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Research Article

BLEPHAROPHIMOSIS PTOSIS EPICANTHUS INVERSUS SYNDROME (BPES)

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A R T I C L E I N F O

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A B S T R A C T

Blepharophimosis ptosis epicanthus inversus (BPES) is a relatively rare congenital disorder, which usually presents with classical eye manifestations. In some cases, it is associated with premature ovarian failure (POF). BPES is of two types, type I and type II. Type I is associated with POF along with eyelid malformations, while Type 2 has only eyelid malformations.

INTRODUCTION

Blepharophimosis ptosis epicanthus inversus syndrome (BPES) is a rare congenital eyelid disorder that is inherited as an autosomal dominant trait, with an estimated incidence of 1 in 50,000 births.[1] Blepharophimosis was first reported by Von Ammon in 1841. Vignes first associated blepharophimosis with ptosis and epicanthus inversus in 1889. It is characterized by shortened horizontal palpebral fissure (blepharophimosis), impaired function of levator palpebrae superior is of upper eyelid (ptosis), a vertical skin fold arising from the lower eyelid that inserts medially into the upper lid (epicanthus inversus) and an increased inner canthal distance (telecanthus).[2] Zlotogra *et al*. in 1983 described two types of BPES, Type I and Type II. Type I is associated with premature ovarian failure (POF) in the affected female, in addition to the classical eye findings, while Type II has only eye features.[3]

The diagnosis of BPES is primarily made by combination of typical facio-ocular features, with clinical and biochemical features of primary ovarian insufficiency. For diagnostic purpose, genetic analysis is not needed. However, both types of BPES are caused by mutations of the forkhead transcriptional factor 2 (FOXL2) gene, that is located on the long arm of chromosome 3 (3q23).[4]

Case report

We report a boy 5 years and his sibling girl of 1year, with BPES syndrome. Both of them had small palpebral fissures (blepharophimosis), drooping eyelids (ptosis) and a skin fold arising from the lower eyelid (epicanthus inversus) and hypertelorism, in which innercanthal and inter pupillary distance were increased. Further their father too had similar presentation which signifies the autosomal dominant mode of inheritance.

The measurements of innercanthal (Table1), interpupillary distance (Table2) and the palpebral fissure length (Table3) of the two children are as follows. Father's measurements could not be done as he was unavailable. The boy had a decreased visual acuity. Girl's visual acuity could not be tested.

We suggested corrective surgery for both children and their father. Due to financial constraint they denied. The children are kept under follow up to detect the occurance of refractive errors, amblyopia and also for the detection of ovarian failure.

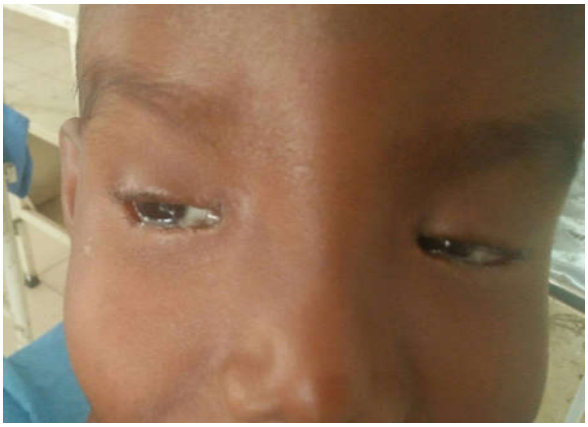

Image 1

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Image 2

Measurements

Table 1

Innercanthal distance	Mean	+2sd	Value
Boy	2.8cm	3.3cm	3.4cm
Girl	2.5cm	3cm	3.1cm

Table 2

Interpupillary distance	Mean	97thcentile	Value
Boy	5cm	5.6cm	5.8cm
Girl	4.6cm	5.25cm	5.3cm

Table 3

Palpebral fissure length	Mean	-2sd	Value
Boy	2.6cm	2.35	1.5cm
Girl	2.2cm	1.95cm	1.4cm

DISCUSSION

First described by Komoto in 1921, blepharophimosis-ptosis-epicanthus inversus syndrome (BPES) is a dominantly inherited disorder characterized by four features that are present at birth. Patients have:

- severe bilateral, symmetric ptosis,
- telecanthus (an abnormally wide intracanthal distance with normal interpupillary distance)
- epicanthus inversus (skin fold arising from the lower eyelid that covers the medial canthus), and
- blepharophimosis (profound narrowing of the palpebral fissure)

Affected individuals are at an increased risk of developing vision problems such as nearsightedness (myopia) or farsightedness (hyperopia) beginning in childhood. They may also have eyes that do not point in the same direction (strabismus) or "lazy eye" (amblyopia) affecting one or both eyes. 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).

There are two types of BPES, which are distinguished by their signs and symptoms. Both types I and II include the eyelid malformations and other facial features. Type I is also associated with an early loss of ovarian function (primary ovarian insufficiency) in women, which causes their menstrual periods to become less frequent and eventually stop before age 40. Primary ovarian insufficiency can lead to difficulty conceiving a child (subfertility) or a complete inability to conceive (infertility).

Genetics

Mutations in the FOXL2 gene cause BPES types I and II. The FOXL2 gene provides instructions for making a protein that is active in the eyelids and ovaries. The FOXL2 protein is likely involved in the development of muscles in the eyelids. Before birth and in adulthood, the protein regulates the growth and development of certain ovarian cells and the breakdown of specific molecules.

It is difficult to predict the type of BPES that will result from the many FOXL2 gene mutations. However, mutations that result in a partial loss of FOXL2 protein function generally cause BPES type II. These 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. These mutations impair the regulation of eyelid development as well as various activities in the ovaries, resulting in eyelid malformation and abnormally accelerated maturation of certain ovarian cells and the premature death of egg cells.[5]

Diagnosis

The characteristic facial features and the autosomal dominant pattern of inheretence is sufficient in making the diagnosis though it can be confirmed by genetic analysis.

Treatment

The treatment of blepharophimosis requires coordination among oculoplastic surgeons, pediatric ophthalmologists, pediatric endocrinologists and genetic counselors. The surgical repair of the eyelid is complex because of the numerous and interdependent eyelid findings. The repair of ptosis is usually addressed first with frontalis suspension. Early surgery for ptosis is advised if the ptosis is severe and amblyogenic. The canthal repair is usually addressed after ptosis, though some advocate canthal repair first. Medial canthoplasty can be accomplished by a combination of flaps and, at times, transnasal wiring. Some also suggest fixing the medial canthus to the periosteum. [6]

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