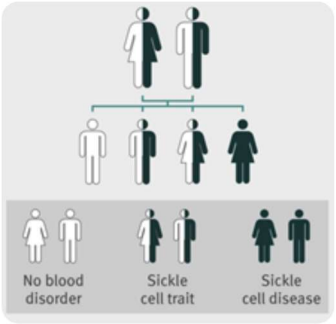
Sickle Cell Disease

Sickle Cell Disease (SCD) is an inherited disease that affects the production of haemoglobin, a protein in red blood cells that carries oxygen throughout the body. An inherited mutation causes people with SCD to not have normal, healthy adult haemoglobin in their red blood cells and instead have an abnormal haemoglobin called sickle haemoglobin. The result is rigid, crescent-shaped, or "sickled" red blood cells that can clump together in blood vessels and block the flow of blood throughout the body. Consequently, SCD can cause frequent episodes of severe pain, weakness, and other serious complications.

Hereditary Pathway
(for a couple, each carrying the sickle cell trait)



Sickle cell disease is a public health priority and a neglected health problem in sub-Saharan Africa, which carries approximately 80% of the global disease burden. The disease takes a heavy toll on children: it is estimated that approximately 1000 children in Africa are born with sickle cell disease every day and more than half will die before they reach the age of five.

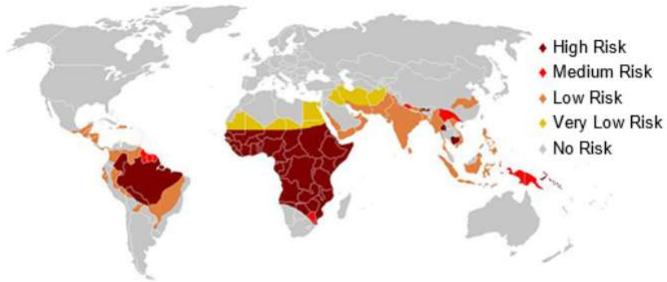
~80%
OF PEOPLE WITH SCD
live in sub-Saharan Africa[5]

~1000
CHILDREN IN AFRICA
are born with SCD every day[6]

>50%
OF CHILDREN WITH SCD
in Africa die before their fifth birthday due to preventable complications[8]

~14 million
NEWBORNS GLOBALLY
will be affected by SCD between 2010 and 2050[7]

Geographical Spread



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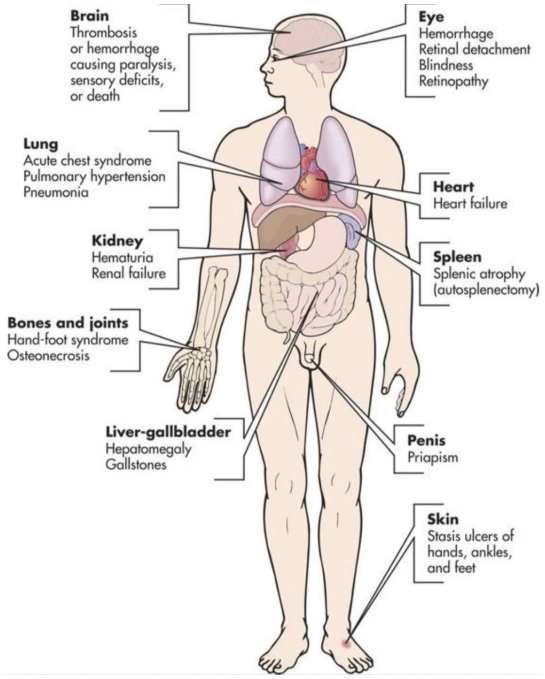
Complications



Primary Care - Responsibilities

- Early treatment of infections to prevent sepsis.
- Prescription of antibiotic prophylaxis
- Ensuring vaccinations are up to date.
- Early referral of pregnant women
- Reproductive advice including contraceptive advice, pre-conceptual testing and partner testing.
- Referral for psychological support and counselling
- Encourage treatment compliance.
- Patient education and self-management of mild painful episodes
- Support during transition and move to further education.

Areas of focus in primary care

- Infection control
- Vaccinations
- Antibiotic Prophylaxis
- Blood pressure monitoring
- Folic Acid
- Vitamin D deficiency
- Acute chest syndrome (ACS)
- Chronic sickle lung disease
- Pulmonary hypertension (PH)
- Renal problems (kidney issues)
- Iron overload
- Stroke

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Treatment

Health maintenance for patients with sickle cell disease starts with early diagnosis, preferably in the new-born period and includes penicillin prophylaxis, vaccination against pneumococcus bacteria and folic acid supplementation.
Treatment of complications often includes antibiotics, pain management, intravenous fluids, blood transfusion and surgery all backed by psychosocial support. Like all patients with chronic disease patients are best managed in a comprehensive multi-disciplinary program of care.

Therapies

1998 - Hydroxyurea

2017 - L-Glutamin (Endary)

2019 - Crizanlizumab (Adakveo)
Voxelotor (Oxbryta)

2020 - Gene editing
- CRISPR Cas 9

Complementary and Alternative Therapies

To reduce the discomfort due to pain the following options may be explored.
- Acupuncture
- Heat treatment
- Hydrotherapy
- Tens machines
- Calm relaxing music
- Rehydration

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The Science

Sickle cell disease is caused by a mutation in the haemoglobin-Beta gene found on **chromosome 11**. Haemoglobin transports oxygen from the lungs to other parts of the body. Red blood cells with normal haemoglobin (haemoglobin-A) are smooth and round and glide through blood vessels.

Foetal haemoglobin (HbF) is an "anti-sickling" haemoglobin that is present before birth in the red blood cells. After birth, the gene that makes foetal haemoglobin turns off, which mostly stops the production of foetal haemoglobin. More foetal haemoglobin in the blood can mean fewer episodes of sickling and pain.

Most therapies including gene editing are based on the simple aim to increase the production of HbF

Summary

sickle cell disease is
- a genetic mutation of Chromosome 11
- an inherited disease
- prevents oxygen from reaching important organs of the body
- hydroxyurea is a drug that increases HbF

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