



GENERALITAT
VALENCIANA



BANCO DE
PATENTES



PRINCIPE FELIPE
CENTRO DE INVESTIGACION

Genomics and Translational Genetics

DESCRIPTION OF THE TECHNOLOGY

The Genomics and Translational Genetics Service (SGGT) is a biotechnological service which integrates the scientific capacity of the CIPF research groups in human genetics, genomics and bioinformatics with the knowledge and experience in genetic diagnosis of a team of CIPF geneticists and physicians.

The SGGT takes a step beyond the genomic analysis that CIPF has been providing since 2005, offering integrated genetic testing and analysis of the human genome moving forward to translational medicine and its application in the clinical praxis.

The SGGT aims to:

- To provide technical and clinical support to health professionals interested in genetic diagnosis, addressing the needs that arise in daily clinical practice. To do this, we have a long history in the genetic analysis of hereditary diseases and, consequently, the experience to resolve the issues that may be rise in this regard.
- To innovate in genetic testing by development of

new tools based on the technology of ultrasequencing (NGS, 'Next Generation Sequencing') in order to accelerate genetic analysis and molecular diagnostics of disorders with a wide genetic heterogeneity.

- To provide researchers with the technical, material and human resources in the field of DNA Sanger sequencing, genomic analysis using microarray (aCGH-Comparative Genomic Hybridization-, Single-Nucleotide Polymorphisms -aSNP- and Copy Number Variations -CNV-) and other related services such as the extraction, purification and concentration and quality control of DNA and RNA samples.
- To investigate the genetic causes and mechanisms underlying inherited disorders and deepen the knowledge currently exists about these diseases, offering collaborations with research groups within the Program in Rare and Genetic Diseases, always with the aim of get new biomarkers and new therapeutic targets.

MARKET APPLICATION SECTORS

Companies, hospitals and medical doctors and researchers interested in genetic diagnosis and mechanisms underlying inherited disorders.

TECHNICAL ADVANTAGES AND BUSINESS BENEFITS

We provide support services to clinical and health care, by specializing in genetic testing of single gene diseases, birth defects, chromosomal and genomic syndromes, intellectual disability and other diseases with genetic component, in order to provide diagnosis and genetic counseling, improving health care for patients and their families. More information available at <http://www.cipf.es/en/web/portada/genomica-y-genetica-traslacional>.

CURRENT STATE OF DEVELOPMENT

SGGT available equipment includes Telstar Mini-V/PCR, NanoDrop ND1000, Veriti 96 wells thermal cycler from Applied Biosystems, 96 capillarABI3730xl sequencer from Applied Biosystems, 2100Bioanalyzer from



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Agilent Technologies, G2565C microarray scanner from Agilent Technologies, TecanHS 4800 Pro Hybridization Station and LC480 Roche for 96 and 284 samples.

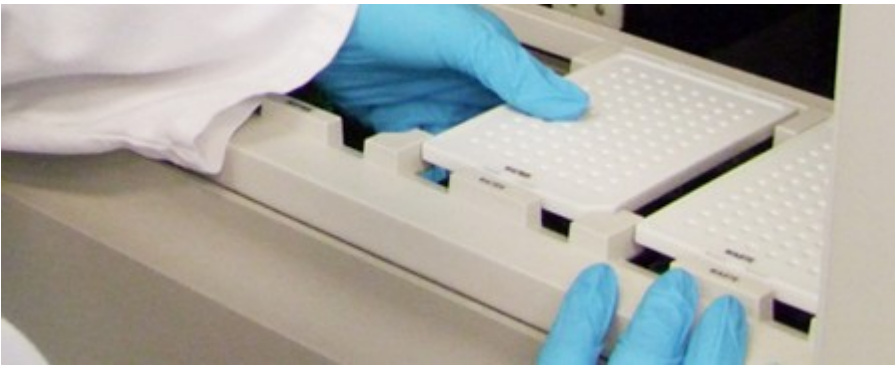
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COLABORATION SOUGHT

Collaboration with companies, hospitals and medical doctors and researchers interested in genetic diagnosis and mechanisms underlying inherited disorders.

RELATED IMAGES



CONTACT

Office of Sponsored Research and Technology Transfer
otri@cipf.es

Principe Felipe Research Center
Eduardo Primo Yufera, 3
46012 - Valencia



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