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BPES Type 2- Revisited

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Abstract
A 5-year-old male child was brought by his parents to the ophthalmology department for routine ocular examination. Examination of both the child as well as his father revealed Blepharophimosis, ptosis, and epicanthus inversus syndrome which is a very rare entity. It as a rare autosomal dominant disorder with a very low worldwide prevalence.

Keywords: BPES, rare, ocular

Introduction
BPES is a rare autosomal dominant disorder. It is caused by mutation in the FOXL2 gene. 50% patients with BPES have an affected parent and 50% of cases are sporadic. [1] This complex eyelid malformation syndrome has four major features, all present at birth: blepharophimosis (narrowing of horizontal aperture of the eyelids), ptosis (drooping of upper eyelid), epicanthus inversus (a skin fold arising from the lower eyelid and running inwards and upwards), and telecanthus (lateral displacement of the inner canthi with normal interpupillary distance).

Case
A 5-year-old male child reported to the department of ophthalmology for routine ocular examination. The parents of the child stated that the child had no ocular complants. He was under treatment from some other department for cervical lymphadenopathy, and was advised to visit an ophthalmologist because he had a different 'eye appearence' as compared to others. The parents said that the child had similar facial features since birth. There was no other significant history. His cycloplegic refraction was not performed as the attendants did not consent for it. His pupillary size, pupillary reactions, ocular movements, intraocular pressure and fundus was within normal limits. Torch examination revealed (figure 1) bilateral severe upper blepharoptosis, absent eyelid crease, bilateral lower eyelids showing epicanthus inversus. There was narrowing of horizontal palpebral apertures in both the eye along with telecanthus. He was diagnosed as a case of Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES)- Type 2. The father (figure 2) also has similar eye findings of blepharophimosis, ptosis, epicanthus inversus and telecanthus. The child was already ordered laboratory investigations in the form of hemogram, renal function test, hepatic function test and electrolytes from some other department. Need for visual aquity testing, genetic testing amd eye surgery was discussed with the parents.

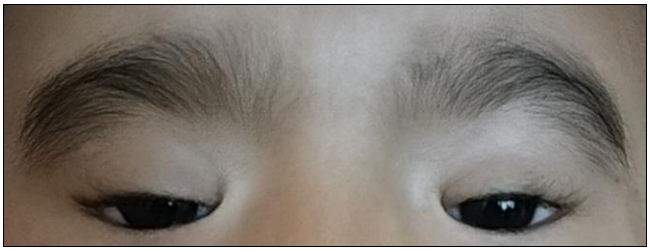

Fig 1

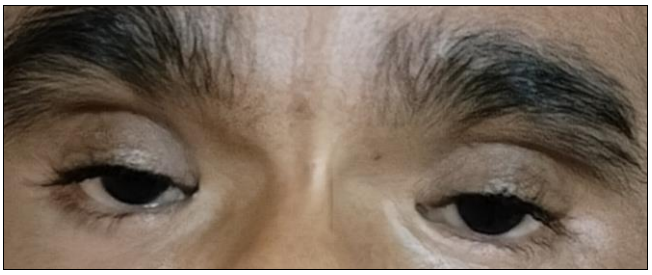

Fig 2

Discussion
BPES type I includes the four major features and primary ovarian insufficiency while BPES type II has only the four major features. [2] The global prevalence of BPES is around 1 in 50,000. Other eye associations with this syndrome are raised arched eyebrows, eyelid margin and lacrimal drainage apparatus abnormalities, squint, microphthalmos, etc. Other abnormalities include a broad flat nasal bridge, high arched palate, protruding or cup shaped ears, female infertility, and cardiac defects. [3] Hypoplastic uterus and small ovaries may be found in some females with BPES-Type 1. Serum estradiol and progesterone are decreased while luteinizing hormone levels are increased. [4] For diagnostic purpose, genetic analysis is not needed. Treatment of BPES is eyelid surgery which involves a medial canthoplasty for correction of the blepharophimosis, epicanthus inversus, and telecanthus at age three to five years, typically followed a year later by ptosis correction. In females, premature ovarian failure is treated with hormone replacement therapy. [5]

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