sistemas genómicos
BIOMEDICINE

BabyTest Plus – Non Invasive Prenatal Testing

BabyTest Plus is a screening test that allows detecting chromosomal abnormalities in the fetus during the pregnancy. It is **the most comprehensive prenatal analysis available on the market, studying all the chromosomes (22 pairs of autosomes, 2 sex chromosomes)**, and detecting amongst others:

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)
- Abnormalities in the sexual chromosomes:
  - Turner syndrome (monosomy X: 45, X)
  - Klinefelter syndrome (47, XXY
  - Triple X
  - Polysomies X
  - XYY karyotype
- Large size microdeletions and microduplications*
- Chromosomal mosaicisms depending on their percentage in cell free fetal DNA*

BabyTest Plus also allows establishing fetal gender.

BabyTest Plus technology

During the pregnancy, short fragments of fetal DNA go through the placenta and circulate in maternal blood with short fragments of the mother's DNA. From a sample of maternal blood, the DNA is extracted and sequenced with Next-Generation Sequencing (NGS) technology. Reads obtained from sequencing are aligned against the corresponding reference genome (Homo sapiens). The reference genome is divided into genomic intervals or bins, and the number of reads present in each of those bins is determined applying various normalisation processes depending on the studied genomic context. Bin counts are subsequently compared with available bin counts coming from unaffected reference samples, so that the gain or loss in the number of copies of each chromosome can be determined.

**In the process of validating our internal data for those conditions*

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BabyTest Plus reliability

BabyTest Plus technical data	
Sensitivity	>99%
Specificity	>99%

At Sistemas Genómicos, we are experts in Next-Generation Sequencing technology, as we were pioneers in using it. BabyTest Plus relies on the experience of our professional teams of Medical Genetics, Bioinformatics and New Technologies, who have studied more than 145.000 genes, near 2.000 gene panels and more than 500 exomes, amongst other studies, and have many scientific publications.

BabyTest Plus indications

BabyTest Plus is recommended for pregnant women who wish to rule out chromosomal abnormalities in their fetus. In particular, it is indicated for women at high risk for those abnormalities (advanced maternal age, history of pregnancies with chromosomal abnormalities, etc.), or with abnormal ultrasound findings or biochemical screening test.

This test should be done from the 10th week of pregnancy**. BabyTest Plus is available for IVF pregnancies, including those with egg donor. It can be performed in twin pregnancies**.

Results interpretation

- **Low risk**: the probability of aneuploidies in the fetus for the analysed chromosomes is low
- **High risk**: there is a higher probability of aneuploidies in the fetus for the analysed chromosomes. The high risk with BabyTest Plus must be confirmed with invasive prenatal testing.
- **Undetermined risk**: it was not possible to determine whether the fetus was affected or healthy, with the analysis. In those cases, it is necessary to test another sample.

Procedure to send samples

1. Obtaining the sample (maternal blood)

   Extract 20 ml of maternal blood in 2 tubes with preservatives for cfDNA (cell-free plasma DNA).
   Identify the tubes with the reference or the identification of the patient.

2. Fill in the NON INVASIVE PRENATAL TESTING REQUEST FORM and sign the INFORMED CONSENT (which should be signed by the doctor or the patient). Annex it to the sample. It is particularly important to indicate the following patient's information: weeks of gestation, weight (Kg) and size (cm) of the patient.

3. Pack the sample well to avoid knocks and brusque changes in temperature; send it to the address below with the request form and the informed consent form*:

Europe and rest of the world

SISTEMAS GENÓMICOS, S.L.
Ronda G. Marconi, 6
Parque Tecnológico de Valencia 46980
Paterna (Valencia) Espagne

Canada and USA

SISTEMAS GENÓMICOS,
4936 Yonge Street
Suite #603 (inside UPS facility)
Toronto, ON M2N 6S3
Canada

*IMPORTANT: The time between extracting the maternal blood sample and its reception in the laboratory should not exceed 72h. The sample should be shipped and maintained at all time at room temperature (6-35ºC). Do not refrigerate or freeze the samples.

*** Consult the specific conditions in the Informed Consent.*

Contact:

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