Sickle Cell Disease

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Prevalence

Sickle trait has a protective effect against malaria; global distribution has emerged from positive selection for this gene mutation.2

Occurrence highest in sub-Saharan Africa and small pockets of the Mediterranean, the Middle East, and India.5

In Cameroon, Republic of Congo, Gabon, Ghana, and Nigeria prevalence 20–30%, in some parts of Uganda 45%.4

Caribbean, South American, and Central American populations more at risk.1

Prevalent in populations where malaria is endemic, particularly African and African-Caribbean origins.2

Key Statistics

- 100,000 Americans have the disease, 12,500–15,000 in England.8
- 1/365 Black or African-American births, 1/160 Hispanic-American births11
- 1/13 Black or African-American babies and 1/79 in the UK are born with sickle cell trait.12
- Patients with SCD in the UK have median survival of 67 years, 58 years in the USA.8
- 1/2,000 births in England.3
- Global estimate 120 million affected by SCD, and 1,000 babies born with the disease each day in Africa alone.4

Current Treatment

Lifelong management:

- Avoid sudden changes in temperature and dehydration to reduce clot risk.7
- Hydroxyurea (hydroxyurea), crizanlizumab, L-glutamine, and voxelotor for pain crises, vaso-occlusive issues, sickling of blood cells, and complications.10
- Long-term antibiotics like penicillin mitigate susceptibility to infection.7
- Blood transfusions: acute, red blood cell, and regular.8

Curative

- Blood and marrow transplant: only current curative option, usually conducted in children unresponsive to other treatment. Cell replacement therapy for production of healthy red blood cells, significant risks involved.
- Gene therapy: CRISPR-Cas9 editing BCL11A in hematopoietic stem cells provides durable engraftment, high foetal haemoglobin expression, and elimination of vaso-occlusive episodes or need for a transfusion.16
- Further experimental testing required, providing generalisable results, before this option is made readily available.16

Unmet Needs

Access to quality treatment:
- Worst affected areas of Africa see 50–90% of children with SCD die before the age of 5 years.12
- In the UK, 90% of children diagnosed survive to adulthood.7

Cost:
- Financial burden is extremely high, a lifetime dependent on treatment $1.7 million USD medical bills.7

Blood and marrow transplants succeed in 85% of children; this must be from a related donor with human leukocyte antigen matched.7

Key:
- SCD: sickle cell disease

References