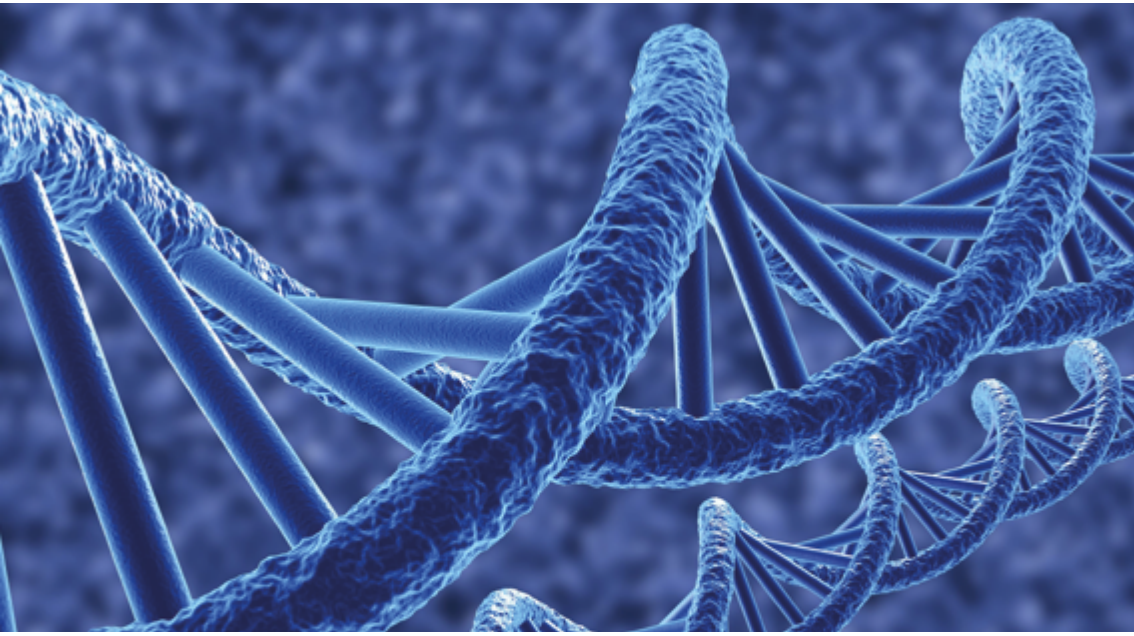
eurofins | Biomnis

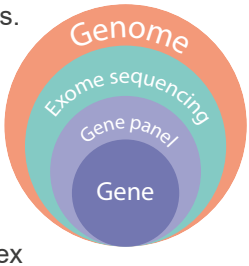


Exome Sequencing in genetic diseases

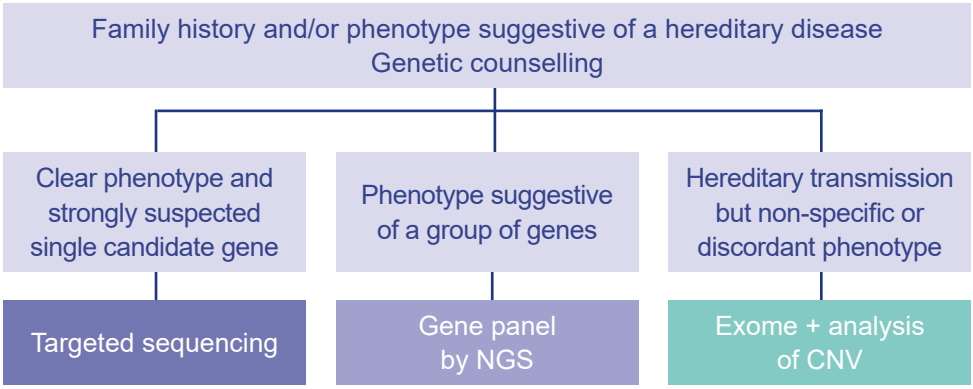
The clinical / clinical pathology partnership that benefits patients

Exome Sequencing in diagnostic practice

The term “genetic disease” embraces a wide range of pathologies. The identification of causal variants facilitates clinical diagnosis and prognosis, patient management, genetic counselling and, in some cases, the adjustment of therapeutic care to individual patient needs.



The conventional step-by-step sequencing approach (gene by gene or panel by panel), which sometimes follows complex decision trees, is gradually being replaced by **exome sequencing as a first-tier technique, in an increasing number of indications**[1-2]**.**



Exome Sequencing makes it possible to sequence, in a single step, all of the coding regions of the human genome (containing more than 85% of pathogenic mutations) **at an affordable price.**

As a first-tier technique, exome sequencing provides:

- time-saving compared to carrying out a series of gene panels,
- a diagnostic test for patients where the clinical examination does not enable the clinician to identify the gene/panel of genes to be tested.
- retrophenotyping in atypical clinical pictures,
- an improved diagnostic yield compared to the gene panel.

The exome sequencing by Eurofins Biomnis

4 weeks

In the laboratory

- A dedicated team of clinical pathologists, technicians and bioinformatics specialists
- Continuous biopathological support, from the implementation of the test to the interpretation of the results.

Sequencing

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

At the end of the analytical process

- An interpretation of an average of 30,000 variants in consultation with the clinician
- Provision of raw data (fastQ, VCF, BAM and quality report) via a secure interface
- Report with detailed results within an optimised period of 4 weeks

Why Eurofins Biomnis?

- Specialised medical pathology laboratory
- ISO 15189 accreditation (CAP standard equivalent)
- Authorisation to perform constitutional genetics tests
- Certified clinical pathologists
- Expertise in sequencing techniques in diagnostic practice

The clinical/clinical pathology partnership that benefits patients

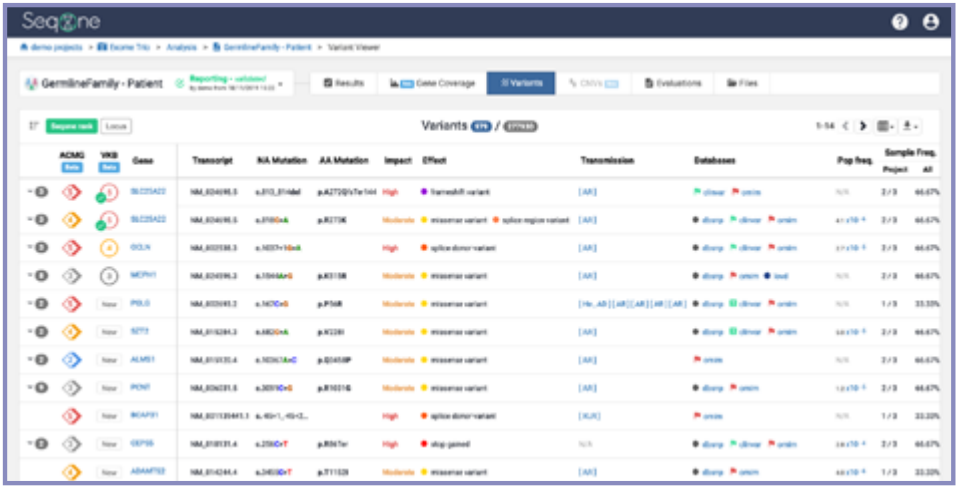
One of the challenges of exome sequencing is the interpretation of variants.

To actively involve partner clinicians and clinical pathologists in the interpretation of data, Eurofins Biomnis provides access to **SeqOne**, a secure bioinformatics platform.

**target CDS Refseq +/- 3 base pairs*
***data calculated from SNV's from NIST002 reference samples, for 40 million pairs of reads generated.*

SeqOne, for optimal diagnostic performance

Thanks to this platform, **a joint interpretation with Eurofins Biomnis or an autonomous interpretation** is possible. This ensures **optimal diagnostic performance** (laboratory experience and literature[3]).



The data is accessible and available to the clinician at all times, which allows:

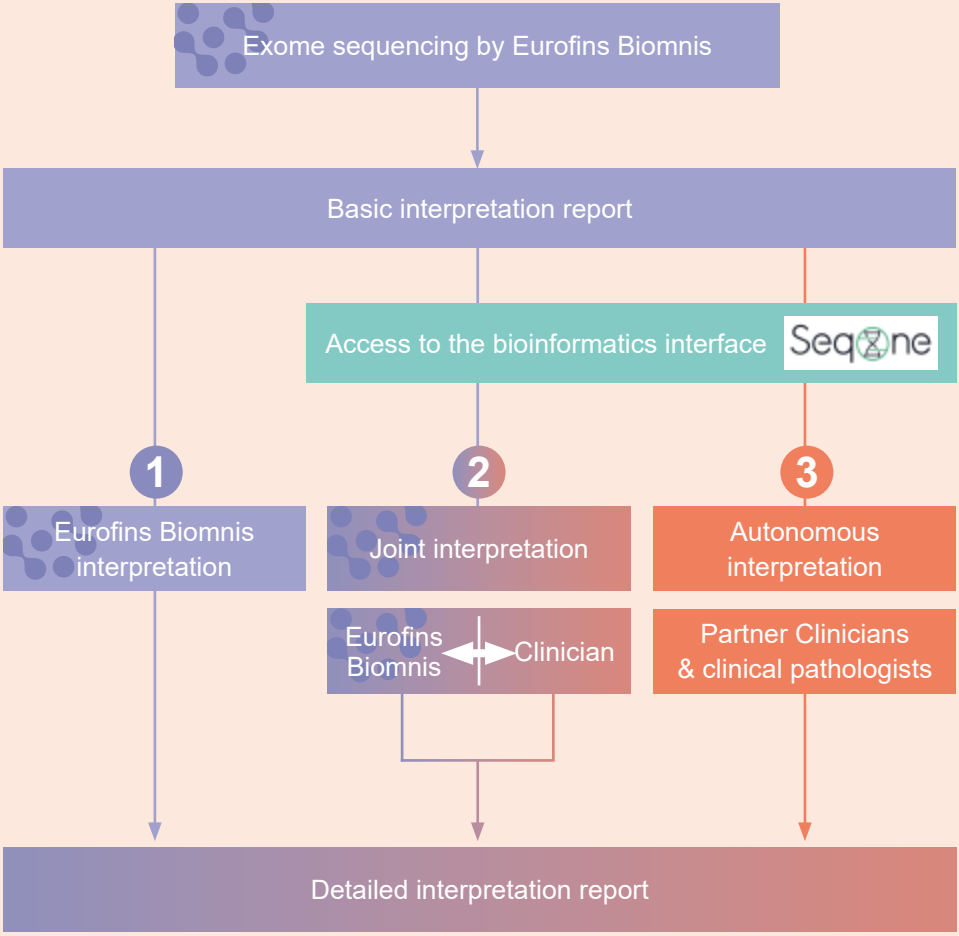
- reanalysis, when if required;
- use of data for research.

Eurofins Biomnis exome add-on modules:

- **CNV detection**
- **Detection of mitochondrial variants**, even with low heteroplasmy
- **Detection of pathogenic variants** in the *MUC1* VNTR
- **Detection of uniparental disomy** (for trio exomes)
- **Pharmacogenetic analysis**
- **HLA typing**

Sequence your exomes with Eurofins Biomnis

3 interpretation options/solutions



References

1. Exome sequencing has higher diagnostic yield compared to simulated disease-specific panels in children with suspected monogenic disorders. Oliver James Dillon et al. European Society of Human Genetics, Feb 18, 2018.

2. Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. Srivastava S et al. Genet Med. 2019 Nov;21(11):2413-2421. doi: 10.1038/s41436-019-0554-6. Epub 2019 Jun 11.

3. Paediatric genomics: diagnosing rare disease in children. Wright CF et al. Nat Rev Genet. 2018 May;19(5):253-268. doi: 10.1038/nrg.2017.116. Epub 2018 Feb 5.

Practical details

	Exome Sequencing & Access to the interpretation interface	
Interpretation level	**Detailed** report	**Basic** report
Turnaround	• **4 weeks** excluding any additional examinations	2 weeks
Indications	• intellectual disability, neurodevelopmental disorders, • syndrome-based disorders, • clinical pictures which do not directly suggest a specific gene test or gene panel, or negative result for these primary tests, • organ damage (kidney, heart, etc.).	
Sample	**Solo:** 5 mL EDTA whole blood or DNA sample **Duo/Trio:** 5mL EDTA whole blood or DNA sample from relative	
Conservation & transport	Room temperature	
Technique	Exome sequencing + bioinformatics pipeline	
Required documents	**B34-INTGB** Test request form available on **www.eurofins-biomnis.com** > Test guide > Analysis code **EXOME**	
Price	Contact us	
Complementary test	Study of relative persons by Sanger sequencing: 5 mL EDTA whole blood or DNA sample from relative	

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