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Fahr syndrome in young boy with hypoparathyroidism.

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Introduction

Fahr syndrome is a rare degenerative disease, characterized by the presence of calcification of the basal ganglia.Autosomal recessive or dominant, variable penetrance.

Usually asymptomatic in the first 2 decades, the disease typically manifests itself either at 30 years of age by the appearance of neuropsychiatric disorders, or at age 60 by progressive dementia with extrapyramidal syndrome.

Observation

I.M aged 09 years addressed in endocrinology for neurological disorders with suspicion of severe hypocalcemia. There is no notion of parental consanguinity. The patient presents with generalized comitials under treatment (troubleshooting) since the 45th day of life with delayed acquisition of gait associated with a mental deficit. Three months prior to the consultation, the patient had a brutal left hemiparesis that was accompanied by a homolateral carpal spasm of progressive accentuation.

The weight of the child at 32 kg (+1 D.S.), the height at 138 cm (+1 D.S.).
Facial dysmorphic syndrome with an elf-like face, depressed nasal ridge, hypertelorism (Fig-1), dental dysgenesis, gingival hypertrophy (Fig-2).

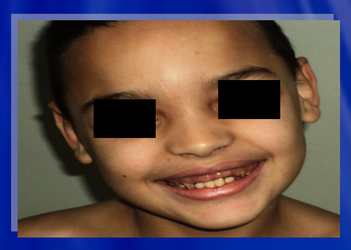

fig-1

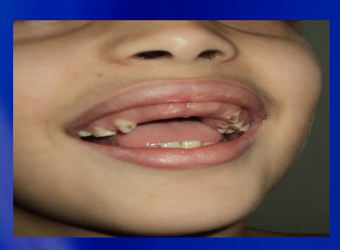

fig-2

Signs of hypocalcemia: paresthesia, cramps, Chvostek present, trousseau positive, dental hypoplasia. The fundus examination is normal and the slit lamp examination does not reveal cataracts.

Medical check-up

Calcémie	55 mg/l Basse	Urée	0.24 g/l
Phosphorémie	95 mg/l Élevée	Créatininémie	08 g/l
PTH N : 12 à 88 pg/ml	2.3 pg/l Basse	Magnésémie	17mg/ml
Vitamine D	Non disponible	Phosphatases alcalines	141 U/l
Protidémie	65 g/l	TSHus	0.53 mU/l
Albuminémie	43.8 g/l	FT4	15 pg/ml

ECG recovers large, pointed and symmetrical T-waves.

EEG reveals an asymmetrical standby pattern, hypovolt to the right, with no critical paroxysmal abnormalities.

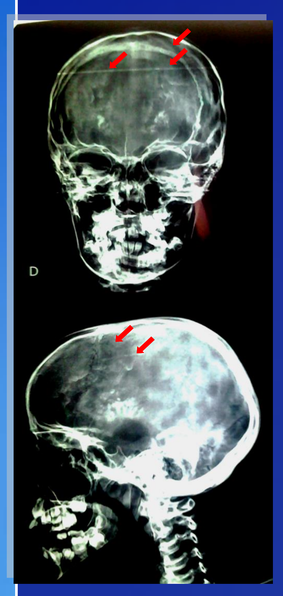

fig-3

Radiography of the skull shows clumps of small right and left cranial calcifications

No osteo-articular lesion on skeletal radiographs (fig-4)

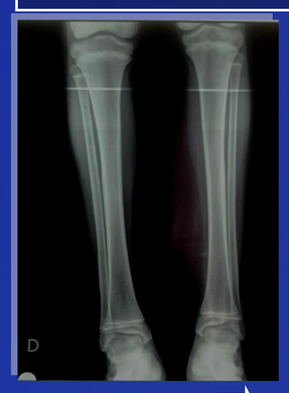

fig-4

Bone age 09 years

According to the method of Greulich & Pyle

(fig-5)

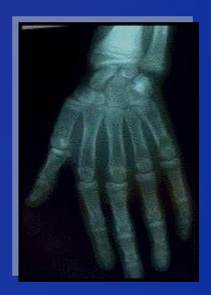

fig-5

Brain Computed Tomography

Large, diffuse and symmetrical calcifications of central grey nuclei, including thalamic, lenticular, caudal and peripheral ventricular paracortical regionstemporo-parietooccipital gyriformes (fig-6)

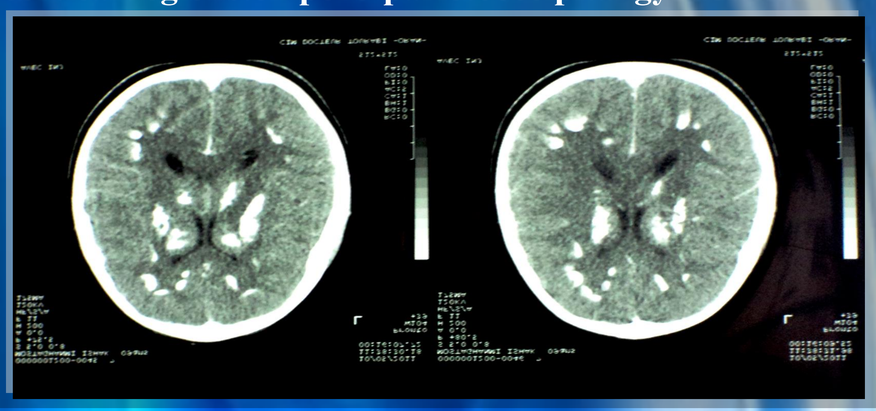

fig-6

The chosen diagnosis: Hypoparathyroidism revealing Fahr syndrome on clinical, biological and radiological data.
A treatment: Combination of calcium (1 g/d) and 1-alpha-hydroxy-vitamin D (2 µg/d).
Evolution: disappearance of neurological disorders after 03 months, normalization of calcemia, phosphoremia and calciuria after 09 months.

Discussion

The pathophysiological mechanisms that contribute to intracerebral calcifications during the SF are not well understood. Most authors suggest a metabolic disorder of oligoglial cells with mucopolysaccharides deposits and secondary onset of vascular, perivascular and calcareous lesions. These calcifications concern the small vessels of the central grey nuclei [1]. Their biochemical analysis showed an organic matrix, consisting of neutral mucopolysaccharides and acids as well as mineral elements (calcium, phosphorus, iron, sulphur, magnesium, aluminium, zinc) [2]. These calcifications are most often manifested by neuropsychic disorders. Hypoparathyroidism is the most common cause of SF-related hypocalcemia.

The hypocalcemia caused by hypoparathyroidism explains the majority of clinical signs (cataract, malabsorption, neuromuscular hyperexcitability, various neurological and neuropsychological signs, psychiatric disorders that can lead to psychosis, various cardiovascular disorders) [3]. It is important not to confuse SF with other conditions that can lead to intracerebral calcification, especially endocrinopathies (hypothyroidism, hypogonadism), systemic pathologies (systemic scleroderma, systemic lupus erythematosus) infections (toxoplasmosis, neurocysticercosis, rubella), various diseases (chronic renal failure, vitamin D intoxication, mitochondrial cytopathies) and primary or calcified secondary brain tumors.

In contrast to the severity of the symptoms it may be responsible for, SF has a good prognosis and correction of phosphocalcic metabolic disorders often leads to significant improvement .

Conclusion

Interest to look for phosphocalcic abnormalities in front of neurological manifestations and / or in the presence of brain calcifications in the child.

CT scan is the best choice for showing a calcification.

Correction of biological disorders = disappearance of symptoms.

Références

1-. Fahr T. Idiopathische Verkalkung der Hirngefässe. Zentralbl Allg Pathol. 1930-1931;50:129–33.
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3-Chouaib N, Rafai M, Belkouch A, Bakkali H, Belyamani L, et al. Découverte fortuite d'un syndrome de Fahr suite à une crise convulsive. Rev Neurol (Paris). 2015 Dec;171(12):894–5

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