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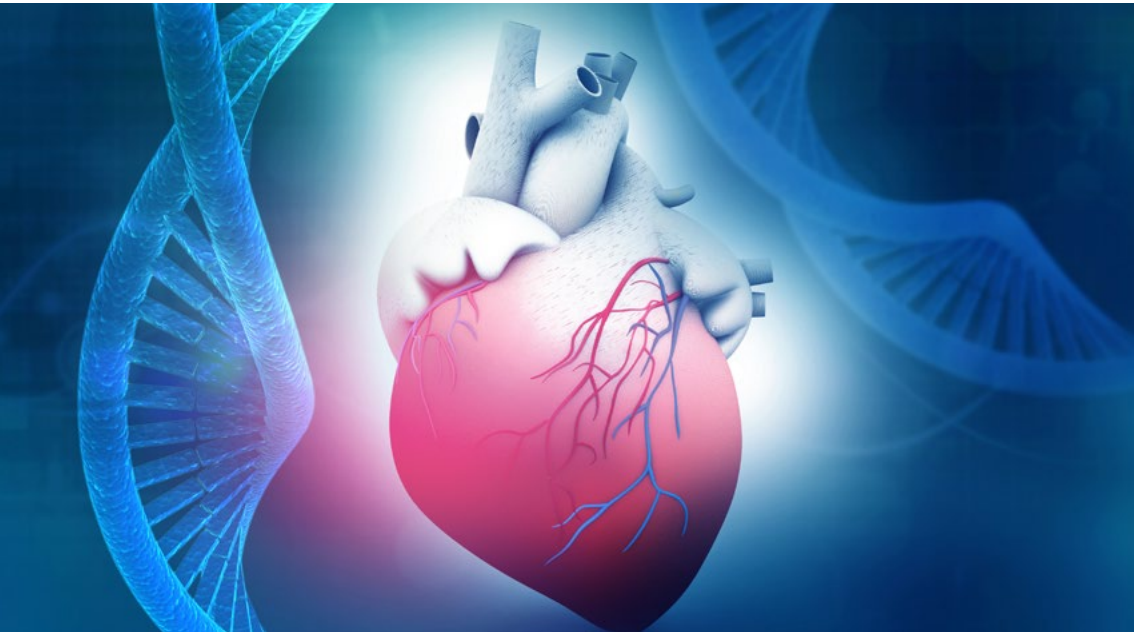

Cardiomyopathies and arrhythmias

The contribution of cardiogenetics in the management and care of patients and their families

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In addition to the well-known cardiovascular risk factors (smoking, high blood pressure, etc.), some cardiac disorders have a genetic origin. These pathologies have essentially an autosomal dominant inheritance. However, for some of them, an X-linked or an autosomal recessive inheritance could also sometimes be observed.

Molecular diagnosis of inherited cardiac diseases could be crucial to allow an appropriate therapeutic strategy, or even personalised care in certain cases, for each patient and their family.

In recent years, molecular approaches were significantly improved, particularly with the development of Next Generation Sequencing (NGS) strategies, which has major applications in cardiogenetics.

This technology can be used to simultaneously sequence a panel of targeted genes involved in a pathology. The result is a selective analysis that facilitates a faster identification of patients with one or more genetic variations.

This panel approach is ideal in view of the clinical, genetic and allelic heterogeneity of inherited cardiac diseases and allows an increase of molecular diagnosis performance.

Impact of genetic screening

In cardiomyopathies and arrhythmias, genetic testing have multiple impacts:

Diagnostic impact

Genetic screening based on a panel of genes improves the diagnostic rate by supporting the analysis of large genes and provides a more exhaustive diagnosis when several gene variants are present in the same patient. The identification of the causal mutation(s) sometimes makes it possible to implement appropriate medical care, significantly reducing the risk of complications for the patient.

Impact on genetic counselling

After identification of a pathogenic or likely pathogenic variation, genetic counselling can be offered to the different relatives within the family.

Impact on prenatal and pre-implantation diagnosis

Identification of pathogenic or likely pathogenic variations can give couples planning a pregnancy information about the risks of disease transmission and in the most severe cases to possibly consider recourse to prenatal diagnosis (PND), a preimplantation diagnosis (PID) or gamete donation.

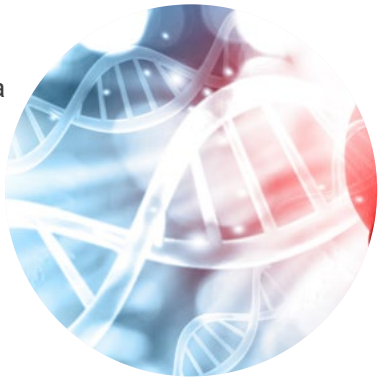
Cardiogenetics by Eurofins Biomnis

Genetic screening requires clinical, genetic as well as scientific and bioinformatics skills.

Building on its expertise and its organisational structures in this area, Eurofins Biomnis offers, after exome sequencing, a bioinformatics analysis which targets a gene panel relating to the patient's pathology. The panels include genes whose variants have been reported to cause the condition in question. These panels are regularly updated according to the progression of medical knowledge and international recommendations for hereditary or rare heart conditions.

Our targeted panels by pathology[1]

- Hereditary Hypertrophic Cardiomyopathy
- Hereditary Dilated Cardiomyopathy
- Hereditary Restrictive Cardiomyopathy
- Non-Compaction of Left Ventricle
- Arrhythmogenic Right Ventricular Dysplasia
- Congenital Long QT Syndrome
- Brugada Syndrome
- Short QT Syndrome
- Jervell Lange-Nielsen Syndrome
- Andersen-Tawil Syndrome
- Timothy Syndrome
- Catecholaminergic Polymorphic Ventricular Tachycardia
- Sudden Cardiac Death
- Cardiac Conduction Disorder



Technique

All coding regions of the genes within the panel are analysed by Next Generation Sequencing on an Illumina Novaseq sequencer.

Sequencing data are further analysed and interpreted via the SeqOne bioinformatics solution.

Performance

- Whole exome coverage: ~22,000 genes and 37.5 Megabases targeted
- Depth > 30X for ~ 98% bases[2]
- > 99% of recall[3]

Why Eurofins Biomnis?

- Specialised clinical diagnostics laboratory
- ISO 15189 accreditation
- Authorisation to perform congenital genetic tests
- Certified clinical pathologists
- Expertise in sequencing techniques for diagnostic practice

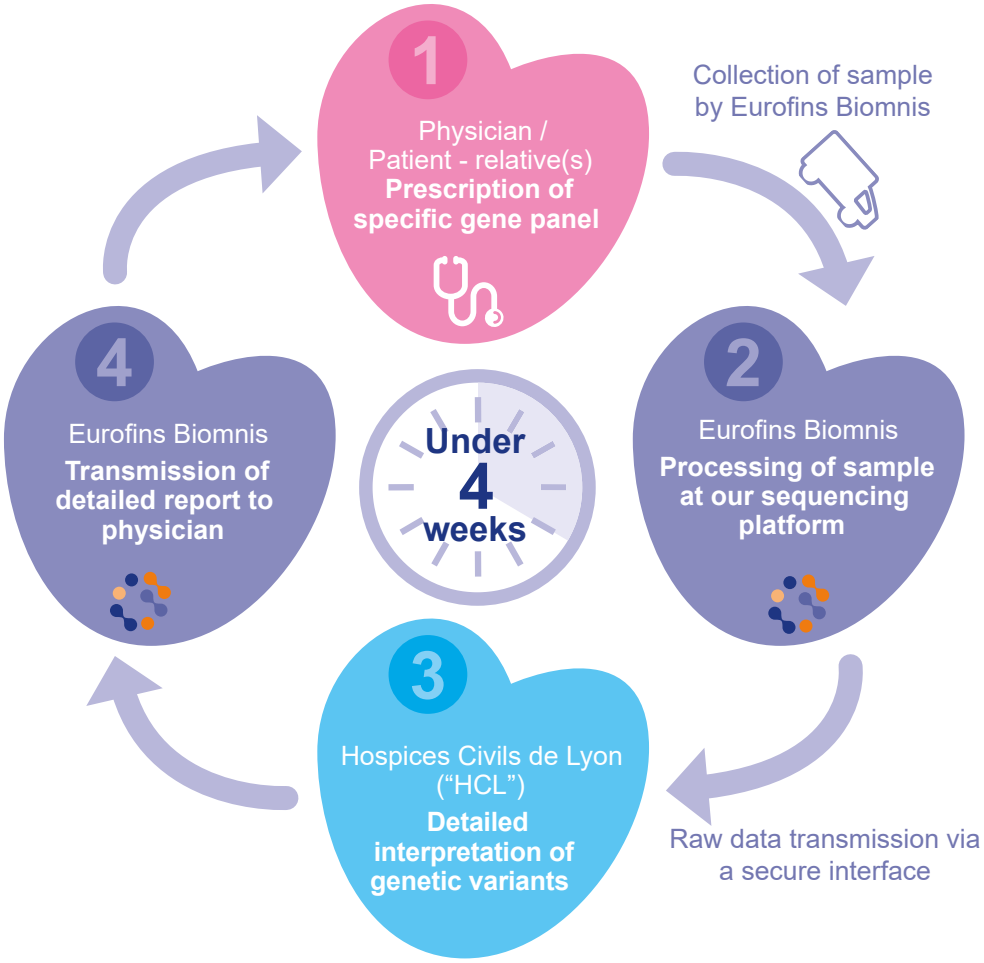
Collaboration with experts for optimal diagnostic results

One of the challenges of exome sequencing is the interpretation of variants.

The accuracy of interpretation and ultimately of the diagnosis is intrinsically linked to the clinical-biological collaboration combining quality of sequencing on the one hand and expertise in the respective pathology on the other.

In order to offer physicians an ever more reliable and precise tool for their therapeutic decisions, **Eurofins Biomnis collaborates with *Hospices Civils de Lyon* ("HCL")**, a teaching hospital group recognised for its expertise in cardiogenetics, in the interpretation of variants.

Perform your targeted gene panels with Eurofins Biomnis and HCL



References

- *Janin, A., Januel, L., Cazeneuve, C. et al. Molecular Diagnosis of Inherited Cardiac Diseases in the Era of Next-Generation Sequencing: A Single Center's Experience Over 5 Years. Mol Diagn Ther (2021). https://doi.org/10.1007/s40291-021-00530-w*
- *Ingles J, et al. Genetic Testing in Inherited Heart Diseases. Heart, Lung and Circulation (2019), https://doi.org/10.1016/j.hlc.2019.10.014*

1. *list of genes on request*
2. *target CDS Refseq +/- 2 base pairs*
3. *Data calculated from SNV's from NIST002 reference samples, for 40 million pairs of reads generated*

Practical details

Test	Cardiomyopathies and hereditary cardiac arrhythmias Targeted gene panels
Analysis code	**EXOME**
Turnaround time	**4 weeks** excluding any additional examinations
Indications	▸ Diagnosis of hereditary cardiomyopathies and arrhythmias ▸ Genetic counselling
Sample	5 mL EDTA whole blood or DNA sample
Storage and transport	Room temperature
Technique	Exome sequencing
Price	Contact us
Required documents	**B61-INTGB** test request form available on **www.euro ins-biomnis.ie** > Test guide > Test code **EXOME**
Supplementary tests	▸ Study of relatives by Sanger sequencing: 5 mL EDTA whole blood or DNA sample from relative ▸ Congenital heart defects: please use the test request form **B34-INTGB** available on **www.eurofins-biomnis.com** > Test guide > Test code **EXOME**

For more information

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