INSTITUTO DE MEDICINA MOLECULAR
FACULDADE DE MEDICINA DA UNIVERSIDADE DE LISBOA

RESEARCH WORK OF CENTRONUCLEAR MYOPATHY CREATES *IN VITRO* PLATFORM TO TEST DRUGS EFFECTIVENESS

Results now published in EMBO Molecular Medicine unravel the mechanism of one of the genes responsible for rare neonatal disease

Lisbon (September 30, 2014): An international team led by Edgar Gomes, of the Myology Institut, Paris and Instituto de Medicina Molecular (IMM), School of Medicine, University of Lisbon, discovered the mechanism behind a gene responsible for the development of centronuclear myopathy (CNM), a genetic disease that affects the skeletal muscles (muscle weakness), a condition mostly attributed to neonatal and with a poor prognosis.

The team led by Edgar Gomes, of the IMM, discovered the mechanism of one (BIN1) of the four genes that have mutations in this pathology, a breakthrough possible through the development of a simple cellular system in which the disease was replicated in cell culture (*in vitro*) and through which the disease cell symptoms were reversed.

To Edgar Gomes this result is a giant step for the understanding of CNM since the study will now allow to quickly finding drugs that are clinically effective in treating this clinical condition.

"Our work suggests that activation of N-WASP by BIN1 can be a new therapeutic approach for the treatment of CNM and other musculoskeletal disorders. Furthermore, the *in vitro* system that we describe in this work provides a platform to screen for potential drugs to ameliorate or treat muscle disorders", underlines the IMM researcher.

The results of this investigation describe a new *in vitro* system for the study of muscular disorders. It was found a novel protein that is regulated by BIN1 (N-WASP) important for nuclear positioning and triad organisation. It was further discovered that N-WASP distribution is disrupted in skeletal muscle fibers from CNM and myotonic dystrophy patients. Furthermore, it was shown that the activation of N-WASP can reverse the pathological features of CNM *in vitro.*

The research paper was published online on September 29 in EMBO Molecular Medicine and can be consulted here.

About Centronuclear myopathy (CNM)

Centronuclear myopathy (CNM) is a rare disease, with about 50 families affected according to data reported in the scientific literature. It is a hereditary neuromuscular disease and its

incidence is estimated to be of 0.06 / 1,000 neonatal with a prognosis of death between three weeks to six months after birth.

About IMM

Instituto de Medicina Molecular (IMM) is a reference biomedical research centre in Portugal, having acquired the special status of Associate laboratory of the Portuguese Ministry of Science and Technology. IMM's mission is to promote basic, translational and clinical biomedical research with the aim to understand the mechanisms of disease and develop novel therapeutic approaches. More at: http://www.imm.fm.ul.pt

For more information please contact:
Liliana de Almeida – Communication and Public Affairs Director
Telephone: +351 217 999 414
Mobile: +351 918 486 058
Email: lsalmeida@medicina.ulisboa.pt