Contents

8 Sickle cell disease

8.1 Introduction

Sickle cell disease is the fastest growing and one of the most common, genetic disorders in England. [1] Sickle cell disease is the name given to a group of hereditary conditions that affect the red blood cells. [2] People with sickle cell disease produce sickle-shaped red blood cells that have a much shorter lifespan than healthy blood cells – 20 days compared to 120 days for a healthy red blood cell. The sickle-shaped blood cells can become occluded in blood vessels, causing a sickle cell crisis (see Box 1 for definition).

Sickle cell disease is a serious condition that can seriously affect a person's life. The average life expectancy for those with sickle cell disease is 40 to 60 years. [2] Sickle cell disease can lead to several other health problems, such as bacterial infections,

bone and joint problems (including impaired growth), folate deficiency, gallstones, epilepsy, priapism,[1] chronic lung disease, neurological diseases, kidney problems, vision problems, or a stroke. [2] People with sickle cell disease who also suffer from depression and anxiety are more likely to suffer from painful episodes. [3] Research has also shown that depressive symptoms in sickle cell disease are associated with poor pain management, multiple blood transfusions, low family income and history of frequent crises. [4]

While the burden of morbidity and mortality at the population level is small, sickle cell disease is a major cause of inequality due to the disproportionate impact on certain ethnic groups (see Section 8.2). While sickle cell disease is less common than other chronic diseases (such as cardiovascular disease, diabetes and cancer) it is associated with higher rates of hospital utilisation, particularly acute care admissions and longer length of stay. [1]

Box 1: Definitions used in this section

Sickle cell trait – a condition where a person carries one of the genes that cause sickle cell, but does not have the condition themselves. They will not develop the disease, but are at risk of having a child with the condition if their partner is also a carrier.

Sickle cell crisis – a painful episode that occurs in sickle cell disease when blood vessels are blocked by sickle-shaped red blood cells.

Thalassaemia – a group of inherited conditions that affect haemoglobin, which is used by red blood cells to carry oxygen around the body. People with thalassaemia produce either no, or too little, haemoglobin. This can make them very anaemic (tired, short of breath and pale).

8.2 Causes and risk factors

Sickle cell disease is caused by a recessive gene. Inheritance of a gene from both parents can cause the red blood cell to form a sickle shape. If the child only inherits a gene for sickle haemoglobin from one parent, they will have the sickle cell trait but they will not have sickle cell disease. If both parents carry the sickle cell trait, their child will have a one in four chance of being born with sickle cell disease, a one in two chance of being a sickle cell carrier themselves, and a one in four chance of not having the disease or the trait.[2]

People who are carriers of sickle cell disease are also at risk of having a child with a blood disorder if their partner is a carrier of a different type of blood disorder, such as the gene for thalassaemia. The sickle cell gene from one parent can combine with a

[1] Long lasting, painful erection that can cause permanent damage if not treated quickly.
[2] If one parent has sickle cell disease and one does not have the disease, all children will be born with sickle cell trait. If one parent has sickle cell disease and one has sickle cell trait, all children will be born with either sickle cell disease or sickle cell trait.

gene for another blood disorder from the other parent, leading to a different type of sickle cell disease.

Sickle cell disease is generally more prevalent in people from Africa, the Caribbean, the Mediterranean, India, Pakistan, south and south-east Asia and the Middle East. Among adults registered with a GP in Hackney and the City of London, 85% of sickle cell sufferers are from a Black ethnic background (see Section 8.4.3 Figure 3)

Sickle cell disease is more common in countries with high malaria prevalence. Studies show that the presence of sickle cell trait can provide some resistance to malaria, which may help to explain this trend. [5]

8.3 Local data and unmet need

In 2017, there were 337 adult Hackney residents (age 18+) recorded as having sickle cell disease by their GP.[3] In the City of London, there were fewer than five adults recorded as having the condition. [6]

In 2016/17, there were 2,344 hospital admissions for sickle cell disease among residents of Hackney and the City of London (age 18+). [7]

Homerton University Hospital NHS Foundation Trust is classified as a high-prevalence trust – that is, more than 2% of antenatal booking blood tests received are screened positive for sickle cell and thalassaemia. [8] Homerton Hospital achieves a very high level of coverage for antenatal sickle cell and thalassaemia screening (see Section 8.6.1 for details of the national screening programme and Section 8.7.1 for local performance). [9] It is likely that this test identifies almost all cases of sickle cell disease locally.

8.4 Inequalities

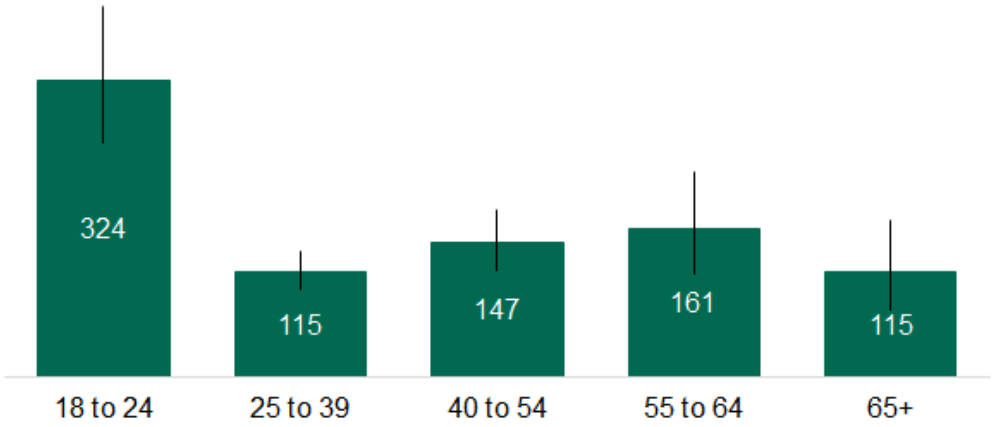
8.4.1 Age

In 2017, about a quarter (23%) of adults with sickle cell disease in Hackney and the City of London were between the ages of 18 and 24.[4] Residents in this age range have the highest rate of sickle cell disease compared to all other age ranges (see Figure 1). [2]

[3] Less than five City of London residents and 153 Hackney residents with sickle cell are under the age of 18. This JSNA chapter focuses on adults.

[4] Due to sickle cell disease being a rare condition, and the small numbers of patients in the City of London, the data for Hackney and the City of London have been presented together.

Figure 1: Rate of GP patients recorded with sickle cell disease in Hackney and the City of London, by age (per 100,000, age 18+, 2017)



Source: Extracted from the local GP register by Clinical Effectiveness Group (CEG), Blizard Institute, April 2017.
Note: Data cover residents of Hackney and the City registered with a GP in Hackney, the City of London, Tower Hamlets and Newham.

8.4.2 Gender

Figure 2 shows that local prevalence of sickle cell disease is similar among men and women.

Figure 2: Rate of GP patients recorded with sickle cell disease in Hackney and the City of London, by gender (per 100,000, age 18+, 2017)

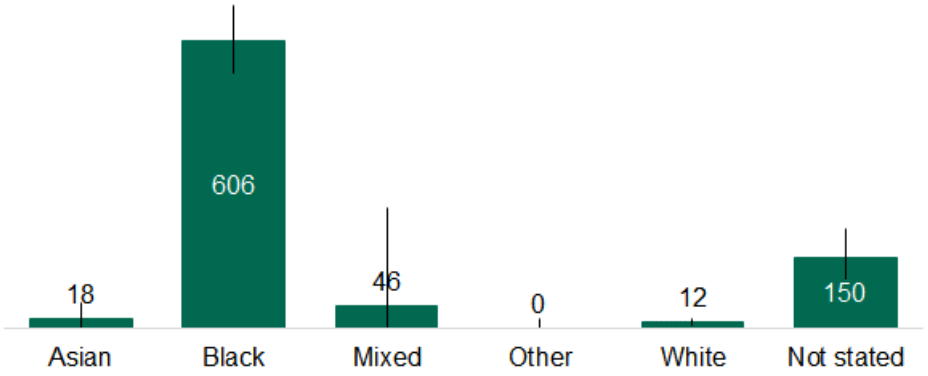
Source: Extracted from the local GP register by CEG, Blizard Institute, April 2017.
Note: Data cover residents of Hackney and the City registered with a GP in Hackney, the City of London, Tower Hamlets and Newham.

8.4.3 Ethnicity

Sickle cell disease predominantly affects people of Black ethnic origin, but can also affect people from the Mediterranean, India, Pakistan, south and south-east Asia and the Middle East. Locally, the vast majority of adults with sickle cell disease are of

Black ethnicity (85%). Black residents are significantly more likely to have sickle cell disease compared to all other ethnic groups (see Figure 3). A small proportion of patients are coded as White (4%) and Asian (1%) ethnicities.

Figure 3: Rate of GP patients recorded with sickle cell disease in Hackney and the City of London, by ethnicity (per 100,000, age 18+, 2017)



Source: Extracted from the local GP register by CEG, Blizard Institute, April 2017.
Note: Data cover residents of Hackney and the City registered with a GP in Hackney, the City of London, Tower Hamlets and Newham.

8.4.4 Sexual identity

There is insufficient information on local rates of sickle cell by sexual identity and orientation to draw local inference.

8.4.5 Disability

People with sickle cell disease are more likely than the general population to have physical and mental disabilities. Sickle cell disease can also lead to several other major health problems, such as pulmonary hypertension, kidney problems, vision problems or strokes.

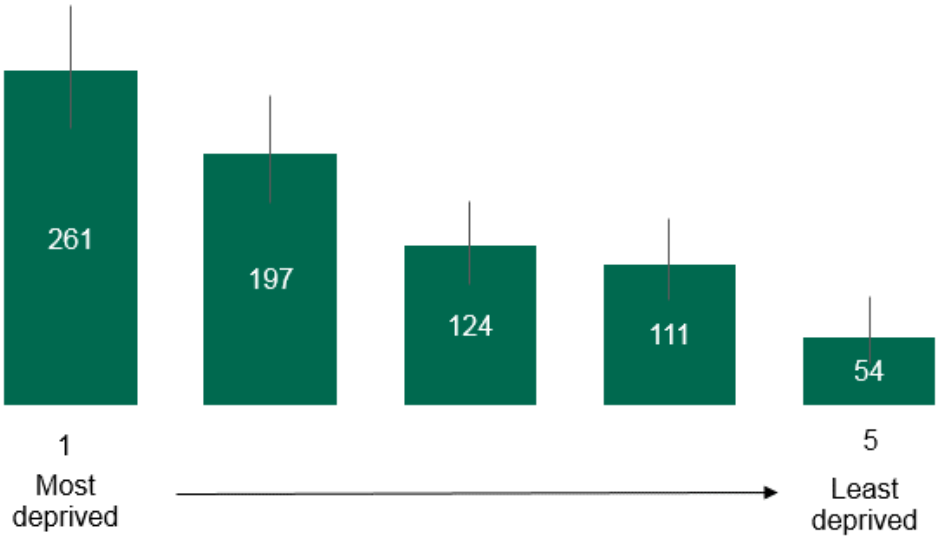
8.4.6 Socio-economic disadvantage

There is a clear social gradient in the prevalence of sickle cell disease, as shown in Figure 4. More than a third of adults living with sickle cell disease in Hackney and the City of London live in the fifth most deprived neighbourhoods, while 5% live in the least deprived.[5] This reflects the fact that people of Black ethnicity are more likely to live within more deprived areas. The impact of reduced schooling due to crises and

[5] Deprivation is defined using the Index of Multiple Deprivation 2015 (IMD). IMD is a measure of relative deprivation for small areas that combines 37 separate indicators, each reflecting a different aspect of deprivation experienced by individuals living in an area.

hospital admissions may affect future employment opportunities, a key driver of socio-economic status.

Figure 4: Rate of GP patients recorded with sickle cell disease in Hackney and the City of London, by deprivation quintile (per 100,000, age 18+, 2017)



Source: Extracted from the local GP register by CEG, Blizard Institute, April 2017.
Note: Data cover residents of Hackney and the City registered with a GP in Hackney, the City of London, Tower Hamlets and Newham.

8.4.7 Location within Hackney and the City

The prevalence of sickle cell disease within Hackney and the City is strongly associated with the proportion of the resident population of Black ethnicity. As reported in the Hackney health and wellbeing ward profiles, prevalence is particularly common in Homerton and Haggerston wards. [10]

Ward level data are not available for the City of London.

8.5 Comparisons with other areas and over time

There are no data available to compare the prevalence of sickle cell disease in Hackney and the City of London with other local authorities, or over time.

Literature exploring hospital admissions for sickle cell disease in 2009/10 found that Hackney and the City had the highest admission rates in England and London at 837 per 100,000 population. This rate was over twice that of the next highest area – Newham, at 388 per 100,000 population. [1]

The number of hospital admissions for sickle cell disease has increased over time locally. In 2012/13, there were 1,735 hospital admissions for sickle cell disease in Hackney and the City residents; in 2016/17, this had increased to 2,344 (Table 1).

This may be due to many factors, such as migration from countries where sickle cell disease is common, increased identification of sickle cell disease patients, and increased life expectancy of patients with sickle cell disease through improved management and treatment. The increase in admissions may also be a result of a drop in the threshold for admissions of patients with sickle cell disease, lack of confidence in managing sickle cell by non-specialists in emergency departments, or patients being discharged too early and then readmitted for the same episode of illness. [1]

Table 1: Number of hospital admissions for sickle cell disease over time (age 18+, 2012/13 – 2016/17)

Financial year	Number of hospital admissions
2012/13	1,735
2013/14	1,973
2014/15	1,899
2015/16	2,284
2016/17	2,344

Source: Hospital episode statistics.
Note: Figures are based on adult residents (age 18+) living in Hackney and the City of London (or those with no residential data registered with a Hackney and the City GP practice) admitted to secondary care for ICD10 code D57 (sickle cell disorders).

The Global Burden of Disease Study estimates that the mortality and morbidity associated with sickle cell disorders and thalassaemia have declined across Greater London between 1990 and 2016. [11]

8.6 Evidence and good practice

8.6.1 Identification and early intervention

The NHS sickle cell and thalassemia (SCT) screening programme was rolled out in 2006 as a linked antenatal and newborn screening programme, to identify the risk of a newborn inheriting sickle cell disease. Symptoms do not usually manifest in babies until they are six months of age, therefore early diagnosis can help to put a care plan in place and help parents to prepare and know what to expect once symptoms begin to appear. [12]

All pregnant women thought to be at risk of being sickle cell carriers should be offered screening for sickle cell by 10 weeks' gestation. The screening involves a blood test and a 'family origin questionnaire' (FOQ), which records the parents' ethnic background. The exact process varies depending on whether the NHS acute trust has a high prevalence of positive screens or not. [13]

- In high-prevalence trusts (where sickle cell disease is estimated to affect more than 1.5 pregnancies per 10,000 births), such as locally (at Homerton University Hospital), all women are offered a blood test and the results are interpreted using information from the FOQ.

- In low-prevalence trusts (where sickle cell disease is estimated to affect fewer than 1.5 pregnancies per 10,000 births), an FOQ is completed and a blood test is then offered only to those women deemed to be at higher risk. [14]

The test is also offered as part of the newborn bloodspot screening programme. [13] This involves a 'heel-prick test' when the baby is five days old, in which a midwife collects four drops of blood by pricking the baby's heel. This blood is tested for nine serious health conditions, including sickle cell disease.[6]

Prenatal testing is available on request for couples who are looking to start a family but do not know their carrier status. As with the antenatal screening programme, an FOQ would be completed for each parent, and a blood sample would be taken to determine the parents' likelihood of being a sickle cell carrier. If both parents were found to be carriers, a viable option is pre-implantation genetic diagnosis – this process is similar to IVF, but the resulting embryos are tested to ensure they don't carry sickle cell disease before they are implanted. [15] Parents also have the option of using a donor egg or sperm.

8.6.2 Treatment, care and support

The frequency and severity of sickle cell crises vary greatly, and triggers can include stress, strenuous exercise, dehydration, or weather conditions. Most crises can be managed at home with pain relief and measures such as staying hydrated, taking a warm bath, or massaging the affected area. However, in some cases the pain or type of crisis (e.g. lung crisis) may not be manageable with pain relief at home, and in such cases, or in cases where there are other complications, sickle cell crises can be treated in hospital. [16]

National Institute for Health and Care Excellence (NICE) guidelines and quality standards have been developed to provide a recommended pathway for treating those with sickle cell disease. [17] The quality statements recommend the following.

- People who present at hospital with an acute painful episode have a pain assessment, a clinical assessment and appropriate pain relief medication within 30 minutes of presentation.
- An assessment of pain relief is carried out every 30 minutes until satisfactory pain relief has been achieved, and then at least every four hours
- People with an acute painful episode who are taking strong opioids are monitored for adverse events every hour for six hours from first administration or increase of dosage, and then at least every four hours.
- People with an acute painful episode are assessed for acute chest syndrome if they have one or more of the following: abnormal respiratory signs or symptoms, chest pain, fever, or signs and symptoms of oxygen deficiency.

[6] The diseases screened for in the heel-prick test are: sickle cell disease, cystic fibrosis, congenital hypothyroidism, phenylketonuria, medium-chain acyl-CoA dehydrogenase deficiency, maple syrup urine disease, isovaleric acidaemia, glutaric aciduria type 1, and homocystinuria.

- Healthcare professionals who care for people with an acute painful episode have access to locally agreed protocols on treatment and management and specialist support from designated centres.
- People with an acute painful episode are given information before discharge on how to continue to manage their current episode.

Regular blood transfusions are often needed to increase the level of normal red bloods cell and treat the anaemia that is caused by the death of the sickle-shaped red cells. This increases the oxygen-carrying capacity of the red blood cells.

A side effect of having repeat transfusions is that people will gradually collect too much iron in the body, which can lead to a dangerous condition known as transfusion-induced iron overload. This excess iron can be removed with iron chelation treatment, although other treatments are available.

Nearly all people who suffer from sickle cell disease have anaemia[7] which is caused by the poor oxygen-carrying capacity of the sickle-shaped red blood cells. [2] Infection risk is very high in sickle cell disease, especially from encapsulated bacterial infections due to splenic infarction (reducing the number of red and white blood cells). People with sickle cell disease are particularly vulnerable to common pathogens such as pneumococcus, haemophilus, and salmonella.

Vaccinations and daily antibiotics can help reduce the risk of contracting many infections – the NHS recommends that adults with sickle cell disease should receive hepatitis B, pneumococcal polysaccharide, haemophilus influenza type B, meningococcal ACWY, meningococcal B, and BCG vaccinations, plus an influenza vaccination every year. [18]

8.7 Services and support available locally

8.7.1 Identification and early intervention

All NHS trusts local to Hackney and the City (including Homerton University Hospital) are classified as high-prevalence trusts, therefore all pregnant women patients at these trusts are offered a blood test as part of the NHS sickle cell and thalassaemia screening programme. Uptake of screening is very high locally – in 2015/16, 99.6% of pregnant women in Hackney and the City were screened and given a conclusive result, above the developmental target of 95%. [13]

8.7.2 Treatment, care and support

Several local hospital services are commissioned by NHS England and available to adults in Hackney and the City with sickle cell disease. These include a specialist

[7] A condition in which there is a deficiency of red cells or of haemoglobin in the blood.

centre for adults at Homerton Hospital, and a specialist centre for adults and children at the Royal London Hospital in Tower Hamlets.

As well as providing inpatient management for adults and specialist laboratory diagnostic services, Homerton University Hospital also provides:[8]

- a drop-in service allowing patients with sickle cell to attend hospital for treatment in a crisis without having to attend A&E
- an automated transfusion exchange service, which allows sickle cell patients to receive blood transfusions faster and less frequently
- a psychology service tailored to adults with sickle cell disease and thalassaemia, to help promote wellbeing and improve quality of life (patients can self-refer and receive support on a range of issues, including pain management in hospital and at home, improving communication with medical staff, adjusting to regular hospital visits, and managing other life problems that can affect coping mechanisms).

There are also several community services commissioned by City and Hackney Clinical Commissioning Group, including:

- a drop-in service for nurse assessment and advice
- advice on welfare, benefits, social care, employment, housing and education
- a therapy clinic providing hydroxycarbamide treatment to help reduce the number of crises and the need for blood transfusions, and chelation treatment to combat iron overload following repeat transfusions.

Sickle cell disease patients can access immunisations through primary care or secondary care.

8.8 Service gaps and opportunities

Blood transfusions form a major part of sickle cell treatment; however, it is crucial that the transfused blood is matched – this is most likely to come from a donor of the same ethnicity. Nationally, however, there is a shortfall of suitable blood donors, and only 1% of blood donors in England are of Black ethnicity, despite the fact that the vast majority of patients with sickle cell are of Black ethnicity. [19] Campaigns have been run to try to increase the number of donors from all ethnic backgrounds, to ensure that blood can be matched as closely as possible in transfusions.

[8] For more detail, visit http://www.homerton.nhs.uk/our-services/services-a-z/s/sickle-cell-service-thalassaemia-service/

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