Short Communication

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# The Management of Common Genetic Blood Disorders

David Gokman*

Department of Molecular Genetics, Weizmann Institute of Science, Israel

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***Corresponding author:**
David Gokman

Gdavid@hotmail.com

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## Introduction

Genes are the blueprint for our bodies, directing our physical and functional characteristics and defining who we are! Let's take a moment to remember that these genes are sometimes responsible for genetic anomalies or illnesses that can have an impact on an individual's health.

When one or more genes are mutated, these genetic disorders develop. A gene mutation can be inherited from one or both parents, or it might be acquired throughout your lifetime. If left untreated, these illnesses can lead to a lifelong battle for many people.

Early diagnosis, on the other hand, can help clinicians plan quick treatment and management choices to improve the quality of life of those who are affected. As a result, it's critical to undergo a full genetic screening of the kid soon after birth to rule out any hidden abnormalities that aren't visible at birth.

## What exactly is Sickle Cell Anaemia, and what causes it?

Sickle cell anaemia is a kind of sickle cell illness in which the protein haemoglobin, which transports oxygen throughout the body, is impaired, putting adequate blood flow at risk. It is an inherited blood disorder caused by mutated genes that are passed down via families.

Normally, red blood cells are disc-shaped and flexible enough to pass through blood channels freely. When a person has sickle cell disease, however, their red blood cells are typically crescent or "sickle" shaped. These cells can impede blood flow to the rest of your body since they can't easily pass through blood arteries.

## Symptoms

Sickle cell disease symptoms commonly occur in early childhood, around the age of 5-6 months. Anaemia (low red blood cell count), infections, swelling in the hands and feet, and pain episodes are all symptoms of this condition. Symptoms differ from one person to the next. Some people only have minor symptoms, while others are routinely admitted to hospitals for more significant problems.

## Diagnosis & Treatment Landscape in India

Couples who know they have the condition or who are 'carriers of the defective gene' should seek genetic counselling and testing to avoid passing the disorder on to their offspring. This understanding aids in making the best reproductive choices for a healthy pregnancy and kid.

Because sickle cell disease is a chronic illness, sufferers must take medications for the rest of their lives. The medications aren't a cure for the ailment, but they can help control the symptoms that come with it. Blood transfusions may be required on a regular basis. A stem cell transplant may be performed depending on the severity of the disease and the availability of donor blood stem cells.

Recent research also reveals that stem cell transplantation are becoming a viable therapy option for Sickle Cell Anaemia. Many stem cell transplants as a curative treatment for sickle cell anaemia have been effective in India's healthcare market, which is said to be as advanced as its western counterparts.

## What Are the Different Forms Of Thalassemia?

Thalassemia is a genetic blood condition in which the body produces insufficient haemoglobin. It is caused by a faulty gene involved in the manufacture of haemoglobin. The percentage of haemoglobin that is not created by default in the body is referred to as 'alpha' or 'beta' in thalassemia

When the body's red blood cells don't have enough haemoglobin, they don't work correctly and don't last as long, resulting in a significantly decreased quantity of healthy red blood cells in the

bloodstream. This disorder affects children who inherit the faulty gene from one or both parents.

However, if the child inherits only one defective gene, then the child has thalassemia minor and is a carrier. This fact, thus, underlines the importance of genetic counselling and prenatal tests in carriers

## Common Symptoms of Thalassemia

Not everyone who is afflicted will have symptoms. In fact, some symptoms may begin to develop later in childhood or adolescence. People with less severe diseases may not realise they have anaemia until they are identified with modest symptoms such as fatigue, yellow skin, delayed growth, or iron overload.

## Diagnosis

In the globe, India has the highest number of children with thalassemia major. With 1 to 1.5 lakh children and about 42 million beta-thalassemia carriers, the figure becomes even more astonishing. Within the first two years of life, the majority of children with moderate to severe thalassemia display symptoms

Anemia and the presence of aberrant haemoglobin can be detected through blood tests. Advanced genetic tests can also be used to diagnose the severity and kind of condition by analysing altered genes. Additionally, the presence of an enlarged spleen may play a role in the diagnosis.

Couples who are thinking about starting a family or who are pregnant can get a genetic carrier screening to see if they are at risk of passing on thalassemia or other genetic diseases to their children.

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