



"WHOLE EXOME" analysis A new diagnostic tool for rare genetic diseases

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Laboratoire Cerba and the University of Bourgogne will be working together to identify new genes involved in rare genetic diseases using whole exome sequencing.

Whole exome sequencing constitutes a new and particularly promising field of investigation in human genetics.

Laboratoire Cerba, is using this diagnostic technique since last summer. Our commitment is the diagnosis of rare constitutional genetic diseases in order to avoid patients and their families a continued diagnostic odyssey in their search for an answer.

The diagnosis of rare genetic diseases is a crucial public health issue given the disabilities and complications associated with them. More than 6,000 diseases have so far been identified and each month five new ones are described in medical literature.

In this context, Laboratoire Cerba and the University of Bourgogne will be undertaking the joint supervision of a doctoral thesis. The aim of this collaborative project is to pool the data of both laboratories obtained by exome sequencing in order to identify new genes responsible for genetic diseases.

As Jean-Marc Costa, clinical pathologist at Laboratoire Cerba and specialist in constitutional molecular genetics, pointed out: "This public-private research partnership will provide further knowledge in the field of rare diseases thanks to the joint patient recruitment of the CHU Dijon Bourgogne and Laboratoire Cerba. It illustrates our commitment to work alongside medical practitioners and patients, to develop research and accelerate the availability of research outcomes to all patients".

According to Professor Laurence Faivre, coordinator for the "developmental anomalies and malformation syndromes" Centre of Reference for Rare Diseases at the CHU Dijon Bourgogne, "this jointly-supervised thesis will enable us to unite our strengths to discover and validate new genes involved in rare diseases. We are delighted with both past and future collaborations with Laboratoire Cerba in this field".

This doctoral thesis is set to begin next January for an initial period of three years. The first results are expected at the end of 2017. This will certainly open a large number of promising perspectives for all the patients and will enable the practitioners to offer both patients and families the very best support possible.

About Laboratoire Cerba

Created in 1967 and accredited in compliance with the NF ISO 15189 standard since 1999, Laboratoire Cerba is the European leader in specialist medical biology.

Integrating more than 40 medical specialties and employing over 600 highly qualified staff on a single site located to the north-west of Paris, on average Laboratoire Cerba handles some 40,000 dossiers daily.

With a state-of-the-art technological platform it is able to offer private and hospital laboratories in over 50 countries a wide range of specialised biological tests in fields such as molecular biology, oncology, allergology, toxicology, hormonology, and infectious and metabolic diseases together with diagnostic analyses for genetic diseases in cytogenetics and molecular genomics.

Laboratoire Cerba ceaselessly works to develop its technological and human potential and has launched numerous collaborative projects with both public and private establishments to ensure it always remains on the leading-edge of innovation. Since 1998, Laboratoire Cerba's pathologists have contributed to over 500 scientific publications.

It is also a founder member of the Cerba HealthCare group, an international medical biology group, working in three complementary sectors:

- routine biology mainly in France, Belgium and Luxembourg with over 300 laboratories, 150 sample collection centres and 50 technical platforms;
- biology for medical specialties with the Cerba platform;
- clinical trial biology, essential to the pharmaceutical and biotechnology industry for the development of new molecules, through its subsidiary BARC today present on five continents.

Recently the Group has diversified into the veterinary biology sector with the creation of a dedicated unit, Cerba Vet.

It now employs nearly 4,300 staff, including 350 pathologists.

<http://www.lab-cerba.com/> - <http://www.cerbahealthcare.com/>

About the CHU Dijon Bourgogne

With more than 7,300 professionals representing over a hundred different professions all working in the interests of the patient, the CHU Dijon Bourgogne responds to all the healthcare needs of those living within the region and beyond. It offers a public service that includes an accident and emergency department, diagnosis, treatment and prevention. With 1,788 beds, it offers some 481,500 days of hospitalisation and accommodation per year. In addition, the establishment initiates and develops research programmes in partnership with other health institutions. Finally, it trains not only future general practitioners and specialist doctors through its role as a university teaching hospital but also several other paramedical professions within its seven schools and training institutes.

A modern structure, the CHU Dijon Bourgogne today figures amongst the establishments of reference at a national level in France. The excellence of its teams has been recognised in several specialisms and for numerous pathologies.

In order to guarantee the best care for its patients, the CHU Dijon Bourgogne has initiated a policy of continuous quality improvement, thus committing to a dynamic approach for its medical, hotel and administrative services. Given its cutting-edge technical platform, the establishment's medical and care teams also work constantly to advance technological innovation.

In addition to medical care and teaching, research also figures as a major element in its institutional policy. The CHU Dijon Bourgogne is therefore involved in a large number of protocols that aim at improving the quality of life of its patients. It notably has a genetics department of reference currently working on the genomics of developmental abnormalities and rare diseases.

<http://www.chu-dijon.fr/fr> - <http://www.translad.org> - <http://www.gad-bfc.org>

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