

THE BRAIN IN PSEUDOHYPOPARATHYROIDISM*

Por P. ALBÚJAR**, R. HUNTER*** & A. DAYAN****

RESUMEN

Comunicamos el caso de una paciente que tuvo rasgos clínicos compatibles con pseudohipoparatiroidismo: convulsiones desde la niñez, deficiencia mental, baja estatura, braquidactilia, tetania, movimientos extrapiramidales coreoatetoides, cataratas, hipocalcemia y posible frecuencia familiar. En la autopsia se halló calcificación de los núcleos basales y cerebelo.

ABSTRACT

We report the case of a female patient with clinical features consistent with pseudohypoparathyroidism: convulsions beginning at childhood, mental deficiency, short stature, brachidactily, tetany, extrapyramidal movements choreoathetoid type, cataracts, hypocalcemia and possible familial occurrence. At autopsy, symmetrical basal ganglia and cerebellar calcification were found.

PALABRAS-CLAVE: Calcificación de núcleos basales, pseudohipoparatiroidismo.

KEY WORDS : Basal ganglia calcification, pseudohypoparathyroidism.

Pseudohypoparathyroidism and Pseudopseudohypoparathyroidism are two conditions of special interest to the neurologist and psychiatrist because they may present with manifestations, that if not properly interpreted, may lead to erroneous diagnoses. Epilepsy for example is so commonly confused with tetany that makes appreciate the classi-

cal advice to determine serum calcium and phosphorus in every new case of epilepsy^{1, 2}.

Our report deals with the case of a patient institutionalized with the diagnosis of insanity, with progressive mental and neurologic deterioration and misinterpreted intracranial calcification.

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** British Council Scholar, Institute of Neurology, University of London.

*** Neurologist.

**** Pathologist, The National Hospital for Nervous Diseases, Queen Square, London.



Fig. 1. Yellow coloured material in shrunken basal ganglia

CASE REPORT

The patient was the third of eleven children born in London of unrelated parents. One brother was said to have fits.

She started to have convulsions at seven years of age which stopped at age 15. She was always backward and unable to attend school.

In 1925, when she was 29, was treated in a general hospital for hysteria. Carpopedal spasm and trismus were observed and her condition improved with calcium lactate.

At age 48 she attempted to commit suicide. She was admitted to Friern Hospital as a case of "insanity and epilepsy". On examination her weight was four feet nine inches (142 cm) and her weight seven and a half stones (47 Kg). She was observed to drag her left leg. Her speech was indistinct and slurred. At times she had difficulty in feeding and swallowing.

At age 50, fits reappeared and continued at irregular intervals. Tremor of hands and writhing movements of protruded tongue and limbs reappeared. Generalized rigidity of cogwheel type was noted. She was delirious and hallucinated. Bilateral cataracts, more marked in the left eye, were found. She became gradually deaf and globally demented.

An skull X-Ray film was reported as showing calcification of choroid plexuses in lateral ventricles. X-Ray of wrists and hands were taken because of Colles fracture. Serum calcium ranged between 7 and 9.4 mg/100ml and serum phosphate was 5.1 mg. Alkaline phosphatase was 11 KA units. Urea and glucose were normal.

The patient became increasingly ataxic, feeble, rigid, akinetic and died at age 75.

At autopsy, the brain was small. On slicing dense calcification involving symmetrically the basal ganglia was found. The



Fig. 2. The calcified tissues of basal ganglion is shown as bluegranulated material

striatum and pallidum were shrunken and largely replaced by yellowcoloured material (Fig.1). The caudate nuclei, putamen, right thalamus more than left, were affected. There was also calcification of deep white matter of parieto-occipital lobes on both sides. Choroid plexuses were normal. Cerebellum showed symmetrical calcification of both dentate nuclei and adjacent white matter. Brain stem showed shrinkage of right cerebral peduncle and pyramidal tract throughout its length. Many major arteries were completely occluded by atheroma. Chemical Analysis: the basal ganglia contained 15.1 g/100g. dry weight Ca". (normal 40-60 mg/100 g)

Microscopically multiple areas of perivascular droplets and confluent calcification in watershed areas of the cerebral cortex, caudate and putamen (Fig.2), deep in cerebral folia and in dentate nuclei were noted. These lesions were associated with neuronal loss and gliosis.

Examination of the rest of the body revealed mild hypertrophy of the heart and generalised atherosclerosis. One parathyroid was found grossly normal.

DISCUSSION

Recapitulation of the clinical history of this patient reveals that she had suffered from tetany and its complications.

The short stature of the patient, calcification of brain and cerebellum and the apparent normal parathyroids are clues that lead to believe that this was probably a case of pseudohypoparathyroidism³. Because this diagnosis came out after postmortem examination, we have no conclusive investigations to support a definite diagnosis, such as proper X-ray examinations of hands and feet, the Ellsworth-Howard test⁴ or excretion of cyclic adenosine 3',5' monophosphate or determination of serum levels of parathormone. The patient had stubby hands but the X-ray films taken be-

cause of a Colles fracture, are not satisfactory for adequate interpretation. The possibility that other members of the same family could also be affected is supported by the fact that one brother had suffered from seizures. Mental retardation⁵ and extrapyramidal signs⁶ are manifestations usually observed in pseudohypoparathyroidism.

Pseudopseudohypoparathyroidism differs from pseudohypoparathyroidism in the biochemical finding of normal serum calcium and lack of tetanic symptoms. The body habitus is similar in the two conditions. In fact they are considered as two forms of the same syndrome to which the name of Albright's Hereditary Osteodystrophy is applied⁷.

True or Idiopathic Hypoparathyroidism seems unlikely because of the patient habitus and calcification of the basal ganglia has rarely been reported with familial incidence as is usually the case of pseudohypoparathyroidism⁸.

In the group called "Familial bilateral calcification of basal ganglia", symptoms are restricted to the nervous system, tetany is absent⁹ and the lesions contain appreciable

amount of iron¹⁰. The eponym Fahr's disease is a vague denomination and its use is not justified¹¹.

Several factors have been considered in the pathogenesis of cerebral calcification. Albright thought that it could be due to plasma supersaturation with phosphorus. It has been suggested that calcification could be the result of parathyroid deficiency but in fact the parathyroid hormone is elevated in pseudohypoparathyroidism¹². Anoxia is a factor that has been seriously considered. In this context the distribution of calcification in boundary zones of major territorial arteries, as in the case described by Norman and Urich¹³, and also observed in our case, suggest the participation of disturbance of arterial flow in the production of abnormal vascular permeability and deposition of calcium salts in selective regions. A generalised disturbance of Ca/P metabolism, however important it may be in the majority of cases, is not the only answer since intracranial corticopallidodentate calcification has been reported in patients with no parathyroid deficiency and, on the other hand, a number of patients with conspicuous parathyroid insufficiency have been reported without calcification.

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