



Press release 28 September 2020

<u>28 September – 2 October</u> <u>The 25th International Annual Congress of the World Muscle</u> <u>Society</u>

Institute of Myology experts step up to the podium

Between 28 September and 2 October, the 25th International Annual Congress of the World Muscle Society, which brings together muscle experts from all over the world, will highlight the work of many researchers from the Institute of Myology, the centre of expertise on muscle and its diseases that was created by AFM-Telethon. From fundamental research to therapeutic advances, no less than 32 communications (e-posters and talks) will be presented by the scientific experts of the Institute of Myology.

Among the selected papers, two important breakthroughs will be presented to the international community on **Thursday, 1 October**:



At 4:30 pm, in the session entitled "OMICs and Al approaches for muscle diseases", Enzo Cohen, a Myology Research Centre post-doctoral fellow from the "Myomatrix & Myonucleus Related Diseases: Genetics & Pathophysiology" team headed by Gisèle Bonne, PhD, Inserm Research Director, will demonstrate how he identified two missense mutations in a patient, triggering a new form of myopathy. This work will help to diagnose patients with the same symptoms, and to generalise this new genetic diagnosis in the Filnemus network (French national health network for neuromuscular diseases).

At 7:30 pm, in the session entitled "From the spinal cord to the *muscle*", Giorgia Querin, MD, PhD, Neurologist, Adult Clinical Trials Platform Coordinating Physician at the Institute of Myology, will detail her study by magnetic resonance imaging (MRI) of the cervical spinal cord carried out in healthy carriers of a genetic mutation (C9Orf72), which can be the origin of amyotrophic lateral sclerosis (ALS) or frontotemporal dementia (FTD), both of which are neurodegenerative diseases. The objective of this study is to identify spinal cord degeneration at an early stage in individuals who do not yet have symptoms of the disease, to predict the risk of developing ALS or FTD, and to treat patients as soon as the first symptoms appear.



 \rightarrow All the information is <u>here</u>



The Institute of Myology, a unique centre of expertise on the muscle

The Institute of Myology was created in 1996 by AFM-Telethon to diagnose and treat patients, and to study diseased muscle, in partnership with five public bodies (the AP-HP, the CEA, Inserm, Sorbonne Université

and the CNRS). This centre of expertise is globally unique and promotes the existence and recognition of the field of myology by bringing together, in a single location, fundamental and applied research, clinical research, physiological assessment, care and teaching. Eight centres, bringing together 250 doctors and researchers, are dedicated to the muscle in all its states, from the national reference centre for the diagnosis, management and monitoring of neuromuscular diseases, to the research centre, a clinical research platform with innovative investigational and measurement tools, and a centre for training and dissemination of knowledge in the field of myology. The Institute is patient-centred and brings together diagnosis, clinical care, assessment and research. https://www.institut-myologie.org/en/

The Muscle Atlas: The first muscle image database to accelerate research, diagnosis and therapeutic approaches

The muscle is a complex organ that can be affected by a wide range of abnormalities. It can take years to diagnose a particular neuromuscular disease, some of which affect only a subset of skeletal muscles, for no particular reason. Today, there is no database in existence that provides an exhaustive and qualified record of muscle biopsies from diseased patients or animals, both treated



and untreated. This is the reason why Bruno Cadot and Dr Norma Beatriz Romero, researchers at the Institute of Myology, had the idea of creating a "Muscle Atlas", i.e. building a database from muscle biopsies analysed, for the most part, in their respective laboratories.

More than 4,000 pictures of muscle biopsies of animal and human origin, all characterised according to the disease and at different stages of the disease, have been recorded by now. This unique database was launched at the

WMS congress and will enable researchers and physicians to work faster in their research and in the diagnosis of neuromuscular diseases. This atlas will be enriched with contributions from researchers and physicians from all over the world in the coming months.

For any question regarding the enrichment of this collection, please contact: <u>nb.romero@institut-myologie.org</u> and <u>b.cadot@institut-myologie.org</u>

Press contacts: Stéphanie Bardon, Marion Delbouis – Email: presse@afm-telethon.fr – Tel.: +33 (0)1 69 47 29 01