



# Role of Technology in Managing Sickle Cell Anemia's Impact on Quality of Life

### Woeklbbier Tangie<sup>\*</sup>

Department of Genetics, University of Madras, Chennai, India

## DESCRIPTION

Sickle cell anemia stands as a poignant example of how genetic mutations can extreme impact human health. This inherited blood disorder, affecting millions worldwide, offers a compelling case study in molecular pathology. By delving into the genetic underpinnings of sickle cell anemia, uncover not only the scientific complexities but also the personal and societal dimensions of living with and managing this condition.

At its core, sickle cell anemia is caused by a single-point mutation in the gene encoding the beta-globin chain of hemoglobin. This mutation leads to the substitution of a glutamic acid residue with valine, resulting in the formation of abnormal hemoglobin known as hemoglobin S. Under conditions of low oxygen levels, HbS molecules can polymerize, distorting red blood cells into characteristic sickle shapes. These sickled cells are rigid, sticky, and prone to clumping, causing blockages in blood vessels and leading to the attribute symptoms of the disease.

#### Clinical manifestations of sickle cell anaemia

The clinical manifestations of Sickle Cell Anemia (SCA) are diverse and can range from mild to severe. Patients often experience recurrent episodes of pain due to vase-occlusive crises, where sickled red blood cells obstruct blood flow and cause tissue damage. Chronic anemia results from the premature breakdown of red blood cells, leading to fatigue and pallor. Additionally, organ damage can occur over time, affecting the spleen, kidneys, lungs, and brain.

**Epidemiology and global impact:** Sickle cell anemia predominantly affects individuals of African, Mediterranean, Middle Eastern, and Indian ancestry. It is estimated that approximately 100,000 Americans and millions worldwide are living with this condition. Regions with high frequencies of the sickle cell trait also face significant health challenges, including increased susceptibility to malaria.

**Diagnostic and therapeutic advances:** Advances in molecular diagnostics have revolutionized the detection and management of sickle cell anemia. Techniques such as DNA analysis and

hemoglobin electrophoresis allow for precise identification of Hobs and other hemoglobin variants. Early diagnosis through new-born screening programs enables prompt intervention and comprehensive care, including prophylactic antibiotics, vaccination protocols, and hydroxyurea therapy to reduce complications and improve quality of life.

#### Advances in treatment and management

Despite the challenges, advances in medical research have led to improved treatments and management strategies for SCA. Early diagnosis through new-born screening programs allows for early intervention and comprehensive care planning. New therapies, including gene therapy and novel medications, are being developed to address the underlying genetic cause of SCA and potentially offer curative treatments in the future.

#### Advocacy and awareness

Raising awareness about SCA is essential to improving the quality of life for individuals and families affected by the disease. Advocacy efforts aim to increase research funding, improve access to healthcare services, and promote policies that support equitable care and resources for all individuals with SCA. Community support groups and patient advocacy organizations play a vital role in providing information, education, and peer support to empower individuals and families affected by SCA.

**Challenges and future directions:** Despite progress, significant challenges remain in the treatment landscape for sickle cell anemia. Curative options such as bone marrow transplantation and emerging gene therapy approaches hold potential but are not universally accessible. Addressing healthcare disparities, improving access to specialized care, and advancing research into novel therapies are critical to enhancing outcomes for individuals living with this complex genetic disorder.

Sickle cell anemia exemplifies the intricate exchange between genetics, pathology, and clinical care. By seperating its genetic basis and understanding its clinical manifestations, we deepen our appreciation for the complexities of genetic disorders. Moving forward, a concerted effort is needed to translate scientific discoveries into tangible improvements in diagnosis,treatment, and ultimately, the quality of life for patients and families affected by sickle cell anemia.

Correspondence to: Woeklbbier Tangie, Department of Genetics, University of Madras, Chennai, India, E-mail Tangie@hotmail.com

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