International Journal of Research in Medical Sciences
Abhilash K. Int J Res Med Sci. 2013 Aug;1(3):283-284
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: 10.5455/2320-6012.ijrms20130825

Case Report

An unusual case of ptosis

Abhilash K*

Associate Professor of Medicine, Sree Gokulam Medical College, Thiruvananthapuram-695011, Kerala, India

Received: 8 May 2013
Accepted: 15 May 2013

***Correspondence:**
Dr. Abhilash K,
E-mail: drabhilashkannan@yahoo.com

ABSTRACT

Blepharophimosis is a condition where the patient has bilateral ptosis with reduced lid size, vertically and horizontally. The nasal bridge is flat and there is hypoplastic orbital rim. Here I am presenting a case of a father & son with blepahrophimosis which could be a part of an uncommon condition called BPES (Blepharophimosis, Ptosis, Epicanthus Inversus, Telecanthus).

Keywords: Blepharophimosis, Ptosis, BPES

INTRODUCTION

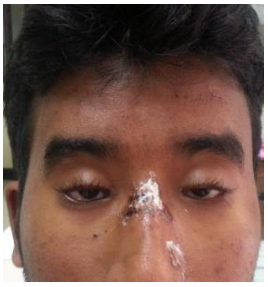
Blepharophimosis is a condition where the patient has bilateral ptosis with reduced lid size, vertically and horizontally. The nasal bridge is flat and there is hypoplastic orbital rim. Both the vertical and horizontal palpebral fissures are shortened.[1] Blepharophimosis syndrome (BPES) is a complex eyelid malformation invariably characterized by four major features: blepharophimosis, ptosis, epicanthus inversus, and telecanthus. BPES type I includes the four major features and premature ovarian failure (POF); BPES type II includes only the four major features.[2]

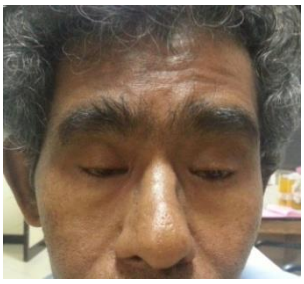
CASE REPORT

A 25 year old male with no significant medical history in the past was admitted in medical ward with short febrile illness and thrombocytopenia. He also had sustained some minor facial injuries because of a fall due to transient giddiness. No history of any loss of consciousness or seizures. No history of any bleeding manifestations.

On examination patient was febrile. A small lacerated wound was present near root of the nose due to the fall. Patient had bilateral ptosis with narrow palpebral fissures which is present since birth. Eyes cannot be opened further by separating the eyelids. His eye movements & vision was normal. All other systems were normal.

Patient's father was the bystander who also had the similar eye findings since birth. No other family members have got similar eye findings. Ophthalmology consultation was done. They diagnosed that the patient is having blepharophimosis. They also detected mild epicanthus inversus & telecanthus. So a literature review for familial cases of blepharophimosis was done which showed a rare condition called Blepharophimosis Syndrome or BPES. BPES are of two types. As type I is associated with premature ovarian failure, we assume that our patient is having BPES type II.





DISCUSSION

Blepharophimosis syndrome (BPES) is a complex eyelid malformation invariably characterized by four major features: blepharophimosis, ptosis, epicanthus inversus, and telecanthus. Epicanthus inversus is a skin fold arising from the lower eyelid and running inwards and upwards. Telecanthus is the lateral displacement of the inner canthi with normal interpupillary distance. BPES type I includes the four major features and premature ovarian failure (POF); BPES type II includes only the four major features. Other ophthalmic manifestations associated with BPES include lacrimal duct anomalies, amblyopia, strabismus, and refractive errors. Minor features include a broad nasal bridge, low-set ears, and a short philtrum. Individuals with BPES and an intragenic disease causing mutation are expected to have normal intelligence.[2] The diagnosis of BPES is primarily based on clinical findings. Occasionally individuals with BPES have cytogenetic rearrangements, such as interstitial deletions and translocations involving 3q23. Molecular genetic testing of *FOXL2*, the only gene currently known to be associated with BPES, is available clinically.

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DOI: 10.5455/2320-6012.ijrms20130825
Cite this article as: Abhilash K. An unusual case of ptosis. Int J Res Med Sci 2013;1:283-4.