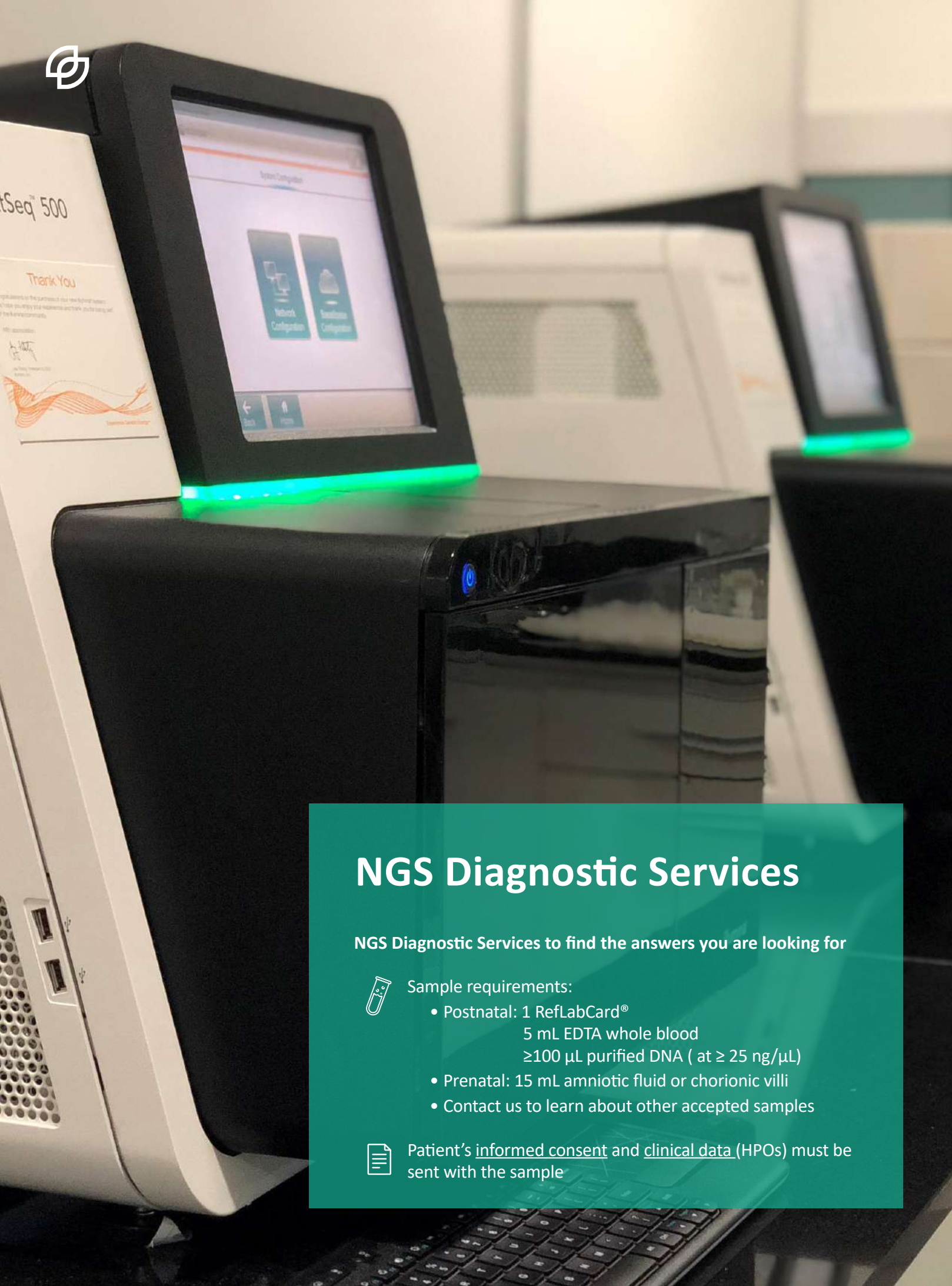
REFERENCE LABORATORY
GENETICS

NGS Diagnostic Services

Find the answers you are looking for
with our NGS Diagnostic services



NGS Diagnostic Services

NGS Diagnostic Services to find the answers you are looking for

Sample requirements:

- Postnatal: 1 RefLabCard®
  5 mL EDTA whole blood
  ≥100 µL purified DNA (at ≥ 25 ng/µL)
- Prenatal: 15 mL amniotic fluid or chorionic villi
- Contact us to learn about other accepted samples

Patient's informed consent and clinical data (HPOs) must be sent with the sample

We offer the most advanced NGS services

Certain patients present a combination of symptoms characteristic of syndromes with several associated genes. In these cases, the study of a single gene is usually an inefficient strategy, resulting in staggered, lengthy and costly diagnostic processes that can end up delaying diagnosis and negatively affecting their quality of life.

At **Reference Laboratory Genetics** we offer the most advanced NGS sequencing services and bioinformatics tools in order to sequence thousands of genes simultaneously and to perform the clinical interpretation of the sequencing data in a reliable, fast and cost-effective way.

RefLab Genome® (WGS)

Despite containing most of the known pathogenic variants, the coding regions of the genes (exons) represent only ~1% of the genome. Recent clinical studies show the cause of certain alterations in non-coding regions, demonstrating the importance of analysing such areas**. RefLab Genome® sequences the coding and non-coding regions of the genome, detecting in a single genetic test, almost any change in a patient's DNA.**

RefLab Exome® (WES)

85% of the known pathogenic variants are found in the coding regions of the genes, the exons. **Focusing on these regions where the disease causing genetic alterations are most likely to be found, RefLab Exome® sequences the whole exome and interprets the sequencing data from a clinical point of view.**

RefLab MedExome® (CES)

RefLab MedExome® is an optimized clinical exome that analyses >5.500 clinically relevant genes with known clinical phenotypes. Our clinical exome allows health professionals to focus on clinically relevant genes avoiding inconclusive results of sequencing genomic regions not related to human diseases. This ensures the highest clinical utility with the shortest turnaround times.

RefLab Global NGS Panels

Our Global NGS Panels are an excellent option for dealing with cases where syndromes with multiple associated genes are suspected as they evaluate multiple genes simultaneously, increasing the diagnostic yield and reducing costs and response times.

RefLab NGS Panels

At Reference Laboratory Genetics, we have developed > 430 NGS panels, covering all medical specialities. Our aim is to provide healthcare professionals with the necessary tools in order to diagnose their patients accurately, and to help them make informed decisions.

Trio analysis available

Extension analysis available

RefLab guarantee

- **Continuous gene update and analytical validation** of all our panels and bioinformatics tools
- **100% customizable panels**
- **Low turnaround times** (<30 days)
- Pre and post analysis **support** provided by our experts in **clinical genetics**
- **Comprehensive, conclusive, transparent and customizable medical reports**
- Highest diagnostic yield and **clinical utility**
- **CNVs analysis.** All detected variants are **confirmed by MLPA** at no additional cost.

Choosing the right diagnostic test

RefLab Genome® (WGS)	• Complex and heterogeneous disorders with unclear or atypical phenotypes • There is no specific genetic test to diagnose the suspected genetic disease • There are inconclusive prior genetic tests
RefLab Exome® (WES)	• Genetically heterogeneous disorders (intellectual disability, metabolopathies, ata-xias, neuropathies...) • There is no specific genetic test to diagnose the suspected genetic disease • There are inconclusive prior genetic tests
RefLab MedExome® (CES)	• Complex phenotypes with multiple associated genes • There is no specific genetic test to diagnose the suspected genetic disease • There are inconclusive prior genetic tests
RefLab Global NGS Panels	• Syndromes with a large number of associated genes are suspected
RefLab NGS Panels	• Syndromes with multiple associated genes are suspected

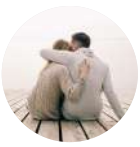
Comprehensive diagnostic testing for all medical specialities

RefLab Exome Intellectual Disability®
Whole exome with increased coverage in genes associated to intellectual disability.
Code: 25954

RefLab Exome Metabolopathies®
Whole exome with increased coverage in genes associated to metabolopathies.
Code: 25954

NeuroRef Global®
Analysis of 1204 genes associated with neurological diseases.
Code: 55620

NeuroRef Epilepsy Plus®
Analysis of 118 genes associated with epilepsy.
Code: 25065



OncoRef Global®
Analysis of 161 genes associated with hereditary cancer.
Code: 57650

CardioRef Global®
Analysis of 192 genes associated with cardiovascular diseases.
Code: 55270

NephroRef Global®
Analysis of 427 genes associated with kidney diseases.
Code: 55580

OphthalmoRef Global®
Analysis of 527 genes associated with ophthalmologic diseases.
Code: 57450

OtoRef Global®
Analysis of 193 genes associated with hearing loss.
Code: 58175

DermaRef Global®
Analysis of 268 genes associated with skin diseases.
Code: 17035

MitoRef®
Analysis of 378 genes associated with mitochondrial diseases.
Code: 54975

CarrierRef®
CarrierRef® helps future parents understand their risks to transmit a recessive genetic disease or X-linked disease to their offspring.
Code: 15063

We add value to genetic diagnosis

For over 40 years the team at **Reference Laboratory Genetics** have been dedicated to offering our clients the most complete catalogue of molecular genetic tests to meet each patient's individual needs.

- Hereditary genetic diseases
- Liquid biopsy and solid tumour analyses
- Oncohematology
- Pharmacogenetics

We offer a comprehensive service for any medical speciality, from the sample to the medical report, providing support at any point in the diagnostic process: collection and transport of samples, genetic counselling, bioinformatics tools and clinical interpretation.

When our clients ask us for a genetic analysis, they get more than just results, they help us increase the collective knowledge of hereditary diseases (all mutations we find are reported to our RefLab Database® and the main public databases).

We strive to provide you with the highest quality

We are UNE-EN ISO 15189 accredited and ISO 9001 and 14001 certified by AENOR. Moreover, we participate in the most extensive external international quality assessment programmes.

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