14.06.2023

Curriculum Vitae

1. Contact data

- Work address: Neurogenetics Clinic & Research Lab, Danish Dementia Research Centre (DDRC), Department of Neurology, Rigshospitalet, Copenhagen University Hospital, Inge Lehmanns Vej, section 8025, 2100 Copenhagen, Denmark.
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2. Education

- MD 1988, University of Copenhagen.
- PhD 1998, University of Copenhagen.
- National Board of Health of Denmark certified specialist in neurology 2002.

3. Recent positions

- 2005-present: Consultant in Neurology at Department of Neurology, Rigshospitalet.
- 2002-2004: Clinician scientist fellowship awarded by the Novo Nordisk Foundation.
- 2005-2006: Clinician scientist fellowship from the Danish Alzheimer Research Foundation.
- 2007-2017 awarded two successive clinician scientist fellowships from the Novo Nordisk Foundation.
- 2016-2018: Clinical associate research professor at Department of Neurology, Rigshospitalet and Department of Clinical Medicine, University of Copenhagen.
- 2018-present: Clinical professor of Neurology with special focus on Inherited Neurodegenerative Diseases at Department of Clinical Medicine, University of Copenhagen, and Department of Neurology, Rigshospitalet.

4. Scientific focus areas

- Clinical and paraclinical aspects including molecular genetics and functional molecular biology of inherited neurodegenerative disorders.

  The exact disease mechanisms of most neurodegenerative conditions are still unknown, and effective treatments are lacking. We have built up a broad range of resources that can provide a paradigm for translational research into neurodegeneration by elucidating common disease mechanisms in rare genetic disorders, especially frontotemporal dementia linked to chromosome 3 (FTD3), spinocerebellar ataxia type 2 (SCA2), and Huntington's disease (HD); diseases with clinical and genetic overlaps and common pathologies with e.g., Alzheimer's disease (AD) and Parkinson's disease (PD).
  We discovered and characterized a mutation in the *CHMP2B* gene causing FTD3. We have shown that CHMP2B mutations affect key degradative pathways in the cell – the autophagy and endosome-lysosome pathways. These pathways are critical for neuronal function and survival and have been implicated in e.g. AD. Defining these pathways will therefore extend well beyond understanding FTD in these rare families and we expect

them to be of fundamental importance to understanding pathways of neurodegeneration and developing new therapeutics for these diseases.
We have recapitulated patient specific brain pathology in fibroblasts and subsequently established proof of principle for a knockdown approach for therapeutics. We have established overlapping druggable cellular phenotypes in patient SCA2 fibroblasts also, and we have expanded our mechanistic studies to more humanized models, namely induced pluripotent stem cell (iPSC)-derived neurons and glia from FTD3, HD and SCA2 patients including gene and drug therapeutic approaches. Our most recent data indicate neuroinflammation to be involved in both FTD3 and HD, which we will investigate further using plasma and cerebrospinal fluid markers as well as iPSC derived neurons and glia. Furthermore, we have recently found increased frequency of Th17.1 cells in the cerebrospinal fluid of HD gene-expansion carriers - which will be an efficacy biomarker for the worldwide first immunomodulatory study in presymptomatic HD, planned to start in our clinic in 2024.

5. International relations

- The international, multidisciplinary Frontotemporal Dementia Research in Jutland Association (FReJA consortium) was established over two decades ago to investigate frontotemporal dementia linked to chromosome 3 (FTD3), which occurs in a large western Jutland FTD family. The team is world leaders in this area with a proven track record (see list of publications). The laboratory studies in the UK are led by Professor of Neurodegenerative Disease, Adrian Isaacs, UCL Queen Square Institute of Neurology. London, UK, and I am heading the studies in Denmark.
- Danish principal investigator in SPATAX, a European research society on inherited ataxia and paraplegia.
- Member of the Steering Committee and adviser for REGISTRY in the European Huntington Disease Network, 2007-2017.
- Danish coordinator of Enroll-HD, which is a worldwide observational study for Huntington's disease.
- Member of European Academy of Neurology, scientific panel, Neurogenetics, appointed by the Danish Neurological Society.

6. Scientific supervision

- Supervision of 15 completed and 4 ongoing PhD projects. Opponent on 7 PhD theses.

7. Management experience and boards

- From 2005, established and expanded the clinical neurogenetics function as part of DDRC, Rigshospitalet. The Neurogenetics Clinic is the only clinic in Denmark offering families and patients with inherited neurodegenerative disorders an in-house combined service encompassing highly specialized diagnostic work-up, genetic counselling, psychological and psychiatric evaluation, and follow-up as well as the translation between clinic and research in the area.
- Research director of the Neurogenetics Clinic & Research Lab, DDRC, (2 post docs, 4 running PhD's and 1 lab. technician).
- PI of several clinical trials in HD

- Member of the NeuroCluster board at University of Copenhagen, 2005-2010.
- From 2014-2020. Appointed by the Neuroscience Centre as a member of the Research Board at Rigshospitalet.
- From 2019-: Representative and member of the board of the European Reference Network - Rare Neurological Diseases (ERN-RND)
- From 2019-: Danish National Genome Center: Successful nomination of neurogenetic patients for whole genome sequencing. Appointed by the Danish Neurological Society as representative in the working group for Clinical application of whole genome sequencing and in 2021 appointed by The Capital Region of Denmark as member of the Specialist network for neurogenetic patients.

8. Funding

- Personal funding: From private foundations: DKK >40.000.000.
  Consortium projects: DKK >60.000.000.

9. Reviews

- The Wellcome Trust, Journal of Neurology, Neurosurgery & Psychiatry, European Journal of Human Genetics, Acta Neurologica Scandinavica, European Journal of Neurology, Annals of Neurology, Neurogenetics, Cerebellum, Human Molecular Genetics, Cell Stem Cell.

10. Communication

- More than 100 invited national and international lectures. Frequent community-based research communication e.g., as lectures, radio interviews and television communication.

11. Publications

- Number of original peer-reviewed publications: 168 (7 as first author, 108 as co-author and 53 as senior author); 6 reviews; 49 consortium publications; 16 book chapters; >170 abstracts; Google Scholar: h-index 44; citations 7740. Web of Science: h-index 34; citations 4799. ORCID: 0000-0003-0453-5582.