GTEST

WHOLE
GENOME
ANALISYS

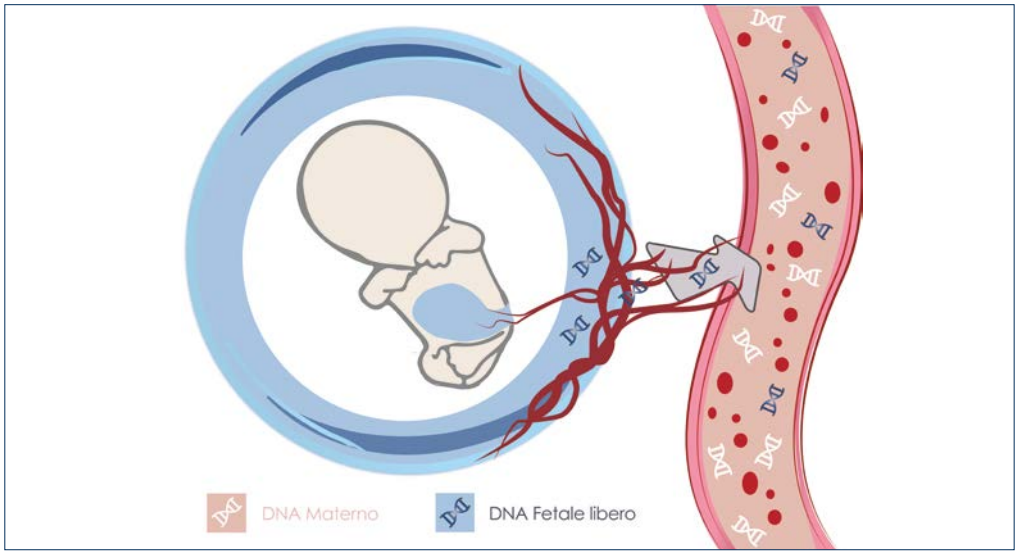
NON INVASIVE PRENATAL DNA TEST

BIOSCIENCE
GENOMICS

Università di Roma
Tor Vergata
spin off

THE GTEST

is a non-invasive prenatal analysis for screening fetal chromosomal diseases.
From a maternal blood sample, important information on the health of the fetus is obtained, without compromising the pregnancy in any way.
Thanks to the next generation sequencing technology (Next Generation Sequencing) and the use of specific proprietary calculation algorithms it is possible to sequence the fragments of the DNA present in the maternal plasma to attribute its belonging to a specific chromosome, thus evaluating aneuploidies and chromosomal anomalies structural.



CELL FREE FETAL DNA FROM MATERNAL BLOOD

During the first weeks of pregnancy, the embryo is fed by a group of cells (trophoblast) that will give rise to the placenta. Some of these cells will "break" naturally and will pour into their mother's bloodstream the DNA contained in them, in the form of fragments, going to make up what is called free fetal DNA.

SEQUENCING TECHNOLOGY

The new generation sequencing (NGS) allows to order all the fragments of free DNA belonging to a specific chromosome and to compare them quantitatively with those normally present in a healthy individual to define the risk (high or low) that the fetus may be affected by chromosomal anomalies object of the test.

TRISOMIES

Trisomies are characterized by the presence of a chromosome more than the couple present in the chromosomal kit of a normal individual. The most common at birth is Trisomy 21, associated with Down Syndrome; Trisomy 18 (Edwards Syndrome), Trisomy 13 (Patau Syndrome), Trisomy 22, Trisomy 16 and Trisomy 9, usually causing premature abortion, endouterin death, are the rarest., perinatal or otherwise of short life expectancy.

SEX CHROMOSOMES ABNORMALITIES

The sex chromosomes aneuploidy are characterized by the absence of a sex chromosome, in the case of Turner's Syndrome, or by the presence of an extra sex chromosome, in the case of Klinefelter Syndrome, of the Jacobs Syndrome and of the XXX Syndrome.

DELETIONS/DUPLICATIONS

Deletions/duplications are unbalanced chromosomal anomalies characterized by the loss (deletion) or duplication of a chromosome's section and consequently of the genes located on the chromosomal fragment.
Some deletions cause rare syndromes, which may be associated with: cardiac abnormalities, facial dysmorphism and labiopalatoschisis, eating disorders in early childhood, changes in the functioning of the gastrointestinal tract and immune system, mental retardation or neuro-cognitive impairment.
The severity of these clinical manifestations varies, from individual to individual, depending on the size and position of the absent chromosomal fragment.

GTEST INCLUDES:

The G-test evaluates the presence of several deletions responsible for the onset of:

Sindrome Cri du Chat
Sindrome da delezione 1p36
Sindrome da delezione 2p33.1
Sindrome da delezione 16p12,2-p11,2
Sindrome di Jacobsen da delezione 11q23
Sindrome di PraderWilli/Angelman 15q11,2
Sindrome Di George 2,12
Sindrome di Van Der Woude

WHOLE GENOME ANALISYS (WGA)

The analysis of the entire genome makes it possible, by applying a proprietary calculation algorithm (FCAPS *), to widen the search for aneuploidies and deletions / duplications to all chromosomes. The WGA Gtest represents the most recent evolution of fetal DNA tests and the overcoming of the obsolete sequencing technologies on single chromosomes (still existing for their low

CLINICAL VALIDATION

The reliability of the G-test was validated through a study involving 146,958 pregnant women. This is the largest study ever published for the validation of a non-invasive prenatal test on fetal DNA from maternal blood.

Sensitivity and specificity data, shown in the table below, refer to 112.669 tests, carried out as part of the study, for which it was possible to obtain a diagnostic check of the result. To date, over 2,500,000 tests performed worldwide are in line with the validation study data.

Trisomia	Sensibilità*	Falsi positivi**	Valore preditivo negativo***
T21	99,17%	0,05%	99,99%
T18	98,24%	0,05%	99,99%
T13	100%	0,04%	99,99%

Zhang et al., Non-Invasive Prenatal Testing For Trisomy 21, 18 and 13 – Clinical Experience from 146,958 Pregnancies. Journal of Ultrasound in Obstetrics and Gynecology, 2015.

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ADVANTAGES

The largest clinical validation in the world

To date, more than 3,000,000 G-test have been performed in over 52 countries. The data obtained indicate:

- A sensitivity greater than 99% (ability to correctly identify a fetus affected by the genetic anomalies object of the test
- Very high sensitivity not only for Trisomy 21 but also for Trisomy 18 and Trisomy 13
- The lowest no call rate between existing fetal DNA tests (only in 0.069% of cases it is impossible to provide the result of the test). The other tests based on selective DNA sequencing have a no call that normally exceeds 5% (5 out of 100 analyzes do not produce a result)
- A very low percentage of false positives (less than 0.05% of cases a high risk result is not confirmed)
- A very low percentage of resampling (only in 2.8% of cases it is necessary to repeat the blood sample to obtain the result of the test) cost). It is therefore possible to perform a non-invasive prenatal screening of the entire chromosomal kit to identify fetal chromosomal abnormalities, even of very small dimensions.

AZIENDA CERTIFICATA
UNI EN ISO 9001:2015

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