Journal of Public Health | pp. 1–5 | doi:10.1093/pubmed/fdx172

How many people have sickle cell disease in the UK?

Elizabeth Dormandy[1], John James[1], Baba Inusa[2], David Rees[3]

[1]Sickle Cell Society, 54 Station Road, London NW10 4UA, UK
[2]Evelina London Children's Healthcare, Guy's and St Thomas' NHS Foundation Trust, London, UK
[3]Department of Molecular Haematology, King's College London and King's College NHS Foundation Trust, London, UK
Address correspondence to Elizabeth Dormandy, E-mail: elizabethdormandy@gmail.com

ABSTRACT

Background Sickle Cell Disease (SCD) is now one of the most common serious genetic condition in England. There is no reliable estimate of the total number of people living with SCD in the UK, to support commissioners and providers of services for people with SCD.

Aim To obtain reliable data on the total number of people living with SCD in the UK in 2016.

Method Information was requested from all national databases known to hold information on the number of people living with SCD in the UK. The information from each data source was first reviewed to estimate likely inaccuracies and then combined to provide a best estimate of people living with SCD in the UK.

Conclusion This process indicated there are are about 14000 people living with SCD in the UK. This is equivalent to 1 in 4600 people.

Keywords blood and immune disorders, epidemiology, genetics

Introduction

Background: Sickle Cell Disease(SCD) is now one of the most common serious genetic condition in England. The condition has a life threatening outcome. However there is no current reliable estimate of the total number of people living with SCD in the UK. This lack of data makes planning appropriate services for those with SCD problematic and has the potential to lead to poor care for those with sickle cell disease.

Aim: To obtain reliable data on the total number of people living with SCD in the UK in 2016. Information was requested on clinically significant cases of sickle cell disease and includes Haemoglobin SS and Haemoglobin SC.

Sickle cell disease (SCD) is a serious lifelong condition. It adversely affects the rheology of red blood cells due to a change in the haemoglobin molecule. When deoxygenated red blood cells are unable to pass freely through blood capillaries they form clusters which can block the blood vessels, resulting in tissue hypoxia and intense pain (known as a sickle crisis). Sickle haemoglobin (HbS) is a haemoglobin variant where the sixth amino acid of the beta globin chain, glutamic acid is replaced by valine. It is inherited as an autosomal recessive condition, and is common in people with family origins from Africa and the Caribbean. The most severe form of SCD is sickle cell anaemia (HbSS).

As well as having attacks of acute pain, children with SCD are at increased risk of infection and stroke. In low-income countries, it is estimated there is a 50% mortality in the first decade of life.[1] In higher income countries more than 94% of children survive into adulthood.[2] Children are, however, at risk from developing long-term complications such as repeated sickle cell crises, severe anaemia, damage to other organs, stroke and infections. Treatment options include prophylactic antibiotics, regular transfusions, therapy with hydroxyurea and education and support to avoid situations that can precipitate a sickle cell crisis.

Newborn screening for SCD has been introduced in countries such as Brazil, UK and the Netherlands.[3–5] Newborn screening is offered to all infants up to 1 year of age in the UK. This screening programme aims to identify all infants

Elizabeth Dormandy, Scientific Advisor
John James, Chief Executive
Baba Inusa, Consultant in Hemoglobinopathies and Children's General Medicine
David Rees, Professor of Paediatric Sickle Cell Disease

with SCD and ensure that they are seen by a hospital doctor, prescribed prophylactic penicillin, regularly reviewed and parents are supported and educated to keep children well.

Globally the number and distribution of people with SCD is changing; resulting from two factors (i) improved survival and (ii) population migration.[6,7] It is important that those responsible for providing clinical services for people with SCD have a reliable estimate of the numbers of people with the condition to ensure that appropriate services are provided. This article collates together information across the UK to provide this information, which will be particularly useful for commissioners and providers of services.

Methods

Information was requested from all national databases known to hold information on the number of people living with SCD in the UK. The data sources are listed in Table 1. They were reviewed to determine the age range covered, cohort or individual data, consented or audit data, the geographical region covered, the time period covered and whether the data source was based on clinically confirmed cases or screen positive cases. All data sources indicated that double counting was possible but unlikely, either because of data checks or the availability of identifiable data. Gender was not available from all data sources and was therefore not analysed further. Socioeconomic status data were not available.

Data sources

There is no single source of data that gives a comprehensive estimate of the current number of people living with SCD in the UK. The following known data sources were contacted and asked to provide information on the number of people with SCD: NHS Sickle Cell and Thalassaemia Screening Programme (https://www.gov.uk/topic/population-screening-programmes/sickle-cell-thalassaemia, 6 December 2017, date last accessed) provides audit data on the number of babies who are reported as screen positive for SCD by newborn screening laboratories in England. These screening data will include some false positive results, and is therefore likely to be an over estimate. The data have been reported annually since 2007 and are collected from all newborn screening laboratories in England.

The National Haemoglobinopathy Registry (NHR) (http://www.nhr.nhs.uk/, 6 December 2017, date last accessed) provides data on the number of people living with SCD in England and is the most complete estimate. However, it is a consented register and requires clinical centres to enter data. Both of these factors are likely to lead to incomplete data. The register started in 2008 and collects data on all ages.

Newborn Outcomes Project (https://www.gov.uk/guidance/newborn-outcomes-project-definition-and-implementation, 6 December 2017, date last accessed) a project with permission to collect non-consented data on the number of children up to the age of 5 living with SCD in England identified by the newborn screening programme. This provides data on confirmed or true positive cases. The data are likely to be more complete than the data from the National Haemoglobinopathy Registry as it is collected without consent on all confirmed cases since 2010. It, however, only contains data on children up to age 5.

Cardiff Sickle Cell and Thalassaemia Centre provided data on all people known to the clinical service with SCD living in Wales. This estimate does not include people with SCD not using the service. There are no data available on people who have SCD but do not use clinical services.

National Specialist and Screening Services, NHS Scotland provided data on numbers of people known to the clinical service with SCD living in Scotland. This service is linked with the Scottish Paediatric and Adult Managed Network This estimate does not include people with SCD not using the service. There are no data available on people who have SCD but do not use clinical services.

The Newborn Screening Programme in Northern Ireland provided data on the number of babies reported as screen positive for SCD (SCD suspected) by the regional newborn screening laboratory during 2012/13–2013/14

Royal Belfast Hospital for Sick Children provided data on numbers of children known to the service with SCD living in Northern Ireland. This estimate does not include children with SCD not using the service. There are no data available on children who have SCD but do not use clinical services.

Although not in the UK, data was also requested from the Republic of Ireland.

Clinical Service, Republic of Ireland provided data on numbers of people known to the service with SCD living in the Republic of Ireland. This estimate does not include people with SCD not using the service.

The data sources and a summary of the data collected are listed in Table A1.

Results

Table 1 lists the estimates provided by the different data sources of people living with SCD in the UK.

Data integration

The estimates cannot be compared directly as they cover different time periods, different age groups as well as different geographies. Therefore, we have reviewed each data source,

Table 1 Estimates of people with sickle cell disease in the UK

Data source	*Numbers*	*Notes*
NHS Sickle Cell and Thalassaemia Screening Programme	1670	Screen positive babies for 5 years 2009/10–2013/14 Screen positive includes HbSS, HbS/ beta thalassaemia, HbS/HPFH, HbSC, HbS/D-Punjab, HbS/E, HbS/O Arab
National Haemoglobinopathy register	9677	This includes people with HbSS, HbSC, HbS/DPunjab, HbS/O Arab, HbS/HPFH, HbS/Lepore, HbSE, HbS β0thal, HbSβ+thal There are 750 children under 5 years old
Newborn outcomes project	1154 Children under 5	(793 SS babies and 361 SC babies)
Cardiff Sickle Cell and Thalassaemia centre	79	This includes HbSS, HbSC, HbS/beta thalassaemia
National Specialist and Screening Services Directorate (NSD), NHS National Services Scotland	168	This includes HbS/beta thalassaemia, HbSC, HbSD and HbSS
Newborn Screening Northern Ireland	<5	Screen positive babies (SCD suspected) for the 2 years 2012/13–2013/14
Royal Belfast Hospital for Sick Children	7	Number of children known to the service with sickle cell disease living in Northern Ireland
Republic of Ireland	358 paediatric 305 SS 45 SC <5S D Punjab <5S β 0 thal 4<5SSβ + thal 68 adult	Paediatric haematology lead Ireland Adult haematology lead Ireland

estimated the inaccuracies around each estimate, compared the estimates and their inaccuracies in a data integration process outlined below.

The most robust data source for the number of people with SCD is from the National Haemoglobinopathy Register (NHR). This indicates there are 9677 people with SCD living in England. However, this is a consented register.

The non-consented data held by the newborn outcomes project shows there are 1154 children up to age 5 in England with SCD. This compares with the 1670 screen positive babies in England recorded for a 5-year period by the newborn screening programme. The difference between the data from the newborn outcomes project and the newborn screening programme is likely to result from the difference between screen positive results (newborn screening data) and confirmed or true positive results (newborn outcomes project reports data). Therefore, for these purposes, the data from the newborn outcomes project are the more reliable estimate of clinical cases. The comparison between these two data sources provides evidence that both estimates are similar, giving further evidence of validity.

The likely size of the underestimate of the numbers on the NHR can be determined by comparing the number of children aged under 5 from the register with the number of children aged under 5 in the newborn outcomes project.

The NHR has identified 814 children aged under 5, compared with 1154 children in the newborn outcomes project. Assuming the number of people not included in the register does not vary by age we can extrapolate from these figures to say the number of people recorded on the register is likely to be 70% (814/1154) of the true figure. Given 9559 people are included on the register, we can estimate the true number of people with SCD is 13 655 for England.

The estimates for the other countries in the UK (Wales, Scotland, Northern Ireland) are based on data from clinical centres. There are limited alternative sources to confirm these data. Assuming these data are correct there are 259 people with SCD in these three countries. There are 350 people living with SCD in the Republic of Ireland. These data have been included in this paper to provide comparison for the data provided by Wales, Scotland and Northern Ireland. They indicate the estimates for Wales, Scotland and Northern Ireland are reasonable estimates.

Combining the figures for the four home countries gives an overall estimate of 14 000 people living with SCD in the UK, equivalent to 1 in 4600 people. Newborn screening identifies 1 in 2000 babies born as screen positive for SCD.[5]

Discussion

Main findings of this study

This report provides the first national estimate of the number of people living with SCD in the UK and Republic of Ireland. It utilizes a range of national data sources and provides a comprehensive and reliable estimate, making it useful to those planning services and ensuring people with SCD are offered high quality care. The data are broken down by country.

What is already known

Data are taken from a range of sources. However, the data cannot be compared directly as the sources cover different time periods, different age groups as well as different geographies.

What this study adds

This study has reviewed each data source, estimated the inaccuracies around each data source, compared the estimates and inaccuracies and integrated the data.

Limitations of this study

The limitations of this report are that it has not possible to report data by area within the four UK countries, nor to report trends over time. Other inaccuracies may arise from patients moving to the UK who were born overseas, and the assumption that the proportion of patients added to the National Haemoglobinopathy Registry is similar across all ages.

This article is an initial step to provide robust data on the number of people living with SCD in the UK. Data were not available from all data sources on trends by time or age. Data on hospital admissions show that the peak age range for hospital admissions in those with SCD is the age range 20–29. These data also showed variation in hospital admission by gender, year and location.[8] Figures on hospital admissions are also an important part of planning services for those with SCD.

This work highlights the need for a unified data capture system that is capable of providing data by area within the four countries and providing trends over time, thus transforming the ability to plan service provision. There need to be pathways to ensure that screen positive babies are seen by a hospital doctor by 3 months of age, that annual specialist reviews of the babies are available, that trans-cranial Doppler Scanning is available and high quality in patient care is available across the country. Similar efforts are needed and occurring internationally to help establish neonatal screening programmes and pathways of care, particularly in Africa and India where the disease is most prevalent.[6]

Estimates suggest around 10 400 people in the UK have cystic fibrosis, with 1 in every 2500 babies born with cystic fibrosis.[9]

Conclusion

National data show there are ~14 000 people living with SCD in the UK. Commissioners and those providing services need to ensure that all people with SCD have access to high-quality care wherever they live in the UK.

Acknowledgements

The Sickle Cell Society extend warm thanks to the following for sharing the following data: National Haemoglobinopathy Register NHS Sickle Cell and Thalassaemia Screening Programme Sickle Cell and Thalassaemia Centre, Cardiff National Specialist and Screening Services Directorate (NSD), NHS National Services Scotland, Newborn Screening Northern Ireland Clinical Services, Royal Belfast Hospital for Sick Children Clinical Service, Republic of Ireland.

References

1 Quinn CT, Rogers ZR, McCavit TL *et al*. Improved survival of children and adolescents with sickle cell disease. *Blood* 2010;**115**(**17**):3447–52.

2 Telfer P, Coen P, Chakravorty S *et al*. Clinical outcomes in children with sickle cell disease living in England: a neonatal cohort in East London. *Haematologica* 2007;**92**:905–12.

3 Brandelise S, Pinheiro V, Gabetta CS *et al*. Newborn screening for sickle cell disease in Brazil: the Campinas experience. *Clin Lab Haematol* 2004;**26**(**1**):15–9.

4 Bouva MJ, Mohrmann K, Brinkman HB *et al*. Implementing neonatal screening for haemoglobinopathies in the Netherlands. *J Med Screen* 2010;**17**(**2**):58–65.

5 NHS SCT Screening Programme. Screening Programme Data Report. https://www.gov.uk/topic/population-screening-programmes/sickle-cell-thalassaemia (6 December 2017, date last accessed).

6 Piel FB, Hay SI, Gupta S *et al*. Global burden of sickle cell anaemia in children under five 2010–2050: modelling based on demographcs, excess mortality and interventions. *PLoS Med* 2013;**10**(**7**):e1001484.

7 Piel FB, Tatem AJ, Huang Z *et al*. Global migration and the changing distribution of sickle haemoglobin: a quantitative study of temporal trends between 1960 and 2000. *Lancet Glob Health* 2014;**2**:e80–89.

8 AlJuburi G, Laverty AA, Green SA *et al*. Trends in hospital admissions for sickle cell disease in England, 2001/02–2009/10. *J Public Health (Bangkok)* 2012;**34**:570–6.

9 Cystic Fibrosis Trust. https://www.cysticfibrosis.org.uk/what-is-cystic-fibrosis/faqs (6 December 2017, date last accessed).

Appendix

Table A1 Data sources and potential limitations

Data source	*Age range*	*Data type*	*Consented*	*Location*	*Time period*	*Estimate based on*
NHS Sickle Cell and Thalassaemia Screening Programme	Newborn babies	Cohort	No	England	2007–present	Screen positive babies
National Haemoglobinopathy register	All ages	Individual	Yes	England	2008–present	Confirmed cases
Screening Wales and Cardiff Sickle Cell and thalassaemia centre	All ages	Cohort	Audit data	Wales	Present	Confirmed cases
NHS National Services Directorate Scotland (NSD)	All ages	Individual	Consented	Scotland	Present	Confirmed cases
Newborn Screening Northern Ireland	Newborn babies	Cohort	Audit data	Northern Ireland	2012/2014	Screen positive babies
Royal Belfast Hospital for Sick Children	Children to age 16	Cohort	Audit data	Northern Ireland	Present	Confirmed cases
Paediatric and Adult Haematology lead Republic of Ireland	All ages	Cohort	Audit data	Republic of Ireland	Present	Confirmed cases
Newborn Outcomes project	0–5	Individual	No	England	2010–present	Confirmed cases—SS and SC disease