



THALASSEMIA SUPPORT FOUNDATION



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comfort and encouragement
to those battling this disorder.

The Thalassemia Support Foundation was founded by patients, parents and friends affected by Thalassemia. The foundation provides hope, comfort and encouragement to those battling this disorder. At the heart of the organization is a strong desire to help improve the quality of life for all patients with Thalassemia. We volunteer our time to organize conferences, raise funds to educate the community, ensure patients and parents know the latest in care, and donate to the work of researchers. The foundation maintains a strong relationship with the medical community that provides diagnoses, treatment and care.

PURPOSE OF ORGANIZATION

Support

1. To help Thalassemia patients and parents understand the disease and the actions around improving the quality of life and finding a cure
2. To ensure patients and parents have access to doctors and information that will allow them to be advocates for their needs.
3. Provide Standards of Care Booklet to parents and patients
4. Provide funds for patients to receive initial examination by expert doctors at one of the Thalassemia Centers
5. Build and maintain a database of Thalassemia patients and parents
6. Organize annual conference in at risk communities to provide updated information on Thalassemia
7. Organize, facilitate, and participate in community based events throughout the year
8. To provide a support system among the thalassemia community (patients, parents, friends, etc.), so that people can get into contact with other people.
9. Have fun

Medical Relationship

1. Build and maintain a close relationships with primary Thalassemia Centers and staffs
2. Encourage doctors who specialize in thalassemia to consult and advise the medical community so that every patient receives the highest quality of care
3. Facilitate events for doctors to share information about the treatment of Thalassemia
4. Support Thalassemia events at Thalassemia Centers

Research Support

1. Support research for improving treatment, diagnosis and cure of Thalassemia

Activist

1. Advocate / Lobby for Thalassemia causes to affect changes in state laws and support
2. Have a clear channel of communications to patients, medical community, and general public about the affects of the disease
3. Connect with the State of California for information on Newborn Screening and assist with information flow to patients for their knowledge and care

Thalassemia is a genetic blood disorder that affects the production of the hemoglobin, the oxygen carrying component of the red blood cell. Because of this, patients have to get blood transfusions, usually every two to three weeks. These blood transfusions are done at a hospital and can take anywhere from six to eight hours or more. After some time, the blood cells break down and leave iron in the patient's body. This iron will bind to the major organs of the body, such as the liver or heart. If left alone, the iron will overload these organs until they will not be able to do their job creating other health problems for the patient.

Thankfully there is a drug called Desferal. Desferal binds to the iron in the patient's body and removes it. This process is called chelation. This drug is usually administered subcutaneously, in places like the stomach or legs, every night up to twelve hours a day. This treatment will start early, usually when a child is between one and two years old.

But these are not the only problems faced by patients with Thalassemia. The psychological and emotional toll put on them by this disease is staggering and can be overwhelming. That is why this organization was founded. To support them and help them.

What is Thalassemia?

Thalassemia is a term that refers to a group of genetic disorders characterized by insufficient production of hemoglobin. There are two proteins involved in the production of hemoglobin, alpha and beta. If there is a deficiency in either of these proteins the red blood cells do not form properly and cannot carry adequate amounts of oxygen to all parts of the body. This then results in organs that are starved for oxygen and unable to function properly.

Is Thalassemia contagious? How would I get Thalassemia?

Thalassemia is a genetic disorder caused by the inheritance of a recessive gene from both parents. You cannot "catch it" from someone. In pregnancies where both parents carry the recessive gene there is a 1 in 4 chance that the child will have the severe form of thalassemia, a 2 in 4 chance that the child will carry the gene for thalassemia and a 1 in 4 chance that the child will neither have the disease or be a carrier.

How do you know if you have Thalassemia?

Thalassemia can be detected in utero if the parents know that they are carriers through a prenatal test called chorionic villus sampling (CVS). If the parents are unaware that they are carriers, blood tests after birth will result in a diagnosis. Symptoms of Thalassemia include the following: paleness of the skin, poor appetite,

failure to grow normally, jaundice, severe anemia, enlargement of the spleen. Thalassemia is usually diagnosed through the use of blood tests showing small abnormally shaped red blood cells and/or a hemoglobin electrophoresis showing abnormal hemoglobin.

Are there different types of Thalassemia?

Alpha thalassemia appears in people whose hemoglobin does not produce enough alpha protein. There are five types of alpha thalassemia:

- *Silent Carrier*
- *Alpha Thalassemia Trait*
- *Hemoglobin H Disease*
- *Hemoglobin H - Constant Spring*
- *Alpha Thalassemia Major*

Beta thalassemia appears in people whose hemoglobin does not produce enough beta protein. There are three types of beta thalassemia:

- *Thalassemia minor or Thalassemia trait* - In this condition the individual has smaller than normal red blood cells but generally has no health problems. Occasionally, those with this condition suffer from mild anemia.
- *Thalassemia Intermedia* - In this condition the lack of beta protein causes more severe anemia and other health problems commonly associated with Thalassemia major (bone deformities and enlarged spleen). These individuals need more regular transfusions but it is typically to improve life not for survival.
- *Thalassemia Major or Cooley's Anemia* - This is the most severe of the beta thalasseemias. These individuals require regular blood transfusions and other medical treatment for survival.

Well, who gets Thalassemia?

Thalassemia can show up in any population of people.

Alpha thalassemia tends to be more common in people with the following ancestry:

- African
- Middle Eastern

- East Indian
- Southeast Asian (Vietnamese, Laotian, Thai, Singaporean, Filipino, Cambodian, Malaysian, Burmese and Indonesian)
- Chinese
- Occasionally Mediterranean (Italian and Greek)

Beta thalassemia tends to be more common in people with the following ancestry:

- Mediterranean (Italian and Greek)
- Iranian
- African
- Southeast Asian
- Chinese

E Beta thalassemia tends to be more common in people with the following ancestry:

- Southeast Asian (Cambodian, Vietnamese and Thai)

Sickle Beta Thalassemia tends to be more common in people with the following ancestry:

- Mediterranean (Italian, Greek and Turk)

How do you treat Thalassemia?

The treatment of thalassemia depends on the severity of the disease for each individual. For those individuals with mild forms of the thalassemia often no treatment is required however as the severity of the disease increases blood transfusions become the primary treatment. The transfusions can occur as frequently as every 2-3 weeks in order to maintain the hemoglobin at near normal levels so as to promote physical growth and general well-being. However this hypertransfusion, as it is referred to, is not without its own risks. The primary one being the risk of iron overload. Our bodies can only process so much iron and the excess iron from the transfusions is stored in the organs, primarily the liver. To help combat iron overload, individuals with thalassemia must employ chelation therapy. This refers to the use of a chelator, Desferal, to remove the excess iron from the system. Desferal is administered subcutaneously and/or intravenously. This drug binds to the iron and the iron is reduced through elimination of bodily functions.

How does Thalassemia affect you?

It is hard to measure specifically how thalassemia will or does affect you. For those who are only carriers or have a mild form of the disease there is often very little affect. They might be slightly anemic or have to have occasional transfusions but otherwise lead "normal" lives.

Individuals who have the severe form of the disease have much to deal with physically and psychologically. Physically, thalassemia patients often suffer from enlargement of the spleen, liver, and/or heart. Their bones become thin and brittle and many will eventually suffer from osteoporosis. Additionally, the bones of the face may become distorted. Individuals with thalassemia are often smaller than their peers due to the fact that they generally grow slower. From a treatment standpoint, the most severe forms of thalassemia require regular blood transfusions and subsequent iron removal through chelation therapy. Each year there are also numerous medical tests to determine the progression of the disease and the success of the current protocol for each individual.

Psychologically, thalassemia is a disease that, if allowed, can control your life. It is difficult to explain to others that you have a genetic blood disorder that requires ongoing medical treatment. It is difficult to have to take responsibility for your medical care. It is difficult to have to spend so much time in the hospital or with doctors. It is difficult to be different.

However, IT IS NOT IMPOSSIBLE!!!

There are many people with thalassemia who are living "normal" lives. They have thalassemia and it is a part of who they are but it is not the only thing in their lives. These individuals make it to adulthood, they go to college, they get married and yes, some of them even have children. How? They have made a commitment to themselves that life is worth living and so they take whatever steps are necessary to make that happen. They are diligent about their treatment. They develop relationships with the medical professional in charge of their care. Most importantly they surround themselves with people who love and support them and with others who have thalassemia and can relate to their experience, especially organizations like the Thalassemia Support Foundation. Get involved, make a difference!

Paul was diagnosed with thalassemia major when he was four months old. At that time, he looked pale, but otherwise appeared to be healthy. Blood tests would prove otherwise.

My husband and I were unaware that we were carriers of this genetic disease; that there was a one in four chance in every pregnancy that a child would be born with thalassemia major. It was even more shocking to us because Paul has an older brother who doesn't even carry the thalassemia trait.

We had no idea what this was or how our lives were about to change. We went to see a specialist on blood disorders. I brought a tape recorder so I could record the visit, afraid I would miss something important. I still have that tape, twenty-five years later.

The specialist told us that Paul's red blood cells were abnormal in shape and size and that he could not reproduce his own blood cells. Because Paul could not reproduce his blood cells, he would need blood transfusions to keep his hemoglobin at a normal level. All this seemed unreal, but at the same time, we wanted his home life to be as normal as possible.

The blood transfusions started when Paul was twelve months old. They have continued every three to four weeks for the last twenty four years and will continue for the rest of his life.

A nurse once told me "you do what you have to do". And we did. There were the constant blood tests. Watching the nurses trying to find a vein in my small child always broke my heart. There were all the late nights at the hospital, waiting for the type and cross, waiting for the blood, waiting for the blood to finish transfusing so we could go home. Always waiting.

And with the transfusions came a high iron count. When Paul was two, I had to learn to insert a needle in Paul's stomach every night so the drug Desferal could excrete the excess iron out of his system through his urine. Just imagine a two year old running around to get away from the treatment and the full family effort it took every night to make sure that he was getting his medication. We can laugh now but it wasn't so funny then.

In patients who have thalassemia the excess iron, from transfusions, binds itself to vital organs if it is not removed through treatments such as Desferal. Over time this excess iron can shorten his life span by causing complications to his organs.

His ferritin level, a measure of the iron in Paul's body, remains fairly low, but liver biopsies are showing that his liver is becoming overloaded with iron, so he is on a more aggressive Desferal treatment.

Years ago a child rarely lived to adulthood. Now they are living productive lives well into their 40's.

At twenty-five years old, Paul is doing quite well. He is married and he and his wife, Melissa, are pursuing their doctorate degrees. Paul now has an implanted portcath that allows him to receive his blood transfusions and Desferal treatment thru the port, instead of through his overused veins. He has taken over the responsibility of scheduling his transfusions and making appointments with home health care nurses for the Desferal treatments. Paul has developed a wonderful relationship with all his doctors and nurses at the hospital.

The hospital trips, doctor appointments, and calls to nurses are a daily reminder that he has to continually monitor his condition. Even a slight fever can mean a trip to the doctor's office to check for infection. We are very fortunate that over the years Paul has taken an active role in his treatment. He is not afraid to question the doctors or make suggestions.

It has not been easy having a child with thalassemia, there are constant worries about his health and worries about his future. Yet over time we have learned to deal with these issues as they come. We have become part of organizations such as the Thalassemia Support Foundation because it's not just Paul who needs to know and be supported by other thalassemia patients; we parents need that support as well. We have done our best to provide Paul with a "normal" life in spite of the circumstances. As part of a close knit Italian family, Paul will always have our love and support.

Thalassemia doesn't just impact the patient; it impacts everyone that knows them.