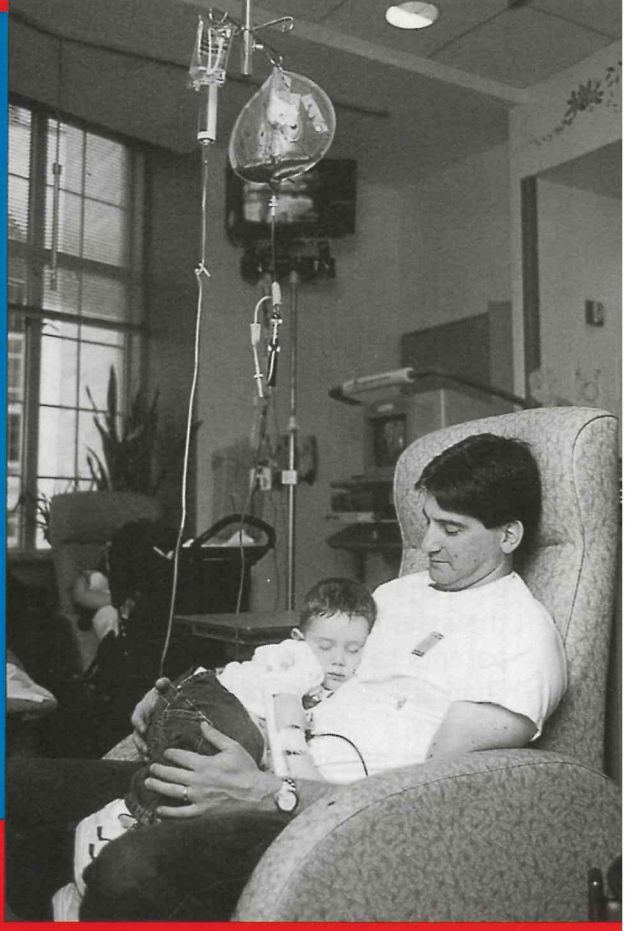
An Introduction

to

Beta-Thalassemia Major



Cooley's Anemia FOUNDATION

Leading the Fight Against Thalassemia

written by Marie B. Martin, RN, and Craig Butler

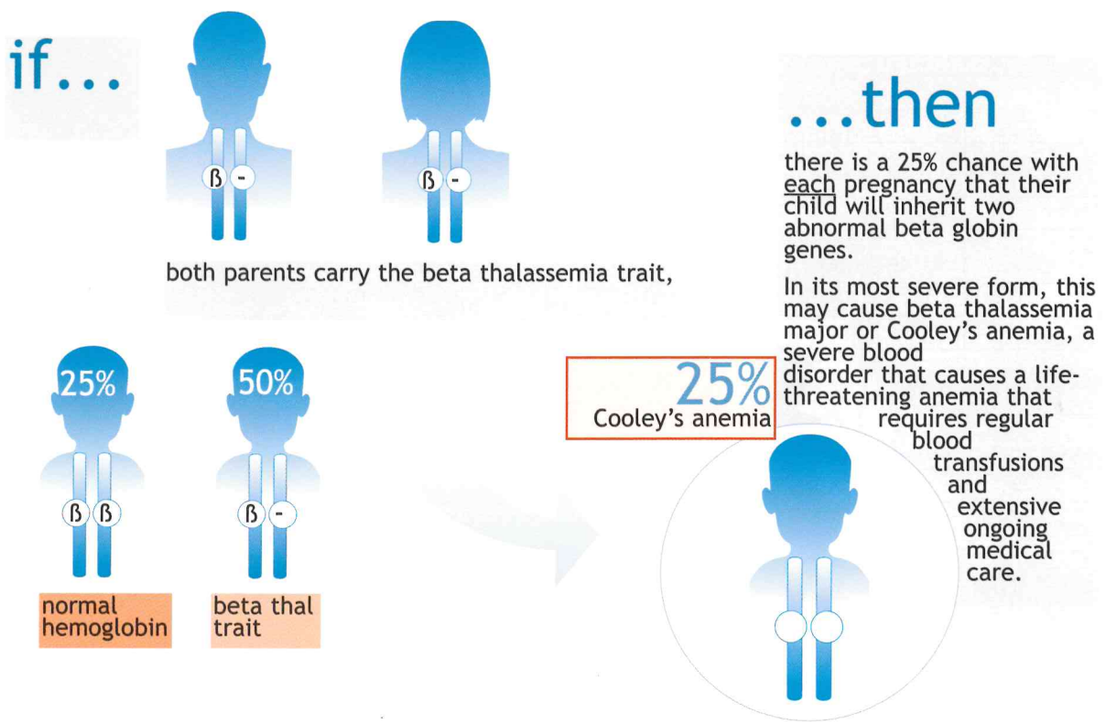
You have been told that your child may have beta-thalassemia major.
This brochure is intended to answer some basic questions about beta-thalassemia major and help you understand what to expect.

What is beta-thalassemia major?

Beta-thalassemia major is a genetic (or "inherited") blood disorder that is sometimes called Cooley's or Mediterranean anemia or sometimes simply called thalassemia. Beta-thalassemia major, the most severe form of the disorder, prevents or greatly reduces the body's ability to produce "adult" hemoglobin and causes anemia. Your child is missing one of the ingredients to make normal adult hemoglobin. Hemoglobin is a part of the red blood cell.

How did my child get it?

Beta-thalassemia major is an inherited disease. In order for a child to get beta-thalassemia major, both parents must carry the trait for thalassemia. If both parents carry the trait (also known as "thalassemia minor"), there is a 1-in-4 chance with each pregnancy that the child will be born with a severe form of the disease.



People who carry the thalassemia trait do not have ill effects from the carrier state and usually are unaware that they carry it. They may be told that they are slightly anemic and have "small red blood cells".

Is it my fault?

No. Just as you cannot control what color eyes your child will inherit, you cannot control whether your child will inherit thalassemia. However, being tested for the trait prior to pregnancy enables a couple to review the results and options with a genetic counselor.

Is my child going to die?

Most children with beta thalassemia major born in the past few decades will live well into adulthood. Medical treatments have improved greatly over the years; there is reason to believe that your child, taking full advantage of the therapies available now and in the future, will live a long, full life.

What is the treatment?

Chronic red blood cell transfusions will begin when your child's hemoglobin is low, or if the child's growth slows, or if the spleen and liver are enlarged. The treatment team at the Thalassemia Center of Excellence in conjunction with you will determine when to begin and how often your child will require transfusions. *(See our "Chronic Transfusion" pamphlet for more information.)*

Another treatment available to some families is Bone Marrow Transplant. We say "some" families because it is recommended that a full sibling who is "HLA matched" be the donor. The treatment center will recommend that your family be tested for a "match" soon after diagnosis. If there is a "matched sibling," you and the treatment center staff will review the options and make a decision for transplant or medical treatment. If you choose bone marrow transplant, your family will be referred to a doctor who is responsible for the procedure.

What are some of the complications of chronic red blood cell transfusions?

The most common complications of transfusions are fever ansd allergic reactions. Allergic reactions are due to proteins in the blood plasma, which can be corrected by washing the donated blood. Fever can be corrected by removing white cells from donated blood, which is now standard practice.

There is a risk of viral infections with transfusions, but the likelihood of transmission is very small because the supply of blood in the U.S. is thoroughly screened. The most well known viruses are hepatitis C and HIV.

Finally, there will be iron overload after a couple of years of transfusion. This is a major complication of transfusions and requires removal of the iron with medication. *(See our Iron Overload pamphlet for more information.)*

What things should I look out for?

The most important things to look for include:

- Your child's color (Is he or she pale?)
- If your child is gaining weight
- If your child's appetite is decreasing
- If his or her belly looks bigger
- If your child is crying a lot
- Irritability
- Poor growth
- Increased sleepiness/fatigue
- Increased infections
- Anything else that is out of the ordinary.

Always call your treatment center with <u>any</u> questions or concerns. <u>NO question is a dumb one!</u>

Where can I turn for help?

The **Cooley's Anemia Foundation** is here to provide you with information you need to deal with thalassemia. Please contact us at **(800) 522-7222** or **info@cooleysanemia.org.**

Recognized **Thalassemia Treatment Centers** have the most highly trained thalassemia experts in the country. Some of these centers are located at:

Children's Healthcare of Atlanta
Children's Hospital Boston
Children's Hospital Los Angeles
Children's Hospital Oakland
Children's Hospital of Philadelphia
Children's Medical Center Dallas
Children's Memorial Hospital (Chicago)
Texas Children's Hospital (Houston)
Weill Medical College of Cornell University (New York)

Many other hospitals are "satellite centers" affiliated with these Centers. Please contact the Cooley's Anemia Foundation for a list of these satellite centers.

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