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CASE REPORT

Intramuscular Hemangioma of the Temporalis Muscle With Incidental Finding of Bilateral Symmetric Calcification of the Basal Ganglia: A Case Report

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We report an 11-year-old boy whose brain computed tomography findings incidentally revealed bilateral basal ganglia calcification. He was symptom-free and had no abnormal neurological findings. He was diagnosed with Fahr's disease based on radiological findings and after excluding other etiologies such as infection, metabolic disorders, congenital malformation and malignancies. Most of the reported cases display an autosomal dominant mode of inheritance. Although Fahr's disease is a rare cause of basal ganglia calcification in children, this disease should be considered in children with a family history of neuropsychiatric disorders.

1. Introduction

Fahr's disease, or idiopathic basal ganglia calcification, is an inheritable disorder characterized by calcification of the basal ganglia and extrastriate regions. It can be autosomal dominant, familial, or sporadic. Most patients remain asymptomatic, but some manifest a variable combination of movement disorders, cognitive impairment, cerebellar signs, speech disorders, psychiatric illness and sensory impairment.

2. Case Report

An 11-year-old boy visited our emergency department because of painful swelling over the right temporalis lasting 1 day. The swelling had been intermittent for 3 months, and was sometimes accompanied by painful sensations. Previously, the swelling resolved spontaneously within 3 hours. The patient denied any pulsatile sensation, weakness, numbness or blurred vision. Trauma and fever were not evident.

On attendance at our emergency department, the patient had a regular heart rate (78 beats/min) and normal blood pressure (105/73 mmHg). An ill-defined, soft, tender, swelling mass, measuring 3 cm × 3 cm × 0.5 cm, was noted over the right temporalis. No local heat, redness, pulsation or bruit was found, and the overlying skin was normal. The patient's head circumference, height and body weight were normal for his age. Neurologic examination did not show any remarkable findings. No tremor, dystonia or choreoathetoid movement was seen.

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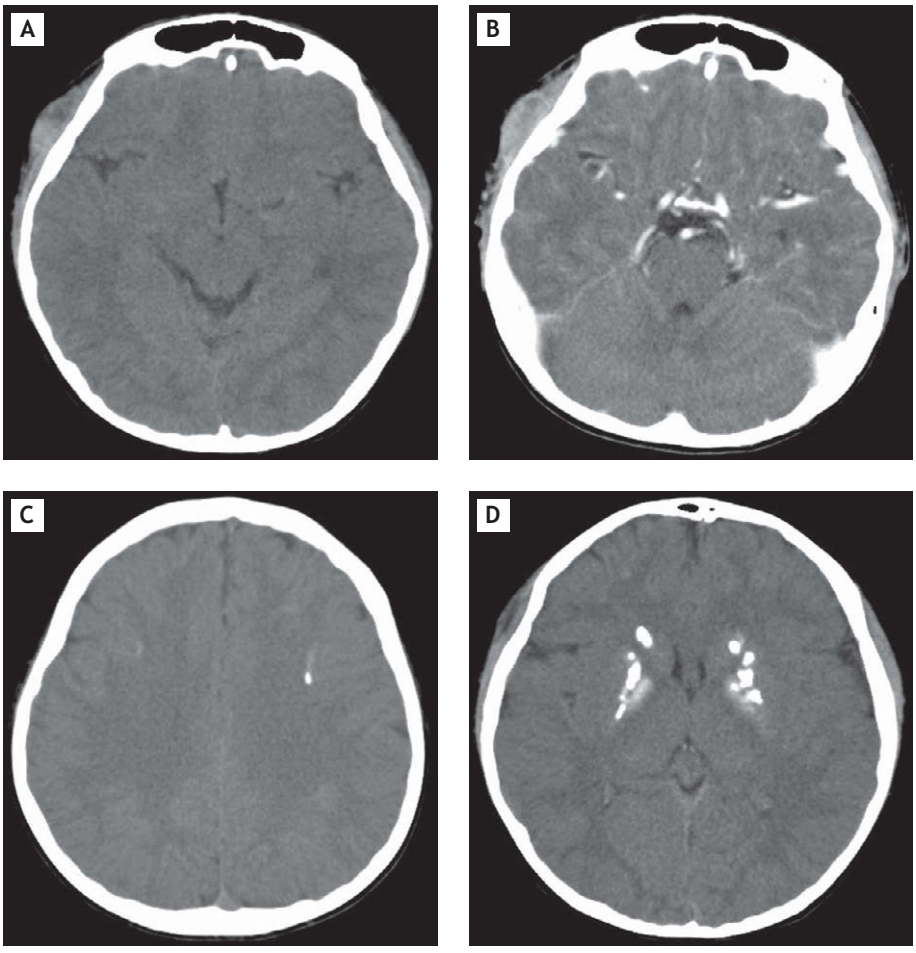

Figure 1 Computed tomography (CT) images of the brain. (A) Precontrast CT shows a poorly demarcated heterogeneous lesion over the right temporalis muscle. (B) Postcontrast CT also shows the heterogeneous right temporalis lesion, which is minimally enhanced. (C) Precontrast CT obtained through the centrum semiovale shows calcification of the left frontal white matter. (D) Precontrast CT obtained through the genu of the internal capsules shows bilateral symmetric calcification of the basal ganglia.

The tentative diagnosis was a vascular or lymphatic lesion over the right temporalis. A computed tomography (CT) scan showed a right temporalis mass, which suggested a hemangioma with bleeding (Figures 1A and 1B). It also revealed multiple symmetrical calcifications over the bilateral basal ganglia and some punctuate calcification over the left frontal white matter (Figures 1C and 1D). The boy received excision of the right temporalis mass, and the pathology turned out to be a cavernous hemangioma.

Biochemistry was performed, and results were as follows: calcium 2.4 mmol/L (normal range, 2.1–2.55 mmol/L), phosphorus 1.9 mmol/L (0.81–1.45 mmol/L), magnesium 0.99 mmol/L (0.65–1.05 mmol/L) and intact parathyroid hormone 21.4 ng/L (15.0–65.0 ng/L). The lactic acid concentration was 1.12 mmol/L (0.5–2.20 mmol/L).

Reviewing the patient's medical history, we found he was born without perinatal insult and had normal growth and development. He had normal intellect and ordinary school performance. There was no family history of epilepsy, dystonia or any other neurologic diseases.

3. Discussion

Basal ganglia calcification is found on 12.5% of routine brain CT scans performed in the general population, with the globus pallidus being the most frequent location.[1,2] However, this finding was observed in only 1–2% of brain CT scans performed in children.[3] The causes of basal ganglia calcification are variable (Table 1), and disorders of calcium metabolism including hypoparathyroidism, pseudohypoparathyroidism or hyperparathyroidism and neoplasms are the main causes. Biochemical investigations to rule out abnormal calcium regulation and metabolic disorders should be made when encountering a child with basal ganglia calcification without an apparent cause.

Table 1 Etiologies of basal ganglia calcification

Endocrine	**Congenital or developmental**
Hypoparathyroidism	Familial idiopathic basal ganglia calcification
Pseudohypoparathyroidism	Down syndrome
Pseudopseudohypoparathyroidism	Neurofibromatosis
Hyperparathyroidism	Tuberous sclerosis
Metabolic	Lipoid proteinosis (hyalinosis cutis)
Mitochondrial disease	Aicardi-Goutieres syndrome
Phenylketonuria type 2	Cockayne's syndrome
Sulfocysteinuria	Oculodentodigital dysplasia
GM1 gangliosidosis	Dyskeratosis congenita
Dihydropteridine reductase deficiency	Cerebro-oculo-facio-skeletal syndrome
Vascular	**Neurodegenerative**
Angiomatous malformation with vein of Galen aneurysm	Pantothenate kinase-associated neurodegeneration
Hematoma	Neuroferritinopathy
Anoxic	Dentatorubropallidoluysian atrophy
Asphyxia or ischemia	**Infectious**
Neoplasm	Congenital (e.g., toxoplasmosis, rubella, CMV, syphilis)
Toxic	Other virus (e.g., VZV, EBV, measles, HIV)
Lead intoxication	Cysticercosis
Radiation therapy	**Inflammatory**
Methotrexate therapy	Systemic lupus erythematosus

CMV=cytomegalovirus; VZV=varicella zoster virus; EBV=Epstein-Barr virus; HIV=human immunodeficiency virus.

Anoxia and congenital infection leading to basal ganglia calcification were unlikely in our patient simply because of his uneventful birth history. Similarly, developmental or syndromic causes were excluded because of the absence of characteristic features or skin stigmata, his normal developmental history, and negative neuroimaging finding, except for the basal ganglia calcification. Neurodegenerative conditions were unlikely because there were no symptoms/signs suggestive of neurodegeneration. Infectious or inflammatory causes or neoplasms were also excluded because there was no exposure history or related symptoms/signs. Meanwhile, biochemical testing precluded metabolic diseases, including disorders of calcium metabolism and mitochondrial disorders. Therefore, since there were no identifiable causes, a diagnosis of idiopathic bilateral basal ganglia calcification was thus made.

Idiopathic basal ganglia calcification, also called Fahr's disease or bilateral striopallidodentate calcinosis, is a rare cause of basal ganglia calcification in children and there are more than 30 terms used to describe the same condition. It is characterized by calcium and other mineral deposits in the vessel wall or in the perivascular space of the basal ganglia and extrastriate regions.[4–6] The diagnosis is established by performing brain CT or magnetic resonance imaging (MRI) and excluding other known causes. CT scans are more sensitive for revealing calcifications, which are usually bilateral and symmetrical, in the basal ganglia, thalamus, dentate nuclei, and centrum semiovale.[7,8] The lesions appear as low-intensity signals on T1-weighted and T2-weighted MRI.

The mode of inheritance can be autosomal dominant, familial or sporadic. The familial form is linked to a locus on chromosome 14q (IBGC1) or another undetermined locus.[9–12] A large proportion of affected persons are asymptomatic. Symptomatic cases are usually aged between 30 and 50 years old at onset and present with cognitive dysfunction, cerebellar signs, dysarthria, extrapyramidal signs, pyramidal signs, psychiatric illness, gait disorder, or sensory impairment. The most common manifestation is movement disorders (55%). Parkinsonism accounts for over half of all movement disorders, while hyperkinetic movement disorders (chorea, tremor, dystonia, athetosis and oro-facial dyskinesia) account for the rest.[13,14] Younger-onset cases may manifest with psychoses, whereas dementia with parkinsonism or choreoathetosis are more typical of later onset. There is great clinical heterogeneity in that the clinical features vary within and between families.[14–19] A registry study[13] showed that men had greater calcium deposits and a higher incidence of symptoms and signs compared with women. The calcification may increase with age, but it remains controversial whether the extent of calcification is correlated with the development of neurologic symptoms. It seems that symptomatic patients have a substantially greater amount of calcification, particularly in the dentate nuclei and centrum semiovale, as well as the total amount.[13]

Here, we have reported a boy with bilateral basal ganglia calcification that was found incidentally in the absence of clinical symptoms. Since neuroimaging studies were not performed in other family members, we could not ascertain whether this was a sporadic case. Having asymptomatic parents, siblings and children is not adequate to conclude that a person is sporadic. CT or MRI scans, or neuropathological evidence showing the absence of bilateral, almost symmetric calcification in the brain is necessary.[20] Follow-up of the case presented here is now needed to learn whether our patient develops more pronounced lesions or shows any related symptoms later in life.

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