# Juvenile parkinsonism with jerky movements: a case report

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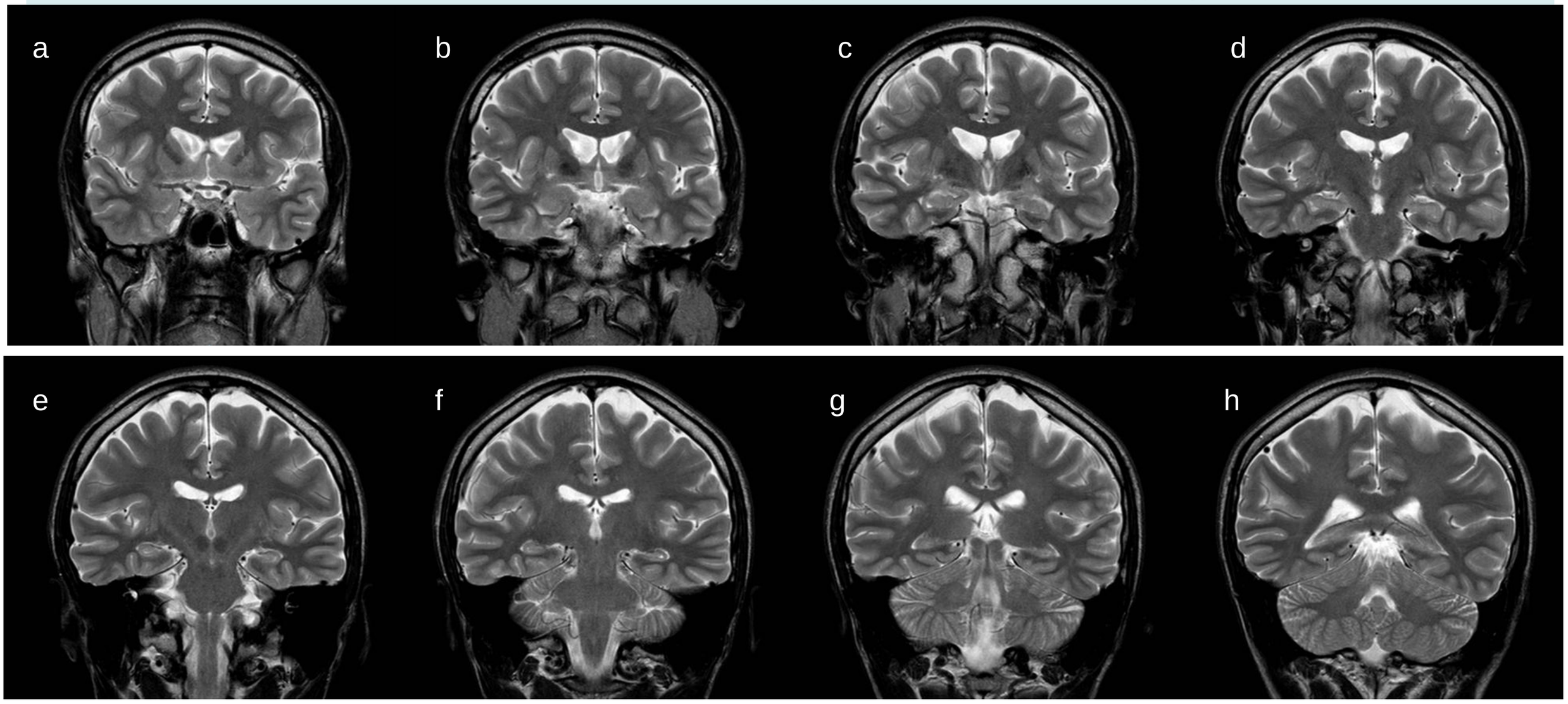
**Introduction:** childhood onset and juvenile movement disorders are rare diseases with great diagnostic complexity and a huge burden for patients and families. Great efforts has been done in these years to discover gene mutations associated with parkinsonism and to identify genotype-phenotype correspondence, as well as response to treatment [1].

**Materials and Methods:** E.N.L. 28 years old, male. Russian nationality.
Since the last month of 2015 the patient manifested postural instability and involuntary, jerky, movements in face and limbs. Relatives also found him to be slower during movements.
In childhood he had vocal tics, which resolved in early adolescents and then reappeared at 15 years, after his father's death. Patient had no burden of these symptoms and seemed unaware of them. His father died in a car crash and was an alcoholic, with some cognitive decline and behavioural disturbancies probably related to his alcohol abuse. The paternal grandfather has been recently diagnosed with a non specified «dementia».
At the neurological examination in january 2017 he did not present cranial nerve alterations, strenght and coordination were normal, muscolar reflexes were brisk and symmetrical. Frequent blinking and sporadic grimaces were observed. While mantaining his arms stretched to the front, he presented jerky movement of hands and fingers. Froment sign was present in both arms but in relaxed condition no rigidity could be found. At finger tapping he presented mild bilateral bradikynesia, with the left arm slightly more involved. Pull test was negative. During walking left arm swing was reduced, the base was slightly wide, but walking in tandem was possible. Hands had frequently twisting movements.
Cognitive evaluation was normal. An acute levodopa challenge test was tried without clinical benefit and the patient refused to try prolonged tretment.
The MRI showed mild bilateral hypointensity in globus pallidus interna and red nucleus in T2 sequencies. A SPECT DAT-scan showed reduced captation bilaterally in basal ganglia.
Blood and urine tests were normal, including copper methabolism, alpha-feto protein, anti-streptolysin O, serum proteins and endocrine functions. Also no signs of autoimmune disorders were found.
Because of the mild parkinsonism with jerky movements of face and hands, combined with imaging findings consistent with iron accumulation in the basal ganglia, we supposed a form of Neurodegeneration with Brain Iron Accumulation (NBIA) [2].
However all those genetic tests turned out to be normal and we re-evaluated the patient in august 2018, finding a mild worsening in gait with a wider base and a more pronounced deterioration in cognitive performance (MoCA score was 22/30, cut off 26). Moreover during the interview his mother described some behavioral abnormalities mainly expressed by self-isolation and apathy.




MRI T2 sequencies: a-h coronal view showing a mild bilateral hypointensity in basal ganglia and a slight enlargement in lateral vetricles.

**Conclusions:** juvenile parkinsonisms are a complex diagnosis. Our first hypothesis of NBIA has been confuted by genetic tests. However the appearance of cognitive and behavioural disturbancies early in the course of the disease and the familial history positive for dementia, could point towards the group of frontotemporal dementias (i.e c9orf72 gene mutations) [3], even in our young patient [4]; those tests are in course, along with analysis of genes commonly related to familial parkinsonisms.

## References

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4 – Murphy NA et al; Age-related penetrance of the *C9orf72*repeat expansion; *Scientific Reports* 7, Article number: 2116(2017).