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New Integrated Medical Genetics

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La Suma de Todos
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NIMGenetics is a Genetic Diagnosis centre authorised by the Department of Health and Consumption of the Community of Madrid, registered in the corresponding Register under number CS 10673.

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TrisoNIM® EXCELLENCE

Non-invasive Prenatal Screening Test

Your baby's reflection

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The identification of the baby's DNA in the mother's blood during pregnancy has revolutionized prenatal diagnosis

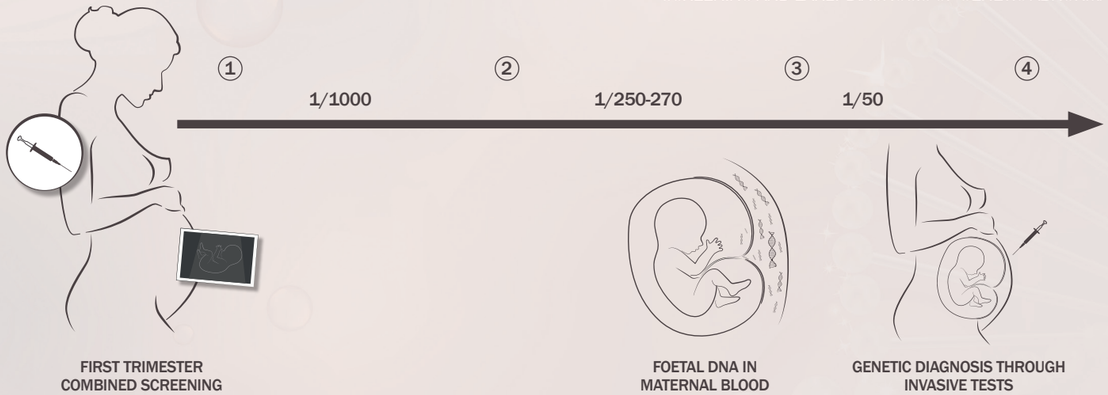
In order to rule out the presence of certain chromosomal abnormalities in the early stages of pregnancy, the first trimester combined screening is carried out. The combination of the result of foetal ultrasound and certain maternal biochemical parameters allows to establish the risk of presenting some of these alterations, with a limited sensitivity.

Once the risk is established, the study of foetal DNA in maternal blood enables the screening for trisomies 21, 18, and 13 (Down syndrome, Edwards, and Patau, respectively) with a sensitivity close to 100%. The high reliability of this test often avoids invasive tests, such as amniocentesis or chorionic villus sampling, which involve a risk of miscarriage.

The exponential advance of technology has made it possible to provide information on other regions of the genome, including microdeletion syndromes. These alterations constitute a broad group of serious pathologies that, despite the incidence of each one of them, individually, is low, their incidence, considered jointly, is high in the population.

Main studies for the evaluation of chromosomal abnormalities during pregnancy

Diagram based on SEGO's recommendations:
SCREENING AND EARLY DIAGNOSIS OF GENETIC ABNORMALITIES



1. At this level of risk, the foetal DNA test complements the results of conventional screening, increasing its reliability in the study of chromosomopathies.
2. A contingent ultrasound test and, if necessary, a foetal DNA test are recommended.
3. The foetal DNA test is the best screening method for chromosome 21 trisomy, according to the SEGO expert consensus.
4. Invasive tests are indicated in the case of a high-risk result in prenatal screening and in the presence of certain ultrasound findings.

• As of week 10, suitable for natural or IVF pregnancies

TrisoNIM® EXCELLENCE

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Foetal DNA in maternal blood test offering an extended study of clinically relevant syndromes that may appear during pregnancy.

The Microdeletion Syndromes, included in TrisoNIM® Excellence, constitute a large group of syndromes with serious clinical repercussions and an overall incidence of 1-2%.

This test also informs you, very early on, of the sex of your baby.

*Results back in 8 days**

ANALYSES ALL CHROMOSOMES

▸ **STUDIES ANEUPLOIDY FOR ALL CHROMOSOMES**

DETECTS:

- Trisomy 21, associated with Down syndrome
- Trisomy 18, associated with Edwards syndrome
- Trisomy 13, associated with Patau syndrome

REPORTS:

- Aneuploidy for sex chromosomes and foetal sex
- Aneuploidies of the remaining autosomal chromosomes

▸ **REPORTS 38 MICRODELETION SYNDROMES**

(*): Business days as of the reception of the sample.

TrisoNIM® EXCELLENCE

• Cutting edge technology

The combination of state-of-the-art sequencing, with more than 25 million readings, and the most powerful analytical system, translates into the highest sensitivity level.

• Reliability

Risk prediction and foetal fraction calculation are performed using a double algorithm*, increasing the accuracy of the analysis.

CHROMOSOMAL ALTERATION	DETECTION CAPACITY	FALSE POSITIVES
T21 (Down´s syndrome)	99,17%	0,05%
T18 (Edwards syndrome)	98,24%	0,05%
T13 (Patau syndrom)	99,99%	0,04%
X0 (Turner´s syndrome)	>95%	-
Detection chromosome Y	>98%	-

Public data: Zhang H et al. Ultrasound Obstet Gynecol 2015;45:530–538

(*): Analysis algorithm with CE-IVD marking for trisomy 21, tested on more than 3 million pregnant women.

• The most comprehensive and complete test

Extended study, with the best selection of microdeletion syndromes in foetal medicine.

For your peace of mind

• With the best prenatal diagnosis platform

A high-risk result after a foetal DNA test must be confirmed by an invasive test. NIMGenetics offers you KaryoNIM® Prenatal, free of charge. This test makes it possible to quickly and effectively establish a genetic diagnosis through the analysis of 124 syndromes.

• Hand in hand with the most qualified experts

Endorsed by the world's most relevant genomic company (BGI), the NIMGenetics team, made up of experts in Medical Genetics, members of the AEDP and the AEGH, is recognized for its specialization in the area of prenatal genetic diagnosis.

AEDP: Spanish Association of Prenatal Diagnosis
AEGH: Spanish Association of Human Genetics

• Certified quality

- The **UNE-EN ISO 15189:2013** accreditation for screening foetal aneuploidies (13, 18, 21, X, and Y chromosomes) and for foetal sex determination in maternal blood by massive sequencing (NGS).
- The **ISO 9001:2015** accreditation for the provision of analysis services for genetic diagnosis in the pre-analytical, analytical, and post-analytical stages for the specialities of genomics, non-invasive prenatal testing and molecular diagnosis.

TrisoNIM® EXCELLENCE

LIST OF REPORTED MICRODELETION SYNDROMES

SYNDROME	#OMIM
1p36 Microdeletion	607872
1p32p31 Microdeletion	613735
2q33.1 Microdeletion	612313
2p12p11.2 Microdeletion	613564
3pterp25 Microdeletion	613792
4p16.3 Microdeletion	194190
4q21 Microdeletion	613509
5q12 Microdeletion	615668
Cri-du-chat Syndrome	123450
5q14.3q15 Microdeletion	612881
6pterp24 Microdeletion	612582
6q11q14 Microdeletion	613544
6q24q25 Microdeletion	612863
Langer-Giedion Syndrome	150230
9p Microdeletion	158170
DiGeorge 2 Syndrome	601362
10q26 Microdeletion	609625
11p11.2 Microdeletion	601224

SYNDROME	#OMIM
Jacobsen Syndrome	147791
WAGRO Syndrome	612469
WAGR Syndrome	194072
Frias Syndrome	609640
14q11q22 Microdeletion	613457
15q26qter Microdeletion	612626
15q26 Microdeletion	142340
15q11q13 Duplication	608636
Prader-Willi Syndrome	176270
Angelman Syndrome	105830
16p12p11 Microdeletion	613408
16q22 Microdeletion	614541
Yuan-Harel-Lupski Syndrome (combination of CMT1A and Potocki-Lupski)	616652
17p13.3 Microdeletion (Miller-Dieker Synd.)	247200
17p13.3 Duplication	613215
17p11.2 Microdeletion (Smith-Magenis Synd.)	182290
17p11.2 Duplication	610883
18q Microdeletion	601808
18p Microdeletion	146390
DiGeorge Syndrome	188400

TrisoNIM® Excellence reports microdeletions sized ≥ 5 Mb, with the exception of DiGeorge Syndrome, where detection capability is up to 3 Mb.
In approximately 30% of cases, Angelman and Prader-Willi Syndromes are produced by genetic alterations not detectable by any foetal DNA in maternal blood test.

Your TrisoNIM® Excellence step by step

1. Consult with your specialist doctor. Based on the advice received, you sign the informed consent.
2. Contact us to coordinate a date for the collection of the sample.
3. At NIMGenetics, we will analyse the sample and issue the report.
4. Visit your specialist doctor for post-test advice.