Congenital Ptosis- Evaluation And Management

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Background

Ptosis, an abbreviation for the term blepharoptosis, refers to vertical narrowing of the palpebral fissure secondary to drooping of the upper eyelid to a lower than normal position. Ptosis is considered congenital if present at birth or if it is diagnosed within the first year of life.Congenital ptosis, or dysmyogenic ptosis, is the most common ptosis seen in childhood. . It comprises a group of diseases in which the ptosis is due to a developmental dystrophy of the levator muscle characterized by fibrosis and deficiency of striated muscle fibers.Congenital ptosis is generally unilateral (70%), but may be bilateral, and can be isolated or associated with disease of one or more of the extra ocular muscles and/or other systemic conditions. More severe forms may involve hypoplasia of the levator palpebrae superioris muscle or tendon with a minimal or absent eyelid crease.. The condition may be associated with anisometropia, astigmatism, strabismus or amblyopia.

Pathophysiology and epidemiology

In most cases of congenital ptosis, a droopy eyelid results from a localized myogenic dysgenesis. Rather than normal muscle fibers, fibrous and adipose tissues are present in the muscle belly, diminishing the ability of the levator to contract and relax. Therefore, the condition is commonly called congenital myogenic ptosis. Congenital ptosis can also occur when the innervation to the levator is interrupted through neurologic or neuromuscular junction dysfunction.Congenital ptosis occurs equally among the different races.Congenital ptosis occurs equally between males and female. Congenital ptosis is usually present at birth but may manifest within the first year of life.

Causes, genetics and associated syndromes

Most cases of congenital ptosis are idiopathic. However, congenital ptosis may occur through autosomal dominant inheritance.Several genes that lead to isolated congenital ptosis or a syndrome involving congenital ptosis have been identified. These include PTOS1, PTOS2, and ZFH-4 which lead to autosomal-dominant forms of isolated congenital ptosis.

In most cases of congenital ptosis, the cause is idiopathic. However, congenital ptosis has been found to be associated with numerous other conditions.

Marcus-Gunn jaw-winking ptosis accounts for approximately 5% of cases of congenital ptosis. It is an example of synkinesis, or an abnormal innervation connecting two groups of normally unrelated muscles. This causes the involved eyelid to retract when the mouth opens and the mandible moves to the opposite side of the ptotic lid. This most often is a result of contraction of the external pterygoid muscle. One may also see the eyelid elevate after teeth clenching and the use of the internal pterygoid muscle.

Blepharophimosis syndrome is an autosomal dominant bilateral condition characterized by severe ptosis, epicanthus inversus, telecanthus, phimosis and ectropion.

A small percentage of patients may have an associated superior rectus weakness because the levator muscle and the superior rectus muscle arise from the same embryologic origin. Less commonly, a third cranial nerve palsy may exist. There are usually signs of aberrant regeneration present, and the pupil may be paradoxically small and nonreactive.

Horner's syndrome is characterized by mild ptosis, miosis and anhydrosis. The ipsilateral lower eyelid may be elevated. Also, because of the lack of sympathetic innervation to the iris melanocyte development, heterochromia may exist. Periorbital tumors, such as plexiform neuroma, lymphoma, leukemia, rhabdomyosarcoma, neuroma, neurofibroma or other deep orbital tumors may produce ptosis or proptosis.

Kearns

Sayre syndrome, a mitochondrial deletion disorder, is characterized by progressive external ophthalmoplegia, heart block, retinitis pigmentosa and central nervous system manifestations. This condition begins in childhood but is rarely present at birth. The conditions are most likely to become symptomatic in the first or second decade of life. Bilateral ptosis is a prominent feature of this syndrome. These patients should get an EKG.

Myasthenia gravis is a defect at the neuromuscular junction that produces unresponsiveness to released acetylcholine, resulting in ptosis. Ptosis tends to be variable. If this diagnosis is being considered, further testing can be done, including an ice test, a Tensilon test or a single fiber electromyography.

Evaluation

History

All pediatric patients presenting with either unilateral droopy eyelid or bilateral droopy eyelids need a thorough examination that includes a medical history, a family history, a history of drug or allergic reactions, and a review of systems.Family photographs can help determine onset or variability of the ptosis. Providing photographs also gives the surgeon a chance to examine the other family members. A patient with a strong family history of congenital ptosis may not need an extensive workup. In severe cases of congenital ptosis in which surgery is needed, historical emphasis should be placed on any anticoagulant use or bleeding disorder to avoid potential complications during surgery. The surgeon should also inquire about a family history of malignant hyperthermia and cardiac disorders. Patients with ptosis and Kearns-Sayre syndrome or chronic progressive external ophthalmoplegia may also have a cardiac conduction disorder. A history of fluctuating ptosis with strabismus may indicate myasthenia gravis.A careful medical history regarding cancer should be obtained. Metastatic or primary orbital tumors can result in malpositioning of the eyelid. A history of trauma with orbital wall fractures can result in pseudoptosis with enophthalmos. Additionally, third cranial nerve palsy from trauma may result in ptosis. A history of drug or allergic reactions may be helpful. Allergic reactions can result in eyelid edema and droopy eyelid.A history of difference in the size of the pupil may be helpful in diagnosing Horner syndrome. Patients with Horner syndrome have ptosis and miosis on the same side. Cervical or apical thoracic tumors can cause damage to the sympathetic chain and result in this condition. Neuroblastoma, which is one of the most common childhood cancers, should be ruled out. A history of dry eyes, intermittent epiphora, or chronic conjunctivitis can indicate a dry eye disorder or corneal surface disease.

Examination

Visual acuity, refractive error, and cycloplegic refraction should be recorded. In infants, the surgeon should make sure that the baby can fixate and follow objects with each eye individually. The patient should be evaluated for strabismus (misalignment) and undergo a dilated fundus examination.

Serial external photographs of the eyes and the face may be included in the patient's record for documentation.

Tear function should be evaluated if any doubt exists about the adequacy of tear production. This evaluation would include a slit-lamp examination with fluorescein stain to examine the cornea, tear meniscus, and tear break-up time. The Schirmer test can also be performed for dry eye syndrome; to do so, a filter paper is applied at the junction of the middle and lateral one third of the lower eyelid.

Corneal sensitivity should be tested if possible. This may be a difficult test in young pediatric patients.

An exophthalmometer can be used to assess relative proptosis or enophthalmos of each eye. In pseudoptosis, a proptosis of the contralateral eye gives the false impression that the normal upper eyelid is droopy.

The pupillary size and the iris color differences between the eyes should be examined for Horner syndrome.

The lid height (palpebral fissure distance) should be observed and measured in millimeters with each eye fixating on a distant target. The distance is the measurement of the greatest width of the palpebral fissure with the patient's eyes in straight gaze.In bilateral ptosis, the amount of ptosis is determined by the marginal reflex distance. This is the distance from the corneal light reflex to the upper lid margin. A normal marginal reflex distance is 4 to 4.5 mm. The lid position in downgaze should be noted. In congenital ptosis, the ptoticlid appears higher in downgaze. Depending on the the palpebral fissure distance, ptosis classified as- mild-2mm moderate - 3mm severe- 4mm

After the palpebral fissure distance is measured, the levator function should be evaluated. The patient looks downward as a ruler is positioned with a mark adjacent to the upper lid margin. With the examiner's hand eliminating any brow action by the patient, the patient looks upward as far as possible without a change in head position. Lid elevation is measured directly from the ruler and is recorded in millimeters of levator function. Levator function is graded as normal- 15 mm or more good- 12-14 mm fair - 5-11 mm poor- 4mm or less

The patient should be examined for Bells phenomenon. The patient closes both eyes tightly as the examiner holds the upper and lower lids apart. If the globe elevates during the forced lid closure, a normal Bell phenomenon is present. This evaluation can help the surgeon to determine the risk of exposure keratopathy following the eyelid surgery.

Careful external examination along with palpation of the eyelids and the orbital rim should be performed. A lid mass can cause extra weight in the lid, resulting in ptosis. Plexiform neuromas, lymphoma, or leukemia can result in an eyelid mass. Rhabdomyosarcoma may present with a mass that is palpable through the lid.

Management-

Traditionally, surgery is delayed until the child is 4 to 5 years old. This allows for more patient cooperation and therefore more accurate preoperative measurements. At this age, the eyelid tissues are larger as well. However, earlier intervention may be required if the pupillary axis is completely covered, putting the patient at risk of deprivation amblyopia or if the patient acquires a significant chin-up position.Congenital ptosis can be corrected by three operative procedures: levator resection by the skin approach, levator resection by the conjunctival approach, which includes the conjunctival Muller's muscle resection and the Fasanella-Servat procedure, and eyebrow suspension of the eyelids (frontalis sling).

Levator resection

Resection and advancement of the levator aponeurosis is a technique often used in correction of ptosis in patients with greater than 5 mm of levator function. This technique is performed via the exposure of the levator aponeurosis through an anterior approach, traditionally using an incision running the entire length of the upper eyelid crease, then advancing the levator aponeurosis by folding or excising the muscle, and reattaching the aponeurosis to the anterior surface of the tarsus . This method results in an elevation in the contour of the upper lid by effectively shortening the levator muscle itself. This approach has the advantages of preserving normal anatomical planes and structures of the eyelid as well as preservation of all elevating structures, including Mueller's muscle and Whitnall's ligament. This procedure may also be carried out using a trans-conjunctival approach or a small skin

incision (8-13 mm) which requires minimal local anesthetic agent, causing less distortion of the tissue arguably leading to better cosmetic results and faster recovery.

Whitnall sling

The Whitnall sling procedure is used to correct severe ptosis with levator function of 3-5 mm. The Whitnall's ligament acts as a support structure for the upper eyelid and superior orbit by functioning as a superior suspensory ligament for the eyelid and lacrimal gland and a pulley off of which the levator muscle gains support and direction.TheWhitnall sling procedure takes advantage of this second support structure of the eyelid. This procedure involves resecting the levator aponeurosis up to the point of Whitnall's ligament (maximal levator resection), and then suturing both Whitnall's ligament and the underlying levator muscle to the superior portion of the tarsal plate. If the suturing of Whitnall's ligament to the tarsus does not provide satisfactory lid elevation, a superior tarsectomy may be performed in addition to the sling procedure. This approach offers many of the same benefits as aponeurosis advancement: preservation of the levator muscle, Müller's muscle, and Whitnall's ligament without altering the structures that produce the three-layer tear film.

Frontalis muscle flap

The frontalis muscle flap procedure is recommended for use in cases of severe ptosis with levator function less than 4 mm. Historically, this procedure evolved from the frontalis sling procedure. The flap procedure involves elevating the innervated frontalis muscle flap, passing it over a pulley created near the insertion of the orbital septum at the superior orbital rim, which redirects the pull of the frontalis to elevate the lid, and then attaching the frontalis muscle to the tarsal plate. The advantages of this technique include eliminating the need for alloplastic or autologous tissue to connect the caudal portion of the frontalis muscle to the eyelid as well as improving the direction of the pull.

Mullerectomy

Müller's muscle-conjunctival resection surgery is another technique that may be used to advance the levator aponeurosis of the upper eyelid to correct ptosis. The success of this procedure is dependent upon the combined function of the Muller's muscle and levator muscle hence use recommended in patients with fairly good muscle function. The Muller's muscle is an involuntary, sympathetically innervated muscle that originates below the levator aponeurosis just distal to the Whitnall's ligament. It attaches to the superior tarsal border by a small tendon and is responsible for an estimated 2-3 mm of eyelid elevation. This particular procedure is recommended for patients who respond well to the phenylephrine test, thereby shortening a responsive Muller's muscle.In this procedure, the conjunctiva and Muller's muscle are separated from the levatoraponeurosis, the anterior aponeurosis is then bluntly dissected and separated from the orbicularis muscle. The aponeurosis and Muller's muscle are then sutured to the anterior aspect of the tarsal plate.

In summary, congenital ptosis presents at birth or within one year of age and can be mild, moderate or severe. It can result in anisometropia, astigmatism, strabismus, abnormal head posture and amblyopia. A thorough history and detailed evaluation is needed and surgery is the mainstay of management.

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