[TO BE INTRODUCED IN THE NATIONAL ASSEMBLY]

A

BILL

to provide compulsory blood test for relatives of Thalassaemia patients.

WHEREAS it is expedient to take concrete steps for controlling the hazardous disease of Thalassaemia;

It is hereby enacted as follows:

1. **Short title and commencement**.- (1) This Act may be called the Compulsory Blood Test of the Relatives of Thalassaemia Patient Act, 2014.

(2) It shall extent to the whole of ICT.

(3) It shall come into force at once.

2. **Definitions**.- In this Act, unless context requires otherwise,-

(a) "Thalassaemia" means a disease in which a child or an adult becomes anemic because of a genetic defect of hemoglobin;

(b) "Thalassaemia Major" means a stage of Thalassaemia which clinically manifests itself as severe anemia requiring treatment with repeated blood transfusion and medicines;

(c) "Thalassaemia Minor/Trait" means Thalassaemia which results in mild anemia and is often misdiagnosed as an iron deficiency anemia if appropriate blood tests are not carried out;

(d) "Genetic Defect" means abnormalities of genes;

(e) "Hemoglobin" is the protein in red blood cells which is responsible for carrying oxygen;

(f) Prenatal diagnosis test carried out during pregnancy;

(g) "Blood Relatives" means directly related aunts and uncles including sisters and brothers of mother and father of the patient, the children of these uncles and aunts and the siblings of the patient;

(h) "Chromosomes" means double helix structures present in the nucleus of the cells, these carry the genes;

(i) "Electrophoresis" means a test used to identify different types of hemoglobin;

(j) "Hemolytic Anemia" An anemia caused as a result of shorter life span of red blood cells;

3. **Description of Thalassaemia.-** Thalassaemia is a hereditary genetic disorder causing impaired production of hemoglobin. The two types of Thalassaemia are:

(1) Thalassaemia Major.
(2) Thalassemia Minor/Trait.

(i) **Thalassaemia Major.-** Thalassaemia which usually becomes symptomatic as a severe, progressive hemolytic anemia during the 2nd and 6th month of life. Normally red blood cells spend 100-120 days in circulation and about 1% are removed from the blood each day and are replaced. In Thalassaemia the rate of red blood cell destruction increase. As a response there is an increase in bone marrow activity in order to replace the destroyed red blood cells.

(ii) However production of red blood cells cannot keep pace with their rate of destruction so that repeat blood transfusions are required. Growth is impaired in these children, puberty rarely occurs, cardiac complication occur and are the cause of death. In transfusion dependent Thalassaemia death usually occurs during the 2nd decade only a few patients survive to their 30's.

(iii) Thalassaemia Minor is Thalassaemia associated with mild anemia. They do not have the symptoms. So that the condition is not diagnosed and the individuals are misdiagnosed as having iron deficiency anemia.

4. **Treatment.-** Patients with Thalassaemia Minor usually do not require treatment. Thalassaemia Major patients however require treatment which includes chronic repeated blood transfusions to combat anemia caused by increase red cell destruction, medication to reduce the burden of iron accumulated in the patient as result of repeated transfusion, operation to remove spleen which is the site where red blood cells destruction occurs.

The prevalence in Pakistan is 3-8%. Pakistan is seeing a large increase in Thalassaemic patients due to lack of screening/genetic counseling. Foci of prevalent groups are present in Pakistan where the disease runs in families. Thalassaemia Major (the clinically more serious disease) is more prevalent where there are higher rates of inter marriages within relatives. There is growing concern that Thalassaemia may become a very serious problem in the next 50 years, one that will burden the blood bank supplies and the health system in general. Each year 5000 children are born with transfusion dependent Thalassaemia Major. A recent study in Thalassaemia (Ahmad et al, 2002) strongly suggests that due to consanguineous marriages Thalassaemia genes are trapped within the family. It was discovered that families with a history of Thalassaemia have a high carrier rate (30%). In the study Thalassaemia was almost nonexistent in families that did not have any known history of Thalassaemia in Pakistan. Thalassaemia in fact is confined to 5-10% of families in Pakistan while 90% are free from Thalassaemia. As Thalassamia in fact is confined to 5% of families and since most of these families are already within the loop of blood transfusion centers either private or public; the

best way therefore to legislate for Thalassaemia. To screen families of indexed patients registered with them. These centers should also provide facilities such as diagnosis during pregnancy for the prevention of Thalassaemia. Screening to identify carriers, genetic counseling and diagnosis during pragnancy of beta Thalassaemia, this can greatly reduce the rate of birth of affected infants and save a lot of families from the anguish and trauma of dealing with a child with the disease.

5. **Transmission of Genetic Defect**: Transmission follows the following pattern:

The child of two (Thalassaemia Minor) carrier parents have 25% (that is 1 chance in 2) of inheriting the abnormal gene from each parent and of being (Thalassaemia Major) which is a more serious type of disease and is fatal.

Whereas it is necessary to make a law to formulate a policy for testing of blood relatives of those children who have Thalassaemia Major and manifest the disease.

6. **The Law:**

Who is to be tested:

(1) All clinics, hospitals and centers handling and treating Thalassaemia patients will ensure that blood relatives of these children are all screened for Thalassaemia Minor/Trait. This test is to be made compulsory for siblings, first cousins and uncles and aunts who are blood relatives.

In this regard it is particularly compulsory for those blood relatives of Thalassamia patients who are getting married to get a pre-marital blood screening to ensure that they are not carrying the Trait.

(2) Ante natal tests are to be carried out on pregnant women who are known carriers and whose partner is also a carrier for the Trait.

(3) All NGOs run centers dealing with Thalassaemia will ensure that they spend at least 10% of their budget on developing facilities for prenatal diagnosis of Thalassamia.

(4) All these clinics, hospitals and centers should counsel relatives of patients on risks of consanguineous marriages and on their chances of having Thalassaemic children.

(5) For pre-marital testing both partners are to have their blood indices done if both the partners have blood reports showing anemia their hemoglobin electrophoresis should be undertaken to ensure that they are not carrying the trait.

(6) Antenatal testing is to be carried out in all pregnant women who are known carriers and whose partners are also known carriers. Diagnosis of the disease is to be carried out and Chinese should have the facilities the facility to carry out tests and and procedure for diagnosis during pregnancy.

7. **Reporting of test result**

The test results are to be reported to those who are tested and if they are carriers (have Thalassaemia Minor/Trait) they are to be given counseling regarding their marrying someone with the same trait and the risk of passing on the disease to their offspring. The test results are to be entered into a data bank for registration of carriers of the Trait.

Antenatal test results are to be reported to the women tested and her partner and if the test is positive the parents are to be advised about the condition of the fetus and offered an option of terminating the pregnancy.

8. **Penalty.-**

(1) In case an NGO clinic, hospital fails to carry out the necessary screening the clinic, hospital will be in negligence of its duty and will be charged a penalty of Rs.100000.

(2) In case the person to be tested does not cooperate and refuses to be tested he can be fined and if charges are proved a penalty of Rs. 100000 is to be imposed. However no charge will be brought if parents refuse an antenatal test provided the hospital or clinic has explained the procedure and the risks associated with and the chance of the parents having affected baby.

(3) It shall be compulsory for clinics, centers and hospitals to provide detailed genetic counseling with information on pattern of disease and trait transmission if clinics do not provide written and oral counseling they will have been deemed to have been negligent of their duty and they will be penalized.

STATEMENT OF OBJECTS AND REASONS

Thalassaemia is a dangerous disease which kills a large number of children in Pakistan every year. It is very much needed to take appropriate steps to stop further spread of this disease and arrange for proper care of those patients who have fallen victim of this disease.

2. The Bill seeks to achieve the aforesaid objectives.

Sd/-
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