Perinatal Genetics Services

Our mission
Our mission is to transform genetic data into useful information to improve the quality and duration of an individual's life, we are experts in the utilization of clinical genomics.

Our Commitment
Veritas is The Genome Company, experts in DNA analysis and interpretation, we are committed to continuous innovation through research and education. Our services to doctors and patients will continually evolve as the global genetic knowledge in the scientific community expands.

Veritas

a LetsGetChecked company

Veritas is a leader in the clinical interpretation of the genome, both at the diagnostic level, under clinical suspicion, and at the predictive medicine level, in healthy individuals.

Veritas also has a scientific and medical team with extensive experience and wide recognition in the field of maternal-fetal medicine.

Dr. Luis Izquierdo López,
MD, MSC, PHD
Director Médico

Doctor of Medicine and Surgery; Extraordinary Doctorate Award from the UCM; Master in Medical Genetics from the University of Glasgow; Medical Genetics Chief of Services in the main University Hospitals in Madrid.

Dr. Vincenzo Cirigliano,
MSC, PhD – CTO
Director Técnico

Internationally recognized for pioneering the development and introduction into clinical routine of innovative molecular tests in prenatal diagnosis, including the rapid diagnosis of aneuploidy by QF-PCR, the introduction of microarrays and non-invasive prenatal diagnosis (NIPT).

From this knowledge of genetics and the clinical reality of prenatal medicine, Veritas offers gynecology and obstetrics units a comprehensive genetic diagnosis service that combines new generation tests with genetic counseling.

my Prenatal

The genomic revolution of the NIPT

Veritas launches a new generation of Non-Invasive Prenatal Test (NIPT), maximizing the screening performance for common trisomies. The test also provides other analysis options to expand the screening including relevant alterations in the fetal genome.

A new generation of NIPT - myPrenatal GenomeScreen

Veritas offers a prenatal screening for the most common aneuploidies (21, 18, 13, X and Y), also offering the possibility to expand the screening to include:

- **Large deletions and duplications (CNVs) of more than 7 Mb,** alterations that may cause several fetal anomalies associated with delayed cognitive development.
- **Aneuploidies in all chromosomes,** associated with fetal loss and other structural alterations.

myPrenatal - Reliable results even with low fetal fraction

myPrenatal bioinformatic algorithm combines the **fetal DNA fraction and sequencing depth** to achieve **highly reliable results** in cases of low fetal fraction, reducing the rate of no-call results.

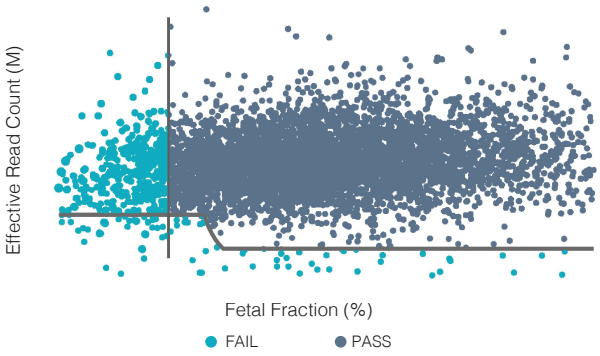
CE-IVD marked and performed in Europe

The test is performed in **our laboratories located in Europe** by an experienced team and is **CE-IVD marked.**

myPrenatal – More accurate results

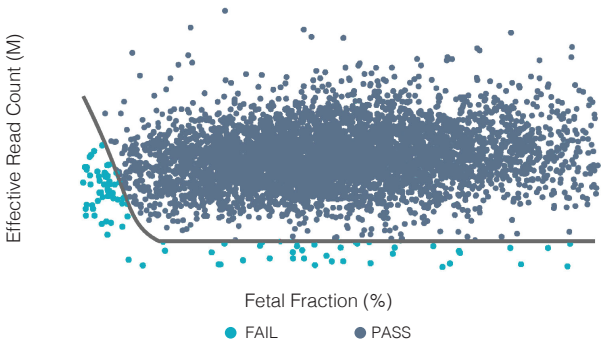
myPrenatal bioinformatic algorithm **assesses fetal fraction and sequencing depth** to deliver the best performance in all cases. In cases with low fetal fraction, results can be delivered with the same accuracy as in cases with a higher fetal fraction, if there is an appropriate sequencing depth. If the fetal fraction is high, the analysis requires a lower sequencing depth.

Other tests



Arbitrary fetal fraction limit of 4%, below this limit no results are delivered. This may occur in about 5% of cases2. The percentage of trisomies in samples with a fetal fraction <4% is significantly higher than in samples with higher fetal fractions[3].

myPrenatal



There is no established limit of fetal fraction. In cases with low fetal fraction with appropriate sequencing depth, it is possible to deliver reliable results, improving the sensitivity for detection of aneuploidies.

Different options for the healthcare provider

Singleton pregnancy

myPrenatal

Trisomy 21
Trisomy 18
Trisomy 13
+ Fetal sex +
Aneuploidies in sex chromosomes X and Y

Twin pregnancy

myPrenatal

Trisomy 21
Trisomy 18
Trisomy 13
+
Determination of Y chromosome presence

+

Singleton and Twin pregnancy

myPrenatal GenomeScreen

Duplications and deletions >7Mb (CNVs)

Aneuploidies in all chromosomes

The aneuploidies in all chromosomes in case of twin pregnancy and the analysis of the CNVs are limited to the autosomal chromosomes (not sexual).

CNVs (Copy Number Variants) larger than 7Mb are generally related to fetal anomalies and developmental delay.

Bibliography:

1) Illumina. VeriSeq NIPT Solution v2 Package Insert. 2020.
2) Norton ME, Jacobsson B, Swamy GK, et al. Cell-free DNA analysis for noninvasive examination of trisomy. N Engl J Med. 2015;372(17):1589-1597.
3) Revello R, Sarno L, Ispas A, et al. Screening for trisomies by cell-free DNA testing of maternal blood: consequences of a failed result. Ultrasound Obstet Gynecol. 2016;47(6):698-704.

myPrenatalWES

myPrenatalWES is an innovative prenatal diagnostic test indicated for pregnancies with risk of presenting a genetic alteration

What is myPrenatalWES?

- myPrenatalWES is a diagnostic service for pregnancies with fetal structural anomalies such as cardiac malformations, short femur, increased nuchal translucency, or intrauterine growth restriction, after ruling out the presence of aneuploidies.

- The test is based on fetal whole exome sequencing enhanced for the detection of CNVs (Copy Number Variants), currently studied by microarrays and pathogenic variants or SNVs (Single nucleotide Variants) in any gene that may be related to fetal anomalies. The aim of the test is the detection of a possible genetic cause in at risk pregnancies performing a single technique.

Why is it important?

The standard procedure in prenatal diagnosis implies the performance of microarrays. In the absence of findings, it is necessary a second assay to sequence specific genes possibly related to the ultrasound findings. This implies a significant delay in diagnosis and eventual therapeutic interventions.

The advanced technology of myPrenatalWES allows both assays to be performed in a single test, shortening turnaround time and maximizing the diagnostic yield.

Possible Outcomes

Variants classified as pathogenic or probably pathogenic based on American College of Medical Genetics and Genomics (ACMG) guidelines (PMID: 25741868) are included in the report. Benign variants or Variants of Uncertain Significance (VUSs) are not reported.

Advantages

Comprehensive genomic analysis	With a single test the analysis of CNVs and SNVs is performed, maximizing the diagnostic yield.
Reduced turnaround time	Turnaround time is shortened as both analyses are performed with the fetal exome sequence data.
Expert professionals	Team with extensive experience in prenatal diagnosis and genetic counseling.
Other tests	In fetuses with a normal result it is possible to request after birth the extended neonatal screening test myNewborn.
Innovative technology	Cutting-edge technology based on whole fetal exome sequencing.

Technical Information

- Whole exome sequencing with enhanced regions with 100x average coverage, sequencing more than 99% of the genes of interest at ≥20x
- In-house variant analysis tool
- The Veritas Intercontinental team includes internationally recognized experts with more than 20 years of experience in prenatal diagnosis and genetic counseling. Pioneers in the development and introduction in Europe of innovative prenatal tests into clinical routine

Pregnancy Loss

One in five pregnancies ends in spontaneous miscarriage.[1] Up to 70% of cases are caused by numeric chromosomal anomalies and around 4% are due to unbalanced chromosomal rearrangements inherited from a parent carrying a balanced chromosomal rearrangement.[2]

Innovation in prenatal medicine

At Veritas we are experts in prenatal medicine innovation. **PregnancyLoss** enables the assessment of the possible underlying genetic cause of pregnancy loss, with a blood sample from the pregnant woman, in a simple and precise way. This information is key for the reproductive genetic counselling, since it allows to establish the risk of presenting the same alteration in future pregnancies.

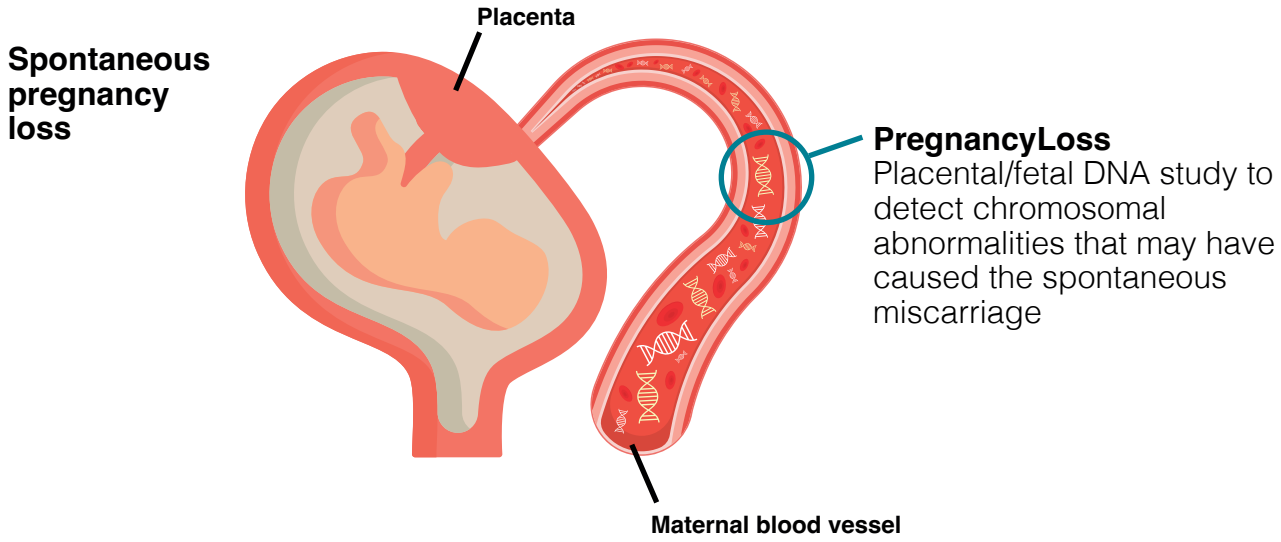
PregnancyLoss evaluates the risk of numerical and structural chromosomal abnormalities in a maternal blood sample, thus avoiding the previously mentioned complications, providing results in most cases. This is key given the impossibility of obtaining a second sample.

The following fetal chromosomal abnormalities are analysed:

- **Aneuploidies in all chromosomes**
- **Deletions and duplications** (CNVs) greater than 7Mb in autosomal chromosomes

When can it be performed

The test can be ordered **as early as 5 weeks of gestation** in pregnancies with spontaneous interruption of fetal development, anembryonic sacs, lack of fetal heartbeat or miscarriage in progress. It is especially recommended in recurrent pregnancy loss.



Results

- Between 50-70% of early pregnancy losses are caused by chromosomal anomalies, although there are other causes such as uterine malformations, thrombophilias, immunological disorders or infections. [2,3]

- Knowing the cause of the spontaneous miscarriage is the first step to be able to determine the risk of recurrence or to evaluate the need for additional tests.

- Balanced structural abnormalities in the parents can be the cause of recurrent miscarriage; evaluating those abnormalities in a first pregnancy loss allows establishing the appropriate medical management to avoid recurrence in future gestations.

Genetic Counselling

Veritas offers a genetic counselling service to support the specialist with the assessment of results or to provide counselling directly to the patient. The main goal is to establish whether additional testing on the parents is necessary based on the results, to try preventing recurrence in future pregnancies.

Bibliography

1. Colley E, et al. Cell-Free DNA in the Investigation of Miscarriage. J Clin Med. 2020;9(11):3428.
2. Yaron Y, et al. Maternal plasma genome-wide cell-free DNA can detect fetal aneuploidy in early and recurrent pregnancy loss and can be used to direct further workup. Hum Reprod. 2020;35(5):1222-1229.
3. El Hachem H, et al. Recurrent pregnancy loss: current perspectives. Int J Womens Health. 2017;9:331-345.

my Newborn

What is myNewborn?

The test allows knowing the newborn risk to develop around **390 diseases with onset in the first years of life.** The test complements the heel prick test, improving the clinical utility.

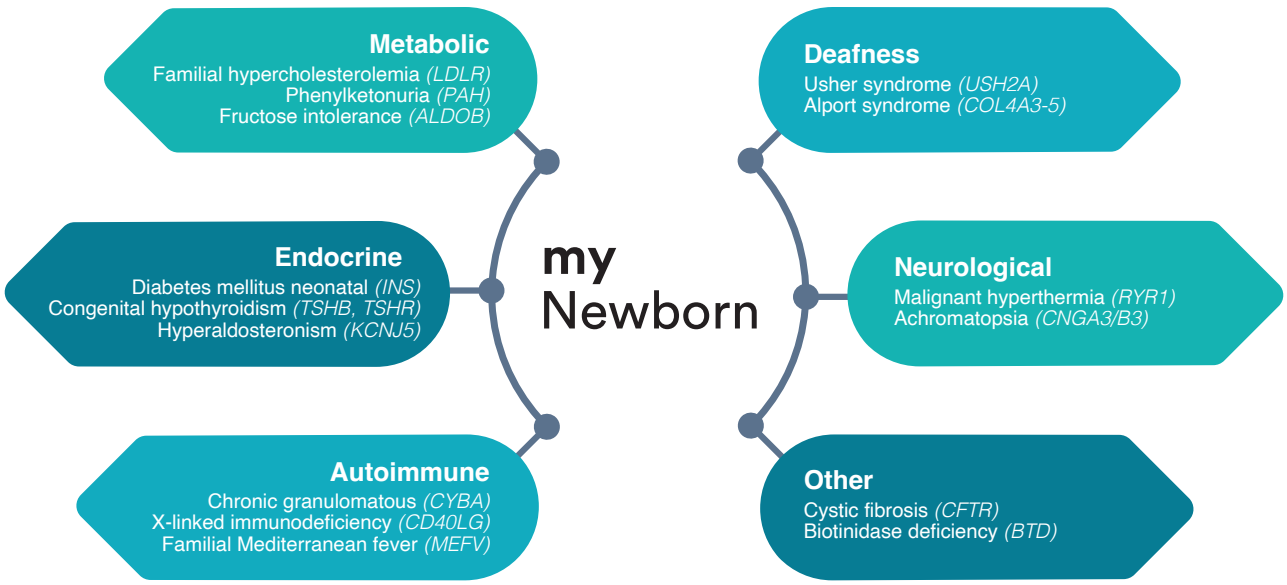
myNewborn entails an expansion of the conventional neonatal biochemical screening test.

What does the test include?

The test analyzes more than **400 genes through Whole Exome Sequencing (WES).**

What type of diseases are included?

- Highly actionable childhood onset diseases.
- Included in the newborn heel prick test.
- Adult onset diseases that are actionable during childhood.
- Common although not treatable diseases, with carrier frequency higher than 1/100.

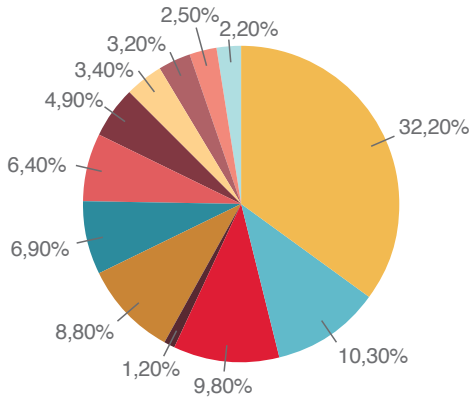


myNewborn clinical application examples

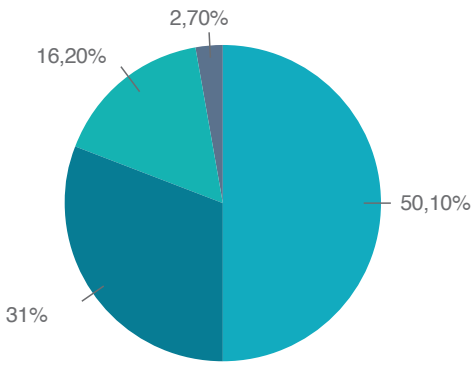
PATHOLOGY/GENE	SYMPTOMATOLOGY	PATIENT MANAGEMENT	
Glucose-6-Phosphate Dehydrogenase Deficiency *(G6PD)*	*Hemolytic anemia when beans, sulfites, specific drugs or food colorings are consumed.*	Dietary modifications and avoid specific drugs	Child without symptomatology
Familial hypercholesterolemia *(LDLR)*	*Early cardiovascular (CV) disease.*	Dietary modification + medication	Prevention of CV disease
Abetalipoproteinemia *(MTTP)*	*Fat absorption inability that alters the nervous system development.*	Low-fat diet + vitamin supplements	Prevention of neuropathy and ataxia

Types of diseases included in myNewborn

Percentage of diseases included in **myNewborn** by category



Classification of the **myNewborn** pathologies by clinical



Metabolic
Syndromic
Pulmonary
Musculoskeletal
Hematologic
Endocrine
Neurological
Cardiovascular
Deafness
Immunological
Cancer
Urogenital
Preventive management
Early management
Dietary management
Other type of intervention

Our perinatal medicine portfolio is part of a broad portfolio of genetic services

PREVENTIVE MEDICINE

myGenome – Whole Genome Sequencing based comprehensive genetic risk screening analysis designed for healthy individuals.

myGeneticRisk – Combined hereditary risk screening analysis of cardiovascular disease and cancer.

myHealthScore – Multi - Polygenic risk score assessment that provides information regarding patient risk for common multifactorial diseases.

DIAGNOSTIC SERVICES

CardioRisk – Hereditary risk screening analysis of inherited cardiovascular disease.

CancerRisk - Hereditary risk screening analysis of multiple types of cancer.

GenomeDx - Whole genome sequencing for genetic diagnosis in complex patients.

ExomeDx - Whole exome sequencing for genetic diagnosis in complex patients.

CustomizedPanels - Exome-based panels, tailored to the clinical case under study.

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