RAMA UNIVERSITY

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FACULTY OF NURSING

THALASSEMIA

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INTRODUCTION

- The thalassemia is a blood disorder passed down through families (inherited) in which the body makes an abnormal form of Hb
- Hemoglobin (Hb) is the protein in RBC's that carries oxygen.
- The disorder results in large no. of RBC's being destroyed, which leads to anemia.

DEFINITION

- Thalassemia is an autosomal -recessive genetic disorder that results in adequate normal Hb production. Whereas IDA affects heme synthesis of globin.
- Thalassemia is a group of diseases that have an autosomal recessive genetic basis involving inadequate production of normal Hb.

INCIDENCE

- Commonly found in members of ethnic groups whose origins are near the Mediterranean Sea and equatorial or near - equatorial regions of Asia; the Middle East, and Africa.
- Commonest group of autosomal recessive disorders in India.
- Carrier frequencies vary from 3% to 17% in different populations, with over 30 million people carrying the defective gene.
- About more than 9000 thalassemic children are born every year in country.

CAUSES

Hb is made up of two proteins:

– Alpha globin,

– Beta globin.

Thalassemia occurs when there is a defect in a gene that helps in control production of one of these proteins. (abnormal Hb synthesis).

Risk factors

Risk factors for thalassemia include: –

- Family history of the disorder.
- Asian, Chinese, Mediterranean, or African-

American ethnicity.

TYPES

A. Two types of thalassemia:

1) Alpha thalassemia:- (most common)

- When a gene or genes related to alpha globin protein are missing or changed (mutated).
- Alpha globin chains are absent / reduced in this type of thalassemia.
- Alpha trait (heterozygous state) is asymptomatic in about 30% of African Americans.
- Alpha thalassemia occur most often in people from Southeast Asia, Middle East , China, And those of African descent.

2) Beta thalassemia:-

- When ‘similar gene defects’ affect the production of beta globin protein.
- Beta globin chains are absent or reduced in this type of thalassemia.
- It mostly occur often in people of Mediterranean origin.
- Chinese , Asians, and Africans are less affected.

B. Both alpha & beta have different subtypes including:

1) Thalassemia major:-

- It is developed by inherited gene defect from both parents.
- A homozygous person has two thalassemic genes, causing a severe condition known as thalassemia major

2) Thalassemia minor:-

- It includes the receiving of faulty gene from the only one parent.
- Those who inherit just one beta gene (heterozygote) have thalassemia minor also called the thalassemia trait, the carrier state of thalassemia.

CLINICAL MANIFESTATIONS

- Individual with alpha-thalassemia may have mild anemia & are typically asymptomatic. (most severe form of alpha thalassemia major causes stillbirth).
- Children born with beta-thalassemia major (Cooley anemia) are normal at birth, but develop severe anemia during first year of life.

- Children also have pain, failure to thrive, frequent infections, diarrhoea, spleenomegaly, hepatomegaly, jaundice from RBC hemolysis, and bone marrow hyperplasia.

Other symptoms are:

- Bone deformities in face.
- Fatigue.
- Growth failure.
- Shortness of breath.
- Yellow skin (jaundice).

DIAGNOSTIC EVALUATION

- History collection.
- Physical examination (can reveal spleenomegaly).
- Blood test including the findings: – RBC's will appear small & abnormal in shape when looked under microscope. – CBC reveals anemia. – A test called Hb electrophoresis shows the presence of an abnormal form of Hb. – A test called mutational analysis can help detect alpha thalassemia.

MANAGEMENT

Medical Management:

- Thalassemia minor usually does not require treatment.
- For thalassemia major, the treatment goals are:

– To provide adequate normal Hb for erythropoiesis and

– To alleviate the effects of iron overload

– Or its treatment goal involves regular blood transfusion & folate supplements).

- During blood transfusion, the client should not take iron supplements (doing so can cause a high amount of iron to build up in body, which can be harmful).
- Chronic transfusions are administered to correct anemia with the targeted Hb level at 9-10 mg/dl .

SURGICAL MANAGEMENT:

- Bone marrow transplantation (especially in children).
- Spleenectomy may be done to decrease transfusion requirements (because RBC's may be sequestered in spleen , if iron supplements used during blood transfusion)

THANK YOU