Press release

Eurofins Biomnis offers a new tool for clinical diagnostics: clinical exome

Lyon, 8 November 2016. The advent of new, high-throughput sequencing techniques, also called NGS (*Next Generation Sequencing*) or MPS (*Massively Parallel Sequencing*) opens up many opportunities in the field of constitutional genetic disorders (rare diseases) and acquired genetic disorders (cancer). By rapidly deciphering an individual's complete DNA and thus generating a wealth of information on genetic heritage, the NGS technique ultimately makes it possible to make a diagnosis, to determine the risk of predisposition to certain diseases, or even to predict the efficacy of a drug treatment (personalized medicine).

Eurofins Biomnis, the European leader in specialized medical pathology, has been an expert in NGS for several years and conducts the technique on a daily basis, in particular in the field of non-invasive prenatal testing for Down's syndrome, Edwards' syndrome and Patau syndrome (NIPT).

Thanks to this technological mastery, **Eurofins Biomnis is the first private medical pathology laboratory in France to offer clinical exome (sequencing & medical interpretation),** an innovative and accurate tool in clinical diagnostics, particularly in the field of intellectual disability.

Clinical exome in the diagnosis of intellectual disability

Intellectual disability is a common disorder. It affects between 1 and 2% of the population (i.e., about 1 million people in France) and has a genetic cause in half of all cases.

Genetic disorders cover a range* of clinically heterogeneous diseases, all caused by one or more variations at the level of the exons.
In over 50% of cases, it is not possible to establish a diagnosis using conventional clinical and biological approaches.

At Eurofins Biomnis, the multidisciplinary team responsible for exome in a routine clinical context study approximately 93 to 97% of a patient's medically relevant genes in a single analysis, thanks to the NGS technique. At the end of the analytical and bioinformatics process, biological interpretation looks at on average 20,000 variations and, in the context of intellectual disability, can identify the genetic abnormality responsible in 23-35% of cases, whereas the conventional approach can only detect the variation in 10-15% of cases.

Identification of the mutation responsible is a considerable breakthrough in the management of intellectual disability. Not only does it help reduce the number of undiagnosed patients, but it also offers doctors a new and reliable tool for making treatment decisions, in order to adapt the specific support and monitoring provided to the patient, or even to propose a targeted and personalized treatment in some case. Clinical exome is also very relevant as an aid to genetic counselling in prenatal diagnostics, where the aim is to determine the risk of passing on the condition.
For many families, clinical exome puts an end to misdiagnosis.

In addition to intellectual disability, Eurofins Biomnis is able to offer clinical exome in France and internationally as routine diagnostics for other disorders, such as autism, epilepsy, neurodevelopmental disorders, cardiomyopathy, metabolic diseases, etc., with the aim of improving the rate of diagnosis of these diseases through the further development of genome sequencing in the near future.

* *All of the coding parts of the genome alone contain 85% of the mutations implicated in genetic disorders. Thus, the human genome consists of about 200,000 exons for 20,687 genes.*

Eurofins Biomnis has great expertise in the field of medical research

The value of NGS has already been evaluated very positively in both medical and socio-economic terms: certainty of the diagnosis, increased diagnostic specificity, more accurate and effective treatment approach, reduced adverse effects of treatment, prevention of disease in carriers, etc. Despite this, its use is still limited because it requires significant investment, specific scientific and clinical expertise, as well as a mastery of bioinformatics.

As we are convinced that this technique will become one of the standard tools in analysis, monitoring and diagnosis in coming years, Eurofins Biomnis is also working together with researchers, the diagnostics sector and the pharmaceuticals industry by offering its expertise in sequencing and clinical exome for all clinical research projects: identification of mutations of interest, help in the understanding of analytical data, and medical pathology-related advice for the interpretation of the results in a variety of areas of human genetics.

Thanks to its dedicated platform, Eurofins Biomnis is able to bring together dedicated sequencing activities and many medical specialties essential to the successful performance of genetic tests and high-tech developments. Using the detailed knowledge of Eurofins Biomnis medical pathologists who are experts in genetics, cytogenetics, haematological oncology, pharmacogenetics and anatomical pathology, as well as the mastery of conventional analytical methods, this multidisciplinary approach bring scientific legitimacy to the developments and commitments undertaken by Eurofins Biomnis in this area.

Innovating today for the health of many tomorrow

Genomic medicine represents a real game-changer for public health, introducing a new approach in the way that we prevent, diagnose and treat diseases, as well as determine a prognosis. This field has in fact been the subject of the "France Genomic Medicine 2025" plan launched by the Prime Minister Manuel Valls, with the aim to grant access to genetic diagnostics throughout the country within 10 years.

To ensure that the medical care of tomorrow can offer the best opportunities to as many people as possible, Eurofins Biomnis is continuing its investments and its development work in this field.

About Eurofins Biomnis

European leader in the sector of specialized medical pathology, Eurofins Biomnis carries out over 32,000 analyses per day from a range of over 2,500 available tests, including specialized tests for which the company has the appropriate authorizations.

Founded in 1897 by Marcel Mérieux, Eurofins Biomnis remains the leader in the field of specialized medical pathology in France. It has kept its position through continuous technological innovation and investment, particularly in areas such as female biology, oncology and personalized medicine, as well as chromosomal and molecular genetics.

The laboratory also stands out through its expertise in clinical research and biobanking, thanks to its dedicated infrastructure.

With 120 years of expertise and innovation in the service of medical pathology, Eurofins Biomnis is the core of the Clinical Diagnostics division in Europe and has its eyes set on its international development.

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