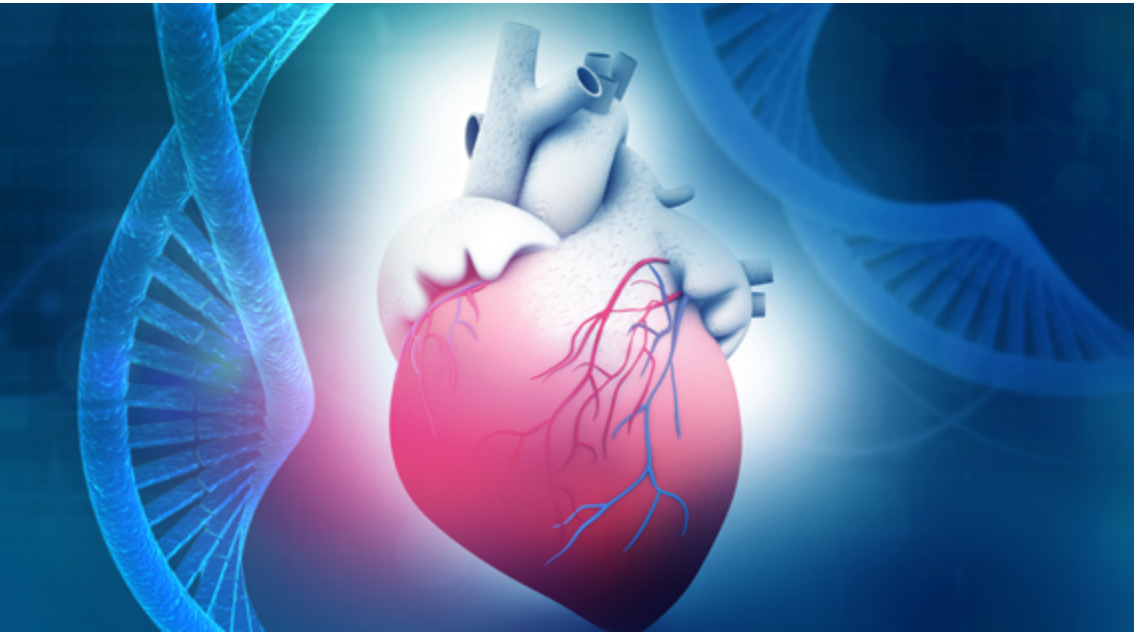
eurofins | Biomnis



Cardiomyopathies and arrhythmias

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

Some heart conditions are genetic and can be passed on in different ways (dominant, recessive or X-linked), sometimes affecting several members of the same family. Identifying the faulty gene(s) involved can help to adapt treatment and even personalise it.

The development of next-generation sequencing (NGS) has revolutionised cardiogenetics. This technology enables the simultaneous analysis of targeted gene panels, adapted to the heterogeneity of heart diseases.

By identifying patients carrying one or more genetic variations more easily and quickly, the panel approach significantly improves diagnostic yield.

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 variation(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, a preimplantation diagnosis or gamete donation.

Pharmacogenetic impact

Therapeutic choices in cardiology can be complex, and cardiologists rely on pharmacogenetic information.

Certain drugs used in cardiology (mavacamten, antiarrhythmic antithrombotics, beta-blockers, statins, etc.) are metabolised by enzymes which activity depends on the presence of genetic variations. Sequencing makes it possible to detect these variations and predict the efficacy and adverse effects of the drugs concerned. It is then possible to determine the dosage and/or molecule to which the patient will respond best.

Cardiogenetics by Eurofins Biomnis

After exome sequencing, Eurofins Biomnis offers a bioinformatic analysis targeting a panel of genes related to the patient's condition. The panels include genes which variations are known in scientific literature to cause the condition in question. They are regularly updated in line with developments in medical knowledge for hereditary or rare heart diseases.

Our targeted panels by pathology[1]

- Hereditary Hypertrophic Cardiomyopathy*
- Hereditary Dilated Cardiomyopathy*
- Hereditary Restrictive Cardiomyopathy*
- Non-Compaction of Left Ventricle*
- Arrhythmogenic cardiomyopathy of the right ventricle/left ventricle/both ventricles*
- Congenital Long QT Syndrome
- Brugada Syndrome
- Short QT Syndrome
- Jervell Lange-Nielsen Syndrome
- Andersen-Tawil Syndrome
- Timothy Syndrome
- Cardiac Conduction Disorder
- Sudden Cardiac Death
- Catecholaminergic Polymorphic Ventricular Tachycardia

*Panels including genes of interest recommended by the French National Authority for Health (HAS) (Evaluation report, February 2025)

Results are available within **4 weeks.**

Technique and performance

Next-generation sequencing (Illumina Novaseq) allows for the analysis of all coding regions of the genes in the panels.

The data are then analysed and interpreted using the SeqOne bioinformatics solution. This approach provides with coverage of the entire exome (~22,000 genes, 37.5 Mb), with ~98% of bases covered at more than 30X[2] and sensitivity greater than 99%[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 performance

In order to provide doctors with an increasingly reliable and accurate tool for therapeutic decisions, **Eurofins Biomnis is partnering with the Hospices Civils de Lyon**, a centre renowned for its experts in cardiogenetics, for the interpretation of variants.

Pharmacogenetics, for the optimisation and personalisation of drug treatments

Exome sequencing makes it possible to define a patient's pharmacogenetic profile based on interindividual genetic variations.

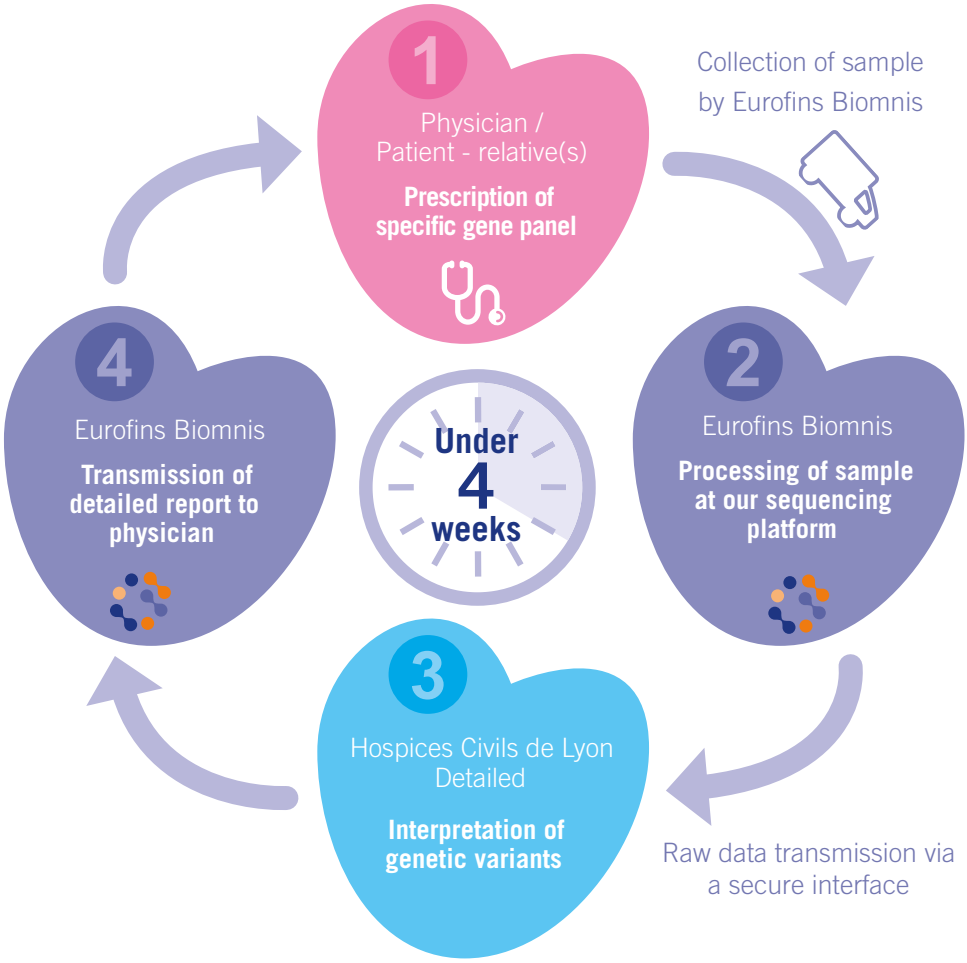
In practice, this pharmacogenetic profile makes it possible to:

- predict a response to a drug,
- explain an adverse effect,
- or guide clinicians in choosing the molecule or dosage.

The profile is carried out using the same sample as for exome sequencing.

Results are available within **4 weeks**.

Perform your targeted gene panels with Eurofins Biomnis and Hospices Civils de Lyon



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

- Rapport d'évaluation "Séquençage haut débit ciblé des panels de gènes dans le diagnostic des cardiomyopathies héréditaires" de la Haute Autorité de Santé, février 2025.

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.eurofins-biomnis.com** > Test guide > Test code **EXOME**
Assosiated tests	▸ Study of relatives by Sanger sequencing ▸ Congenital heart defects ▸ Additional pharmacogenetic profile ▸ Hereditary transthyretin amyloidosis - Molecular study of the TTR gene

For more information

Eurofins Biomnis
International Division
Tél. : +33 (0)4 72 80 23 85
E-mail : international@biomnis.eurofinseu.com

eurofins | Biomnis

Eurofins Biomnis
17/19 avenue Tony Garnier
BP 7322 - 69357 LYON Cedex 07 - FRANCE
www.eurofins-biomnis.com

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