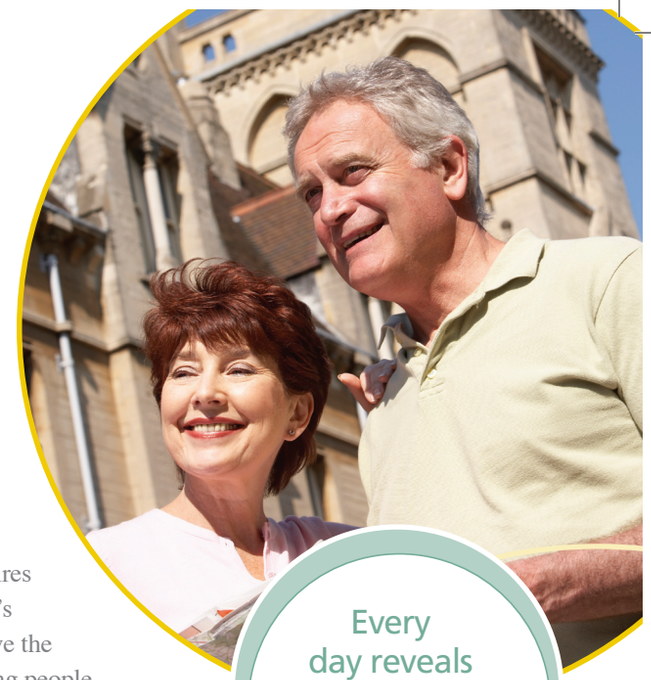
Thalassemia



Every day reveals a more promising future.

Thalassemia is a genetic disorder in which one of the two proteins that make up hemoglobin in red blood cells is deficient. Hemoglobin helps carry oxygen through the blood to all parts of the body. Most forms of thalassemia produce a chronic lifelong anemia that begins in early childhood and often must be treated with frequent transfusions. Significant, often life-threatening complications are common in the most severe forms.

Cooley's anemia (beta thalassemia major) is a severe form of thalassemia that requires regular, often monthly, blood transfusions. An estimated 1,000 people have Cooley's anemia in the United States, and an unknown number are carriers – people who have the genetic trait and can pass it on to their children. Thalassemia is most common among people of Mediterranean descent, such as Italians and Greeks, and is also found among people from the Arabian Peninsula, Iran, Africa, Southeast Asia, and Southern China. Because many affected families are recent immigrants belonging to these ethnic groups they face cultural and language challenges that may impede their ability to seek appropriate care and understand the resources available for living with thalassemia.

Because of the need for frequent blood transfusions, people with thalassemia are at increased risk for exposure to transfusion-related infections. Additionally, because there is no natural way for the body to eliminate iron, the iron in transfused blood cells builds up and becomes toxic to tissues and organs, particularly the liver and heart. Iron overload can typically result in early death from organ failure. Preventing iron overload requires regular treatment with medicines to rid the body of the excess iron. Unfortunately, these medicines must be administered through a needle under the skin which can be painful, thus limiting some people's ability to comply with this treatment.

A Public Health Approach

Epidemiology, Surveillance, and Research

The Centers for Disease Control and Prevention's (CDC) Thalassemia Data and Blood Specimen Collection System collects health information that will provide a better understanding of how to reduce or prevent the complications of thalassemia.

Currently, seven Thalassemia Treatment Centers participate in a CDC blood safety and health monitoring program. As part of this program, participants donate blood specimens to be screened for HIV and hepatitis A, B, and C. This repository of tested blood samples allows CDC to facilitate rapid investigation when emerging blood-borne pathogens are identified. In addition, clinical data are collected that can be used to describe the health status and extent of complications of people with thalassemia.

Data collection efforts increase the power to detect emerging infections and provide a more comprehensive view of the clinical characteristics and complications experienced by people with thalassemia nationwide. This knowledge of thalassemia will play a vital role in developing new research ideas and methods to optimize health outcomes for people with this condition.

Current research initiatives focus on several areas:

- The role of comprehensive health care services for thalassemia treatment as a means to prevent complications of the condition.
- The effectiveness of blood safety and surveillance efforts.

National Center on Birth Defects and Developmental Disabilities