

Summary of Research Program:

My research interests are focused on gene discovery and elucidation of molecular pathways underlying molecular mechanisms in cognitive decline caused by neurodegeneration and protein aggregation disorders. My current research projects involve both Mendelian (X-linked Parkinson and Spasticity) and complex disorders (such as Alzheimer's disease).

We analyze familial forms of diseases using exome sequencing and linkage mapping approaches. Candidate variants are validated using live patient cells and post-mortem tissue. Further functional insights are gained by generation of model cell lines and/or transgenic mice. The strategy allowed uncovering various disease mechanisms central to pathogenesis: protein misfolding and aggregation, deletion/duplication of genic regions and tissue-specific alternative splicing.



Olena Korvatska, PhD

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