ISSN: 0975-8585

Research Journal of Pharmaceutical, Biological and Chemical Sciences

A Family of Blepharophimosis Ptosis Epicanthus Inversus Syndrome (BPES).

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Abstract

Blepharophimosis Ptosis Epicanthus Inversus Syndrome (BPES) is a rare autosomal dominant disorder primarily affecting the eyelids and mid-face structures. It is characterized by four main features: blepharophimosis (narrowing of the eye opening), ptosis (drooping of the upper eyelid), epicanthus inversus (an upward fold of skin of the lower eyelid near the inner corner of the eye), and telecanthus (increased distance between the inner corners of the eyes). BPES was first described by Dr. Komoto in 1921 and is subdivided into two main types: Type I, associated with premature ovarian failure, and Type II, which presents with only the characteristic facial features without ovarian involvement. BPES is a condition with significant ophthalmic implications, particularly if not addressed early, as it may result in amblyopia (lazy eye). Early diagnosis and appropriate management are crucial for preventing vision impairment. The condition often requires surgical correction to improve both functional and aesthetic outcomes. This case report describes a family affected by BPES, highlighting the clinical presentations and outcomes of four family members who presented to the ophthalmology outpatient department.
Keywords: blepharophimosis, ptosis, epicanthus inversus, BPES.

https://doi.org/10.33887/rjpbcs/2024.15.5.37

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ISSN: 0975-8585

INTRODUCTION

Blepharophimosis Ptosis Epicanthus Inversus Syndrome (BPES) is a rare genetic disorder characterized by the presence of blepharophimosis (narrowing of the eye opening), ptosis (drooping of the eyelid), epicanthus inversus (upward fold of the lower eyelid), and telecanthus (increased distance between the inner corners of the eyes) [1, 2]. BPES is inherited in an autosomal dominant pattern and can be classified into two types: Type I, associated with premature ovarian failure, and Type II, limited to facial features without reproductive involvement [3, 4]. This case report highlights a family of four members affected by BPES, including a father and his three children, who presented with visual impairment and drooping eyelids. The female members of the family also exhibited signs of premature ovarian failure. All family members showed typical ocular features of BPES without other systemic abnormalities. Early diagnosis and intervention, including ptosis correction surgery and genetic counselling, are critical in preventing long-term visual impairment and improving quality of life. The report emphasizes the importance of timely surgical intervention, hormonal therapy for females, and genetic counselling in managing BPES effectively [4].

CASE SERIES

This case report discusses four individuals from a single family diagnosed with BPES. The family, consisting of a father and his three children, residents of Nandurbar, presented to the ophthalmology outpatient department with complaints of bilateral eyelid drooping and visual impairment since childhood. None of the patients had a history of systemic disease, ocular trauma, ocular surgeries, or allergies to medications.

Case 1: 64-year-old Male (Father)

The first case involved a 64-year-old male, the father of the three other patients. He presented with complaints of progressive diminution of vision in both eyes and drooping of the eyelids since childhood. On clinical examination:

Right Eye: Visual acuity was finger counting (FC) at 3 meters.

Left Eye: Visual acuity was perception of light (PL) with projection of rays (PR).

Bilateral moderate ptosis was noted, along with **blepharophimosis** and **epicanthus inversus**.

The anterior segment and fundus examinations were within normal limits for both eyes.

Case 2: 40-year-old Female (First Child)

The second case involved the 40-year-old first child of the family, a female who presented with complaints of reduced vision and drooping eyelids. On examination:

Right Eye: Visual acuity was 6/60.

Left Eye: Visual acuity was 6/24.

She exhibited **bilateral moderate ptosis**, **blepharophimosis**, and **epicanthus inversus**.

Both anterior segments and fundi were within normal limits.

Additionally, she was noted to be nulligravida and had amenorrhea, indicating primary ovarian failure, a hallmark of Type I BPES.

Case 3: 38-year-old Male (Second Child)

The third case involved a 38-year-old male, the second child, who presented with similar complaints of reduced vision and eyelid drooping. On examination:

ISSN: 0975-8585

Right Eye: Visual acuity was 6/18.

Left Eye: Visual acuity was 6/24.

He also had **bilateral moderate ptosis**, **blepharophimosis**, and **epicanthus inversus**.

The anterior segment and fundus examinations were within normal limits in both eyes.

Case 4: 30-year-old Female (Third Child)

The fourth case involved a 30-year-old female, the third child, who presented with progressive diminution of vision and drooping eyelids. On examination:

Right Eye: Visual acuity was 6/24.

Left Eye: Visual acuity was 6/24.

She had **bilateral moderate ptosis**, **blepharophimosis**, and **epicanthus inversus**.

The anterior segment and fundus examinations were within normal limits in both eyes.

Like her elder sister, she was nulligravida and had amenorrhea, suggestive of **primary ovarian failure**.

Case 1: 64-year-old Male (Father)

Case 2: 40-year-old Female (First Child)

ISSN: 0975-8585

Case 3: 38-year-old Male (Second Child)

Case 4: 30-year-old Female (Third Child)

DISCUSSION

BPES is a rare congenital disorder that follows an autosomal dominant inheritance pattern, with affected individuals typically exhibiting the classic features of blepharophimosis, ptosis, epicanthus inversus, and telecanthus. The syndrome results from mutations in the **FOXL2** gene, located on chromosome 3q23. This gene plays a critical role in the development of the eyelids and ovaries, which explains the association of Type I BPES with premature ovarian failure.

In our case series, all four family members displayed the hallmark features of BPES, including bilateral blepharophimosis, ptosis, and epicanthus inversus. However, only the female members exhibited signs of premature ovarian failure, which is consistent with the presentation of Type I BPES. The presence of visual impairment in all family members, despite the absence of systemic or other facial abnormalities, underscores the importance of early diagnosis and intervention to prevent long-term visual complications such as amblyopia.

BPES may be associated with other extraocular findings, such as broad nasal bridges, arched palates, and ear abnormalities. In rare cases, mental retardation has been reported, though this was not observed in our family series. In this family, there were no significant facial abnormalities or mental

disorders, and the patients' anterior segments and fundi were largely normal, apart from the visual acuity issues related to the ptosis and blepharophimosis.

The management of BPES typically involves a combination of surgical and non-surgical interventions. Medial canthoplasty is often performed to correct telecanthus, while ptosis correction surgery improves both the aesthetic appearance and the visual field. For female patients with Type I BPES, hormone replacement therapy is recommended to address ovarian insufficiency. Early genetic counselling and regular ophthalmic evaluations are essential to optimize long-term outcomes.

In this family, counselling was provided to inform the patients about the nature of their condition, the risks of amblyopia, and the available treatment options. Ptosis correction surgery and medial canthoplasty were discussed as potential interventions to improve their visual fields and facial appearance. The female patients were also advised to seek further evaluation and management for their primary ovarian failure.

CONCLUSION

BPES is a rare genetic disorder that affects the oculofacial region and, in some cases, the ovaries. It presents a significant challenge due to its potential to cause visual impairment if not treated early. Surgical interventions such as ptosis correction and medial canthoplasty, combined with hormonal therapy for patients with ovarian insufficiency, are effective management strategies. Early diagnosis, family counselling, and genetic testing are key components in managing this condition and improving the overall quality of life for affected individuals. In the case of the family presented here, surgical intervention is planned to enhance the visual field and correct the aesthetic concerns associated with the condition. The importance of timely intervention and the need for public awareness cannot be overstated, especially in regions where access to specialized care may be limited.

This family's case highlights the importance of genetic counselling, early surgical correction, and ongoing ophthalmic follow-up to prevent amblyopia and other visual complications associated with BPES. The positive outlook for the family emphasizes that, with appropriate management, individuals with BPES can achieve significant improvements in both function and appearance.

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