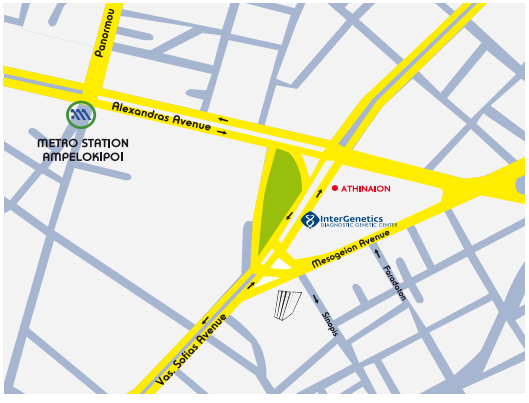

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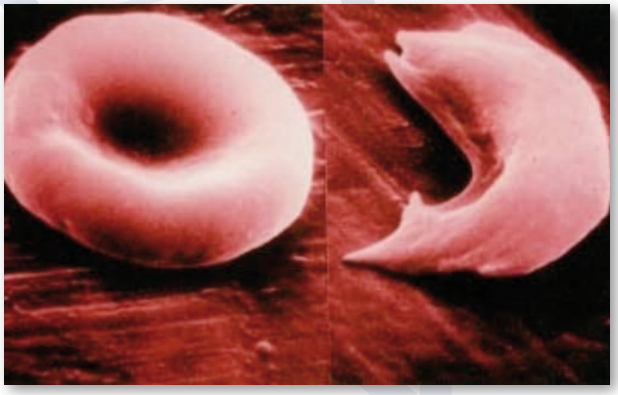
GENETIC TESTING FOR HEMOGLOBINOPATHIES



- ✓ *genetic testing covers >99,99% of mutations in the Greek population and in other ethnic groups*
- ✓ *comprehensive testing led to the discovery of novel disease mutations*
- ✓ *successful therapy of affected children through transplantation from a histocompatible sibling, following preimplantation genetic diagnosis (PGD)*

The thalassemias (α- and β-thalassemia) and hemoglobinopathies (e.g., sickle cell anemia) are generally the most common genetic disease in the Mediterranean, the Arab world, South-East Asia, India and other countries. They comprise of α-thalassemia and β-thalassemia, depending on the type of hemoglobin chain that is affected. The isolation and characterization of the α-type and β-type genes of hemoglobin about 35 years ago, has allowed us to analyze the genes and uncover the genetic lesions which lead to the disease.

These lesions, called mutations, are responsible for the pathological expression of the α-or β-type chain of hemoglobin, leading to the corresponding type of thalassemia (α- or β-).

Although most carriers, but not all, can be detected by simple blood tests, knowledge of the disease mutations is necessary for genetic counseling of reproduction and for prenatal diagnosis.

Why and how is the test performed

The test involves detailed analysis for mutations in a person's DNA in order to detect carriers or affected individuals. The number of mutations in the α- and β-type genes of hemoglobin, which may cause the disease, is quite large and are associated with different disease severity.

When both copies of the gene harbor a mutation, then the person is a homozygote (same mutation in both copies), or double heterozygote (different mutation in each copy), and are both affected.

By analyzing the DNA of these genes it is therefore possible to accurately determine whether a person is actually a carrier or affected and to identify the specific mutation(s) involved. The identification of the mutations in the parents is very important, since only then can we proceed to the detection of these mutations in the fetus through molecular prenatal testing and determine if it is healthy or affected.

Genetic testing involves the detection of mutations and deletions/rearrangements of the α- and β-type genes, through a combination of advanced molecular genetics techniques. The testing strategy applied permits the detection of all possible genetic lesions associated with the disease.

Note: It is important that both parents undergo simple hematological testing (blood count, iron and ferritin levels, hemoglobin electrophoresis and HbA2 quantitation) prior to pregnancy, so that their possible carrier status is revealed at an early stage.