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NEUROPSYCHIATRIC PHENOTYPE OF BETA-PROPELLER PROTEIN-ASSOCIATED NEURODEGENERATION (BPAN)

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Introduction: Neurodegeneration with brain iron accumulation (NBIA) comprises an array of progressive brain disorders presenting with neurological and psychiatric symptoms, sometimes accompanied by intellectual impairment and behavioural problems. Clinically, according to time of onset and deterioration gradient, the NBIAs can be subdivided into two major groups: early onset / rapid, and late onset / slow. To date, nine genetic types of NBIA can be identified.
Recently, a novel NBIA was described showing an additional specific neuroimaging pattern in the substantia nigra together with global cerebral atrophy, originally termed Static Encephalopathy of childhood with Neurodegeneration in Adulthood (SENDA). Shortly thereafter, it became clear that SENDA is caused by *de novo* heterozygous mutations in the WDR45-gene located in Xp11.2 that was named Beta-propeller Protein Associated Neurodegeneration (BPAN).
Objectives: Investigating the neuropsychiatric phenotype of BPAN.
Aims: Diagnostic evaluation of three adult female patients.
Methods: Detailed neuropsychiatric examination of 3 patients with genetically proven BPAN.
Results: All patients had a moderate to severe intellectual disability. In their early thirties autistic-like and depressive symptoms paralleled progressive motor and cognitive decline, ultimately resulting in wheelchair dependency. MRI of the brain demonstrated significant iron deposition in the globus pallidus and the substantia nigra as well as cerebral atrophy. Dystonia of the extremities and parkinsonian symptoms were most prominent. Treatment with levodopa/carbidopa was not effective.
*Conclusion:*The neuropsychiatric phenotype of BPAN is characterized by symptoms from the autistic and anxiety spectrum that, however, should be functionally interpreted as resulting form motor deterioration and severe cognitive inflexibility.

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