(AS PASSED BY 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 and to make a law for testing of blood relatives of those persons who have thalassaemia and manifest the disease;

It is hereby enacted as follows:-

Short title and commencement.- (1) This Act may be called the Compulsory Blood Test of the Relatives of Thalassaemia Patient Act, 2017.

(2) It extends to the whole of Islamabad Capital Territory.

(3) It shall come into force at once.

2. **Definitions.-** In this Act, unless the 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 or 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" means 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; and

(j) "hemolytic anemia'' means an anemia caused as a result of shorter life span of red blood cells.

3. **Test of thalassaemia mandatory.-** (1) All clinics, hospitals and centers notified by the Government for handling and treating thalassaemia patients shall ensure that blood relatives of children having thalassaemia are all screened for thalassaemia minor or trait or referred to centers having requisite facility for screening. 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 thalassaemia patients who are getting married to get a pre-marital blood screening to ensure that they are not carrying the trait. It will be ensured by the designated Hospital to provide counseling to those blood relatives of the Thalassaemia patients who are getting married and it will not be a public document.

(2) Antenatal 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 non-governmental organizations (NGOs) running centers dealing with thalassaemia shall ensure that they spend at least 10% of their budget on developing facilities for prenatal diagnosis of thalassaemia.

(4) All 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 electrophoreses should be undertaken to ensure that they are not carrying the trait.

(6) Diagnosis of the disease is to be carried out through such facilities so as to carry out tests and procedure for diagnosis during pregnancy.

(7) In case a non-governmental organization, clinic or hospital fails to carry out the necessary action they shall face administrative action from the concerned Ministry. An amount of rupees fifty thousand shall be imposed on the non-governmental organization, clinic or hospital who fails to carry out the test.

4. **Reporting of test result.-** (1) The test results are to be reported to those who are tested and if they are carriers (have thalassaemia minor or 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.

(2) Antenatal test result 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.

STATEMENT OF OBJECTS AND REASONS

Thalassaemia is a hereditary genetic disorder causing impaired production of hemoglobin. The two types of thalassaemia are-

a) Thalassaemia major 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 increases. As a response there is an increase in bone marrow activity in order to replace the destroyed red blood cells. However production of red blood cells cannot keep pace with their rate of destruction so that repeated blood transfusion is required. Growth is impaired in these children, puberty rarely occurs, cardiac

complication occurs 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; and

b) 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.

2. Patients with thalassaemia minor usually do not require treatment. Thalassaemia major patients however require treatment which includes chronic repeated blood transfusion 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.

3. The prevalence in Pakistan is 3-8%. Pakistan is seeing a large increase in thalassaemic patients due to lack of screening and 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 (Ahmed 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 thalassaemia 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 is 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 pregnancy 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 a disease.

4. 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.

5. 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 step further spread of this disease and arrange for proper care of those patients who have fallen victim of this disease.

6. The Bill seeks to achieve the aforesaid objectives.

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