PRINCIPE FELIPE
CENTRO DE INVESTIGACION

Seminario CIPF

GDAP1 links mitochondrial dynamics to insulin signaling in Charcot-Marie-Tooth neuropathy

Speaker: Víctor López del Amo
Developmental Biology And Neuromuscular Diseases Models
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Abstract: One of the genes involved in Charcot Marie Tooth disease (CMT), an inherited peripheral neuropathy, is GDAP1 which encodes a protein anchored to the mitochondrial outer membrane. We recently published (doi: 10.1093/hmg/ddu416.) that there is a true ortholog of this gene in Drosophila, which we have named Gdap1.

A metabolomic study using nuclear magnetic resonance (NMR), determined that up- and down-regulation of Gdap1 results in a systemic attenuation of the insulin pathway, which is probably caused by abnormal mitochondria-ER contact as the result of too much or too little Gdap1 activity.

We translated our Drosophila results to neuronal mammalian cell culture to study the function of different proteins related with insulin signaling route. Overexpression of GDAP1 mutations showed a poor response after insulin treatment in the phosphorylation cascade of some proteins related with insulin pathway.

These results point to a coexistence between neurodegeneration and metabolism imbalance with mitochondria as main protagonist. This new pathway could be important for CMT caused by GDAP1 mutations and for other diseases related with mitochondria.

C/ Eduardo Primo Yúfera, 3 (junto Oceanográfico) · 46012 VALENCIA (Spain) · Tel: +34 96 328 96 80 - Fax: +34 96 328 97 01 / www.cipf.es / CIF G-46/923421

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