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Review: Overview on Thalassemia

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Abstract
Thalassemia is an inherited autosomal recessive blood disorder. Which results in excessive destruction of Red blood cells and further leads to anemia. Thalassemia'sare prevalent Worldwide with 16,800 death and 4,39,000 severe cases in 2015. According to 2017 data 80 million are carriers of beta thalassemia, Beta thalassemia is more common in children. Thalassemia is caused by variant or missing genes that affects how the body make hemoglobin. Which people having thalassemia Make less hemoglobin and fewer Circulating red blood cells than normal results in mild or severe anemia.
Keywords: Thalassemia, Hemoglobin, Cooley Anemia, Amniocentesis and Bone marrow transplant.

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I. Introduction

The name thalassemia derived from a combination of two Greek words: Thalassa means the sea (Cooley) and anemia (weak blood) another term found in literature although in frequently is cooley's anemia of the name of Prof, Cooley Thomas, a pediatrician in USA who first described the clinical characteristics of this disorder in patients of Italian Origin. Thalassemia are blood disorder that are inherited from parents to offspring, which patients carry the genes of thalassemia it may be show symptoms in early age or later age that can results in the abnormal formation of hemoglobin and red blood cells being destroyed, which leads to anemia. It is caused by variant or missing genes that affects how the stem cells produced Hemoglobin, In this disorder the shape or size of Red blood cells is changed or abnormal. Mutation in the DNA or the cells that produce Hemoglobin this hemoglobin involving the HB1 and HB2. This is most commonly inherited in a mendelian recessive fashion, asian, Chinese, African and American ethnicity.

Types

1. Alpha thalassemia
- Alpha thalassemia minor
- Alpha thalassemia major

2. Beta Thalassemia
- Beta thalassemia minor
- Beta thalassemia major

Clinical Manifestations

Thalassemia clinical features depends upon the types of the thalassemia. In Alpha thalassemia may have mild anemia and typical asymptomatic (Most severe form of alpha thalassemia major cause birth. Beta thalassemia children born with beta thalassemia major they are normal at birth, but develop severe anemia in first year of age. Beta- Thalassemia is also known as (Cooley Anemia) Fatigue and weakness, Pale skin or jaundice, Poor appetite, Dark urine and lethargy these all are the some common sign and symptoms. Protruding abdomen with enlarge spleen and liver, Abnormal facial bones and poor growth and Bone marrow hyperplasia these are the sever sign and symptoms seen in thalassemia major condition.

Diagnostic Evaluation

History collection, Physical examination (can reveal spleenomegaly), Blood test including the findings: RBC's will appear small and abnormal in shape when looking under microscope, CBC reveal anemia. Hb electrophoresis test show the presence abnormal form of Hb and with the help of Mutational analysis detect alpha thalassemia. Amniocentesis (fetal diagnosis for a specific type or thalassemia) and Molecular diagnostics test.

Management

Medical treatment: Thalassemia minor usually not required treatment but Thalassemia major required treatment according to patients conditioon or it's symptoms - Blood transfusion, Iron chelation therapy, Genetic testing and counseling. Surgical treatment: Bone marrow transplantation (especially in children), Spleenectomy rriay be done to decrease the transfusion requirements (because RBC's may be sequestered in spleen if iron supplement is used during blood transfusion.

II. Conclusion

Thalassemia are inherited disorder; beta Thalassemia has high severity, presented by Mild to severe anemia. Diagnosis by the Complete blood count (CBC), Blood Smear, Iron studies, Hemoglobinopathy, DNA analysis (Genetic testing), and Prenatal testing (Genetic testing of amrtiotic fluid). Alpha thalassemia major causes s0llbirth. Treatment of thalassemia depends upon the severity and regular blood transfusion, iron chelation therapy, bone marrow transplantation etc. Prevention of this by premarital screening carrier detection, genetic counseling before family planning and prenatal testing. Prognosis of beta thalassemia major is very poor but we can increase the rate of survival by the some therapy like iron chelation therapy, blood transfusion and bone marrow transplantation.

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