UNIVERSITÀ
DI TRENTO
Dipartimento di
Biologia Cellulare, Computazionale e Integrata - CIBIO

CIBIO EXTERNAL seminar

JUNE 21ST at 3 p.m. Room B109, Povo 2

ZEBRAFISH AVATARS: TOWARDS DIAGNOSIS AND PERSONALIZED THERAPIES FOR RARE DISEASE PATIENTS

Victor Mulero

Dpto. Biología Celular e Histología / Facultad de Biología - Universidad de Murcia
Instituto Murciano de Investigación Biosanitaria Pascual Parrilla (Murcia Biomedical Research | Institute Pascual Parrilla)
Centro de Investigación Biomédica en Red de Enfermedades Raras (Spanish Biomedical Research Center for Rare Diseases)

Between 6,000 and 8,000 different rare diseases affect an estimated 30 million people in the EU. Most of RD debut in childhood and are chronic and disabling diseases, many of them degenerative, considerably reducing quality and life expectancy. The average estimated time that elapses between the appearance of the first symptoms and the achievement of the diagnosis is 5 years. In one in five cases, 10 or more years elapse before the correct diagnosis is achieved. The delay in diagnosis has various consequences: not receiving any support or treatment (40.9%), receiving inadequate treatment (26.7%) and worsening of the disease (26.8%). Our research group have developed a unit within the University of Murcia and the Murcia Institute for Biomedical Research (IMIB) led by Drs. Mulero and Cayuela dedicated to the diagnosis and development of personalized therapies for patients with RD (ZEBER). ZEBER uses a unique model, the zebrafish, which makes it possible to quickly and cheaply identify the pathogenic mutations that cause the RD and the development of personalized treatments. Zebrafish constitutes an ideal platform to discover the biological significance of new variants in genes, as well as in the search for orphan drugs by reversing a specific phenotype. This will be illustrated with a patient with type I interferonopathy caused by a gene encoding a nuclear corepressor.

APTAMERS AS THERAPY FOR CONGENITAL NEUTROPENIA DISEASES

Maria Luisa Cayuela

Instituto Murciano de Investigación Biosanitaria Pascual Parrilla | Centro de Investigación Biomédica en Red de Enfermedades Raras
Hospital Clinico Universitario Virgen de la Arrixaca (University Hospital Virgen de la Arrixaca)

Dyskeratosis congenita (DC) is a rare disease caused by mutations in telomerase complex or telomeric proteins. Mutations in telomerase are part of the genes panel that cause bone marrow disorders. This enzyme consists mainly of a protein component (TERT) and an RNA component (TERC). The mutation of Telomerase RNA component, TERC, causes myelodysplastic syndrome and even acute myeloid leukemias. Recently and thanks to the use of the zebrafish model, we have shown that TERC has a fundamental role in the formation of the myeloid lineage and this new function is exerted independently of that performed at the telomeres. We now know that TERC regulates the expression of the major genes controlling myelopoiesis by binding to regulatory regions and recruiting the transcription machinery to their promoters. In-depth knowledge of TERC's mechanism of action has allowed us to design aptamers (small oligonucleotides) that mimic the behavior of the entire Telomerase RNA (TERC) molecule, providing an alternative to other current drugs for neutropenias and Myelodysplastic Syndrome (MDS).

Contacts
Department of Cellular, Computational
and Integrative Biology - CIBIO
Via Sommarive, 9
38123 - POVO (TN)
comunicazione.cibio@unitn.it