Different neuropsychological profile in patients with primary familial brain Calcification.

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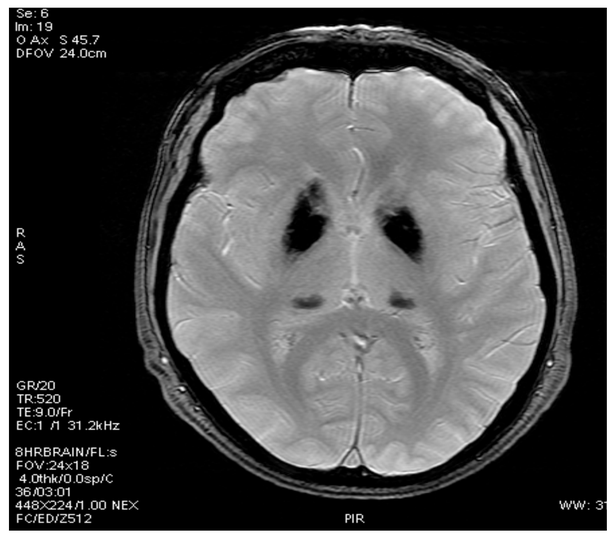
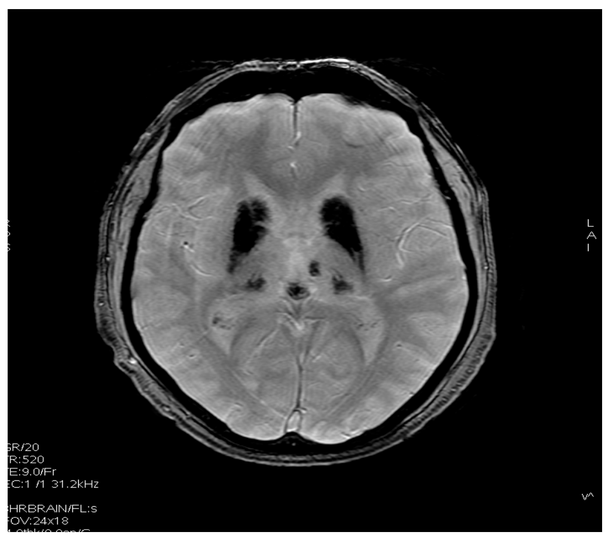
Background: Idiopathic Basal Ganglia Calcification (IBGC) is a rare neurodegenerative disorder of unknown etiology. Also known as Fahr's disease, IBGC is clinically heterogeneous. Patients display a variety of clinical manifestations including movement disorders, cognitive impairment and psychiatric symptoms with diverse severity and ages of onset. In some cases, patients can remain asymptomatic throughout life.

Case description: We reported here a complete neuropsychological assessment of two brothers: G. D. 54 years old (patient 1) and F.D. 51 years old (patient 2) of a family of south Italy in which a novel mutation in SLC20A2 gene was detected (heterozygous deletion c.21_21delG (p.L7Ffs*10). Cognitive functions were evaluated using the Mini Mental State Examination; Token Test, WEIGL'S Test, Colored Progressive Matrices, Stroop Test; Frontal Assessment Battery, Phonemic Verbal fluency. The non cognitive assessment was performed through the Apathy Evaluation Scale, Beck Depression Inventory and Hamilton Anxiety Rating Scale. MRI showed that both patients had the same radiological pattern of bilateral minerals accumulation in the basal ganglia. Despite the evidence of the same gene mutation and radiological findings, the behavioral and cognitive profile were different. Test results, indeed, showed that patient 1 had pathological performances in executive functions (Stroop test and verbal fluency) and mild depression and apathy. Patient 2, otherwise, had no cognitive deficits, but he developed psychiatric symptoms (aggression and anxiety behavior, difficulty in collaborating, reduced insight ability).



PATIENT 1



PATIENT 2

Conclusion: Our cases suggest the existence of a heterogeneity genothype-phenothype for the novel gene mutation in SLC20A2 and that the same mutation can result in a very broad phenotypic variation.

References:
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