REFERENCE LABORATORY GENETICS

Pablo Iglesias, 57 – Polígono Gran Via Sur
08908 L'Hospitalet de Llobregat (Barcelona)
Tel. 932 593 700 – Fax. 932 845 000

VeriRef Gold®

NONINVASIVE PRENATAL SCREENING FOR THE DETECTION OF ANEUPLOIDIES AND DELETIONS/DUPLICATIONS (CNVs) IN MATERNAL BLOOD

High Risk of Sex Chromosome Trisomy (XYY)

Request No.:

Client:

Analysis code:

Patient Name:

Date of Birth: Patient Ref.:

IVF: Gestional Age:

Draw Date:

Sample Arrival Date: Report Date:

Number of fetuses: 1

RESULT: Sex chromosome trisomy is detected (XYY)

Chromosomes	Result	Interpretation
Chromosome 21	No aneuploidy detected	Compatible with two copies of chromosome 21
Chromosome 18	No aneuploidy detected	Compatible with two copies of chromosome 18
Chromosome 13	No aneuploidy detected	Compatible with two copies of chromosome 13
Chromosome X/Y	Aneuploidy detected	Compatible with one copy of chromosome X and two copies of chromosome Y (XYY)
Rest of chromosomes	No aneuploidy detected	Compatible with two copies of the rest of chromosomes
CNVs	No variants are detected	Compatible with normality

Fetal sex: Male

Fetal fraction: 5 %

A high risk result should be confirmed from an amniotic fluid sample. In these cases we offer a free confirmation by QF-PCR or CGH array.

METHODOLOGY

Whole genome analysis of fetal cfDNA by means of MPS (Massive Parallel sequencing). Illumina NextSeq® 500 Sequencer and last generation bioinformatic analysis used.

OBSERVATIONS

VeriRef Gold® is a laboratory test that detects, from the DNA released by the placenta in maternal blood, aneuploidies on all chromosomes and CNVs (deletions and duplications) on autosomal chromosomes

This test has the following yields in the case of simple pregnancies:

Chromosome	Sensibility	Specificity
21	>99.9% (130/130)	99.9% (1982/1984)
18	>99.9% (41/41)	99.9% (1995/1997)
13	>99.9% (26/26)	99.9% (2000/2002)
Monosomy X	95.0% (19/20)	99.9% (1961/1963)
XX	>99.9% (995/995)	99.8% (995/997)
XY	>99.9% (966/966)	>99.9% (966/966)
Rest of chromosomes	96.4% (136/141)	99.8% (2001/2005)

Deletions and duplications	Sensibility	Specificity
CNVs	74.1% (20/27)	99.8% (2000/2004)

This test is valid in case of in vitro fertilization and in gestations from oocyte donation.

Noninvasive prenatal testing (NIPT) based on cell-free DNA analysis from maternal blood is a screening test; it is not diagnostic. Test results must not be used as the sole basis for diagnosis. Further confirmatory testing is necessary prior to making any irreversible pregnancy decision.

LIMITATIONS: Currently, in twin pregnancies it is only possible to detect aneuploidies on chromosomes 13, 18 and 21, and the presence of the Y chromosome. In simple pregnancies the reported sexual abnormalities are as follows: XO, XXX, XXY and XYY. This test does not detect: CNVs of size <7Mb, CNVs on sex chromosomes, polyploidies (such as triploidies), or balanced chromosomal rearrangements. A negative result does not eliminate the possibility of pregnancy presenting other genetic conditions or congenital defects. The presence of large duplications or deletions greater than 75% of the chromosome size are interpreted as aneuploidy of the complete chromosome. A false positive or negative may be due to the presence of maternal chromosomal alterations, a high index of maternal body mass, recent transfusion in the mother, mosaicism confined to placenta or the existence of an evanescent twin or fetal death.