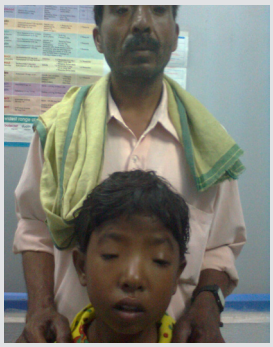
SPOT DIAGNOSIS (IMAGE GALLERY)



FAMILIAL PTOSIS

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A 10 years old girl presented to the pediatric outpatient department for the complaints of low grade fever for 1 month. On examination cervical and axillary glands were enlarged. However the most peculiar finding was the typical phenotypic appearance of the patient as well as her father {fig1}. The features were narrowing of the eye opening, inner canthal folds, lateral displacement of inner canthi and drooping upper eyelids. Father also informed that the various other family members from every generation were suffering from the same complaint.

What is the diagnosis?

Blepharophimosis, ptosis, and epicanthus inversus syndrome {also called BPES} is a rare genetic disorder that mainly affects the development of the eyelids. People with this condition have a narrowing of the eye opening {blepharophimosis}, droopy eyelids {ptosis}, and an upward fold of the skin of the lower eyelid near the inner corner of the eye {epicanthus inversus}. In addition, there is an increased distance between the inner corners of the eyes {telecanthus}. Because of these eyelid malformations, the eyelids cannot open fully, and vision may be limited. There are two types of BPES. Type I consists of the four major features of blepharophimosis, ptosis, epicanthus inversus, and telecanthus, plus premature ovarian failure. So type I is associated with menstrual problems and infertility. Type II consists of only the eyelid malformations. The prevalence of BPES is unknown. Mutations in the FOXL2 gene cause BPES types I and II. Approximately 12 percent of people with BPES do not have an identified FOXL2 gene mutation; the cause of the condition in these people is unknown. This condition is inherited in an autosomal dominant pattern.

References

1. Blepharophimosis, ptosis, and epicanthus inversus syndrome {BPES} – Medpedia. Available at website: wiki.medpedia.com, Blepharophimosis,_ptosis,_and_epicanthus_inversus_syndrome_{BPES}. Accessed on 11th January 2011
2. Blepharophimosis, ptosis, epicanthus inversus syndrome. Available at website: children.webmd.com, blepharophimosis-ptosis-epicanthus-inversus-syndrome. Accessed on 11th January 2011

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