

A Case Report on Ocular Albinism

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Abstract: Ocular albinism is an inherited condition characterized by reduced ocular pigmentation and developmental abnormalities that may affect vision. Foveal hypoplasia and nystagmus are common features that limit visual acuity, while cataract can cause further deterioration. This case report describes a 50-year-old male with ocular albinism, foveal hypoplasia, pendular nystagmus, and bilateral nuclear cataract. The patient presented with reduced vision of 5/60 in both eyes. Clinical examination revealed iris and fundus hypopigmentation, while optical coherence tomography demonstrated foveal hypoplasia. Sequential cataract surgery was performed using phacoemulsification with intraocular lens implantation in both eyes. The postoperative recovery was uneventful, and best-corrected visual acuity improved to 6/24 in both eyes. Although the underlying foveal abnormality restricted further visual improvement, cataract removal provided a meaningful improvement in vision. This case highlights the importance of identifying ocular developmental abnormalities before cataract surgery and counselling patients about realistic visual outcomes. Cataract surgery can improve vision in patients with ocular albinism, even when pre-existing foveal hypoplasia limits the final visual acuity.

Keywords: Ocular Albinism, Foveal Hypoplasia, Bilateral Cataract, Phacoemulsification, Visual Acuity

1. Introduction

Albinism comprises a heterogeneous group of inherited disorders characterized by reduced or absent melanin pigmentation. It originates from the Latin word "albus" which means white. Ocular involvement may occur predominantly, in eyes as in ocular albinism or may be associated with variable cutaneous hypopigmentation in oculocutaneous forms. The first group is less common. So proper ophthalmic and dermatological evaluation is needed.

The ocular manifestations include iris transillumination or hypopigmentation, fundus hypopigmentation, foveal hypoplasia, nystagmus, reduced visual acuity or abnormal optic nerve development. Foveal hypoplasia is an important cause of reduced visual potential in patients with albinism.

The diagnosis of ocular albinism is primarily clinical and may be supported by detailed anterior segment examination, Optic Coherent Tomography (OCT), and genetic testing if indicated. OCT is particularly useful for identifying and grading foveal hypoplasia.

As age progresses cataract may coexist with albinism and can further reduce vision. Recognition of the underlying developmental ocular abnormalities is important when counselling the patients regarding the expected visual outcome following cataract surgery.

We report a case of an adult male with clinical features of ocular albinism, foveal hypoplasia and bilateral visually significant cataract who underwent sequential cataract surgery with improvement in visual acuity.

2. Case Report

A 50-year-old male presented to the ophthalmology outpatient department with complaints of defective vision in both eyes. There was no history of ocular trauma or previous ocular surgery.

On examination, his visual acuity was 5/60 in both eyes. Pendular nystagmus was present. The eyelids were within normal limits. The cornea was clear in both eyes. The iris shows hypopigmentation. The pupils were normal in size and reaction to light.

Slit-lamp examination revealed **nuclear sclerosis grade 2-3 cataract in both eyes.**



Figure 1: Post cataract surgery BE

On dilated fundus examination, the fundus shows generalized hypopigmentation and absent foveal reflex suggestive of foveal hypoplasia.

In view of visually significant bilateral cataract, the patient was counselled regarding the expected vision outcome particularly in view of the underlying foveal hypoplasia and nystagmus.

Cataract surgery was subsequently performed in both eyes by Phacoemulsification with implantation of an intraocular lens.

The postoperative period was uneventful. Following surgery, the patient was pseudophakic in both eyes. Best-corrected visual acuity improved to **6/36 in both eyes**, with no further improvement on pinhole examination.

The patient was advised regular follow-up and appropriate postoperative care.

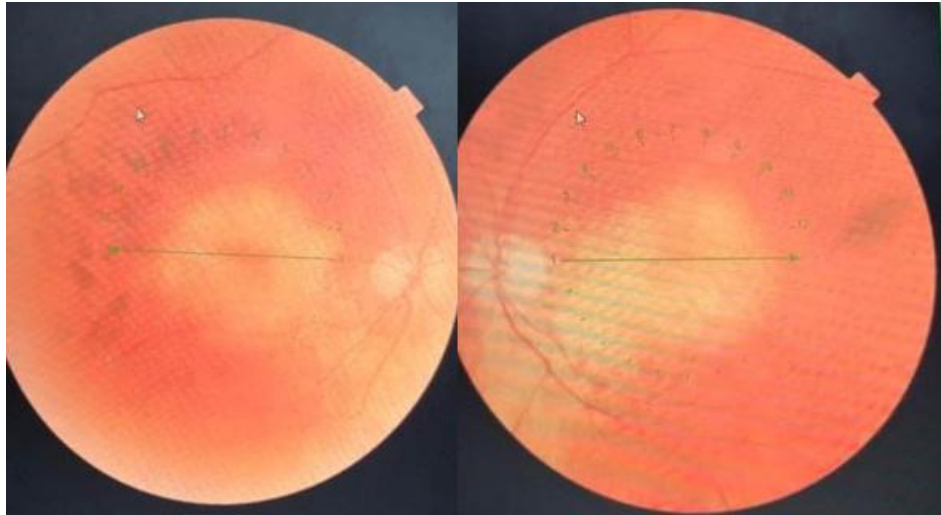


Figure 2: BE fundus pic. showing foveal hypoplasia

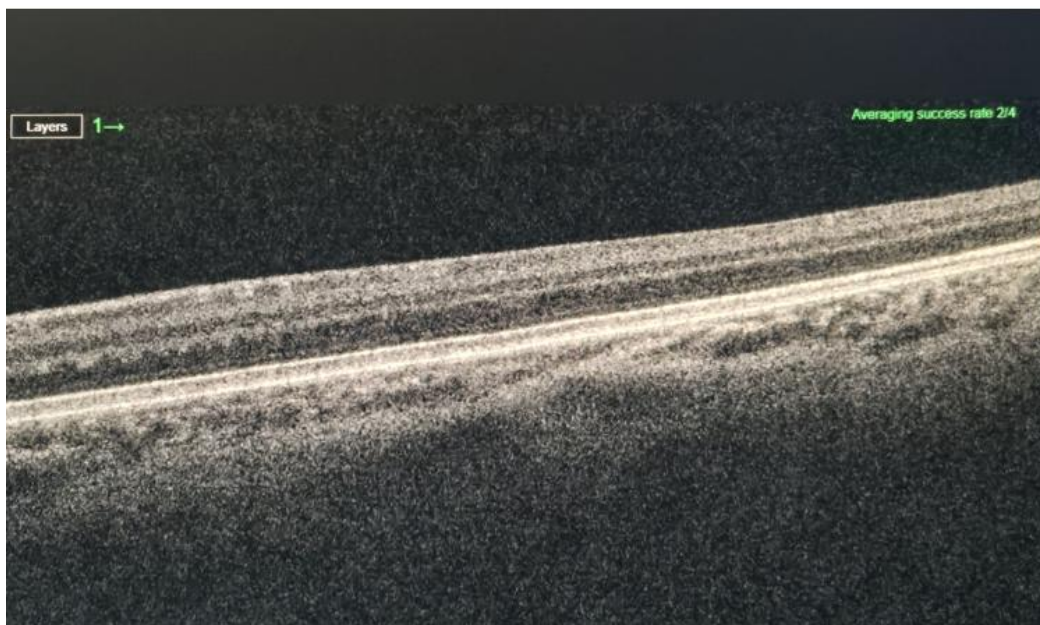


Figure 3: (a) LE OCT showing foveal hypoplasia

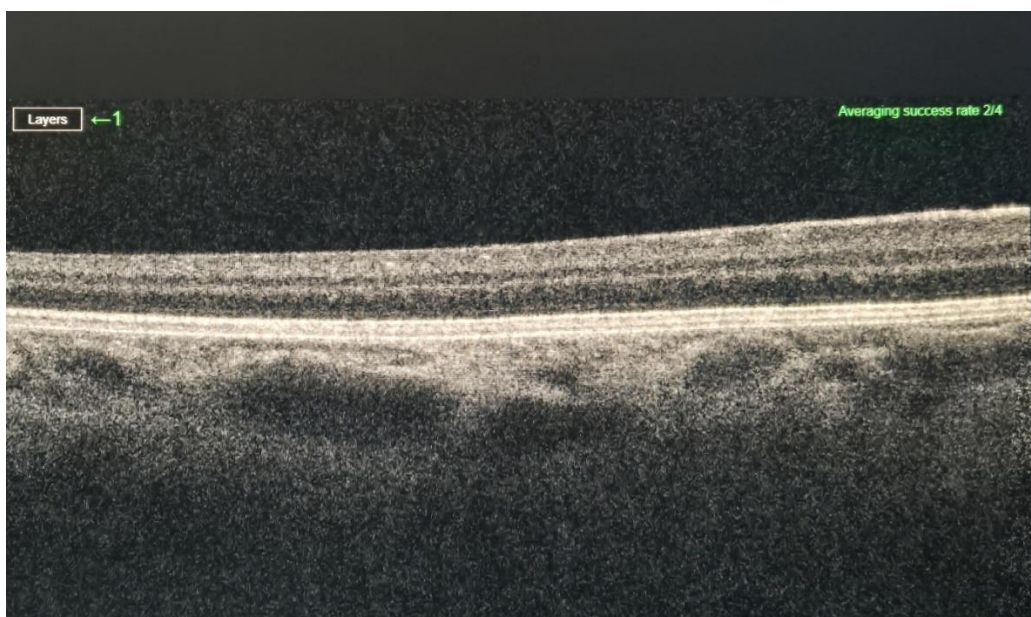


Figure 3: (b) RE OCT showing foveal hypoplasia

3. Discussion

Ocular albinism is characterized by abnormalities in ocular pigmentation and its development. The reduction in melanin during ocular development can affect several structures, particularly the fovea and optic disc.

Foveal hypoplasia is a vital structural abnormality associated with albinism and contributes significantly to the reduced visual acuity. Clinically it may be suspected by the absence of the normal foveal reflex and can be demonstrated more accurately with optical coherence tomography (OCT)

Nystagmus is another common manifestation and may be present from early childhood. Iris hypopigmentation and fundus hypopigmentation are useful clinical clues to the diagnosis.

In the present case, the combination of iris hypopigmentation, nystagmus, fundus hypopigmentation and foveal hypoplasia was suggestive of ocular albinism. The presence of bilateral nuclear cataract provided an additional, potentially treatable cause of visual impairment.

An important consideration in cataract surgery in patients with albinism is that the postoperative visual acuity may remain limited by the pre-existing foveal hypoplasia and other developmental abnormalities. Therefore, appropriate counselling is essential.

Despite these limitations, the improvement from 5/60 to 6/36 following cataract surgery demonstrates that removal of the lenticular opacity can provide clinically meaningful visual improvement in appropriately selected patients

4. Conclusion

Ocular albinism should be considered in patients with hypopigmented iris, nystagmus, fundus hypopigmentation and reduced visual acuity. Foveal hypoplasia may substantially limit the visual potential. However, coexisting cataract represents a treatable cause of further visual Deterioration, cataract surgery can result in significant functional visual improvement, although patients should be counselled regarding the limitations imposed by underlying foveal developmental abnormalities. This case highlights importance of early recognition of characteristic ocular features, appropriate management of treatable lesions and realistic counselling regarding visual prognosis are essential in patients with ocular albinism

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