SUNDAY OBSERVER

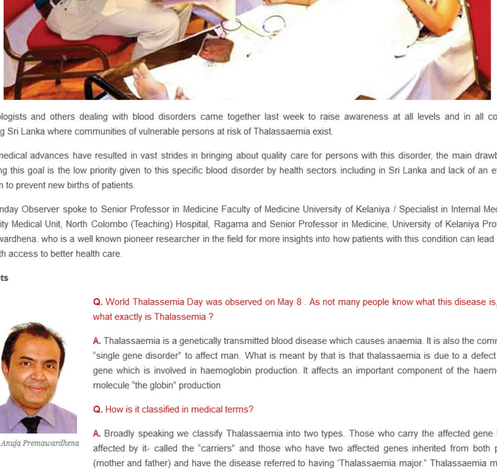
Sunday, 14 May 2023

Bridging care gaps vital to help Thalassemia patients lead normal lives - Pioneer researcher in Thalassemia, Prof Anuja Premawardhena

BY CAROL ALOYSIUS

Health

14 May, 2023

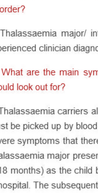


Hematologists and others dealing with blood disorders came together last week to raise awareness at all levels and in all countries including Sri Lanka where communities of vulnerable persons at risk of Thalassaemia exist.

While medical advances have resulted in vast strides in bringing about quality care for persons with this disorder, the main drawback to achieving this goal is the low priority given to this specific blood disorder by health sectors including in Sri Lanka and lack of an effective program to prevent new births of patients.

The Sunday Observer spoke to Senior Professor in Medicine Faculty of Medicine University of Kelaniya / Specialist in Internal Medicine , University Medical Unit, North Colombo (Teaching) Hospital, Ragama and Senior Professor in Medicine, University of Kelaniya Prof Anuja Premawardhena. who is a well known pioneer researcher in the field for more insights into how patients with this condition can lead normal lives with access to better health care.

Excerpts



Prof. Anuja Premawardhena

Q. World Thalassemia Day was observed on May 8 . As not many people know what this disease is, tell us what exactly is Thalassemia ?

A. Thalassaemia is a genetically transmitted blood disease which causes anaemia. It is also the commonest "single gene disorder" to affect man. What is meant by that is that thalassaemia is due to a defect of one gene which is involved in haemoglobin production. It affects an important component of the haemoglobin molecule "the globin" production

Q. How is it classified in medical terms?

A. Broadly speaking we classify Thalassaemia into two types. Those who carry the affected gene but not affected by it- called the "carriers" and those who have two affected genes inherited from both parents (mother and father) and have the disease referred to having "Thalassaemia major." Thalassaemia major is. very serious disease as it leads to very severe anaemia and presents in very early childhood.

If not treated by regular blood transfusions the child would die. That's the very opposite of the "carrier." They are completely normal! Unless diagnosed by a blood test, or in the extremely unfortunate scenario where a child is born into the family with Thalassaemia major they will not be picked up. In addition to these polar opposite conditions; somewhere in between lies a group of patients who may require an occasional blood transfusion due to Thalassaemia.

That group is referred to as Thalassaemia intermedia. In Sri Lanka the commonest type of Thalassaemia patients who seek treatment have "beta thalassaemia major. There is also an important sub group of thalassaemia patients who have a disease called Hameoglbin E beta thalassaemia. They account for about a third of all patients who attend thalassaemia centres for treatment.

Q. Do you see an increase in the number of Thalassaemia cases in recent years?

A. There is no increase in the number of patients diagnosed in Sri Lanka. The annual number of new patients diagnosed in the island remains around 40-60. What concerns us is that we don't have an effective way to prevent the new births.

Q. What is the main mode of Thalassemia transmission?

A. It is transmitted genetically in what is referred to as autosomal recessive manner. The carrier male and carrier female (who are clinically silent ie: normal) produces a child with severe disease

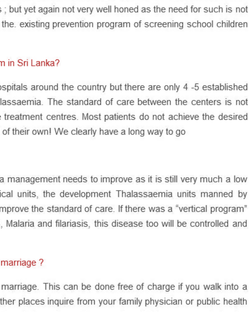
Q. How widely prevalent is it in Sri Lanka and where is it most widely spread?

A. The distribution of Thalassaemia in the country is linked to the historical prevalence of malaria in the country. In Sri Lanka the highest prevalence is found in the districts of Anuradhapura, Kurunegala, Badulla, Monaragala, Ampara. No region is spared though.

Q. Can it be mistaken for any other blood disorder? What are the distinguishing characteristics between Thalassemia and any other blood disorder?

A. Thalassaemia major/ intermedia causes anaemia. It can be mistaken for any number of diseases which cause anaemia. For an experienced clinician diagnosing thalassaemia is not difficult.

Q. What are the main symptoms of this condition that a high risk person should look out for?



A. Thalassaemia carriers also referred to as traits have no symptoms. They must be picked up by blood tests. Those with the severe disease have such severe symptoms that there is no real need to educate anyone about them! Thalassaemia major presents in early childhood (usually between 3 months to 18 months) as the child becomes so ill that the mother invariably brings it to hospital. The subsequent diagnosis is easy for the clinicians.

The real problem is to pick those of us who have thalassaemia carrier state. In Sri Lanka the overall prevalence of the beta thalassaemia carrier state is around 3 percent. It reaches almost 10 percent in some areas referred to earlier. This means over half a million of us are carriers for the disease. The risk for them lies in marriage. Unknowingly marrying another carrier might result in an absolute tragedy. The birth of a Thalassaemia major child.

Q. What will happen to those who do not take any treatment or fail to come on the day they should?

A. Early death. Thalassaemia major is a fatal disease; unless treated well. So access to good , standard care is essential to prolong survival. If blood transfusions and chelation is done optimally most patients do very well. They live long. Life expectancy and quality of life depends on access to quality care.

Q. Since parents are the main carriers of this disease, what are the chances of their children getting the disease if 1) both parents 2. Only one If only one of the parents is a carrier of Thalassaemia?

A. If both parents are carriers there is a 25 percent chance of any child being born with the severe disease. This is a very high risk. If one is a carrier and the other is not; there is no (zero) risk of a patient being born. So if you are a carrier for Thalassaemia: the message is simple: make sure that your partner is not a carrier for Thalassaemia.

Q. In some countries most patients are screened for Thalassemia if they are in the high risk group before marriage and advised accordingly by the medical practitioner. Is there a similar scheme in Sri Lanka?

A. A high risk group would be family members including extended family members of an identified Thalassaemia major child or even a newly diagnosed "carrier." Every time a diagnosis is made the people involved inform the family members to be present for screening. This needs to happen voluntarily. Unlike for disorders like TB where a public health inspector (PHI) is involved in "contact" tracing there is no active case tracing. What we realize is that voluntary presentation for screening even within the immediate family members is not very high.

Q. The International Thalassaemia Day 2023 theme is "Be Aware. Share. Care: Strengthening Education to Bridge the Thalassaemia Care Gap. What is its significance to Lankans afflicted by this condition in particular?

A. Education of the public is crucial to win the battle against Thalassaemia. It should be fought on two fronts. The awareness of the condition called "Thalassaemia" and that all young people should get themselves checked for their "Thalassaemia status" prior to marriage must be inculcated into our society. Being a " thalassaemia carrier" has and should not have any stigma attached to it. It would allow you to make a sensible choice in a partner in marriage. The "second front" is the lack of awareness among the health care workers. Advances in understanding of the way to treat thalassaemia major patients has allowed those affected by the disease to live almost near normal lives. This knowledge unfortunately has not filtered down to some medical practitioners; which is a major obstacle to treating patients well.

Q. How well prepared is our Health Ministry to face this challenge considering our limited resources?

A. Resources are always going to be a restricting force. To treat a Thalassaemia patient for one year the Ministry of Health spends 2500 US$ per year (calculated in 2015). To this number every year 40-60 more patients will be added.. A recent policy brief exercise conducted in Sri Lanka found that a much-improved version of the prevention programme for Thalassaemia than that which exists now would still save more money for the state rather than treating patients. (i.e.: will be more cost effective). All this would need careful planning, prioritisation and resource allocation. Thalassaemia is still not up in the pecking order!

Q. So do you have any suggestions on the best cost effective solution?

A. The most cost effective strategies for prevention of thalassaemia; without a doubt is what is referred to as the "Cyprus Model. "This is where Thalassaemia screening happens early in pregnancy and for those couples where both parents are carriers; prenatal diagnosis is offered. If the foetus is affected the choice of termination is discussed. In Sri Lanka with the current legal system this option would not be feasible. The techniques for prenatal diagnosis is established in a few centres ; but yet again not very well honed as the need for such is not there. Hence, in the current context we have recently suggested intensifying the. existing prevention program of screening school children with better central control.

Q. What are the gaps you see in the present Thalassemia patient care system in Sri Lanka?

A. There are massive gaps. Patients with the disease are treated over 27 hospitals around the country but there are only 4 -5 established Thalassaemia centres. There is only one center for adult patients with Thalassaemia. The standard of care between the centers is not uniform. The availability of essential treatment is not uniform between these treatment centres. Most patients do not achieve the desired targets of treatment any time in their lives and not necessarily due to any fault of their own! We clearly have a long way to go

Q. How would you like to fill them?

A. Amongst the health care professionals the awareness about Thalassaemia management needs to improve as it is still very much a low priority disease. Rather than managing patients in general paediatric/medical units, the development Thalassaemia units manned by specially trained staff; with an interest and dedication to the "subject" would improve the standard of care. If there was a "vertical program" controlled by a separate directorate in the Ministry of Health as it has for TB, Malaria and filariasis, this disease too will be controlled and contained very well.

Q. Have you a message for the public, especially to those on the threshold of marriage ?

A. All young people should be aware of their Thalassaemia "status" prior to marriage. This can be done free of charge if you walk into a Thalassaemia centre in Kurunegala, Ragama, Badulla or Anuradhapura. In other places inquire from your family physician or public health inspector or any health care provider about how you can get yourself tested and thereafter make use of available resources to lead a normal quality life.