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STATIC ENCEPHALOPATHY OF CHILDHOOD WITH NEURODEGENERATION IN ADULTHOOD (SENDA) AS CAUSE OF DYSTONIA AND PARKINSONISM

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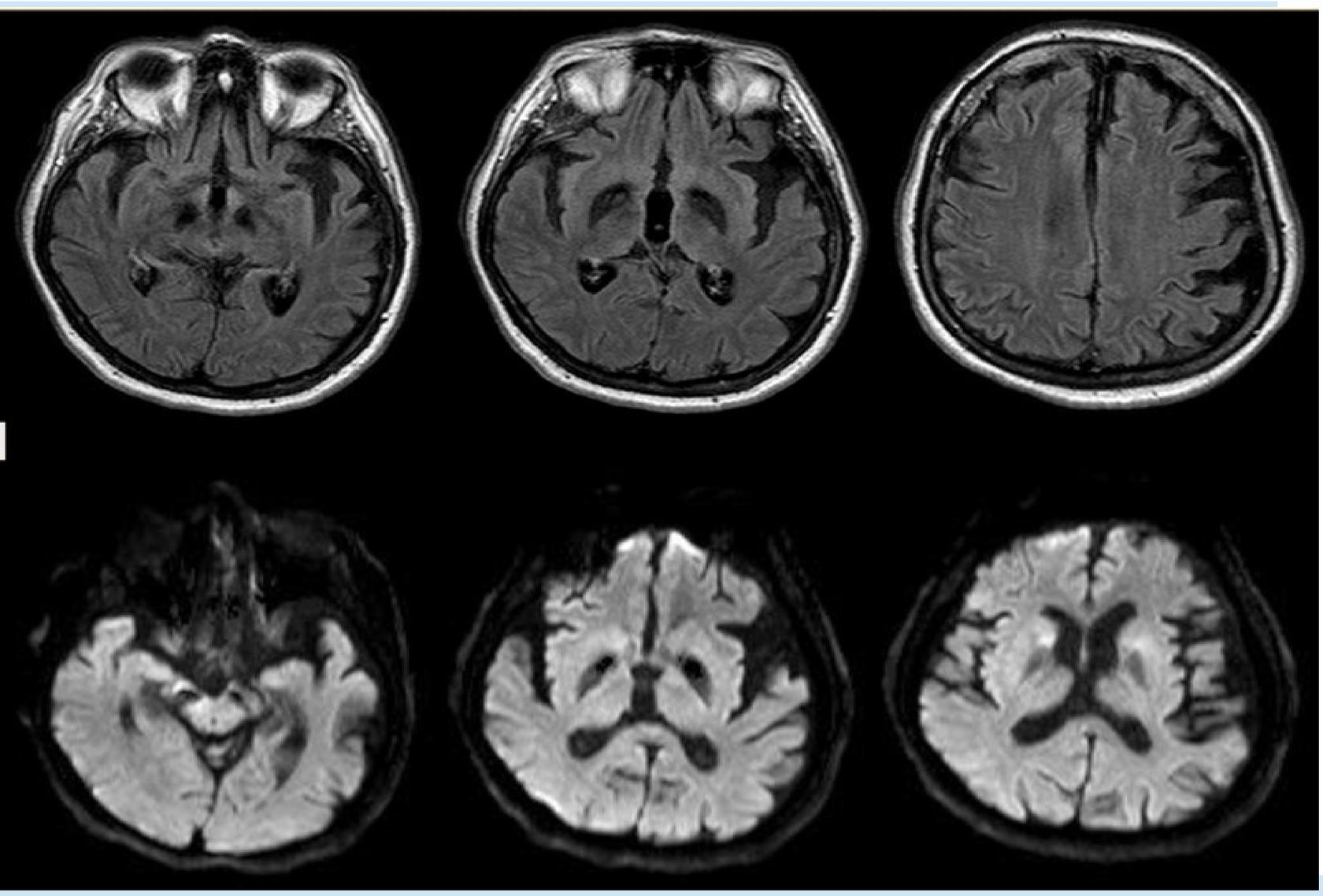
Neurodegeneration with brain iron accumulation **(NBIA**) encompasses a group of neurodegenerative disorders that show radiographic evidence of iron deposition in the basal ganglia.

BPAN (Beta-propeller Protein Associated Neurodegeneration) is a rare X-linked dominant disease that accounts for approximately 1-2 % of all cases of NBIA.
It is associated with mutations in the WDR45 gene, responsible for generating a protein involved in intracellular autophagy.

The aim of this report is to describe clinical and neuroradiological features and time course of a case of BPAN, and to underlie that, even if rare, NBIA must be considered in the differential diagnosis process when evaluating an adult patient with atypical extrapyramidal features.

We report the case of a 60-year-old female with mild psychomotor retardation since childhood, who developed, at age 59, rapid progressive cognitive dysfunctions, severe dystonia of the neck, dysphagia, and parkinsonism with impairment in deambulation, leading the patient bedridden in about a year. The patient had no family history of neurodegenerative disorders.
A detailed diagnostic work-up including complete clinical and laboratory examinations and neuroradiological investigations has been done.



A brain 3T-MRI revealed T2-weighted hypointense signal in the substantia nigra and globus pallidus, consistent with iron deposition and T1-weighted imaging showed hyperintensity of the substantia nigra with a central band of hypointensity (Fig).
Genetical screening for NBIA detected a pathological mutation in *WDR45 gene*, responsible of BPAN named also “static encephalopathy of childhood with neurodegeneration in adulthood” (***SENDA***).

CONCLUSIONS

Our patient had a non-progressive developmental delay in early childhood followed by an extrapyramidal syndrome with very rapid progression, appeared at the end of the fifth decade, much later in life than previous cases reported in literature.

Dystonia and parkinsonism in BPAN do not present exclusively in late twenties/ early thirties early, but can present much later.

Therefore, **BPAN must be considered in the differential diagnosis of adult parkisonisms**.

Further studies focused on WDR45 gene mutations and their role in compromising autophagy could contribute to move toward pathogenesis-targeted therapies and to better understand pathologic mechanisms that lead to human neurodegenerative diseases.

REFERENCES

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