Blepharophimosis syndrome: Case Report

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Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) is an uncommondysmorphic syndrome ,which primarily affect the soft tissues of the mid-face.

It comprises of

- narrowing of the eye opening (blepharophimosis)
- droopy eyelids (ptosis)
- an upward fold of the skin of the lower eyelid near the inner corner of the eye (epicanthus inversus)

Also, there is an increased distance between the inner corners of the eyes (telecanthus). Because of these eyelid abnormalities, the patients generally maintain a chin up posture with taut brows in order to see clearly, because they are unable to open eyelids completely.

People with BPES may also have distinctive facial features including a broad nasal bridge, low-set ears, or a shortened distance between the nose and upper lip (a short philtrum), and are at an increased risk of developing visual symptoms related to myopia or hyperopia.

This condition is categorised into two types, Type I being associated with primary ovarian failure along with the facial abnormalities, whereas the Type II having no such systemic associations.

BPES types I and II were each mapped on the long arm of chromosome 3 to the FOXL2 gene. The FOXL2 gene provides instructions for making a protein that is active in the eyelids and ovaries.The mutations probably impair regulation of normal development of muscles in the eyelids, resulting in malformed eyelids that cannot open fully. Mutations that lead to a complete loss of FOXL2 protein function often cause BPES type I and partial loss lead to BPES type 2

This condition is typically inherited in an autosomal dominant pattern, with only one copy of the altered gene sufficient to cause the disorder.

We managed two patients with blepharophimosis syndrome aged 10 and 13 years.

Pre-operative clinical assessment:

Both the patients presented at our institute with complaints of drooping of upper eyelids since birth, as stated by their parents. On examination, we found them to be having bilateral ptosis, epicanthus inversus and telecanthus. The BCVA in case 1 was 6/9 and the case 2 was at 6/12 in both eyes. Tha anterior and posterior segment did not show any abnormalities on slit lamp examination.

Surgical procedure:

Both the cases were operated by the same surgeon and a similar surgical approach after proper informed consent from the parents. General anaesthesia was given in both the cases. The correction was done in two stages, where telecanthus and epicanthus inversus was repaired in the first stage and ptosis repair done in

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another stage 6 months later.

Epicanthal fold and telecanthus was repaired using the Mustarde' double Z plasty approach. The site of intended medial canthus was marked using a sterile marking pen as point A. The skin was pulled taut towards midline to obliterate the epicanthal fold and the existing medial canthus was marked as point B. The two marks were joined and their midline was bisected by two short lines at 60^0 from which two more lines directed at 45^0 were drawn towards the intended medial canthus. This Z shaped marking pattern was incised, undermined and retracted with stay sutures. The subcutaneous tissue including the Orbicularis muscle was cut to expose the medial canthal tendon. Thereafter, the periosteum over the nasal bone was exposed and the canthal tendon was tied over the posterior lacrimal crest with the help of 4-0 silk double armed sutures. No transnasal wiring was done in both the cases. The skin was then closed with 5-0 silk sutures, which were removed on day 10 postoperatively.

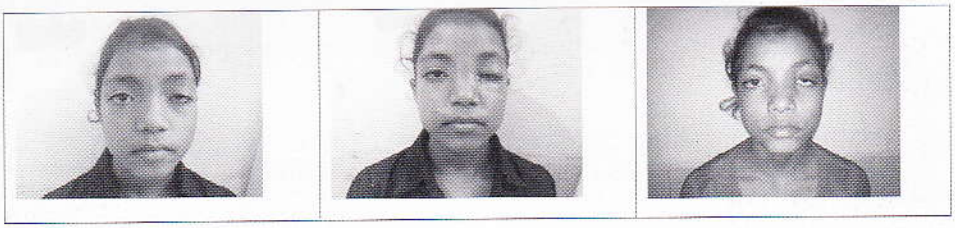
The ptosis repair is scheduled to be repaired in a second stage 6 months later by Frontalis sling surgery. Both the cases were followed postoperatively and the amount of telecanthus and repaired was measured at day 1, day 10 and advised follow up at 3 months thereafter.

Results:

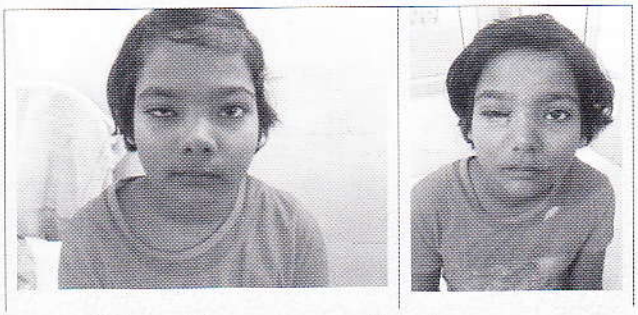
Both the cases showed good correction of the telecanthus, as measured postoperatively.

Discussion:

The treatment of blepharophimosis syndrome requires a combination of proper patient education about the disease entity along with support from pediatric endocrinologists and genetic counselors in the medical management of the disease. The surgical management require a deft oculoplastic approach as this syndrome is associated with varied dysmorphic features. Timely surgical intervention is advised in order to preserve the visual acquity, else delay can lead to the development of amblyopia in severe cases. Many surgeons these days prefer one stage repair of both telecanthus, epicanthus and ptosis. This has also shown equivalent results and thus the choice of repair rests with the clinical judgement and expertise of the operating surgeon.



CASE 1 : At presentation, on day 1 and day 10 postoperatively.



CASE 2: at presentation and Day 10 postoperatively