REFERENCE LABORATORY GENETICS

ENAC ENSAYOS Nº 1065/LE2112

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VeriRef®

NONINVASIVE PRENATAL SCREENING OF ANEUPLOIDIES IN MATERNAL BLOOD

High Risk of Trisomy 18

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: It was detected a trisomy of chromosome 18

Chromosomes	Result	Interpretation
Chromosome 21	No aneuploidy detected	Compatible with two copies of chromosome 21
Chromosome 18	Aneuploidy detected	Compatible with chromosome 18 trisomy
Chromosome 13	No aneuploidy detected	Compatible with two copies of chromosome 13
Chromosome X/Y	No aneuploidy detected	Compatible with two copies of sex chromosomes (XX)

Fetal sex: Female

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.

POWERED BY illumina®

METHODOLOGY

Analysis of free fetal DNA by MPS (Massive Parallel sequencing) of the complete genome, Illumina NextSeq® 500 Sequencer and State-of-the-Art Bioinformatics Methods.

OBSERVATIONS

VeriRef® is a laboratory test that analyzes, from it is DNA released by the placenta in maternal blood, the risk of aneuploidies on chromosomes 13, 18, 21, X and Y in the fetus. This test presents the following performances:

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)

This test is valid in case of in vitro fertilization and in pregnancy from egg 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: This test is designed as 'screening' for chromosomal aneuploidies and is validated for chromosomes 13, 18, 21, X and Y. It has been validated for single and twin pregnancies of gestational age equal to or greater than 10 weeks. Currently, it is not possible to detect aneuploidies of sex chromosomes in twin pregnancies. No other possible chromosomal, subchromosomal or genetic alterations are detected by the test used. A false positive or negative may be due to the presence of maternal chromosomal alterations, a high maternal body mass index, placental confined mosaicism, or the existence of an evanescent/deferred twin (See yield table).