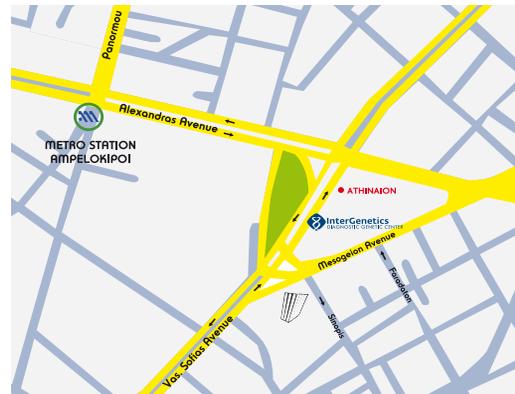
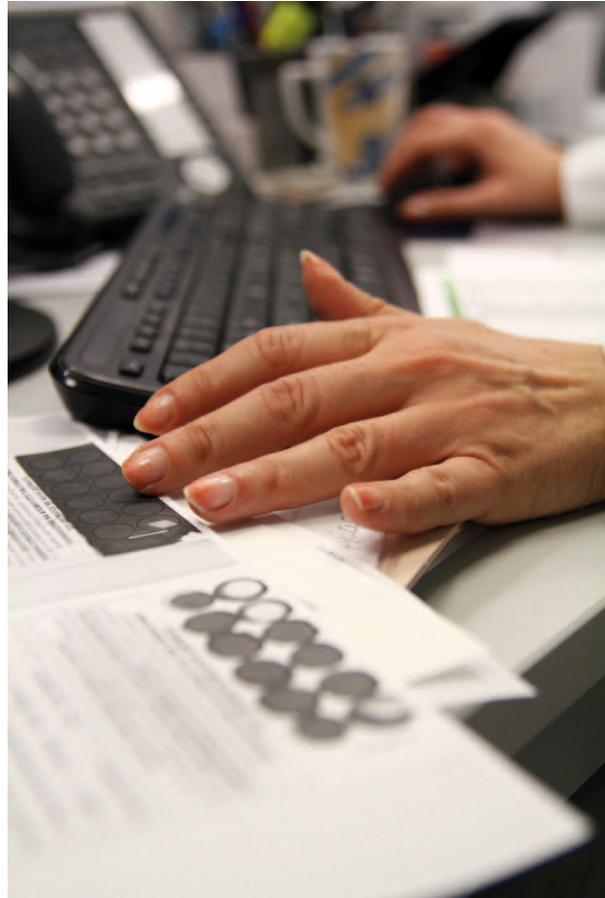


It is important to stress that sampling through a maternal dry blood sample, utilizing the special paper card, is an internationally accepted and fully scientifically documented procedure, which ensures the stability of the sample and therefore the accuracy of the measurement, and is applied by some of the largest and most reputable laboratories in the world.



InterGenetics
DIAGNOSTIC GENETIC CENTER

120, Vas. Sofias Av.,
11526 Athens, Greece

T/ (+30) 2107705010 • 2107756588

T/ (+30) 2107705125 • 2104177919

F/ (+30) 2107705011

info@intergenetics.eu
www.intergenetics.eu

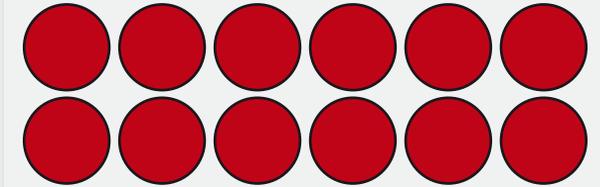


© InterGenetics 2013



InterGenetics
DIAGNOSTIC GENETIC CENTER

DRY TEST A SAFE METHOD FOR DETERMINING THE RISK FOR DOWN SYNDROME DURING PREGNANCY



- ✓ *measurement of the levels of PAPP-a, free β -hCG, AFP and uE3 from a maternal dry blood sample*
- ✓ *established method, internationally accepted and scientifically validated*
- ✓ *longstanding experience, (>133,000 samples) and detailed statistical data*
- ✓ *allows for simultaneous testing from the same sample for the common mutations F508del of cystic fibrosis and 35delG of hereditary deafness*

This screening test calculates the risk for Down syndrome (trisomy 21) and Edwards syndrome (trisomy 18) in pregnancy, taking into account the age of the mother, the levels of biochemical markers produced during pregnancy and detected in the maternal blood, with the aid of a special software.

The test is applied to dry blood samples of the mother and the biochemical markers are measured by automated ELISA techniques.

In the 1st trimester (10 to 13 weeks), the test measures the levels of the biochemical markers free β -hCG (beta-human chorionic gonadotropin) and PAPP-a (placental protein). The results are then evaluated in combination with the provided nuchal translucency (NT) measurement of the fetus with the help of dedicated software in order to determine the final risk.

In the 2nd trimester (16 to 22 weeks), the test measures the levels of the biochemical markers free β -hCG (beta-human chorionic gonadotropin), aFP (alpha-fetoprotein) and uE3 (unconjugated estriol). The results are then evaluated with the help of dedicated software in order to determine the final risk.

The results are always given in the form of a probability and the threshold for a high risk pregnancy with Down's syndrome is 1/250. Above this threshold (e.g. probability 1/200 or 1/100, etc.) the pregnancy is characterized as high risk and prenatal chromosomal diagnosis is recommended by through chorionic villi (1st trimester) or amniotic fluid (2nd trimester) sampling, which provide absolute diagnostic accuracy.

Our considerable experience over the years reveals that 4% of the tests performed in 1st trimester

and 6% in the 2nd trimester result in a high risk assessment. However, even in a case with a low-risk result, birth of a child with Down syndrome or other chromosomal abnormalities cannot be excluded, since the test has the ability to detect in the 1st trimester about 87% of pregnancies in combination with NT measurement, or 62% without NT, and 68% in the 2nd trimester.

The "dry test" allows, from the same sample, the simultaneous detection of:

1. the F508del mutation of cystic fibrosis

Cystic fibrosis (CF) is one of the most common genetic disorders, with a carrier frequency in the Greek population of about 4% (1/25 people).

There are numerous mutations responsible for the disease (>1,500), but one specific mutation, F508del, is found in Greece with a frequency of approximately 53% in patients and a carrier frequency in the general population of about 2% (1/50 people).

Through the analysis of this mutation we can detect approximately half of the carriers of the disease and in case of a positive finding it is important immediately test the father (for at least 85% of Greek mutations), to determine whether there is a need to perform prenatal diagnosis early in pregnancy.

F508

2. the 35delG mutation of autosomal non-syndromic recessive deafness

Prelingual non-syndromic deafness, i.e. the form that occurs prior to the development of lingual communication in a child), has a genetic etiology in >50% of cases and is primarily caused by recessive mutations.

There are more than 100 genes implicated in deafness. However, mutations in only one of these, the gene for connexin 26 (GJB2), are present in more than 65% of patients with non-syndromic hearing loss.

In fact, a single mutation, called 35delG, represents >90% of all pathological gene mutations and has a carrier frequency of about 3.5% in the general population (1/28 persons).

