Exophthalmos as the first and the only one clinical manifestation of fibrous dysplasia

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ABSTRACT

Introduction: Fibrous dysplasia (FD) of bone is a rare congenital disease characterized by a focal proliferation of fibrous tissue in the bone marrow, leading to osteolytic lesions, deformities and fractures.
Purpose: To present a case of exophthalmos as the one clinical manifestation of fibrous dysplasia.
Case: A 9-year-old girl with exophthalmos of the left eye was underwent the ophthalmological examination and computed tomography. The both eyes had a visual acuity of 20/20. The visual field was normal. On computed scan, the patient had a characteristic, pageotic appearance.
Conclusion: Exophthalmos may be the only one clinical finding in fibrous dysplasia and should be a concern because of the possibility of visual impairment when the disease is progressive. The proper diagnosis may be necessary for the choice medical or surgical treatment and may be effective in preventing visual loss in FD patients. Long-term radiological and pediatric monitoring is essential. Therefore the ophthalmologist plays an important role in the early diagnosis of fibrous dysplasia.
Key words: exophthalmos; fibrous dysplasia; vision

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INTRODUCTION

Fibrous dysplasia (FD) of bone is a rare congenital disease characterized by a focal proliferation of fibrous tissue in the bone marrow, leading to osteolytic lesions, deformities and fractures [1]. It may account for about 2.5% of bone disorders and 7% of so-called benign bone tumors.

The fibroblastic expansion of affected bones may lead to pain, swelling, and disfigurement. The disease may be restricted to one site in the skeleton (monostotic) or may affect multiple sites (polyostotic). Polyostotic FD in association with cutaneous pigmentation (cafe-au-lait spots) and precocious puberty is known as the McCune-Albright syndrome [2, 3].

Several breakthroughs in the understanding of the pathophysiology have been made in the past 10 years. It is known recognized that FD is caused by missense mutations occurring post-zygotically in the gene coding for the α-subunit of the stimulatory G-protein, mGs, in the guanine nucleotide binding, alpha stimulating (GNAS) complex locus in chromosome 20q13 [4]. The two described mutations (replacement of the arginine residue in codon 201 by either cysteine or histidine), which are dominant acting, are responsible for a somatic mosaic, and the resulting as a consequence. Activation of the Gsα/ PKA/CREB pathway induces c-fos over-expression in FD lesions [5-8]. This mutation results in osteoblastic differentiation defects, and bone resorption is often increased.

The prevalence of the disease in unknown. Monostotic forms probably represent 60% of all patients with FD, and the proportion of patients with McCune-Albright syndrome is likely to be less than 5% of all individuals with FD [9].

FD is often asymptomatic. In many patients, however, the disease is diagnosed because of bone pain, bone deformity or fragility fracture. The first symptoms often appear during childhood, but the first bone pain or fracture may be observed in the third, fourth or even fifth decade of life. First symptoms appear before the age of 15 years in 80% of patients.

Among FD of the entire body, craniofacial FD is relatively rare, and it is known that ocular problems such as visual loss, diplopia and proptosis occur in 20-35% of patients with FD of this region [10-13].

In this study, we present a case of exophthalmos as the one clinical manifestation of FD.

CASE PRESENTATION

In 2005, a healthy 10-year-old girl was seen in the Department of Pediatric Ophthalmology Medical University of Bialystok, Poland, with a history of exophthalmos of left eye of 3 years' duration. There was no precocious puberty, cutaneous pigmentary changes, or family history of polyposis or colonic carcinoma.

On physical examination, the visual acuity was 20/20 in both eyes and there was no relative afferent pupillary defect. Hertel measurements showed 6 mm of left exophthalmos. Tonometry revealed intraocular pressure of 18 mm Hg on the both eyes. The results of slit-lamp, funduscopic and visual field examination were normal in both eyes. Computed tomography performed in May 2005 (Aquillion, Toshiba; resolution 300 dpi) of the orbits revealed diffuse involvement of multiple cranial bones by fibrous dysplasia with involvement of the left orbit, nasal cavity, sinuses, and showed replacement of the left orbital roof and the lesser wing of the sphenoid by dysplastic bone with alternating areas of lucency and increased density ("pagetoid" lesion), and narrowing of the left optic canal (Fig. 1).

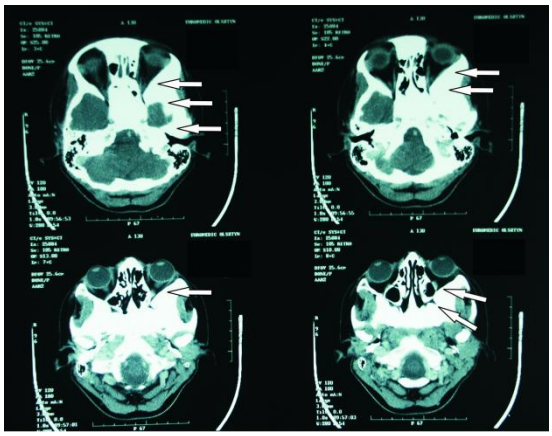

Figure 1. Computed tomographic scans through the orbits shows diffuse involvement of multiple cranial bones (sphenoid and zygomatic) by fibrous dysplasia with involvement of the left orbit, nasal cavity, and sinuses.

She has remained stable over six years of follow-up. The results of radiological, ophthalmic and pediatric examinations were without progression.

DISCUSSION

FD is an uncommon disease characterized by replacement of normal cancellous bone by fibrous tissue and immature woven bone. It is not a disease confined to adolescence but may continue into adulthood, and even middle age [10,11]. Although benign, it is slowly progressive (9). Malignant transformation is rare, occurring in only

0.5% of cases [12]. Treatment options include radiation and surgery [14]. However, radiotherapy increases the incidence of malignant transformation to 44%, making surgery the only acceptable treatment modality [9,15].

The most notable ocular complication of FD involving the sphenoid bone is a visual loss secondary to optic nerve compression. Because the optic nerve lacks the potential to regenerate, damage in the nerve may be permanent and the progressive nature of this disease can lead to irreversible blindness. FD with visual compromise has two clinical courses. Chronic type manifests gradual visual deterioration caused by mechanical compression by FD itself. In contrast, acute type manifests sudden of visual function caused by acute compression by either hemorrhage into FD, cyst formation of formation of mucocele [16, 17]. Our patient can be classified as chronic type, and this type is a very good indication for surgical intervention, but the parents didn't permit. However, prompt surgical intervention should be strongly considered in cases of acute visual loss.

In conclusion, we report a case of FD that showed exophthalmos as the only one clinical finding in this disease. The proper diagnosis may be necessary for the choice medical or surgical treatment and may be effective in preventing visual loss in FD patients. Long-term neurological, otolaryngological, orthopedic monitoring is essential. Therefore, the ophthalmologist plays an important role in the early diagnosis of FD.

Conflict of interest

None of the authors have any conflicts of interest.

Financial Disclosure

None declared.

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