Tajuk Kursus : ONE-DAY WORKSHOP; UPDATES IN LABORATORY DIAGNOSIS OF THALASSEMIA AND HAEMOGLOBINOPATHY

Tarikh : 1 December 2022, Thursday

Tempat : AC Hotel by Marriott Kuala Lumpur

Anjuran : MMLHS (MEMBER OF MALAYSIAN LABORATORY HAEMATOLOGY SOCIETY)

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SITI NADIAH BINTI AZMI

What is Thalassemia?

1. Thalassemia is an inherited blood disorder. It affects your body's ability to produce normal hemoglobin. Hemoglobin is a protein in red blood cells. It allows your red blood cells to transport oxygen throughout your body, nourishing your body's other cells.
2. If you have thalassemia, your body produces fewer healthy hemoglobin proteins, and your bone marrow produces fewer healthy red blood cells. The condition of having fewer red blood cells is called anemia. As red blood cells serve the vital role of delivering oxygen to tissues in your body, not having enough healthy red blood cells can deprive your body's cells of the oxygen they need to make energy and thrive.
3. Thalassemia is genetic disorder with broad spectrum of clinical presentation from silent carrier up to TdT thalassemia or born with hydrops fetalis.
4. Familiar with common types of beta mutation in our population, correlate with clinical phenotype or disease severity in our populations.
5. Interaction between alpha and beta thalassemia with some of variants in our population- it is significant.

What causes thalassemia?

1. Haemoglobin consists of four protein chains:
    - two alpha globin chains
    - two beta globin chains.

    Each chain — both alpha and beta — contains genetic information, or genes, passed down from your parents. Think of these genes as the "code" or programming that controls each chain and (as a result) your haemoglobin. If any of these genes are defective or missing, you'll have thalassemia.

2. Alpha globin protein chains consist of four genes, two from each parent.
3. Beta globin protein chains consist of two genes, one from each parent.
4. The thalassemia you have depends on whether your alpha or beta chain contains the genetic defect. The extent of the defect will determine how severe your condition is.

What are the types of thalassemia?

Thalassemia is classified as:

- trait
- minor
- intermedia
- major

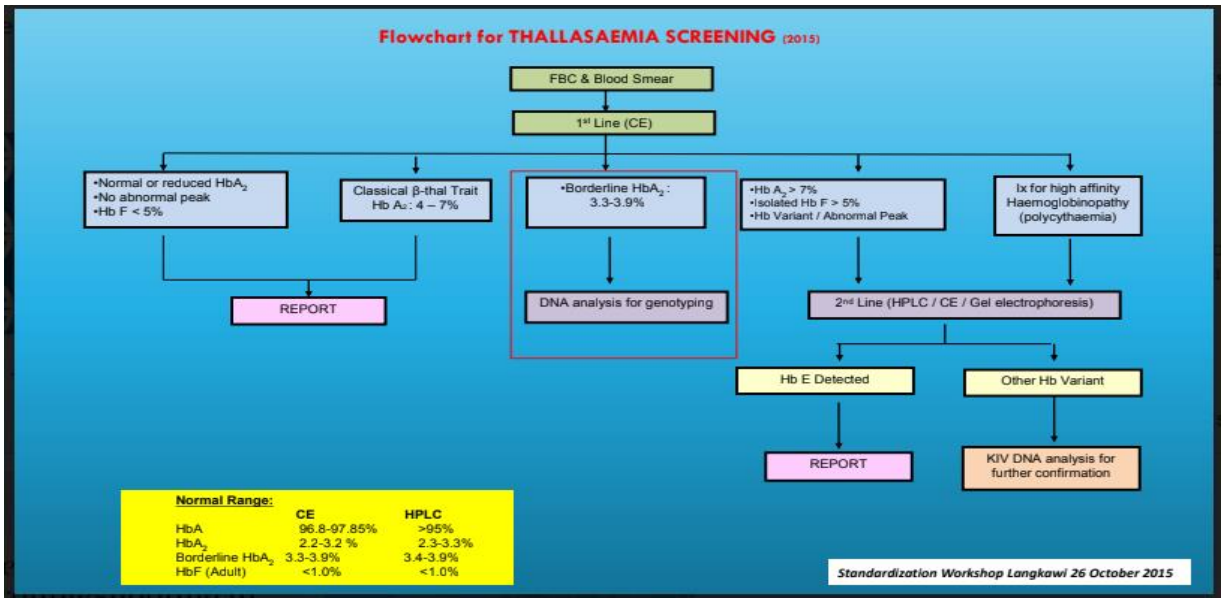
There are two types of thalassemia:

- alpha thalassemia
- beta thalassemia

How is thalassemia diagnosed?

Blood tests to diagnose thalassemia:

1. A complete blood count (FBC/FBP) that includes measures of hemoglobin and the quantity (and size) of red blood cells. People with thalassemia have fewer healthy red blood cells and less hemoglobin than normal. They may also have smaller-than-normal red blood cells.
2. A reticulocyte count (a measure of young red blood cells) may indicate that your bone marrow isn't producing enough red blood cells.
3. Studies of iron will indicate whether the cause of your anemia is an iron deficiency or thalassemia.
4. HbH inclusion screening
5. Haemoglobin electrophoresis is used to diagnose beta thalassemia.
6. Hb quantitation of CE and HPLC
7. Molecular analysis.



Screening and prevention

Must be done at intensive level:

- Screening for carriers
- Genetic counselling
- Facilities for prenatal diagnosis