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Pre-marital Screening for Thalassaemia:

IS IT RELEVANT IN MALAYSIA?

Muhammad Al-Fateh Mohd Yusof
Amrieel Nuqman Mohd Din
Aina Safiyya Shahreel Riza
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PREFACE

The IIUM MBBS SEMINAR SERIES are e-books based on Phase 1 Kulliyyah of Medicine, IIUM, MBBS student seminar presentations. As part of the curriculum, all students are required to present one seminar. Topics covered are varied and not necessarily related to medicine.

This e-book is based on the topic of thalassaemia. It touches on the the disease, its clinical manifestations, screening tests, and the relevance of pre-marital screening in Malaysia.

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INTRODUCTION

Thalassaemia is a group of inherited blood disorders characterised by impaired haemoglobin production, resulting in chronic anaemia and associated complications. To understand thalassaemia, it is crucial to first appreciate the fundamentals of haemopoiesis and the structure of haemoglobin, both of which are central to its pathophysiology.

Hematopoiesis is the process by which the body produces blood cells, including red blood cells (RBCs), white blood cells (WBCs), and platelets. This process is crucial for maintaining oxygen transport, immune defense, and blood clotting. It occurs in specific locations that change as we age. During early embryonic development, hematopoiesis begins in the yolk sac, where primitive blood cells are formed. By the second trimester, it shifts to the liver and spleen, which become the primary sites of blood cell production in the fetus. By the time of birth, the bone marrow takes over as the dominant site for hematopoiesis. In adults, active hematopoiesis occurs primarily in the red bone marrow of flat bones, such as the sternum, pelvis, and vertebrae, as well as the proximal ends of long bones like the femur and humerus.

Haemoglobin (Hb) synthesis is an integral part of red blood cell production. It occurs in the erythroblasts of the bone marrow and involves two main components: heme and globin chains. Heme is synthesised in the mitochondria and cytoplasm of developing erythrocytes, requiring iron as a key element. Globin chains are produced in the ribosomes. The type of haemoglobin varies throughout life: some of it is Foetal haemoglobin (HbF) predominates from mid-gestation to birth and has a high affinity for oxygen to support the foetus. After birth, adult haemoglobin (HbA) gradually becomes predominant, consisting of two alpha and two beta chains.

In summary, haematopoiesis is a dynamic process that begins in the yolk sac, shifts to the liver and spleen during foetal development, and transitions to the bone marrow after birth. Haemoglobin synthesis supports oxygen transport by combining heme and globin chains, adapting to the body’s needs throughout life.

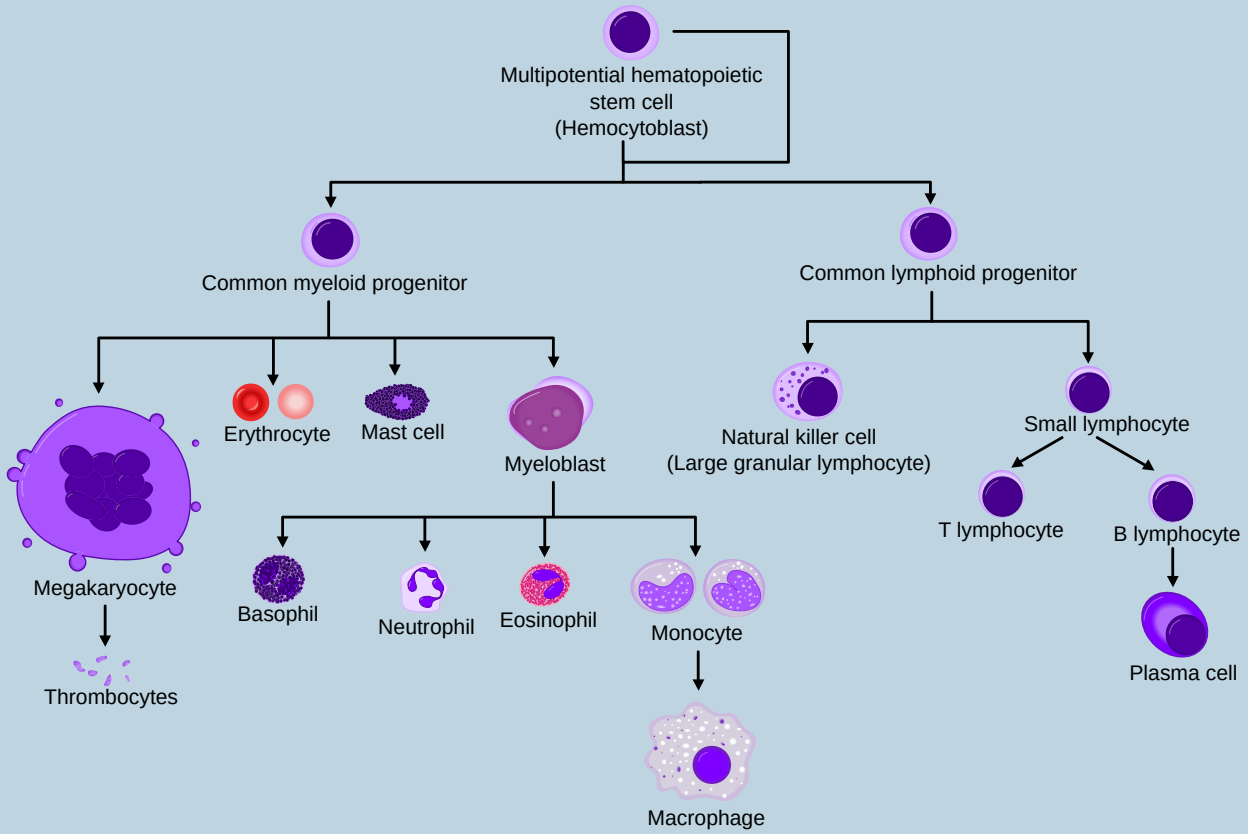


Figure 1: Haematopoiesis

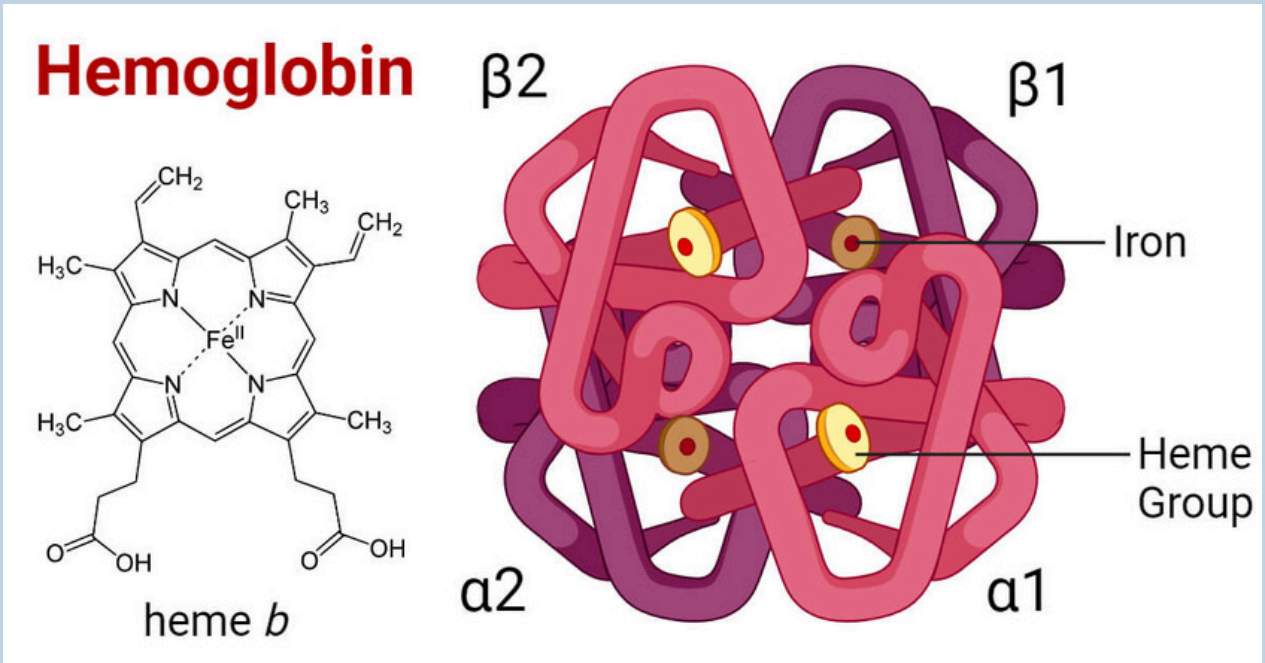
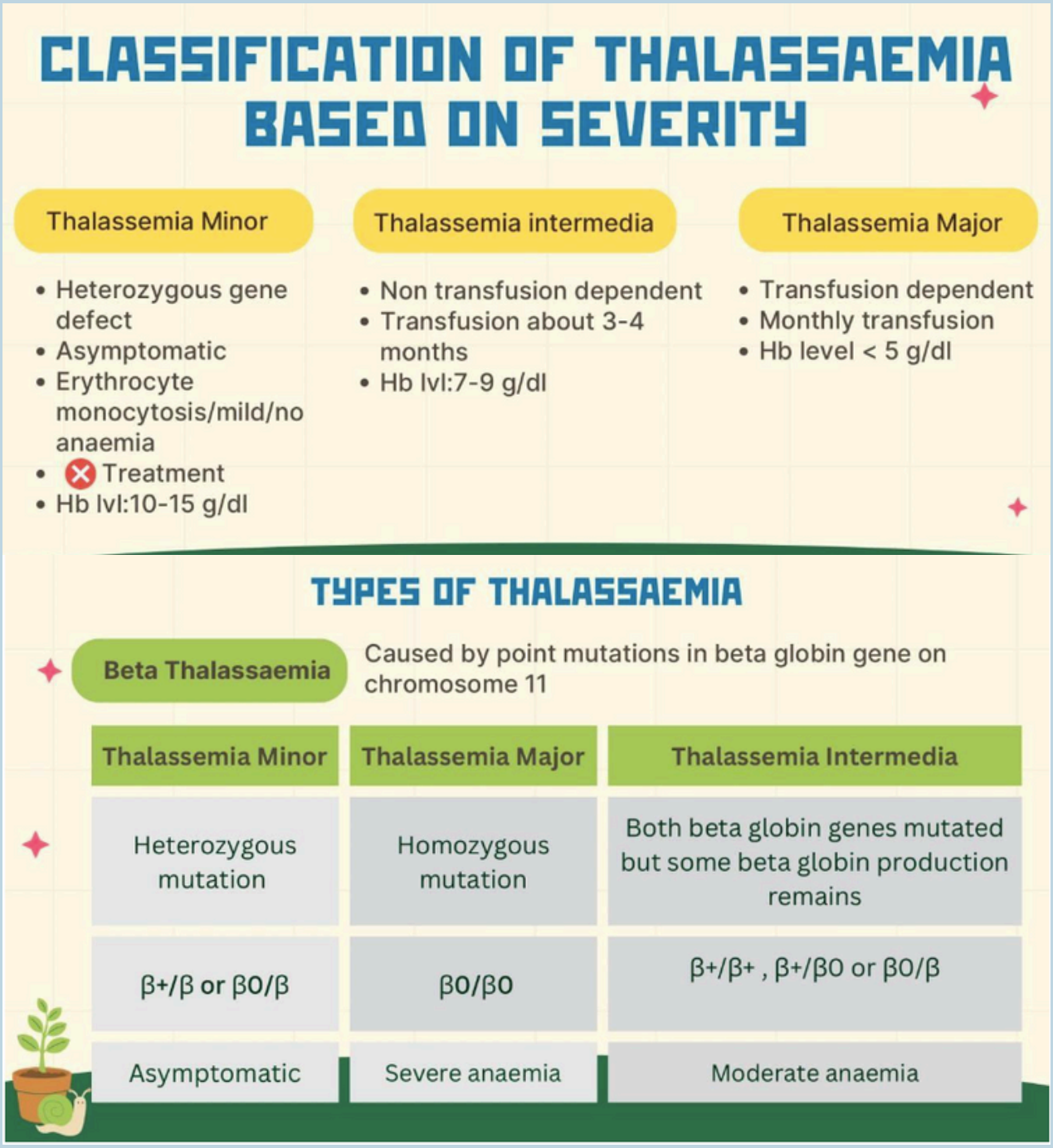
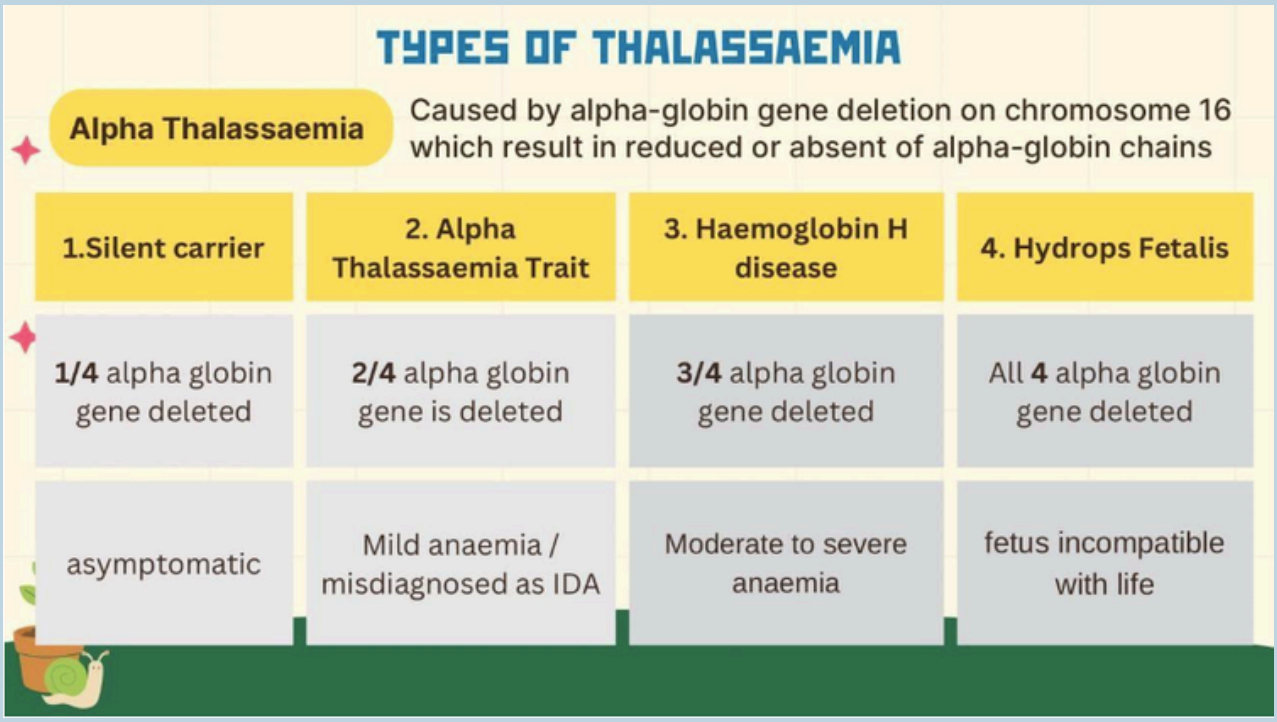


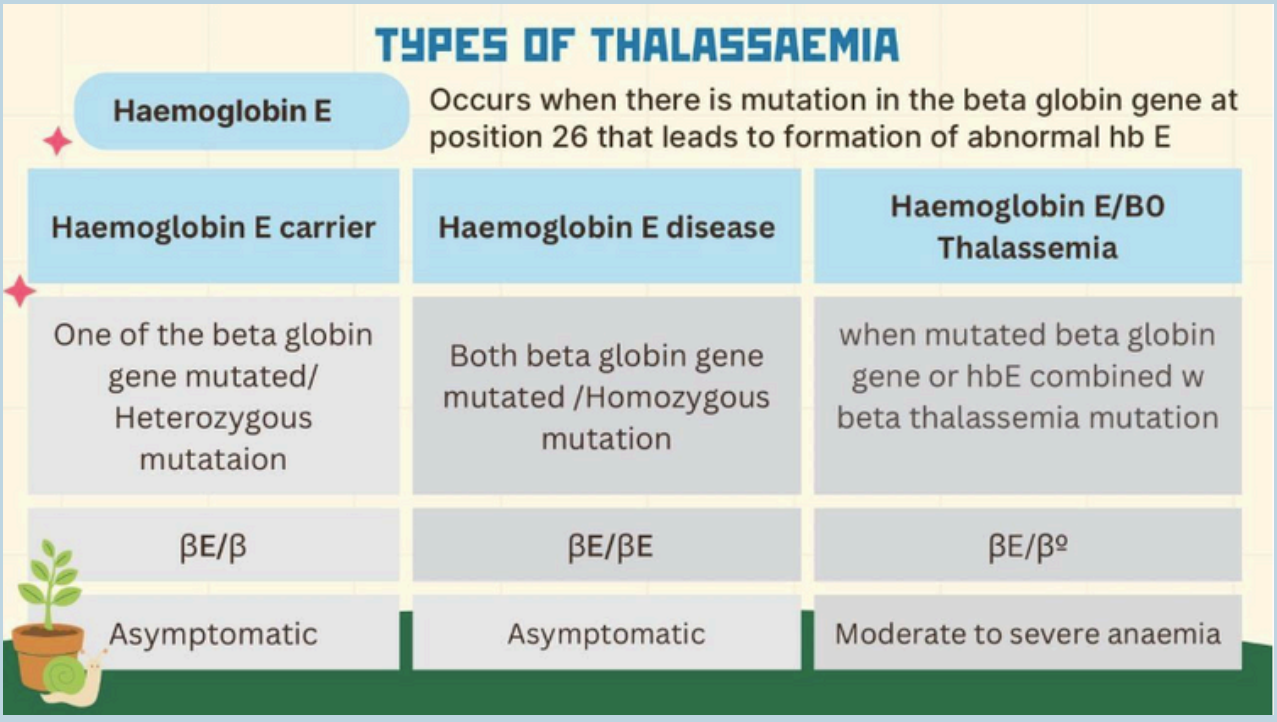
Figure 2: Haemoglobin structure

CLASSIFICATION OF THALASSAEMIA

Thalassaemia is defined as a heterogeneous group of disorders resulting in reduced production of either alpha or beta globin chains.




TYPES OF THALASSAEMIA
Alpha Thalassaemia
Caused by alpha-globin gene deletion on chromosome 16 which result in reduced or absent of alpha-globin chains
1.Silent carrier
2. Alpha Thalassaemia Trait
3. Haemoglobin H disease
4. Hydrops Fetalis
1/4 alpha globin gene deleted
2/4 alpha globin gene is deleted
3/4 alpha globin gene deleted
All 4 alpha globin gene deleted
asymptomatic
Mild anaemia / misdiagnosed as IDA
Moderate to severe anaemia
fetus incompatible with life


TYPES OF THALASSAEMIA
Haemoglobin E
Occurs when there is mutation in the beta globin gene at position 26 that leads to formation of abnormal hb E
Haemoglobin E carrier
Haemoglobin E disease
Haemoglobin E/B0 Thalassemia
One of the beta globin gene mutated/ Heterozygous mutataion
Both beta globin gene mutated /Homozygous mutation
when mutated beta globin gene or hbE combined w beta thalassemia mutation
βE/β
βE/βE
βE/βº
Asymptomatic
Asymptomatic
Moderate to severe anaemia

THALASSEMIA IN MALAYSIA

Thalassemia is a major public health concern worldwide based on Word Health Organisations(WHO). Thalassemia is also a major public health concern in Malaysia. In South Asia, 6.8% from the population are actually thalassemia carriers and in Malaysia 4.5-5% from the populations are carriers.

The Malaysian Thalassemia Registry (MTR) was officially launched on 12 May 2007 involving 137 hospitals available for thalassemia screening and 110 hospitals available as treatment centers including Hospital Tengku Ampuan Afzan (HTAA), University Malaya Medical Center and Hospital Tengku Ampuan Rahimah (HTAR). In the MTR website, all of the patients' data from the day they were diagnosed were recorded by research assistants in various regional centers in Malaysia. Besides that, other data such as the clinical characteristics, signs and symptoms, lab results findings, type of treatments the patient received and complications from were also recorded.

Based on MTR the prevalence of thalassemia from 2007-2019 was around 8178 cases. However, this is just the total number of cases that registered in the program, thus the total number of cases from those years might actually be higher.

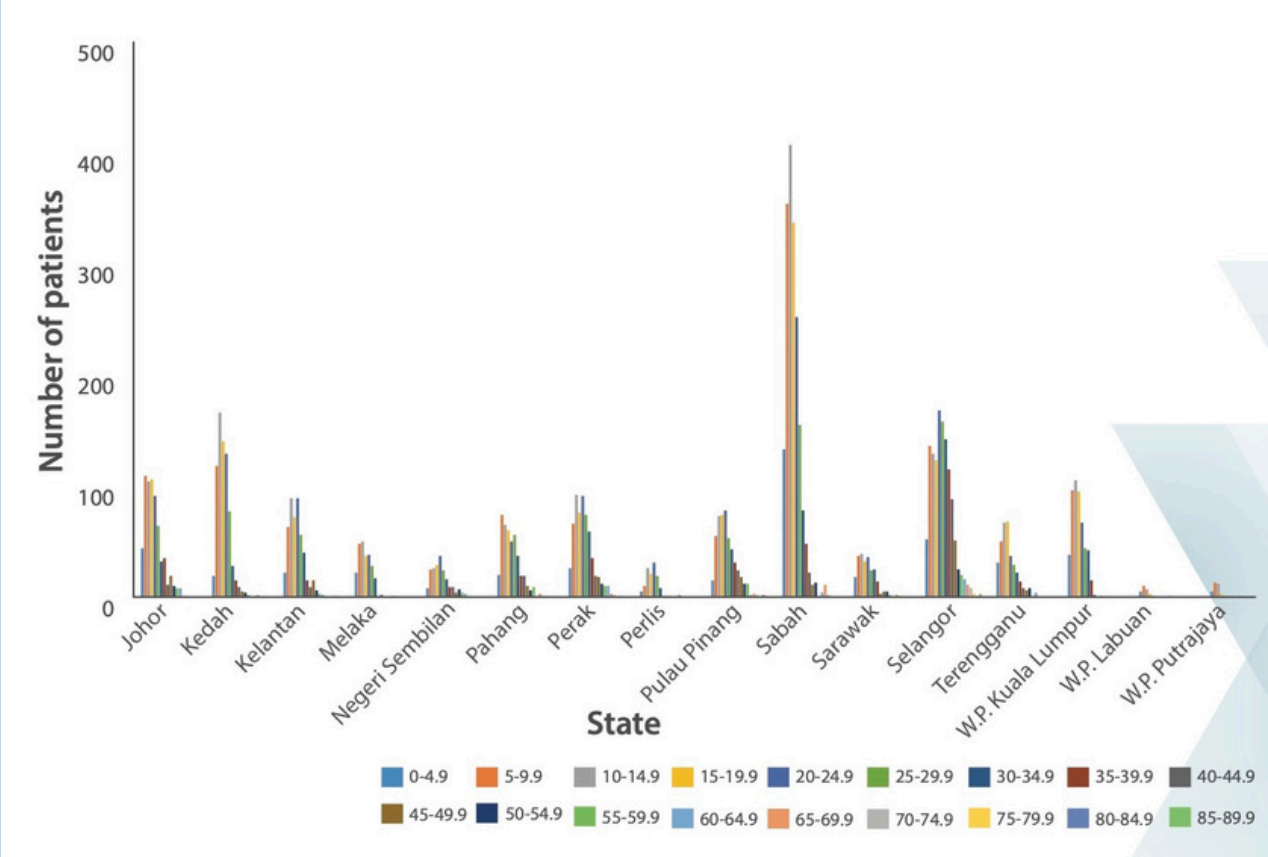


Figure 3: Distribution of Thalassaemia Patients in Malaysia According to Age Group

Source: Annual Report of the Malaysian Thalassemia Registry 2019

According to the MTR the mean patient age was 20.9 ± 13.3 years. Sabah has the highest total number of cases reported followed by Selangor and Kuala Lumpur.

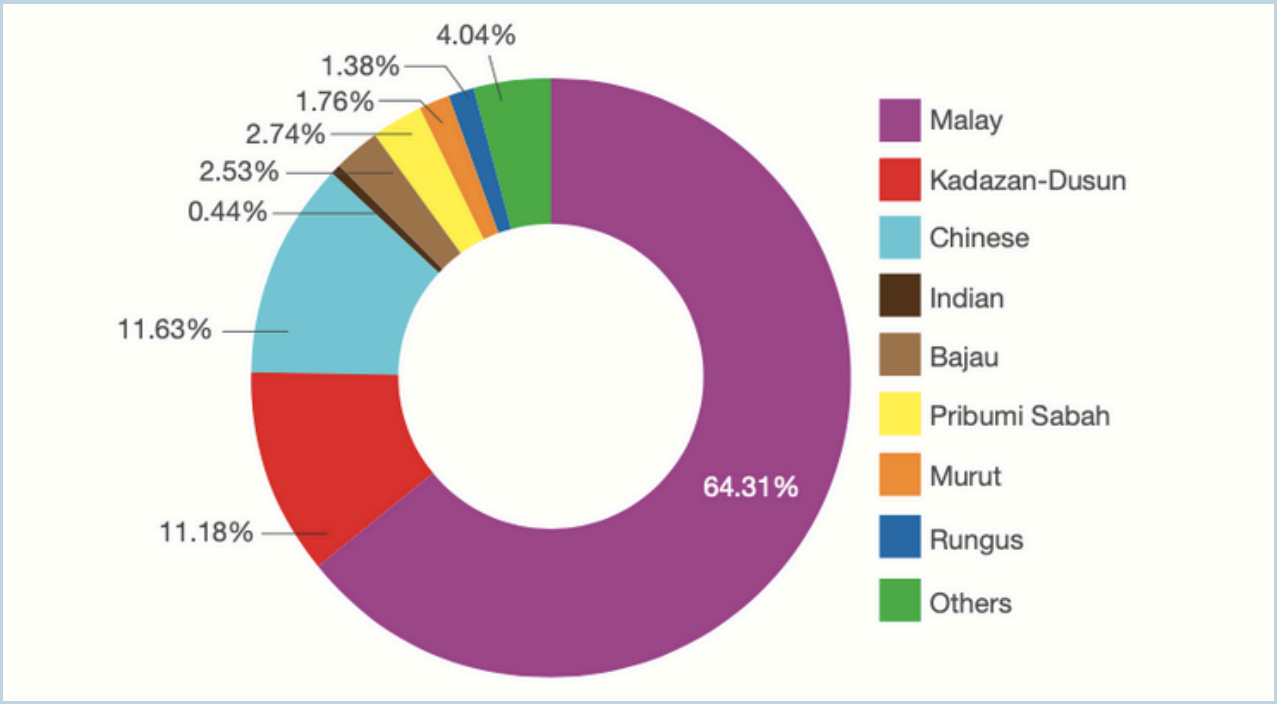


Figure 4: Distribution of Thalassaemia Patients in Malaysia by Ethnic Group

Source: Annual Report of the Malaysian Thalassemia Registry 2019

As we all know Malaysia consists of many beautiful ethnic groups such as Malay, Chinese, Indian and others. According to the data from MTR, Malays have the highest cases reported with 5259 out of 8178 cases followed by Chinese with 951 cases and Kadazan Dusun with 914 cases. This may account for the high number of thalassemia in Sabah.

State	Total Number of Patients	Malay		Chinese		Indian		Kadazan-Dusun	
		No.	%	No.	%	No.	%	No.	%
Peninsular Malaysia	6078	5044	82.99	764	12.57	35	0.58	42	0.69
Sabah + Labuan	1856	109	5.87	96	5.17	1	0.05	868	46.77
Sarawak	244	106	43.44	90	36.89	0	0.00	4	1.64
Total	8178	5259		950		36		914	

Note: Percentage (%) is calculated based on the geographic regional total.

Table 1: Distribution of Patients by Major Ethnic Groups Based on Modified Geographical Regions

Source: Annual Report of the Malaysian Thalassemia Registry 2019

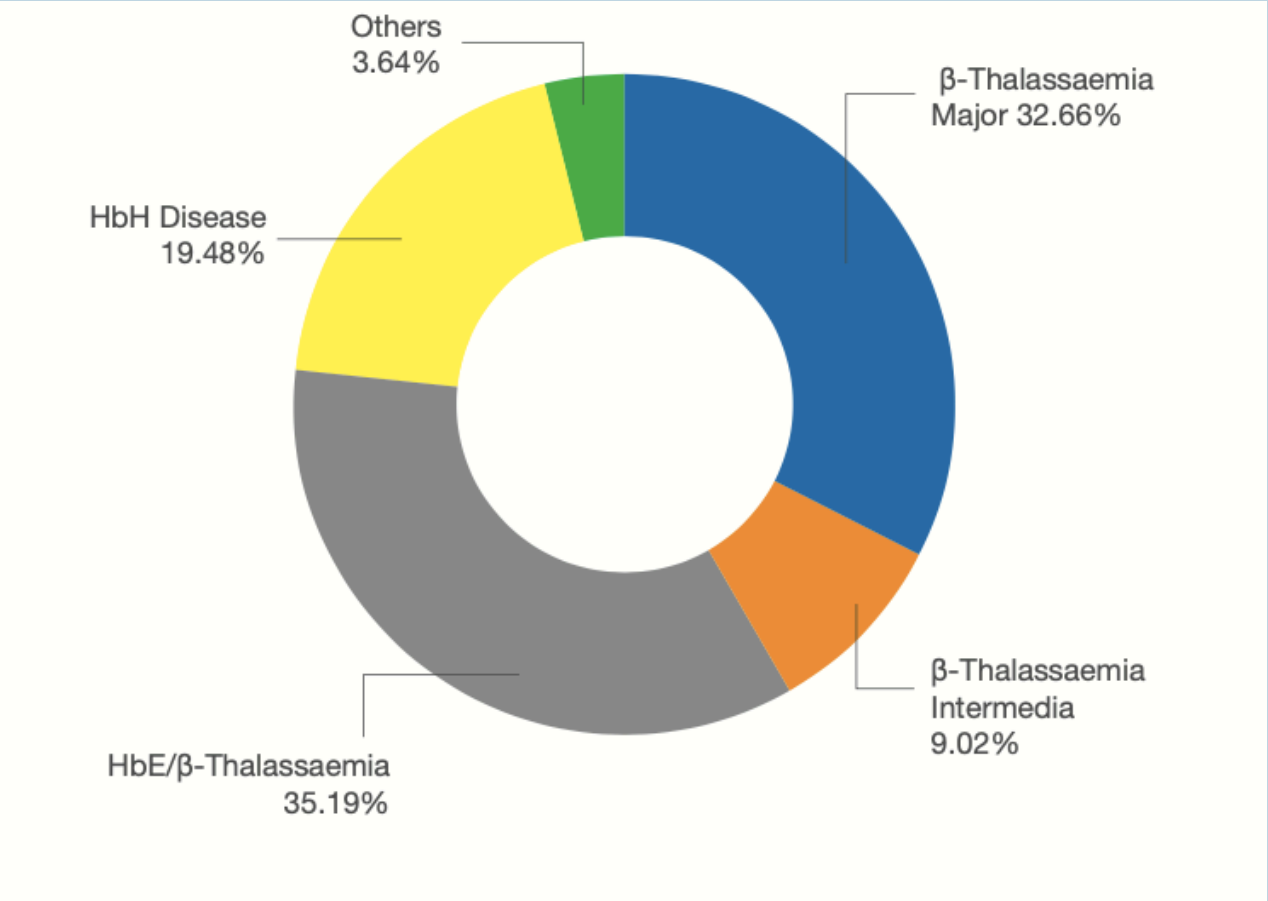


Figure 5: Distribution of Patients in Malaysia by Diagnosis in 2019

Source: Annual Report of the Malaysian Thalassemia Registry 2019

The interaction of HbE with β-thalassaemia results in HbE/β-thalassaemia, an extremely heterogeneous clinical condition. This is the most common form of β-thalassaemia in Southeast Asia. In Malaysia the most common is HbE/β-thalassaemia with 2878 cases, beta thalassemia 2671 cases and Hbh Disease with 1593 cases.

Year	Total Number of Patients	Number of NTDT Patients	Number of TDT Patients	Total Number of Transfusions
2019	8178	3460	4718	17468
2018	7984	3455	4529	33497
2017	7882	3392	4490	27226
2016	7605	3235	4370	21230
2015	7217	3014	4203	17570
2014	6805	2787	4018	15649
2013	6386	2554	3832	15173
2012	5973	2345	3628	15160
2011	5547	2146	3401	13011
2010	5164	1962	3202	16885

Note: Some missing data for year 2019.

Table 2: Distribution of Patients According to Transfusion Requirement by Year

Source: Annual Report of the Malaysian Thalassemia Registry 2019

One of the principles for treatment and management for thalassemia patients is blood transfusion depending on the severity of their cases. In MTR data, Malaysia has 4178 Transfusion Dependent Thalassemia (TDT) cases needing blood transfusions for every 2 to 3 weeks while the remaining 3460 cases are Non Transfusion Dependent Thalassemia (NTDT) patients.

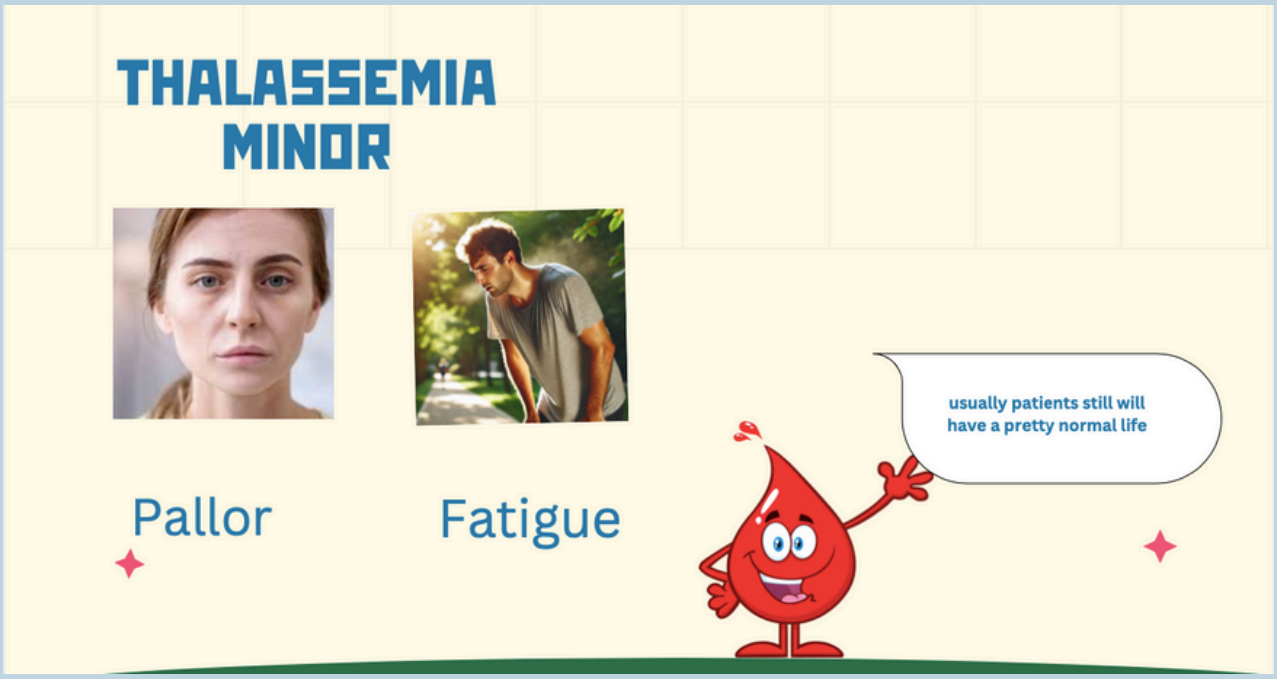
State	Number of Patients
Johor	36
Kedah	32
Kelantan	34
Melaka	5
Negeri Sembilan	10
Pahang	48
Perak	47
Perlis	14
Pulau Pinang	15
Sabah	318
Sarawak	21
Selangor	82
Terengganu	28
W.P. Kuala Lumpur	90
W.P. Labuan	4
W.P. Putrajaya	1
Total	786

Table 3: Cumulative Reported Deaths by State from 2007 until October 2019

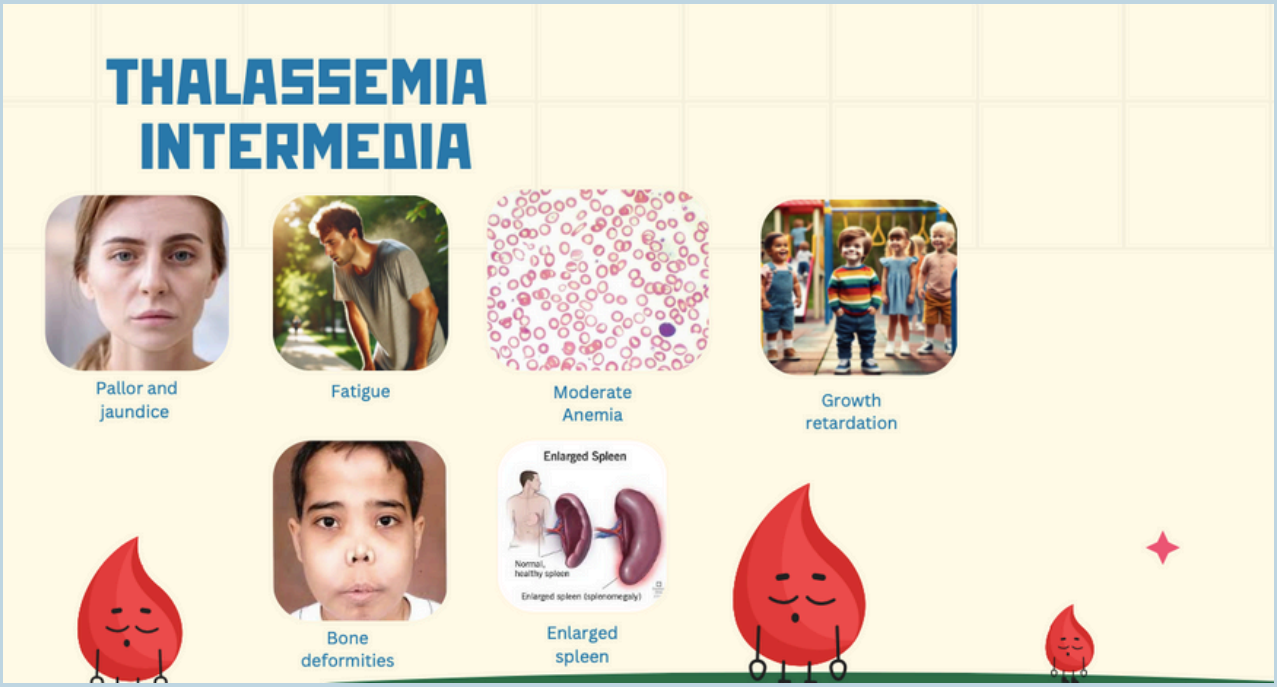
Source: Annual Report of the Malaysian Thalassemia Registry 2019

There are 786 total deaths reported in the MTR with highest cases recorded in Sabah followed by Selangor and Kuala Lumpur. The total number of thalassemia cases in Malaysia has been decreasing each year. This is because of the successful initiative from the Ministry of Health which is the screening program targeting 4 high school students. The program for high school thalassemia screening started in 2016 while in Malaysia the screening program began in 2004. Moreover, there is also increasing awareness among the public regarding the importance of screening to ensure the health of their future children.

SIGNS AND SYMPTOMS OF THALASSAEMIA



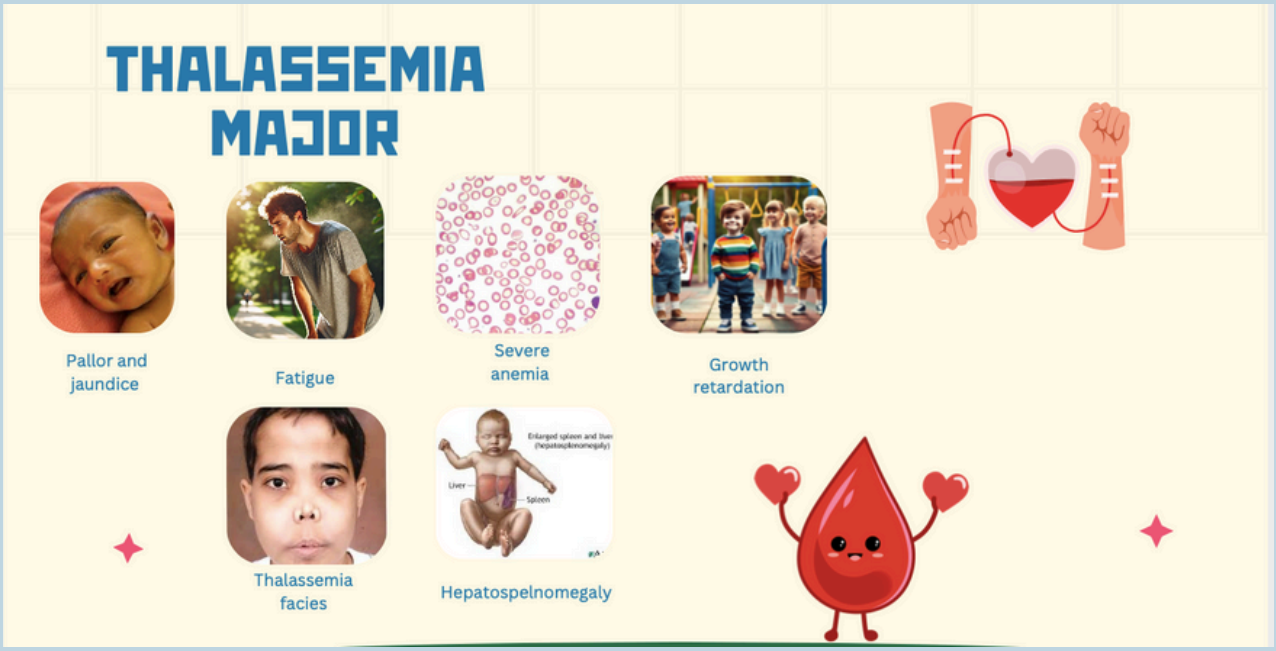
Thalassemia minor (also called thalassemia trait) is typically a mild form of thalassemia, and individuals may either be asymptomatic or have mild symptoms. Most individuals are asymptomatic and lead normal lives without any complications. Symptoms, if present, are often non-specific, such as mild tiredness or slight paleness.



Thalassemia intermedia is a moderate form of thalassemia that falls between thalassemia minor and thalassemia major in severity. It usually manifests with variable clinical symptoms depending on the genetic mutations and the degree of anaemia. Unlike thalassemia major, patients with thalassemia intermedia can often maintain some level of haemoglobin production without regular blood transfusions, though they may require medical management in certain situations.

While patients with thalassemia intermedia do not typically require regular transfusions, certain situations can exacerbate anemia and lead to worsening symptoms, such as:

- Infections
- Pregnancy
- Increased haemolysis during stress or illness



Thalassemia major (also known as Cooley's anemia) is the most severe form of thalassemia. It typically presents early in life and requires regular medical care, including frequent blood transfusions, to manage the symptoms and prevent complications.

Symptoms of severe anaemia usually appear within the first 6–24 months of life and may include:

- Profound fatigue.
- Weakness and lethargy.
- Pallor (pale skin).

Patients also have failure to thrive, jaundice, bone deformities, splenomegaly, hepatomegaly, delayed puberty, darkening of skin and short stature.

Frequent blood transfusions and increased intestinal iron absorption lead to excessive iron in the body, which can affect:

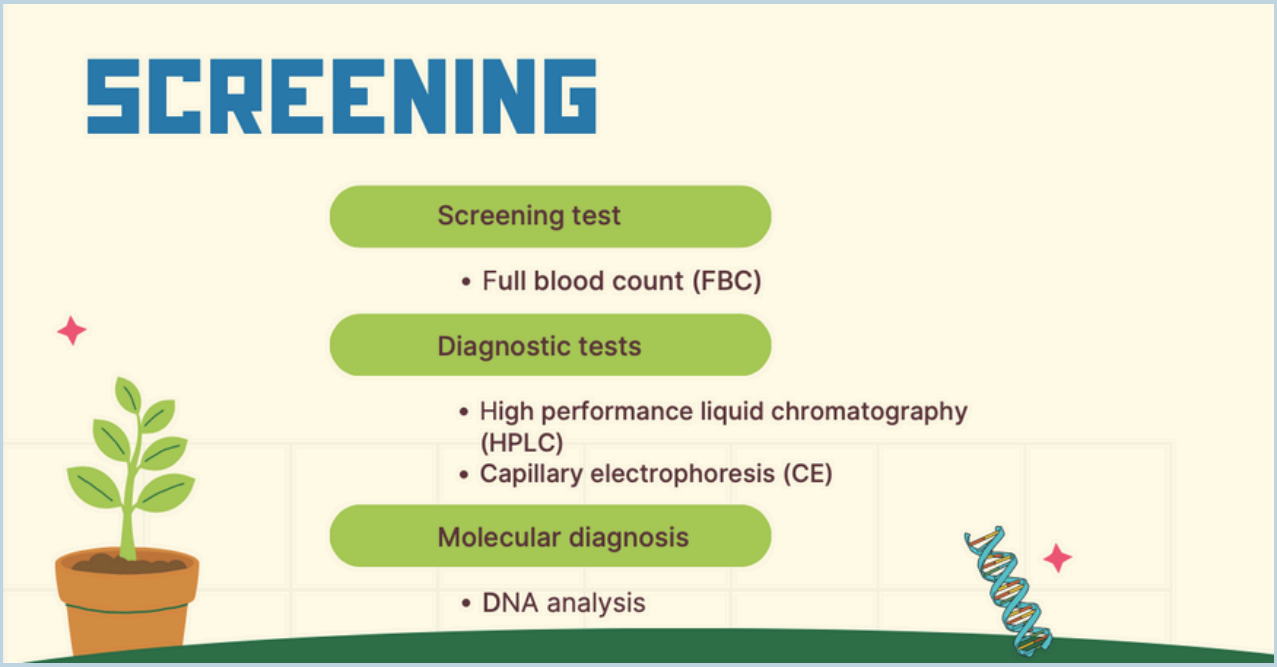
- The heart (cardiomyopathy, arrhythmias, heart failure).
- The liver (liver damage, cirrhosis).
- Endocrine organs (diabetes, hypothyroidism, hypogonadism).

Severe anaemia and iron overload can strain the cardiovascular system can lead to heart failure and pulmonary hypertension.

If left untreated or inadequately managed, thalassemia major can lead to life-threatening complications, including severe heart or liver disease, endocrine dysfunction, osteoporosis and fractures.

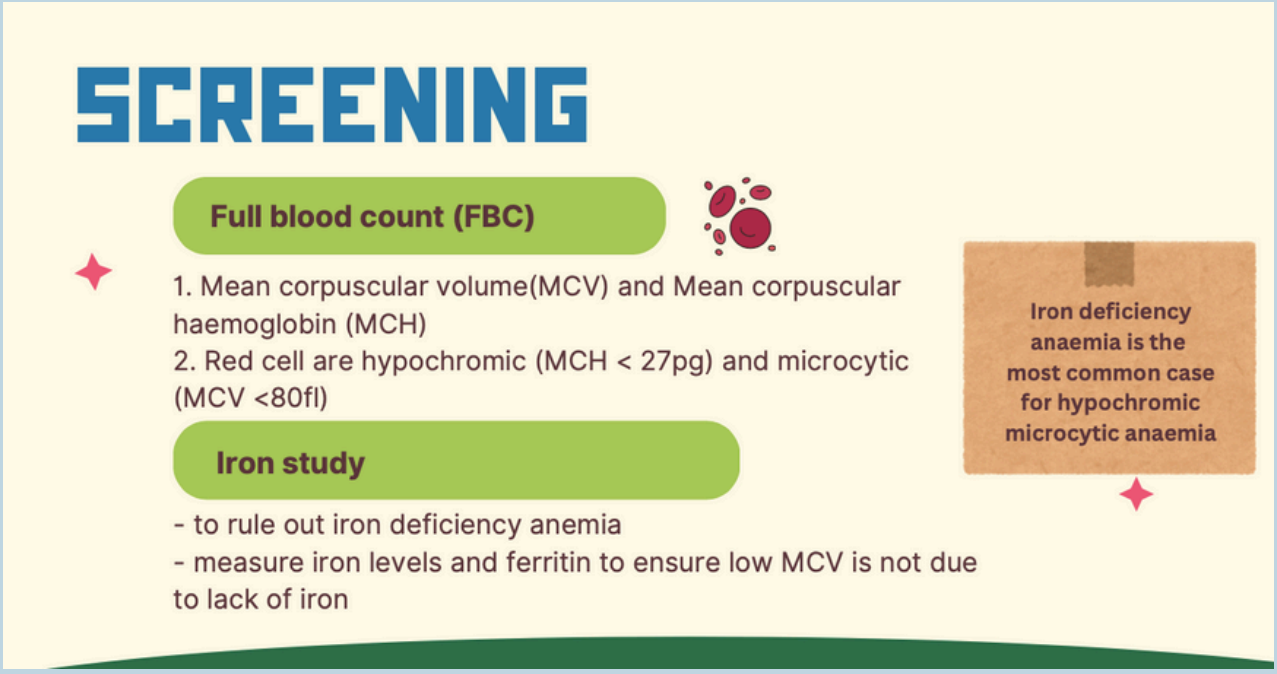
SCREENING OF THALASSAEMIA

Thalassaemia screening involves several steps to ensure accurate diagnosis and identification of the condition. The process begins with a blood test to perform a full blood count (FBC), which provides a detailed analysis of blood components. If the FBC results are abnormal, further diagnostic tests are carried out for confirmation. These include High-Performance Liquid Chromatography (HPLC) and Capillary Electrophoresis (CE), which are used to identify abnormal haemoglobin types. These tests allow for the precise quantification of haemoglobin variants such as HbA2 and HbF. In thalassaemia, HbA2 and HbF levels may be elevated, making these tests particularly useful for detecting beta-thalassaemia.

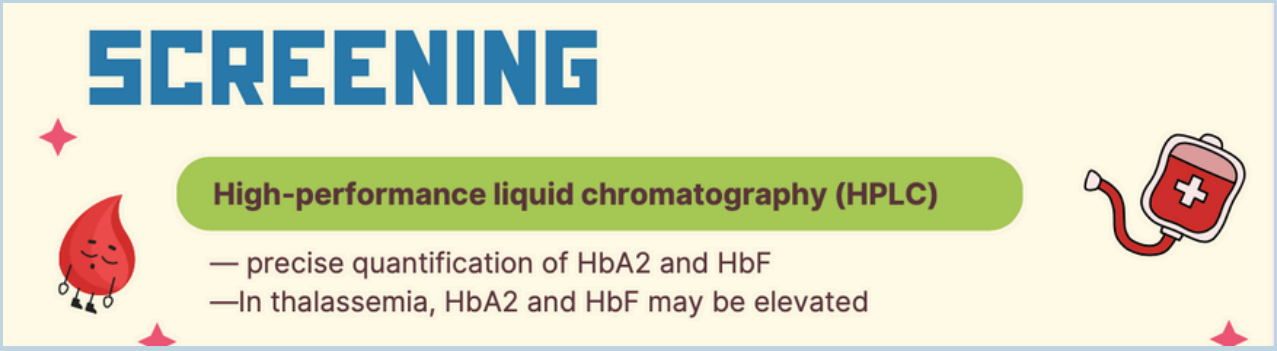


Additionally, molecular diagnosis through DNA analysis is conducted to confirm specific genetic mutations responsible for thalassaemia. This test is particularly important for identifying carrier status and more complex forms of the condition, such as alpha-thalassaemia.

The FBC results are analysed based on two key parameters: the Mean Corpuscular Volume (MCV), which indicates the average size of red blood cells, and the Mean Corpuscular Haemoglobin (MCH), which measures the average haemoglobin content in red blood cells. Abnormal FBC results are characterised by low haemoglobin levels, an MCV of less than 80 femtolitres, and an MCH of less than 27 picograms. These findings suggest hypochromic microcytic anaemia, a hallmark of thalassaemia. Following this, an iron study is performed to rule out iron deficiency anaemia (IDA), as it is the most common cause of hypochromic microcytic anaemia. The iron study measures serum iron and serum ferritin levels to determine if the low MCV and MCH values are due to iron deficiency rather than thalassaemia.



Ruling out IDA is a crucial step since it shares similar characteristics with thalassaemia, such as hypochromic microcytic anaemia. Differentiating between these two conditions ensures accurate diagnosis and appropriate management. For advanced diagnostic testing, HPLC provides precise measurements of HbA2 and HbF levels, which may be elevated in thalassaemia, indicating an abnormal proportion of haemoglobin. Molecular testing through DNA analysis identifies specific gene mutations responsible for thalassaemia, which is critical for confirming carrier status and diagnosing complex forms of the disease. By following this systematic approach, thalassaemia screening ensures accurate diagnosis, enabling proper management and counselling for affected individuals and carriers.



THALASSAEMIA MODE OF INHERITANCE

Thalassaemia is inherited in an autosomal recessive manner, where the severity of the condition depends on the specific genetic mutations inherited from both parents. If one parent is a carrier (thalassaemia minor), the child has a 50% chance of being a carrier, but no risk of developing severe disease unless both parents are carriers. When two carriers marry, there is a 25% chance of thalassaemia major, a 50% chance of the child being a carrier, and a 25% chance of no mutation inheritance. The inheritance pattern becomes more complex in cases involving different types of thalassaemia, such as alpha-thalassaemia, beta-thalassaemia, and haemoglobin E. For instance, marriages between HbE carriers and beta-thalassaemia carriers can result in HbE-Beta-Thalassaemia, which ranges from mild to severe.

Parent Combination	Possible Offspring	Symptoms
Beta-Thal Carrier + Non-Carrier	50% Beta-Thal Carrier, 50% Normal	No symptoms in offspring.
Beta-Thal Carrier + Beta-Thal Carrier	25% Beta-Thal Major, 50% Beta-Thal Carrier, 25% Normal	Severe symptoms (Beta-Thal Major) or asymptomatic carrier.
Beta-Thal Major + Non-Carrier	100% Beta-Thal Carrier	All children are asymptomatic carriers.
Beta-Thal Major + Beta-Thal Carrier	50% Beta-Thal Major, 50% Beta-Thal Carrier	Severe symptoms in 50% (Beta-Thal Major).
Beta-Thal Major + Beta-Thal Major	100% Beta-Thal Major	All offspring have severe symptoms.

Parent Combination	Possible Offspring	Symptoms
Alpha-Thal Carrier (1 deletion) + Non-Carrier	50% Alpha-Thal Carrier, 50% Normal	No symptoms in offspring.
Alpha-Thal Carrier (2 deletions) + Non-Carrier	50% Alpha-Thal Trait (2 deletions), 50% Normal	Mild anaemia possible in Alpha-Thal Trait offspring.
Alpha-Thal Carrier (1 deletion) + Alpha-Thal Carrier (1 deletion)	25% Alpha-Thal Major (4 deletions), 50% Alpha-Thal Trait (2 deletions), 25% Normal	Severe symptoms (hydrops fetalis) in Alpha-Thal Major; mild anaemia in Alpha-Thal Trait.
Alpha-Thal Trait (2 deletions) + Alpha-Thal Trait (2 deletions)	25% HbH Disease, 50% Alpha-Thal Trait, 25% Normal	Severe anaemia (HbH Disease) in 25%; mild anaemia in Alpha-Thal Trait.
Alpha-Thal Trait + HbH Disease	50% HbH Disease, 50% Alpha-Thal Trait	Moderate to severe anaemia.
HbH Disease + HbH Disease	25% Hb Bart's (Hydrops Fetalis), 50% HbH Disease, 25% Alpha-Thal Trait	Fatal in Hydrops Fetalis; moderate to severe anaemia in HbH Disease.

Parent Combination	Possible Offspring	Symptoms
HbE Carrier (HbAE) + Non-Carrier	50% HbE Carrier, 50% Normal	No symptoms.
HbE Carrier (HbAE) + HbE Carrier (HbAE)	25% HbEE Disease, 50% HbE Carrier, 25% Normal	Mild anaemia in HbEE Disease.
HbE Carrier (HbAE) + Beta-Thal Carrier	25% HbE-Beta-Thalassaemia, 25% HbE Carrier, 25% Beta-Thal Carrier, 25% Normal	HbE-Beta-Thalassaemia ranges from mild to severe anaemia.
HbE Carrier (HbAE) + Beta-Thal Major	50% HbE-Beta-Thalassaemia, 50% Beta-Thal Carrier	Moderate to severe anaemia in HbE-Beta-Thalassaemia.
HbE-Beta-Thal + Non-Carrier	50% HbE Carrier, 50% Beta-Thal Carrier	Mild anaemia possible in carriers.
HbE-Beta-Thal + HbE Carrier (HbAE)	50% HbE-Beta-Thalassaemia, 50% HbE Carrier	Severity depends on genetic mutation; ranges from mild to moderate anaemia.
HbE-Beta-Thal + Beta-Thal Carrier	Complex combinations, often leading to severe anaemia ↓	Severe forms like HbE-Beta-Thalassaemia or Beta-Thal Major likely.

MANAGEMENT OF THALASSAEMIA

MANAGEMENT OF THALASSAEMIA MINOR

Thalassaemia minor is generally asymptomatic and requires minimal medical intervention. However, patients should be informed about their carrier status and the implications for family planning, particularly the risk of passing the gene to offspring. Therefore, genetic counselling is essential for couples in which one or both partners are carriers, particularly to inform them of the risks of having children with thalassemia major.

MANAGEMENT OF THALASSAEMIA INTERMEDIA

Thalassemia intermedia presents with moderate anaemia and complications that are less severe than thalassemia major but may still require medical attention. The management aims to prevent complications and improve quality of life. Regular monitoring of haemoglobin levels, growth, and development is essential, especially in children. Patients are often prescribed folic acid supplementation to support red blood cell production.

Blood transfusions are generally required only intermittently, during periods of physiological stress such as infections, pregnancy, or surgery, or when haemoglobin levels drop significantly. Unlike thalassemia major, transfusions are not typically needed on a regular basis unless complications arise. Chronic transfusions or increased intestinal iron absorption may lead to iron overload, in which case iron chelation therapy is started.

Splenomegaly, common in thalassemia intermedia, may require intervention if it causes discomfort or worsens anaemia. Splenectomy is considered if the spleen significantly impacts red blood cell survival or haemoglobin levels. Patients with thalassaemia intermedia are also at risk of complications such as thromboembolic events, requiring anticoagulation therapy in specific cases. Bone health issues, including osteopenia and osteoporosis, are addressed with calcium and vitamin D supplementation or bisphosphonates. Screening and management of pulmonary hypertension are recommended if symptoms develop. Genetic counselling is an integral part of care, particularly for couples at risk of having children with thalassemia major, providing them with information on options such as prenatal diagnosis or assisted reproductive techniques. These measures aim to manage symptoms, reduce complications, and enhance the overall well-being of patients with thalassaemia intermedia.

MANAGEMENT OF THALASSAEMIA MAJOR

The management of thalassaemia major focuses on regular blood transfusions, iron chelation therapy, and long-term monitoring to prevent complications and improve survival. Regular blood transfusions are the cornerstone of treatment, typically required every 2–4 weeks, to maintain haemoglobin levels between 9–10 g/dL. This helps prevent severe anaemia and its associated complications, such as growth delays and organ damage. However, repeated transfusions lead to iron overload, which necessitates iron chelation therapy to remove excess iron and prevent damage to vital organs. Common chelators include deferoxamine, deferasirox, and deferiprone, with treatment tailored to the patient's needs and monitored using serum ferritin levels and MRI scans of the liver and heart.

Splenectomy may be performed in patients with severe splenomegaly or when the spleen significantly increases red blood cell destruction, although the procedure carries risks such as increased susceptibility to infections. Vaccinations and prophylactic antibiotics are essential for patients who undergo splenectomy.

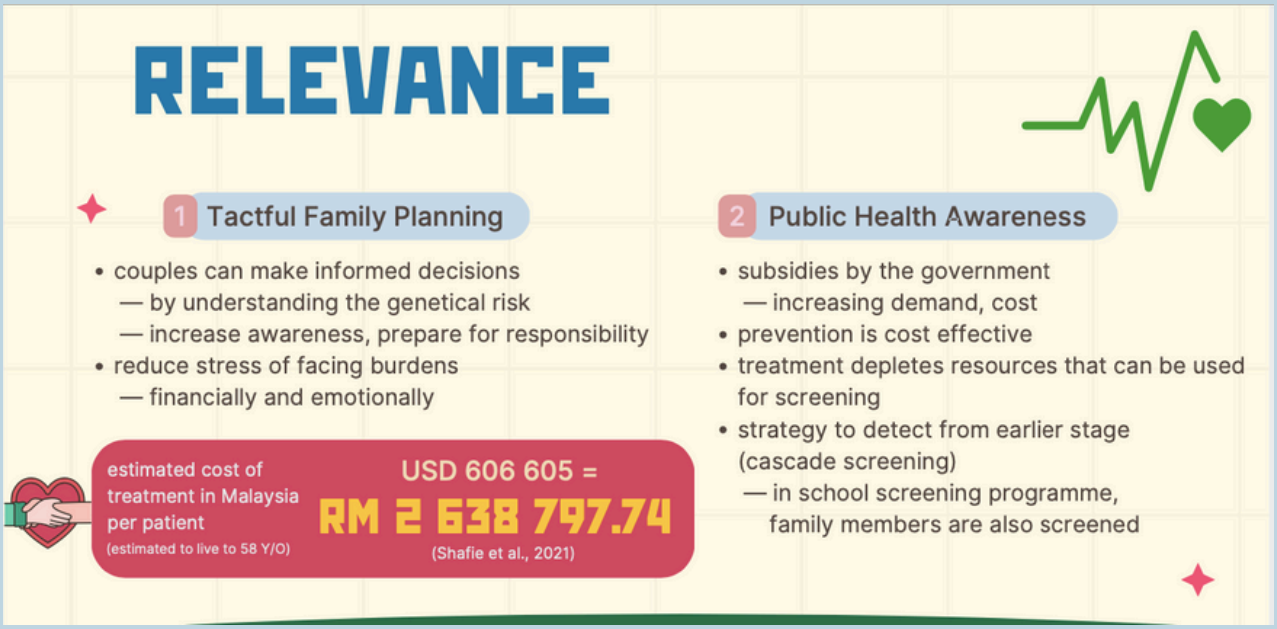
Bone marrow or stem cell transplantation offers a potential cure for thalassaemia major, particularly in patients under 16 years old with an HLA-matched donor, though the procedure is costly and carries significant risks. Regular monitoring is vital to manage complications of iron overload, including cardiomyopathy, liver disease, and endocrine dysfunctions such as diabetes and hypothyroidism.

Supportive care, including monitoring of growth, development, and psychosocial support, is integral to ensuring quality of life. Genetic counselling plays a crucial role in helping families understand the inheritance pattern and risks of the disease, particularly for those planning pregnancies. Emergency care is required for acute complications like severe anaemia or iron toxicity. With advances in treatment, life expectancy has improved significantly for individuals with thalassaemia major, but lifelong management remains essential to address the chronic nature of the disease.

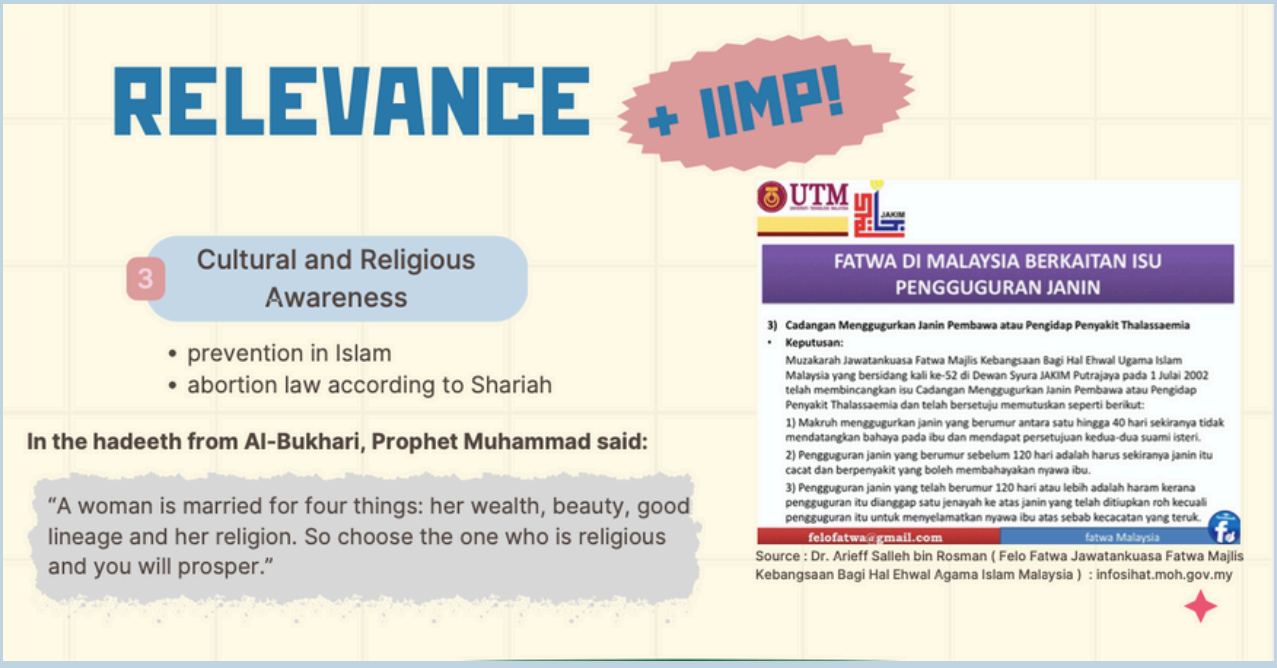
Aspect	Thalassaemia Minor	Thalassaemia Intermedia	Thalassaemia Major
Anaemia Management	Rarely needed	Periodic transfusions as needed	Regular transfusions required
Iron Chelation	Rarely needed	If iron overload occurs	Essential with transfusions
Splenectomy	Not required	If splenomegaly impacts anaemia	Common for severe splenomegaly
Complication Management	Minimal	Thrombosis, bone health, etc.	Extensive monitoring required
Genetic Counselling	For carriers	For carriers and affected families	For families and long-term planning

CONCLUSION

The Malaysian Thalassaemia Screening Programme is a cornerstone of the nation's efforts to tackle the significant public health burden of thalassaemia. Established in 2004 by the Ministry of Health (MOH), the programme was developed in response to the high prevalence of thalassaemia carriers in Malaysia—estimated at around 4.5% of the population—and the increasing demand for blood transfusions and chelation therapy among patients with thalassaemia major. Drawing from successful models in countries like Cyprus and Sardinia, the initiative aimed to reduce the incidence of severe thalassaemia cases through early detection and preventive measures.



The programme's implementation underscores its relevance to public health, tactful family planning, and cultural and Islamic values. Screening is made widely accessible through schools, government clinics, and hospitals, targeting adolescents and couples planning for marriage. By identifying carriers early, the programme empowers individuals and couples to make informed reproductive decisions, significantly reducing the risk of children inheriting severe forms of thalassaemia. This aligns with the Islamic principle of maslahah (public benefit), as well as cultural values that prioritise family health and community well-being. Genetic counselling provided as part of the programme supports these efforts by offering guidance that respects personal and religious beliefs, ensuring that decisions are informed and compassionate.



The programme's impact has been substantial, improving carrier detection rates and reducing new cases of thalassaemia major. Advances in diagnostic tools, such as High-Performance Liquid Chromatography (HPLC) and DNA analysis, have enhanced the accuracy of screenings. Public awareness campaigns, often conducted in collaboration with NGOs like the Malaysian Thalassaemia Association, have further amplified the programme's reach. However, challenges persist, including addressing gaps in awareness among rural populations and overcoming stigma associated with carrier status.

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Thalassemia Screening

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This e-book is based on the topic of thalassaemia. It touches on the the disease, its clinical manifestations, screening tests, and the relevance of pre-marital screening in Malaysia.

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