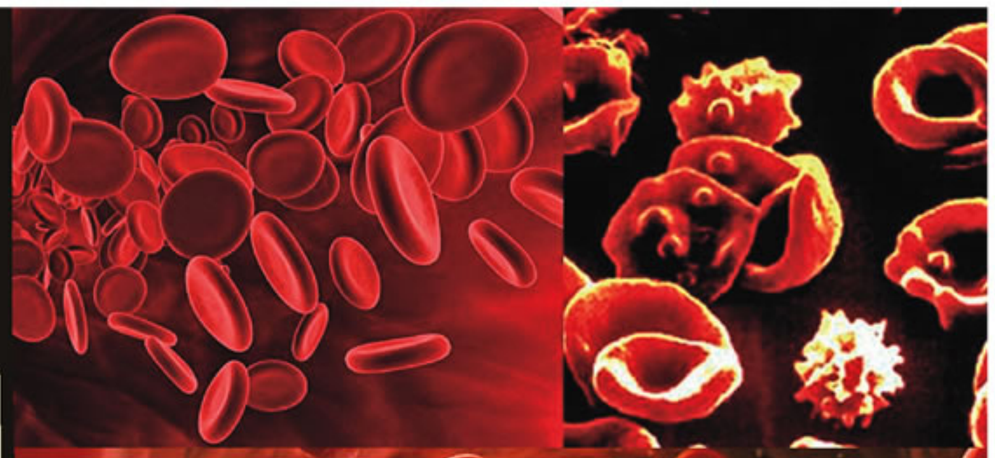
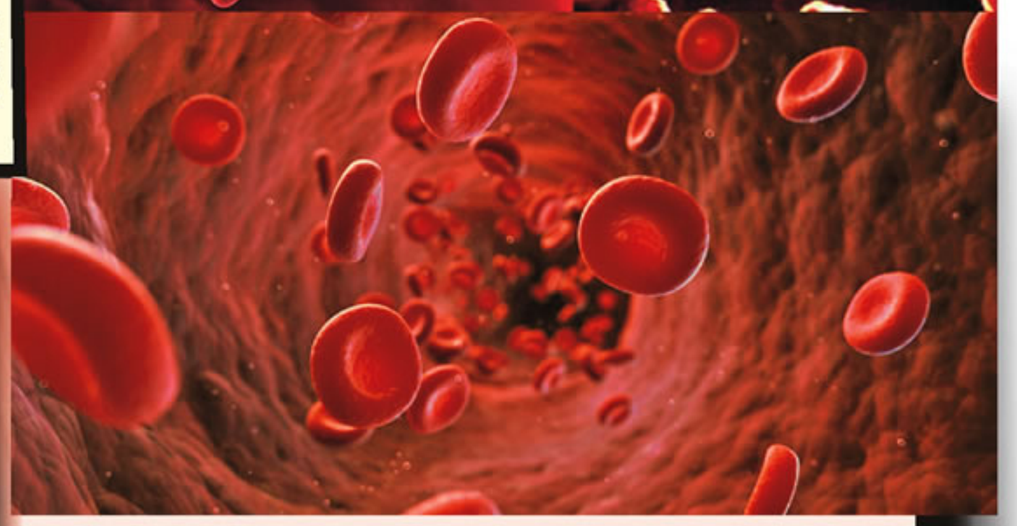
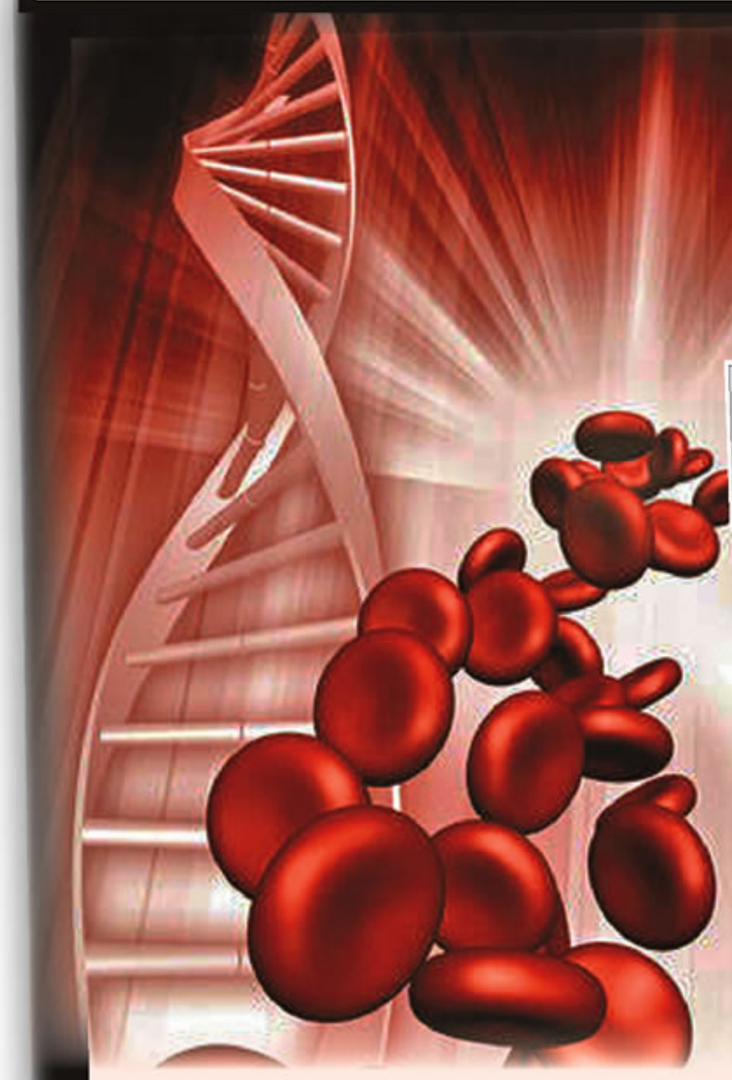
YOUR BLOOD LIFE





NADIRA GUNATILLEKE

Dr Sachith Mettananda, MBBS(Col) DCH(Col) MD(Paed)(Col) DPhil(Oxon) Lecturer and Consultant Paediatrician Faculty of Medicine and North Colombo Teaching Hospital, Ragama speaks about Thalassemia, one of the preventable, incurable and manageable (with life long blood transfusion and medical treatment) diseases which affect a human being for his/her life time while putting an unbearable financial and mental burden for the parents of the patient.



Health WATCH

Lecturer and Consultant Pediatrician Faculty of Medicine and Ragama North Colombo Teaching Hospital **Dr Sachith Mettananda.**

*** Thalassaemia is a preventable disease**

*** The best way to prevent thalassaemia is by stopping marriage/child births between two thalassaemia carrier adults**

*** If one partner is found to be carrying an abnormal thalassaemia gene (thalassemia carrier) he/she should be careful to select a partner who is not a thalassaemia carrier.**

*** If a thalassaemia carrier individual marries a non-carrier individual, there is no risk of having children with thalassaemia**

*** It is advisable that every couple get themselves tested for thalassaemia carrier state before starting courtships / marriage or at least before getting pregnant.**

*** Thalassaemia carrier state can be easily identified by simple blood tests which includes FBC and haemoglobin HPLC test. These tests can be done free of charge at any thalassaemia center in the country.**

Q: What is Thalassaemia?

A: Thalassaemia is a disease of blood which results in profound destructions of red blood cells. In humans, oxygen is carried around the body by the molecule known as haemoglobin which is found inside red blood cells. Hemoglobin is composed of two types of proteins which are known as alpha-globin and beta-globin.

In thalassaemia, production of beta-globin protein is reduced and this reduction leads to continuous premature destructions of red blood cells within the bone marrow (the site where all blood cells are produced) and within circulation.

Destruction of red blood cells compromises the body's ability to supply oxygen to its organs which leads to symptoms of thalassaemia.

Thalassaemia is genetically transmitted and the gene responsible is beta-globin gene. All humans have two beta-globin genes (each inherited from either parent) and individuals with thalassaemia have mutations (abnormalities) in both beta-globin genes.

Q: How about symptoms and diagnosis?

A Children with thalassaemia are perfectly normal at birth. These infants begin to show symptoms of thalassaemia around six months of age. As already mentioned, red blood cells of these babies are destroyed which leads to the clinical state known as 'anaemia' (i.e. not enough haemoglobin in the body).

Patients with thalassaemia develop symptoms of anaemia such as paleness (pallor) of the body, feeding difficulties, poor weight gain, difficulty in breathing and lethargy. In addition the liver and the spleen are enlarged and these kids will have changes in bones which include bossing of the forehead and prominence of cheek bones.

The diagnosis of thalassaemia is straight forward and can be done through few blood tests. Full blood count (FBC) test will show low haemoglobin level and the blood film (blood picture) will demonstrate smaller and less haemoglobinised red blood cells with some abnormal cells.

The definitive diagnosis is made through Haemoglobin HPLC which is freely available in thalassaemia centres (eg: Kurunegala, Ragama, Anuradhapura and Badulla) around the country.

Q: What are the treatments available in Sri Lanka at the moment?

A Blood transfusion is the only routinely available treatment for thalassaemia at present in Sri Lanka. Children with thalassaemia need regular monthly blood transfusions to keep the amount of haemoglobin at safe levels and to sustain life. In addition, patients with thalassaemia are at increased risk of iron overload and iron deposition in body organs therefore, requires treatment to remove excess iron from the body.

Currently three iron chelating medications are available, namely desferrioxamine, deferasirox and deferiprone. All these medications are given free of charge to all the patents with thalassaemia in Sri Lanka. Despite treatment patients with thalassaemia develop numerous complications therefore, invariably have a reduced life expectancy and a poor quality of life.

The only cure for thalassaemia is bone marrow transplantation and has been performed in a limited number of patients in private hospitals in Sri Lanka. However, even in developed countries, bone marrow transplantation is only available to a minority of patients who have suitable donors.

Q: How can we prevent Thalassemia ?

A Thalassaemia is a preventable disease. As already mentioned, thalassaemia occurs when a child inherits two mutated (abnormal) beta-globin genes from his/her parents. The parents of patients with thalassaemia are healthy and normal as only one (out of two) beta-globin genes are mutated (abnormal). Those individuals who has only one mutated (abnormal) beta-globin gene are known as thalassaemia carriers. A patient with thalassaemia could only be born to a couple who are both thalassaemia carriers. The best way to prevent thalassaemia is by stopping marriage between two thalassaemia carrier adults or not giving birth to children if two thalassaemia carriers decide to get married. Such couples should only adopt a child from another family if they badly need a baby.

It is estimated that 2.5% of Sri Lankan population (500,000 people) are carriers for thalassaemia.

Fortunately, thalassaemia carrier state can be easily identified by simple blood tests which includes FBC and haemoglobin HPLC test. These tests can be done free of charge at any thalassaemia centre in the country. It is advisable that every young adult get tested for thalassaemia carrier state before marriage. If an individual is free of abnormal thalassaemia gene there is no restrictions in finding the partner. If one is found to be carrying an abnormal thalassaemia gene (thalassemia carrier) he/she should be careful to select a partner who is not a thalassaemia carrier. If a thalassaemia carrier individual marries a non-carrier individual, there is no risk of having children with thalassaemia. In this way, by stopping marriages between two thalassaemia carriers, birth of children will thalassaemia can be prevented and the disease can be eliminated.