NATIONALT VIDENSCENTER FOR DEMENS

# Neurogenetics

Read about DDRC's research projects on neurogenetics.

## Huntington's disease

In a longitudinal study we are following a large cohort of Huntington's disease gene mutation carriers and family controls. We are exploring endophenotypes, especially neuropsychological and neuropsychiatric changes and their relation to biomarkers in serum, CSF and by applying different imaging modalities.

Further we have generated patient-derived induced pluripotent stem cells (iPSCs), which are differentiated into relevant neurons, astrocytes and microglia to explore cellular pathogenic pathways.

**Contact**
Consultant neurologist, clinical professor, research director Jørgen E. Nielsen
Joergen.Erik.Nielsen.01@regionh.dk (Please note that this email service is not encrypted.)

## Hereditary spastic paraplegias and spinocerebellar ataxias

A focus area is phenotype/genotype correlations and in spinocerebellar ataxia type 2 (SCA2) especially the generation of patient-derived iPSCs and their isogenic controls. From iPSCs we are establishing hindbrain neurons/organoids to functionally clarify the role of mitochondria and calcium signaling as well as autophagocytic processes in the pathogenesis of SCA2.

**Contact**
Consultant neurologist, clinical professor, research director Jørgen E. Nielsen
Joergen.Erik.Nielsen.01@regionh.dk (Please note that this email service is not encrypted.)

NATIONALT VIDENSCENTER FOR DEMENS

## Inherited frontotemporal dementia and Alzheimer's disease

In a large Danish family with early-onset FTD a pathogenic variant was identified in the *CHMP2B* gene. Encompassing six generations and more than 500 members, the kindred constitutes a unique resource for studying genetic FTD.

The mutation causes c-terminal truncation of the CHMP2B protein, which is involved in intracellular transport and sorting of autophagocytosed material. In addition, studies indicate a neuroinflammatory component in the degenerative process in the brain of deceased patients as well as in transgenic mice.

We have generated patient-specific iPSC-derived neurons and microglia as well as isogenic controls from FTD and AD patients to further study autophagy and inflammation by applying high resolution microscopy, co-culturing experiments cross-relating to markers in serum and CSF.

**Contact**
Consultant neurologist, clinical professor, research director Jørgen E. Nielsen
Joergen.Erik.Nielsen.01@regionh.dk (Please note that this email service is not encrypted.)

Latest update: 14. December 2020