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Genetic disorder Thalassemia: Causes and Prevention

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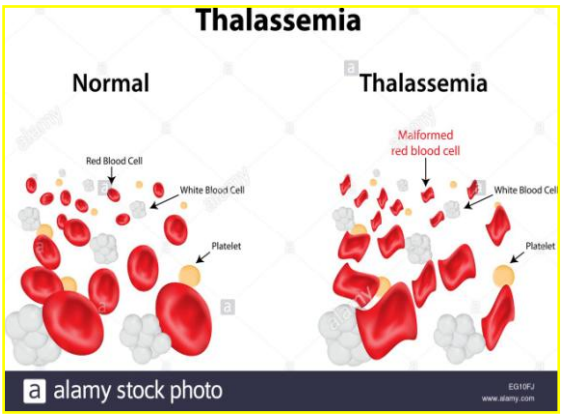
ABSTRACT

This paper presents the depiction of the millions of Thalassemia patients of the society who inherit this genetic disease as a result of ignorance spread by their parents and in the society. Thalassemia is a genetic disorder therefore 10% of the world's Total Thalassemic patients are found in India. The concern is that one out of every 30 children is born with Thalassemia trait/carrier of mutated gene.With specific application to a group of people those living relatively long term with a genetic disorder whichever passed on parents to their off-springs. Approximately 20% of the world population carries alpha and beta Thalassemia. 56000 people of the world are suffering from Thalassemia major, out of which 30000 need blood transfusions on regular basis. 3000 children in the world die every year due to uncontrolled iron overload of the age at 10 to 20. The child with the affected gene can be prevented from being born with chronic vellus sampling during 11-13 month of pregnancy.

1. Introduction

Humans are an integral part of the society as his action and interaction determined the structure of the society.Any society can function well only if, when parts contribute in its development. If an important part of the younger generation of a society cannot contribute to the development of the society due to its inadequacy, then how society can sustain the goal of development? The eminent psychologist rightly said that man's behavior is influenced by two forces as heredity and environment.The biological characteristics are transmitted to children by their parents. Heredity traits are intrinsic when the association of two parent cells, that come together when a male sperm fertilizes a female egg. So, the chromosomes and genes of parents determined the characteristic of their children.Thalassemia is a genetic blood disorder which is passed on parents to children. Due to lack of awareness in Indian continental 10% of total world Thalassemia population is suffering with this chronic disease. In the absence of informationparents could not know about their Thalassemia status and after the birth of Thalassemia major child, they come to know about this diseases. Gynecologists furthermoredon'task to diagnose thedisease, due to which children are being born with thalassemic genes. If gynecologist writes to scrutinize HB Electrophoresis or HBA2 test during pregnancy period in between 11- 13 weeks of pregnancy, then the disease can be crushed.



Source: https://ghr.nlm.nih.gov/condition/beta-thalassemia

Since 1925 when American pediatrician Thomas Cooley first described about Thalassemia, our level of information has increased so much that we can crushthe disease nip in the bud. Due to this fatal disease, 1 child in every 45 minutes and 10000 children in one consecutive year are being born from affected gene of Thalassemia.Normal hemoglobin called HB A consists of four protein chain –two alphaglobin and two betaglobin. The two major types of Thalassemia Alpha and Beta are named after defects in these protein chains. Two genes (one from each parent) are needed to make enough Beta protein. Beta Thalassemia occurs if one or both genes are mutated.

Types of Beta Thalassemia: - It can be bifurcated into three types...

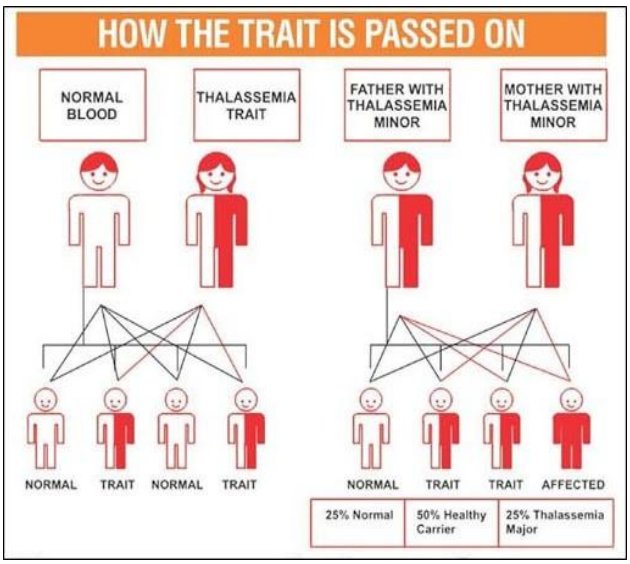
Minor Thalassemia, (heterozygous)-Thalassemia minor has only one copy of the affected gene associated with one perfectly normal beta chain gene. However person with Thalassemia minor have a normal blood iron level unless they are iron deficient for other reasons.

Intermedia Thalassemia (Non Transfusion dependent thalassemic) - in this stage symptoms are found less and requires less blood or after five to six years.

Genotype	Hb A	Hb A2	Hb F
Normal	Normal	Normal	Normal
Silent Carrier	Normal	Normal	Normal
β Thalassemia Minor	Dec	Normal to Increased	Normal to Increased
β Thalassemia Intermedia	Dec	Normal to Increased	Increased
β Thalassemia Major	Dec	Usually Increased	Increased

Source: http://www.medical-labs.net/beta-thalassemia-types-with-hemoglobin-comparison-1332/

Major Thalassemia (Homozygous Cooley's anemia) -the most dangerous condition in which the symptoms of disease begin to appear a few months after birth. The child born with Thalassemia major has two affected genes and no normal beta- chain gene. The size of RBC in affected person usually have smaller and survival rate of Red blood cells is 15 to 20 days comparison to normal man which RBCsurvive 120 days .Thereby, the patient's body is reduced to blood and he has to resorted blood transfusion after every three months. By which the iron element gets accumulated (Iron Overload) in individual body and many parts of the body can be affected due to overload such as liver, kidney and heart and condition of reversal damage usually known as cirrhosis occurred in which DNA of the liver cell damaged and increased the risk of liver cancer. The patient has to resort iron chelation therapy which is very painful and expensive too.



Source: https://thalassaemia.org.cy/education/learn-about-thalassaemia/haemoglobin-disorders/beta-thalassaemia/

Who carries Thalassemia:-Now days this chronic disease has taken the whole world to its grip. Beta Thalassemia is dominantly prevalent in Mediterraneanregion such as Greece, Italy, Spain and others, but it has been overcome through Africa, North and South America and in Asian continent. 3- 8% population of Asia is found in the grip of this chronic disease such as India, Nepal, Pakistan, Bangladesh, china, Malaysia and Thailand.

Sign,Symptoms and complication of Thalassemia:-Thalassemia major is the severe stage in which sign and symptoms such as severe anemia and other health problems usually occur within the first year of birth.

- A pale appearance
- Poor appetite
- Jaundice
- Dark urine which sign that red blood cells are breaking down
- The enlargement of spleen liver and heart
- Bone problem especially with bone in the face

Complication- Good medical care as well as family support can make the life of thalassemic qualitative and prolong. That's why family and thalassemic patients must be prepared to face the following problems that may occur with time. Blood transfusions increase the amount of iron element in the body and due to the size of individual liver, heart and spleen increases which lead heart failure, arrhythmias (disease related to heart beat), heart attacks, cirrhosis in liver.Spleen acts as a filter for blood as part of the immune system. Old blood cells are recycled in the spleen, platelets and white blood cells are stored there. It helps to fight certain kinds of bacteria that cause pneumonia and meningitis. Due toiron overload in the spleen,it enlarged in size. Patients who have had their spleen removed are at even higher risk of infection as they no longer have infection fighting organ.Thalassemic patients have to fight againstosteoporosis in which bones are weak and brittle usually breaks easily.

Screening and prevention:-Thalassemia is a genetic disorder that is transferred from the parents to their children. So by taking into consideration this chronic disease, every individual should be get their Thalassemia status checked (HB Electrophoresis or HBA2 test) before planning marriage. If married, then both husband and wife should get genetic counseling from genetic department before family planning. If both husband and wife are Thalassemia minor/ carrier by status and knew the fact after the baby is conceived, within 11 to 13 weeks of pregnancy, chronic vellus sampling (CVS) can be detected that the being born is not affected by Thalassemia gene. Awareness campaigns are being organized in India on large scale both by Government and NGO in favor of voluntary premarital screening to detect carriers of thalassemic and marriage between both carriers are strongly discouraged.

The treatment of Thalassemia major-treatment for Thalassemia relied on the type and severity of the disorder. People who are carriers of beta Thalassemia trait have mild or no symptoms, need not any treatment, but those who have moderate (Intermedia) and the patients of major and moderate Thalassemia have to take regular blood transfusions, iron chelation therapy and folic acid supplements. Regular blood transfusions for Thalassemia major (often every 2 to 4 weeks) helps them to maintain normal hemoglobin and red blood cell levels, whichever lead affected people to enjoy normal activities and feel better. Regular blood transfusion can lead to a build of iron in the blood. This is known as iron overload and this condition can damages the liver, heart and other parts of the body. Hence iron chelation therapy comprising deferasirox and kelfer have to be taken according to fix dose determined by the physician.Folic acid and calcium tablets have to take once daily for the production of red bloods cells and strengthen to avert osteoporosis respectively. This is life saving treatment but carries a risk of transmitting infections and viruses (HCV and HIV).

Bone marrow transplantation is the only treatment by which this disease can be cured. Bone marrow is the soft, fatty tissue inside your bones which produces blood cells. In bone marrow translations damaged or affected bone marrow replaced with healthy bone marrow stem cell. It is a procedure that in which a graft is implanted that takes one year plant to tree. For bone marrow transplantation, first of all DNA of thalassemic is matched with his siblings if dissimilarity found in DNA than parents are given priority for HLA typing. IfDNA does

not match with both sibs and parents, then external DNA registry are accessed and transplantation is done when DNA matches usually called unrelated donor transplantation. If there is a regression after bone marrow transplant, Thalassemia comes back and will have to do transplantation Immediately with the same donor of other ones which matches HLA typing. Critical situation occurred when there is rejection in bone marrow transplantation, bone marrow suddenly becomes empty so that neither the person himself nor the donor's cell comes and it threatens life and has to transplant immediately. Bone marrow donor is not harmed and he gets recover in 5 to 10 days.

Financial aids- In most hospitals, blood and medicines are given free by the government. On average which the annual expenditure from 50000 to 200000 Indian rupees. Indian railways and most of the state governments provide free travel pass for thalassemic patients and one attendant.The amount spent in the treatment of a thalassemic person is outside the purview of income tax. CBC, HCV, HIV, KFT, LFT and Serum ferritin blood tests are done by the medical staff on regular basis. Assistance of five lakh andthree lakh is provided by Delhi chief minister fund and prime minister of India respectively.

Living with-Survival and quality of life has been improved for people who have Intermedia and major Thalassemia. Since maximum people are getting blood transfusion, due to blood screening infection rate has been reduced respectively, iron chelation therapy is available that are easier for some people to take and last but not the least thalassemic are being cured through bone marrow transplantation.

Problems faced in society: - If the first baby born with thalassemic major due to lack of awareness then the allegation of deficiency is mostly framed on wife. The facility of blood transfusions is available only in post graduate institutes, for which the patient and his / her guardians' have to leave the house early in the morning for the hospital or treatment institute and the whole process takes 10 to 15 hours. Due to this, children's higher education is also affected because before choosing college or university they have to investigate that would they get all the essential fundamental medical facilities for the treatment. A person suffering from Thalassemia disease is always worried that if the employer finds out the fact that they are Thalassemic then the employer will either expel them or not offer work. The central govt. of India has given relief to the thalassemic people and have been included them in disabled category by passing the law "The Right of Person with Disabilities Bill- 2016".It is a good initiative from the central government, but thalassemic have pave a long way towards improvements in the treatment facilities.

Centers of Treatments: - Information about Thalassemia treatment centers and hospitals in India is given below.

- All India institute of medical science (AIIMS New Delhi)
- Christian medical college and hospital (CMC Vellore)
- Thalassemia ad sickle cell society in Hyderabad
- Thalassemia children welfare association at post graduate institute of medical education and research(PGIMER)
- Sanjay Gandhi post graduate institute of medical sciences in Lucknow

2. Conclusion

On the basis of above discussion, it may be concluded that Thalassemia is a severe illness related to human blood. It is mostly based on heredity. Major symptoms of it are, pale appearance, poor appetite, jaundice, dark urine which sign that red blood cells are breaking down, the enlargement of spleen liver and heart and bone problem especially with bone in the face etc. Its care and diagnosis of Thalassemia may depends upon its stage and intensity. It needs regular transfusion of blood under medical supervision. India being a developing nation, has not so affordable and advanced health care facilities. But some NGOs are contributing a lot towards. The present govt. is sustaining its vital efforts towards advancement of health care facilities but a lot remains to be done.

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