

Thalassemia: Genetically Transmitted Blood Disorder

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Alpha Thalassemia is most common Inherited Blood Disorder in the Asian/Pacific Islander population. Beta Thalassemia is common in the Mediterranean population. Autosomal recessive in Inheritance pattern.

Thalassemia is a blood disorder passed down through families (inherited) in which the body makes an abnormal form of hemoglobin. The deficient synthesis of one or more of the normal blood globin chains. Blood disorder ranging from minor to severe. The disorder results in excessive destruction of red blood cells leading to Anemia. Alpha Thalassemia occurs when a mutation in the gene that codes for alpha globin results in reduced or absent production of alpha globin. Beta Thalassemia occurs with a corresponding change in beta globin genes. Therefore Thalassemias are a result of quantitative mutation in globin genes. The instructions for alpha globin production are present in duplicate, two genes on each chromosome 16 for a total of 4. The instructions for beta globin production are on chromosomes 11, one gene on each chromosome for a total of two.

Genetic Counseling and screening is done to identify individuals at risk for a specific disorder. In Genetic Consultation the patient and the genetic specialist or counselor review family histories to determine whether there are any diseases which may recur in other relatives. Also explains fully the risks for diseases and conditions which have occurred in the family and all test and procedures that should be performed.

Genetic Counseling is the communication process of providing information and support to individuals and families with a diagnosis and or risk of occurrence of an inherited disorder. It is needed at diagnosis and during adolescence, prior to and after any genetic testing prior to pregnancy and/or easily in pregnancy as possible. Annual follow-ups and needed to reinforce teaching.

If both parents carry a hemoglobinopathy trait, the risk is 25% for each pregnancy for an affected child.

Concern is increasing that Thalassemia may become a very serious problem in the next 50 years, one that will burden the World Blood Bank supplies and the Health Systems in general.

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