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Ophthalmic Features in Patients of Oculocutaneous Albinism

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Abstract
Purpose - *to study the ophthalmic features in patients of oculocutaneous albinism.*
Methods- *This was a prospective observational study that involved 14 eyes of 7 patients with oculocutaneous albinism complaining of diminution of vision. Complete ophthalmic examination was done in diffuse light followed by direct opthalmoscopic examination and optical coherence tomography.*
Results-*There were 4 females and 3 males .Ophthalmic features in patients of oculocutaneous albinism include hypopigmented eyebrows and eyelashes , reduced visual acuity, photophobia and nystagmus. On slit lamp examination there is iris transillumination due to iris hypopigmentation.There is red reflex in pupillary area. 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 nystagmus and photophobia. Hypopigmentation of eyebrow and eyelashes along with iris hypopigmentation leading to iris transillumination was present in most of the patients.OCT and ophthalmoscopy shows foveal hypoplasia and pale background and prominence of choroidal vcasculature.*
Keywords: *Oculocutaneous albinism, nystagmus, hypopigmentation, iris transillumination, foveal hypoplasia.*

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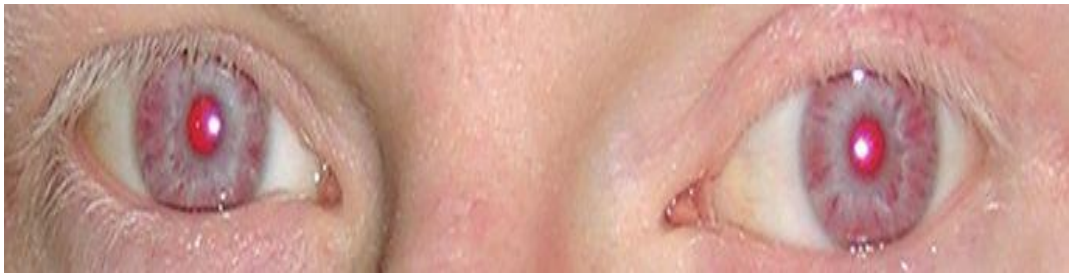
I. Introduction

Oculocutaneous albinism (OCA) is a group of inherited disorders of melanin biosynthesis characterized by a generalized reduction in pigmentation of hair, skin and eyes. The prevalence of all forms of albinism varies considerably worldwide and has been estimated at approximately 1/17,000, suggesting that about 1 in 70 people carry a gene for OCA[1]. The clinical spectrum of OCA ranges, with 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. At least four genes are responsible for the different types of the disease (*TYR*, *OCA2*, *TYRP1* and *MATP*). Diagnosis is based on clinical findings of hypopigmentation of the skin and hair, in addition to the characteristic ocular symptoms. Due to the clinical overlap between the OCA forms, molecular diagnosis is necessary to establish the gene defect and OCA subtype. Differential diagnosis includes ocular albinism, Hermansky-Pudlak syndrome, Chediak-Higashi syndrome, Griscelli syndrome, and Waardenburg syndrome type II. Carrier detection and prenatal diagnosis are possible when the disease causing mutations have been identified in the family. Correction of strabismus and nystagmus is necessary and sunscreens are recommended. Regular skin checks for early detection of skin cancer should be offered. Persons with OCA have normal lifespan, development, intelligence and fertility.

- In OCA1A the hair, eyelashes and eyebrows are white, and the skin is white and does not tan. Irises are light blue to almost pink, and fully translucent . Pigment does not develop and amelanotic nevi may be present. The symptoms do not vary with age or race. Visual acuity is 1/10 or less, and photophobia is intense.[2]



Eyes from a patient with OCA. Irises are almost pink, and fully translucent.

- In OCA1B, the hair and skin may develop some pigment with time (after 1 to 3 years), and blue irises may change to green/brown. Temperature-sensitive variants manifest as having depigmented body hairs, and pigmented hairs on hands and feet due to lower temperatures. Visual acuity is 2/10. This phenotype was previously known as yellow albinism.
- In OCA2, the amount of cutaneous pigment may vary, and newborn nearly always have pigmented hair. Nevi and ephelids are common. Iris color varies and the pink eyes seen in OCA1A are usually absent. Visual acuity is usually better than in OCA1, and can reach 3/10. In Africans, brown OCA is associated with light brown hair and skin, and gray irises. Visual acuity may reach 3/10.
- OCA3 results in Rufous or red OCA in African individuals, who have red hair and reddish brown skin (xanthism). Visual anomalies are not always detectable, maybe because the hypopigmentation is not sufficient to alter the development.
- OCA4 cannot be distinguished from OCA2 on clinical findings.

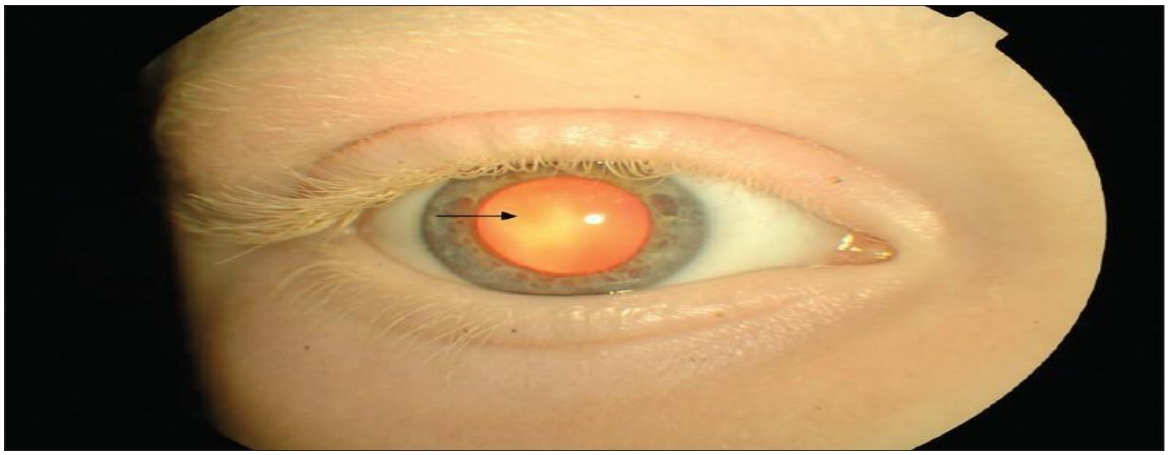
CLINICAL FEATURES

Poor vision: Vision can range from normal for those minimally affected to legal blindness or worse (vision less than 20/200) for those with more severe forms of albinism. 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.

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.

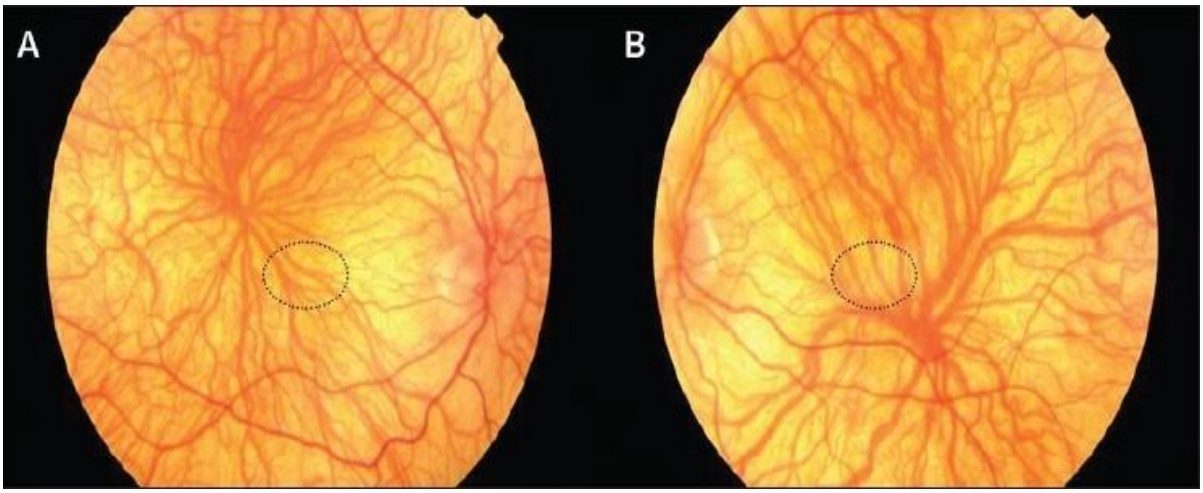
Refractive Errors: Both far-sightedness (hyperopia) and near-sightedness (myopia) can occur, and astigmatism is very common.

Nystagmus: Refers to rhythmic, involuntary, conjugate eye movement. Affected infants may have large amplitude with low frequency pattern of eye movement starting at 2-3 months of age, later changing to a pendular form without distinct fast or slow phases. Eye muscle surgery may be considered to reduce nystagmus.



Iris transillumination: The iris (the colored area in the center of the eye) in albinism has little to no pigment to screen out stray light coming into the eye. On slit lamp exam, the examiner may detect speckled or diffuse transillumination defect. This finding, while common with albinism, is not specific as iris transillumination occurs in diseases unrelated to albinism such as pseudoexfoliation, pigment dispersion syndrome, megalocornea, iris atrophy, and Axenfeld-Rieger spectrum. When present in an otherwise normal individual, this finding may indicate carrier status of a hypomelanotic gene mutation.
The iris may be translucent and the margin of the crystalline lens may be visible on transillumunation during slit lamp examination.

Foveal hypoplasia (absence of a foveal pit): In albinism, the retina (the surface inside the eye that receives light) does not develop normally before birth and in infancy because of inappropriate retinal pigment epithelium (RPE) pigmentation that is required for macular development. Optical coherence tomography (OCT) can demonstrate an absence of the foveal pit and the loss of normal thinning of the retina. Also, the foveal avascular zone is small or nonexistent with vessels crossing the area 2 disc diameter temporal to the optic disc margin. Foveal hypoplasia is the single most important contributor to poor vision in albino patients.[5]



The degree of skin and hair hypopigmentation varies with the type of OCA.
Other features include hypopigmentation of eyebrow and eyelashes.
A characteristic finding is misrouting of the optic nerves, consisting in an excessive crossing of the fibres in the optic chiasma, which can result in strabismus and reduced stereoscopic vision [3]. The abnormal crossing of fibres can be demonstrated by monocular visual evoked potential [4]. Absence of misrouting excludes the diagnosis of albinism.

II. Method and material

This was a prospective observational study that involved 14 eyes of 7 patients with oculocutaneous albinism complaining of diminution of vision. Patients were recruited from the OPD of MLB MEDICAL college, Jhansi ,Uttar Pradesh and were followed from 1st October 2019 - 1st march 2020 . It 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

1. 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 ocular systemic diseases(like diabetes) that could affect the retina.
2.Patients with other retinal disorders
3.Patients with 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, refraction using Topcon autorefractometer and best corrected visual acuity (VA) measurement. All patients had complete ophthalmic examination including biomicroscopic slit lamp examination , fundus examination with 90D lens and fundusphotography and optical coherence tomography.

Optical coherence tomography examination was done through dilated pupils, OCT examination was done through a dilated pupil using commercially available Cirrus HD-OCT Model 4000 - Carl Zeiss Meditec, Inc., Dublin,California, USA or Spectralis OCT Heidelberg Engineering.

III. Results

A total of 14 eyes of 7 patients were studied. We included eyes with complaint of diminution of vision. There were 3 males and 4 females and 60% of the studied eyes were the right eyes.

All eyes had one or more features typical of oculocutaneous albinism (nystagmus, photophobia, strabismus, iris transillumination, foveal hypoplasia)

Table1: Ophthalmic features in patients of oculocutaneous albinism

Features	Total %
Poor vision	99
Refractive error	90
Photophobia	85
Nystagmus	82
Strabismus	54
Iris transillunination	84
Prominent choroidal vasculature	80
Foveal hypoplasia	78
Pale retina	62

IV. Discussion

OK Sreelatha study shows that the incidence of oculocutaneous albinism in Oman is 1 in 30,000 live births.[6] There are various types of albinism . The main clinical features are hypopigmentation of hair, skin, and eyes. Ophthalmic manifestations include photophobia, nystagmus, defective vision, and squint. A team of biologists in brown university has discovered the way in which a specific genetic mutation appears to lead to the lack of melanin production underlying a form of albinism. About 1 in 40,000 people worldwide have type 2 oculocutaneous albinism, which has symptoms of unsually light hair and skin coloration, vision problems, and reduced protection from sunlight-related skin or eye cancers. S Biswas**,** I C Lloyd Oculocutaneous albinism (OCA) is a heterogenous group of autosomal recessive disorders affecting melanin synthesis, characterised by congenital hypopigmentation of the skin, hair, and eyes. Reduced visual acuity, photophobia, iris transillumination, foveal hypoplasia, nystagmus, and an abnormal decussation of nerve fibres at the optic chiasm are common features[7]. Creel D in his study showed a characteristic finding in oculocutaneous albinism is misrouting of the optic nerves, consisting in an excessive crossing of the fibres in the optic chiasma, which can result in strabismus and reduced stereoscopic vision[8].Bouzas EA showed the abnormal crossing of fibres can be demonstrated by monocular visual evoked potential [9]. Absence of misrouting excludes the diagnosis of albinism. All four types of OCA are inherited as autosomal recessive disorders. Thus, the parents of an affected child are obligate carriers, the recurrence risk for another affected child is 25%, and healthy sibs are at 67% risk of being carriers. Offspring of an affected person are obligate carriers. Carriers are asymptomatic.In most cases, there is no previous family history of albinism but the condition does occur in individuals of two generations of a family, so called pseudodominance, and is due to an affected person having children with a person who is a carrier.Carrier detection and prenatal diagnosis are possible when the disease causing mutations have been identified in the family. Both disease causing mutations in an affected person have to be identified and established to be on the paternal and maternal chromosome, respectively, before prenatal diagnosis can be performed in pregnancies at 25% risk for an affected child. The testing can be done on DNA extracted from chorion villus sampling (CVS) at 10–12 weeks gestation or on DNA extracted from cultured amniocytes. Preimplantation diagnosis using molecular genetic analysis is also possible in principle, but to our knowledge, this has not been carried out. Previously, prenatal diagnosis has been performed on skin biopsies from the fetus [10,11]

V. Conclusion

Individuals with oculocutaneous albinism have photophobia , nystagmus and refractive error.Most of the patients have iris transillumination and foveal hypoplasia with prominent choroidal vessels in fundus examination . Because of tyrosine mutation and melanin deficiency patients have hypopigmentation of skin and hairs and ocular structures. Due to foveal hypoplasia patients have poor vision .Regular ophthalmic examination should be done and screening for skin cancers should also be done periodically.

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