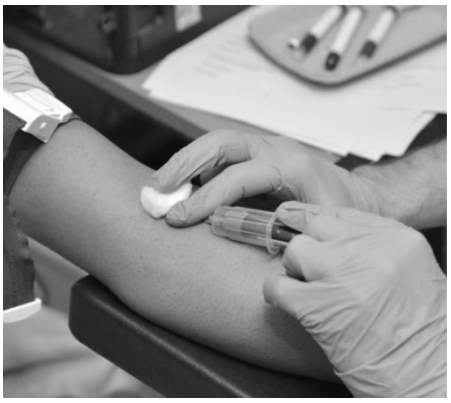
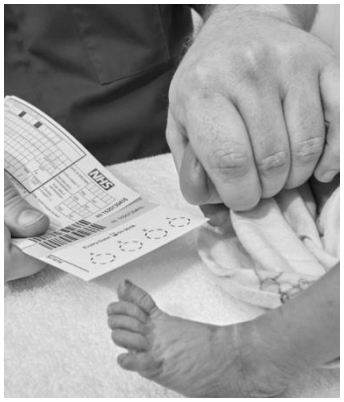
Public Health England

NHS

NHS Sickle Cell and Thalassaemia Screening Programme

Data report 2016 to 2017: 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-leading 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 Social Care, and a distinct delivery organisation with operational autonomy. We provide government, local government, the NHS, Parliament, industry and the public with evidence-based professional, scientific and delivery expertise and support.

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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.

www.gov.uk/topic/population-screening-programmes
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Executive summary

This report presents screening data for the NHS Sickle Cell and Thalassaemia Screening Programme for the financial year 1 April 2016 to 31 March 2017. This year the programme screened approximately 677,000 pregnant women for sickle cell and thalassaemia, and approximately 667,500 newborn babies for sickle cell disease.

Coverage remains high in both antenatal and newborn screening. Sub-regions are a sub-set of regions, allowing us to report data at a more granular level. Antenatal coverage was above the achievable level of 99% in all sub-regions except the North West, which was close at 98.8%. Coverage of newborn screening continues to improve and more Clinical Commissioning Groups (CCGs) were at or above the achievable level for key performance indicator (KPI) NB1. This year we have also started to report data on coverage for movers-in through KPI NB4.

Data for KPI ST2 shows that the acceptable level of pregnant women having a screening result available by 10 weeks and 0 days gestation is met nationally. The median performance for this KPI is above the acceptable level in all regions except in London, but there is wide variation in performance within both regions and sub-regions. While there was an improvement in the proportion of women tested by 10 weeks and 0 days, this doesn't appear to have had a positive impact on early prenatal diagnostic (PND) testing. The proportion of PND tests performed by 12 weeks and 6 days gestation remains at approximately 40%, compared to 50% in the year 2014 to 2015. This delay in screening can have a significant impact on people's lives, as shown through the recently published parents' stories. The programme is introducing a new KPI to measure the number of at risk women offered PND testing by 12 weeks and 0 days. This new KPI should identify whether there is a delay in the offer of PND testing, which may be causing PND testing to take place at a later gestation.

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

Just under 40% of newborn screen positives had no information provided for their age at first visit to a paediatrician, but of those that did have information approximately 90% were seen by 90 days of age. For comparison, data from the National Congenital Anomalies and Rare Disorders Registration Service (see Appendix A) indicates that 77.7% of affected babies were seen in clinic by 3 months of age.

1. Introduction

1.1. About the NHS Sickle Cell and Thalassaemia Screening Programme

Our aim 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 and Service Specification no.18: NHS Sickle Cell and Thalassaemia Screening Programme. Data is collected using spreadsheet-based data templates. The data is checked on receipt and, if required, the relevant laboratory is contacted for any clarifications that are required.

We request 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 and on FOQ completion is collected as key performance indicators (KPIs). The annual KPI data excludes providers where data has not been submitted for all four quarters in that year. The National Congenital Anomalies and Rare Disorders Registration Service (NCARDRS), collects patient-identifiable PND data from the laboratories, which can be matched to other data sources to improve the quality and completeness of the data reported.

While the screening programme covers only England, screening data is provided by the newborn laboratories in Scotland, Wales, and Northern Ireland. However, these countries are not included in the ethnicity figures, as Scotland uses different ethnic categories and Wales and Northern Ireland do not routinely collect ethnicity data. Data is compared for consistency and clarifications are sought if required.

Data is presented by financial year (1 April to 31 March) unless stated otherwise. The year ‘2016 to 2017’, for example, refers to the financial year ‘1 April 2016 to 31 March 2017’.

2. Response rates and data quality

Response rates

132 of the 141 expected data returns were received (93.6% response rate). The programme received data from all 4 prenatal diagnostic (PND) laboratories (100% response rate). The programme received data from all 13 newborn screening laboratories in England, and from the laboratories in Scotland, Wales, and Northern Ireland (100% response rate).

Data quality

Antenatal screening data

The data presented in this report represents the data provided by the antenatal screening laboratories or through key performance indicator (KPI) submissions. These figures may differ from those reported elsewhere.

Not all laboratories were able to submit data for all fields that were requested. Data returns were excluded where providers were unable to submit data so as to not bias reported rates which depend on aggregating these figures. Where exclusions were made, these are identified below the relevant charts and tables. Some laboratories are unable to match mother results to father results and so cannot provide the number of at risk couples. As a result, the reported number is likely to be an under-estimate of the true number of at risk couples.

Prenatal diagnostic (PND) testing data

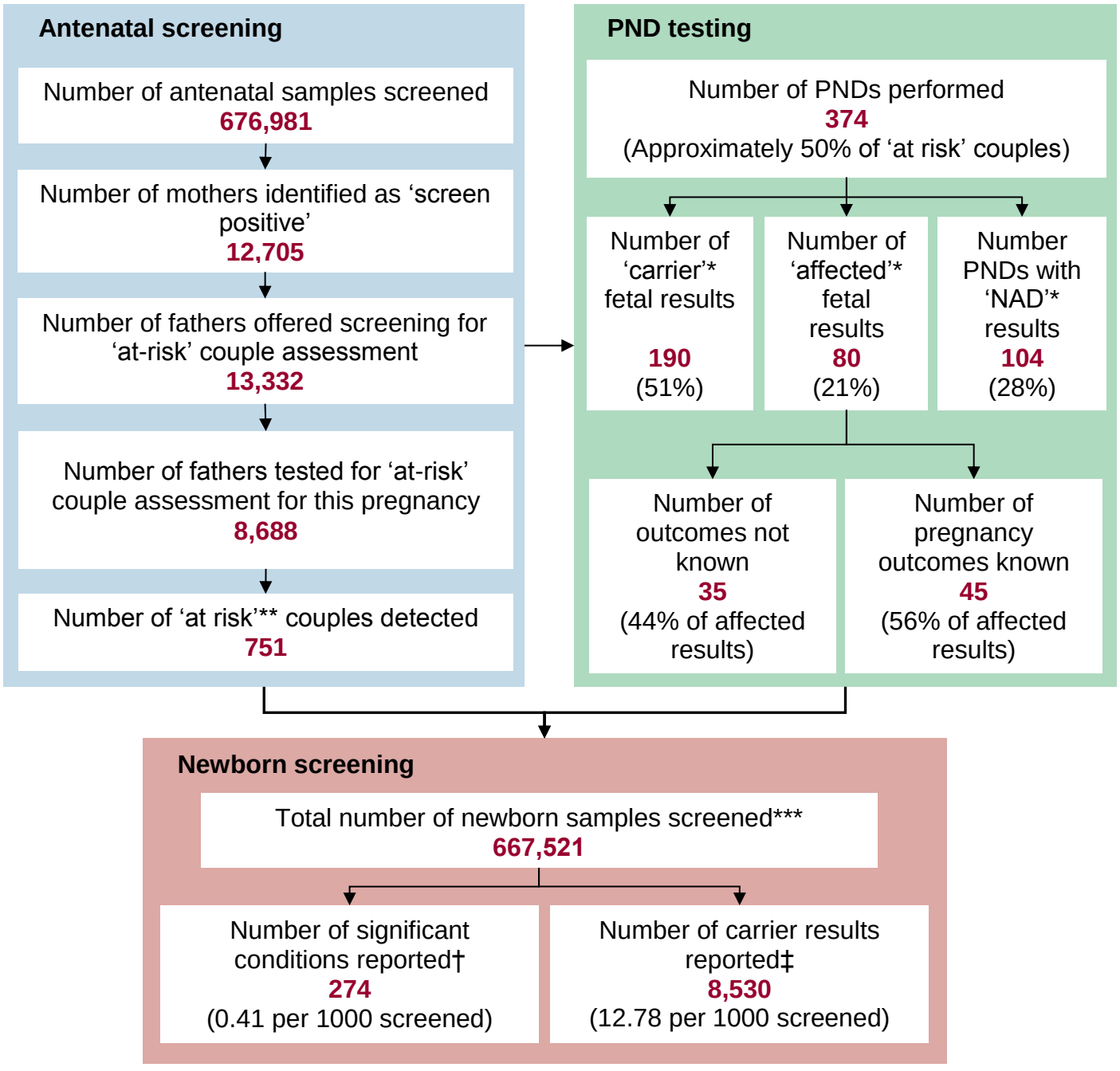
There remain gaps in the data, with 3 PND tests (0.8%) not having gestation at PND recorded and 35 (45%) affected result not having the pregnancy outcome recorded.

Newborn screening data

Newborn laboratories report on samples rather than babies tested. Data by region and by ethnicity are collected separately which can lead to discrepancies when comparing the figures.

The programme requests data on laboratory processes and timeliness of entry into care for screen positive babies. There were, however, some gaps in the numbers for age at receipt of sample in the laboratory and for age at first visit to a paediatrician at a specialist health team or local health team.

3. 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.
** 'At risk couples' comprises cases where there is a 1 in 4 chance of an affected fetus. This number excludes low-risk cases and cases where the father was not available for testing.
***Includes both tested and declines

4. Antenatal screening and PND testing for sickle cell and thalassaemia

4.1. Overview

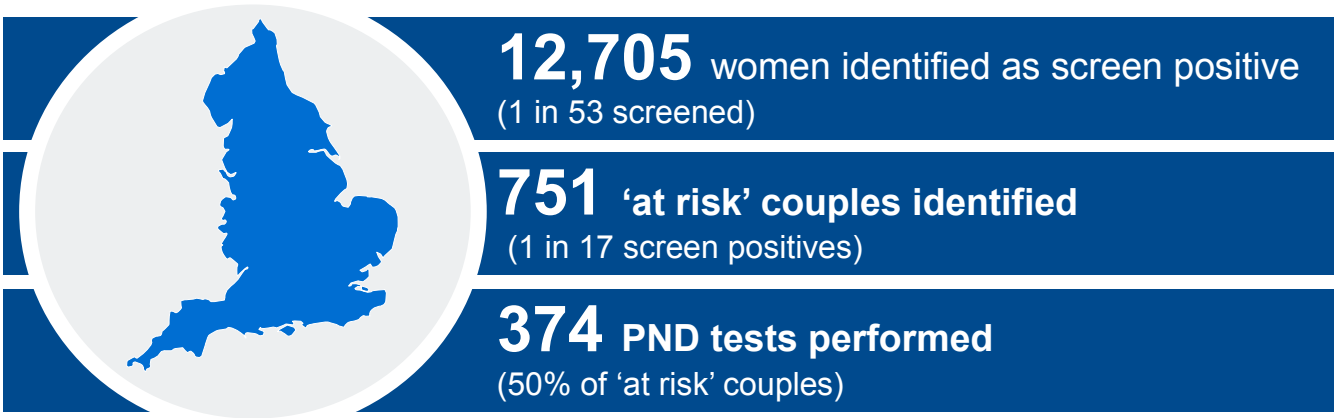
Antenatal coverage

99.3% **across the whole of England**

(range 94.5% – 100.0%)

FOQ completion

97.3% **of samples had a completed family origin questionnaire (FOQ) attached**



Father uptake

64.2% **of specimens requested from babies' biological father were received**

Antenatal declines

0.37% **of women booked decline screening**

Timeliness

53.1% of pregnant women tested by 10 weeks and 0 days gestation

37.5% of PND tests performed by 12 weeks and 6 days where gestation known

4.2. Antenatal coverage

Coverage is calculated as the number of women tested as a proportion of the number of women eligible for screening. KPI ST1 collects this data on a quarterly basis. Annual data is derived from the quarterly returns, but any provider that did not provide data in one or more quarter in that year is excluded. The thresholds for KPI ST1 set an acceptable level of 95% and an achievable level of 99%.

Table AN-1. Coverage of antenatal screening, 2016 to 2017: England

Sub-region	Completeness: Submitted all 4 quarters/ no.of providers	Eligible women	Tested women	Coverage (%)
East Midlands	7 / 9	47,746	47,422	99.3
East of England	16 / 18	70,242	69,589	99.1
London	25 / 25	154,264	153,896	99.8
North East	2 / 8	9,978	9,877	99.0
North West	15 / 22	77,007	76,057	98.8
South East	18 / 19	98,220	97,841	99.6
South West	17 / 17	60,092	59,473	99.0
West Midlands	12 / 14	58,093	57,578	99.1
Yorkshire & The Humber	9 / 13	47,984	47,783	99.6
England total	121 / 145	623,626	619,516	99.3

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

Figure AN-1. Trends in antenatal coverage, 2014 to 2015 – 2016 to 2017: England

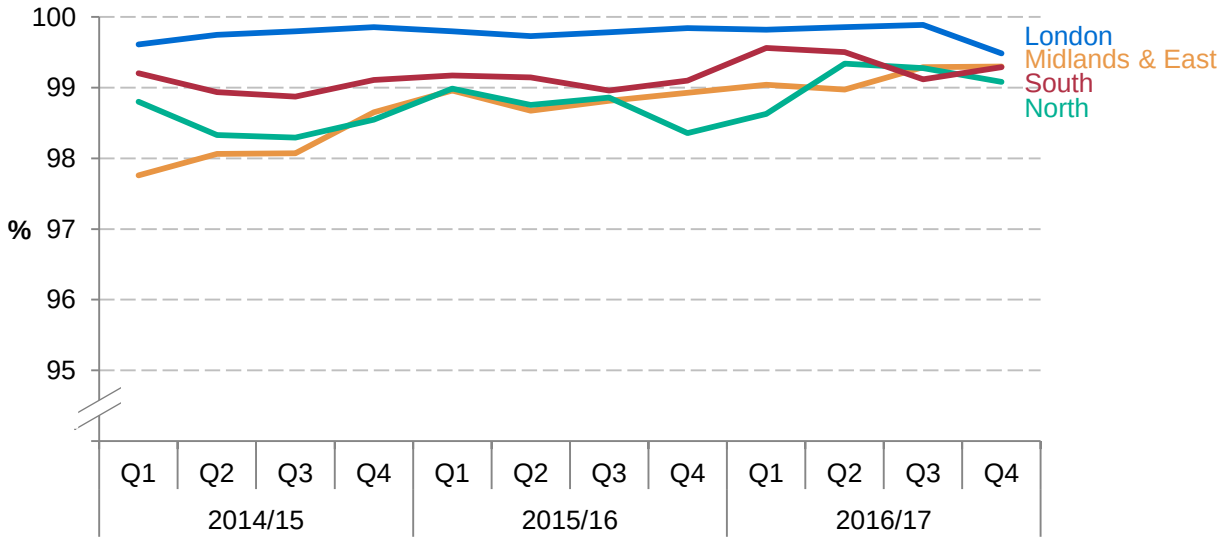
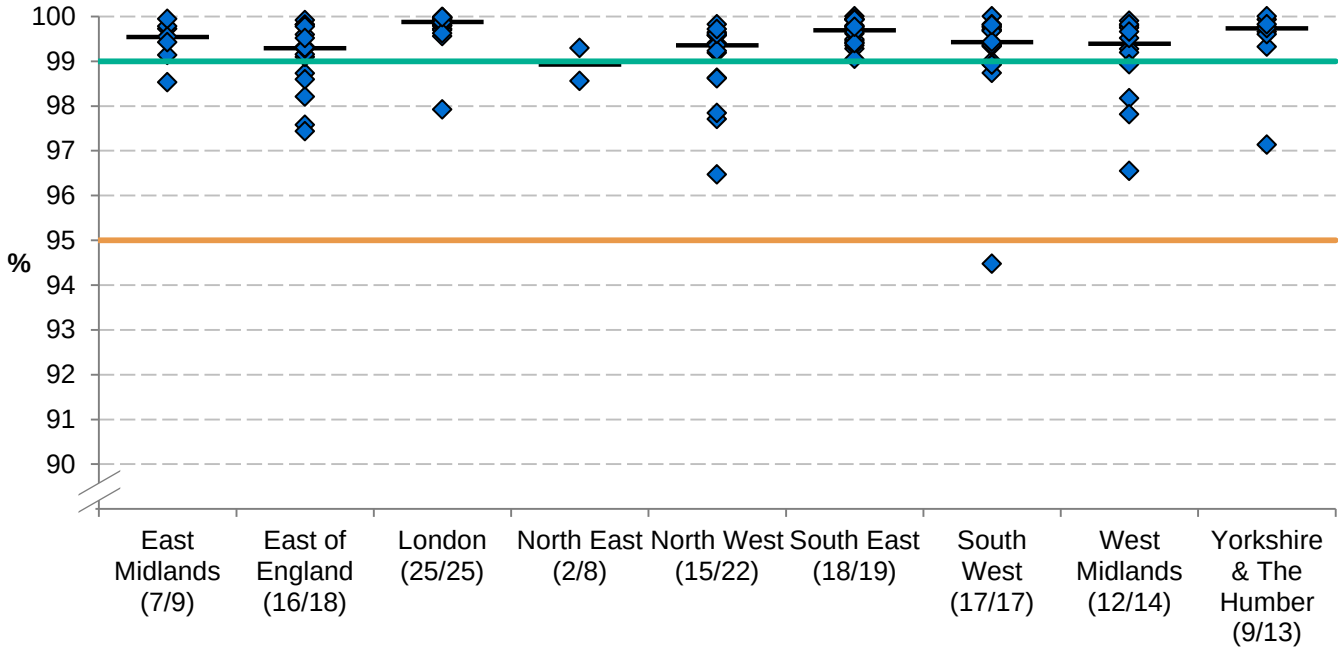


Figure AN-2. Variation in coverage of antenatal screening by sub-region, 2016/17: England by sub-region



The black horizontal markers represent the median value for each sub-region. The orange reference line represents the acceptable level for this KPI, and the green reference line represents the achievable level for this KPI. The numbers in brackets represent the number of providers that submitted data for this KPI out of the number of expected returns.

4.3. Numbers screened and detected in antenatal screening

In 2016 to 2017 there were 676,981 bookings reported by the screening laboratories, of which 12,705 were identified as screen positive (approximately 1 in 53 women screened). Of these, 751 pregnancies were identified as being at risk of an affected fetus (approximately 1 in 17 screen positive women) based on the results of both biological parents. We would expect the number of at risk couples to be approximately four times the number of newborn screen positive results (FS, FSC, FS-other, and FE results), plus 4 times the number of babies with an F-only results, plus terminations of affected pregnancies following PND testing. This gives an estimate of approximately 1,200 at risk couples. The lower number of at risk couples identified in the antenatal laboratory data may be due to couples where the baby's biological father's status is unknown, or where parents declined screening.

Table AN-2. Numbers screened and detected, 2016 to 2017: England

Sub-region	Returns received/ requested	Antenatal screening samples	Screen positive (Scr+)		At risk couples	
		n	n	% of samples	n	% of Scr+
East of England	15 / 16	74,022	1,036	1.40	55	5.31
East Midlands	9 / 9	56,544	865	1.53	48	5.55
London	22 / 24	130,425	5,201	3.99	327	6.29
North East	8 / 8	31,307	235	0.75	17	7.23
North West	16 / 19	78,598	1,125	1.43	65	5.78
South East	21 / 21	101,671	1,239	1.22	71	5.73
South West	17 / 17	62,157	464	0.75	24	5.17
West Midlands	12 / 14	73,859	1,687	2.28	98	5.81
Yorkshire and The Humber	12 / 13	68,398	853	1.25	46	5.39
Total England	132 / 141	676,981	12,705	1.88	751	5.91

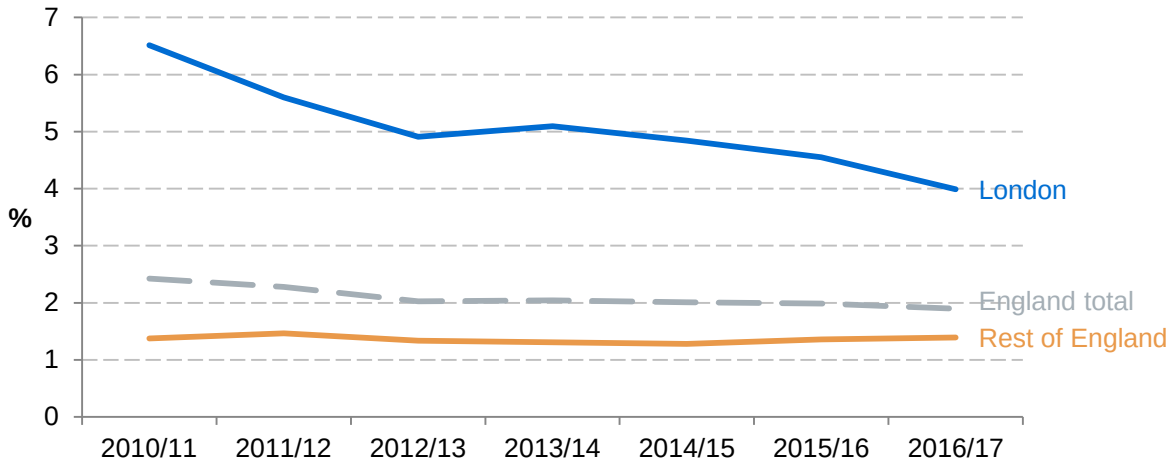
Table AN-3. Numbers screened and detected, 2016 to 2017: high prevalence areas

Sub-region	Returns received/ requested	Antenatal screening samples	Screen positive (Scr+)		At risk couples	
		n	n	% of samples	n	% of Scr+
East of England	4 / 5	22,431	565	2.52	38	6.73
East Midlands	5 / 5	35,565	700	1.97	39	5.57
London	22 / 24	130,425	5,201	3.99	327	6.29
North East	1 / 1	6,643	81	1.22	4	4.94
North West	6 / 7	45,045	922	2.05	58	6.29
South East	7 / 7	37,414	725	1.94	45	6.21
South West	2 / 2	11,217	153	1.36	6	3.92
West Midlands	7 / 7	50,412	1,499	2.97	85	5.67
Yorkshire and The Humber	5 / 5	32,224	548	1.70	36	6.57
Total England	59 / 63	371,376	10,394	2.80	638	6.14

Table AN-4. Numbers screened and detected, 2016 to 2017: low prevalence areas

Sub-region	Returns received/ requested	Antenatal screening samples	Screen positive (Scr+)		At risk couples	
		n	n	% of samples	n	% of Scr+
East of England	11 / 11	51,591	471	0.91	17	3.61
East Midlands	4 / 4	20,979	165	0.79	9	5.45
London	0 / 0	-	-	-	-	-
North East	7 / 7	24,664	154	0.62	13	8.44
North West	10 / 12	33,553	203	0.61	7	3.45
South East	14 / 14	64,257	514	0.80	26	5.06
South West	15 / 15	50,940	311	0.61	18	5.79
West Midlands	5 / 7	23,447	188	0.80	13	6.91
Yorkshire and The Humber	7 / 8	36,174	305	0.84	10	3.28
Total England	73 / 78	305,605	2,311	0.76	113	4.89

Figure AN-3. Screen positive women as a percentage of antenatal screening samples received by laboratory, 2010 to 2017



Exclusions based on missing or unavailable data: 2010/11: 2; 2011/12: 1; 2012/13: 1; 2013/14: 2; 2014/15: 1; 2015/16: 1; 2016/17: 1.

4.4. The family origin questionnaire

Data on the proportion of antenatal sickle cell and thalassaemia samples submitted to the laboratory accompanies by a completed family origin questionnaire (FOQ) is collected as a key performance indicator (ST3) on a quarterly basis. Annual data is derived from the quarterly returns, but exclusions are made for any provider that did not provide data in one or more quarter in that year. The thresholds for KPI ST3 set an acceptable level of 95% and an achievable level of 99%.

Table AN-5. Completion of FOQ by sub-region, 2016 to 2017: England

Sub-region	Completeness: Submitted all 4 quarters/ no.of providers	Antenatal screening samples	Samples with completed FOQ	% FOQ completion
East Midlands	9 / 9	55,078	56,059	98.3
East of England	17 / 18	74,708	77,104	96.9
London	25 / 25	147,707	153,120	96.5
North East	8 / 8	28,694	29,178	98.3
North West	20 / 22	87,698	90,841	96.5
South East	17 / 19	92,181	93,570	98.5
South West	17 / 17	60,057	61,574	97.5
West Midlands	14 / 14	77,900	80,325	97.0
Yorkshire & The Humber	13 / 13	68,515	70,024	97.8
England total	140 / 145	692,538	711,795	97.3

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

Figure AN-4. Trends in FOQ completion, 2014 to 2015 – 2016 to 2017: England

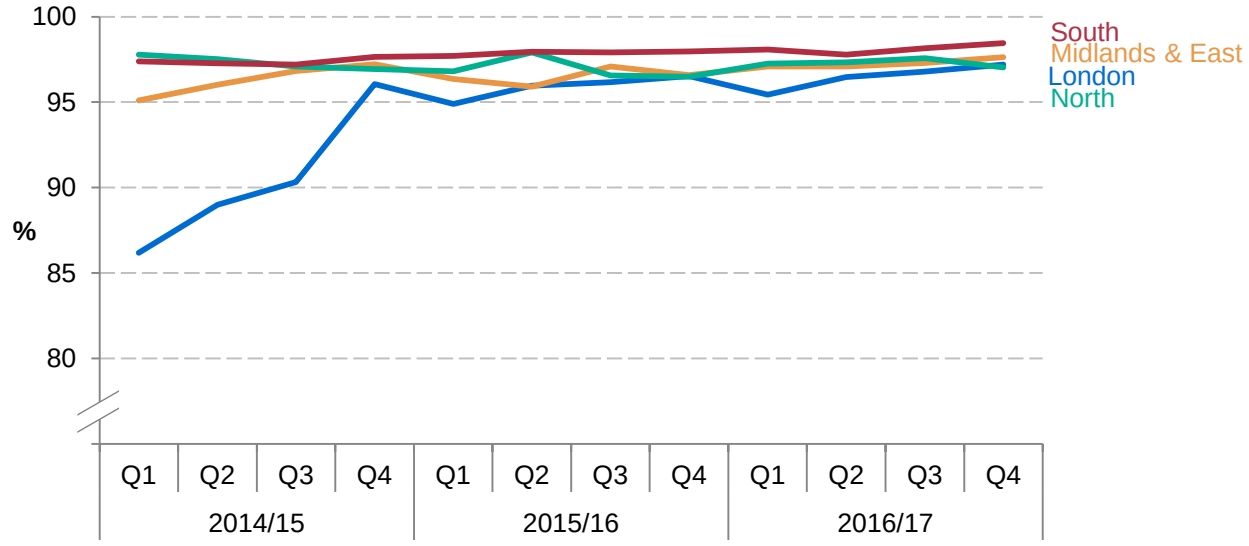
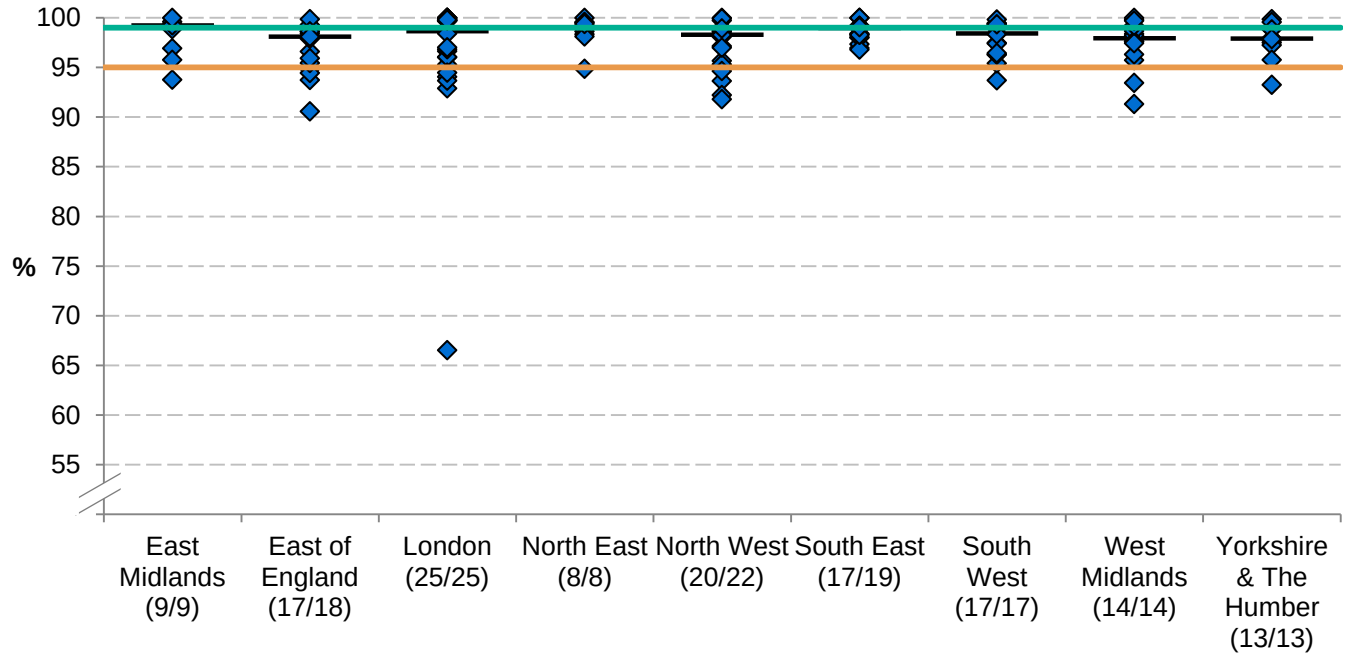


Figure AN-5. Variation in completion of FOQ by sub-region, 2016 to 2017: England



The black horizontal markers represent the median value for each sub-region. The orange reference line represents the acceptable level for this KPI, and the green reference line represents the achievable level for theis KPI. The numbers in brackets represent the number of providers that submitted data for this KPI out of the number of expected returns.

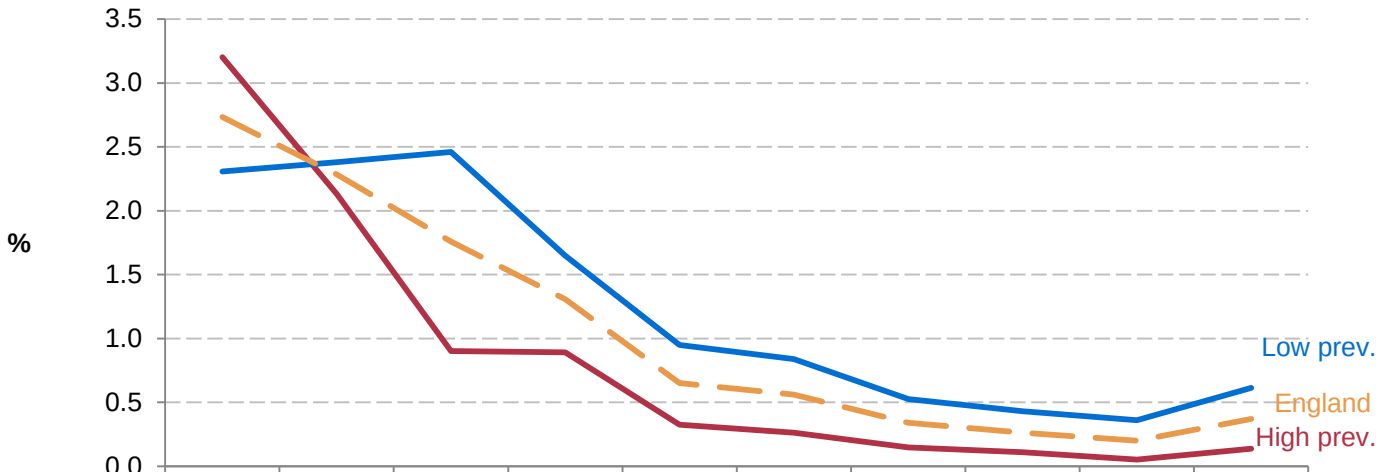
4.5. Declined screening tests for sickle cell and thalassaemia

Table AN-6. Declined screening by sub-region, 2014 to 2015 – 2016 to 2017: England

Sub-region	2014/15			2015/16			2016/17		
	Antenatal screening samples	Declines	% of samples	Antenatal screening samples	Declines	% of samples	Antenatal screening samples	Declines	% of samples
East Midlands	54,544	85	0.16	51,822	67	0.13	56,544	56	0.10
East of England	71,349	403	0.56	74,006	300	0.41	74,022	287	0.39
London	126,075	60	0.05	124,816	28	0.02	102,338	37	0.04
North East	31,907	140	0.44	31,512	57	0.18	31,307	66	0.21
North West	72,244	57	0.08	79,913	71	0.09	72,086	880	1.22
South East	100,551	156	0.16	97,350	90	0.09	89,847	49	0.05
South West	62,128	558	0.90	54,813	558	1.02	54,959	808	1.47
West Midlands	70,229	35	0.05	81,099	37	0.05	73,859	41	0.06
Yorkshire and The Humber	71,620	240	0.34	70,605	126	0.18	68,398	83	0.12
England Total	660,647	1,734	0.26	665,936	1,334	0.20	623,360	2,307	0.37

Exclusions based on missing or unavailable data: 2014/15: 12; 2015/16: 8; 2016/17: 9.

Figure AN-6. Trends in declined screening as a percentage of antenatal screening samples received, 2007 to 2008 – 2016 to 2017: England by prevalence



	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14	2014/15	2015/16	2016/17
High prev.	3.20	2.13	0.90	0.89	0.33	0.26	0.15	0.11	0.05	0.14
Low prev.	2.31	2.38	2.46	1.65	0.95	0.84	0.53	0.43	0.36	0.61
England	2.73	2.28	1.76	1.31	0.65	0.56	0.34	0.26	0.20	0.37

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; 2016/17: 9.

4.6. Testing of the baby's biological father

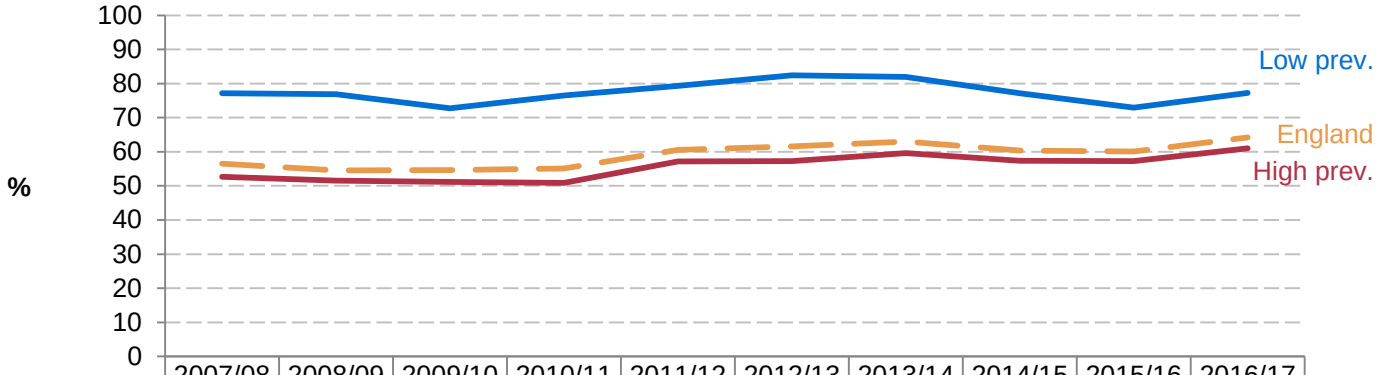
If a woman is screened positive, the baby's biological father should be offered testing to determine the risk to the pregnancy. If the baby's biological father is not available for testing, it is not possible to accurately assess the risk status of the pregnancy and the screen positive woman should be offered prenatal diagnostic (PND) testing as if they were at risk. It is estimated that this group of women accounts for approximately 36% of screen positive women (calculated from the number of screen positive women minus the number of father specimens received).

Table AN-7. Uptake of testing of the baby's biological father, 2014 to 2015 – 2016 to 2017

Sub-region	2014/15			2015/16			2016/17		
	Fathers requested	Fathers received	% uptake	Fathers requested	Fathers received	% uptake	Fathers requested	Fathers received	% uptake
East Midlands	718	504	70.19	850	596	70.12	900	697	77.44
East of England	1,016	734	72.24	1,171	760	64.90	1,102	693	62.89
London	7,225	3,646	50.46	6,340	3,205	50.55	5,291	3,044	57.53
North East	242	194	80.17	258	191	74.03	242	192	79.34
North West	1,082	719	66.45	1,137	735	64.64	1,174	758	64.57
South East	1,356	957	70.58	1,321	938	71.01	1,354	904	66.77
South West	489	366	74.85	603	391	64.84	512	448	87.50
West Midlands	1,591	1,004	63.10	1,693	1,066	62.97	1,745	1,104	63.27
Yorkshire and The Humber	829	665	80.22	938	713	76.01	936	673	71.90
England total	14,548	8,789	60.41	14,311	8,595	60.06	13,256	8,513	64.22

Exclusions based on missing or unavailable data, the proportion was greater than 100%, or there were no screen positive cases: 2014/15: 8; 2015/16: 2; 2016/17: 2.

Figure AN-7. Trends in uptake of testing of the baby's biological father, 2007 to 2008 – 2016 to 2017: England by prevalence



	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14	2014/15	2015/16	2016/17
High prev.	52.7	51.5	51.1	50.9	57.1	57.3	59.6	57.4	57.2	61.1
Low prev.	77.2	76.9	72.8	76.5	79.3	82.4	82.0	77.2	73.0	77.3
England	56.5	54.6	54.6	55.1	60.5	61.6	63.0	60.4	60.1	64.2

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; 2016/17: 2.

‘At risk’ couples are identified based on the results of both of the baby’s biological parents. 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 was not available for testing or the laboratory is unable to link their results to the mother’s results.

‘High risk’ pregnancies are those represented by the dark orange boxes in the breakdown table in Appendix A. The light orange boxes represent low risk pregnancies, and the white boxes represent minimal risk pregnancies. 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.

Table AN-8. Breakdown of pregnancy risk for screen positive women, 2016 to 2017

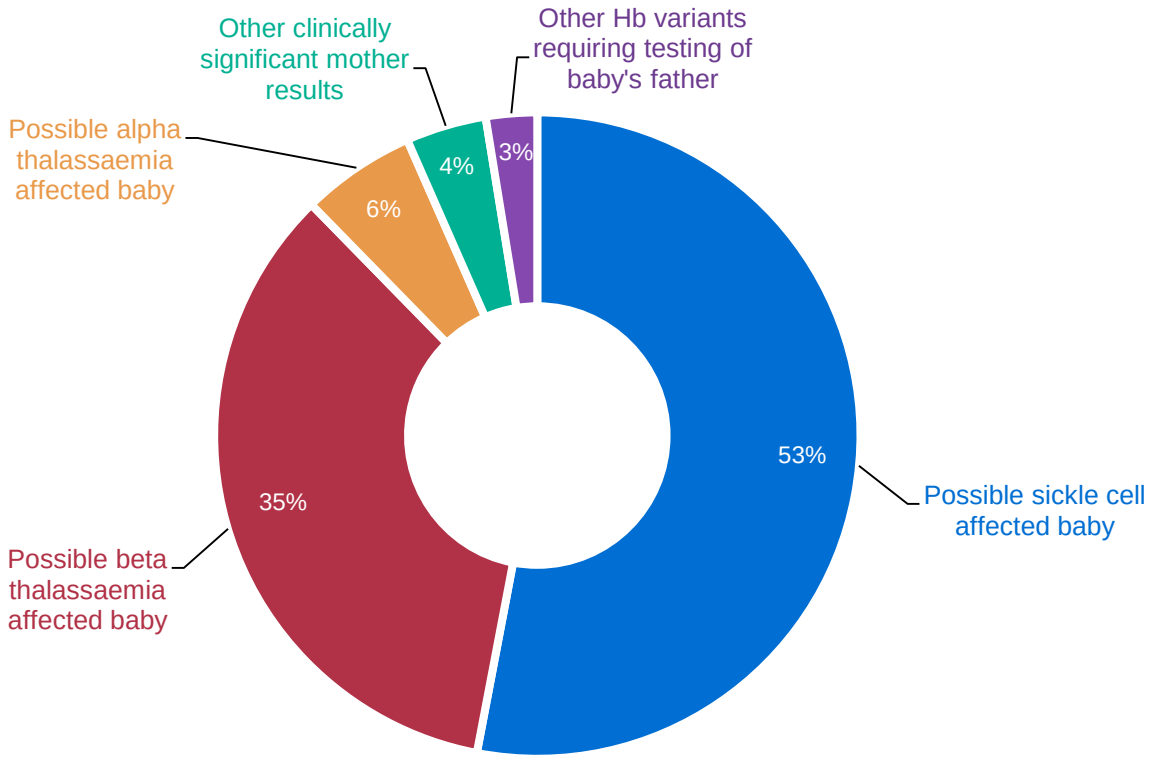
	Mother's screening result	Risk to pregnancy				Totals			
		High Risk	Low/ minimal risk	Father not a carrier	Father result not available	Total with result	Total for group	Rate/ 1000 samples received	% of screen positives
Possible sickle cell affected baby	Hb S	506	146	2,410	2,025	5,087	6,765	9.99	53.0
	Hb D	*	*	499	126	664			
	Hb C	52	56	480	413	1,001			
	Hb O-Arab	*	*	12	*	13			
Possible beta thalassaemia affected baby	βThalassaemia	132	81	2,590	721	3,524	4,427	6.54	34.7
	δβ thalassaemia	*	*	65	19	93			
	Hb E	13	38	585	163	799			
	Hb Lepore	*	*	8	*	11			
Possible alpha thalassaemia affected baby	High risk alpha0	32	28	475	196	731	731	1.08	5.7
Other clinically significant mother results	HPFH/Compound heterozygous/ donor egg/bone marrow transplant	15	29	314	157	515	515	0.76	4.0
Other Hb variants requiring testing of baby's father		-	14	241	72	327	327	0.48	2.6
Totals		753	437	7,679	3,896	12,765	12,765	18.86	100.0

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 women for whom father testing was requested that were reported by the same laboratories included here was 13,256 (96% included in the breakdown) and 748 high risk couples (100% included in the breakdown). 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.

Figure AN-8. Screen positive women, broken down by risk to the pregnancy, 2016 to 2017



Based on 12,765 of the 13,256 (96%) screen positive women reported by the laboratories in 2016/17.

4.7. Offer of screening early in pregnancy

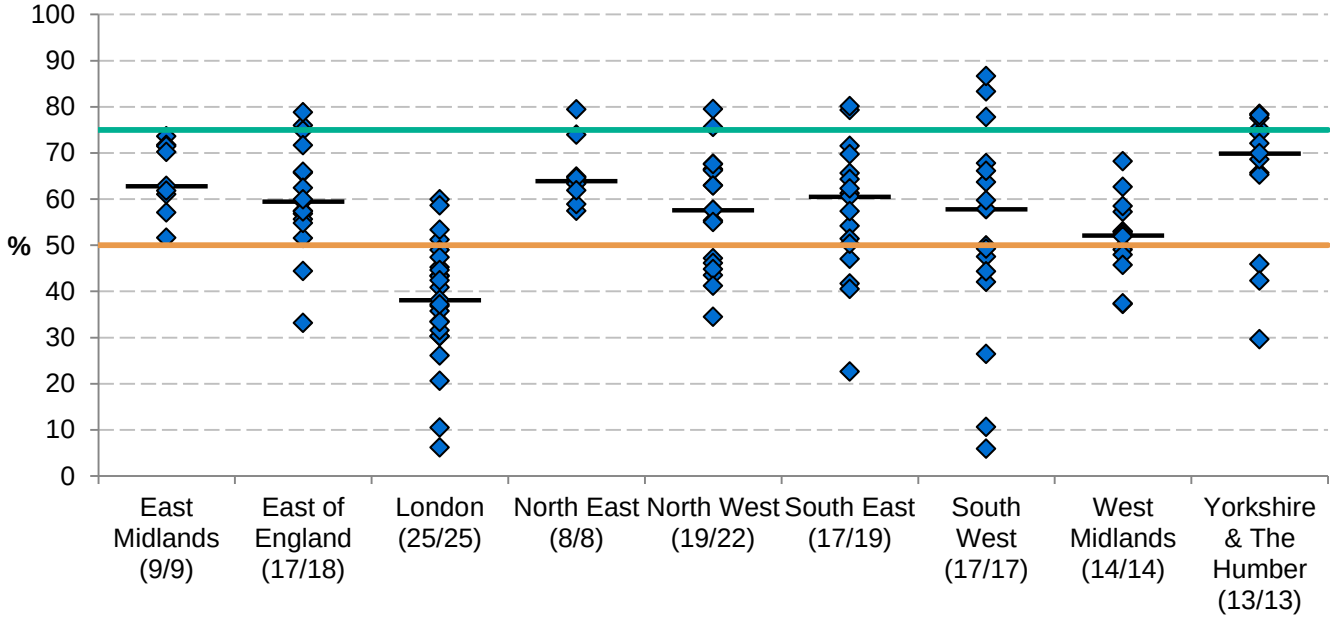
Antenatal screening

KPI ST2 collects data on the proportion of antenatal screening samples tested by 10 weeks and 0 days gestation on a quarterly basis. Annual data is derived from the quarterly returns, but exclusions are made for any provider that did not provide data in one or more quarter in that year. The thresholds for KPI ST2 set an acceptable level of 50% and an achievable level of 75%.

Table AN-9. Antenatal screening samples tested by 10 weeks + 0 days gestation, 2016 to 2017

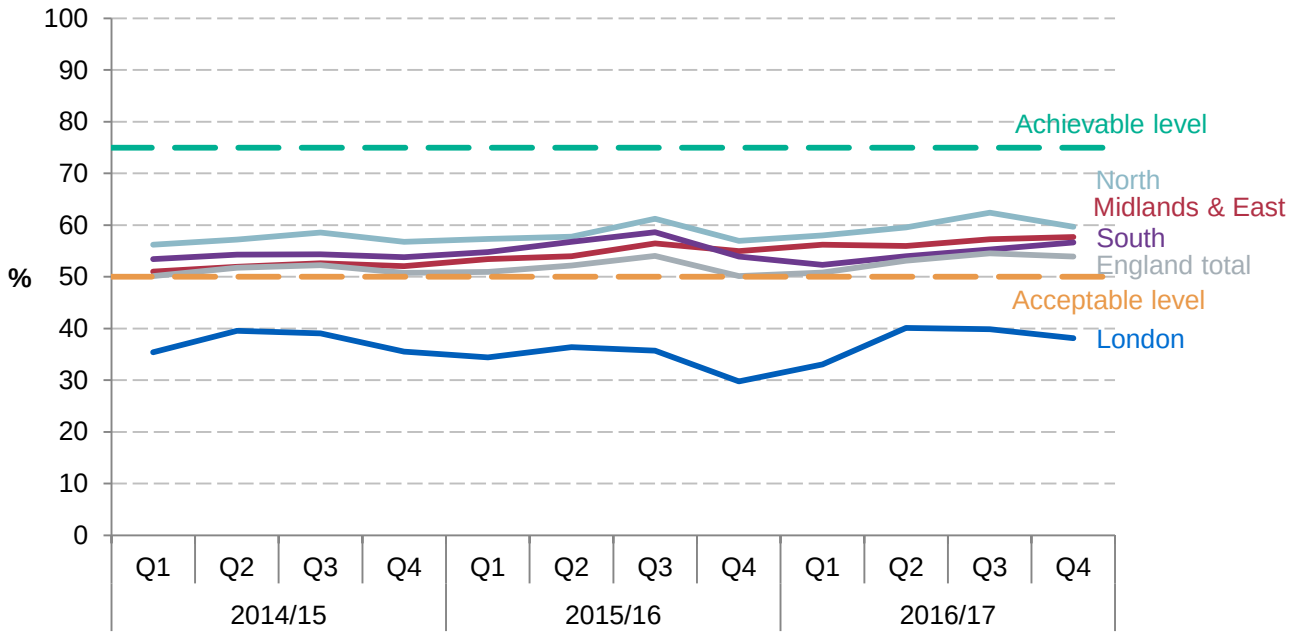
Region	Sub-region	Samples received by laboratory	Tested by 10 weeks + 0 days	%
London	London	149,171	56,282	37.7
Midlands & East	East Midlands	56,186	35,815	63.7
	East of England	76,039	44,686	58.8
	West Midlands	80,544	40,349	50.1
North	North East	29,210	18,493	63.3
	North West	87,747	48,114	54.8
	Yorkshire & The Humber	69,921	45,326	64.8
South	South East	93,108	53,728	57.7
	South West	61,465	30,627	49.8
England		703,391	373,420	53.1

Figure AN-9. Variation in testing by 10 weeks and 0 days by sub-region, 2016 to 2017: England



The black horizontal markers represent the median value for each sub-region. The orange reference line represents the acceptable level for this KPI, and the green reference line represents the achievable level for theis KPI. The numbers in brackets represent the number of providers that submitted data for this KPI out of the number of expected returns.

Figure AN-10. Trends in antenatal screening samples tested by 10 weeks + 0 days gestation by region, 2014 to 2015 – 2016 to 2017



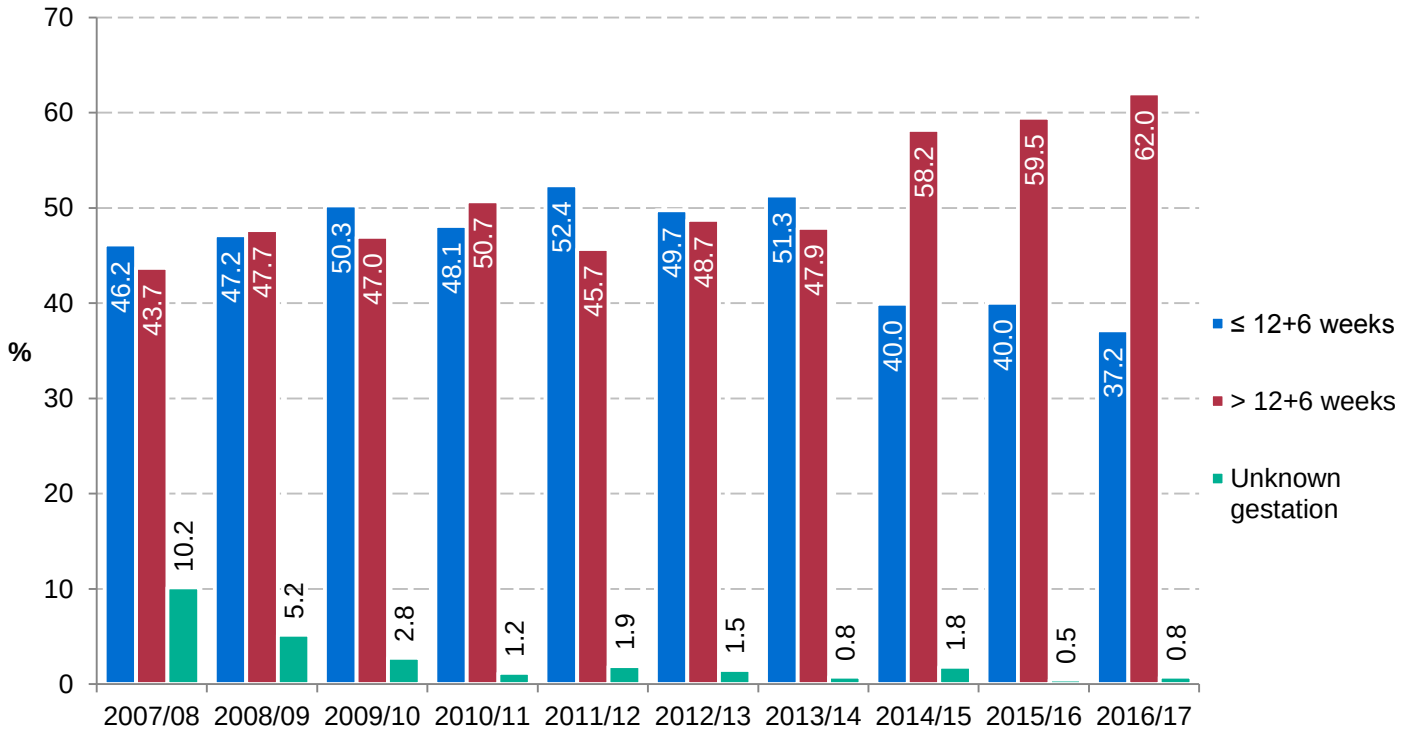
Exclusions where data was note returned in all four quarters: 2014/15: 16; 2015/16: 11; 2016/17: 6.

Prenatal diagnosis

Table PND-1. Gestation at PND test, 2014 to 2015 – 2016 to 2017

Gestation at PND test	2014/15		2015/16		2016/17	
	n	%	n	%	n	%
<12+6 weeks	173	40.0	163	40.0	139	37.2
13+0 - 14+6 weeks	121	27.9	106	26.0	110	29.4
≥15+0 weeks	131	30.3	136	33.4	122	32.6
Unknown gestation	8	1.8	2	0.5	3	0.8
Total	433	100.0	407	100.0	374	100.0

Figure AN-11. Proportion of PND tests performed by gestation, 2007 to 2008 – 2016 to 2017



4.8. Numbers tested and detected in prenatal diagnostic testing

Figure PND-1. Number of PND tests performed by laboratory, 2007 to 2008 – 2016 to 2017

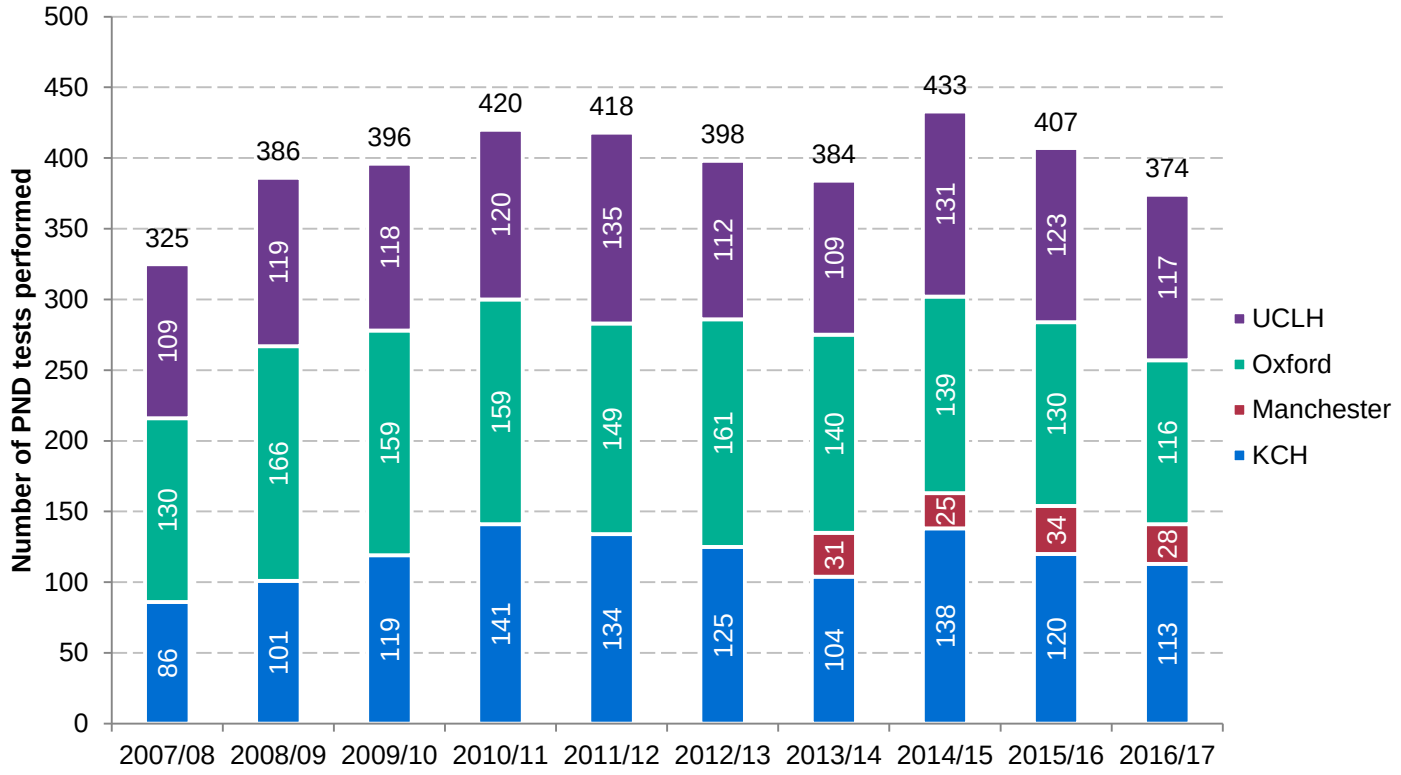


Table PND-2. Breakdown of PND fetal results by condition, 2014 to 2015 – 2016 to 2017

Fetal result†	PND result/risk	2014/15	2015/16	2016/17
Affected	Sickle Cell affected	99	80	65
	Thalassaemia affected	23	24	13
	Other	1	0	2
Carrier	Sickle Cell carrier	155	125	146
	Thalassaemia carrier	39	46	38
	Other	12	8	6
NAD	Risk for Sickle Cell	89	97	84
	Risk for Thalassaemia	15	26	20
	Risk not known	0	0	0
Inconclusive/result not known	All risks	0	1	0
Total		433	407	374

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.

4.9. Prenatal diagnostic results by family origin

Figure PND-2. Number of PND tests by mother's family origins, 2014 to 2015 – 2016 to 2017

	2014/15		2015/16		2016/17	
Mother's family origin	n	%	n	%	n	%
African	220	50.81	201	49.4	195	52.1
Caribbean	104	24.02	88	21.6	80	21.4
Indian	12	2.77	17	4.2	10	2.7
Pakistani	4	0.92	7	1.7	3	0.8
Cypriot/Mixed Cypriot	7	1.62	9	2.2	7	1.9
Other Asian	30	6.93	33	8.1	29	7.8
Southern & Other European	10	2.31	11	2.7	10	2.7
Middle Eastern	5	1.15	8	2.0	9	2.4
Mixed/Other	5	1.15	15	3.7	8	2.1
Not Known	36	8.31	18	4.4	23	6.1
Total	433	100.00	407	100.0	374	100.0

4.10. 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.

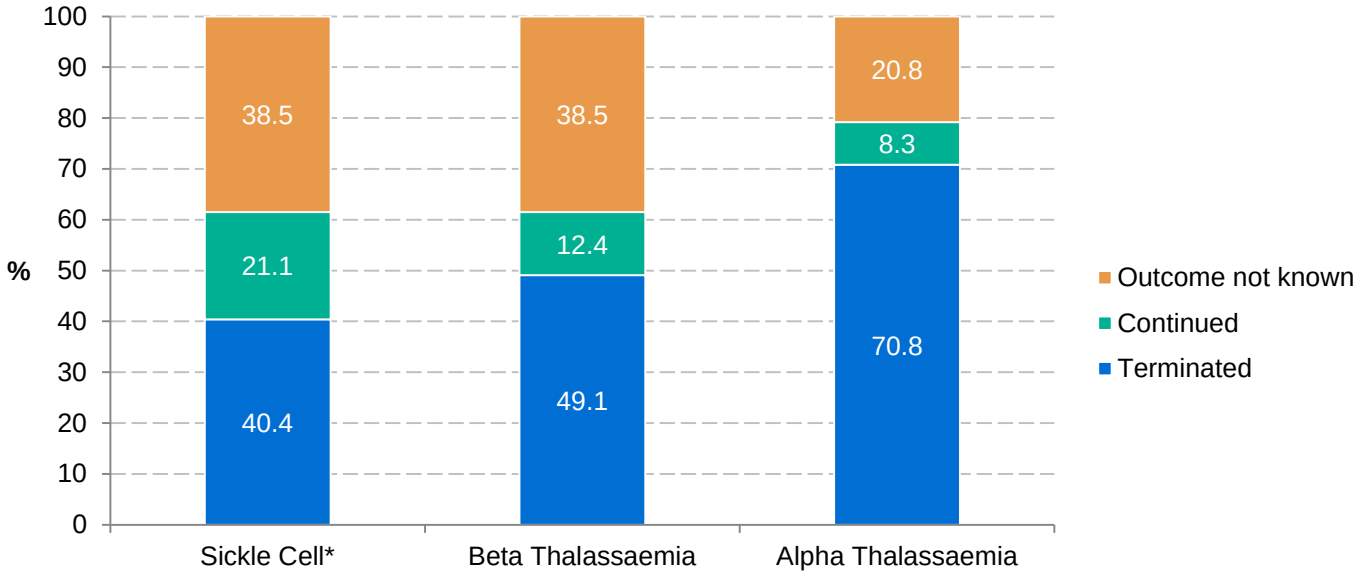
Table PND-3. Outcomes for pregnancies with affected fetal results at PND 2014 to 2015 – 2016 to 2017

Condition	Pregnancy outcome	2014/15 % of total identified with condition	2015/16 % of total identified with condition	2016/17 % of total identified with condition
Sickle Cell	Continued	27.3	15.0	18.5
	Terminated	38.4	46.3	32.3
	Not Known	34.3	38.8	46.2
Beta Thalassaemia	Continued	22.7	18.2	30.0
	Terminated	40.9	54.5	40.0
	Not Known	36.4	27.3	30.0
Alpha Thalassaemia	Continued	0.0	0.0	0.0
	Terminated	100.0	100.0	100.0
	Not Known	0.0	0.0	0.0
Total Affected		123	104	80

Other haemoglobin variants and miscarriage outcomes have been excluded.

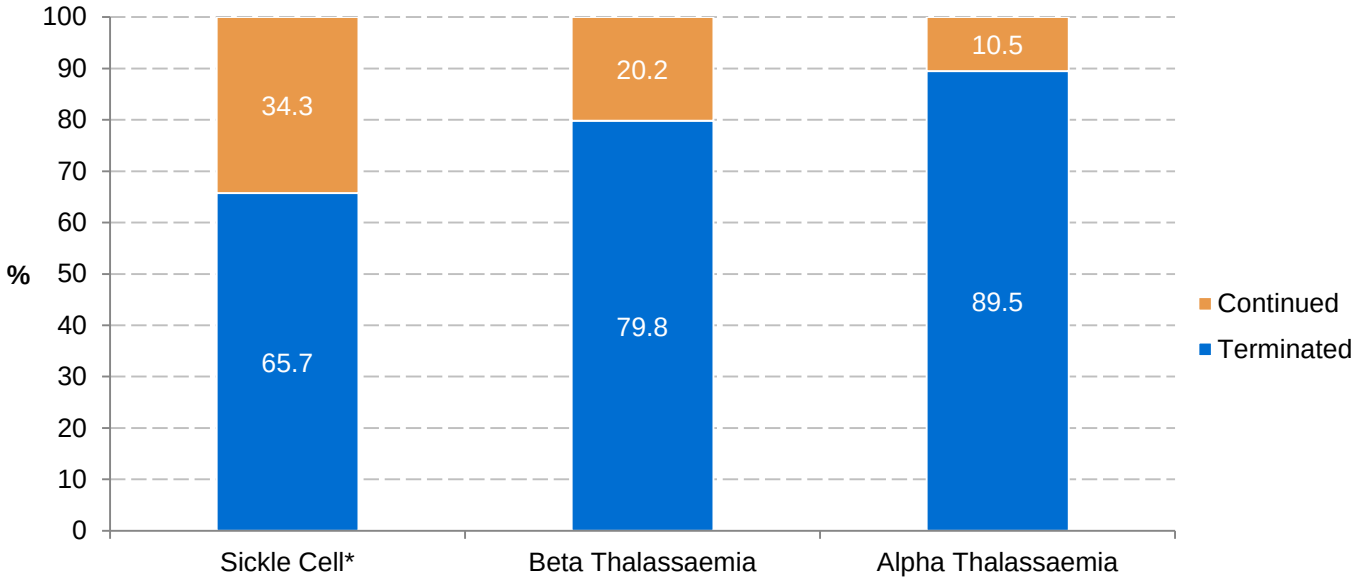
Please note that alpha thalassaemia rates are based on small numbers and should be interpreted with caution.

Figure PND-3. Outcomes for pregnancies with 'affected' diagnosis at PND, 2008 to 2009 – 2016 to 2017



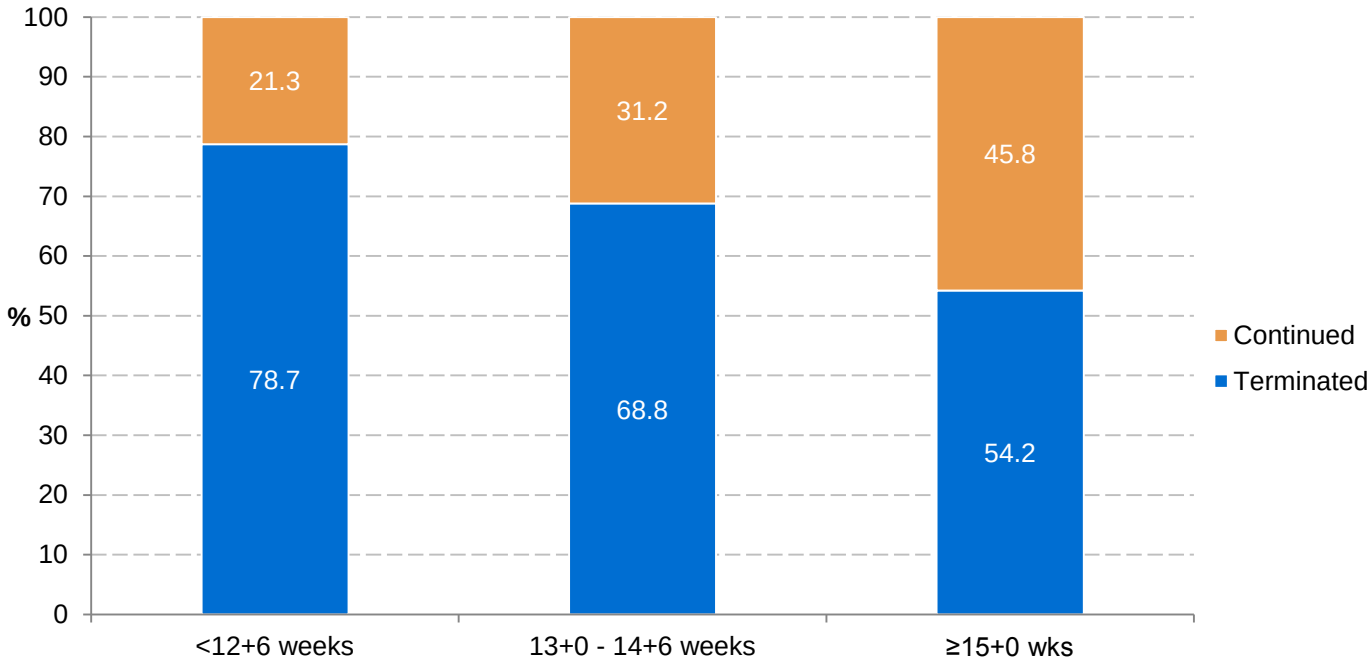
*The "Sickle Cell" category includes HbSS, HbSC, HbS/beta thalassaemia, and HbS+other variant requiring clinical follow-up. Excludes miscarriage outcomes due to small numbers.

Figure PND-4. Outcomes for pregnancies with 'affected' diagnosis at PND (known outcomes only), 2008 to 2009 – 2016 to 2017



*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. Outcomes by gestation at PND (known outcomes only), 2008 to 2009 – 2016 to 2017



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

5. Newborn screening for sickle cell disease

Overview

Newborn coverage (born and resident in the CCG)

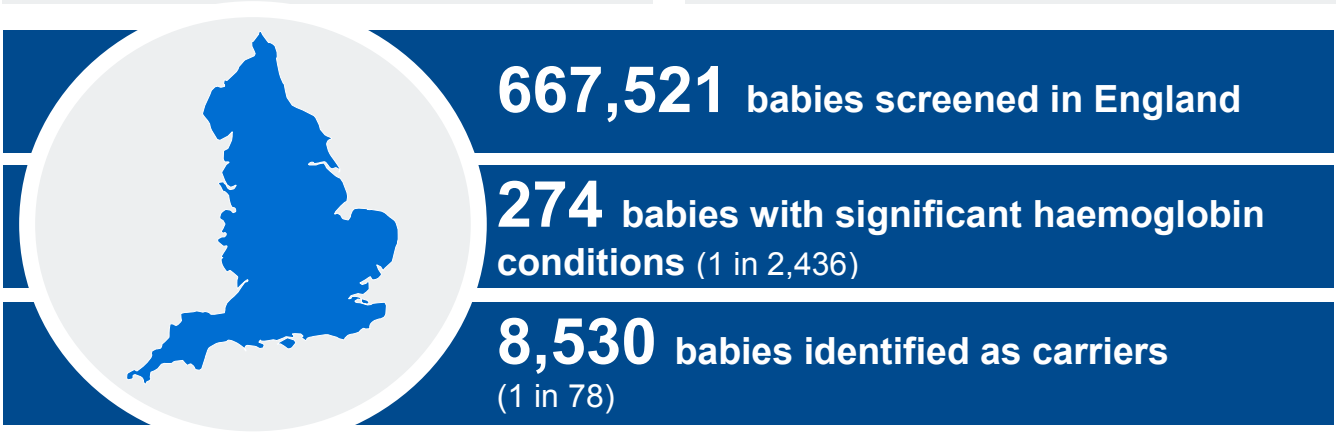
96.5% in England

(range 79.4% – 99.9%)

Newborn coverage (movers-in to the CCG)

87.1% in England

(range 47.5% – 100.0%)



667,521 babies screened in England

274 babies with significant haemoglobin conditions (1 in 2,436)

8,530 babies identified as carriers (1 in 78)

Post-transfusion samples

1.8 per 1,000 babies screened had a sample taken after having a blood transfusion

Newborn declines

2.3 per 1,000 babies reported by laboratories as having declined screening for sickle cell disease

Timeliness

95.8% of samples taken by 8 days of age

90.1% of babies had first visit to a paediatrician ≤90 days (where data provided)

5.1. Newborn coverage

Newborn screening KPI NB1 collects coverage data 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. 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.

Please note that the coverage figures from KPI NB1 only include those born and resident in the sub-region and will not include movers-in. KPI NB4 collects data on coverage of movers-in, using an effective timeframe of 21 calendar days of notification to the CHRD of movement in.

Table NB-1. Coverage of newborn screening (born and resident population), 2016/17: England

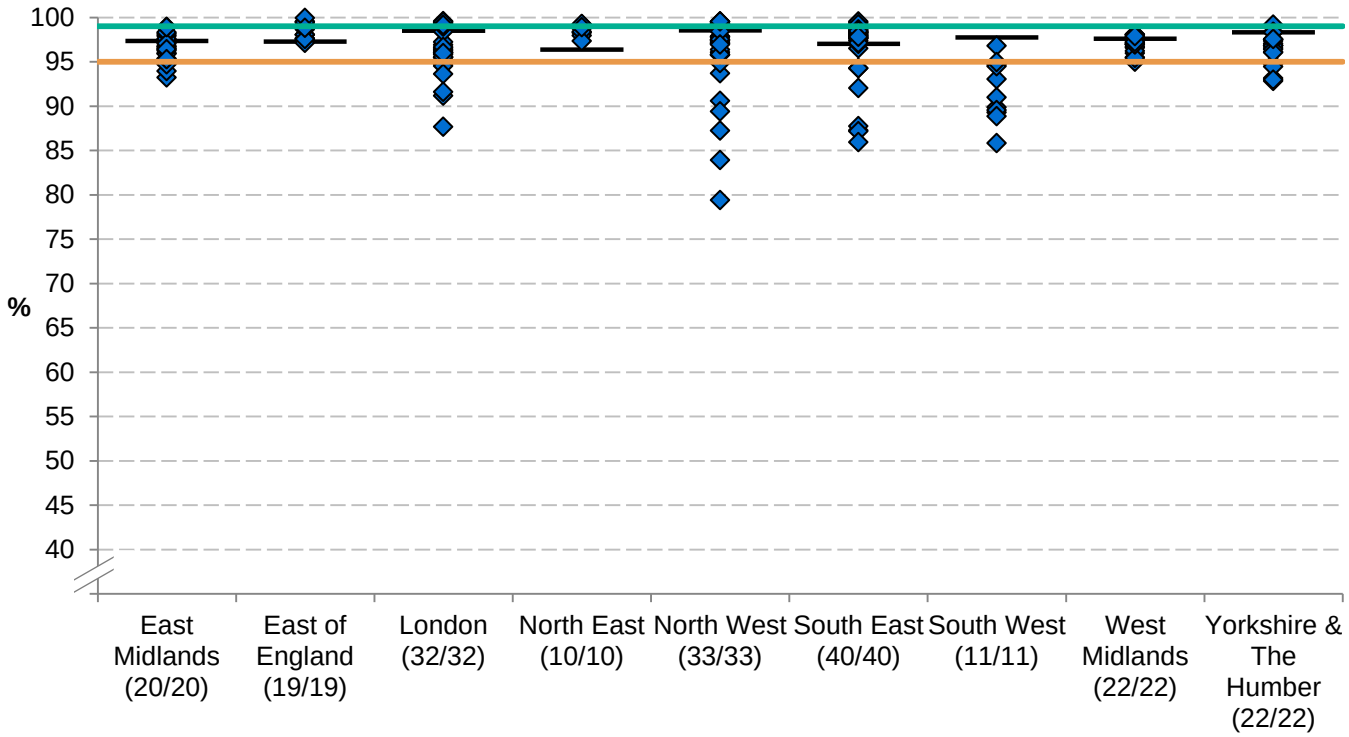
Sub-region	Completeness: Submitted all 4 quarters/ no.of providers	Tested babies	Eligible babies	Coverage (%)
East Midlands	20 / 20	47,188	49,084	96.1
East of England	19 / 19	63,840	64,946	98.3
London	32 / 32	109,784	114,069	96.2
North East	10 / 10	26,553	26,912	98.7
North West	33 / 33	76,515	79,202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

Table NB-2. Coverage of newborn screening (movers-in), 2016 to 2017: England

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

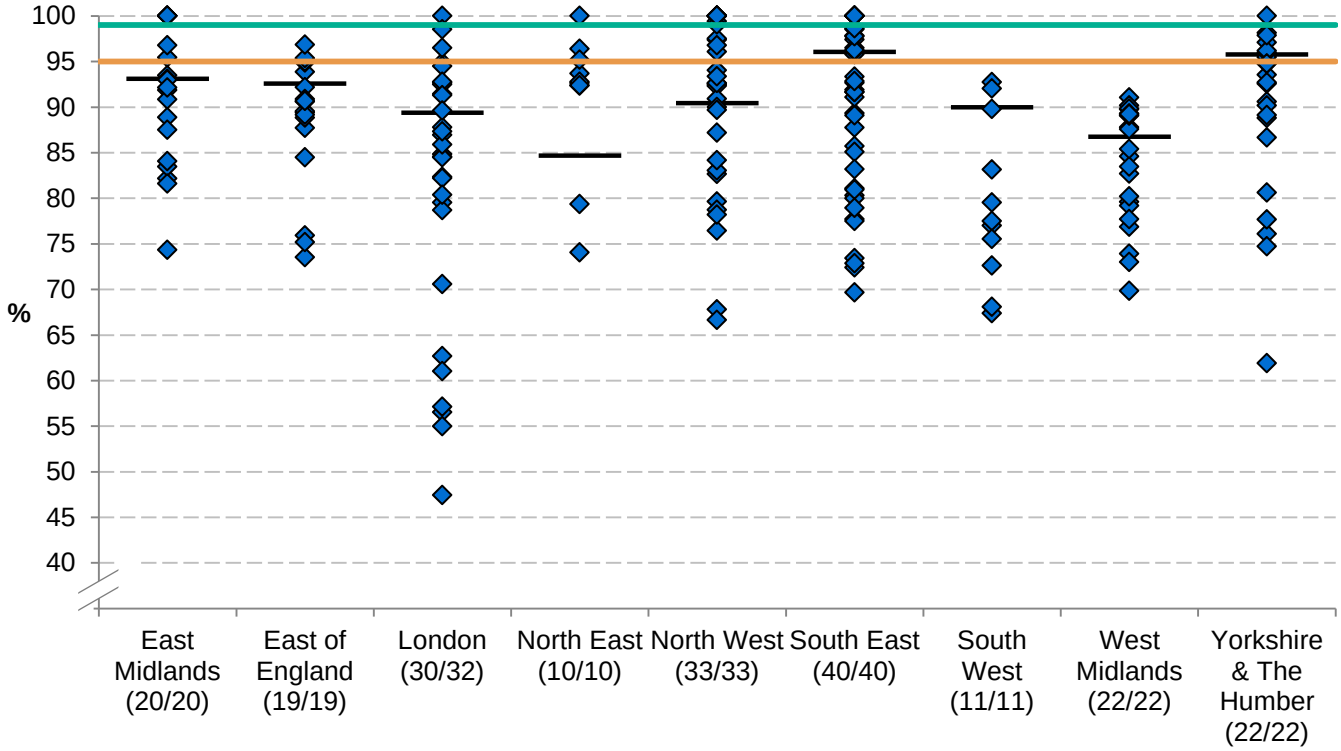
*2 providers excluded where data was not returned in all four quarters.

Figure NB-1. Variation in coverage of newborn screening (born and resident in the CCG) by sub-region, 2016 to 2017: England by sub-region



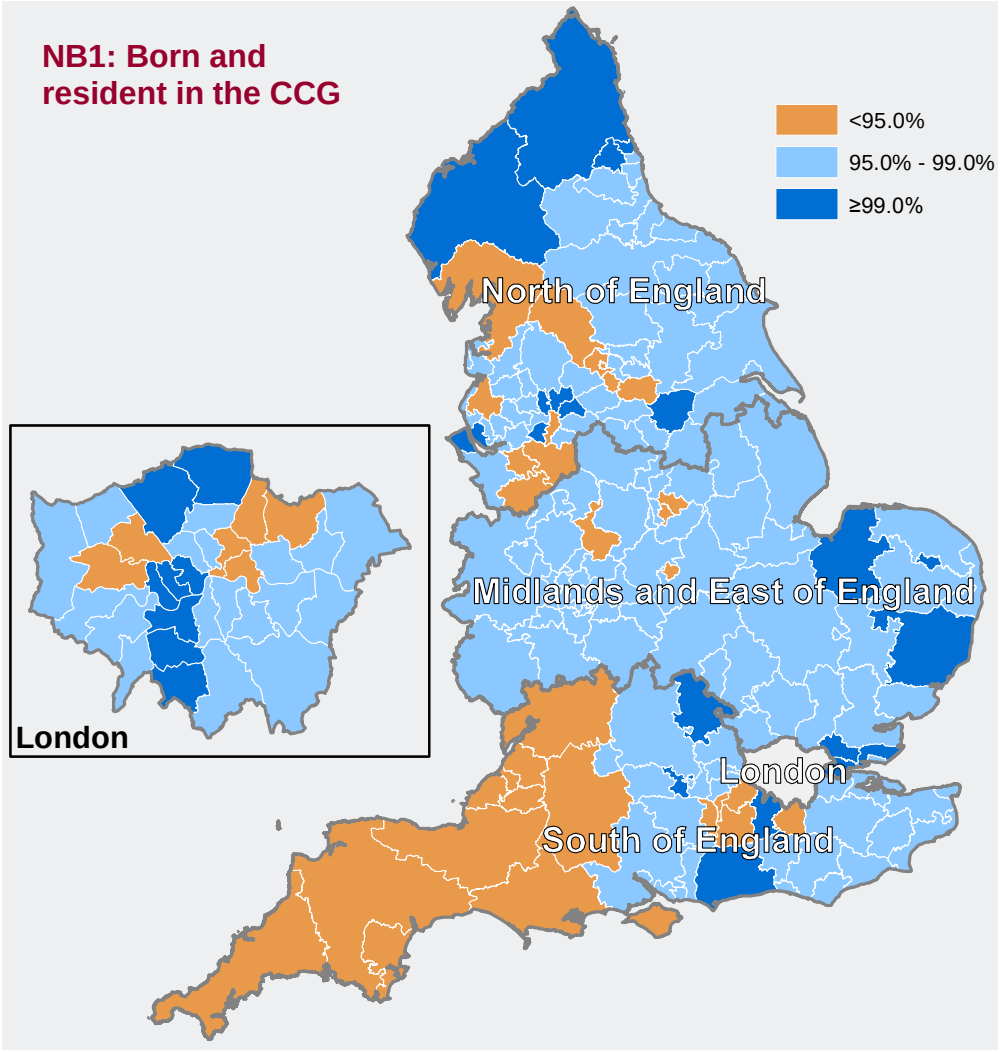
The black horizontal markers represent the median value for each sub-region. The orange reference line represents the acceptable level for this KPI, and the green reference line represents the achievable level for theis KPI.

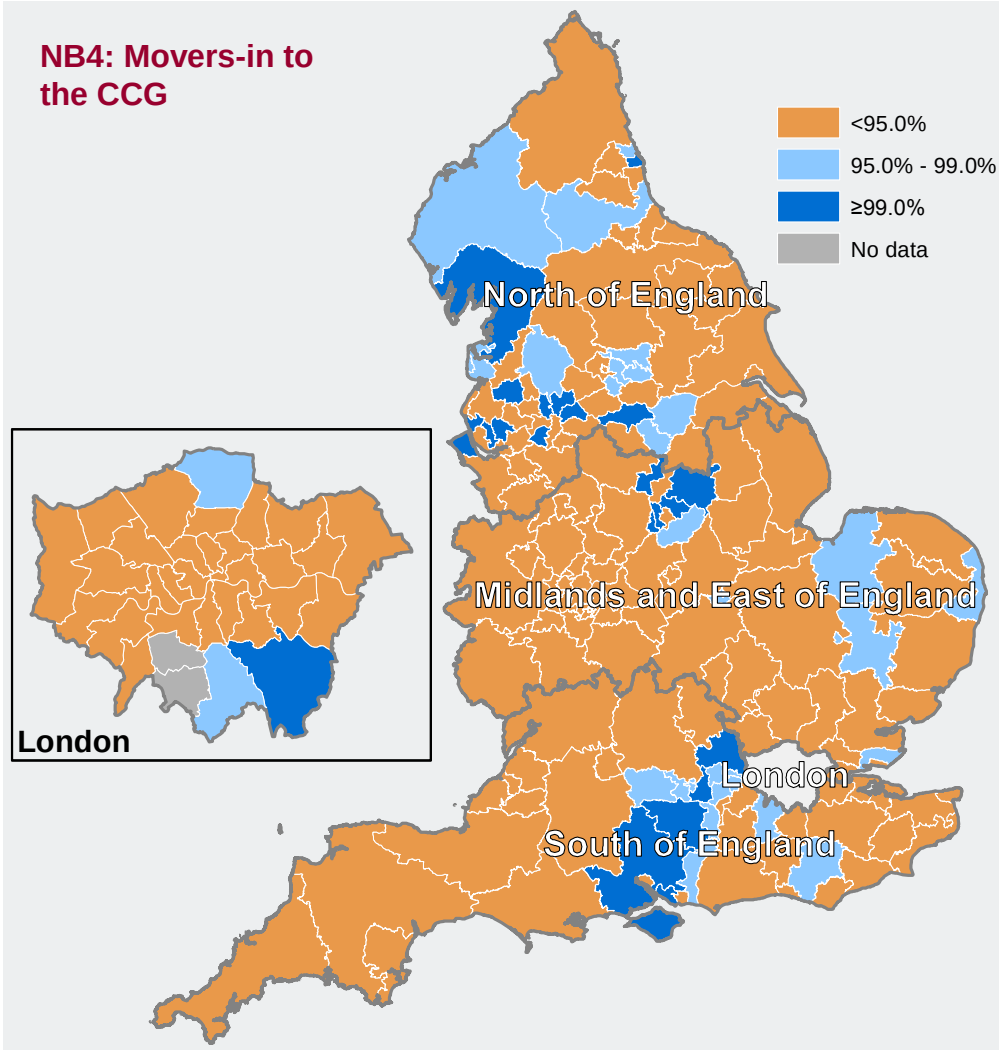
Figure NB-2. Variation in coverage of newborn screening (movers-in to the CCG) by sub-region, 2016 to 2017: England by sub-region



The black horizontal markers represent the median value for each sub-region. The orange reference line represents the acceptable level for this KPI, and the green reference line represents the achievable level for theis KPI.

Figure NB-3. Coverage for newborn screening, 2016 to 2017: England by CCG





5.2. Numbers screened and results

Significant conditions comprise FS, FSC, FS-other and FE results. Carrier results comprise FAS, FAC, FAD, FAE and other haemoglobin variants.

Table NB-3. Numbers and rates of significant conditions and carrier results, 2016 to 2017

Region	Significant Conditions			Carriers			No. of babies Screened (tested + declines)
	n	Rate/1000	1 in x	n	Rate/1000	1 in x	
North	33	0.19	5,396	1268	7.12	140	178,068
South	30	0.19	5,183	1099	7.07	141	155,498
Midlands & East	65	0.34	2,923	2142	11.27	89	189,999
London	139	1.05	957	3903	29.35	34	132,959
Unknown region	7	0.64	1,563	118	10.79	93	10,997
England Total	274	0.41	2,436	8,530	12.78	78	667,521
Scotland	*	0.06	18,122	211	3.88	258	54,365
Wales†	*	0.09	10,925	0	-	-	32,776
Northern Ireland	0	-	-	43	1.77	565	24,311
UK total	280	0.36	2,782	8,784	11.28	89	778,973

*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.

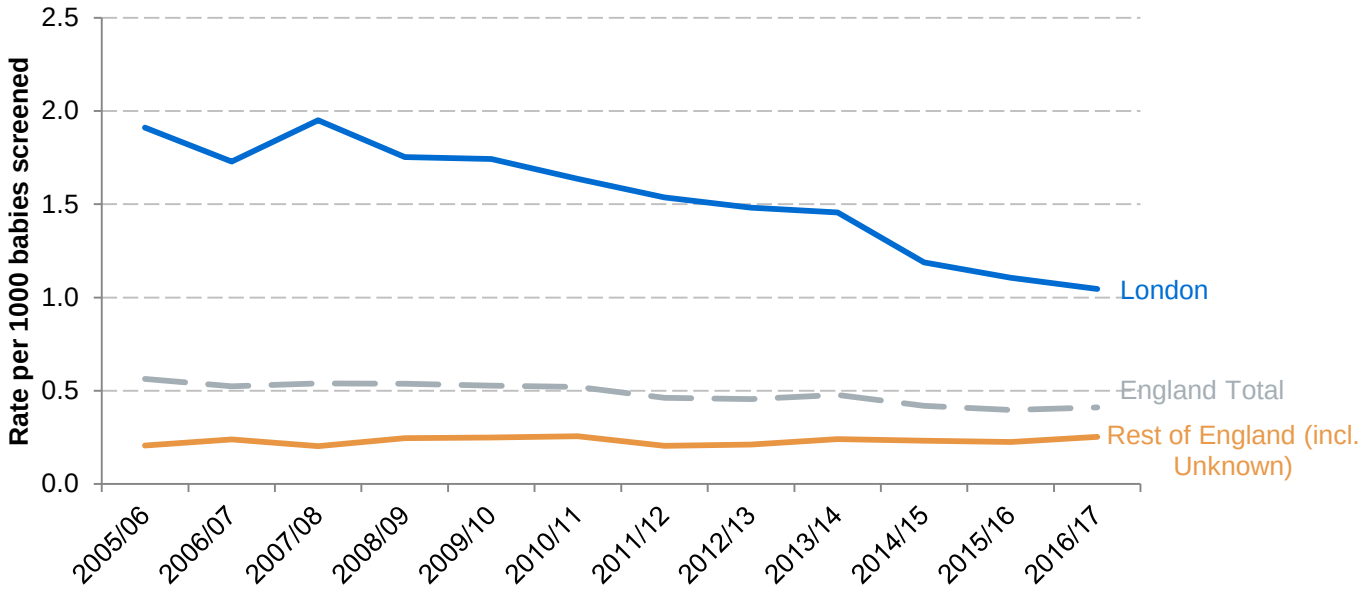
Table NB-4. Breakdown of newborn screening results, 2016 to 2017

Region	Significant Conditions					Carriers					Trans-fused	Declined	Normal+ Abnormal	Babies screened (tested + declined)
	FS	FSC	FS-Other	FE	F-only	FAS	FAC	FAD	FAE	Other				
North	24	*	6	*	7	815	154	162	131	6	223	181	177,887	178,068
South	16	14	*	*	*	685	156	107	142	9	337	258	155,240	155,498
Midlands & East	44	17	*	*	7	1,354	346	222	217	*	255	450	189,549	189,999
London	95	32	*	9	9	2,733	570	205	389	6	205	331	132,628	132,959
Unknown region	5	*	*	*	*	79	21	9	8	*	189	233	10,705	10,997
England Total	184	66	11	13	25	5,666	1,247	705	887	25	1,209	1,453	666,009	667,521
Scotland	*	*	*	*	*	153	18	18	21	*	56	52	54,313	54,365
Wales†	*	*	*	*	*	*	*	*	*	*	20	184	32,592	32,776
Northern Ireland	*	*	*	*	*	28	*	5	8	*	29	222	24,089	24,311
UK total	189	67	11	13	25	5,847	1,267	728	916	26	1,314	1,911	777,003	778,973

*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.

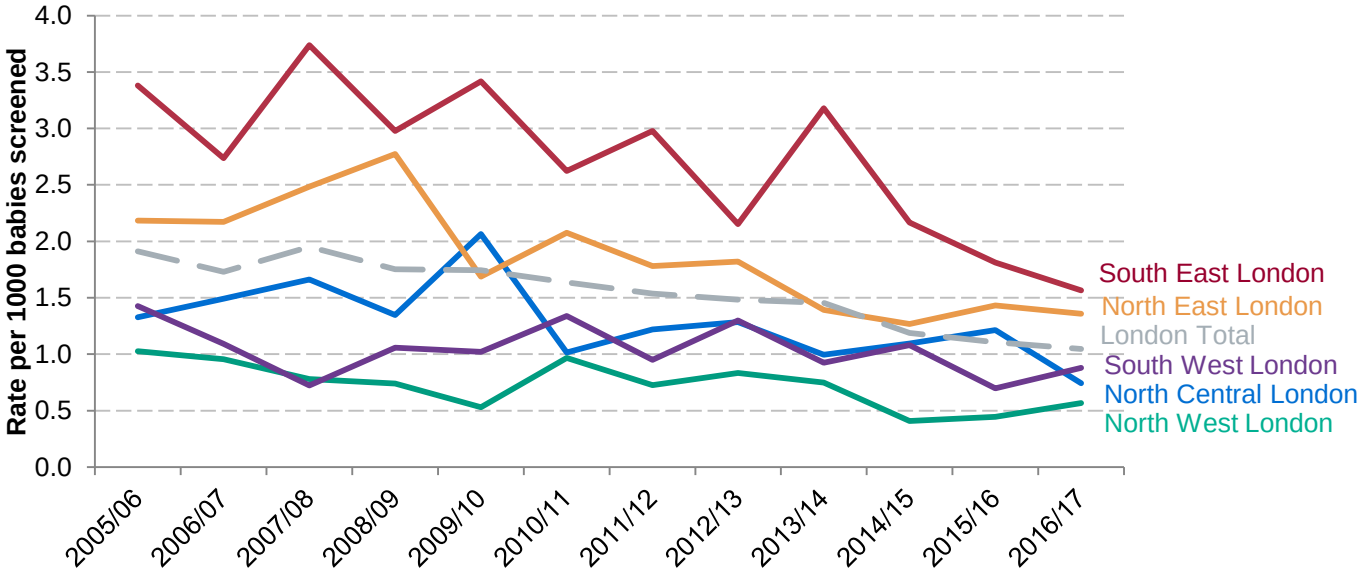
The Wales newborn screening protocol is designed to detect only the disease states of SCD. However, those cases that are identified from the newborn screening process and subsequently determined to be carriers of SCD are referred for follow-up.

Figure NB-4. Trends in rates of babies identified with a significant condition, 2005 to 2006 – 2016 to 2017



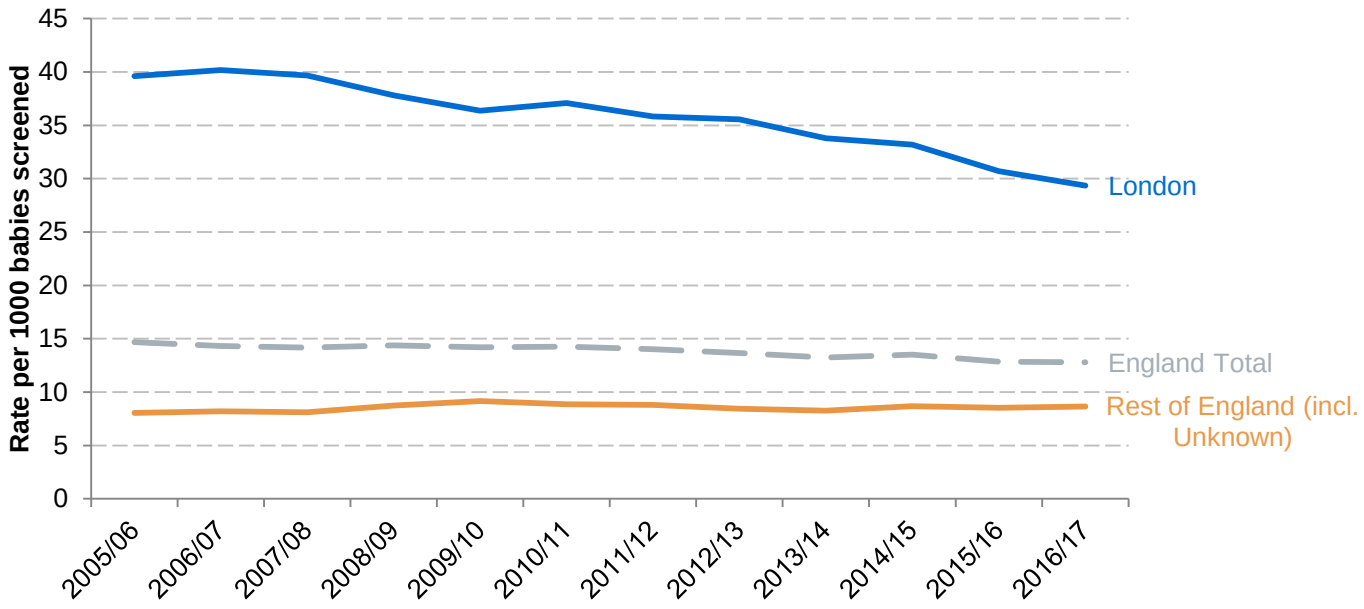
Significant conditions comprise FS,FSC, FS Other and FE.

Figure NB-5. Trends in rates of babies identified with a significant condition, 2005 to 2006 – 2016 to 2017: London sectors (pre-2006 SHA)



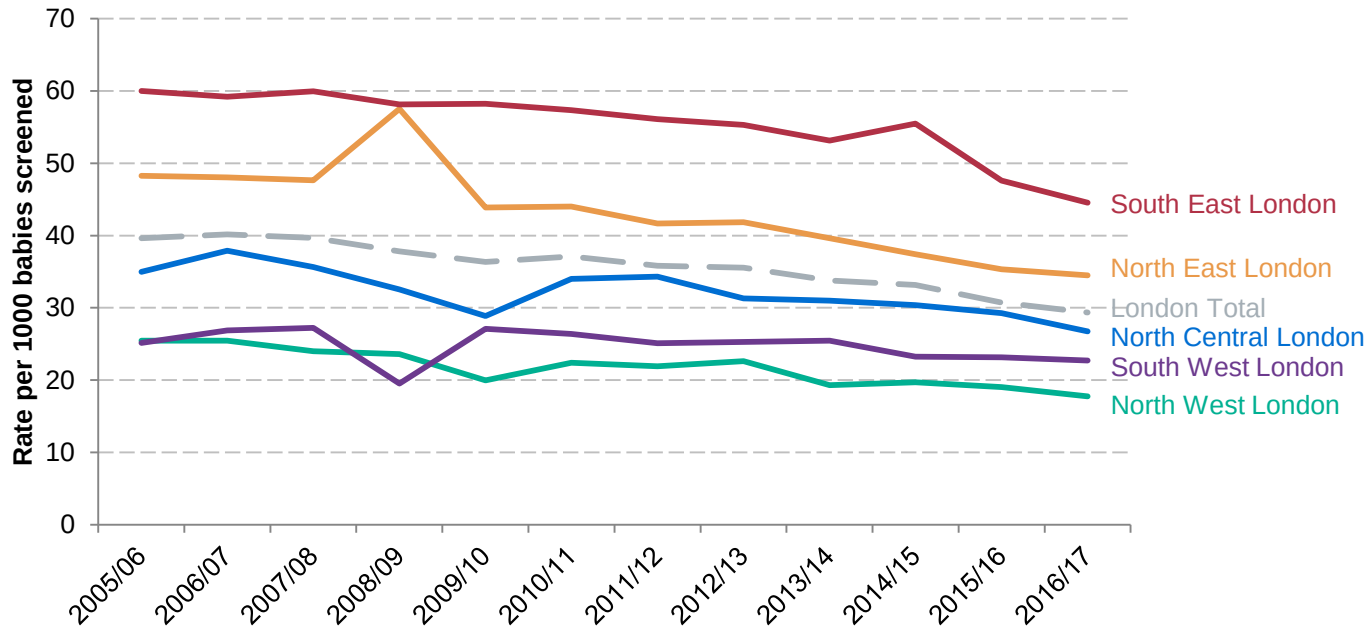
Significant conditions comprise FS,FSC, FS Other and FE.

Figure NB-6. Trends in rates of babies with carrier results, 2005 to 2006 – 2016 to 2017



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

Figure NB-7. Trends in rates of babies with carrier results, 2005 to 2006 – 2016 to 2017: London sectors (pre-2006 SHA)



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

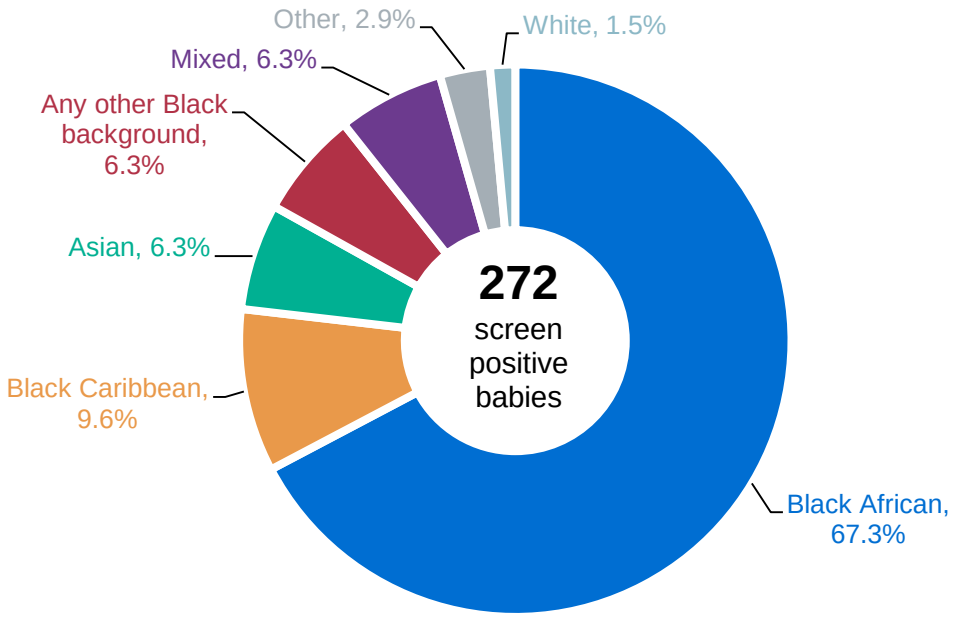
5.3. Results by ethnicity

Table NB-5. Numbers and rates of significant conditions and carrier results by ethnic category, 2016 to 2017

Ethnic Category	Significant Conditions			Carriers			No. of babies Screened (tested + declines)
	n	Rate/ 1000	1 in x	n	Rate/ 1000	1 in x	
White	4	0.01	118,676	742	1.56	640	474,704
Mixed	17	0.41	2,439	1,598	38.54	26	41,461
Asian	17	0.23	4,381	1,208	16.22	62	74,471
Black Caribbean	26	4.35	230	728	121.72	8	5,981
Black African	183	7.69	130	3,283	137.99	7	23,791
Any other Black background	17	4.23	236	417	103.76	10	4,019
Other*	8	0.20	5,108	538	13.17	76	40,923
England Total	272	0.41	2,446	8,514	12.80	78	665,350

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

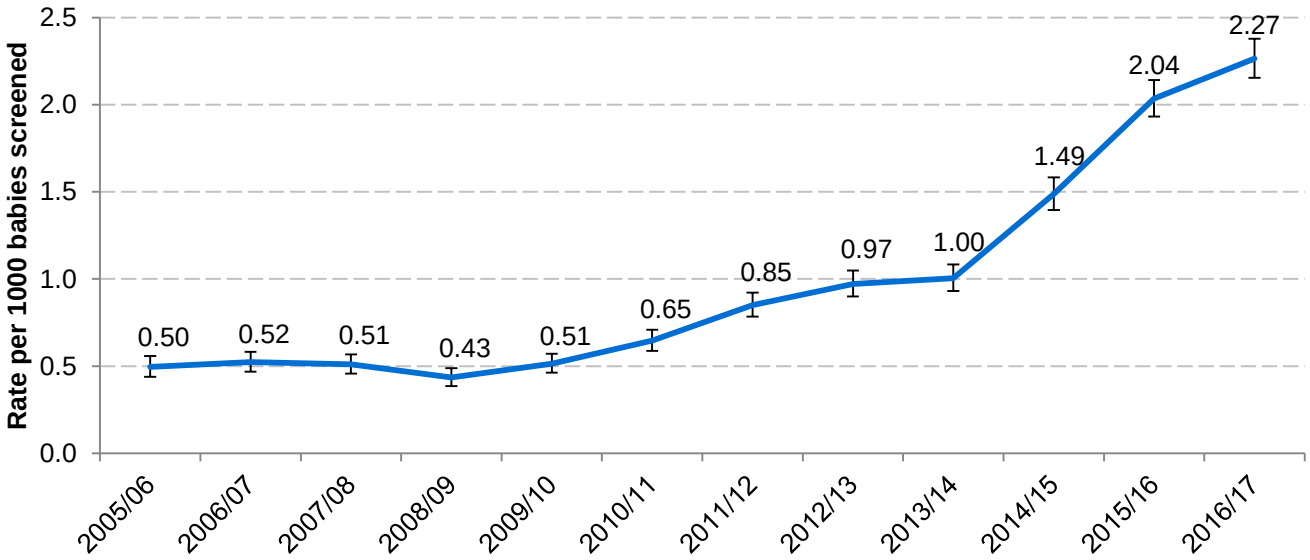
Figure NB-8. Breakdown of screen positive babies by ethnic category, percentage of all screen positives, 2016 to 2017: England



5.4. Declined screening test

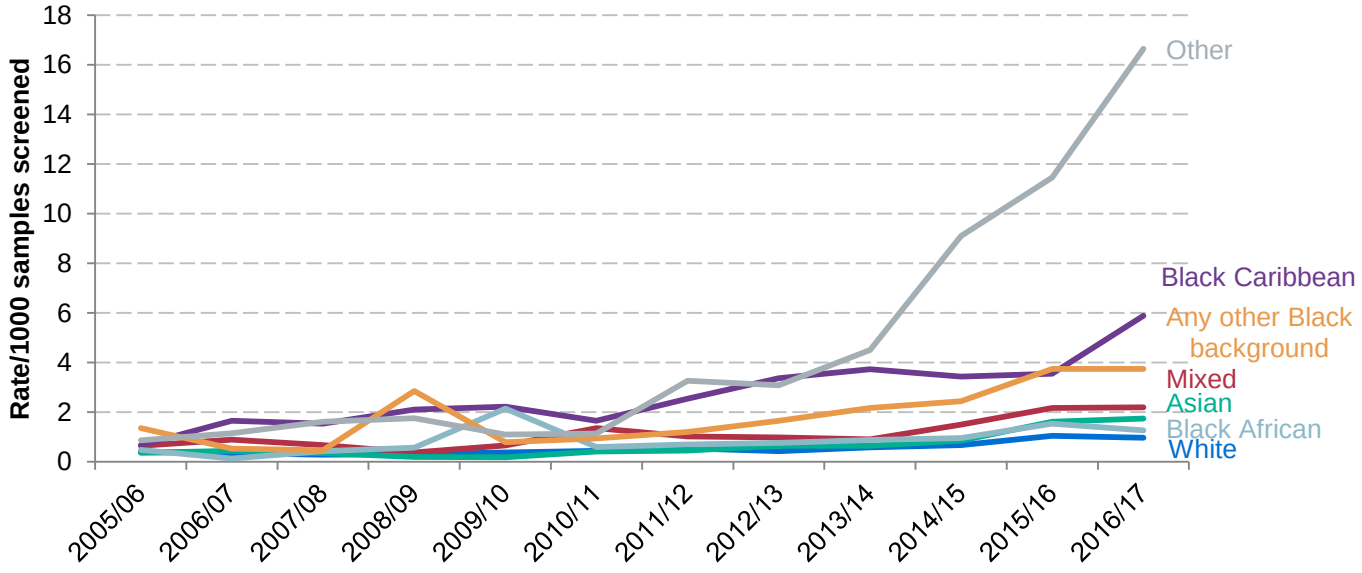
There appears to be a continuation of the increase in the rate of declined tests, and the rate is now at 2.27 per 1,000 babies screened. It is difficult to identify the causes as the reason for declining is not recorded, but some potential explanations include mover-in babies who may have been tested elsewhere or it may be due to better reporting now that there is a laboratory sub-code for declines.

Figure NB-9. Declined screening tests for sickle cell disease, 2005 to 2006 – 2016 to 2017



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 1st July 2006.

Figure NB-10. Declined screening tests for sickle cell disease by ethnic category, 2005 to 2006 – 2016 to 2017: England

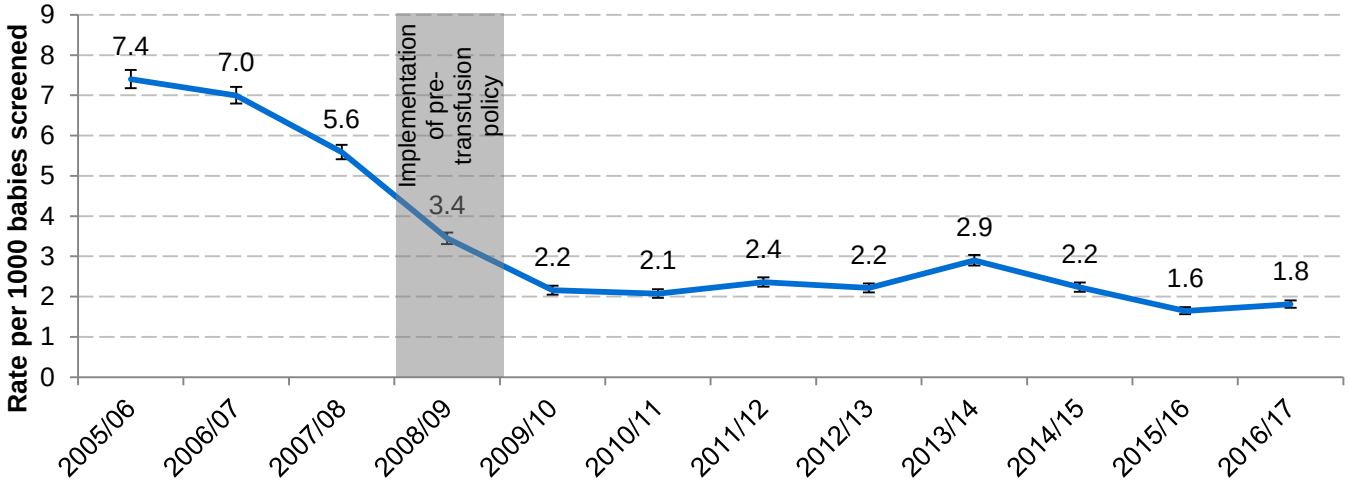


5.5. Post-transfusion testing

Table NB-6. Number and rates of post-transfusion samples reported by newborn laboratories, 2014 to 2015 – 2016 to 2017

Sub-region	2014/15			2015/16			2016/17		
	n	Total screened	Rate/ 1000	n	Total screened	Rate/ 1000	n	Total screened	Rate/ 1000
East Midlands	88	48,552	1.81	82	48,583	1.69	91	47,477	1.92
East of England	55	69,581	0.79	44	71,375	0.62	63	72,605	0.87
London	432	130,388	3.31	269	130,150	2.07	205	132,959	1.54
North East	40	26,387	1.52	37	28,596	1.29	60	28,805	2.08
North West	133	85,327	1.56	164	87,106	1.88	50	81,792	0.61
South East	298	102,999	2.89	131	102,284	1.28	101	103,299	0.98
South West	35	57,861	0.60	35	53,466	0.65	236	52,199	4.52
West Midlands	104	69,870	1.49	149	70,676	2.11	101	69,917	1.44
Yorkshire and the Humber	149	65,406	2.28	124	68,462	1.81	113	67,471	1.67
Unknown	146	6,046	24.15	64	7,102	9.01	189	10,997	17.19
England total	**1,480**	**662,417**	**2.23**	**1,099**	**667,800**	**1.65**	**1,209**	**667,521**	**1.81**

Figure NB-11. Rates of post-transfusion samples, 2005 to 2006 – 2016 to 2017



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 1st July 2006; Transfused 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.

Table NB-7. Numbers detected through DNA testing for transfused babies reported by DNA testing laboratories, 2012 to 2013 – 2016 to 2017

	2012/13	2013/14	2014/15	2015/16	2016/17
Total Specimens received per Quarter	1,343	1,160	1,123	1,198	1,071
Number of Negative results (HbS not detected)	1,319	1,140	1,106	1,183	1,054
Number of Positive Heterozygotes	21	20	16	15	17
Number of Positive Homozygotes	*	0	*	0	0

*Numbers less than 5 have been suppressed

Table NB-8. Number of post-transfusion samples received from each screening laboratory, 2016 to 2017

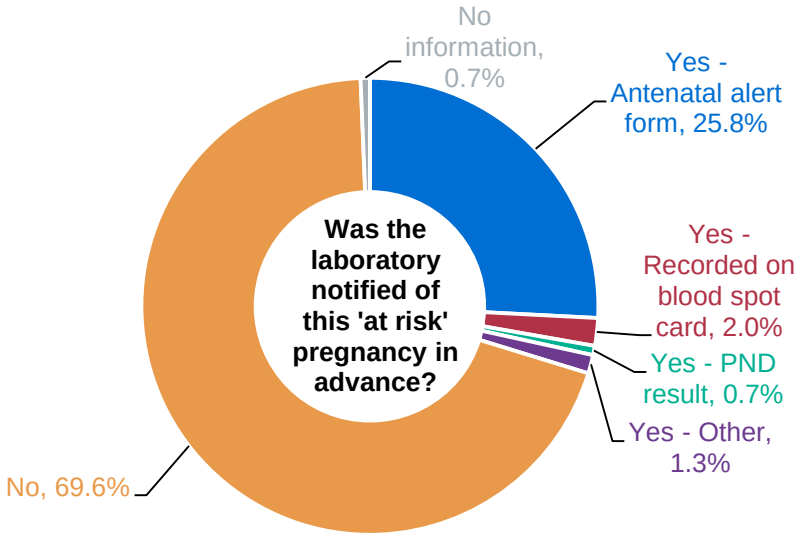
DNA testing laboratory	Newborn Laboratory	Number of samples
King's College Hospital	Bristol	27
	Cambridge	33
	GOS & CMH	181
	Oxford	24
	Portsmouth	70
	South East Thames	171
	South West Thames	61
Sheffield	Leeds	76
	Liverpool	61
	Manchester	60
	Newcastle	55
	Sheffield	150
	West Midlands	102
England total		1,071

5.6. 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-12. Proportion of screen positive babies where the laboratory was notified in advance of newborn screening, 2016 to 2017: England



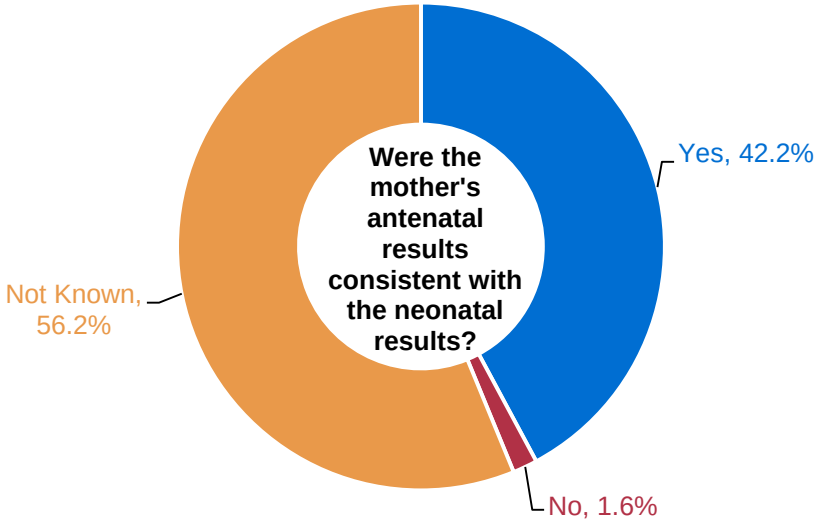
Another link between antenatal and newborn screening is through antenatal screening results being available to the newborn screening laboratory at the time of testing.

Table NB-9. Screen positive babies for whom antenatal results were available at the time of testing, 2016 to 2017

Antenatal results recorded on blood spot card?	Yes		No		Not Known		Total*
	n	%	n	%	n	%	
Mother's antenatal results recorded	70	22.9	207	67.6	29	9.5	306
Father's antenatal results recorded	70	22.9	207	67.6	29	9.5	306

*Includes FS,FSC, FS Other, FE, and F-only results.

Figure NB-13. Consistency of antenatal and neonatal screening results, 2016 to 2017



Timeliness of clinical referral

Newborn Blood Spot (NBS) Screening Programme standard 4 is for the sample to be taken on day 5 and in exceptional circumstances between day 5 and day 8 (day of birth is day 0). The thresholds for this standard are 90% as an acceptable level and 95% as an achievable level. Standard 5 (timely receipt of a sample in the newborn screening laboratory) requires 95% of samples to arrive in the laboratory within 3 working days of sample collection as an acceptable level, and 99% as an achievable level.

Table NB-10. Timeliness of reporting affected newborn results, 2016 to 2017

Laboratory	No. of screen positives*	Sample ≤8 days		Sample received by lab ≤3 days		Clinical referral by 28 days†		Time between sample taking and clinical referral (days)		
	n	n	%	n	%	n	%	Min	Max	Median
Bristol	3	3	100.0	2	66.7	2	100.0	7	20	13.5
Cambridge	3	3	100.0	3	100.0	3	100.0	12	17	14
GOS & CMH	107	101	94.4	80	74.8	102	98.1	4	19	9
Leeds	22	21	95.5	16	72.7	19	90.5	3	28	6
Liverpool	2	2	100.0	2	100.0	2	100.0	1	1	1
Manchester	14	14	100.0	11	78.6	14	100.0	3	16	9.5
Newcastle	0	-	-	-	-	-	-	-	-	-
Oxford	11	10	90.9	8	72.7	11	100.0	8	17	11
Portsmouth	4	4	100.0	4	100.0	4	100.0	14	17	15
Sheffield	10	10	100.0	8	80.0	9	90.0	10	29	14
South East Thames	59	56	94.9	49	83.1	55	96.5	4	22	8
South West Thames	29	28	96.6	22	75.9	29	100.0	5	26	12
West Midlands	42	41	97.6	33	78.6	41	97.6	5	24	12.5
England Total	306	293	95.8	238	77.8	291	95.1	1	29	10
Scotland	4	4	100.0	3	75.0	4	100.0	2	6	3
Wales	3	3	100.0	1	33.3	3	100.0	12	14	13
Northern Ireland	0	-	-	-	-	-	-	-	-	-
UK total	313	300	95.8	242	77.3	298	95.2	1	29	10

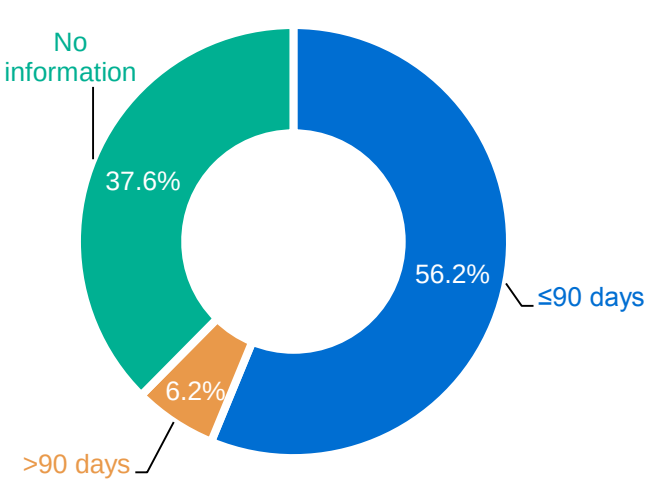
†Excludes 7 cases where where data was missing or the age at clinical referral given was smaller than the age at sample. These exclusions are reflected in the reported percentages in this column.

*This includes F-only cases, which are likely beta thalassaemia affected babies

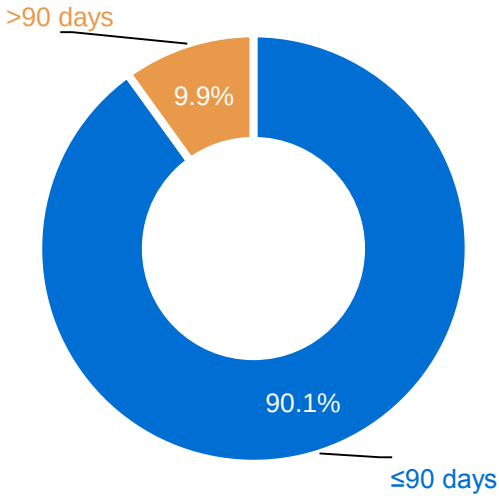
Age at first visit to a paediatrician

Standard 9 (timely receipt into haemoglobinopathy centres) sets an acceptable level of 90% and an achievable level of 95% of screen positive babies to attend their first clinical appointment by 90 days of age.

Figure NB-14. Age of screen positive babies at first visit to paediatrician at specialist health team or local health team, 2016 to 2017



These figures include F-only cases, which are likely to be beta thalassaemia affected babies.



Excludes 115 babies for whom no information was submitted; These figures include F-only cases, which are likely to be beta thalassaemia affected babies.

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 2 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).

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

The National Congenital Anomaly and Rare Disease Registration Service (NCARDRS), part of Public Health England, collects patient-identifiable data without the need for individual informed consent with permission from the National Information Governance Board under section 251 the NHS Health ACT 2006 and the authority of the Health Service (Control of Patient Information) Regulations 2002 (CAG ref: CAG 10-02(d)/2015).

NCARDRS collects named data for babies and children with sickle cell disorders or thalassaemia to assess:

- the health of affected babies or children
- timeliness of entry into care and start of their treatment
- their antenatal screening history

Between 1 April 2016 and 31 March 2017 there were 265 screen positive babies born in England (excluding those with insignificant diagnosis, death not ascribed to sickle cell, and known to have migrated or were born abroad). Of those:

- 225 babies have confirmed sickle cell of which 77.7 % (175/225) were seen in clinic by 3 months
    - 42/225 were not seen within 3 months
    - 8/225 are missing the date
- 26 babies have confirmed thalassemia of which 80.7% (21/26) were seen in clinic by 3 months
    - 1/26 was not seen by 3 months
    - 4/26 are missing the date
- 14 cases have an unconfirmed diagnosis of which 35.7% (5/14) were seen in clinic by 3 months
    - 2 were not seen by 3 months
    - 7/14 are missing the date

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

		Father's test result												
		Hb S	βThal	db thal	Hb Lepore	Hb D	Hb C	Hb E	Hb O-Arab	HPFH	High risk alpha0	Compound Hetero-zygous	Other	Not a carrier	Father result not available
Mother's test result	Hb S														
	βThal														
	db thal														
	Hb Lepore														
	Hb D														
	Hb C														
	Hb E														
	Hb O-Arab														
	HPFH														
	High risk alpha0														
	Compound Heterozygous														
	Egg donor/bone marrow transplant														