UNIVERSITÀ
DI TRENTO
Department of
Cellular, Computational and Integrative Biology - CIBIO

PhD Colloquia 2022

An opportunity to address, explore and exchange information on scientific topics with scientists.

Povo 1, Room A221
Via Sommarive 9, Povo (TN)

Friday, 27th May 2022 - h 9:00

MASSIMO PANDOLFO

Department of Neurology and Neurosurgey
McGill University, Montreal (CA)

Boosting Frataxin expression to treat Friedreich Ataxia

Friedreich ataxia (FRDA) is the most common form of hereditary ataxia in Europe. It is caused by deficiency of the frataxin protein, leading to impaired mitochondrial biogenesis of Fe/S clusters, loss of activity of multiple Fe/S enzymes, mitochondrial dysfunction, and altered iron metabolism. In most cases, frataxin deficiency is due to impaired transcription of the FXN gene triggered by homozygous intronic expanded GAA repeats via epigenetic changes. Data from cellular and animal models support restoration of frataxin levels as potential therapy for FRDA. This can be accomplished in principle by enhancing the transcriptional activity of the endogenous FXN gene or via the introduction of a frataxin-expressing transgene. The use of epigenetic modifiers and AAV-based gene therapy are currently close to or into clinical testing.
Challenges include targeting all affected cells before they are lost, establish sufficient but not excessive frataxin levels, avoid systemic or local immune reactions, and allow multiple treatments if needed.

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