# PROJECT ABSTRACTS – PHD CALL: SEPTEMBER 2026

## a. Laboratory for Targeted Therapy of Autoimmune Diseases (Dr. Luca Perico)

Area of Research: **Molecular mechanisms of ageing, mitochondrial biology, inflammation and senescence.**

Ageing is associated with progressive mitochondrial dysfunction and chronic low-grade inflammation across multiple organs, but the molecular links between these processes remain incompletely understood. This project will investigate whether the age-associated release of mitochondrial RNA (mtRNA) into the cytosol drives chronic inflammation, cellular senescence, and progressive heart and kidney dysfunction. Its main objective is to establish a mechanistic link between mitochondrial dysfunction, cytosolic mtRNA accumulation, and age-related organ damage. The work will combine molecular and cellular studies in cardiomyocytes and renal tubular epithelial cells with analyses of tissues from ageing mice to identify actionable therapeutic targets. Innovative strategies will also be evaluated to preserve mitochondrial integrity, promote cytosolic mtRNA degradation, and selectively eliminate senescent cells. The expected outcome is the development of new approaches to limit inflammaging, preserve cardiac and renal function, and promote healthy ageing.

## b. Center for Human Genetics (Dr. Marina Noris)

Area of Research: **Integrative Multi-Omics and Patient-Derived Models to Elucidate Disease Mechanisms in Undiagnosed Rare Disorders.**

By integrating multi-omics data with patient-derived models, the project aims to uncover pathogenic mechanisms and advance precision medicine for undiagnosed rare diseases. Despite the widespread use of Whole-Exome Sequencing (WES), many patients with rare diseases remain without a definitive molecular diagnosis, highlighting the need for complementary functional genomics approaches. This PhD project will



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investigate unresolved cases by integrating transcriptomics, spatial transcriptomics, and functional validation. RNA sequencing of patient-derived fibroblasts will be used to identify aberrant splicing, gene expression dysregulation, and the functional consequences of non-coding variants or variants of uncertain significance. Spatial transcriptomics will then be applied to human tissue samples to combine gene expression with cellular and histological context, enabling the identification of disease mechanisms, biomarkers, and potential therapeutic targets. Finally, candidate genes and pathways emerging from these multi-omics analyses will be functionally validated using patient-derived cellular models to investigate the molecular and cellular consequences of candidate variants and their contribution to disease pathogenesis.

Preferred academic and scientific backgrounds: Master's degree in biology or biotechnology. Requested expertises includes molecular biology, cell culture, DNA and RNA processing.

### c. Laboratory of Traumatic Brain Injury and Neuroprotection (Dr. Elisa Zanier)

Area of Research: **Translational research on chronic sequelae of acute brain injury.**

Traumatic brain injury (TBI) can cause cognitive, memory, emotional, and physical disturbances, which can persist or even worsen years after the initial event.

As such, TBI represents a leading risk factor for neurodegenerative disease, with unique potential to contribute mechanisms leading to development of Alzheimer's disease related dementias (ADRD).

To understand the transition from TBI to ADRD, there's an urgent need for animal models mirroring human conditions. Many existing models only capture some TBI-ADRD features, and the progression of these characteristics remains poorly defined. In response to this need, the project aims at developing a TBI-ADRD model exploiting genetic diversity, integrating neuroinflammatory factors, and incorporating major contributing factors to progressive polyproteinopathy.