



PRENATAL DIAGNOSIS: ARRAY CGH

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Prenatal diagnosis



Worldwide, millions of individuals are affected by dominant or recessive genetic mutations. In order to avoid the transmission of severe pathogenic genetic variants and to enable early detection of genetic disorders, prenatal testing is offered.

There are two types of prenatal diagnosis techniques: Non-invasive and Invasive ones.

Non-invasive techniques

Non-invasive procedures are used to detect general disease or deformations and don't have particular risk of abortions.

Rh safe and Prenatal safe are two of them.

Invasive techniques

These techniques have 1% risk of abortion because they take the cells from the amniotic fluid (Amniocentesis) from the uterus or from the chorionic villus (Chorionic villus sampling).

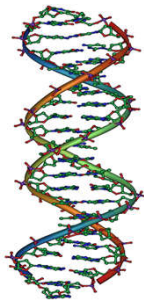
HOW CAN SCIENTISTS ANALYSIS THE FETAL DNA?

Several technologies have been invented in order to examine the fetus DNA.

There are general or specific analysis that can use fast methods, so the result would be ready sooner, or slower methods which are very precise so also microdeletions and micro mutations can be detected.

The traditional way to analyse the fetus DNA is the karyotype but more specific techniques are being developed.

One of them is the microarray CGH, we want to examine what they are and how do they work.

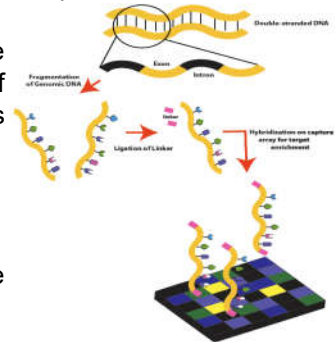


ARRAY CGH



What is it?

Array CGH is a significant advance in technology that allows detection of chromosomes imbalances that are too small to be detected by looking down the microscope. Karyotyping is only as good as the resolution of a microscope and is not able to detect subtle chromosomes changes. These smaller alterations, often called submicroscopic alterations because they cannot be seen down the microscope, can still disrupt growth and development. These very small changes are often called microdeletions and micro-duplications. It compares the fetus DNA with a control DNA sample and identifies differences between the two sets of DNA. In this way deletions and duplications can be identified.



How does array CGH work?

A microarray works by exploiting the ability of a DNA molecule or strand to bind specifically to or hybridize to, another DNA molecule. The microarray comprises tens of thousands of short sequences of DNA arranged in a precise grid on a glass slide called a chip. DNA from the patient is "digested" (chopped up into short lengths or fragments), then these fragments are labeled with a colored fluorescent dye. Reference DNA, From a person, or pool of people, with no genetic abnormalities, is labeled with a different colored fluorescent dye.

The fluorescent dyes commonly used are red and green. Reference and patient samples are mixed together and applied to the chip and hybridization takes place- the fragments of DNA hybridize with their matching probes on the array. The chip is then scanned in a machine called a microarray scanner which measures the amount of red and green fluorescence on each probe. The microarray scanner together with computer analytical software calculates the ratio of the red to green fluorescent dyes to determine whether, for the piece of DNA represented by each probe. The patient sample has the correct amount of DNA (shown as yellow), too much DNA (a duplication) which would be shown by too much red, or too little DNA (a deletion) shown by too much green.

