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# Unilateral Acorea with Posterior Embryotoxon an Incidental Finding in Adult - Case Report

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**Abstract:** ***Acorea is a rare condition with a congenital absence of a pupil. It can present as familial or sporadic mode; however, the exact mode of inheritance remains unknown, with a likelihood of autosomal dominant inheritance. It leads to stimulus deprivation amblyopia (SDA) in the affected eye. If detected early in life, it can be managed surgically by pupilloplasty. There are several documented congenital anomalies of the pupil, such as dyscoria, anisocoria, microcorea, heterochromia, polycorea, etc. In this communication we delineate an unusual case of a 40-year-old female with unilateral acorea in association with posterior embryotoxon.***

**Keywords:** Acorea, Posterior Embryotoxon, Pupilloplasty, Acorea, congenital absence of pupil, autosomal dominant, stimulus deprivation amblyopia

## 1. Introduction

Acorea is a congenital anomaly of pupil characterised by absence of aperture in iris tissue. (1) It is one of the rare congenital anomalies of pupil among many others.

Understanding the developmental of Iris:

In the human embryo, eyes develop through a complex and delicate process. Interruption of this process leads to malformations of the eye or related structures that can result in both functional and aesthetic deficits. Congenital ocular malformations are relatively rare. Eye development occurs in the human embryo from approximately the third week through the tenth week of gestation. Ocular tissues are of mesodermal and ectodermal origin. The retina, ciliary body, optic nerves, and iris are derived from neuroepithelium. (2)

During the fourth week of pregnancy, upon neural tube closure, the optic grooves are transformed into optic vesicles, the optic vesicles develop into the optic cup, the inner layer of which forms the retinal pigment epithelium, with the retina being formed by the outer layer. The iris and ciliary body develop from the middle part of the optic cup. As the optic cup invaginates, the lens placode forms through the thickening of the ectoderm and eventually forms the lens vesicle upon separation from the ectoderm. (2)

**Pathophysiology**
Congenital ocular defects arise through a variety of mechanisms and in association with many different syndromes and diseases. The Pax-6 gene has been identified as one of vital importance to the initiation of eye development and its proper progression. Many genetic mutations and environmental factors are implicated in ocular malformations. (2)

During embryonic period Iris develop from anterior most part of the optic canal/ neural crest and iris muscle develop from mesodermal tissues Anterior segment structures developing from the neural crest are corneal stroma, endothelium, iris stroma and trabecular meshwork, hence due to disturb in neural crest development anterior segment dysgenesis occurs and can lead to different manifestation in form of abnormality in iris tissues, cornea and trabecular meshwork. It manifested in form of iris abnormalities, glaucoma and corneal opacities in different combination of Iridocorneal dysgenesis syndrome / Anterior segment dysgenesis.

Since acorea is also a part of iris abnormality, it is due to defect in migration of 3rd wave of neural crest precursor to iris stroma during intrauterine life. (3)

Physiology: Due to absence of opening light cannot enter in eye leading to stimulus deprivation amblyopia.

Differential diagnosis: It should be differentiated from other anomalies like Persistent pupillary membrane, which is commonly occurring congenital anomaly because of anterior remnants of tunica vasculosalentis and appear as strands of connective tissue, usually it is a very thin membrane and does not obstruct light and usually an incidental finding. but in some cases, it may present as thick membrane and can

lead to amblyopia and need surgical excision or ND yaglysis.(4)

In the case reported, we see a rare anomaly acorea, along with unusual combination of posterior embryotoxon and absence of strabismus.

In this case we also noticed prominent thick white line which is posterior embryotoxon. (figure 1)

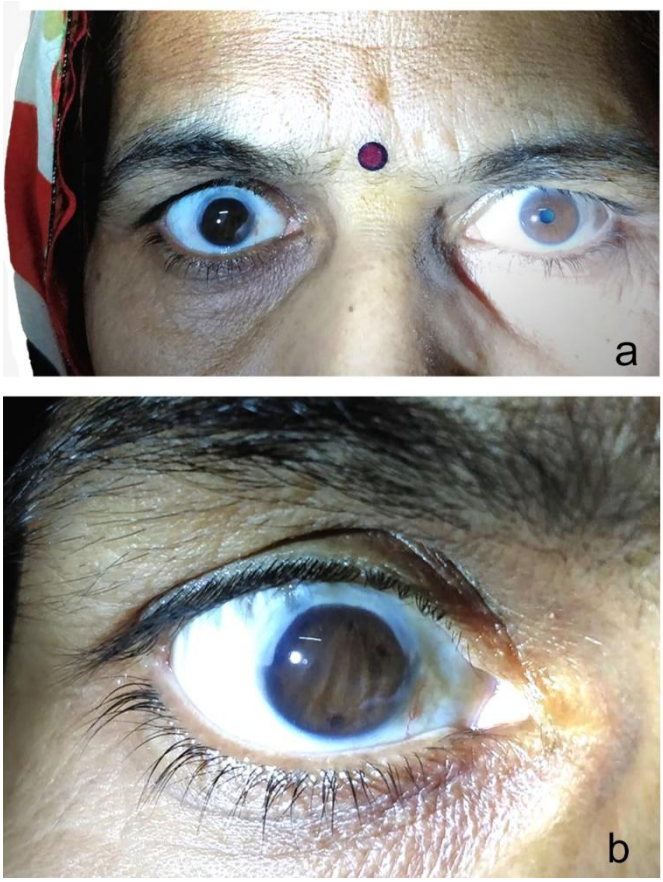


**Figure 1: (a)** Posterior Embryotoxon**; (b)** Close up of the Eye

Posterior embryotoxon is a clinical and histologic term referring to displacement of Schwalbe's line anterior to the limbus in the cornea. Axenfeld's anomaly, which is characterized by the anterior displacement of the Schwalbe's line, thereby causing posterior embryotoxon.

## 2. Case Study

A 42-year female, housewife, mother of three children aged 20, 16 and 14 years respectively, of low socio-economic status, residence of Indore, Madhya Pradesh. Un-educated, came to the eye department with complaint of watery eye with, itching and discomfort in left eye. She, also mentioned difficulty seeing the near and small objects clearly and sharply since almost one year.

On detailed history taking she told absence of vision in right eye since birth, and dismissed any history of trauma of any kind or any other related ocular morbidity. No significant history of any other systemic infection could be traced that lead to loss of vision.

On observation, the patient had acorea in the right eye and allergy in the left.

On asking family history for acorea, the patient's response was negative. From both paternal as well as maternal side, also no case reported in any of her children.

On Detailed eye examination there was No PL / PR in Right Eye. While, vision was normal 6/6, with glasses N6 in her left eye.

Right eye Slit lamp examination revealed thick iris tissue presenting at pupillary area with no pupillary aperture. Iris structure was also abnormal.

On attempting to search for pupil we did gonioscopy, since embryotoxon was present it was unclear and not very reliable,

Applanation tonometery was normal in both the eyes.

Strabismus, which is common in cyclopes vision, was absent in the patient.

To further investigate, a USG B Scan (figure 2b) was done for the right eye, which revealed non-cataractous normal lens with clear vitreous and attached retina.

Anterior segment OCT revealed open angle structures with no abnormality. Other visible thickening, posterior embryotoxon and occlusion of light. (figure 2a)

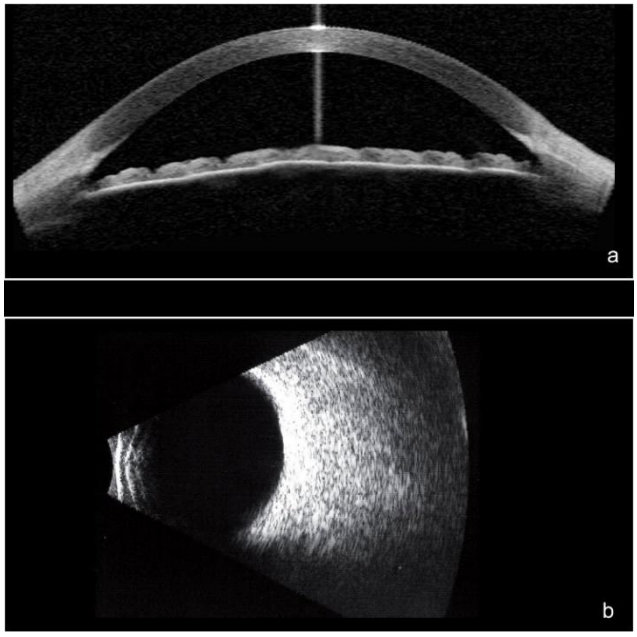


**Figure 2 (a):** Anterior Segment Optical Coherence Tomography (AS-OCT)**; (b)** USG B-Scan (Brightness Scan)

All systemic investigations including blood test, serum electrolyte, ECG, LFT, RFT were normal.

No systemic problem was detected,
Ruling out the differential diagnosis of 'Persistent pupillary membrane", and absence of pupillary opening with folds of iris tissue at center of iris which is a cardinal feature of acoria, we were able to diagnose as a case of unilateral

acorea with posterior embryotoxon.

We suggested pupiloplasty, with explaining about amblyopia but, patient unwillingness to undergo surgical management prevented us to explore the intervention and try regaining the vision.

## 3. Discussion

It is a rare case of acorea along with posterior embryotoxon, to our knowledge if the case was detected at an early age the results of pupiloplasty would have prevented amblyopia and could have restored functional vision of the patient, which could have some favourable effects on patient's life.

Not much is known about acorea, usually it is sporadic, although it may be hereditary too. Exact genetic pattern of inheritance is unknown but Autosomal dominant inheritance is most probable cause. (1)

Its association with Iridocorneal dysgenesis in isolation indicates that PAX 6 genes may be defective and during 7th month of intrauterine life defective migration of neural crest cells can lead to this type of defect. It can be associated with other ocular abnormalities but systemic abnormalities are mostly not associated. Acorea can be associated with microphthalmos, cataract, and iridocorneal dysgenesis. (1)

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## Author Profile

**Dr. Vandana Jain**, completed her MBBS in 2001 from NSCB Medical College, Jabalpur, and then done her post-graduation from esteemed institute of MGM Medical College, Indore. She has done her anterior segment fellowship from Aravind eye hospital, Puducherry, Medical retina and laser course from Aravind eye hospital Madurai, as well as Ratan tata phaco fellowship from Sankara netralaya, Chennai. At present she is a senior consultant in government sector ESIC hospital, Indore since 2009 and serving the neediest persons of society. She has done many cataract and other anterior segment surgeries during this period.

**Dr. D Rajakannan,** an esteemed alumnus of Annamalai University with an MBBS degree and a specialization in Ophthalmology from Aravind Hospital, Madurai, has dedicated over 25 years to clinical practice in both private and public sector hospitals. Throughout his illustrious career, he has served millions of patients and has been recognized with numerous national and state awards for his innovative contributions, including stem cell grafting and crutch glasses. Currently, he leads the Ophthalmology Department at ESIC, MH & ODC, Indore guiding his subordinates on the path to success and continuing to make significant strides in his field.

**Dr. Priyanka,** an accomplished ophthalmologist, completed her M.B.B.S from DAVV University, India in 2013, and went on to pursue M.S Ophthalmology from Malwanchal University, India in 2018. During her tenure as an Assistant Professor at IMCHRC, Indore, she developed a keen interest in anterior segment surgeries. This led her to complete a fellowship in Comprehensive Ophthalmology from Shivanand Mission Eye Hospital, Gujarat, India. With a decade of work experience under her belt and thousands of successful surgeries and procedures to her credit, she has been serving the public as a Consultant Ophthalmologist at ESIC hospital, Indore since 2021.

**Dr. Saurabh,** a distinguished alumnus of "Jamia Hamdard", one of India's top 20 universities, completed his graduation in Occupational Therapy with a focus on Ergonomics in 2011. He further pursued Post Graduation in Occupational Therapy (Neurological Disorders), with his research centered on Tele-rehabilitation. This pioneering study on Tele-rehabilitation in the country was submitted to "Guru Gobind Singh Indraprastha University", Delhi, and served as the foundation for setting up a tele-rehab unit at a leading institute in New Delhi. With over 15 years of experience and a passion for knowledge sharing, he currently heads the Occupational Therapy Department at the ESIC MH & ODC, Indore, since November 2017. His recent work unveiled the current status of occupational therapy practices in India

### Contribution Details

The manuscript has been read and approved by all the authors, that the requirements for authorship as stated earlier in this document have been met, and each author believes that the manuscript represents honest work.

| | Contributor 1 | Contributor 2 | Contributor 3 | Contributor 4 |
|---|---|---|---|---|
| Concepts | ✔ | ✔ | | |
| Design | ✔ | | | |
| Definition of intellectual content | ✔ | ✔ | | |
| Literature search | ✔ | | ✔ | |
| Data acquisition | ✔ | | ✔ | |
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