

# [Beta thalassemia: causes and treatments](https://assignbuster.com/beta-thalassemia-causes-and-treatments/)

Thalassemia is an inherited autosomal recessive blood disease that affects the body’s ability to produce haemoglobin, the protein that red blood cells use to carry oxygen. It reduces the rate of production of globin chains that make up haemoglobin, which are either alpha (Î±) chain or beta(Î²) chain. Beta-Thalassemia Major disorder occurs when both the beta globin chain are affected by mutation or deletion, resulting in the abnormalities in the formation, size and shape of red blood cell. Patients suffering from beta-thalassemia have to endure the pain from severe anemia, fatigue, enlarged spleen, and also bone problems besides jaundice. Thalassemia is found in almost every country worldwide, however it is more commonly found in Mediterranean and Asians. The data from the National Thalassemia Registry shows that currently there are 4, 768 thalassemia patients registered in Malaysia. Based on the screening conducted by Ministry of Health, the carrier rate averages at about 5%, which means about one in 20 Malaysians carry the thalassemia gene. The diagram attached in the next page shows an overview of the pathogenesis of Î² thalassemia. Thalassemia can be treated and prevented by many different screening test and treatments. However, thalassemia is not curable yet. So, what are the current available treatments to help relieve the disorder symptoms?

## A POSSIBLE SOLUTION

The treatment for thalassemia depends on the severity of the conditions. For example, people who are carriers or have a mild Î± or Î² thalassemia trait which usually does not have any symptoms, need little or no treatment. People with moderate and severe condition of thalassemia need treatments as soon as possible or else they are under dangerous situation that can cause fatal.

### BLOOD TRANSFUSION

As the red blood cells of thalassemic patients are little and not healthy, it couldn’t carry out its normal function of carrying oxygen efficiently which may cause the patient to have difficulties in breathing and other related problems. Therefore, the most effective treatment for these patients is blood transfusion therapy.

Blood transfusion therapy is the main treatment for thalassemic patient. It is given through a needle into the vein, which gives healthy red blood cells with normal haemoglobin. Since the lifespan of the red blood cell for thalassemic patient is less than 120 days, the patient requires a regular blood transfusion. A person with moderate thalassemia needs blood transfusion when the body is under stress such as during infections. However, people with major thalassemia, Cooley’s anemia, need more regular blood transfusion which is every 2 to 4 weeks. 9 Basically, there are reasons why thalassemic patients need regular red blood cells transfusion. First of all, it is to correct anemia and make sure that tissues get normal amount of oxygen and carry out body functions. It is proven by research done by the scientists and doctors, where a thalassemic patient who receives a regular transfusion could live up to 50years compared to the one not receiving any treatment whom could survive only until childhood. Regular transfusion also helps to keep the haemoglobin level near normal and the production of ineffective haemoglobin could be stopped. 7, 12 This treatment helps to improve child’s growth and well-being and usually prevents heart failure. 1

The blood transfusion in thalassemic patients is not the same as normal blood transfusion. As we know, blood contains different types of components and this component has its advantage that the components are in small volumes with higher concentrations of a particular product for specific indications. Therefore, thalassemic patients are given packed red blood cells instead of whole blood to improve the oxygen carrying capacity. 13

However, regular blood transfusion leads to another medical problem known as iron overload. Regular blood transfusion creates an excess of iron in the body because the haemoglobin in red blood cell is an iron rich protein. As there is no natural way of eliminating it from the body, it is stored primarily in spleen, liver, endocrine organs, and heart, and becomes toxic in tissues and organ. Iron overload can cause these organs to malfunction and damages it if untreated. 14, 15, 16 In order to remove the iron overload, a therapy known as iron chelation is done. A test called serum ferritin test is carried out to determine the amount of iron in the body as it is an important iron-storage protein. Injections of Desferrioxamine (Desferal) is given to the patient 5-7 nights per week with 8-12 hours supply which is done during sleeping time. It is injected under the skin from a small pump. Desferal works by carrying out excess iron through urine. 22 Thus, iron level in the body can be reduced.

So, in conclusion, the main treatment for thalassemic patients is regular blood transfusion and iron chelation therapy as blood transfusion makes the iron stored in the body overloaded. However, one sad thing about what thalassemic patients undergoes is that they started doing this since they are infant and continues this painful treatment for the rest of their lives and endure the side effects of both treatment.

## SOCIAL AND ECONOMIC IMPLICATIONS

People suffering from thalassemia seem to have some emotional and psychological problems that disturb their routine lives. As we know, the people suffering from thalassemia have certain physical symptoms such as enlarged spleen which makes the stomach looks big and the head of the patient would be slightly in oval shape as the diploetic fibres in skull extend from the internal lamina. This makes them feel awkward and shy to meet people and other people also might look at them in different way.

Based on the experience of Noor Hafiza Noor Hamdan, 33 a thalassemic patient interviewed for The Star newspaper, she said that : ‘ Before this, I was shy and solemn, and I did not like to smile…. When people smile at me, I would ask myself, why? Maybe it’s because I have a sickness’ this lines are quoted from what she said in the interview. This shows that thalassemic patients lack of confidence and ashamed of themselves being in that condition, so they interact less with the public. This makes them feel lonely and leads to depression, another psychological disorder. Some people outside even discriminate them.

Many thalassemic patients suffer in getting jobs besides being neglected by the parents and also the public. According to Pn. Noor Hafiza, she was not lucky with several other employers before working in a fast food outlet as she was not given the job opportunity after she showed her medical check-up which states that she has thalassemia. 17 Some patients even tend to commit suicide as they couldn’t cope with the pain and treatments that gives them more pain. Patients who couldn’t cope with the treatment which requires not only money but also almost half of their hours in a day makes them think that it’s better to die rather than suffer and being a burden to the family. Besides that, they feel embarrassed by their limited ability to do things by their own. 4, 19

On the other hand, economic implications also give a big impact to the patient’s life. Beta- thalassemia major patients require a regular blood transfusion of every 2 weeks, which cost them a lot of money. Not only transfusion, patients have to undergo many blood test and to be forgotten, the iron chelation therapy. Desferal is a controlled medicine that is very expensive medicine. This is one of the reasons why thalassemic patient couldn’t continue their treatment.

## 1997 1998 (as from February)

Thalassaemia association RM138. 90 RM153. 00

Goverment purchase (via Remedi) RM157. 20 RM157. 20

Pharmacy/clinic RM190. 00 RM209. 00

HUKM ‘ non-formulary purchase’ RM194. 00

Customer purchase RM211. 00 RM261. 00

This research done by Prof. Elizabeth George shows the Desferal’s price increase from year 1997 to 1998, by 2009, the price had increased in the range of from RM500 to over RM1000 which is definitely not affordable for low class and middle class families. Thus, it becomes a financial burden to the family. As the patient gets older, the blood requirement increases as well as the Desferal. This directly increases the cost of living for a thalassemic patient. 20 There is no other way for the patients besides taking Desferal as without it, they have an average lifespan of only 20years only. 5 WORD COUNT: 1463words

## BENEFITS AND RISK

Benefits of blood transfusion are that it helps the body to have new and healthy red blood cell with normal haemoglobin. As we knew, thalassemic patients suffer from abnormal red blood cells with mutated or deleted haemoglobin, hence their red blood cells couldn’t carry out its function and creates health problems such as the most severe ones is anemia. With this blood transfusion, a person could live longer to an average lifespan of 50years besides growing normally. Blood transfusion also helps the patient’s bones to form normally. As blood is given externally, the bone marrow does not have to work hard to produce red blood cell so it can concentrate on the formation of proper and normal bones. On the other hand, iron chelation helps to remove iron and prevents the organs like endocrine organs and liver from being damaged. 21

However, there are risks in both treatments. The most common risk of blood transfusion is infections and rejection. The patient has to get the same blood group in order to avoid rejection. There are chances of infection during transfusion either due to the blood itself or improper handling of the needle and syringe. Although a person receives regular Desferal, over time, the iron will be deposited in the spleen and liver and causes it to be damaged. Besides that, the patient has to cope with the side effects of both blood transfusion and Desferal which includes nausea, dizziness, and problems in hearing. 22

## ALTERNATIVE SOLUTION

SPLENECTOMY

Splenectomy is a surgical way of removing spleen. Old and damaged red blood cells are removed in the spleen. Since the red blood cells in a thalassemic patient are weakened, it is destroyed when passed through spleen. This makes the body to have fewer red blood cells. Removing spleen can help to lower the number of red blood cells that are lost, besides being removed due to the enlargement which causes pain to the patient. Splenectomy is done to patient above 11years old and after the removal, patient might need antibiotics to prevent infections as without spleen a person is more prone to infections. Patients can also get pneumococcal vaccine (Pneumovax) before the surgery to prevent infections. 2

BONE MARROW STEM CELLTRANSPLANT

Bone marrow transplant is another possible solution for thalassemia patient. The first successful transplant was performed in 1981 to a six-month baby from Italy. 23 Bone marrow transplant involves the replacement of healthy stem cells as the patient’s stem cells are not producing healthy blood cells. The transplant procedure is similar to blood transfusion where it is infused through central nervous catheter. The stem cells migrate to the bones from the blood stream to stimulate the production of new bone marrow that migrates to the large cavities of bones and produce new blood cells. 24 The most commonly used stem cell is the embryonic stem cell because it is easier to extract and have more advantage compared to adult stem cell as it is totipotent and not yet undergo differentiation and determination. 25 However, there are risks in getting stem cell transplant.

First of all, risk of getting a suitable stem cell that matches the body to prevent rejections. Besides that, risk of infection is another problem during the process. According to Dr. Revathy Raj, Consultant Paediatric Haemato Oncologist, Apollo Speciality Hospital, Chennai, as the immune system will be lowered by immunosuppressant drug to reduce the risk of rejection, the body is now more prone to other infections. 6, 26

## EVALUATION

http://www. penthal. org/thalassaemia. htm is the website from a non-profit organization that aimed to give information about thalassemia not only to the patients but also to those who wants to know about it. In my opinion, this website is trustable and reliable as the information is factual and valid. This organization is an active group and always updated with latest new. Besides that, they are reviewed by some doctors and also origins from Penang, which means the information is more specific to Malaysians. It is also trustable as the basic informations on thalassemia given are the same as other resources used in this report such as http://www. medindia. net/patients/patientinfo/Thalassemia. htm , http://www. thalassemiapatientsandfriends. com/index. php? topic= 1571. 0

Information are also obtained from a paper cut : The Star Online, “ My blood is Different” which is based on the personal experience of Noor Hafiza Noor Hamdan and some information from Ministry of Health’s Thalassemia website, www. mytalasemia. net. my. The experience shared by the patient gives a lot of reliable information on treatments and their risks. The Ministry of Health’s website is also very helpful in providing all kind of information by using videos and slides shows which makes it more clear and understandable. However, it gives only a brief and limited input regarding thalassemia as its main purpose is to provide brief information about this disease.

It is hard to find any specific book related to thalassemia as the disease is still considered as ‘ new’ and unpopular in Malaysia. Most of the book sources that I referred are books related to genetics and pathology as thalassemia is related to both. However, the book, Robbins Basic Pathology, 7th Edition by Vinay Kumar and others as stated in bibliography is the most reliable and useful among all. The diagram taken from the book is very useful in explaining the pathogenesis of Î²-thalassemia as it couldn’t be found in any website I explored. Besides that, the explanations given in the book are also simple, clear and understandable. For example this sentence that describes about Î² thalassemia:

Î’-thalassemia, associated with total absence of Î²-globin chain, and characterized by reduced synthesis of the chain in homozygous state. Genes obtained from thalassemic patients has revealed more than 100 different mutations responsible for it.