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Thalassemia: An Inherited Autosomal Recessive Disorder of Human Blood

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A B S T R A C T

Thalassemia is a form of inherited autosomal recessive blood disorders characterized by abnormal formation of hemoglobin. Thalassemia is caused by variant or missing genes that affect how the body makes hemoglobin, the protein in red blood cells that carries oxygen. There has been a rise in concern for this genetic disorder of blood especially in children and many awareness campaigns and initiatives in greater interest of public health are undertaken in India in association with various hospitals and NGOs.

Keywords: Blood disorder, Gene, Thalassemia

A R T I C L E I N F O

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1. Introduction

People with thalassemia make less hemoglobin and have fewer circulating red blood cells than normal, which results in mild or severe microcytic anemia. The abnormal haemoglobin formed results in improper oxygen transport and destruction of red blood cells [1]. Thalassemia can cause significant complications, including iron overload, splenomagaly, bone deformities, and cardiovascular illness. Thalassemia may confer a degree of protection against malaria (specifically, malaria caused by the protozoan parasite *Plasmodium falciparum*. Various thalassemias

resemble another genetic disorder affecting hemoglobin, sickle-cell disease.[2,3]

Cause

Both α- and β-thalassemias are often inherited in an autosomal recessive manner. Cases of dominantly inherited α- and β-thalassemias have been reported. For the autosomal recessive forms of the disease, both parents must be carriers for a child to be affected. The risk is 25% for each pregnancy for an affected child when both parents carry a hemoglobinopathy trait.

Incidence of the trait

An estimated 60-80 million people in the world carry the β-thalassemia trait. Genetic counseling and genetic testing are recommended for families who carry a thalassemia trait. Countries such as Bangladesh, Nepal and Pakistan are seeing a large increase of thalassemia patients due to lack of genetic counseling and screening. Concern is increasing that thalassemia may become a very serious problem in the next 50 years. An estimated 1,000 people live with thalassemia major in the United States, and an unknown number of carriers. Because of the prevalence of the disease in countries with little knowledge of thalassemia, access to proper treatment and diagnosis can be difficult.

2. Therapeutic correction

Multiple blood transfusions can result in iron overload. Medications with deferoxamine, deferiprone or deferasirox for iron overload related to thalassemia may be treated by chelation therapy.[4] These treatments have resulted in improved life expectancy in those with thalassemia major [4]. Bone marrow transplantation may offer the possibility of a cure in young people who have an HLA-matched donor [5]. Success rates have been in the 80–90% range [5]. Common side effects of medications include: nausea, vomiting and diarrhea. It however is not effective in everyone and is probably not suitable in those with significant cardiac issues related to iron overload [4].

Directives with Recommendation

There is no evidence from randomised controlled trial to support zinc supplementation in thalassemia [6]. Mortality from the procedure is about 3%.[7]. There are no randomized controlled trials which have tested the safety and efficacy of non-identical donor bone marrow transplantation in persons with β- thalassemia who are dependent on blood transfusion [8].

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