Press release
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Eurofins Biomnis uses Whole Exome Sequencing to detect Copy Number Variation (CNV), improving diagnostic performance for patients with genetic diseases

Eurofins Biomnis, European leader in specialised clinical pathology and in genomic sequencing expertise, is now using *Whole Exome Sequencing* (WES) to detect *Copy Number Variation* (CNV). The losses or gains of chromosomal material associated with CNVs can be responsible for genetic diseases. Until now, when there was a suspicion of a genetic pathology, two tests were performed one after the other to determine whether the abnormality was chromosomal in origin or related to a gene mutation. It is now possible by means of WES to answer these two questions with a single DNA analysis. This creates a time saving for both patients and clinicians by cutting out a lot of diagnostic guesswork.

The combination of clinical pathology with bioinformatics for high quality screening

When screening for a genetic disease, several tests can be performed to find a diagnosis for the patient's condition. If healthcare professionals suspect a chromosomal abnormality, a cytogenetic analysis is performed. Two technical approaches are possible: the karyotype and the ACPA (chromosomal analysis by DNA chip (or SNP array / CGH array)). And if a small order variation is suspected (from one nucleotide to dozens of nucleotides), a high throughput sequencing method is used. Until recently, it was necessary to do the two tests one after the other. Eurofins Biomnis now offers – based on the *Whole Exome Sequencing* technique – a molecular genetics method that detects chromosomal and genetic abnormalities in a single test. Eurofins Biomnis is the first private sector clinical laboratory in France to have gained sufficient expertise in sequencing, bioinformatics[1] analysis and diagnostic interpretation to offer this option.

The high performance of Eurofins Biomnis *Whole Exome Sequencing* analyses (sensitivity, coverage and depth ...) makes it possible to replace ACPA and karyotype (in the exploration for unbalanced abnormalities). However, the technological leap forward does not stop there: it is also possible to identify abnormalities too small to be detected with cytogenetic techniques, thus increasing the diagnostic yield.
Eurofins Biomnis can also count on the expertise of its dedicated exome sequencing team. The team consists of four bioinformaticians, four scientific managers and six clinical pathologists with the qualifications, experience and skills to analyse and interpret the results.

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[1] **Eurofins Biomnis uses its SeqOne platform for the analysis and interpretation of data from exomic sequencing.*

Faster and less expensive results

By using exome sequencing to detect CNVs, Eurofins Biomnis helps reduce patient waiting times for diagnoses.
This is because the method provides comprehensive information in a single step and on a single sample, with enhanced diagnostic performance. It also reduces the turnaround time for the results. Instead of two analyses previously performed, one is now sufficient. For patients, this means a significant reduction in the waiting time for results, and for clinicians, a reduction in the number of consultations and samples collected before reaching a diagnosis.

In addition, screening for genetic diseases incurs significant costs for the health system. The single-stage analysis means these costs can be reduced.

For both healthcare professionals and patients, this approach represents a real step forward as it is now possible to diagnose genetic diseases at an earlier stage and to implement personalised care for each patient.

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About Eurofins Biomnis

European leader in the specialised medical biology sector, Eurofins Biomnis performs more than 32,000 analyses per day on a panel of more than 2,500 examinations, including specialised procedures for which it has all the necessary approvals. Established in 1897 by Marcel Mérieux, Eurofins Biomnis is the market leader in clinical biology in France thanks to constant innovation and technological investment, particularly in the fields of women's biology, oncology and personalised medicine as well as chromosomal and molecular genetics. With 120 years of expertise and innovation in the service of medical biology, Eurofins Biomnis – the European platform of the Clinical Diagnostics division of the Eurofins Group – is consistently growing its international business.
www.eurofins-biomnis.com

About Eurofins - the world leader in bioanalysis

About Eurofins - the world leader in bioanalysis Eurofins analyses in the service of life and health. With a workforce of over 50,000 persons and a network of over 800 laboratories in over 50 countries, the Eurofins Group companies offer a rich portfolio of over 200,000 analytical methods. Eurofins Scientific shares are listed on the Euronext Paris stock market.