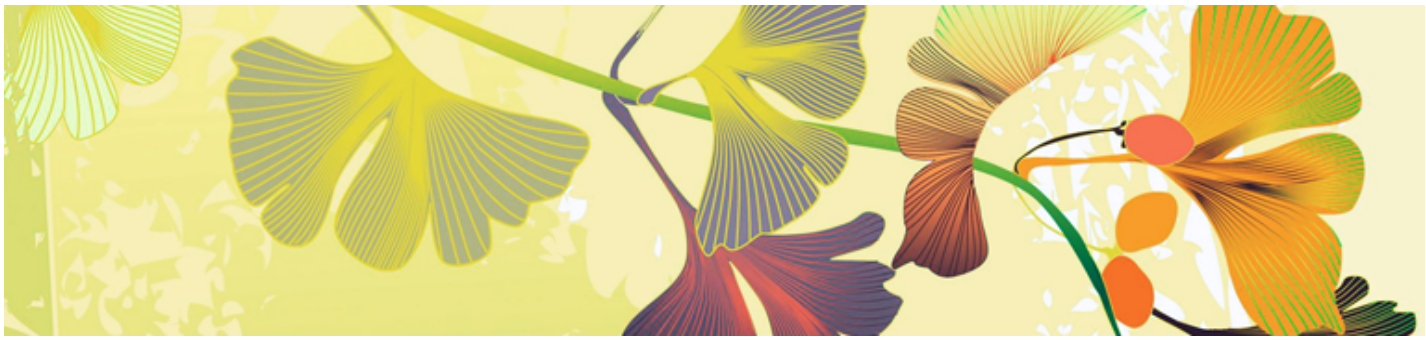

Thalassemia地中海性貧血(英文)

Definition

Thalassemia is a blood disorder passed down from family blood line; such an inherited disease is abnormal hemoglobin for the protein in red blood cell carries oxygen that destroys red blood cells and anemia.

Causes

Hemoglobin is made of two proteins: Alpha globin and beta globin. Thalassemia occurs when there is a defect in a gene that helps control production of these proteins.

There are two main types of thalassemia :

• Alpha thalassemia occurs when a gene or genes related to the alpha globin protein are missing or changed (mutated).

• Beta thalassemia occurs when similar gene defects affect production of the beta globin protein.

Alpha thalassemias occur most common in persons from southeast Asia, Middle East, China, and in those of African descent.

Beta thalassemias occur in persons of Mediterranean origin, and to a lesser extent, Chinese, other Asians, and African Americans.

There are many forms of thalassemia. Each type has many different subtypes. Both alpha and beta thalassemia include the following two forms :

- Thalassemia major
- Thalassemia minor

You must inherit the defective gene from both parents to develop thalassemia major.

Thalassemia minor occurs if you receive the defective gene from only one parent. Persons with this form of the disorder are carriers of the disease and do not have symptoms.

Beta thalassemia major is also called Cooley's anemia.

Risk factors for thalassemia include :

- Asian, Chinese, Mediterranean, or African American ethnicity
- Family history of the disorder

Symptoms

The most severe form of alpha thalassemia major causes stillbirth (death of the unborn baby during birth of the late stages of pregnancy).

Children born with thalessemia major (Cooley's anemia) are normal at birth, but develop severe anemia during the first year of life.

Other symptoms include:

- Bone deformities in the face
- Fatigue
- Growth failure
- Liver and spleen swelling
- Shortness of breath
- Yellow skin (jaundice)

Persons with the minors form of alpha and beta thalassemia have small red blood cells (that can be seen under a microscope), but no symptoms.

Treatment

There is no need to treat thalassemia minor.

Treatment for thalassemia major often involves regular blood transfusions and folate supplements.

If you receive blood transfusions, you should not take iron supplements. Doing so can

cause a high amount of iron to build up in the body, which can be harmful.

Persons who receive significant numbers of blood transfusions need a treatment called chelation therapy to remove iron from the body.

Bone marrow transplant may help treat the disease in some patients, especially children.

Outlook (Prognosis)

Severe thalassemia can cause early death due to heart failure a, usually between ages 20 and 30. Frequent blood transfusions with therapy to remove iron from the body helps improve the outcome.

Less severe forms of thalassemia usually do not result in a shorter life span.

Possible Complications

Untreated thalassemia major leads to heart failure and liver problems, and makes a person more likely to develop infections.

Blood transfusions can help control some symptoms, but may result in too much iron which can damage the heart, liver, and endocrine system.

Prevention

Genetic counseling and prenatal screening may be available to those with a family history of this condition who are planning to have children.