Institute of Molecular Genetics
of the Czech Academy of Sciences

Regular Wednesday IMG seminar

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Laboratory of RNA Biology

"Characterization and targeted rescue of a novel PRPF31 mutation in retinitis pigmentosa"

Retinitis pigmentosa (RP) is an inherited retinal disease characterized by photoreceptor loss. Mutations in the PRPF31 gene cause approximately 10% of cases of autosomal dominant RP. In this project, we characterize a novel intronic pathogenic variant in the PRPF31 gene. We provide evidence that the novel RP mutation activates a cryptic splice site, inducing the expression of an abnormal transcript. The resulting protein is unstable, and patient cells exhibit lower expression of PRPF31 and other splicing factors. To correct aberrant RNA splicing, we applied modified antisense oligonucleotides (ASOs) to improve PRPF31 splicing in patient cells. We differentiated the patients' induced pluripotent stem cells (iPSCs) into retinal pigment epithelium (RPE) cells and demonstrated that ASO treatment improves PRPF31 splicing and increases PRPF31 protein levels in patient's RPE cells. These findings suggest that ASO-based splicing correction is a promising therapy for this currently untreatable disease.

The seminar will be held

on Wednesday 25 February 2026 at 15:00

in the Milan Hašek Auditorium at IMG

(Institute of Molecular Genetics of the Czech Academy of Sciences, Vídeňská 1083, Prague 4)