Paris and Issy-les-Moulineaux (France), 19 July 2021

METAFORA BIOSYSTEMS, AP-HP AND CERBA HEALTHCARE PRESENTED THE FINDINGS OF THE VALIDATION STUDY FOR THE METAGLUT1 TEST AT THE 3RD EUROPEAN CONFERENCE ON GLUT1 DEFICIENCY SYNDROME

The METAglut1™ test is the result of a collaborative initiative between METAFORA biosystems, the teams at AP-HP and over 30 clinical trial sites, and Cerba HealthCare. Its aim is the early diagnosis of this rare disease, which still remains underdiagnosed.

The De Vivo disease, or GLUT1 deficiency syndrome, is a rare and debilitating neurological disease that is still relatively unknown among the medical community, but it *can* be treated. It is estimated[1] that more than 3,000 people suffer from the De Vivo disease in France, and over 90% of them have currently gone undiagnosed. In GLUT1-deficient patients, the transporter necessary for glucose uptake by brain cells is impaired. The decreased glucose level in the brain causes epileptic seizures, abnormal movements and developmental delay. However, unlike many genetic diseases, treatment is available for De Vivo disease in the form of a high-fat diet (known as a ketogenic diet), which significantly improves symptoms in patients. Molecules designed to bypass the deficiency related to the lack of glucose supplied to the brain are also currently being assessed. Diagnosing the disease as early as possible so that proper treatment can be established is therefore of crucial importance.

This new blood diagnostic test promises to help quickly (within 48h) and easily identify affected children and adults, compared to the current diagnostic tests which rely on an invasive procedure (lumbar puncture) or complex genetic analysis.

The partners announced the key findings of a multicentre validation study at the 3rd European GLUT1D Conference on Friday 11 June 2021. The study highlighted similar performance between the diagnostic test and the more invasive standard approach – lumbar puncture – for detecting this metabolic abnormality. In particular, the test resulted in almost 100% specificity and its sensitivity was identical to the lumbar puncture at around 80%.

"This rapid and precise blood diagnostic test, readily available to all physicians and, especially, paediatricians and neurologists, is a major milestone for patients with this rare disease, as their diagnosis and treatment will be much faster," stated Dr Fanny Mochel, Principal Investigator for the study, geneticist and Head of the Reference Centre for Adult Neurometabolic Diseases at AP-HP Pitié-Salpêtrière Hospital.

"METAglut1 offers patients and their families major prospects in the fight against delayed diagnosis, which can mean years before they get to the bottom of their symptoms, despite treatments being available," explained Magali Sorret, Founder and President of the ASDGLUT1 patient support group.

Thanks to this new and innovative blood test, it will be possible to screen for the disease in any patient showing learning difficulties and/or epilepsy and/or abnormal movements.

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[1] https://ghr.nlm.nih.gov/condition/glut1-deficiency-syndrome and Symonds et al. *Incidence and phenotypes of childhood-onset genetic epilepsies: a prospective population-based national cohort*. Brain. 2019 Aug 1;142(8):2303-2318.

"We are delighted to have been supporting Metafora and the clinicians since the test was in its very first development phases, and especially to have participated in this major multicentre study. The consortium has successfully demonstrated how robust this innovative test is on a large scale, which means it will be distributed nationwide," explained Jérôme Sallette, Chief Scientific Officer at Cerba HealthCare.

This validation study of the diagnostic performance of METAglut1 was conducted under the Forfait Innovation programme, which is a French Coverage with Evidence Development scheme for innovative *in vitro* diagnostic medical devices or procedures in the early stages of development. It is awarded subject to completing a study to confirm the value of the technology. This is the first diagnostic technology to benefit from the Forfait Innovation as part of its validation.

"We are grateful to all our partners for the quality of their work and for their involvement, which made this ambitious study possible. The Forfait Innovation offered a particularly suitable framework for us to see it through, and we would like to thank the French Ministry of Social Affairs and Health as well as the HAS for the trust they have placed in us," concluded Vincent Petit, CEO of Metafora biosystems.

About METAFORA biosystems

METAFORA biosystems develops a unique proprietary platform able to detect abnormal metabolism at the cell level. Innovative reagents and softwares power the technology platform and allow to identify metabolic reprogramming which is part of numerous disorders.

METAFORA's approach to cell metabolism has been published in numerous scientific and medical articles in leading journals, and the company is now developing innovative solutions for in vitro diagnostics (IVD). It is geared to develop new solutions which address medical unmet needs. The market launch of its first test is happening in 2021 and 2022 in Europe, and its R&D pipeline in oncology aims at positioning METAFORA as a key player in the diagnostics market.

http://www.metafora-biosystems.com

About Cerba HealthCare

Cerba HealthCare, a leading player in medical diagnosis, aims to support the evolution of health systems towards more prevention. It draws on more than 50 years of expertise in clinical pathology to better assess the risk of diseases development, detect and diagnose diseases earlier, and optimize the effectiveness of personalized medicine.

Every day, on 5 continents, the Group's 9 600 employees sustain the transformation of medicine, driven by one deep conviction : to advance diagnosis is to advance health.

Cerba HealthCare, *enlightening health*.
www.cerbahealthcare.com

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