A CASE OF BLEPHAROPHIMOSIS SYNDROME(BPES)

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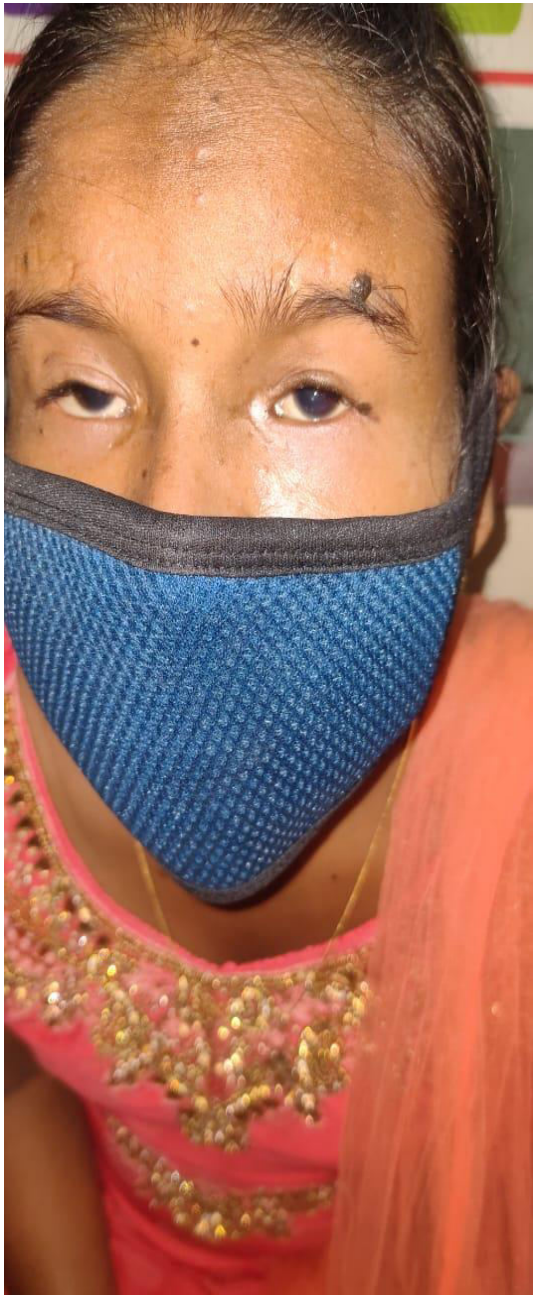
- A 17 year old female presented with drooping of upper eyelids in both eyes since early childhood.
- She has past history of corrective procedure surgery for both eyelids 5 years back at Aravind eye hospital, madhurai.
- No history of birth trauma.
- developmental milestones normally attained
- Attending college
- Menstrual h/o: regular.
- Family history: her father and younger brother have similar appearance of the eyelids in both eyes
- No h/o consanguinity.

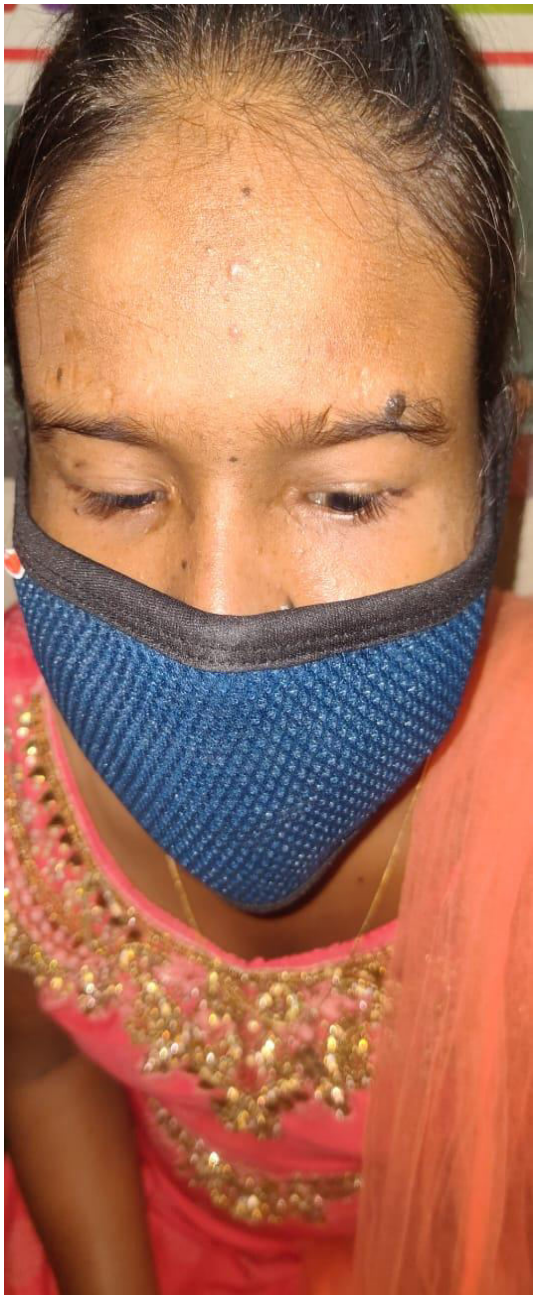
Local examination:

- Head posture: midline,
- Facial symmetry: b/l symmetrical
- Eyes are orthophoric
- EOM: Uniocular and binocular movements are normal in both eyes
- No chin elevation
- Inter medial canthal distance : 36mm
- IPD : 56mm
- Epicanthus inversus present

Parameters	RE	LE
VA	6/6	6/6
EYEBROWS	Position is slightly elevated with deep wrinkling of the forehead	Position is slightly elevated with deep wrinkling of forehead
EYELIDS	Multiple SCARS PRESENT ABOVE THE EYELID probably due to sling surgery	MULTIPLE SCARS PRESENT ABOVE THE EYELID probably due to sling surgery
EYELASHES	NORMAL	NORMAL
CONJUNCTIVA	NORMAL	NORMAL
CORNEA	CLEAR	CLEAR
AC	NORMAL	NORMAL
IRIS	NORMAL	NORMAL
PUPIL	NSRL	NSRL
LENS	clear	clear

PARAMETERS	RE	LE
PALPEBRAL FISSURE VERTICAL HEIGHT (No increase in PFH on jaw movements)	6mm	7mm
HORIZONTAL WIDTH	20mm	21mm
MRD1	3mm	4mm
MRD2	3mm	3mm
BELLS PHENOMENON	Good	Good
LPS ACTION	3mm	4mm





Discussion:

- Blepharophimosis ptosis epicanthus inversus syndrome (BPES) is a rare congenital oculofacial disorder in an autosomal dominant pattern. Diagnosis is primarily made by presence of a combination of typical oculofacial features and primary ovarian insufficiency in type I.
- Mutations of FoxL2Gene located on long arm of chromosome3 (3q23)
- FoxL2Gene appears to be involved in development of eyelid muscles as well as in growth and development of ovarian cells.
- In the above case only oculofacial feature ptosis with normal menstrual history and a diagnosis of type II BPES is made, and genetic counselling was advised

References :

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