Ten years of newborn screening for sickle cell disease in England

Authors C. Coppinger, M. Charlton, L. Oni

Public Health England leads the NHS Screening Programmes

INTRODUCTION

2016 marks the 10th anniversary of the full roll-out of newborn screening for sickle cell disease (SCD) in England. The programme commenced in 2002.

We've screened more than 7 million newborn babies for SCD between April 2006 and March 2016.

Babies are screened for SCD as part of the newborn blood spot screening test. Of the 7 million screened, just over 3,600 (approximately 1 in 2,000) have SCD screen positive results.

SCD is the name for a group of genetically inherited conditions that affect the quality of haemoglobin and the capacity to carry oxygen around the body. The most serious form of the disease is sickle cell anaemia.

Screening for sickle cell disease in newborn babies identifies infants affected by the condition, with the aim of minimising childhood morbidity. Children with the condition are at risk of severe illness or even death from infections.
Early identification of the condition allows for preventive management and treatment to begin promptly.

Despite detection of thalassaemia not being part of the programme, we expect beta thalassaemia major to be detected and reported. In the past 10 years, nearly 270 cases of beta thalassaemia major have been identified (approximately 1 in 27,000).

SCREENING RESULTS

Results by region 2005-2016

Region	Significant Conditions					Non-significant conditions			Carriers					Total Screened		
	FS	FSC	FS-Other	FE	F-only	FC	FD	Other	FAS	FAC	FAD	FAE	Other	Transfused	Declined	Normal+ Abnormal
East Midlands	91	30	3	6	7	11	3	7	2,739	612	554	373	219	1,334	215	549,180
East of England	152	63	14	10	25	11	18	16	4,612	1,048	543	835	498	1,632	918	741,015
London	1,528	571	56	85	65	166	51	81	36,024	7,770	2,110	4,083	1,275	6,031	1,271	1,411,487
North East	28	2	0	4	1	1	4	2	703	90	143	271	271	547	166	307,612
North West	155	29	41	18	42	8	18	110	4,585	803	860	998	708	2,752	705	948,965
South Central	87	52	7	5	13	15	12	19	2,883	669	551	594	426	968	380	519,445
South East Coast	62	18	4	6	6	6	2	9	2,320	441	376	593	339	2,688	326	570,118
South West	33	17	5	4	0	8	1	4	1,496	407	408	406	212	1,086	585	612,562
West Midlands	161	43	7	21	59	21	18	3	5,886	1,482	1,373	1,130	28	3,401	758	774,512
Yorkshire and the Humber	74	27	31	5	47	13	37	16	2,913	488	804	676	773	1,912	581	725,798
Unknown region	20	8	0	0	3	1	2	21	649	159	91	92	76	2,882	408	120,744
England Total	2,391	860	168	164	268	261	166	288	64,810	13,969	7,813	10,051	4,825	25,233	6,313	7,281,438

Data is collected annually from the 13 newborn screening laboratories in England via spreadsheet-based data return templates, and in the future will also be collected from the laboratories in Scotland, Wales, and Northern Ireland.

Rates by region 2005-2016

Region	Significant Conditions			Carriers			No. of babies Screened
	n	Rate/ 1000	1 in x	n	Rate/ 1000	1 in x	
East Midlands	130	0.24	4,224	4,497	8.19	122	549,180
East of England	239	0.32	3,100	7,536	10.17	98	741,015
London	2240	1.59	630	51,262	36.32	28	1,411,487
North East	34	0.11	9,047	1,478	4.80	208	307,612
North West	243	0.26	3,905	7,954	8.38	119	948,965
South Central	151	0.29	3,440	5,123	9.86	101	519,445
South East Coast	90	0.16	6,335	4,069	7.14	140	570,118
South West	59	0.10	10,382	2,929	4.78	209	612,562
West Midlands	232	0.30	3,338	9,899	12.78	78	774,512
Yorkshire and the Humber	137	0.19	5,298	5,654	7.79	128	725,798
Unknown region	28	0.23	4,312	1,067	8.84	113	120,744
England Total	3583	0.49	2,032	101,468	13.94	72	7,281,438

Current data collection templates are available at https://www.gov.uk/government/collections/sickle-cell-and-thalassaemia-screening-data-collection .

We would like to thank the newborn screening laboratories for all their efforts in providing screening data over the last ten years. We would also like to thank the Specialist Nurses, Midwives, Centres and those involved in antenatal and newborn screening in England.

SCREENING RESULTS BY ETHNIC CATEGORY

Rates by ethnic category 2005-2016

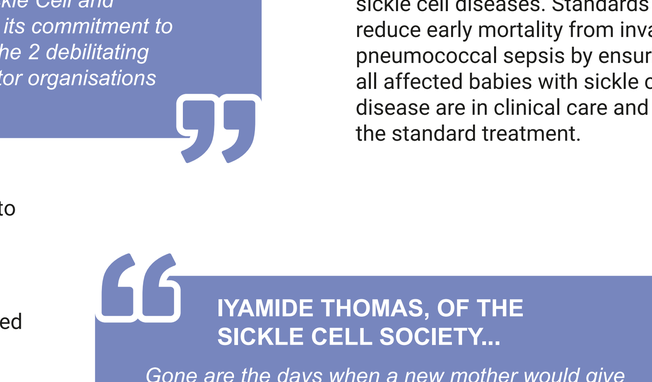
Ethnic Category	Significant Conditions			Carriers			No. of babies Screened
	n	Rate/ 1000	1 in x	n	Rate/ 1000	1 in x	
A - White British	37	0.01	118,117	7,814	1.79	559	4,370,333
B - White Irish	2	0.12	8,011	64	3.99	250	16,021
C - Any other White background	4	0.01	120,055	1,473	3.07	326	480,219
D - White and Black Caribbean	67	0.86	1,169	5,011	63.98	16	78,327
E - White and Black African	51	1.12	897	3,870	84.64	12	45,725
F - White and Asian	4	0.05	20,389	1,549	18.99	53	81,557
G - Any other mixed background	59	0.47	2,128	3,878	30.89	32	125,548
H - Indian	30	0.15	6,611	2,705	13.64	73	198,323
J - Pakistani	13	0.05	20,140	3,114	11.89	84	261,823
K - Bangladeshi	95	0.98	1,019	4,584	47.35	21	96,811
L - Any other Asian background	30	0.30	3,356	1,610	15.99	63	100,686
M - Black Caribbean	377	5.45	183	8,477	122.56	8	69,165
N - Black African	2,167	8.81	114	35,828	145.59	7	246,090
P - Any other Black background	157	5.09	196	3,246	105.30	9	30,825
R - Chinese	0	0.00	-	209	5.84	171	35,771
S - Any other ethnic category	75	0.46	2,170	3,807	23.39	43	162,760
Z - Not stated	299	0.56	1,793	8,955	16.70	60	536,227
England Total	3,467	0.50	2,001	96,194	13.87	72	6,936,211

Results by ethnic category 2005-2016

Ethnic Category	Significant Conditions					Non-significant conditions			Carriers					Total screened		
	FS	FSC	FS-Other	FE	F-Only	FC	FD	Other	FAS	FAC	FAD	FAE	Other	Transfused	Declined	Normal+ abnormal
A - White British	24	4	7	2	2	3	3	48	2,698	820	1,793	537	1,966	9,637	1,665	4,370,333
B - White Irish	1	1	0	0	0	0	0	2	38	6	9	4	7	51	34	16,021
C - Any other White background	3	0	0	1	5	1	5	9	760	166	202	112	233	834	760	480,219
D - White and Black Caribbean	42	13	12	0	0	10	0	4	3,606	1,257	31	43	74	222	71	78,327
E - White and Black African	34	13	4	0	1	3	0	1	3,222	561	20	15	52	125	46	45,725
F - White and Asian	2	1	0	1	1	1	0	7	160	31	213	1,064	81	155	101	81,557
G - Any other mixed background	40	12	7	0	2	4	0	16	2,466	631	154	525	102	356	144	125,548
H - Indian	20	2	2	6	18	0	29	10	741	36	1,281	359	288	501	142	198,323
J - Pakistani	4	0	2	7	164	0	85	13	175	16	2,077	489	357	627	144	261,823
K - Bangladeshi	0	0	0	95	10	0	2	15	38	30	253	4,135	128	264	36	96,811
L - Any other Asian background	11	0	0	19	22	2	6	24	379	34	324	717	156	287	72	100,686
M - Black Caribbean	230	129	16	2	1	31	0	13	6,157	2,186	15	24	95	390	166	69,165
N - Black African	1,573	515	79	0	8	157	0	25	30,256	5,111	37	17	407	1,149	211	246,090
P - Any other Black background	110	41	6	0	1	12	1	4	2,611	561	15	15	44	146	53	30,825
R - Chinese	0	0	0	0	3	0	0	42	18	5	2	110	74	87	15	35,771
S - Any other ethnic category	41	14	10	10	7	7	8	24	2,355	486	216	558	192	446	182	162,760
Z - Not stated	171	93	18	17	21	22	27	29	5,662	1,234	726	850	483	4,697	2,138	536,227
England total	2,306	838	163	160	266	253	166	286	61,342	13,171	7,368	9,574	4,739	19,974	5,980	6,936,211

SCD is most common in people of African, Caribbean, Middle Eastern, Mediterranean and Asian ancestry BUT can affect anyone from any population.

Percentage of all affected babies



SERVICE USERS' FEEDBACK

ANTENATAL WOMAN

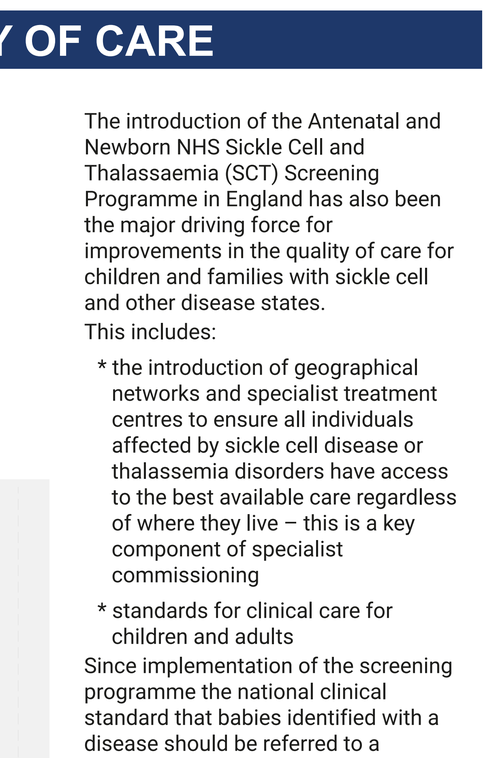
It is important to do the antenatal test so you know it prepares you, you know...
We had the baby tested (prenatal diagnosis), if I had not had that and had to wait until the baby was born I would have been a nervous wreck mentally and emotionally right through my pregnancy, I could not (have) coped with that.

MOTHER OF A NEWBORN

If we did not test we would not know and may miss something when she is sick…
by testing you know what to expect, you are better prepared. If you don't know well… how will you know your child needs special care? I am glad I knew so I can care for her properly?

MOTHER OF A CHILD WITH SCD

I think people need to know we must not be ignorant, it helps you prepare for the child, you can prevent complications if you know, if you don't, if child is sick, you will not know how to look after him properly, otherwise when your child is sick you get a shock to find he has sickle cell disease later, it's dangerous, really dangerous.



IMPROVING AWARENESS

Research in 2004 showed awareness of SCD and thalassaemia was low. The national screening programme established relationships with patient organisations including the Sickle Cell Society (SCS) and the UK Thalassaemia Society (UKTS). These relationships allowed the programme to understand the user perspective and raise awareness through successful outreach work in communities most affected by the conditions.

IYAMIDE THOMAS, OF THE SICKLE CELL SOCIETY...

By investing in a public outreach programme for sickle cell and thalassaemia, the NHS Sickle Cell and Thalassaemia Programme showed its commitment to the communities most affected by the 2 debilitating conditions and to the voluntary sector organisations that represented them.

The NHS newborn sickle cell and thalassaemia screening (SCT) programme aims to achieve the lowest possible childhood death rate and to minimise childhood morbidity from sickle cell diseases. Standards aim to reduce early mortality from invasive pneumococcal sepsis by ensuring that all affected babies with sickle cell disease are in clinical care and receiving the standard treatment.

In September 2010 we started a project to evaluate the outcomes of the linked antenatal and newborn screening programme; we collect data to assess

* the health of babies or children affected with sickle cell disorders or thalassaemia
* timeliness of entry into care and start of treatment for affected babies or children
* antenatal screening history

The programme currently has approval to collect named data without consent and the SCS and UKTS play an important role in the governance of the project and working with parents to share the lessons learned from the project, for example the importance of taking antibiotics.

IYAMIDE THOMAS, OF THE SICKLE CELL SOCIETY...

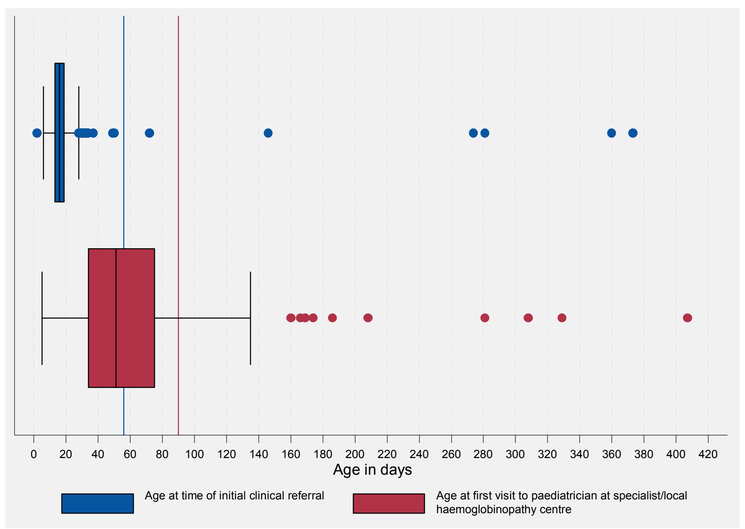
Gone are the days when a new mother would give birth to a child with sickle cell and ask 'what's that'?

IMPROVING QUALITY OF CARE

KATE RYAN, CONSULTANT HAEMATOLOGIST...

There is always room for improvement but I think we can confidently say that the outlook for a child born in 2016 with sickle cell disease is so much better than for a child born before the introduction of newborn screening.

Age at referral/first visit (2014/15 - 2015/16)



The introduction of the Antenatal and Newborn NHS Sickle Cell and Thalassaemia (SCT) Screening Programme in England has also been the major driving force for improvements in the quality of care for children and families with sickle cell and other disease states.

This includes:

* the introduction of geographical networks and specialist treatment centres to ensure all individuals affected by sickle cell disease or thalassemia disorders have access to the best available care regardless of where they live – this is a key component of specialist commissioning
* standards for clinical care for children and adults

Since implementation of the screening programme the national clinical standard that babies identified with a disease should be referred to a designated local healthcare professional by 8 weeks of age is being met in 99% of cases and 85% are seen in a specialist treatment centre by 3 months.

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