MRI Spectrum of Neurodegeneration with Brain Iron Accumulation: A Case Series of Varied Clinical Phenotypes

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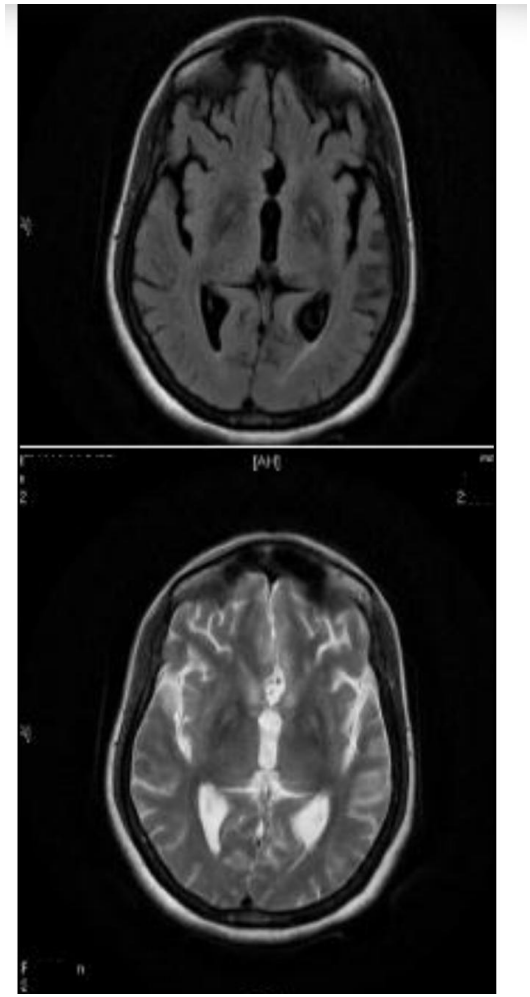
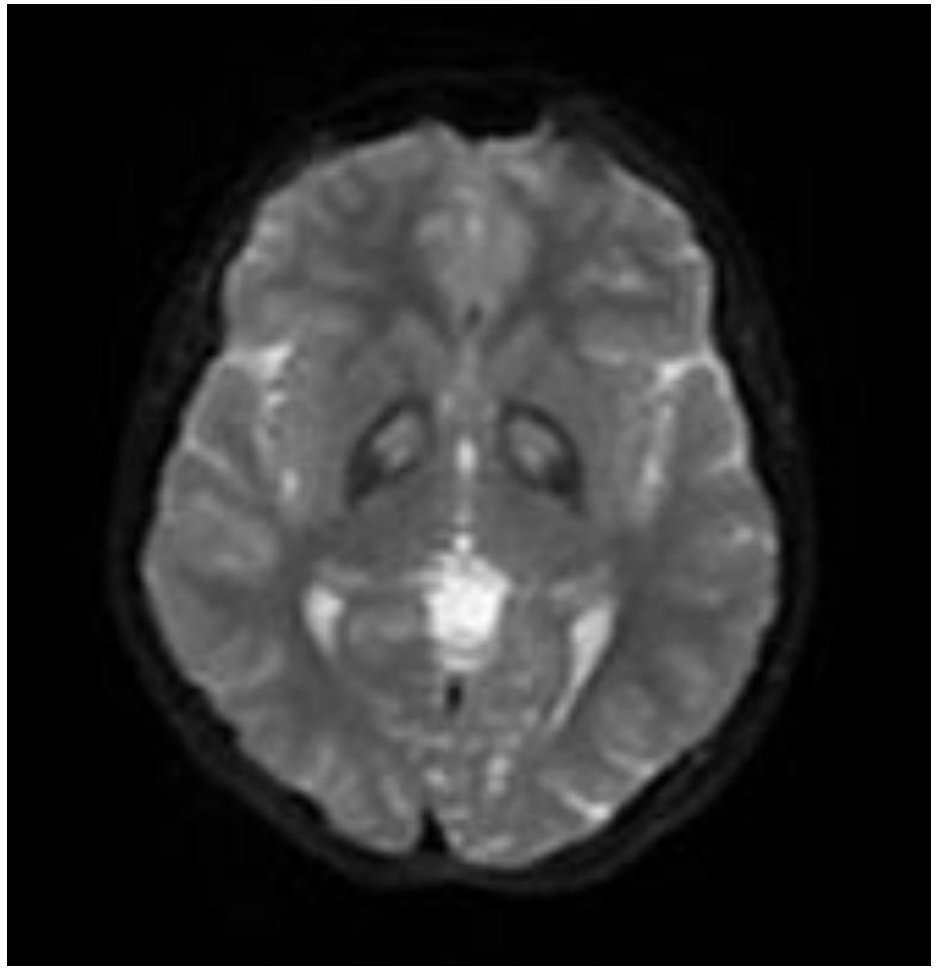
Aim:

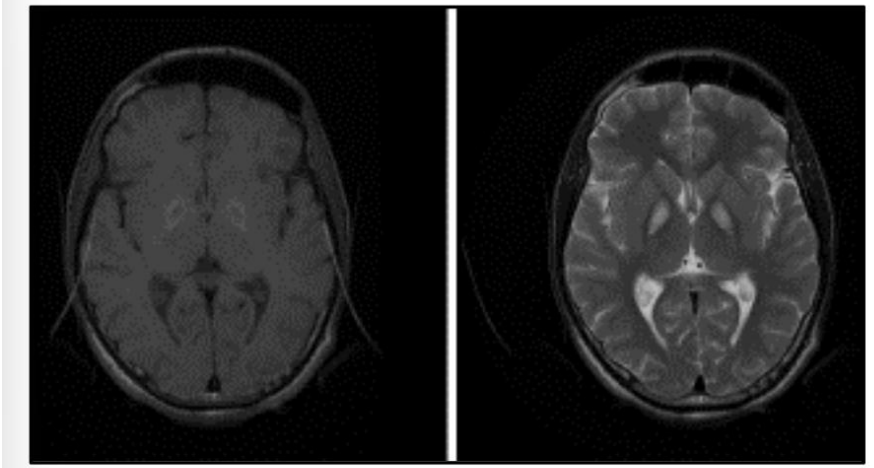
- This study aims to illustrate the diverse clinical presentations and characteristic MRI findings in patients with NBIA.

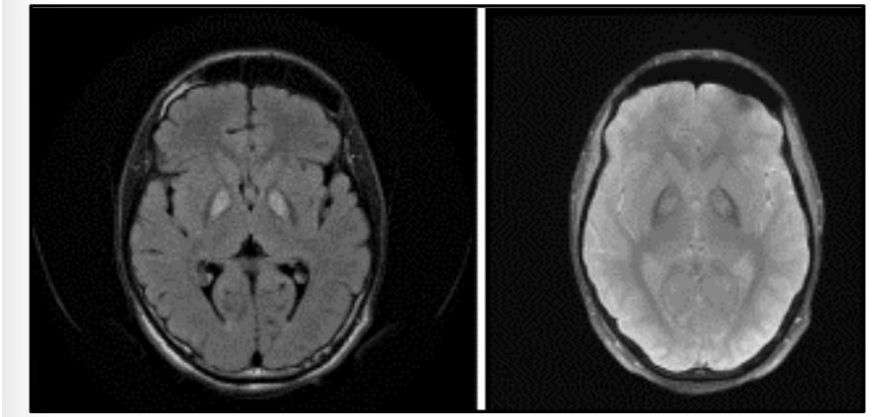
Materials & Methods:

- This is a retrospective descriptive case series of ten patients diagnosed with NBIA based on clinical evaluation and characteristic MRI findings, particularly the “Eye-of-the-Tiger” sign. Detailed history, neurological examination, and MRI brain imaging and genetic testing were performed. Family history and consanguinity were noted to assess potential genetic linkage.









Results:

- The cohort included patients aged 12 to 60 years, with a male-to-female ratio of 1:1. Clinical presentations varied from behavioral abnormalities, psychiatric symptoms, and cognitive decline to extrapyramidal signs such as dystonia, bradykinesia, tremors, and ataxia. Six patients showed the pathognomonic “Eye-of-the-Tiger” sign on MRI. Three patients were from the same family, suggesting genetic inheritance. One asymptomatic case with MRI findings supports pre-symptomatic detection. A patient with Kayser-Fleischer (KF) rings indicated overlapping features with Wilson disease, highlighting diagnostic complexity. Six patients were positive for PKAN mutation. One patient had ADCY5 mutation.

Conclusion:

- NBIA encompasses a spectrum of disorders with heterogeneous clinical phenotypes and characteristic MRI features. Early recognition and imaging play a crucial role in diagnosis, especially in familial and consanguineous contexts. This case series emphasizes the diagnostic value of MRI in guiding clinical suspicion and enabling timely intervention and counseling.