Clinical aspects of PKAN and possible treatment based on deep brain stimulation.

PKAN is one of the most common forms of NBIA, accounting for approximately half of all NBIA cases. Traditionally, PKAN patients have been divided into typical and atypical according to age at onset, but the clinical spectrum represents a continuum. Dystonia, spasticity, tremor, parkinsonism are the major motor features of the disease, in motor symptom are mainly represented by cognitive impairment, psychiatric disturbances and retinopathy; epilepsy may occur at later stage of the disease. The course of the disease is progressive, with a more rapid evolution the earlier the onset of symptoms. A dramatic deterioration (dystonic state) is a not uncommon and life-threatening complication. Treatment involves both pharmacological and surgical approaches. Pallidal deep brain stimulation is commonly used as a treatment for dystonia; however, the response is not always satisfactory and new surgical techniques and targets have been explored in recent years.

The clinical features of a large single-centre case series of paediatric and adult PKAN patients with a long follow-up will be illustrated, with a focus on unusual or peculiar clinical aspects and on the outcome after surgical treatment including different approaches.