Living with and Managing Sickle Cell and Thalassaemia
Please note, people's experience of an illness differs, and some people might experience milder symptoms despite having the same type of sickle cell or thalassaemia genotype.

Sickle Cell
The most common symptom of sickle cell is the severe pain known as a '**crisis**' which can require strong painkillers such as morphine. Anaemia results as the sickle cells get destroyed and over time organ damage can result. Key aspects of managing the condition include pain killers, blood transfusions, Folic acid and penicillin. Individuals should try to avoid crisis triggers such as infections, dehydration, cold temperatures and stress and maintain a healthy diet which has good sources of iron.

Thalassaemia
Living with thalassaemia involves ongoing medical treatment, such as regular blood transfusions and chelation therapy to manage iron overload. Key aspects of managing the condition include being compliant to treatment, maintaining a healthy diet, exercising regularly, avoiding infections, and staying up to date with vaccinations. Adhering to treatment schedules is crucial to prevent complications like organ damage and fatality.

Cure for Sickle Cell or Thalassaemia
The primary cure for sickle cell or transfusion-dependent β-thalassemia are bone marrow or stem cell transplants. These transplants replace a person's faulty blood-forming stem cells with healthy ones from a donor but require a close match and carry risks. A new one-off gene therapy cure known as Casgevy or Exa-cel now approved by the NHS uses CRISPR-technology to edit a patient's own stem cells to produce healthy haemoglobin, offering a potential cure. It is important to note a person who is cured with the above treatments can still pass on the thalassaemia or sickle cell gene to any offspring they have.

Tests that can help 'at-risk' Couples
Pre-implantation genetic testing (PGT) is available in the UK and helps couples who are at risk of having a child with a serious genetic condition. It is a special type of in vitro fertilisation (IVF) in which embryos are created outside of the body and are tested for the genetic condition in the family. Only embryos which are not affected by the genetic condition are placed into the womb, ensuring the baby is not born with the condition.

Non-invasive Prenatal Testing (NIPT) (unlike CVS and amniocentesis) is not yet available in UK as a prenatal diagnostic test for sickle cell or thalassaemia as its accuracy still needs validation. It involves assessment of fragments of DNA called cell free DNA in the maternal blood. In some cases, NIPT can be performed in pregnancy for e.g. Down's syndrome.

Where to Get Help and Advice

Sickle Cell Society
54 Station Rd
London NW10 4UA
T: 020 8961 7795
W: www.sicklecellsociety.org
E: info@sicklecellsociety.org
Facebook: Sickle Cell Society UK
Instagram / X: @SickleCellUK

UK Thalassaemia Society
19 The Broadway
London N14 6PH
T: 020 8882 0011
W: www.ukts.org
E: office@ukts.org
Instagram / X: @ukts2025

Useful Links
https://www.sicklecellsociety.org/supportgroups/
https://www.sicklecellsociety.org/living-with-sickle-cell-or-beta-thalassaemia-trait-summary/
https://youtu.be/TMh3WsKv33c
https://www.sicklecellsociety.org/resource/its-in-our-genes/

Sickle Cell Society
www.sicklecellsociety.org

Sickle Cell
https://www.nhs.uk/conditions/sickle-cell-disease/carriers/
https://www.nhs.uk/conditions/sickle-cell-disease/

UK Thalassaemia Society
https://ukts.org/

Thalassaemia
https://ukts.org/alpha-thalassaemia/
https://ukts.org/b-thalassaemia-major/
https://www.nhs.uk/conditions/thalassaemia/carriers/
https://www.nhs.uk/conditions/thalassaemia/

PRECONCEPTION TESTING FOR SICKLE CELL AND THALASSAEMIA

The Sickle Cell Society and UK Thalassaemia Society are national charities that have produced this leaflet particularly for individuals of child-bearing age who have not yet had children. It will give you current information on testing for sickle cell and thalassaemia genes **before** you and your partner are expecting a baby. It also contains information on support after testing and on living with the conditions. **Preconception testing** is a specific blood test carried out **before pregnancy** to establish whether you carry any genes for sickle cell or thalassaemia.

The United Kingdom has an established NHS Sickle Cell and Thalassaemia Screening Programme that provides the option of screening for parents-to-be (antenatal) and newborn babies. The Programme's mission is to:

- Support people to make informed screening choices during pregnancy & before conception
- Improve infant health through prompt identification of affected babies
- Provide high quality and accessible care for individuals with sickle cell disease and thalassaemia
- Promote greater understanding and awareness of sickle cell and thalassaemia and the value of screening

Sickle Cell and Thalassaemia

What is Sickle Cell?
Sickle cell disease or disorder is a collective term describing a number of different but similar genetically inherited blood conditions in which a mutated (i.e. changed) form of haemoglobin (the protein that carries oxygen and gives blood its red colour) causes the normally round blood cells to change to a sickle or 'half-moon' shape after giving up their oxygen, blocking their flow around the body. This results in severe pain ('crisis'), anaemia and long-term organ damage.

Your haemoglobin genotype shows the combination of the two genes you have inherited for making haemoglobin. The most common combination is **HbAA** which involves the two usual haemoglobin genes. Other genotype examples are **HbSS** (sickle cell anaemia), **HbSC** (Sickle Haemoglobin C disease) and **HbSβthal** (Sickle beta-thalassaemia), indicating that two unusual genes have been inherited for making haemoglobin and resulting in the three common types of sickle cell disease.

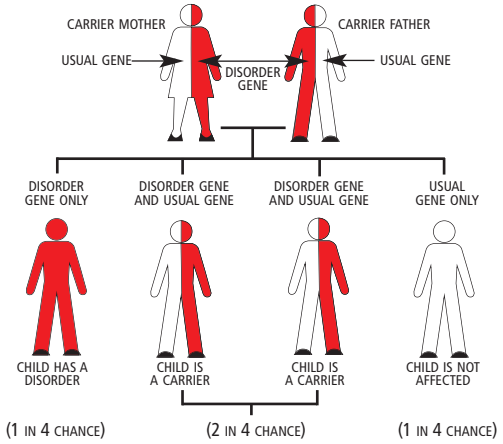
Who does Sickle Cell affect?
Sickle cell **mainly** affects people who originate from Africa, The Caribbean, Middle East, Mediterranean and India. This is because the sickle gene mutation is thought to have been a protective measure against malaria. Sickle cell is less common in white Europeans. In the United Kingdom (UK), approximately 18,500 people are living with sickle cell and almost 300 babies are born with the condition each year.

What is Thalassaemia?
Thalassaemia is a genetically inherited blood disorder that affects the body's ability to produce haemoglobin and healthy red blood cells. The body makes less haemoglobin than normal. This can lead to severe anaemia, requiring regular blood transfusions and long-term health care.

Who does Thalassaemia affect?
It **mainly** affects people of Mediterranean, south Asian, southeast Asian and Middle Eastern origin. In the UK, approximately 2770 people are living with thalassaemia and around 30 babies are born with the condition each year. If your haemoglobin genotype is **β^0/β^0** it means you have Beta Thalassaemia Major also known as Transfusion Dependent Thalassaemia which is a very serious form of thalassaemia.

Sickle Cell and Thalassaemia Inheritance and 'Carrier' Status
HbAS, **HbAC**, or **HbAβ^0** are examples of genotypes that indicate you are sickle cell, haemoglobin C or thalassaemia 'carrier' status (also known as 'trait') respectively. You **do not** have the condition but can have an affected child with someone who is also a carrier.
How Can Sickle Cell or Thalassaemia be Inherited?
If both parents are carriers of an unusual or 'disorder' gene as indicated below, for example haemoglobin genotype **HbAS** or **HbAβ^0** then each time they're expecting a child, there is a:

- 1 in 4 chance (25%) the child could inherit the disorder. This could be either sickle cell anaemia (**HbSS**) or Beta Thalassaemia Major **β^0/β^0** depending on if the parents' haemoglobin genotype were **HbAS** or **HbAβ^0** respectively.
- 2 in 4 chance (50%) the child could be a sickle cell carrier (**HbAS**) or thalassaemia carrier (**HbAβ^0**)
- 1 in 4 chance (25%) the child could inherit the usual haemoglobin genotype (**HbAA**).



It is important to note that the above example is for individuals who are carriers only and that the inheritance probabilities (i.e. chances) are different if parents have different haemoglobin genotypes such as **HbAA**, **HbSS** or **β^0/β^0** for example. Resources listed at the back of this leaflet will show you the inheritance patterns for other genotypes such as **HbAC** carriers or alpha thalassaemia. Alpha plus thalassaemia is harmless and alpha zero thalassaemia may have more serious implications. Couples with at least one unusual gene each are known as '**at risk**' and can be fast tracked in the Screening Programme pathway.

Why it is important to get tested?
It is important to get tested for your genotype for the following reasons:

- Informed reproductive choices – knowing if you're a carrier enables individuals to work out the risk of their baby inheriting a serious genetic condition
- You may wish to tell close family members so they might test too
- Health information, as being a carrier can have implications (e.g. during general anaesthetic, high altitude, extreme exercise)
- Understanding if you have any symptoms of carrier status and taking actions to help

What is the screening test used?
If you want to be tested for your haemoglobin genotype, you will need to have a special blood test called *haemoglobin electrophoresis* and a full blood count.

Where can you get tested?
Before pregnancy or planning a baby, you can ask your GP for the screening blood test, particularly if you know of close family members who have sickle cell or thalassaemia condition or are carriers of the gene. In **some** parts of the country there are NHS Sickle Cell and Thalassaemia centres (https://www.sicklecellsociety.org/supportgroups/) and they might also arrange preconception testing for you if you live in their catchment area. Please note that some GPs **might** charge for the test, particularly if you are not entitled to NHS services or are not at risk of being a carrier.

Help with getting tested
If you're having difficulty getting a screening blood test, please contact the Sickle Cell Society or UK Thalassaemia Society Helplines who might be able to advocate on your behalf.

What next if you are a 'carrier'?
If you are a 'carrier' of sickle cell or thalassaemia (i.e. have the 'trait') you can get information from the Sickle Cell Society or UK Thalassaemia Society respectively. You can also get counselling support from the nurses at an NHS Sickle Cell and Thalassaemia centre if there's one in your area. Below is the link to a very informative screening video podcast with panellists from the Societies, NHS Sickle Cell and Thalassaemia Screening Programme and service users: https://youtu.be/TMh3WsKv33c
See contact details for the Societies and other useful website links at the back of the leaflet.