· Case report ·

# A distinct and sporadic case of congenital fibrosis of the extraocular muscles

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## Abstract

• AIM: To report a distinct and sporadic case of congenital fibrosis of the extraocular muscles(CFEOM).
• METHODS:A case report.
• RESULTS: A 3-year-old Chinese boy who presented with exotropia and ptosis was diagnosed with CFEOM. Abnormal optic nerves were depicted by fundus examination, neuroradiology and electrophysiology. Pathological study showed muscle fibers were atrophic degeneration.
• CONCLUSION: Our case highlights that CFEOM may have abnormal optic nerve and nontypical eye movement. The pathological study could help to find out the muscle fibers' alterations.
• KEYWORDS:children; strabismus; surgery
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## INTRODUCTION

Congenital fibrosis of the extraocular muscles (CFEOM) is characterized by congenital restrictive ophthalmoplegia. Its prevalence rate is 1/230000. A 3-year-old Chinese boy who presented with exotropia and ptosis was diagnosed with CFEOM. Abnormal optic nerves were depicted by fundus examination, neuroradiology and electrophysiology. Pathological study showed muscle fibers were atrophic degeneration.

## CASE REPORT

A 3-year-old Chinese boy presented with abnormal eye position and ptosis right after his birth. The family history and consanguinity marriage history were negative. Horizontal and vertical movements were absent in the right eye. In the left eye adduction and vertical movements were absent, abduction was severely limited, and the globe slightly retracted during attempted abduction. The lower lids were pulled down during jaw opening (Figure 1). The pupils' diameter was 4 mm with negative light reflex. The optic discs were pale (Figure 2).

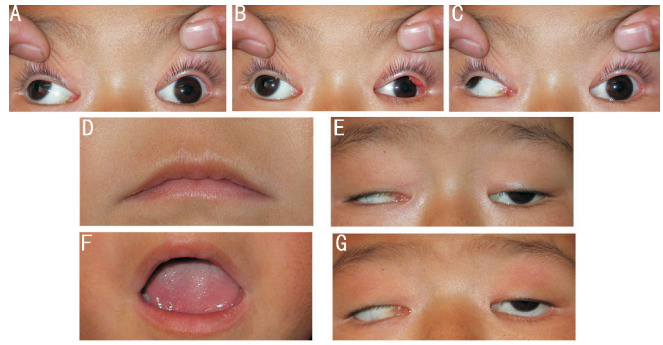


**Figure 1** A, C: In primary position of the right eye, exotropia and hypertropia existed, accompanied by pendular nystagmus. The left eye position was normal. B: The left globe slightly retracted during attempted abduction. D-G: Bilateral ptosis with the upper eyelids covered to the horizontal level of the pupil's center. D, E: In primary position, the palpebral fissures narrowed with jaw closing. F, G: Lower lids were pulled down during jaw opening.

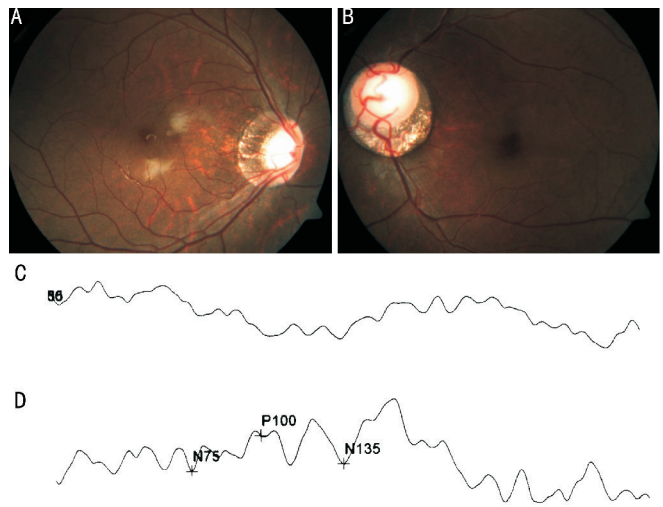


**Figure 2** A, B: The optic discs were pale, and the left optic disc was hypoplastic (B); C: The VEP wave shapes were flat in the right eye; D: The left eye's latencies were normal, while the VEP amplitudes were decreased. In the left eye, the latent periods at N75, P100, and N135 were 76ms, 110ms, 150ms respectively; the amplitudes at Ap-100 and An-135 were 7. 13μV and 5. 83μV respectively.

The cycloplegic refraction result was +0. 50DS/-5. 00 DC×60 in the right eye and -0. 50 DS in the left. After anesthesia, forced duction test showed abduction was positive, with minimal movements, and the other ductions were negative. Magnetic resonance imaging (MRI) of the orbits and brain showed the eyes were not round. The right optic nerve took a circuitous course, which compressed the neighbouring medial

rectus (Figure 3). Visual evoked potential (VEP) wave shapes were abnormal in both eyes (Figure 2).

The patient underwent medial rectus muscle resection of his right eye to correct strabismus. The resected tissue was used as a biopsy specimen. Pathological study showed atrophic degeneration existed in the muscle fibers (Figure 4). Four weeks after surgery, the right eye in primary position was almost in the normal position.

## DISCUSSION

Congenital fibrosis of the extraocular muscles (CFEOM) is characterized by congenital restrictive ophthalmoplegia affecting muscles in the oculomotor and trochlear nerve distribution. Its prevalence rate is 1/230000[1]. It has different phenotypes of CFEOM1, CFEOM2, and CFEOM3. CFEOM2, characterized by exotropia and bilateral blepharoptosis, is an autosomal recessive disease. The ocular motor defect is congenital and non-progressive. It has been reported that miosis with no light reflex presents in a consanguineous pedigree[1]. Our case met clinical criteria for CFEOM2. The patient had bilateral blepharoptosis and exotropia accompanied by nystagmus in the right eye. The ocular motility was severely limited or absent in both eyes. As a sporadic case, the patient's pupils were normal size with absent light reflex. His optic discs were pale. The left globe was slightly retracted without narrowing of the palpebral fissure during attempted abduction. His lower lids were pulled down rather than the upper lids were lifted during jaw opening. Ophthalmic problems associated with sporadic CFEOM include choroidal coloboma, microphthalmos with corneal opacity, congenital cataract, retinitis pigmentosa, optic atrophy, optic disc hypoplasia, and Marcus Gun jaw winking[3-6].

The MRI of the brain and orbits depicted nonspherical eye shapes and abnormal right optic nerve. Compared to the left eye, the right eye's rectus muscles were thinner. It has been reported that oculomotor nerve is absent and extraocular muscles innervated by it are reduced in volume[7]. Other types of CFEOM sometimes reveal volume reduction of optic nerve, anomalous oculomotor nerve and extraocular muscles[8]. The VEPs showed anomalous changes in both eyes, which indicated abnormalities existed in the visual pathway. This result could be explained by the abnormal right optic nerve in MRI and the hypoplastic left optic disc.

There are recommendations for CFEOM's treatment. Amblyopia should be aggressively treated prior to surgery. Blepharoptosis surgery should be delayed until the extraocular muscle surgery has been completed[9]. As the patient had abnormal optic nerves, the vision could hardly be increased. We performed cosmetic surgery and got amazing follow-up outcome.

Pathological study showed muscle fibers were atrophic degeneration. The skeletal muscle fiber had fat and hyaline degeneration in fibrous tissue, unequal diameters and moderately variable shapes. Muscle fibers showed clustering phenomenon, and enzymatic activity was increased. Nuclear ingression, nuclear chain and spiral-shaped muscle fibers were observed. Glycogen was reduced or deficient. Lymphocyte infiltration was scattered. Fibrous tissue was markedly hyperplastic. Histopathological studies of extraocular muscles from sporadic cases of CFEOM often reveal the presence of multiple dense bands of connective tissue with some areas of normal striated muscle tissue. The areas of muscle with fibrous tissue are characterized by reduced number of fibers that mix into the fibrotic Tenon's capsule. The muscle myofibrils appear abnormal, and the mitochondria show degenerative changes[4]. Lipid vacuoles are also found. Inflammatory cells

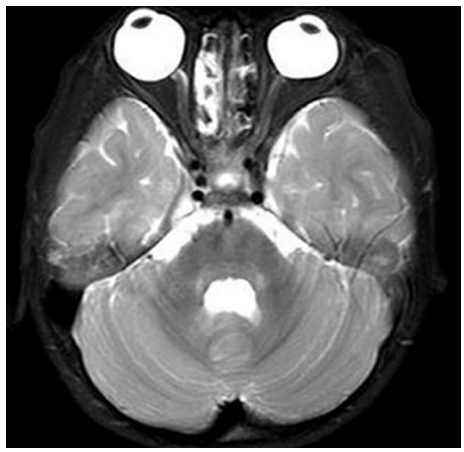

**Figure 3 MRI of the brain and orbits. Compared to the left eye, the right eye's medial, superior and inferior rectus muscles were thinner, and the fluid in the optic nerve sheath cavity was greater.**

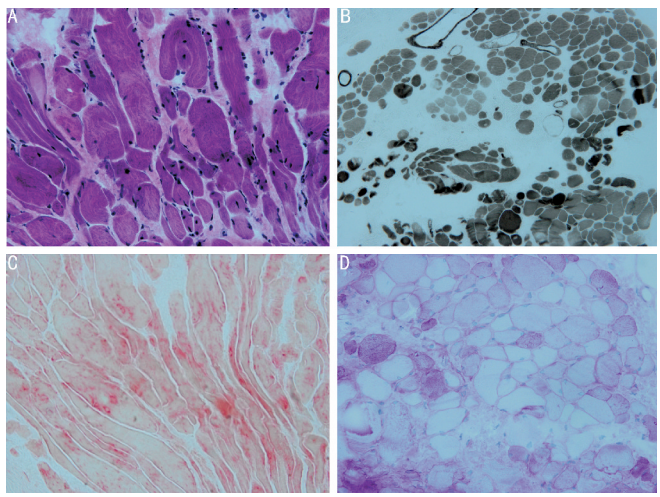


**Figure 4 Histopathology with magnifications at ×100 (A), ×200 (B), ×400 (C, D)** A: HE staining showed the skeletal muscle fiber had fat and hyaline degeneration in fibrous tissue, unequal diameters and moderately variable shapes. Lymphocyte infiltration was scattered. Fibrous tissue was markedly hyperplastic; B: In ATPase staining, type II muscle fiber showed clustering phenomenon; C: In ACPase staining, enzymatic activity in muscle fibers was increased; D: In PAS staining, glycogen in muscle fibers was reduced or deficient.

are absent. The rectus muscles often have anomalous insertions that appear to fall short of their destination[9].

In conclusion, this case highlights that CFEOM may have abnormal optic nerve and nontypical eye movement. The pathological study could help to find out the muscle fibers' alterations.

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## 先天性眼外肌纤维化 1 例

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**摘要**

**目的**:报道 1 例少见的先天性眼外肌纤维化(congenital fibrosis of the extraocular muscles,CFEOM)病例。

**方法**:病理报告。

**结果**:一名 3 岁的中国男孩表现为眼外斜和上睑下垂,被诊断为 CFEOM。眼底、神经影像学、电生理检查发现异常视神经。病理学显示眼外肌萎缩变性。

**结论**:本病例提示 CFEOM 可合并异常视神经和非典型眼运动。病理学检查有助于发现眼外肌纤维的病变。

**关键词**:儿童;斜视;手术