

# An Introduction



to  
**Beta-  
Thalassemia  
Intermedia**

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This brochure is intended to answer some basic questions about beta-thalassemia intermedia and help parents understand what to expect if their child has been diagnosed with thalassemia intermedia.

### What is beta-thalassemia intermedia?

Beta-thalassemia intermedia is a genetic (or “inherited”) blood disorder that is sometimes called Cooley’s or Mediterranean anemia or sometimes simply called thalassemia. Beta-thalassemia intermedia, the milder form of the disorder, 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.

Thalassemia intermedia is less clinically severe than beta-thalassemia major. Often, people just use the term “thalassemia” to refer to any person with either thalassemia intermedia or thalassemia major. The distinction is the need for chronic red blood cell transfusions for “major” and no or intermittent transfusions for intermedia. (The DNA testing that helps determine thalassemia cannot reliably predict whether a child is major or intermedia; that determination is dependent upon the transfusion needs of the individual.)

### How did my child get it?

Thalassemia intermedia is an inherited disease. In order for a child to get thalassemia intermedia, 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 the 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”.

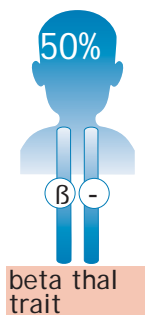
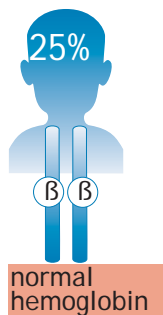
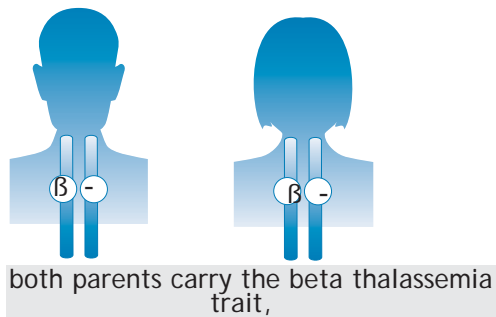
### Is it my fault?

No. Just as you cannot control what color eyes your child will inherit, you cannot control whether your child will inherit thalassemia. You can, however, be tested prior to pregnancy then review the results with a genetic counselor.

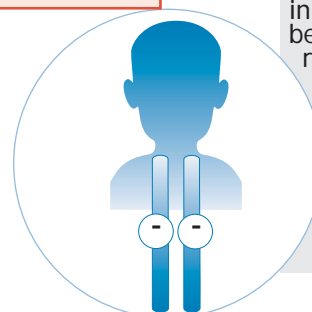
### What is the treatment?

Unlike in thalassemia major, where the degree of severity of anemia is more uniform from patient to patient, there is a great deal of variation in the severity of the anemia

if...



25%  
thalassemia major or  
intermedia



...then

there is a 25% chance with each pregnancy that their child will inherit two abnormal beta globin genes, resulting in thalassemia major or thalassemia intermedia.

associated with thalassemia intermedia. All patients with thalassemia intermedia experience some form of decreased hemoglobin levels.

In children with thalassemia intermedia, there are many decisions to be made by the health care team and the family in terms of transfusion. Frequently, your child's doctor will only give transfusions if the hemoglobin falls below a certain value on several occasions. Other times the team will decide to give chronic transfusion for a certain time period and then re-evaluate. These decisions depend on the well-being of your child and how he or she feels. It is hard to predict in what category your child will fall; therefore, the child will require frequent check-ups with your doctor.

Some patients are able to function normally at lower hemoglobin levels; however, it is important that a minimum hemoglobin level be maintained in order to prevent complications (*see below*).

### *If my child is not frequently transfused, what happens?*

Your child's red blood cells don't live as long as normal red blood cells and when they "die," the iron they carry is deposited in the organs. Your child's body is trying to make up for its low hemoglobin by creating many more red blood cells than is typical - and thus depositing more iron. The "dead" red blood cells are destroyed in the spleen, a filtration system for the body. As the spleen absorbs all the old blood, it grows, and the abdomen will look big. When the spleen gets big, the hemoglobin goes down because the cells are being filtered in the spleen. Sometimes, if the spleen is removed, the hemoglobin will go back up one to two grams.

If a child does not have regular transfusions, the bone marrow must overcompensate to make enough red blood cells to maintain an adequate level of hemoglobin. (Think of the bone marrow as a factory always working overtime.)

When the bone marrow has to work overtime to create more and more red blood cells, this

"hyperactivity" affects the bones themselves, and can cause them to become distorted, thinner and more easily broken. This hyperactivity also can lead to a need for greater intake of certain vitamins, such as folic acid, vitamin D and calcium.

Often, the bone marrow decides it needs help in creating all of these excess blood cells, and so other parts of the body that don't normally create blood cells - such as the chest area and the spine - are called upon to join in.

If bone marrow activity and changes in skeletal structure become significant and result in symptoms, the patient may be a candidate for chronic transfusion therapy.

### *What are some of the complications that may develop with thalassemia intermedia?*

There are a number of complications that may or may not occur, depending upon the severity of the anemia and upon the course of treatment that is best for your child.

Let's assume your child does not require transfusions. You may notice some "bony changes" in the face; the forehead or the cheek bones may appear to protrude, or he or she may appear to have "buck teeth". These changes are due to the bone marrow working extra hard to make red blood cells. This extra work causes the cells to "expand" in these areas. As your child gets older, he or she may experience broken bones more easily. Again, this is due to the extra work in the bone marrow.

Some patients develop hypersplenism, if their bodies attempt to create more red blood cells outside the bone marrow.

Although thalassemia intermedia are not chronically transfused as are thalassemia major patients, they still may develop iron overload. They therefore should avoid foods with high iron concentrations and iron supplements. If their iron levels reach a certain level, iron chelation therapy may be required.

Delayed puberty, impaired growth, gallstones and osteoporosis may also occur in older

children with thalassemia intermedia. There is also some evidence that thalassemia intermedia patients, especially those who have had their spleens removed, may have a slightly greater risk of blood clots.

Because there are so many potential complications associated with thalassemia, patients should undergo an annual “comprehensive care evaluation.” This should be performed at a Thalassemia Center of Excellence, each of which has a team of experts from different disciplines with significant experience in thalassemia.

### *Are there treatments that can increase my hemoglobin so that I don't have to get transfused?*

Some experiments with chemotherapy classes of drugs, such as hydroxyurea, have increased the amount of fetal hemoglobin in a patient, possibly increasing the baseline hemoglobin one to two grams or reducing transfusion requirements.

Some patients have been prescribed drugs such as Procrit or Aranesp, that stimulate the creation of red blood cells; however, red blood cells created in this manner still contain defective hemoglobin. As a result, the patient simply makes more “defective” red blood cells, which do not alleviate the anemia.

Other medical therapies are being explored which attempt to increase the hemoglobin via means other than transfusion. At the current time, however, transfusion remains the most effective way of raising or maintaining hemoglobin levels for most patients.

### *What things should I look out for?*

If you notice your child looking especially pale or jaundice (yellow) or if they are sleepier than usual, call your care center. This may mean that your child is more anemic than usual. These symptoms may be seen if your child is sick or has a fever. If an older child complains of upper abdominal (belly) pain, especially after eating fatty foods, you should contact your treatment center, as your child may be developing gallstones. This happens because of hemolysis (breaking down) of the red blood

cells. You should also monitor your child's growth process, as well as report any complaints of pain in his/her bones.

### *Where can I turn for help?*

The Cooley's Anemia Foundation is always ready to provide you with the information you need to deal with thalassemia. Please contact us at  
(800) 522-7222  
or  
[info@cooleysanemia.org](mailto:info@cooleysanemia.org).



The Thalassemia Centers of Excellence have the most highly trained thalassemia experts in the country. They are located at:

[Children's Hospital Boston](#)  
[Children's Hospital Los Angeles](#)  
[Children's Hospital Oakland](#)  
[Children's Hospital of Philadelphia](#)  
[Children's Memorial Hospital \(Chicago\)](#)  
[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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