UNIVERSITÀ DI PAVIA

Dipartimento di Medicina Molecolare

Postdoc position

2D and 3D modelling of neurodevelopmental diseases using patient-derived iPSCs

The project builds on the large cohort of iPSC lines produced from patients with Joubert syndrome (JS), a neurodevelopmental disorder characterised by a common cerebellar malformation and associated with mutations in a high number of causative genes. iPSC lines derived from patients will be differentiated and compared to healthy and isogenic controls to identify the effect of specific human mutations on neural lineage formation. The approach includes neuronal differentiation in monolayer format and the production of 3D cerebellar organoids. PiggyBac vectors will be used to compare the effect of WT and mutated versions of the genes of interest on cell phenotype, and the resulting cell phenotypes will be characterised through single-cell sequencing and advanced cell imaging approaches.

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To apply: https://pica.cineca.it/unipv/medmol-2023-b08/

Key points:

- **Patient cell reprogramming to iPSCs**
- **Neuronal differentiation in 2D and 3D organoids**
- **Genetic modification to study the effect of patient mutations**
- **Single cell transcriptomics and advanced microscopy**
- **Collaboration with Italian and European research groups**