PREMARITAL HEMOGLOBINOPATHY SCANNING
INFORMED CONSENT (2)

Mediterranean anaemia or, in medical terms, Thalassemia is a hereditary blood disease which is encountered in the countries surrounding the Mediterranean and various other parts of the world where the disease spread due to migrations. The disease is characterized with the faulty synthesis of haemoglobins, a component of erythrocytes in blood, and, therefore, the inefficient production erythrocytes. According to the World Health Organization (WHO) data, there are around 266 million haemoglobinopathy carriers around the world. In Turkey, hereditary blood diseases are significant public health issues with, in particular, thalassemia and sickle-cell anaemia. Every year, hundreds of children are born with the disease which causes material and non-material damage to families and the society. The reason why the disease is frequently observed in Turkey is that Anatolia was home to various races and cultures throughout centuries and that kin marriage was common. (25.1%) Kin marriages are mostly seen (70%) to take place between first-degree relatives. This, as a result, increases the prevalence of hereditary diseases in the society.

Various types of thalassemia are transmitted from parents to children through genes. Depending on the affected genes in parents, the types of thalassemia encountered in children vary as follows:

1-Thalassemia Minor (Carrier Type): This is a thalassemia type which can be identified with simple blood tests but does not show any significant symptom and accompany a normal length and quality of life. The individuals with this disease are quite healthy people and do not encounter any problem except for mild anaemia.

2-Thalassemia Major (Patient Type) : This is a severe type of the disease. It is encountered in children conceived of marriages where both mother and father are carriers of the disease. Clinical findings generally appear between the 6th month and age 2. The disease may cause pale skin, weakness, lack of appetite, poor feeding, disturbance, abdominal distension due to hepato- and splenomegaly, changes in bones starting with the facial bones and cranium, and a typical facial appearance. The disease may develop heart failure due to severe anaemia which may be seen suddenly in early periods. Frequent and regular blood transfusion is required to prevent heart failure. Otherwise, patient may pass away within a couple of years.

3-Thalassemia Intermedia (Light Disease Type) : This is a type which does not grant a completely healthy life like the carrier type but generally starts at a further age and allows normal growth and development without regular blood transfusion. Although the disease shows clinical onset at a later age (after 4), the cases with the disease may indicate hepato- and splenomegaly as well as bone changes.

It should be noted that there may be silent carriers even though Haemogram and HPLC (blood tests) indicate normal results.

Protective treatment plays a significant role in the fight against the disease. However, it is difficult to treat these patients to the best. Today, there are not so many patients who have the opportunity to undergo bone marrow transplantation which is the definitive treatment for thalassemia. This is a very expensive and a difficult process. Therefore, the treatment methods applied in many countries are currently blood transfusion and iron chelation (the latter is a process applied to reduce iron levels in blood). Nevertheless, thalassemia is **preventable** once parents are found to carry the disease. After receiving prenatal diagnosis, two individuals who carry thalassemia can have a healthy child. A baby can be diagnosed whether he or she is healthy by taking blood while still in mother's womb. Prenatal diagnosis is performed during the first months of pregnancy. However, the genes of parents bearing the disease must be identified before pregnancy starts. Therefore, future parents should visit a health institution before pregnancy or as soon as they think of having a child. If, in consequence of prenatal diagnosis, unborn child is healthy, pregnancy may continue. Otherwise, parents may decide to terminate pregnancy.

I, (patient's name), ……. have taken the Premarital Haemoglobinopathy scanning.
I have discovered that I'm a carrier of the disease and received necessary consultancy services in this regard. (To be written by the patient by hand) (I have READ, UNDERSTOOD and AGREE to the foregoing.)
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OR
I, (patient's name), …… or my prospective husband/wife (full name) …… have taken the
Premarital Haemoglobinopathy scanning. We have discovered that we are both Suspected/Confirmed Carriers of the disease. We received necessary consultancy services in this regard and were informed about the risks thereof. (To be written by the patients by hand) (We have
READ, UNDERSTOOD and AGREE to the foregoing.)
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Patient's Full Name	**Patient's Full Name**
Signature, Date	**Signature, Date**
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Legal provisions concerning the subject:

1-LAW NO. 3960 CONCERNING THE FIGHT AGAINST HEREDITARY DISEASES

2-REGULATION FOR THE PROGRAM OF HEMOGLOBINOPATHY CONTROL AS A HEREDITARY BLOOD DISEASE AND FOR DIAGNOSIS AND TREATMENT CENTRES