Review Article

FARH Syndrome: About a Case Diagnosed at the Abass University Center NDAO

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ABSTRACT

This was a 69-year-old woman, seen for a progressive bilateral decline in visual acuity. Brain CT performed during neurology follow-up revealed the presence of calcifications of the basal ganglia. She had been known and treated for hypertension for 10 years. She underwent a total thyroidectomy and suffered a fracture in her left hip. Ophthalmological examination showed corrected visual acuity of 6/10 P2 in the right eye and 8/10 P2 in the left eye, with early cortical cataract in both eyes. Neurological examination showed extrapyramidal syndrome. The visual field revealed a deep, more or less confluent, bilateral peripheral deficit. Visual evoked potentials showed bilateral partial cortical damage, associated with moderate axonal damage more accentuated in the periphery in both eyes. The electrocardiogram showed left atrial hypertrophy. In biology, we noted: hypocalcemia, hyperphosphatemia, hypomagnesemia , reduced vitamin D, PTH < 1.00 ng / l . Thus the diagnosis of FS was retained. We started a treatment based on levothyroxine, calcium supplementation, vitamin D3, magnesium, a muscle relaxant and optical correction. After 3 months of treatment the evolution was marked by a correction of phosphocalcic metabolism disorders and neuropsychological signs.

Conclusion: FS is a rare pathology and must be differentiated from FARH disease. CT is the gold standard for making the diagnosis.

INTRODUCTION

FARH Syndrome (FS) is defined radiologically by the presence of symmetrical, non-ateriosclerotic bilateral intracerebral calcifications located in the basal ganglia, cerebellar nuclei and most often associated with phosphocalcic disorders [1]. The pathophysiology remains poorly understood; it must be distinguished from FARH disease which corresponds to calcifications of the basal ganglia without abnormality of phosphocalcic metabolism and which can be genetic or sporadic [2]. The clinical manifestations are polymorphous with a clear predominance of neuropsychiatric disorders [3]. Ophthalmological damage during FS is rare and often unrecognized [4], its evolution without treatment can compromise the patient's visual prognosis.

The aim of our study was to report a case of FS and to review the literature.

OBSERVATION

This was a 69-year-old woman, seen for a bilateral progressive decreased visuel acuity. Three months earlier, she was followed for an ischemic stroke in neurology. Brain CT performed during follow-up revealed the presence of calcifications of the

basal ganglia and the cerebellum (Figure 1,2). She was known and treated for hypertension for 10 years (eperindopril/amlodipine). The father was known to have glaucoma. She underwent a total thyroidectomy for nodular goiter in 2016, and had a fractured left hip in 2017.

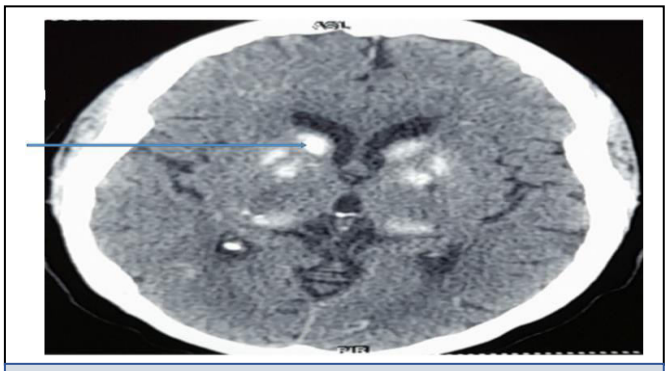

Figure 1: Brain CT showing basal ganglia calcifications (blue arrows).

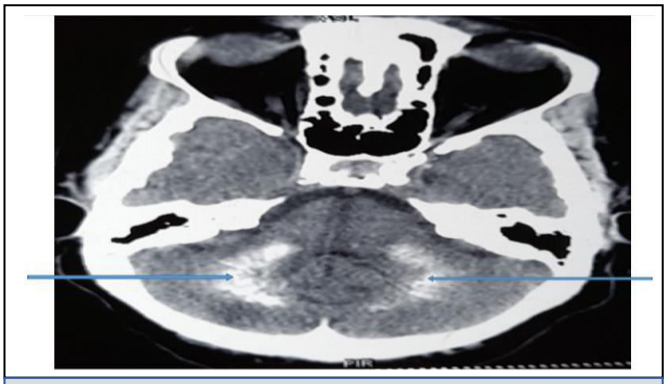

Figure 2: Brain CT showing calcifications of the cerebellum (blue arrows).

Ophthalmological examination showed corrected visual acuity of 6/10 P2 in the Right Eye (RE) and 8/10 P2 in the Left Eye (LE), an incipient cortical cataract, and an ocular tone of 20 mmHg, in both eyes. The neurological examination showed an extrapyramidal syndrome (rest tremor, dyskinesia, muscular hypertonia). Examination of the other devices was unremarkable. The visual field revealed a deep, more or less confluent peripheral deficit (Figure 3,4). Visual Evoked Potentials (VEP) showed bilateral partial cortical damage, associated with moderate axonal damage more accentuated in the periphery of the eyes (Figure 5).

Glaucoma mode OCT was normal (Figure 6,7). Pachymetry showed a thin thickness (475 µm at RE and 486 µm at LE).

The electrocardiogram showed left atrial hypertrophy. Ultrasound of the urinary tract was normal. In biology, we noted: hypocalcemia at 70.1 mg/ l, hyperphosphatemia at 63 mg/l, hypomagnesemia at 15.4 mg / l, vitamin D reduced at 29 ng /l, PTH < 1, 00 ng /l. Thus the diagnosis of FARH syndrome was retained. We initiated a treatment based on levothyroxine, calcium supplementation (2 g/day), vitamin D3 (calcifediol: 12 drops/day), magnesium, a muscle relaxant and optical correction. After 3 months of treatment the evolution was marked by a correction of phosphocalcic metabolism disorders and neuropsychological signs.

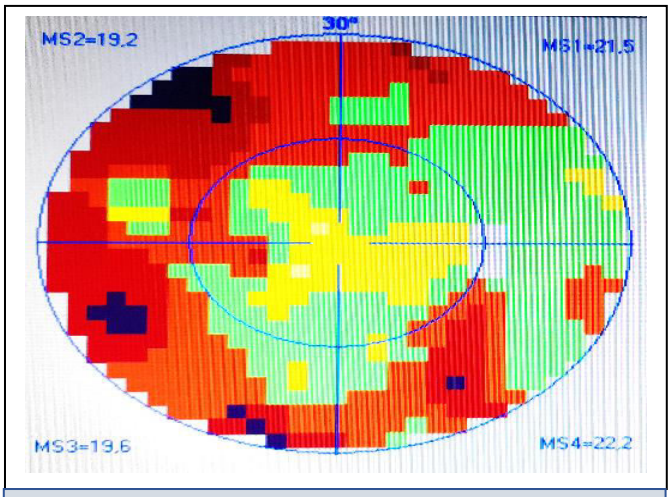

Figure 3: Visual field of the left eye with discontinuous relative peripheral deficits.

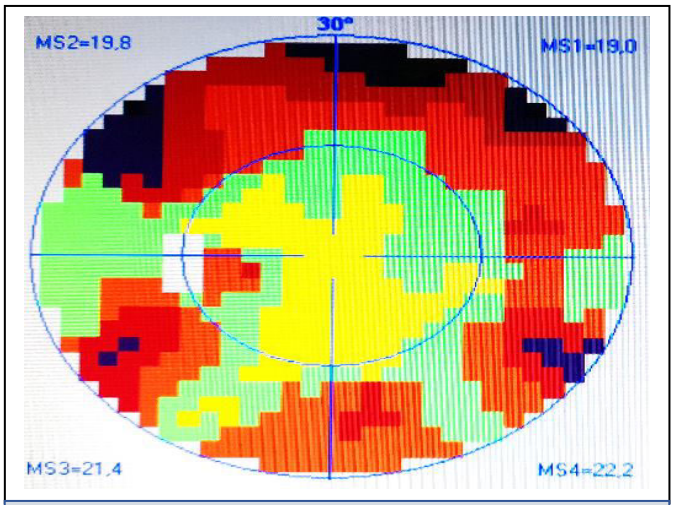

Figure 4: Visual field of the right eye showing discontinuous relative peripheral scotomata.

DISCUSSION

FS occurs preferentially in patients with dysparathyroidism , primarily hypoparathyroidism [5]. It can remain asymptomatic and be discovered during cerebral radiological explorations [6]. FS can result in polymorphic and non-specific neurological

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manifestations. Ophthalmological manifestations are rarer during this pathology [5]. It was the first case diagnosed in the department. Winner [7] found that the frequency of calcifications in the elderly group was significantly higher than that in the young group. It affects men more than women, with a sex ratio of 2 according to studies [8]. These calcifications affect the small vessels of the basal ganglia. In our study, the patient was seen for a progressive bilateral distant and near decreased visuel acuity, which is consistent with the Riani work. [4] where bilateral decreased visuel acuity of progressive installation was their main reason for consultation. The most common ocular involvement is a bilateral posterior subcapsular cataract related to chronic hypocalcemia and concerns FS secondary to hypoparathyroidism [9].

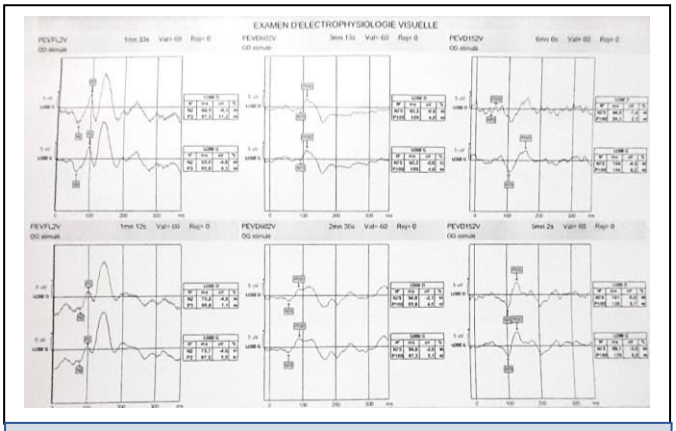

Figure 5: Visual evoked potential in both eyes showing bilateral partial cortical damage.

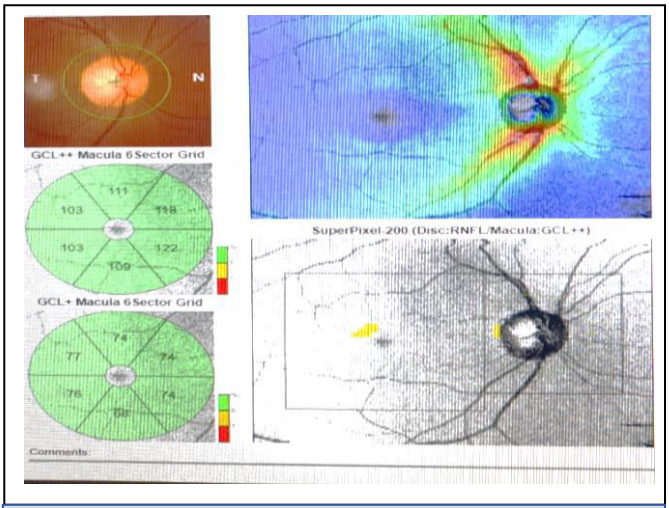

Figure 6: OCT of the right eye normal.

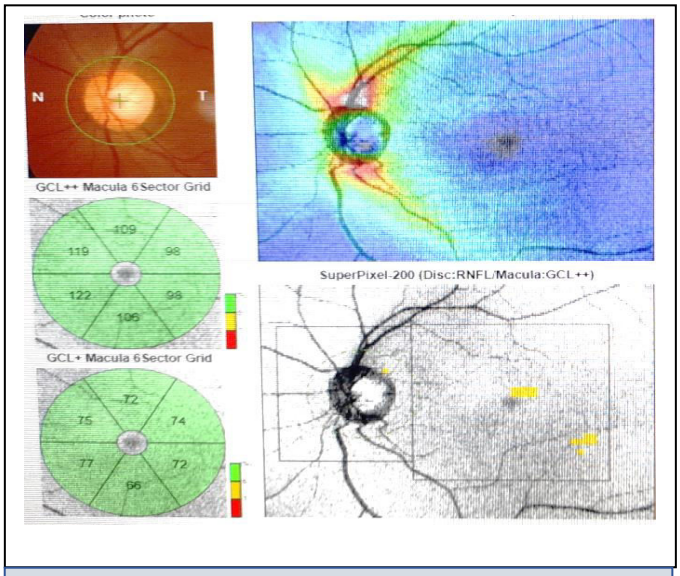

Figure 7: OCT of the left eye normal.

The incidence of extrapyramidal syndromes in pseudohypoparathyroidism is 12.5% [10]. The symptoms of hypocalcemia are closely linked to the calcium level, and will be all the more obvious if the hypocalcemia is severe and sudden in onset. Pereira GS [1] reports generalized neuromuscular irritability with muscle cramps and tetany in a patient. In a study by Otheman [11], the patient presented a fairly classic picture of paranoid schizophrenia, the existence of an association between calcifications of the basal ganglia and schizophrenia was mentioned [12]. With Manyam [13], VEP were normal in all his patients. Koller 's study. [14] and Puvanendran [15], and 2% over a period of 16 months for Fenelon 's team [16]. This is to underline the relevance of this examination of choice, which can detect asymptomatic cases. It is the reference examination, the most sensitive for detecting calcifications [13]. In a study carried out at the Rabat military hospital, the phosphocalcium balance was disturbed in 87.5% of cases with severe hypocalcemia ranging from 0.9 mmol /l to 1.5 mmol /l. They also note hyperphosphatemia between 1.9 mmol /l and 2.5 mmol/l .

In FS, replacement treatment with calcium therapy combined with vitamin D therapy ensures improvement in the case of dysparathyroidism , the main objective of which is to correct phosphocalcic disorders [17]. In adults, the dose of calcium supplementation is usually between 1 and 6 g/day in 1 to 4 doses. In children, it is 500 to 1000 mg/day or 40 to 60 mg/kg/day depending on age [18]. This supplementation can

be provided in the form of calcium carbonate. Until the calcium pool has been replenished, calcium supplementation is necessary [18]. Antipsychotic treatments have an important place when psychiatric symptoms are prominent and disabling [19]. Etiological treatment is necessary if the calcifications are secondary to systemic diseases, secondary infections or other pathologies. Therefore, asymptomatic patients should be monitored.

CONCLUSION

FS is a rare pathology and must be differentiated from FARH disease. CT is the reference examination to demonstrate basal ganglia calcifications and must be systematic in any patient with dysparthyroidism. The functional ocular and neuromuscular impairment can compromise the patient's quality of life. The treatment East purely symptomatic.

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