Bilateral striopallidodentate calcinosis. A presentation in the form of facial dystonia and frontotemporal dementia

Dear Editor:

In 1860, Delacour was the first to report vascular calcifications in the basal ganglia in a male who presented rigidity, weakness in his lower limbs, and tremor. Although known as Fahr’s disease, this is a misnomer, since this latter author reported a case of calcinosis in the basal ganglia that does not correspond to what we currently understand this term to mean. Since the calcifications exhibit a preference for the basal ganglia and dentate nucleus, it would seem to be more appropriate to refer to it as bilateral striopallidodentate calcinosis (BSPDC).

Calcification of the basal ganglia has been associated with more than 30 diseases, including infections, metabolic disorders, and genetic syndromes. The incidence of calcifications in the basal ganglia in neuroimaging studies is 0.6%, although most of them are very small and generally confined to the globus pallidus. They present clinically as extrapyramidal, cerebellar, and cognitive manifestations, with an inherited autosomal dominant variant in most cases and another, sporadic variant.

Sixty-six year old female, hypertensive, with bradycardia-tachycardia syndrome for which she had a definitive pacemaker. She had not undergone thyroid surgery nor had she received treatment with dopaminergic antagonists. No family history of dementia or extrapyramidal disorders.

The patient presents a bilateral hemifacial spasm for the last 5 years associated with jaw-closing dystonia that was treated with botulinic toxin with clear improvement of symptoms for 4 months. Over the course of the last 3 years, the patient has developed cognitive impairment with apathy, withdrawal, executive dysfunction, and emotional lability with loss of personal care.


Neuropsychological evaluation: mild attention disorder and temporospatial disorientation, mild impairment of reading comprehension, mild deficit of episodic memory and long-term information recall. Frontal dysfunction with perseveration in graphic sequences, highly concrete thinking, limited working memory and sequencing errors in written expression.

Analyses: no alterations of interest in the blood test, coagulation, vitamin B12, folic acid, glucose, electrolytes, kidney function, liver enzymes, thyroid hormones, antinuclear antibodies, and blood proteins. Total and free parathyroid hormone, total calcium, calcium ions, and phosphorus were all normal. Genetic study for Huntington’s disease, negative. X-rays of hands and feet: absence of subchondral bone cysts. Cranial CT (Figure 1): cortico-subcortical retraction with slight frontal predominance. Bilateral calcinosis at the level of the basal ganglia, thalamus, and dentate nuclei of the cerebellum.

The brain is especially well-protected against different toxins thanks to the existence of the blood-brain barrier. However, subcortical nuclei are vulnerable to several different minerals; thus, the accumulation of copper causes Wilson’s disease; the accumulation of iron produces Hallervorden-Spatz’s disease; the accumulation of organic mercury is the cause of Minamata’s disease, and the accumulation of manganese, Parkinsonism. Different disorders produce calcification of the basal ganglia; however, the reason as to why these systemic processes bring about focal deposit in the basal ganglia is unknown. The finding of calcification in the basal ganglia in
neuroimaging study requires that hypoparathyroidism be ruled out, since it is the most common explanation for this finding (70-80%).

A registry with many BSPDC patients has revealed that only 68% of the cases were symptomatic and 32% were asymptomatic. The most common clinical manifestation is movement disorders, present in 55% of the cases. Of these disorders, 57% were Parkinsonism, 19% chorea, 8% tremor, 8% dystonia, 5% athetosis, and 3% consisted of orofacial dyskinesia.

Our patient had a negative clinical family history and one sibling who had been studied for peripheral vertigo did not present calcifications in the basal ganglia on the cranial CT performed, although a hereditary basis cannot be ruled out entirely.

In a neurological study of patients with BSPDC, frontal executive function tests revealed alterations. The cognitive impairment reported by other authors include slowed thinking, poor concentration and attention, and impaired verbal and non-verbal memory with normal language, abstraction, and praxis; as a result, frontal subcortical dementia is thought to be the dementia most commonly detected in these patients.

The presence of frontal subcortical dementia associated with calcification of the basal ganglia forces us to rule out other diseases such as Nasu-Hakola’s disease in which there are subchondral bone cysts. Frontal dementia with calcifications in the basal ganglia has also been described; it is a sporadic form of pre-senile dementia with neurofibrillary tangles, but without senile plaques in Japanese patients and that receives the acronym DTNC. The course of BSPDC is unfavourable, moving toward progressive worsening. Response to different treatments is poor or temporary.

The selective elimination of the calcium deposits in the brain without affecting those in bones and other tissues appears to be an impossible task.

References


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