Medicina Fetal
Ginecologia
Obstetrícia

Non-Invasive Prenatal Test (NIPT)

What is the Harmony test?

The Harmony test analyzes cell free DNA in maternal blood and gives a strong indication of whether the fetus is at high or low risk of having trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome) or trisomy 13 (Patau syndrome). It also analyses sex chromosomes, checking baby's gender.

- The test identifies 99%, but not all, of the fetuses with trisomy 21, 97% of fetuses with trisomy 18 and 92% of fetuses with trisomy 13.

How is the test performed?

- The test consists in take a sample of maternal blood.

What are the test types?

There are 2 types of tests.

- The simple one, assesses only chromosomal abnormalities (chromosomes 21, 18,13 and sex).
- The full NIPT also assesses microdeletions of genes – the number and which ones will depend on the lab chosen.

What is trisomy 21, 18 or 13?

In humans, there are 23 types of chromosomes and most people have a pair of each one of these chromosomes (therefore a total of 46 chromosomes). In trisomy, there are three rather than two of a particular chromosome (total of 47 chromosomes). The most common trisomies are those of chromosomes 21, 18 and 13.

- Trisomy 21 is found in about 1 in 700 births and the risk increases with maternal age. The condition is associated with intellectual disabilities and some physical defects, most commonly heart abnormalities. The life expectancy is about 60 years.
- Trisomies 18 and 13 are found in about 1 in 7,000 births and the risk increases with maternal age. The conditions are associated with severe mental handicap and several physical defects. Most affected individuals die before or soon after birth and they rarely survive beyond the first year of life.

When do I expect to get the results?

- In most labs, it takes up to two weeks to get the results back.
- In about 5% of cases the test does not give a result. This is due to technical problems with the analysis of the sample and does not suggest that there is a problem with the baby. Most of the labs will let you repeat the test (at no cost) and there is a 50% chance that the test will give a result.

What would the results show?

- If the Harmony test shows that there is a high risk that the fetus has trisomy 21 or 18 or 13 it does not mean that the fetus definitely has one of these defects. If you want to be certain if the fetus has one of these defects you should have CVS or amnio.
- If the Harmony test shows that there is a low risk (less than 1 in 10,000) that the fetus has trisomy 21 or 18 or 13 it is unlikely that the fetus has one of these defects.

*Quality of results depend on the fetal fraction obtain

Do I need to have any other tests?

- The Harmony test does not provide information on other rare chromosomal abnormalities. If the scan at 11-13 weeks shows a high nuchal translucency (more than 3.5 mm) or major defects, such as exomphalos, holoprosencephaly, heart abnormalities or megacysis, the risk for some rare chromosomal defects may be high. In such cases you may choose to have CVS or amnio.
- The Harmony test does not provide information on physical defects, such as heart or brain abnormalities and spina bifida, or fetal growth. It is therefore advisable that you still have ultrasound scans at 11-13 weeks and at 20-22 weeks to examine the fetal anatomy and at the 3rd trimester to examine the fetal growth.