CMX Seminar

Leveraging zebrafish to understand congenital brain and muscle disorders

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Dept of Neuroscience and Cell Biology
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August 21, 2024 (Wed) 14:00~15:00

Shinryokaikan Memorial Hall A, Medical C3 Commons 4F

The main goal of Dr. Manzini's research is to bridge the genetics and mechanisms of disease to identify genes that are essential for human brain development and to define the molecular mechanisms underlying neurodevelopmental disorders focusing on autism, intellectual disability, and neuromuscular disorders. The Manzini laboratory combines human genetics with molecular, cellular, and behavioral approaches in murine and zebrafish models to link human genetics to cell biology, intracellular signaling, and behavior.

Her seminar will focus on the most severe forms of congenital muscular dystrophy (CMD), the dystroglycanopathies, that are also associated with heterogeneous brain and eye malformations. Dystroglycanopathies are caused by mutations in several genes that converge on the synthesis of a specialized glycan on a-dystroglycan (a-DG), a transmembrane protein required for tissue differentiation and maintenance. The goal of her research for the past 15 years has been to identify the genetic causes of dystroglycanopathies and other severe muscle conditions also affecting the brain. Her laboratory then uses zebrafish to recapitulate the human mutations to understand the role of these genes in the muscle, eye, and brain and to develop novel therapies for different disease presentations. She will present recent advances in understanding the pathogenesis of these disorders leveraging the power of zebrafish models.

Sponsored by: Center for Medical Transformation (CMX)

In charge: Department of Physiology and Cell Biology (Professor Toru Takumi)

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