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Fahr's syndrome revealed by convulsive seizures. A case report

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ABSTRACT

Fahr's syndrome is a fairly rare antomo-clinic entity, it is defined by the existence of bilateral and symmetrical cerebral calcifications, especially of the central grey nuclei and is often associated with dysparathyroidism. We report the case of a young 29-year-old patient who presented with seizures and headaches. A CT scan of the brain showed calcifications of the central grey nuclei, the periventricular and subcortical white matter as well as the dentate cerebellar nuclei. The biological assessment showed hypoparathyroidism and hypocalcaemia. Substitutive medical treatment based on calcium and vitamin D allowed a clear rapid clinical improvement.

Keywords
Fahr's syndrome, seizures, hypo-parathyroidism

INTRODUCTION

Fahr's syndrome, described in 1930 by Théodor Fahr, is defined radiologically by the presence of striato-pallido serrated calcificatons, no arteriosclerotic, bilateral and symmetrical (1). It is a rare condition characterized by a clinical polymorphism made essentially of neuropsychiatric disorders. The etiologies are dominated by dysparathyroidism. Fahr's triad is defined by the association of bilateral and symmetrical intracerebral calcifications of the basal ganglia, disorders of phosphor-calcium metabolism and neuropsychiatric manifestations (2).

CLINICAL CASE

A 29-year-old female patient was seen in consultation 3 years ago for episodes of non-clonic and very invigorating convulsive seizures followed by a brief loss of consciousness and partial functional of the left hemibody which resolved spontaneously in 30 minutes. All this evolved in a context of chronic headaches. The interrogation found a history of the death of his older brother in the context of convulsive seizures after several years.

The clinical examination found a sluggish patient, communicates very little and presents a frozen look at times. MRI and Brain CT (Pictures A, B, C) showed multiple symmetrical and bilateral intracerebral calcifications of the basal ganglia (striatum and thalamic nuclei),

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periventricular and subcortical white matter, and dentate cerebellar nuclei. This radiological appearance was strongly suggestive of Fahr's syndrome.

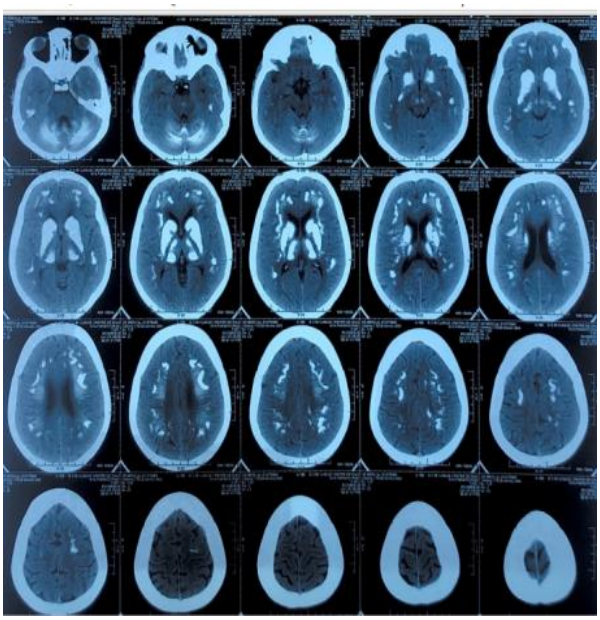

Figure 1. Initial brain scan in axial section without injection of contrast product.

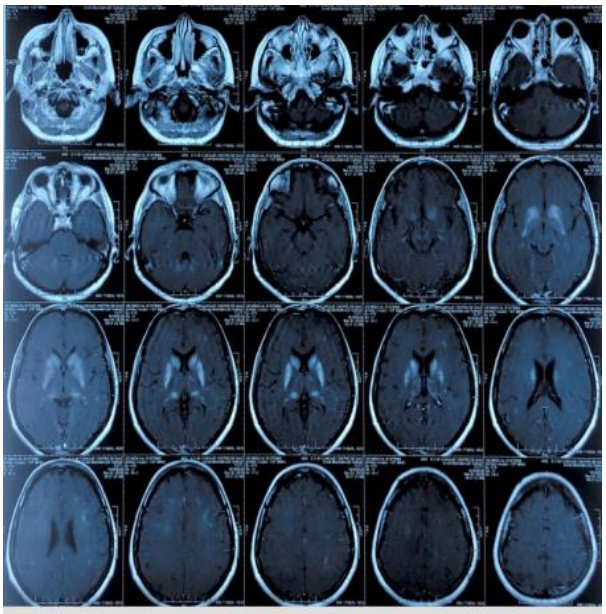

Figure 2. Initial IMR in axial section without injection of contrast product.

Laboratory tests revealed a profound hypoparathyroidism and hypocalcemia (Table N°1). The rest of the assessment, in particular the blood count (CBC), 24-hour proteinuria, thyroid assessment, cervical ultrasound, abdominal and pelvic ultrasound, returned to normal.

The patient was put on an anticonvulsant (sodium valproate at 30 mg/kg/day), on calcium (2.5 g/day) and vitamin D (2 µg/day) and an adapted diet, in particular in terms of calcium intake.

The evolution during these 3 years was favorable and marked by the complete cessation of the crises even under low doses of 15 mg/kg/day of sodium valproate, the headaches decreased considerably, the calcemia was corrected, the parathyroid hormone remained low and imaging unchanged.

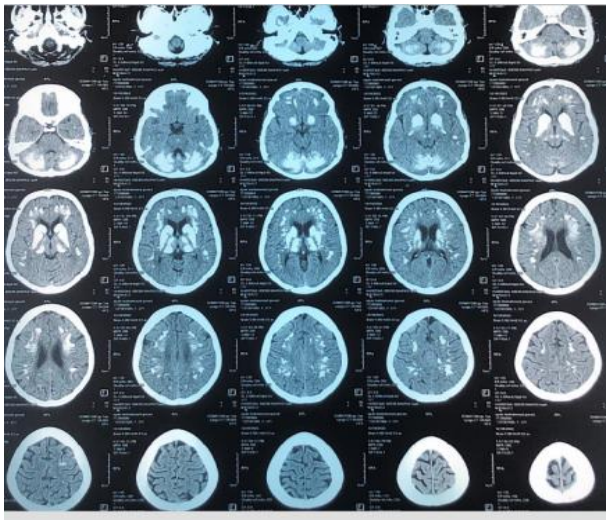

Figure 3. Brain scan after 3 years of medical treatment.

Table 1. Evolution of the biological assessment under medical treatment.

	Initial Value	At 18 months	At 36 months
Calcemia	50,90 mg/l	103,04 mg/l	102,60 mg/l
Parathyroid hormone	5,54 pg/ml	2,24 pg/ml	5,19 pg/ml
Vitamin D	36,82 ng/ml	41,78 ng/ml	23,61 ng/ml
Phosphoremia	61,4 mg/l	56,12 mg/l	-
Magnesemia	15,77 mg/l	26,68 mg/l	-

Discussion

Intracerebral calcifications have been described by several authors since 1855 before Théodor Fahr (1930) gave his name to the syndrome (1). Fahr's syndrome is a rare anatomo-clinical entity,

determined by the presence of intracerebral, bilateral and symmetrical, non-arteriosclerotic calcifications, located in the central gray nuclei. It is usually associated with disorders of calcium phosphate metabolism, and mainly with hypoparathyroidism (3). The physiopathological mechanisms of intracerebral calcifications in Fahr's syndrome are poorly understood. Most authors mention a metabolic disorder of oligoglial cells with deposits of muco-polysaccharides and secondary appearance of vascular and perivascular lesions and calculus encrustations. (4).

It is important to distinguish Fahr's syndrome from Fahr's disease, the latter is a genetic disease to be sought in front of a Fahr's syndrome without found etiology (5) and would be autosomal recessive or dominant (6).

The occurrence of calcifications is possible from infant age (5) to advanced age (7). The average age of symptom manifestation would be around 29 years (8) as in the case of our patient. The male sex would be more affected (1, 8, 9).

Fahr's syndrome can remain asymptomatic and be discovered during brain imaging (3, 10). The clinical manifestations are polymorphic (3, 11, 12) and are made up of neuropsychiatric disorders, essentially cognitive disorders, psychiatric disorders and abnormal movements. Neurological signs are dominated by seizures (1, 4, 5, 9, 13) as in the case of our patient in whom it was the discovery of the syndrome following episodes of seizures. Parkinsonian syndrome (7, 11), myoclonus (12), cerebellar syndrome, intracranial hypertension syndrome and pyramidal syndrome are also described in neurological manifestations. Neuropsychiatric and cognitive signs may be indicative of the syndrome (5). Dermatological (14), ophthalmological (cataract and exophthalmos) (15) and signs related to hypocalcaemia (muscle cramps, tetany attacks) should be sought in the context of Fahr's syndrome.

The cerebral scanner makes it possible to make the diagnosis of calcifications. These calcifications are bilateral and symmetrical and may involve the central gray nuclei, the cortex, the vessels and the cerebellar gray nuclei (1).

The phosphor-calcic balance, constantly disturbed, manifests itself essentially by hypoparathyroidism, hypocalcaemia and hyperphosphoremia (1).

The treatment is medical, based on the treatment of the cause and that of the symptoms. Phospho-calcic disorders (in particular hypocalcemia) and hypovitaminosis D are treated by the replacement intake of calcium and vitamin D due to 1 to 2g/day of calcium in adults, 40 to 60 mg/Kg/ day in children and 1µg/day of OH-Vitamin D3 (3, 4, 1, 13). As in our patient, the case of epilepsy, antiepileptic drugs could be offered while avoiding those that can induce or aggravate hypocalcemia. Extrapyramidal manifestations generally respond poorly to dopa therapy (5).

The evolution under treatment is very favorable with complete disappearance of clinical manifestations and biological abnormalities, from the first months of medical treatment (11, 7). The precocity of the medical treatment is a guarantee of the considerable improvement of the neurological clinical signs in particular the convulsive crises, the cerebellar syndrome and even extrapyramidal. Mental disorders, when established, are not improved by medical treatment, nor are cerebral calcifications (16).

Conclusion

Fahr's syndrome is a rare pathology and its patho-physiological mechanisms are not yet well understood. Despite its clinical polymorphism, the prognosis remains good, especially if medical treatment is started early. Hence, the interest of looking for anomalies of calcium phosphate metabolism and intracerebral calcifications in the event of neuropsychiatric disorders.

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