SNS COLLEGE OF NURSING

Saravanampatti (po), coimbatore.

DEPARTMENT OF NURSING

COURSE NAME : BSC (NURSING) II YEAR

SUBJECT :MEDICAL SURGICAL NURSING I

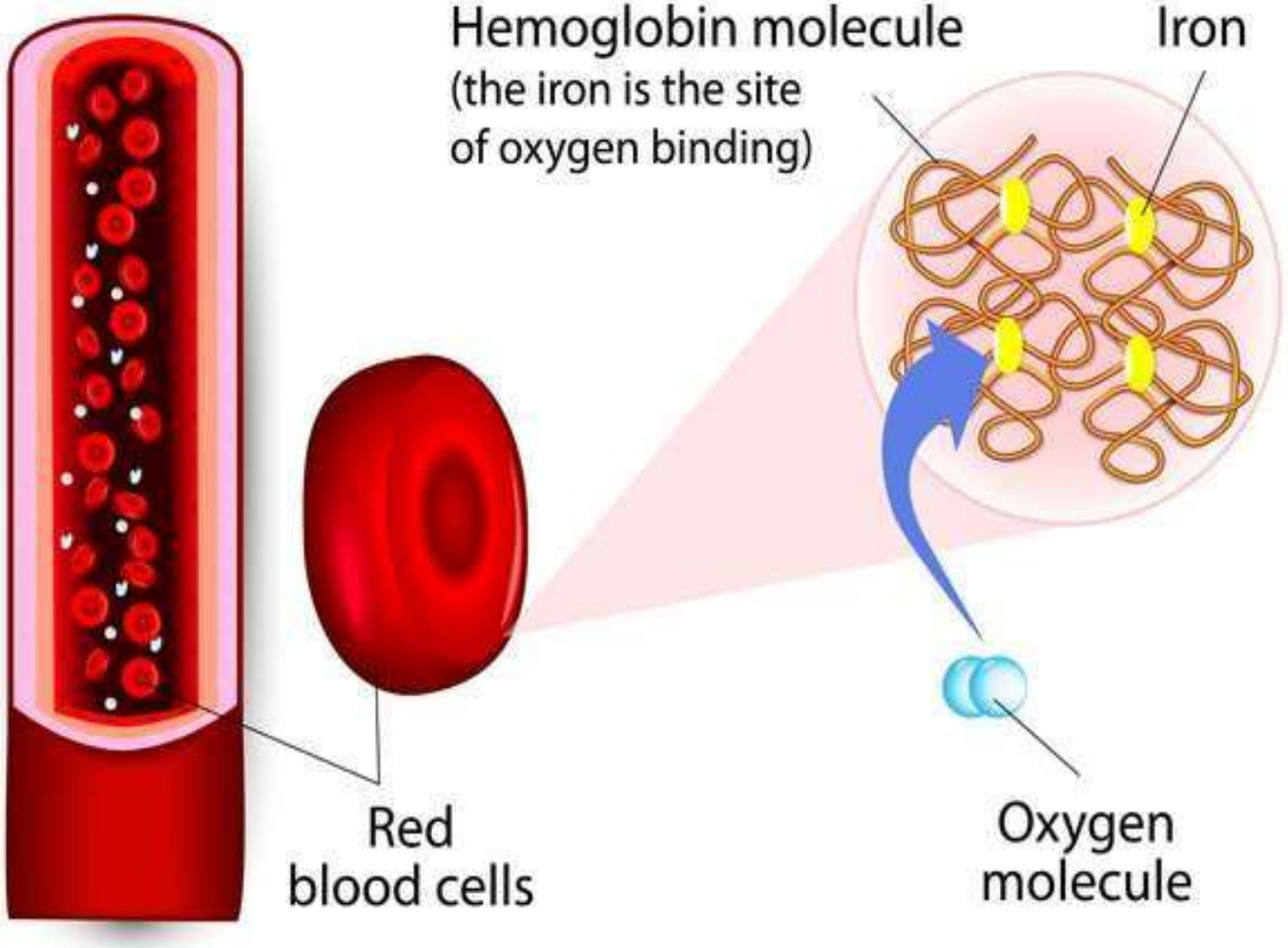
UNIT: V CARE OF PATIENTS WITH BLOOD DISORDERS

TOPIC : Thalassemia

INTRODUCTION

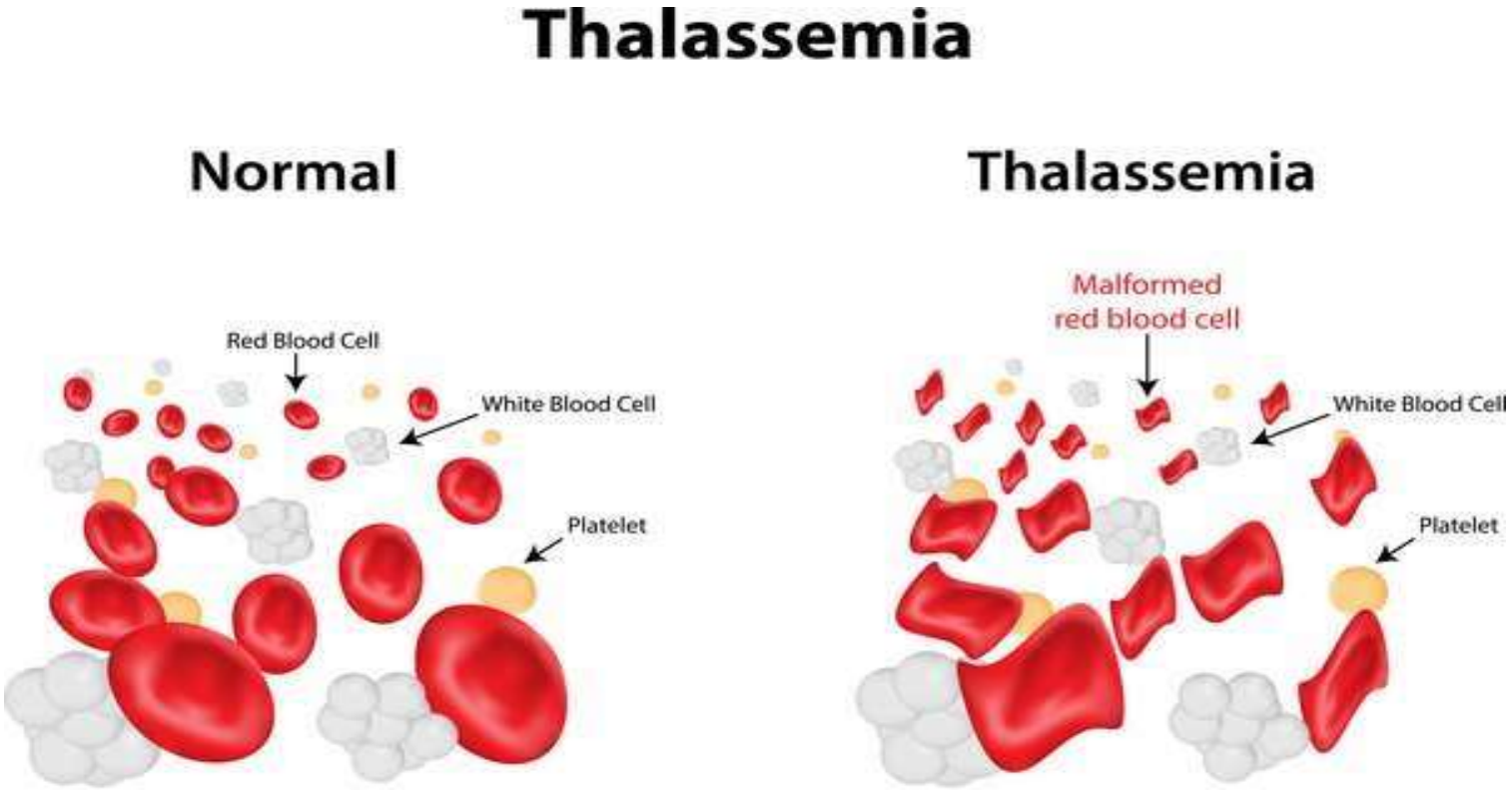
- **The Thalassemia Thalassemia Thalassemia is a blood disorder**
- **passed .**
- **down through families (inherited) in which the body makes**
- **an abnormal form of Hemoglobin (hb). It leads to Anemia**
- **due to deceased Erythrocyte production.**

HEMOGLOBIN


Hemoglobin molecule
(the iron is the site
of oxygen binding)
Iron
Red
blood cells
Oxygen
molecule

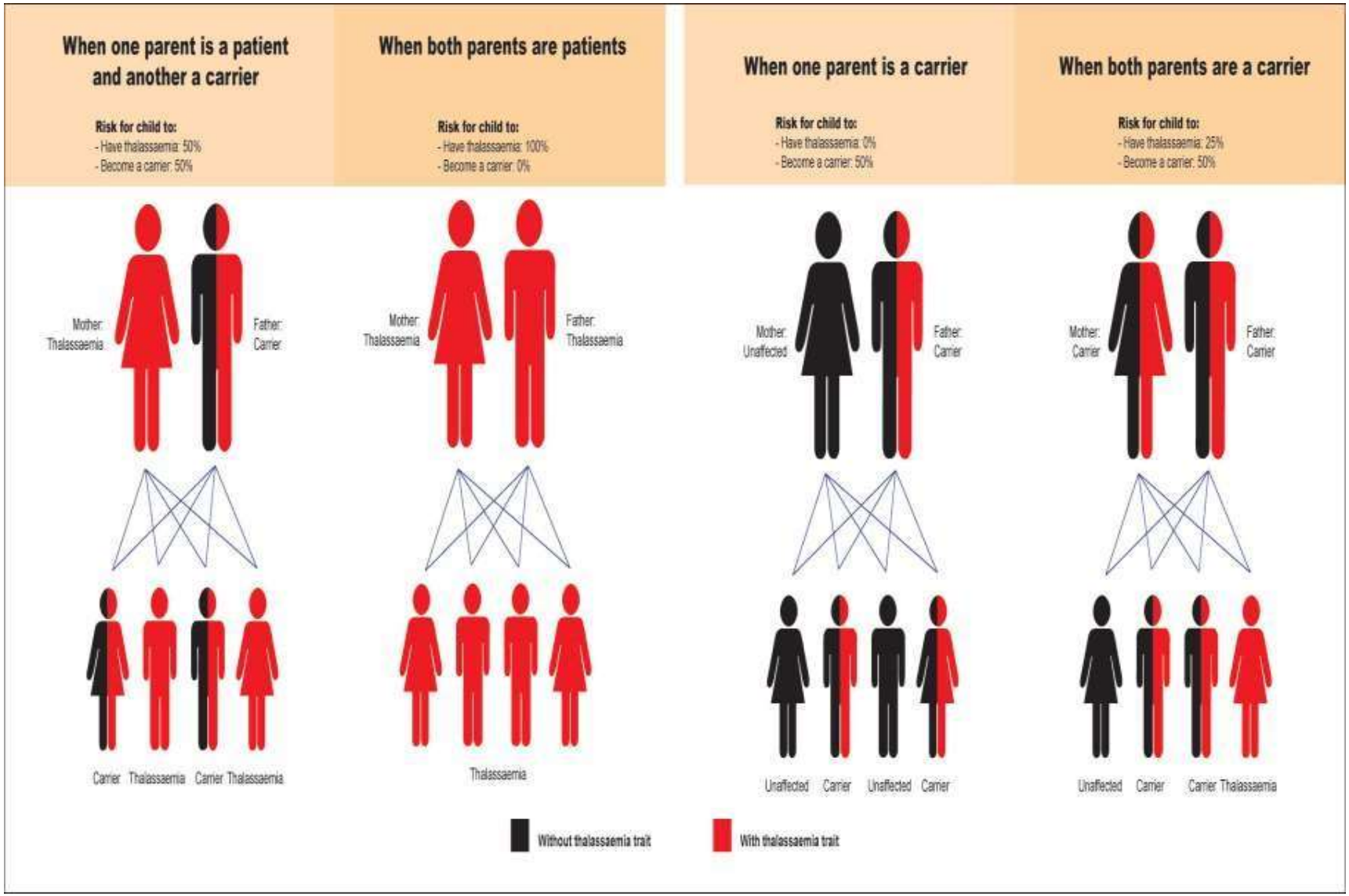
DEFINITION

Thalassemia is an autosomal recessive genetic disorder that results in inadequate production of normal haemoglobin in the blood.



INCIDENCE

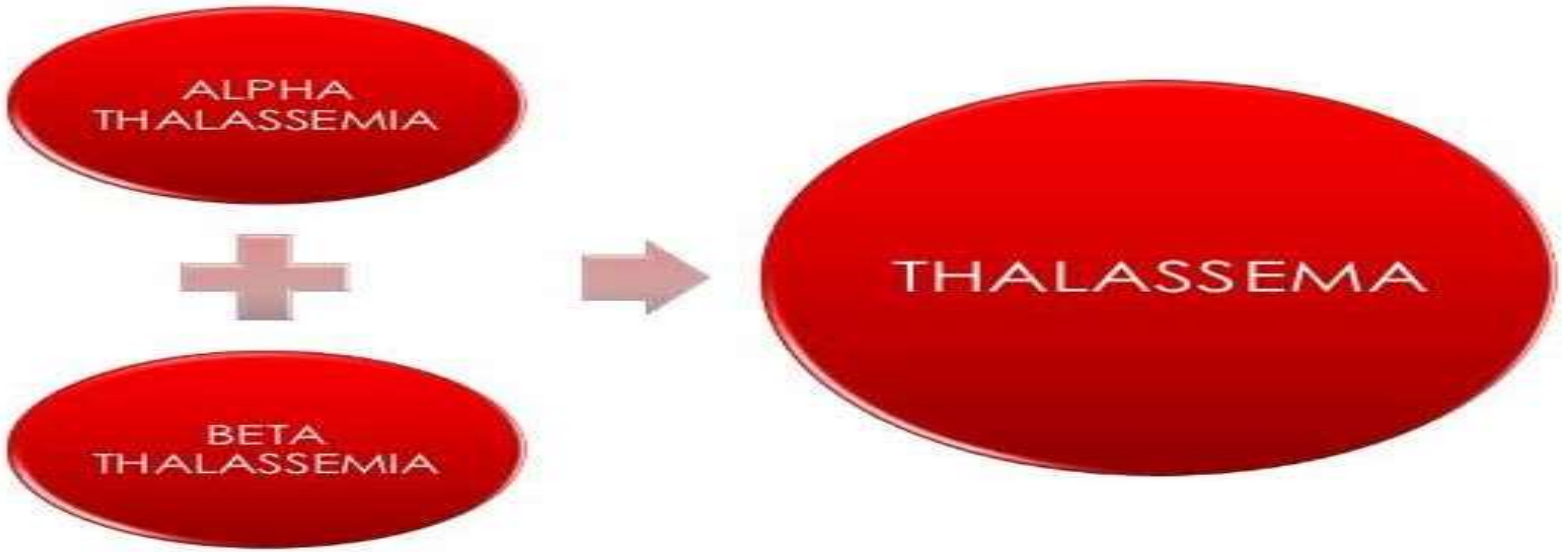
- Commonly found in members of ethnic groups whose origins are near the mediterranean sea and equatorial or near - equatorial regions of asia; the middle east, and africa.
- Commonest group of autosomal recessive disorders in india.
- Carrier frequencies vary from 3% to 17% in different populations, with over 30 million people carrying the defective gene.
- About more than 9000 thalassemic children are born every year in country.


When one parent is a patient and another a carrier
Risk for child to:
- Have thalassaemia: 50%
- Become a carrier: 50%
When both parents are patients
Risk for child to:
- Have thalassaemia: 100%
- Become a carrier: 0%
When one parent is a carrier
Risk for child to:
- Have thalassaemia: 0%
- Become a carrier: 50%
When both parents are a carrier
Risk for child to:
- Have thalassaemia: 25%
- Become a carrier: 50%
Mother: Thalassaemia
Father: Carrier
Carrier Thalassaemia Carrier Thalassaemia
Mother: Thalassaemia
Father: Thalassaemia
Thalassaemia
Mother: Unaffected
Father: Carrier
Unaffected Carrier Unaffected Carrier
Mother: Carrier
Father: Carrier
Unaffected Carrier Carrier Thalassaemia
Without thalassaemia trait
With thalassaemia trait

RISK FACTORS

Risk factors for Thalassemia include:

- Family history of the Thalassemia disorder.
- Asian, chinese, mediterranean, african or american ethnicity.

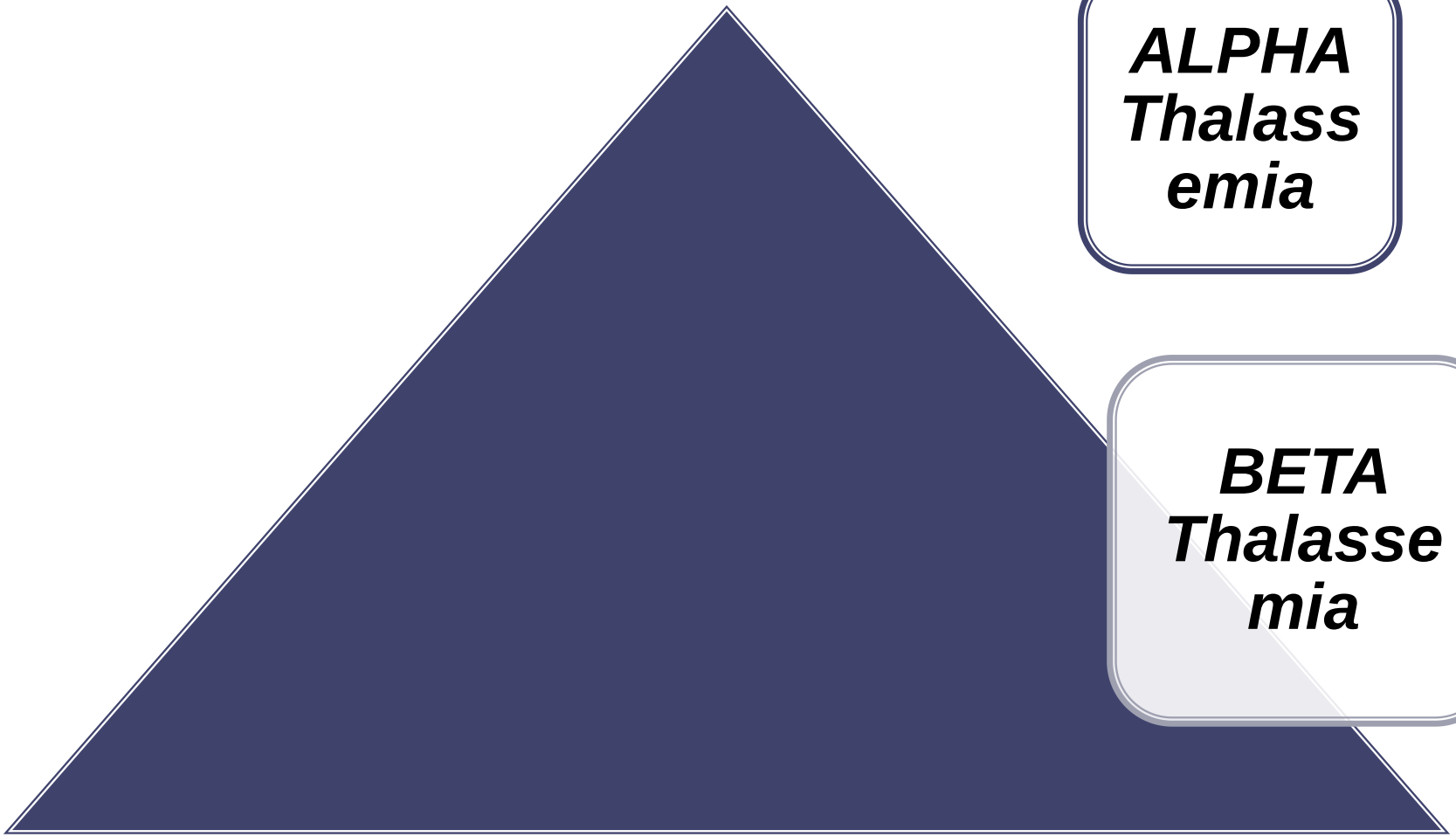


CAUSES

- 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)
- Thalassemia is caused by mutations in the dna of cells. The mutations associated with Thalassemia are passed from parents to children.

TYPES OF THALASSEMIA



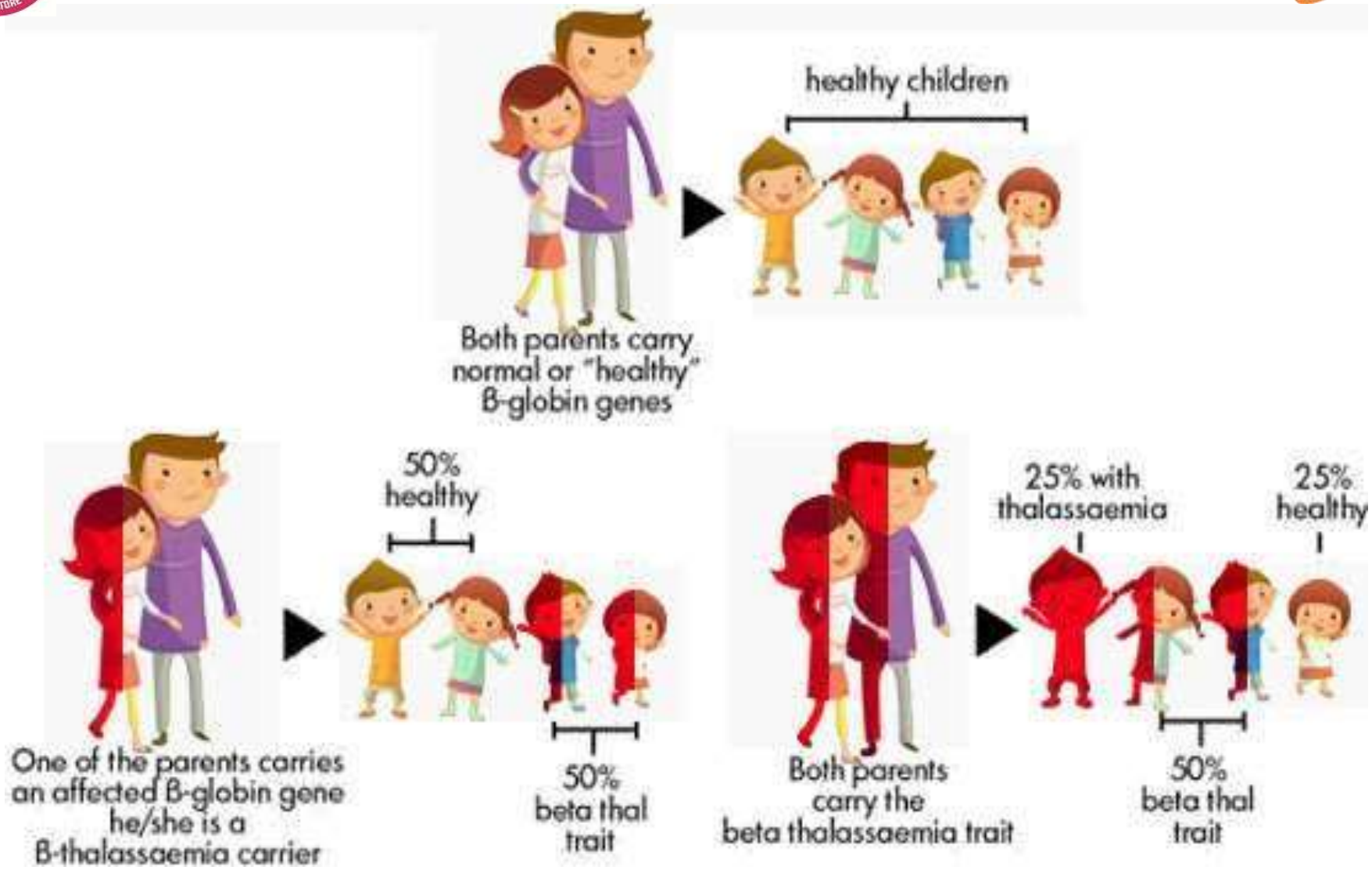
TYPES OF THALASSEMIA

1) alpha Thalassemia:-

- It is the most common type of Thalassemia
- 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 Thalassemia occurs most often in people from southeast asia, middle east , china, and those of african descent.

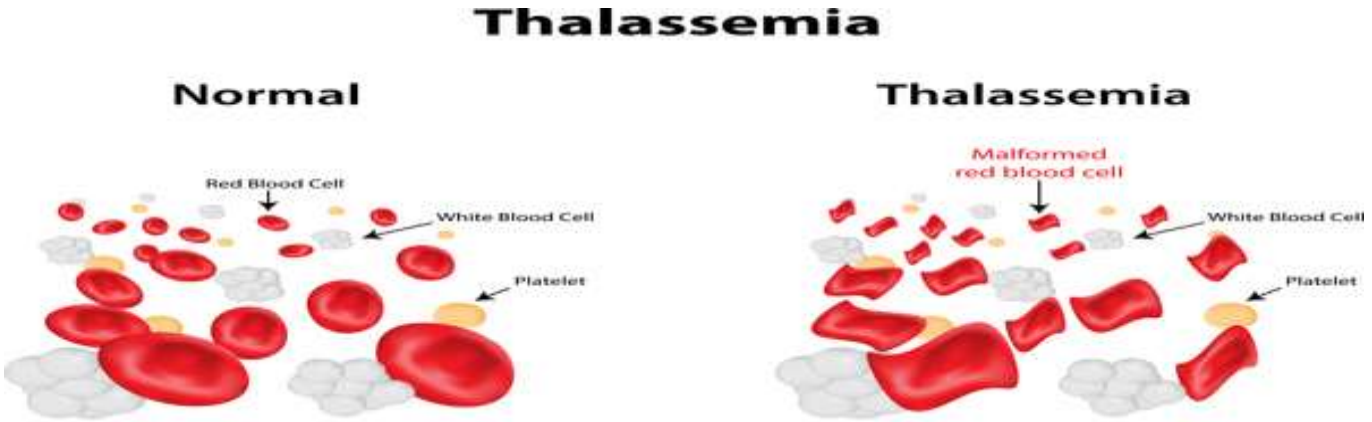
2) beta Thalassemia:-

- 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.

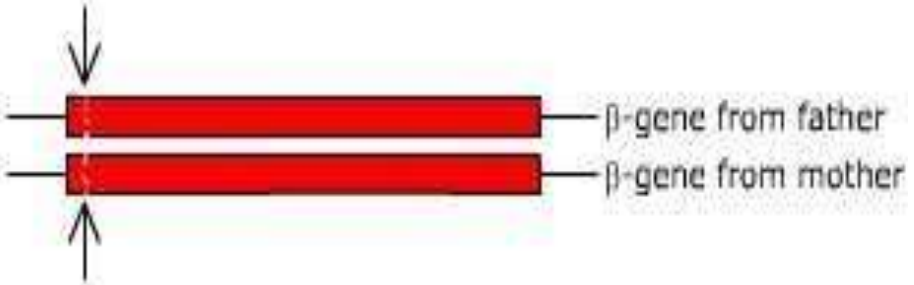

healthy children
Both parents carry normal or "healthy" β-globin genes
50% healthy
One of the parents carries an affected β-globin gene he/she is a β-thalassaemia carrier
50% beta thal trait
Both parents carry the beta thalassaemia trait
25% with thalassaemia
25% healthy
50% beta thal trait

1.THALASSEMIA MAJOR

- Those who inherit both beta- genes (homozygote) have Thalassemia major, which results in a profound and life threatening Thalassemia.
- Beta Thalassemia major is also called cooley Thalassemia



With a mutation on one of the two ß-globin genes, a carrier is formed with lower protein production, but enough hemoglobin



Without a mutation enough Hemoglobin

No thalassemia carrier

With one mutation less Hemoglobin

β-thalassemia carrier without illness, but less hemoglobin (slight aneamia)

With two mutations no β-globin

β-thalassemia major patient with severe aneamia

2. Thalassemia minor:-

•It includes the receiving of faulty gene from the only one parent.

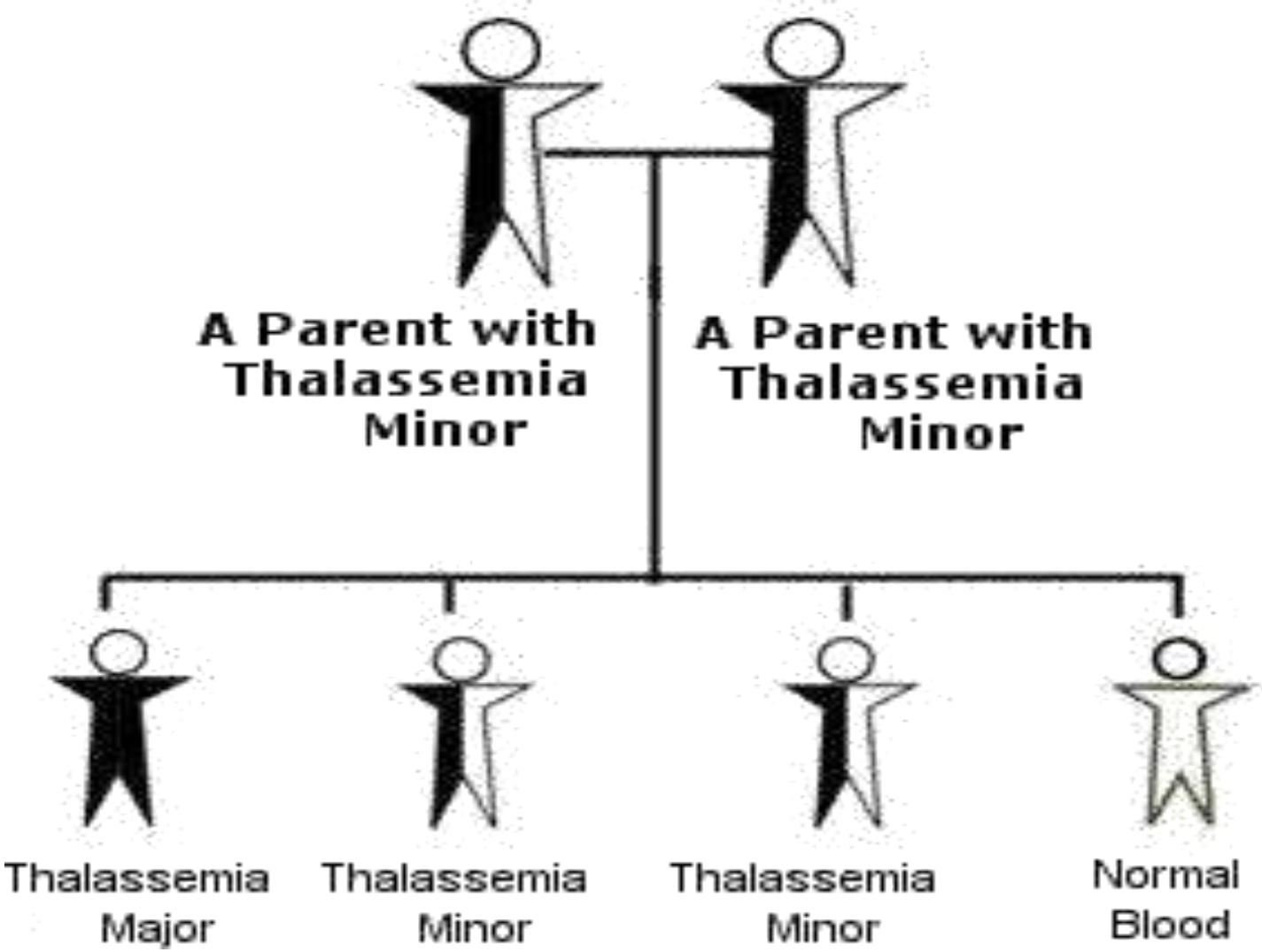
•Those who inherit just one beta gene (heterozygote)

• have Thalassemia minor also called the Thalassemia trait,

•the carrier state of Thalassemia.

•A person who is heterozygous has one thalassemic gene

•and one normal gene and is said to have Thalassemia minor (or thalassemic trait), which is a mild form of disease.


A Parent with Thalassemia Minor
A Parent with Thalassemia Minor
Thalassemia Major
Thalassemia Minor
Thalassemia Minor
Normal Blood

PATHOPHYSIOLOGY

Mutation in alpha-globin gene

Defected gene inheritance

One beta -globin gene is mutated

Existence of a mutation in both beta-genes

Minor disruption in beta -globin synthesis

Significant impairment of beta globin synthesis

Marked reduction in Hb production

Alpha-Thalassemia

Thalassemia major

Thalassemia minor

Profound Thalassemia (Beta –Thalassemia)

Hemolysis results from an alpha & beta globin chains, which are normally paired

The excess unpaired alpha or beta globin chains aggregat

Form a precipitatethat damages RBC membranes

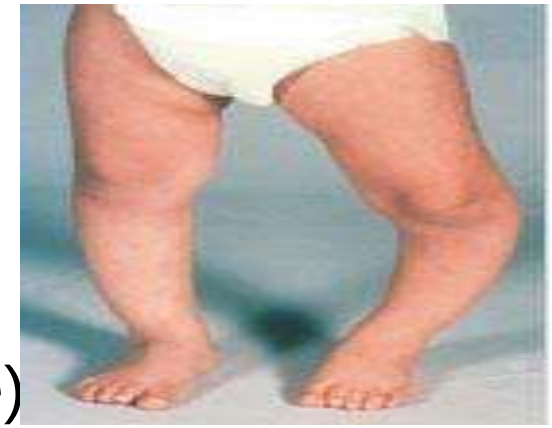
Intravascular hemolysis

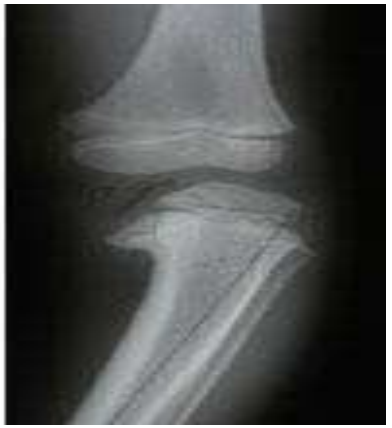
CLINICAL MANIFESTATIONS:

Individual with Alpha-Thalassemia may have mild Thalassemia & are typically asymptomatic.(Most severe form of alpha Thalassemia major causes *stillbirth*).

- Individuals born with Beta-Thalassemia major (Cooley Thalassemia) are normal at birth, but develop *severe Thalassemia* during first year of life.
- They may appear normal at birth because fetal hb contain no beta-globin; in first few months, manifestations of severe Thalassemia begin to appear.

- Patientsmay also have pain, failure to thrive, frequent infections, diarrhoea, splenomegaly, 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)

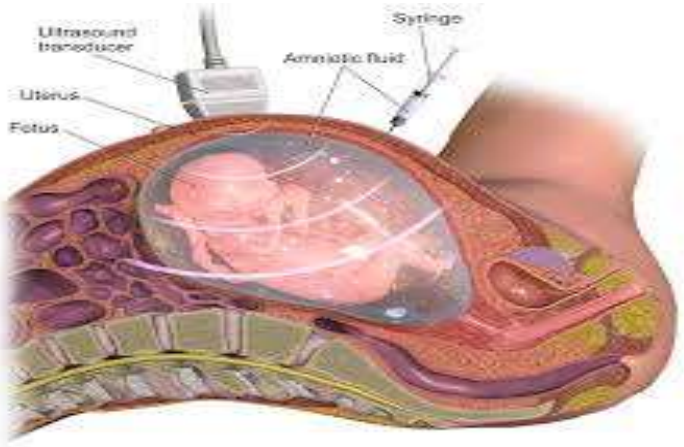




DIAGNOSTIC EVALUATION

- 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 Thalassemia.
  - A test called Hb electrophoresis shows the presence of an abnormal form of Hb.
  - A test called mutational analysis can help detect alpha Thalassemia.

- Amniocentesis (fetal diagnosis for a specific type of Thalassemia).
- Molecular diagnostic tests can determine whether a mutation is present after 8 weeks of gestation.



MANAGEMENT

Medical management:

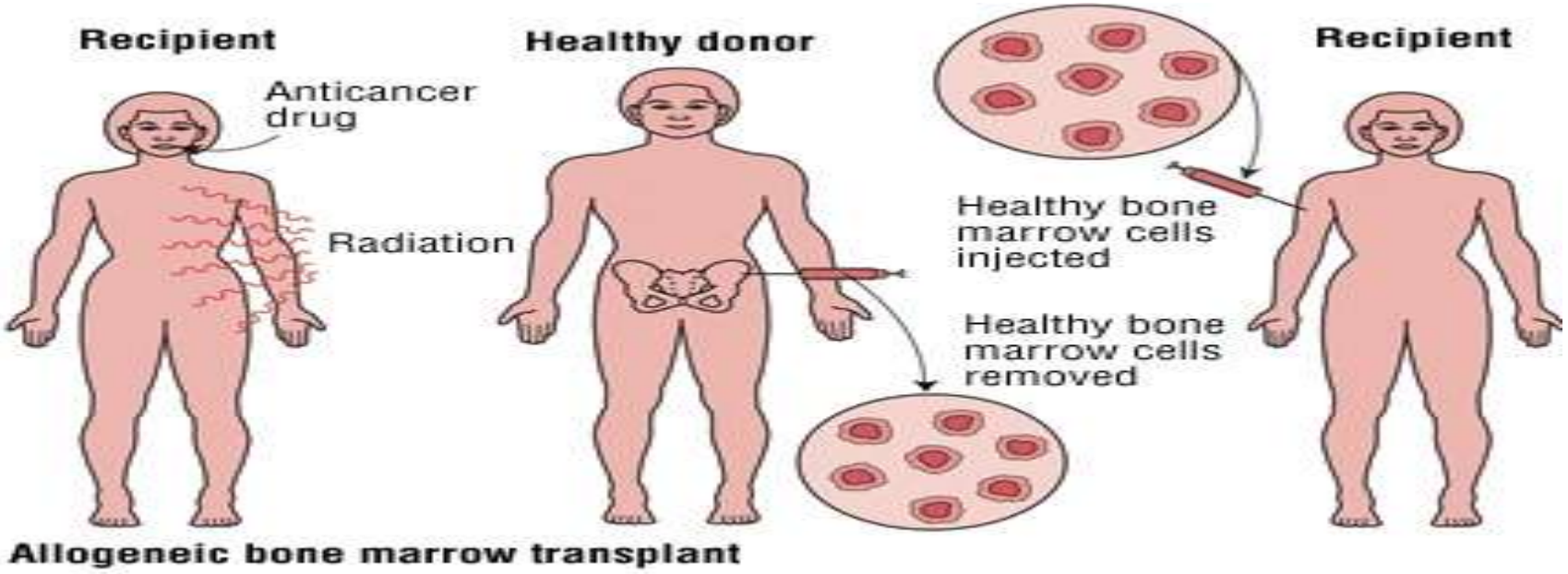
- Thalassemia minor usually does not require treatment.
- For Thalassemia major, the treatment goals are:
  - To provide adequate normal hb for erythropoiesis and
  - To alleviate the effects of iron overload
  - Or its treatment goal involves regular blood transfusion & folate supplements).

- 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 Thalassemia with the targeted hb level at 9-10 mg/dl
- Genetic counseling and testing for families should be encouraged .

SURGICAL MANAGEMENT

Bone marrow transplantation (especially in children).

Splenectomy may be done to decrease transfusion requirements (because RBC's may be sequestered in spleen , if iron supplements used during blood transfusion)



NURSING MANAGEMENT

- Assess the client's general conditions.
- Monitor vital signs.
- Monitor intake output.
- Manage fluid overload.
- Monitor regular blood transfusion to keep hb at 10.5 gm/dl.
- 24 hoursurine collection after chelating therapy to estimate amount of iron excreted.
- Reassure the orange color of urine is normal (can show dehydration).

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Nursing diagnosis:

1. Risk for infection related to decreased resistance secondary to hypoxia.
2. Impaired nutritional pattern less than body requirement related to inadequate nutritional intake and anorexia.
3. Activity intolerance related to impaired oxygen transport.
4. Disturbed body image related to skeletal changes.

SUMMARY

- Introduction.
- Definition.
- Incidence.
- Causes.
- Types.
- Pathophysiology.
- Clinical features.
- Diagnostic evaluation.
- Management.

References

1. Janice L.Hinkle,Kerry H.Cheever(2018) Brunner and Suddarth's Text Book of Medical Surgical Nursing ,14th edition,south asian edition,vol II,Wolters Kluwer.pg no. 925-93

2. Lizy Sonia & Shaina sharma (2016).Medical Surgical Nursing-prep Manual for Undergraduates,voll,Thomson ress,India,Elsevier publishers.pg no.521

Assessment

- Define Thalassemia.
- List the causes of Thalassemia.
- Explain the types of Thalassemia.
- Describe the pathophysiology of each type of Thalassemia.
- Enumerate the clinical features of Thalassemia
- Outline the diagnostic evaluation.
- Explain in detail the management of Thalassemia.