

Lyon, the 22th of January 2020

## **EUROFINS BIOMNIS AND SEQONE PARTNER TO IMPROVE CLINICAL ACCESS TO WHOLE EXOME TESTING**

**Eurofins Biomnis, a leading medical interpretation lab and SeqOne, the provider of advanced genomic interpretation tools today announced a partnership aimed at facilitating access to Whole Exome genomic testing. The partnership allows Eurofins Biomnis to provide clinicians treating hereditary diseases access to fast and affordable Whole Exome sequencing, together with state-of-the-art interpretation tools that facilitate the interpretation of the data. Thanks to this partnership, a clinician can, working with a Eurofins Biomnis genomic specialist, obtain a detailed genomic analysis report that allows him to diagnose patients more quickly and accurately.**

Eighty percent of rare diseases are genetic in origin and result from a small number of genetic mutations. Identifying these variations is essential in diagnosing the diseases and can dramatically improve patient care.

### **Making Whole Exome sequencing more affordable to improve diagnostic efficiency**

Since 2017, Eurofins Biomnis has offered "Whole Exome" sequencing which, starting from a simple blood test, provides information on all the genes relevant to a diagnosis. Using Whole Exome sequencing more than 30% of patients can be diagnosed with a single. Test, which represents a significant improvement over traditional gene panel approaches that result in diagnoses in only 10% - 15% of cases.

Until now and despite its superior performance, the use of "Whole Exome" sequencing has been limited. Obstacles have included the high costs, as well as the limited availability of adequate scientific, clinical and technical means, necessary to identify the causal mutation among the tens of thousands of mutations detected in "Whole Exome" sequencing.

### **Harnessing artificial intelligence to better identify and interpret mutations**

The partnership between Eurofins Biomnis and SeqOne combines best-in-class sequencing with the most efficient tools to facilitate the interpretation of the results. Thanks to this, a detailed report of the results to be obtained in under six weeks.

Whole Exome sequencing generates on average 200 GB of raw data and reveals tens of thousands of mutations. The SeqOne platform uses artificial intelligence techniques to explore academic research databases and cross-reference this information with the test results. This allows the platform to

automatically rank each patient's mutations in order of diagnostic relevance thus helping to identify the right diagnosis. The easy-to-use cloud-based system offers biologists and clinicians the most effective genomic interpretation tool on the market. "SeqOne was designed to make the most complex genetic analysis accessible to as many people as possible," said Nicolas Philippe, CEO of SeqOne.

Dr Ines Harzallah of the chromosomal molecular genetics service of CHU Saint-Etienne explains the advantages of having access to a high performance genomic interpretation platform like SeqOne thus: "We are convinced that Whole Exome sequencing will help improve our diagnostics. Nevertheless, this test is complex and expensive today. The Eurofins Biomnis/SeqOne combined offer provides an excellent quality of sequencing coupled with an intuitive bioinformatics analysis notably thanks to the automatic help in the classification of variants and all at an attractive price. Our experience shows that, in practical terms, it saves precious time".

### **Do-it-yourself or collaborative variant interpretation options available**

When a healthcare professional requests a Whole Exome analysis from Eurofins Biomnis, they automatically have access to the SeqOne platform, at no additional cost. There are three options for interpreting the results:

- interpretation of the variants by Eurofins Biomnis,
- a joint interpretation carried out with a Eurofins Biomnis clinical pathologist, or
- interpretation of the data independently.

The co-interpretation model that ensures optimal diagnostic yield because it combines the scientific knowledge of Eurofins Biomnis biologists specializing in genetics with that of the clinician. For Laure Raymond, the Eurofins Biomnis clinical pathologist responsible for Whole Exome test and interpretation, "the partnership with SeqOne is part of Eurofins Biomnis' strategy to actively involve clinicians in the interpretation of exomes". The number of patients without diagnosis is reduced and clinicians have the opportunity to grow their expertise in genomic medicine thanks to the intuitive SeqOne interpretation tool and interaction with our experts.

Thanks to its logistics network, Eurofins Biomnis is able to guarantee patients and the medical profession availability of this medical test anywhere in France with more countries slated to come onstream soon.

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#### **About Eurofins Biomnis**

European leader in the sector of specialised clinical pathology, Eurofins Biomnis carries out over 32,000 analyses per day from a range of over 2,500 available tests, including specialised tests for which the company has the appropriate authorisations. Founded in 1897 by Marcel Mérieux, Eurofins Biomnis is the leader in specialised clinical pathology in France. It has maintained this leading position through continuous technological innovation and investment, particularly in areas such as female biology, oncology and personalised medicine, as well as chromosomal and molecular genetics. With 120 years of expertise and innovation in the service of clinical pathology, Eurofins Biomnis forms the core of the Clinical Diagnostics division of the Eurofins Group in Europe and is actively expanding in worldwide markets. [www.eurofins-biomnis.com](http://www.eurofins-biomnis.com)

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#### About SeqOne

SeqOne develops state-of-the-art genomics analysis tools for clinical applications in the fields of cancer and rare disease. Its flagship product, SeqOne | Platform is a cloud-based end-to-end solution that dramatically reduces the turnaround time and cost required to deliver accurate genetic analyses for use in mainstream medicine. In the short time since it has launched its platform secured a wide user base in a diverse range of healthcare establishments including hospitals and private sector testing labs. It has won numerous awards including the prestigious iLab award and the ARC cancer foundation's Hélène Stark prize. SeqOne is supported by the SATT AxLR and the Montpellier BIC incubator. For more information: <https://seqone.com>