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Ophtalmology

Pulsatile Exophthalmos Revealing Spheno-Orbital Agenesis Associated with Von Recklinghausen's Disease

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Abstract | **Case Report**

Ophthalmological manifestations during Von Recklinghausen's disease are rare [1]. Only a few cases have been reported in the international literature. We report the case of patient with pulsatile exophthalmos revealing spheno-spheno-orbital agenesis associated with Von Recklinghausen's disease.
Keywords: Pulsatile exophthalmos, Von Recklinghausen's disease, spheno-orbital agenesis.

INTRODUCTION

As a rule pulsating exophthalmos is of vascular etiology, which may be of traumatic, tumoral or malformative origin [2].

Unilateral pulsating exophthalmos, not accompanied by intracranial bruit in von Recklinghausen's disease is an extremely rare manifestation and is implicitly accompanied by changes in the wing of the sphenoid. The rare occurrence of this clinical entity justifies our report on a case recently admitted to our ophthalmology department.

CASE REPORT

Miss F L, aged 32, consulted for an irrepressible and painless right-sided unilateral exophthalmos with limitation of all ocular movements, without loss of visual acuity. The general examination of the patient revealed the presence of two soft temporo-palpebral masses, one right and the other left, as well as multiple café-au-lait spots. A cranio-orbital and facial spiral CT scan was performed, in axial sections (Fig. 1), with 2D coronal and sagittal reconstructions, and 3D volume rendering (fig. 2), showed the absence of the large wing of the right sphenoid associated with an orbital meningocele repressing and compressing the optic nerve and oculomotor muscles.

The diagnosis of neurofibromatosis type1 with agenesis of the great wing of the sphenoid was retained in this patient.

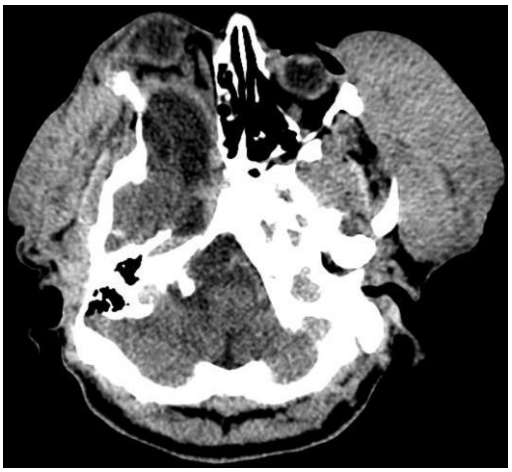

Fig-1: Orbito-cerebral TDM, showing agenesis of the large wing of the right sphenoid with orbital meningocele, associated with bilateral plexiform temporo pavascular neuromas

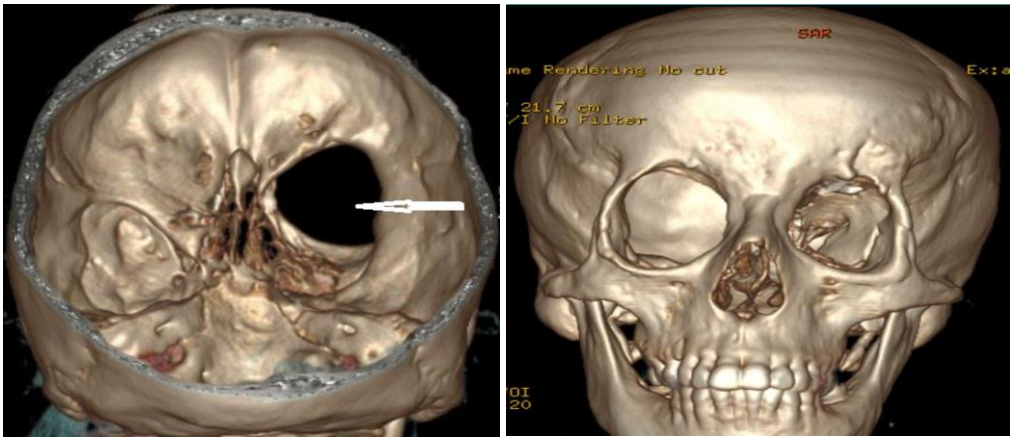

Fig-2: 3D volume reconstruction, showing the agenesis of the large wing of the right sphenoid

DISCUSSION

Pulsatile exophthalmos is a globular protrusion often resulting from a vascular pathology type cavernous carotid fistula, rarely found in the context of neurofibromatosis type 1 or Von-Recklinghausen [1].

Von-Recklinghausen disease is the most common phacomatosis, characterized by the presence of neurofibromas which are benign and heterogeneous tumors of the sheath of peripheral nerves disseminated in different territories (skin, subcutaneous tissue, deep tissue). Cephalic lesions represent 3 to 7% of the attacks during Von Recklinghausen neurofibromatosis [1-3].

The orbito-palpebral region is the preferred site of cephalic involvement during neurofibromatosis type I, characterized by neurofibromas like plexiform neuroma of the upper eyelid that are pathognomonic of NF1. It is associated with other orbitofacial manifestations, the most frequent being represented by hypertrophy of the ipsilateral hemiface and spheno-orbital dysplasia [1,3].

The latter achieves a total thinning or dehiscence of the sphenoid constitutive elements at the origin of a facio-orbital deformity with widening of the orbit, the superior orbital fissure and the temporal fossa, responsible for communication between the frontotemporal parenchyma and the bottom of the orbit performing a meningoencephalic hernia.

Clinically, it associates a plagiocephaly with a pulsatile exophthalmia which remains rare, due to the cerebro-meningeal expansion in the upper part of the orbit [4]. The dysplasia of the large sphenoid wing is associated with a plexiform neuroma in 50 to 100% of cases, which explains the progressive worsening of dysplasia with age [5].

Radiographically, standard radiography is a valuable aid for the detection of bone abnormalities during spheno-orbital agenesis, objecting to the absence of the large wing of the sphenoid, with enlargement of the orbit and elevation of the small wing of the sphenoid and roof of the orbit [6].

These bone lesions are better analyzed by TDM, which represents the main imaging medium for spheno-orbital dysplasia, showing partial or total dysplasia or a simple thinning of the large wing of the sphenoid [7].

MRI makes it possible to better study nerve tumors, in particular the plexiform neurofibroma, which appears to be relatively hyposignal relative to the muscle in T1, in hypersignal T2 if the lesion is bulky, a central hypo signal producing a characteristic cocarde appearance, the enhancement is variable: central, diffuse, peripheral, or target [8].

The surgical treatment of spheno-orbital dysplasia remains reserved for complicated cases of a meningoencephalic hernia that has led to significant exophthalmia or a reduction in visual acuity by compression of the optic nerve. However, Morax and Coll [9] propose a neurosurgical approach with reduction of meningoencephalic hernia and reconstruction of the orbital apex using bone grafts.

CONCLUSIONS

Orbito-sphenoid dysgenesis is a little known cause of exophthalmos. It must be evoked in the context of neurofibromatosis since the exophthalmos is pulsatile. Conversely, its finding must lead to a clinical and radiological examination for signs suggestive of recklinghausen neurofibromatosis.

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