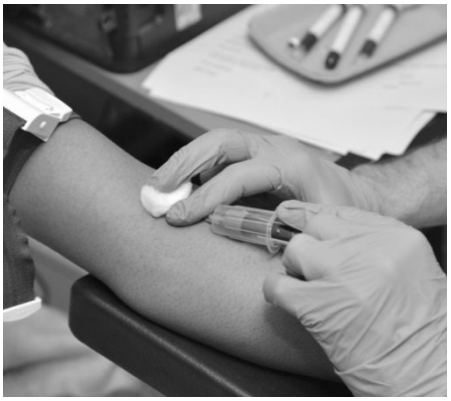
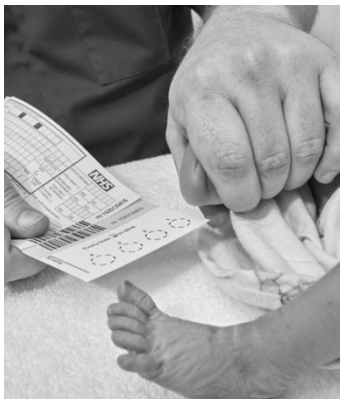
Public Health England

Protecting and improving the nation's health

NHS Sickle Cell and Thalassaemia Screening Programme

Data report 2017 to 2018


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.

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

About PHE screening

Screening identifies apparently healthy people who may be at increased risk of a disease or condition, enabling earlier treatment or informed decisions. National population screening programmes are implemented in the NHS on the advice of the UK National Screening Committee (UK NSC), which makes independent, evidence-based recommendations to ministers in the 4 UK countries. PHE advises the government and the NHS so England has safe, high quality screening programmes that reflect the best available evidence and the UK NSC recommendations. PHE also develops standards and provides specific services that help the local NHS implement and run screening services consistently across the country.

www.gov.uk/topic/population-screening-programmes
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Prepared by: NHS Sickle Cell and Thalassaemia Screening Programme
For queries relating to this document, please contact: phe.screeninghelpdesk@nhs.net

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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 2017 to 31 March 2018. The report contains data submitted by antenatal screening laboratories, prenatal diagnostic (PND) laboratories, newborn screening laboratories and newborn DNA testing laboratories. Detailed presentation of screening standards data is not included in this report, as this is contained within the annual antenatal standards data report.

In 2017 to 2018 approximately 650,000 pregnant women were screened for sickle cell, thalassaemia and other haemoglobin variants, and around 652,500 newborn babies were screened for sickle cell disease.

The proportion of women screening positive in antenatal screening in England remained stable in 2017 to 2018 compared to previous years. In screen positive women, the most common identified risk to pregnancy was a baby born with a possible sickle cell condition (52% of screen positive women), followed by a baby born with a possible beta thalassaemia condition (34% of screen positive women).

There were 366 PND tests performed in 2017 to 2018, a similar number to in 2016 to 2017. The proportion of PND tests performed at less than or equal to 12 weeks + 6 days gestation increased slightly in 2017 to 2018, but remained lower than between 2008 to 2009 and 2013 to 2014.

In England in 2017 to 2018, approximately 1 in 2500 babies screened for sickle cell disease were positive for significant conditions, and 1 in 80 were carriers. The rates of babies screening positive for significant conditions and carrier results has decreased since 2005 to 2006. These decreases have been predominantly driven by decreases in London. However, over the last three years in London, rates of babies screening positive for significant conditions has stabilised, whilst rates of carrier results continue to decrease. There is a continuing trend of increases in the rate of declines for newborn screening for sickle cell disease, with the rate in 2017 to 2018 being 2.84 per 1,000 babies screened. The reason for this increase is not clear, but it may be due to improved reporting of declines or declines for mover-in babies who are older babies and may have been tested elsewhere.

1. Introduction

1.1. About the NHS Sickle Cell and Thalassaemia (SCT) Screening Programme

Our aim is to provide a linked antenatal and newborn programme of high quality screening and care to:

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

1.2. Methodology

The SCT screening programme has 9 screening standards. Screening standards provide reliable and timely information about the quality of the screening programme, across the whole screening pathway. These standards are submitted by maternity services (standards 1,2, 3, 5 and 7), antenatal screening laboratories (standard 4) and the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) (standards 6, 8 and 9). The SCT screening standards are presented in the annual antenatal standards report, alongside data tables which provide local and regional breakdowns. In addition to the SCT screening standards, some standards within the Newborn blood spot (NBS) screening programme are also relevant to the SCT screening programme. These include standards relating to coverage and blood spot test processes, such as timely sample collection. The NBS screening standards are presented in annual reports. As the screening standards are presented in these reports, the focus of this report is data that is separate to the screening standards. The collection of this data is described below.

Timely annual data returns are required from all antenatal and newborn screening laboratories in line 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.

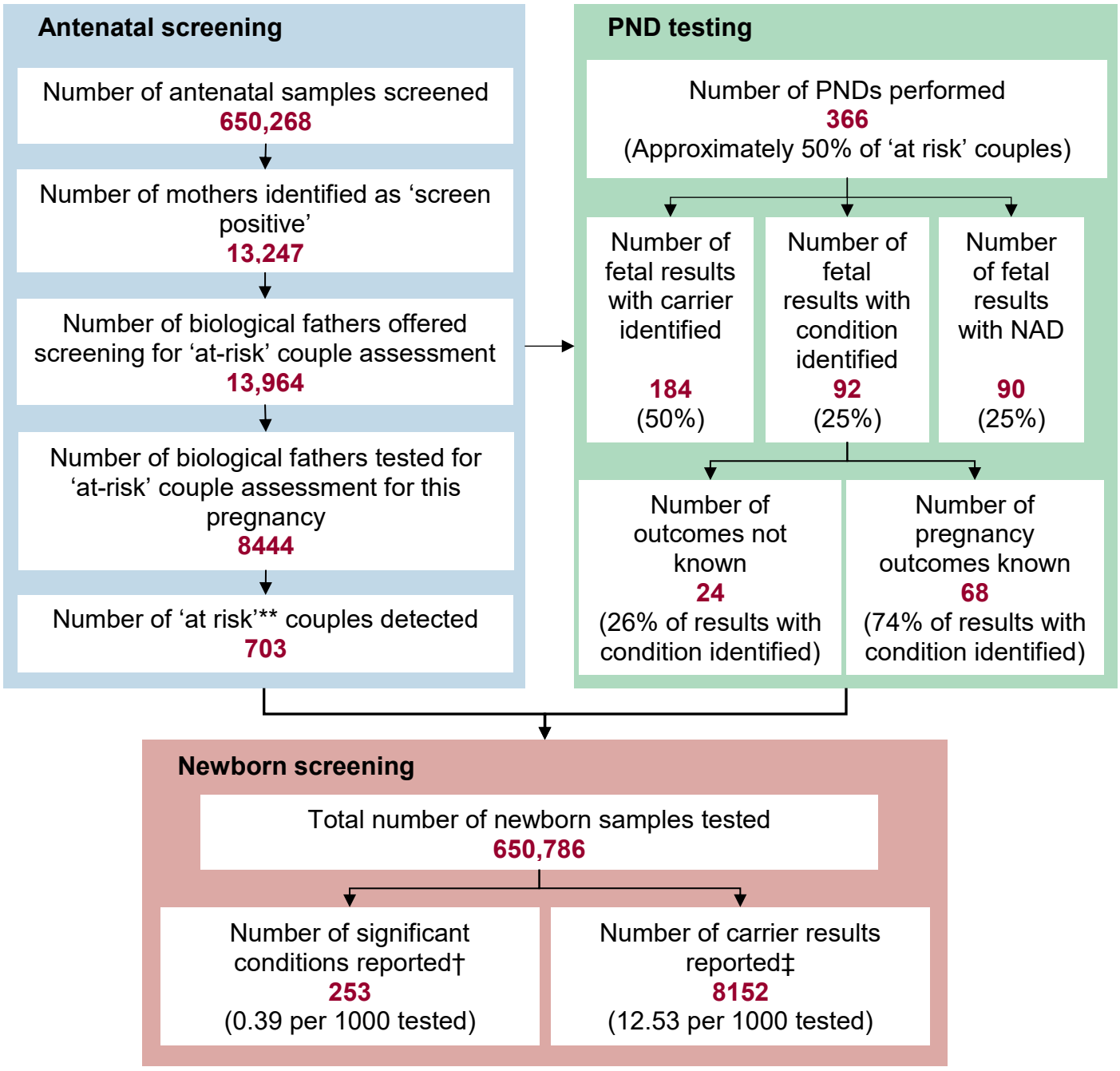
While the screening programme covers only England, screening data is provided by the newborn laboratories in Scotland, Wales and Northern Ireland. However, these countries are not included in the ethnicity figures, as Scotland uses different ethnic categories, and Wales and Northern Ireland do not routinely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

NCARDRS provides a de-personalised dataset of PND test data to the programme. This data is collected from PND laboratories and verified by NCARDRS.

The 2 newborn DNA testing laboratories in England also submit aggregated data to the programme annually.

Data is presented by financial year (1 April to 31 March) unless stated otherwise. The year ‘2017 to 2018’, for example, refers to the financial year ‘1 April 2017 to 31 March 2018’.

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. It is therefore not recommended that rates are calculated based on the above figures.
*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 or higher of the fetus being affected by a serious haemoglobinopathy. This number excludes pregnancies at risk of less serious disorders and cases where the father was not available for testing.

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

3.1. Antenatal coverage

Coverage for antenatal screening is calculated as the number of women tested as a proportion of the number of women eligible for screening. This is a screening standard (SCT-S01). The England performance for antenatal screening coverage in 2017 to 2018 was 99.6%. More detailed presentation of antenatal coverage data can be found in the annual antenatal standards data report.

3.2. Antenatal screening laboratory data quality and completeness

Out of 144 expected laboratory returns, there were 133 received for 2017 to 2018 (92.4% response rate). The expected data returns are based on the maternity provider served by the screening laboratory, and one screening laboratory may serve more than one provider.

Not all laboratories were able to submit data for all fields that were requested. Data fields were excluded where providers were unable to submit data. Where exclusions were made, these are identified below the relevant charts and tables. Some laboratories are unable to match the mother results to biological 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.

3.3. Numbers screened and detected in antenatal screening

In 2017 to 2018, after data exclusions were applied, there were 640,389 antenatal care bookings reported in the included returns, of which 12,778 were identified as screen positive (approximately 1 in 50 women screened). Of these screen positive women, 698 pregnancies were identified as being ‘at risk’ of the baby inheriting a sickle cell or thalassaemia condition, (approximately 1 in 18 screen positive women) based on the results of both parents. These ‘at risk’ pregnancies are those represented by the dark orange boxes in the breakdown table in Appendix A. We would expect the number of ‘at risk’ couples to be approximately 4 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 result, 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 excluded data and non-returns, couples where the baby’s biological father’s status is unknown, or where parents declined antenatal screening.

Table 1: Numbers screened and detected, England, 2017 to 2018

Region	Returns included/ expected^	Antenatal screening samples	Screen positive (Scr+)		At risk couples	
		n	n	% of samples	n	% of Scr+
London	21/26	118,904	5,538	4.66	334	6.03
Midlands and East	36/39	199,254	3,402	1.71	169	4.97
North	35/41	174,185	2,236	1.28	114	5.10
South	35/38	148,046	1,602	1.08	81	5.06
England total	**127/144**	**640,389**	**12,778**	**2.00**	**698**	**5.46**

Table 2: Numbers screened and detected, high prevalence areas, England, 2017 to 2018

Region	Returns included/ expected^	Antenatal screening samples	Screen positive (Scr+)		At risk couples	
		n	n	% of samples	n	% of Scr+
London	21/26	118,904	5,538	4.66	334	6.03
Midlands and East	17/18	109,079	2,691	2.47	145	5.39
North	12/13	86,721	1,645	1.90	80	4.86
South	7/9	42,129	793	1.88	47	5.93
England total	**57/66**	**356,833**	**10,667**	**2.99**	**606**	**5.68**

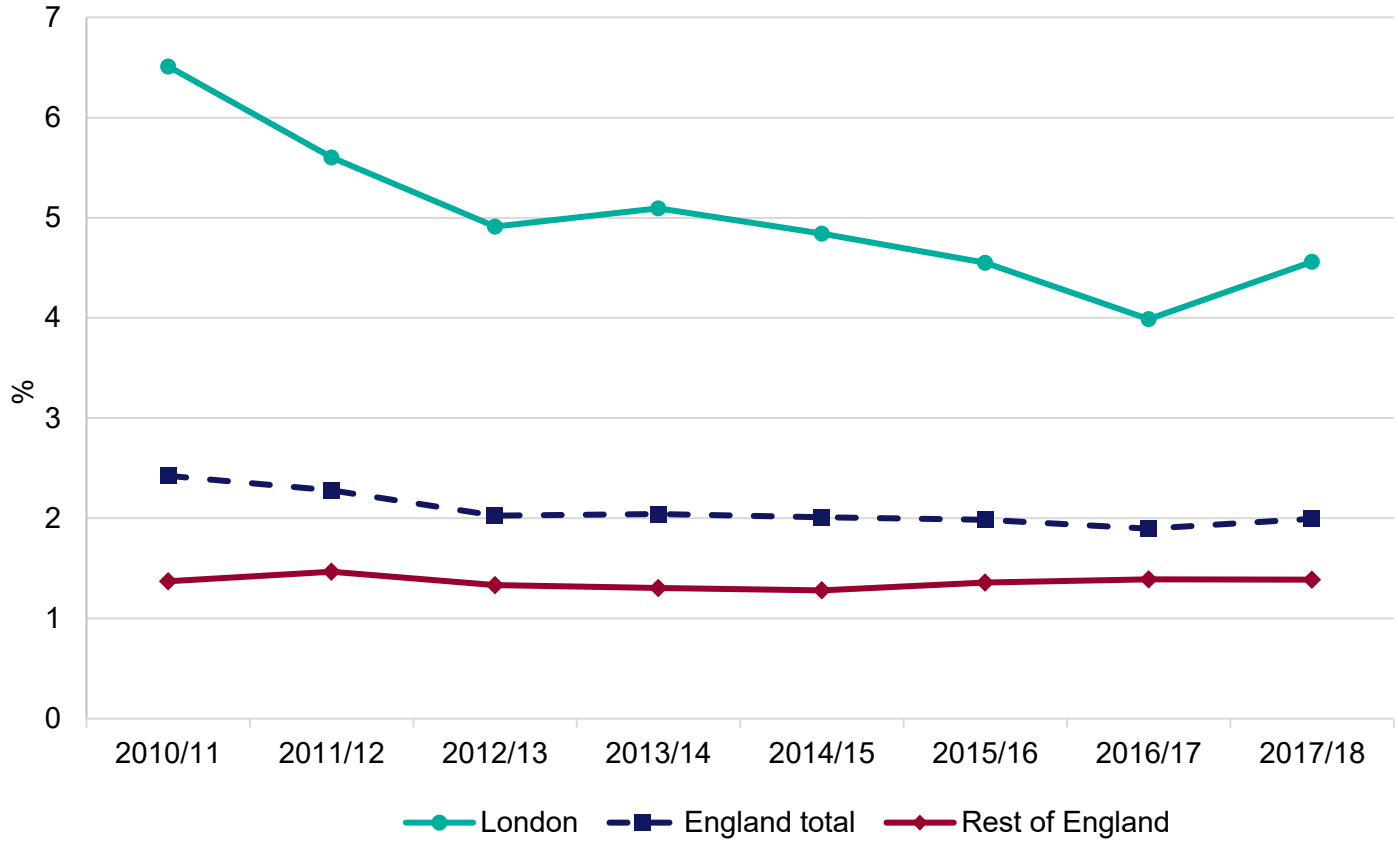
Table 3: Numbers screened and detected, low prevalence areas, England, 2017 to 2018

Region	Returns included/ expected^	Antenatal screening samples	Screen positive (Scr+)		At risk couples	
		n	n	% of samples	n	% of Scr+
London	0/0	-	-	-	-	-
Midlands and East	19/21	90,175	711	0.79	24	3.38
North	23/28	87,464	591	0.68	34	5.75
South	28/29	105,917	809	0.76	34	4.20
England total	**70/78**	**283,556**	**2,111**	**0.74**	**92**	**4.36**

^Expected returns are not included when no data return was received or when exclusions to received data have been made. Received data returns are only included above if data for number of samples, number of screen positives and number of at risk couples were all accepted.

In London, there was a small increase in the percentage of antenatal screening samples that were screen positive in 2017 to 2018 compared to the previous year (Figure 1). This followed a period of consistent decreases in the percentage of screen positives in London since 2010 to 2011. In the rest of England, the percentage of screen positives has remained stable over recent years.

Figure 1: Screen positive women as a percentage of antenatal screening samples received by laboratory, England, 2017 to 2018



Returns not included as data not received or excluded: 2010/11: 3; 2011/12: 2; 2012/13: 3; 2013/14: 2; 2014/15: 4; 2015/16: 6; 2016/17: 10; 2017/18: 14.

3.4. The family origin questionnaire

Screening standard 3 (SCT-S03) relates to the proportion of antenatal samples that are submitted to the laboratory accompanied by a completed Family Origin Questionnaire (FOQ). The England performance for this standard in 2017 to 2018 was 97.6%. More detailed presentation of this data can be found in the annual antenatal standards data report.

3.5. Declined antenatal screening tests for sickle cell and thalassaemia

Personal informed choice is an important element of population screening, and as such, screening tests for sickle cell and thalassaemia may be declined for various reasons. Table 4 shows, after data exclusions, the number of women who declined antenatal screening by region and for the whole of England as a proportion of booking blood samples tested. National rates of declines decreased in 2017 to 2018 following a small increase seen in 2016 to 2017. It is important to note that there were a higher number of data exclusions in 2017 to 2018 compared to previous years, which may have affected comparisons between years. Whilst antenatal screening laboratories have reported the data shown in table 4, the number of declines is also reported by maternity services in data returns for the coverage standard, SCT-S01. Differences between declines reported by laboratories and maternity providers have been noted, and the programme is currently conducting a piece of work to investigate this further and determine how declines will be published in future.

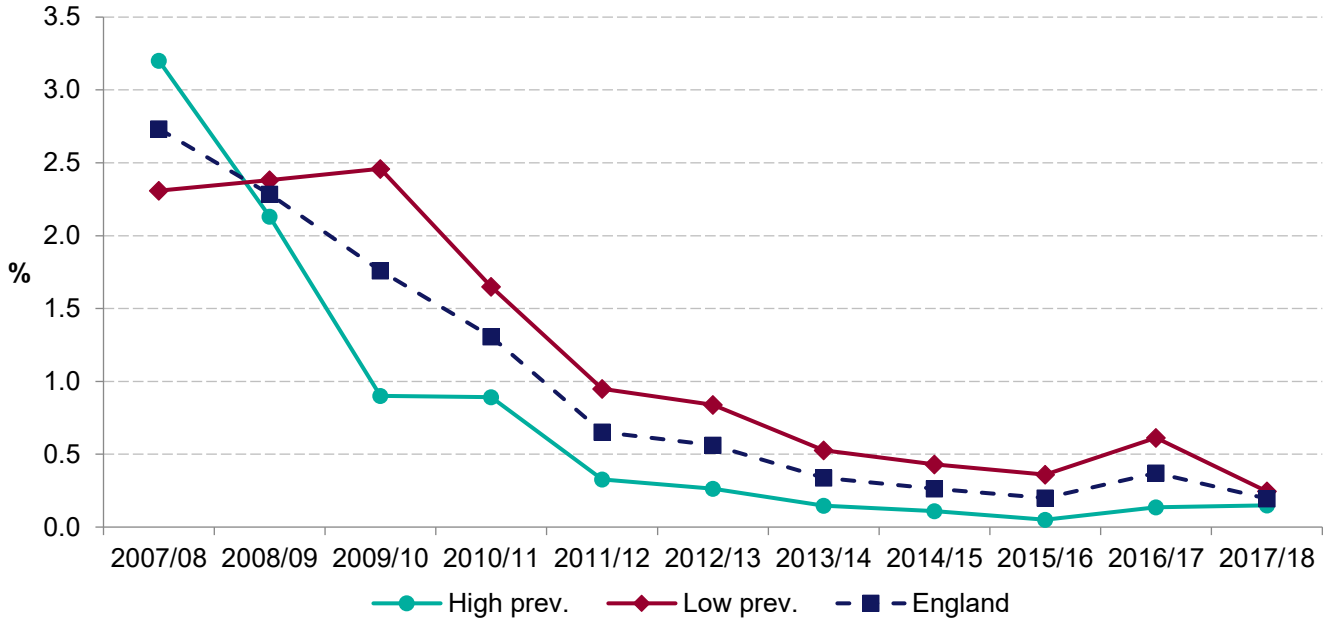
Table 4: Declined antenatal screening by region, England, 2015 to 2016 – 2017 to 2018

Region	2015 to 2016			2016 to 2017			2017 to 2018		
	Antenatal screening samples	Declines	% of samples	Antenatal screening samples	Declines	% of samples	Antenatal screening samples	Declines	% of samples
London	124,816	28	0.02	102,338	37	0.04	82,840	44	0.05
Midlands and East	206,927	404	0.20	204,425	384	0.19	191,000	252	0.13
North	182,030	254	0.14	171,791	1,029	0.60	162,541	571	0.35
South	152,163	648	0.43	144,806	857	0.59	134,773	249	0.18
England total	**665,936**	**1,334**	**0.20**	**623,360**	**2,307**	**0.37**	**571,154**	**1,116**	**0.20**

Returns not included as data not received or excluded: 2015/16: 13; 2016/17: 8; 2017/18: 29

Figure 2 shows the change over time in rates of declined antenatal screening since 2007 to 2008, broken down by high and low prevalence areas. Rates have followed a similar pattern, with year on year decreases, apart from in 2016 to 2017 where a small increase was seen. In 2017 to 2018 rates of declines were similar in high and low prevalence areas. This follows a period since 2008 to 2009 where rates of declines were consistently lower in high prevalence areas.

Figure 2: Trends in declined antenatal screening as a percentage of antenatal screening samples received, England, by prevalence, 2007 to 2008 – 2017 to 2018



Returns not included as data not received or excluded: 2007/08: 40; 2008/09: 46; 2009/10: 42; 2010/11: 18; 2011/12: 15; 2012/13: 16; 2013/14: 15; 2014/15: 15; 2015/16: 13; 2016/17: 18; 2017/18: 29

3.6. 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 more difficult to accurately assess the baby's risk of inheriting sickle cell and thalassaemia. In this situation, women are counselled and offered PND. 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 biological father specimens received).

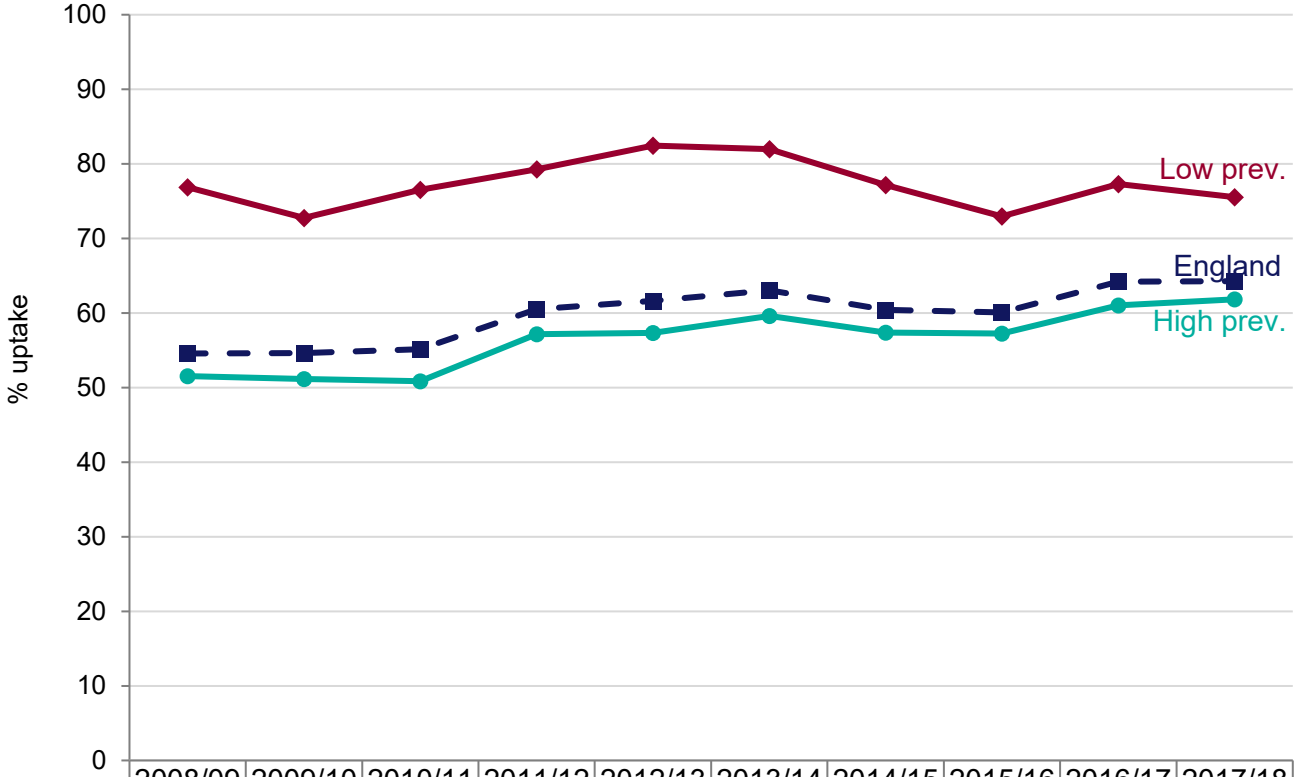
The uptake of biological father testing was stable between 2016 to 2017 and 2017 to 2018, following a small increase in uptake in both high and low prevalence areas in 2016 to 2017.

Table 5: Uptake of testing of the baby's biological father, England, 2015 to 2016 – 2017 to 2018

Region	2015 to 2016			2016 to 2017			2017 to 2018		
	Father samples requested	Father samples available	% uptake	Father samples requested	Father samples available	% uptake	Father samples requested	Father samples available	% uptake
London	6,340	3,205	50.6	5,291	3,044	57.5	5,361	2,947	55.0
Midlands and East	3,714	2,422	65.2	3,747	2,494	66.6	3,580	2,449	68.4
North	2,333	1,639	70.3	2,352	1,623	69.0	2,375	1,681	70.8
South	1,924	1,329	69.1	1,866	1,352	72.5	1,823	1,367	75.0
England total	**14,311**	**8,595**	**60.1**	**13,256**	**8,513**	**64.2**	**13,139**	**8,444**	**64.3**

Returns not included as data not received or excluded: 2015/16: 7; 2016/17: 11, 2017/18: 18
Father samples available includes results known from historical records in line with guidance in the Handbook for Laboratories.

Figure 3: Trends in uptake of testing of the baby's biological father, England, by prevalence, 2008 to 2009 – 2017 to 2018



	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14	2014/15	2015/16	2016/17	2017/18
High prev.	51.5	51.1	50.9	57.1	57.3	59.6	57.4	57.2	61.0	61.8
Low prev.	76.9	72.8	76.5	79.3	82.4	82.0	77.2	73.0	77.3	75.5
England	54.6	54.6	55.1	60.5	61.6	63.0	60.4	60.1	64.2	64.3

Returns not included as data not received or excluded: 2007/08: 23; 2008/09: 15; 2009/10: 22; 2010/11: 7; 2011/12: 9; 2012/13: 10; 2013/14: 11; 2014/15: 11; 2015/16: 7; 2016/17: 11; 2017/18: 18
% uptake will include father results known from historical records in line with guidance in the Handbook for Laboratories.

‘At risk’ couples are identified based on results from the baby’s biological parents. Breakdown data is requested on both biological mother and father results to identify the specific risk of an affected pregnancy. This information also allows us to separate 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 the results to the mother’s results.

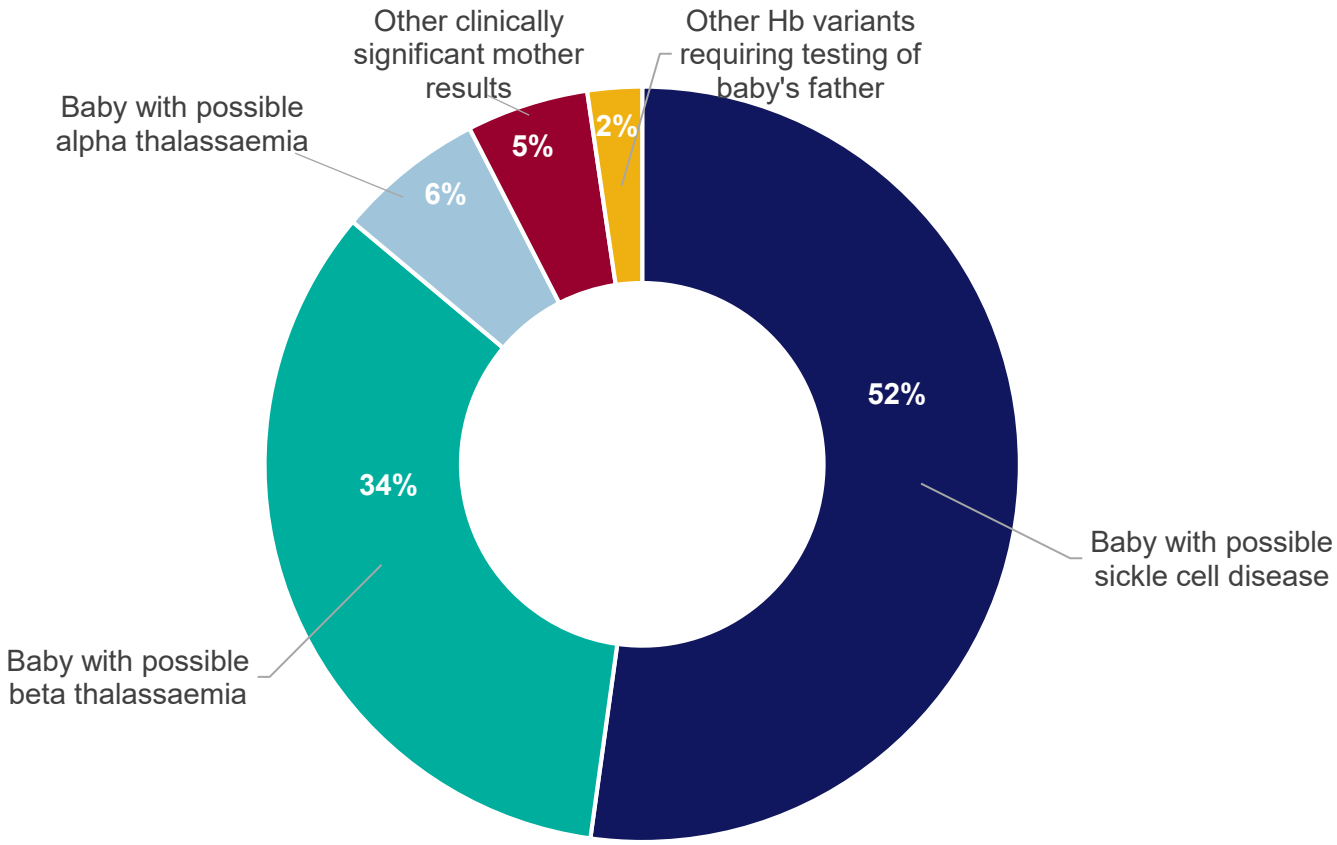
‘High risk’ pregnancies are those where there is a 1 in 4 chance or higher of the fetus having a serious haemoglobinopathy, and are represented by the dark orange boxes in the breakdown table in Appendix A. The light orange boxes represent low risk pregnancies, which are at risk of less serious haemoglobin disorders, and the white boxes represent minimal risk pregnancies. Women with beta thalassaemia results are included in the ‘baby with possible beta thalassaemia’ group in the table. However, HbS/beta thalassaemia causes sickle cell disease, and these cases are included in the ‘high risk’ category.

Table 6: Breakdown of pregnancy risk for screen positive women, England, 2017 to 2018

	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
Baby with possible sickle cell disease	Hb S	452	109	2,453	2,167	5,181	6,862	10.88	52.2
	Hb D	4	41	468	145	658			
	Hb C	55	54	462	433	1,004			
	Hb O-Arab	1	1	14	3	19			
Baby with possible beta thalassae-mia	β thalassae-mia	126	70	2,476	814	3,486	4,457	7.01	33.9
	δβ thalassae-mia	0	5	56	23	84			
	Hb E	5	38	655	161	859			
	Hb Lepore	0	2	21	5	28			
Baby with possible alpha thalassae-mia	High risk alpha0	29	23	484	298	834	834	1.27	6.3
Other clinically significant mother results	HPFH/ Compound heterozygo-us /donor egg/bone marrow transplant	15	27	414	231	687	687	1.03	5.2
Other Hb variants requiring testing of baby's father		-	13	226	68	307	307	0.49	2.3
Totals		687	383	7,729	4,348	13,147	13,147	20.68	100.0

Note: 'Mother's screening results' include both cases where the mother is a carrier and where she has a condition.
The above includes data from 126 included returns out of the 144 expected returns. All included laboratories provided a complete breakdown for all screen positive women.
Only data returns where the number of samples and a breakdown of results are both available have been used to calculate the rate.

Figure 4: Screen positive women, broken down by risk to the pregnancy, England, 2017 to 2018



3.7. Offer of screening early in pregnancy

Early antenatal screening for sickle cell and thalassaemia is important as this will maximise the opportunity for parents to make a personal informed choice. Where parents choose to have PND, advanced gestational age may limit reproductive choices.

Antenatal screening

Screening standard 2 (SCT-S02) collects data on the proportion of pregnant women having antenatal sickle cell and thalassaemia screening for whom a result is available ≤ 10 weeks + 0 days gestation. The England performance for this standard in 2017 to 2018 was 55.9%.

Prenatal diagnosis (PND)

Screening standard 6 (SCT-S06) collects data on the proportion of PND tests performed ≤12 weeks + 6 days gestation. The performance for England in 2017 to 2018 for this standard, after exclusions were made due to missing data, was 41.6%.

More detailed presentation of screening standards 2 and 6 can be found in the annual antenatal standards data report.

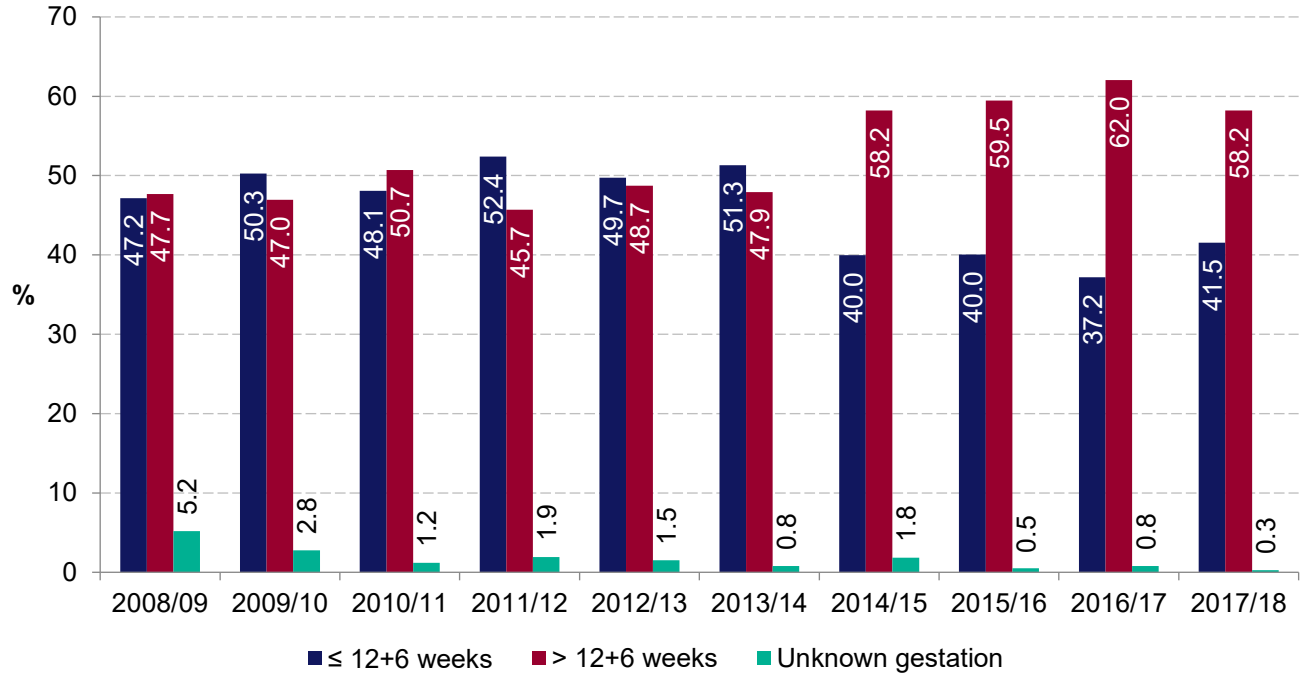
Additional data related to PND tests over time are presented below. This demonstrates that the proportion of PND tests performed ≤ 12 + 6 weeks gestation, increased slightly in 2017 to 2018 compared to the previous year, but remains lower than in the years up to 2013 to 2014.

Table 7: Gestation at PND test, England, 2014 to 2015 – 2016 to 2017

Gestation at PND test	2015/16		2016/17		2017/18	
	n	%	n	%	n	%
<12+6 weeks	163	40.0	139	37.2	152	41.5
13+0 - 14+6 weeks	106	26.0	110	29.4	88	24.0
≥15+0 weeks	136	33.4	122	32.6	125	34.2
Unknown gestation	2	0.5	3	0.8	1	0.3
Total	407	100.0	374	100.0	366	100.0

Gestation relates to the gestation on the day of fetal sampling.

Figure 5: Proportion of PND tests performed by gestation, England, 2008 to 2009 – 2017 to 2018



3.8. Numbers tested and detected in prenatal diagnostic testing

Figure 6: Number of PND tests performed, by laboratory, England, 2008 to 2009 – 2017 to 2018

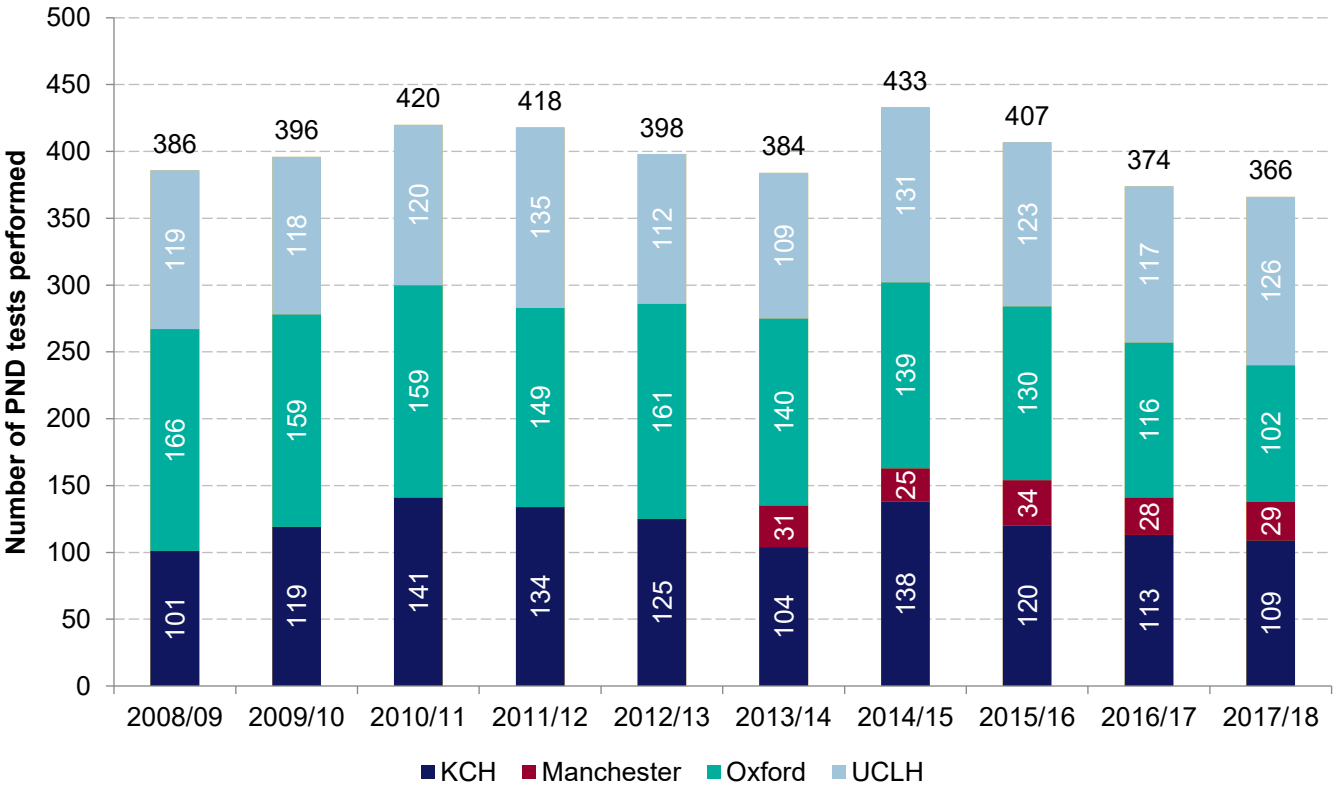


Table 8: Breakdown of PND fetal results by condition, England, 2015 to 2016 – 2017 to 2018

Fetal result	PND result	2015/16	2016/17	2017/18
Haemoglobinopathy	Hb SS	61	55	55
	Hb SC	15	8	12
	Hb S/Beta thalassaemia	4	2	4
	Hb S + other	0	0	2
	Alpha Thalassaemia	2	3	0
	Beta Thalassaemia	22	10	19
	Other	0	2	0
Carrier	Hb AS	125	146	130
	Hb AC	7	3	13
	Alpha thalassaemia carrier	4	2	6
	Beta thalassaemia carrier	42	36	32
	Other Hb carrier	1	3	3
No abnormality detected (NAD)	**PND risk**			
	Risk for Sickle Cell	91	77	65
	Risk for Thalassaemia	26	20	12
	Risk for Sickle Cell/Beta thalassaemia	6	7	12
	Risk not known	0	0	1
Inconclusive/result not known	All risks	1	0	0
Total		**407**	**374**	**366**

'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 9: Number of PND tests by mother's family origins, England, 2015 to 2016 – 2017 to 2018

Mother's family origin	2015/16		2016/17		2017/18	
	n	%	n	%	n	%
African / African-Caribbean	291	71.5	273	73.0	254	69.4
South Asian	46	11.3	33	8.8	26	7.1
South East Asian / Other Asian	11	2.7	9	2.4	9	2.5
Other Non-European	15	3.7	14	3.7	10	2.7
Southern and other European	20	4.9	16	4.3	14	3.8
Mixed	5	1.2	5	1.3	7	1.9
Northern European/United Kingdom (White)	1	0.2	1	0.3	3	0.8
Not Known	18	4.4	23	6.1	43	11.7
Total	**407**	**100.0**	**374**	**100.0**	**366**	**100.0**

Mother's family origin is recorded in data received from PND laboratories. Categories have then been assigned to mother's family origin based upon the categories used in the FOQ.

The data received from PND laboratories do not currently allow African and African-Caribbean family origins to be accurately separated. The programme will work with laboratories to ensure that African and African-Caribbean family origins can be reported separately in future.

3.10. Pregnancy outcomes

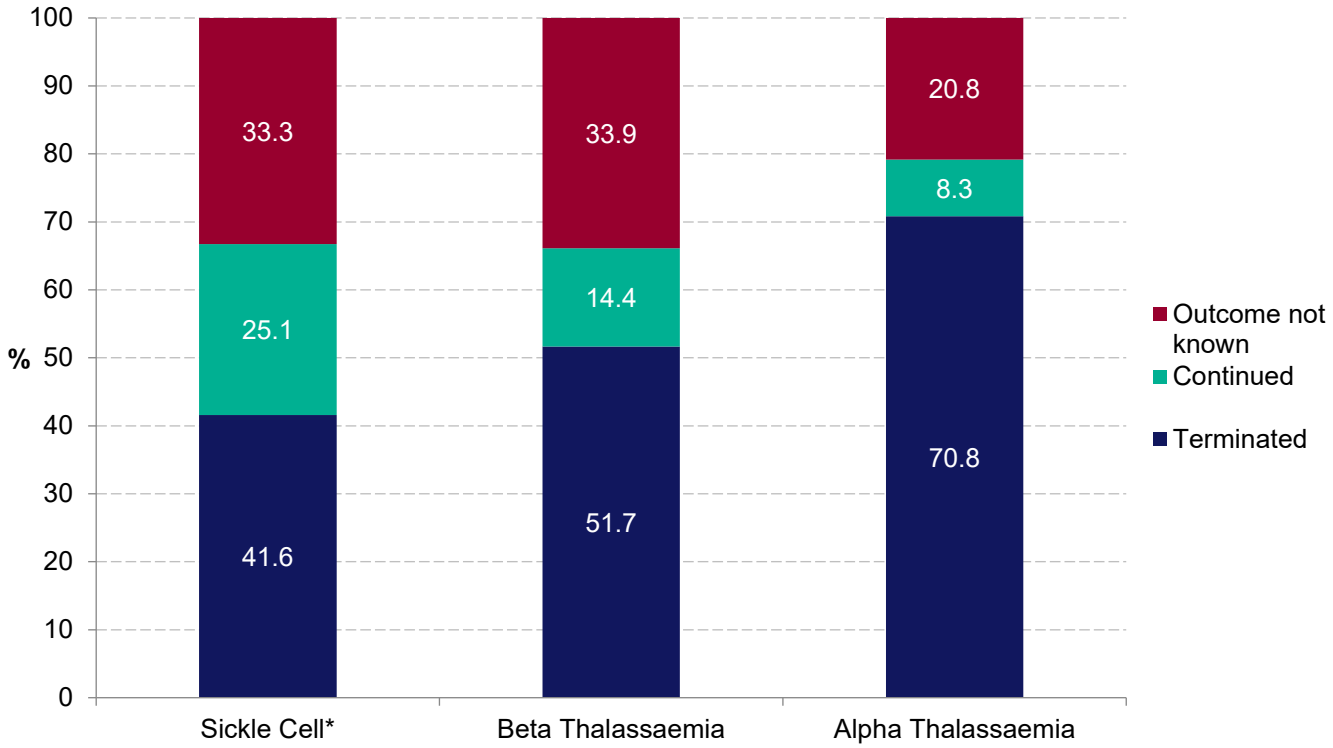
One of the aims of the SCT screening programme is to ensure personal informed choice. The screening programme collects data on pregnancy outcomes following PND testing to assess what choices women and couples make following the test. There are gaps in the data, with the pregnancy outcome not known for 26% of affected pregnancies in the 2017 to 2018 dataset.

Table 10: Outcomes for pregnancies with haemoglobinopathy fetal results at PND, England, 2015 to 2016 – 2017 to 2018

Condition	Pregnancy outcome	2015/16 % of total identified with condition	2016/17 % of total identified with condition	2017/18 % of total identified with condition
Sickle Cell*	Continued	17.5	33.9	32.4
	Terminated	46.3	43.6	36.6
	Not known	36.3	22.6	31.0
Beta Thalassaemia	Continued	22.7	20.0	31.6
	Terminated	54.6	70.0	57.9
	Not known	22.7	10.0	10.5
Alpha Thalassaemia	Continued	0.0	0.0	-
	Terminated	100.0	100.0	-
	Not known	0.0	0.0	-

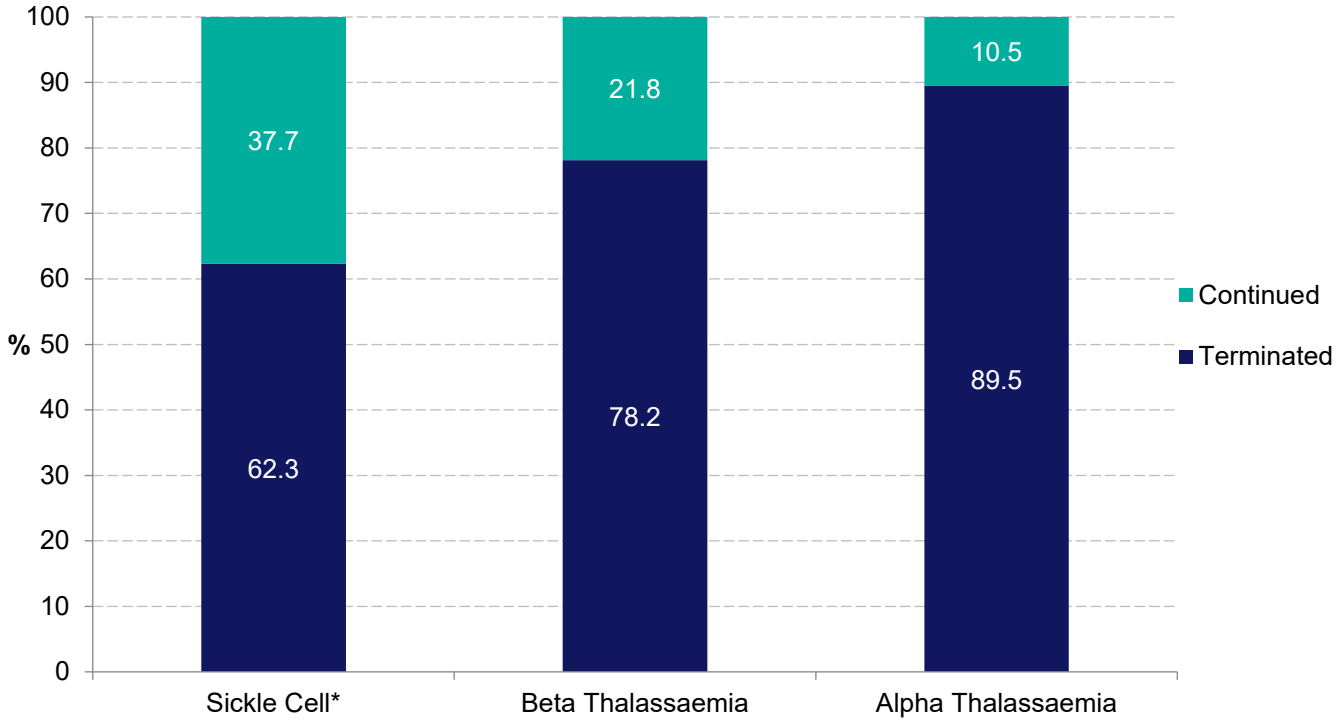
*The "Sickle Cell" category includes HbSS, HbSC, HbS/beta thalassaemia, and HbS+other variant requiring clinical follow-up.
Other haemoglobin variants are not presented and miscarriage outcomes have been excluded.
Please note that alpha thalassaemia percentages are based on small numbers and should be interpreted with caution.

Figure 7: Outcomes for pregnancies with haemoglobinopathy diagnosis at PND, England, 2008 to 2009 – 2017 to 2018



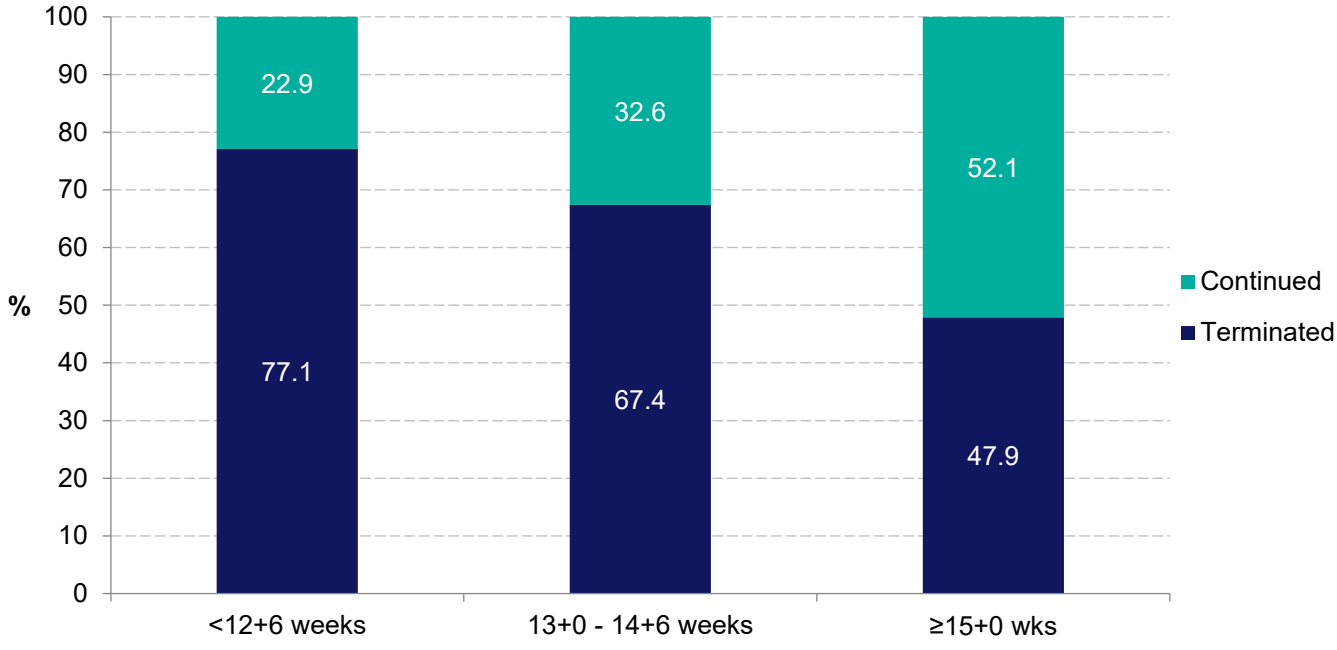
*The "Sickle Cell" category includes HbSS, HbSC, HbS/beta thalassaemia, and HbS+other variant requiring clinical follow-up.
Excludes miscarriage outcomes due to small numbers.

Figure 8: Outcomes for pregnancies with haemoglobinopathy diagnosis at PND (known outcomes only), England, 2008 to 2009 – 2017 to 2018



*The "Sickle Cell" category includes HbSS, HbSC, HbS/beta thalassaemia, and HbS+other variant requiring clinical follow-up.
Excludes miscarriage outcomes, and 319 cases where pregnancy outcome was not known.

Figure 9: Outcomes for pregnancies with haemoglobinopathy diagnosis at PND, by gestation at PND (known outcomes only), England, 2008 to 2009 – 2017 to 2018



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

4. Newborn screening for sickle cell disease

4.1. Newborn coverage

Babies are screened for sickle cell disease (SCD) as part of the NHS Newborn Blood Spot (NBS) screening programme. Newborn screening coverage is collected by the NBS programme through screening standard 1a and 1b (NBS-S01a and NBS-S01b).

NBS-S01a relates to the proportion of babies registered within the clinical commissioning group (CCG) both at birth and on the last day of the reporting period who are eligible for NBS screening and have a conclusive result recorded on the child health information service system (CHISS) at less than or equal to 17 days of age. The performance for this standard, coverage of newborn screening (born and resident population), in England in 2017 to 2018 was 96.7%.

NBS-S01b relates to the proportion of babies eligible for NBS screening who changed responsible CCG in the reporting period, or have moved in from another UK country or abroad and have a conclusive result recorded on the CHISS at less than or equal to 21 calendar days of notifying the child health record department (CHRD) of movement in. The performance for this standard, coverage of newborn screening (movers-in), in England in 2017 to 2018 was 90.0%.

The NBS screening programme annual reports present this data in more detail.

4.2. 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 11: Numbers and rates of significant condition and carrier screening results, newborn blood spot screening for sickle cell disease, United Kingdom, 2017 to 2018

	Significant conditions			Carriers			
Region	n	Rate/1000	1 in x	n	Rate/1000	1 in x	Babies tested
London	139	1.07	935	3,673	28.26	35	129,967
Midlands and East	56	0.29	3,417	2,185	11.42	88	191,340
North	35	0.21	4,833	1,165	6.89	145	169,140
South	14	0.10	10,447	977	6.68	150	146,255
Unknown region	9	0.64	1,565	152	10.79	93	14,084
England total	**253**	**0.39**	**2,572**	**8,152**	**12.53**	**80**	**650,786**
Scotland	2	0.04	26,278	229	4.36	229	52,555
Wales†	2	0.06	15,966	-	-	-	31,932
Northern Ireland	0	-	-	38	1.66	603	22,923
UK total	**257**	**0.34**	**2,950**	**8,419**	**11.10**	**90**	**758,196**

Region is based upon the reported maternity provider, CCG or CHRD of the baby. The geography used differs according to the submitting laboratory.

Babies identified to be from Scotland by newborn screening laboratories in England have been included within the Scotland totals.

Babies identified to be from Isle of Man or overseas by newborn screening laboratories have not been included in the above.

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

Data is based on samples received into newborn screening laboratories in 2017 to 2018, apart from for Portsmouth laboratory where data is based on samples from babies born in 2017 to 2018, and for Wales, where data is based on babies tested from the cohort of eligible babies born in 2017 to 2018. For two laboratories in England, data provided is based on samples received rather than babies tested.

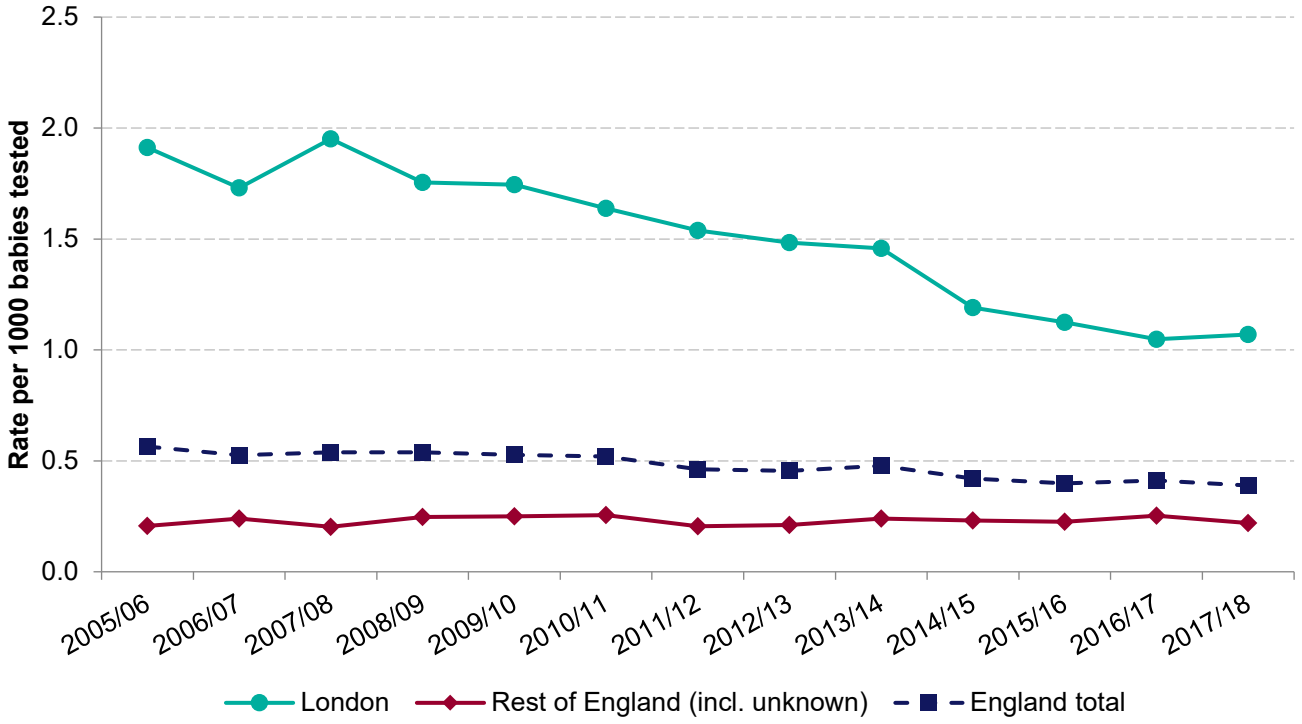
Table 12: Breakdown of newborn screening results, newborn blood spot screening for sickle cell disease, England, 2017 to 2018

Region	Significant Conditions					Carriers					Post trans-fused	De-clined	Babies tested	Babies offered screening (tested + declined)
	FS	FSC	FS-Other	FE	F Only	FAS	FAC	FAD	FAE	Other				
London	92	37	4	6	9	2,510	568	209	382	4	190	445	129,967	130,412
Midlands and East	38	12	2	4	9	1,352	334	250	246	3	267	621	191,340	191,961
North	23	2	7	3	2	707	128	146	184	0	304	281	169,140	169,421
South	9	4	1	0	3	611	132	103	118	13	94	252	146,255	146,507
Unknown region	5	2	2	0	2	111	15	13	10	3	96	257	14,084	14,341
England total	167	57	16	13	25	5,291	1,177	721	940	23	951	1,856	650,786	652,642

Region is based upon the reported maternity provider, CCG or CHRD of the baby. The geography used differs according to the submitting laboratory.
Babies identified to be from Isle of Man or overseas by newborn screening laboratories have not been included in the above.
Data is based on samples received into newborn screening laboratories in 2017 to 2018, apart from for Portsmouth laboratory where data is based on samples from babies born in 2017 to 2018. For two laboratories in England, data provided is based upon samples received rather than babies tested.

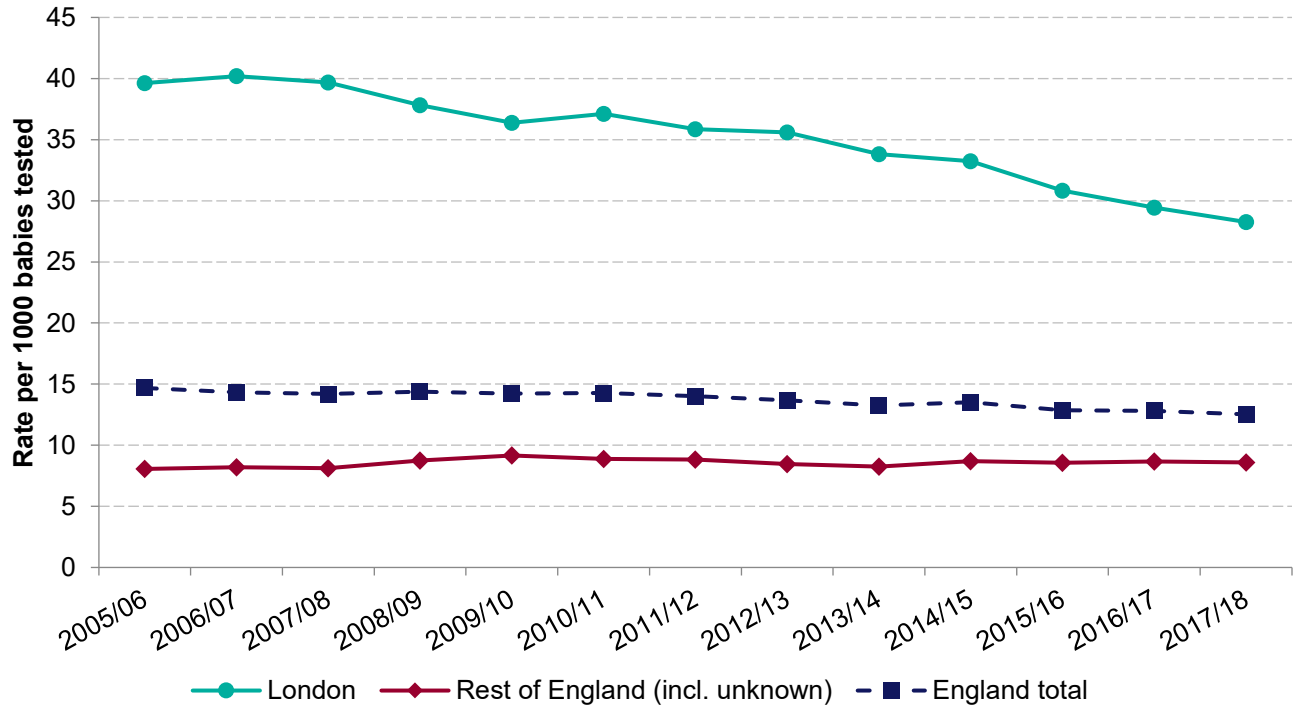
The rates of babies identified with a significant condition in England have decreased from approximately 0.6 per 1000 babies tested in 2005 to 2006 to 0.4 per 1000 babies tested in 2017 to 2018 (Figure 10). This decrease has been driven by decreases in London, particularly in the years up to 2014 to 2015. Over the past 3 years the rate of babies identified with a significant condition in London has stabilised, and this has also been reflected in the England rate. Outside of London, the rates of babies identified with a significant condition has remained stable since 2005 to 2006. The trends in rates of babies identified with carrier results have followed a similar pattern to rates of significant conditions, although rates in London have continued to decrease in recent years (Figure 11).

Figure 10: Trends in rates of babies identified with a significant condition, 2005 to 2006 – 2017 to 2018



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

Figure 11: Trends in rates of babies with carrier results, 2005 to 2006 – 2017 to 2018



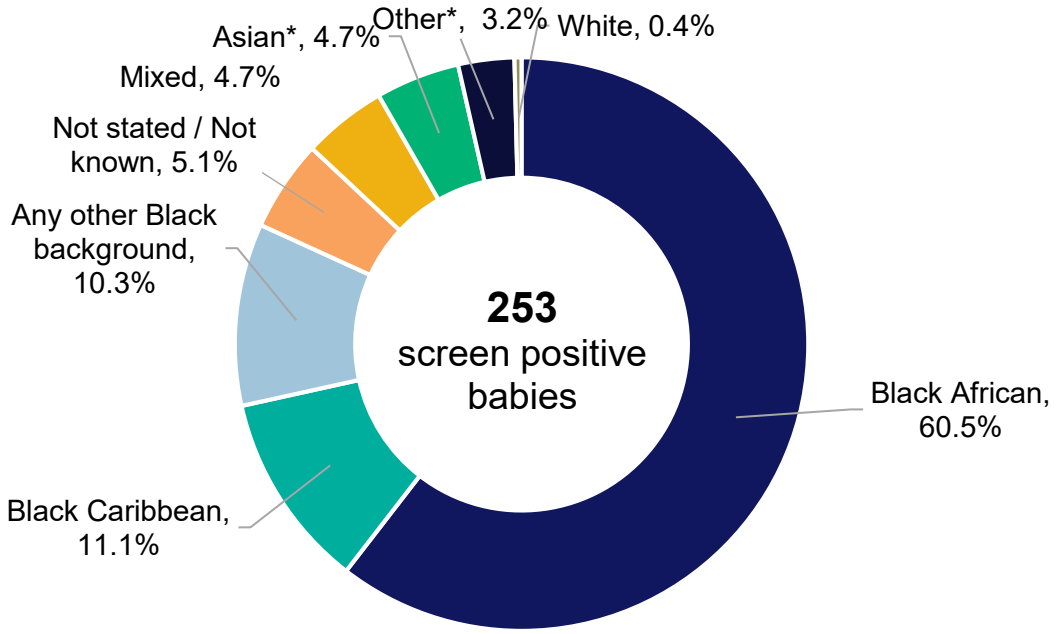
Carrier results comprise FAS, FAC, FAD, FAE and other carriers

4.3. Results by ethnicity

Table 13: Numbers and rates of significant conditions and carrier screening results by ethnic category, 2017 to 2018: English laboratories

Ethnic category	Significant conditions			Carriers			No. of babies tested
	n	Rate/1000	1 in x	n	Rate/1000	1 in x	
White	1	0.00	464,631	735	1.58	632	464,631
Mixed	12	0.29	3,414	1,559	38.05	26	40,970
Asian*	12	0.17	5,984	1,235	17.20	58	71,808
Black Caribbean	28	4.88	205	670	116.87	9	5,733
Black African	153	6.71	149	3,050	133.85	7	22,786
Any other Black background	26	6.73	149	422	109.21	9	3,864
Other*	8	0.43	2,344	229	12.21	82	18,749
Not stated / Not known	13	0.56	1,779	253	10.94	91	23,133
England total	**253**	**0.39**	**2,576**	**8,153**	**12.51**	**80**	**651,674**

Figure 12: Breakdown of screen positive babies by ethnic category, percentage of all babies that screen positive for significant conditions, 2017 to 2018: English laboratories



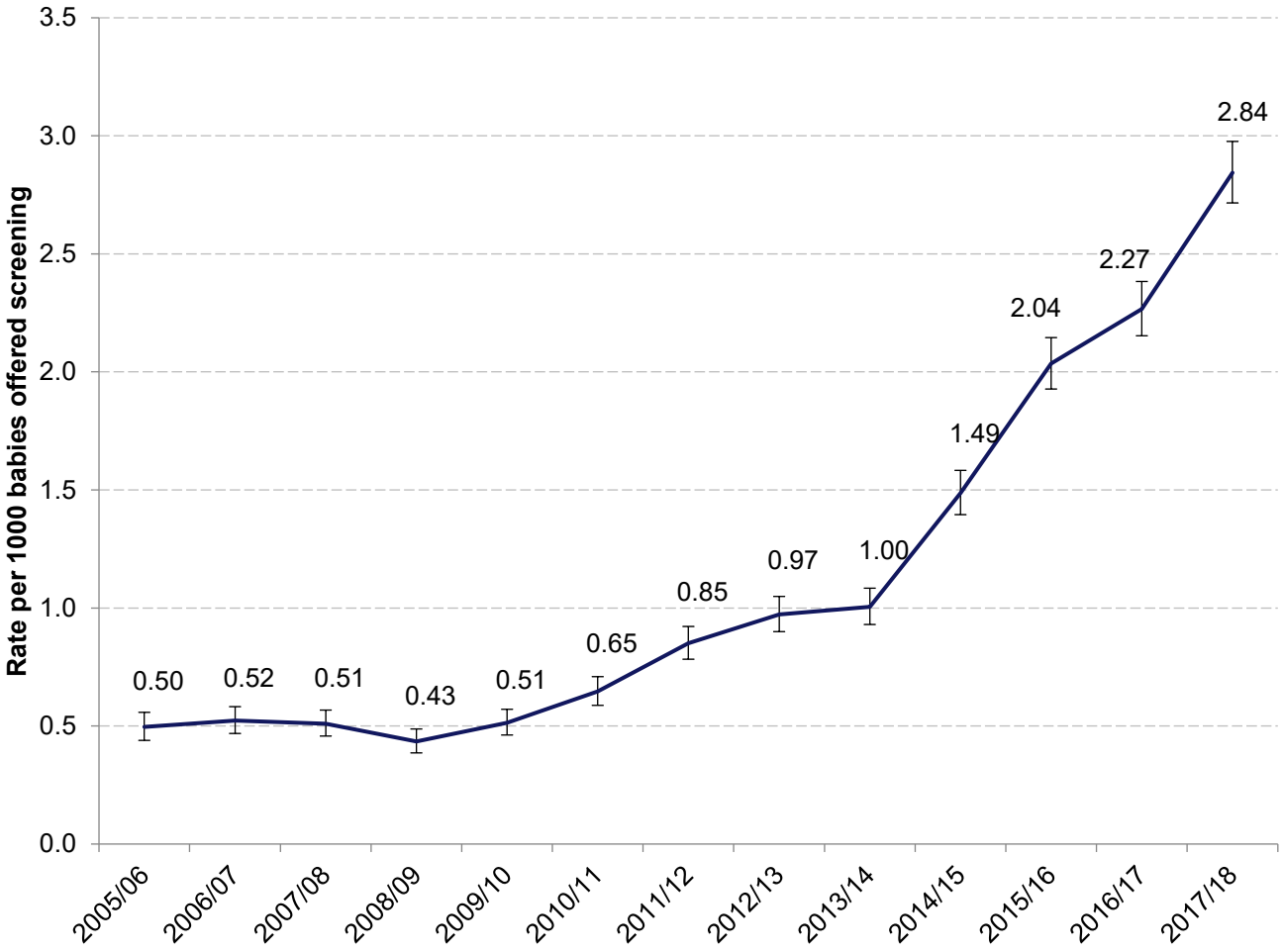
Data in Table 13 and Figure 12 includes all babies tested in English laboratories. Therefore, babies from outside England but tested within an English laboratory will be included in the above.

*'Asian' includes 'Indian', 'Pakistan', 'Bangladeshi' and 'Any other Asian background'. 'Other' includes 'Chinese' and 'Any other ethnic category'

4.4. Declined screening test

There has been a continuation of the increase in the rate of declined tests, with the rate in 2017 to 2018 being 2.84 per 1000 babies offered screening. The reason for this increase is not clear, but it may be due to improved reporting of declines or declines for mover-in babies who are older babies and may have been tested elsewhere.

Figure 13: Declined screening tests for sickle cell disease, England, 2005 to 2006 – 2017 to 2018

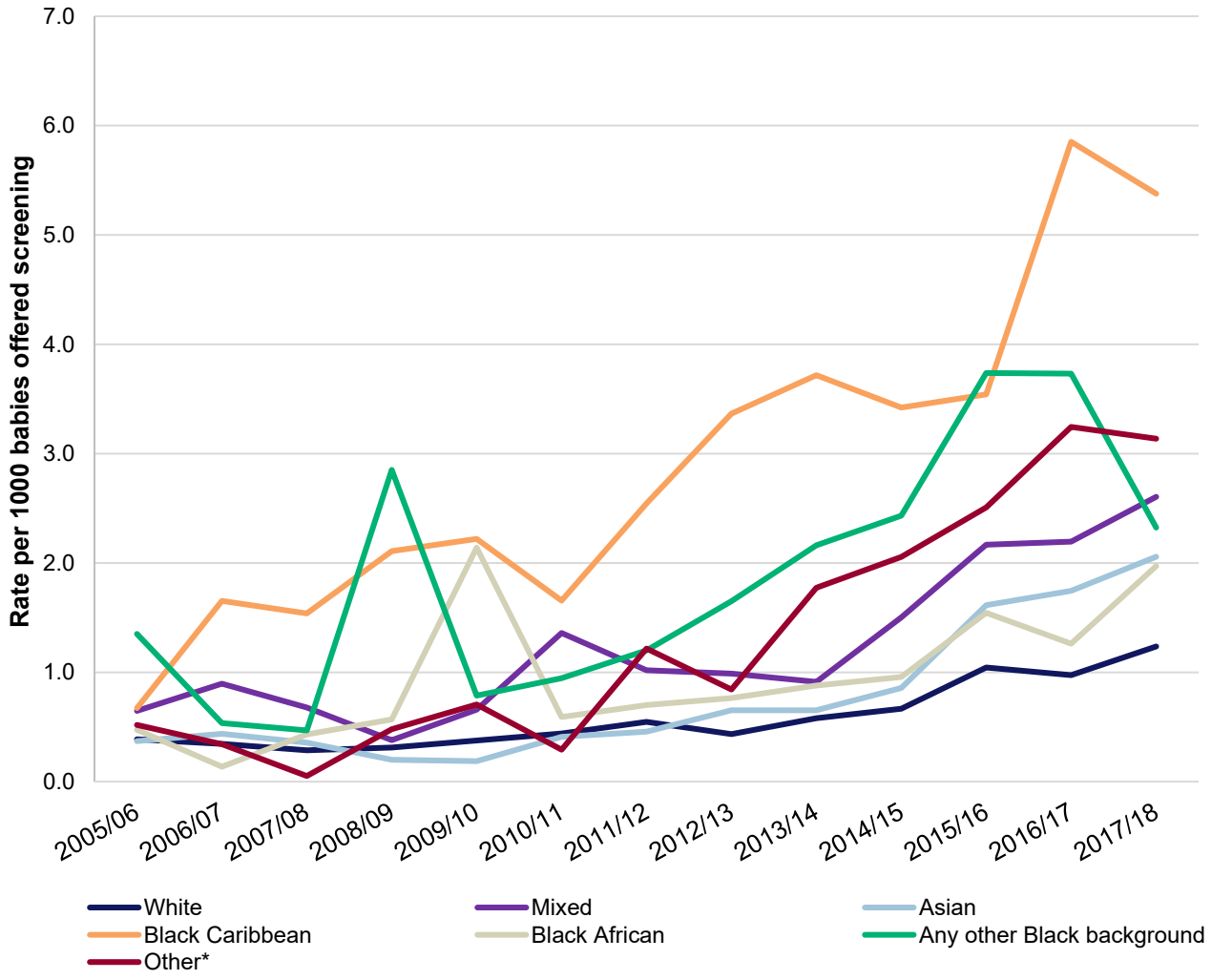


Bristol laboratory data for first half of 2005 to 2006 not included and Oxford and Portsmouth laboratories data not included for whole of 2005 to 2006; Oxford laboratory data starts from 1st July 2006.

Figure 14 shows the trends in rates of declined screening tests, by ethnicity, for babies where ethnicity is reported. In 2017 to 2018 the highest decline rates were in the 'Black Caribbean' and 'Other' ethnic categories, although decline rates for both these ethnic categories decreased slightly compared to the previous year.

Although not shown in figure 14, the rates of declined screening tests for babies where ethnicity was not stated or not known has increased from approximately 0.9 per 1000 babies offered screening in 2005 to 2006, to 37 per 1000 babies offered screening in 2017 to 2018. This is due to decreases in the number of babies with ethnicity not stated or not known, and increases in the declines recorded for these babies over the same period. This may be due to changes in the recording of both ethnicity and declines. The recording of ethnicity may also be more likely to be absent for babies where the test has been declined.

Figure 14: Declined screening tests for sickle cell disease by ethnic category, England, 2005 to 2006 – 2017 to 2018



The above includes all babies tested in English laboratories. Therefore, babies from outside England but tested within an English laboratory will be included in the above.
*'Other' includes the 'Chinese' and 'Any other ethnic category'
Babies with not stated / not known ethnic category have not been included in the above.
Bristol laboratory data for first half of 2005 to 2006 not included and Oxford and Portsmouth laboratories data not included for whole of 2005 to 2006; Oxford laboratory data starts from 1st July 2006.

4.5. Post-transfusion testing

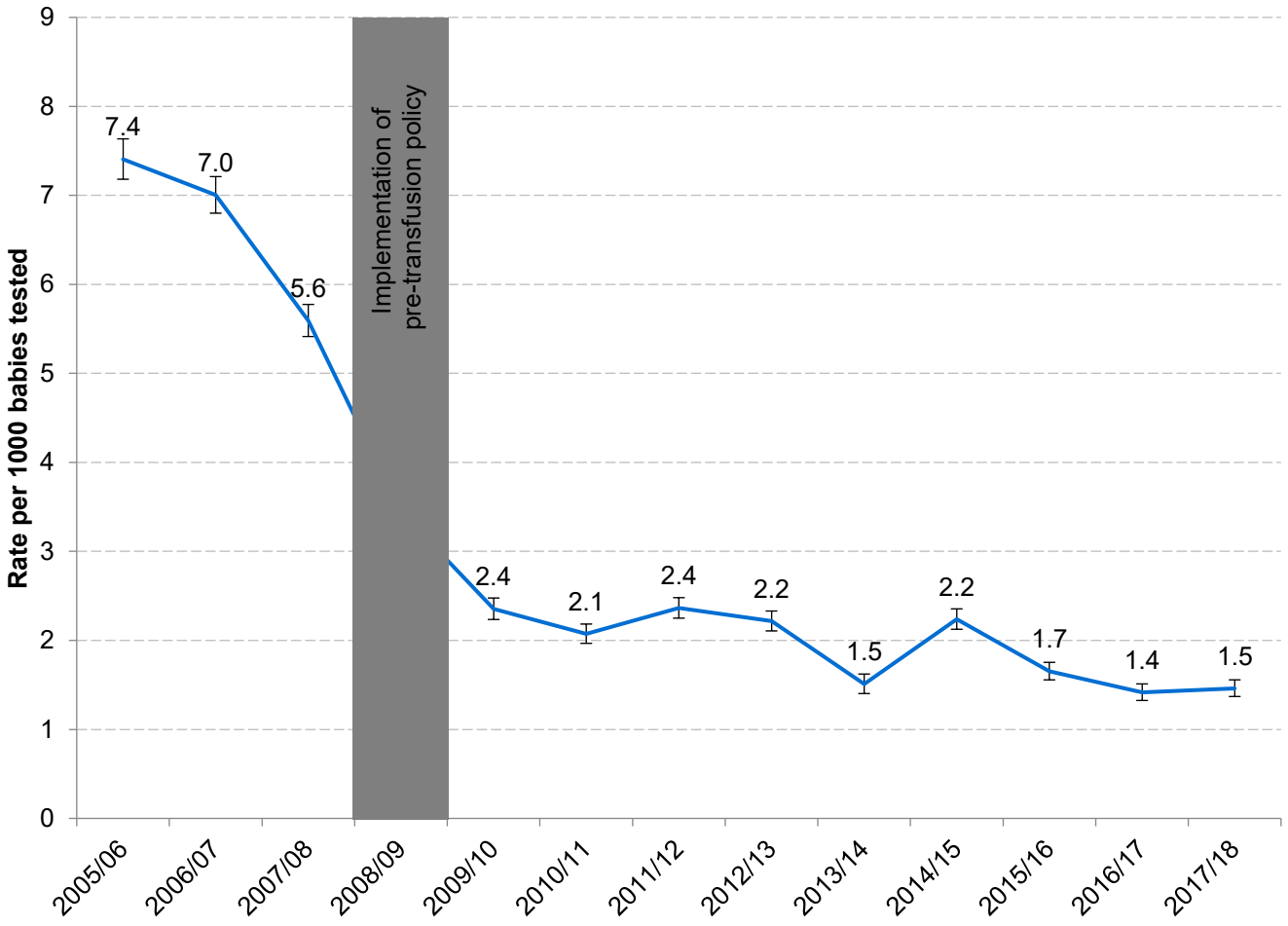
Haemoglobin analysis is not suitable for testing samples from transfused babies as transfused red cells can survive up to 120 days in circulation. It is therefore important that pre-transfusion samples are taken in line with newborn blood spot sampling guidelines. The NBS programme introduced a pre-transfusion sample policy in 2008, as detailed in the guidelines for newborn blood spot sampling, which requires that blood spots should be taken for SCD screening before blood transfusion. Figure 15 demonstrates that the incidence of post-transfusion samples has decreased since this policy was introduced, and the rates in England have remained stable over the last 3 years.

Table 14: Number and rates of post-transfusion samples reported by newborn screening laboratories, England, 2015 to 2016 – 2017 to 2018

Region	2015/16			2016/17			2017/18		
	n	Total tested	Rate / 1000	n	Total tested	Rate / 1000	n	Total tested	Rate / 1000
London	255	129,055	1.98	205	132,619	1.55	190	129,967	1.46
Midlands and East	278	194,243	1.43	256	193,207	1.33	267	191,340	1.40
North	331	184,099	1.80	223	150,425	1.48	304	169,140	1.80
South	174	152,304	1.14	127	151,437	0.84	94	146,255	0.64
Unknown region	64	6,948	9.21	93	10,701	8.69	96	14,084	6.82
England total	**1,102**	**666,649**	**1.65**	**904**	**638,389**	**1.42**	**951**	**650,786**	**1.46**

Post-transfusion sample data from Liverpool laboratory for 2016 to 2017 is not available and thus excluded from the above.

Figure 15: Rates of post-transfusion samples, England, 2005 to 2006 to 2017 to 2018



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;
Data from Manchester laboratory for 2009/10, from GOSH for 2013/14 and from Liverpool for 2016/17 is not available. Data for South East Thames in 2013/14 excluded for data quality reasons.

According to the guidelines for newborn blood spot sampling, where it is not possible to take a pre-transfusion sample, DNA testing is required to mitigate the risk of a missed baby. 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 Tables 15 and 16.

Table 15: Numbers detected through DNA testing for transfused babies reported by newborn DNA testing laboratories, England, 2013 to 2014 – 2017 to 2018

	2013/14	2014/15	2015/16	2016/17	2017/18
Total Specimens received per year	1,160	1,123	1,198	1,071	1,012
Number of Negative results (Hb S not detected)	1,140	1,106	1,183	1,054	992
Number of Heterozygotes	20	16	15	17	19
Number of Homozygotes	0	1	0	0	0

Table 16: Number of post-transfusion samples received from each screening laboratory, England, 2017 to 2018

Newborn DNA testing laboratory	Newborn Screening Laboratory	Number of samples
King's College Hospital	Bristol	31
	Cambridge	43
	Great Ormond Street	208
	Oxford	10
	Portsmouth	56
	South East Thames	96
	South West Thames	39
Sheffield	Leeds	83
	Liverpool	72
	Manchester	68
	Newcastle	59
	Sheffield	138
	West Midlands	109
England total		**1,012**

4.6. Blood spot test processes

The Newborn Blood Spot (NBS) Screening Programme has 3 screening standards relating to the blood spot test that are relevant to newborn screening for sickle cell disease.

Screening standard 4 (NBS-S04) specifies that the blood spot sample should be taken on day 5, and in exceptional circumstances between day 6 and day 8 (day of birth is day 0). The England performance for this standard in 2017 to 2018 was 84.0%.

Screening standard 5 (NBS-S05) relates to the proportion of blood spot samples that are received less than or equal to 3 working days of sample collection. The England performance for this standard in 2017 to 2018 was 94.3%.

Screening standard 6 (NBS-S06) collects the proportion of blood spot samples that require repeating due to an avoidable failure in the sampling process. This standard is a reverse polarity indicator (this means that a lower percentage indicates better performance). Performance for this standard in 2017 to 2018 was 2.5%.

The NBS screening programme annual reports present this data in more detail.

Within the SCT screening programme, screening standard 8 (SCT-S08) relates to the reporting of newborn screen positive results to parents. The standard collects the proportion of parents receiving newborn screen positive results at less than or equal to 28 days of age. In 2017 to 2018 the England performance for this standard was 65.9%. This data is also presented in the annual antenatal standards data report.

4.7. Entry into care

SCT Screening standard 9 (SCT-S09) collects the proportion of babies with a positive screening result who are seen at a paediatric clinic or discharged for insignificant results by 90 days of age. Performance for this standard in England in 2017 to 2018 was 83.7%. More detailed presentation of this data can be found in the annual antenatal standards data report.

Abbreviations

CCG	Clinical commissioning group
CHISS	Child health information service system
CHRD	Child health record department
FOQ	Family Origin Questionnaire
Hb	Haemoglobin – see glossary for haemoglobin variants
HPFH	Hereditary persistence of fetal haemoglobin
MCH	Mean cell haemoglobin
NAD	No abnormality detected
NBS	Newborn blood spot
NCARDRS	National Congenital Anomaly and Rare Disease Registration Service
PHE	Public Health England
PND	Prenatal diagnosis
SCD	Sickle cell disease
SCT	Sickle cell and thalassaemia
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 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 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 Hb Barts hydrops fetalis, and the mother runs the risk of obstetric complications, particularly in the third trimester of pregnancy. 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 1 in 4 chance or higher of the fetus having a serious haemoglobinopathy, based on antenatal screening results for both parents. Where the mother carries or has a significant haemoglobinopathy and the father is not available for testing, or where father results cannot be linked to mother results, are considered to be ‘at risk’, but are not counted as an ‘at risk’ couple. The number of ‘at-risk’ couples includes ‘high-risk’ pregnancies (see below).

Beta thalassaemia major
A severe anaemia caused by inheritance of two beta thalassaemia genes, resulting in a lack of normal haemoglobin production. This only starts to cause symptoms when the baby is a few months old. 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.

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.

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

Haemoglobinopathy
Mild or serious diseases that can occur in people who have inherited 2 haemoglobin gene variants (see 'variant' below). The most common haemoglobinopathies screened for include:

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

'High-risk' pregnancies
Pregnancies that are identified as having a 1 in 4 chance or higher of the fetus being affected by a serious haemoglobin disorder. These are identified based on the combinations of mother and father antenatal test results (represented by the dark orange boxes on the antenatal data return, see Appendix A).

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), Hb SC disease, and Hb S/beta thalassaemia. Sickle cell diseases can cause episodes of acute pain (crisis), anaemia, increased risk of infections, and chest problems.

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).

Appendices

Appendix A: 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														