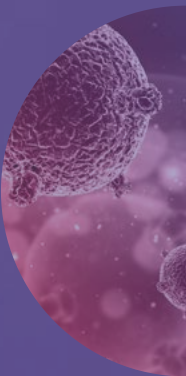


# COMPREHENSIVE SOLUTIONS IN **GENETICS**



**360°**

 **eurofins**

Specialties  
Clinical Diagnostics

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## Who are we?

**Eurofins Specialties Clinical Diagnostics** is part of **Eurofins Scientific**, present in 65 countries and a world leader in clinical analysis, genomics, food, environment and pharmaceuticals.

**Eurofins Specialties Clinical Diagnostics** with more than 30 years of experience, is currently the largest laboratory in Spain, leading the management of intra-hospital laboratories in the country, with a presence in 67 hospitals providing 24-hour service 365 days a year.

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## Our value proposition

### 360° SERVICE

We offer services to all medical specialties, responding to all the needs that arise in daily clinical practice.

### FULL INTEGRATION

Expert support both pre-test and post-test, with reports with clinical interpretation and unified web-based platform.

### ALL AVAILABLE TECHNIQUES

NGS (gene panels, exome, genome), MLPA, FISH, qPCR, ddPCR, arrays, liquid biopsy and more.

### MULTIDISCIPLINARY TEAM

>200 experts (doctors, biologists, bioinformaticians) at the service of your site.

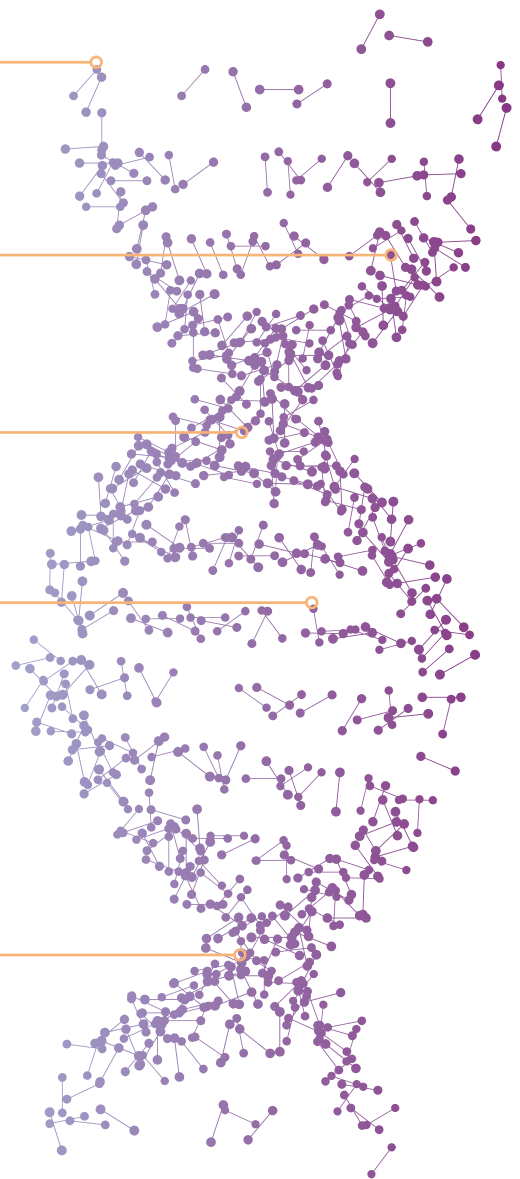
Our team of experts is at the service of the clinician for any consultation needed in the interpretation of the result, obtaining the most efficient results of applied genomics.

Genetic counselling with AEGH accredited personnel.

### ACCREDITED QUALITY

CE-IVD marking, ISO 9001 certification, ISO 15189 accreditation and participation in international quality control programmes.

**Quality, our raison d'être.**



# Integral Solutions

## 3.1 MEDICAL SPECIALITIES

The clinical genetics unit deals with all medical specialties, providing diagnostic solutions for each pathology.

### CARDIOLOGY

- Cardiomyopathies: Hypertrophic, dilated
- Long QT
- Short QT
- Brugada Syndrome
- Arrhythmogenic syndromes and channelopathies
- Diseases of the aorta and other vessels

### ENDOCRINOLOGY

- Diabetes: MODY, Mellitus...
- Low stature
- Tumours
- Hormone deficits
- Obesity
- Hypogonadism
- Rickets
- Overgrowth

### INTERNAL MEDICINE

- Immunodeficiencies
- Hyperthermia
- Autoimmune diseases
- Rare diseases
- Familial hypercholesterolemia

### NEUROLOGY

- Ataxias
- Paraplegia
- Dementias (Alzheimer's)
- Neuropathies (Charcot-Marie-Tooth)
- Neurodegenerative diseases
- Movement disorders (Parkinson's disease)
- Epilepsies

### NEUROPAEDIATRICS

- Developmental delay
- Autism
- Intellectual disability
- Fragile X Syndrome
- Duchenne Dystrophy
- Williams syndrome
- Prader Willi/Angelman Syndrome
- Epilepsies

### OPHTHALMOLOGY

- Retinitis pigmentosa
- Alport syndrome
- Stargard disease
- Usher syndrome
- Macular dystrophy
- Oculocutaneous albinism
- Retinal dystrophy

### GYNAECOLOGY & OBSTETRICS

- NIPT
- Prenatal diagnosis
- Skeletal dysplasia panel
- Noonan Syndrome
- Di George Syndrome
- Karyotype
- Array CGH

### REPRODUCTIVE MEDICINE

- Carrier test
- DGP
- Embryo biopsy
- FISH and sperm fragmentation

### ONCOLOGY

- Oncohematology
- Therapeutic targets
- Minimal residual disease
- Liquid biopsy

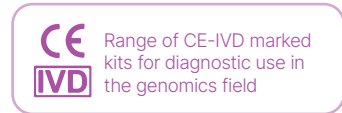


### 3.2 SERVICES

**In order to provide a 360° service** in genetics, Eurofins offers a set of support areas to cover all the needs that may arise in daily clinical practice and laboratory workflows, always following the highest quality standards.

## DIGITAL PLATFORMS & TECHNOLOGY TRANSFER

**Eurofins** is aware of the different needs that genetics laboratories have, therefore, in addition to offering our end-to-end services by delivering a final report to the applicant, we offer the following services:



#### GeneSGKits

CE-IVD kits for NGS library preparation and on-site processing in hospitals or laboratories. The laboratory will have a variety of products to address different diagnostic approaches in the field of genetics.

#### GeneSystems

Cloud-based CE-IVD platform for NGS data analysis (panels, exome and genome). It allows the laboratory to obtain relevant information for diagnosis or research, covering the entire data analysis process in a simple and rigorous way.

#### Sequencing service

In those cases where the laboratory does not have sequencing capacity, Eurofins makes this resource available to the laboratory, sending the raw data to the laboratory for local analysis with our GeneSystems software.

#### Drylab services

Aimed at those laboratories that have sequencing capacity, but do not have the resources to analyse the data. Eurofins has a team of experts who analyse the data and provide personalised reports with clinical interpretation.

## MEDICAL GENETICS & GENETIC COUNSELLING

We have a team of medical geneticists who provide support both in the clinical integration of cases and by offering medical genetics consultations for those patients who require it. On the other hand, we offer both pre-test and post-test support from our genetic counselling team to solve any questions you may have.

## R+D+i UNIT

Eurofins is committed to Research, Development and Innovation, supporting translational medicine, in order to incorporate the latest innovations resulting from the results of national and international research projects in which it participates into the service of our clients.

#### Applied research:

- Development of new panels based on emerging publications and clinical guidelines.
- Continuous internal validation: meta-analysis of performance, reproducibility and sensitivity.
- Strategic collaborations with universities, hospitals and international consortia.



## PHARMACOGENOMICS

Eurofins aims to offer personalised and precise medicine through its wide range of diagnostic tests, its therapeutic targets unit and its extensive pharmacogenetics service.

We offer tailor-made services with different technologies adapted to each drug, using MASSArray, NGS and Sanger Hot-spot.

### Available solutions:

- **Antineoplastics**
- **Cardiology:** statins, lipid-lowering, antiplatelet and antiplatelet drugs
- **Gastroenterology:** Proton pump inhibitors
- **Treatment of infections:** antivirals, antifungals, interferons.
- Immunosuppressants
- **Anti-inflammatory drugs:** NSAIDs
- 75 drug Response Panel
- Panel on psychotropic drugs
- Chronic Pain Drug Response Panel
- 25-drug response panel for cancer therapy drugs
- **Others (on request):**
  - Pharmacogenetic study (with interpretation)
  - Pharmacogenetic GENOTYPE (without interpretation)
  - **GLOBAL PGx GENE PANEL:** 161 active ingredients associated with more than 300 medicines

## MICROBIOME

Microbiome studies offer several advantages for understanding gut and general health. They can identify imbalances in the gut microbiota, detect gastrointestinal pathogens, assess intestinal permeability and, in some cases, even study the endometrial microbiome.

Our shotgun metagenomic sequencing technology allows the complete study of the genome of all microorganisms that make up the vaginal and intestinal ecosystem. This avoids the amplification bias inherent in conventional 16S ribosomal gene studies, which often only focus on a small portion (less than 20%) of the 16S rRNA gene.

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## Techniques

We have all the techniques available in the field of genetics to respond to all the clinical and therapeutic needs of our clients following the highest quality standards:

- NGS
- CNVs studies
- FISH
- Array Genotyping
- Sanger Sequencing
- Methylation studies
- Array CGH
- MLPA
- Specific variants
- Exomes
- Expansions
- Genome
- NGS Panels
- Karyotype

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## Benefits for your organisation



### SPEED AND FLEXIBILITY

Response times from 10 days (germ cell panels) to 12 days (CGP)



### TURNKEY SOLUTION

From sample collection to delivery of results and genetic counselling



### SCALABILITY AND CUSTOMISATION

Adapt services and panels to your centre's portfolio



### 24/7 SUPPORT

Coverage in hospitals and laboratories, with multi-channel customer service



### LEADING TECHNOLOGIES

NovaSeq 6000, NextSeq, Ion S5, MinION



### RIGOROUS QUALITY CONTROL

Coverage, uniformity and reading depth metrics



### COMPREHENSIVE LOGISTICS

Sample collection, refrigerated shipments and tracking

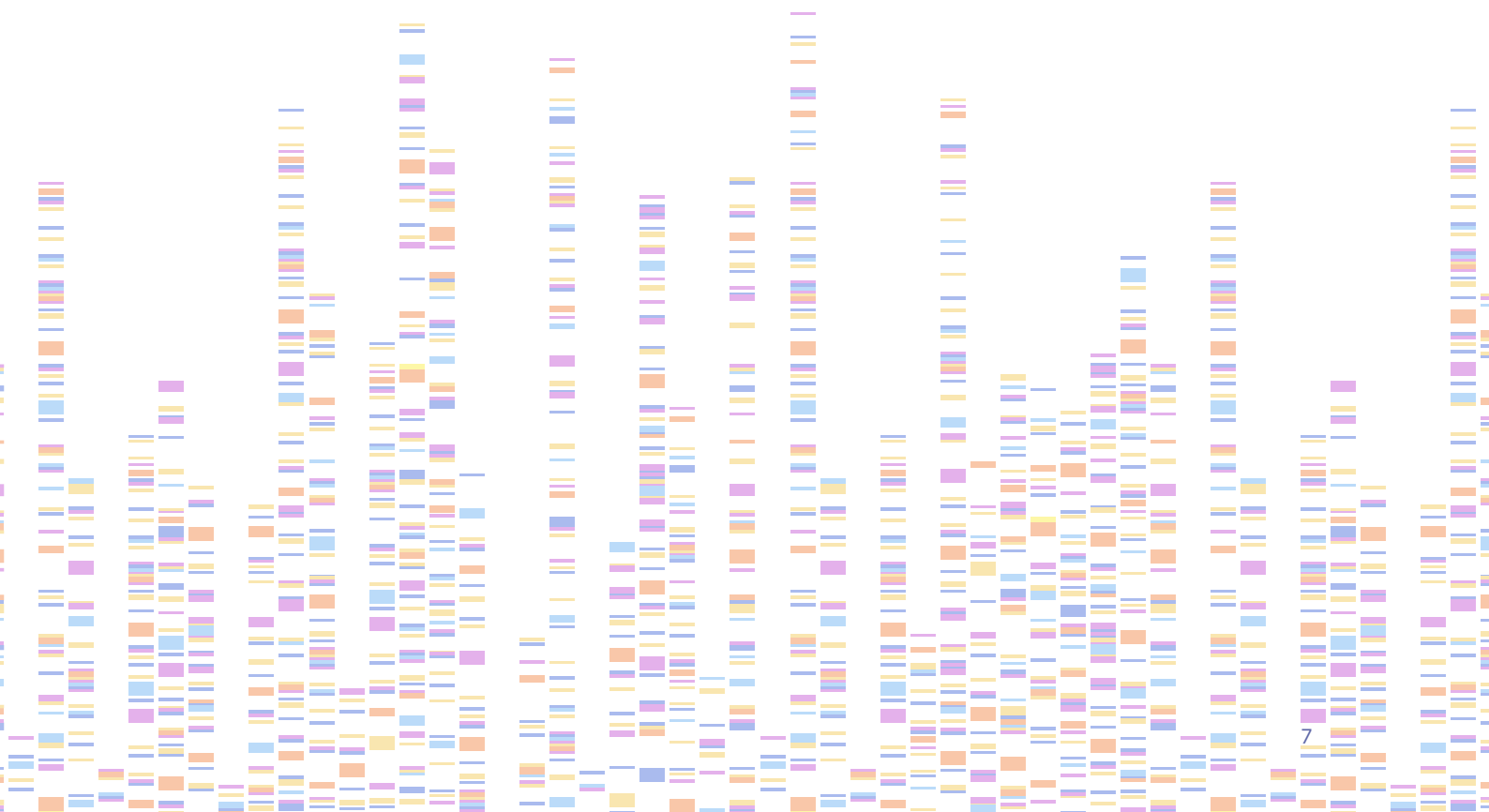
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## Quality

### ISO 15189

Technical competence in the following studies:

- NIPT
- Exome - NGS Panels
- Preimplantation Genetic Testing PGT
- Embryo biopsy
- Sanger Sequencing
- Fragment analysis
- MLPA
- Karyotypes
- FISH





Specialties  
Clinical Diagnostics

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