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RESEARCH ARTICLE

BILATERAL CONGENITAL PTOSIS WITH CONGENITAL FIBROSIS OF EXTRAOCULAR MUSCLES (CFEOM) IN MONOZYGOTIC TWINS

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Abstract

Cases of congenital ptosis are rare, that too occurring in twins. We are presenting a case of mono zygotic twins with bilateral ptosis with congenital fibrosis of extraocular muscles (CFEOM). A 5-year-old boy child and his twin brother who are monozygotic twins presented with bilateral ptosis. Both children had been diagnosed as having congenital fibrosis of extraocular muscles (CFEOM). Parents have refused surgical intervention for the extraocular muscle weakness. They were advised, if the twins complained of any visual abnormality, they should be brought to the ophthalmologists for further assessment.

Introduction:-

The incidence of congenital ptosis world wide is unknown; only 30% of the cases involve both eyes.[1]

Causes of congenital ptosis can be divided into isolated (simple ptosis) and ptosis associated with other ocular findings or systemic disorders.[2]

Examples of ptosis associated with other disorders include blepharophimosis-ptosis epicanthus inversus syndrome (BPES), congenital fibrosis of extra-ocular muscles (CFEOM), Horner's syndrome, congenital myasthenic syndrome, Marcus Gunn jaw winking, third cranial nervepalsy and upper eyelid mass (for example plexiform neurofibroma, dermoid cyst).[2]

Case presentation

A 5-year-old boy child and his twin brother who are monozygotic twins presented with bilateral ptosis. The ptosis was present at birth but only noted by the parents at 2 months of life. During this presentation, the parents denied any visual problems. Both children had been diagnosed as having congenital fibrosis of extraocular muscles (CFEOM).

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.

The parents were reassured on this eye condition as their parents refused for surgical intervention for the extraocular muscle weakness. However, they were advised, if the twins complained of any visual abnormality, they should be brought to the ophthalmologists for further assessment.

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We are unable to produce photos for this case report as parents objected for taking photos of their children

Discussion:-

Ptosis is defined as drooping of the upper eyelidand 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 thiscase; congenital fibrosis of extraocular muscles(CFEOM). Other causes are blepharophimosis ptosis-epicanthus inversus syndrome (BPES), congenital myasthenic syndrome or chronic obstructive external ophthalmoplegia.[2]

CFEOM is a congenitally rare eye condition that ischaracterized by non-progressive ophthalmoplegiawith or without ptosis. Its prevalence is1:230,000.[3] There are three subtypes, CFEOM1,CFEOM2, and CFEOM3. CFEOM1 clinically involves bilateral ptosis and downward rotationof the eyes in primary position,while CFEOM2 is bilateral ptosis with outward rotation of the eye (exotropia). CFEOM3 is variable clinical presentation. Both CFEOM1 andCFEOM3 autosomal dominant while CFEOM2 isautosomal recessive.

Examination require complete eye assessment including assessment of visual acuity, refraction, fundoscopic and slit lamp examinationsas 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 is occlusion of the visual axis, risk of amblyopia from astigmatism due to compression on the ptoticlids onto the cornea, excessive or unacceptable abnormal head posture, or cosmetic concerns.There are numerous approaches that can be usedto surgically repair ptosis, and the three common approaches are Muller's muscle conjunctival resection (MMCR), frontalis sling and levatoradvancement.[4]Surgery can also improve alignmentof the eyes but does not restore normal function. A successful surgical repair can often producegood cosmetic and functional results. However upto 50% of children may require another surgery in8-10 years' time.[1]

The parents of the patients declined surgical intervention for the moment and have defaulted follow up. The role of ophthalmolgists 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 tocompensate for the ptosis or amblyopia.[3]

Another important issue is the genetic counseling for the parents as this condition may be autosomaldominant 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.[3]

Continued education,counselling and support is needed for this type of rare condition.

Conclusion:-

Congenital fibrosis of extraocular muscle(CFEOM) is rare genetic disorder 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.

In 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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