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

17 MARCH

4 P.M.
ROOM B107 - POVO 2

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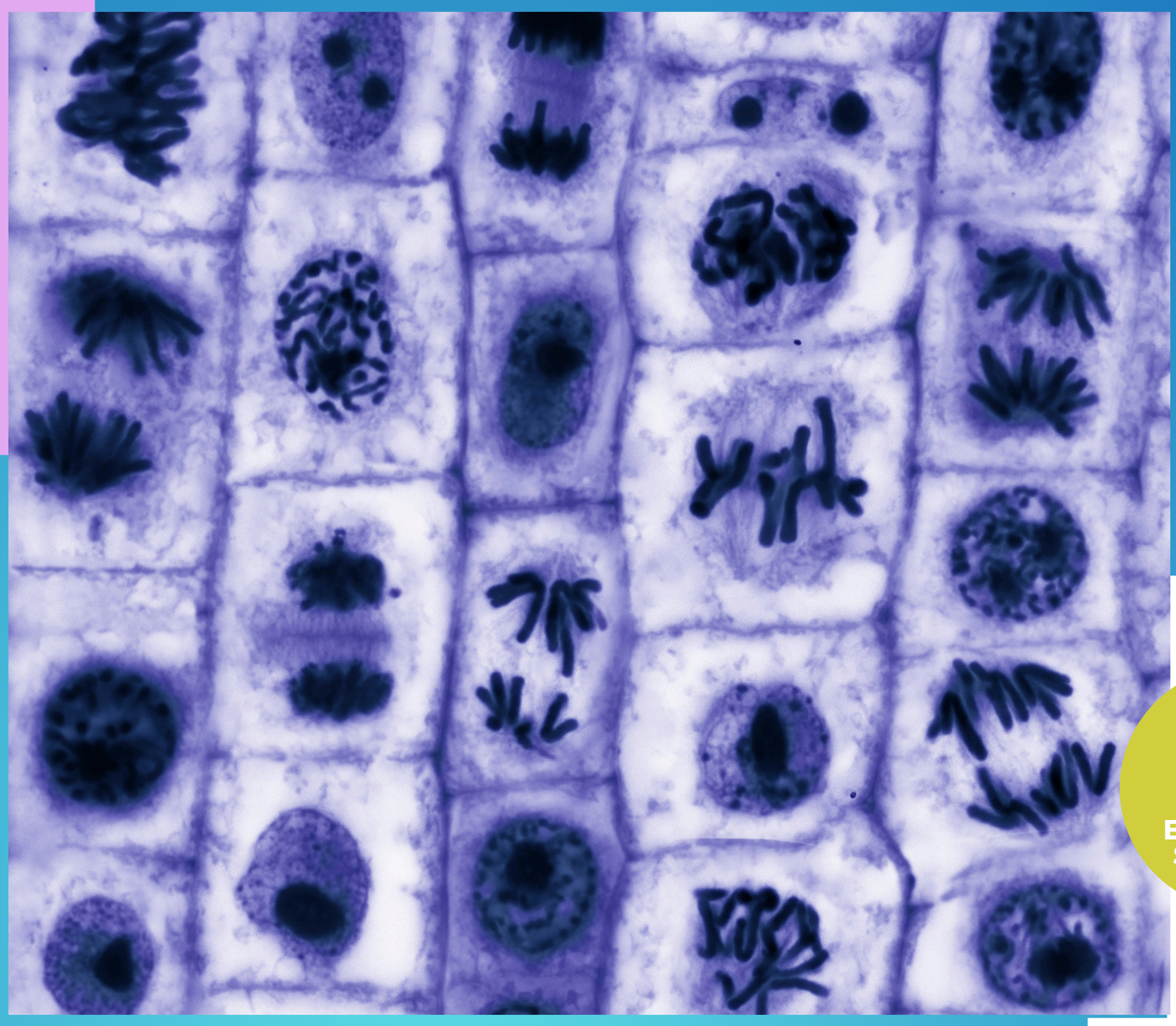

EXTERNAL SEMINAR

PREVENTING AMYOTROPHIC LATERAL SCLEROSIS: RISK, RESILIENCE AND PRESYMPTOMATIC BIOLOGY

Monogenetic adult-onset neurodegenerative diseases have been considered **deterministic**. In genetic forms of **Amyotrophic lateral sclerosis** (ALS, also known as motor neuron disease, MND), incomplete and family-specific penetrance indicates that pathogenic rare variants are **insufficient** to explain disease expression. This implies a more complex biological architecture – in which rare variants establish **vulnerability**, whilst common genetic variants, ageing and broader systemic factors determine **when compensatory mechanisms fai**l. Better understanding of the determinants of risk, prediction of who will develop ALS and the timing of disease onset is essential to preventing ALS **as targeted genetic therapies become a reality**.

In this seminar, I will describe work to understand the drivers of ALS risk **including common and rare variant interactions** and **systemic metabolic factors**. I will review **longitudinal biomarker trajectories** that are refining prediction of ALS onset and discuss necessary future developments to enable **prevention of ALS**.