Fahr syndrome in young boy with hypoparathyroidism.

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

LM 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 convulsions 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).

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

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
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<tbody>
<tr>
<td>Calcium</td>
<td>65 mg/l</td>
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<tr>
<td>Phosphorus</td>
<td>14 mg/l</td>
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<tr>
<td>PTH</td>
<td>12 pg/ml</td>
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<tr>
<td>Vitamin D</td>
<td>43 μg/d</td>
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<tr>
<td>Albumine</td>
<td>43.8 g/l</td>
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ECG recovers large, pointed and symmetrical T-waves.

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

Brain Computed Tomography

Large, diffuse and symmetrical calcifications of central grey nuclei, including thalamic, lenticular, caudal and peripheral ventricular para-cortical regiontemporo-parietooccipital gyriformes.

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.

Evolution: disappearance of neurological disorders after 03 months, normalization of calcemia, phosphemia 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 oligodendrocyte 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 neuropsychiatric 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 (hyperparathyroidism, hypogonadism), systemic pathologies (systemic scleroderma, systemic lupus erythematosus) infections (toxoplasmosis, neurocytomecosis, 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.

References