وزارة الصحة
Ministry of Health

Genetic Blood Disorders

Genetic blood disorders are a group of diseases that are passed down from parents to their children. Certain blood disorders are caused by abnormalities in the composition and components of red blood cells due to genetic defects, resulting in red blood cells that are unable to perform their normal functions. The main types of genetic blood disorders are thalassemia, and sickle cell anemia.

These disorders are passed down from parent to child through the genes carried on the chromosomes. When both parents have a genetic defect, there's a 25% chance that each child will be born with the disease. However, if one parent is healthy and the other parent is carrying the trait, then it's possible for the disease to get passed down to some children, making them carriers of the disease.

Genetic blood disorders in Saudi Arabia:

The prevalence of genetic blood disorders (sickle cell anemia and thalassemia) varies among the different regions of the Kingdom. The highest rates are recorded in the eastern and southern regions, while the rates are low in the central and northern regions of the Kingdom. According to the annual reports released by the Ministry of Health (Healthy Marriage Program) from 1425H till the end of 1430H, the recorded incidence of sickle cell anemia was 0.27%, whereas the incidence of thalassemia was 0.05%.

What is sickle cell anemia?

Sickle cell anemia is a genetic blood disorder that results in an abnormality in the oxygen-carrying protein hemoglobin found in red blood cells due to a genetic defect. In sickle cell anemia, the red blood cells become rigid and sticky and are shaped like sickles or crescent moons. These irregularly shaped cells can

get stuck in small blood vessels, which can slow or block blood flow and oxygen to parts of the body resulting in symptoms like severe pain, shortness of breath and others.

Symptoms:

- Periodic episodes of pain, called crises, in different parts of the body depending on the location where sickle-shaped red blood cells break apart and block microvessels. For example, pain can occur in pain can occur in the abdomen, joints, or one of the limbs.
- Chronic anemia.
- Frequent infections.
- Symptoms of malnutrition, short stature and delayed growth.
- Bone deformities.
- Lethargy and fatigue.

Complications:

Complications occur due to the breakdown of red blood cells, and the blockage of microvessels, and these complications include:

- Heart attacks and strokes.
- Increased infections.
- Jaundice .
- Gallstones.
- Vision disorders and blindness.
- Delayed growth in children.

Treatment:

- Treatment for sickle cell anemia is usually aimed at avoiding crises, relieving symptoms and preventing complications, as well as improving the patient's ability to cope with the disease.

- Sickle cell anemia patients are in need of continual care to prevent recurrence of complications and deterioration of their health status.
- Sickle cell anemia patients are given folic acid supplements to stimulate the production of red blood cells.
- To relieve pain during a sickle crises, the patient is treated with pain-relieving medications, and fluids.
- In certain cases, patients may respond to over-the-counter pain-relieving medication, while others may require stronger prescription medications, such as morphine and meperidine under medical supervision.
- Treatment with hydroxyurea may reduce the frequency of painful crises and respiratory symptoms such as chest pain and shortness of breath.
- Routine and seasonal vaccines such as seasonal influenza vaccines are extremely important to prevent infections, especially for children with sickle cell anemia because their infections can be severe.
- Patient does not need blood transfusions on a regular basis, as well as in emergency crises.
- Patient's eyes may be affected, which can lead to partial or complete blindness; therefore, sickle cell anemia patients should make sure to visit an eye doctor on a regular basis.
- Bone marrow transplant, also known as stem cell transplant, can be used to treat sickle cell anemia.

What is Thalassemia?

Thalassemia, also known as Mediterranean anemia, is a genetic blood disorder characterized by less hemoglobin and fewer red blood cells in the body than normal. This condition is very common in the Mediterranean region, and it occurs due to a defect in the genetic makeup of hemoglobin.

Types and Symptoms of Thalassemia:

1. Alpha Thalassemia:

Hemoglobin consists of four chain genes of the alpha type, two from the father and two from the mother. Alpha thalassemia occurs when one or more of the four alpha chain genes fails to function. The severity of the condition depends on how many of these chains are defective. If only one chain gene is defective then the patient does not experience any signs or symptoms of thalassemia. A person with this condition is called a "silent carrier".

If two alpha chain genes are defective, then the patient develops what is known as minor alpha thalassemia. A person with this condition exhibits mild symptoms, and it may not be detected with a blood test.

If three alpha chain genes are defective, the patient develops severe anemia, and the symptoms range from moderate to severe. This condition is called Hemoglobin H disease. A blood test for this condition shows small deformed red blood cells. A person with this condition may suffer from an enlarged spleen, bone deformities due to its increased activity to compensate for damaged red cells. The patient needs to have blood transfusions to be able to live normally.

If four alpha chain genes are defective, then the patient develops what is known as major alpha thalassemia which can result in fetal death before or after birth.

2. Beta Thalassemia:

Hemoglobin consists of two beta chains, each of which is inherited from a parent, beta thalassemia occurs when a mutation occurs in these chains, and based on the number of mutations that affect the chains two types of thalassemia occur:

- **Thalassemia minor**: This condition occurs when only one of the beta chains is defective, and it results mild symptoms similar to those of a mild anemia that can be detected with a routine blood test.
- **Thalassemia major/Thalassemia**: This condition occurs when both beta chains are defective. The patient suffers from severe anemia, bone

deformity and spleen enlargement; the patient also needs regular blood transfusions to be able to lead a normal life.These symptoms do not appear when the baby is born; But begin to appear during the first two years of life.

Complications:

Patients of thalassemia major and hemoglobin H disease may develop many complications if they do not undergo proper treatment in a timely manner, such complications include:

- Slowed growth rates in children.
- Enlarged spleen and swollen abdomen.
- Bone deformities.
- Worsening anemia and a sense of fatigue and constant tiredness.

Treatment:

- Patients of thalassemia major and hemoglobin H disease require regular and ongoing care to prevent various complications of thalassemia such as recurrent infections and deterioration of health, which can lead to death if not treated properly.
- Patients need to undergo regular blood transfusions to increase the level of red blood cells in the blood, and avoid anemia.
- Patients are given folic acid supplements to stimulate the production of red blood cells.
- Some thalassemia patients may need to undergo chelation therapy to balance the rate of iron accumulation due to persistent red cell breakage, which can be done with the help of a medication called Desferal.
- Patients of thalassemia major may need to undergo a splenectomy to to remove the spleen.
- Bone marrow transplant can be used to treat patients who are diagnosed early before developing complications.

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Prevention of Genetic Blood Disorders:

Undergoing a comprehensive premarital screening and consulting a genetic counselor can help reduce the odds of transmitting genetic blood disorders to future generations. Premarital screenings can reveal if the couples who are planning to get married have any genetic defects or if they are asymptomatic carriers of certain diseases.

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