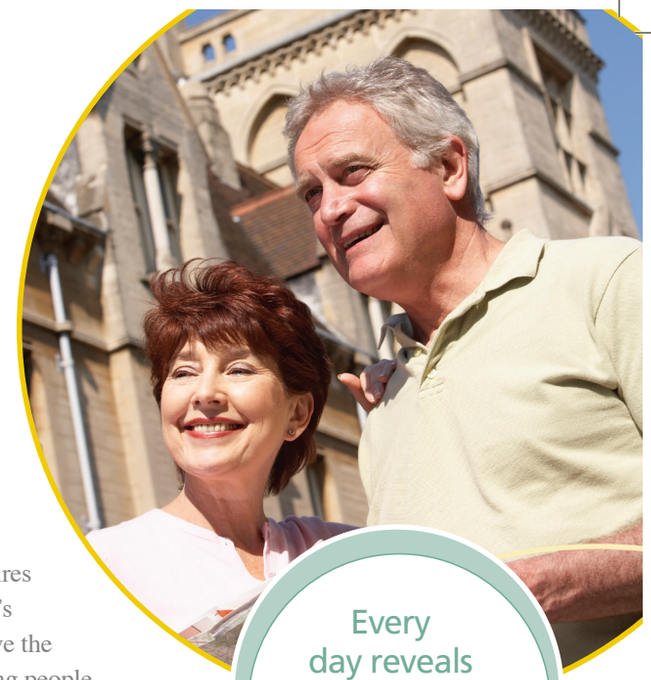
Thalassemia



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.

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Thalassemia

- The efficacy and adequacy of prevention and research activities.
- The evaluation of best practices to determine which measures, treatment, and follow-up protocols maximize optimal health outcomes.

Informatics and Infrastructure

CDC supports data collection and analysis for research activities on thalassemia. Because each health care provider may care for a very small number of people with thalassemia in his or her practice, it is necessary to collect data from many providers and settings so that enough data will be available to provide meaningful information.

It is important to design and offer data collection systems that are flexible and minimize the burden of data reporting. To meet this requirement, informatics systems provide the capability for investigators and collaborators to submit data in a variety of formats, including paper forms, electronic submission to a website, and export of data from third-party software or information systems. Additionally, a sound informatics infrastructure can help to coordinate and align data from multiple sources within a health care setting, such as clinical and laboratory data.

Health Education and Health Literacy

As with many of the blood disorders, no cure exists for thalassemia. Therefore, prevention efforts are focused on early identification and the promotion of health behaviors that prevent or lessen complications of this disease. CDC funds and works with our community-based partner, the Cooley's Anemia Foundation (CAF), to support outreach and education activities for people with thalassemia. CDC has collaborated with CAF to develop and translate educational materials for patients and their families and to provide educational materials to community-based providers and service organizations. Focus groups have been used to better understand the issues related to living with thalassemia and have provided useful information for improving education and support programs. This work has resulted in multilingual and culturally appropriate educational materials about the CDC blood safety initiative for more than 1,000 patients and their families. CDC has also sought feedback from the thalassemia community to better understand how to help people live with their treatment regimens.

Other CDC activities include:

- Delivering consistent health messages about preventing complications, complying with treatment regimens, and enhancing the quality of life for people with thalassemia and their families.
- Participating in outreach activities to identify new patients and provide access to services for underserved populations.
- Developing specific materials for nurses to educate families about thalassemia management and its complications.

Laboratory Capacity and Support

A priority for CDC is enhancing laboratory research capacity in the community by providing collaborating investigators with services such as subject-matter expertise, technical support, and laboratory analysis. CDC's hematologic laboratories conduct research and provide diagnostic services to people with thalassemia. Currently, CDC is serving as the research and service laboratory for blood safety monitoring by conducting testing for hepatitis A, B, and C and HIV for all participants enrolled in the thalassemia surveillance programs. A bank of blood specimens given regularly by participants is stored for future study of agents or conditions of importance to persons with thalassemia, such as emerging blood-borne infections.

For more information, please visit our website at www.cdc.gov/blooddisorders.

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