

## **Clinical genetics of tauopathies**

### **Z. Wszolek**

*Neurology, Mayo Clinic Florida, USA*

A discovery of *MAPT* gene mutations in 1998 opened the road to understanding of pathophysiological implications of tauopathies. The list of tauopathies is growing and includes progressive supranuclear palsy, corticobasal degeneration, Pick disease, chronic traumatic encephalopathy, globular glial tauopathy, argyrophilic grain disease, primary age-related tauopathies, and frontotemporal dementia with Parkinsonism linked to chromosome 17. Alzheimer disease, the most common neurodegenerative disorder is considered to be both amyloidopathy and tauopathy. Many of the newer tauopathies can be diagnosed only on pathological grounds. Some of them are clearly the genetic disorders usually transmitted in autosomal dominant fashion. As a matter of fact the younger the symptomatic disease onset is, the higher the chance that this tauopathy is of inherited form. Interestingly in about a half of autopsied *LRRK2* gene mutation carriers, the tau pathology is present. The *LRRK2* Parkinson disease (PD) is the most common form of genetic PD described so far. The carriers of *LRRK2* mutations usually clinically present with classic PD phenotype, and their illness is usually well responsive to medical and surgical therapies utilized in PD. During my lecture I will discuss the clinical presentations of common tauopathies and their genetic forms.