Journal of Hematology & Thromboembolic Diseases

OPEN ACCESS Freely available online

Short Communication

THALASSEMIA

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ABSTRACT

Thalassemia is an autosomal recessively inherited blood disorder which results from alteration in the rate of globin chain production which impedes normal heamoglobin synthesis. Thalassemia is caused by mutations in the DNA of cells that make heamoglobin — the substance in red blood cells that carries oxygen throughout your body. The mutations associated with thalassemia are passed from parents to children.

Keywords: Thalassemia, Heamoglobin, Mutations, Red blood cells

INTRODUCTION

Haemoglobin molecules are made of chains called alpha and beta chains that can be affected by mutations. In thalassemia, the production of either the alpha or beta chains are reduced, resulting in either alpha-thalassemia or beta-thalassemia [1].

In alpha-thalassemia, the severity of thalassemia you have depends on the number of gene mutations you inherit from your parents. The more mutated genes, the more severe your thalassemia.

In beta-thalassemia, the severity of thalassemia you have depends on which part of the heamoglobin molecule is affected.

SYMPTOMS

Fatigue

Weakness

Pale or yellowish skin

Facial bone deformities

Slow growth

Abdominal swelling

Dark urine

Some babies show signs and symptoms of thalassemia at birth; others develop them during the primary two years of life. Some people that have just one affected heamoglobin gene do not have thalassemia symptoms [2].

CAUSES

Thalassemia is caused by mutations in the DNA of cells that make heamoglobin — the substance in red blood cells that carries oxygen throughout your body. The mutations related to thalassemia are passed from parents to children [2].

Heamoglobin molecules are made of chains called alpha and beta chains that can be affected by mutations. In thalassemia, the assembly of either the alpha or beta chains are reduced, leading to either alpha-thalassemia or beta-thalassemia.

Alpha-thalassemia, the severity of thalassemia you have depends on the number of gene mutations you inherit from your parents. The more mutated genes, the more severe your thalassemia.

In beta-thalassemia, the severity of thalassemia you've got depends on which a part of the haemoglobin molecule is affected.

RISK FACTORS

Factors that increase your risk of thalassemia include:

Family history of thalassemia: - Thalassemia is passed from parents to children through mutated haemoglobin genes.

Certain ancestry: - Thalassemia occurs most frequently in African Americans and in people of Mediterranean and Southeast Asian descent.

COMPLICATIONS

Possible complications of moderate to severe thalassemia include:

Iron Overload: - People with thalassemia can get an excessive amount of iron in their bodies, either from the disease or from frequent blood transfusions. Too much iron may result in damage to your heart, liver and system, which incorporates hormone-producing glands that regulate processes throughout your body.

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Received: July 27, 2020; **Accepted**: July 29, 2020; **Published**: August 04, 2020

Citation: Thode S (2020) THALASSEMIA. J Hematol Thrombo Dis 8: 306. DOI: 10.24105/2329-8790.2020.8.306

Infection: - People with thalassemia have an increased risk of infection. This is especially true if you've had your spleen removed.

Bone Deformities: - Thalassemia can make your bone marrow expand, which causes your bones to widen. This can end in abnormal bone structure, especially in your face and skull. Bone marrow expansion also makes bones thin and brittle, increasing the prospect of broken bones.

Enlarged spleen: - The spleen helps your body fight infection and filter unwanted material, like old or damaged blood cells. Thalassemia is usually amid the destruction of an outsized number of red blood cells. This causes your spleen to enlarge and work harder than normal. An enlarged spleen can make anaemia worse, and it can reduce the life of transfused red blood cells. If your spleen grows too big, your doctor might suggest surgery to get rid of it.

Slowed Growth Rates: - Anemia can both slow a child's growth and delay puberty.

Heart Problems: - Congestive heart failure and abnormal heart rhythms can be associated with severe thalassemia.

TREATMENT

Chronic anemia in thalasemic patients is managed with.

Blood transfusion:- The transfusion are given once a month using washed filtered or frozen red cells neocytes for transfusion can be used with aim of reducing transfusion frequency

Iron chelation:- desferoxamine is used for iron chelation as there will be iron overload.it is given either iv or subcutaneously as it is poorly absorbed from gastrointestinal tract [3].

Deferiprone is an oral chelating agent which is less effective than deferoxamine in preventing organ damage.it may cause arthritis, neutropenia, and agranulocytosis so careful monitoring is required.

Deferasirox is another oral chelating agent that has similar efficacy as deferoxamine.

Ascorbic acid supplementation in small doses wil increase iron chelation with desferoxamine

Spleenectomy it is done in cases of hypersplenism.

Haemopoietic stemcell transplantation..it is the only curative treatment for thalassemia

PREVENTIONS

Genetic counselling is needed for the couple and their family to prevent the birth of other children with thalassemia major.

Prenatal testing can be used to detect thalassemia major in the foetus.

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