

BPES

Subjects: **Genetics & Heredity**

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Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) is a condition that mainly affects 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 abnormalities, the eyelids cannot open fully, and vision may be limited.

genetic conditions

1. Introduction

Other structures in the eyes and face may be mildly affected by BPES. 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).

2. Frequency

The prevalence of BPES is unknown.

3. Causes

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.

3.1. The gene associated with Blepharophimosis, ptosis, and epicanthus inversus syndrome

- FOXL2

4. Inheritance

This condition is typically inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In some cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

5. Other Names for This Condition

- blepharophimosis syndrome
- blepharophimosis, ptosis, and epicanthus inversus
- BPES

References

1. Allen CE, Rubin PA. Blepharophimosis-ptosis-epicanthus inversus syndrome(BPES): clinical manifestation and treatment. *Int Ophthalmol Clin*. 2008Spring;48(2):15-23. doi: 10.1097/IIO.0b013e3181694eee. Review.
2. Beysen D, De Jaegere S, Amor D, Bouchard P, Christin-Maitre S, Fellous M, Touraine P, Grix AW, Hennekam R, Meire F, Oyen N, Wilson LC, Barel D, Clayton-Smith J, de Ravel T, Decock C, Delbeke P, Ensenuer R, Ebinger F, Gillesen-Kaesbach G, Hendriks Y, Kimonis V, Laframboise R, Laissue P, Leppig K, Leroy BP, Miller DT, Mowat D, Neumann L, Plomp A, Van Regemorter N, Wiczorek D, Veitia RA, De Paepe A, De Baere E. Identification of 34 novel and 56 known FOXL2 mutations in patients with Blepharophimosis syndrome. *Hum Mutat*. 2008Nov;29(11):E205-19. doi: 10.1002/humu.20819.

3. Beysen D, De Paepe A, De Baere E. FOXL2 mutations and genomic rearrangements in BPES. *Hum Mutat.* 2009 Feb;30(2):158-69. doi: 10.1002/humu.20807. Review.
4. Beysen D, Raes J, Leroy BP, Lucassen A, Yates JR, Clayton-Smith J, Ilyina H, Brooks SS, Christin-Maitre S, Fellous M, Fryns JP, Kim JR, Lapunzina P, Lemyre E, Meire F, Messiaen LM, Oley C, Splitt M, Thomson J, Van de Peer Y, Veitia RA, DePaepe A, De Baere E. Deletions involving long-range conserved nongenic sequences upstream and downstream of FOXL2 as a novel disease-causing mechanism in blepharophimosis syndrome. *Am J Hum Genet.* 2005 Aug;77(2):205-18.
5. Choi KH, Kyung S, Oh SY. The factors influencing visual development in blepharophimosis-ptosis-epicanthus inversus syndrome. *J Pediatr Ophthalmol Strabismus.* 2006 Sep-Oct;43(5):285-8. doi: 10.3928/01913913-20060901-03.
6. D'haene B, Nevado J, Pugeat M, Pierquin G, Lowry RB, Reardon W, Delicado A, García-Miñaur S, Palomares M, Courtens W, Stefanova M, Wallace S, Watkins W, Shelling AN, Wieczorek D, Veitia RA, De Paepe A, Lapunzina P, De Baere E. FOXL2 copy number changes in the molecular pathogenesis of BPES: unique cohort of 17 deletions. *Hum Mutat.* 2010 May;31(5):E1332-47. doi: 10.1002/humu.21233.
7. Di Pietromaria A, Benayoun BA, Todeschini AL, Rivals I, Bazin C, Veitia RA. Towards a functional classification of pathogenic FOXL2 mutations using transactivation reporter systems. *Hum Mol Genet.* 2009 Sep 1;18(17):3324-33. doi: 10.1093/hmg/ddp273.
8. Taylor A, Strike PW, Tyers AG. Blepharophimosis-ptosis-epicanthus inversus syndrome: objective analysis of surgical outcome in patients from a single unit. *Clin Exp Ophthalmol.* 2007 Apr;35(3):262-9.

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