

Blepharophimosis

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INTRODUCTION

Blepharophimosis, first reported by von Ammon¹ in 1841, is defined as a palpebral aperture that is small both horizontally and vertically. Although it is associated with a wide variety of conditions, the term "**Blepharophimosis**" is frequently used synonymously with the **blepharophimosis** syndrome (also called the **blepharophimosis**, ptosis, epicanthus inversus syndrome or BPES).²

CLINICAL FEATURES

The syndrome was first described as a triad of **blepharophimosis**, ptosis and epicanthus inversus by Komoto³ in 1921. Later the same year, Dimitry⁴ reported a family with **blepharophimosis** with the complete triad and noted the dominant inheritance in an extensive documentation of the family (Fig. 1). He coined the term "**Blepharophimosis**,"



Fig. 1: Father and both children are affected by the **blepharophimosis** syndrome. Youngest child has had a brow suspension procedure 2 weeks previously to prevent amblyopia.

from the Greek *blepharon* (eyelid) and *phimosis* (stricture). Kohn and Romano⁵ noted the presence of telecanthus and added it to the syndrome. Many other associated features have since been described including hypoplasia of the tarsal plates, lengthening of the lateral canthal tendon, epiblepharon, lower lid retraction and ectropion, flat supraorbital rim, punctal and canalicular anomalies, female infertility and dysmorphic facial features. It is now recognized that the **blepharophimosis** syndrome encompasses a complex array of signs with no characteristic clinical appearance. Mild expressions of this syndrome are the most common; Callahan⁶ and Mustardé⁷ noted the occurrence of large epicanthal folds even in the mildest cases. Gene localization studies have recently suggested that the location of the **blepharophimosis** gene is at 3q2.⁸

The reduced size of the palpebral apertures is caused by a combination of telecanthus, upper lid ptosis and a variable degree of epicanthus inversus. Mustardé⁹ coined the term "Telecanthus" to describe an abnormally large intercanthal distance with a normal interpupillary distance. The degree of **blepharophimosis** is variable. The normal adult horizontal fissure may be reduced from the normal 25–30 mm to as little as 20 mm.¹⁰ The medial canthus is displaced in most cases due to an unusually long medial canthal tendon.

INDICATIONS AND CONTRAINDICATIONS

Many of the abnormalities seen in the **blepharophimosis** syndrome require correction. Early surgery is recommended to minimize the psychological trauma of school, although the final results may be better in older children and adults.¹¹ In the presence of severe ptosis with occlusion of the pupillary axis, early ptosis surgery is indicated.

Historically, the defects that have proven to be the most difficult to treat have been the combination of epicanthus inversus and telecanthus. Unlike other forms of epicanthus, the inversus type rarely improves spontaneously with nasal development. Canthal surgery is necessary for cosmetic improvement as well as for lengthening the palpebral fissure to allow correction of the ptosis. In most cases, the nasal bones are normal in width and only soft tissue correction of the telecanthus is required.

Correction of the epicanthal fold and telecanthus lengthens the palpebral fissure, which may have a bowstring effect on the upper lid, worsening the ptosis. We have noticed that release of vertical tethering of the epicanthal fold sometimes compensates for the narrowing effect and may reduce the ptosis. The unpredictability of the effect of correcting the epicanthus on the ptosis demands that the ptosis be repaired secondarily. When the visual axis is not occluded by the ptotic eyelid, we prefer to perform the medial canthoplasty at 3–4 years of age, followed by ptosis surgery 3 or more months later to allow soft-tissue relaxation and correction prior to the child commencing school.

Surgical Techniques

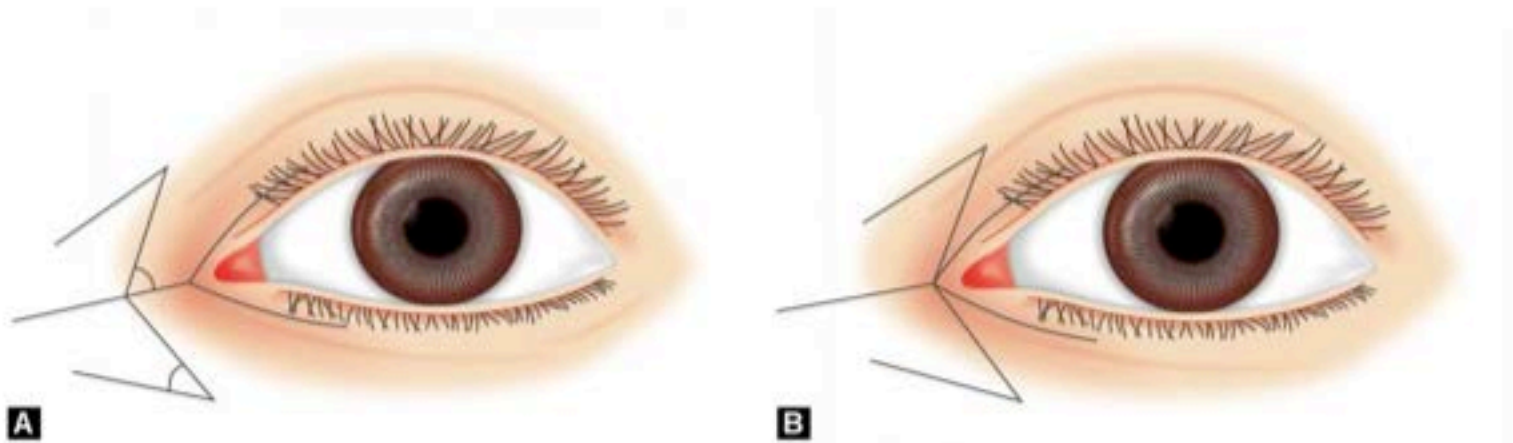
Medial canthal surgery is performed to correct epicanthus and telecanthus. Many techniques have been described. The aim of surgery is to achieve good cosmetic and functional results. A brief review of the major developments in the surgical correction of the medial canthal deformity helps in the understanding of currently popular techniques.

Rogman¹² was the first to realize that the epicanthal fold arises secondary to shortage rather than surplus of vertical skin. Verwey¹³ appreciated the necessity for rearrangement of tissues rather than resection when he designed a Y- to

V-tissue rearrangement, which was an important step in the development of the modern repair as it allows reduction of vertical tension. The Y-V procedure uses the plastic surgery concept of advancing one point on the skin toward another, at the same time allowing the tissue on either side of the Y to be relaxed. This procedure remains a major part of all the modern techniques for correction of medial canthal defects.

Hughes¹⁴ improved upon Verwey's Y-V procedure by extensively undermining and removing the fibrofatty tissues surrounding the medial canthal tendon to form a depression in the medial canthal region. Hughes' stem of the "Y" extended almost to the dorsum of the nose, resulting in marked scarring. Callahan⁶ later modified this procedure, but undercorrection and unsightly scars were common. This procedure has the disadvantage of leaving a strongly concave scar on the nasal skin, with a tendency to form a secondary fold. Mustardé¹⁵ and Anderson¹⁶ (Figs. 2A and B) have both overcome these problems by avoiding extension of the stem of the "Y" onto the nasal skin and by keeping the "Z" plasty scars as close to the medial canthal angle as possible.

Hughes recommendation to shorten the medial canthal tendon was a major advance.¹⁴ He shortened the tendon by using a catgut suture, which was passed through the medial canthus and the periosteum and tied over a rubber peg. Johnson^{10,11} observed that the use of absorbable sutures was inadequate because the considerable tension would prevent adequate healing by the time the suture absorbed. He recommended the use of 5-0 steel on a half-curved cutting needle to shorten the medial canthal tendon. He also stressed the importance of not having any dead space under the skin and of applying firm pressure to allow adequate contouring of the transposed flaps.



Figs. 2A and B: (A) The Mustardé "jumping man" four-flap technique; (B) The Anderson five-flap technique combining double Z-plasties with Y-V flap.

Lessons learned from the evolution of the various techniques have allowed modern surgeons to correct the basic defects underlying the medial canthal deformity, namely a shortage of skin in the vertical meridian, excess deep medial canthal fibrofatty tissues and a lengthened medial canthal tendon. Based on the principles, we designed the five-flap technique for the correction of epicanthus inversus and telecanthus in **blepharophimosis**.¹⁶

The Five-Flap Technique

The five-flap technique is performed under general anesthesia in children. A mark is made on the epicanthal fold at the site of the present canthus. A further mark is made at the site of the desired final position of the canthus. This point is one-half the distance from the center of the pupil to the center of the nasal bridge. A straight line is drawn between these two points.

Paramarginal lines are marked along the upper and lower eyelids and connected to the initial point on the epicanthal fold, thereby forming a "Y" configuration. A curved line is drawn on the epicanthal fold through the angle of the Y and is equal in length on each side. This line forms the central member of two "Z" configurations. The final limbs of the Z are then constructed, roughly parallel to the paramarginal marks. The angle on the final arm may vary from 45° to 60° depending on the amount of effect necessary. This configuration creates a double Z-plasty formed on the stem of the Y. We call this procedure the "five-flap technique" because of the four flaps involved

in the Z-plasty and the one additional flap of the Y-V technique.

Lacrimal probes are placed in the canaliculi to prevent canalicular and sac injury. A No. 15 Bard-Parker scalpel is used to make the incisions on the markings (Fig. 3). The flaps are gently retracted with small skin hooks and undermined with scissors (Fig. 4). Removal of the excess muscle, adipose and fibrous tissues between the skin, and elongated medial canthal tendon helps to reduce the epicanthal folds and facilitates posteromedial placement of the medial canthus. We believe that the hypertrophic tissues underlying the skin are at least partially responsible for the epicanthal folds. Extensive resection of the excess tissue in the medial canthal region provides a better correction of the telecanthus and minimizes the need for transnasal wiring.

The medial canthal tendon is usually elongated in these patients. The tendon may be plicated, resected and advanced, or a transnasal wire may be placed. A 4-0 polyglactin 910 suture on a strong half-circle needle is placed at the angle of the medial canthus to firmly engage the anterior fibers of the most lateral portion of the medial canthal tendon (Fig. 5). The same suture is then passed through the insertion of the medial canthal tendon or periosteum on the frontal process of the maxilla as far posterior and medial as possible (Fig. 6). A further suture may be used for reinforcement. When these sutures are tied, the canthus is moved to a position that is more medial, superior and posterior.

In severe cases in which the bone prevents the canthus from being adequately moved medially, a standard transnasal wiring with bone removal may be required. Some



Fig. 3: Five flaps following incision and undermining of orbicularis muscle.



Fig. 4: Flaps are retracted gently with skin hooks to expose the medial canthal structures.

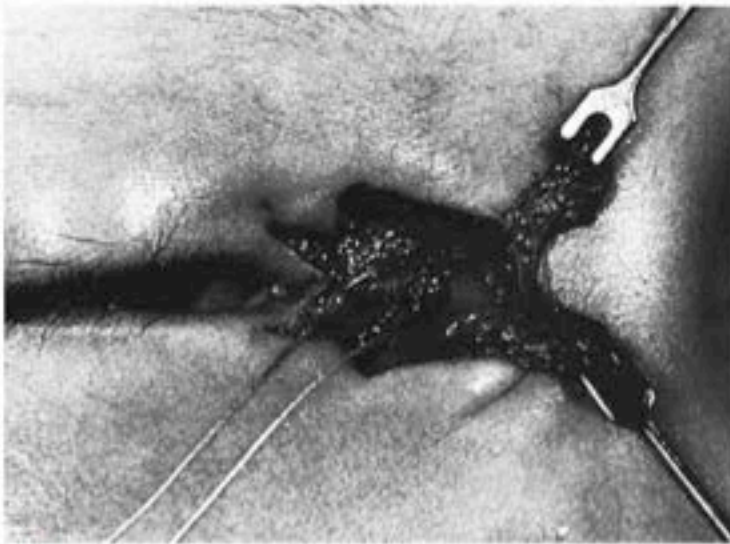


Fig. 5: Polyglactin 910 suture (4-0) is passed through the most lateral part of the medial canthal tendon.

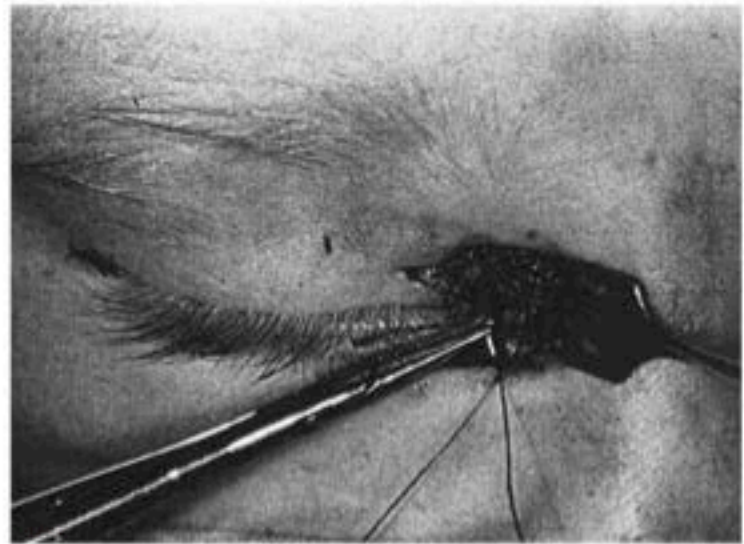


Fig. 6: Suture has been placed in lateral part of medial canthal tendon and periosteum as far posterior and medial as possible.



Fig. 7: The medial canthal suture has been tightened and the flaps have been transposed on the right. The left side demonstrates the excised deep muscle and adipose and fibrous tissue.



Fig. 8: Transposed flaps are sutured.

surgeons also believe that cutting the tendon and shortening it will result in a firmer anchoring of the tendon.

Once the medial canthus is in the desired position, the flaps are transposed (Fig. 7). The Y-V flap is closed by first placing a strong 4-0 polyglactin 910 suture in the apex of the V with a bite to the bony insertion of the medial canthal tendon to hold the skin as medial and posterior as possible. The apices of the transposed Z-plasty flaps are sutured first, followed by the flaps, using 6-0 or 7-0 chromic catgut (Fig. 8). Antibiotic steroid eye ointment is placed over the incisions for 1 week.

The five-flap technique uses a logical geometric flap design with simple construction of the flaps. Meticulous measuring, angle plotting and trimming of flaps are avoided. Maximum advantage is taken of the Y-V flap in combination with double Z-plasty. The five-flap technique is easier to understand than the Mustardé procedure. We believe that removal of excess muscle and fibrofatty tissues overlying the medial canthal tendon may be as important as the type of medial canthoplasty performed in **blepharophimosis**. The removal of these tissues allows maximal medial and posterior placement of medial canthal tissue, achieving

a more natural and cosmetically pleasing concavity in the medial canthus. In our patients, the average increase in fissure length at 1 year after surgery was 6.1 mm. The effect on the degree of ptosis was not predictable, some patients worsened, others improved and some remained the same.

TECHNICAL ALTERNATIVES AND PITFALLS

Mustardé has made a significant contribution to the logical analysis of the underlying defects in the blepharophimosis syndrome. His four-flap technique described in 1959 was a great advance over earlier procedures and is still probably the most commonly used procedure for the correction of the medial canthal deformity.¹⁵

Four-flap Technique

The interpupillary distance is measured, and the intended positions of the new canthi are marked at approximately one-half the interpupillary distance. The fold is then obliterated by pulling the skin toward the nose; a second point is marked at the canthus and the two points are connected. At the halfway point along this line, two lines are drawn at 60° from the horizontal, each equal to the original line (< 2 mm). From these lines, backcuts of the same length are drawn at 45°. Paramarginal extensions are then drawn of the same length [(original line < 2 mm) Fig. 2A]. The incisions are made through the skin and orbicularis, and the flaps are retracted. Deep fat and muscle are removed, and the canthal tendon is identified. The periosteum is exposed at the proposed new site, and the canthal tendon is cut and sutured to the new site in mattress fashion using a white 4-0 silk

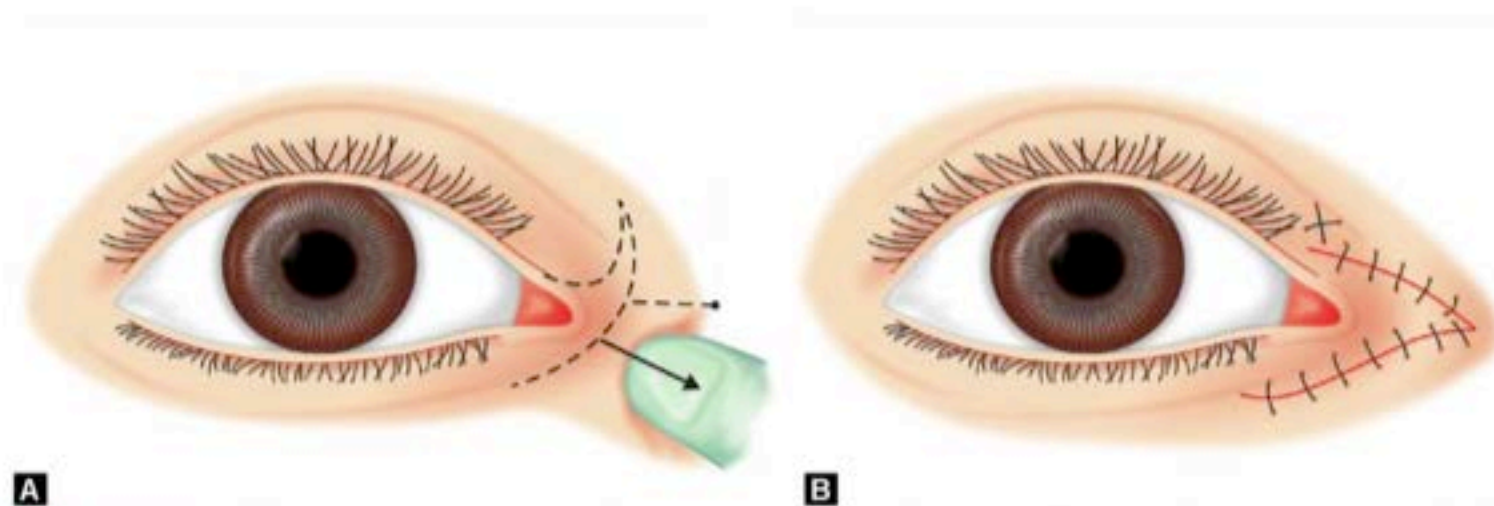
suture. The flaps are transposed and a 4-0 chromic suture is inserted. Excess skin at the angles and tips is trimmed to obtain a tidy inset. The flaps are sutured with interrupted 6-0 chromic catgut.

This technique required meticulous measurements and mapping of the lines and angles. It is difficult to understand the basis of Mustardé's flap design and one often needs to refer to his description prior to marking out flaps at surgery. The quadrilateral flaps created by this design are difficult to transpose and frequently have to be trimmed.

The Del Campo Technique

In 1984, Del Campo¹⁷ described a transposition flap technique to treat epicanthal folds and recommended it because of its effectiveness and simplicity. The principal is one of an asymmetric Y-V plasty. It is a quicker procedure with fewer flaps and may be ideal for the occasional blepharophimosis surgeon. However, it leaves a more noticeable scar extending medially to the medial canthal angle.

In the correction of epicanthus inversus, the epicanthal fold is marked first. The center of this is connected to the position of the intended new medial canthus. A further curvilinear line is drawn from the superior end of the epicanthal fold toward the upper lid border, ending 2 mm above the superior punctum (Figs. 9A and B). Telecanthus is corrected by plicating the tendon. A further suture is passed in the midportion of the internal aspect of the flap and sutured to the periosteum to secure its new position and to prevent a bowstring effect. The transposition of the flap is completed (Figs. 9A and B).



Figs. 9A and B: The Del Campo technique: (A) Curvilinear line drawn from superior end of fold stopping 2 mm short of upper punctum; (B) Transposed flaps.

Ptosis and Visual Function

The incidence of visual and motility problems regarding the blepharophimosis syndrome was largely ignored in the literature until Beaconsfield and Collin¹⁸ presented a review of 101 cases. Amblyopia (defined for the purpose of the study as vision of 20/40 or less) was found in a remarkable 56.4% of cases and strabismus was found in 26.7%.¹⁹ There was a high incidence of amblyopia in severe bilateral and unilateral ptosis. Based on their results, the authors advocated earlier surgery in patients with severe bilateral ptosis and in asymmetric ptosis and close orthoptic follow-up with early ptosis surgery where necessary in the others.

The ptosis may be severe with little or no demonstrable levator function and an absent upper-lid skin crease. When sufficient levator function is present, an external or posterior levator resection is performed in the standard way. Gore-Tex or supramid frontalis suspension procedures may be indicated at a very early age in cases of severe ptosis with poor levator function. A more definitive procedure is carried out using autogenous fascia lata when the child is 4 years of age or older when adequate fascia lata may be harvested. Banked irradiated fascia lata is a viable option to autogenous fascia lata, although the former is associated with a worse long-term success rate.

Lateral Canthal Defects

Moderate lateral ectropion can be treated with a lateral tarsal strip procedure,²⁰ together with release of the lower lid retractors and the inferior orbital septum, which is usually shortened in these cases. In more severe ectropion, there may be no alternative to the use of full-thickness skin grafts as first advocated by Johnson.^{10,11} Several procedures have been described for the opening of the palpebral aperture at the lateral canthus. Many of these give little lengthening and further loosen the support of the lower lid. A red lateral eyelid margin because of exposed conjunctiva is a further complication.

Mustardé recommends repeated medial canthoplasty as a secondary procedure to obtain further correction of blepharophimosis and suggests that the lateral canthi should not be violated.

OUTCOMES

Many deformities requiring surgical correction may be seen in the blepharophimosis syndrome.²¹ Careful assessment of

the visual development is required especially in the presence of severe ptosis.²² Early ptosis surgery with the use of temporary sling materials may be necessary. If visual development is not at risk, correction of the epicanthus inversus and telecanthus are undertaken at 3 years of age using the Anderson five-flap or Del Campo technique followed by ptosis correction 3–6 months later. It is important to remove excess muscle and deep tissue in the medial canthal region in addition to shortening or tightening the medial canthal tendon.

Note: Written in 1995 by Blupendra CK Patel and Richard Anderson. Re-edited by Wade Brock, MD in 2012.

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