Conference: Cellular and Molecular Drivers of RNA Toxicity in Myotonic Dystrophy Type 1 (DM1)

DM1 is the most common form of muscular dystrophy, caused by an expanded CTG repeat in the DM protein kinase (DMPK) gene. The nuclear retention of the mutant mRNA leads to RNA toxicity, particularly in myocytes. Muscle wasting and weakness, and fatigue are the most problematic symptoms for DM1 patients. Respiratory failure and cardiac arrhythmias are the leading cause of death in DM1. Our ultimate goal is to elucidate critical drivers of pathologies associated with RNA toxicity and to identify potential therapy targets that may mitigate morbidity and mortality associated with DM1.

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Lunes 20 de junio, 9.45
Salón de Grados, Facultad de Farmacia UV (click in the map)

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Organizes:

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About the speaker:
A physician-scientist interested in human genetics, medical research and translational medicine. He was one of the discoverers of the mutation that stems DM1 and helped establish the concept of RNA toxicity. He is the Head of the Division of Clinical Genomics, the Medical Director of the Molecular Diagnostics Laboratory.