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A STUDY ON OCULAR MANIFESTATIONS OF OCULO-CUTANEOUS ALBINISM IN PATIENTS ATTENDING REGIONAL EYE HOSPITAL, KURNOOL.

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ABSTRACT **Aim :** To study ocular manifestations of oculo-cutaneous albinism patients attending Regional Eye Hospital, Kurnool. **Methods:** A total of 60 patients diagnosed with albinism, from newborn to 40 years of age and of either sex, attending Regional Eye Hospital, Kurnool during the period from August 2021 to August 2022 were included in the study. A complete ocular examination was done. Patients with other ocular pathology and patients with other hereditary and metabolic disorders were excluded from the study. **Results :** Out of 60 patients, Refractive errors were seen in 51 patients, poliosis was seen in 41 patients, iris defects were seen in 40 patients, nystagmus was seen in 32 patients, lateral displacement of medial canthus was seen in 32 patients and fundus changes were seen in all patients. **Conclusion :** As ocular albinism is an inherited disorder, counselling should be offered and affected families should be advised about visual rehabilitation to meet their needs.

KEYWORDS : Albinism, Foveal hypoplasia, Nystagmus, Iris heterochromia, Refractive errors.

INTRODUCTION

Albinism is a genetically determined heterogenous group of disorder of melanin synthesis typically resulting in no pigmentation or reduced pigmentation in which either the eyes alone (ocular albinism) or the eyes, skin, hair (oculocutaneous albinism) may be affected.

Inheritance is usually x linked recessive but occasionally autosomal recessive.

MATERIALS AND METHODS:

Data has been collected from patients attending Regional Eye Hospital, Kurnool during the period from August 2021 to August 2022. Sample size: 60

Type of study: Prospective observational study.

Inclusion Criteria:

- Patients diagnosed with albinism with age group of neonates to 40 yrs.
- Patients of either sex.

Exclusion Criteria:

- Patients with other ocular pathology.
- Patients with other hereditary and metabolic disorders.

After obtaining informed consent, detailed history was taken and complete ocular examination was done. All patients who were included in the study underwent Visual acuity testing using Snellen chart. Both presenting and best-corrected vision after refraction were measured for each eye using the Snellen chart. Objective refraction was performed with the help of a streak retinoscope, followed by subjective acceptance with which the best-corrected acuity was measured and recorded. External eye examination and anterior segment examination were done with the help of a slit lamp biomicroscope. For all patients, pupils were dilated with mydriatics and after dilatation, a fundus examination was done. Fundus picture was taken using a Zeiss fundus camera in patients with ocular albinism.

RESULTS:

Table 1: Various manifestations of ocular albinism.

Manifestation	Frequency	Percentage
Refractive Errors	51	85%
Poliosis	41	68.3%
Lateral displacement of medial canthus	32	53.3%
Iris defects	40	66.6%
Nystagmus	32	53.3%
Fundus changes	60	100%
Skin changes	60	100%

DISCUSSION:

Oculo-cutaneous albinism has 2 subtypes. Individuals with Tyrosinase negative albinism lack melanin pigment throughout all ocular structures throughout life.

Individuals with Tyrosinase positive albinism synthesize variable amounts of melanin pigment.

OCULAR FEATURES:

REDUCED VISUAL ACUITY:

The degree of vision loss is variable and is related to the type of albinism. Tyrosinase-negative albino individuals have poorer vision compared to tyrosinase positive.

NYSTAGMUS:

Nystagmus is usually horizontal and symmetrical between the two eyes.

REFRACTIVE ERRORS:

Emmetropia is rare in albinism and high refractive errors such as myopia, hyperopia and astigmatism are common.

Figure 1 **Figure 2**

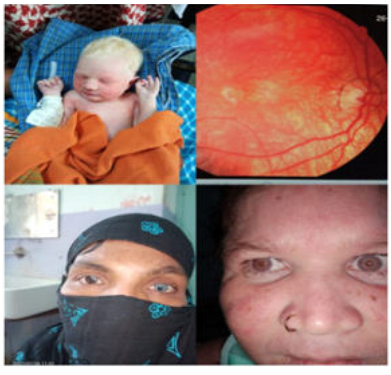

Figure 3 **Figure 4**

Figure 1: Newborn with oculocutaneous albinism.
Figure 2: Fundus picture showing foveal hypoplasia.
Figure 3: Female with iris heterochromia.
Figure 4: Female with oculocutaneous albinism with a red-colored pupil.

IRIS:
Iris is diaphanous and translucent giving rise to a pink appearance, it may be blue or dark brown with variable translucency and hypochromic irides with segmental or total heterochromia.

FUNDUS:
It lacks pigment and shows conspicuously large choroidal vessels. There is foveal hypoplasia with an absence of a foveal pit and poorly formed perimacular vascular arcades.

Other features include lateral displacement of medial canthi, squint, absence of stereopsis, etc.

CONCLUSION:
Patients with ocular albinism had various presentations, so every finding has to be considered and evaluated. Foveal hypoplasia appears to be the most significant vision-limiting factor in ocular albinism. As it is an inherited disorder, counseling should be offered and affected families should be advised about visual rehabilitation. Low vision aids improved the visual functions of majority of people with oculo-cutaneous albinism. Therefore, early referral to low vision clinic is necessary to monitor the potential enhancement in their visual functions.

Despite medical advances in many metabolic conditions, a curve for ocular albinism remains difficult to achieve.

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