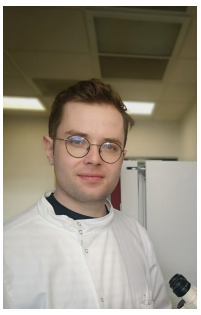
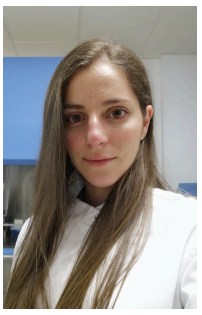
5th April 2022 | 4PM CET VIRTUAL

iPSZÜRICH

A LECTURE SERIES FOCUSED ON INDUCED PLURIPOTENT STEM CELLS

funded by a GRC Grant, UZH





Maria Georgious

Rob Atkinson

Newcastle University

Prof. Majlinda Lako's Retinal Stem Cell Research Laboratory, Newcastle University

Why does faulty splicing affect the retina?

Splicing refers to the removal of introns from pre-mRNA. It is an essential step in eukaryotic gene expression carried out by spliceosomes. The heart of the spliceosome contains a group of proteins known as pre-mRNA processing factor (PRPFs). Paradoxically, PRPF mutations cause retinal disease (retinitis pigmentosa) despite their ubiquitous expression.

An international and interdisciplinary team of researchers led by Professor Majlinda Lako used a combination of cellular, molecular, biochemical, and bioinformatics approaches to assess the impact of PRPF mutations on human retinal cells. Human retinal tissue was generated using induced pluripotent stem cells (iPSCs) with mutations in spliceosome gene *PRPF31*. The results show *PRPF31* mutations promote formation of intracellular protein aggregates. These protein aggregates accumulate over time and affect cell survival. Our results highlight the usefulness of iPSCs in modelling and discovering treatments for retinal disease.

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