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Short Communication

Neurodegeneration with Brain Iron Accumulation (NBIA) in a Child

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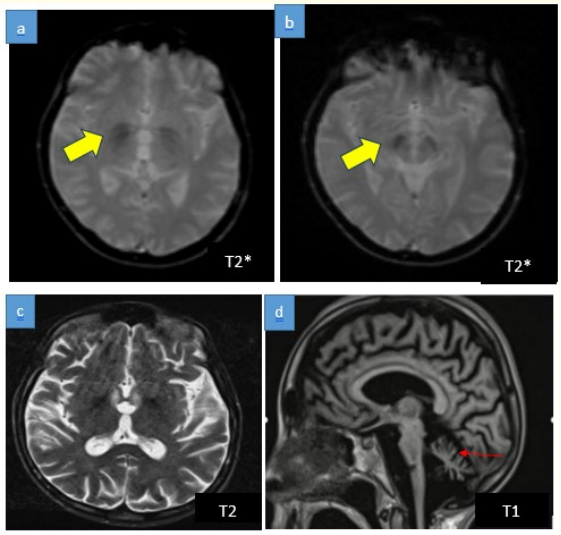
Neurodegeneration with brain iron accumulation (NBIA) is a group of hereditary disorders characterized by abnormal iron accumulation in the basal ganglia, mainly in the globus pallidus (GP) and substantia nigra (SN) [1]. Several autosomal recessive NBIA syndromes can present during childhood, with pantothenate kinase-associated neurodegeneration (PKAN; caused by mutations in the PANK2 gene) and phospholipase A2 group 6-associated neurodegeneration (PLAN; linked to genetic defects in PLA2G6) being the most common [2].

We present the case of a 14-year-old child with psychomotor developmental delay since birth (slowed acquisition of developmental milestones), who later developed progressive weakness, loss of independent walking at the age of 5, as well as dystonia, dysarthria accompanied by ataxia, and ideomotor apraxia (patient is wheelchair-bound). Ophthalmological examination revealed no particular abnormalities.



Figure

For our case, the MRI shows bilateral and symmetrical signal abnormalities of the basal ganglia (pallidum, internal capsules) and substantia nigra on T2* GRE (Figure a and b) and T2 sequences (Figure c and d). Significant atrophy of the cerebellar vermis and hemispheres is a common feature and usually precedes iron accumulation (Figure e, red arrow).

Magnetic resonance imaging (MRI) is sensitive to the presence and concentration of non-heme iron in the brain and is currently the modality of choice for the investigation and differential diagnosis of NBIA syndromes [3].

We find: T1: Three-dimensional T1 images are used to look for changes in brain morphology and atrophy. T1-weighted images are useful for detecting metal deposits such as copper and iron molecular complexes, which appear hyperintense. T2/FLAIR: To assess changes in white matter, gliosis, and cavitations. T2/SWI*: To detect microscopic iron deposits, particularly hypointensities in the bilateral globus pallidus [4].

Biological findings vary depending on the type of NBIA. A genetic study detects the causal genetic mutation, which is the only means of confirming the diagnosis.

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