

Congenital ptosis: Etiology and its management

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Abstract

The ptosis is drooping of the upper eyelid below its normal position 2mm below the superior corneal limbus in primary gaze. It may hinder partially or completely the upper visual field and cosmetically unacceptable. According to age of onset ptosis can be categorized as congenital or acquired. Congenital ptosis is if it is present at birth or diagnosed during the first year of life and can be treated by resection of the levator muscle aponeurosis and the frontalis suspension. The levator muscle function is the main parameter to decide the surgical technique. Frontalis suspension and the supra-maximal resection of the levator muscle is required with weak levator function but if its function is above 4 or 5mm, the resection of the levator aponeurosis is preferred.

Introduction

Ptosis or blepharoptosis (“toe-sis”) refers to the unilateral or bilateral drooping of the upper eyelid. It usually occurs due to partial or complete dysfunctional levator palpebrae superioris(LPS) and the Muller’s muscle that elevate the upper eyelid.⁽¹⁾ The anatomical structures involved in upper eyelid elevation include three muscles: LPS, Müller’s smooth muscle and frontalis muscle innervated by the oculomotor nerve, sympathetic innervation and facial nerve respectively.

Ptosis is characterised by abnormal positioning of the upper eyelid below its normal level, 0.5-2mm below the superior limbus in the primary gaze.⁽²⁾ Normally upper eyelid covers the corneal limbus by 2 mm at the 12 o’clock position and the difference between normal position of lid and upper eyelid margin with ptosis was first parameter used by Beard⁽³⁾ for its classification as mild (1.52 mm), moderate (3 mm) and severe (4 mm).

Classification of ptosis

Ptosis can be classified as congenital or acquired based on age of onset if present at birth or within the first year of life is congenital ptosis. Usually this is an isolated entity and rarely may be non-isolated associated with other findings.

Ptosis which present after one year is termed acquired ptosis which may be an isolated or non-isolated. On basis of etiology ptosis may also be classified: aponeurotic, neurogenic, myopathic, neuromuscular, neurotoxic, traumatic, mechanical and pseudoptosis. Its early diagnosis and treatment prognosticate its management.

In United States the frequency of congenital ptosis has not been reported and 75 percent of the cases are unilateral.⁽⁴⁾ The majority of congenital ptosis is due to myogenic dysgenesis of the LPS in which fibrous and adipose tissue are present in the muscle belly.⁽⁵⁾ This impairs the ability of muscle to contract and elevate the eyelids.⁽⁶⁾ Congenital ptosis is mainly isolated and does not affect vision. Drooping of eyelid may occlude all or

part of the pupil and may interfere with vision, resulting in amblyopia in severe ptosis.⁽⁷⁾ Ptosis can result in astigmatism due to compression of the eye ball by ptotic lid.⁽⁸⁾ In cases of severe bilateral congenital ptosis compensatory head posture and chin up position may be adopted by children to obtain good vision. Some children may lift the ptotic eyelid mechanically to see clearly.⁽⁹⁾ Congenital ptosis shows autosomal dominant inheritance.⁽¹⁰⁾ Genetic or chromosomal defects are likely due to common familial occurrences.

Isolated congenital ptosis

Simple congenital ptosis: It is most common type of congenital ptosis due to a dystrophy of the LPS. The child usually manifest with a lid lag on down-gaze and poorly formed or higher than normal lid crease with lagophthalmos rarely.

Synkinetic ptosis: An aberrant innervation of the levator muscle by the mandibular branch of the trigeminal nerve, congenital ptosis results in Marcus Gunn Jaw-Winking Syndrome rarely. In this syndrome, brisk upper lid retraction is seen during mastication, jaw thrusting to the contralateral side, chewing, smiling, jaw protrusion or sucking due to ipsilateral pterygoid muscle contraction.⁽¹¹⁾ This phenomenon is seen in infants while bottle- or breastfed.

Aponeurotic ptosis: If aponeurosis fails to insert on the anterior surface of the tarsus or from birth trauma following forceps delivery results in congenital aponeurotic defects. Depending on where the aponeurosis is affected, skin crease may remain normal or high without lid lag on down-gaze with good levator function.

Non-isolated congenital ptosis: Superior rectus weakness may be associated with congenital ptosis as embryological development of the levator and superior rectus muscles are nearby.

Blepharophimosis Syndrome: It is rare autosomal dominant condition. The characteristic features are blepharophimosis, telecanthus, epicanthus inversus and

ptosis.⁽¹²⁾ With this disorder amblyopia association is very high.⁽¹³⁾ Other systemic associations include ovarian failure, arched palate and cardiac defects.

Neurogenic Ptosis: In infants ptosis, miosis, anhidrosis and progressive heterochromia can be seen in Horner's Syndrome. The lighter colored iris is seen on affected side. The lesion may be occur along the oculosympathetic pathway. It is important to rule out etiologies⁽¹⁴⁾ such as congenital varicella, vascular lesions of the internal carotids or subclavian artery and tumors of the neck and mediastinum.

Congenital Third Cranial Nerve Palsy: It may be partial or complete. It may present with ptosis with inability to elevate, depress or adduct the eye with pupillary dilatation. Pupil may be paradoxically small and non-reactive as signs of aberrant regeneration. Other causes of ptosis in children

- Traumatic birth
- Duane syndrome where the lateral rectus muscle acquires an innervation from the third cranial nerve despite of sixth nerve. Enophthalmos with apparent ptosis may result although the synkinesis produced does not involve lid innervations.
- Periorbital tumor or other deep orbital tumors may produce proptosis with ptosis.
- Congenital Fibrosis of the Extraocular Muscles(CFEOM) is a non-progressive, autosomal dominant ocular disorder, resulting in fibrosis of the extraocular muscles.⁽¹⁵⁾ This disorder is characterized by bilateral ptosis, external ophthalmoplegia, with a compensatory backward tilt of the head. The disorder could be due to myopathic or neurogenic etiology.
- Kearns-Sayre syndrome which begins in childhood is characterized by progressive external ophthalmoplegia, retinitis pigmentosa, heart block, central nervous system manifestations. It presents with bilateral ptosis and generally becomes symptomatic in the first or second decade of life.⁽¹⁶⁾
- Myotonic dystrophy is an autosomal dominant disorder characterized clinically by myotonia and progressive muscle weakness.⁽¹⁷⁾ Patients may present with bilateral ptosis, polychromatic cataracts, gonadal atrophy or premature thinning and loss of hair. Myotonic dystrophy causes generalized weakness, usually beginning in the muscles of the hands, feet, neck, or face with further progression to other muscle groups like heart. Symptoms may appear at any time from infancy to adulthood.
- Myasthenia gravis causes unilateral or bilateral fluctuating ptosis which may be seen in children.
- Pseudoptosis — Decreased orbital mass due to unilateral smaller eye, fat atrophy, blowout fracture which may produce the apparent ptosis due to decreased orbital volume.

Clinical features of congenital ptosis: Congenital ptosis often results due to failure of embryonic development of the LPS muscle. This muscle is initially formed from the superior rectus muscle during embryogenesis and reaches its normal position around the fourth month of pregnancy when first abnormality appear. Müller's muscle also develop and affected at this stage. In congenital myogenic ptosis LPS fibres are dystrophic and replaced by fibrous tissue.⁽¹⁸⁾

Congenital ptosis has different characteristics depending on the position of gaze as it is accentuated in upgaze but in downgaze it shows lid lag as the muscle does not relax normally and absence of eyelid crease in most cases.⁽¹⁹⁾(Fig. 1)



Fig. 1: Showing left eye congenital ptosis

The initial evaluation of children with congenital ptosis includes an assessment of marginal reflex distance (MRD-1), LPS excursion, height of the upper eyelid crease, Bell's phenomenon, and the presence of conditions such as Marcus Gunn syndrome and associated vertical strabismus.⁽²⁾ A significant decrease in LPS function (4 mm or less) is usually observed. Although ptosis is not considered a progressive condition, children with ptosis have a higher incidence of amblyopia (14-23%) and other developmental visual disorders such as myopia, astigmatism, anisometropia, torticollis and strabismus.⁽²⁰⁾

Ptosis is unilateral in 70% of cases and can be associated with abnormalities in one or more extraocular muscles or systemic diseases. More severe cases involve hypoplasia of the LPS or its aponeurosis, with an absent or attenuated eyelid crease.⁽²¹⁾

Measurement of LPS function, i.e. eyelid excursion, is the most important parameter while deciding the surgical technique. Accurate measurement require child's cooperation. Cycloplegic retinoscopy should be done to identify refractive errors and amblyopia. In infant's fixation, maintenance and light following should be noted. Pupillary examination and iris color differences among both eyes should be examined to rule out Horner's syndrome. Extraocular muscle movement should be evaluated as in CPEG, there may be extraocular muscle weakness together with ptosis. Evaluation of strabismus if present. Dilated fundus examination should rule out any posterior

segment pathology, such as abnormal retinal pigmentation seen in Kern Sayre's syndrome.

Various techniques for surgical correction of congenital ptosis

When there is no risk or sign of amblyopia, surgical correction can be performed at the age of 3-5 years, when eyelid structures are well developed and the fascia lata can be removed. If amblyopia is present, ptosis correction should be performed at earliest using an alloplastic material as a temporary sling until the patient is mature enough for autologous fascia lata grafting.

Levator aponeurosis resection: In the traditional approach to aponeurosis resection, a horizontal incision measuring approximately 20-22 mm is made on the skin to dissect the orbicularis muscle and the septum is opened. Aponeurosis is identified as the pink-white structure underneath the eyelid fat is then resected at the intended height and subsequently advanced up to the middle third of the tarsal plate with three U-shaped sutures positioned in the central, medial and lateral regions.⁽²⁾

In order to better identify and individualise the aponeurosis, 2% lidocaine with a vasoconstrictor (1:200000 adrenaline) can be injected in the subconjunctival space to separate the LPS aponeurosis from the conjunctiva-Müller's muscle complex easily. A small horizontal incision of the aponeurosis in the upper third of the tarsus to disconnect it from the tarsus followed by medial and lateral extension. Oblique cuts made vertically along the medial and lateral horns of the aponeurosis help to move it anteriorly. A Berke forceps can be used to manipulate the aponeurosis. Non-absorbable sutures should be used, as absorbable sutures can lead to late surgical failure.⁽²²⁾

An eyelid crease or fold can be created by suturing the upper and lower margin of the skin with the aponeurosis by three to four sutures. The major question in this kind of procedure is the amount of aponeurosis to be resected, which is highly dependent on surgeon's experience. The resection can be small (10-13 mm), medium (14-20 mm), or large (e"21 mm); LPS function and the degree of ptosis should be used as parameters. The resection table proposed by Beard⁽²⁾ can be used to determine resection size.

Two variants of technique

1. Whitnall's ligament suspension: the aponeurosis is resected up to Whitnall's ligament and the tarsus is sutured directly to the ligament. This procedure is indicated with LPS function of 4-5 mm. Dissecting the medial and lateral pillars of the ligament can compromise its supporting role as Whitnall's ligament works as a mobile "sleeve" for the LPS muscle, turning its horizontal force into a vertical force for the upper eyelid.⁽²³⁾ The eyelid eversion can be seen if the suture is placed too close to the lower border of the tarsus.

2. Supramaximal resection of the aponeurosis includes resection of the aponeurosis and LPS muscle >30mm. Its adhesion is resected medially and laterally to release the posterior part of the ligament without any damage to the superior rectus muscle.⁽²⁴⁾ This is an alternative to frontalis suspension that avoids the risk of infection and extrusion and does not require removal of the fascia lata, thus avoiding an additional scar.

Frontalis suspension

Frontalis suspension is widely used to correct congenital ptosis with poor LPS function and good frontalis muscle function (Fig. 2). It can correct blepharophimosis syndrome and neurogenic ptosis also.⁽²⁵⁾ The procedure connects the frontalis muscle to the upper eyelid. Skin incisions are placed at the tarsus and eyebrow to insert the sling material in the suborbicularis plane.⁽²⁰⁾ The material is placed anteriorly to the orbital septum plane to raise eyelid towards the eyebrow instead of along the eye surface, decreasing the interaction between the eyelid and the cornea. A "hood" can also be formed under the pre-tarsal and pre-septal skin, delaying eyelid lowering in downgaze.⁽²¹⁾

The sling material may become loose and reduce its function and effectiveness. In Asian patients eyelid inversion following frontalis suspension surgery is seen commonly.⁽²⁶⁾

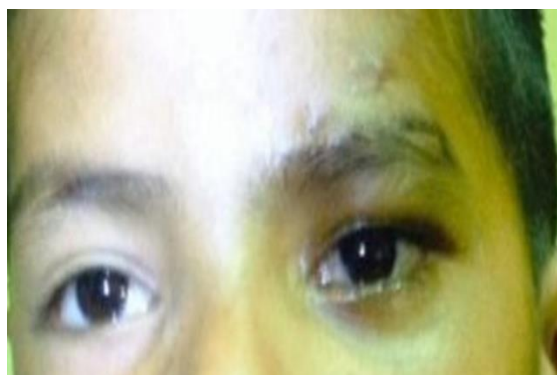


Fig. 2: Showing left eye ptosis correction after frontalis sling surgery

Endogenous versus exogenous materials

The most commonly used materials are preserved or non-preserved fascia lata or temporal fascia, the palmaris longus tendon, and the umbilical vein. Different exogenous materials have been used for suspension, such as silicone, nylon, collagen, silk, and stainless steel sutures and Mersilene, Supramid and Gore-Tex mesh.⁽²⁷⁾

Autologous fascia lata requires a second surgical incision but it minimise risk of infection, extrusion or rupture. The patient should be at least 3 years old for sufficient leg size for removing an appropriate fascia.⁽²⁶⁾ A fascia lata allograft is another option, but in

long term it shows higher (8-63.2%) rates of recurrence compared to autologous fascia lata (0.8-5%). Preserved fascia lata produce a permanent effect, but it can also be absorbed prematurely and involves the risk of transmitting infections. While several authors consider the fascia lata to be the best material for frontalis suspension,⁽²⁰⁾ others prefer silicone.⁽²⁸⁾

Silicone is a readily accessible, adjustable and elastic material, which makes it convenient for frontalis suspension in conditions mild Bell's phenomenon such as chronic progressive external ophthalmoplegia, myasthenia gravis and third cranial nerve palsy. A silicone suture on the tarsus has the benefits of lower migration and thus lower recurrence rates, and is also important for creating an eyelid crease.⁽²⁸⁾

Fascia lata was considered the best material until 2005, but recently silicone is preferred due to its superior cosmetic outcomes and lower recurrence rates.⁽²⁹⁾ Still, prospective randomised trials are required to confirm the superiority of silicone over the fascia lata⁽²⁹⁾ and to compare silicone with other materials.

Studies have shown that nylon, Mersilene and polytetrafluorethylene (PTFE or "Gore-tex") also have good acceptance, but show varying rates of extrusion, infection, and granuloma formation.⁽²⁷⁾

Autologous fascia lata is preferred material and is considered the gold-standard procedure.⁽²⁰⁾ Second-best option is preserved fascia lata.⁽³⁰⁾ Even though eyelid height, contour, and creasing seem to be satisfactory in the early postoperative period, the cosmetic outcome can change over time, mainly in terms of symmetry of eyelid height in unilateral cases and eyelid creasing in unilateral and bilateral cases, even when the functional outcome remains good.

Sling Design

Various suture designs can be used, including: single triangle, double triangle, single rhomboid (Friedenwald-Guyton procedure), double rhomboid (Iliff procedure), double trapezoid (Wright procedure), single pentagon (Fox procedure), and double pentagon (Crawford procedure).⁽³⁰⁾ Single triangle procedure is preferred for pointed eyebrows and the pentagonal or rhomboid procedures are preferred for equally elevated eyebrows. Single rhomboid procedure is preferred for small children, as it prevents postoperative eyelid folds.⁽²⁰⁾ The Crawford procedure is recommended for fascia lata grafting, and the Fox procedure is recommended for alloplastic material. Crease incisions showed better outcomes regarding eyelid contour and crease symmetry than supraciliary incisions.⁽³¹⁾

Bilateral versus unilateral surgery in severe unilateral ptosis

Some authors recommend bilateral frontalis suspension for the treatment of unilateral congenital ptosis for achieving symmetry when closing the eyes, blinking, and gazing downward.⁽²⁶⁾ However, bilateral

surgery puts both eyes at risk of postoperative complications such as lagophthalmos, exposure keratitis, upper eyelid entropion, eyelash ptosis, absent eyelid crease, excess skin, and superior oblique muscle palsy. Other authors suggest that unilateral surgery preserving the healthy side is more likely to be accepted by parents, as well as being a shorter procedure with lower risks. The presence of spontaneous eyebrow elevation on the affected side preoperatively can be predictive of successful unilateral frontalis suspension. Unilateral surgery is recommended for unilateral congenital ptosis with poor LPS function and without amblyopia. Patients with amblyopia are at risk of under-correction when subjected to unilateral surgery, therefore bilateral frontalis suspension is the procedure of choice in these cases.⁽²⁶⁾

Indications for surgical techniques

Current recommendations for congenital ptosis correction vary, but frontalis suspension is recommended for children younger than 3-4 years of age with poor LPS function. The options for children with LPS function under 3 mm include frontalis suspension, frontalis muscle flap, and Whitnall's ligament suspension. The LPS muscle can be resected or advanced in patients whose LPS function is > 5 mm. Whitnall's ligament suspension (with or without tarsectomy) can be performed in cases of relapse after frontalis suspension, and vice versa.

The greatest difficulties occur when indicating surgery for cases with LPS function between 5 and 7 mm, since resection of the LPS aponeurosis may not be sufficient. Alternatives include frontalis suspension, supramaximal LPS resection and Whitnall's ligament suspension alone or combined with superior tarsectomy.⁽²⁸⁾

Marcus Gunn syndrome

Marcus Gunn syndrome is a trigeminal-oculomotor synkinesis consisting of unilateral congenital ptosis in which the eyelid retracts when the ipsilateral pterygoid muscle is stimulated. This stimulation can occur when the patient opens his/her mouth, chews, sucks, smiles, moves the jaw laterally to the affected and/or unaffected site, and contracts the sternocleidomastoid muscle. Although bilateral cases exist rarely. The condition's prevalence is similar for both genders and sides of the face, being observed in 2-13% of patients with congenital ptosis. It is believed to be caused by a deviation of one branch of the fifth cranial nerve to the third cranial nerve, but other cranial nerves can also be involved. Some patients learn to control the position and excursion of the affected eyelid.⁽²⁵⁾

It is classified according to upper eyelid excursion during mouth stimulation, measured in millimetres: mild (<2 mm), moderate (2-5 mm), and severe (>5 mm). If it causes functional or cosmetic impairment, surgical treatment should be considered, such as LPS

excision in the affected site and weakening or excision of the contralateral LPS, followed by bilateral frontalis suspension. For patients who do not want to undergo bilateral surgery or non-amblyogenic patients, the procedure can be performed only in the affected side, although it can result in eyelid asymmetry in downgaze. Patients submitted to bilateral surgery show better upper eyelid symmetry in the primary position of gaze.⁽³²⁾

Associated changes such as treatable amblyopia (23-59% of patients), vertical strabismus (23-48%) and horizontal strabismus (34%) should be resolved beforehand.⁽³²⁾ If the jawwinking synkinesis causes only minor cosmetic problems it can be ignored, and ptosis treatment should be done by simply using the appropriate techniques for each degree of LPS function. If the synkinesis is moderate to severe and causes problems to the patient, it should be taken into account of surgical treatment. For mild synkinesis, treatment includes observation, LPS resection, or the Fasanella-Servat procedure.⁽²⁵⁾

The LPS aponeurosis has a number of connections underneath Whitnall's ligament and divides the lacrimal gland into its orbital and palpebral sections. Therefore, LPS excision is not always complete, and the connections between the LPS and the eyelid can be restored. This is why some authors recommend excising the aponeurosis and the terminal LPS associated with bilateral frontalis suspension. This would lead to a better outcome due to the symmetrical weakness of upper eyelids, symmetrical frontalis suspension, and the use of the frontalis muscle to elevate both upper eyelids.⁽³²⁾

Complications of ptosis surgery

The most common complication is under-correction which can result from improper resection, incorrect identification of anatomical structures, excessive scarring, or improper suturing.⁽²¹⁾ Over-correction results in incomplete eyelid occlusion and is a rare complication of congenital ptosis correction, but it can occur when the eyelid is sutured to Whitnall's ligament or with excessively shortened orbital septum. Although it is desirable postoperatively, symmetrical eyelid height can expose the cornea, therefore a Frost suture is recommended at the end of surgery and removed after 48 hours.

Other complications include transient or permanent diplopia in cases of residual third cranial nerve palsy, adverse reaction to anaesthetics, infection, mild keratitis and corneal abrasion secondary to improper suturing, reactions to alloplastic materials, or suture abscess. Absent or low eyelid crease can be secondary to an improper incision or a failure in crease formation, and eyelid margin distortions can be secondary to asymmetric aponeurosis advancement.⁽²¹⁾ Late complications include eyelid asymmetry and foreign-body sensation. Eyelash ptosis, entropion and excessive

skin folding can also occur. Fixation of the fascia to the lower tarsus reduces the risk of entropion, but it can lead to a euryblepharon-like opening of the margin. Creating an eyelid crease is also important. In order to prevent skin folding, excessive skin should be removed as appropriate.⁽²⁶⁾

Conclusion

The major techniques used in the treatment of congenital ptosis are resection of the LPS aponeurosis and frontalis suspension. When amblyopia is present the ptosis needs to be corrected early; otherwise, it can be corrected after three years of age.

Measurement of LPS function, i.e. eyelid excursion, is the most important parameter when choosing the surgical technique. Frontalis suspension is indicated when LPS function is poor; alternatively, supramaximal resection of the LPS muscle can also be employed. Aponeurosis resection is the preferred technique in patients whose LPS function is around 4-5 mm.

Outcomes are less effective in patients with blepharophimosis syndrome than in simple congenital ptosis. Bilateral frontalis suspension is still the most used technique, although supramaximal resection of the LPS can also be considered for such cases. For Marcus Gunn syndrome, maximal resection of the LPS in combination with frontalis suspension produces the best results. Under-correction is the most frequent complication, followed by crease deformities, lagophthalmos, keratopathy, implant extrusion and granuloma formation with alloplastic materials in frontalis suspension.

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