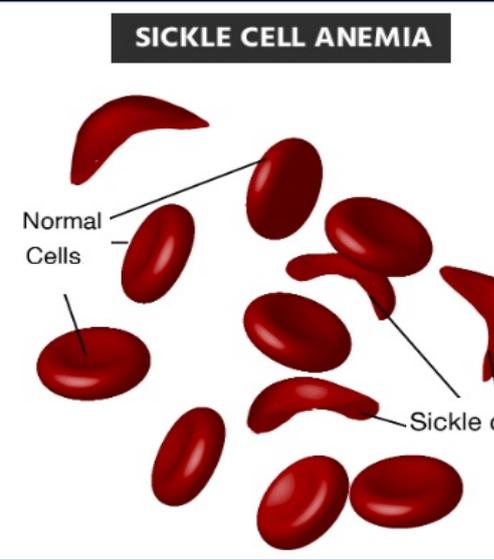
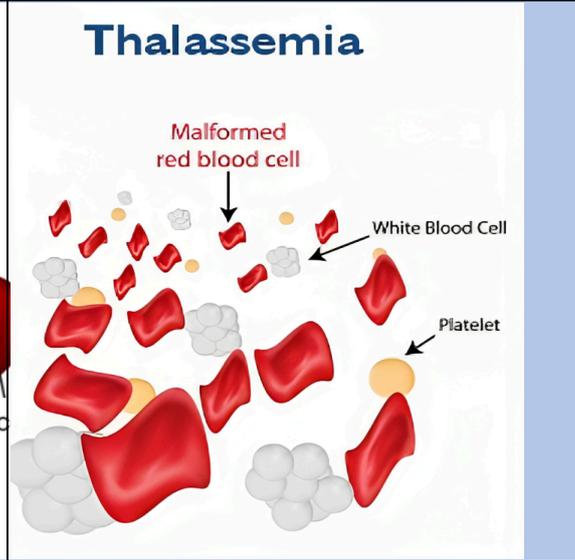


Gene Therapy for Sickle Cell Anaemia

Why in news?

The first therapy based on gene editing technology Crispr-Cas9 for [sickle cell disease](#) and thalassaemia has been approved in UK.

What is sickle cell anaemia and thalassaemia?

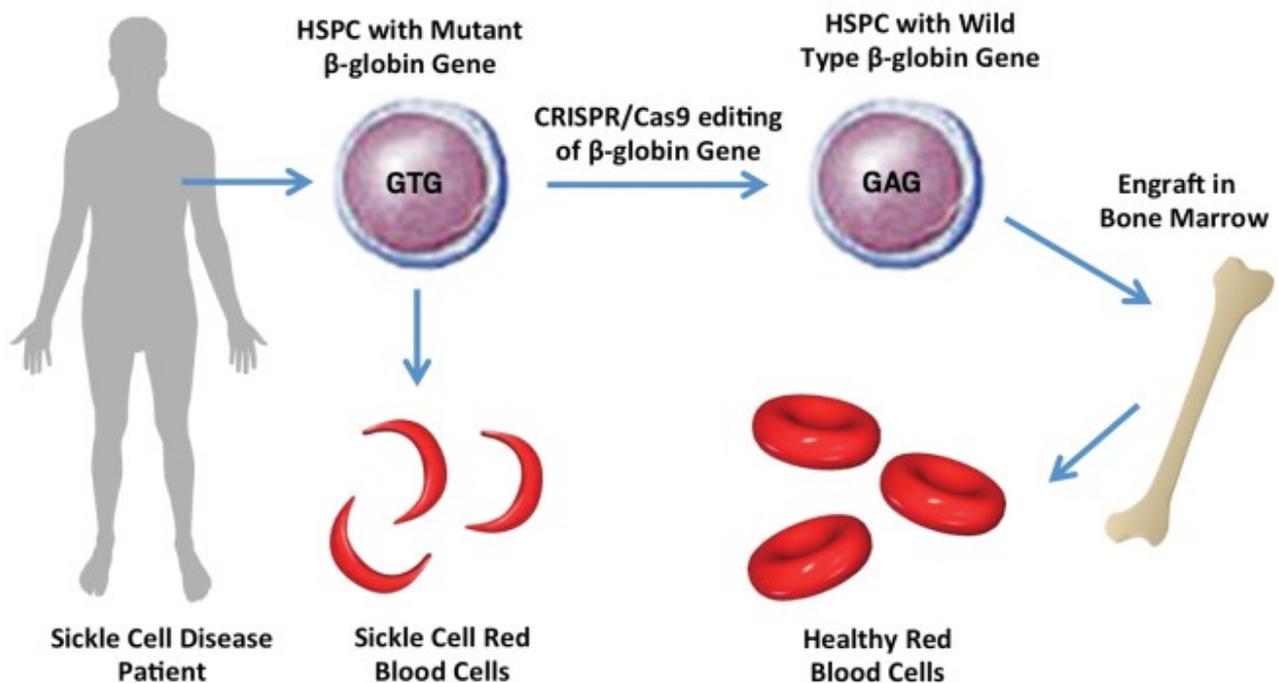
| About | Sickle Cell Anaemia | Thalassaemia |
|------------------------------------|--|---|
| Disease | An inherited blood disorder where people who inherit a pair of genes from both parents experience symptoms like severe anaemia. | |
| Effect on haemoglobin chain | Caused by a mutation in the haemoglobin- β gene found on Chromosome 11 affecting <i>only the beta chain</i> | Production of <i>either the alpha or beta chains</i> is reduced resulting in either alpha-thalassaemia or beta-thalassaemia |
| Haemoglobin production | Mutation in haemoglobin chains makes them into a <i>crenate shape</i> under low oxygen level | Caused by reduced production of haemoglobin chains |
| Effects | Pain, fever, infection, stroke and organ damage | Fatigue, shortness of breath, irregular heartbeats and need blood transfusions throughout their life |
| Status in India | An estimated 30,000-40,000 children in India are born with the disorder every year. | India has the largest number of children with thalassaemia (about 1-1.5 lakh). |
| |  <p>SICKLE CELL ANEMIA</p> <p>Normal Cells</p> <p>Sickle c</p> |  <p>Thalassaemia</p> <p>Malformed red blood cell</p> <p>White Blood Cell</p> <p>Platelet</p> |
| Treatment | Treated by blood transfusions, iron supplements, or stem cell transplants. | |

GLOBAL BURDEN OF SICKLE CELL DISEASE



What is Casgevy?

- Casgevy is the 1st licensed therapy in the world based on the gene editing technology [Crispr-Cas9](#).
- **Apheresis** - It is a *one-time treatment* for which the doctor has to first collect blood stem cells from the bone marrow using a process called **apheresis** (filtering out the blood for different components).
- The cells are then sent to the manufacturing site where it takes about 6 months for them to be edited and tested.
- **Gene editing** - The therapy uses the patient's own blood stem cells, which are precisely edited using **Crispr-Cas9**.
 - So far, the only permanent treatment has been a bone marrow transplant, for which a closely matched donor is needed.
- A gene called **BCL11A**, which is crucial for switching from foetal to adult haemoglobin, is targeted by the therapy.
 - Foetal haemoglobin (naturally present in everyone at birth), does not carry the same abnormalities as adult haemoglobin.
- The therapy uses the body's own mechanisms to start producing more of foetal haemoglobin, alleviating the symptoms of the two conditions.
- **Side effects**- They are similar to those associated with autologous stem cell transplants, including nausea, fatigue, fever and increased risk of infection.



What are the pros and cons of this treatment?

| Significance | Challenges |
|---|--|
| Efficacy- It restores haemoglobin production and alleviates symptoms in most patients. | Limited authorization- It is currently approved in the UK only and is being reviewed by other regulatory bodies. |
| Pain reliever- It reduces the need for blood transfusions and pain crises in the patients. | Health inequity- It is expensive, thereby limiting the accessibility in poor countries. |
| Reliable- No serious safety concerns were reported, but long-term effects are still being monitored. | Inaccuracy- There are concerns with potential off targets effects of CRISPR editing, which could cause unwanted changes in other parts of the genome. |

References

1. [Indian Express- Sickle cell breakthrough](#)
2. [Live Science- World's first CRISPR therapy has been approved](#)