Institute of Molecular Genetics
of the Czech Academy of Sciences

Regular Wednesday IMG seminar

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"Aberrant variants of splicing factor Prpf8 perturb homeostasis of mature cerebellum"

A subset of patients suffering from a familial blindness disorder known as retinitis pigmentosa (RP) carry inborn mutations in a component of the splicing machinery pre-mRNA processing factor 8 (PRPF8), the disease-provoking mechanism are nonetheless unclear. We established two novel alleles of murine Prpf8 that genocopy RP-causative PRPF8 variants. Homozygous mutant animals of both Prpf8 mutant strains developed early-onset neurodegenerative disease that progressively abolished the granule cell pool in mature cerebellum. Transcriptomic analyses thereby indicated perturbations alternative spicing of protein-coding genes, and moreover, altered biogenesis and/or mis-splicing of specific subclasses of circular RNAs that might be essential to maintenance of synapses and hence neural vitality. Deregulation of the circular transcriptome by aberrant variants of a core spliceosomal protein may thus represent a novel pathological mechanism that can derail cellular homeostasis in splicing factor-linked disease.

The seminar will be held on-line at

https://cesnet.zoom.us/j/95531377591

on Wednesday 26th January 2022 at 15:00

The seminar can also be attended in the Milan Hašek Auditorium at IMG

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

Please note

All participants in the Milan Hašek Auditorium (except the speakers) must have their mouths and noses covered by respirators. All participants are bound to observe the current sanitary regulations and directives established by the Czech Government.