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HALLERVORDEN-SPATZ SYNDROME

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Key-word: Brain, abnormalities

Background: A 28-year-old man was referred to the neurology department of our hospital with difficulty of social interaction, impairment in carrying out daily life activities and muscle rigidity. He had a history of head trauma 3 years ago. Neurological examination revealed bradykinesia, hypophonic speech, resting and postural tremor, rigidity, spasticity, hyperreflexia and psychosis.

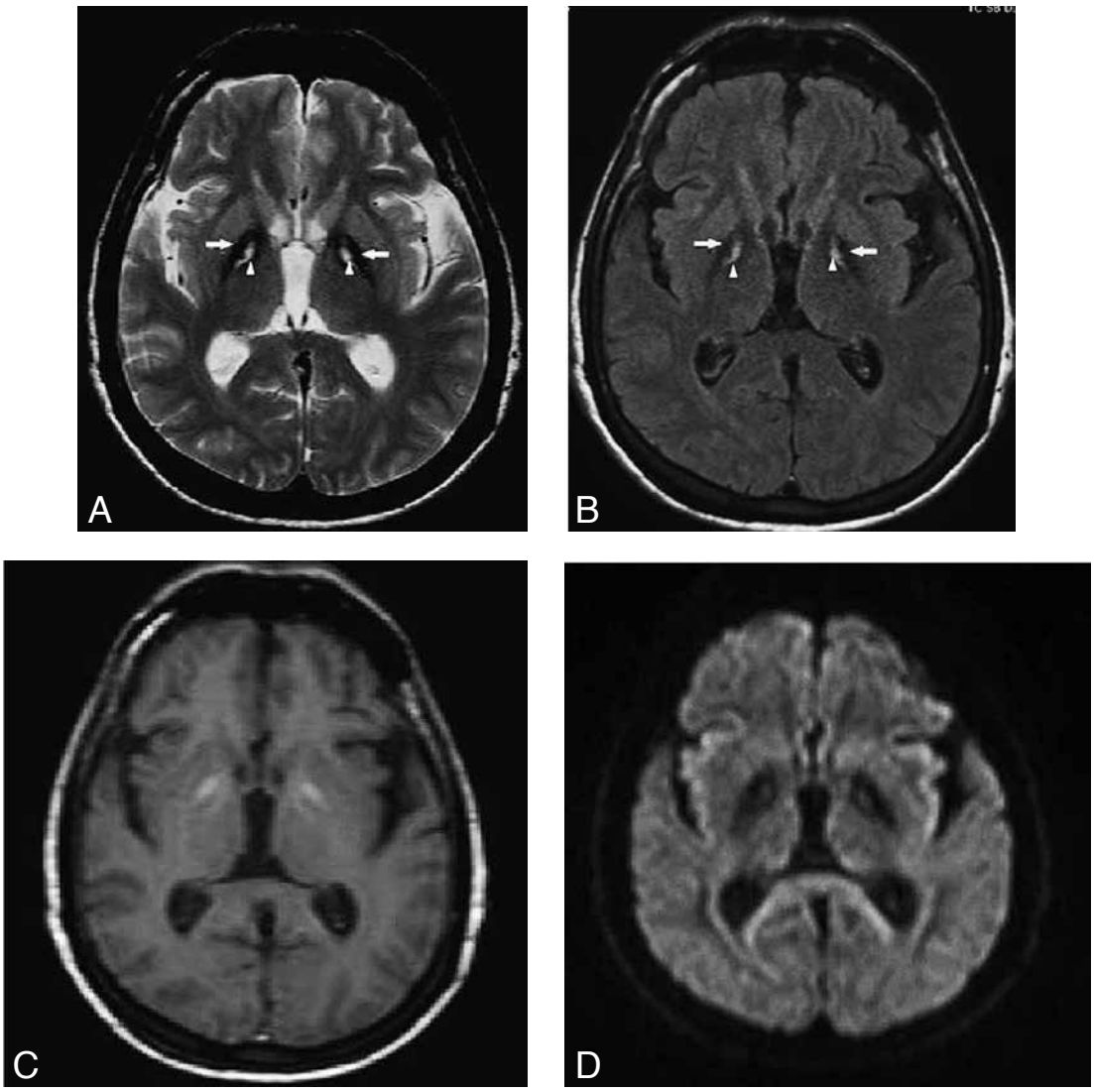

Fig. 1A | 1B / 1C | 1D

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Work-up

MRI examination of the brain (Fig. 1) included an axial T2-weighted turbo spin echo image (A), an axial FLAIR image (B) which demonstrated the classic "eye-of-the-tiger" sign, a sign that refers to a marked hypointensity of the globus pallidus bilaterally (arrows), containing a small hyperintense area at the center (arrowheads). Axial T1-weighted turbo spin echo image (C) revealed only very mild hypointensity in the globus pallidus with an area of more pronounced hyperintensity at the central part of the lesion. On echo-planar axial diffusion weighted image (D), no restricted diffusion is seen at the level of the globus pallidus.

Radiological diagnosis

Based on the clinical findings (extrapyramidal symptoms) and the characteristic "eye-of-the-tiger" sign on MRI the diagnosis of *Hallervorden-Spatz syndrome* was made.

Discussion

Hallervorden-Spatz syndrome (HSS) is a rare, autosomal recessive degenerative disorder often characterized by childhood onset of progressive symptoms including intellectual impairment and extra- pyramidal signs such as tremor, dystonia and bradykinesia. In our case, the clinical finding is characterized by intellectual impairment as psychosis and progressive extrapyramidal signs.

Hallervorden and Spatz first described the disease in 1922 as a form of familial brain degeneration characterized by iron deposition in the brain. The term neurodegeneration with brain iron accumulation type1 has been used for this condition, and the most recently used term for the same condition is pantothenate kinase-associated neurodegeneration (PKAN). Two clinical forms are classified: early-onset, rapidly progressive disease, and late-onset, slowly progressive disease. Our case concerned the late onset form as the symptoms began 3 years ago and the disease was further slowly progressive. Pathologically, there is neuronal loss and gliosis affecting mainly the internal segment of the globus pallidus and substantia nigra. PKAN is associated with intracellular and extracellular iron deposition in the globus pallidus and substantia nigra. MRI shows bilateral high signal intensity surrounded by a region of low signal intensity on T2-weighted MRI in the medial globus pallidus, creating the characteristic feature of the 'eye-of-the-tiger', which corresponds to the pathological findings. The hypointensity on T2-weighted image is caused of iron deposition while the central hyperintensity is secondary to gliosis, demyelination, neuronal loss, axonal swelling and spongiosis.

Individuals exhibiting any of the above listed symptoms are often tested using MRI for a number of neuro-related disorders. As PKAN is a disease prominently evident in the brain, MRI is very useful for diagnosis. Radiological studies have demonstrated pathological levels of iron deposition in the globus pallidus and pars reticulata of the substantia nigra in patients diagnosed with this syndrome. Other disorders affecting basal ganglia are Leigh's disease, mitochondrial encephalopathies, infantile bilateral necrosis and Wilson's disease. These diseases more frequently affect the putamen rather than the globus pallidus. The other diseases of iron deposition in the basal ganglia and "eye-of-the-tiger"-sign include aceruloplasminemia and neuroferritinopathy.

There is no cure of PKAN. Most pharmacologic treatments are symptomatic. Iron chelating agents have been used somewhat successfully in retarding the disorder. The patient in the presented case was treated with dopaminergic, antipsychotic therapy and physiotherapy.

Hallervorden-Spatz disease is diagnosed by its specific brain MR imaging changes, even in the very early stages in the disease, it is distinguished from other diseases by history and clinical findings.

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