Rare disease

Congenital orbital fibrosis associated with fibrosis of extraocular muscle

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Summary

Congenital orbital fibrosis is a non-familial, unilateral, distinct clinical entity, characterised by the presence of a diffusely infiltrating orbital mass and is extremely a rare disease. Congenital orbital fibrosis with combined fibrosis of extraocular muscles have not been reported previously. We treated an 8-year-old boy with the presence of a diffusely infiltrating orbital mass and fibrosis of extraocular muscles with secondary involvement of extraocular muscles. Clinical examination revealed left exotropia, hypotropia and fibrosis of extraocular muscle, an irregular, retrobulbar mass located within the orbit, incorporating the optic nerve, medial, superior, inferior and lateral rectus muscle. The CT, MRI and light microscopic studies confirmed the diagnosis. We performed exploration of the orbit, release and biopsy of scar tissue and strabismus surgery. Unlike other reported cases, our case was a progressive congenital disorder with combined fibrosis of extraocular muscle.

BACKGROUND

Unilateral fibrosis, blepharoptosis and enophthalmos syndrome was first described by Leone *et al* in 1972.¹ Since then 18 cases have been reported worldwide in the literature.^{2–8} Mavrikakis *et al*⁷ identify this syndrome as a distinct clinical entity and use the general term congenital orbital fibrosis instead of unilateral fibrosis, blepharoptosis and enophthalmos syndrome. This disorder occurs in infancy as unilateral blepharoptosis, proptosis or enophthalmos, limitation of the extraocular movements, strabismus and decreased vision, usually due to amblyopia. Unlike other reported cases, our case was a progressive congenital disorder with combined fibrosis of extraocular muscle.

CASE PRESENTATION

An 8-year-old boy was referred to the clinic with abnormal left extraocular movements since birth. The patient was born uneventfully at full term with no significant medical or family history. On examination, he was found to be visually attentive with right eye. Uncorrected visual acuity was 40/60 ocular dexter (OD) and 20/60 ocular sinister (OS). Cycloplegic refraction was +1.50+1.00×90 OD and +2.50+3.00×90 OS. There was a downward and axial displacement of the left globe associated with 10 mm of medial scleral show. There was a 65-prism diopter left exotropia and a 35-prism diopter left hypotropia in primary gaze. Note the ipsilateral proptosis 2 mm. Extraocular movement examination revealed fibrosis of extraocular muscle and complete absence of adduction and minimal elevation of the left eye. Corneal reflexes were used for this measurement. The extraocular movements of the right eye were normal (figure 1).

INVESTIGATIONS

Ultrasound examination was normal. He underwent two CT scans at the age of 5 (figure 2A,B) and 8 (figure 3A,B). Repeat CT scan showed obvious change of the morphology

of the left orbit. CT demonstrated the normal orbital wall and an intraconal soft-tissue mass in the left orbit abutting the posterior aspect of the globe. The mass incorporated the optic nerve, the medial, lateral, superior and inferior rectus muscle. The globe was proptotic and inferiorly displaced.

MRI showed proton density image of left orbit. Corresponding axial MRI of the mass in left orbit show T1-weighted isointense to muscles (figure 4A), T2-weighted (figure 4B) isointense to muscles and gadolinium-enhanced T1-weighted (figure 4C) hyperintensity. Coronal MRI T2-weighted showed the mass isointense to muscles incorporated the optic nerve, the medial, lateral, superior and inferior rectus muscle (figure 4D).

DIFFERENTIAL DIAGNOSIS

- Orbital vascular malformation
- Congenital general fibrosis of extraocular muscles

TREATMENT

This boy underwent exploratory surgery via lateral and inferior fornix conjunctivae incision and biopsy. We found the inferior and lateral rectus muscle was fibrosis. The lateral and inferior rectus muscle was released from scar tissue. Meanwhile lateral rectus muscle suspension surgery was performed. Primary position of eye was normal after postoperative (figure 5). Histopathology of excised orbital fat demonstrated dense focal fibrosis without inflammation (figure 6).

OUTCOME AND FOLLOW-UP

During the 10 months of follow-up after surgery, the patient had no evidence of local recurrence and the primary position of the eye was normal.

DISCUSSION

We report the first case of congenital orbital fibrosis with combined fibrosis of extraocular muscle. Congenital

Figure 1 An 8-year-old boy presented with an absence of adduction and minimal elevation of the left eye since birth. Note the ipsilateral proptosis, hypoglobus and exotropia.

Figure 2 CT scan was performed at the age of 5. (A) CT axis demonstrated a heterogeneous retrobulbar density in the left orbit obscuring the optic nerve, and the medial and lateral rectus muscles with associated proptosis. The bony orbit appeared to be within normal limits. (B) CT coronal demonstrated a heterogeneous retrobulbar density in the left orbit obscuring the optic nerve, and the medial and lateral rectus muscles. The appearance of the right eye, orbit and orbital contents were normal.

Figure 3 CT scan was performed at the age of 8. (A) CT axis demonstrated a heterogeneous retrobulbar density in the left orbit obscuring the optic nerve, and the medial and lateral rectus muscles with associated proptosis. The bony orbit appeared to be within normal limits. (B) CT coronal demonstrated a heterogeneous retrobulbar density in the left orbit obscuring the optic nerve, and the medial, superior, lateral and inferior rectus muscles. The appearance of the right eye, orbit and orbital contents were normal.

Figure 4 Distinctive MRI characteristics of left orbit. Corresponding axial MRI of the mass in left orbit show T1-weighted isointense to muscles (A), T2-weighted (B) isointense to muscles and gadolinium-enhanced T1-weighted (C) hyperintensity. MRI coronal (D) T2-weighted sequences show isointense to muscles.

orbital fibrosis is a non-familial, unilateral, distinct clinical entity, characterised by the presence of a diffusely infiltrating orbital mass with secondary involvement of extraocular muscles resulting in variable symptomatology due to the cicatricial process. Our case with proptosis and fibrosis of extraocular muscle did not demonstrate blepharoptosis and enophthalmos. CT scan shows the presence of an irregular, diffusely infiltrating, retrobulbar mass located within the orbit. The optic nerve, the medial, lateral, superior and inferior rectus muscle was involved in our patient. Of the 18 previously reported cases, 14 had supportive radiological imaging, either CT or MRI.^{5–11} Of these, 12 had an associated diffusely infiltrating orbital mass, while the remaining two had an adjacent benign ethmoidal sinus tumour with encroachment into the orbit, one of which was a mesenchymoma. These two cases should be considered separately from the other cases.⁵ The medial rectus was involved in nine cases, inferior rectus in seven, the superior rectus in four, the superior

oblique in three, the lateral rectus in two strabismus, both horizontal and vertical, was present in all the patients in this series.

Unlike other reported cases, our patient had unilateral orbital fibrosis with proptosis, fibrosis of extraocular muscle and abnormal movements of the extraocular muscles. Meanwhile, the diffusely infiltrating orbital mass was progressive. Although treatment of these patients is difficult, visual rehabilitation can be achieved. This is accomplished with occlusion for amblyopia and spectacle correction of refractive errors. The goal of strabismus surgery is to release muscle adhesions and to achieve optimal alignment in the primary position.⁴ Our patient was performed exploration of the orbit, release and biopsy of scar tissue and strabismus surgery.

The aetiology of congenital orbital fibrosis is unknown and is likely related to prenatal insults. No family history was forthcoming to substantiate a familial disease in our

Figure 5 An 8-year-old boy presented with normal primary position of eye after postoperative.

Figure 6 Tissues obtained from the orbital apex adjacent to the extraocular muscles demonstrate fibrosis tissues (H&E, original magnification $\times 200$).

patient. In addition, there was no history of trauma, infection, or inflammation in childhood.

Our case demonstrates congenital orbital fibrosis to be progressive congenital disease and sometimes associated with fibrosis of extraocular muscle. The objective of treatment is to correct strabismus, ptosis and amblyopia.

Learning points

- ▶ Congenital orbital fibrosis with combined fibrosis of extraocular muscle is a rare disease.
- ▶ The synergy of detailed history, careful ocular and systemic examination, CT and MRI are essential in the diagnosis.
- ▶ The objective of treatment is to correct strabismus, ptosis and amblyopia.

Competing interests None.

Patient consent Obtained.

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