Beta Thalassemia Major

What is Beta Thalassemia Major?

Beta thalassemia major, also called "Cooley's Anemia," is a genetic disorder affecting the red blood cells. It mainly affects people of Mediterranean, Middle Eastern, Asian and South Asian descent.

Red blood cells contain a protein called "hemoglobin" that helps to carry oxygen throughout the body. Hemoglobin is made of alpha globin chains and beta globin chains. Patients with beta thalassemia have a genetic abnormality that prevents them from making enough beta globin chains, and so they cannot make hemoglobin properly.

How did my child get it?

Beta thalassemia major is an inherited disease, which means it is passed from parent to child. Normally, children get one beta gene from each parent. If one of these beta genes is abnormal, the child will have beta thalassemia trait. In thalassemia trait, the child will have small red blood cells and mild anemia, but no clinical symptoms. If both beta genes the child receives are abnormal, the child will have beta thalassemia major. In thalassemia major, the child cannot make properly functioning red blood cells, and will have life-threatening anemia.

What are the symptoms?

Children born with beta thalassemia major are usually fine right after they are born. However, within the first two years of life they begin to have decreased energy levels and poor growth. This is the point at which treatment is started. The life expectancy of a patient with beta thalassemia major depends largely on the patient's compliance with the prescribed treatment. A person who works together with the hematology team and takes the proper steps to care for themselves will have the best chance of living a long, productive life.

How is it treated?

Transfusions

Children and adults with beta thalassemia major require blood transfusions to survive. Blood transfusions begin when symptoms first appear and are then given every 3-4 weeks for life. The amount of blood your child receives will depend on your child's body size and hemoglobin level. Blood is given through an IV in the hematology clinic, and takes about 4 hours.

It is important to know some of the risks of transfusions. The most common risk is a reaction to the transfusion. Your child will be monitored closely for signs of a reaction and you will be educated about what to watch for at home. There is also an extremely rare risk for infection with HIV, hepatitis B or other viruses. In Canada there are very strict rules in place to screen donors and test the blood before it reaches your child, so the risk of getting an infection from a blood product is very low. However, as an increased precaution the hematology team will monitor your child's blood work for these viruses annually.

Iron Chelation Therapy

Blood transfusions contain large amounts of iron, much more than what we normally get in our diet or in supplements. The body has no way to get rid of this extra iron, so it builds up in the heart and liver, causing damage and eventually organ failure. Your child will be at risk for iron overload because of his/her monthly transfusions. To prevent iron overload and preserve organ function, your child must do "iron chelation therapy." There are three iron chelators available in Canada: Desferal, Jadenu and Ferriprox. Your hematology team will discuss with you which medicine is best suited for your child. For more information on iron chelators, see the BC Children's Hospital handout Iron Chelation Therapy.

Bone Marrow Transplant

Bone marrow transplant (BMT) is a complex procedure and is the only way to cure thalassemia. Chemotherapy is given to destroy the bone marrow, and is replaced with new bone marrow from a healthy, matched donor. This new bone marrow does not have the abnormal beta gene, and can produce healthy red blood cells and cure the thalassemia. This procedure does come with many serious and life-threatening risks, and requires the patient to remain in hospital for several weeks. Only certain patients are eligible. For more information contact your hematology team.

When should I call the doctor?

Call your child's hematology team if you notice your child:

- Has unusually yellow skin or eyes
- Looks more pale than usual
- Is more sleepy or tired than normal
- Has fever, flushing, hives or rash within 24 hours of a blood transfusion

Where can I find more information?

Cooley's Anemia Foundation - www.cooleysanemia.org

Northern California Comprehensive Thalassemia Center - www.thalassemia.com

Thalassemia International Federation - www.thalassemia.org.cy

Thalassemia Foundation of Canada - www.thalassemia.ca

KidsHealth - http://kidshealth.org/parent/medical/heart/beta_thalassemia.html#

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