Fertility Design
GYNAECOLOGIST - FERTILITY SPECIALIST
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Chromosome Analysis, Peripheral Blood (karyotyping)

The genetic material that is transferred from generation to generation is composed by structures called chromosomes. Human beings have approximately 23.000 genes on 46 chromosomes. In somatic cells, chromosomes appear as 23 homologous pairs to form the diploid number of 46. There are 22 pairs of matching chromosomes, the autosomes and one pair of sex chromosomes. Throughout the blood test investigation of karyotyping, it is able to identify genetic problems and examine chromosomes in a sample of cells, distinguishing the cause of disorder or disease. The classic Karyotype is not able to detect very small deficiencies (micro-deficiencies, microdeletions) and very small duplications (micro-duplications). This possibility is provided by the Extended Control (karyotype pair 100%).

The examination can identify

- Numerical abnormalities such as TRISOMY 21(Down syndrome)
- Syndromes as Di George, Williams, Miller Dieker, Prader Willi/Angelman
- The existence of possible deficiencies and / or doubling across all chromosomes
- Telomere deficiency syndromes: the deficiency in the ending parts of the chromosome that often associated with mental retardation. Telomeres are DNA-protein structures that protect chromosome ends
- The investigation of specific genes associated with more than 120-disease syndromes

The analytical and detecting ability of the karyotype pair (100% karyotype pair) is 100 to 1,000 times greater than that of the classic karyotype and detects micro-defects and microdeletions comparing to the classical karyotype. The examination of karyotyping 100%, despite the fact that offers a greater depth of analysis than the classic karyotype, has limits to the degree of analytic ability. Thus, it cannot detect diseases due to even smaller size changes at the genetic material (point mutations).Associated with certain genetic diseases, such as sickle cell anemia or cystic fibrosis.

Cases that the aforementioned test is considered necessary:

- In cases of miscarriages and primarily abortion (> 3)
- In cases of osteoarthritis

- In cases where the woman gave birth to an embryo, with proven chromosomal disorder
- Repeated failed IVF procedures

The examination is performed by taking peripheral blood from the fetuses and the results are usually given after, approximately, 15 working days.

Normal Results

- Females: 44 autosomes and 2 sex chromosomes (XX), written as 46, XX
- Males: 44 autosomes and 2 sex chromosomes (XY), written as 46, XY