Sickle Cell Disease

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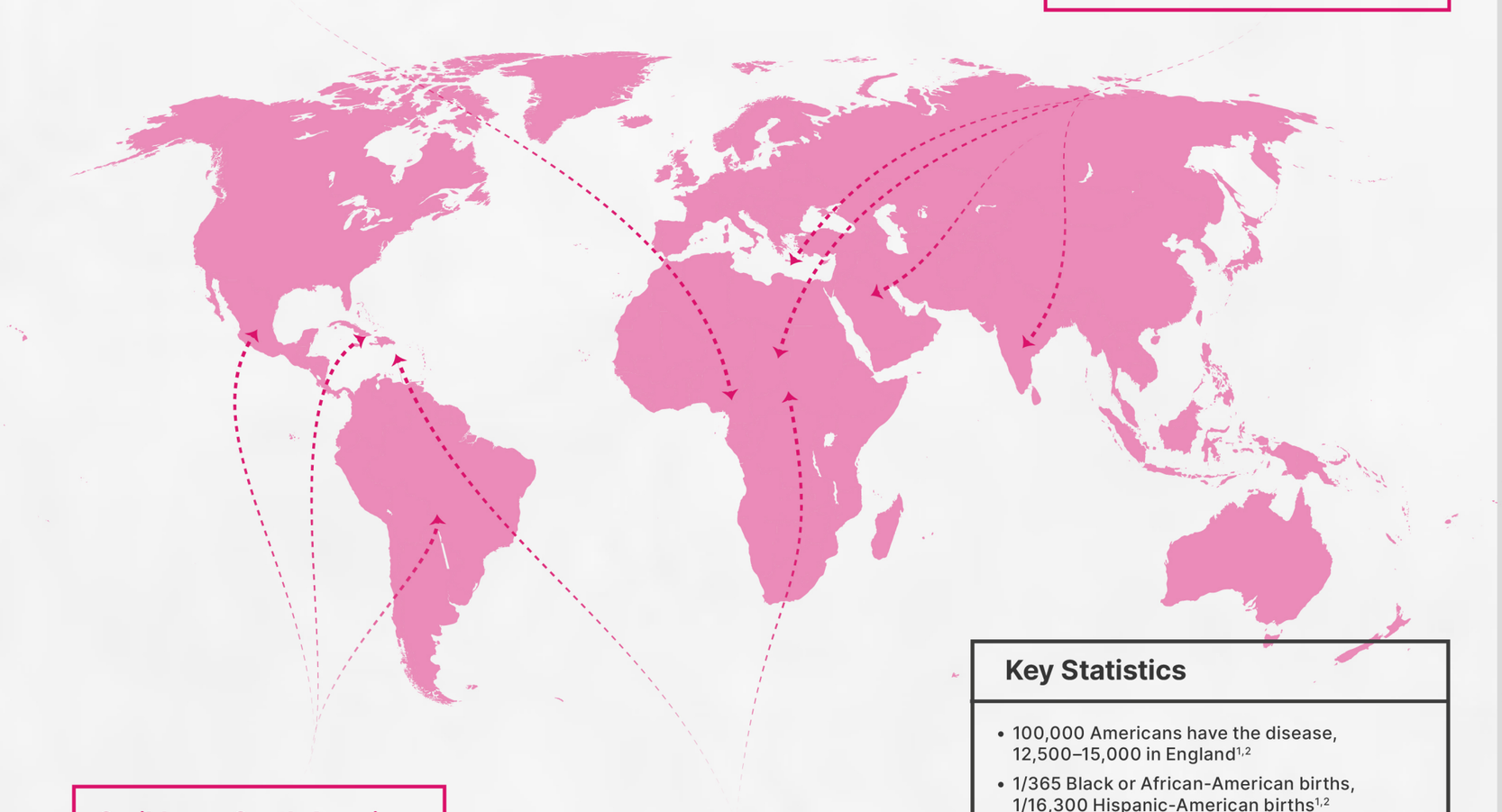
Infographic

Prevalence

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

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]



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

SCD: sickle cell disease

Key Statistics

- 100,000 Americans have the disease, 12,500–15,000 in England[1,2]
- 1/365 Black or African-American births, 1/16,300 Hispanic-American births[1,2]
- 1/13 Black or African-American babies and 1/79 in the UK are born with sickle cell trait[1-3]
- Patients with SCD in the UK have median survival of 67 years, 58 years in the USA[6]
- 1/2,000 births in England[3]
- Global estimation 120 million affected by SCD, and 1,000 babies born with the disease each day in Africa alone[9]

Current Treatment

Lifelong management:

- Avoid sudden **changes in temperature** and **dehydration** to reduce clot risk.[7]
- **Hydroxycarbamide (hydroxyurea)**, **crizanlizumab**, **L-glutamine**, and **voxelotor** for pain crises, vaso-occlusive issues, sickling of blood cells, and complications.[7,8]
- **Long-term antibiotic**s 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.[7]
- Gene therapy: **CRISPR-Cas9 editing** *BCL11A* in hematopoietic stem cells provides durable engraftment, high foetal haemoglobin expression, and elimination of vaso-occlusive episodes or a need for transfusion.[10]
- **Further experimental testing required**, providing generalisable results, before this option is made readily available.[10]

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.[4,6] In the UK, 99% of children diagnosed survive to adulthood.[6]

Cost:
Financial burden is extremely high, a lifetime dependent on treatment ≈$1.7 million USD medical bills.[11]

Blood and marrow transplants succeed in 85% of children; this must be from a related donor with human leukocyte antigen matched.[8]

Adults:
interventions are focused on children, but millions of adults live with the disease globally, awaiting a cure and new treatment options made readily available.

Limitations:

1. Difficulty finding donor matches
2. 15% still unsuccessful
3. risk of graft versus host disease and immune rejection of transplant
4. procedural complications such as infections and seizures
5. 5% undergoing this procure die.[8]

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