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Ophthalmology

OPHTHALMOLOGICAL MANIFESTATIONS OF OCULAR ALBINISM

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ABSTRACT **Purpose-**To study the ophthalmic manifestation in patients of oculocutaneous albinism. Methods- This was a prospective observational study that involved 12 eyes of 6 patients with oculocutaneous albinism complaining of diminution of vision. Complete ophthalmic examination was done in diffuse light followed by direct ophthalmoscopic examination and then further examination was taken forward by doing optical coherence tomography (OCT). **Results-**There were 4 males and 2 females albinism include hypopigmented eyebrows and eyelashes,reduced visual acuity, photophobia and nystagmus. On slit lamp examination there is iris transillumination due to iris hypopigmentation. Fundus findings include clear view of choroidal vessels, pale retina , foveal hypoplasia and indistinct optic disc margin. **Conclusion-**All the patients with oculocutaneous albinism had poor vision with mostly nystagmus seen and few showed photophobia. Hypopigmentation of eyebrow and eyelashes along with iris hypopigmentation was seen and iris transillumination was present in most of the patients when examined under slit lamp biomicroscopy. OCT and ophthalmoscopy shows foveal hypoplasia (absence of a foveal pit) with pale background.

KEYWORDS : Pendular nystagmus, Iris transillumination, Foveal hypoplasia strabismus

INTRODUCTION

Albinism (origin - Latin word albus[1] which means white) refers to a group of hereditary disorders with an abnormality of melanin synthesis or distribution. There are various types of albinism The most commonly thought of presentation is that of oculocutaneous albinism (OCA[2]). Till date, seven types of nonsyndromic albinism (OCA1 to OCA7) have been described. The main clinical features are hypopigmentation of hair, skin, and eyes. Ophthalmic manifestations include photophobia, nystagmus, defective vision, and squint. The OCA1A being the most severe type with a complete lack of melanin production throughout life, while the milder forms OCA1B, OCA2, OCA3 and OCA4 show some pigment accumulation over time. All four types of OCA are inherited as autosomal recessive disorders.It is estimated to occur worldwide at a range of 1 in 5,000 to 1 in 40,000 population[3,4]

- ✓ OCA1: Prevalence is 1 in 40,000 worldwide but one of the most common forms in America and China (70% of cases)
- ✓ OCA2: The most common worldwide (1:39,000). Prevalence as follows: African Americans (1:10,000), overall Americans (1:36,000), Sub-Saharan Africa (1:3,900)
- ✓ OCA3: Prevalence is 1: 8500 of African individuals primarily in southern Africa. It can also be seen in Pakistani, German, Indian, and Japanese populations.
- ✓ OCA4: Prevalence is 1:100,000 but accounts for 24% of Japanese OCA. It is also described in German, Turkish, Indian, Korean, Chinese, Danish, and Moroccan, populations.
- ✓ OCA5: Very rare, only mentioned in a case report of a Pakistani family
- ✓ OCA6: Very rare, case reports in a Chinese family and a man from eastern India
- ✓ OCA7: Very rare

- ➢ Hermansky-Pudlak syndrome (HPS): Prevalence is 1:500,000 worldwide but 1:1800 in Puerto Rico
- ➢ Chediak-Higashi syndrome (CHS): Very rare (less than 500 cases published in the past 20 years)
- ➢ Angelman syndrome (AS) and Prader-Willi syndrome (PWS): Prevalence of AS (1:12,000 to 20,000) and PWS (1:15,000) are higher than the OCAs. However, only approximately 1% of AS and PWS sufferers have contiguous gene deletions that lead to OCA2-like presentation
- ➢ Oculocutaneous albinism (Oa1): Prevalence is 1:50,000

Clinical Features

- ➢ Strabismus due to misrouted optic nerve fibres at the chiasm
- ➢ Photophobia : Sensitivity to bright light and glare can occur due to scattering of light within the eye. Patients may prefer to wear sunglasses to reduce their sensitivity to light
- ➢ Refractive Errors: Both myopia and hyperopia can occur, and astigmatism is very common
- ➢ Poor visual acuity: Near vision is often better than distance vision. Generally, those who have the least amount of pigment (i.e. most severely affected) have the poorest vision.
- ➢ Pendular nystagmus
- ➢ Foveal hypoplasia (absence of a foveal pit): Because the retinal pigment epithelium (RPE), which is necessary for macular formation, has an inappropriate pigmentation in albinism, the retina does not develop correctly before birth or during infancy. Optical coherence tomography (OCT) can demonstrate an absence of the foveal pit and the loss of normal thinning of the retina. Foveal hypoplasia is the single most important contributor to poor vision in albino patients[5]
- ➢ Reduced stereopsis (depth perception) and binocular vision due to strabismus
- ➢ Reduced iris pigmentation
- ➢ Iris transillumination: On slit lamp examination as light reflects off the retina and back through the iris
- ➢ Due to hypomelanosis of the retinal epithelium, the retina turns yellow or orange and loses its characteristic red look. Prominent choroidal veins are also present.
- ➢ Patients with albinism show an asymmetry of VEP between the two eyes secondary to misrouting of optic pathways.

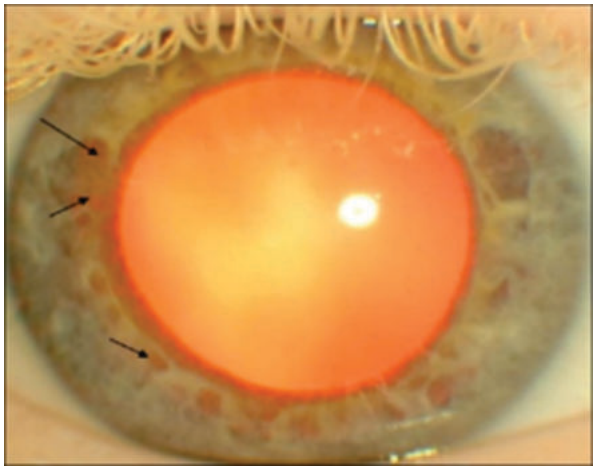


Figure 1: Iris Transillumination (arrows) Due To Escape Of Reflected Light From The Retina Through The Iris.[6]

MATERIAL AND METHOD

This was a prospective observational study that involved 12 eyes of 6 patients with oculocutaneous albinism complaining of diminution of vision. Patients were selected after all the criterias were fulfilled, from Dept.of Ophthalmology of M.L.B. Medical college, Jhansi ,Uttar Pradesh and were followed from 1st November 2022 - 1st April 2023. The study was performed under the Helsinki Declaration of 1975, as revised in 2000. The necessary permission from the Ethical and Research Committee was obtained for the study.

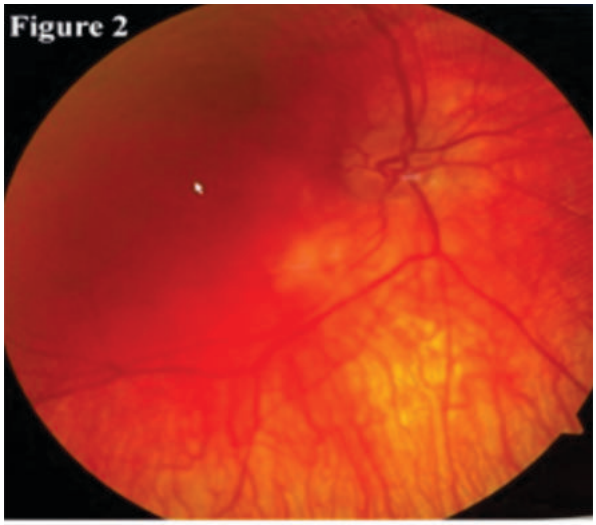
Inclusion Criteria

All patients who presented to the OPD of MLB medical College Jhansi with the complaint of dimunition of vision and nystagmus who were found to have oculocutaneous albinism were included.

Exclusion Criteria

1. Patients with systemic diseases were excluded from the study as they can affect retina too.
2. Patients with other retinal disorders
3. Patients with any recent intraocular surgery
4. Patients with the history of trauma
5. Mentally or physically unfit patients

All patients were subjected to a detailed history taking including family history too and best corrected visual acuity (BCVA) was given. All patients had complete ophthalmic examination including biomicroscopic slit lamp examination, fundus examination and fundus photography and optical coherence tomography. Optical coherence tomography examination was done in dilated pupil using commercially available Cirrus HD-OCT Model 4000 - Carl Zeiss Meditec, Inc., Dublin,California, USA or Spectralis OCT Heidelberg Engineering.



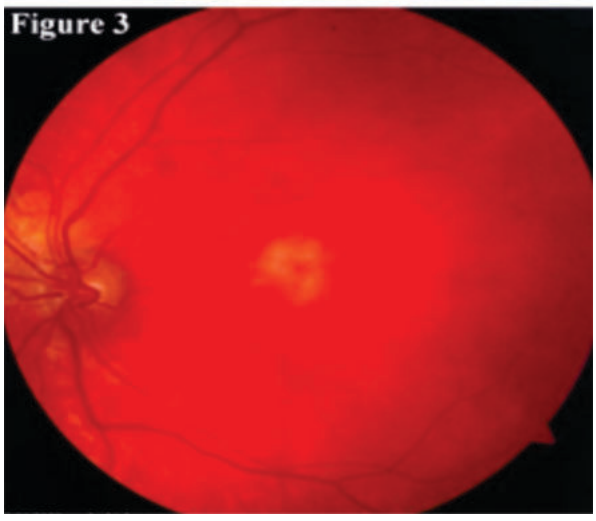


Figure 2 And 3: Albinic Fundus Of Right And Left Eye Respectively Of The Patient Who Presented To Our Department In M.L.B Medical College,jhansi

RESULT

A total of 14 eyes of 7 patients were studied. We included eyes with complaint of diminution of vision. There were 4 males and 3 females and mostly it was seen that right eye was more involved than the left eye. All eyes showed features typical of oculocutaneous albinism (nystagmus, photophobia, strabismus, iris transillumination)

CLINICAL FEATURES	TOTAL (IN %)
Poor visual activity	98
Refractive error	90
Photophobia	72
Nystagmus	88
Strabismus	60
Iris transillumination	88
Foveal hypoplasia	88

DISCUSSION

Albinism is a group of heritable conditions associated with decreased or absent melanin in ectoderm-derived tissues (most notably the skin, hair, and eyes), yielding a characteristic decrease in skin pigmentation. Albinism, in any of its forms, is the result of heritable mutations that lead to defective melanocytes, unable to properly synthesize melanin or distribute it through dermal tissues. The exact number of melanocytes in the skin are preserved, unlike conditions such as piebaldism and vitiligo, where melanocytes are absent. The disorder can be divided into two groups: oculocutaneous albinism (OCA),the most common condition among hypopigmentation disorders with varying degrees of involvement of the eyes, hair, and skin; the second group is the less common group called oculocutaneous albinism (OA) with disease involvement limited to the eyes. Albinism has classically been organized into two broad categories, tyrosinase-positive (mild to moderate) and tyrosinas-negative (severe) albinism.

Nitisinone is an FDA-approved inhibitor of tyrosine degradation for hereditary tyrosinemia. Brooks and colleagues[7] hypothesized that the relative deficiency of tyrosinase in OCA type 1 may be ameliorated by increasing the concentration of tyrosine with nitisinone and thus improving pigmentation with OCA In mice studies, nitisinone indeed improved pigmentation in fur and irides, suggesting potential for benefit in humans affected by OCA 1.

A group by Martinez-Garcia et al attempted to supplement L-DOPA, an intermediate metabolite of melanin synthesis pathway in mouse models. Various experiments suggested that L-DOPA supplementation may help overcome visual abnormalities associated with albinism[8] Phase 2 clinical trial is underway in the US to study the effects of L-Dopa supplementation in human subjects

A Japanese group suggested the use of aminoglycosides that can read through non-sense mutations for a potential treatment measure for common mutations found in albinism Patients with albinism should be strongly encouraged to limit their sun exposure by avoiding outdoor occupations and by using sun-protective products like appropriate clothing, large-brim hats, sunglasses, and sunscreen agents. Patients with albinism are at higher risk for squamous cell and basal cell cancers in sun-exposed areas.

DNA from cultured amniocytes or DNA collected from chorion villus sampling (CVS) at 10–12 weeks of gestation can both be used for the testing. The use of molecular genetic analysis for preimplantation diagnosis is theoretically also feasible, however to our knowledge. Skin samples taken from foetuses have previously been used for prenatal diagnosis[9,10]

CONCLUSIONS

In this study it was also seen that Individuals having oculocutaneous albinism presented with photophobia, nystagmus and refractive error. Most of the patients showed iris transillumination and foveal hypoplasia with prominent choroidal vessels in fundus examination Patients with melanin deficiencies and tyrosine mutations exhibit hypopigmented skin, hair, and ocular tissues. Due to foveal hypoplasia patients have poor vision .Regular ophthalmic examination should be done and screening for skin cancers should also be done periodically as they are more prone for skin cancers.

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