

**Case report:**

**A Rare Case of Twins with Bilateral Congenital Ptosis – A Case Report**

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**Abstract:**

Cases of congenital ptosis are rare, let alone bilateral and occurring in twins. There are many diagnostic dilemmas, often needing genetic testing and advanced imaging such as magnetic resonance imaging. This case report will look at the case of a twin with this interesting feature who are presented to a primary care clinic. It will center around how the diagnosis of congenital fibrosis of extraocular muscles was reached and what management options are available for this twin.

**Keywords:**Twins, congenital ptosis, primary care, surgery, blepharoptosis.

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**Introduction**

The incidence of congenital ptosis worldwide is unknown; only 30% of the cases involve both eyes.<sup>1</sup> Causes of congenital ptosis can be divided into ptosis that occurs in isolation (simple ptosis) and ptosis associated with other ocular findings or systemic disorders.<sup>2</sup> Examples include blepharophimosis-ptosis-epicanthus inversus syndrome (BPES), congenital fibrosis of extra-ocular muscles, Horner's syndrome, congenital myasthenic syndrome, Marcus Gunn jaw winking, third cranial nerve palsy and upper eyelid mass (for example plexiform neurofibroma, dermoid cyst).<sup>2</sup> Determining the causes will enable proper pre-operative assessment and treatment of the underlying cause with accurate prognostication.

**Case presentation**

A 3-year-old boy presented with upper respiratory tract infection symptoms for two days in a primary care clinic in Selangor, Malaysia. On examination, it was noted that the child and his twin brother have bilateral ptosis. The ptosis was present at birth but only noted by the parents at one month of life. During this presentation, the parents denied any visual problems such as reflective errors, strabismus or abnormal head posture. Both

children had been diagnosed as having congenital fibrosis of extraocular muscles (CFEOM) by the pediatrics and ophthalmology team. This was based on the clinical presentation. Parents have refused genetic testing and any further surgical intervention to address the extraocular muscle weakness.

Examination of both eyes revealed severe ptosis with slight chin lift and excessive use of frontalis muscles to compensate the ptosis. The visual acuity of the patients was normal with restriction of lateral gaze (Figure 1, 2).



Figure 1 : Eyes inspection of the first twin.

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Figure 2 : Eyes inspection of the second twin.

The parents were reassured on this eye condition. However, they were advised, if the twins complained of any visual abnormality, they should be brought urgently to the ophthalmologists or primary care physicians for further assessment.

### **Discussion**

Ptosis is defined as drooping of the upper eyelid and can be categorized as minimal (1-2 mm), moderate (3-4 mm) or severe(>4mm). It may either be acquired or congenital and unilateral or bilateral.

Bilateral causes of ptosis include as in this case; congenital fibrosis of extraocular muscles (CFEOM). Other causes are blepharophimosis-ptosis-epicanthus inversus syndrome (BPES), congenital myasthenic syndrome or chronic obstructive external ophthalmoplegia.<sup>2</sup>

CFEOM is a congenitally rare eye condition that is characterized by non-progressive ophthalmoplegia with or without ptosis. Its prevalence is 1:230,000.<sup>3</sup> There are three subtypes, CFEOM1, CFEOM2, and CFEOM3. CFEOM1 clinically involves bilateral ptosis and downward rotation (infraduction) of the eyes in primary position, while CFEOM2 is bilateral ptosis with outward rotation of the eye (exotropia). CFEOM3 is a variable clinical presentation. Both CFEOM1 and CFEOM3 autosomal dominant while CFEOM2 is autosomal recessive. Tukel syndrome is CFEOM3 phenotype with congenitally missing fingers or toes (oligodactyly or oligosyndactyly).<sup>4</sup>

Examination would require complete eye assessment including assessment of visual acuity, refraction, fundoscopic and slit lamp examinations as well as assessment of pupillary reactivity.

Treatment will aim at partially correcting the extraocular weakness to ensure vision that is functional as well as addressing the ptosis itself. Surgery for ptosis will be indicated if there are occlusion of the visual axis, risk of amblyopia from astigmatism due to compression on the ptotic lids onto the cornea, excessive or unacceptable abnormal head posture, or cosmetic concerns. There are numerous approaches that can be used to surgically repair ptosis, and the three common approaches are Muller's muscle conjunctival resection (MMCR), frontalis sling and levator advancement.<sup>5</sup> Surgery can also improve alignment of the eyes but does not restore normal function.<sup>4</sup> A successful surgical repair can often produce good cosmetic and functional results. However up to 50% of children may require another surgery in 8-10 years' time.<sup>1</sup>

This is a rare genetic case found in primary care with patients presenting with URTI. During the writing of this case, the authors found that the parents of the patients have declined any surgical intervention for the moment and have defaulted follow up. The role of primary care here is to continue seeing the patients for these types of cases and at the same time to look for ophthalmic complications such as worsening visual problems, amblyopia, dry eyes and musculoskeletal complications due to abnormal head posture to compensate for the ptosis or amblyopia.<sup>3</sup>

Another important issue is the genetic counselling for the parents as this condition may be autosomal dominant or recessive. It is important to relay to the parents and other family members on preconception and premarital counselling as there have been cases of CFEOM in consanguineous marriage.<sup>3</sup>

Hence, facilitating combined care with the relevant departments for these patients is essential based on their progress. Continued education, counselling and support is needed for this type of rare condition.

### **Conclusion**

Congenital fibrosis of extraocular muscle (CFEOM) is rare genetic disorders which presents with bilateral ptosis in infancy and among toddlers. If it is left untreated, complications such

as amblyopia and musculoskeletal problems can arise. Genetic counselling is important due to its autosomal dominant or recessive feature.

Cases seen in primary care are usually common cases<sup>6-7</sup>, however occasionally there are instances of very rare genetically linked conditions. Therefore, primary care physicians should familiarise themselves with such cases. In some cases where the patients or their guardians refuse for further assessment or treatment, follow-up should be advised to them in order to monitor for any potential complications, which may arise at any stage.

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### **Conflict of Interest**

No conflict of interest has been disclosed by the authors.

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