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FAHR Syndrome: An Unusual Case in the ICU

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ABSTRACT :

Fahr syndrome is a rare micro-vessel disease, whose diagnosis is often badly clarified, with unelucidated physiopathology secondary to a myriad of mutations and others aetiology. It can be clinically defined as bilateral brain calcinosis of the basal ganglia in the presence of neuropsychiatric and extrapyramidal disorders.

We will report the case of a 23-year-old patient with chronic headache, that had atypical clinic and to whom the diagnosis was fortunate. We aimed to present an incidental diagnose of Fahr's syndrome in a patient with mutism to raise awareness of this new clinical feature.

Key-Words: *Fahr's syndrome. Brain calcinosis. Basal ganglia, emergency medicine, Intensive care.*

Introduction:

The prevalence of Fahr's syndrome is more or less unknown <1/1,000,000 (1). due to insufficient investigations of first-degree relatives of the patients. Fahr's disease is a rare, neurological disorder secondary to an autosomal dominant Character (2);

There are four genes which have been implicated as the molecular basis of Fahr disease (SLC20A2, PDGFRB, PDGFB, and XPR1) (3.4), loss of function mutation in the gene SLC20A2 encoding type 3 sodium-dependent phosphate transporter 2 (PiT2) on chromosome 8p (40%), a mutation in gene XPR1 that encodes for a retroviral receptor with phosphate export function on chromosome 1q (2%), a mutation in the gene which encodes the receptor for members of the platelet-derived growth factor family- gene PDGFRB on chromosome 5q (2%), and gene PDGFB on chromosome 22q (11%). 46% of cases have some unknown gene mutations. Other loci that have been linked to Fahr disease include IBGC1 locus at chromosome 14q, a locus at chromosome 2q, and another one at chromosome 8 (5). The exact etiologies of this syndrome are unknown, but calcium metabolism disorders, toxins, infections, genetic causes, hypoparathyroidism, and pseudohypoparathyroidism may cause this syndrome (3.6).

It has a really polymorphous clinical manifestations: seizures, rigidity, and dementia with characteristic calcification of the basal ganglia (7).

In this case report, we aimed to present an incidental diagnose of Fahr's syndrome in a patient with mutism, which is an atypical clinical presentation.

Case Report:

We will report the case of a 23-year-old patient, with a medical history of asthma and chronic headache. He came to the emergency room for alteration of consciousness with a Glasgow score at 10/15. At the admission, the patient presented mutism, with no neurological signs of localization and reactive pupils.Blood pressure was 100/50 mmHg, Heart rate was 85 bpm, respiratory frequency was 20 cycles per min, and 99% saturation with ambient air. The biological tests were normal. The electrocardiogram was without abnormality. The brain scan revealed extensive, bilateral and symmetrical calcifications in the basal ganglia.

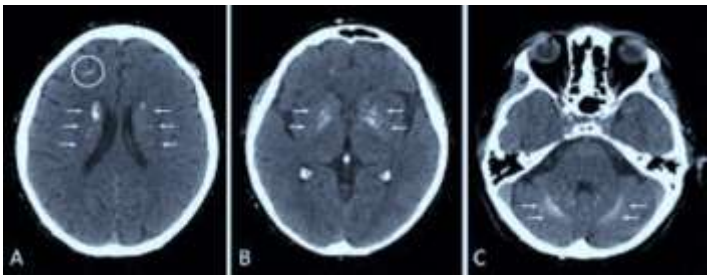

Fig : CT-scan of the patient showing calcifications of the basal ganglia

Discussion:

In the case of our patient, he had clinical feature that was described by literature such as headache , and cognitive deterioration and some went missing such as dementia stroke. . On the other hand, we have not observed other symptoms reported in different studies such as hallucinations, Mood disorders, Anxiety, obsessive behaviors, Irritability, Aggression, Personality disorder, Parkinsonism and movement disorders Seizures, Headache, Vertigo, Paresis, Stroke Syncope, Ataxia, Dysarthria Tremor, Orthostatic hypotension (7). Furthermore, the patient presented, a mutism an unusual manifestation of Fahr syndrome.

Not described in literature, the diagnostic criteria are progressive neurological disorders in the bilateral basal ganglia detected by the imaging methods in the 4th decades of life.(8) in the case of our patient The CT-scan fit to the literature descriptions (8) but started at the very young age of 23year-old.

In the case that we are discussing, there was no metabolic abnormality nor a familial history of similar cases. The absence of biochemical abnormalities and somatic features suggesting mitochondrial or other systemic disease (9)

The pathogenesis of Fahr syndrome is not fully understood. The defects in the transport of iron atoms and tissue damage caused by free radicals cause the onset of calcification (4.10).

Conclusion:

In Fahr's syndrome no specific aetiology agent has been identified yet. Which make the diagnosis difficult for clinicians. Moreover , the exact mechanism remains mystical and the polymorphous clinical syndrome and the apparition of new clinical feature might be an indicator to another mutation that could be incriminated.

However, since mutism is not described in literature, it spots the hypothesis of a new mutation that could lead to Fahr's syndrome.

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