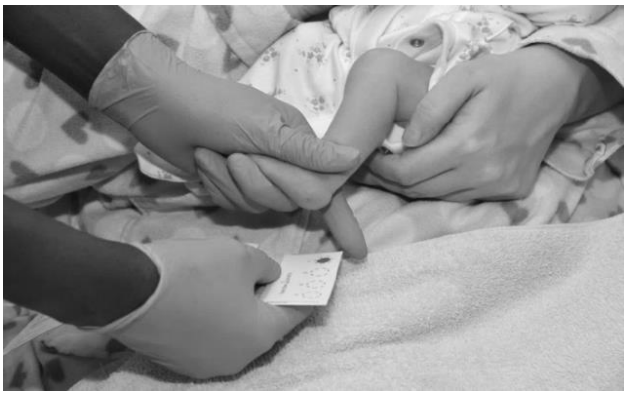
NHS

Screening Programmes

Sickle Cell and Thalassaemia

Data Report 2013/14

Trends and performance analysis



Acknowledgements

The programme would like to thank all those who have provided data for inclusion in this report for all their hard work. In particular we would like to thank the antenatal, prenatal diagnostic and newborn laboratories, as well as the maternity units.

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Published March 2015
PHE publications gateway number: 2014808

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Executive summary

In 2013/14 approximately 731,000 women were screened antenatally for sickle cell disease and thalassaemia conditions, and of these approximately 15,000 women (2%) were identified as screen positive. There were 353 prenatal diagnostic (PND) tests performed, which represents approximately 40% of the number of 'high risk' couples identified in antenatal screening.

There were approximately 668,000 babies screened through newborn blood spot screening. Of these, 319 were identified as affected by significant conditions (approximately 1 in 2,000 babies screened) and 8,850 were identified with carrier results (approximately 1 in 76 babies screened).

There has been an improvement in the proportion of antenatal screening tests performed by 10 weeks gestation, from approximately 46% in 2012/13 to approximately 50% nationally in 2013/14. This meets the acceptable standard for the first time. However, the variation between high and low prevalence areas remains the same each year with the proportion of women tested by 10 weeks gestation approximately 10% lower in high prevalence areas than in low prevalence areas. This could mean that women who are at greater risk of being screen positive are less likely to be offered screening before 10 weeks gestation. Similarly, there is variation in completion of the family origin questionnaire (FOQ) between high and low prevalence areas.

There has been a decrease in the proportion of samples not tested due to a known previous result with national rates comparable to those from 2007/08. This may be linked to a change in programme guidance on re-testing women in subsequent pregnancies which recommends that women need not be re-tested in the same or subsequent pregnancy provided that there are two or more previous results from an accredited laboratory. Anecdotal reports suggest that some laboratories may be opting to re-test all samples.

There has been an improvement in the completion of gestational data for PND testing, although completion of pregnancy outcome data has declined. Approximately half of PND tests are performed by 12 weeks and six days, which is consistent with figures for previous years. However, approximately 30% of PND tests are being performed after 15 weeks gestation. Early PND testing is dependent on early antenatal screening, unless the parents' results are already known, in order to identify the risk to the pregnancy. It is therefore important that efforts continue to improve early antenatal screening to move towards the achievable standard of 75%.

Pregnancy outcome data covering six years show that in approximately 67% of cases where there was a sickle cell affected result, and in approximately 87% of cases with a beta thalassaemia affected result, parents opted to terminate the pregnancy.

There were 668,117 babies reported as being screened by newborn screening laboratories in 2013/14. This represents a decrease in numbers screened compared to previous years, but this drop is also

reflected in ONS birth figures for 2013. Significant conditions are most prevalent in black African and black Caribbean backgrounds, but these conditions are not exclusive to these ethnic categories. In 2013/14 five affected cases were identified as white British, or one in 64 cases identified with significant conditions.

Declined newborn screening tests for sickle cell disease have continued to increase and are now at approximately one per 1,000 babies screened. Nationally the rates for post-transfusion samples appear to have increased, although this appears to mainly be due to an increase in the South East Coast region.

Whilst beta thalassaemia is not currently screened for in newborn screening, F-only cases are picked up as a by-product of screening for sickle cell disease. These are probable beta thalassaemia major cases and require follow-up. In 2013/14 there were 32 F-only cases reported by the newborn laboratories.

The Sickle Cell and Thalassaemia (SCT) Screening Programme has expanded the data requested on timeliness of newborn screening to include anonymised data on screening outcomes. Data for these new fields were requested for 2013/14 so that the programme could assess whether they are appropriate, but will only be reported from 2014/15. Preliminary findings from incomplete data based on 292 babies with significant conditions of F-only results suggest that the median age for reporting a screen positive result is 15 days. This suggests that the system is capable of meeting the 28 day standard for informing parents of results.

Abbreviations

AN	Antenatal
CCG	Clinical Commissioning Group
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
NBBS	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
PND	Prenatal Diagnosis
SCD	Sickle Cell Disease
SCT	Sickle Cell and Thalassaemia
SHA	Strategic Health Authority
UK NSC	United Kingdom National Screening Committee

Glossary

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

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

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

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

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

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

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

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

Haemoglobin disease:
Mild or serious diseases that can occur in people who have inherited two haemoglobin gene variants. The most common haemoglobin diseases are sickle cell diseases and thalassaemia disorders, also called haemoglobinopathies. 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).

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

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

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

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

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

List of charts and tables

Antenatal Screening

Prenatal Diagnostic Testing

Newborn Screening

1. Introduction

1.1. About the NHS Sickle Cell and Thalassaemia Screening Programme

The NHS Sickle Cell and Thalassaemia (SCT) Screening Programme uses blood tests to screen newborn babies and pregnant women for two serious inherited blood disorders – sickle cell disease and thalassaemia major.

The NHS Sickle Cell and Thalassaemia Screening Programme was set up in England in 2001 following a Government commitment in the NHS Plan in 2000 and is the world's first linked antenatal and newborn screening programme.

Our mission statement is to develop a linked programme of high quality screening and care in order to:

- support people to make informed choices during pregnancy and before conception
- improve infant health through prompt identification of affected babies
- provide high quality and accessible care throughout England
- promote greater understanding and awareness of the disorders and the value of screening

Public Health England (PHE) is responsible for the NHS Screening Programmes. PHE is an executive agency of the Department of Health and works to protect and improve the nation's health and wellbeing, and reduce health inequalities.

1.2. Methods

Timely annual data returns are required from all screening laboratories in accordance with the laboratory guidance. Data are collated by the laboratories and submitted to the Sickle Cell and Thalassaemia Screening Programme via spreadsheet-based data return templates. On receipt, the data are checked for any discrepancies or aspects that would benefit from clarification and if needed are followed up with the relevant laboratory.

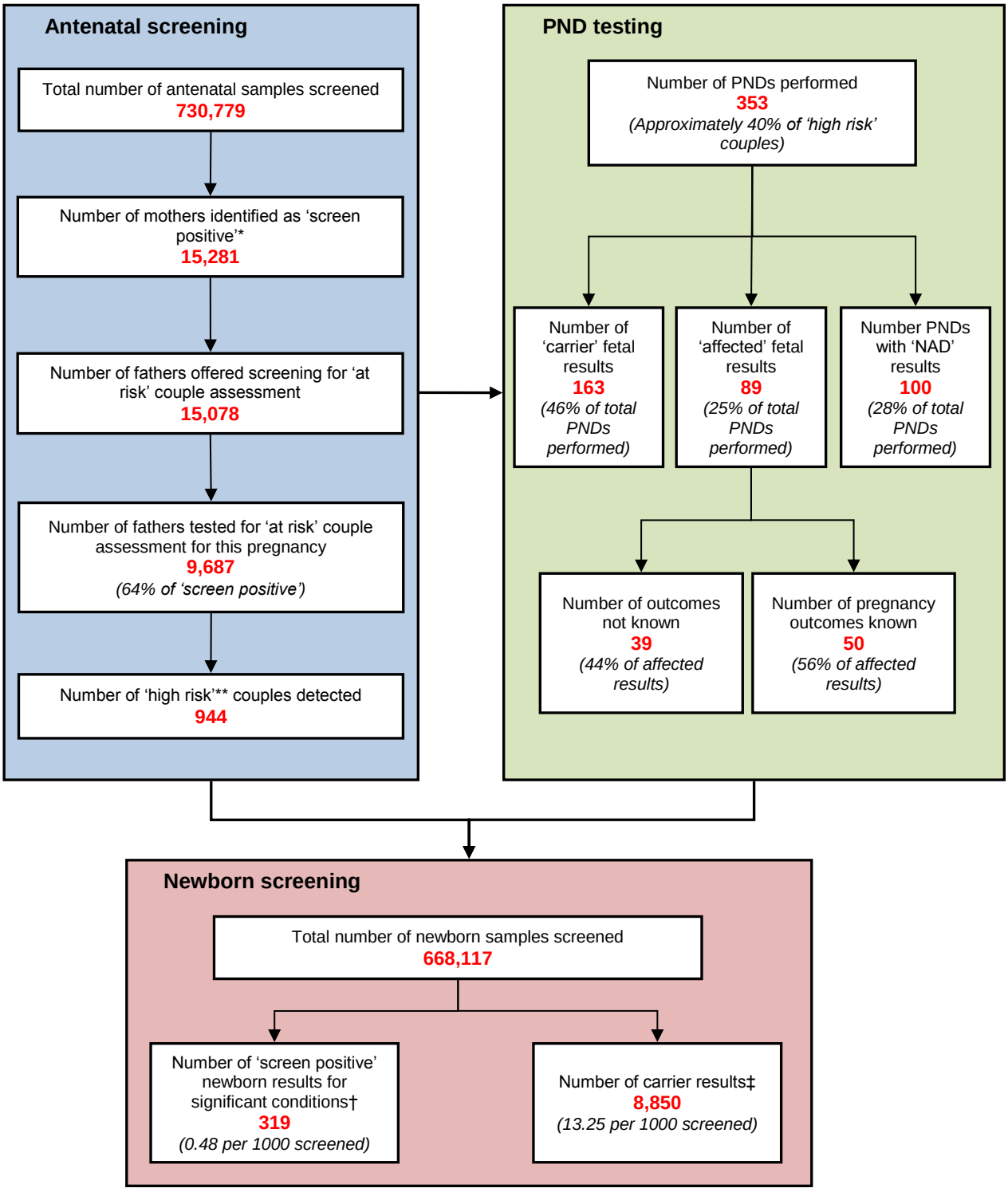
For the antenatal laboratories in particular, we recognise the difficulty of data collection in the absence of standardised data collection tools and IT systems. We try to ask for limited data and work hard to justify all data requests, ensuring there are no gaps and no duplication across the screening pathway and between screening programmes.

PND data are requested several months after the requests for data are sent to the antenatal and newborn laboratories. This is to allow time for complete gestation in all pregnancies in order to give a more complete set of data on pregnancy outcomes following PND testing.

The newborn data received by the programme sometimes include data for areas outside of England. These are excluded in our analysis. Prevalence data by region and by ethnicity are compared and laboratories contacted for clarification if inconsistencies are found.

Current versions of the antenatal and newborn data returns can be found at sct.screening.nhs.uk/datacollection.

2. Overview of national screening figures



Note: These figures represent total numbers reported and numbers may differ from those where exclusions could be made based on missing or unavailable data.
**'Screen positive' in antenatal screening includes both sickle cell haemoglobin variants and thalassaemia results.*
†'Significant conditions' in newborn screening comprises FS, FSC, FS Other and FE.
‡'Carrier results' in newborn screening comprises FAS, FAC, FAD, FAE and other carriers.
*** 'High risk' comprises cases where both parents are carriers or affected and there is a high risk that the baby will be affected by a significant condition. This number excludes low risk cases and cases where the father was not available for testing.*

3. Antenatal screening data

3.1. Response rates and data quality

Response rate:

The SCT screening programme received all 142 data returns requested across England. We would like to thank all those involved in providing data to the programme.

Data quality:

A total of 730,779 booking bloods were reported for 2013/14. Whilst data returns were received from all laboratories, not all laboratories were able to provide complete data for all of the requested fields. To reduce bias when reporting rates, exclusions have been made where data were missing. This means that some figures may differ when comparing charts and tables. Where exclusions have been made, these are specified in the relevant footnotes. The number of exclusions required in 2013/14 were comparable to those in 2012/13.

Figures on booking bloods tested by 10 weeks are often dependent on completion of the FOQ to obtain gestational information. This means that the figures presented on timeliness of testing offer a base rate, but actual proportions achieving this standard may be higher.

The number of father specimens received may not include cases where the father's results were previously known, and so the rates for father uptake may in fact be higher than those shown. Some laboratories are unable to match mother results to father results and so can't provide the number of high risk couples. As a result, the actual number of high risk couples is likely to be higher than reported.

Some laboratories use figures that are provided by maternity units to determine the number of booking bloods received as they are unable to distinguish between antenatal and non-antenatal specimens. This may distort the figures slightly as maternity units may refer samples to more than one laboratory and so the number of booking bloods received may appear higher than it actually is. Some laboratories cover more than one hospital and we ask for separate data returns for each hospital covered. As a result, the number of laboratories represented where data are broken down to this level may be higher than the actual number of laboratories that provide screening for sickle cell and thalassaemia.

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

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

3.2. Numbers screened and detected

National:

Table AN-1 shows the antenatal screening figures by region for England in 2013/14. These figures represent the totals reported and no exclusions are made in this table based on missing or incomplete data. In 2013/14 a total of 730,779 antenatal samples were identified by the laboratories, of which 15,281 women (one in 48 women screened) were identified as carriers of haemoglobin variants which could result in the pregnancy being at risk for sickle cell disease or a thalassaemia condition. The number of screen positive women includes cases where the woman had a known previous screen positive result, cases where a donor egg was used or there was a bone marrow transplant, and other haemoglobin variant carriers where testing of the baby's father was recommended.

'High risk' couples comprise pregnancies where both parents are identified as either carriers or as affected and there is a high risk that the baby will be affected by a significant condition. In 2013/14 there were 944 pregnancies (one in 16 screen positive women) identified as at high risk of an affected pregnancy. This figure excludes cases where the father was not available for testing, or where the father's result cannot be matched with the mother's result to determine risk. It is therefore estimated that the actual number of high risk pregnancies is higher. We would expect the number of high risk couples in antenatal screening to be approximately four times the number of newborn screen positive cases identified with significant conditions (FS, FSC, FS-Other and FE results) plus four times the number of babies with an F-only newborn result (which are potential beta thalassaemia affected results), plus terminations of affected pregnancies following PND testing, giving an estimate of approximately 1,450 high risk pregnancies.

It should be noted that some high prevalence laboratories provide screening services for low prevalence maternity services. While we request that data from laboratories are provided separately by site, some laboratories cannot differentiate between samples received, which could affect the figures.

Table AN-1. Antenatal screening results by region, 2013/14: England

Region	No. of Labs	Booking bloods received (BBs)	Booking bloods tested by 10 wks		FOQ Attached		Screen 'positive' women		Screen 'negative' women		Result pending		High risk couples identified	
	Submitted / Total Labs	n	n	% of BBs	n	% of BBs	n	% of BBs	n	% of BBs	n	% of BBs	n	% of screen 'positive women
East of England	17 / 17	81,303	35,221	43.32	79,228	97.45	1,120	1.38	66,422	81.70	13	0.02	65	5.80
East Midlands	8 / 8	52,367	33,769	64.49	51,408	98.17	674	1.29	51,581	98.50	*	0.00	43	6.38
London	24 / 24	146,281	43,162	29.51	120,096	82.10	7,653	5.23	134,484	91.94	24	0.02	505	6.60
North East	10 / 10	33,171	13,666	41.20	31,672	95.48	220	0.66	31,517	95.01	3	0.01	12	5.45
North West	19 / 19	90,926	37,244	40.96	87,955	96.73	1,148	1.26	87,421	96.15	31	0.03	61	5.31
South Central	10 / 10	52,918	27,779	52.49	49,199	92.97	814	1.54	51,947	98.17	16	0.03	54	6.63
South East Coast	10 / 10	58,525	11,041	18.87	57,291	97.89	649	1.11	53,105	90.74	10	0.02	27	4.16
South West	17 / 17	65,585	29,285	44.65	65,054	99.19	449	0.68	60,829	92.75	*	0.00	22	4.90
West Midlands	15 / 15	77,809	25,453	32.71	75,228	96.68	1,512	1.94	74,665	95.96	6	0.01	91	6.02
Yorkshire and The Humber	12 / 12	71,894	39,082	54.36	70,489	98.05	1,042	1.45	70,196	97.64	9	0.01	64	6.14
Total England	**142 / 142**	**730,779**	**295,702**	**40.46**	**687,620**	**94.09**	**15,281**	**2.09**	**682,167**	**93.35**	**115**	**0.02**	**944**	**6.18**

**Numbers less than 3 have been suppressed*

High prevalence areas:

Table AN-2 shows the antenatal screening figures for high prevalence areas for 2013/14 as reported by the antenatal laboratories. A total of 392,592 antenatal samples were identified in high prevalence areas in 2013/14. Of these, 12,750 women (one in 31 women screened) were identified as screen positive as carriers of haemoglobin variants which could result in the pregnancy being at risk for sickle cell disease or thalassaemia, including women with a known previous result. In high prevalence areas 825 pregnancies (one in 16 screen positive women) were identified as being at high risk for these conditions.

Table AN-2. Antenatal screening results by region, 2013/14: High prevalence areas

Region	No. of Labs	Booking bloods received (BBs)	Booking bloods tested by 10 wks		FOQ Attached		Screen 'positive' women		Screen 'negative' women		Result pending		High risk couples identified	
	Submitted / Total Labs	n	n	% of BBs	n	% of BBs	n	% of BBs	n	% of BBs	n	% of BBs	n	% of screen 'positive women
East of England	5 / 5	25,887	8,102	31.30	25,055	96.79	599	2.31	24,987	96.52	*	0.00	40	6.68
East Midlands	4 / 4	31,282	20,595	65.84	30,641	97.95	534	1.71	30,701	98.14	*	0.00	33	6.18
London	24 / 24	146,281	43,162	29.51	120,096	82.10	7,653	5.23	134,484	91.94	24	0.02	505	6.60
North East	1 / 1	6,574	*	0.00	6,538	99.45	88	1.34	6,477	98.52	*	0.00	5	5.68
North West	7 / 7	51,555	19,880	38.56	49,340	95.70	969	1.88	49,587	96.18	20	0.04	54	5.57
South Central	6 / 6	31,829	17,535	55.09	28,391	89.20	576	1.81	31,196	98.01	10	0.03	46	7.99
South East Coast	2 / 2	11,147	3,393	30.44	11,001	98.69	222	1.99	10,770	96.62	*	0.00	10	4.50
South West	2 / 2	12,121	4,158	34.30	12,036	99.30	192	1.58	11,876	97.98	*	0.00	10	5.21
West Midlands	7 / 7	47,363	12,135	25.62	45,149	95.33	1,314	2.77	44,450	93.85	*	0.00	76	5.78
Yorkshire and The Humber	3 / 3	28,553	12,648	44.30	27,285	95.56	603	2.11	27,746	97.17	8	0.03	46	7.63
Total England	**61 / 61**	**392,592**	**141,608**	**36.07**	**355,532**	**90.56**	**12,750**	**3.25**	**372,274**	**94.82**	**64**	**0.02**	**825**	**6.47**

**Numbers less than 3 have been suppressed*

Low prevalence areas:

Table AN-3 shows the antenatal screening figures by region for low prevalence areas for 2013/14 as reported by the antenatal laboratories. A total of 338,187 antenatal samples were identified in low prevalence areas in 2013/14. Of these, 2,531 women (one in 134 women screened) were identified as screen positive as carriers of haemoglobin variants which could result in the pregnancy being at risk for sickle cell disease or thalassaemia, including women with a known previous result. In low prevalence areas 119 pregnancies (one in 21 screen positive women) were identified as being at high risk for these conditions.

Table AN-3. Antenatal screening results by region, 2013/14: Low prevalence areas

Region	No. of Labs	Booking bloods received (BBs)	Booking bloods tested by 10 wks		FOQ Attached		Screen 'positive' women		Screen 'negative' women		Result pending		High risk couples identified	
	Submitted / Total Labs	n	n	% of BBs	n	% of BBs	n	% of BBs	n	% of BBs	n	% of BBs	n	% of screen 'positive women
East of England	12 / 12	55,416	27,119	48.94	54,173	97.76	521	0.94	41,435	74.77	13	0.02	25	4.80
East Midlands	4 / 4	21,085	13,174	62.48	20,767	98.49	140	0.66	20,880	99.03	*	0.00	10	7.14
London	0 / 0	-	-	-	-	-	-	-	-	-	-	-	-	-
North East	9 / 9	26,597	13,666	51.38	25,134	94.50	132	0.50	25,040	94.15	3	0.01	7	5.30
North West	12 / 12	39,371	17,364	44.10	38,615	98.08	179	0.45	37,834	96.10	11	0.03	7	3.91
South Central	4 / 4	21,089	10,244	48.58	20,808	98.67	238	1.13	20,751	98.40	6	0.03	8	3.36
South East Coast	8 / 8	47,378	7,648	16.14	46,290	97.70	427	0.90	42,335	89.36	10	0.02	17	3.98
South West	15 / 15	53,464	25,127	47.00	53,018	99.17	257	0.48	48,953	91.56	*	0.00	12	4.67
West Midlands	8 / 8	30,446	13,318	43.74	30,079	98.79	198	0.65	30,215	99.24	4	0.01	15	7.58
Yorkshire and The Humber	9 / 9	43,341	26,434	60.99	43,204	99.68	439	1.01	42,450	97.94	*	0.00	18	4.10
Total England	**81 / 81**	**338,187**	**154,094**	**45.56**	**332,088**	**98.20**	**2,531**	**0.75**	**309,893**	**91.63**	**51**	**0.02**	**119**	**4.70**

**Numbers less than 3 have been suppressed*

3.3. Bookings tested by 10 weeks

The target for antenatal testing is an offer of testing by 10 weeks gestation. To offer informed choice, a series of tests may be required: for the mother, the father and, if required, on the unborn baby. If tests show that the baby is at risk of inheriting a major haemoglobin disorder then the parents need time to receive counselling and consider their options, with a target for all tests to be completed by 12 weeks and six days of gestation.

The proportion of booking bloods tested by 10 weeks of gestation links to KPI ST2 (timeliness of testing) and with programme standard AP1. It is important to note that standard AP1 specifies an offer by 10 weeks, whereas these data identify samples actually tested by 10 weeks. This means that these figures are using a stricter measure than that identified in the programme standards, as it is not currently possible for all maternity services to collect data on the offer of test.

Figures on booking bloods tested by 10 weeks are often dependent on completion of the FOQ to obtain gestational information (see 3.1 Response rates and data quality).

Table AN-4 shows the proportion of booking bloods tested by 10 weeks of gestation in high and low prevalence areas covering a three-year period. In this period the proportion tested by 10 weeks was consistently higher in low prevalence areas than in high prevalence areas, but an increase has been reported each year and the national figures show nearly half of booking bloods tested by 10 weeks in 2013/14.

The data indicate that in high prevalence areas where there is a greater risk of being screen positive a carrier of sickle cell disease or thalassaemia, pregnant women are less likely to be offered screening before 10 weeks gestation (45% tested by 10 weeks in high prevalence areas compared to 55% in low prevalence areas). This could have an impact on equality and access to screening. This gap of approximately 8% can be seen in each of the three years shown in Table AN-4.

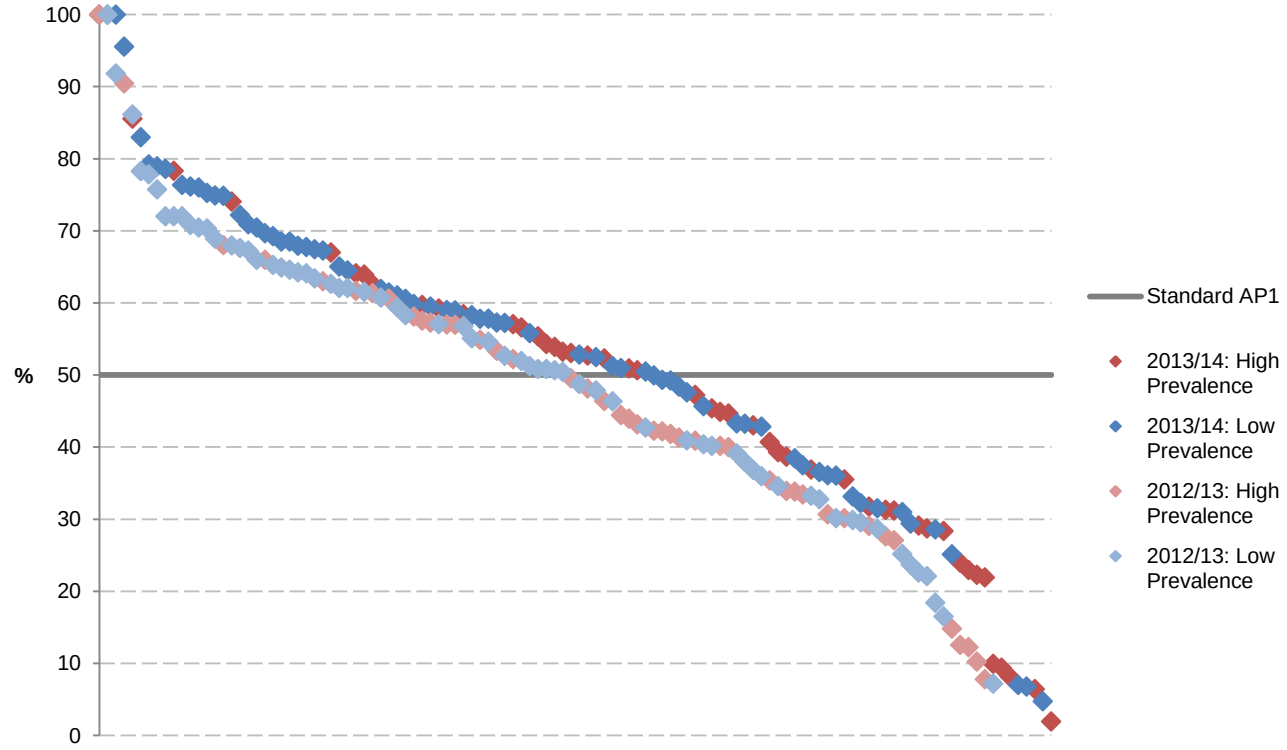
Table AN-4. Antenatal booking bloods tested by 10 weeks gestation, 2013/14: England by prevalence

Prevalence	2011/12			2012/13			2013/14		
	Booking bloods (BBs) received	BBs tested by 10 wks	% of BBs	Booking bloods (BBs) received	BBs tested by 10 wks	% of BBs	Booking bloods (BBs) received	BBs tested by 10 wks	% of BBs
High prevalence areas	302,004	121,349	40.18	302,348	125,509	41.51	306,470	137,503	44.87
Low prevalence areas	245,768	120,250	48.93	267,547	136,205	50.91	281,377	153,774	54.65
Total England	**547,772**	**241,599**	**44.11**	**569,895**	**261,714**	**45.92**	**587,847**	**291,277**	**49.55**

Exclusions based on missing or unavailable data for the data fields shown: 2011/12: 36; 2012/13: 27; 2013/14: 25.

Figure AN-1 shows a breakdown of the figures for 2013/14 by laboratory, comparing them to those for 2012/13. Whilst a greater number of laboratories appear to be achieving the 50% acceptable level for programme standard AP1, it also appears that those currently below the standard level are improving compared to the previous year.

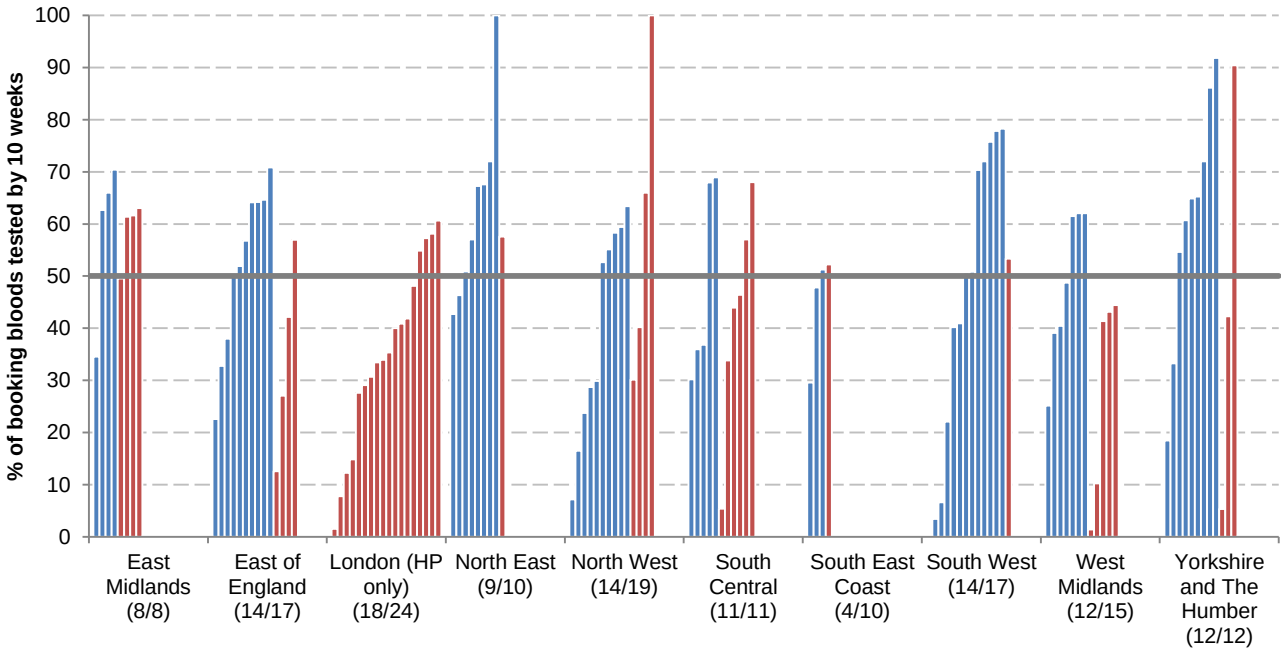
Figure AN-1. Percentage of antenatal booking bloods tested by 10 weeks by laboratory, 2012-14: England by prevalence



Each marker represents one laboratory.
The reference line represents the 50% acceptable level for programme standard AP1.
The rate for the whole of England was 44% in 2011/12, was 46% in 2012/13 and was 50% in 2013/14.
**2013/14 data excludes 25 data returns, 2012/13 data excludes 27 data returns based on missing or unavailable data.*

Figure AN-2 shows a breakdown of the figures for 2013/14 by laboratory grouped by region. This includes only laboratories that were able to provide information on both the total number of booking bloods and the number of samples tested by 10 weeks.

Figure AN-2. Percentage of antenatal booking bloods tested by 10 weeks by region, 2013/14: High and low prevalence areas



Each bar represents one laboratory. Red bars represent high prevalence areas and blue bars represent low prevalence areas.
The reference line represents the 50% acceptable level for programme standard AP1.
The rate for the whole of England is 46%
The numbers below the region represents how many of the laboratories that provided data were received were able to provide complete data on booking bloods tested by 10 weeks, e.g. "4/10" shows that of the 10 returns received, 4 included complete data for both fields.
Excludes 25 laboratories where data on booking bloods tested by 10 weeks or the total number of booking was missing or unavailable.

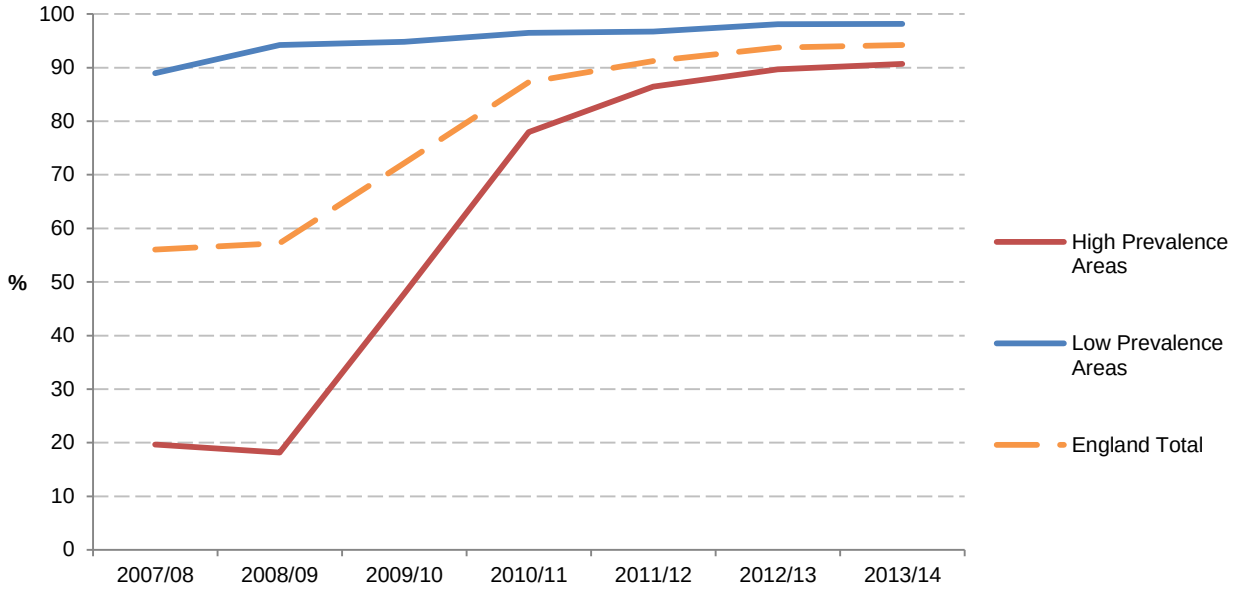
3.4. Samples with FOQ attached

The family origin questionnaire (FOQ) is used as a screening tool in both high and low prevalence areas. The proportion of booking bloods received with a FOQ attached links to programme standard AO1aiii and KPI ST3 (completion of FOQ).

Figure AN-3 shows the proportion of booking bloods received with a FOQ attached in both high and low prevalence areas across England between 2007 and 2014. The proportion has continued to improve in both high and low prevalence areas, and as a whole high prevalence areas are now achieving the 90% acceptable level for programme standard AO1aiii.

In high prevalence areas approximately 91% of booking bloods had a FOQ attached in 2013/14, compared to approximately 98% in low prevalence areas. Aggregated data for the whole of England shows that approximately 94% of booking bloods had a FOQ attached. Samples arriving without a completed FOQ could lead to unnecessary testing, and tests are performed using implied consent in the absence of information on the FOQ.

Figure AN-3. Booking bloods received with a FOQ attached, 2007-14: England by prevalence



	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14
High prevalence areas	19.6	18.2	47.8	78.0	86.4	89.7	90.7
Low prevalence areas	88.9	94.2	94.8	96.5	96.7	98.1	98.2
England total	56.0	57.2	72.2	87.3	91.2	93.7	94.2

Exclusions based on missing or unavailable data for the data fields shown: 2007/08: 36; 2008/09: 23; 2009/10: 19; 2010/11: 8; 2011/12: 1; 2012/13: 2; 2013/14: 2.

Table AN-5 shows the number and rates of samples received with a FOQ attached between 2011 and 2014 by region and for the whole of England (both high and low prevalence areas). Aggregated data show that outside of London, all regions are achieving the acceptable level for programme standard AO1aiii, and most are achieving over 95%.

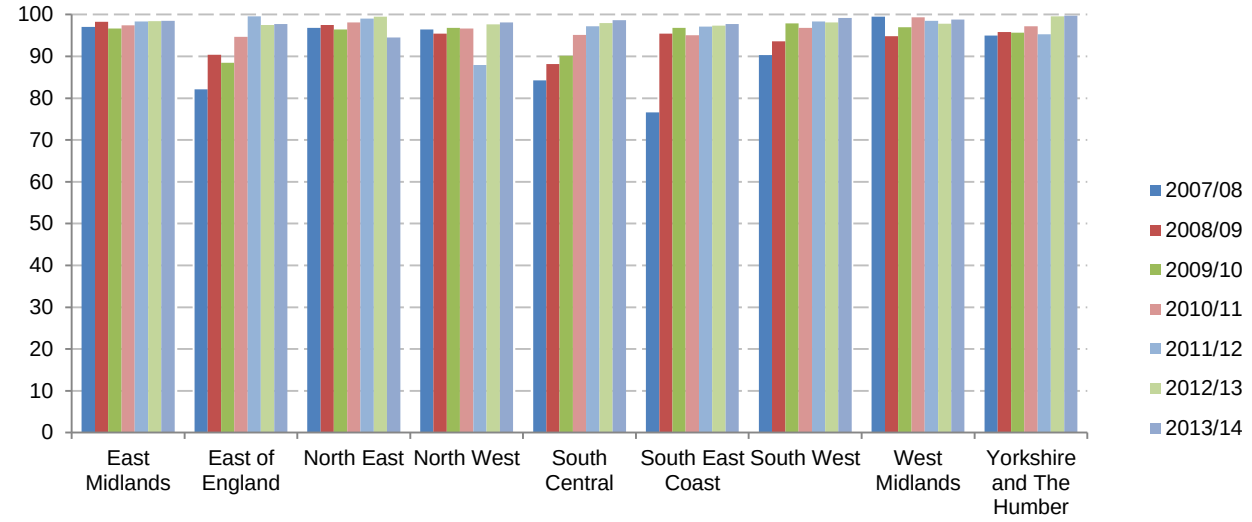
Table AN-5. Booking bloods received with a FOQ attached by region, 2011-14: England

Region	2011/12			2012/13			2013/14		
	Booking Bloods (BBs)	FOQ attached	% of BBs	Booking Bloods (BBs)	FOQ attached	% of BBs	Booking Bloods (BBs)	FOQ attached	% of BBs
East Midlands	55,195	53,653	97.2	53,016	52,238	98.5	52,367	51,408	98.2
East of England	81,767	79,752	97.5	78,123	75,433	96.6	81,303	79,228	97.4
London	142,595	117,395	82.3	125,135	105,123	84.0	141,535	116,404	82.2
North East	34,319	33,697	98.2	33,288	32,799	98.5	33,171	31,672	95.5
North West	90,653	82,461	91.0	91,004	86,892	95.5	90,926	87,955	96.7
South Central	54,676	50,710	92.7	51,803	48,351	93.3	52,918	49,199	93.0
South East Coast	44,716	43,508	97.3	57,885	56,366	97.4	58,525	57,291	97.9
South West	67,534	66,452	98.4	65,570	64,458	98.3	65,585	65,054	99.2
West Midlands	79,565	66,029	83.0	77,760	70,691	90.9	77,809	75,228	96.7
Yorkshire and The Humber	72,381	66,270	91.6	71,773	68,751	95.8	71,894	70,489	98.0
England Total	**723,401**	**659,927**	**91.2**	**705,357**	**661,102**	**93.7**	**726,033**	**683,928**	**94.2**

Exclusions based on missing or unavailable data for the data fields shown: 2011/12: 1; 2012/13: 2; 2013/14: 2.

Figure AN-4 shows these rates for low prevalence laboratories only, showing trends for each region. Some labs appear to have shown an increase in the proportion of booking bloods received with a FOQ attached, while others appear to have rates consistent with previous years.

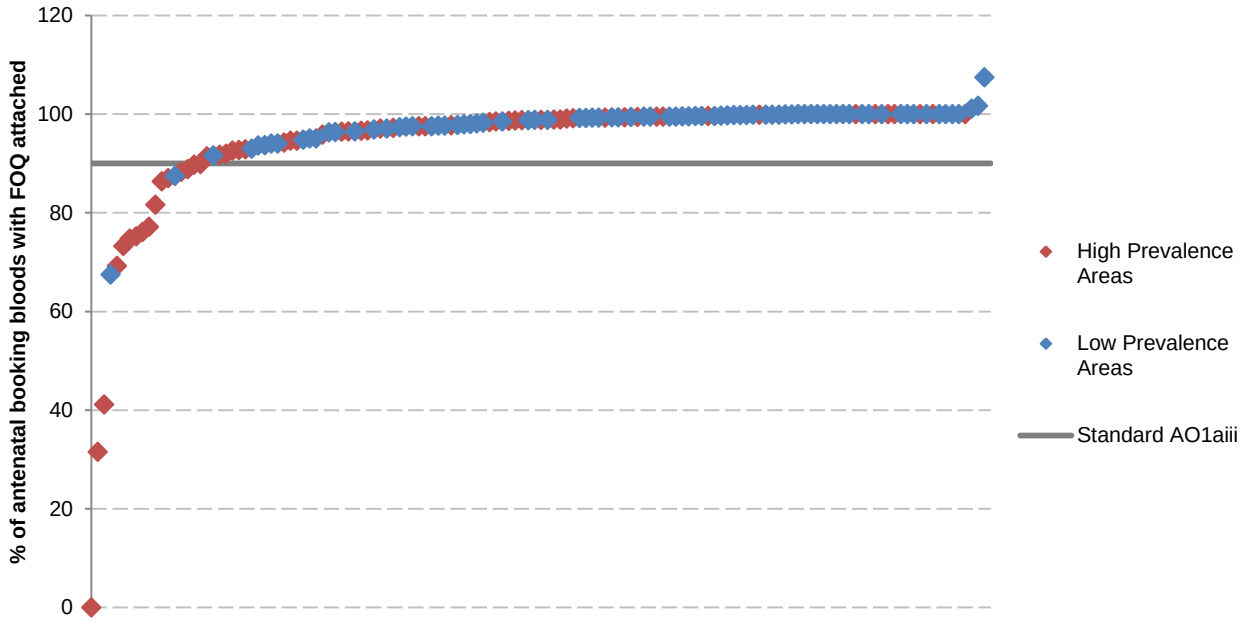
Figure AN-4. Booking bloods received with a FOQ attached, 2007-14: Low prevalence areas by region



Exclusions based on missing or unavailable data for the data fields shown: 2007/08: 5; 2008/09: 5; 2009/10: 5; 2010/11: 2; 2011/12: 1; 2012/13: 0; 2013/14: 0.

Figure AN-5 shows the proportion of booking bloods received with a FOQ attached by each laboratory across the whole of England in 2013/14, highlighting high and low prevalence areas. The majority of low prevalence areas are exceeding the 90% acceptable level for programme standard AO1aiii, but there are a number of laboratories falling below this level. Rates over 100% indicate data quality issues.

Figure AN-5. Percentage of antenatal booking bloods with a FOQ attached, 2013/14: England by laboratory



Each marker represents one laboratory.
The reference line represents the 90% acceptable level for programme standard AO1iii.
The rate for the whole of England is 94%.
**Excludes 2 laboratories where either data on booking bloods with an FOQ attached or the total number of booking bloods was missing or unavailable.*

Table AN-6 shows the usage of the FOQ in identifying potential carriers of alpha zero thalassaemia for the whole of England covering 2008-2014. These figures are broken down by high and low prevalence areas in Table AN-7 and Table AN-8.

Cases where the MCH is less than 25pg are potentially carriers of alpha zero thalassaemia. If the FOQ shows that the woman has a high risk family origin they are considered to be at high risk of being carriers of alpha zero thalassaemia. If no FOQ information is provided, laboratories are unable to exclude this risk which may lead to unnecessary testing.

Table AN-6. Use of the FOQ in determining women at high risk of being alpha zero carriers, 2008-14: England

Year	Booking bloods received (BBs)	FOQ attached		MCH < 25pg		High risk alpha0	
	n	n	% of BBs	n	% of FOQ attached	n	% of BBs
2008/09	554,216	317,009	57.20	17,826	5.62	4,817	0.87
2009/10	526,343	393,606	74.78	16,344	4.15	2,075	0.39
2010/11	659,060	574,641	87.19	20,557	3.58	3,532	0.54
2011/12	692,845	634,051	91.51	21,247	3.35	3,212	0.46
2012/13	691,310	647,252	93.63	23,881	3.69	2,834	0.41
2013/14	685,932	649,943	94.75	20,777	3.20	2,609	0.38
Total for six year period	**3,809,706**	**3,216,502**	**84.43**	**120,632**	**3.75**	**19,079**	**0.50**

Exclusions based on missing or unavailable data for the data fields shown: 2008/09: 29; 2009/10: 28; 2010/11: 11; 2011/12: 9; 2012/13: 7; 2013/14: 8.

Table AN-7. Use of the FOQ in determining women at high risk of being alpha zero carriers, 2008-14: High prevalence areas

Year	Booking bloods received (BBs)	FOQ attached		MCH < 25pg		High risk alpha0	
	n	n	% of BBs	n	% of FOQ attached	n	% of BBs
2008/09	271,815	51,375	18.90	12,937	25.18	4,031	1.48
2009/10	236,843	119,305	50.37	11,463	9.61	1,179	0.50
2010/11	328,967	255,854	77.77	14,861	5.81	2,185	0.66
2011/12	369,639	317,377	85.86	15,589	4.91	1,911	0.52
2012/13	364,656	326,730	89.60	17,522	5.36	2,089	0.57
2013/14	352,930	322,773	91.46	13,710	4.25	1,742	0.49
Total for six year period	**1,924,850**	**1,393,414**	**72.39**	**86,082**	**6.18**	**13,137**	**0.68**

Exclusions based on missing or unavailable data for the data fields shown: 2008/09: 21; 2009/10: 22; 2010/11: 7; 2011/12: 4; 2012/13: 3; 2013/14: 6.

Table AN-8. Use of the FOQ in determining women at high risk of being alpha zero carriers, 2008-14: Low prevalence areas

Year	Booking bloods received (BBs)	FOQ attached		MCH < 25pg		High risk alpha0	
	n	n	% of BBs	n	% of FOQ attached	n	% of BBs
2008/09	282,401	265,634	94.06	4,889	1.84	786	0.28
2009/10	289,500	274,301	94.75	4,881	1.78	896	0.31
2010/11	330,093	318,787	96.57	5,696	1.79	1,347	0.41
2011/12	323,206	316,674	97.98	5,658	1.79	1,301	0.40
2012/13	326,654	320,522	98.12	6,359	1.98	745	0.23
2013/14	333,002	327,170	98.25	7,067	2.16	867	0.26
Total for six year period	**1,884,856**	**1,823,088**	**96.72**	**34,550**	**1.90**	**5,942**	**0.32**

Exclusions based on missing or unavailable data for the data fields shown: 2008/09: 8; 2009/10: 6; 2010/11: 4; 2011/12: 5; 2012/13: 4; 2013/14: 2.

3.5. Tests not performed due to a known previous result

Current programme guidance states that women do not need to be tested again in the same or a subsequent pregnancy, provided that there are two or more previous results from an accredited laboratory, the red cell indices remain the same and can be used for a reliable interpretation, and the woman's identification has three or more matching data items (see the *Handbook for Laboratories, third edition*, available at http://sct.screening.nhs.uk/standardsandguidelines, for full guidance).

Table AN-9 shows the number and rates of pregnant women who were not tested due to a known previous result in England by region. These figures combine previous screen positive and negative results and exclusions are only made where both of these fields were missing or unavailable. In 2013/14 approximately 3% of women screened had a known previous result, or one in 30 women screened.

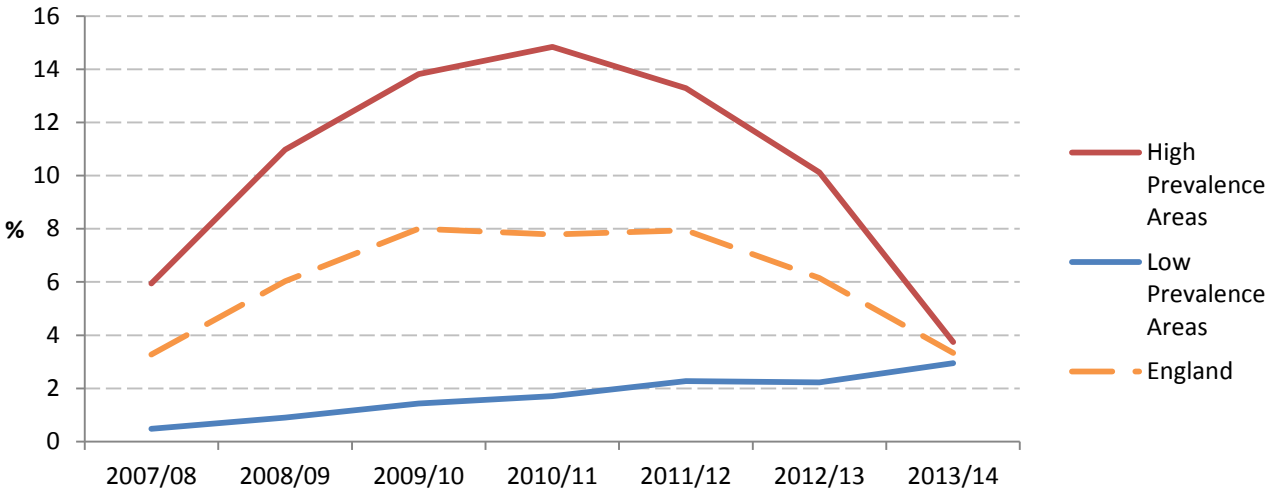
Table AN-9. Pregnant women where testing was not indicated due to a known previous result, 2011-14: England by region

Region	2011/12			2012/13			2013/14		
	Booking bloods received (BBs)	Known previous results	% of BBs	Booking bloods received (BBs)	Known previous results	% of BBs	Booking bloods received (BBs)	Known previous results	% of BBs
East Midlands	49,991	6,492	12.99	40,763	6,191	15.19	52,367	2,972	5.68
East of England	76,421	2,508	3.28	72,604	2,332	3.21	78,659	1,231	1.56
London	100,504	9,016	8.97	79,069	3,006	3.80	68,158	337	0.49
North East	29,059	2,680	9.22	33,288	2,882	8.66	33,171	432	1.30
North West	53,813	3,024	5.62	71,164	2,730	3.84	65,168	1,309	2.01
South Central	54,676	10,353	18.94	44,986	4,033	8.97	44,856	2,019	4.50
South East Coast	38,166	1,224	3.21	45,054	1,277	2.83	47,340	386	0.82
South West	54,831	1,256	2.29	53,555	791	1.48	50,456	687	1.36
West Midlands	72,422	6,728	9.29	68,040	8,298	12.20	75,513	7,926	10.50
Yorkshire and The Humber	68,503	4,249	6.20	50,528	2,832	5.60	44,443	1,388	3.12
England Total	**598,386**	**47,530**	**7.94**	**559,051**	**34,372**	**6.15**	**560,131**	**18,687**	**3.34**

Exclusions based on missing or unknown data on number of booking bloods received or where data on both previous screen positive and previous screen negative were missing or unavailable: 2011/12: 27; 2012/13: 31; 2013/14: 34.

Figure AN-6 shows trends in the proportion of women not tested due to a previously known test result by prevalence and for the whole of England. Rates remain higher in high prevalence areas than in low prevalence areas. Rates appear to have increased in low prevalence areas, but there has been a decline in high prevalence areas. However if the low prevalence outlier shown in Figure AN-7 at 82% is removed, the rate for low prevalence areas decreases to approximately 1.1%. The decrease may be related to the change in guidance on re-testing women in subsequent pregnancies which requires two previous results as described above.

Figure AN-6. Percentage of pregnant women where testing was not indicated due to a known previous test result, 2007-14: England by prevalence

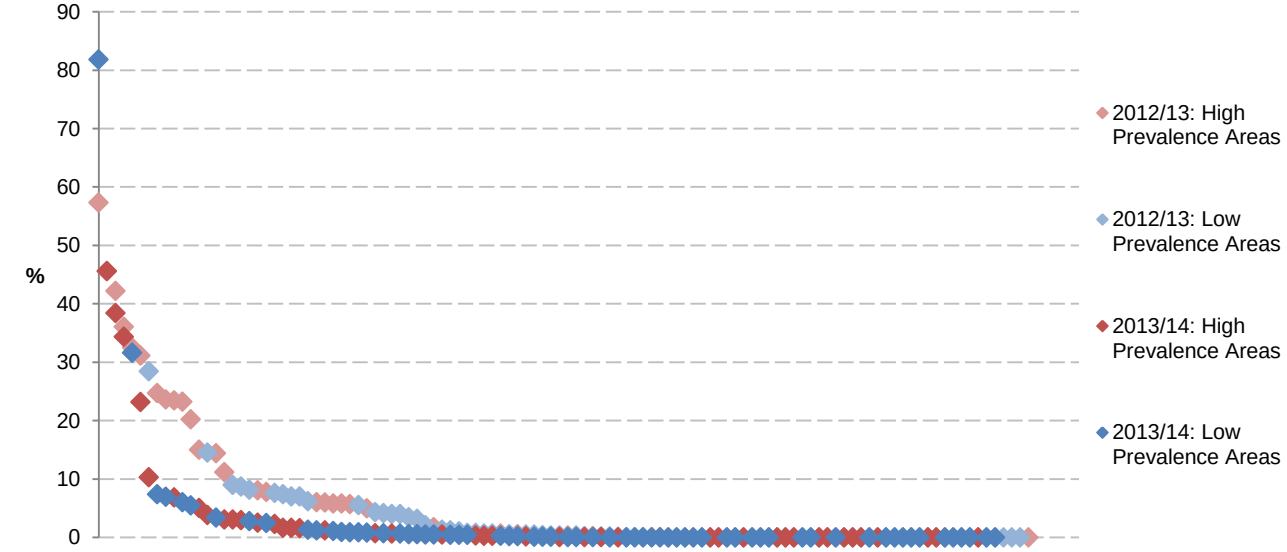


	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14
High prevalence areas	6.0	11.0	13.8	14.8	13.3	10.1	3.7
Low prevalence areas	0.5	0.9	1.4	1.7	2.3	2.2	3.0
England total	3.3	6.0	8.0	7.8	7.9	6.1	3.3

Exclusions based on missing or unavailable data: 2007/08: 35; 2008/09: 50; 2009/10: 45; 2010/11: 33; 2011/12: 27; 2012/13: 31; 2013/14: 34.

Figure AN-7 shows the proportion of women not tested due to a previously known test result by laboratory and by prevalence. The decline seen in Figure AN-6 is seen across all laboratories with the exception of one low prevalence outlier at 82%. A number of laboratories report zero per cent of mothers as having a known previous test result, which could be an indication of laboratories not linking up (or not being able to link) their data to identify previous test results. It may also indicate that laboratories may be re-testing all samples.

Figure AN-7. Percentage of pregnant women where testing was not indicated due to a known previous result, 2012-14: England by laboratory



Exclusions where data on mothers not tested due to a previous test or the total number of booking bloods were missing or unavailable: 2012/13: 31; 2013/14: 34.

3.6. Declined screening tests

Table AN-10 shows the number and proportions of pregnant women that declined antenatal screening for sickle cell and thalassaemia, by region and for the whole of England between 2011 and 2014. In 2013/14 approximately 0.3% booking bloods received nationally were identified as having declined screening, varying between 0.07% and 1.16% regionally.

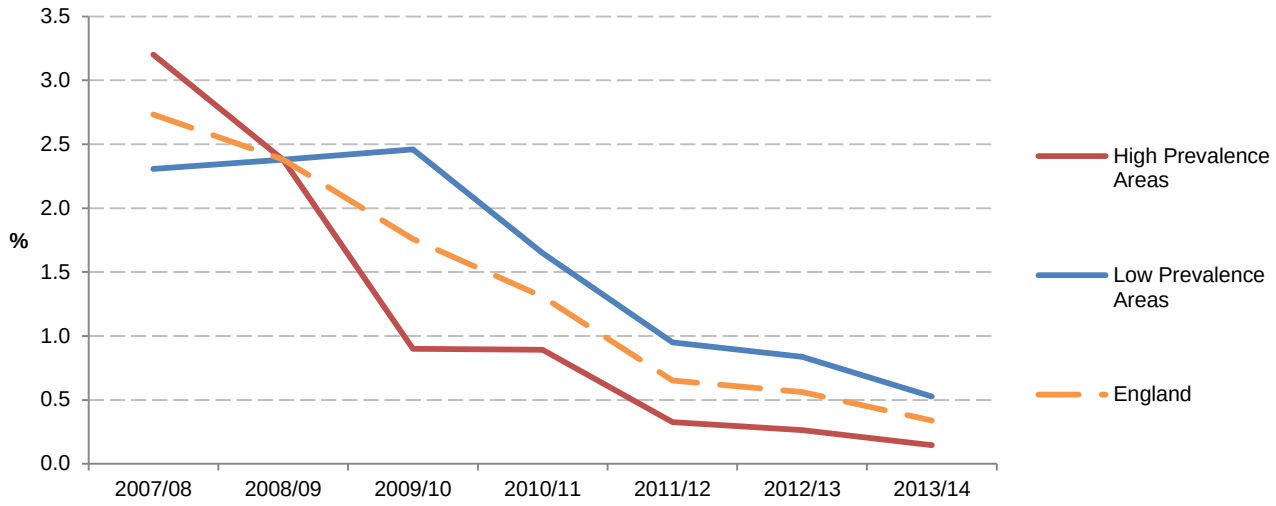
Table AN-10. Declined tests by region, 2011-14: England

Region	2011/12			2012/13			2013/14		
	Booking bloods received (BBS)	Declined testing	% of BBs	Booking bloods received (BBS)	Declined testing	% of BBs	Booking bloods received (BBS)	Declined testing	% of BBs
East Midlands	55,195	164	0.30	53,016	210	0.40	52,367	93	0.18
East of England	81,767	844	1.03	78,123	580	0.74	81,303	426	0.52
London	102,155	184	0.18	103,108	76	0.07	105,994	49	0.05
North East	34,319	385	1.12	33,288	317	0.95	33,171	166	0.50
North West	75,842	477	0.63	75,294	555	0.74	73,911	121	0.16
South Central	50,484	225	0.45	47,740	80	0.17	48,783	79	0.16
South East Coast	54,925	109	0.20	52,783	258	0.49	50,834	67	0.13
South West	58,273	860	1.48	61,527	810	1.32	61,516	715	1.16
West Midlands	74,944	449	0.60	73,336	191	0.26	73,353	50	0.07
Yorkshire and The Humber	72,381	604	0.83	71,773	571	0.80	71,894	448	0.62
England Total	**660,285**	**4,301**	**0.65**	**649,988**	**3,648**	**0.56**	**653,126**	**2,214**	**0.34**

Exclusions based on missing or unavailable data: 2011/12: 14; 2012/13: 14; 2013/14: 15.

Figure AN-8 shows trends in the proportion of booking bloods received where testing was declined between 2007 and 2013, aggregated by prevalence. The proportion of reported declined tests has continued to decrease each year, but remains higher in low prevalence areas than in high prevalence areas.

Figure AN-8. Declined tests as a percentage of booking bloods received, 2007-14: England by prevalence

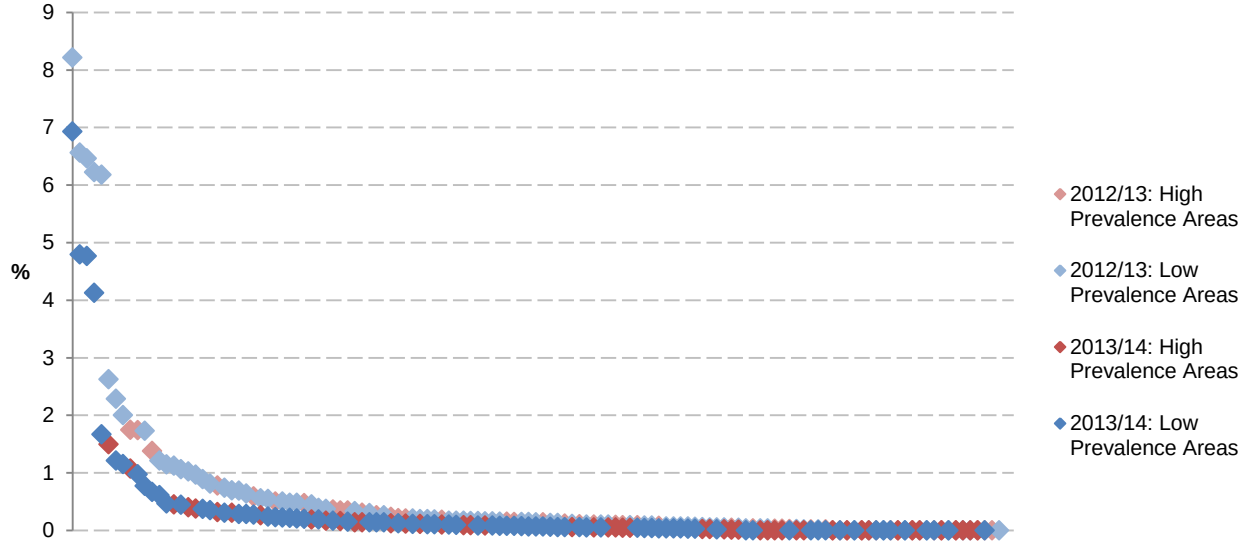


	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14
High prevalence areas	3.20	2.38	0.90	0.89	0.33	0.26	0.15
Low prevalence areas	2.31	2.38	2.46	1.65	0.95	0.84	0.53
England total	2.73	2.38	1.76	1.31	0.65	0.56	0.34

Exclusions based on missing or unavailable data: 2007/08: 40; 2008/09: 47; 2009/10: 32; 2010/11: 17; 2011/12: 14; 2012/13: 14; 2013/14: 15.

Figure AN-9 shows the percentage of pregnant women who declined testing for sickle cell disease and thalassaemia by laboratory and by prevalence for 2012/13 and 2013/14. This shows that the decrease in the proportion of declined tests appears to have occurred across all trusts, including both high and low prevalence areas.

Figure AN-9. Percentage of women that declined testing, 2012-14: England by laboratory



Each marker represents one laboratory.
Exclusions where data on mothers that declined testing or the total number of booking bloods were missing or unavailable: 2012/13: 14; 2013/14: 15.

3.7. Testing of the baby's father

Programme standard AP2ii requires that all fathers of carrier women's babies are to be offered information about counselling and testing. Table AN-11 shows the uptake of father testing by region and across England in 2013/14.

In some regions the number of father specimens requested is greater than the number of screen positive women. This could be due to local variation in policy for father testing. In some cases it may also be the case that midwives collect specimens from both parents if they are both available at the initial booking, before the mother's haemoglobinopathy status is known. Another explanation may be that in cases where the mother's results are inconclusive a specimen is requested from the baby's father. It is possible that the number of father specimens requested for some laboratories does not include cases where father testing was not indicated due to a known previous result.

Table AN-11. Uptake of father testing, 2013/14: England by region

Region	Booking bloods received (BBs)	Screen positive women		Father specimens requested		Father specimens received		'High risk' couples	
	n	n	% of BBs	n	% of screen positive women	n	% of fathers requested	n	% of fathers received
East Midlands	52,367	674	1.29	732	108.61	581	79.37	43	7.40
East of England	81,303	1,120	1.38	1,090	97.32	735	67.43	65	8.84
London	141,535	7,210	5.09	7,259	100.68	3,929	54.13	471	11.99
North East	33,171	220	0.66	217	98.64	186	85.71	12	6.45
North West	73,143	928	1.27	904	97.41	595	65.82	54	9.08
South Central	52,918	814	1.54	800	98.28	690	86.25	54	7.83
South East Coast	58,525	649	1.11	665	102.47	496	74.59	27	5.44
South West	56,531	429	0.76	406	94.64	296	72.91	19	6.42
West Midlands	74,159	1,498	2.02	1,497	99.93	1,058	70.67	89	8.41
Yorkshire and The Humber	71,894	1,042	1.45	944	90.60	678	71.82	64	9.44
England total	**695,546**	**14,584**	**2.10**	**14,514**	**99.52**	**9,244**	**63.69**	**898**	**9.71**

Excludes 8 laboratories where data on the number of booking bloods received, number of screen positive women, data on father specimens requested or received, or the number of 'high risk' couples was missing or unavailable.

In England 14,514 father specimens were reported as requested, which approximately equates to the number of screen positive women (although this does not take into account regional variation and areas reporting more than 100% screen positive women with father specimens requested). Of these, 9,244 father specimens were received by laboratories, or approximately 64% of those requested. Of the father specimens received, 898 (approximately 10%) were identified as being carriers and the pregnancies at high risk for a sickle cell disease or thalassaemia affected baby.

It is not possible to assess the risk status of the pregnancy in cases where the baby's father was not available for testing. These are considered to be 'at risk', but are not included here in the number of 'high risk' couples. These are estimated to account for approximately 36% of screen positive women (calculated from the number of screen positive women minus the number of father specimens received). As a result, the actual number of 'high risk' pregnancies is likely to be higher than the figures shown in these data.

Table AN-12 shows the number and rates of father uptake from 2011/12 to 2013/14 by region. Father uptake varies between regions, ranging between approximately 55% in London and approximately 86% in the North East in 2013/14.

Table AN-12. Uptake of father testing, 2011-14: England by region

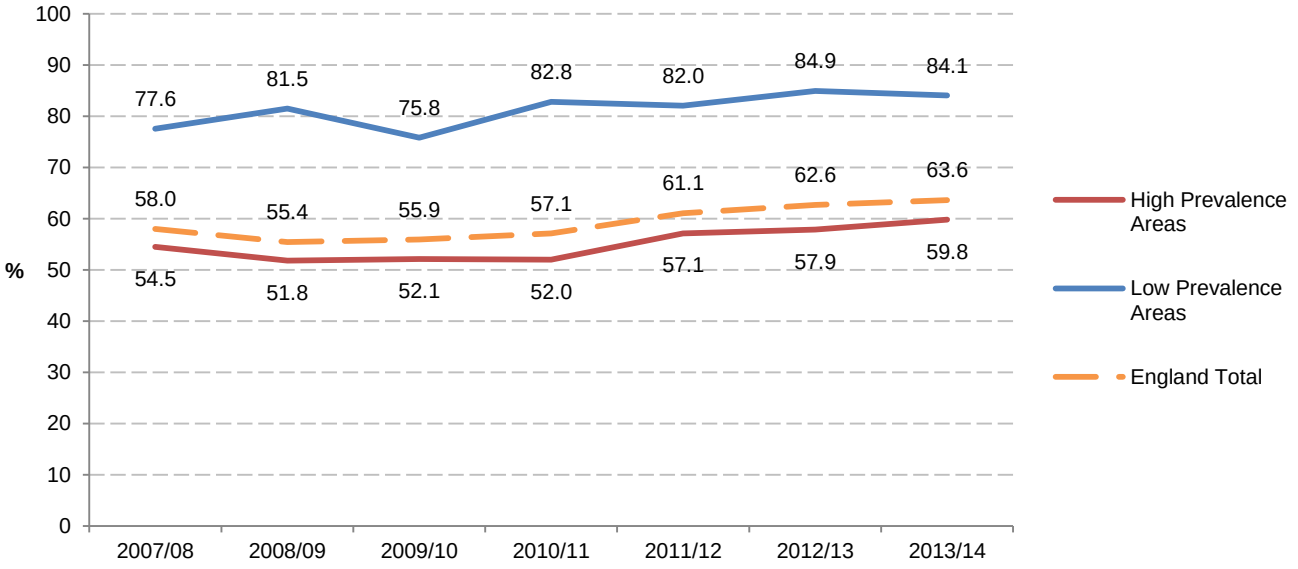
SHA	2011/12			2012/13			2013/14		
	Fathers requested	Fathers received	% uptake	Fathers requested	Fathers received	% uptake	Fathers requested	Fathers received	% uptake
East Midlands	721	577	80.03	713	555	77.84	732	581	79.37
East of England	1,027	694	67.58	1,057	693	65.56	1,090	735	67.43
London	8,296	4,290	51.71	6,218	3,154	50.72	7,705	4,201	54.52
North East	214	187	87.38	256	208	81.25	217	186	85.71
North West	826	583	70.58	996	740	74.30	904	595	65.82
South Central	997	735	73.72	836	631	75.48	800	690	86.25
South East Coast	577	443	76.78	725	543	74.90	665	496	74.59
South West	458	338	73.80	430	338	78.60	406	296	72.91
West Midlands	1,521	872	57.33	1,434	942	65.69	1,497	1,058	70.67
Yorkshire and The Humber	859	746	86.85	890	688	77.30	944	678	71.82
England total	**15,496**	**9,465**	**61.08**	**13,555**	**8,492**	**62.65**	**14,960**	**9,516**	**63.61**

Exclusions based on missing or unavailable data: 2011/12: 6; 2012/13: 6; 2013/14: 6.

Figure AN-10 shows the national rates for father uptake between 2007/08 and 2013/14 by prevalence. Across England there has been a small increase in father uptake each year since 2008/09, from approximately 55% to approximately 64%. Rates appear to have increased in high prevalence areas and father uptake is now at approximately 60%. A small decline can be seen in low prevalence areas in 2013/14, but rates remain higher than in high prevalence areas.

Father uptake figures may have been affected by the change in guidance for re-testing babies' fathers in subsequent pregnancies in line with the guidance for re-testing pregnant women (see 3.5 Tests not performed due to a known previous result). If fathers have been tested previously but decline to have a second test in this pregnancy, these would be included as having declined testing and so the reported rates for father uptake may be lower.

Figure AN-10. Uptake of father testing, 2007-14: England by prevalence



Exclusions based on missing or unavailable data: 2007/08: 19; 2008/09: 10; 2009/10: 7; 2010/11: 3; 2011/12: 6; 2012/13: 6; 2013/14: 6.

The SCT programme requests breakdown data on mother and father results to identify the specific risk of an affected pregnancy. This information also allows us to separate out sickle cell and thalassaemia screen positive results. Table AN-13 shows the risk status of pregnancies for antenatal screening by the mother's results for 2013/14. Not all laboratories have been able to provide these data or to provide complete data, which means that this breakdown does not include all screening encounters in this period. However, of the 15,281 screen positive women identified in antenatal screening, 14,328 (94%) are included in this breakdown, and of the 944 'high risk' couples identified, 817 (87%) are included here. This represents an improvement in data completion for this breakdown when compared to the previous year, where completion was at 93% and 85% respectively.

Of the 14,328 screen positive women included in the breakdown, 7,739 (54%) had clinically significant haemoglobin variants where there was a risk of a baby with sickle cell disease (comprising HbS, HbD, HbC, and HbO^{Arab}). There were 6,145 screen positive women (43%) identified with results that could lead to a thalassaemia affected baby (comprising beta thalassaemia, delta beta thalassaemia, those at high risk of being carriers of alpha zero thalassaemia, HbE and Hb Lepore). The remaining 444 screen positive women (3%) were identified with compound heterozygous results including one of more of the screen positive results for sickle cell or thalassaemia, cases where a donor egg was used or where there was a bone marrow transplant, cases identified with Hereditary Persistence of Fetal Haemoglobin (HPFH), or other haemoglobin variants where testing of the baby's father was recommended.

'High risk' pregnancies are those that are represented by the dark orange boxes in the breakdown table in Appendix Two. Low risk pregnancies are represented by the light orange boxes, and minimal risk pregnancies are represented by the white boxes in the breakdown.

Table AN-13. Breakdown of pregnancy risk for screen positive women, 2013/14: England

		Risk to pregnancy					Totals		
	Mother's screening result	High Risk	Low Risk	Minimal Risk	Father not a carrier	Father result not available	Total number of mothers with result	Total for group	Rate/1000 BBs received
Possible sickle cell affected baby	Hb S	567	8	67	2,386	2,791	5,819	7,739	10.59
	Hb D	*	-	42	541	144	728		
	Hb C	63	-	49	508	564	1,184		
	Hb O-Arab	*	*	*	5	3	8		
Possible beta thalassaemia affected baby	βThalassaemia	142	*	63	2,539	936	3,680	4,763	6.52
	δβ thalassaemia	*	*	*	28	11	41		
	Hb E	12	*	59	730	224	1,026		
	Hb Lepore	*	*	*	6	10	16		
Possible alpha thalassaemia affected baby	High risk alpha0	23	-	25	528	806	1,382	1,382	1.89
Other clinically significant mother results	HPFH/Compound heterozygous/donor egg/bone marrow transplant	9	4	23	272	128	436	436	0.60
Other Hb variants requiring testing of baby's father	Other Hb variants requiring testing of baby's father	-	-	*	8	*	8	8	0.01
	Totals	817	14	329	7,551	5,617	14,328	14,328	

Note: Mothers' results combine both carrier and affected results

**Numbers less than 3 have been suppressed*

Not all laboratories were able to provide complete breakdown data for all screen positive women. For comparison, the total number of screen positive women reported by laboratories was 15,281 (94% included here) and 944 high risk couples (87% included here). The figure for rate per 1000 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.

4. Prenatal diagnostic (PND) testing data

4.1. Response rates and data quality

Response rate:

Data were received from all three PND laboratories, including pregnancy outcome data for the period 1st April 2013 – 31st March 2014. We would like to thank the PND laboratories for their efforts in submitting this data.

Data quality:

The proportion of PND tests with an unknown or missing gestation has continued to decrease, and in 2013/14 approximately 99.9% of tests had information on gestation at time of test. However, the proportion of PND tests resulting in an affected pregnancy that had a known pregnancy outcome (i.e. whether the couple continued the pregnancy, miscarried, or opted for termination) has continued to decrease. In 2013/14 approximately 44% of affected results had an unknown outcome, compared to 39% in 2012/13, 34% in 2011/12, and 20% in 2010/11.

4.2. Numbers tested and detected

Table PND-1 shows the number of PND tests performed by each PND laboratory by year since 2007/08. PND tests for patients who were outside of England or who were private patients were excluded. Table PND-2 shows the number of PND tests performed by the mother's region and by year.

Table PND-1. Number of PNDs performed, 2007-14: England by laboratory

PND laboratory	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14
King's College Hospital	86	101	119	141	134	125	104
Oxford	130	166	159	159	149	161	140
University College London Hospital	109	119	118	120	135	112	109
Total	**325**	**386**	**396**	**420**	**418**	**398**	**353**

Table PND-2. Number of PNDs performed, 2007-14: England by regions

Region	2007/08		2008/09		2009/10		2010/11		2011/12		2012/13		2013/14	
	n	%	n	%	n	%	n	%	n	%	n	%	n	%
East Midlands	14	4.3	-	0.0	9	2.3	18	4.3	12	2.9	4	1.0	3	0.8
East of England	23	7.1	20	5.2	30	7.6	30	7.1	22	5.3	21	5.3	16	4.5
London	205	63.1	183	47.4	229	57.8	266	63.3	249	59.6	195	49.0	189	53.5
North East	5	1.5	-	0.0	8	2.0	4	1.0	4	1.0	*	0.3	*	0.3
North West	18	5.5	-	0.0	26	6.6	22	5.2	21	5.0	*	0.3	*	0.6
South Central	16	4.9	3	0.8	15	3.8	15	3.6	12	2.9	*	0.3	4	1.1
South East Coast	5	1.5	9	2.3	9	2.3	12	2.9	12	2.9	17	4.3	7	2.0
South West	*	0.6	-	0.0	4	1.0	11	2.6	4	1.0	*	0.3	*	0.3
West Midlands	18	5.5	-	0.0	28	7.1	16	3.8	21	5.0	3	0.8	5	1.4
Yorkshire and the Humber	10	3.1	*	0.3	15	3.8	16	3.8	11	2.6	4	1.0	*	0.3
Unknown Region	9	2.8	170	44.0	23	5.8	10	2.4	50	12.0	150	37.7	124	35.1
England total	**325**	**100.0**	**386**	**100.0**	**396**	**100.0**	**420**	**100.0**	**418**	**100.0**	**398**	**100.0**	**353**	**100.0**

Numbers may differ from those presented in previous years as the regions are derived from CCGs rather than PCTs as in previous years.
**Number less than three have been suppressed.*

Table PND-3 shows the number of affected, carrier, and 'NAD' (no abnormality detected) results by year. The figures each year reflect the expected 25:50:25 ratio of affected, carrier, and NAD results respectively. Table PND-4 breaks these figures down by PND result or risk to the pregnancy.

Table PND-3. PND fetal results, 2007-14: England

Fetal result	2007/08		2008/09		2009/10		2010/11		2011/12		2012/13		2013/14	
	n	%	n	%	n	%	n	%	n	%	n	%	n	%
Affected	90	27.7	84	21.8	102	25.8	95	22.6	101	24.2	85	21.4	89	25.2
Carrier	152	46.8	201	52.1	198	50.0	224	53.3	207	49.5	213	53.5	163	46.2
NAD	78	24.0	96	24.9	96	24.2	100	23.8	104	24.9	96	24.1	100	28.3
Inconclusive/ Missing result	5	1.5	5	1.3	*	0.0	*	0.2	6	1.4	4	1.0	*	0.3
England total	**325**	**100.0**	**386**	**100.0**	**396**	**100.0**	**420**	**100.0**	**418**	**100.0**	**398**	**100.0**	**353**	**100.0**

**Numbers less than three have been suppressed.*

Table PND-4. Breakdown of PND fetal results by condition, 2007-14: England

Fetal result	PND result/risk	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13	2013/14
Affected	Sickle Cell affected	66	60	79	66	85	68	69
	Thalassaemia affected	24	24	23	29	15	17	19
	Other	*	*	*	*	*	*	*
Carrier	Sickle Cell carrier	110	150	152	174	154	169	124
	Thalassaemia carrier	42	51	46	50	52	42	39
	Other	*	*	*	*	*	*	*
NAD	Risk for Sickle Cell	33	58	72	80	63	52	77
	Risk for Thalassaemia	22	25	24	20	6	18	22
	Risk not known	23	13	*	*	35	26	*
Inconclusive†	All risks	5	5	*	*	6	4	*
Total		325	386	396	420	418	398	353

†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.
Alpha and beta thalassaemia cases are grouped due to the small number of alpha thalassaemia cases.
**Numbers less than three have been suppressed.*

4.3. Gestation data

Programme standard AO1requires a minimum of 50% of all prenatal diagnoses to be performed by 12 weeks and six days of gestation, and 75% as an achievable standard. Table PND-5 shows the gestation at which PND tests were performed between 2007/08 and 2013/14.

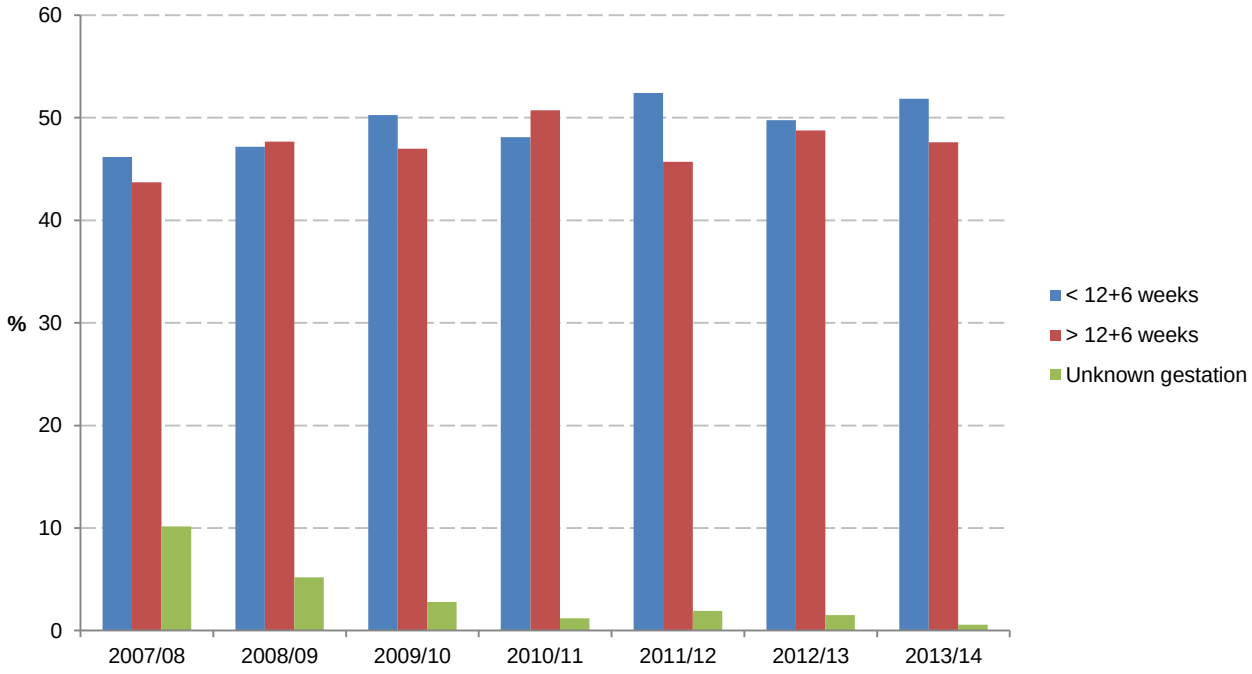
Table PND-5. Gestation at sample for PND, 2007-14: England

Gestation	2007/08		2008/09		2009/10		2010/11		2011/12		2012/13		2013/14	
	n	%	n	%	n	%	n	%	n	%	n	%	n	%
<12+6 weeks	150	46.2	182	47.2	199	50.3	202	48.1	219	52.4	198	49.7	183	51.8
13+0 - 14+6 weeks	49	15.1	65	16.8	76	19.2	105	25.0	93	22.2	71	17.8	63	17.8
≥15+0 weeks	93	28.6	119	30.8	110	27.8	108	25.7	98	23.4	123	30.9	105	29.7
Unknown gestation	33	10.2	20	5.2	11	2.8	5	1.2	8	1.9	6	1.5	*	0.6
Total	325	100.0	386	100.0	396	100.0	420	100.0	418	100.0	398	100.0	353	100.0

**Numbers less than three have been suppressed.*

Figure PND-1 shows the proportion tested before and after 12 weeks and six days of gestation, and the proportion with an unknown gestation. The proportions have been fairly steady each year with approximately half tested by 12 weeks and six days. It is important to continue efforts to ensure that the acceptable standard is maintained and to work towards the achievable standards of 75% in order to ensure early testing to facilitate parents to make informed choices. The proportion with an unknown gestation has decreased each year, indicating an improvement in the quality of gestation data.

Figure PND-1. Percentage of PND tests performed by gestation and year, 2007-14: England



4.4. Results by ethnicity

Table PND-6 shows the number of PND tests performed each year by the mother's ethnic grouping (as reported to the laboratories) between 2007/08 and 2013/14. As in previous years, mothers from an African background accounted for approximately half of all PND tests performed in 2013/14. In 2013/14 it appears that the proportion identified with a Caribbean background has increased, but this may be due to differences in how their ethnic background was described as the number in the 'mixed/other' category appears to have decreased by a similar proportion. Mixed ethnicities can include combinations of ethnic groupings which prevents categorisation into these groupings. Those with an unknown ethnicity account for nearly 20% of PND tests performed in 2013/14.

Table PND-6. Number of PND tests by mother's ethnicity, 2007-14: England

Mother's ethnic grouping	2007/08		2008/09		2009/10		2010/11		2011/12		2012/13		2013/14	
	n	% of total	n	% of total	n	% of total	n	% of total	n	% of total	n	% of total	n	% of total
African	158	48.6	184	47.7	212	53.5	240	57.1	211	50.5	180	45.2	162	45.9
Caribbean	15	4.6	22	5.7	22	5.6	23	5.5	12	2.9	14	3.5	61	17.3
Indian	18	5.5	9	2.3	14	3.5	16	3.8	9	2.2	9	2.3	3	0.8
Pakistani	16	4.9	13	3.4	16	4.0	19	4.5	*	0.5	*	0.5	4	1.1
Cypriot/Mixed Cypriot	10	3.1	10	2.6	10	2.5	11	2.6	7	1.7	4	1.0	6	1.7
Other Asian	15	4.6	22	5.7	23	5.8	16	3.8	50	12.0	16	4.0	35	9.9
Southern & Other European	6	1.8	6	1.6	6	1.5	5	1.2	3	0.7	8	2.0	6	1.7
Middle Eastern	10	3.1	9	2.3	12	3.0	5	1.2	4	1.0	4	1.0	4	1.1
Mixed/Other	*	0.6	8	2.1	*	0.3	8	1.9	97	23.2	77	19.3	11	3.1
Not Know n	75	23.1	103	26.7	80	20.2	77	18.3	23	5.5	84	21.1	61	17.3
Total	**325**	**100.0**	**386**	**100.0**	**396**	**100.0**	**420**	**100.0**	**418**	**100.0**	**398**	**100.0**	**353**	**100.0**

**Numbers less than three have been suppressed.*

4.5. Pregnancy outcomes

Data collection for pregnancy outcomes for PND testing began in 2008/09. Table PND-7 shows the number of PND tests with each pregnancy outcome where an affected pregnancy was identified, and Table PND-8 shows these figures separated out by condition. Completeness of outcome data has decreased in recent years (see 4.1 Response rates and data quality) and in 2013/14 44% of affected results had missing outcome data.

Table PND-7. Number of PND tests with affected results by outcome, 2008-14: England

Outcome	2008/09		2009/10		2010/11		2011/12		2012/13		2013/14	
	n	%	n	%	n	%	n	%	n	%	n	%
Continued	9	10.8	19	18.6	26	27.4	18	18.0	13	15.7	13	14.6
Terminated	27	32.5	41	40.2	50	52.6	48	48.0	38	45.8	37	41.6
Not Known	47	56.6	42	41.2	19	20.0	34	34.0	32	38.6	39	43.8
Total	**83**	**100.0**	**102**	**100.0**	**95**	**100.0**	**100**	**100.0**	**83**	**100.0**	**89**	**100.0**

Cases where the pregnancy miscarried have been excluded due to the small numbers involved.

Table PND-8. Outcomes for pregnancies with affected fetal diagnoses at PND, 2008-14: England by condition

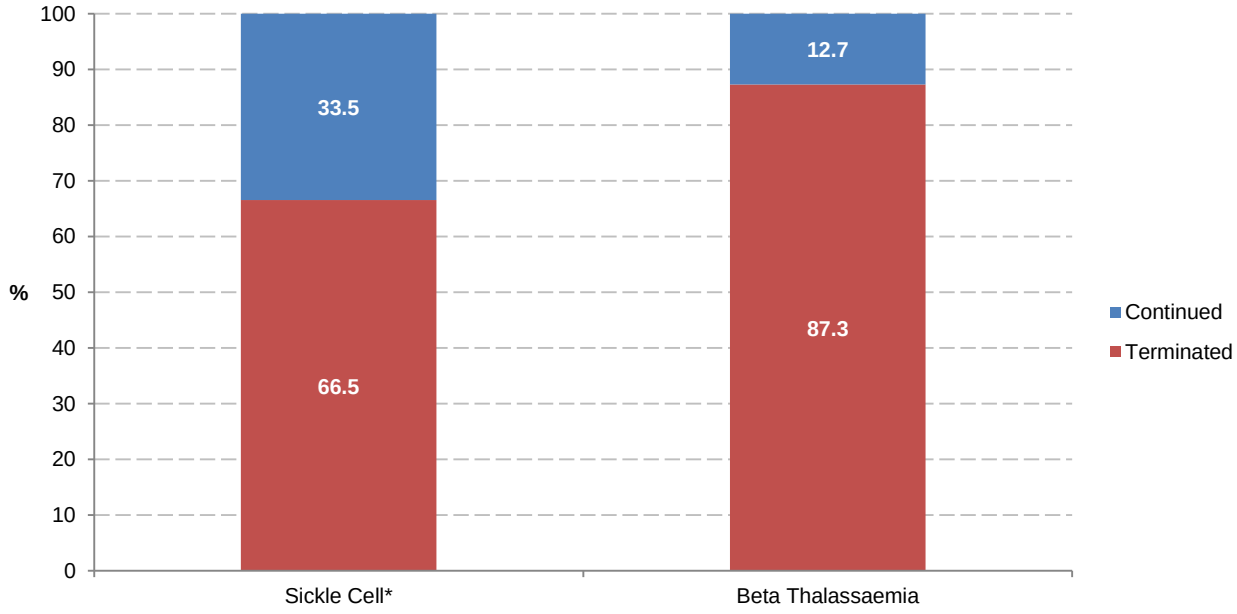
Condition	Pregnancy outcome	2008/09		2009/10		2010/11		2011/12		2012/13		2013/14	
		n	% of total identified with condition	n	% of total identified with condition	n	% of total identified with condition	n	% of total identified with condition	n	% of total identified with condition	n	% of total identified with condition
Sickle Cell	Continued	8	13.3	18	22.8	21	31.8	17	20.0	12	17.6	12	17.4
	Terminated	17	28.3	30	38.0	31	47.0	38	44.7	28	41.2	31	44.9
	Miscarried	*	1.7	*	0.0	*	0.0	*	0.0	*	2.9	*	0.0
	Not Known	34	56.7	31	39.2	14	21.2	30	35.3	26	38.2	26	37.7
Beta Thalassaemia	Continued	*	0.0	*	4.8	4	16.0	*	8.3	*	7.1	*	6.3
	Terminated	8	38.1	9	42.9	17	68.0	8	66.7	8	57.1	5	31.3
	Miscarried	*	0.0	*	0.0	*	0.0	*	8.3	*	0.0	*	0.0
	Not Known	13	61.9	11	52.4	4	16.0	*	16.7	5	35.7	10	62.5
Alpha Thalassaemia	Continued	*	33.3	*	0.0	*	25.0	*	0.0	*	0.0	*	0.0
	Terminated	*	66.7	*	100.0	*	50.0	*	66.7	*	66.7	*	33.3
	Miscarried	*	0.0	*	0.0	*	0.0	*	0.0	*	0.0	*	0.0
	Not Known	*	0.0	*	0.0	*	25.0	*	33.3	*	33.3	*	66.7
Total Affected		**84**		**102**		**95**		**101**		**85**		**89**	

**Numbers less than three have been suppressed*
Affected results for other haemoglobinopathies have been excluded.

Figure PND-2 shows the proportion of PND tests with an affected result where parents opted to continue the pregnancy or opted to terminate the pregnancy, using aggregated data covering 2008/09 – 2013/14. Alpha thalassaemia results and miscarriages are excluded as the numbers are small, as are cases where the pregnancy outcome was not known.

Of the PND tests performed where the outcome was known (56% of affected results), approximately 33.5% of cases with sickle cell affected results opted to continue the pregnancy while 66.5% opted to terminate. Approximately 13% of cases with beta thalassaemia affected results opted to continue the pregnancy while 87% opted to terminate.

Figure PND-2. Outcomes for pregnancies with affected fetus diagnosis at PND, 2008-14: England by condition



The "Sickle Cell" category includes cases where the result was 'sickle cell and thalassaemia'
Excludes alpha thalassaemia cases, miscarriage outcomes, and 213 cases where pregnancy outcome was not known.

Table PND-9 shows pregnancy outcomes by gestation at which the PND test was performed for affected sickle cell and beta thalassaemia results. This includes data between 2008/09 and 2013/14. Where couples opted to terminate the pregnancy following a beta thalassaemia affected result, the majority were tested by 12 weeks and six days gestation (40 tested by 12 weeks and six days compared to 14 tested at 13 weeks or later). For sickle cell affected results where the parents opted to terminate, 82 were tested by 12 weeks and six days and 89 were tested at 13 weeks or later.

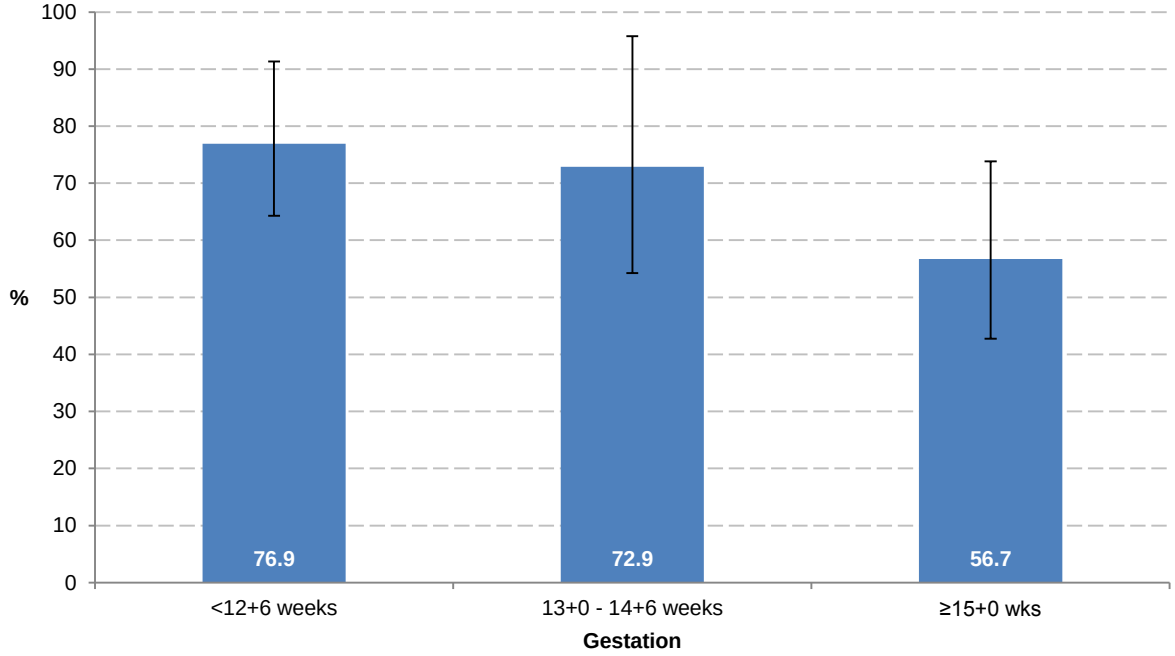
Table PND-9. Gestation at PND for affected results with a known outcome, 2008-14: England

		<12+6 weeks	13+0 - 14+6 weeks	≥15+0 wks
Condition	Outcome	n	n	n
Sickle Cell	Continued	29	17	40
	Terminated	82	40	49
Beta Thalassaemia	Continued	7	*	*
	Terminated	40	9	5

**Numbers less than three have been suppressed.*

Figure PND-3 shows the proportion of affected results where the parents opted to terminate the pregnancy, shown with 95% confidence intervals by gestation for all conditions. These data indicate that the later in pregnancy that PND tests are performed, the lower the percentage of parents that opt to terminate the pregnancy.

Figure PND-3. Percentage of affected results where parents opted to terminate, by gestation at PND for known pregnancy outcomes, 2008-14



Excludes cases where the gestation at PND was unknown, and cases where the pregnancy outcome was unknown.

5. Newborn screening data

5.1. Response rates and data quality

Response rate:

Data were received from all 13 newborn screening laboratories in England. We would like to thank all those involved in collecting and submitting these data to the screening programme.

Data quality:

Newborn laboratories report on 'results' which may differ from the number of babies tested. Data by region and by ethnicity are collected separately, which can lead to discrepancies when comparing the figures. Potential causes for these differences can include samples from outside of England being excluded in the regional data where these could be identified, but not in the ethnicity data. For the 2013/14 data collection year the data fields requested were expanded to include screening outcome data. As this was the first year that outcome data were requested in this format, we asked that laboratories provide this information if they can. This data will be fed back to the laboratories and will be used to assess the suitability of the data fields requested, but these data is not included here.

5.2. Numbers screened

Laboratory data report 668,117 babies screened in 2013/14. Birth figures from the Office of National Statistics (ONS) can offer a point of comparison to the laboratory data to provide validation for numbers screened, shown in Table NB-1. Nationally there is a discrepancy of 0.54% between these two datasets. This discrepancy could be accounted for in part by the different periods covered (the ONS data covers the 2013 calendar year, whereas the laboratory data covers the 2013/14 financial year 1 April – 31 March); by declined screening tests; and babies reported with an unknown region. The 'unknown' region category includes cases where the laboratories provide figures as 'out of region' or 'unknown region' and so cannot be attributed to a specific region. Laboratory data may include repeat tests and babies born abroad who moved to England up to one year of age.

Table NB-1. Comparison of ONS birth figures and number of babies screened as reported by newborn screening laboratories, 2013/14: England by region

Region	Data from newborn laboratories*	ONS figures†	Discrepancy (%)
East of England	69,082	71,309	3.12
East Midlands	48,382	52,895	8.53
London	130,373	128,332	-1.59
North East	27,896	28,961	3.68
North West	84,384	86,372	2.30
South East	103,123	102,190	-0.91
South West	57,940	58,710	1.31
West Midlands	70,773	71,188	0.58
Yorkshire and The Humber	67,820	64,560	-5.05
Unknown Region	8,344	-	-
England Total	**668,117**	**664,517**	**-0.54**

**Data collected from the 13 newborn laboratories in England. This data covers the financial year 2013/14.*

†Data from ONS (Live Births by Area of Usual Residence 2013, found at http://www.ons.gov.uk/ons/publications/re-reference-tables.html?edition=tcm%3A77-320857). This data covers the 2013 calendar year.

5.3. Newborn screening results

Laboratory data for 2013/14 identified 668,117 babies as having been screened, of whom 319 (one in 2,094) were identified as screen positive for a significant condition, and 8,850 (one in 75) were identified as carriers. Table NB-2 shows a breakdown of newborn screening results by haemoglobinopathy result for 2013/14 including both significant conditions and carrier results.

Table NB-2. Babies screened and newborn screening results, 2013/14: England by region

	Significant Conditions					Carriers					Total Screened		
Region	FS	FSC	FS-Other	FE	F-only	FAS	FAC	FAD	FAE	Other Carrier	Transfused	Declined	Normal+ Abnormal
East Midlands	10	4	*	*	*	241	61	46	35	*	94	*	48,382
East of England	16	5	*	*	4	393	95	49	74	18	22	124	69,082
London	128	49	3	10	10	3,081	688	230	407	4	522	161	130,373
North East	*	*	*	*	*	54	8	18	31	23	36	18	27,896
North West	15	*	*	*	5	462	83	80	91	5	137	75	84,384
South Central	14	3	*	*	3	298	70	50	63	*	33	32	51,413
South East Coast	8	*	*	*	*	226	42	48	53	*	619	49	51,710
South West	4	*	*	*	*	167	50	45	41	*	30	54	57,940
West Midlands	16	3	*	*	4	511	131	124	115	*	115	93	70,773
Yorkshire and the Humber	7	3	3	*	5	266	41	74	68	*	96	45	67,820
Unknown region	*	*	*	*	*	56	19	*	7	*	236	21	8,344
England Total	219	72	8	20	32	5,755	1,288	766	985	56	1,940	672	668,117

**Numbers less than three have been suppressed.*

Significant conditions:

Significant conditions comprise FS, FSC, FS-Other and FE. Table NB-3 shows the number and rates of babies identified with a significant condition for each year between 2011 and 2014.

In 2013/14 there were 319 babies identified with a significant condition, which equates to 0.48 per 1000 babies screened or one in 2,094 babies screened. Prior to 2011/12 there were approximately 360 babies identified with a significant condition through newborn screening each year, which equates to between 0.52 and 0.56 per 1000 babies screened each year between 2005 and 2011.

Rates across England range between 0.07 per 1000 (one in 13,948) babies screened in the North East and 1.46 per 1000 (one in 686) babies screened in London. Babies born with a significant condition were identified in all regions in England, although approximately 60% of these babies were in London.

Newborn screening does not specifically test for beta thalassaemia major. However, F-only cases are probable beta thalassaemia cases and require follow-up testing. There are approximately 20 to 30 F-only cases reported each year, and in 2013/14 there were 32 cases reported.

Figure NB-1 shows the geographical prevalence of babies identified with a significant condition per 1000 babies screened in 2013/14.

Table NB-3. Trends in the number of babies identified with significant conditions, 2011-14: England by region

Region	2011/12			2012/13			2013/14		
	n	Total screened	Rate/ 1000	n	Total screened	Rate/ 1000	n	Total screened	Rate/ 1000
East Midlands	8	50,901	0.16	8	49,898	0.16	16	48,382	0.33
East of England	18	72,091	0.25	21	72,421	0.29	21	69,082	0.30
London	205	133,245	1.54	195	131,424	1.48	190	130,373	1.46
North East	*	28,122	0.07	*	28,966	0.03	*	27,896	0.07
North West	21	88,405	0.24	25	87,369	0.29	19	84,384	0.23
South Central	18	54,286	0.33	13	53,352	0.24	20	51,413	0.39
South East Coast	8	53,521	0.15	7	51,682	0.14	9	51,710	0.17
South West	4	60,811	0.07	7	59,938	0.12	8	57,940	0.14
West Midlands	21	72,970	0.29	21	72,559	0.29	20	70,773	0.28
Yorkshire and the Humber	10	69,457	0.14	11	69,613	0.16	13	67,820	0.19
Unknown region	5	9,469	0.53	3	8,216	0.37	*	8,344	0.12
England total	**320**	**693,278**	**0.46**	**312**	**685,438**	**0.46**	**319**	**668,117**	**0.48**

**Numbers less than three have been suppressed.*

Figure NB-1. Babies identified with a significant condition per 1000 babies screened, 2013/14: England by region

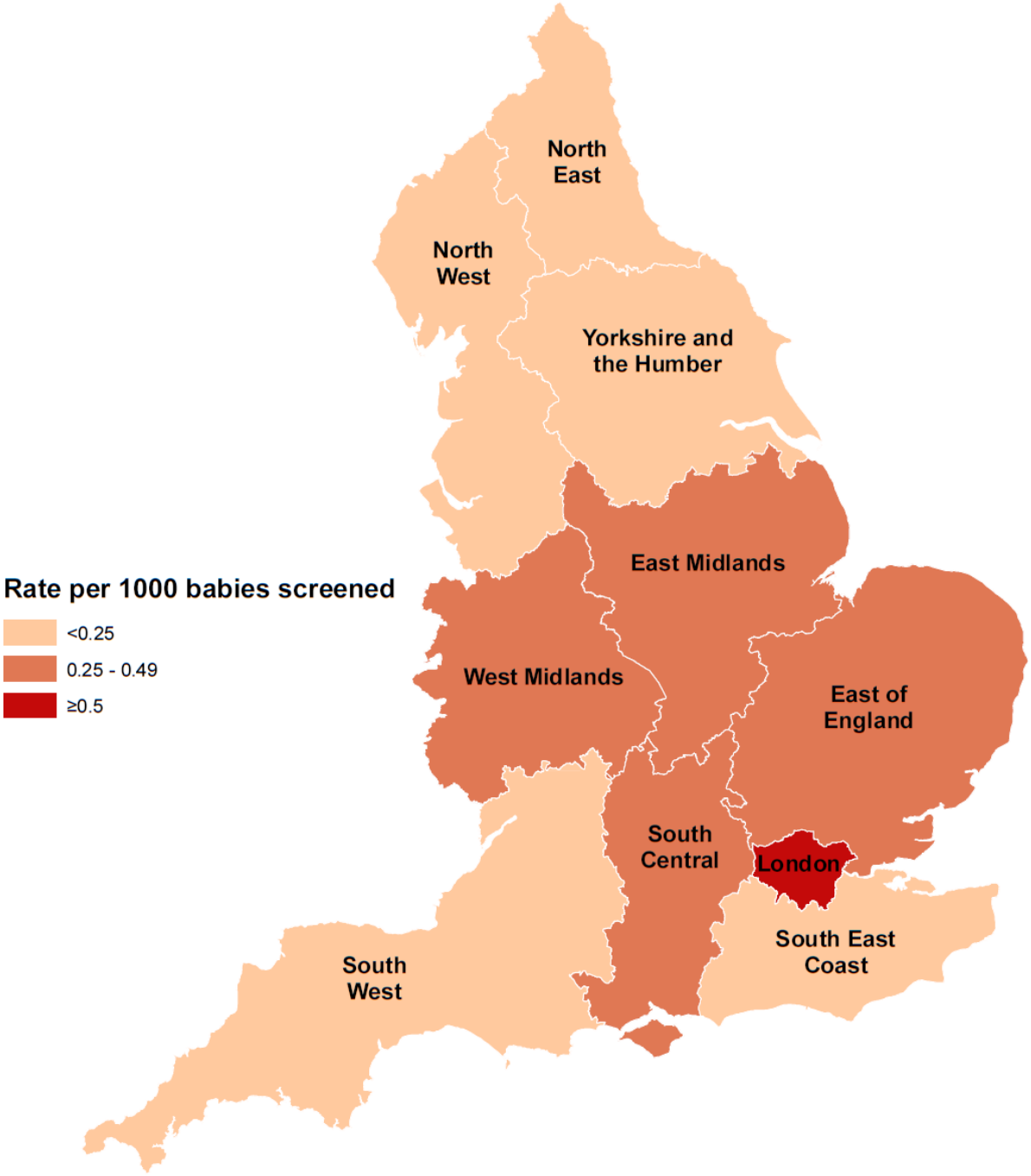
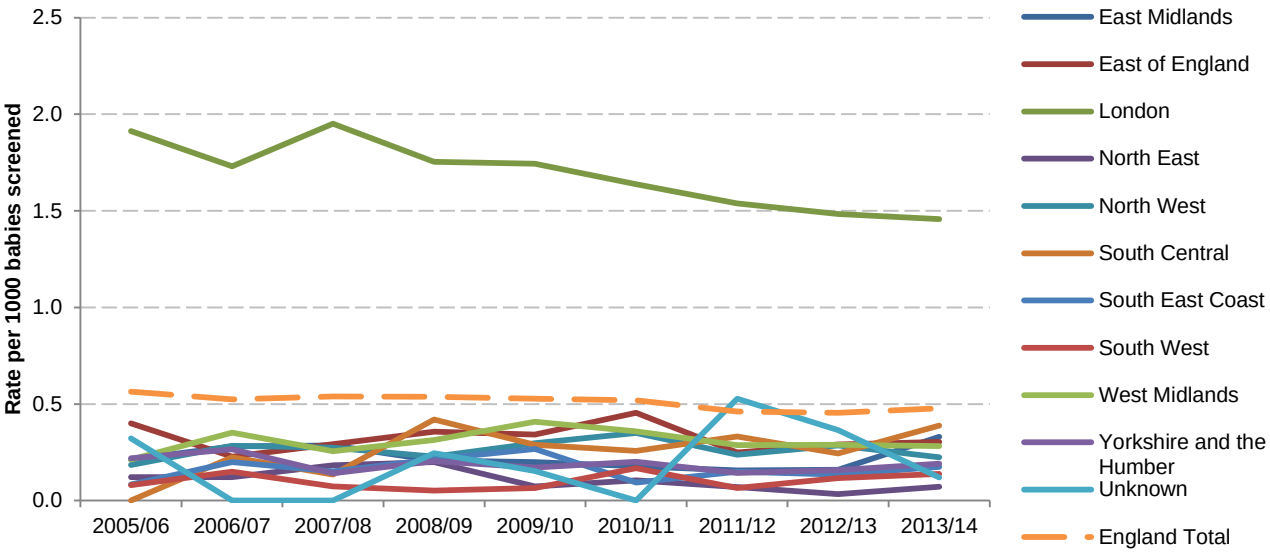


Figure NB-2 shows the trends in rates of babies identified with a significant condition for England by region. Figure NB-3 compares the rates for London with those for the rest of England (including cases where the region is unknown).

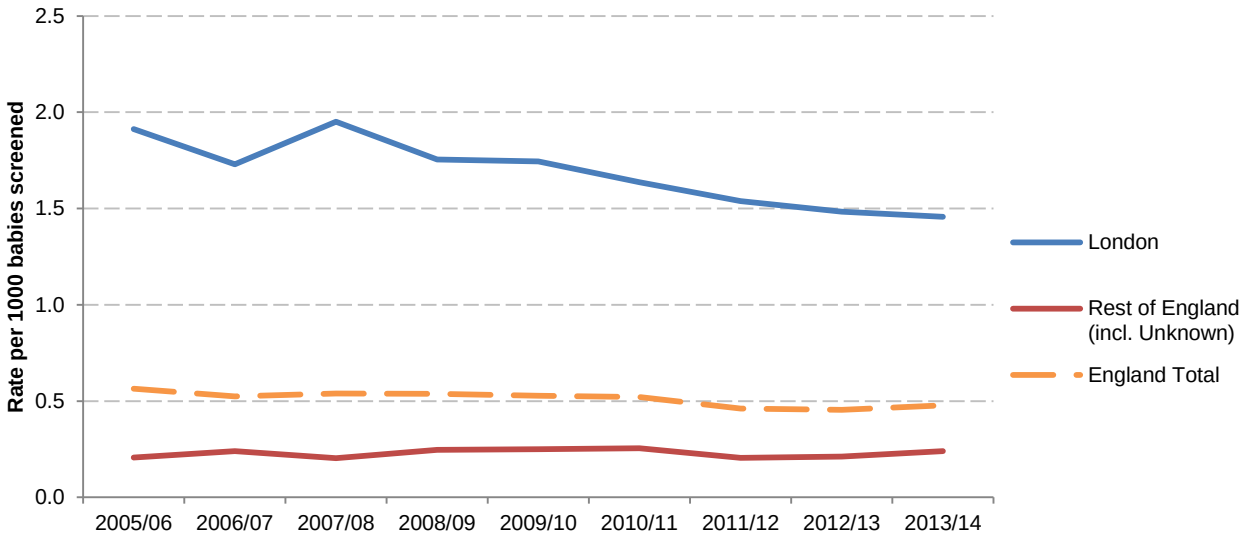
The rates for the whole of England appear steady at approximately 0.5 per 1000 babies screened with some slight variation between years. The rates in London are consistently higher than in the rest of England, although there appears to be an overall decline in rates for London.

Figure NB-2. Trends in babies identified with a significant condition, 2005-14: England by region



Significant conditions comprise FS,FSC, FS Other and FE.
**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-3. Trends in babies identified with a signficant condition, 2005-14: London and the rest of England



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

Carriers:

Carrier results comprise FAS, FAC, FAD, FAE and other haemoglobin variants. Table NB-4 shows the number and rates of babies with a carrier result between 2011 and 2014. In 2013/14 there were 8,850 babies identified as carriers, which equates to 13.25 per 1000 babies screened or one in 76 babies screened. Rates across England ranged between approximately one in 208 babies screened in the North East and one in 30 babies screened in London.

Figure NB-4 shows the geographical prevalence of babies identified with a carrier result per 1000 babies screened in 2013/14 by region.

Table NB-4. Trends in the number of babies identified with carrier results, 2011-14: England by region

Region	2011/12			2012/13			2013/14		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
East Midlands	398	50,901	7.82	444	49,898	8.90	383	48,382	7.92
East of England	728	72,091	10.10	706	72,421	9.75	629	69,082	9.11
London	4,778	133,245	35.86	4,679	131,424	35.60	4,410	130,373	33.83
North East	122	28,122	4.34	157	28,966	5.42	134	27,896	4.80
North West	817	88,405	9.24	665	87,369	7.61	721	84,384	8.54
South Central	542	54,286	9.98	564	53,352	10.57	481	51,413	9.36
South East Coast	404	53,521	7.55	357	51,682	6.91	369	51,710	7.14
South West	351	60,811	5.77	323	59,938	5.39	305	57,940	5.26
West Midlands	939	72,970	12.87	900	72,559	12.40	881	70,773	12.45
Yorkshire and the Humber	529	69,457	7.62	477	69,613	6.85	450	67,820	6.64
Unknown	110	9,469	11.62	96	8,216	11.68	87	8,344	10.43
England Total	**9,718**	**693,278**	**14.02**	**9,368**	**685,438**	**13.67**	**8,850**	**668,117**	**13.25**

Figure NB-4. Babies identified with a carrier result per 1000 babies screened, 2013/14: England by region

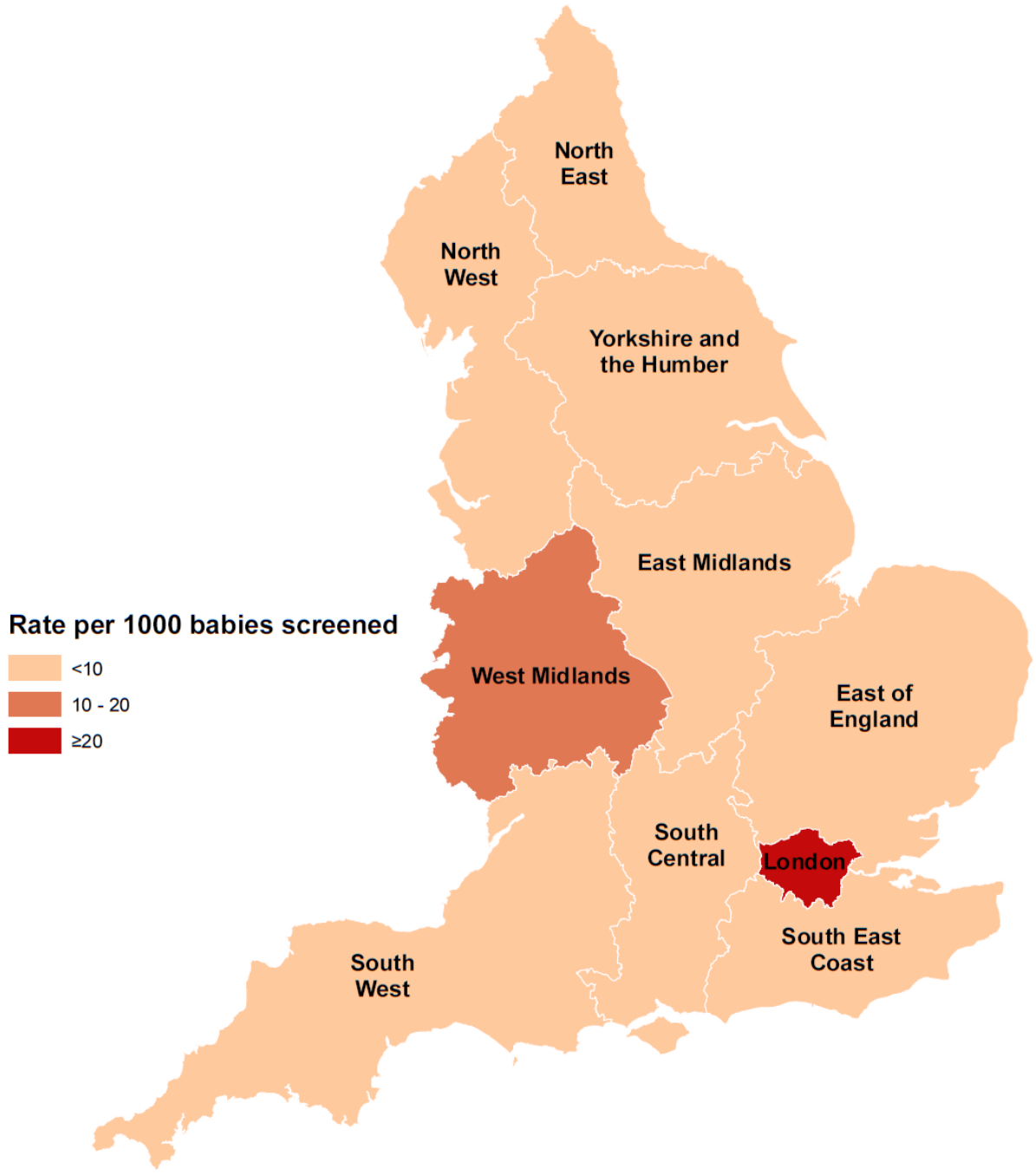
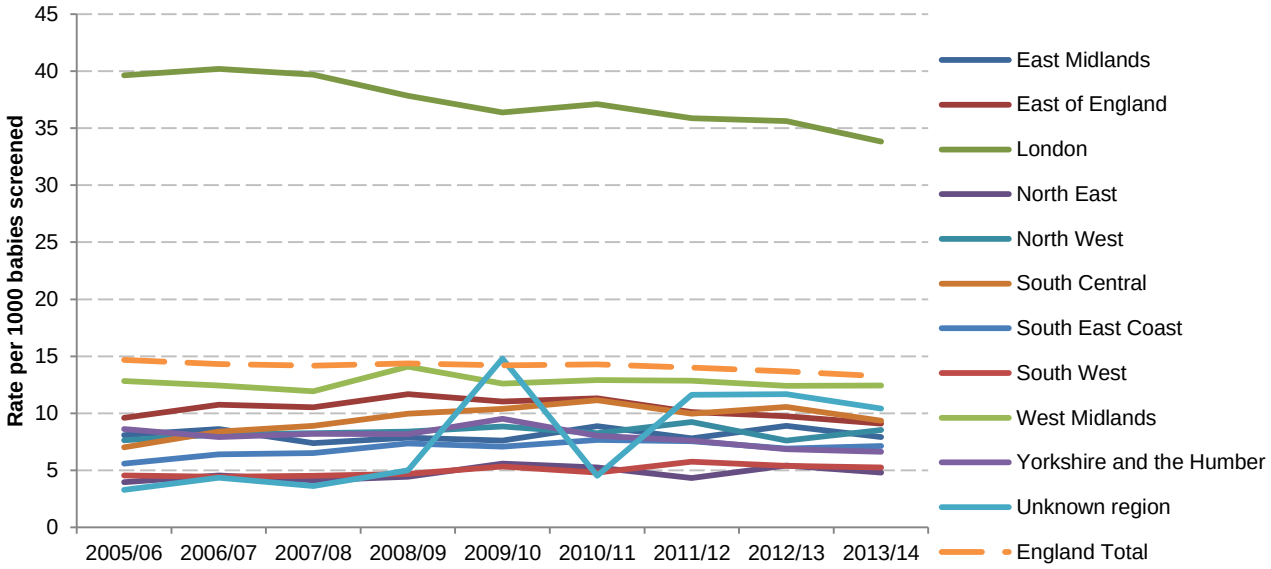


Figure NB-5 shows the trends in rates of babies identified with a carrier result for England by region. Rates in London are consistently higher in each year than other regions in England. Figure NB-6 shows the rates for London, broken down by London sectors (pre-2006 SHAs), and Figure NB-7 shows the rates by region excluding London.

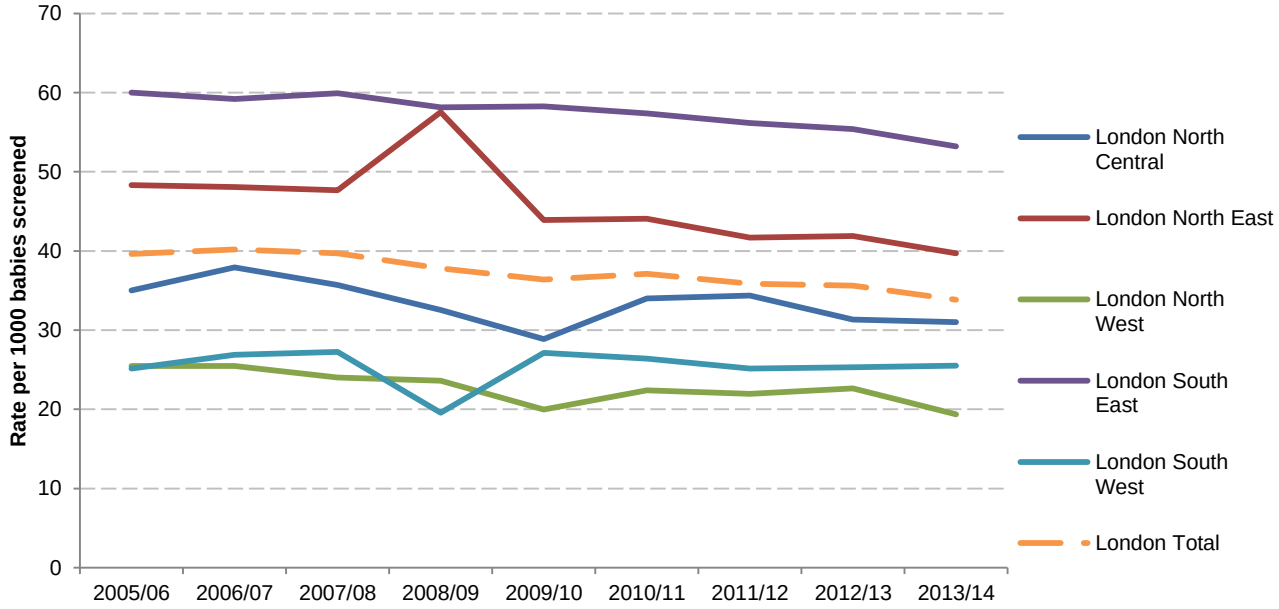
Carrier numbers are higher than those for significant conditions, which makes the rates more steady when comparing between years. Outside of London, rates are consistently highest in the West Midlands in this nine-year period and lowest in the South West and North East.

Figure NB-5. Trends in babies identified as carriers, 2005-14: England by region



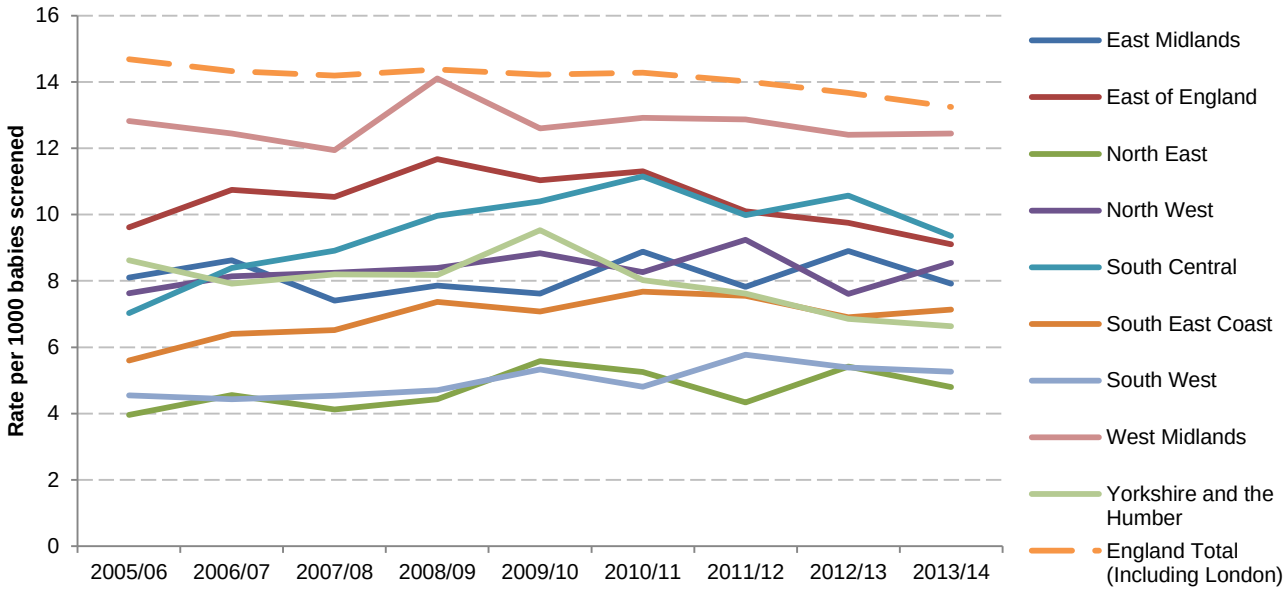
Carrier results comprise FAS, FAC, FAD, FAE and other carriers.
**Bristol data for first half of 2005/06 not included and Oxford and Portsmouth data not included for whole of 2005/06; Oxford data starts from 1st July 2006.*

Figure NB-6. Trends in babies identified as carriers, 2005-14: London sectors (pre-2006 SHAs)



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

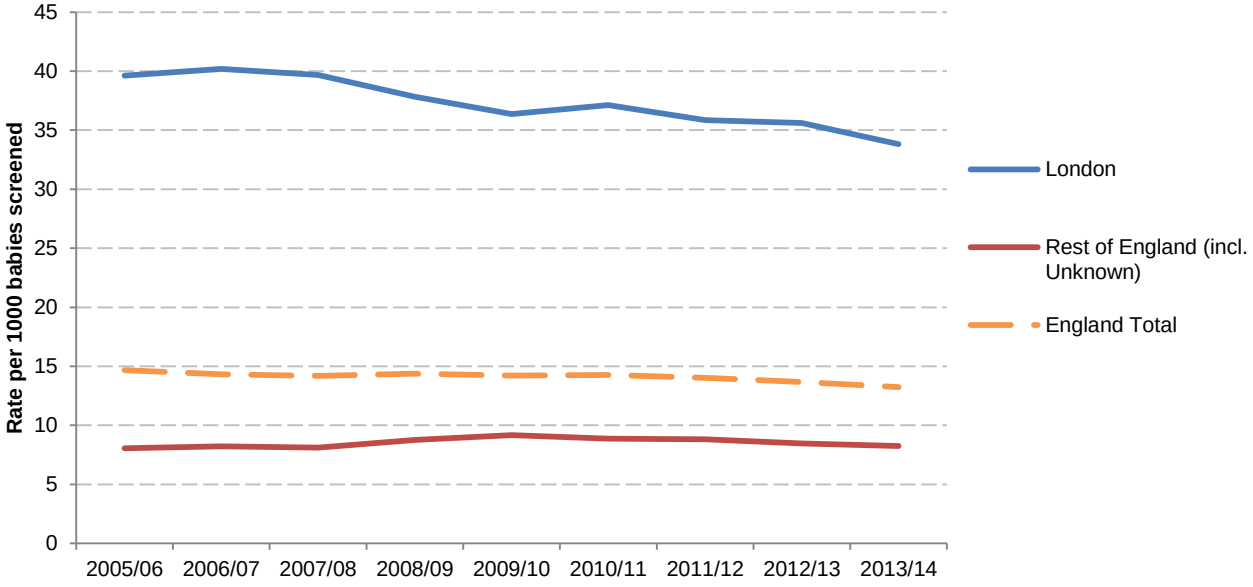
Figure NB-7. Trends in babies identified as carriers, 2005-14: England by region (excluding London)



Carrier results comprise FAS, FAC, FAD, FAE and other carriers.
**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-8 compares the rates for London, where they are highest in England, to those for the rest of England (including cases where the region is unknown). Carrier rates for the whole of England appear stable across this period ranging between 13 and 15 per 1000 babies screened. There appears to be a continuation of the decline in rates in London, ranging from 40 per 1000 babies screened in 2006/07 to 34 per 1000 babies screened in 2013/14. There is also some indication of a decline in the rates for the rest of England, although this appears smaller than in London.

Figure NB-8. Trends in babies identified as carriers, 2005-14: London and the rest of England



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

5.4. Results by ethnicity

Newborn screening figures by ethnicity can differ slightly from the figures shown by region (see 5.1 Response rates and data quality). Table NB-5 shows the number and rates of babies identified with a significant condition in the three years since 2011/12, and Table NB-6 shows similar data for carrier results in the same period.

Babies reported as black African accounted for approximately 60% of significant conditions detected and approximately 38% of carrier results detected in 2013/14. The next largest group is babies reported as black Caribbean, which accounted for 10% of significant conditions and 9% of carriers. Whilst sickle cell disease is more common in people from these ethnic categories, it is not confined to these groups and in 2013/14 1.6% of affected babies and 8.9% of carriers were identified as white British, white Irish, or any other white background.

Table NB-5. Numbers of babies identified with significant conditions, 2011-14: England by ethnicity

Ethnic category	2011/12			2012/13			2013/14		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
A - White British	3	443,448	0.01	5	434,514	0.01	5	413,538	0.01
B - White Irish	*	1,671	0.00	*	1,723	0.00	*	1,910	1.05
C - Any other white background	*	47,675	0.00	*	49,386	0.00	*	57,664	0.00
D - White and black Caribbean	9	7,581	1.19	8	7,803	1.03	10	8,042	1.24
E - White and black African	6	4,407	1.36	3	4,348	0.69	5	5,080	0.98
F - White and Asian	*	8,056	0.00	*	10,495	0.00	*	8,545	0.12
G - Any other mixed background	4	12,340	0.32	3	10,735	0.28	8	13,404	0.60
H - Indian	4	21,736	0.18	4	21,560	0.19	*	21,149	0.05
J - Pakistani	*	27,262	0.04	*	27,855	0.00	3	27,595	0.11
K - Bangladeshi	7	9,096	0.77	9	9,856	0.91	8	9,166	0.87
L - Any other Asian background	3	9,869	0.30	*	8,078	0.25	4	10,868	0.37
M - Black Caribbean	38	6,668	5.70	30	7,402	4.05	32	6,163	5.19
N - Black African	195	24,294	8.03	196	22,244	8.81	191	22,770	8.39
P - Any other black background	20	3,330	6.01	17	4,838	3.51	25	3,233	7.73
R - Chinese	*	3,687	0.00	*	3,951	0.00	*	3,213	0.00
S - Any other ethnic category	5	16,828	0.30	10	18,615	0.54	5	14,234	0.35
Z - Not stated	25	46,071	0.54	24	43,490	0.55	19	42,863	0.44
England total	**320**	**694,019**	**0.46**	**311**	**686,893**	**0.45**	**319**	**669,437**	**0.48**

**Numbers less than three have been suppressed.*

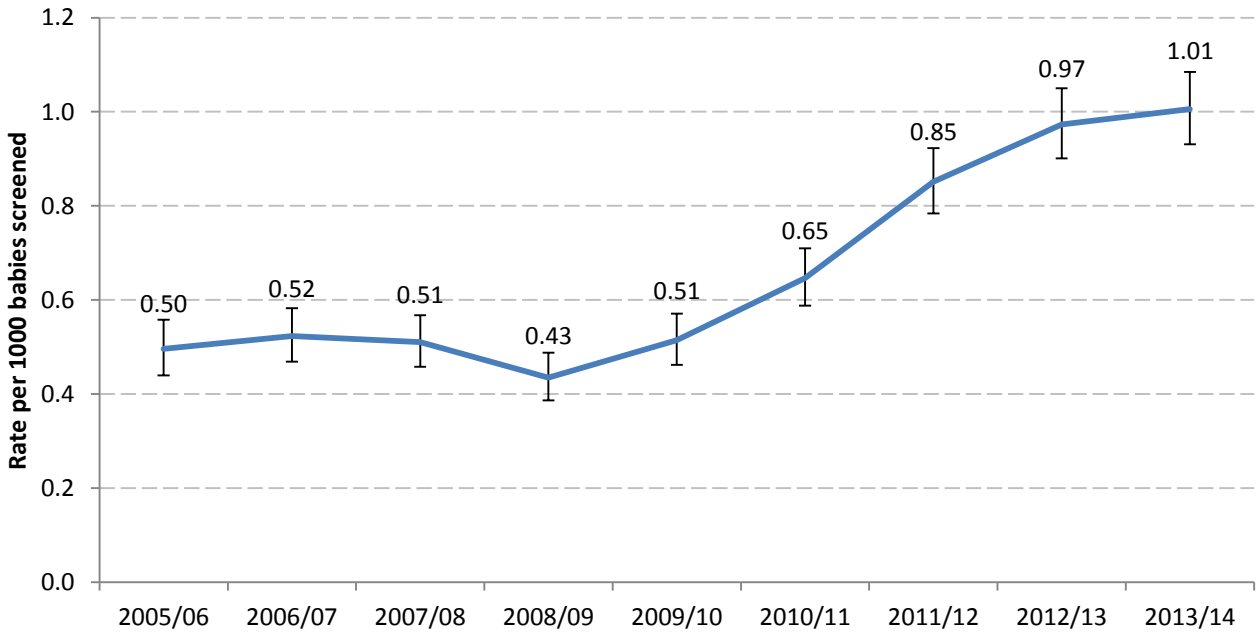
Table NB-6. Numbers of babies identified with carrier results, 2011-14: England by ethnicity

Ethnic category	2011/12			2012/13			2013/14		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
A - White British	957	443,448	2.16	762	434,514	1.75	609	413,538	1.47
B - White Irish	5	1,671	2.99	3	1,723	1.74	25	1,910	13.09
C - Any other white background	171	47,675	3.59	139	49,386	2.81	158	57,664	2.74
D - White and black Caribbean	464	7,581	61.21	457	7,803	58.57	506	8,042	62.92
E - White and black African	375	4,407	85.09	359	4,348	82.57	384	5,080	75.59
F - White and Asian	168	8,056	20.85	146	10,495	13.91	139	8,545	16.27
G - Any other mixed background	327	12,340	26.50	352	10,735	32.79	395	13,404	29.47
H - Indian	328	21,736	15.09	314	21,560	14.56	262	21,149	12.39
J - Pakistani	351	27,262	12.88	293	27,855	10.52	305	27,595	11.05
K - Bangladeshi	416	9,096	45.73	420	9,856	42.61	468	9,166	51.06
L - Any other Asian background	182	9,869	18.44	158	8,078	19.56	147	10,868	13.53
M - Black Caribbean	850	6,668	127.47	843	7,402	113.89	750	6,163	121.69
N - Black African	3,467	24,294	142.71	3,483	22,244	156.58	3,369	22,770	147.96
P - Any other black background	396	3,330	118.92	412	4,838	85.16	391	3,233	120.94
R - Chinese	27	3,687	7.32	29	3,951	7.34	11	3,213	3.42
S - Any other ethnic category	401	16,828	23.83	495	18,615	26.59	245	14,234	17.21
Z - Not stated	838	46,071	18.19	746	43,490	17.15	693	42,863	16.17
England total	**9,723**	**694,019**	**14.01**	**9,411**	**686,893**	**13.70**	**8,857**	**669,437**	**13.23**

5.5. Declined screening tests

Figure NB-9 shows the rate per 1000 babies screened that declined testing in England between 2005 and 2014. There were approximately 672 declined screening tests in 2013/14, which equates to approximately 1 per 1000 babies screened. There has been an increase each year in the number of reported declined tests in the past six years, from 0.43 per 1000 babies screened in 2008/09 to 1.01 per 1000 babies screened in 2013/14.

Figure NB-9. Declined screening for sickle cell disease, 2005-14: England



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.

5.6. Post-transfusion testing

Routine techniques are not suitable for testing samples from transfused babies as transfused red cells can survive up to 120 days in circulation. It is therefore important that pre-transfusion samples are taken in accordance with standards.

Table NB-7 shows he number and rates of post-transfusion samples between 2011 and 2014. In 2013/14 there were 1,940 post-transfusion samples reported which require follow-up DNA testing, which equates to approximately three per 1000 babies screened. No data on post-transfusion samples were identified by the Great Ormond Street Hospital (GOSH) laboratory for 2013/14, so this figure is likely to be an under-estimate.

Rates by region appear consistent with previous years with the exception of the South East Coast region where rates have increased from approximately four per 1000 babies screened to approximately 12 per 1000 babies screened.

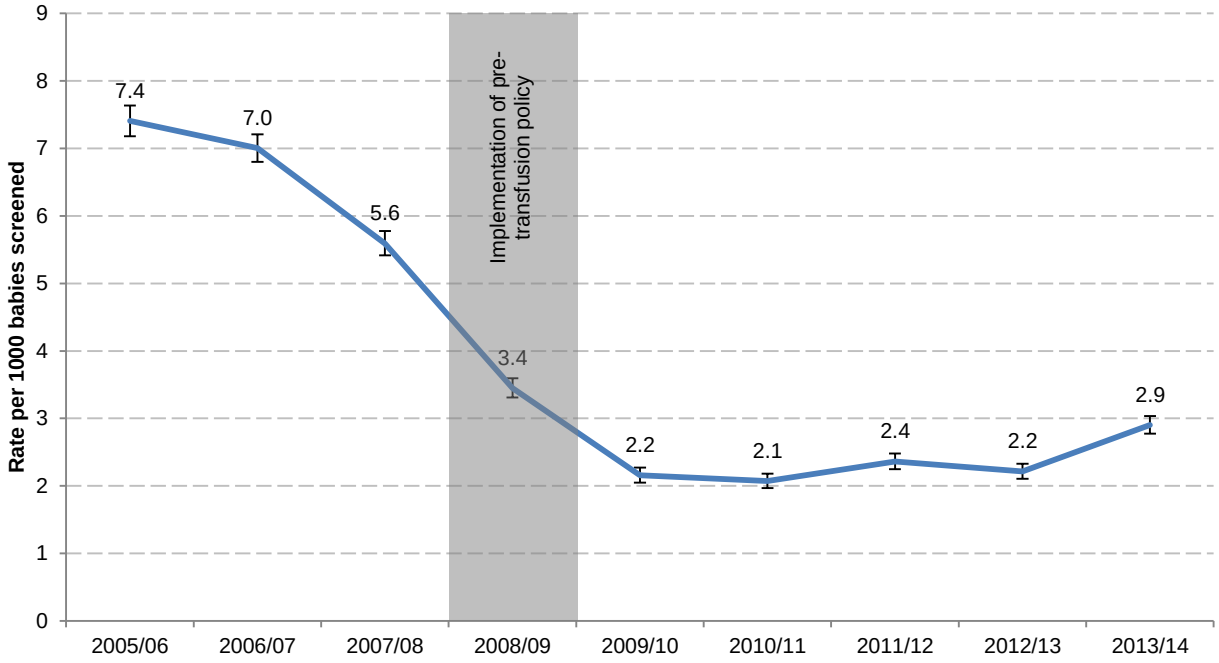
Table NB-7. Post-transfusion samples, number and rate per 1000 babies screened, 2011-14: England by region

Region	2011/12			2012/13			2013/14		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
East Midlands	94	50,901	1.85	89	49,898	1.78	94	48,382	1.94
East of England	83	72,091	1.15	96	72,421	1.33	22	69,082	0.32
London	344	133,245	2.58	368	131,424	2.80	522	130,373	4.00
North East	35	28,122	1.24	44	28,966	1.52	36	27,896	1.29
North West	224	88,405	2.53	137	87,369	1.57	137	84,384	1.62
South Central	34	54,286	0.63	42	53,352	0.79	33	51,413	0.64
South East Coast	195	53,521	3.64	199	51,682	3.85	619	51,710	11.97
South West	42	60,811	0.69	34	59,938	0.57	30	57,940	0.52
West Midlands	213	72,970	2.92	182	72,559	2.51	115	70,773	1.62
Yorkshire and the Humber	145	69,457	2.09	133	69,613	1.91	96	67,820	1.42
Unknow n region	229	9,469	24.18	195	8,216	23.73	236	8,344	28.28
England total	**1,638**	**693,278**	**2.36**	**1,519**	**685,438**	**2.22**	**1,940**	**668,117**	**2.90**

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

Figure NB-10 shows the national rates for each year since 2005. Since the programme implemented the pre-transfusion policy in 2008/09 rates for post-transfusion samples have remained steady at approximately 2.2 per 1000 babies screened, although an increase can be seen in 2013/14. This increase may reflect the higher figures reported for the South East Coast region (see Table NB-7).

Figure NB-10. Post-transfusion samples, rate per 1000 babies screened, 2005-14: England



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.

Post-transfusion samples require DNA testing to mitigate the risk of a missed baby. Data on DNA testing for transfused babies from the laboratories at King's College Hospital and Sheffield Children's Hospital is shown in Table NB-8. In 2013/14 there were 1,160 samples tested by these laboratories. Since Q3 of 2009/10 there have been 6,190 samples tested, of which five were found to be screen positive for sickle cell disease (approximately one in 1,238 babies screened using DNA testing).

Table NB-8. Numbers detected through DNA testing for transfused babies, 2009-14

	2009/10†	2010/11	2011/12	2012/13	2013/14	Total
Total Specimens received per Quarter	493	1674	1520	1343	1160	6190
Number of Negative results (HbS not detected)	483	1650	1497	1319	1140	6089
Number of Positive Heterozygotes	10	24	21	21	20	96
Number of Positive Homozygotes	*	*	*	3	*	5
Number of Results pending	*	*	*	*	*	*
Number rejected due to lack of identifiers	*	*	*	*	*	*

†2009/10 data is for Q3 and Q4 only

**Numbers less than three have been suppressed*

5.7. Timeliness of reporting results

Newborn Blood Spot Screening (NBBS) Programme standard 4 (timely sample collection) is for the sample to be taken on day five and in exceptional circumstances between day five and day eight (day of birth is day zero). This standard has a 95% acceptable threshold and 99% achievable threshold. Standard 5 (timely receipt of a sample in the newborn screening laboratory) sets an acceptable threshold of 99% of samples to arrive in the laboratory within four working days of sample collection and an achievable threshold of 99% of samples to arrive in the laboratory within three working days.

Sickle Cell and Thalassaemia (SCT) Screening Programme standard NP3 (timely communication of positive screening results) requires 90% of sickle cell disease results to be communicated to parents by four weeks of age.

Table NB-9 shows timeliness figures for newborn babies identified with a significant condition in newborn screening, including F-only cases which are probable beta thalassaemia affected cases. Turn-around-time is calculated from the number of days between the age at sample and the age at which the positive result was reported. Turn-around times for Bristol are not shown because data on age at clinical referral were missing, and for Newcastle because timeliness data were not included in the data returned to the Programme Centre.

Table NB-9. Timeliness of reporting affected newborn results, 2013/14: England by laboratory

Laboratory	No. of screen positives	Sample ≤8 days		Sample received by lab within 4 days		Clinical referral by 28 days		Turn-around time		
	n	n	%	n	%	n	%	Min	Max	Average
Bristol	*	*	100	*	100	†	N/A	†	†	†
Cambridge	*	*	100	*	100	*	100	13	13	13
GOS & CMH	109	103	94	99	91	106	97	6	46	10
Leeds	13	12	92	11	85	13	100	5	23	11
Liverpool	7	7	100	6	86	7	100	5	10	7
Manchester	17	16	94	17	100	17	100	7	16	10
Newcastle	†	†	N/A	†	N/A	†	N/A	†	†	†
Oxford	20	17	85	17	85	16	80	9	26	14
Portsmouth	5	*	80	*	60	*	80	14	32	19
Sheffield	22	21	95	20	91	18	82	12	31	18
South East Thames	89	83	93	73	82	83	93	3	86	12
South West Thames	32	32	100	23	72	32	100	4	24	14
West Midlands	24	24	100	20	83	23	96	8	25	14
England Total	343	324	94	294	86	320	93	3	86	12

†*Missing data*

**Numbers less than five have been suppressed*

6. Key Performance Indicators (KPIs)

Background to Key Performance Indicators (KPIs)

The Sickle Cell and Thalassaemia Screening Programme has three antenatal KPIs and shares one newborn KPI with the Newborn Blood Spot Screening Programme. Newborn KPIs NB1 and NB2 are also relevant to SCT screening.

For more information on KPI data collection and reporting, please see www.screening.nhs.uk/kpi.

KPI Code	KPI Description	Acceptable level	Achievable level
ST1	The proportion of pregnant women eligible for antenatal sickle cell and thalassaemia screening for whom a conclusive screening result is available at the day of report (the day on which data to support an audit or performance return are collated)	≥95.0%	≥99.0%
ST2	The proportion of women having antenatal sickle cell and thalassaemia screening for whom a conclusive screening result is available by 10 weeks' gestation	≥50.0%	≥75.0%
ST3	The proportion of antenatal sickle cell and thalassaemia samples submitted to the laboratory which are supported by a completed Family Origin Questionnaire (FOQ)	≥90.0%	≥95.0%
NB1	The proportion of babies registered within the CCG both at birth and on the last day of the reporting period who are eligible for newborn blood spot screening and have a conclusive result recorded on the Child Health Information System within an effective timeframe. For this KPI, PKU is used as a proxy for all tests and the test must be completed by 17 days of age.	≥95.0%	≥99.9%
NB2	The percentage of babies from whom it is necessary to take a repeat blood sample due to an avoidable failure in the sampling process.	≤2.0%	≤0.5%
NB3	The proportion of newborn blood spot screening results which are screen negative for all five conditions, available for communication to parents within six weeks of birth	95.0%	98.0%

Table KPI-1 shows a breakdown of KPI data for each quarter in the 2013/14 financial year with national performance levels for each.

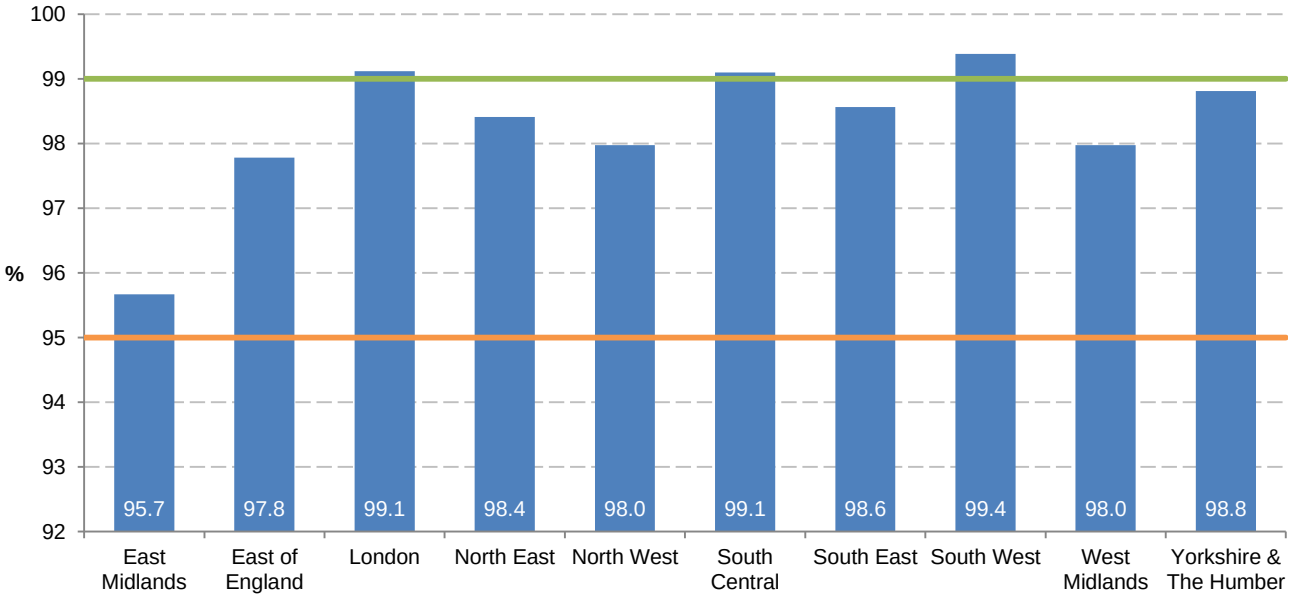
Table KPI-1. KPI figures by quarter, 2013/14

KPI	Q1			Q2			Q3			Q4			Total for year		
	Num	Denom	%	Num	Denom	%	Num	Denom	%	Num	Denom	%	Num	Denom	%
ST1	137,500	140,016	98.2	142,224	144,739	98.3	138,884	140,701	98.7	150,872	152,937	98.6	569,480	578,393	98.5
ST2	80,739	165,909	48.7	84,462	165,765	51.0	86,194	164,441	52.4	87,374	176,682	49.5	338,769	672,797	50.4
ST3	167,595	175,974	95.2	168,029	176,877	95.0	169,363	177,733	95.3	176,615	183,870	96.1	681,602	714,454	95.4
NB1	122,073	129,682	94.1	132,412	142,418	93.0	142,916	154,084	92.8	137,590	143,137	96.1	534,991	569,321	94.0
NB2	3,564	108,316	3.3	4,194	128,191	3.3	4,010	144,145	2.8	3,752	132,210	2.8	15,520	512,862	3.0
NB3	127,342	128,007	99.5	138,892	139,615	99.5	150,322	151,384	99.3	139,779	140,797	99.3	556,335	559,803	99.4

The KPI data presented here is aggregated data for Q1 – Q4 2013/14. Completeness of data and response rates vary in each quarter and for each KPI. Where complete cohort data cannot be provided, these figures are excluded.

KPI ST1 is defined as the number of women tested as a proportion of eligible women. The acceptable threshold for this KPI is greater than or equal to 95% and the achievable level is greater than or equal to 99%.

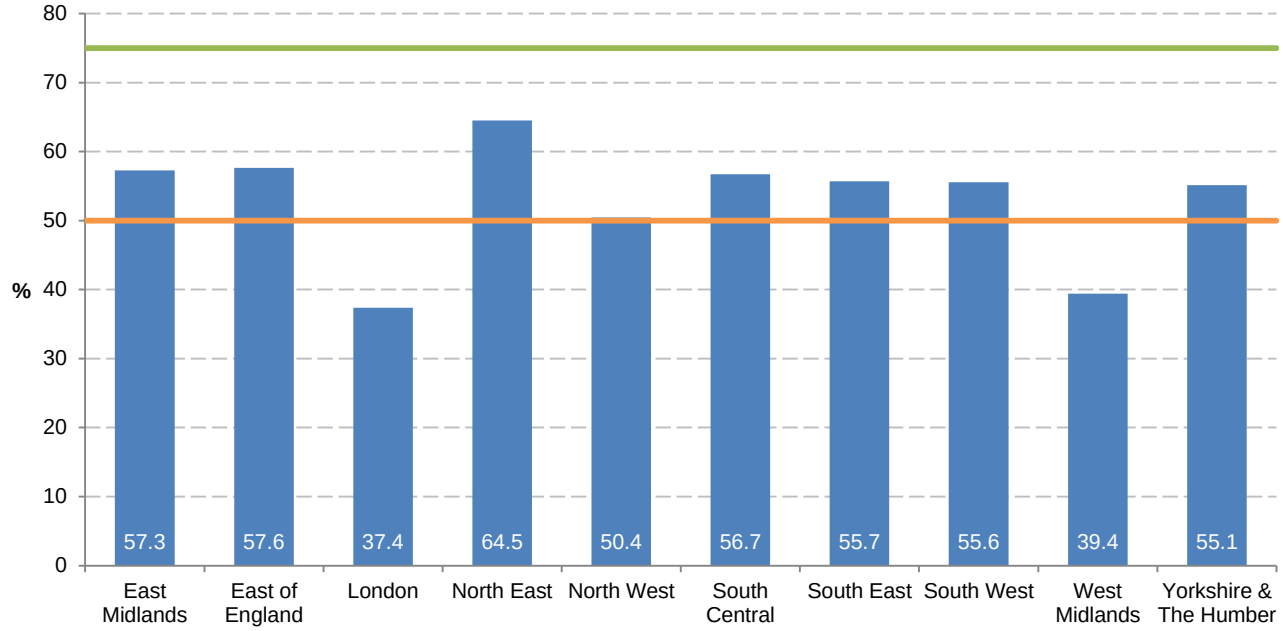
Figure KPI-1. ST1 - Coverage by region, 2013/14: England



The lower reference line represents the acceptable level for this KPI, and the upper reference line represents the achievable level for this KPI.

KPI ST2 is defined as the number of women tested by 10 weeks gestation as a proportion of women for whom a sample was received at the laboratory. The acceptable threshold for this KPI is greater than or equal to 50% and the achievable level is greater than or equal to 75%.

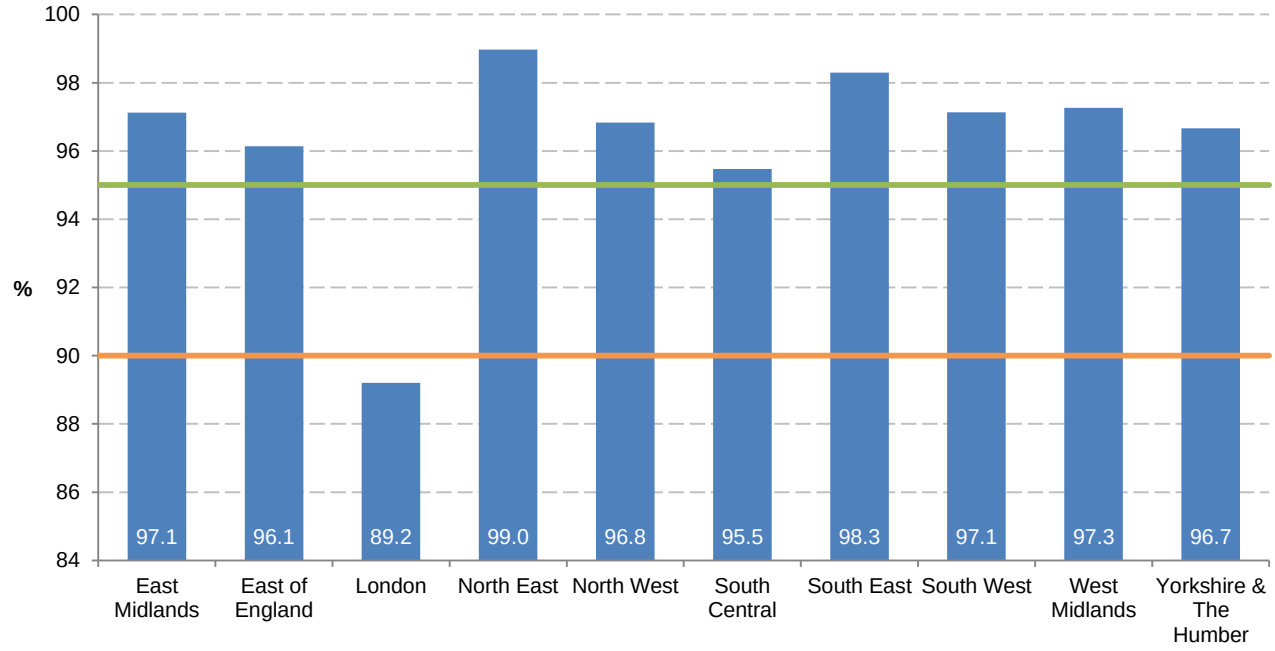
Figure KPI-2. ST2 - Timeliness of test by region, 2013/14: England



The lower reference line represents the acceptable level for this KPI, and the upper reference line represents the achievable level for this KPI.

KPI ST3 is defined as the number of laboratory requests with a completed FOQ form as a proportion of laboratory requests. The acceptable threshold for this KPI is greater than or equal to 90% and the achievable level is greater than or equal to 95%.

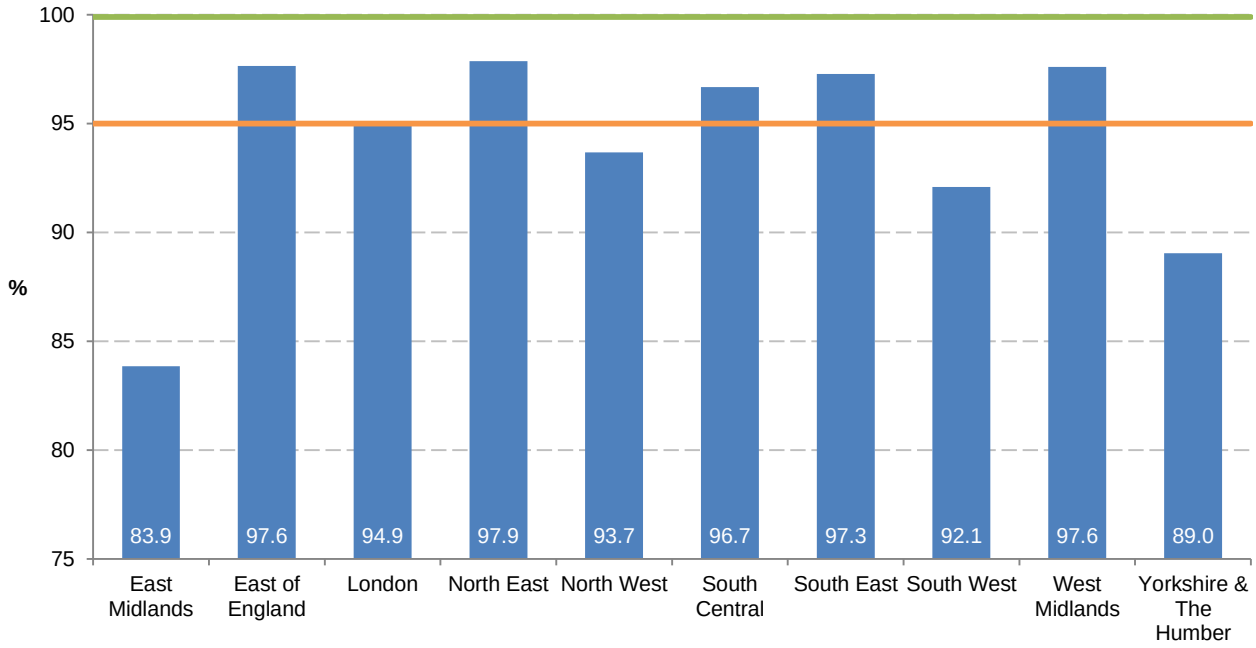
Figure KPI-3. ST3 - Completion of FOQ, 2013/14: England



The lower reference line represents the acceptable level for this KPI, and the upper reference line represents the achievable level for this KPI.

KPI NB1 is defined as the number of babies tested as a proportion of eligible babies. The acceptable threshold for this KPI is greater than or equal to 95% and the achievable level is greater than or equal to 99.9%.

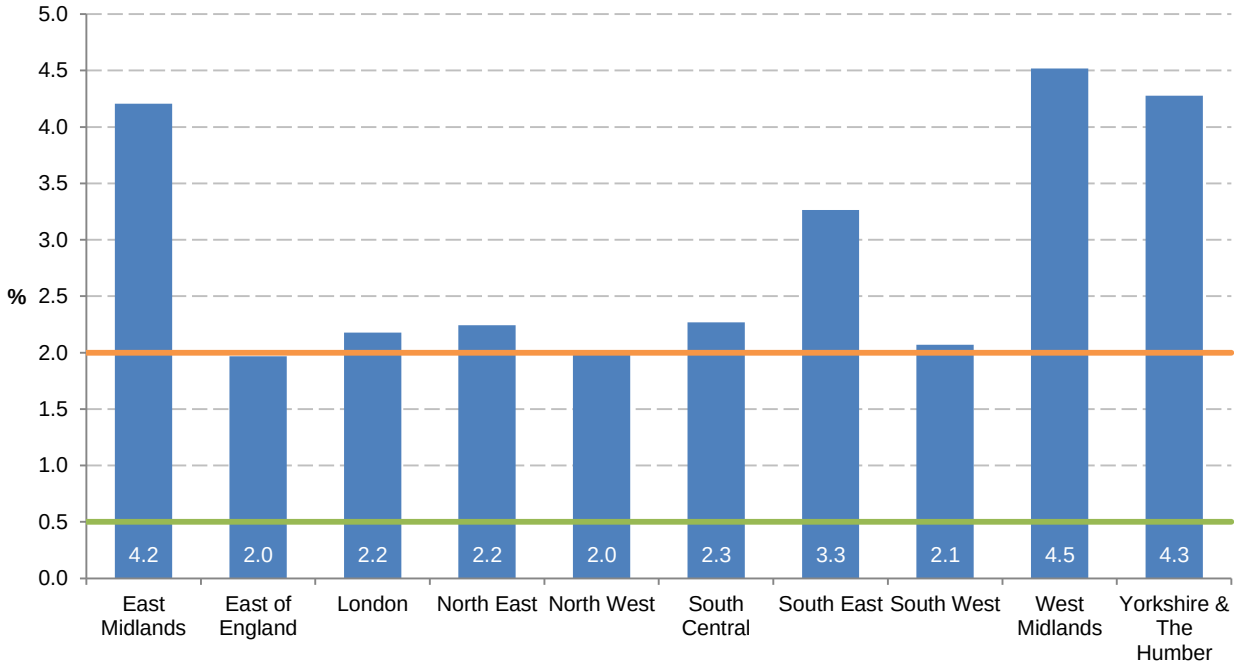
Figure KPI-4. NB1 - Coverage by region, 2013/14: England



The lower reference line represents the acceptable level for this KPI, and the upper reference line represents the achievable level for this KPI.

KPI NB2 is defined as the number of avoidable repeats as a proportion of the number of initial blood samples. The acceptable threshold for this KPI is less than or equal to 2.0% and the achievable level is less than or equal to 0.5%.

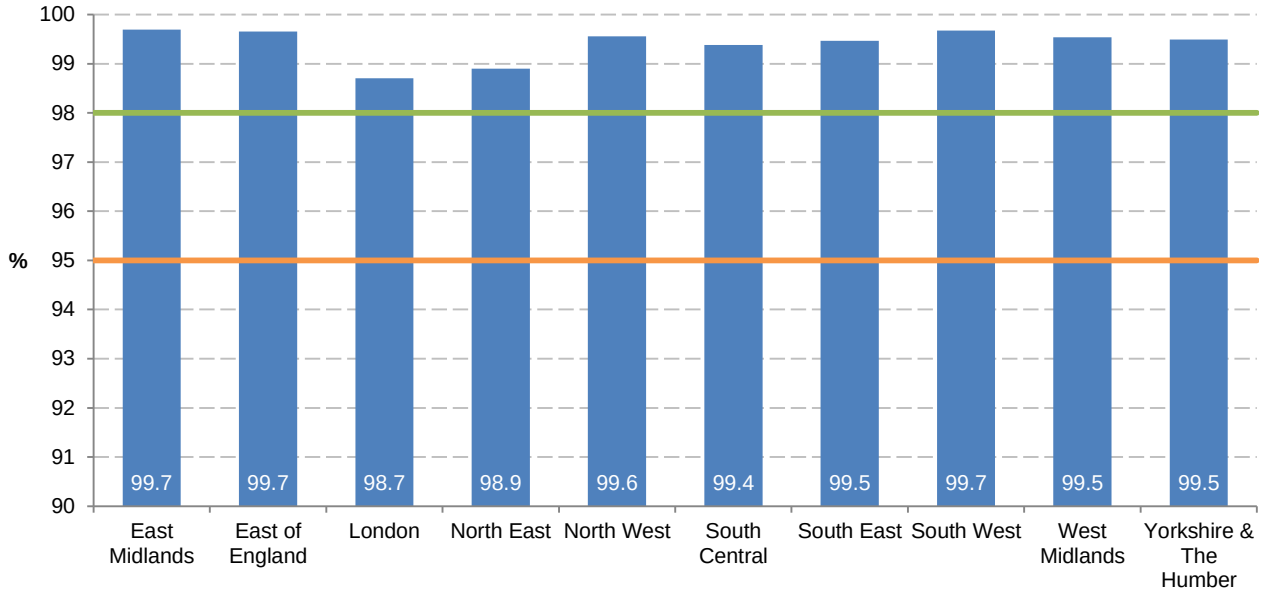
Figure KPI- 5. NB2 - Avoidable repeat tests, 2013/14: England



The upper reference line represents the acceptable level for this KPI, and the lower reference line represents the achievable level for this KPI.

KPI NB3 is defined as the number of results available for communication by six weeks as a proportion of the number of babies that are screen negative for all conditions screened for. The acceptable threshold for this KPI is 95% and the achievable level is 98%.

Figure KPI-6. NB3 - Timeliness of result availability, 2013/14: England



The lower reference line represents the acceptable level for this KPI, and the upper reference line represents the achievable level for this KPI.

7. Appendices

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

Universal newborn screening for sickle cell disease and beta thalassaemia has been available in England since 2005.

In September 2010, the NHS Sickle Cell and Thalassaemia Screening Programme started a project to assess the outcomes of the linked antenatal and newborn screening programme. We are collecting identifiable data on babies, or children under age 5, with sickle cell disorders or beta thalassaemia. This project will assess:

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

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

Between 1 April 2013 and 31 March 2014 there were 300 screen positive babies born. Of these:

- 268 babies had suspected sickle cell disease of which 88% were seen in clinic by 3 months
- 32 had suspected beta thalassaemia of which 85% were seen by 3 months

Babies were excluded from this cohort where there were clinically insignificant cases, births abroad, and deaths which were not ascribed to sickle cell disease.

Data collection has been a lengthy and protracted process. This has been in part due to manual processes and varying numbers of babies across the country (with the greatest number in London), and in part due to differing methods used for notification to specialist and community centres. The process needs to be streamlined and simplified to avoid duplication.

It has been proposed that a single data collection form is used both by the lab for notification and by the community centre as part of the referral process to the clinician. The updated forms are available at sct.screening.nhs.uk/evaluation. The clinician would be expected to complete the relevant clinical data and return it to the project administrator using nhs.net email, to ensure confidentiality. These changes have been supported by the programme's information governance and clinical group and a jointly signed letter by the programme and the newborn blood spot laboratory lead will be sent to the newborn labs. This new approach will aim to improve data quality and completeness. In the future there will also be a data analysis and quality assurance group which will oversee ongoing developments of the programme.

Appendix Two: Antenatal data return form part two - 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														