Press release

Eurofins Biomnis offers new diagnostic tool for rare diseases

Lyon, 20 February 2017. More than 8,000 rare diseases have now been identified[1]. More than 3 million individuals are affected by these pathologies in France and 1 in 2 of the sufferers is a child.

80% of these rare diseases are of genetic origin and result from a mutation (or modification) of a single major gene or a combination of genetic mutations. They are expressed in a wide variety of symptoms that not only differ from one disease to another but also from one patient to another with the same disease, which explains the complexity and duration of the analyses required for their diagnosis. It is estimated that at present one sufferer in three is undiagnosed. The search for a diagnosis often is long and difficult for patients and their families.

Just few days before the 10th International Rare Disease Day, improving access to information, diagnosis and appropriate care for persons with rare diseases is more than ever a major public health issue.

[1] *A disease is said to be rare when it affects less than 1 person in 2,000.*

The medical exome in the diagnosis of rare diseases

The genome, which is unique to each person, is the sum of all the genetic material in the body. The genome consists of more than 20,000 genes, themselves consisting in part of exons. The role of exons is to synthesize the various proteins essential to the proper functioning of all our organs. The individual exons together go to form the exoma, which alone contains 85% of the anomalies responsible for rare genetic diseases.

At the Eurofins Biomnis laboratory, the scientific and medical team responsible for the “medical exoma” test is now able to identify around 93-97% of all medically relevant genes in a patient in a single analysis with the aid of a high-throughput sequencing technique[2], from a single blood sample. On completion of the test, sequencing of the exome makes it possible to identify the genetic abnormality responsible for the respective condition in 23 to 35% of the cases, compared with conventional clinical/biological approaches which can only detect such anomalies in 10 to 15% of cases.

Not only does complete sequencing of the exoma reduce the number of undiagnosed patients, but it also provides physicians with a new, reliable tool for making treatment decisions, enabling optimization of the support and follow-up of the individual patient, and in some cases even opening up the prospect of selective, customized treatment. Clinical exome sequencing has also proven extremely valuable as an aid to genetic counselling in prenatal diagnostics, where the aim is to determine the risk of occurrence or of passing on hereditary conditions.
For many families, the medical exoma puts an end to diagnostic guesswork.

With its mastery of these scientific and technological techniques, Eurofins Biomnis is the first private medical laboratory in France to offer the medical exome and backed by its efficient organization and daily collection network, assure all patients and the medical profession of complete continuity of support both in France and internationally.

[2] *High-throughput sequencing, also known as Next Generation Sequencing (NGS), is a molecular biology technique that rapidly decodes the subject’s complete DNA and generates a wealth of information about his or her genetic make-up.*

The expertise of Eurofins Biomnis in the service of medical research

In relation to rare diseases, raising awareness of the risk of misdiagnosis is as important as the search for treatments. In fact, nearly 97% of these pathologies are so-called “orphan” diseases for which no treatment is available.

The use of new generation sequencing techniques is still limited because it requires significant investment, great scientific and clinical expertise, and advanced skills in bioinformatics. The involvement of specialized medical biology laboratories in partnership with academic research institutes and public health care providers is therefore essential.

Eurofins Biomnis is working alongside researchers, the diagnostics sector and the pharmaceuticals industry by offering its expertise in sequencing and clinical exome sequencing for clinical research projects designed to identify new approaches to treating these numerous diseases.

Innovating today for the health of many tomorrow

Genomic medicine is a major challenge in public healthcare. This challenge was addressed the “France Genomic Medicine 2025” plan launched by the former Prime Minister Manuel Valls, with the aims of improving understanding of genetic mechanisms and mutations including previously unknown ones and securing access to genetic diagnostics throughout France within 10 years.

To ensure that the medical care of tomorrow can offer the best opportunities to as many as possible, Eurofins Biomnis is committed to integrating genomic medicine in the management of rare diseases and in particular is continuing to invest and pursue development work in this field with a view to improving the rate of diagnosis of these pathologies thanks to the sequencing of the genome[3] in the near term.

[3] *Genome sequencing will improve and extend exoma coverage and thus increase diagnostic effectiveness.*

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.
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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