7.28 Sickle Cell Disease

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What is sickle cell disease?

Sickle cell disease is the fastest growing genetically inherited condition in the UK affecting over 1 in 2,000 births and over 12,000 adults are living with the condition in the UK.

In Barking and Dagenham the number of residents with Sickle Cell Disease is increasing. Sickle cell disease and trait have been becoming progressively more visible amongst the populations of outer north east London (ONEL)[1], most notably in Barking and Dagenham. This reflects the growing diversity of the population and the fact that sickle cell disease is now the most common serious genetic disorder in England affecting over 1 in 2,000 live births[2].

Sickle cell disease describes a group of conditions caused by the 'sickle' mutation of the haemoglobulin molecule (Hb). Hb is responsible for transporting oxygen in the red blood cells of humans, and the sickle mutation reduces the ability of the red blood cell to carry oxygen.

Clinically significant sickle cell disease is a result of individuals inheriting the sickle gene from both parents or a sickle gene and another specific variant. Sickle cell carrier status, also sometimes called sickle cell trait, occurs when individuals only inherit one sickle gene, carriers rarely have clinical symptoms except in extremis and so are rarely aware they carry the gene unless screened.

Sickle cell disease is more common amongst individuals from Africa, the Mediterranean, Middle East, parts of India, the Caribbean and South and Central America. This reflects the relationship between malaria and sickle cell disease. Prevalence of the sickle cell gene is significantly higher in areas with a history of malaria and this is because sickle cell trait protects against malaria, hence conferring a survival advantage in areas when malaria is a significant cause of death.

Sickle cell disease most commonly presents with painful crises due to the deformed blood cells clogging up small blood vessels and starving the surrounding tissue of oxygen. Repeated crises can cause organ damage which can occur in any organ in the body. Other presentations include overwhelming sepsis, acute chest syndrome, priapism, lung disease, pulmonary hypertension, renal disease, recurrent chronic leg ulceration, visual loss and stroke[3]. Sickle cell disease is unpredictable with random crises and variable severity which adds to the psychological impact of a chronic disease and the challenge of managing education, employment and social interaction.

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[1] Outer north east London consists of the London Boroughs of Barking & Dagenham, Havering, Redbridge and Waltham Forest

[2] Sickle cell disease in childhood: Standards and guidelines for clinical care. 2nd Ed. Oct. 2010. NHS Screening Programmes. p5

[3] Standards for the clinical are of adults with sickle cell disease in the UK. Sickle Cell Society. 2008. p16

Sickle cell disease can also lead to premature death and the median age of death for men with sickle cell disease is 42 years and for women is 48 years[4].

Sickle cell disease can be diagnosed with a single blood test. There has been a national antenatal and newborn screening programme since 2001 in England, although this obviously does not identify sickle cell disease in new arrivals or individuals born before 2001.[5, 6]

National standards have been published for health services for adults and children with sickle cell disease in 2008 and 2010. The aim of sickle cell disease management is to improve survival and reduce the frequency, duration and severity of painful crises and other complications. This involves prophylaxis through immunisation and antibiotics (Penicillin V), drug treatment and in some cases blood transfusion, lifestyle support and management, psychological and social/welfare support.

Sickle cell disease is a chronic disease with a highly variable pattern of presentation which requires clear and coherent service delivery and support across primary, secondary and tertiary health services and social care to improve the quality and life expectancy for people affected by the condition.

What's the local picture and how do we compare?

The demographic profile of outer north east London has been evolving over the last ten years and the increasing diversity is reflected in the growth in the case load of patients living with and affected by sickle cell disease.

Estimated demand was modelled using the generic North East London prevalence estimate of 2.18 significant conditions[7] per 1,000 babies screened[8] applied to the GLA 2011 population estimated for the ONEL boroughs[9]. The age band was capped at 74years which is possibly still over-estimating the life-expectancy of people living with sickle cell disease. This estimated a total patient case load of 1,366 patients living in ONEL boroughs with clinically significant sickle cell disease.

Table 7.28.1: Estimated Sickle Cell Disease Case Load in outer north east London boroughs

Borough	Projected No. new births	Estimated Case Load	Projected Population 1 to 8 yrs	Estimated Case Load	Projected Population 19 to 74 yrs	Estimated Case Load	Total Estimated Case Load
Barking & Dagenham	3,724	8.1	45,722	99.7	118,546	258.4	366.2
Havering	2,836	6.2	48,569	105.9	163,416	356.2	468.3
Redbridge	4,196	9.1	60,531	132.0	179,089	390.4	531.5
Total	10,756	23.4	154,822	337.5	461,051	1005.1	1366.1

[4] Mortality in Sickle Cell Disease. Life expectancy and risk factors for early death. Platt OS, et al. The New England Journal of Medicine, 09 June 1994, vol. /is. 330/23(1639-44), 0028-4793
[5] http://sct.screening.nhs.uk/standardsandguidelines
[6] http://sct.screening.nhs.uk/
[7] Significant conditions comprise the following results: FS, FSC, FS other and FE (F, foetal haemoglobin; S, S haemoglobin; C, C haemoglobin; E, E haemoglobin).
[8] Positive screening and carrier results for the England-wide universal newborn sickle cell screening programme by ethnicity and area for 2005–07. Streetly A et al. J Clin Pathol 2010;63:626-629 doi:10.1136/jcp.2010.077560 Table 1
[9] GLA 2005 extrapolations for 2011 population

Demand is increasing and the total caseload at BHRUT has increased from 575 in 2009 (288 adults and 263 children) to 748 in 2014 (406 adults and 342 children) (Table 7.28.2). It is also worth noting that the service currently manages around 85 patients for other areas including Newham, Waltham Forest Essex.

Table 7.28.2: Activity at BHRUT in 2014

Borough	Paediatric Register	Adult Register	Total Case Load
Barking & Dagenham	200	173	373
Havering	52	88	140
Redbridge	59	92	151
Other	32	53	85
Total	343	406	749

The modelling is limited somewhat by the differing impact of ethnicity on the three related boroughs. Havering has a significantly smaller proportion of the ethnic minority populations which are associated with sickle cell disease, and in contrast Barking and Dagenham has a larger proportion of these populations. However, there is a still a substantial gap between the estimated and actual populations, especially for adults, and this may well reflect the impact of the national screening program increasing awareness of children with the condition, and the relatively low levels of screening and identification amongst adults.

Modelling was done for Barking and Dagenham to estimate the number of potential carriers of sickle cell disease in the population. Although this group rarely have complications or clinical presentations it is an important risk factor during pregnancy, labour and surgical procedures. The modelling estimated that around 2,500 people are carriers of the sickle cell gene in Barking and Dagenham and this is over double the number of carriers identified by BHRUT, suggesting a substantial gap in identification of carriers in the community. The data from BHRUT antenatal screening programme has estimated the carrier rates locally to be 1 in 14 for Barking and Dagenham, 1 in 50 for Havering and 1 in 32 for Redbridge.

An audit undertaken by the service at BHRUT over a 3 month period in 2013/14 found that there were 77 attendances by adults, of whom 51 went to Accident and Emergency and 26 to the Day Unit. 36 patients were admitted from A&E and 11 form the Day Unit. During the same time period 20 paediatric cases were reviewed by A&E, of whom 15 were admitted. This audit compares with one undertaken in 2010 when there were 58 A&E attendances (47 adults and 11 children), and demonstrates the increasing need for care.

Between 2007 and 2009 there were two deaths in which sickle cell disease was recorded in the death certification and therefore could be identified in the reports.

Antenatal sickle cell and thalassaemia screening[10]

There are two key performance indicator for antenatal sickle cell and thalassaemia screening, ST1 and ST2:

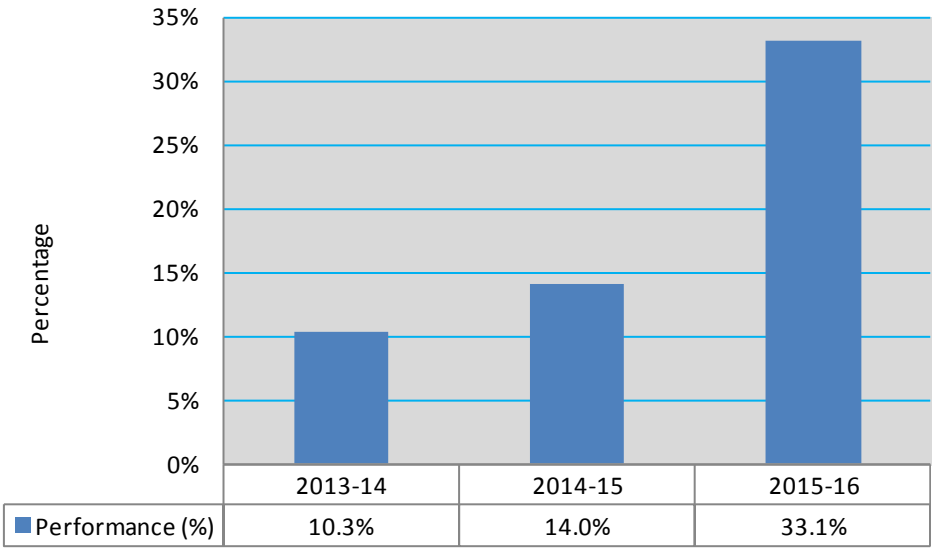
ST1: Antenatal sickle cell and thalassaemia screening coverage: The proportion of pregnant women eligible for antenatal sickle cell and thalassaemia screening for whom a conclusive screening result is available at the day of report.

BHRUT's performance rate for ST1 in the past three years from 2013-14 to 2015-16 where above the achievable threshold of 99%.

ST2: Antenatal sickle cell and thalassaemia screening – timeliness of test: The proportion of women having antenatal sickle cell and thalassaemia screening for whom a conclusive screening result is available by 10 weeks' gestation.

There has been some improvement in BHRUT's performance on ST2 during the last three years (illustrated in Figure 7.28.1) but it's performance is still behind the acceptable performance threshold of 50%. In 2014-15 the BHRUT performance level was 14%, the 8th lowest between 130 service providers nationally with available data. In quarter-3 of 2015-16 BHRUT's performance increased to 41.4%, the 32nd lowest between 141 service providers, however it would be better to see the outcome of the annual performance once it has published.

Figure 7.28.1 Antenatal sickle cell and thalassaemia screening – timeliness of test, BHRUT, 2013-14 to 2015-16*



Source: PHE, 2016, NHS screening programmes
**2015-16 data is the average of 3 quarters (the 4th quarter is not available yet).*

[10] *PHE, 2016, "NHS screening programmes: national data reporting" [online] available from: https://www.gov.uk/government/collections/nhs-screening-programmes-national-data-reporting [Last accessed: 29 Jul. 16]*

Local services for people with sickle cell disease

BHRUT currently provide specialist haematology services and a specialist nurse led community service based in King George Hospital with twice weekly clinics at Barking Hospital (B&D), weekly clinics at King George Hospital (Redbridge) and a monthly Saturday clinic here and weekly clinics at MyPlace (Havering). There is a lead consultant for adults and children with sickle cell disease, although she has substantial other responsibilities within the haematology department.

Paediatric patients have annual trans-cranial Doppler scans done by BHRUT. Approximately 120 are done each year and they are seen by the consultant as part of a normal appointment. Approximately 1 in 5 paediatric patients have an abnormal scan and require more frequent follow up scans. These children are often the group that then require long term transfusions to prevent a stroke and therefore move from an out-patient procedure to a day case tariff.

Obstetric patients are seen in a joint consultant led clinic and in 2010 14 women with sickle cell disease delivered under BHRUT care. The obstetric management of sickle cell disease is intensive, with additional ultrasound screening and regular blood tests, and women are seen around 12 to 14 times in clinic during their pregnancy.

The out-patient service is funded through national tariff payments for outpatients and the service in 2009 had 700 paediatric outpatient appointments and 500 adult appointments, excluding those for trans-cranial Doppler screening. These numbers are likely to have increased year on year with the increasing number of people on the sickle cell register.

In addition NHS Barking and Dagenham CCG supports additional provision through:

- Funding for the specialist nurse and administration for follow-up of the positive cases identified through the new born and antenatal linked screening program.
- Funding for welfare and benefits advice provided by a specialist advisor alongside the outpatient clinic.

Views of local people with sickle cell disease

As part of a review of the service for people with sickle cell disease in 2010 a range of stakeholders were identified and engaged with to discuss the service provision and models of care, these included patients, professionals and commissioners.

A specific patient engagement event was held in February 2011 with the local Sickle Cell and Thalassaemia Support Group which included representatives from the national sickle cell support organisation.

The feedback from the stakeholders was:

- Strong patient loyalty to current service at BHRUT and viewed generally as a competent and coherent unit.
- Recognition that current service is stretched and limited, particularly because of administration and lack of specialist nurses.
- Some really positive local service development is already happening around day case management of transfusions but not clear how this translated into tariff payments.
- Generally a lack of awareness of growth in demand for these services amongst clinicians outside the service and the impact of the changing population demographic.
- Highly varied views on ability of primary care and general practice to offer support to patients with haemoglobinopathies, with patients having least confidence in GPs to understand and meet their needs.
- Patients would like access to appointments in the afternoon and early evening (4-7pm).
- Some support for telephone and email advice and consultations but limited.
- Very strong call for more education and training of professionals across secondary and primary care.

Since the engagement work in 2011, a community sickle cell disease service has been commissioned from BHRUT as part of developing the service to better meet the needs of the local population. Work has also been commissioned to better support children living with, and affected by, sickle cell disease through a new A to Z resource developed by the local Sickle Cell and Thalassaemia Support Group and the Lead for Haemoglobinopathies at BHRUT for the under 5 year age group and, in 2013, by Medikidz to support children aged 8-12 years living with sickle cell disease.

The Barking and Dagenham 2012-15 Health and Wellbeing Strategy established a focus on improving services for people living with sickle cell disease in the first year of the Health and Wellbeing Partnership. The sickle cell service for Barking and Dagenham was commissioned and has brought additional focus across the borough to this area.

Recommendations for Commissioners

Health and Wellbeing Board partner organisations should continue to work collaboratively to signposting to increase awareness of sickle cell disease across front line staff in health, education, social care, voluntary sector and police settings. Partners should focus on good outcomes for individuals with Sickle Cell Disease and Thalassaemia.