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# **BIOMARKERS:**

**GENOMICS FOR PRECISION MEDICINE**

# INCREASE SUCCESS, IMPROVE INSIGHTS, INCREASE VALUE



**T**he characterisation of genomic variation is of increasing diagnostic value in many diseases like cancer, neurological diseases, and other pathologies. Specific genomic variants predict drug response or resistance (genomics-driven medicine). Furthermore, changes in gene expression and modification of DNA and RNA can provide valuable insight into biological processes, thus supporting strategic development decisions such as: patient stratification; drug target identification; evaluation of drug competitiveness; and the treatment effect size. The EATRIS Genomics Centres provide all of the tools of modern functional genomics and access to the international research and clinical communities. EATRIS institutions are equipped with state-of-the-art technologies for Next Generation Sequencing and genomics, expression analysis, microarrays, qPCR, and more. The EATRIS platform has available a panoply of analytical methods of genome and gene expression studies that have been adapted to biomarker assessment in tissue and bodily fluids. EATRIS supports the biomedical research, healthcare, and pharmaceutical industries in developing new competitive products (e.g. validation of new diagnostic kits).

## **A selection of EATRIS institutes**

Instituto Ramón y Cajal (IRYCIS), [Madrid, Spain](#)

IRCCS Fondazione Pascale, [Napoli, Italy](#)

Institute for Molecular Medicine Finland (FIMM), [Helsinki, Finland](#)

The Medical Research Institute of the Hospital La Fe (IIS-La Fe), [Valencia, Spain](#)

Fundacion Jimenez Diaz Institute for Medical Research (IIS-FJD), [Madrid, Spain](#)

Fondazione IRCCS Istituto Nazionale dei Tumori, [Milan, Italy](#)

Central European Institute of Technologies (CEITEC), [Brno, Czech Republic](#)

Hospital La Paz Institute for Health Research (IdiPAZ), [Madrid, Spain](#)

Centro di Riferimento Oncologico di Aviano (CRO Aviano), [Aviano, Italy](#)

August Pi i Sunyer Biomedical Research institute (IDIBAPS), [Barcelona, Spain](#)

Institute of Molecular and Translational Medicine (IMTM), [Olomouc, Czech Republic](#)

Instituto de Investigación Sanitaria - INCLIVA, [Valencia, Spain](#)

BioDonostia Health Research Institute, [San Sebastian, Spain](#)

SNP&SEQ Technology platform, National Genomics Infrastructure/Science for Life Laboratory at Uppsala University, [Uppsala, Sweden](#)

## Access leading academic expertise

- DNA/RNA sequence analysis for diagnostic purposes;
- Patient stratification by DNA/RNA sequence analysis;
- Patient stratification by transcript/ncRNA/miRNA assessment;
- Pan-European population analyses;
- Transcriptomics;
- Identification and validation of candidate biomarkers (i.e. miRNA) for disease diagnostics, monitoring of heterogeneity, and progression;
- Identification and validation of markers of drug resistance/sensitivity/toxicity/efficacy; and
- Epigenetics testing for chemotherapy resistance.

## How does EATRIS add value to your portfolio?

- Single point of access - from marker identification, collection to analytical testing, assay development; and validation;
- Expert advice at the highest scientific level for the development of biomarkers and clinical expertise – cooperative design of development plan;
- Consulting experts for the optimal use of available biosamples;
- Fast access to clinical samples and well annotated clinical data;
- Access to multisite clinical trials;
- Access multiple sites for clinical validation of your diagnostic (panel);
- Biomarker identification, validation, ready for qualification;
- Data stewardship;
- Reference diagnostics; and
- Inter-laboratory comparisons

## Key technologies

### ACCESS TO HIGH-QUALITY SEQUENCING AND PROFILING TECHNOLOGIES

- Expression Analysis (transcripts, mRNA, microRNA, ncRNA)
- Next Generation Sequencing and Mapping
- Microarray
- PCR, Heat Pulse Extension – PCR, droplet PCR, high-throughput automated qPCR
- Sequence Analysis
- Sequencing, Pyrosequencing Next Generation Sequencing
- Real Time PCR, droplet digital PCR (ddPCR), high-throughput automated qPCR genotyping
- HRM followed by sanger sequencing
- Epigenetics
- Bisulfite sequencing
- Methylation Specific PCR (MSP)
- Telomere length determination

### BIOINFORMATICS

- Alignment, assembly and polymorphism detection
- Detection of structural variants
- Gene expression quantification
- Detrimental effect analysis

## Regulatory/QA aspects of the services and infrastructure

- Access to biological samples adhering to current regulatory and QA requirements;
- Compliance with regulatory and industry standards;
- Standardised protocols by core technical labs;
- Clinical and technological excellence;
- Implementation of new emerging technologies and methodologies;
- Involvement of medical laboratories with ISO 15189 quality management systems; and
- Regulatory expertise for in vitro medical devices CE marking procedures (CE IVD).