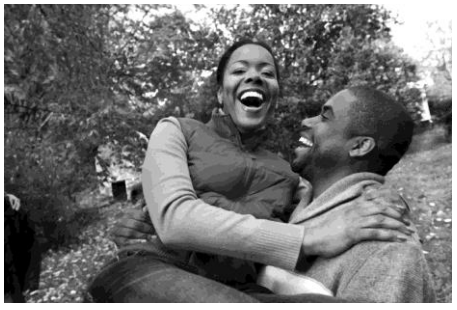
UK National Screening Committee

NHS Screening Programmes

Sickle Cell and Thalassaemia

Data Report 2012/13
Trends and performance analysis



Sickle Cell and Thalassaemia Screening Programme

sct.screening.nhs.uk

Acknowledgements

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NHS Sickle Cell and Thalassaemia Screening Programme
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Executive summary

In England approximately 720,000 pregnant women were screened for sickle cell and thalassaemia, of whom approximately 15,000 (approximately 2%) were identified as screen positive. There were 398 prenatal diagnostic (PND) tests performed of which 21% had an affected result, 54% had a carrier result, and 24% had a 'no abnormality detected' result.

There were approximately 685,000 newborn babies screened, of whom 312 (0.46 per 1000 babies screened) were identified with a significant condition and 9,368 (13.67 per 1000 babies screened) were identified with a carrier result.

There has been an increase in the number of sites meeting the 50% acceptable standard for screening offer by 10 weeks of gestation. Nationally the rate is 46%, compared to 44% in 2011/12.

The proportion of samples with a Family Origin Questionnaire (FOQ) attached has increased in both high and low prevalence areas, and high prevalence areas are now close to the 90% acceptable standard.

In clinical practice, use of the FOQ has achieved cost savings by ensuring that high risk women are not missed for screening, reducing unnecessary re-screening of women with inconclusive results and allowing more accurate father screening.

There has been an increase in the proportion of fathers offered testing for whom a sample is received by the laboratory, and is now at approximately 63%.

The number of PND tests performed accounts for approximately half of the number of 'high risk' pregnancies identified in antenatal screening. Trend data covering the five-year period 2008–2013 shows that approximately 66% of parents of babies affected by sickle cell disease, and 80% of parents affected by beta thalassaemia, choose not to continue the pregnancy.

The standard for 50% of PND tests performed before 12 weeks and 6 days of pregnancy is being met, but 31% of PND tests performed are taking place at 15 weeks gestation or later. Data suggests that more parents opted to terminate in cases where testing occurred earlier in the pregnancy.

In newborn screening, the rates of post-transfusion samples being taken has declined due to the policy of taking a pre-transfusion sample in babies on admission to specialist units. In the four years since implementation of this policy, rates have been steady, ranging between 2.1 per 1000 babies screened and 2.4 per 1000 babies screened. It is therefore important that the policy for DNA testing for transfused babies continues and that staff are reminded of the requirement for pre-transfusion samples so that levels of DNA testing are kept low.

The executive summary from the Newborn Blood Spot Screening Programme is included in Appendix One and covers the whole of the United Kingdom.

Abbreviations

AN	Antenatal
FOQ	Family Origin Questionnaire
Hb	Haemoglobin – see glossary for haemoglobin variants
HES	Hospital Episodes Statistics
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
UKNSC	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 Afro-Caribbeans 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 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

The UK National Screening Committee and NHS Screening Programmes are part of Public Health England (PHE), an executive agency of the Department of Health. PHE was established on 1 April 2013 to bring together public health specialists from more than 70 organisations into a single public health service.

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 followed up with the relevant laboratory.

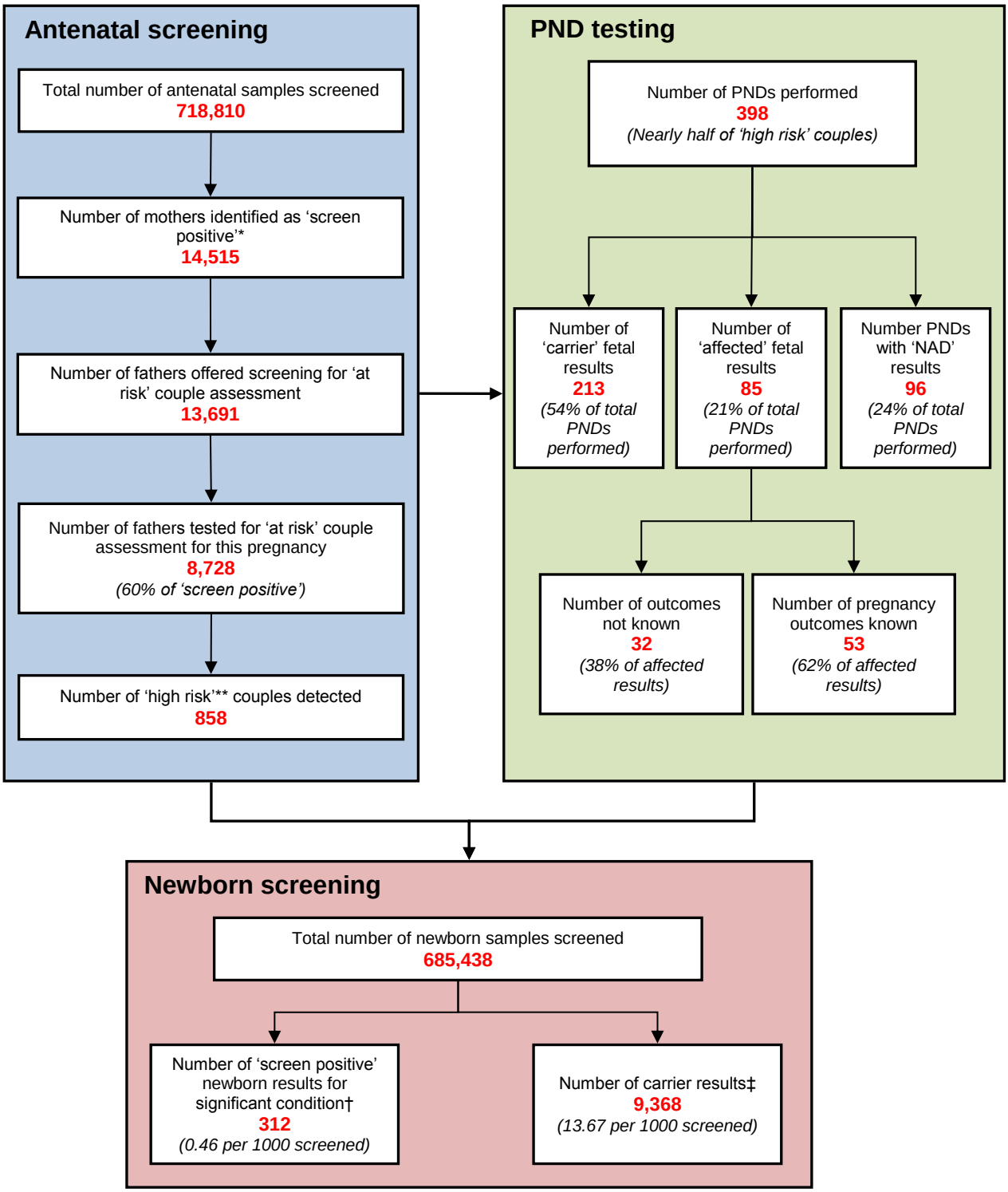
For the antenatal laboratories in particular, we recognise the difficulty of data collection in the absence of standardised data collection tools and IT systems. We try to ask for limited data and work hard to justify all data requests, ensuring there are no gaps and no duplication across the screening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

Response rate:

In England 143 data returns out of 145 were received. Data were not received for Newham University Hospital or for Whipps Cross Hospital. These laboratories (both within Barts Health NHS Trust) have merged with the Royal London and were unable to provide figures for these sites for this year. We have been informed that it should be possible to provide data for the whole Trust in the future.

Data quality:

A total of 718,810 booking bloods were reported. As in previous years there were some gaps in the data provided where some laboratories were unable to provide data for certain data 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.

There were fewer exclusions made compared to last year for the data fields for booking bloods tested by 10 weeks gestation and for booking bloods with a FOQ attached. However more exclusions were made for data on those not tested due to previous tests. The number of exclusions made for those not tested due to declines, and for father uptake, was the same as in 2011/12. A higher proportion of screen positive women was included in the breakdown data (93% in 2012/13 compared to 90% in 2011/12), but a lower proportion of high risk couples (85% in 2012/13 compared to 88% in 2011/12).

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

The number of father specimens received may not include cases where the father was tested previously and not re-tested in this pregnancy, 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 that 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.

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.

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.

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 would not be considered to be screen positive for sickle cell or thalassaemia.

3.2. Numbers screened and detected

National:

Table AN-1 shows the antenatal screening figures by region for England in 2012/13. There are no exclusions made based on missing or incomplete data in this table. In 2012/13 a total of 718,810 antenatal samples were identified in the annual laboratory data returns. Of these, 14,515 women (one in 50 women screened) were identified as carriers for sickle cell disease or for thalassaemia. The number of screen positive women includes cases where the woman had a known previous screen positive result and cases where a donor egg was used (requiring testing of the baby's father).

'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 2012/13 there were 858 pregnancies (one in 17 screen positive women) identified as at high risk for these conditions. 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 the risk, and so we estimate 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 for 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, giving an estimate of approximately 1,400 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, 2012/13: 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	78,123	28,223	36.13	75,433	96.56	1,010	1.29	66,534	85.17	*	0.00	59	5.84
East Midlands	8 / 8	53,016	32,242	60.82	52,238	98.53	677	1.28	49,137	92.68	*	0.00	28	4.14
London	24 / 26	138,588	35,965	25.95	105,123	75.85	6,817	4.92	127,362	91.90	29	0.02	447	6.56
North East	10 / 10	33,288	17,048	51.21	32,799	98.53	259	0.78	32,977	99.07	3	0.01	12	4.63
North West	19 / 19	91,004	29,817	32.76	86,892	95.48	1,221	1.34	84,081	92.39	19	0.02	63	5.16
South Central	11 / 11	51,803	23,882	46.10	48,351	93.34	788	1.52	49,614	95.77	39	0.08	48	6.09
South East Coast	10 / 10	57,885	7,486	12.93	56,366	97.38	724	1.25	52,448	90.61	23	0.04	42	5.80
South West	17 / 17	65,570	24,888	37.96	64,458	98.30	440	0.67	59,805	91.21	6	0.01	21	4.77
West Midlands	15 / 15	77,760	25,207	32.42	70,691	90.91	1,663	2.14	68,646	88.28	*	0.00	82	4.93
Yorkshire and The Humber	12 / 12	71,773	36,956	51.49	68,751	95.79	916	1.28	68,415	95.32	9	0.01	56	6.11
Total England	**143 / 145**	**718,810**	**261,714**	**36.41**	**661,102**	**91.97**	**14,515**	**2.02**	**659,019**	**91.68**	**132**	**0.02**	**858**	**5.91**

**Numbers less than 3 have been suppressed*

High prevalence areas:

Table AN-2 shows the antenatal screening figures for high prevalence areas for 2012/13 as reported by the antenatal laboratories. In 2012/13 a total of 381,531 antenatal samples were identified. Of these, 11,963 women (one in 32 women screened) were identified as carriers for sickle cell disease or for thalassaemia, including women with a known previous result. In high prevalence areas 756 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, 2012/13: 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	25,707	6,279	24.43	24,339	94.68	573	2.23	24,921	96.94	*	0.00	42	7.33
East Midlands	4 / 4	31,587	18,886	59.79	31,146	98.60	535	1.69	30,981	98.08	0	0.00	22	4.11
London	24 / 26	138,588	35,965	25.95	105,123	75.85	6,817	4.92	127,362	91.90	29	0.02	447	6.56
North East	1 / 1	6,944	3,997	57.56	6,583	94.80	115	1.66	6,708	96.60	0	0.00	7	6.09
North West	7 / 7	50,893	16,598	32.61	47,720	93.77	1,014	1.99	47,784	93.89	12	0.02	52	5.13
South Central	6 / 6	30,761	14,488	47.10	27,743	90.19	614	2.00	30,114	97.90	18	0.06	42	6.84
South East Coast	2 / 2	10,796	2,973	27.54	10,530	97.54	198	1.83	10,462	96.91	0	0.00	17	8.59
South West	2 / 2	11,585	4,025	34.74	11,500	99.27	164	1.42	11,219	96.84	0	0.00	11	6.71
West Midlands	7 / 7	46,588	11,597	24.89	40,213	86.32	1,410	3.03	40,740	87.45	*	0.00	77	5.46
Yorkshire and The Humber	3 / 3	28,082	10,701	38.11	25,255	89.93	523	1.86	25,739	91.66	6	0.02	39	7.46
Total England	**61 / 63**	**381,531**	**125,509**	**32.90**	**330,152**	**86.53**	**11,963**	**3.14**	**356,030**	**93.32**	**68**	**0.02**	**756**	**6.32**

**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 2012/13 as reported by the antenatal laboratories. In 2012/13 a total of 337,279 antenatal samples were identified. Of these, 2,552 women (one in 132 women screened) were identified as carriers, including those with a known previous screen positive test result. In low prevalence areas 107 pregnancies (one in 25 screen positive women) were identified as being at high risk for these conditions.

Table AN-3. Antenatal screening results by region, 2012/13: 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	52,416	21,944	41.87	51,094	97.48	437	0.83	41,613	79.39	0	0.00	17	3.89
East Midlands	4 / 4	21,429	13,356	62.33	21,092	98.43	142	0.66	18,156	84.73	*	0.00	6	4.23
London	0 / 0	-	-	-	-	-	-	-	-	-	-	-	-	-
North East	9 / 9	26,344	13,051	49.54	26,216	99.51	144	0.55	26,269	99.72	3	0.01	5	3.47
North West	12 / 12	40,111	13,219	32.96	39,172	97.66	207	0.52	36,297	90.49	7	0.02	11	5.31
South Central	5 / 5	21,042	9,394	44.64	20,608	97.94	174	0.83	19,500	92.67	21	0.10	6	3.45
South East Coast	8 / 8	47,089	4,513	9.58	45,836	97.34	526	1.12	41,986	89.16	23	0.05	25	4.75
South West	15 / 15	53,985	20,863	38.65	52,958	98.10	276	0.51	48,586	90.00	6	0.01	10	3.62
West Midlands	8 / 8	31,172	13,610	43.66	30,478	97.77	253	0.81	27,906	89.52	0	0.00	5	1.98
Yorkshire and The Humber	9 / 9	43,691	26,255	60.09	43,496	99.55	393	0.90	42,676	97.68	3	0.01	17	4.33
Total England	**82 / 82**	**337,279**	**136,205**	**40.38**	**330,950**	**98.12**	**2,552**	**0.76**	**302,989**	**89.83**	**64**	**0.02**	**102**	**4.00**

**Numbers less than 3 have been suppressed*

3.3. Bookings tested by 10 weeks

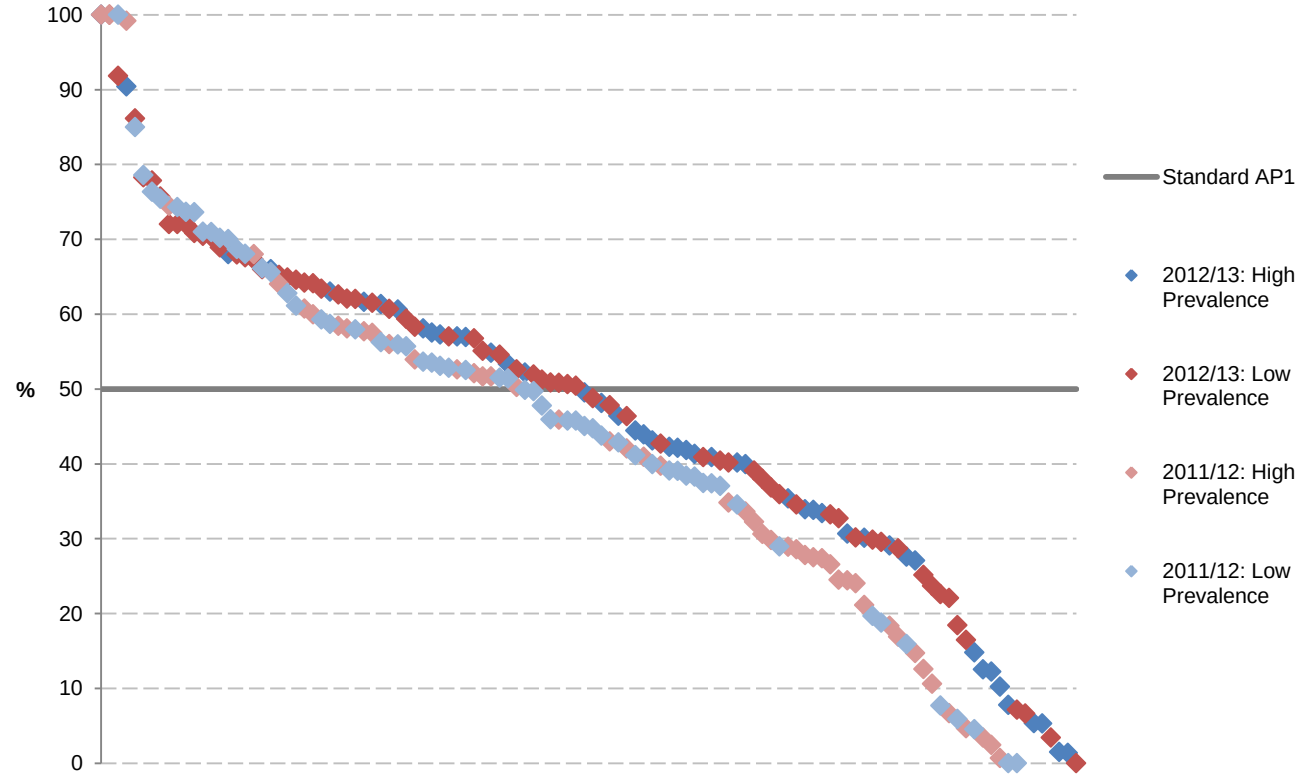
The target for antenatal testing is an offer of testing by 10 weeks gestation. To offer informed choice, a series of tests may be required: for the mother, the father and, if required, on the unborn baby. If tests show that the baby is at risk of inheriting a major haemoglobin disorder then the parents 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 maternity services to collect data on the offer of test.

National figures (without any exclusions where data were missing or incomplete) are shown in Table AN-1, Table AN-2, and Table AN-3. Across the whole of England, without exclusions, approximately 36% of samples were tested by 10 weeks of gestation, while in high and low prevalence areas this figure was 33% and 40% respectively.

Figure AN-1 shows a breakdown of the proportion of booking bloods tested by 10 weeks for high and low prevalence areas for 2011/12 and 2012/13. This chart excludes 27 data returns for 2012/13 and 36 data returns for 2011/12 where data on booking bloods tested by 10 weeks or the total number of booking bloods received were missing or unavailable. More laboratories appear to be meeting the acceptable level for standard AP1 compared to last year. With exclusions made for missing or unavailable data, the national proportion of booking bloods tested by 10 weeks was 46% in 2012/13, compared to 44% in 2011/12.

Figure AN-1. Percentage of antenatal booking bloods tested by 10 weeks by laboratory, 2011-13: England by prevalence

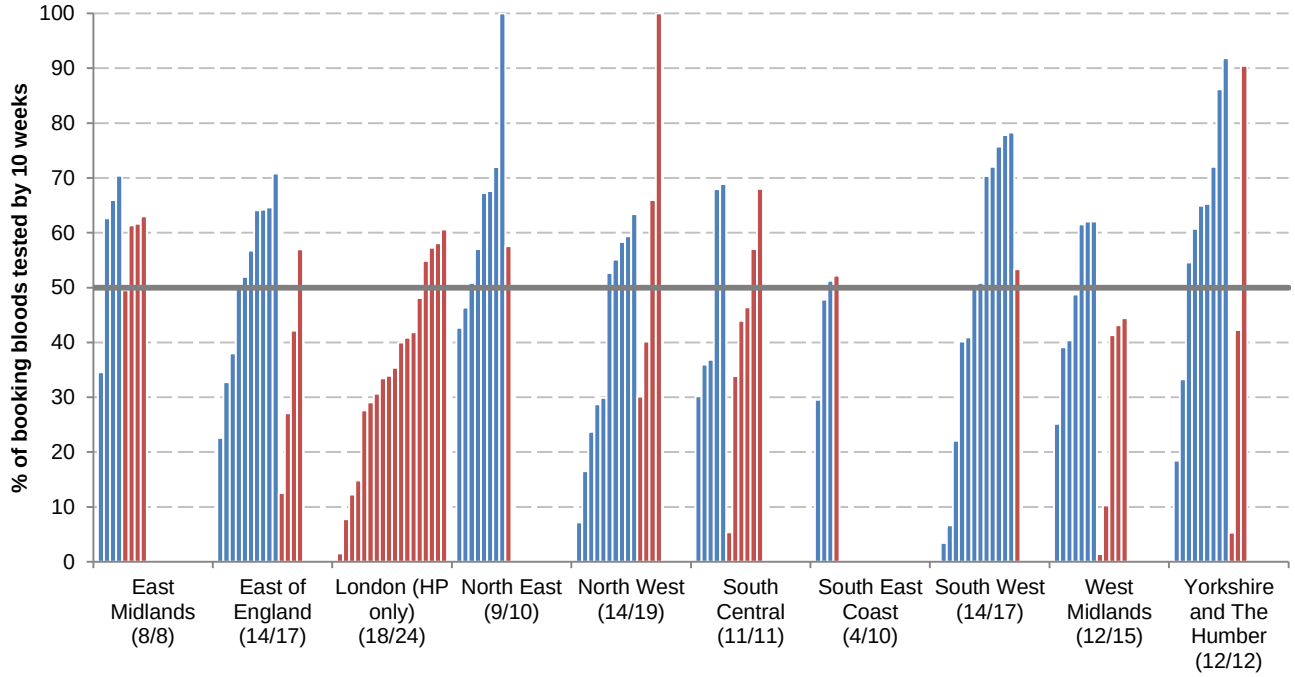


Each marker represents one laboratory.
The reference line represents the 50% acceptable level for Programme standard AP1.
The rate for the whole of England was 37% in 2010/11, was 44% in 2011/12, and was 46% in 2012/13.
2012/13 data excludes 27 laboratories, 2011/12 data excludes 36 laboratories and 2010/11 data excludes 62 laboratories based on missing or unavailable data.

Figure AN-2 shows a breakdown of the percentage of samples tested by 10 weeks by laboratory and by region across England. This chart also excludes 27 data returns as in Figure AN-1. Of the laboratories that are able to provide information on both the number of samples tested by 10 weeks and the total number of booking bloods, there is variation in performance between Trusts within regions, between regions, and between high and low prevalence areas.

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

Figure AN-2. Percentage of antenatal booking bloods tested by 10 weeks, 2012/13: 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 those Trusts for which data returns 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 27 laboratories where data on booking bloods tested by 10 weeks or the total number of booking was missing or unavailable.

Hospital Episodes Statistics (HES) contain details of all admissions to NHS hospitals in England and offers another source of gestation at booking. Table AN-4 shows HES figures for first antenatal assessments carried out by 10 weeks of gestation for 2012/13. These figures show that where gestation was known, 36% of women in England had their first antenatal assessment by 10 weeks gestation. This compares with 35% in 2011/12, 32% in 2010/11 and 29% in 2009/10, indicating a year-on-year increase in earlier testing.

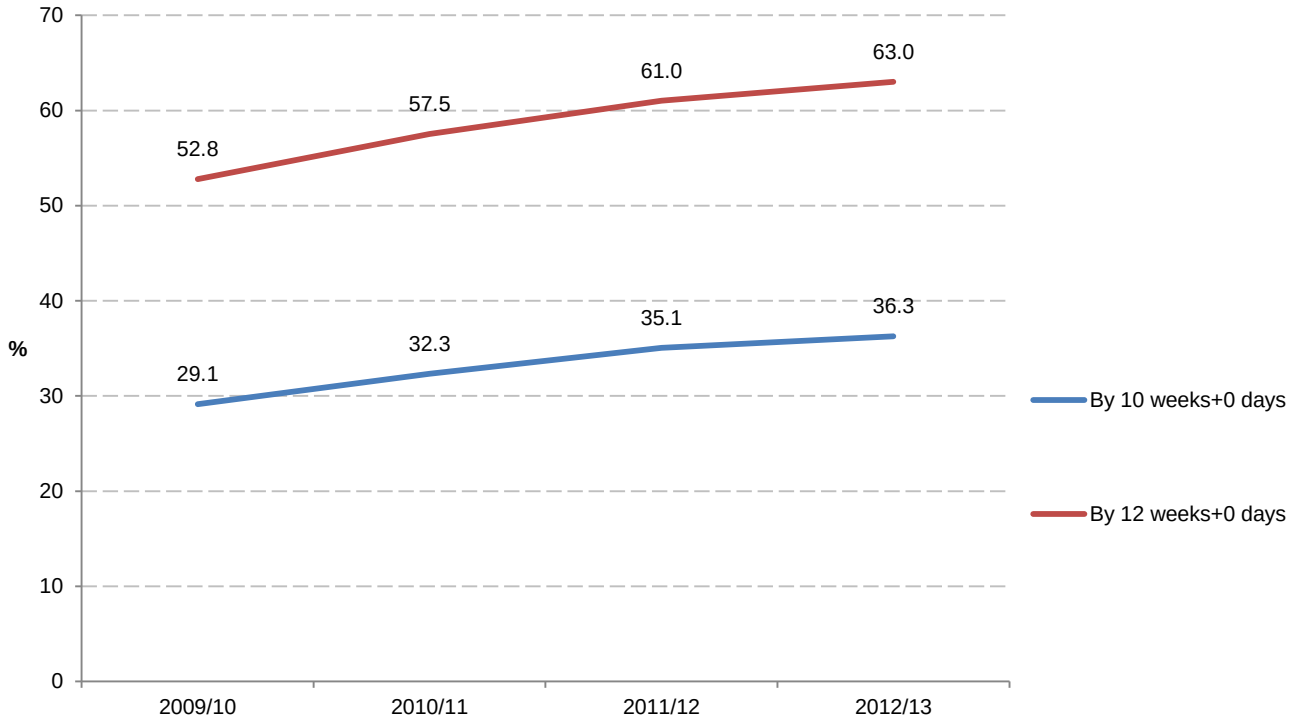
Figure AN-3 shows the proportion of first antenatal assessments by 10+0 weeks and by 12+0 weeks over a four-year period, showing this increase each year.

Table AN-4. First antenatal assessments by 10 weeks and by 12 weeks gestation, 2012/13: England by region

Region	By 10 weeks+0 days		By 12 weeks+0 days		Total known gestations		Total pregnancy episodes (incl. unknown gestations)
	n	% of known gestations	n	% of known gestations	n	% of pregnancy episodes	n
East Midlands	17,103	42	27,242	67	40,615	86	47,016
East of England	25,460	47	39,304	73	54,058	77	70,183
London	24,355	25	52,876	54	97,305	72	135,108
North East	13,762	47	20,756	71	29,128	94	31,094
North West	24,204	35	42,632	62	68,755	77	89,149
South Central	15,028	39	27,720	71	38,949	81	47,945
South East Coast	10,684	33	20,269	63	32,161	59	54,755
South West	21,774	47	34,767	75	46,392	78	59,746
West Midlands	12,686	34	20,993	57	37,077	51	73,141
Yorkshire and The Humber	12,883	28	22,476	49	45,979	73	63,118
England	177,939	36	309,035	63	490,419	73	671,255

Source: Hospital Episode Statistics (HES), The Health and Social Care Information Centre.

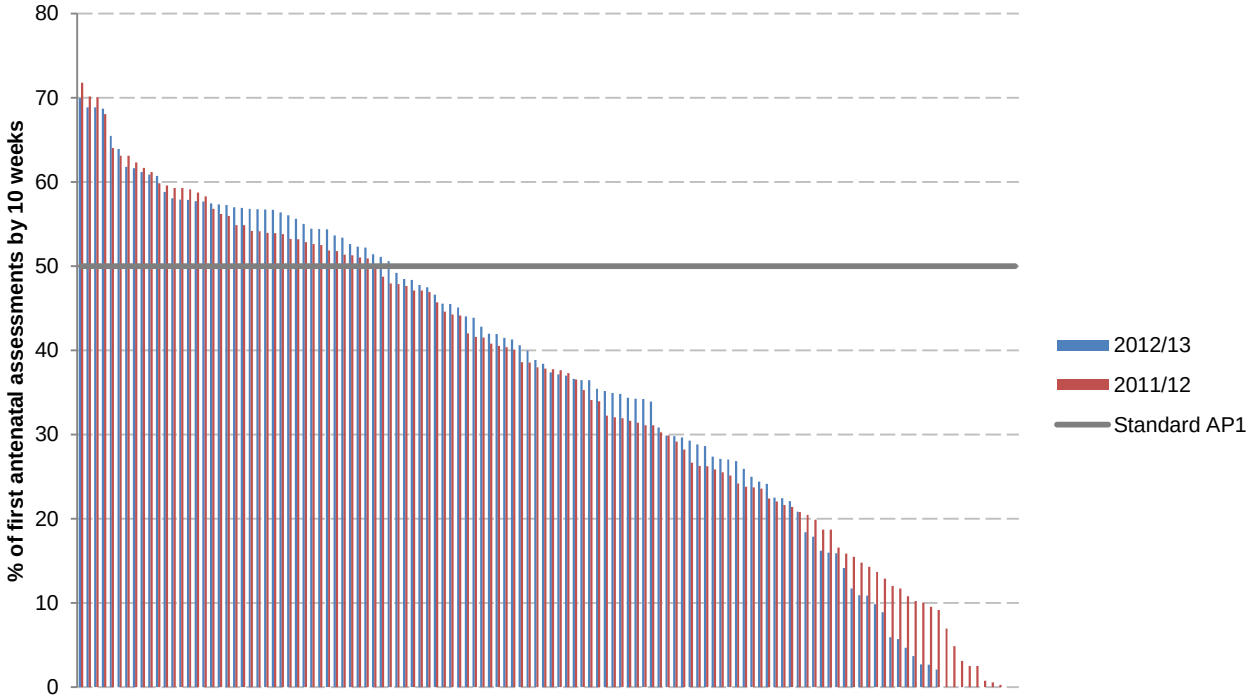
Figure AN-3. First antenatal assessments by 10 weeks and by 12 weeks gestation, 2009-13: England



Source: Hospital Episode Statistics (HES), The Health and Social Care Information Centre.

Figure AN-4 shows the percentage of first antenatal assessments by 10 weeks by Trust for both 2011/12 and 2012/13. On the whole an improvement can be seen, with more Trusts achieving the 50% acceptable level for standard AP1. There are fewer Trusts included for 2012/13 compared to the previous year which may in part account for the appearance of lower performance levels for those below the 20% line.

Figure AN-4. Percentage of first antenatal assessments within 10 weeks gestation, 2011/12 and 2012/13: England by Trust



The reference line represents the 50% acceptable level for Programme Standard AP1.
Source: Hospital Episode Statistics (HES), The Health and Social Care Information Centre.

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).

Table AN-5 shows the usage of the FOQ in identifying potential carriers of alpha zero thalassaemia for the whole of England for 2008-13. Data returns are excluded where data on booking bloods received, FOQ attached, MCH <25pg or the number of high risk alpha zero cases was missing or unavailable. These figures are broken down for high and low risk areas in Table AN-6 and Table AN-7.

In high prevalence areas close to 90% of booking bloods had a FOQ attached in 2012/13, compared to 98% in low prevalence areas. The proportion for the whole of England was 94%. 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.

Table AN-5. Use of the FOQ in determining women at high risk of being alpha zero carriers, 2008-13: England

Year	Booking bloods received (BBs)	FOQ attached		MCH < 25pg (potential alpha0 carrier)		High risk alpha0 (High risk of carrying 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
Total for four year period	3,123,774	2,566,559	82.16	99,855	3.89	16,470	0.53

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.

Table AN-6. Use of the FOQ in determining women at high risk of being alpha zero carriers, 2008-13: High prevalence areas

Year	Booking bloods received (BBs)	FOQ attached		MCH < 25pg (potential alpha0 carrier)		High risk alpha0 (High risk of carrying 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
Total for four year period	1,571,920	1,070,641	68.11	72,372	6.76	11,395	0.72

Exclusions based on missing or unavailable data for the data fields shown: 2008/09: 21; 2009/10: 22; 2010/11: 7; 2011/12: 4; 2012/1

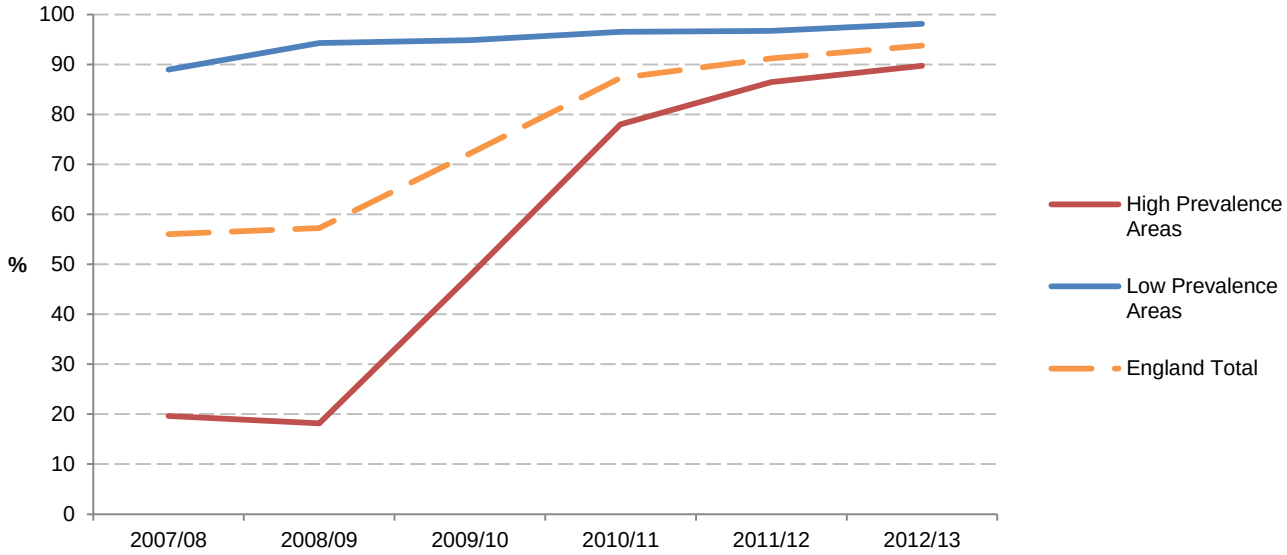
Table AN-7. Use of the FOQ in determining women at high risk of being alpha zero carriers, 2008-13: Low prevalence areas

Year	Booking bloods received (BBs)	FOQ attached		MCH < 25pg (potential alpha0 carrier)		High risk alpha0 (High risk of carrying 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
Total for four year period	1,551,854	1,495,918	96.40	27,483	1.84	5,075	0.33

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:

Figure AN-5 shows the proportion of booking bloods received with a FOQ attached in both high and low prevalence areas across England between 2007 and 2013. Exclusions are made where data on the number of booking bloods received or the number of samples received with a FOQ attached were missing or unavailable. The number of exclusions made has reduced each year, indicating an improvement in data quality. The proportion of booking bloods with a FOQ attached has continued to improve in both high and low prevalence areas, and for the first time high prevalence areas as a whole are close to the 90% acceptable standard for programme standard AO1aiii.

Figure AN-5. Booking bloods received with a FOQ attached, 2007-13: England by prevalence



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.

Table AN-8 shows the numbers and rates of samples received with a FOQ attached by region between 2010 and 2013 for the whole of England (both high and low prevalence areas). Exclusions are made for each year where data on the number of booking bloods received or the number of samples with a FOQ attached were missing or unavailable so as to not bias the rates.

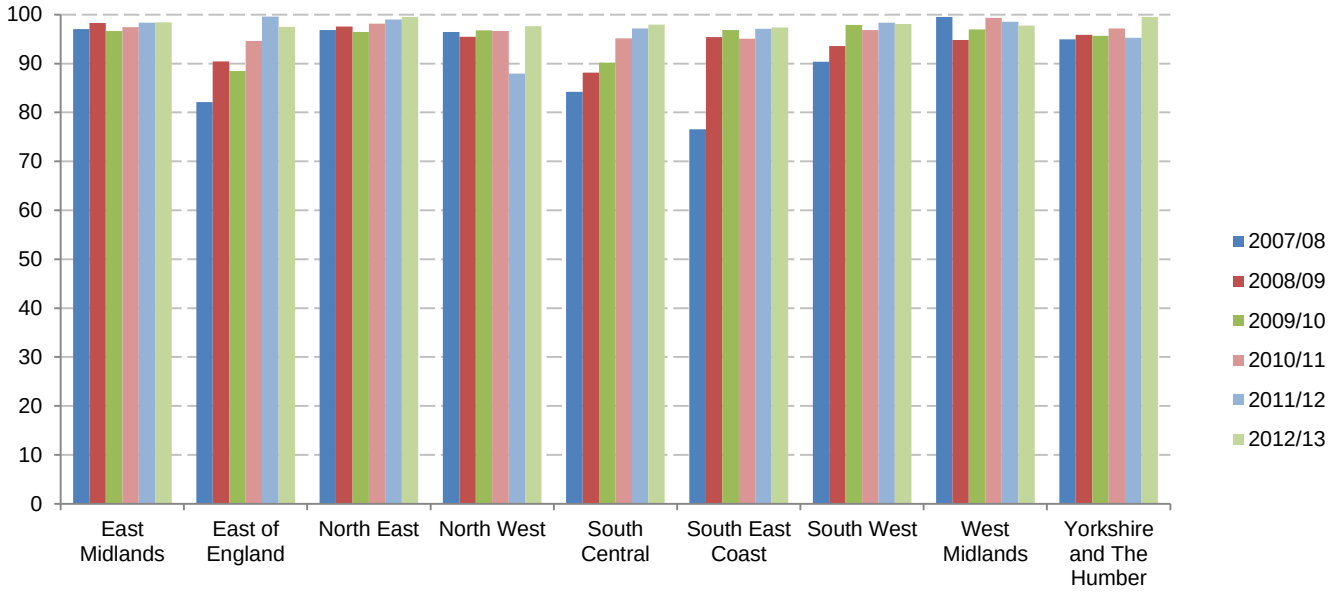
Table AN-8. Booking bloods received with a FOQ attached by region, 2010-13: England

Region	2010/11			2011/12			2012/13		
	Booking Bloods (BBs)	FOQ attached	% of BBs	Booking Bloods (BBs)	FOQ attached	% of BBs	Booking Bloods (BBs)	FOQ attached	% of BBs
East Midlands	54,224	46,307	85.4	55,195	53,653	97.2	53,016	52,238	98.5
East of England	80,942	75,460	93.2	81,767	79,752	97.5	78,123	75,433	96.6
London	112,262	78,564	70.0	142,595	117,395	82.3	125,135	105,123	84.0
North East	34,920	33,961	97.3	34,319	33,697	98.2	33,288	32,799	98.5
North West	90,093	83,296	92.5	90,653	82,461	91.0	91,004	86,892	95.5
South Central	49,516	42,572	86.0	54,676	50,710	92.7	51,803	48,351	93.3
South East Coast	43,879	41,709	95.1	44,716	43,508	97.3	57,885	56,366	97.4
South West	64,254	61,942	96.4	67,534	66,452	98.4	65,570	64,458	98.3
West Midlands	74,301	65,808	88.6	79,565	66,029	83.0	77,760	70,691	90.9
Yorkshire and The Humber	63,187	53,160	84.1	72,381	66,270	91.6	71,773	68,751	95.8
England Total	667,578	582,779	87.3	723,401	659,927	91.2	705,357	661,102	93.7

Exclusions based on missing or unavailable data for the data fields shown: 2010/11: 8; 2011/12: 1; 2012/13: 2.

Figure AN-6 shows these rates for low prevalence laboratories only, showing trends for each region. Whilst there is some variation between years within regions, an overall increase in booking bloods with a FOQ attached can be seen in each region.

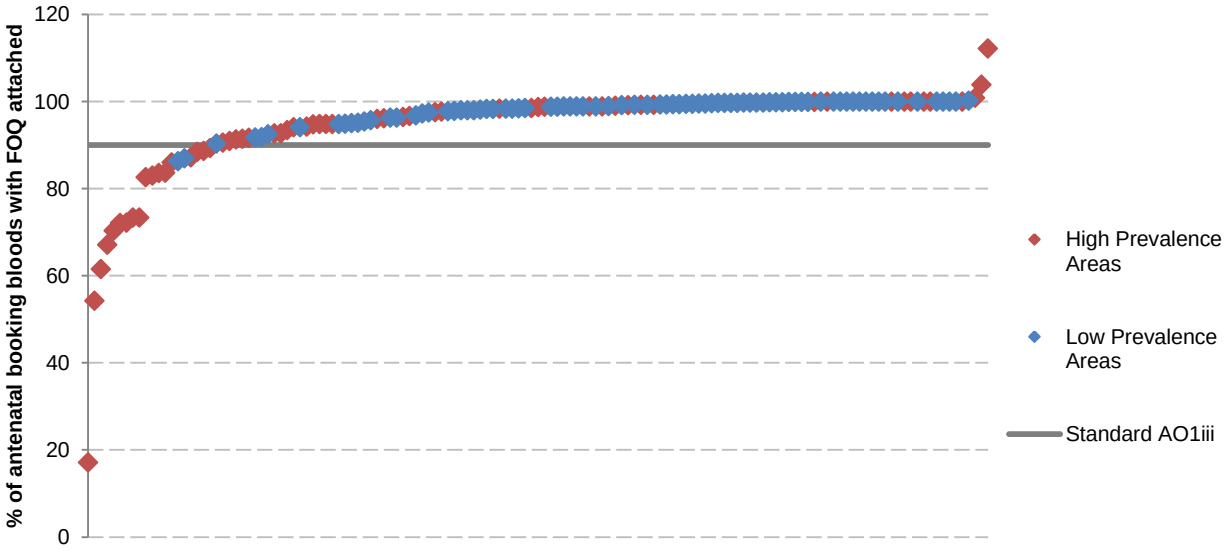
Figure AN-6. Booking bloods received with a FOQ attached, 2007-13: Low prevalence areas by region



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.

Figure AN-7 shows the proportion of booking bloods received with a FOQ attached by each laboratory across the whole of England in 2012/13, highlighting high and low prevalence areas. Nearly all of the low prevalence areas are achieving the 90% acceptable level for programme standard AO1iii, and more high prevalence areas are achieving this standard compared to previous years. Rates over 100% may indicate a data quality issue.

Figure AN-7. Percentage of antenatal booking bloods with a FOQ attached, 2012/13: 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 a FOQ attached or the total number of booking bloods was missing or unavailable.

3.5. Tests not performed due to a known previous result

Table AN-9 shows the number and rates of pregnant women who were not tested due to a known previous test result in England by region. These figures combine previous screen positive and negative results and exclude data returns where both of these fields were missing or unavailable. In 2012/13 approximately 6% of women screened had a known previous result, or one in 16 women screened.

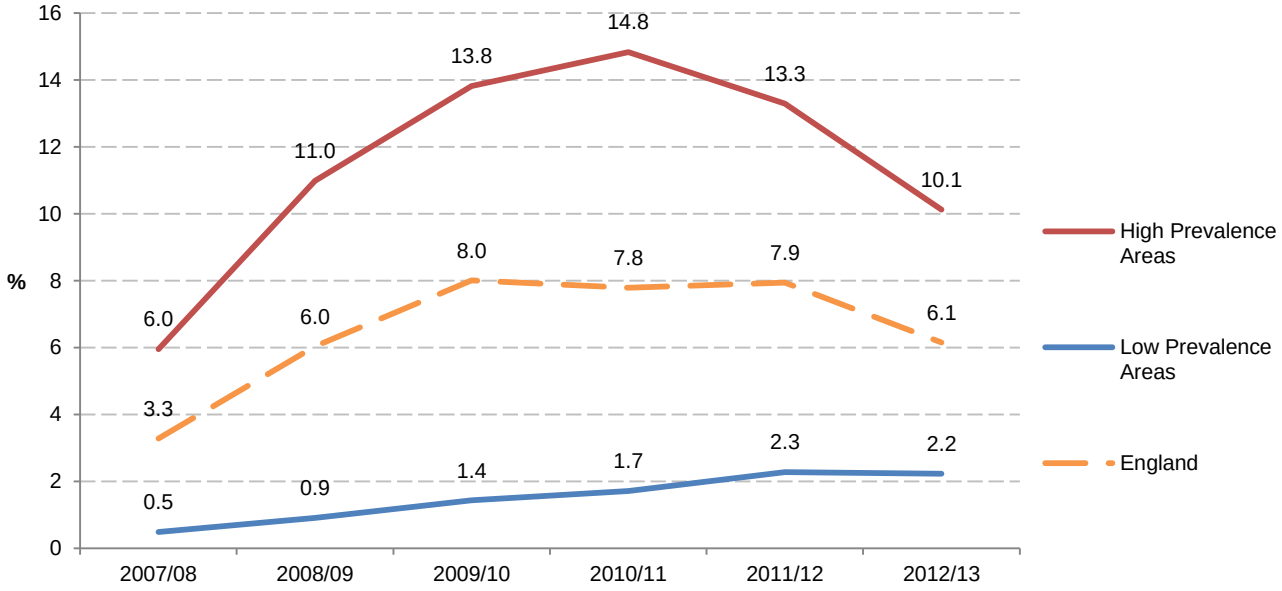
Table AN-9. Pregnant women where testing was not indicated due to a known previous result, 2010-13: England by region

Region	2010/11			2011/12			2012/13		
	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	38,118	6,029	15.82	49,991	6,492	12.99	40,763	6,191	15.19
East of England	76,474	1,981	2.59	76,421	2,508	3.28	72,604	2,332	3.21
London	101,977	11,056	10.84	100,504	9,016	8.97	79,069	3,006	3.80
North East	33,113	2,365	7.14	29,059	2,680	9.22	33,288	2,882	8.66
North West	59,407	2,570	4.33	53,813	3,024	5.62	71,164	2,730	3.84
South Central	45,779	6,056	13.23	54,676	10,353	18.94	44,986	4,033	8.97
South East Coast	24,761	801	3.23	38,166	1,224	3.21	45,054	1,277	2.83
South West	44,133	642	1.45	54,831	1,256	2.29	53,555	791	1.48
West Midlands	60,450	5,949	9.84	72,422	6,728	9.29	68,040	8,298	12.20
Yorkshire and The Humber	48,909	4,055	8.29	68,503	4,249	6.20	50,528	2,832	5.60
England Total	533,121	41,504	7.79	598,386	47,530	7.94	559,051	34,372	6.15

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: 2010/11: 33; 2011/12: 27; 2012/13: 31.

Figure AN-8 shows trends in the proportion with a previously known test result by prevalence and for the whole of England. Rates are consistently higher in high prevalence areas than in low prevalence areas. Nationally there appears to have been a decline in the proportion of booking bloods with a previously known result compared to previous years. There appears to have been a decline in high prevalence areas, whereas rates have been rising in low prevalence areas.

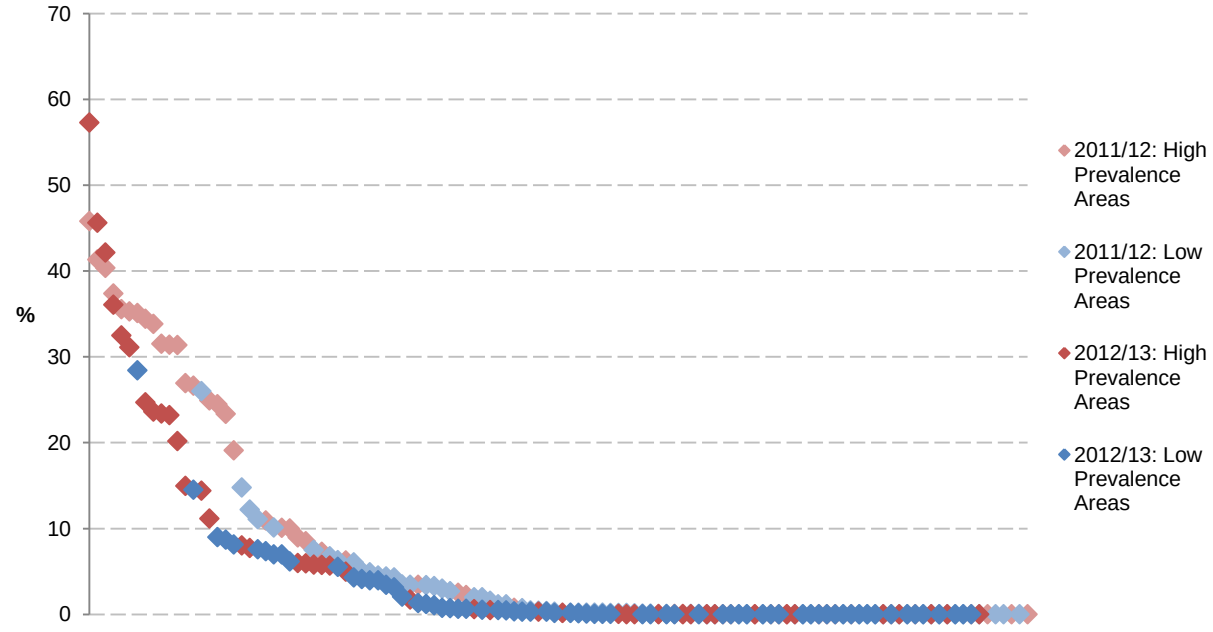
Figure AN-8. Percentage of pregnant women where testing was not indicated due to a known previous test result, 2007-13: England by prevalence



Exclusions based on missing or unknown data on booking bloods received or where data on both previous screen positive and previous screen negative were missing or unavailable: 2007/08: 35; 2008/09: 50; 2009/10: 45; 2010/11: 33; 2011/12: 27; 2012/13: 31.

Figure AN-9 shows the distribution of known previous test results across laboratories and by prevalence. The decline shown in Figure AN-8 can also be seen here. Rates are generally higher in high prevalence areas than in low prevalence areas, accounting for nearly all of those above 10%. A number of laboratories show 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.

Figure AN-9. Percentage of pregnant women where testing was not indicated due to a known previous result, 2011-13: 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: 2011/12: 27; 2012/13: 31.

3.6. Declined screening tests

Table AN-10 shows the number and rates of pregnant women who declined antenatal screening by region, covering the whole of England between 2010 and 2013. These figures exclude data from laboratories where data on the number of booking bloods received or tests that were declined missing or unavailable.

In 2012/13 approximately 0.6% of booking bloods received were identified as having declined testing for sickle cell disease and thalassaemia. The rates vary by region, ranging between 0.07% in London and 1.32% in the South West.

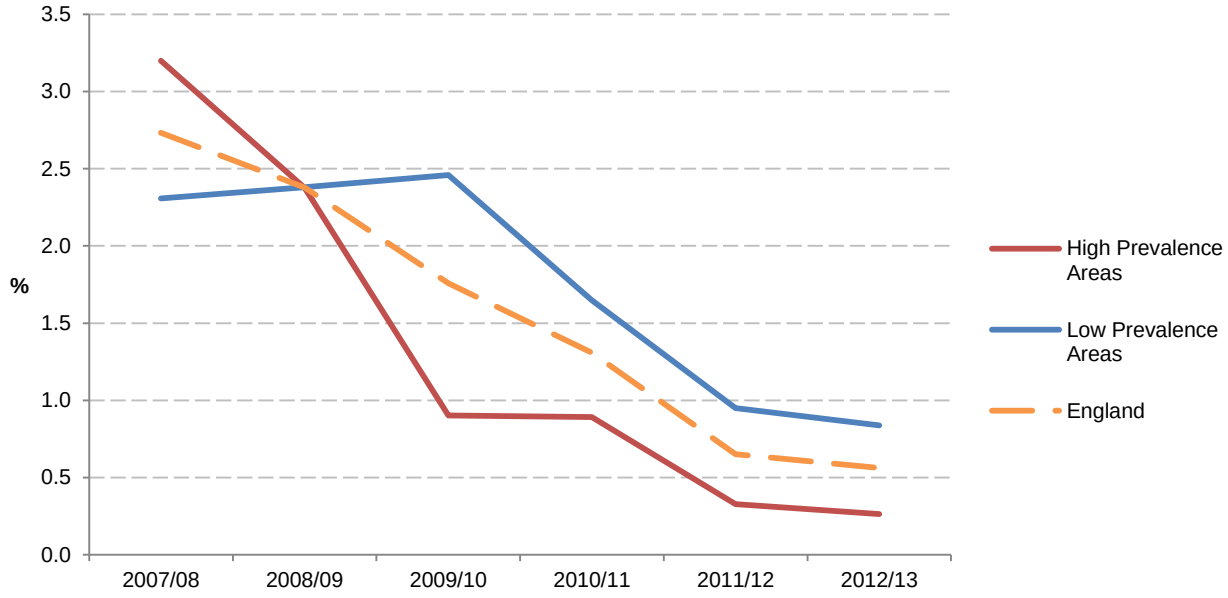
Table AN-10. Declined tests by region, 2010-13: England

Region	2010/11			2011/12			2012/13		
	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	54,224	259	0.48	55,195	164	0.30	53,016	210	0.40
East of England	80,942	1,866	2.31	81,767	844	1.03	78,123	580	0.74
London	92,786	204	0.22	102,155	184	0.18	103,108	76	0.07
North East	34,920	757	2.17	34,319	385	1.12	33,288	317	0.95
North West	75,933	2,159	2.84	75,842	477	0.63	75,294	555	0.74
South Central	45,779	297	0.65	50,484	225	0.45	47,740	80	0.17
South East Coast	58,491	28	0.05	54,925	109	0.20	52,783	258	0.49
South West	55,360	1,103	1.99	58,273	860	1.48	61,527	810	1.32
West Midlands	65,740	285	0.43	74,944	449	0.60	73,336	191	0.26
Yorkshire and The Humber	70,597	1,340	1.90	72,381	604	0.83	71,773	571	0.80
England Total	634,772	8,298	1.31	660,285	4,301	0.65	649,988	3,648	0.56

Exclusions based on missing or unavailable data: 2010/11: 17; 2011/12: 14; 2012/13: 14.

Figure AN-10 shows trends in the proportion of booking bloods received where testing was declined between 2007/08 and 2012/13. The proportion of declined tests appears to have declined each year since 2009/10, and has been consistently lower in high prevalence areas than in low prevalence areas.

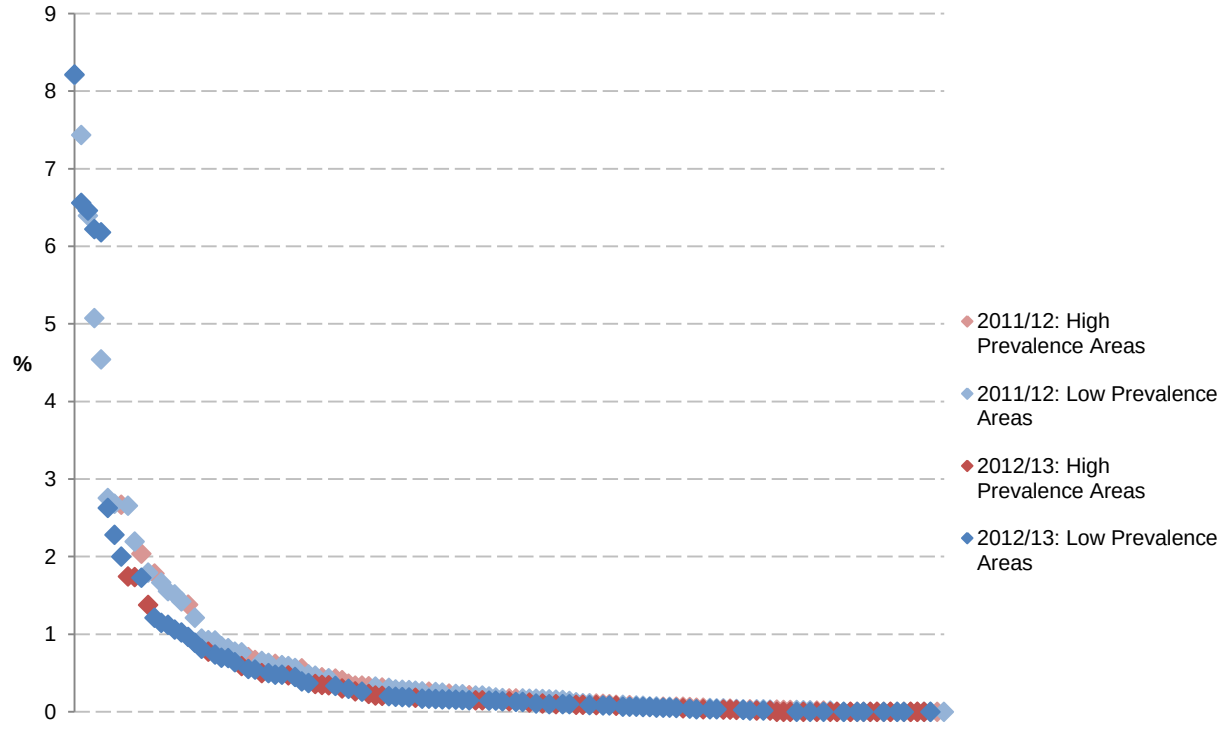
Figure AN-10. Declined tests as a percentage of booking bloods received, 2007-13: England by prevalence



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.

Figure AN-11 shows the percentage of pregnant women who declined testing for sickle cell disease and thalassaemia by laboratory and by prevalence for 2011/12 and 2012/13. There appear to be a few laboratories that had higher rates of declined testing compared to other laboratories.

Figure AN-11. Percentage of women that declined testing, 2011-13: England by laboratory



Each marker represents one laboratory.

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 2012/13. There are eight exclusions made 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 were missing or unavailable.

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 may 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 2012/13: 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	53,016	677	1.28	713	105.32	555	77.84	28	5.05
East of England	78,123	1,010	1.29	1,057	104.65	693	65.56	59	8.51
London	125,172	5,981	4.78	6,218	103.96	3,154	50.72	404	12.81
North East	33,288	259	0.78	256	98.84	208	81.25	12	5.77
North West	79,353	1,039	1.31	890	85.66	681	76.52	63	9.25
South Central	51,803	788	1.52	836	106.09	631	75.48	48	7.61
South East Coast	57,885	724	1.25	725	100.14	543	74.90	42	7.73
South West	63,395	437	0.69	430	98.40	338	78.60	21	6.21
West Midlands	67,846	1,529	2.25	1,417	92.67	925	65.28	82	8.86
Yorkshire and The Humber	71,773	916	1.28	890	97.16	688	77.30	56	8.14
England total	681,654	13,360	1.96	13,432	100.54	8,416	62.66	815	9.68

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.

Across England 13,432 father specimens were requested, which equates approximately (allowing for regional variation, including those greater than 100%) to the number of screen positive women identified. Of these, laboratories reported that 8,416 samples were received, or approximately 63% of those requested. Of the father specimens received, 815 (approximately 10%) were identified as being carriers and the pregnancies at high risk for a sickle cell disease or thalassaemia affected baby. Father test uptake varies between regions, between approximately 51% in London and approximately 81% in the North East.

It is not possible to assess the father's risk status in cases where he was not available for testing, and so the pregnancy will be deemed 'at risk'. These cases are not included in the number of 'high risk' couples, but account for approximately 37% 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 2010/11 to 2012/13 by region. Exclusions are made where data on the number of booking bloods received, the number of father specimens requested, or the number of father specimens received, were missing or unavailable.

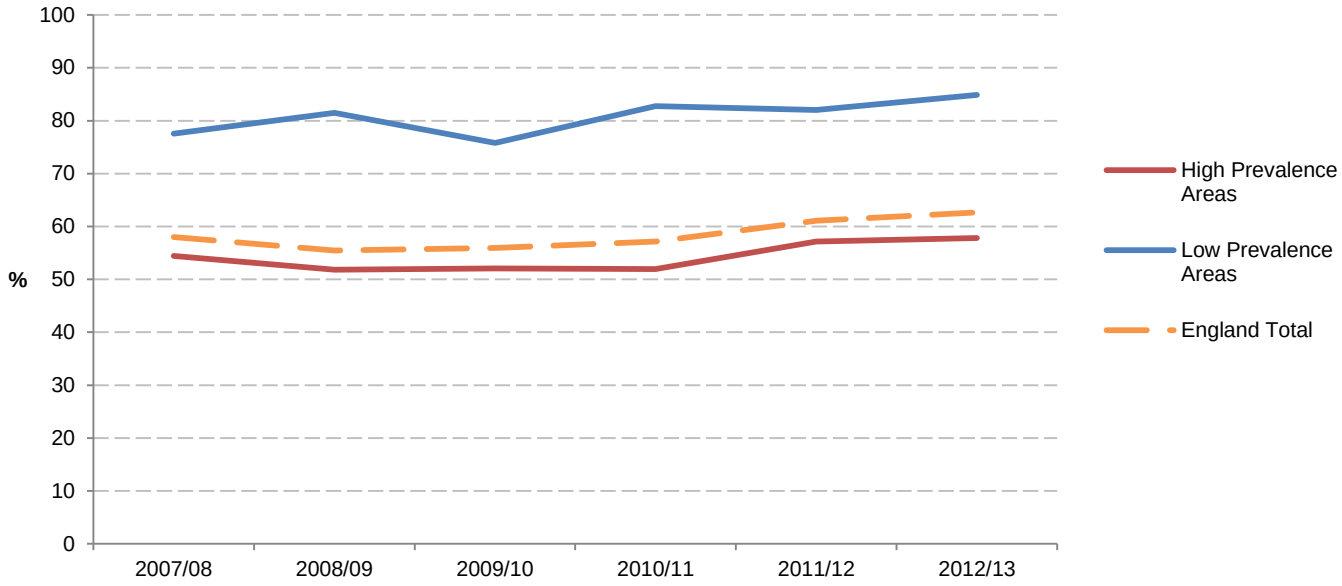
Table AN-12. Uptake of father testing, 2010-13: England by region

SHA	2010/11			2011/12			2012/13		
	Fathers requested	Fathers received	% uptake	Fathers requested	Fathers received	% uptake	Fathers requested	Fathers received	% uptake
East Midlands	671	421	62.74	721	577	80.03	713	555	77.84
East of England	1,110	701	63.15	1,027	694	67.58	1,057	693	65.56
London	8,774	4,218	48.07	8,296	4,290	51.71	6,218	3,154	50.72
North East	254	218	85.83	214	187	87.38	256	208	81.25
North West	822	592	72.02	826	583	70.58	996	740	74.30
South Central	785	618	78.73	997	735	73.72	836	631	75.48
South East Coast	763	546	71.56	577	443	76.78	725	543	74.90
South West	480	366	76.25	458	338	73.80	430	338	78.60
West Midlands	1,499	770	51.37	1,521	872	57.33	1,434	942	65.69
Yorkshire and The Humber	641	577	90.02	859	746	86.85	890	688	77.30
England total	15,799	9,027	57.14	15,496	9,465	61.08	13,555	8,492	62.65

Exclusions based on missing or unavailable data: 2010/11: 1; 2011/12: 3; 2012/13: 3.

Figure AN-12 shows the national rates for father uptake between 2007/08 and 2012/13 by prevalence. Across England the percentage of father uptake appears to have increased since 2008/09, from approximately 55% to approximately 63% uptake. In high prevalence areas father uptake has ranged between approximately 55% – 58%, while in low prevalence areas it has ranged between approximately 75% – 85% uptake.

Figure AN-12. Uptake of father testing, 2007-13: 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.

The Programme requests breakdown data on mother and father results to identify the specific risk of an affected pregnancy. Table AN-13 shows the risk status of pregnancies for antenatal screening by the mother's results for 2012/13. 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 14,515 screen positive women identified in antenatal screening, 13,564 (93%) are included in this breakdown, and of the 858 'high risk' couples identified, 731 (85%) are included here.

Of the 13,564 screen positive women included in the breakdown, 7,360 (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 5,691 screen positive women (42%) 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 513 screen positive women (4%) were identified with compound heterozygous results including one or more of the above results, cases where a donor egg was used or where there was a bone marrow transplant, or identified with Hereditary Persistence of Fetal Haemoglobin (HPFH).

High risk pregnancies are those that are represented by the dark orange boxes on the breakdown 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, 2012/13: 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
Possible sickle cell affected baby	Hb S	511	9	70	2,267	2,750	5,607	7,360
	Hb D	7	-	45	449	158	659	
	Hb C	49	-	34	442	555	1,080	
	Hb O-Arab	0	0	0	7	7	14	
Possible thalassaemia affected baby	βThalassaemia	126	0	50	2,094	1,019	3,289	5,691
	δβ thalassaemia	0	*	*	29	8	40	
	High risk alpha0	17	-	24	564	913	1,518	
	Hb E	7	*	42	606	179	836	
	Hb Lepore	0	*	0	3	3	8	
Other clinically significant mother results	HPFH/Compound heterozygous/donor egg/ bone marrow transplant	14	11	21	305	162	513	513
	Totals	731	25	288	6,766	5,754	13,564	13,564

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 14,515 (93% included here) and 858 high risk couples (85% included here).

4. Prenatal diagnostic (PND) testing data

4.1. Data quality and methods of data collection

Response rate:

Data were received from all three PND laboratories, including pregnancy outcome data, for the period 1st April 2012 – 31st March 2013. We would like to commend the PND laboratories for this excellent response rate.

Data quality:

The proportion of PND tests where the gestation was missing or unknown has decreased, and in 2012/13 approximately 99% of PND tests performed had information on gestation at test. However, the proportion of tests performed where the outcome (i.e. whether the couple continued the pregnancy, miscarried, or opted for termination) was unknown has increased. In 2012/13, of all PND tests performed, 40% had a missing outcome, compared to 32% and 37% in 2010/11 and 2011/12 respectively. Of the affected pregnancies identified, outcome data were available for 62%, compared to 66% in 2011/12 and 80% in 2010/11.

4.2. Numbers tested and detected

Table PND-1 shows the numbers of PND tests performed by each PND laboratory by year since 2007.

Table PND-1. Number of PNDs performed, 2004-13: England by laboratory

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

Table PND-2 shows the number of PND tests performed by the mother's region by year since 2007/08.

Table PND-2. Number of PNDs performed, 2007-13: England by region

Region	2007/08		2008/09		2009/10		2010/11		2011/12		2012/13		Total
	n	%	n	%	n	%	n	%	n	%	n	%	
East Midlands	13	4.0	-	0.0	9	2.3	18	4.3	12	2.9	4	1.0	84
East of England	24	7.4	20	5.2	31	7.8	30	7.1	22	5.3	21	5.3	195
London	205	63.1	184	47.7	232	58.6	267	63.6	250	59.8	195	49.0	1,881
North East	5	1.5	*	0.3	8	2.0	4	1.0	4	1.0	*	0.3	35
North West	18	5.5	-	0.0	26	6.6	22	5.2	21	5.0	*	0.3	127
South Central	15	4.6	*	0.5	14	3.5	14	3.3	12	2.9	*	0.3	97
South East Coast	5	1.5	9	2.3	9	2.3	12	2.9	12	2.9	17	4.3	69
South West	3	0.9	*	0.3	5	1.3	12	2.9	4	1.0	*	0.3	38
West Midlands	18	5.5	-	0.0	28	7.1	16	3.8	21	5.0	3	0.8	139
Yorkshire and the Humber	10	3.1	*	0.3	15	3.8	16	3.8	11	2.6	4	1.0	95
Region unknown	9	2.8	168	43.5	19	4.8	9	2.1	49	11.7	150	37.7	458
Total	325	100.0	386	100.0	396	100.0	420	100.0	418	100.0	398	100.0	3,218

** Numbers less than 3 have been suppressed.*

Table PND-3 shows the number of affected, carrier and 'NAD' (No Abnormality Detected) results for each year since 2007/08. In each year the results reflect the expected 25:50:25 ratio of affected, carrier, and NAD results respectively.

Table PND-3. PND fetal results, 2007-13: England

Fetal result	2007/08		2008/09		2009/10		2010/11		2011/12		2012/13	
	n	% of total	n	% of total	n	% of total	n	% of total	n	% of total	n	% of total
Affected	90	27.7	84	21.8	102	25.8	95	22.6	101	24.2	85	21.4
Carrier	152	46.8	201	52.1	198	50.0	224	53.3	207	49.5	213	53.5
NAD	78	24.0	96	24.9	96	24.2	100	23.8	104	24.9	96	24.1
Inconclusive/ Missing result	5	1.5	5	1.3	0	0.0	*	0.2	6	1.4	4	1.0
Total	325	100.0	386	100.0	396	100.0	420	100.0	418	100.0	398	100.0

** Numbers less than 3 have been suppressed.*

Table PND-4 provides a breakdown of these results by condition or risk between 2007/08 and 2012/13.

Table PND-4. Breakdown of PND fetal results by condition, 2007-13: England

Fetal result	PND result/risk	2007/08	2008/09	2009/10	2010/11	2011/12	2012/13
Affected	Sickle Cell affected	66	60	79	66	85	68
	Thalassaemia affected	24	24	23	29	15	17
	Other	0	0	0	0	*	0
Carrier	Sickle Cell carrier	110	150	152	174	154	169
	Thalassaemia carrier	42	51	46	50	52	42
	Other	0	0	0	0	*	*
NAD	Risk for Sickle Cell	33	58	72	80	63	52
	Risk for Thalassaemia	22	25	24	20	6	18
	Risk not known	23	13	0	0	35	26
Inconclusive†	All risks	5	5	0	*	6	4
Total		325	386	396	420	418	398

†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.
** Numbers less than 3 have been suppressed.*
Alpha and beta thalassaemia cases are grouped due to the small number of alpha thalassaemia cases.

4.3. Gestation data

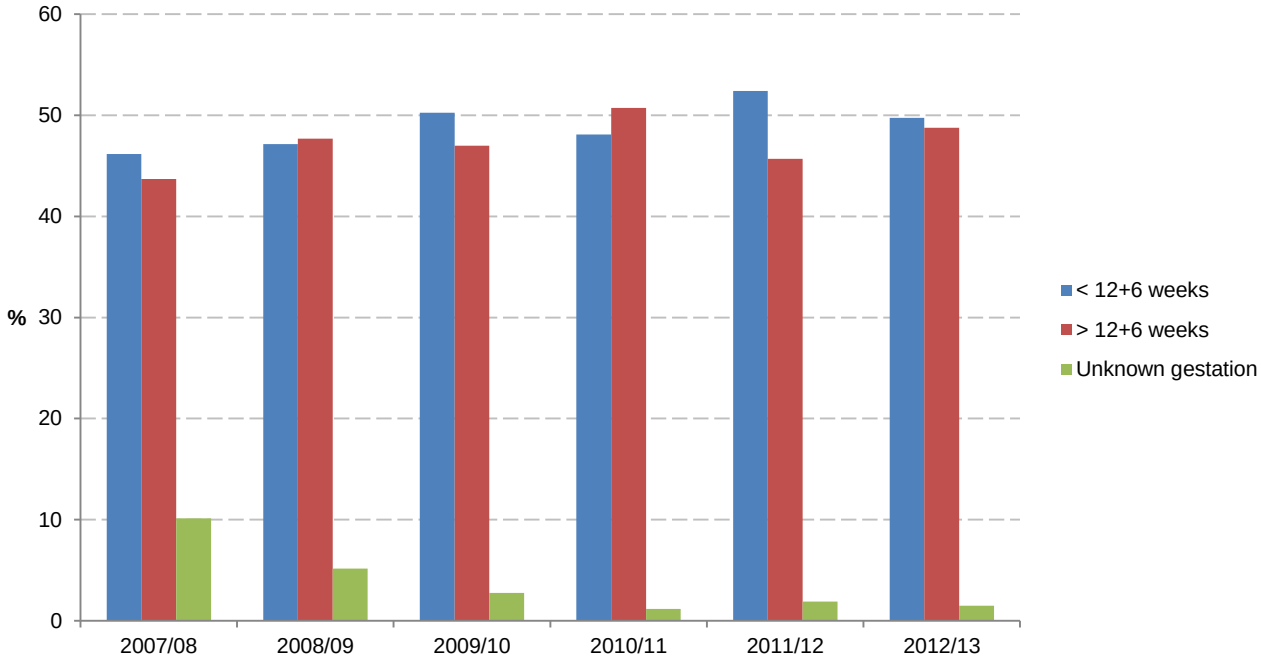
Programme standard AO1b requires 50% of all prenatal diagnoses to be performed by 12 weeks and six days of gestation as a minimum standard, and 75% as an achievable standard. Table PND-5 shows the gestation at which PND tests were performed between 2007/08 and 2012/13.

Table PND-5. Gestation at sample for PND, 2007-13: England

Gestation	2007/08		2008/09		2009/10		2010/11		2011/12		2012/13	
	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
13+0 - 14+6 weeks	49	15.1	65	16.8	76	19.2	105	25.0	93	22.2	71	17.8
≥15+0 weeks	93	28.6	119	30.8	110	27.8	108	25.7	98	23.4	123	30.9
Unknown gestation	33	10.2	20	5.2	11	2.8	5	1.2	8	1.9	6	1.5
Total	325	100.0	386	100.0	396	100.0	420	100.0	418	100.0	398	100.0

Figure PND-1 shows the proportion tested before and after 12 weeks and six days and the proportion with an unknown gestation. The proportions have been fairly steady each year at approximately 50% of the PND tests performed. It is important to continue efforts to ensure that the acceptable standard is maintained and to work towards the achievable standard of 75% in order to ensure early testing to facilitate parents to make informed choices.

Figure PND-1. Percentage of PND tests performed by gestation and year, 2007-13: England



4.4. Results by ethnicity

Table PND-6 shows the number of PND tests performed each year by mother's ethnic grouping (as reported to the laboratories) between 2007/08 and 2012/13. In 2012/13 mothers from an African background accounted for nearly half of all PNDs performed and mothers from a Caribbean background accounted for approximately 4% of all PNDs performed in the year. Mothers from mixed or other ethnic backgrounds accounts for nearly 20% of PND tests performed. Mixed ethnicities can include combinations of ethnic groupings which prevents categorisation into these groups.

Table PND-6. Number of PND tests by mother's ethnicity, 2007-13: England

Mother's ethnic grouping	2007/08		2008/09		2009/10		2010/11		2011/12		2012/13	
	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
Caribbean	15	4.6	22	5.7	22	5.6	23	5.5	12	2.9	14	3.5
Indian	18	5.5	9	2.3	14	3.5	16	3.8	9	2.2	9	2.3
Pakistani	16	4.9	13	3.4	16	4.0	19	4.5	*	0.5	*	0.5
Cypriot/Mixed Cypriot	10	3.1	10	2.6	10	2.5	11	2.6	7	1.7	4	1.0
Other Asian	15	4.6	22	5.7	23	5.8	16	3.8	50	12.0	16	4.0
Southern & Other European	6	1.8	6	1.6	6	1.5	5	1.2	3	0.7	8	2.0
Middle Eastern	10	3.1	9	2.3	12	3.0	5	1.2	4	1.0	4	1.0
Mixed/Other	*	0.6	8	2.1	*	0.3	8	1.9	97	23.2	77	19.3
Not Know n	75	23.1	103	26.7	80	20.2	77	18.3	23	5.5	84	21.1
Total	325	100.0	386	100.0	396	100.0	420	100.0	418	100.0	398	100.0

**Numbers less than 3 have been suppressed*

4.5. Pregnancy outcomes

The collection of data on pregnancy outcomes for PND tests began in 2008/09. Table PND-7 shows the total number of PND tests performed by pregnancy outcome where an affected baby was identified.

Table PND-7. Number of PND tests with affected results by outcome, 2008-13: England

Outcome	2008/09		2009/10		2010/11		2011/12		2012/13	
	n	%	n	%	n	%	n	%	n	%
Continued	9	10.7	19	18.6	26	27.4	18	17.8	13	15.3
Terminated	27	32.1	41	40.2	50	52.6	48	47.5	38	44.7
Miscarriage	*	1.2	0	0.0	0	0.0	*	1.0	*	2.4
Not Known	47	56.0	42	41.2	19	20.0	34	33.7	32	37.6
Total	84	100.0	102	100.0	95	100.0	101	100.0	85	100.0

**Numbers less than 3 have been suppressed.*

Table PND-8 shows PND tests with affected results by pregnancy outcome for each condition as numbers and as a proportion of those identified with each condition.

Table PND-8. Outcomes for pregnancies with affected fetal diagnoses at PND, 2008-13: England by condition

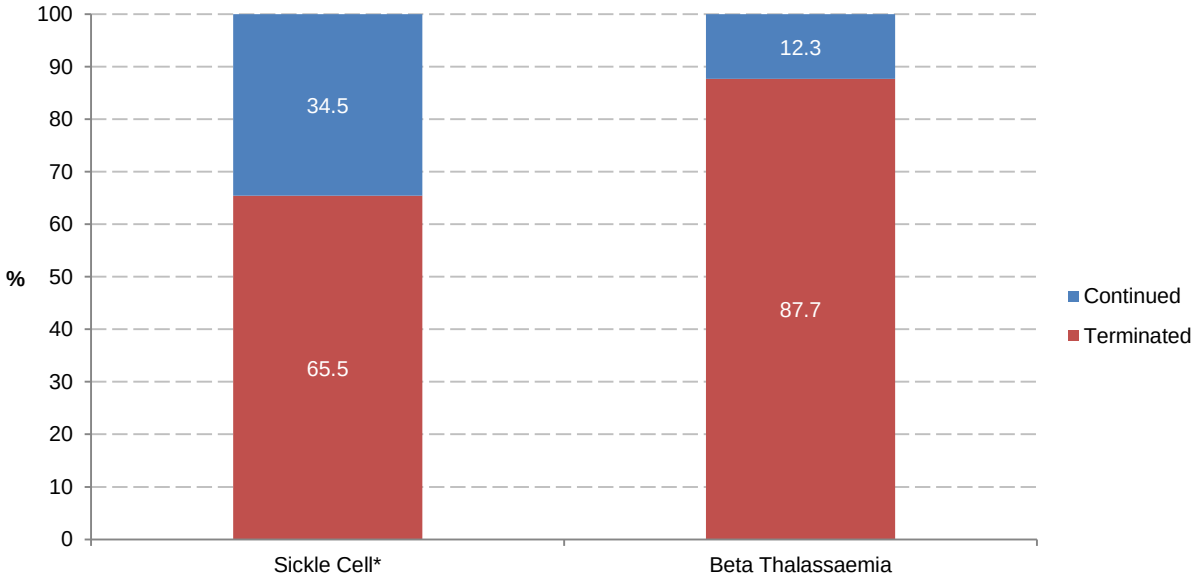
Condition	Pregnancy outcome	2008/09		2009/10		2010/11		2011/12		2012/13	
		n	% of total identified with condition	n	% of total identified with condition	n	% of total identified with condition	n	% of total identified with condition	n	% of total identified with condition
Sickle Cell	Continued	8	13.3	18	22.8	21	31.8	17	20.0	12	17.6
	Terminated	17	28.3	30	38.0	31	47.0	38	44.7	28	41.2
	Miscarried	*	1.7	0	0.0	0	0.0	0	0.0	*	2.9
	Not Known	34	56.7	31	39.2	14	21.2	30	35.3	26	38.2
Beta Thalassaemia	Continued	0	0.0	*	4.8	4	16.0	*	8.3	*	7.1
	Terminated	8	38.1	9	42.9	17	68.0	8	66.7	8	57.1
	Miscarried	0	0.0	0	0.0	0	0.0	*	8.3	0	0.0
	Not Known	13	61.9	11	52.4	4	16.0	*	16.7	5	35.7
Alpha Thalassaemia	Continued	*	33.3	0	0.0	*	25.0	0	0.0	0	0.0
	Terminated	*	66.7	*	100.0	*	50.0	*	66.7	*	66.7
	Miscarried	0	0.0	0	0.0	0	0.0	0	0.0	0	0.0
	Not Known	0	0.0	0	0.0	*	25.0	*	33.3	*	33.3
Total Affected		84		102		95		100		85	

**Numbers less than 3 have been suppressed.*

Affected results for other haemoglobinopathies have been excluded

Figure PND-2 shows the proportion of PND tests with an affected result that opted to continue the pregnancy and opted to terminate the pregnancy in the period 2008/09 to 2012/13. This excludes alpha thalassaemia results and miscarriages as the numbers are low and cases where the pregnancy outcome was not known. Of the PND tests performed where an affected sickle cell result was identified and there was a known outcome, 65.5% opted to terminate the pregnancy and 34.5% opted to continue the pregnancy. Of the PND tests performed where an affected beta thalassaemia result was identified, 87.7% opted to terminate and 12.3% opted to continue the pregnancy.

Figure PND-2. Outcomes for pregnancies with affected fetus diagnosis at PND, 2008-13: England by condition



**"Sickle Cell" includes cases where the result was sickle cell and thalassaemia*
Excludes alpha thalassaemia cases, miscarriage outcomes, and 174 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, combining figures for 2008/09 to 2012/13. For affected results for beta thalassaemia where the couple opted to terminate, the majority were tested by 12 weeks and six days of gestation (38 tested compared to 11 tested after this gestational period). For affected results for sickle cell where the couple opted to terminate, 65 were tested by 12 weeks and six days, compared to 76 tested after this gestational period.

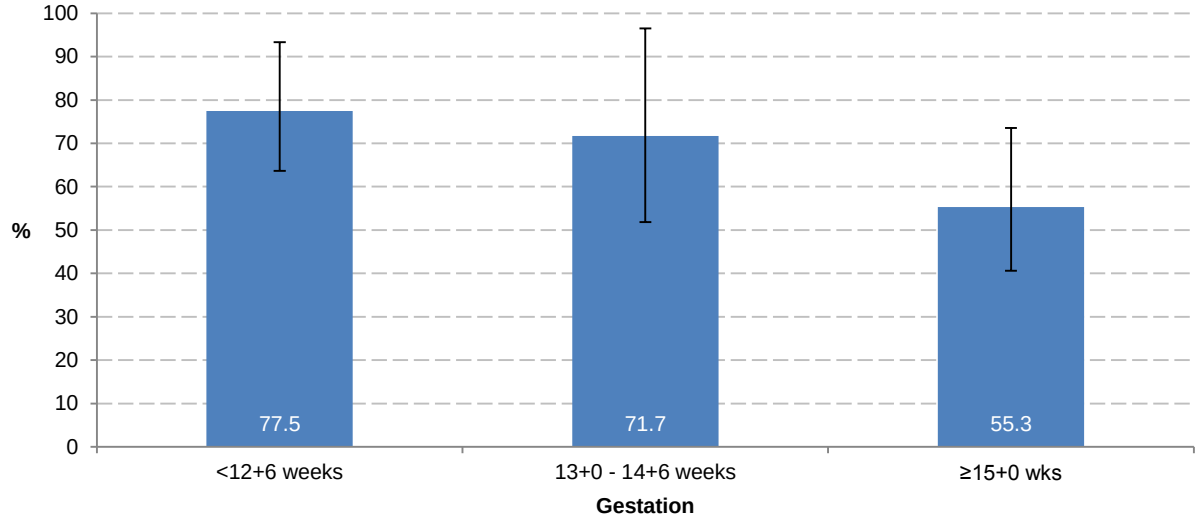
Table PND-9. Gestation at PND for affected results with a known outcome, 2008-13: England

		<12+6 weeks	13+0 - 14+6 weeks	≥15+0 wks
Condition	Outcome	n	n	n
Sickle Cell	Continued	23	15	36
	Terminated	65	34	42
Beta Thalassaemia	Continued	6	*	0
	Terminated	38	7	4

**Numbers less than 3 have been suppressed.*
Excludes affected PND tests where the outcome was unknown, where there was a miscarriage, or where gestation at PND was unknown.

Figure PND-3 shows the proportion of affected results where the parents opted to terminate the pregnancy, shown by gestation for all conditions combined with 95% confidence intervals shown. 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.

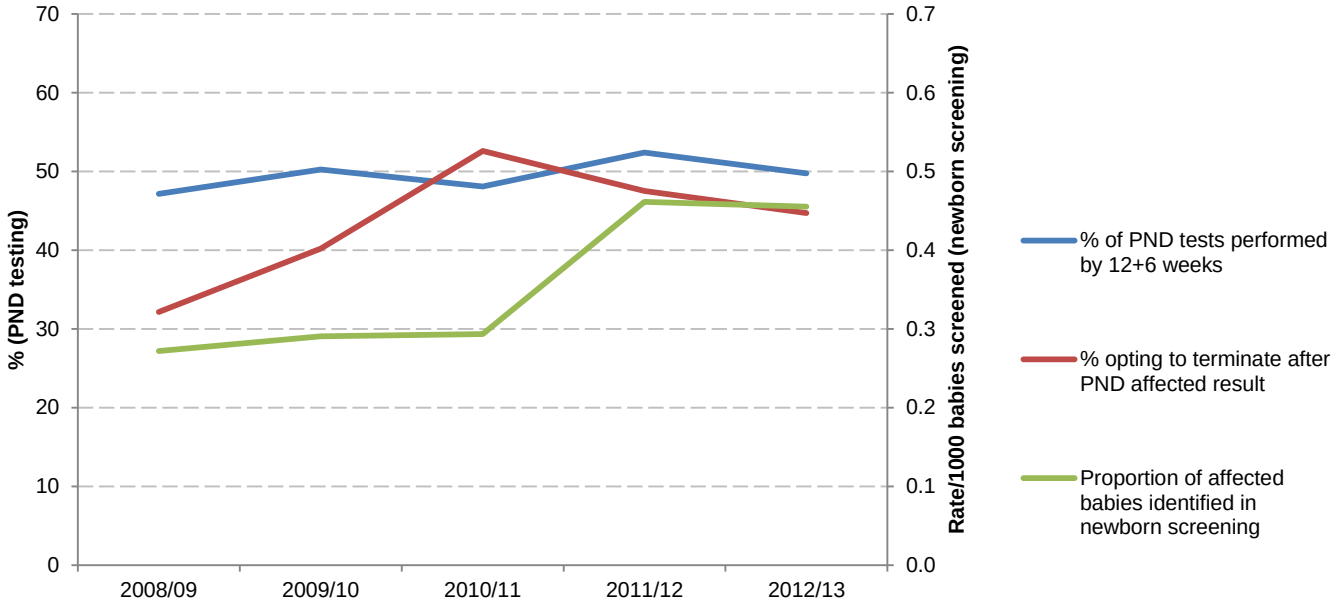
Figure PND-3. Percentage of affected results where parents opted to terminate, by gestation at PND for known pregnancy outcomes, 2008-13



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

Figure PND-4 shows a comparison of the proportion of PND tests performed by 12 weeks and six days, the proportion of affected PND results where parents opted to terminate the pregnancy, and the proportion of affected babies identified in newborn screening. There appears to be a correlation between the percentage of PND tests performed by 12 weeks and six days and the proportion of affected results opting to terminate the pregnancy, which would indicate the benefits of early screening. However, there is not currently sufficient data to identify if there is a link between the proportion opting to terminate and the proportion of babies identified with a significant result in newborn screening.

Figure PND-4. Comparison of early PND testing, terminations for affected results and affected results in newborn screening, 2008-13: England



5. Newborn screening data

5.1. Data quality and methods of data collection

Response rate:

The sickle cell and thalassaemia data return templates were sent to the 13 newborn laboratories in England and there was a 100% response rate. We would like to commend the newborn laboratories for this excellent response rate.

Data quality:

Newborn laboratories report on 'results' which may differ from the number of babies tested. Data by region and data by ethnicity are collected separately. This can lead to discrepancies when comparing the figures which can be a result of exclusions being made, for example for samples from areas outside of England. As a result some figures may not match when comparing the figures by region and by ethnicity, although these discrepancies are relatively small.

5.2. Numbers screened

Laboratory data for 2012/13 identified 685,438 babies as having been screened, of whom 312 were identified as screen positive for a significant condition and 9,368 were identified as carriers.

Comparing these data with birth figures from the ONS provides some data validity and shows a discrepancy of 1.27% between datasets. Differences between the ONS figures and those reported by the screening laboratories could be accounted for by the different periods of time covered (laboratory data covers financial years whereas ONS data covers calendar years), declined screening, and screened babies reported with an unknown region.

Table NB-1. Comparison of ONS birth figures and number of babies screened reported by newborn screening laboratories, 2012/13: England by region

Region	Data from newborn laboratories*	ONS figures†	Discrepancy (%)
East Midlands	49,898	55,645	10.33
East of England	72,421	74,571	2.88
London	131,424	134,186	2.06
North East	28,966	30,291	4.37
North West	87,369	89,211	2.06
South Central	53,352	53,957	1.12
South East Coast	51,682	53,901	4.12
South West	59,938	61,131	1.95
West Midlands	72,559	73,940	1.87
Yorkshire and The Humber	69,613	67,408	-3.27
Unknown	8,216	-	-
England Total	685,438	694,241	1.27

**Data collected from the 13 newborn laboratories in England. This data covers the financial year 2012/13 and excludes cases where screening was declined.*

†Data from ONS (Live Births by Area of Usual Residence 2012, found at http://www.ons.gov.uk/ons/publications/re-reference-tables.html?edition=tcm%3A77-320857). This data covers the 2012 calendar year.

The 'unknown' region category indicates cases where the laboratories provide figures as 'out of region' or 'unknown PCT' and so cannot be attributed to a specific region.

5.3. Newborn screening results

Significant conditions:

Significant conditions comprise FS, FSC, FS-Other and FE variants. Table NB-2 shows the number and rates of babies identified with a significant condition for each year between 2010 and 2013.

Table NB-2. Trends in the number of babies identified with significant conditions, 2010-13: England by region

Region	2010/11			2011/12			2012/13		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
East Midlands	9	50,764	0.18	8	50,901	0.16	8	49,898	0.16
East of England	30	66,048	0.45	18	72,091	0.25	21	72,421	0.29
London	216	131,913	1.64	205	133,245	1.54	195	131,424	1.48
North East	3	28,560	0.11	*	28,122	0.07	*	28,966	0.03
North West	31	88,581	0.35	21	88,405	0.24	25	87,369	0.29
South Central	14	54,258	0.26	18	54,286	0.33	13	53,352	0.24
South East Coast	5	53,274	0.09	8	53,521	0.15	7	51,682	0.14
South West	10	59,399	0.17	4	60,811	0.07	7	59,938	0.12
West Midlands	26	72,404	0.36	21	72,970	0.29	21	72,559	0.29
Yorkshire and the Humber	14	69,382	0.20	10	69,457	0.14	11	69,613	0.16
Unknown	0	13,731	0.00	5	9,469	0.53	3	8,216	0.37
England Total	358	688,314	0.52	320	693,278	0.46	312	685,438	0.46

**Numbers less than 3 have been suppressed*

In 2012/13 there were 312 babies identified with a significant condition, which equates to 0.46 per 1000 babies screened, or approximately one in 2,200 babies screened. Prior to 2011/12 there were approximately 360 babies identified with a significant condition through newborn screening each year.

Across England rates range between 0.03 per 1000 babies screened (approximately one in 29,000 babies screened) in the North East and 1.48 per 1000 babies screened (approximately one in 674) in London. Babies born with a significant condition were identified in all regions, although approximately 63% of these babies were screened in London.

Newborn screening does not specifically test for beta thalassaemia major. However, F-Only cases are likely beta thalassaemia cases and will require follow-up testing. The number of F-Only cases is approximately 20 each year and has ranged between 19 in 2010/11 and 25 in 2012/13.

Figure NB-1 shows the geographical prevalence of babies identified with a significant condition per 1000 babies screened in 2012/13 by region.

Figure NB-1. Babies identified with a significant condition per 1000 babies screened, 2012/13: England by region

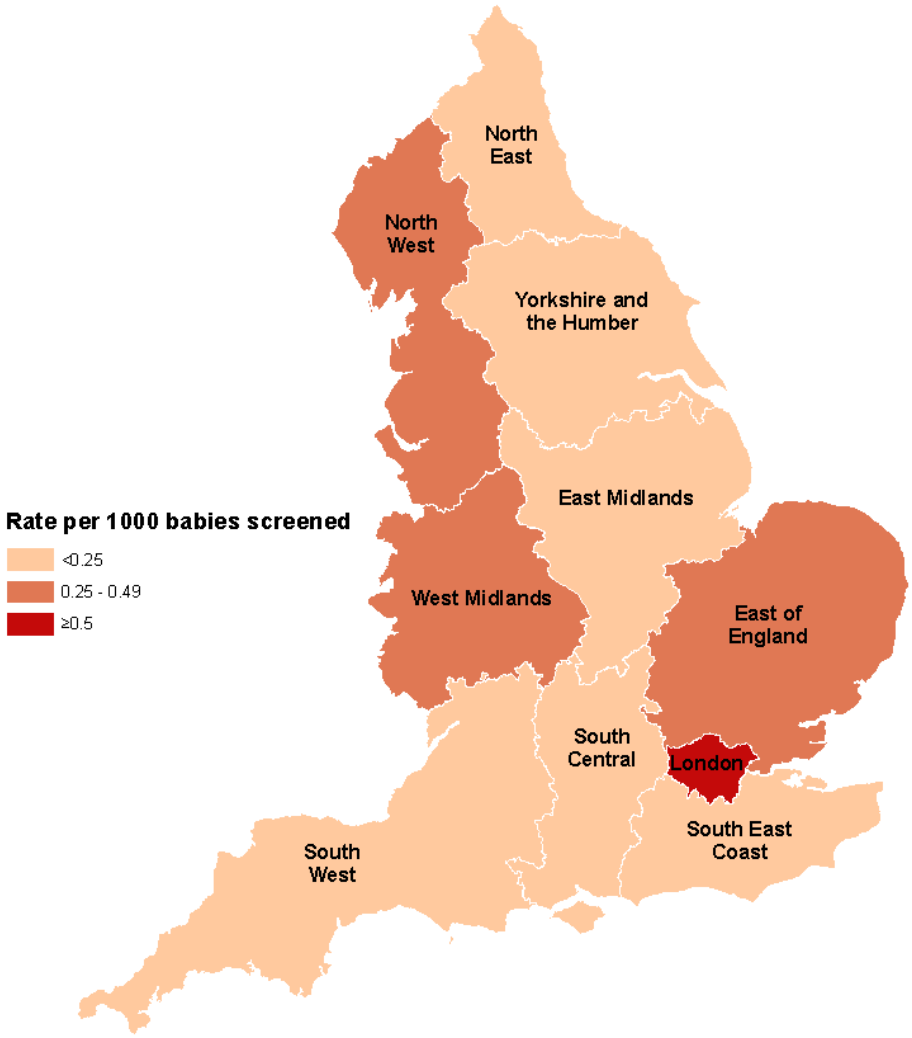
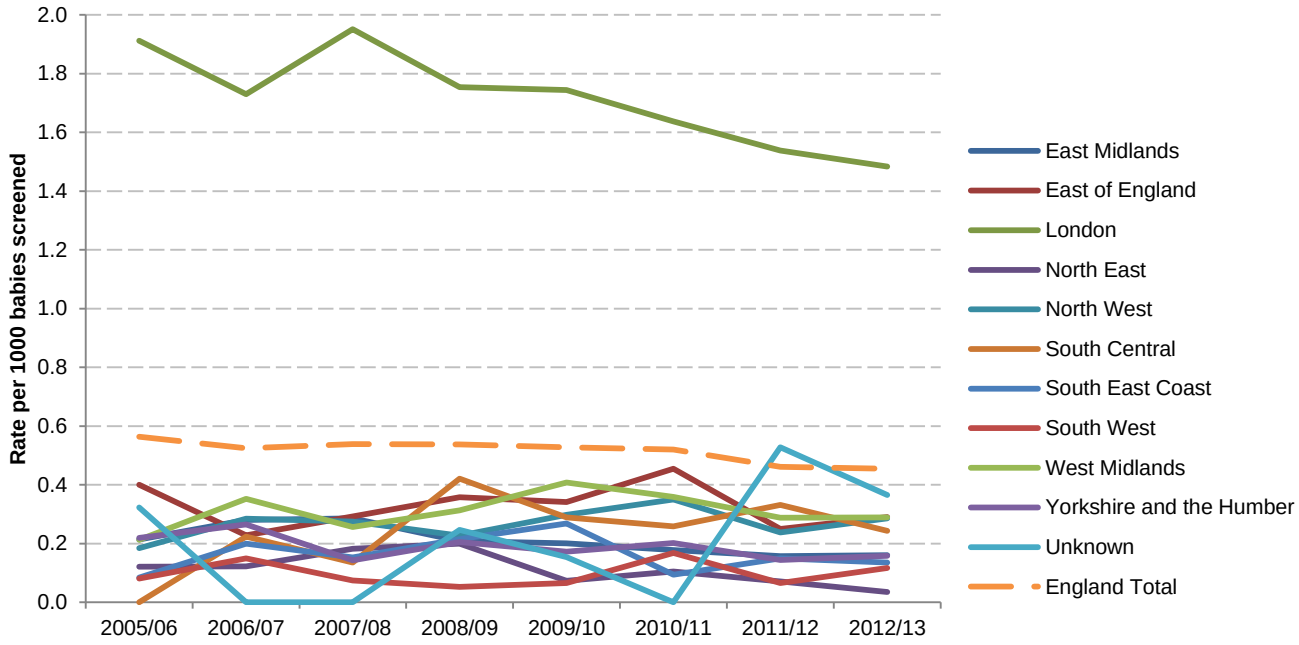


Figure NB-2 shows the trends in rates of babies identified with a significant condition for England by region. Figure NB-3 compares the rates for London with those for the rest of England (including cases where the region is unknown).

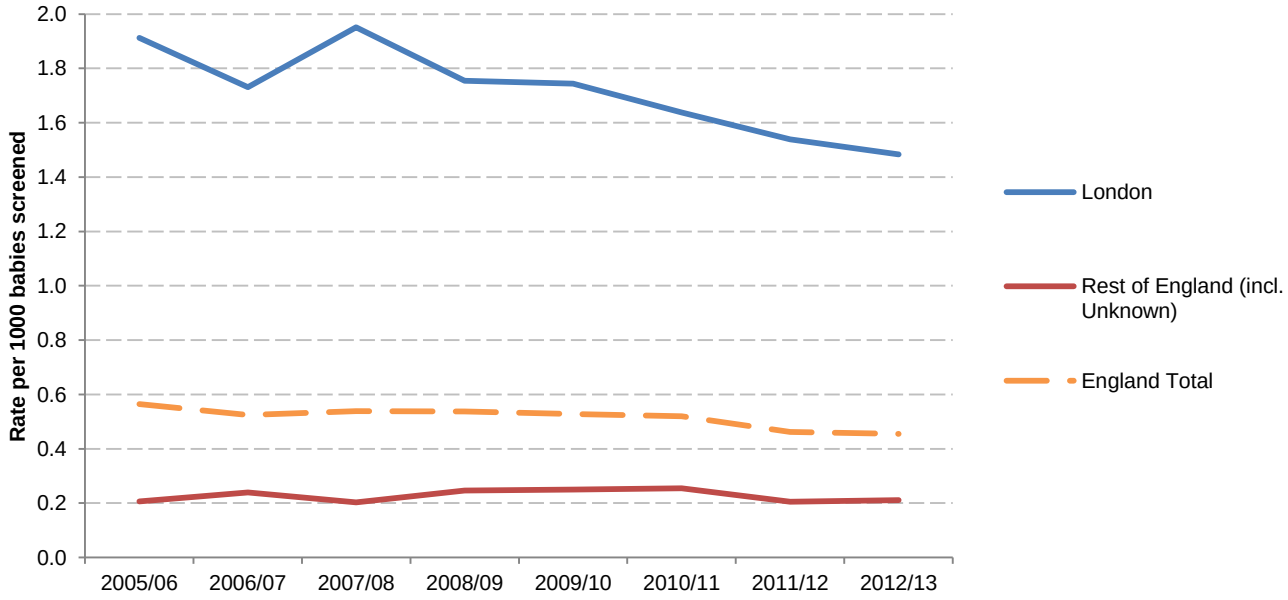
The rates for the whole of England appear steady at approximately 0.5 per 1000 babies screened over the eight-year period with slight variations between years. The rates in London are consistently higher than in other parts of England, but there is an indication of a continued decline in London and a slight increase in other parts of England, which could reflect population movements.

Figure NB-2. Trends in babies identified with a significant condition, 2005-13: 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 significant condition, 2005-13: 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 carrier haemoglobin variants. Table NB-3 shows the number and rates of babies with a carrier result between 2005 and 2013. In 2012/13 there were 9,368 babies identified as carriers, which equates to approximately 14 per 1000 babies screened, or one in 73 babies screened. Across England the rates ranged between one in 186 in the South West and one in 28 in London.

Table NB-3. Trends in the number of babies identified with carrier results, 2005-13: England by region

Region	2010/11			2011/12			2012/13		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
East Midlands	451	50,764	8.88	398	50,901	7.82	444	49,898	8.90
East of England	747	66,048	11.31	728	72,091	10.10	706	72,421	9.75
London	4,896	131,913	37.12	4,778	133,245	35.86	4,679	131,424	35.60
North East	150	28,560	5.25	122	28,122	4.34	157	28,966	5.42
North West	732	88,581	8.26	817	88,405	9.24	665	87,369	7.61
South Central	605	54,258	11.15	542	54,286	9.98	564	53,352	10.57
South East Coast	409	53,274	7.68	404	53,521	7.55	357	51,682	6.91
South West	286	59,399	4.81	351	60,811	5.77	323	59,938	5.39
West Midlands	935	72,404	12.91	939	72,970	12.87	900	72,559	12.40
Yorkshire and The Humber	557	69,382	8.03	529	69,457	7.62	477	69,613	6.85
Unknown	62	13,731	4.52	110	9,469	11.62	96	8,216	11.68
England Total	9,830	688,314	14.28	9,718	693,278	14.02	9,368	685,438	13.67

Figure NB-4 shows the geographical prevalence of babies identified with a carrier result per 1000 babies screened in 2012/13 by region.

Figure NB-4. Babies identified with a carrier result per 1000 babies screened, 2012/13: England by region

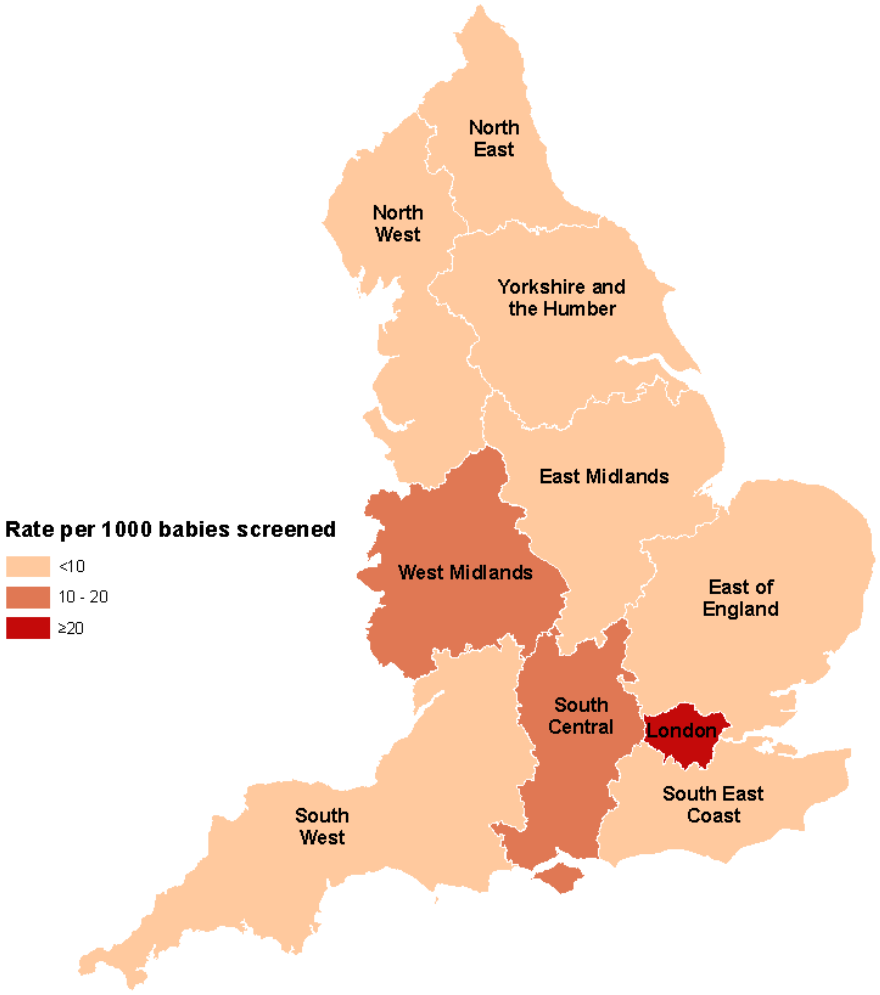
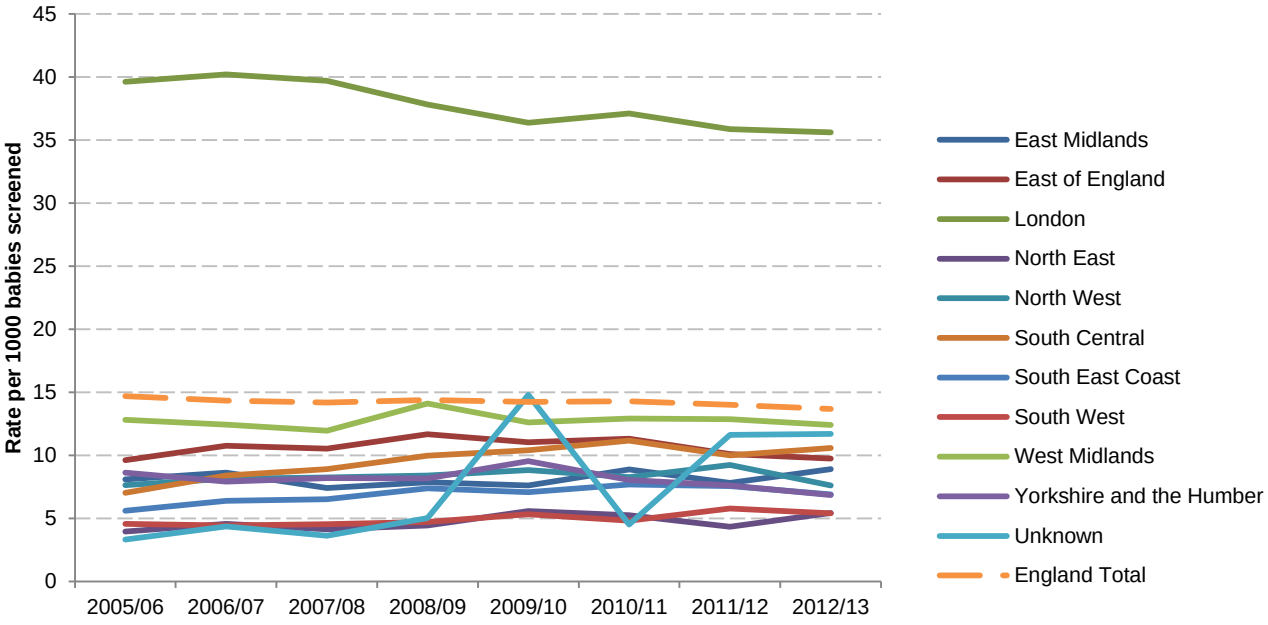


Figure NB-5 shows the trends in rates of babies identified with a carrier result for England by region. Figure NB-6 shows these data for London only with a breakdown for London sectors (pre-2006 SHAs) and Figure NB-7 shows data for England excluding London and cases where the region is unknown. Figure NB-8 compares the rates for London with those for the rest of England (including cases where the region is unknown).

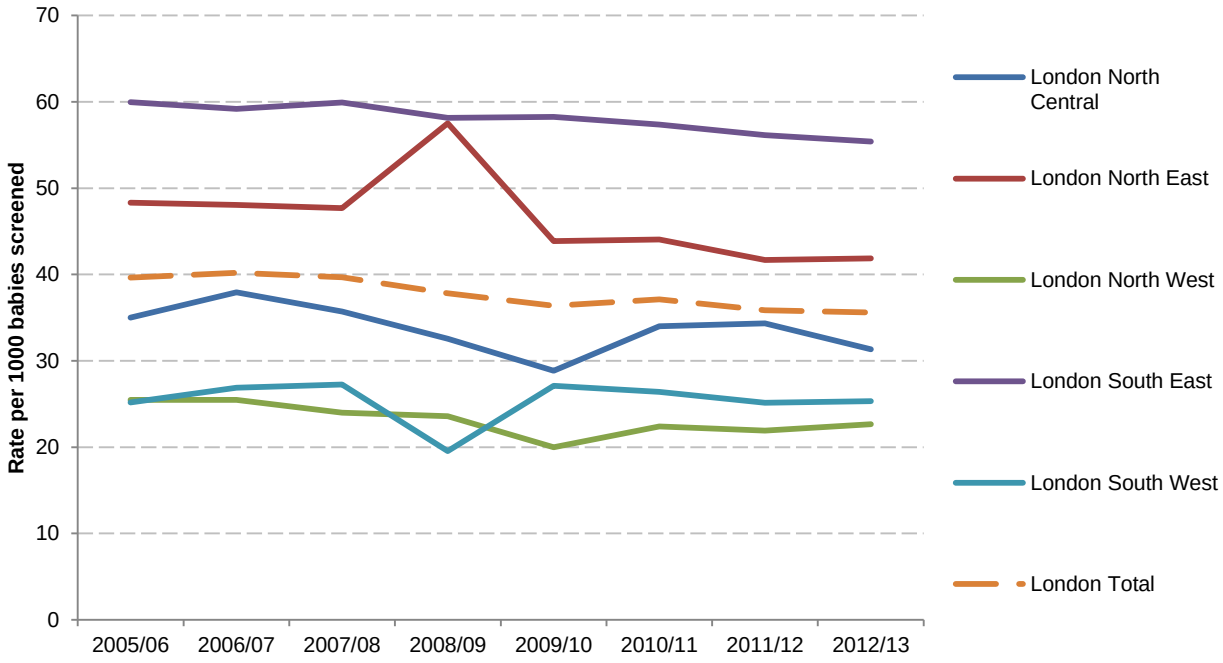
Carrier numbers are higher than those for significant conditions, making the rates more reliable. As a result, carrier rates appear more stable across the eight-year period when compared to rates for babies identified with a significant condition. As with the figures for significant conditions, rates in London are consistently higher than in other parts of England. Carrier rates for the whole of England appear stable, but again there is some indication of a slight decline in London and an increase in other parts of England.

Figure NB-5. Trends in babies identified as carriers, 2012/13: England by region



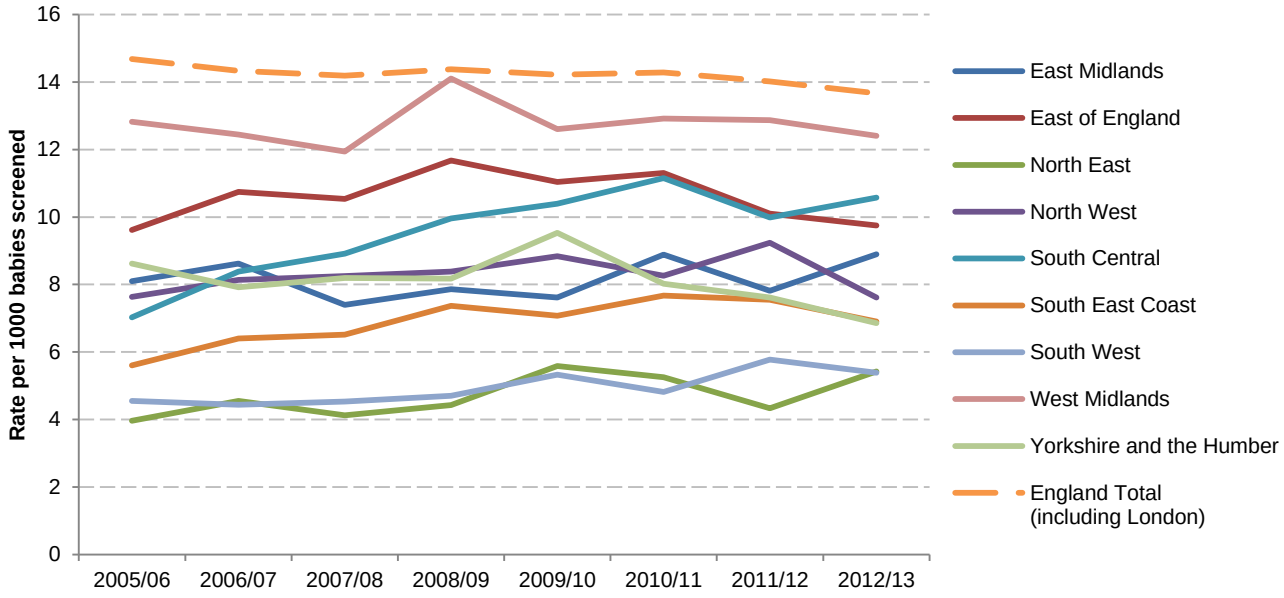
Carrier results comprise FAS, FAC, FAD, FAE and other carriers.
**Bristol data for first half of 2005/06 not included and Oxford and Portsmouth data not included for whole of 2005/06; Oxford data starts from 1st July 2006.*

Figure NB-6. Trends in babies identified as carriers, 2005-13: London sectors (pre-2006 SHAs)



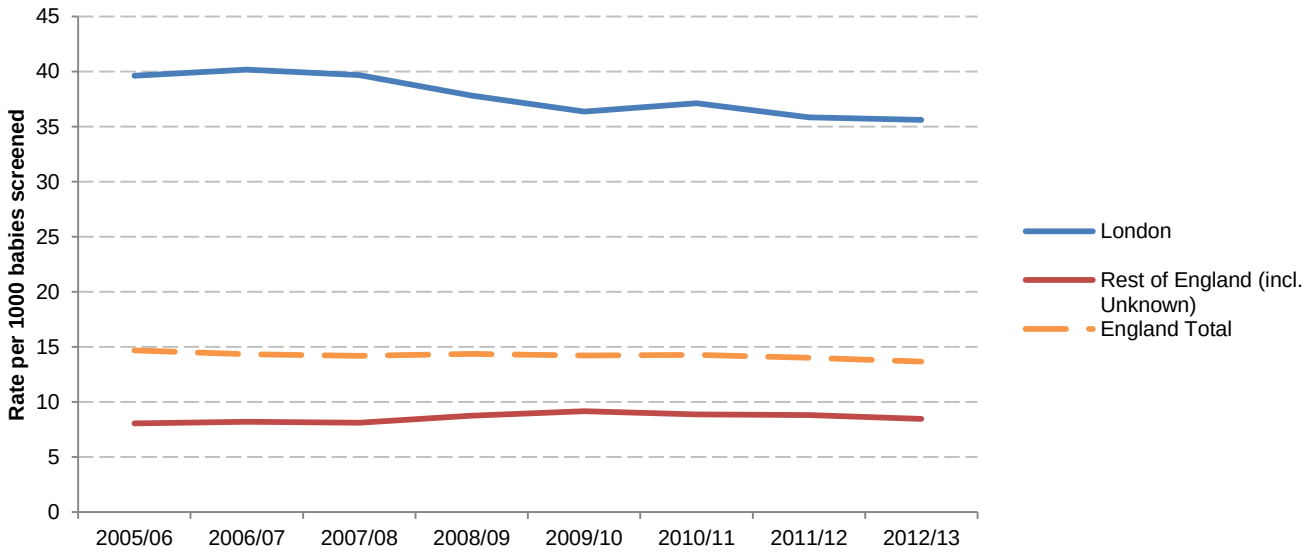
Carrier results comprise FAS, FAC, FAD, FAE and other carriers.

Figure NB-7. Trends in babies identified as carriers, 2005-13: 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. Trends in babies identified as carriers, 2005-13: London and the rest of England



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

Table NB-4 shows a breakdown of newborn screening results by haemoglobinopathy result for 2012/13 including both significant conditions and carrier results.

Table NB-4. Babies screened and newborn screening results, 2012/13: England by region

Region	Significant Conditions					Carriers							
	FS	FSC	FS-Other	FE	F-only	FAS	FAC	FAD	FAE	Other Carrier	Transfused	Declined	Total Screened
East Midlands	6	*	0	0	*	270	62	69	43	0	89	0	49,898
East of England	12	6	*	*	*	448	88	51	77	42	96	90	72,421
London	131	47	7	10	7	3,351	661	197	381	89	368	145	131,424
North East	*	0	0	0	0	88	6	10	24	29	44	28	28,966
North West	17	4	3	*	4	409	65	84	93	14	137	85	87,369
South Central	8	5	0	0	3	319	62	56	78	49	42	13	53,352
South East Coast	5	*	0	0	0	208	31	37	62	19	199	32	51,682
South West	5	*	*	0	0	157	50	38	37	41	34	45	59,938
West Midlands	16	4	*	0	5	534	145	120	100	*	182	79	72,559
Yorkshire and the Humber	6	*	3	0	3	269	50	67	56	35	133	85	69,613
Unknown	3	0	0	0	0	60	19	7	4	6	195	65	8,216
England Total	210	73	17	12	25	6,113	1,239	736	955	325	1,519	667	685,438

**Numbers less than 3 have been suppressed.*

5.4. Results by ethnicity

Newborn screening figures by ethnicity differ slightly from the figures by region (see 5.1 - Data quality and methods of data collection). Table NB-5 shows the number and rates of babies identified with a significant condition in the three years since 2010/11, and Table NB-6 shows similar data for carrier results in the same period.

Babies reported as Black African accounted for approximately 63% of significant conditions detected and approximately 37% of carrier results detected in 2012/13. While sickle cell disease is more common in the Black African, Black Caribbean and Any Other Black background ethnic categories, it is not confined to these groups and in 2012/13 1.6% of babies with significant conditions detected were declared as White British.

Table NB-5. Numbers of babies identified with significant conditions, 2010-13: England by ethnicity

Ethnic Category	2010/11			2011/12			2012/13		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
A - White British	4	443,856	0.01	3	443,448	0.01	5	434,514	0.01
B - White Irish	0	1,477	0.00	0	1,671	0.00	0	1,723	0.00
C - Any other White background	0	45,844	0.00	0	47,675	0.00	0	49,386	0.00
D - White and Black Caribbean	5	7,520	0.66	9	7,581	1.19	8	7,803	1.03
E - White and Black African	*	4,414	0.45	6	4,407	1.36	3	4,348	0.69
F - White and Asian	0	7,859	0.00	0	8,056	0.00	0	10,495	0.00
G - Any other mixed background	*	11,089	0.18	4	12,340	0.32	3	10,735	0.28
H - Indian	3	19,392	0.15	4	21,736	0.18	4	21,560	0.19
J - Pakistani	*	27,259	0.04	*	27,262	0.04	0	27,855	0.00
K - Bangladeshi	14	9,153	1.53	7	9,096	0.77	9	9,856	0.91
L - Any other Asian background	4	9,970	0.40	3	9,869	0.30	*	8,078	0.25
M - Black Caribbean	40	7,234	5.53	38	6,668	5.70	30	7,402	4.05
N - Black African	233	25,353	9.19	195	24,294	8.03	196	22,244	8.81
P - Any other Black background	15	3,173	4.73	20	3,330	6.01	17	4,838	3.51
R - Chinese	0	3,757	0.00	0	3,687	0.00	0	3,951	0.00
S - Any other ethnic category	10	16,770	0.60	5	16,828	0.30	10	18,615	0.54
Z - Not stated	26	46,209	0.56	25	46,071	0.54	24	43,490	0.55
England Total	359	690,329	0.52	320	694,019	0.46	311	686,893	0.45

**Numbers less than 3 have been suppressed.*

Table NB-6. Numbers of babies identified with carrier results, 2010-13: England by ethnicity

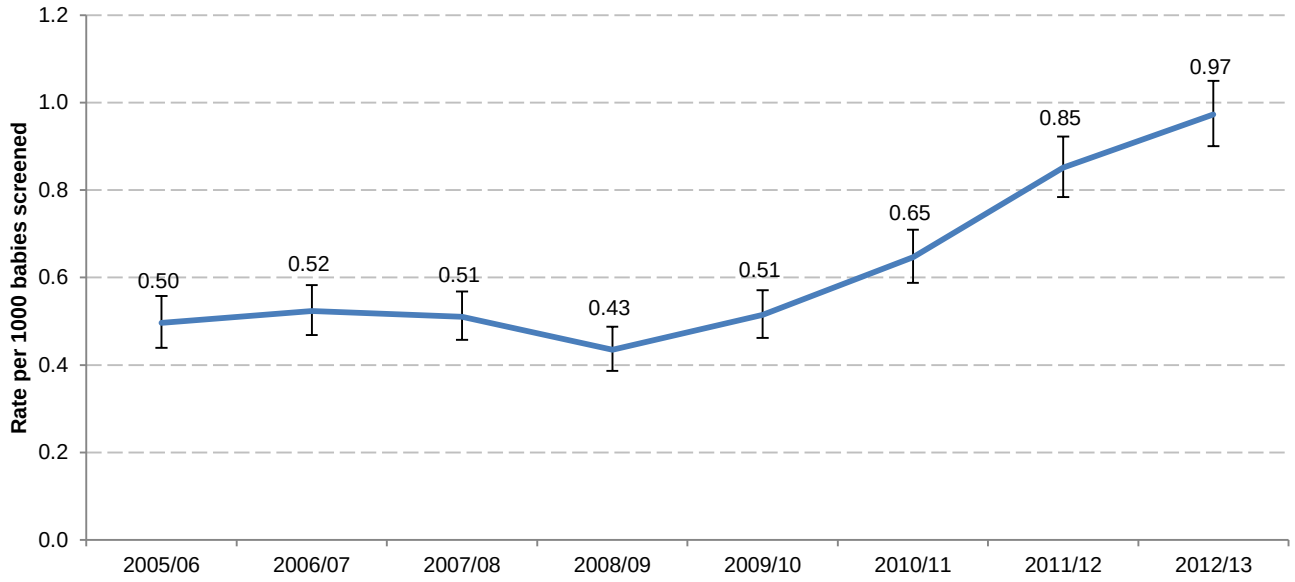
Ethnic Category	2010/11			2011/12			2012/13		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
A - White British	980	443,856	2.21	957	443,448	2.16	762	434,514	1.75
B - White Irish	*	1,477	1.35	5	1,671	2.99	3	1,723	1.74
C - Any other White background	150	45,844	3.27	171	47,675	3.59	139	49,386	2.81
D - White and Black Caribbean	495	7,520	65.82	464	7,581	61.21	457	7,803	58.57
E - White and Black African	339	4,414	76.80	375	4,407	85.09	359	4,348	82.57
F - White and Asian	177	7,859	22.52	168	8,056	20.85	146	10,495	13.91
G - Any other mixed background	396	11,089	35.71	327	12,340	26.50	352	10,735	32.79
H - Indian	286	19,392	14.75	328	21,736	15.09	314	21,560	14.56
J - Pakistani	338	27,259	12.40	351	27,262	12.88	293	27,855	10.52
K - Bangladeshi	437	9,153	47.74	416	9,096	45.73	420	9,856	42.61
L - Any other Asian background	174	9,970	17.45	182	9,869	18.44	158	8,078	19.56
M - Black Caribbean	897	7,234	124.00	850	6,668	127.47	843	7,402	113.89
N - Black African	3,629	25,353	143.14	3,467	24,294	142.71	3,483	22,244	156.58
P - Any other Black background	349	3,173	109.99	396	3,330	118.92	412	4,838	85.16
R - Chinese	22	3,757	5.86	27	3,687	7.32	29	3,951	7.34
S - Any other ethnic category	406	16,770	24.21	401	16,828	23.83	495	18,615	26.59
Z - Not stated	773	46,209	16.73	838	46,071	18.19	746	43,490	17.15
England Total	9,850	690,329	14.27	9,723	694,019	14.01	9,411	686,893	13.70

**Numbers less than 3 have been suppressed.*

5.5. Declined screening tests

In 2012/13 there were a total of 667 declined tests across England, or 0.97 per 1000. Figure NB-9 shows the national rates for each year between 2005/06 and 2012/13, and indicates an increase in the number of cases where screening was declined since 2008/09.

Figure NB-9. Declined screening for sickle cell disease, 2005-13: 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.

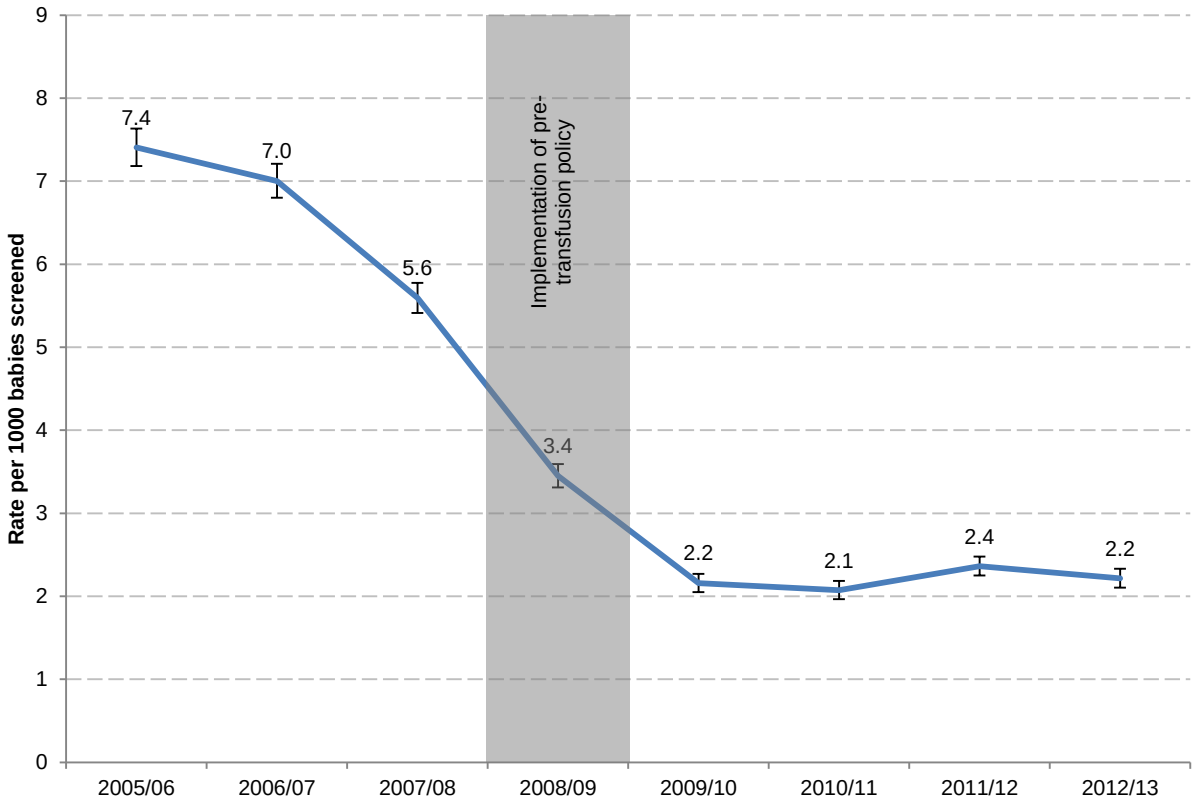
Table NB-7 shows the number and rates of post-transfusion samples between 2010 and 2013 by region. In 2012/13 there were 1,519 post-transfusion samples received, or 2.2 per 1000 babies screened. The rates by region range between 0.57 per 1000 in the South West and 3.85 per 1000 in the South East Coast region. There were 195 post-transfusion samples where the region is unknown.

Figure NB-10 shows the national rates for each year since 2005. Over this period there has been a steady decrease in the rate of post-transfusion samples for newborn screening which has been the result of much hard work. There was a small increase in this rate in 2011/12, but in 2012/13 the rate decreased to 2.2 per 1000 babies screened.

Table NB-7. Post-transfusion samples, number and rate per 1000 babies screened, 2010-13: England by region

Region	2010/11			2011/12			2012/13		
	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000	n	Total Screened	Rate/ 1000
East Midlands	72	50,764	1.42	94	50,901	1.85	89	49,898	1.78
East of England	101	66,048	1.53	83	72,091	1.15	96	72,421	1.33
London	305	131,913	2.31	344	133,245	2.58	368	131,424	2.80
North East	57	28,560	2.00	35	28,122	1.24	44	28,966	1.52
North West	192	88,581	2.17	224	88,405	2.53	137	87,369	1.57
South Central	45	54,258	0.83	34	54,286	0.63	42	53,352	0.79
South East Coast	111	53,274	2.08	195	53,521	3.64	199	51,682	3.85
South West	81	59,399	1.36	42	60,811	0.69	34	59,938	0.57
West Midlands	216	72,404	2.98	213	72,970	2.92	182	72,559	2.51
Yorkshire and the Humber	47	69,382	0.68	145	69,457	2.09	133	69,613	1.91
Unknown	200	13,731	14.57	229	9,469	24.18	195	8,216	23.73
England Total	1,427	688,314	2.07	1,638	693,278	2.36	1,519	685,438	2.22

Figure NB-10. Post-transfusion samples, rate per 1000 babies screened, 2005-13: England



As a failsafe to ensure that transfused babies are offered appropriate screening and to mitigate the on-going risks of a missed baby, the Sickle Cell and Thalassaemia Screening Programme worked with the Newborn Blood Spot Screening Programme to introduce a pilot process of DNA testing for babies which was to run between November 2009 and March 2014, and some positive cases have been identified through this failsafe measure. Between November 2009 and March 2013 there were 5,030 samples tested at King's College Hospital and Sheffield Children's Hospital using DNA techniques, of which five were found to be screen positive for sickle cell disease (approximately one per 1000 babies screened using DNA techniques). Since the programme introduced a policy for pre-transfusion samples and DNA testing, there has been a reduction in the proportion of post-transfusion samples being received by the laboratories, and the rates have now levelled out at approximately 2.2 per 1000 babies screened. Future funding for DNA testing for transfused babies will be provided by NHS England.

For more information on DNA testing for transfused babies, see sct.screening.nhs.uk/transfusedbabies.

Table NB-8. Numbers detected through DNA testing for transfused babies, 2009-13

Collection year	2009/10		2010/11				2011/12				2012/13				Total for period
Quarter (financial)	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	
Total Specimens received per Quarter	143	350	371	457	455	391	358	351	390	421	333	366	315	329	5,030
Number of Negative results (HbS not detected)	139	344	366	454	446	384	350	341	387	419	330	357	310	322	4,949
Number of Positive Heterozygotes	4	6	5	3	9	7	8	9	3	*	3	7	4	7	76
Number of Positive Homozygotes	0	0	0	0	0	0	0	*	0	*	0	*	*	0	5
Number of Results pending	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Number rejected due to lack of identifiers	*	0	0	0	0	0	0	0	0	0	0	0	0	0	*

**Numbers less than 3 have been suppressed.*

5.7. Timeliness of reporting results

Laboratories have been asked to provide data on timeliness of reporting results for newborn screening for sickle cell disease. Newborn Blood Spot Screening Programme standard 4 (timely sample collection) is for the sample to be taken on day 5 and in exceptional circumstances between day five and day eight (day of birth is day zero). Standard 5 (timely receipt of a sample in the newborn screening laboratory) is for all samples to arrive in the laboratory within four days of sample collection. Sickle Cell and Thalassaemia 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 the timeliness figures for each laboratory. This includes all significant conditions and not just sickle cell disease results, and includes F-only cases. Turn-around time is calculated as the number of days between the age at sample and the age at which the positive result was reported. Turn-around times for Portsmouth are not shown because no data were provided for the age at which the positive result was reported.

Table NB-9. Timeliness of reporting affected newborn results, 2012/13: England by laboratory

	No. of screen positives	Sample ≤8 days		Sample received by lab within 4 days		Positive result by 28 days		Turn-around time		
Laboratory	n	n	%	n	%	n	%	Min	Max	Average
Bristol	3	3	100	3	100	3	100	4	6	5
Cambridge	*	*	100	*	100	*	100	11	11	11
GOS & CMH	123	122	99	7	6	123	100	6	17	10
Leeds	11	10	91	10	91	11	100	4	10	7
Liverpool	5	3	60	5	100	5	100	3	8	5
Manchester	24	24	100	20	83	23	96	10	28	16
New castle	*	0	0	*	100	*	100	4	4	4
Oxford	14	13	93	12	86	14	100	8	23	14
Portsmouth	5	5	100	4	80	†	-	-	-	-
Sheffield	12	11	92	9	75	10	83	13	45	20
South East Thames	62	60	97	50	81	59	95	3	34	9
South West Thames	39	38	97	22	56	38	97	11	23	15
West Midlands	26	26	100	20	77	22	85	7	35	16
England Total	326	316	97	164	50	310	95	3	45	12

†*No data provided for this field*

**Numbers less than 3 have been suppressed*

6. Key Performance Indicators

Background to Key Performance Indicators

Key Performance Indicators (KPIs) give a high level overview of the quality of screening programmes at key points on the screening pathway. They contribute to the quality assurance of screening programmes, but are not in themselves sufficient to quality assure or performance manage screening services.

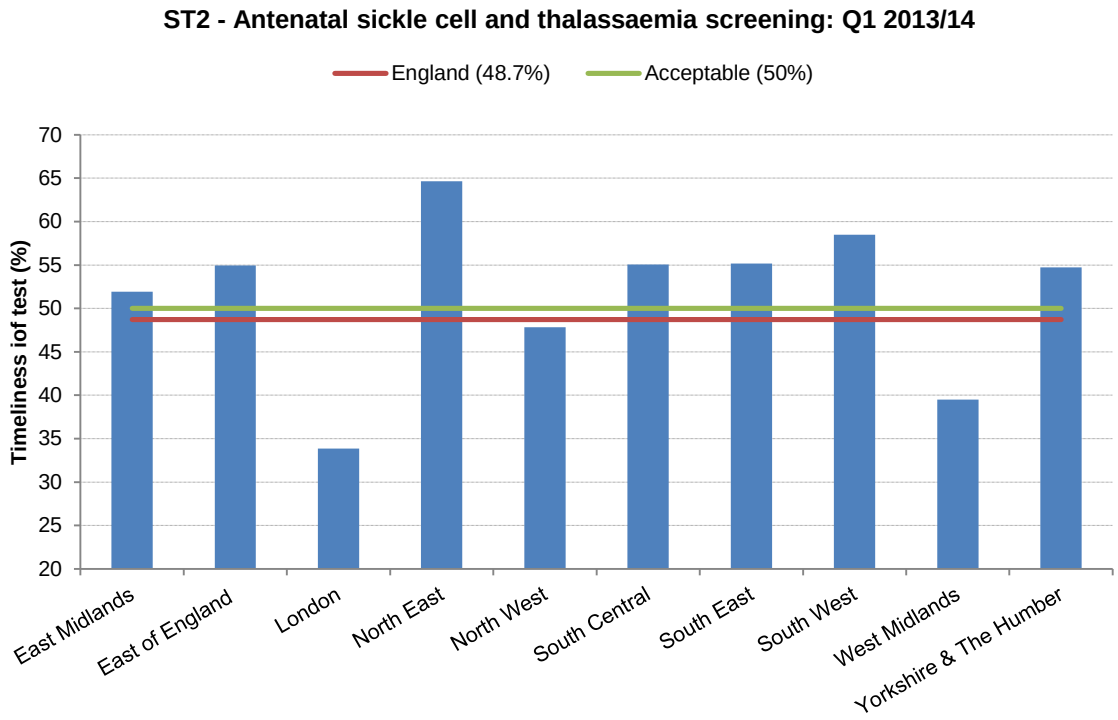
KPI data covering quarter 1 of 2013/14 (1 April to 30 June 2013) have been published and can be accessed at http://www.screening.nhs.uk/kpi/reports/2013-14. It is important to note that these data cover only one quarter and are in a different reporting year than the other data presented in this report.

The Sickle Cell and Thalassaemia Screening Programme has three antenatal KPIs and shares one newborn KPI with the Newborn Blood Spot Screening Programme:

KPI Code	KPI Description
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)
ST2	The proportion of women having antenatal sickle cell and thalassaemia screening for whom a conclusive screening result is available by 10 weeks' gestation
ST3	The proportion of antenatal sickle cell and thalassaemia samples submitted to the laboratory which are supported by a completed Family Origin Questionnaire (FOQ)
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

Key findings

- In this quarter coverage for sickle cell and thalassaemia (ST1) was high at 98.2%.
- The acceptable level for ST2 is 50%. The England average was 48.7%, with London (33.9%), West Midlands (39.5%), and North West (47.8%) reporting below 50% (see chart below).
- The acceptable level for ST3 is 90%. Across England 128 Trusts out of 146 achieved this level and 14 did not, of which nine were in London. One Trust submitted data higher than 100%, three did not submit data and two Trusts withdrew data.
- The report focus for NB3 is maternity service, but it is reported by old PCT boundaries and 24 organisations did not submit data or had data withdrawn. Of those that submitted data, performance was high with all organisations achieving greater than the acceptable level of 95% and 33 achieving 100%



The proportion of women having antenatal sickle cell and thalassaemia screening for whom a conclusive screening result is available by 10 weeks' gestation.

7. Appendices

Appendix One: UK Newborn Blood Spot data report 2012/13: Executive Summary

During the fiscal year of 2012-13 just over 810,000 babies had blood spot sample taken and of these, 1,442 babies had a positive screening result.

A newborn screening study took place in 2012-13 in five areas (Yorkshire, the East Midlands, the West Midlands, the North West of England and some parts of London and the South East). Five additional rare disorders were included in the study: maple syrup urine disease, homocystinuria (pyridoxine unresponsive), isovaleric acidaemia, glutaric aciduria type 1 and long-chain hydroxyl acyl-CoA dehydrogenase deficiency. The study began with babies tested on or after 16th July 2012 and ended with those tested on or before 19th July 2013, and was subsequently extended to 31st September 2014. On the basis of this study, the UK National Screening Committee is consulting on its recommendation to expand the current newborn blood spot screening programme to include:

- homocystinuria (HCU)
- maple syrup urine disease (MSUD)
- glutaric aciduria type 1 (GA1)

The NHS Newborn Blood Spot Screening Programme has continued to work with the Sickle Cell and Thalassaemia Screening Programme and awarded Northgate Information Solutions Ltd the contract to implement the Newborn Blood Spot Failsafe Solution. This will identify babies that may have missed newborn blood spot screening and possibly mothers and babies postnatal care. The project has won the Health Service Journal award for the 'Efficiencies in Clinical Support Services' category.

Overall newborn screening performance continues to improve in general for use of the NHS number, bar coded label, timely collection and despatch of the blood spot sample.

Although the number of avoidable repeat requests has decreased in some areas, this remains an area for concern and the blood spot programme is working to address the high number of samples repeated for avoidable reasons, which continues to cause unnecessary distress to families and waste NHS resources.

Referral times and entry into care remain good for PKU, MCADD, CHT and CF babies screened positive on the first sample with 94% of babies within this meeting the national standards. Timeliness is less good for those screened on second sample and the programme aims to pinpoint where these delays are happening.

For the first time, screening data has been mapped to the CF, MCADD and PKU protocols for the UK. Unfortunately due to the varied TSH cut off levels used across laboratories, data was not able to be mapped to the CHT protocol.

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														