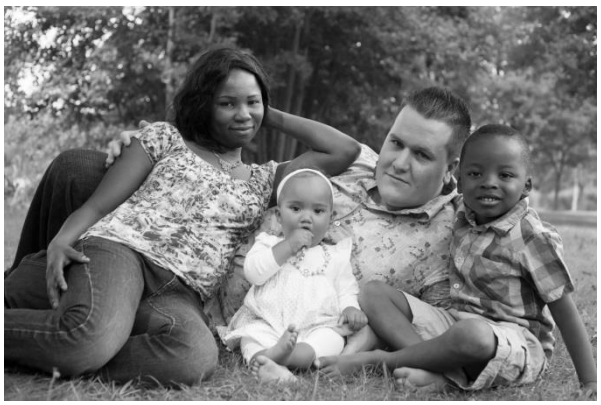
Public Health England

NHS

NHS Sickle Cell and Thalassaemia Screening Programme

Data report 2015/16: trends and performance analysis





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. We do this through world-class science, knowledge and intelligence, advocacy, partnerships and the delivery of specialist public health services. We are an executive agency of the Department of Health, and are a distinct delivery organisation with operational autonomy to advise and support government, local authorities and the NHS in a professionally independent manner.

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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

In 2015/16 approximately 706,000 women received antenatal screening for sickle cell disease and thalassaemia conditions in England. Of these, just under 14,000 women were reported to be screen positive (1 in 51 samples screened), and 772 high risk couples (1 in 18 screen positive women) were identified based on the results from both the mother and the baby's biological father. National antenatal coverage for sickle cell and thalassaemia (SCT) screening was 99.1% in 2015/16, up from 98.9% in 2014/15. The majority of trusts in England were above the 95% acceptable threshold for key performance indicator (KPI) ST1.

Completion of the family origin questionnaire (FOQ) continues to improve and is now at 97% in 2015/16, up from 96% in 2014/15. The majority of trusts reported FOQ completion to be above the 95% acceptable level. Rates of declined antenatal screening have continued to fall each year and are now at 2 per 1,000 booking bloods received. Father uptake remains at approximately 60% nationally, but the rates appear to be falling in low prevalence areas.

Early testing in antenatal screening is important as there may be a series of tests required in order to offer women and couples informed choices. Programme standards state that pregnant women should receive screening by 10 weeks gestation, and that testing (including a prenatal diagnostic test if chosen) should be completed by 12 weeks and six days gestation. Data from KPI ST2 shows that just over half of pregnant women are being screened by 10 weeks gestation. Rates appear steady over the last two years in most regions, but there appears to be a decline in London where prevalence is highest.

'At risk' couples should be offered prenatal diagnostic (PND) testing to identify whether the fetus is affected by sickle cell disease or thalassaemia conditions. In the last two years 40% of PND tests were performed by the 12 weeks and six days standard, representing a decline of approximately 10% on previous years. There has been a corresponding increase of 10% in the proportion tested in the 13th and 14th weeks, and the proportion tested in the 15th week or later remains at approximately 30%.

This year marks the 10th anniversary of full roll-out of newborn screening in July 2006. We have included some figures in this report to show the numbers screened and detected in the 11 years since data collection began in 2005/06. In this period 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 781,166 for the whole of the UK. 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 was 51 days.

1. Introduction

1.1. About the NHS Sickle Cell and Thalassaemia Screening Programme

The NHS Sickle Cell and Thalassaemia Screening Programme was set up in England in 2001 following a government commitment in the NHS Plan in 2000 and is the world's first linked antenatal and newborn screening programme. Our mission statement is to develop a linked programme of high quality screening and care in order to:

- ensure a high quality, accessible screening programme throughout England
- support people to make informed choices during pregnancy and ensure timely transition into appropriate follow up and treatment
- improve infant health through prompt identification of affected babies and timely transition into clinical care
- promote greater understanding and awareness of the conditions and the value of screening

1.2. Methodology

Timely annual data returns are required from all screening laboratories in accordance with laboratory guidance[1] and 'Service Specification no.18: NHS Sickle Cell and Thalassaemia Screening Programme'[2]. Data is collated and submitted via spreadsheet-based data templates. On receipt the data is checked and, if required, the data is clarified with the relevant laboratory.

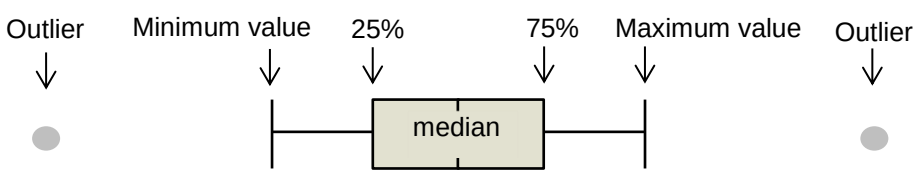
For the antenatal laboratories in particular, we recognise the difficulty of data collection in the absence of standardised data collection tools and IT systems. We try to ask for limited data and work hard to justify all data requests, ensuring there are no gaps and no duplication across the screening pathway and between screening programmes. Data on tests by 10 weeks (standard AP1) and data on FOQ completion (standard AO1aiii) are collected as key performance indicators (KPIs) through a separate process. More information on the KPI data collection can be found on gov.uk.

Trusts are excluded in the annual KPI data where data has not been submitted for all four quarters in that year. Exclusions may, however, differ between years where multiple years are shown. PND data is requested several months after the requests for antenatal and newborn data to include pregnancy outcomes. It is planned that in future years the PND data will be collected via the National Congenital Anomalies and Rare Disorders Registration Service (NCARDRS), which should improve completion of outcomes data for positive results.

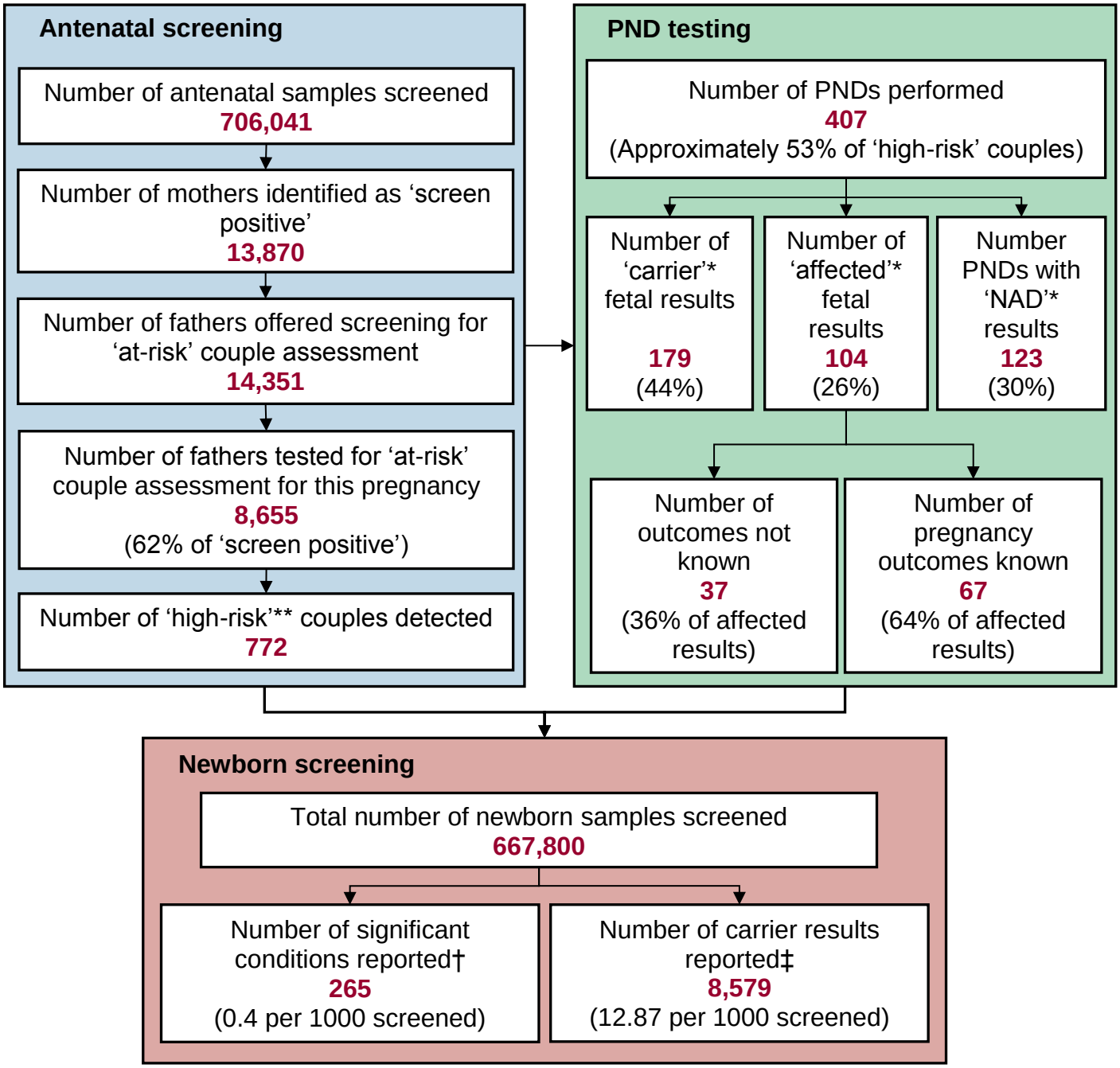
The newborn data reported for England excludes cases from outside of England. Prevalence data by region and by ethnicity is compared and laboratories are contacted for clarification if inconsistencies are found. While the screening programme only has a remit for England, this year we have included data from the newborn laboratories in Scotland, Wales, and Northern Ireland. These are, however, excluded from the ethnicity data as Scotland uses different categories for ethnicity, and the Wales and Northern Ireland laboratories do not routinely collect data on ethnicity.

Current versions of the antenatal and newborn data returns are available on gov.uk. Data is presented by financial year (1 April to 31 March) unless stated otherwise. The year '2014/15', for example, refers to the financial year '1 April 2014 to 31 March 2015'.

'Box and whisker' plots are used to show variation in performance between trusts. This type of chart shows the highest and lowest values, as well as the median value and the interquartile range (containing half of the values):



2. Overview of national screening figures



Note: These figures represent total numbers reported and numbers may differ from those elsewhere where exclusions have been made based on missing or unavailable data.

*Excludes cases where the result was not included in the data return.

†'Significant conditions' in newborn screening comprises FS, FSC, FS Other and FE.

‡'Carrier results' in newborn screening comprises FAS, FAC, FAD, FAE and other carriers.

** 'High risk' comprises cases where both parents are carriers or affected and there is a high risk that the baby will be affected by a significant condition. This number excludes low-risk cases and cases where the father was not available for testing.

3. Antenatal screening and prenatal diagnostic testing data

3.1. Response rates and data quality

Response rate

The screening programme received 134 of the expected 139 antenatal data returns (96%). Data was received from all three prenatal diagnostic (PND) laboratories.

Data quality

Antenatal screening data

The data presented in this report represents the data provided by the antenatal laboratories unless specified otherwise. We are aware that figures may differ from those from other data sources.

A number of laboratories were unable to provide data for some of the data fields requested. Where this was the case, exclusions have been made so as to not bias the reported rates (for example if the numerator is provided and the denominator is not, national rates would appear higher than they actually are, or if the denominator is provided and the numerator is not, rates would appear lower than they actually are). Where exclusions have been made, these are identified below the associated charts and tables.

Figures on the number of tests by 10 weeks can be dependent on complete information on the FOQ to obtain gestational information. This means that the figures reported on timeliness of testing may offer a base rate, but actual numbers tested by 10 weeks may be higher.

Some laboratories are unable to match mother results to father results and so cannot provide the number of high-risk couples. As a result, the actual number of high-risk couples is likely to be higher than reported.

Some laboratories are unable to distinguish between antenatal and non-antenatal specimens and so use figures provided by maternity units to determine the number of booking bloods received. This may distort the figures slightly as maternity units may refer samples to more than one laboratory and so the number of booking bloods received may appear higher than it actually is.

Some laboratories cover more than one hospital. In these cases, we ask for separate data returns for each hospital covered.

We are aware that the number of screen positive and screen negative women, plus pending results, does not add up to the number of booking bloods received. This is due to the way that data is provided by some laboratories. The difference may be accounted for, to a degree, by the inclusion of other haemoglobinopathy variants which are outside the scope of the SCT screening programme.

Prenatal diagnostic (PND) testing data

Non-identifiable patient-level data is provided by the PND laboratories, but there are some gaps in the data. Approximately 0.5% of PND tests performed did not have information on gestational age, and approximately 36% of affected results did not have any information on pregnancy outcomes (whether the couple continued the pregnancy, miscarried, or opted for termination).

The Manchester laboratory started carrying out PND tests in 2013/14, but was not included in data for previous years. Data from the Manchester laboratory has now been added to the historical data to improve data completeness, which means that figures reported this year will differ from those previously reported.

3.2. Antenatal coverage

Antenatal coverage data is collected as key performance indicator (KPI) ST1, and is collected on a quarterly basis. Performance against this KPI is calculated as the number of tested women as a proportion of the eligible population. More information on KPI definitions can be found on gov.uk. Annual data is derived from the quarterly data, but exclusions are made for any trust that did not provide data in one or more quarter in that year.

Table AN-1 shows a summary of the annual data for ST1 for the last two years. Nearly all regions have either seen a small improvement or have remained the same compared to last year. The thresholds for this KPI are set at 95% as an acceptable level and 99% for an achievable level. Of the 10 sub-regions shown, all are above the acceptable level and six are above the achievable level.

Table AN-1. Coverage of antenatal screening by sub-region 2014/15 to 2015/16

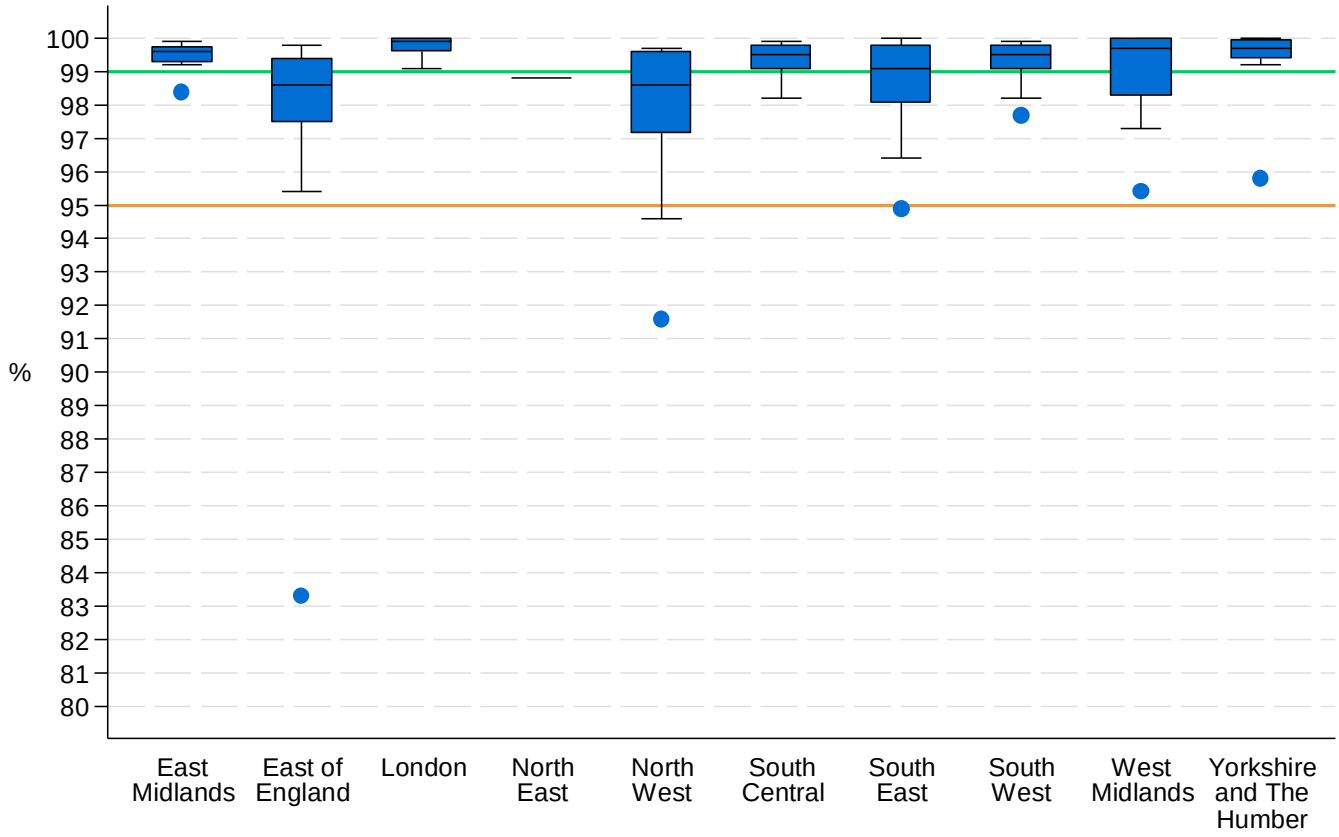
Sub-region	Completeness (2015/16)	2014/15			2015/16		
	Submitted all 4 quarters / no. of trusts	Eligible women	Tested women	%	Eligible women	Tested women	%
East Midlands	8 / 9	34,522	33,923	98.3	49,486	49,131	99.3
East of England	17 / 18	71,398	70,075	98.1	70,539	69,305	98.3
London	24 / 24	139,264	138,923	99.8	153,180	152,856	99.8
North East	1 / 8	6,735	6,574	97.6	5,621	5,553	98.8
North West	16 / 22	75,034	73,641	98.1	78,325	77,015	98.3
South Central	7 / 7	41,807	41,602	99.5	40,788	40,493	99.3
South East	11 / 12	64,240	63,181	98.4	59,664	58,861	98.7
South West	17 / 17	62,298	61,929	99.4	63,675	63,283	99.4
West Midlands	9 / 14	49,045	48,094	98.1	52,621	52,213	99.2
Yorkshire and the Humber	8 / 13	40,777	40,483	99.3	42,466	42,247	99.5
England total	118 / 144	585,120	578,425	98.9	616,365	610,957	99.1

*Exclusions where data was not returned in all four quarters: 2014/15: 29; 2015/16: 26.

Figure AN-1 shows the variation between the highest and lowest performers in each region and the interquartile range (containing half the values) for each region. Half of

the sub-regions have an interquartile range above the 99% achievable level. Of the five regions where the interquartile range falls below the achievable, only one region has trusts that report coverage below the acceptable level (excluding outliers). It appears that the best performing sub-regions also have the least variation in performance between trusts. It should be noted that the figures for the North East are based on a single trust that was able to report matched-cohort data for all four quarters in 2015/16.

Figure AN-1. Antenatal coverage 2015/16: England by sub-region



Excludes 26 trusts that did not provide data for all four quarters.

3.3. Numbers screened and detected in antenatal screening

National

Table AN-2 shows the number of booking bloods received by laboratories in each region, the number of screen positive women detected, and the number of high risk couples identified. 'High risk' couples comprise pregnancies where both biological parents are identified as either carriers or as affected and there is a high risk that the baby will be affected by a significant condition.

This figure excludes cases where the baby's biological father was not available for testing, or where the father's result cannot be matched to the mother's result by the laboratory in order to determine the risk to the pregnancy. This means that the actual

number of high risk couples is expected to be higher. The numbers where the father was not available for testing or could not be matched to mother results are included in section 3.6 ‘Testing of the baby’s biological father’.

In 2015/16 there were 706,041 booking bloods reported by laboratories, of which 13,870 were identified as screen positive (approximately 1 in 51 women screened). Of these 772 pregnancies were identified as high risk (approximately 1 in 18 screen positive women).

We would expect the number of high risk couples in antenatal screening to be approximately four times the number of newborn screen positive cases identified with significant conditions (FS, FSC, FS-Other, and FE results), plus four times the number of babies with an F-only newborn result (which are potential beta thalassaemia affected results), plus terminations of affected pregnancies following PND testing. This gives an estimate of approximately 1,200 high risk pregnancies.

Table AN-2. Numbers screened and detected by sub-region 2015/16: England

Sub-region	No. of Labs	Booking bloods received (BBs)	Screen 'positive' women		Screen 'negative' women		Result pending/ inconclusive result		High risk couples identified	
	Submitted/ Total Labs	n	n	% of BBs	n	% of BBs	n	% of BBs	n	% of screen positive
East of England	16 / 16	78,869	1,106	1.40	67,480	85.56	48	0.06	73	6.60
East Midlands	9 / 9	56,299	829	1.47	55,158	97.97	0	0.00	45	5.43
London	21 / 24	136,876	6,228	4.55	130,726	95.51	44	0.03	309	4.96
North East	8 / 8	31,512	255	0.81	27,307	86.66	12	0.04	11	4.31
North West	18 / 19	83,820	1,115	1.33	81,126	96.79	60	0.07	60	5.38
South Central	8 / 9	48,162	658	1.37	46,683	96.93	31	0.06	34	5.17
South East Coast	12 / 12	54,374	590	1.09	46,324	85.20	5	0.01	42	7.12
South West	17 / 17	64,425	555	0.86	59,445	92.27	1	0.00	17	3.06
West Midlands	14 / 14	81,099	1,650	2.03	78,440	96.72	11	0.01	93	5.64
Yorkshire and the Humber	11 / 11	70,605	884	1.25	69,076	97.83	1	0.00	88	9.95
Total England	134 / 139	706,041	13,870	1.96	661,765	93.73	213	0.03	772	5.57

High prevalence areas

Table AN-3 shows the number of booking bloods received in high prevalence areas, the number of screen positive women detected, and the number of high risk couples identified.

In 2015/16 there were 382,575 booking bloods reported by laboratories, of which 11,512 were identified as screen positive (approximately 1 in 33 women screened). Of these 651 were identified as high risk (approximately 1 in 18 screen positive women).

Table AN-3. Numbers screened and detected by sub-region 2015/16: high prevalence areas

Sub-region	No. of Labs	Booking bloods received (BBs)	Screen 'positive' women		Screen 'negative' women		Result pending/ inconclusive result		High risk couples identified	
	Submitted/ Total Labs	n	n	% of BBs	n	% of BBs	n	% of BBs	n	% of screen positive
East of England	5 / 5	24,609	634	2.58	23,090	93.83	0	0.00	50	7.89
East Midlands	5 / 5	35,214	697	1.98	34,473	97.90	0	0.00	38	5.45
London	21 / 24	136,876	6,228	4.55	130,726	95.51	44	0.03	309	4.96
North East	1 / 1	6,302	104	1.65	6,198	98.35	0	0.00	3	2.88
North West	7 / 7	48,082	965	2.01	45,678	95.00	60	0.12	54	5.60
South Central	5 / 5	28,791	473	1.64	27,624	95.95	28	0.10	27	5.71
South East Coast	2 / 2	10,937	254	2.32	10,425	95.32	0	0.00	17	6.69
South West	2 / 2	12,088	152	1.26	11,854	98.06	0	0.00	6	3.95
West Midlands	7 / 7	51,243	1,431	2.79	48,974	95.57	11	0.02	83	5.80
Yorkshire and The Humber	3 / 3	28,433	574	2.02	27,252	95.85	1	0.00	64	11.15
Total England	58 / 61	382,575	11,512	3.01	366,294	95.74	144	0.04	651	5.65

Low prevalence areas

Table AN-4 shows the number of booking bloods received in low prevalence areas, the number of screen positive women detected, and the number of high risk couples identified.

In 2015/16 there were 323,466 booking bloods reported by laboratories, of which 2,358 were identified as screen positive (approximately 1 in 137 women screened). Of these, 121 were identified as high risk (approximately 1 in 20 screen positive women).

Table AN-4. Numbers screened and detected by sub-region 2015/16: low prevalence areas

Sub-region	No. of Labs	Booking bloods received (BBs)	Screen 'positive' women		Screen 'negative' women		Result pending/ inconclusive result		High risk couples identified	
	Submitted/ Total Labs	n	n	% of BBs	n	% of BBs	n	% of BBs	n	% of screen positive
East of England	11 / 11	54,260	472	0.87	44,390	81.81	48	0.09	23	4.87
East Midlands	4 / 4	21,085	132	0.63	20,685	98.10	0	0.00	7	5.30
London	0 / 0	-	-	-	-	-	-	-	-	-
North East	7 / 7	25,210	151	0.60	21,109	83.73	12	0.05	8	5.30
North West	11 / 12	35,738	150	0.42	35,448	99.19	0	0.00	6	4.00
South Central	3 / 4	19,371	185	0.96	19,059	98.39	3	0.02	7	3.78
South East Coast	10 / 10	43,437	336	0.77	35,899	82.65	5	0.01	25	7.44
South West	15 / 15	52,337	403	0.77	47,591	90.93	1	0.00	11	2.73
West Midlands	7 / 7	29,856	219	0.73	29,466	98.69	0	0.00	10	4.57
Yorkshire and The Humber	8 / 8	42,172	310	0.74	41,824	99.17	0	0.00	24	7.74
Total England	76 / 78	323,466	2,358	0.73	295,471	91.35	69	0.02	121	5.13

3.4. The family origin questionnaire

Samples with a family origin questionnaire attached

The family origin questionnaire (FOQ) is used as a screening tool in both high and low prevalence areas, and it is important that a FOQ form is completed for each sample. This year we have moved to reporting on data for KPI ST3 rather than requesting this data separately from the laboratories. This change has been because the KPI data is now more robust, and we want to avoid duplication in data requests where possible to reduce the burden of data collection. As we cannot directly compare the KPI figures to the laboratory figures presented in previous reports, only two years of data are shown here.

Table AN-5 shows the number of antenatal samples received, and of these how many had a completed FOQ attached, by sub-region. The thresholds for this KPI are an acceptable level of 95% and an achievable level of 99%. All sub-regions are above the acceptable level, but no region as a whole is yet reaching the achievable level.

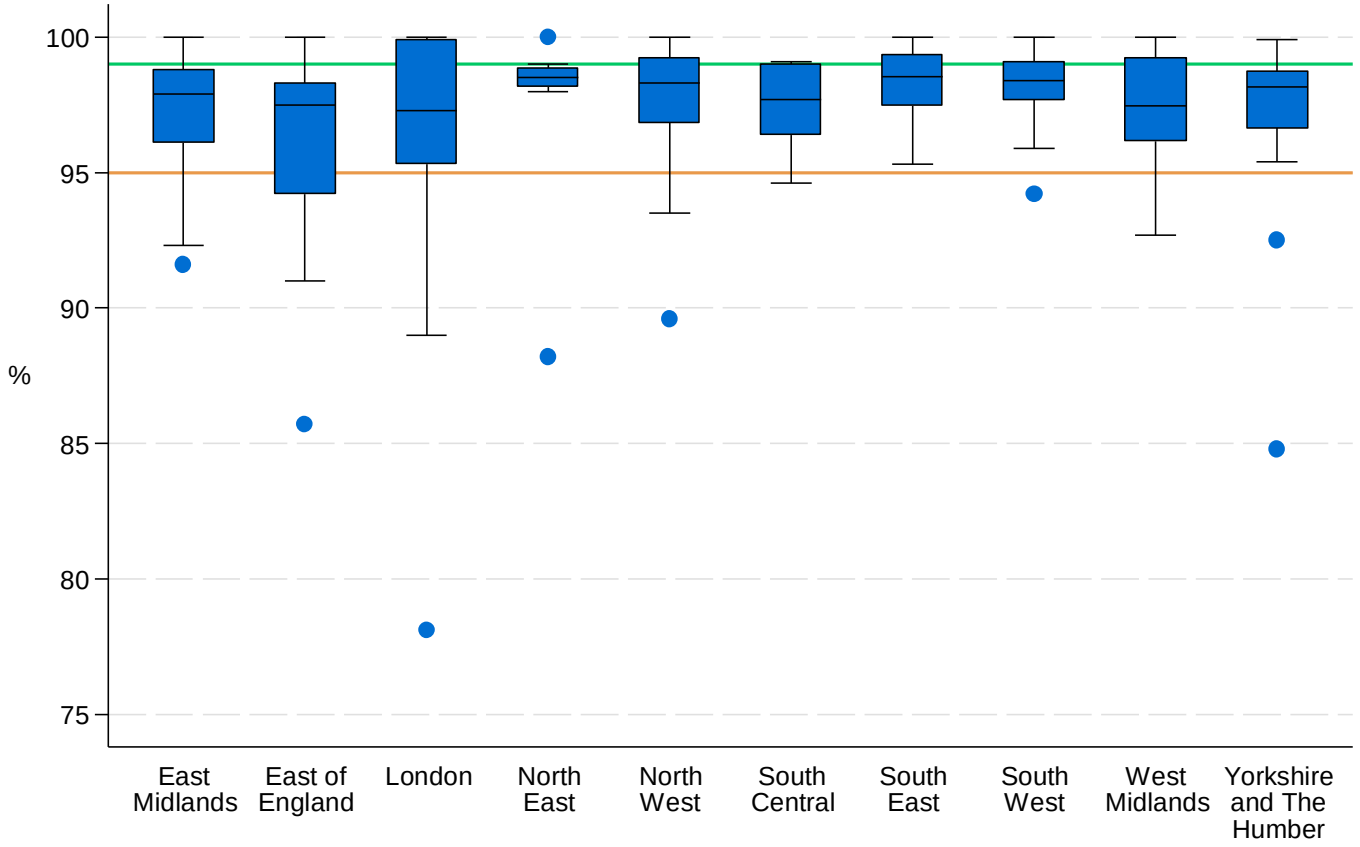
Table AN-5. Antenatal samples received with a completed FOQ by sub-region 2014/15 to 2015/16: England

Sub-region	2014/15			2015/16		
	Antenatal samples received	Completed FOQ attached	% of samples received	Antenatal samples received	Completed FOQ attached	% of samples received
East Midlands	55,313	53,422	96.6	54,976	53,309	97.0
East of England	68,286	64,847	95.0	75,303	72,086	95.7
London	127,878	115,622	90.4	130,263	124,913	95.9
North East	27,463	27,087	98.6	30,811	30,004	97.4
North West	83,703	80,852	96.6	86,203	83,457	96.8
South Central	42,491	40,899	96.3	41,751	40,657	97.4
South East	64,680	63,160	97.6	64,415	63,240	98.2
South West	65,268	63,860	97.8	63,956	62,628	97.9
West Midlands	77,447	75,353	97.3	75,101	72,766	96.9
Yorkshire & The Humber	71,786	70,118	97.7	68,214	66,091	96.9
England total	684,315	655,220	95.7	690,993	669,151	96.8

Exclusions where data was not returned in all four quarters: 2014/15: 11; 2015/16: 9.

Figure AN-2 shows variation in performance against this KPI by sub-region. Of the 10 sub-regions all but one have the whole of the interquartile range above the 95% acceptable level. For four sub-regions all trusts (excluding outliers) are above the acceptable threshold, and a number of trusts are reporting performance above the achievable level for this KPI.

Figure AN-2. Variation in FOQ completion by sub-region, 2015/16



Excludes 9 trusts that did not provide data for all four quarters.

3.5. Declined screening tests

Choice is an important element of population screening and as such screening tests for sickle cell and thalassaemia may be declined for a number of different reasons. Table AN-6 shows the number of women who declined antenatal screening by sub-region and for the whole of England as a proportion of booking bloods tested. National rates for declines have decreased each year since 2013/14 and are now at 0.2% (or 2 per 1,000) for 2015/16, compared to 0.34% in 2013/14.

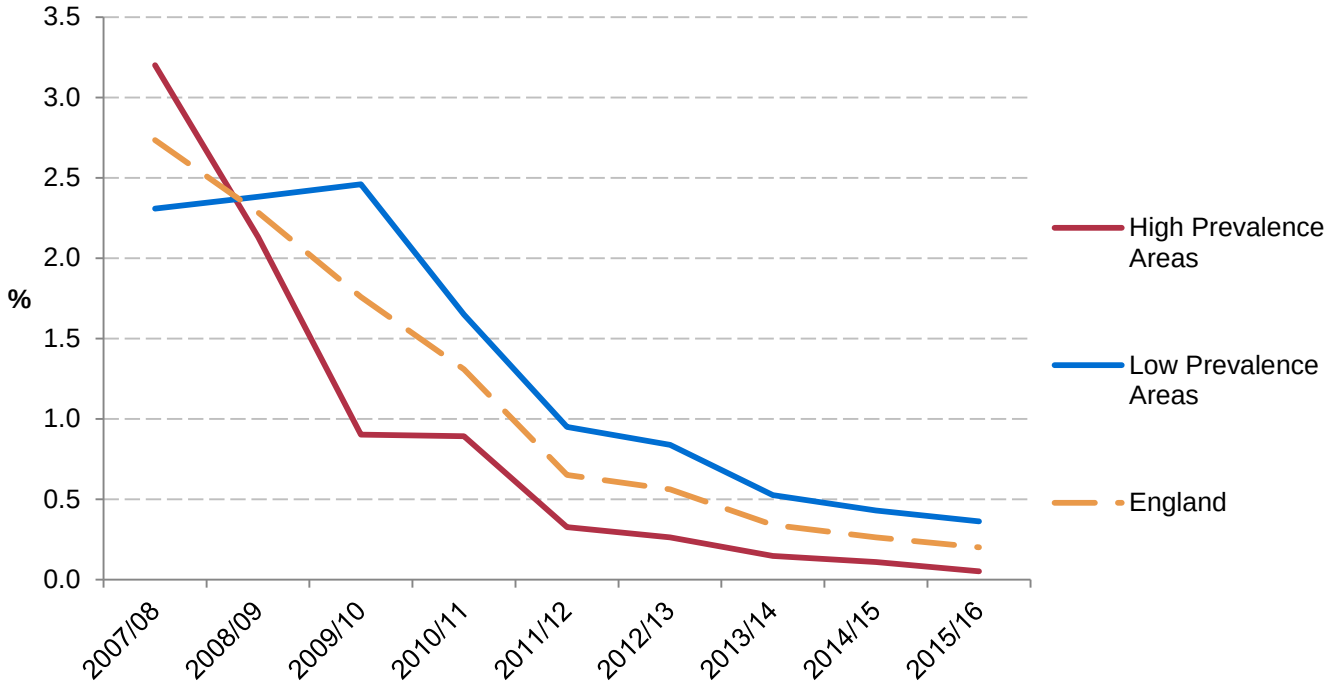
Table AN-6. Declined SCD screening tests by sub-region 2013/14 to 2015/16

Sub-region	2013/14			2014/15			2015/16		
	Booking bloods received (BBS)	Declined testing	% of BBs	Booking bloods received (BBS)	Declined testing	% of BBs	Booking bloods received (BBS)	Declined testing	% of BBs
East Midlands	52,367	93	0.18	54,544	85	0.16	51,822	67	0.13
East of England	81,303	426	0.52	71,349	403	0.56	74,006	300	0.41
London	105,994	49	0.05	126,075	60	0.05	124,816	28	0.02
North East	33,171	166	0.50	31,907	140	0.44	31,512	57	0.18
North West	73,911	121	0.16	72,244	57	0.08	79,913	71	0.09
South Central	48,783	79	0.16	49,302	59	0.12	48,162	17	0.04
South East Coast	50,834	67	0.13	51,249	97	0.19	49,188	73	0.15
South West	61,516	715	1.16	62,128	558	0.90	54,813	558	1.02
West Midlands	73,353	50	0.07	70,229	35	0.05	81,099	37	0.05
Yorkshire and The Humber	71,894	448	0.62	71,620	240	0.34	70,605	126	0.18
England Total	653,126	2,214	0.34	660,647	1,734	0.26	665,936	1,334	0.20

Exclusions based on missing or unavailable data: 2013/14: 15; 2014/15: 12; 2015/16: 8.

Figure AN-3 shows the change over time in rates of declined screening since 2007/08, broken down by high and low prevalence areas. Rates have dropped each year in both high and low prevalence areas, but rates for declined screening are consistently lower in high prevalence areas compared to low prevalence areas.

Figure AN-3. Declined tests for SCD as a percentage of booking bloods received 2007/08 to 2015/16



Exclusions based on missing or unavailable data: 2007/08: 40; 2008/09: 46; 2009/10: 32; 2010/11: 17; 2011/12: 14; 2012/13: 14; 2013/14: 15; 2014/15: 12; 2015/16: 8.

	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14	2014/15	2015/16
High prevalence areas	3.20	2.13	0.90	0.89	0.33	0.26	0.15	0.11	0.05
Low prevalence areas	2.31	2.38	2.46	1.65	0.95	0.84	0.53	0.43	0.36
England total	2.73	2.28	1.76	1.31	0.65	0.56	0.34	0.26	0.20

3.6. Testing of the baby's biological father

If a woman is identified as screen positive, the baby's biological father should be offered testing to determine the risk to the pregnancy. If the baby's biological father is not available for testing, it is not possible to accurately assess the risk status of the pregnancy and the screen positive woman should be offered prenatal diagnostic (PND) testing as if they were at risk. It is estimated that this group of women accounts for approximately 38% of screen positive women (calculated from the number of screen positive women minus the number of father specimens received).

Father uptake is calculated from the number of specimens requested from the biological fathers and the number of specimens received. Table AN-7 shows father uptake for antenatal testing for the last three years. In 2015/16 the national rate has remained at approximately 60% for the second consecutive year. Uptake varies between sub-

regions, ranging between approximately 50% in London to approximately 79% in South Central.

Table AN-7. Uptake of testing of the baby's biological father by sub-region 2013/14 to 2015/16

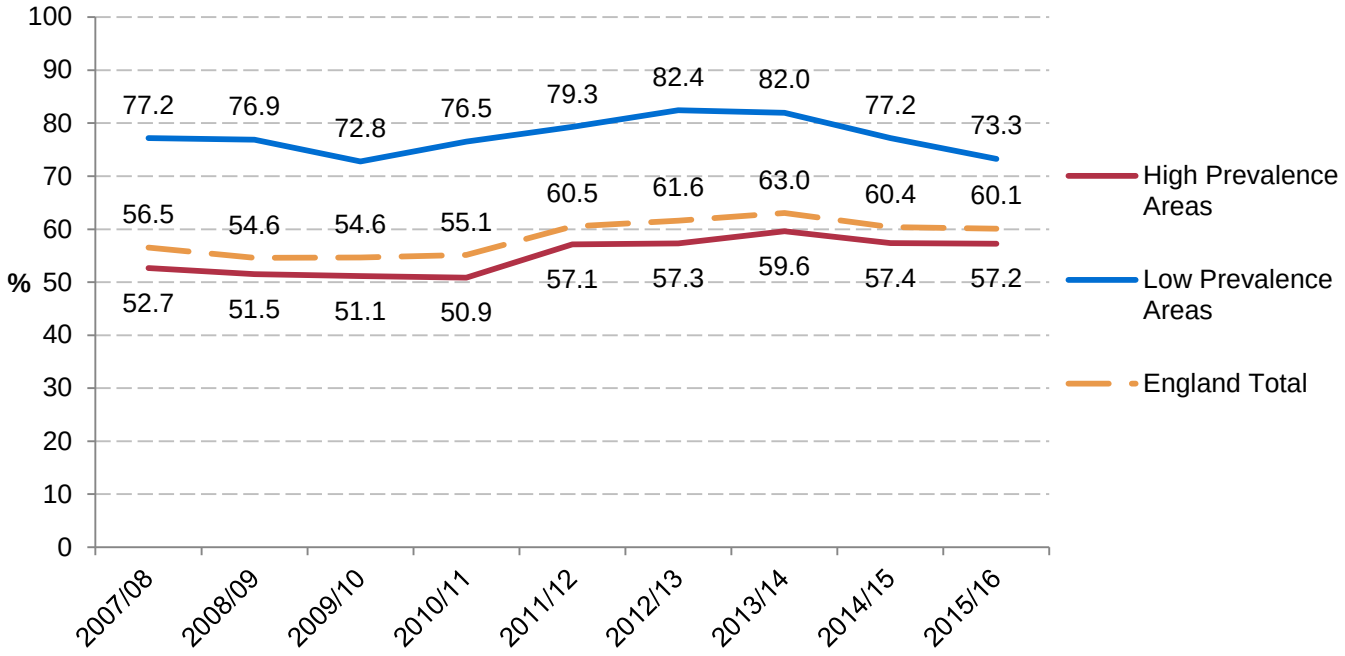
Sub-region	2013/14			2014/15			2015/16		
	Father samples requested	Father samples received	% uptake	Father samples requested	Father samples received	% uptake	Father samples requested	Father samples received	% uptake
East Midlands	867	680	78.43	718	504	70.19	850	604	71.06
East of England	1,072	703	65.58	1,016	734	72.24	1,171	760	64.90
London	7,705	4,201	54.52	7,225	3,646	50.46	6,340	3,205	50.55
North East	217	186	85.71	242	194	80.17	258	191	74.03
North West	904	595	65.82	1,082	719	66.45	1,137	735	64.64
South Central	556	471	84.71	746	564	75.60	695	552	79.42
South East Coast	665	496	74.59	610	393	64.43	626	386	61.66
South West	402	282	70.15	489	366	74.85	603	391	64.84
West Midlands	1,497	1,058	70.67	1,591	1,004	63.10	1,693	1,066	62.97
Yorkshire and The Humber	936	669	71.47	829	665	80.22	938	713	76.01
England total	14,821	9,341	63.03	14,548	8,789	60.41	14,311	8,603	60.11

Exclusions based on missing or unavailable data, the proportion was greater than 100%, or there were no screen positive cases: 2013/14: 11; 2014/15: 8; 2015/16: 2.

Figure AN-4 shows the uptake of testing by babies' biological fathers since 2007/08, both nationally and separated into high and low prevalence areas. National rates have remained at approximately 60% and high prevalence areas have remained at approximately 57%. In low prevalence areas, however, there appears to be a drop in father uptake, from 82% in 2012/13 and 2013/14 down to 73% in 2015/16.

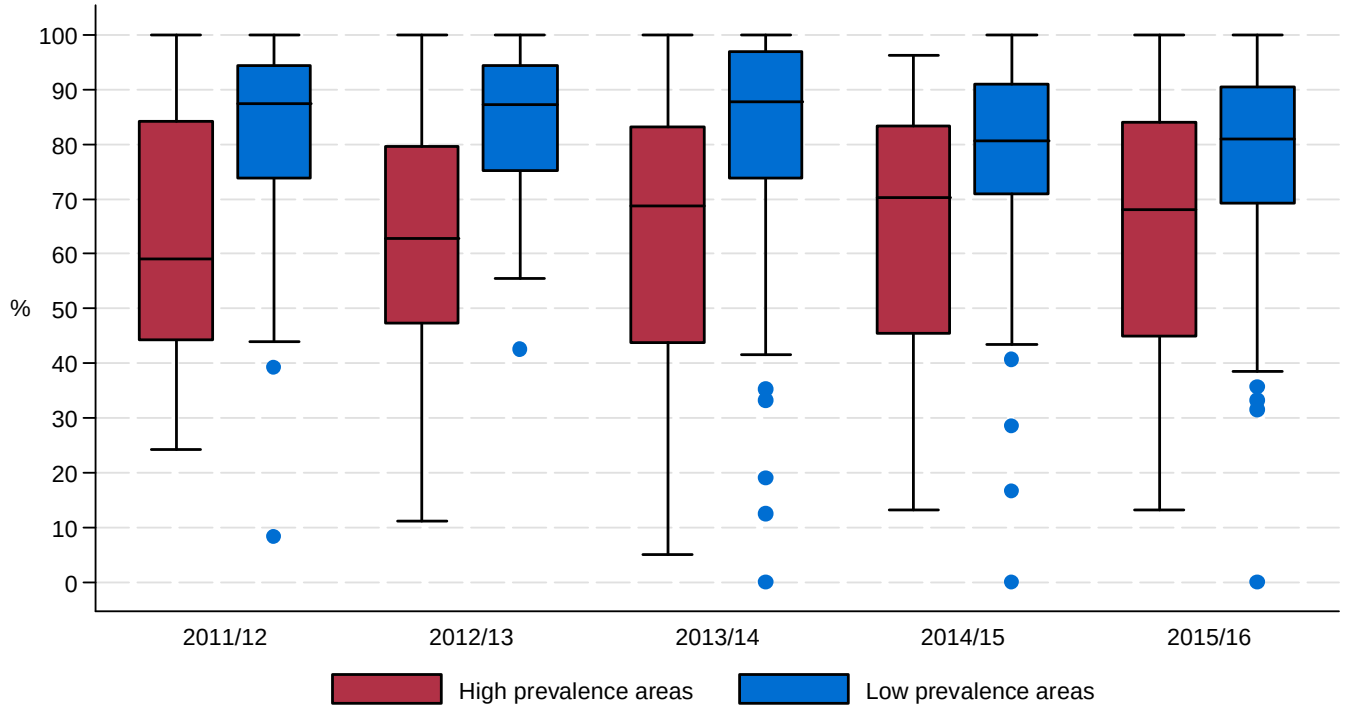
Figure AN-5 shows variation in uptake of testing for babies' biological father in the last five years. While the interquartile ranges for 2015/16 look similar to those for the previous year, in low prevalence areas there appears to be wider variation and more outliers compared to 2014/15.

Figure AN-4. Uptake of testing of the baby's biological father by prevalence 2007/08 to 2015/16



Exclusions based on missing or unavailable data, the proportion was greater than 100%, or there were no screen positive cases: 2007/08: 23; 2008/09: 15; 2009/10: 12; 2010/11: 6; 2011/12: 8; 2012/13: 8; 2013/14: 11; 2014/15: 8; 2015/16: 2.

Figure AN-5. Variation in uptake of testing for baby's biological father by prevalence 2011/12 to 2015/16



Exclusions based on missing or unavailable data, the proportion was greater than 100%, or there were no screen positive cases: 2011/12: 8; 2012/13: 8; 2013/14: 11; 2014/15: 8; 2015/16: 2.

'High risk' couples are identified based on both the baby's biological mother and father results. Breakdown data is requested on mother and father results in order to identify the specific risk of an affected pregnancy. This information also allows us to separate out sickle cell and thalassaemia screen positive results, and to identify cases where the baby's biological father wasn't available for testing or the laboratory can't link their results to the mother's results.

Table AN-8 shows screen positive results broken down by the risk to the pregnancy (based on the baby's biological father's results). Not all laboratories were able to provide complete breakdown data, so not all screen positive women are included. Data is included for 98% of screen positive women, and 94% of high risk couples.

Table AN-8. Breakdown of pregnancy risk for screen positive women 2015/16

		Risk to pregnancy				Totals		
	Mother's screening result	High Risk	Low/ minimal risk	Father not a carrier	Father result not available	Total number of mothers with result	Total for group	Rate/ 1,000 BBs received
Possible sickle cell affected baby	Hb S	474	102	2,374	2,632	5,582	7,451	10.55
	Hb D	*	*	479	159	674		
	Hb C	69	48	489	566	1,172		
	Hb O-Arab	*	*	12	10	23		
Possible beta thalassaemia affected baby	βThalassaemia	148	78	2,525	1,004	3,755	4,831	6.84
	δβ thalassaemia	*	4	31	13	48		
	Hb E	4	57	734	212	1,007		
	Hb Lepore	*	*	14	7	21		
Possible alpha thalassaemia affected baby	High risk alpha0	22	30	515	454	1,021	1,021	1.45
Other clinically significant mother results	HPFH/Compound heterozygous/ donor egg/bone marrow transplant	10	20	280	148	458	458	0.65
Other Hb variants requiring testing of baby's father		-	26	197	89	312	312	0.44
Totals		729	400	7,650	5,294	14,073	14,073	19.93

Note: 'Mother's screening results' include both cases where the mother is a carrier and where she is affected by a condition

*Numbers are suppressed to mask small numbers

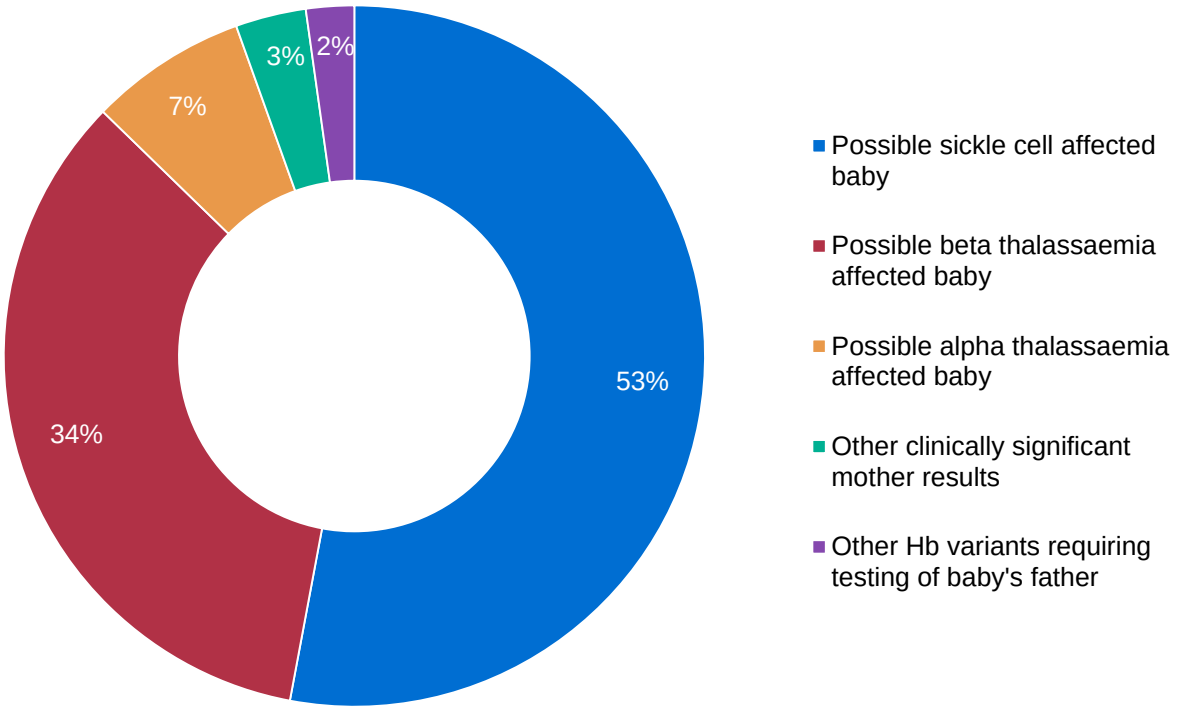
Not all laboratories were able to provide complete breakdown data for all screen positive women. For comparison, the total number of screen positive women reported by laboratories was 14,351 (98% included here) and 772 high risk couples (94% included here). The figure for rate per 1,000 booking bloods received is based on the number of booking bloods reported by laboratories with no exclusions made. The rates are therefore likely to be an under estimate.

'High risk' pregnancies are those represented by the dark orange boxes in the breakdown table in 'Appendix B: Antenatal data return form part 2 – breakdown of screen positive women'. Low risk pregnancies are represented by the light orange boxes, and minimal risk pregnancies by the white boxes.

Women with beta thalassaemia results are included in the 'possible beta thalassaemia affected baby' group in this table. However, HbS/beta thalassaemia is a sickle cell condition and these cases are included in the 'high risk' category.

Figure AN-6 shows the proportion of screen positive women at risk for each condition. Of the 14,073 women included in the breakdown data, 53% were at risk for sickle cell disease, 34% were at risk for beta thalassaemia, and 7% were at risk for alpha thalassaemia.

Figure AN-6. Screen positive women broken down by risk to the pregnancy 2015/16



Based on 14,073 of the 14,351 (98%) screen positive women reported by the laboratories in 2015/16.

3.7. Offer of screening early in pregnancy

The screening programme aims to support people to make informed choices during pregnancy. To be able to offer informed choice, there are two parts of antenatal screening. Pregnant women are offered antenatal screening for sickle cell and thalassaemia, and if they screen positive then testing should be offered to the baby's biological father. If the couple are identified as being at risk of having an affected pregnancy, they should be offered prenatal diagnostic (PND) testing of the fetus.

When offering informed choice, there are some important considerations to take into account. Women/couples who are at risk of an affected pregnancy may have strong beliefs that influence their decisions about having PND testing or about terminating the pregnancy. As such, they may wish to take these decisions privately before announcing the pregnancy, and so it is important that screening is offered as early as possible in the pregnancy.

Programme standard AO1b sets a target of prenatal diagnosis being completed (where the offer of PND testing was accepted) by 12 weeks and six days gestation. In order to allow time for all the tests for the mother, biological father, and fetus to be completed, programme standard AP1 sets a target of antenatal screening of pregnant women to be completed by 10 weeks and 0 days gestation.

Antenatal screening

As in section 3.4 ‘The family origin questionnaire’, this year we are reporting on data for KPI ST2 (timeliness of test) rather than requesting this data separately from the laboratories. The thresholds for this standard are 50% as the acceptable level and 75% as the achievable level.

Figures on gestation at antenatal screening can be dependent on completion of the FOQ to obtain gestational information. The numbers shown here may, therefore, appear lower than they actually are if the information on the FOQ was incomplete.

Table AN-9 shows the number and proportion of women tested by 10 weeks and 0 days gestation for the last two years. In 2015/16 national performance against this standard was 51.8%, with performance varying between regions, ranging from 34% in London to 63.7% in South Central. Of the 10 sub-regions, six were above the acceptable level, but no sub-region as a whole is yet reaching the achievable level.

Figure AN-7 shows the variation of performance against this standard broken down by sub-region. The median performance varies between each sub-region, and there appears to be wide variation in performance within some sub-regions. However, there are also a number of trusts that are above the achievable level in five of the sub-regions.

Figure AN-8 shows the variation of national performance comparing each quarter over the last two years. The median performance appears broadly comparable each quarter and there appears to be some slight fluctuation in the size of the interquartile range. In 2015/16 the bottom end of the range appears to getting wider, but this appears to be due to the reduction in the number of outliers. The top end of the range is also widening which indicates that there are trusts achieving higher levels for this standard. In 2015/16

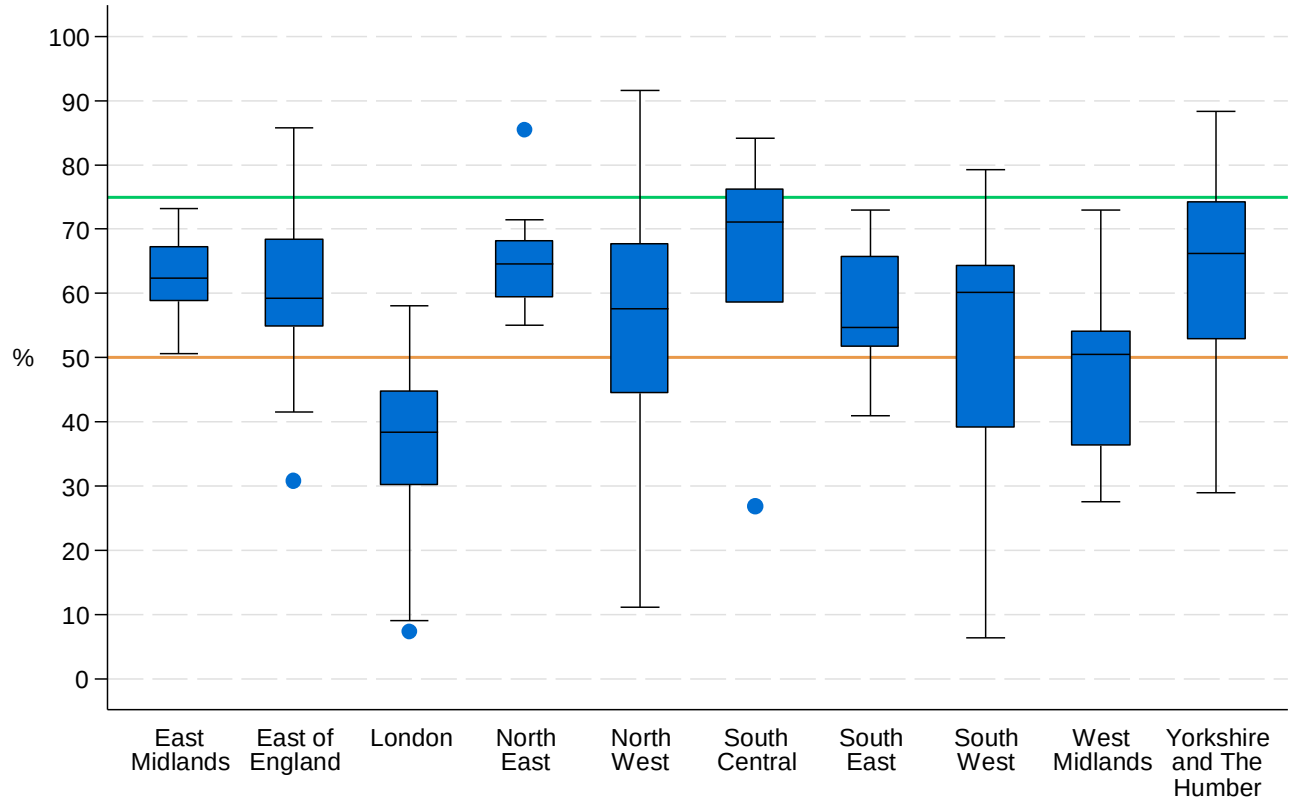
no trusts reported 0% of tests completed by 10 weeks, representing an improvement on 2014/15.

Table AN-9. Antenatal samples tested by 10+0 weeks gestation 2014/15 to 2015/16

Sub-region	2014/15			2015/16		
	Tested by 10+0 weeks	Samples received	%	Tested by 10+0 weeks	Samples received	%
East Midlands	33,481	54,633	61.3	34,337	55,165	62.2
East of England	37,832	67,513	56.0	43,049	73,801	58.3
London	44,125	118,052	37.4	46,430	136,450	34.0
North East	18,790	31,268	60.1	19,571	30,893	63.4
North West	41,580	78,258	53.1	43,672	83,143	52.5
South Central	24,057	42,557	56.5	26,622	41,768	63.7
South East	34,464	59,406	58.0	32,219	55,094	58.5
South West	31,364	64,518	48.6	31,265	63,945	48.9
West Midlands	28,718	70,545	40.7	34,249	75,054	45.6
Yorkshire & The Humber	40,012	65,952	60.7	41,328	65,245	63.3
England total	334,423	652,702	51.2	352,742	680,558	51.8

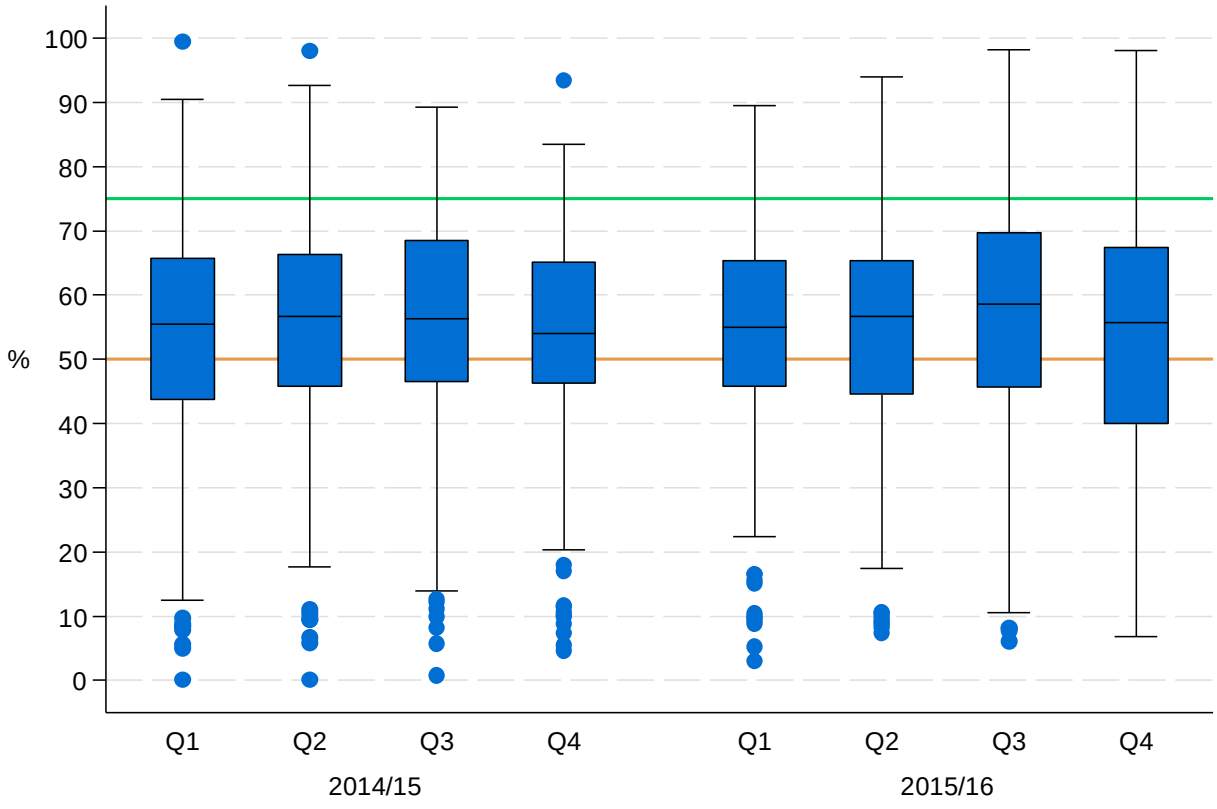
Exclusions where data was not returned in all four quarters: 2014/15: 16; 2015/16: 11.

Figure AN-7. Variation in testing by 10 weeks by sub-region 2015/16



Excludes 11 trusts where data was not returned in all four quarters.

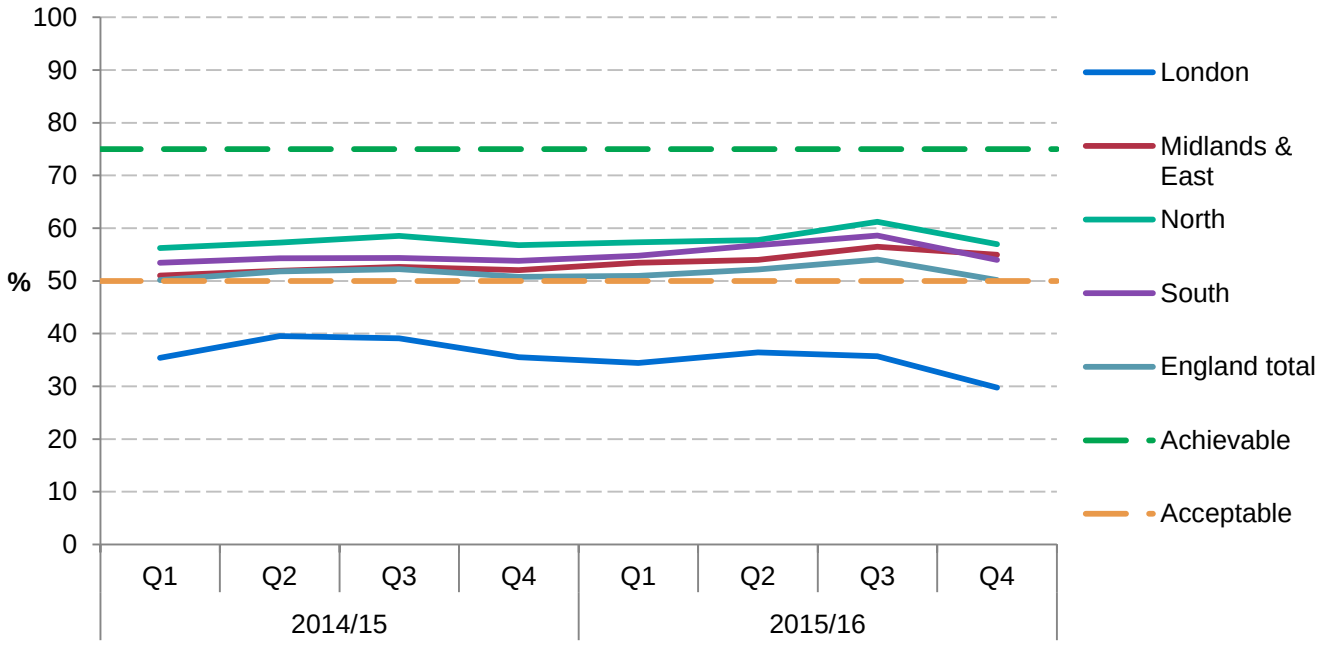
Figure AN-8. Variation in testing by 10 weeks by quarter 2014/15 to 2015/16



Exclusions where data was not returned in all four quarters: 2014/15: 16; 2015/16: 11.

Figure AN-9 shows the trends over time by region for 2014/15 to 2015/16. While the performance for regions outside of London appears to be fairly stable, there appears to be more of a decline in London.

Figure AN-9. Antenatal samples tested by 10+0 weeks by region, 2014/15 to 2015/16



Exclusions where data was not returned in all four quarters: 2014/15: 29; 2015/16: 26.

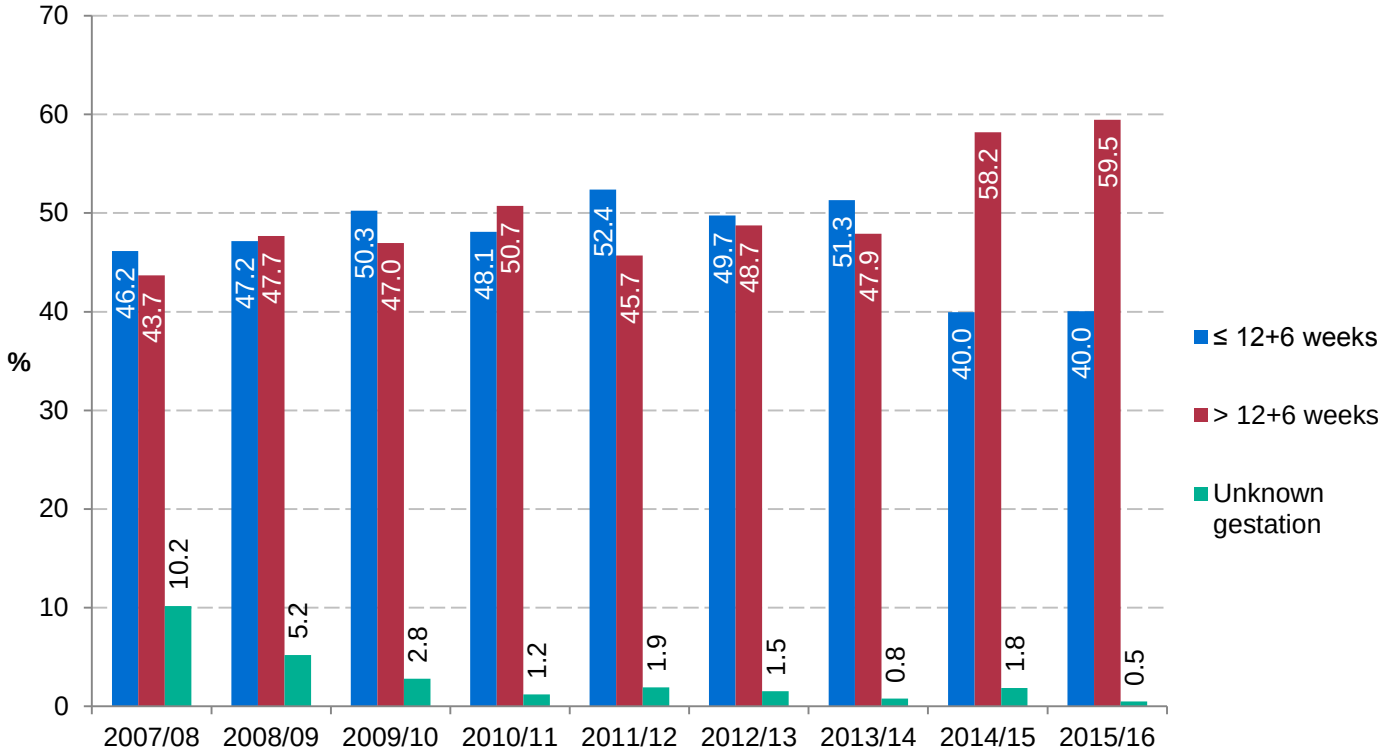
Prenatal diagnostic (PND) testing

Programme standard AO1b sets an acceptable level of 50% of all prenatal diagnoses to be performed by 12 weeks and six days gestation, and an achievable level of 75%. Table PND-1 shows the number and proportion of PND tests done before and after 12 weeks and six days gestation in the five-year period 2011/12 to 2015/16, and Figure PND-1 shows the proportions tested before and after this gestation since 2007/08. Prior to 2014/15 approximately half of PND tests were performed by 12 weeks and six days. However, in the last two years this has dropped to 40% of PND tests performed. As with last year, tests performed in the 13th and 14th week have increased by approximately 10% compared to rates before 2014/15 and the proportion tested in the 15th week or later remains at approximately 30%.

Table PND-1. Gestation at sample for PND, 2011/12 to 2015/16

Gestation	2011/12		2012/13		2013/14		2014/15		2015/16	
	n	%	n	%	n	%	n	%	n	%
<12+6 weeks	219	52.4	198	49.7	197	51.3	173	40.0	163	40.0
13+0 - 14+6 weeks	93	22.2	71	17.8	72	18.8	121	27.9	106	26.0
≥15+0 weeks	98	23.4	123	30.9	112	29.2	131	30.3	136	33.4
Unknown gestation	8	1.9	6	1.5	3	0.8	8	1.8	2	0.5
Total	418	100.0	398	100.0	384	100.0	433	100.0	407	100.0

Figure PND-1. Proportion of PND tests performed by gestation 2007/08 to 2015/16



3.8. Numbers tested and detected in prenatal diagnostic testing

Prenatal diagnostic (PND) testing should be offered to couples identified as 'at risk' in antenatal screening, and to carrier or affected women for whom the baby's biological father is not available for testing. Figure PND-2 shows the number of PND tests performed each year since 2007/08, broken down by PND laboratory. Numbers will differ from those previously reported as these figures now include data from the Manchester laboratory which started performing PND testing in 2013/14. In 2015/16 there were 407 tests performed, which appears consistent with previous years where there have been approximately 400 PND tests performed each year.

Figure PND-2. Number of PND tests performed, 2007/08 to 2015/16

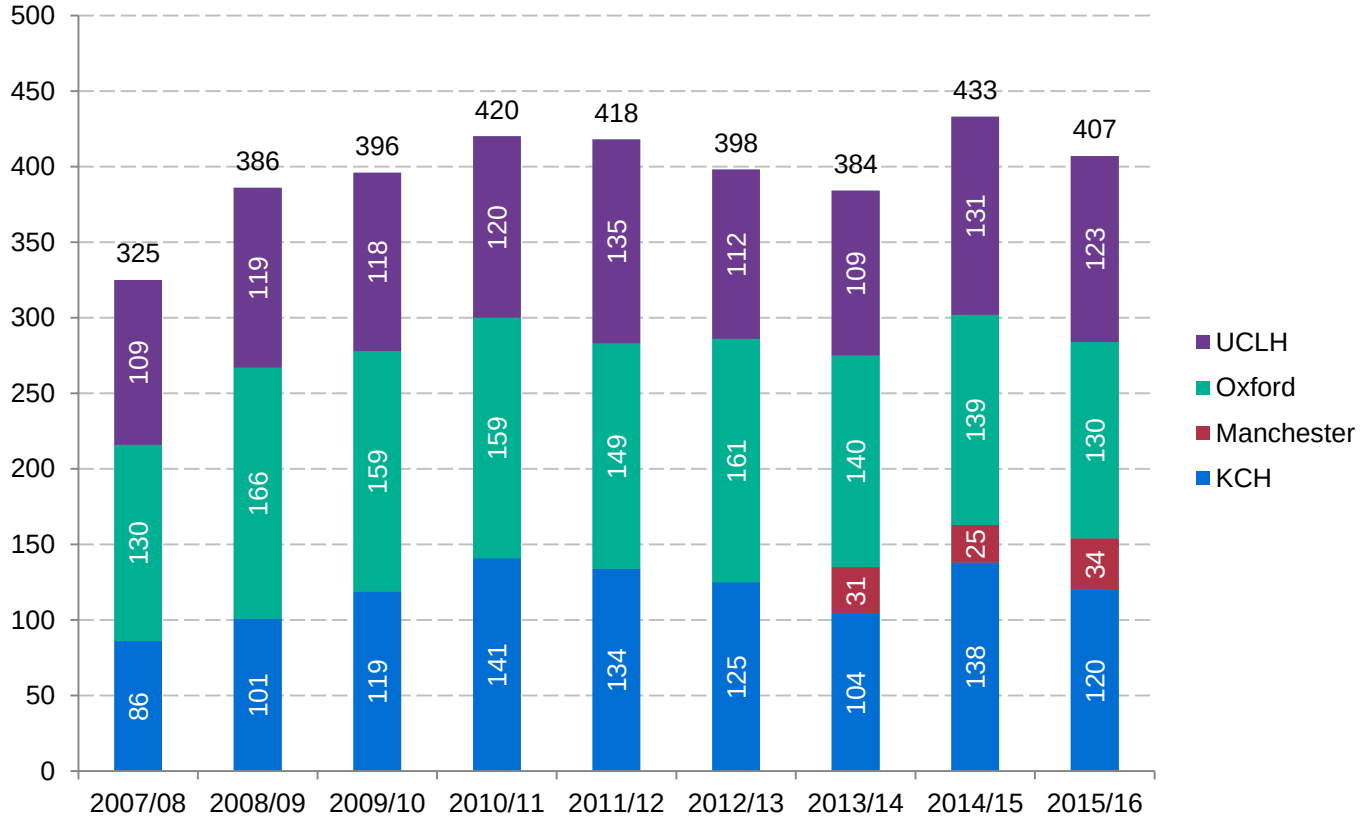


Table PND-2 breaks down the number of PND tests performed in the last five years by sub-region as a proportion of all tests performed in that year. Each year, approximately half of all PND tests performed were from London.

Table PND-3 shows the number of affected, carrier, 'no abnormality detected' (NAD), and inconclusive results, broken down by fetal result or risk to pregnancy. In 2015/16 104 PND tests had affected results, 179 had carrier results, 123 had NAD results, and one had inconclusive results.

Table PND-2. Number of PNDs performed by region 2015/16

Region	2011/12		2012/13		2013/14		2014/15		2015/16	
	n	%	n	%	n	%	n	%	n	%
North	36	8.6	6	1.5	35	9.1	25	5.8	36	8.8
South	28	6.7	19	4.8	12	3.1	14	3.2	17	4.2
Midlands & East	55	13.2	28	7.0	24	6.3	32	7.4	33	8.1
London	249	59.6	195	49.0	189	49.2	230	53.1	193	47.4
Unknown Region	50	12.0	150	37.7	124	32.3	132	30.5	128	31.4
England Total	418	100.0	398	100.0	384	100.0	433	100.0	407	100.0

Table PND-3. Breakdown of PND fetal results by condition 2011/12 to 2015/16

Fetal result	PND result/risk	2011/12		2012/13		2013/14		2014/15		2015/16	
		n	%	n	%	n	%	n	%	n	%
Affected	Sickle Cell affected	87	20.8	68	17.1	76	19.8	99	22.9	80	19.7
	Thalassaemia affected	14	3.3	17	4.3	19	4.9	23	5.3	24	5.9
	Other	1	0.2	0	0.0	1	0.3	1	0.2	0	0.0
Carrier	Sickle Cell carrier	139	33.3	160	40.2	127	33.1	155	35.8	125	30.7
	Thalassaemia carrier	53	12.7	41	10.3	40	10.4	39	9.0	46	11.3
	Other	14	3.3	12	3.0	13	3.4	12	2.8	8	2.0
NAD	Risk for Sickle Cell	63	15.1	52	13.1	83	21.6	89	20.6	97	23.8
	Risk for Thalassaemia	6	1.4	18	4.5	23	6.0	15	3.5	26	6.4
	Risk not known	35	8.4	26	6.5	1	0.3	0	0.0	0	0.0
Inconclusive/ result not known	All risks	6	1.4	4	1.0	1	0.3	0	0.0	1	0.2
Total		418	100.0	398	100.0	384	100.0	433	100.0	407	100.0

Alpha and beta thalassaemia cases are grouped due to the small number of alpha thalassaemia cases.

†'Sickle Cell affected' includes HbSS, HbSC, HbS/beta thalassaemia, and HbS+other variant requiring clinical follow-up; 'Sickle Cell carrier' includes HbAS results 'Thalassaemia' includes both alpha and beta thalassaemias as well as HPFH results; 'Other' includes other haemoglobinopathy variants; 'Inconclusive' results include both those declared as "inconclusive" in the data returns and those where the data was not of a quality to determine a result with certainty; 'Not known' includes cases where no data was provided by the PND laboratory.

3.9. Prenatal diagnostic results by family origin

Table PND-4 shows the number of PND tests performed broken down by mother's family origins. In 2015/16 approximately half of PND tests performed had African family origins, and approximately 20% had Caribbean family origins. The 'mixed/other' category includes cases where multiple family origins were given, or where the family origins did not fit in to one of the other categories.

Table PND-4. Number of PND tests by mother's family origins 2011/12 to 2015/16

Mother's family origin	2011/12		2012/13		2013/14		2014/15		2015/16	
	n	%	n	%	n	%	n	%	n	%
African	211	50.5	180	45.2	187	48.7	308	71.1	201	49.4
Caribbean	12	2.9	14	3.5	62	16.1	16	3.7	88	21.6
Indian	9	2.2	9	2.3	3	0.8	12	2.8	17	4.2
Pakistani	2	0.5	2	0.5	5	1.3	4	0.9	7	1.7
Cypriot/Mixed Cypriot	7	1.7	4	1.0	6	1.6	7	1.6	9	2.2
Other Asian	50	12.0	27	6.8	36	9.4	30	6.9	33	8.1
Southern & Other European	3	0.7	8	2.0	6	1.6	10	2.3	11	2.7
Middle Eastern	4	1.0	4	1.0	6	1.6	5	1.2	8	2.0
Mixed/Other	97	23.2	66	16.6	12	3.1	5	1.2	15	3.7
Not Known	23	5.5	84	21.1	61	15.9	36	8.3	18	4.4
Total	418	100.0	398	100.0	384	100.0	433	100.0	407	100.0

3.10. Pregnancy outcomes

One of the aims of antenatal screening for sickle cell and thalassaemia is to offer couples informed choice. The screening programme collects data on pregnancy outcomes following PND testing to assess what choices couples make following PND testing. Table PND-5 shows the number of PND tests with an affected result, broken down by condition and pregnancy outcome. Numbers with alpha thalassaemia affected results are small, and so rates for this condition should be interpreted with caution. It should also be noted that a large number of PND tests do not have a known pregnancy outcome which may also affect the rates shown.

Table PND-5. Outcomes for pregnancies with affected fetal results at PND 2013/14 to 2015/16

Condition	Pregnancy outcome	2013/14 % of total identified with condition	2014/15 % of total identified with condition	2015/16 % of total identified with condition
Sickle Cell	Continued	19.7	27.3	15.0
	Terminated	46.1	38.4	46.3
	Not Known	34.2	34.3	38.8
Beta Thalassaemia	Continued	6.3	22.7	18.2
	Terminated	31.3	40.9	54.5
	Not Known	62.5	36.4	27.3
Alpha Thalassaemia	Continued	0.0	0.0	0.0
	Terminated	33.3	100.0	100.0
	Not Known	66.7	0.0	0.0
Total Affected (n)		96	123	104

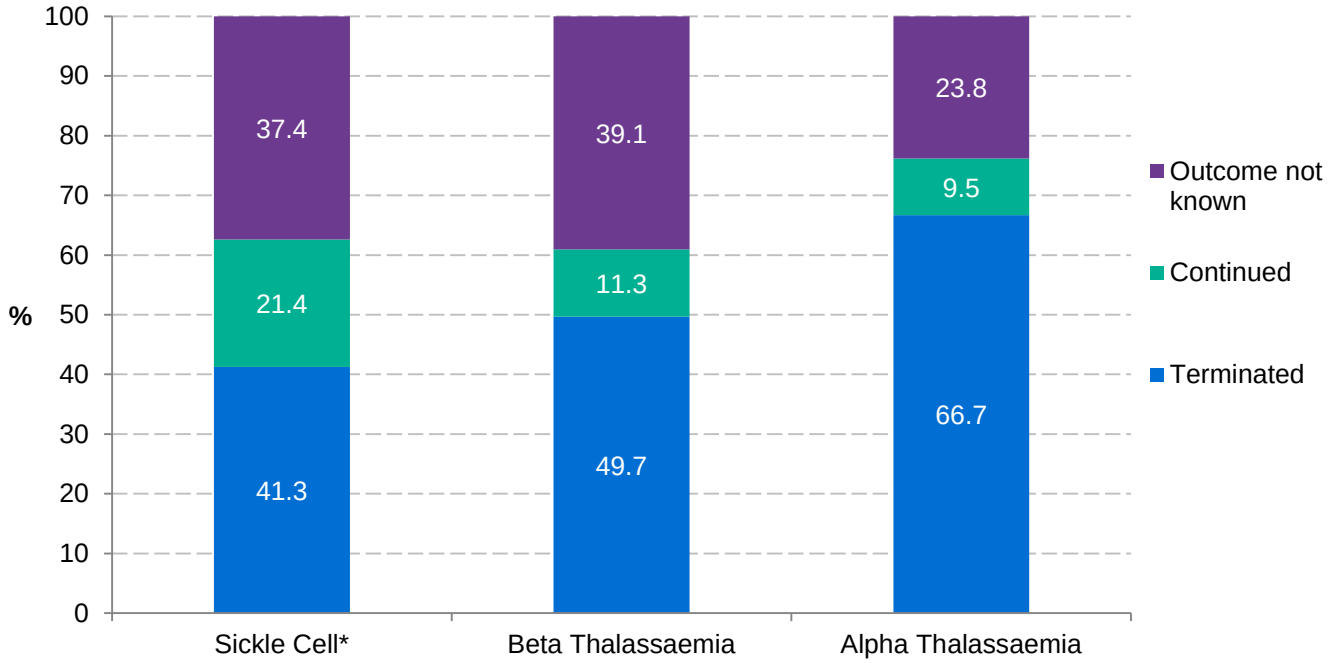
Other haemoglobin variants and miscarriage outcomes have been excluded.

Please note that alpha thalassaemia rates are based on small numbers and should be treated with caution.

Figure PND-3 shows the proportion of affected PND results where parents opted to either continue or terminate the pregnancy. These figures cover the eight-year period since 2008/09 and exclude cases of miscarriage due to small numbers. Figure PND-4 shows this information for PNDs with a known pregnancy outcome only. Of the PND tests with a sickle cell affected diagnosis and a known outcome, approximately 66% opted to terminate, compared to 82% with a beta thalassaemia diagnosis and 88% with an alpha thalassaemia diagnosis.

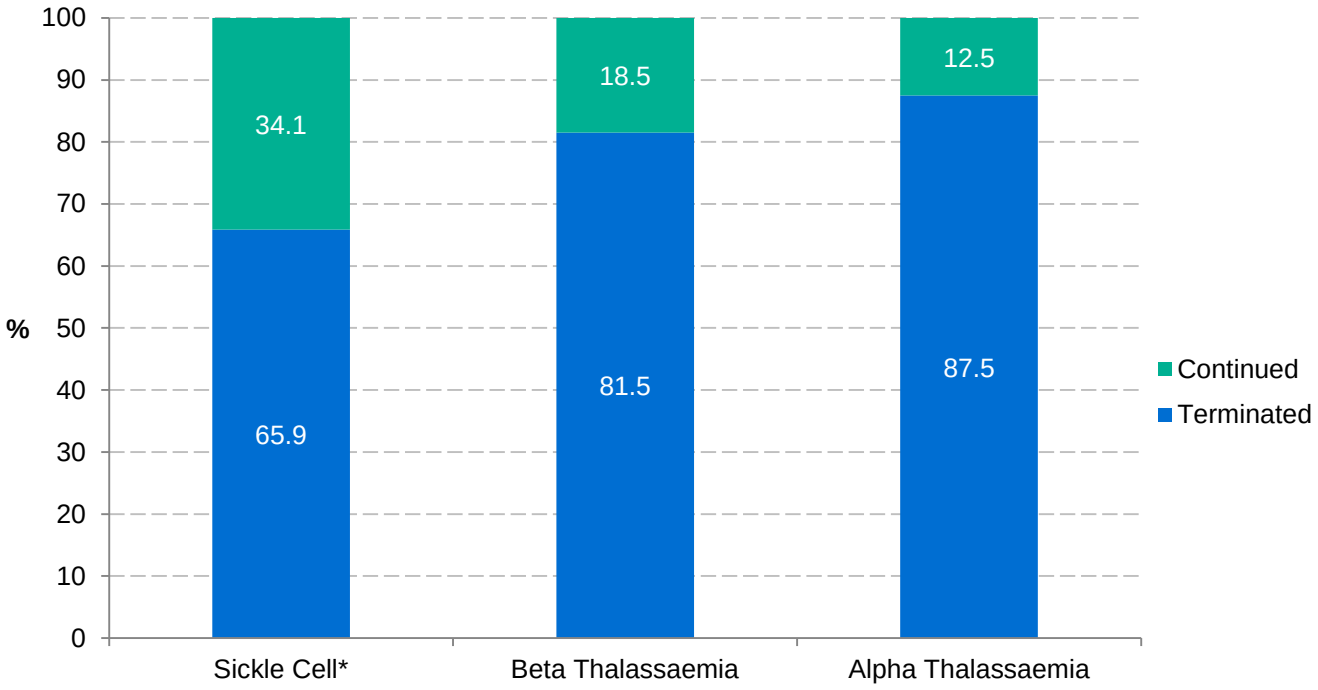
Figure PND-5 shows pregnancy outcomes by gestation at PND test, excluding miscarriages, since 2008/09. Where there were affected results, it appears that a greater proportion opted to terminate if they were tested earlier in the pregnancy than those tested later in the pregnancy. This may indicate that the later in pregnancy that PND testing takes place, the less likely parents are to choose to terminate.
Figure PND-6 shows this information for PNDs with a known pregnancy outcome only.

Figure PND-3. Outcomes for pregnancies with 'affected' diagnosis at PND 2008/09 to 2015/16



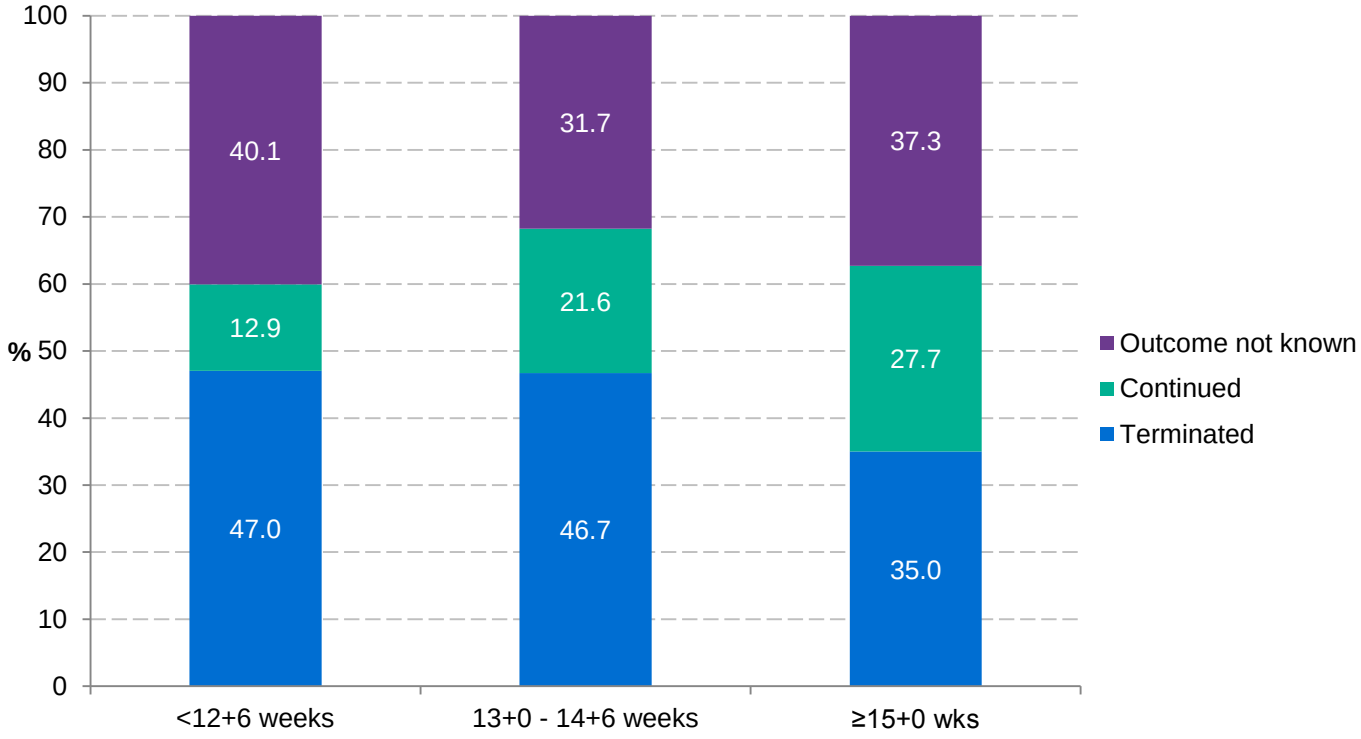
*The 'Sickle Cell' category includes HbSS, HbSC, HbS/beta thalassaemia, and HbS+other variant requiring clinical follow-up. Excludes miscarriage outcomes due to small numbers.

Figure PND-4. Outcomes for pregnancies with 'affected' diagnosis at PND (known outcomes only) 2008/09 to 2015/16



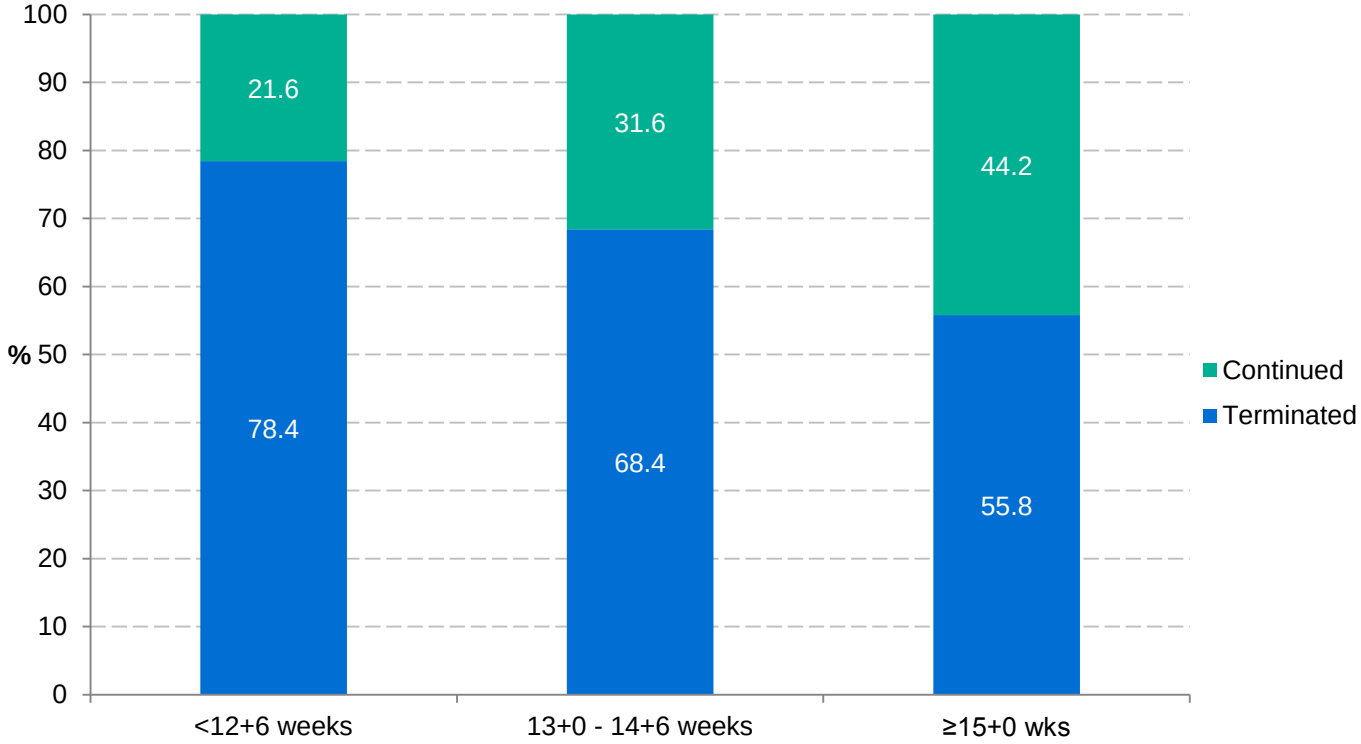
*The 'Sickle Cell' category includes HbSS, HbSC, HbS/beta thalassaemia, and HbS+other variant requiring clinical follow-up. Excludes miscarriage outcomes, and 295 cases where pregnancy outcome was not known.

Figure PND-5. Proportion of affected results with known pregnancy outcomes, grouped by gestation, by outcome, 2008/09 to 2015/16



Excludes miscarriage outcomes and cases where the gestation at PND was unknown.

Figure PND-6. Proportion of affected results with known pregnancy outcomes, grouped by gestation, by outcome (known outcomes only), 2008/09 to 2015/16



Excludes unknown and miscarriage outcomes, and cases where the gestation at PND was unknown.

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4. Newborn screening data

4.1. Response rates and data quality

Response rate

Data was received from all 13 newborn screening laboratories in England, and this year we have also included data from the Scotland, Wales, and Northern Ireland laboratories. We would like to thank all those involved in collecting and submitting this data to the screening programme.

Data quality

Newborn laboratories report on ‘samples’ which may differ from the number of babies tested. Data by region and by ethnicity are collected separately, which can lead to discrepancies when comparing the figures.

Birth figures from the Office of National Statistics (ONS) offer a data quality check by comparing numbers of babies born with numbers of samples screened. ONS figures report on calendar years whereas the laboratory figures report on financial years. So these datasets do not exactly match.

Other differences may be accounted for by samples tested where their region was unknown (babies reported by the laboratories as ‘out of region’ or ‘unknown region’), repeat tests, and babies who were born abroad who moved to England up to one year of age. However, comparing these two datasets shows these figures to be broadly similar, offering some data validation for the laboratory figures. The ‘total screened’ figure combines the number of normal and abnormal results with the number of declines.

The programme requests data on laboratory processes and timeliness of entry into care for screen positive babies. In 2015/16 there were 293 babies accounted for in the timeliness data, compared to 292 in the regional data (including F-only cases). The difference of one is due to an FS case that was accounted for in the timeliness data but not in the regional data. There were also some differences in how some of the screen positives were reported, with one laboratory reporting a case as FSC in the regional data and FS-other in the timeliness data, and another laboratory reporting a case as FSC in the regional data and FS in the timeliness data. Completeness of timeliness data varies between data fields, and cases for which no information was given are identified in section 4.7 ‘Laboratory processes and entry into care’.

Table NB-1. Comparison of ONS birth figures and number of samples screened reported by newborn screening laboratories 2015/16

Sub-region	Data from newborn laboratories*	ONS figures†	Discrepancy (%)
East of England	71,375	72,505	1.56
East Midlands	48,583	53,641	9.43
London	130,150	129,615	-0.41
North East	28,596	28,400	-0.69
North West	87,106	85,838	-1.48
South East	102,284	102,703	0.41
South West	53,466	58,033	7.87
West Midlands	70,676	69,806	-1.25
Yorkshire and The Humber	68,462	63,858	-7.21
Unknown Region	7,102	-	-
England Total	667,800	664,399	-0.51
Scotland	55,616	55,098	-0.94
Wales	33,181	33,279	0.29
Northern Ireland	24,569	24,215	-1.46
UK total	781,166	776,991	-0.54

*Data collected from the newborn laboratories in England, Scotland, Wales, and Northern Ireland. This data covers the financial year 2015/16.

†Data from ONS (Live Births by Area of Usual Residence 2015, found at: www.ons.gov.uk/ons/rel/vsob1/births-by-area-of-usual-residence-of-mother--england-and-wales/index.html). This data covers the 2015 calendar year.

4.2. Newborn screening coverage

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

Table NB-2 shows a summary of the annual data for KPI NB1 for the last two years. The thresholds for this KPI are set at 95.0% as an acceptable level and 99.9% as an achievable level. While there have been some small changes in sub-regions between 2014/15 and 2015/16, the national rate remains at approximately the same level. Of the

10 sub-regions six are above the acceptable threshold for this KPI. No region as a whole is yet reaching the achievable level.

It should be noted that the coverage figures from the KPI data only include those born and resident in the sub-region and will not include movers-in.

Table NB-2. Coverage of newborn screening by sub-region 2014/15 to 2015/16

Sub-region	2014/15			2015/16		
	Babies tested within 17 days	Babies eligible for newborn screening	% of eligible population	Babies tested within 17 days	Babies eligible for newborn screening	% of eligible population
East Midlands	49,754	53,099	93.7	50,351	53,757	93.7
East of England	54,821	55,701	98.4	56,606	57,756	98.0
London	89,585	92,122	97.2	98,962	102,689	96.4
North East	20,902	21,457	97.4	25,281	25,825	97.9
North West	56,780	58,573	96.9	69,553	74,245	93.7
South Central	33,043	33,849	97.6	46,748	47,871	97.7
South East	29,768	32,151	92.6	34,636	35,953	96.3
South West	36,735	39,491	93.0	51,059	55,426	92.1
West Midlands	44,992	45,803	98.2	64,826	66,441	97.6
Yorkshire & The Humber	51,012	55,496	91.9	57,715	61,394	94.0
England total	467,392	487,742	95.8	555,737	581,357	95.6

Exclusions where data was not returned in all four quarters: 2014/15: 47; 2015/16: 13.

Figure NB-1 shows coverage broken down by CCG for 2015/16, showing coverage for newborn screening geographically. Birmingham CrossCity was the only CCG in England that is reporting coverage at the achievable level.

Figure NB-2 shows the variation in coverage by sub-region. While some sub-regions show wide variation in performance, all trusts in two sub-regions reported above 95% coverage in 2015/16.

Figure NB-1. Newborn coverage 2015/16 by CCG

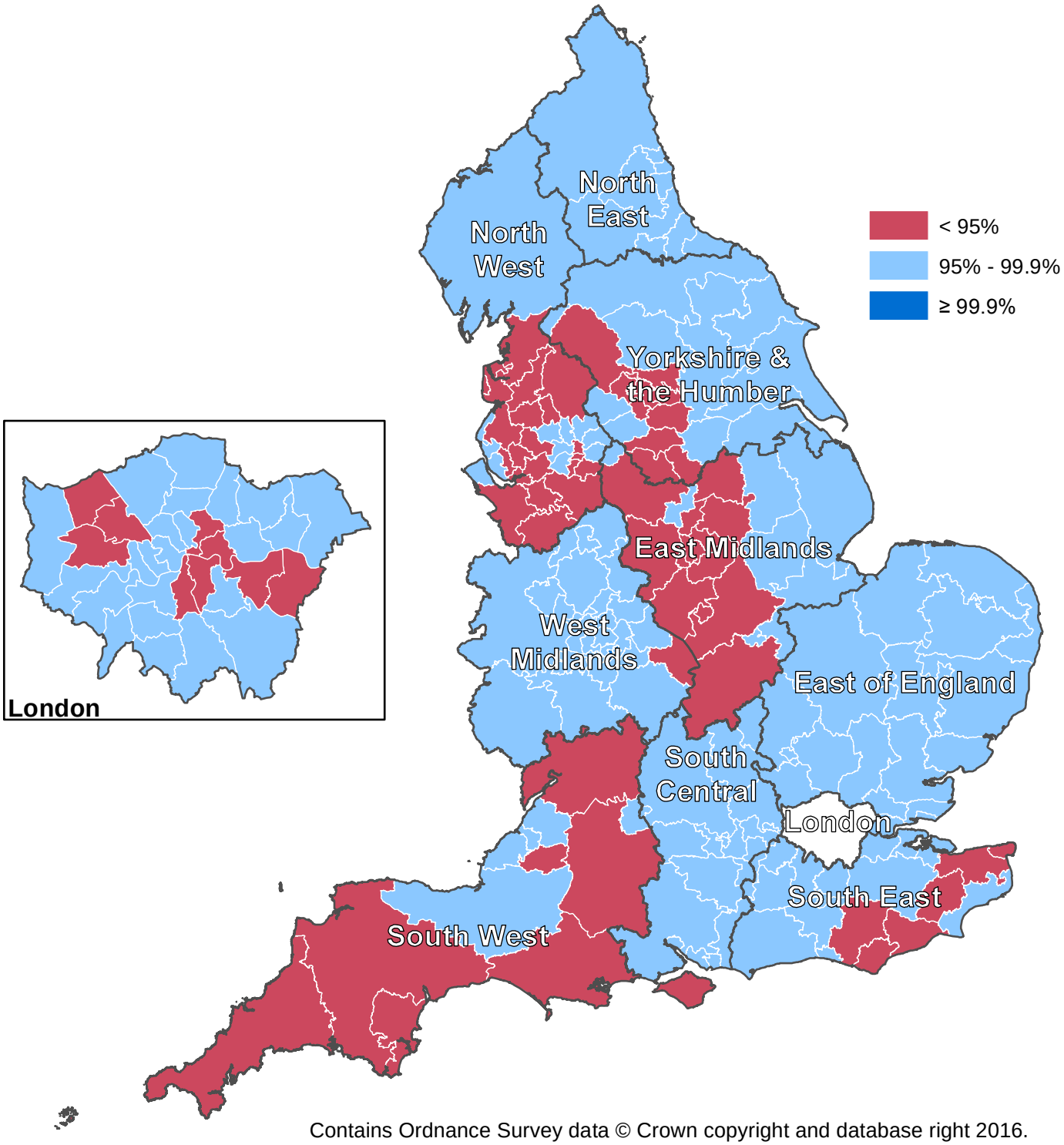
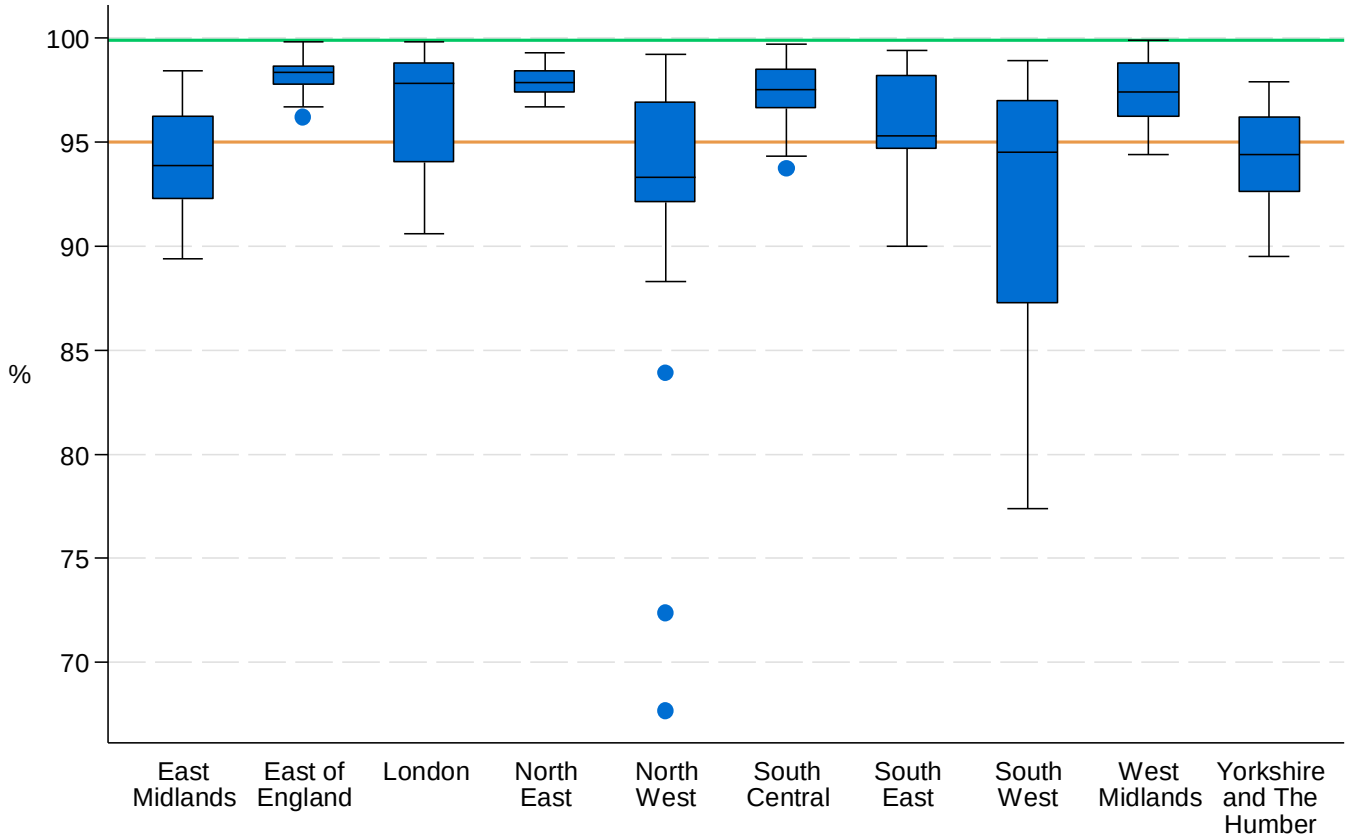


Figure NB-2. Newborn coverage 2015/16: England by sub-region



Excludes 13 trusts where data was not returned in all four quarters.

4.3. Numbers screened and results

Numbers screened

This year marks the 10th anniversary of full roll-out of newborn screening in July 2006. This year we have included figures showing numbers screened and detected in the 11 years since data collection began in 2005/06. Table NB-3 shows the numbers screened and detected by the screening programme in England. In this period 7.3 million babies have been screened in England. Of these, just over 3,500 babies (1 in 2032) were identified with significant conditions, and approximately 101,500 (1 in 72) were identified as carriers.

Table NB-4 shows the numbers screened and detected by the screening programme in 2015/16 only, and includes data for Scotland, Wales and Northern Ireland. In this year, 667,800 babies were screened in England (including the 1,359 where the parents declined testing), and 781,116 in the whole of the UK.

Table NB-3. Samples screened and newborn screening results 2005/06 to 2015/16

Region	Significant Conditions					Carriers					Total Screened		
	FS	FSC	FS-Other	FE	F-only	FAS	FAC	FAD	FAE	Other	Transfused	Declined	Normal+ Abnormal
North	257	58	72	27	90	8,201	1,381	1,807	1,945	1,752	5,211	1,452	1,982,375
South	182	87	16	15	19	6,699	1,517	1,335	1,593	977	4,742	1,291	1,702,125
Midlands & East	404	136	24	37	91	13,237	3,142	2,470	2,338	745	6,367	1,891	2,064,707
London	1,528	571	56	85	65	36,024	7,770	2,110	4,083	1,275	6,031	1,271	1,411,487
Unknown region	20	8	0	0	3	649	159	91	92	76	2,882	408	120,744
England Total	2,391	860	168	164	268	64,810	13,969	7,813	10,051	4,825	25,233	6,313	7,281,438

Table NB-4. Samples screened and newborn screening results 2015/16

Region	Significant Conditions					Carriers					Total Screened		
	FS	FSC	FS-Other	FE	F-only	FAS	FAC	FAD	FAE	Other	Transfused	Declined	Normal+ Abnormal
North	15	8	*	*	7	867	134	155	179	27	325	269	183,895
South	12	10	*	*	*	660	138	88	126	17	166	269	155,481
Midlands & East	46	13	*	5	13	1,252	295	205	203	*	275	355	190,279
London	96	34	*	12	5	2,743	653	195	362	44	269	312	129,838
Unknown region	*	*	*	*	*	143	28	33	25	5	64	154	6,948
England Total	173	67	6	19	27	5,665	1,248	676	895	95	1,099	1,359	666,441
Scotland	*	*	*	*	*	159	21	17	19	*	40	45	55,571
Wales †	*	*	*	*	*	*	*	*	*	*	32	139	33,042
Northern Ireland	*	*	*	*	*	*	*	9	8	*	32	175	24,394
UK total	178	69	6	19	30	5,854	1,275	702	922	95	1,203	1,718	779,448

*Numbers are suppressed to mask small numbers

† The Wales newborn screening protocol is designed to detect only the disease states of Sickle Cell Disorder. However, any carriers identified from the screening process are referred for follow-up.

Significant conditions

Significant conditions comprise FS, FSC, FS-other and FE results. Table NB-5 shows the number and rates of significant conditions detected since 2005/06. Of the 7.3 million babies screened in this period, 3,583 were identified with significant conditions, or 1 in 2,034.

Table NB-5. Number and rates of significant conditions detected 2005/06 to 2015/16

Sub-region	Significant Conditions			No. of babies Screened
	n	Rate/ 1000	1 in x	
East Midlands	130	0.24	4,226	549,395
East of England	239	0.32	3,104	741,933
London	2,240	1.59	631	1,412,758
North East	34	0.11	9,052	307,778
North West	243	0.26	3,908	949,670
South Central	151	0.29	3,443	519,825
South East Coast	90	0.16	6,338	570,444
South West	59	0.10	10,392	613,147
West Midlands	232	0.30	3,342	775,270
Yorkshire and the Humber	137	0.19	5,302	726,379
Unknown region	28	0.23	4,327	121,152
England Total	3,583	0.49	2,034	7,287,751

Table NB-6 shows these figures for the last three years. Both numbers and rates for significant conditions appear to have fallen in this three-year period, from 319 (0.48 per 1,000) in 2013/14 to 265 (0.40 per 1,000) in 2015/16. Broken down by sub-region, rates in 2015/16 ranged between 1 in 904 in London and 1 in 28,596 in the North East.

While newborn screening does not specifically test for beta thalassaemia major, F-only cases, which are probable beta thalassaemia affected babies, are identified as a by-product of screening for sickle cell disease. There are approximately 20 to 30 F-only cases reported each year, and in 2015/16 there were 27 F-only cases reported in England, and 30 for the whole of the UK.

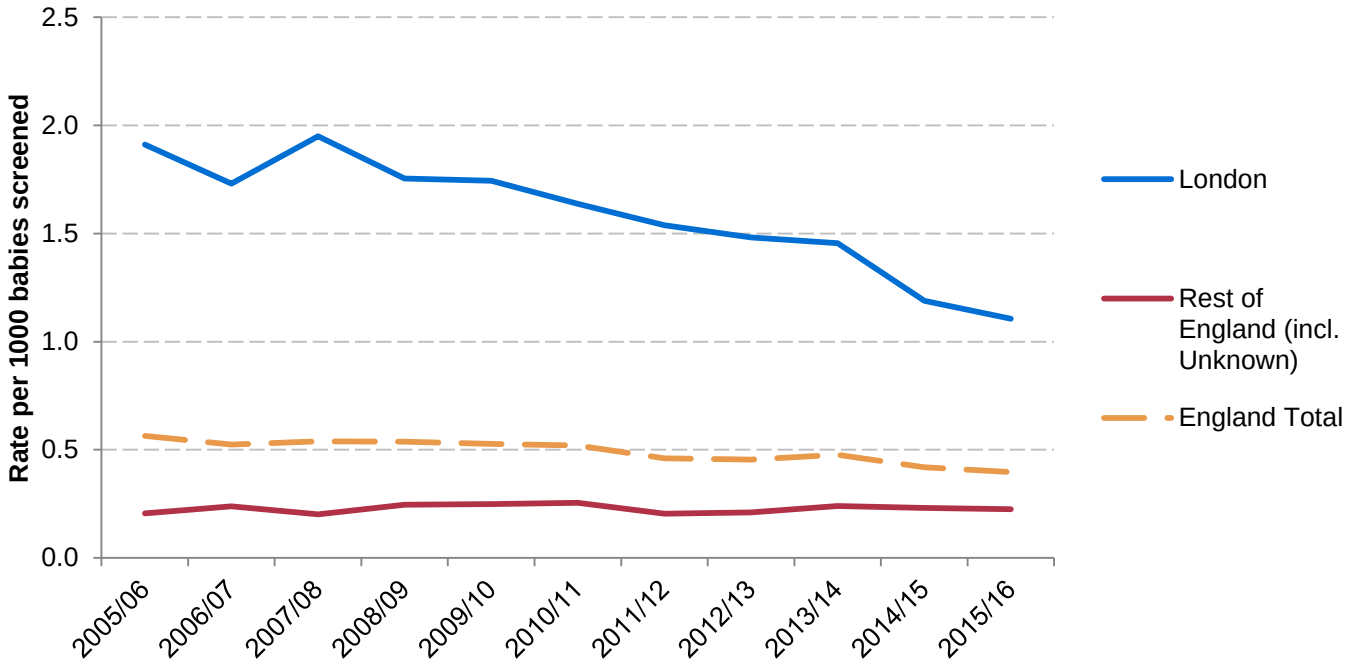
Table NB-6. Trends in significant conditions 2013/14 to 2015/16

Region	2013/14				2014/15				2015/16			
	n	Total screened	Rate/ 1000	1 in x	n	Total screened	Rate/ 1000	1 in x	n	Total screened	Rate/ 1000	1 in x
North	34	180,238	0.19	5,301	44	177,120	0.25	4,025	26	184,164	0.14	7,083
South	37	161,198	0.23	4,357	31	160,860	0.19	5,189	22	155,750	0.14	7,080
Midlands & East	57	188,454	0.30	3,306	44	188,003	0.23	4,273	67	190,634	0.35	2,845
London	190	130,534	1.46	687	155	130,388	1.19	841	144	130,150	1.11	904
Unknown region	1	8,365	0.12	8,365	4	6,046	0.66	1,512	6	7,102	0.84	1,184
England total	319	668,789	0.48	2,097	278	662,417	0.42	2,383	265	667,800	0.40	2,520
Scotland	-	-	-	-	-	-	-	-	3	55,616	0.05	18,539
Wales	-	-	-	-	-	-	-	-	4	33,181	0.12	8,295
Northern Ireland	-	-	-	-	-	-	-	-	0	24,569	0.00	-
UK total	-	-	-	-	-	-	-	-	272	781,166	0.35	2,872

Data for Scotland, Wales, and Northern Ireland only available from 2015/16.

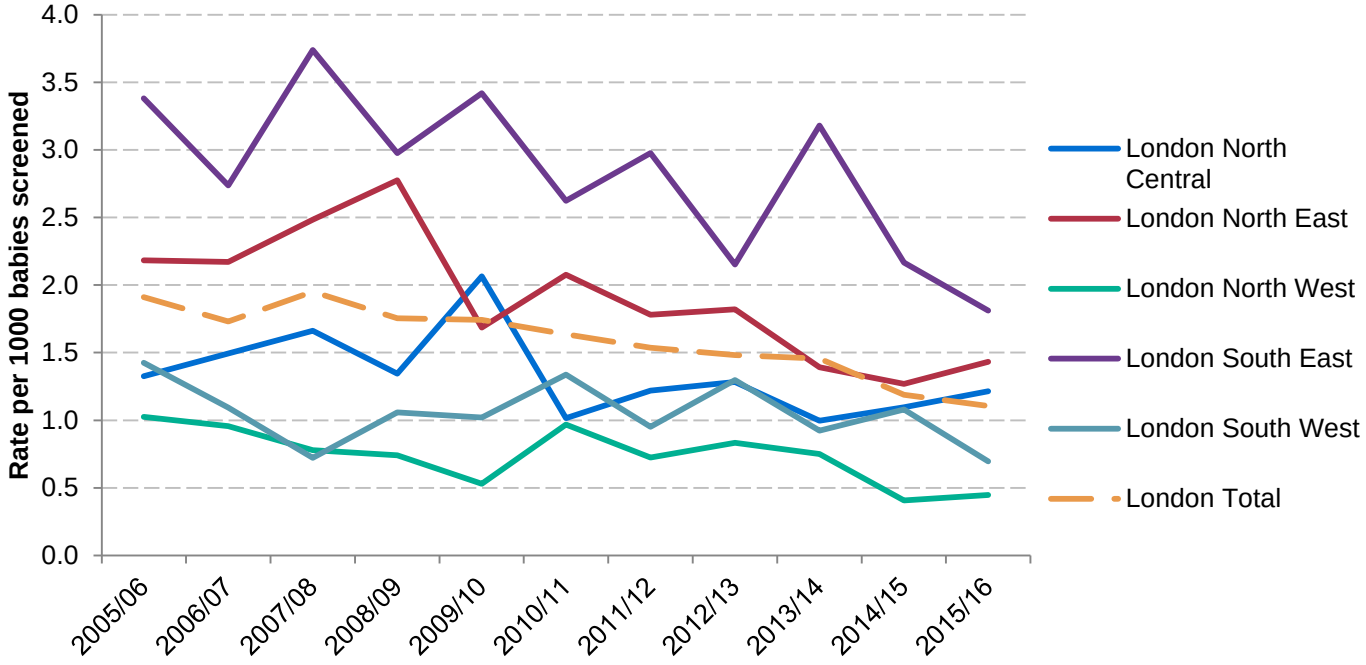
Figure NB-3 compares the rates of babies identified with a significant condition over time between London (where sickle cell disease is most prevalent) and the rest of England. While the rates in the rest of England appear relatively steady over the whole period, there appears to be a continued decline in rates in London each year. Figure NB-4 shows a breakdown of these rates over time by London sector (pre-2006 SHA).

Figure NB-3. Trends in rates of babies identified with a significant condition 2005/06 to 2015/16



Significant conditions comprise FS,FSC, FS Other and FE.

Figure NB-4. Trends in rates of babies identified with a significant condition 2005/06 to 2015/16: London sectors (pre-2006 SHAs)



Significant conditions comprise FS,FSC, FS Other and FE.

Carriers

Carrier results comprise FAS, FAC, FAD, FAE and other haemoglobin variants. The Wales newborn screening protocol is designed to detect only the disease states of SCD. However, those cases that are identified from the newborn screening process and subsequently determined to be carriers of SCD are referred for follow-up.

Table NB-7 shows the number and rates of carriers detected in England since newborn screening was introduced in 2005. Of the 7.3 million babies screened in this period, 101,468 were identified as carriers, or 1 in 72 babies screened.

Table NB-8 shows these figures for the last three years. There appears to have been a slight drop in carrier numbers and rates in 2015/16 compared to previous years, from 8,942 (1 in 74) in 2014/15 to 8,579 (1 in 78) in 2015/16. Broken down by sub-region, rates in 2015/16 ranged between 1 in 33 in London and 1 in 482 in the South West.

Table NB-7. Number and rates of carriers detected 2005/06 to 2015/16

Sub-region	Carriers			No. of babies Screened
	n	Rate/ 1000	1 in x	
East Midlands	4,497	8.19	122	549,395
East of England	7,536	10.16	98	741,933
London	51,262	36.29	28	1,412,758
North East	1,478	4.80	208	307,778
North West	7,954	8.38	119	949,670
South Central	5,123	9.86	101	519,825
South East Coast	4,069	7.13	140	570,444
South West	2,929	4.78	209	613,147
West Midlands	9,899	12.77	78	775,270
Yorkshire and the Humber	5,654	7.78	128	726,379
Unknown region	1,067	8.81	114	121,152
England Total	101,468	13.92	72	7,287,751

Table NB-8. Trends in carrier results 2013/14 to 2015/16

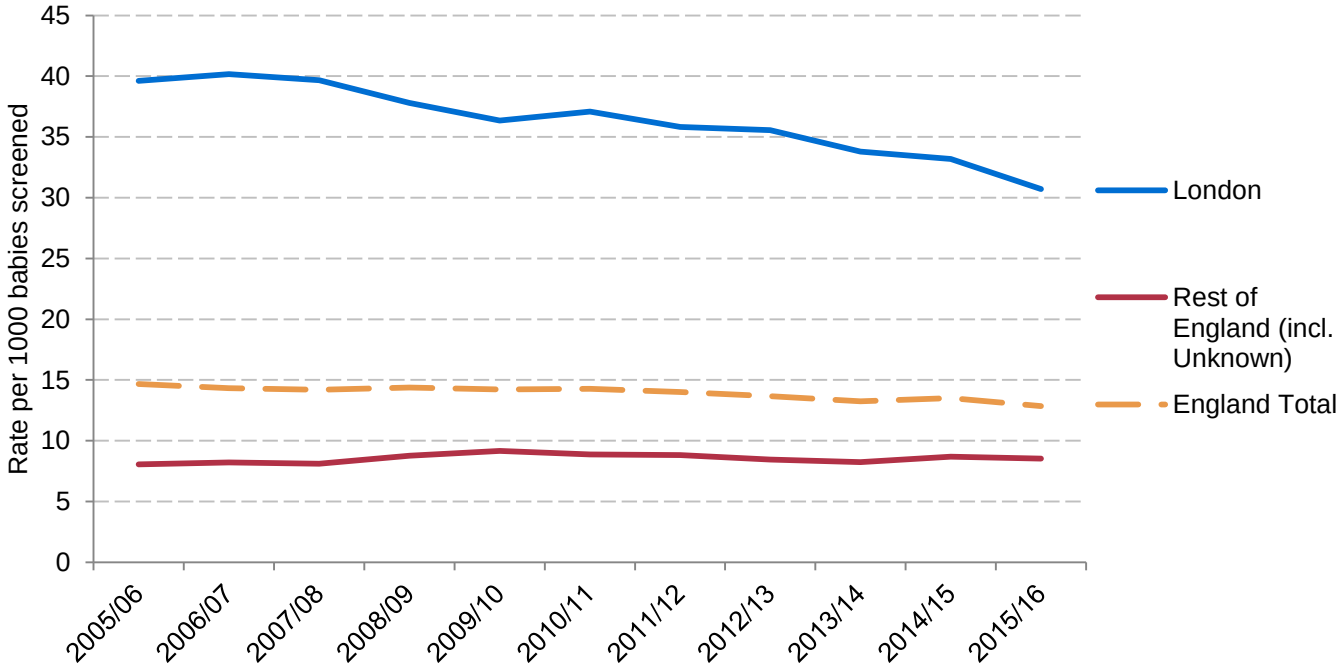
Sub-region	2013/14				2014/15				2015/16			
	n	Total screened	Rate/ 1000	1 in x	n	Total screened	Rate/ 1000	1 in x	n	Total screened	Rate/ 1000	1 in x
East Midlands	383	48,382	7.92	126	395	48,552	8.14	123	431	48,583	8.87	113
East of England	629	69,206	9.09	110	660	69,581	9.49	105	625	71,375	8.76	114
London	4,410	130,534	33.78	30	4,326	130,388	33.18	30	3,997	130,150	30.71	33
North East	134	27,914	4.80	208	115	26,387	4.36	229	169	28,596	5.91	169
North West	721	84,459	8.54	117	762	85,327	8.93	112	723	87,106	8.30	120
South Central	481	51,445	9.35	107	532	51,300	10.37	96	443	47,673	9.29	108
South East Coast	369	51,759	7.13	140	378	51,699	7.31	137	475	54,611	8.70	115
South West	305	57,994	5.26	190	305	57,861	5.27	190	111	53,466	2.08	482
West Midlands	881	70,866	12.43	80	926	69,870	13.25	75	901	70,676	12.75	78
Yorkshire and the Humber	450	67,865	6.63	151	482	65,406	7.37	136	470	68,462	6.87	146
Unknown	87	8,365	10.40	96	61	6,046	10.09	99	234	7,102	32.95	30
England Total	8,850	668,789	13.23	76	8,942	662,417	13.50	74	8,579	667,800	12.85	78
Scotland	-	-	-	-	-	-	-	-	216	55,616	3.88	257
Wales †	-	-	-	-	-	-	-	-	4	33,181	N/A	N/A
Northern Ireland	-	-	-	-	-	-	-	-	49	24,569	1.99	501
UK total	-	-	-	-	-	-	-	-	8,844	747,985	11.82	85

Data for Scotland, Wales, and Northern Ireland only available from 2015/16.

† The Wales newborn screening protocol is designed to detect only the disease states of Sickle Cell Disorder. However, any carriers identified from the screening process are referred for follow-up. Due to the different protocol followed the UK total does not include the carrier data from Wales.

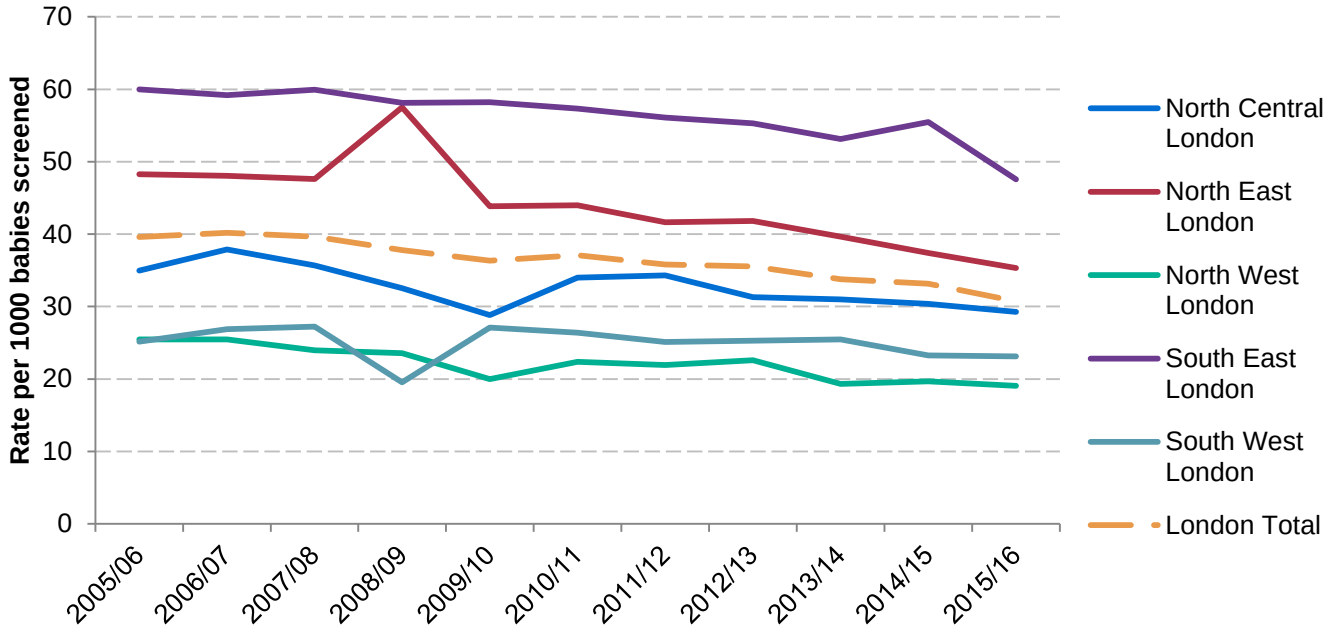
Figure NB-5 compares the rates of babies with carrier results over time between London and the rest of England. As with the rates detected with significant conditions, rates appear to be declining in London while they appear relatively steady in the rest of England. Figure NB-6 shows a breakdown of these rates over time by London sector (pre-2006 SHA).

Figure NB-5. Trends in rates of babies identified with a carrier result 2005/06 to 2015/16



Carrier results comprise FAS, FAC, FAD, FAE and other carriers.

Figure NB-6. Trends in rates of babies identified with carrier results 2005/06 to 2015/16: London sectors (pre-2006 SHAs)



Carrier results comprise FAS, FAC, FAD, FAE and other carriers.

4.4. Results by ethnicity

Newborn screening figures by ethnic category differ slightly from the figures shown by sub-region (see 4.1 'Response rates and data quality'). Table NB-9 shows the numbers and rates of babies detected with significant conditions and carrier result between 2005/06 and 2015/16. In this period, screen positive babies were reported for all ethnic categories except one (Chinese), and carrier results were reported for all ethnic categories. Table NB-10 shows these figures for 2015/16 only.

Figure NB-7 shows the proportion of screen positive babies reported for each ethnic category grouping between 2005/06 and 2015/16. Black ethnic categories accounted for approximately 78% of screen positive babies, but these conditions are not restricted to this group and 1.2% of affected babies were reported as White, approximately 5% as Asian, 5% as mixed or multiple ethnic groups, and 11% as other ethnic groups.

Table NB-9. Number and rates of babies detected with significant conditions and carrier results 2005/06 to 2015/16

Ethnic Category	Significant Conditions			Carriers			No. of babies Screened
	n	Rate/ 1000	1 in x	n	Rate/ 1000	1 in x	
White	43	0.01	113,233	9,351	1.92	521	4,869,032
Mixed	181	0.55	1,832	14,308	43.16	23	331,519
Asian	168	0.26	3,917	12,013	18.26	55	658,037
Black Caribbean	377	5.44	184	8,477	122.27	8	69,331
Black African	2,167	8.80	114	35,828	145.46	7	246,301
Any other Black background	157	5.08	197	3,246	105.12	10	30,878
Other*	374	0.51	1,971	12,971	17.60	57	737,093
England Total	3,467	0.50	2,002	96,194	13.86	72	6,942,191

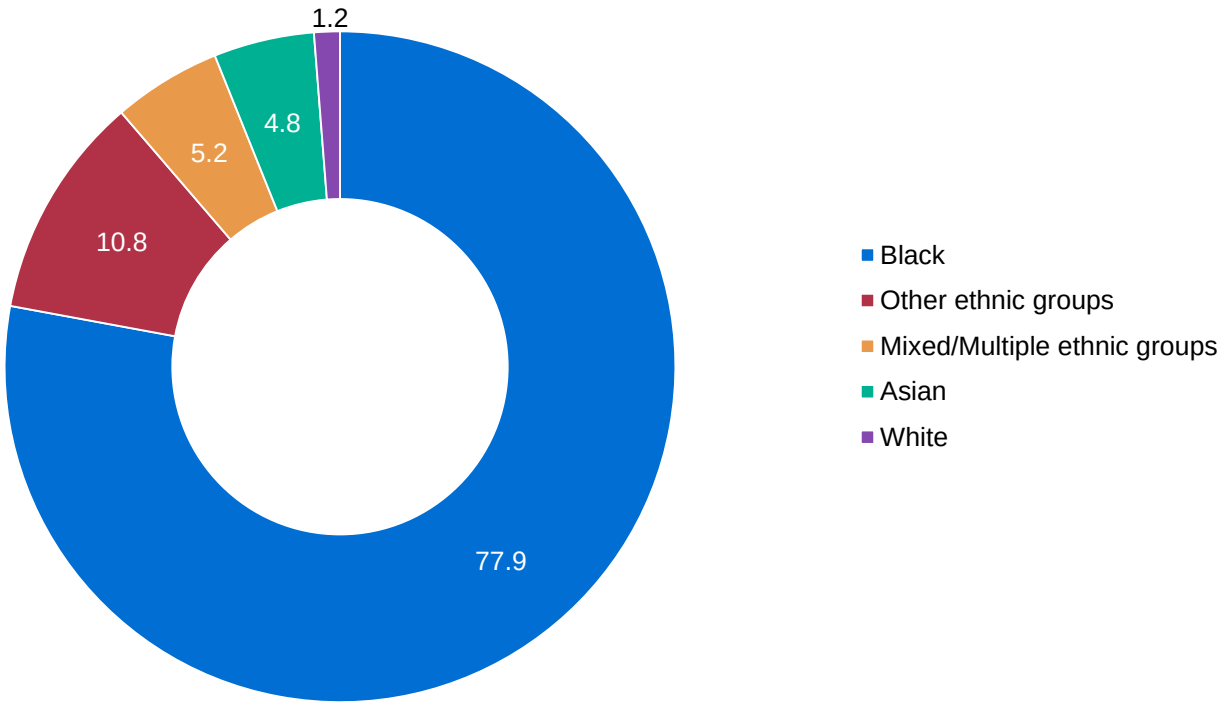
*'Other' includes the 'Chinese', 'Any other ethnic category', and 'Not stated' ethnic categories

Table NB-10. Number and rates of babies detected with significant conditions and carrier results 2015/16 only

Ethnic Category	Significant Conditions			Carriers			No. of babies Screened
	n	Rate/ 1000	1 in x	n	Rate/ 1000	1 in x	
White	4	0.01	119,412	738	1.55	647	477,647
Mixed	12	0.30	3,309	1,605	40.42	25	39,711
Asian	17	0.24	4,157	1,107	15.66	64	70,676
Black Caribbean	21	3.72	269	676	119.75	8	5,645
Black African	182	8.02	125	3,253	143.37	7	22,689
Any other Black background	13	4.05	247	333	103.71	10	3,211
Other*	17	0.32	3,090	878	16.71	60	52,530
England Total	266	0.40	2,527	8,590	12.78	78	672,109

*'Other' includes the 'Chinese', 'Any other ethnic category', and 'Not stated' ethnic categories

Figure NB-7. Percentage of all affected babies broken down by ethnic category 2005/06 to 2015/16

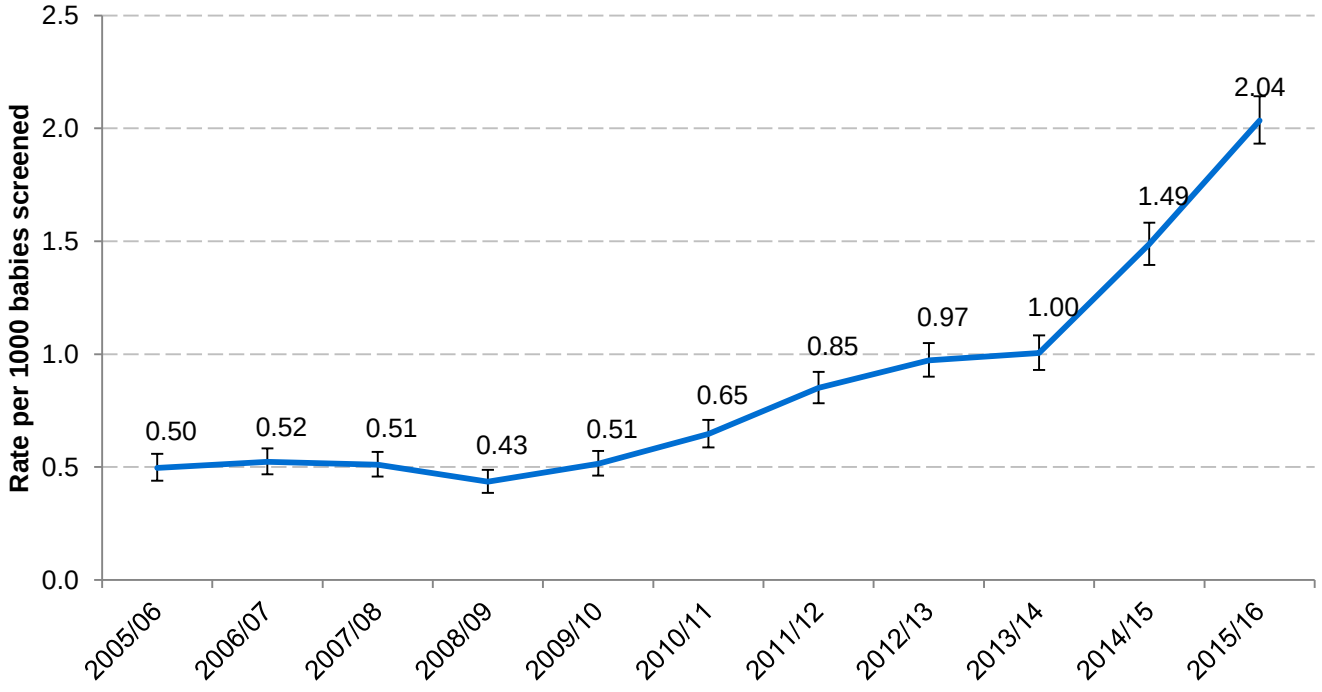


4.5. Declined screening tests

In 2015/16 there were 1,361 declined screening tests, which equates to approximately 2 per 1,000 babies screened. Figure NB-8 shows the rate of declined tests per 1,000 babies screened since 2005/06. There appears to be a continuation of the increase in the rate of declined tests, and the rate is now at 2.04 per 1,000 babies screened. It is difficult to identify the causes for this increase in declines as the reason for declining is not recorded, but some potential explanations include mover-in babies who may have been tested elsewhere and therefore have testing declined. It may relate to better reporting by laboratories of declines now that there is a laboratory sub-code for this. Or it may be that some of these declines are declined repeat samples rather than declined screening entirely.

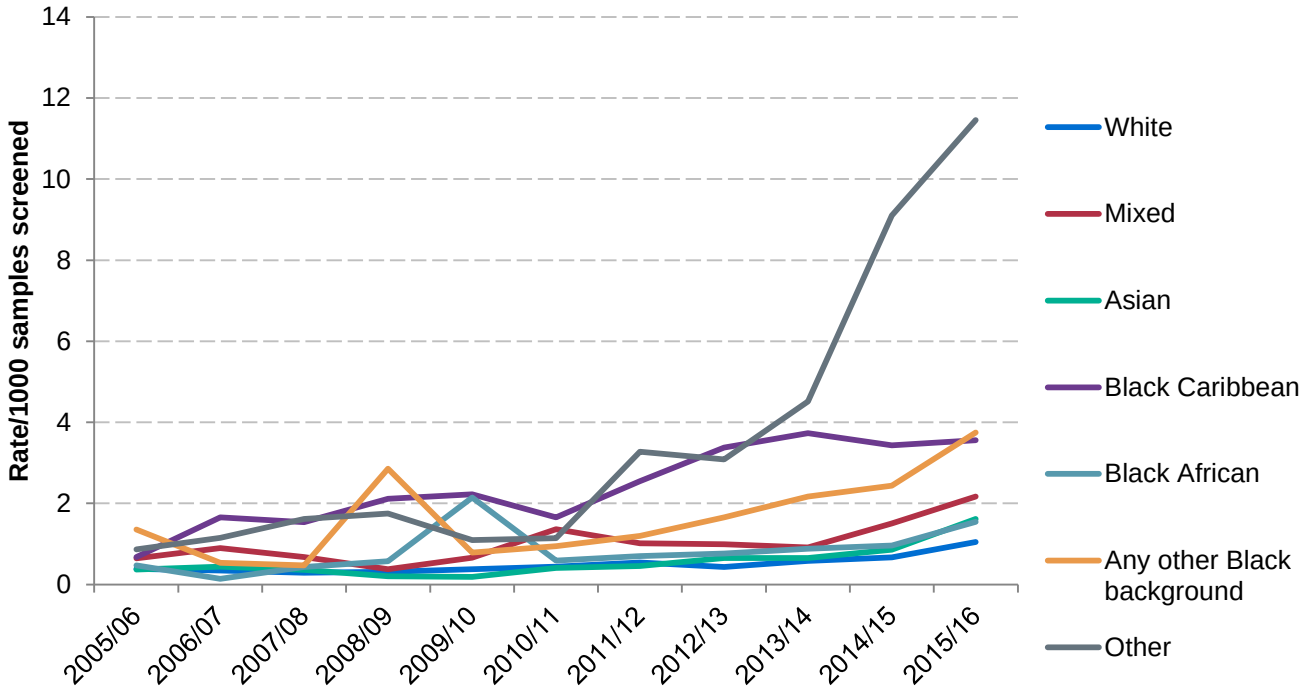
Figure NB-9 shows the decline rate per 1,000 babies screened broken down by ethnic category and Figure NB-10 shows these figures broken down by sub-region. Of the numbers that declined screening, 40% did not have ethnicity recorded, and 11% had an unknown sub-region.

Figure NB-8. Declined screening tests for sickle cell disease 2005/06 to 2015/16



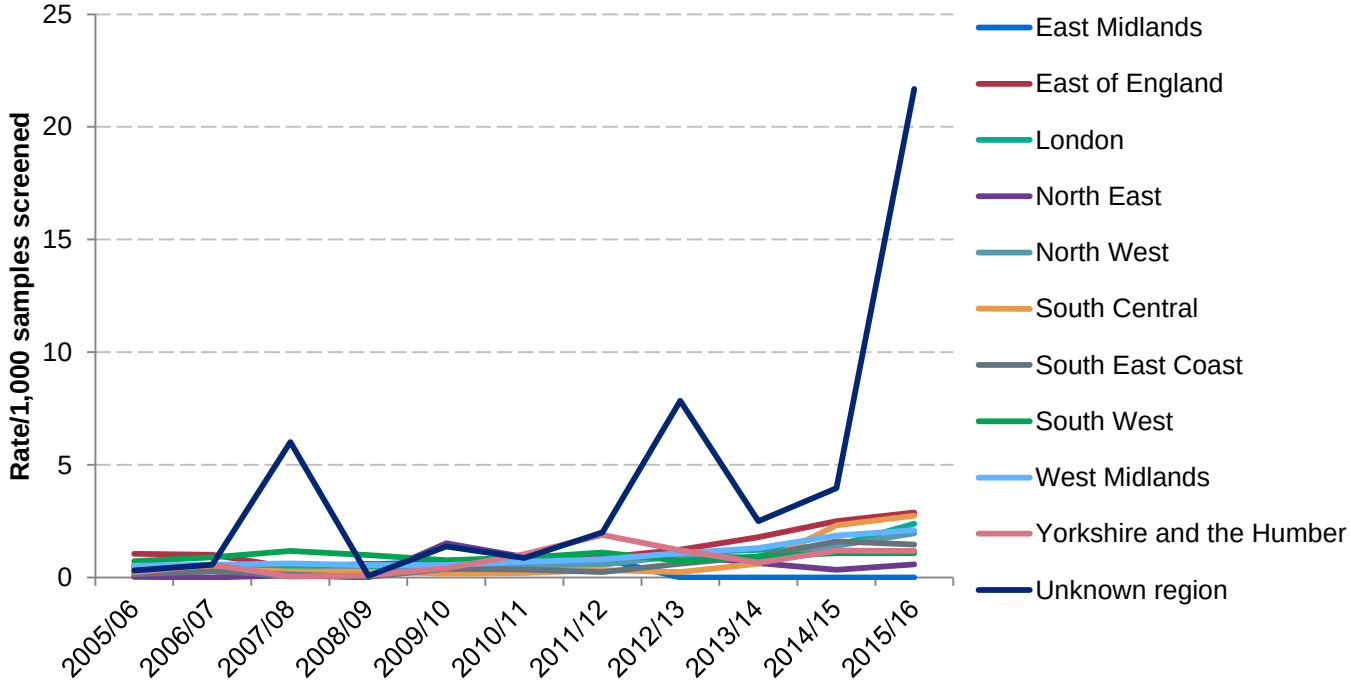
Bristol data for first half of 2005/06 not included and Oxford and Portsmouth data not included for whole of 2005/06; Oxford data starts from 1 July 2006.

Figure NB-9. Declined screening tests for sickle cell disease by ethnic category 2005/06 to 2015/16



Bristol data for first half of 2005/06 not included and Oxford and Portsmouth data not included for whole of 2005/06; Oxford data starts from 1 July 2006.
'Other' comprises 'Chinese', 'Any other ethnic category' and 'Not stated' ethnic categories.

Figure NB-10. Declined screening tests for sickle cell disease by sub-region 2005/06 to 2015/16



Bristol data for first half of 2005/06 not included and Oxford and Portsmouth data not included for whole of 2005/06; Oxford data starts from 1 July 2006.

4.6. Post-transfusion testing

Routine techniques are not suitable for testing samples from transfused babies as transfused red cells can survive up to 120 days in circulation. It is therefore important that pre-transfusion samples are taken in accordance with newborn blood spot sampling guidelines. The programme introduced a pre-transfusion sample policy in 2008 which requires that blood spots should be taken for SCD screening prior to blood transfusion[3].

Table NB-11 shows the number and rates of post-transfusion samples in the last three years. In 2015/16 there were 1,099 post-transfusion samples reported by the laboratories, or 1.65 per 1,000 samples screened. Rates vary between regions, ranging from 0.62 per 1,000 in the East of England to 2.11 in the West Midlands. The rate for babies with an unknown region was nine per 1,000 samples in 2015/16.

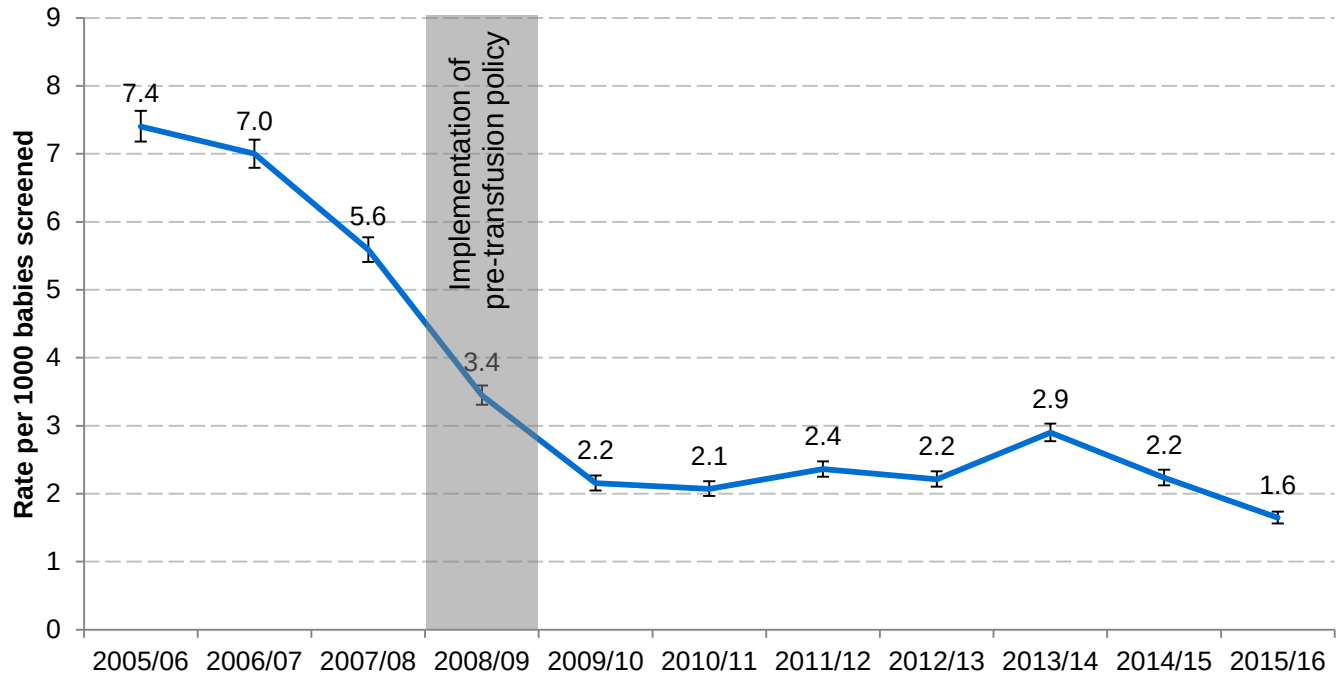
Figure NB-11 shows the change in rates over time between 2005/06 and 2015/16, showing the effect of the implementation of the pre-transfusion policy in 2008/09. Rates are at their lowest level in this period at 1.6 per 1,000 babies screened. The apparent increase in post-transfusion rates in the South East Coast sub-region has now declined and the rates now appear consistent with the other sub-regions.

Table NB-11. Number and rate of post-transfusion samples 2013/14 to 2015/16

Sub-region	2013/14*			2014/15			2015/16		
	n	Total screened	Rate/ 1,000	n	Total screened	Rate/ 1,000	n	Total screened	Rate/ 1,000
East Midlands	94	48,382	1.94	88	48,552	1.81	82	48,583	1.69
East of England	22	69,206	0.32	55	69,581	0.79	44	71,375	0.62
London	522	130,534	4.00	432	130,388	3.31	269	130,150	2.07
North East	36	27,914	1.29	40	26,387	1.52	37	28,596	1.29
North West	137	84,459	1.62	133	85,327	1.56	164	87,106	1.88
South Central	33	51,445	0.64	42	51,300	0.82	46	47,673	0.96
South East Coast	619	51,759	11.96	256	51,699	4.95	85	54,611	1.56
South West	30	57,994	0.52	35	57,861	0.60	35	53,466	0.65
West Midlands	115	70,866	1.62	104	69,870	1.49	149	70,676	2.11
Yorkshire and the Humber	96	67,865	1.41	149	65,406	2.28	124	68,462	1.81
Unknown	236	8,365	28.21	146	6,046	24.15	64	7,102	9.01
England total	1,940	668,789	2.90	1,480	662,417	2.23	1,099	667,800	1.65

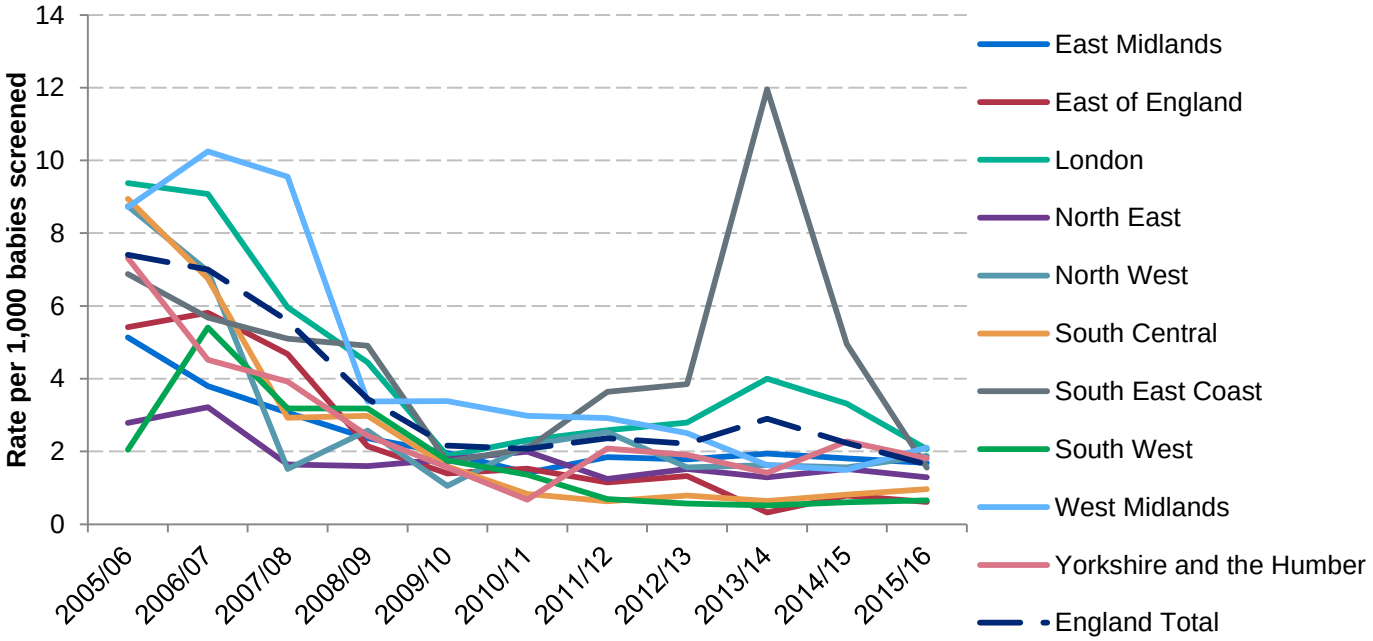
*Transfusion data for GOSH for 2013/14 not separated out from the 'normal+abnormal' figure and so not included here.

Figure NB-11. Rates of post-transfusion samples 2005/06 to 2015/16



Bristol data for first half of 2005/06 not included and Oxford and Portsmouth data not included for whole of 2005/06; Oxford data starts from 1 July 2006; Transfusion data from Manchester laboratory for 2009/10 not available; Transfusion data for GOSH for 2013/14 not separated out from the 'normal+abnormal' figure and so not included here.

Figure NB-12. Rates of post-transfusion samples by region 2005/06 to 2015/16



Bristol data for first half of 2005/06 not included and Oxford and Portsmouth data not included for whole of 2005/06; Oxford data starts from 1 July 2006; Transfusion data from Manchester laboratory for 2009/10 not available; Transfusion data for GOSH for 2013/14 not separated out from the 'normal+abnormal' figure and so not included here.

Where it is not possible to take a pre-transfusion sample, DNA testing is required to mitigate the risk of a missed baby[3]. DNA testing is provided by laboratories at King's College Hospital and Sheffield Children's Hospital, and the figures from these laboratories are shown in Table NB-12. Numbers of specimens reported by the DNA laboratories are different compared to the number of post-transfusion samples reported by the newborn screening laboratories (1,198 DNA tests compared to 1,099 post-transfusion samples reported by the screening laboratories in 2015/16).

Since DNA testing for transfused babies started there have been six positive homozygous cases identified through DNA testing. Table NB-13 shows the number of post-transfusion samples received by the DNA testing laboratories from newborn screening laboratories.

Table NB-12. Numbers detected through DNA testing for transfused babies 2010/11 to 2015/16

	2010/11	2011/12	2012/13	2013/14	2014/15	2015/16	Total
Total Specimens received per Quarter	1,674	1,520	1,343	1,160	1,123	1,198	7,313
Number of Negative results (HbS not detected)	1,650	1,497	1,319	1,140	1,106	1,183	7,195
Number of Positive Heterozygotes	24	21	21	20	16	15	112
Number of Positive Homozygotes	6 cases in 6-year period						

Table NB-13. Number of post-transfusion samples received broken down by newborn screening laboratory 2015/16

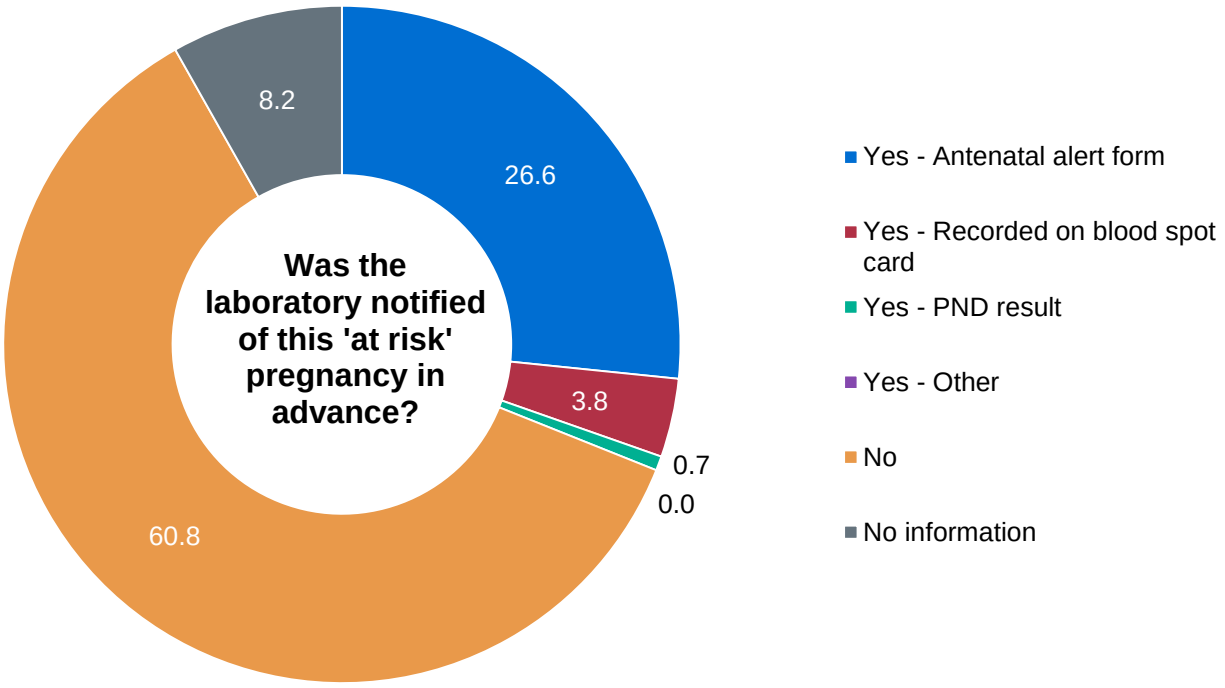
DNA testing laboratory	Newborn Laboratory	Number of samples
King's College Hospital	Bristol	50
	Cambridge	20
	GOS & CMH	162
	Oxford	38
	Portsmouth	64
	South East Thames	167
	South West Thames	83
Sheffield Children's Hospital	Leeds	72
	Liverpool	88
	Manchester	94
	Newcastle	44
	Sheffield	156
	West Midlands	160
England total		1,198

4.7. Laboratory processes and entry into care

Links between antenatal and newborn screening

Notification of at risk pregnancies to newborn laboratories provides a link between antenatal and newborn screening. Figure NB-13 shows the proportion of screen positive babies where the newborn laboratory was notified in advance, and how they were notified. Data completion has improved compared to last year when 32% of screen positive babies had no information provided. Of the 293 babies included in the timeliness data for England in 2015/16, laboratories had been notified in advance for 31% and not notified for 61%. Of the notifications that had been received, the majority were by antenatal alert form. This chart only shows data for England as the numbers for Scotland, Wales, and Northern Ireland are small.

Figure NB-13. Proportion of screen positive babies where the laboratory was notified in advance of newborn screening in England 2015/16



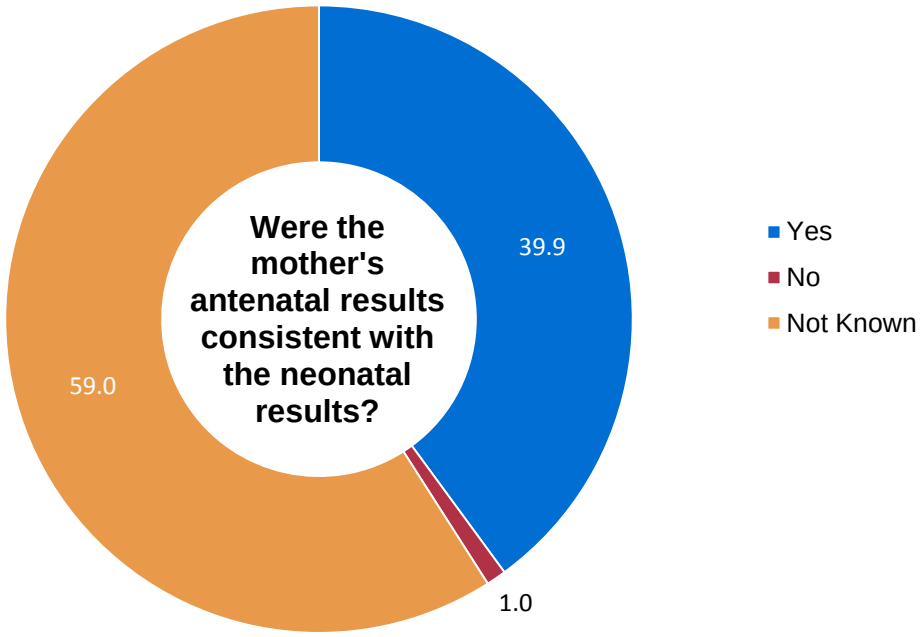
Another link between antenatal and newborn screening is through antenatal screening results being available to the newborn screening laboratory at the time of testing. Table NB-14 shows the numbers and proportions of screen positive babies for whom antenatal results were available at the time of testing. Only 16% were reported as having antenatal results available at the time of testing in 2015/16, but it should also be noted that no information was provided for 57% of screen positive babies. For comparison, last year only 20% had no information for this metric.

Table NB-14. Screen positive newborn babies for whom antenatal results were available at the time of testing 2015/16

Antenatal results recorded on blood spot card?	Yes		No		Not Known		Total
	n	%	n	%	n	%	
Mother's antenatal results recorded	46	15.7	81	27.6	166	56.7	293
Father's antenatal results recorded	46	15.7	81	27.6	166	56.7	293

Figure NB-14 shows the proportion of neonatal results that were consistent with the mother's antenatal result. Of the screen positive babies identified in 2015/16, 40% were reported to be consistent, but nearly 60% had no information provided by laboratories.

Figure NB-14. Consistency of antenatal and neonatal screening results 2015/16



Timeliness of clinical referral

Newborn Blood Spot (NBS) Screening Programme standard 4[4] is for the sample to be taken on day five and in exceptional circumstances between day five and day eight (day of birth is day zero). The thresholds for this standard are 95% as an acceptable level and 99% as an achievable level. Standard 5 (timely receipt of a sample in the newborn screening laboratory) requires 99% of samples to arrive in the laboratory within four working days of sample collection as acceptable, and within three working days as achievable.

SCT Screening Programme standard NP3[5] requires 90% of sickle cell disease results to be communicated to parents by four weeks of age. Table NB-15 shows the timeliness figures for newborn babies identified with a significant condition or with F-only results. Liverpool reported no screen positive babies in 2015/16, and Great Ormond Street and Central Middlesex hospitals did not provide data on age at clinical referral. The median age at time of clinical referral was 16 days, suggesting that programme standard NP4 (effective follow-up of infants with positive screening results) is both realistic and achievable.

Table NB-15. Timeliness of reporting affected newborn results, 2015/16

	No. of screen positives*	Sample ≤8 days		Clinical referral by 28 days†	
Laboratory	n	n	%	n	%
Bristol	2	1	50.0	2	100.0
Cambridge	7	7	100.0	6	85.7
GOS & CMH	112	106	94.6	No data provided	
Leeds	16	16	100.0	16	100.0
Liverpool	0	-	-	-	-
Manchester	15	14	93.3	15	100.0
Newcastle	1	1	100.0	1	100.0
Oxford	12	12	100.0	12	100.0
Portsmouth	6	6	100.0	6	100.0
Sheffield	18	16	88.9	15	83.3
South East Thames	51	48	94.1	49	96.1
South West Thames	23	22	95.7	22	95.7
West Midlands	30	30	100.0	30	100.0
England Total	293	279	95.2	174	59.4
Scotland	6	6	100.0	6	100.0
Wales	4	4	100.0	4	100.0
Northern Ireland	0	-	-	-	-
UK total	303	289	95.4	184	60.7

†Excludes 114 cases where data was missing or age at clinical referral was smaller than the age at sample.
*This includes F-only cases, which are likely beta thalassaemia affected babies

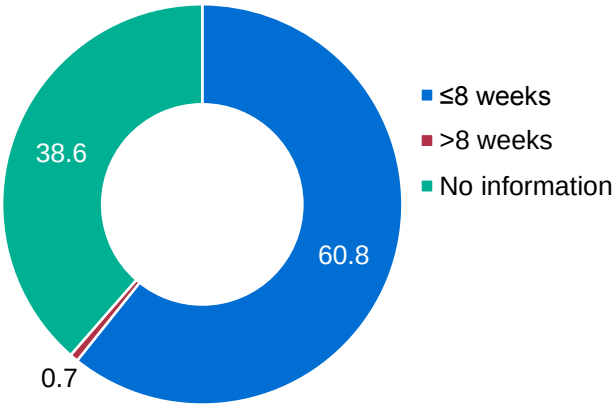
Age at first visit to specialist health team or local health team

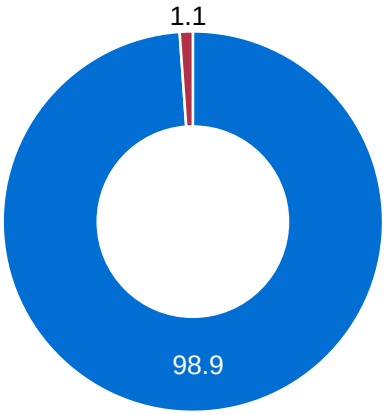
SCT programme standard NP4 is for babies identified with positive screening results to be referred by eight weeks to a designated healthcare professional and to attend a local clinic by three months of age. The thresholds for both timeframes are set at 90% for the acceptable level and 95% for the achievable level.

Figure NB-15 shows the proportion of screen positive babies with an initial referral to specialist services within eight weeks of age, both with and without any exclusions for 2015/16. This year, 113 babies (39%) had information missing for this field, compared to last year when there was only one baby with no information. With exclusions based on missing data, 99% of babies with information provided had their initial referral to specialist services by eight weeks of age.

Figure NB-16 shows the proportion of screen positive babies that had their first visit to a paediatrician at a specialist health team (SHT) or a local health team (LHT) in 2015/16. There were 202 babies (69%) for whom laboratories provided no information for this data field.

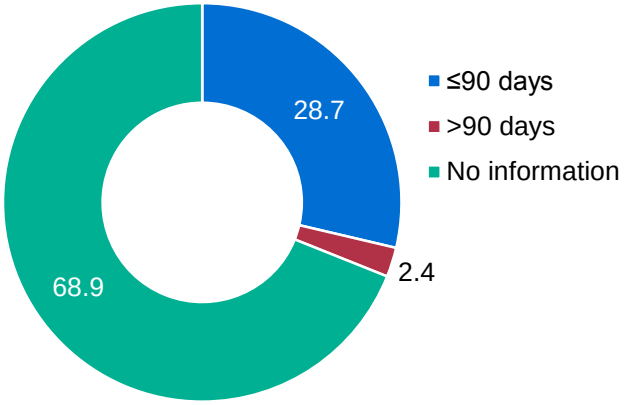
Figure NB-15. Age of screen positive babies at time of initial referral to specialist services (%) 2015/16

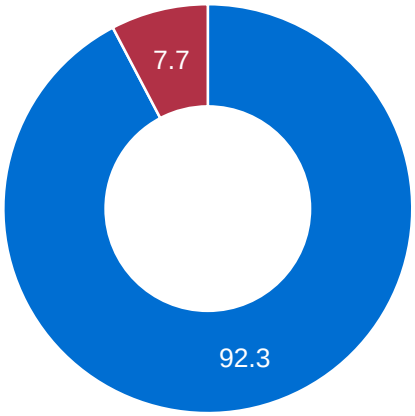




Excludes 113 babies for whom no information was submitted.

Figure NB-16. Age of screen positive babies at first visit to paediatrician at SHT or LHT (%) 2015/16



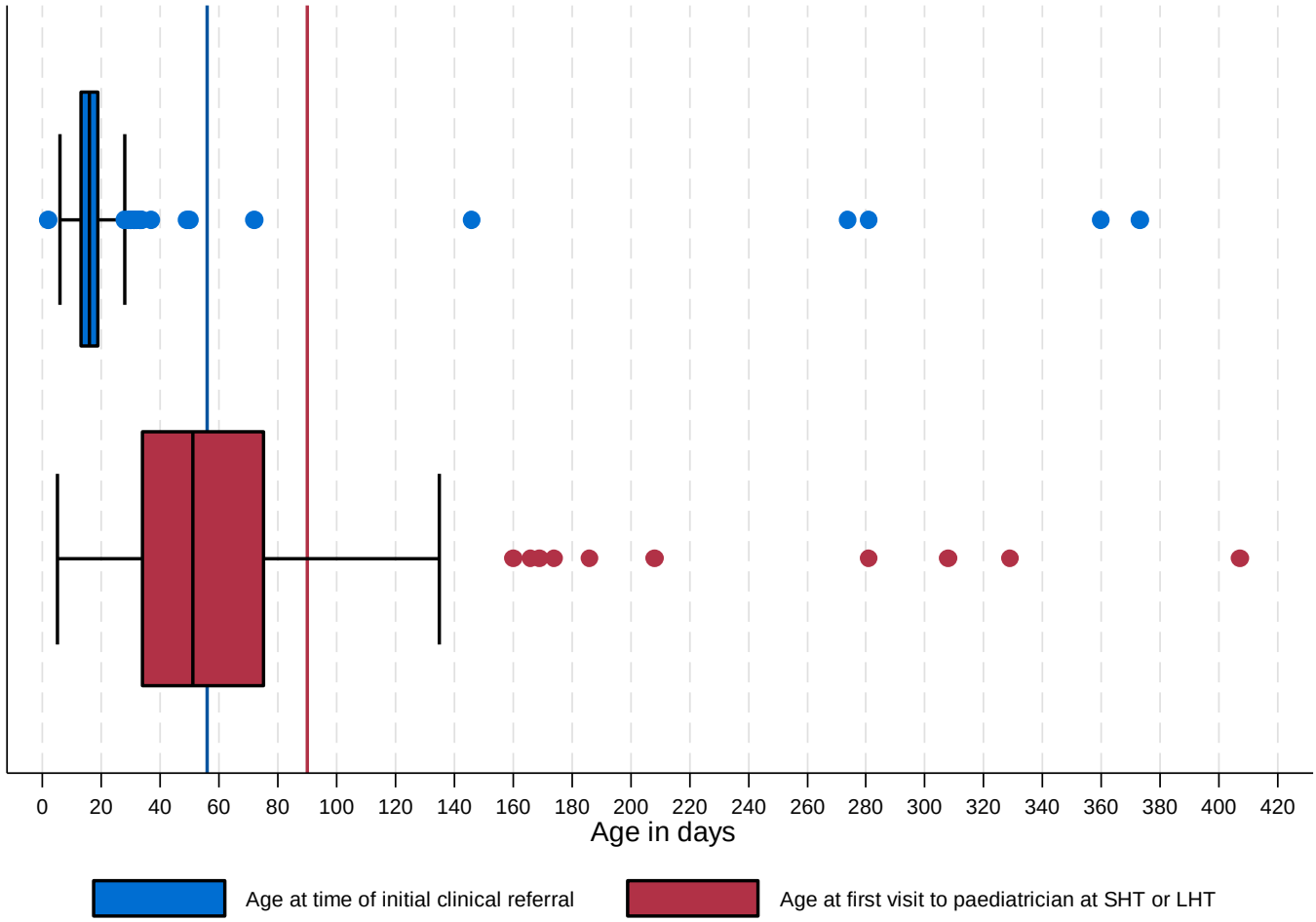


Excludes 202 babies for whom no information was submitted.

Figure NB-17 shows the variation in the age at time of initial clinical referral and at first visit to a paediatrician at a SHT or LHT in 2014/15 and 2015/16. The blue reference line represents the eight-week standard for initial clinical referral and the red reference line represents the three-month standard for first visit to a paediatrician (using 90 days to represent three months).

Given that there is a significant proportion of missing data for both fields for 2015/16 it is difficult to draw conclusions. However, based on the data reported and making exclusions where data was missing, in this two-year period only six babies were reported as having their initial referral to specialist services after 56 days (eight weeks). The median age for this standard was 16 days of age. In this same period, the majority of babies for whom data was provided had their first visit to a paediatrician by 90 days of age (three months). Of the 301 babies for whom data is available, 266 (88%) had their first visit by 90 days. The median age at first visit was 51 days.

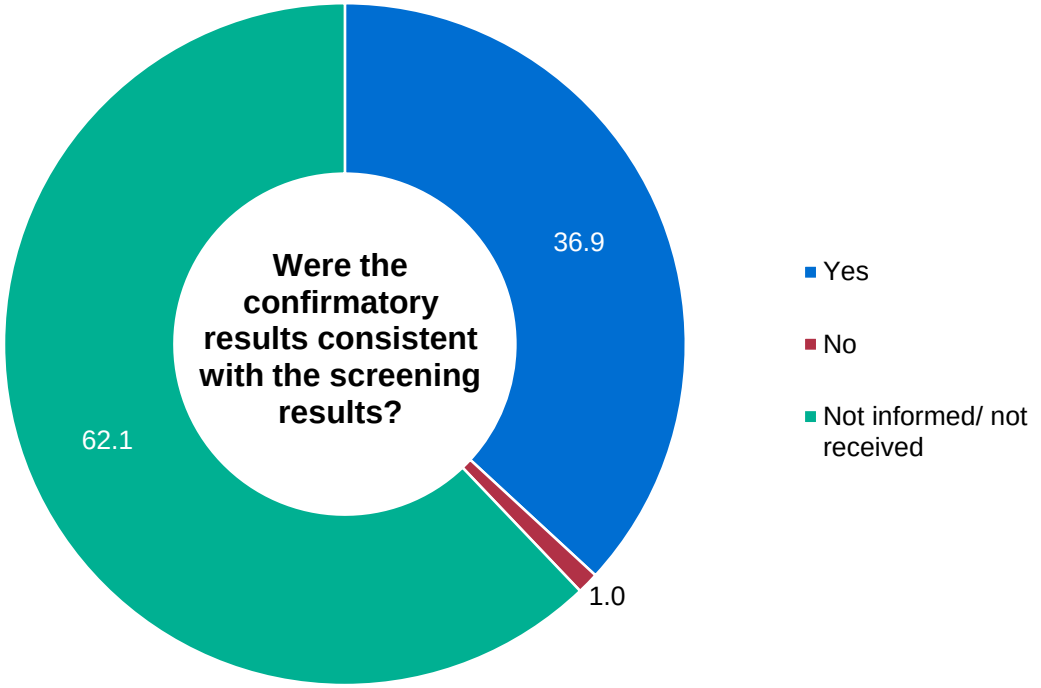
Figure NB-17. Variation in age at time of initial clinical referral and at first visit to paediatrician at SHT or LHT 2014/15 to 2015/16



Consistency of results

Newborn screening requires confirmatory testing, which involves taking and testing a second sample from the baby and comparing these results to the screening result. Figure NB-18 shows that 37% of screen positives had consistent confirmatory result, and that 62% had no information provided for this data field. There were three babies for whom confirmatory testing results were not consistent with screening results. Of these, two were reported by the laboratories as being subsequently found to be carriers rather than affected. The other case had no further information given.

Figure NB-18. Consistency of newborn and confirmatory results (%) 2015/16



Abbreviations

AN	Antenatal
CCG	Clinical commissioning group
CHRD	Child health record department
FOQ	Family Origin Questionnaire
Hb	Haemoglobin – see glossary for haemoglobin variants
HP	High prevalence
HPFH	Hereditary persistence of fetal haemoglobin
KPI	Key performance indicator
LP	Low prevalence
MCH	Mean cell haemoglobin
NAD	No abnormality detected
NB	Newborn
NBS	Newborn blood spot
NICE	National Institute for Health and Clinical Excellence
NIGB	National Information Governance Board
ONS	Office for National Statistics
PCT	Primary care trust
PHE	Public Health England
PKU	Phenylketonuria
PND	Prenatal diagnosis
SCD	Sickle cell disease
SCT	Sickle cell and thalassaemia
SHA	Strategic health authority
UK NSC	United Kingdom National Screening Committee

Glossary

Alpha plus thalassaemia (-α/αα or -α/-α)
This is found in all ethnic groups, with a high carrier frequency in populations in some parts of Africa, in the Caribbean and in South and Southeast Asia. Even if both partners are carriers, there is no risk to the fetus. Homozygous alpha plus thalassaemia is not a clinically significant disorder with respect to genetic or obstetric complications, but can cause diagnostic confusion with carriers of alpha zero thalassaemia or iron deficiency.

Alpha thalassaemia major, or Hb Barts hydrops fetalis (--/--)
A severe anaemia that affects the fetus. No normal fetal haemoglobin is produced and this leads to stillbirth or neonatal death.

Alpha zero thalassaemia (--/αα)
This carries the potential for a clinically significant disorder if both parents are carriers. If both parents are carriers of alpha zero thalassaemia, there is a risk of having a fetus with alpha thalassaemia major and the mother runs the risk of obstetric complications, particularly in the third trimester of pregnancy. The mutations are almost always due to a gene deletion. If one partner carries alpha zero thalassaemia and the other alpha plus thalassaemia, then there is a risk of having a child with Hb H disease. Prenatal diagnosis is not usually indicated for Hb H disease.

‘At-risk’ couples
Pregnancies identified with a potential risk of an affected baby, based on antenatal screening results for both parents. Cases where the father is not available for testing or where father results cannot be linked to mother results are also considered to be ‘at risk’ for an affected pregnancy. The number of ‘at-risk’ couples includes ‘high-risk’ couples (see below).

Beta thalassaemia major
A severe anaemia caused by inheritance of two beta thalassaemia genes, resulting in a lack of normal haemoglobin production. Treatment by regular blood transfusions and drugs to remove excess iron leads to long-term survival. Some affected children can be ‘cured’ by bone marrow transplantation.

Carrier (also referred to as trait)
An individual who carries a single altered gene where two altered genes are required for an individual to be affected with a condition that may require treatment. The carrier can pass on the gene to their offspring. The most common haemoglobin carrier states in the UK are Hb S, C, D, E and beta thalassaemia.

Family origins
A term used to describe a person’s ancestry.

Haemoglobin
The substance in our blood that carries oxygen around the body. Hb A is normal adult haemoglobin and Hb F is fetal haemoglobin.

Haemoglobin disease
Mild or serious diseases that can occur in people who have inherited two haemoglobin gene variants (see 'variant' below). These are also called haemoglobinopathies. The most common haemoglobin diseases screened for include:

- HbSS (sickle cell anaemia)
- HbSC disorder
- HBS/Beta thalassaemia
- Beta thalassaemia major
- E/beta thalassaemia

'High-risk' couples
Pregnancies that are identified as having a high risk of an affected baby. These are identified based on the combinations of mother and father antenatal test results which are considered to indicate a high risk of an affected baby (represented by the dark orange boxes on the antenatal data return, see Appendix Two).

Prevalence
The proportion of people in a population who have an attribute or a given disease.

Sickle cell disease
A group of inherited diseases that are characterised by sickling of red blood cells when there is a shortage of oxygen. The most common sickle cell diseases are sickle cell anaemia (Hb SS), haemoglobin SC disease, and haemoglobin S/beta thalassaemia. Sickle cell diseases can cause episodes of acute pain (crisis), anaemia, increased risk of infections, and chest problems. They can be life threatening, particularly for young children.

Thalassaemia major
A group of inherited conditions caused by a reduction in the amount of haemoglobin produced. People with a thalassaemia condition have various degrees of severe anaemia.

Variant
A change from the usual, for example, in a gene or protein. A variant haemoglobin gene may result in sickle or another type of haemoglobin in the body.

Haemoglobin variants include:

- Hb S – sickle haemoglobin
- Hb C – haemoglobin C
- Hb D – haemoglobin D

- Hb E – haemoglobin E

Examples of newborn screening results include FS (baby with fetal and sickle haemoglobins – probable sickle cell disease) and FAS (baby with fetal, adult, and sickle haemoglobins – probable sickle cell carrier).

List of charts and tables

Antenatal screening

Prenatal diagnostic testing

Newborn screening

Appendices

Appendix A: Update on the Newborn Outcomes Project: an evaluation of the linked antenatal and newborn screening programme

In September 2010, the NHS Sickle Cell and Thalassaemia Screening Programme started a project to assess the outcomes of the linked antenatal and newborn screening programme. We are collecting identifiable data on babies, or children under age five, with sickle cell disorders or beta thalassaemia.

This project will assess:

- the health of babies or children affected with sickle cell disease or thalassaemia
- timely entry to care and start of treatment of affected babies or children
- a review of the mother's antenatal screening history

The main rationale for the project is to reduce early mortality from invasive pneumococcal sepsis by ensuring that all affected babies with sickle cell disease are in clinical care and receiving the standard treatment. The programme currently has approval to collect named data without consent. We are collecting anonymised data alongside this data to assess its viability as a long-term exit strategy for the project, to provide assurance that all babies who need clinical care are receiving it. There is also an expectation that clinicians will enrol every newborn with sickle cell disease and clinically significant beta thalassaemia onto the National Haemoglobinopathy Registry if parents give consent, which may provide an alternative exit strategy.

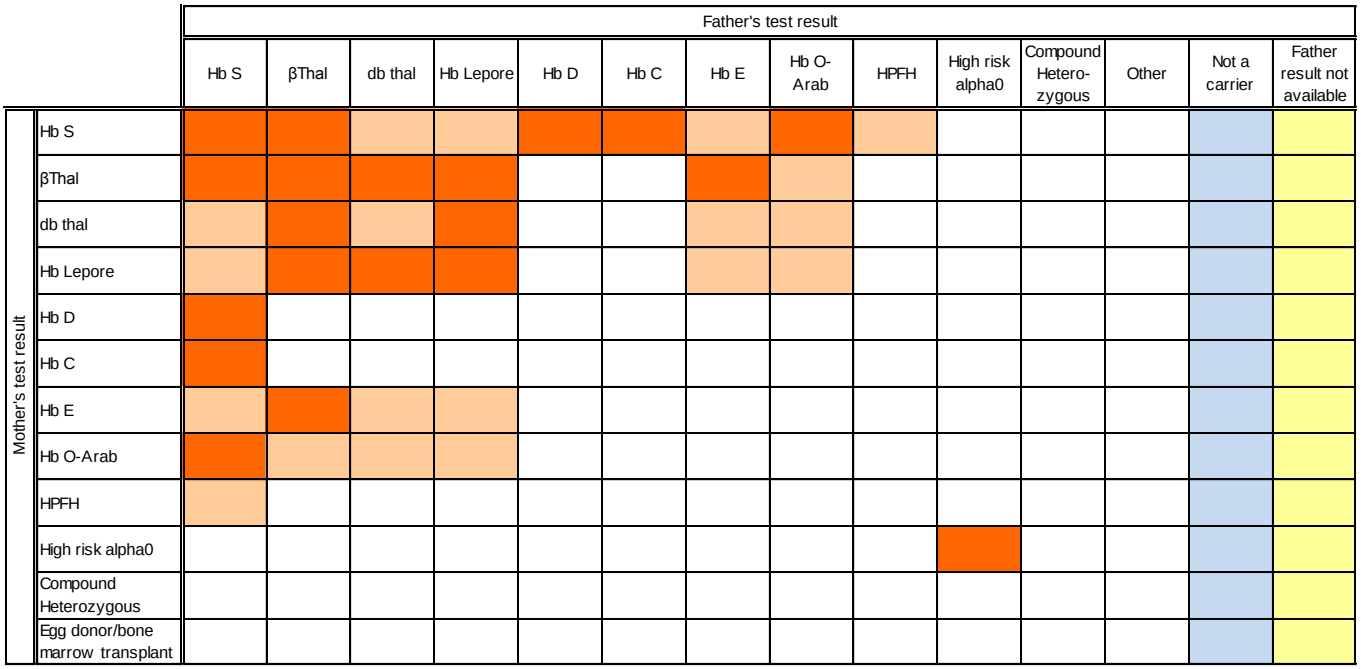
Between 1 April 2015 and 31 March 2016 there were 266 screen positive babies born in England. Of these:

- 229 babies had confirmed sickle cell disease of which 198 (86.5%) were seen in clinic by three months
- 34 babies had confirmed beta thalassaemia of which 26 (76.5%) were seen in clinic by three months
- three cases had an unconfirmed diagnosis

Babies were excluded from this cohort if they were presented with a clinically insignificant diagnosis, they had migrated or were born abroad, and any deaths which were not ascribed to sickle cell disease. Data collection has been a lengthy and protracted process. This has been in part due to manual processes and varying numbers of babies across the country (with the greatest number in London), and in part due to differing methods used for notification to specialist and community centres. The process needs to be updated and simplified to avoid duplication of requests.

In April 2017, responsibility for storing the data will transfer to the National Congenital Anomalies and Rare Disorders Registration Service (NCARDRS). For more information and to access the data collection form see the project pages on gov.uk.

Appendix B: Antenatal data return form part 2 – breakdown of screen positive women



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