

Congenital unilateral fibrosis, blepharoptosis, and enophthalmos syndrome

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Abstract

Congenital fibrosis syndrome is a rare disorder associated with restricted ocular movements, ptosis, and enophthalmos. Imaging may assist in ruling out orbital bony deformity and other pathology in surrounding structures. This report describes the use of combined positron-emission tomography/computed tomography scan to assist with the diagnosis and management of a patient with congenital fibrosis syndrome.

Key words: Blepharoptosis, Enophthalmos, Fibrosis, Positron-emission tomography, Tomography, X-ray computed

Introduction

Congenital unilateral fibrosis, blepharoptosis, and enophthalmos syndrome is the rarest form of congenital fibrosis of the extraocular muscles. The condition manifests in infancy by unilateral ptosis, enophthalmos, and strabismus of variable degrees. Only 13 patients have been reported in the literature.¹⁻⁴ Computed tomography (CT) and magnetic resonance imaging (MRI) may show a non-specific infiltrating orbital mass on the affected side. Biopsy is usually performed to confirm the diagnosis. This report is of the use of combined positron-emission tomography/CT (PET/CT) to assist the diagnosis and management of a patient with congenital fibrosis syndrome.

Case report

A 32-month-old boy presented with left ptosis since birth. His parents also noted that he had mild esotropia. He was delivered after an uncomplicated pregnancy. The family history was unremarkable.

Ocular examination showed blepharoptosis in the left eye with a 3-mm difference in vertical fissure height. There was no jaw-wrinkling phenomenon. Levator function was 10 mm and 13 mm in the left and right eyes, respectively. There was 1 mm of enophthalmos over the left eye. There was a 10-PD left esotropia. Ocular motility examination showed mild limitation in left eye abduction and depression (**Figure 1**). Atropinized refraction was +2 D in the right eye (visual acuity by Cardiff, 6.0/9.5) and +2.5 D in the left eye (visual acuity, 6.0/9.5). There was no scar over the eyelids, and anterior segment, papillary reaction, and fundal examination were all normal.

CT and MRI showed left globe enophthalmos. There was an ill-defined soft tissue density mass over the superomedial aspect of the left orbit arising from the superior rectus, levator palpebrae superioris, superior oblique, and lateral rectus muscles of the left eye, involving both the muscle bulk and the origins (**Figure 2**). The mass measured 8 x 6 mm, and was contrast enhancing. There was no bone erosion detected in the left orbit. Blood test results, including thyroid function, complete blood count, and erythrocyte sedimentation rate, were normal. Whole body F18-fluorodeoxyglucose (FDG) PET/CT scan was done to rule out any malignant infiltrative lesions such as leukemia or rhabdomyosarcoma in the orbit. There was no increase in FDG activity in the left orbital mass (**Figure 3**).

The boy was diagnosed with congenital unilateral fibrosis, blepharoptosis, and enophthalmos syndrome. The parents were reluctant for him to undergo surgery. No biopsy was done and he was managed conservatively. He has been followed up for 3 years and his condition is stable.

Discussion

Congenital fibrosis of the extraocular muscles is a rare condition. There are 5 subtypes: general fibrosis syndrome;

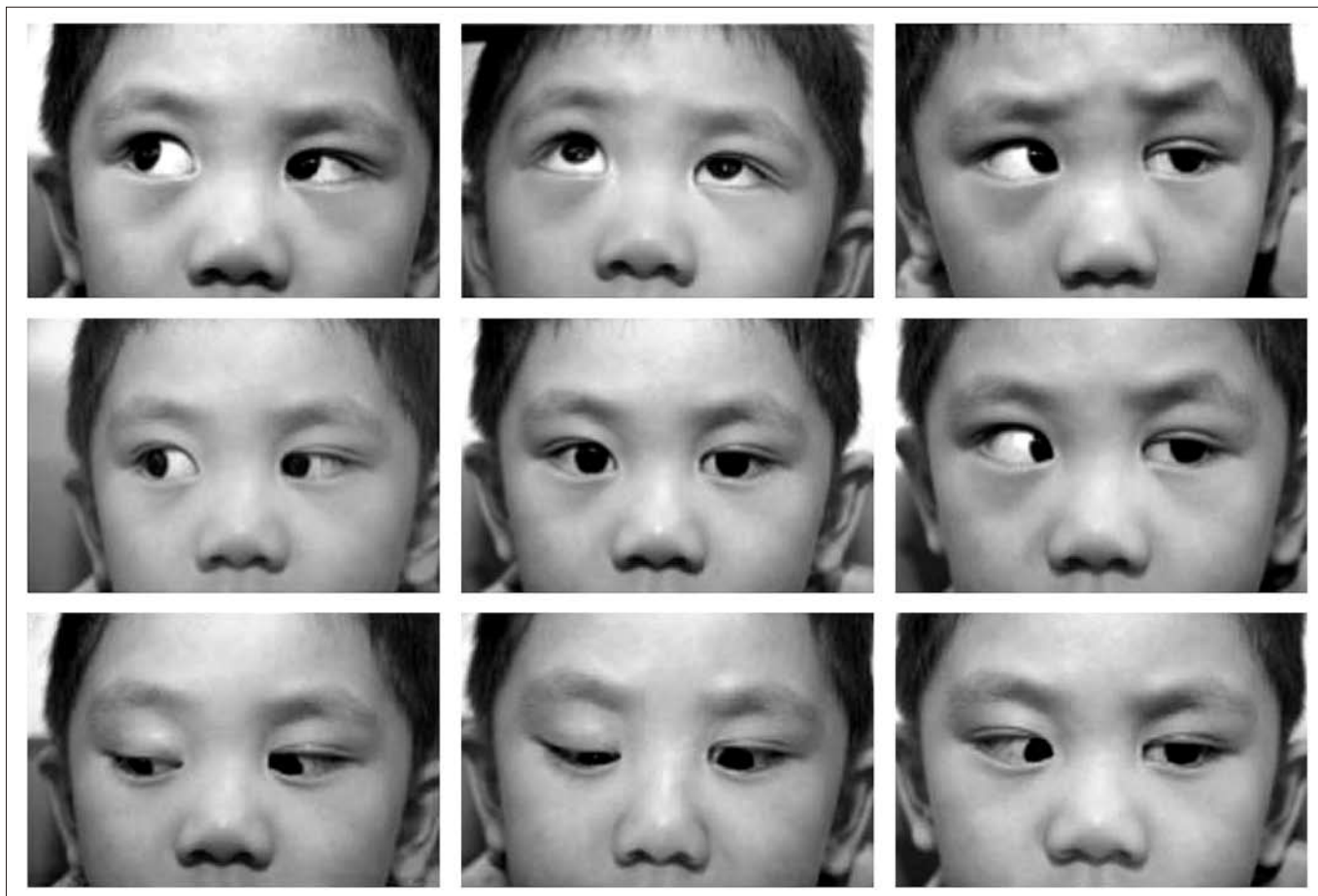


Figure 1. The patient at presentation showing left eye ptosis with enophthalmos and abnormal extraocular movements.



Figure 2. Precontrast T1-weighted axial magnetic resonance imaging showing an ill-defined soft tissue density mass over the superomedial aspect of the left orbit (arrow).

fibrosis of the inferior rectus with blepharophimosis; strabismus fixus; vertical retraction syndrome; and unilateral fibrosis, blepharoptosis, and enophthalmos syndrome.² The latter is the rarest form, with only 13 patients being reported in the literature.¹⁻⁴

Unilateral enophthalmos and blepharoptosis were the salient features for all the reported patients. Other variable associations were noticed. All 3 patients reported by Effron et al had scars over the lower lid, suggesting prenatal orbital

penetration injury as the cause of fibrosis.¹ In another series reported by Hertle et al, only 1 patient had a lower lid scar and the authors believed that the scar was representative of partial clefting syndrome.² There were no scars in the eyelids of the patient in this report. This patient had mild esotropia; other strabismus patterns have been reported in the literature depending on which extraocular muscles were involved.^{1,2} Ethmoidal sinus tumor was also found to be associated with this syndrome in 2 patients.^{1,4}

CT and MRI are used for investigation of this condition, but there are no reported pathognomonic imaging features to confirm the diagnosis radiologically. These investigations may show a variable degree of extraocular muscle thickening. An ill-defined intraconal or extraconal mass, which may or may not have contrast enhancement, will show up on a CT scan. On MRI, an ill-defined mass would be isointense to the muscle cone. Other malignant conditions such as leukemia, lymphoma, or rhabdomyosarcoma would also have similar imaging findings.

For this patient, the radiological findings were non-specific, and usually an open biopsy is needed to rule out malignancy and to confirm the diagnosis. However, F18-FDG PET/CT scan can help to distinguish the nature of the intraorbital mass. Malignant infiltrative masses such as leukemia, lymphoma, or rhabdomyosarcoma would cause an increase in uptake of 18-FDG, which will show up as a hot spot in the

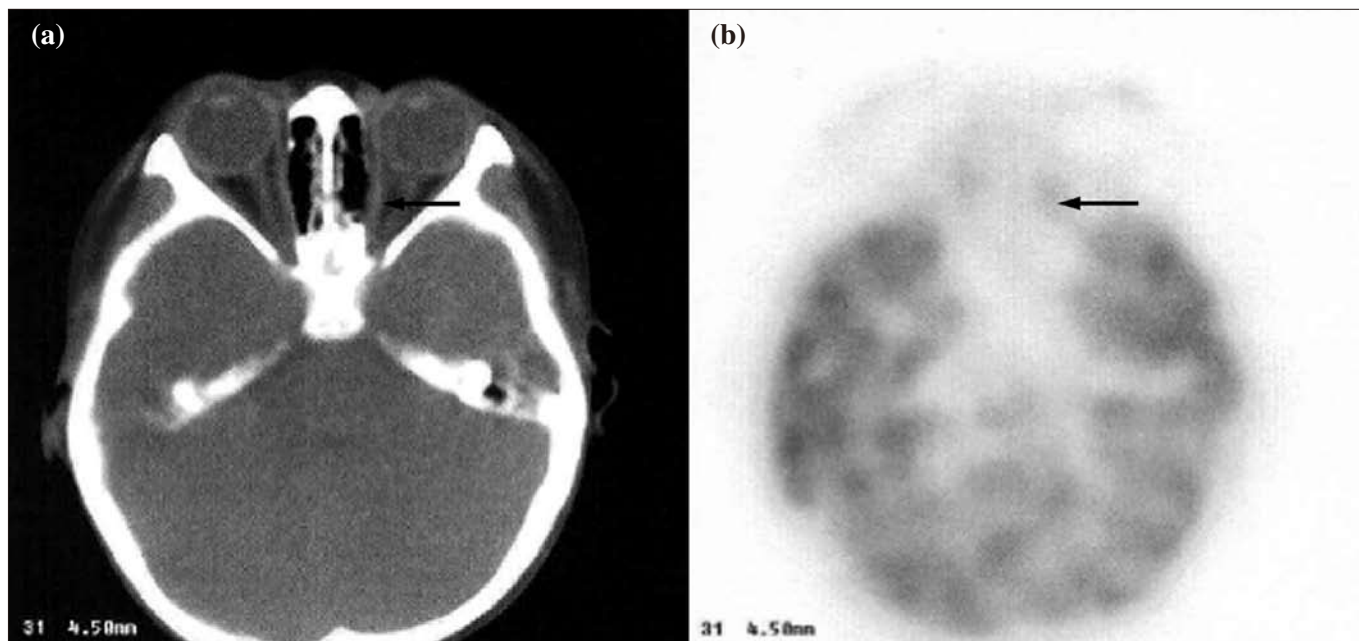


Figure 3. Combined positron-emission tomography/computed tomography scan showing (a) the ill-defined mass; and (b) no increase in F18-fluorodeoxyglucose activity (arrow).

PET/CT scan, while fibrotic tissue would show up as a cold spot, as it would not take up 18-FDG.⁵

A conservative approach to the management was adopted for this patient, as there was no amblyopia and enophthalmos was minimal. However, the management of this rare disease

should be tailored for individual patients. Visual acuity and refractive errors should be monitored, and treatment for amblyopia should be commenced when indicated.² Ptosis surgery can also be considered for patients with the visual axis obscured. Correction of enophthalmos can be corrected with a wedge operation.³

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