

Sickle Cell Anemia: A Review

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Background

- monogenic disease, mutated hemoglobin S (HbS)
- Alters morphology of red blood cells (RBC) into a “sickle” shape (Figure 1)
- Abnormal RBC shape impedes blood flow and leads to inability to carry oxygen
- Occurs in 1/365 African-American births in the U.S

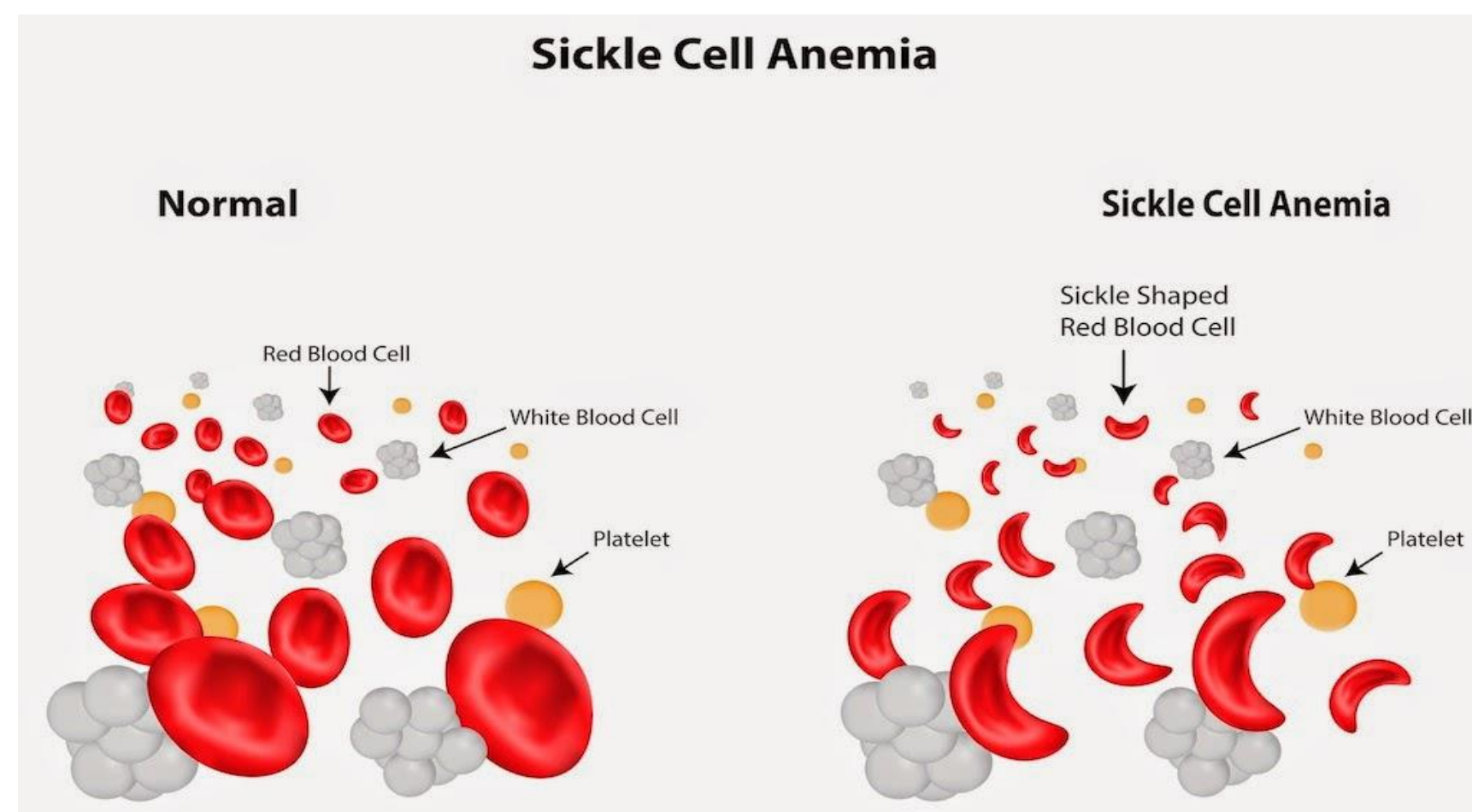


Fig 1. Comparison of Normal Red Blood Cell Morphology vs. Sickle Shaped Red Blood Cell

Genetics

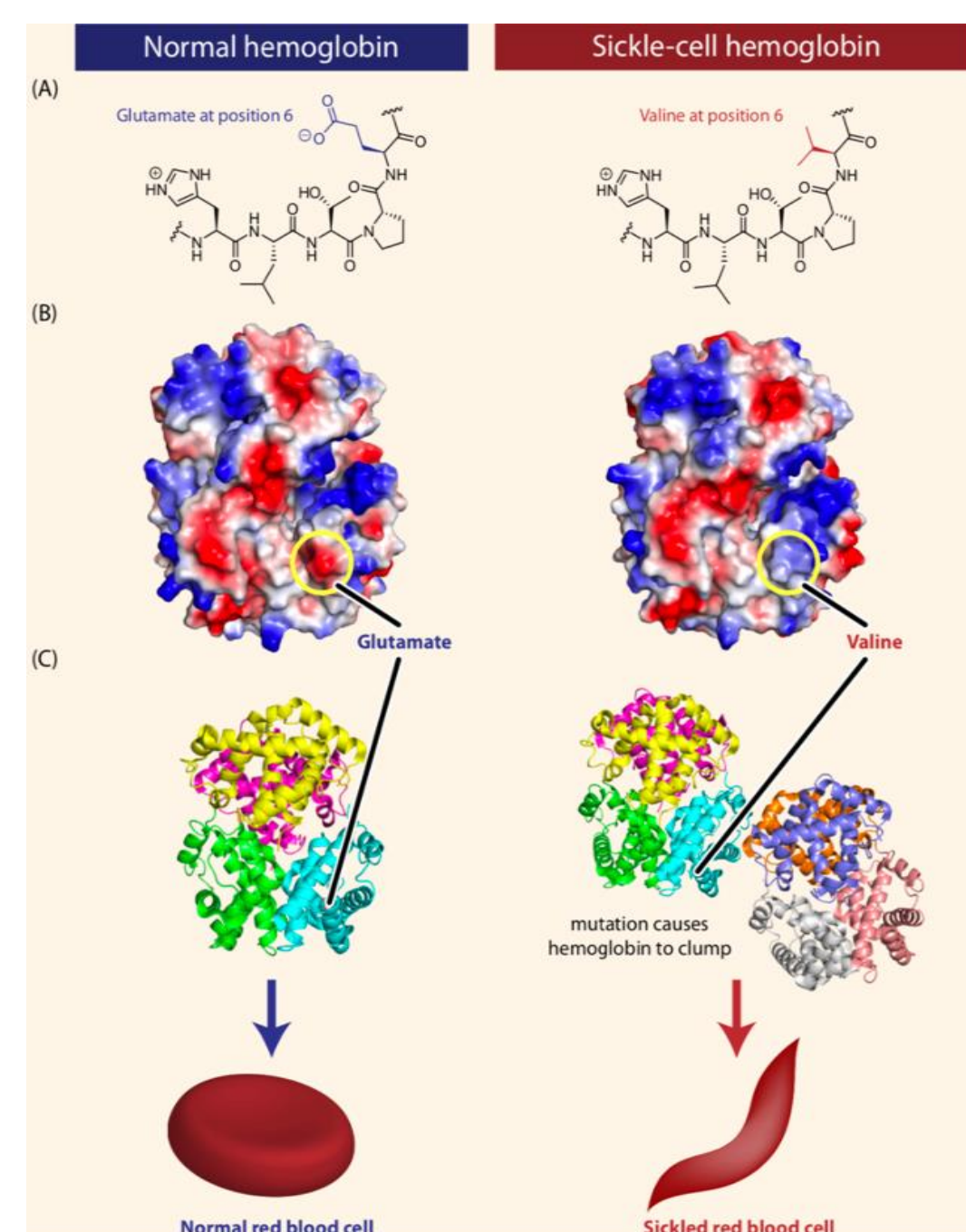


Fig 2. Mutated Hemoglobin due to Single Amino Acid Substitution

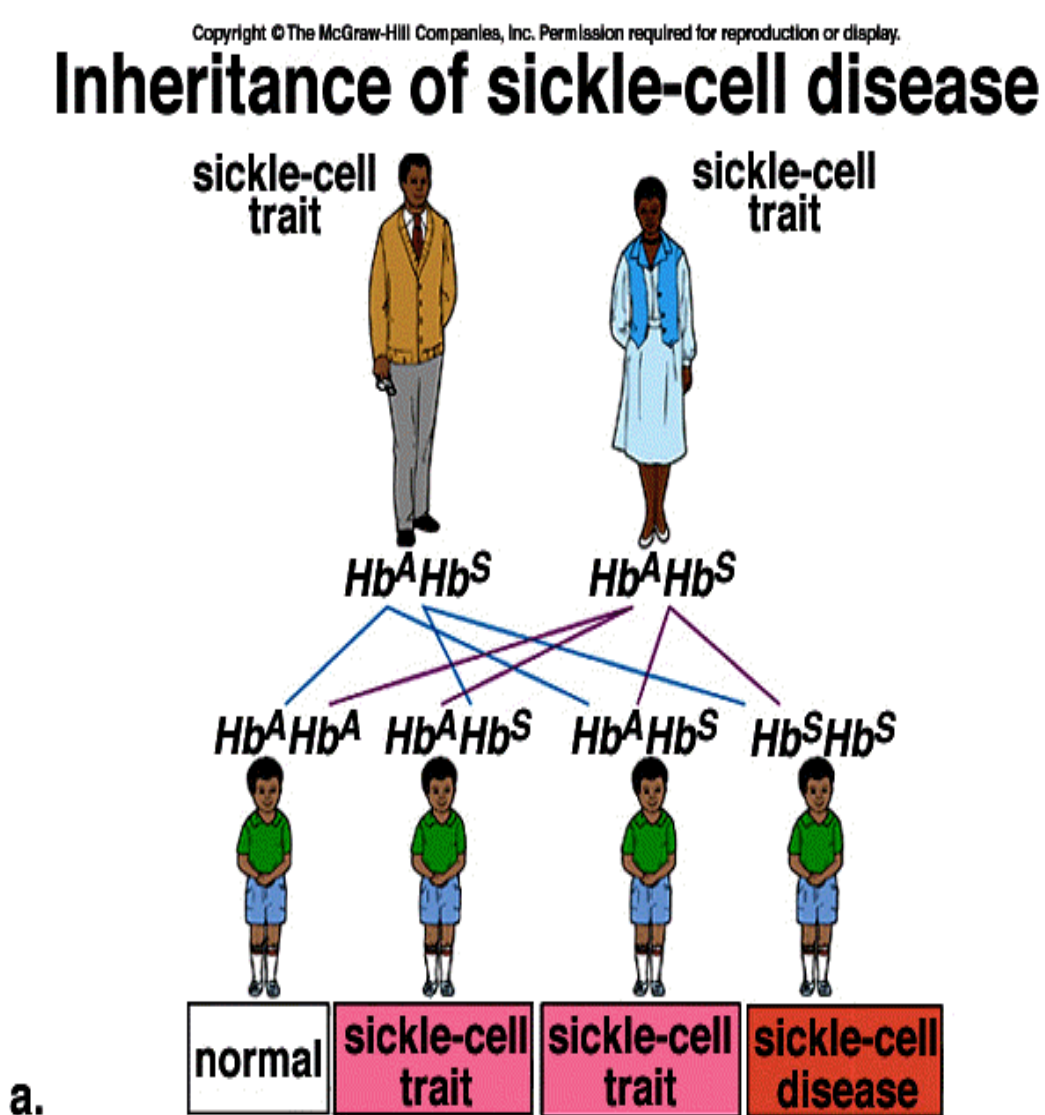


Fig 3. Autosomal Recessive Inheritance of Sickle-cell Disease

Symptoms

- Vasooclusive crisis
 - Stroke
 - Chronic vs acute pain
- Nervous & pulmonary system

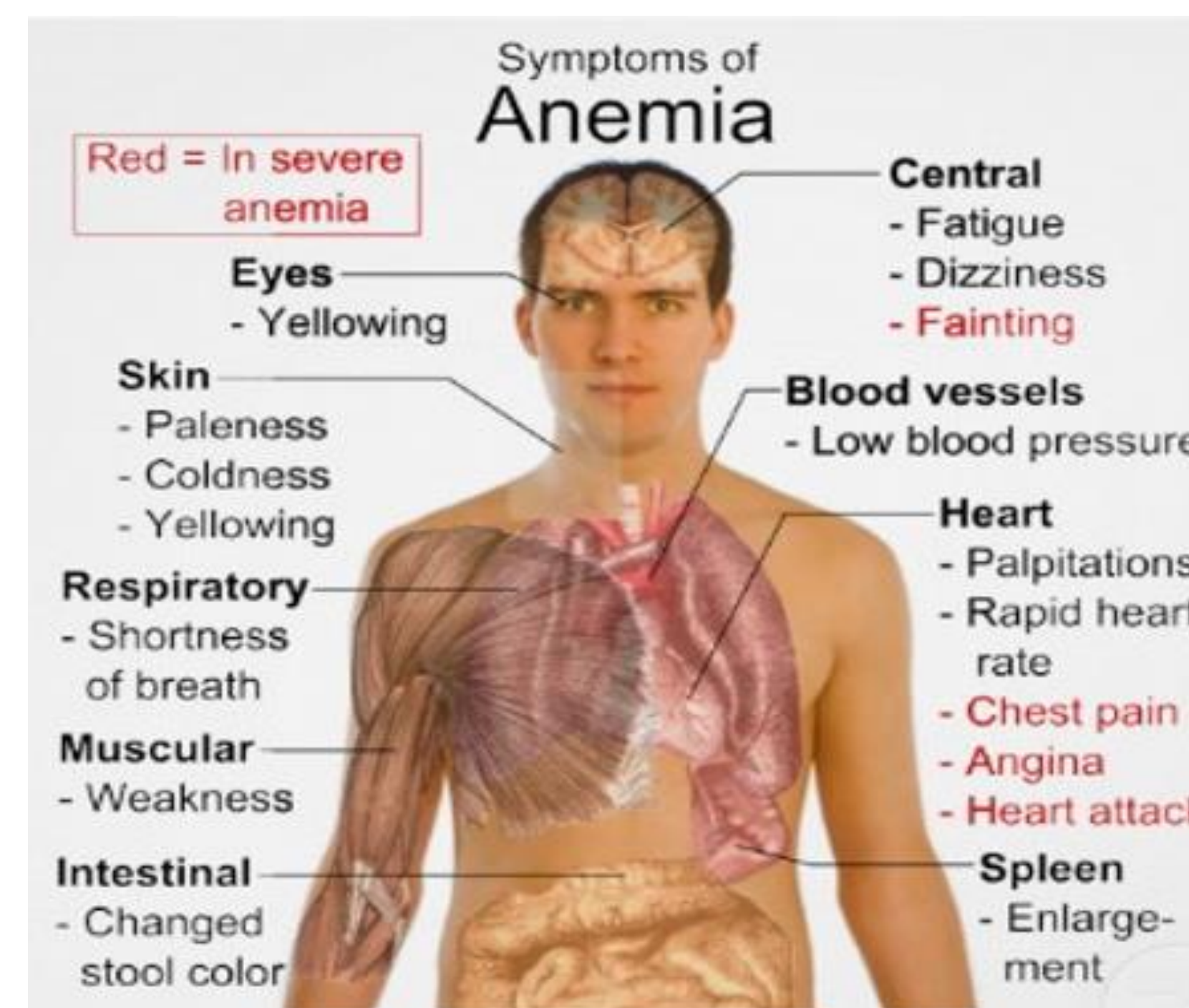


Fig 4: Bodily Symptoms of Sickle Cell Anemia

Case Study: Stroke

According to a study done by Domingos et. al.

- SOD2 Val16Ala
- Superoxide dismutase (SOD)

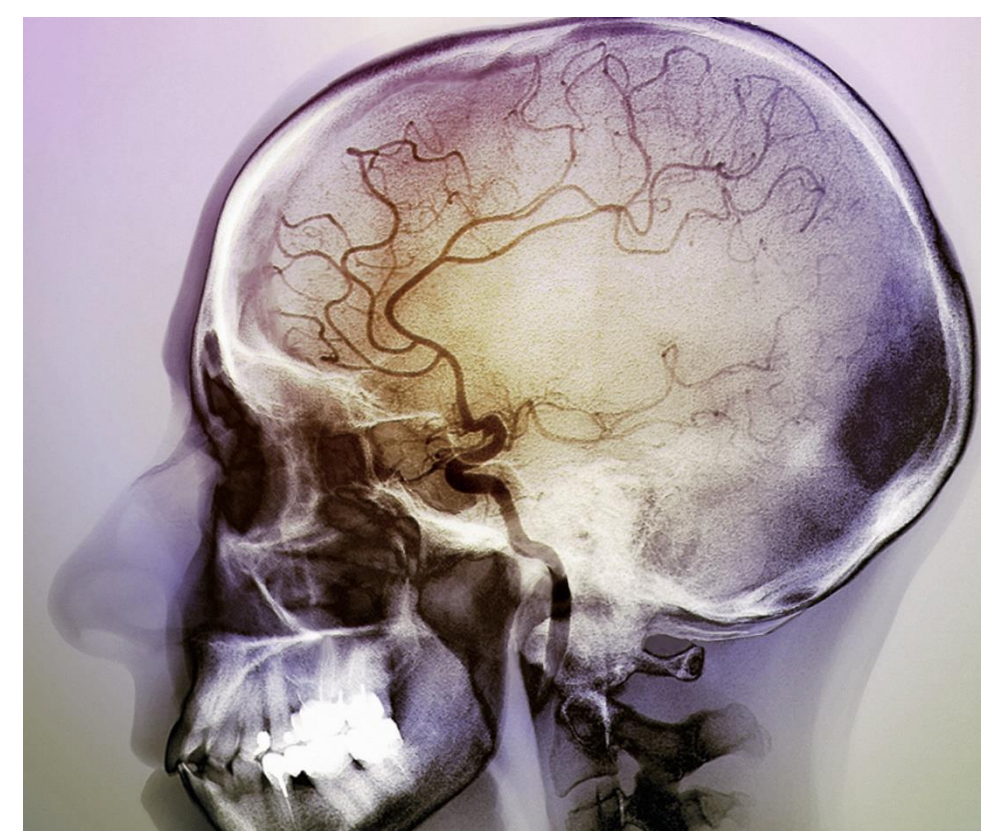


Fig 5: Cranial Image of Ischemic Stroke Patient



Fig 6: Sickle cell patient receiving gene therapy

Current and Future

- hydroxyurea/opioids
 - Increases hemoglobin gamma to replace mutated beta hemoglobin
- transfusion therapy
 - Acute pain (acute chest syndrome)
- stem cell transplant
 - Improved pain intensity, physical function and pain impact
- gene therapy
 - Completely replace mutation

References

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