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Oculocutaneous Albinism

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ABSTRACT

Oculocutaneous albinism (OCA) is a group of inherited autosomal recessive disorders characterized by reduced or absent melanin production, affecting pigmentation in the skin, hair, and eyes. This leads to significant visual impairments, including foveal hypoplasia (underdevelopment of the central retina), nystagmus (involuntary eye movements), and refractive errors such as myopia and astigmatism. This report discusses the case of a 14-year-old girl from Chitwan, Nepal, with a known history of OCA, who presented with blurred vision and photophobia. Ophthalmologic examination revealed intermittent exotropia (outward deviation of the eyes), nystagmus, and abnormal fundus vascularization.

She was diagnosed with OCA-associated visual impairment and prescribed corrective glasses for myopic astigmatism, along with tinted lenses to manage photophobia.

This case highlights the need for early diagnosis, refractive correction, and regular follow-up to optimize visual function and quality of life in individuals with OCA. Multidisciplinary care is essential for long-term management.

Keywords: Blepharophimosis syndrome, BPES, palpebral.

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INTRODUCTION

Oculocutaneous albinism (OCA) is a group of autosomal recessive disorders caused by mutations in genes involved in melanin biosynthesis, leading to hypopigmentation of the skin, hair, and eyes.[1] The lack of melanin in the eyes results in foveal hypoplasia, leading to reduced visual acuity, along with misrouting of the optic nerve fibers, which affects depth perception and binocular vision.[2] The clinical severity of OCA varies based on the type. OCA1, caused by mutations in the TYR gene, is the most severe form, where melanin production is completely absent or significantly reduced throughout life. In contrast, OCA1B, OCA2, OCA3, and OCA4 allow for some pigment accumulation over time, resulting in a milder phenotype.[3] While different types of OCA arise from distinct genetic mutations, their clinical presentations often overlap, making genetic testing crucial for accurate diagnosis and management.[4]

CASE REPORT

A 14-year-old girl from the Chitwan District, Nepal, with a known history of oculocutaneous albinism, visited the pediatric department of Bharatpur Eye Hospital (BEH). Her primary complaints were blurred vision in both eyes and light sensitivity (photophobia), which she has experienced since childhood. She was born at 38 weeks of gestation via normal vaginal delivery, with a birth weight of 2.5 kg, and she did not require NICU admission or oxygen supplementation. A family history was noted as her twin sister also had the condition. On examination, her visual acuity was 6/60 in both eyes, improving to 6/36 with pinhole testing. Dry retinoscopy revealed -1.75 cylinder at 180 degree in BE. Cycloplegic retinoscopy was also performed which revealed +2.00 spherical with -3.00 cylinder at 180degree in RE and +3.50 cylinder at 80degree in LE. The orthoptics evaluation revealed a intermittent exotropia X(T) of 25 degrees by Hirschberg test (HBT) with nystagmus in both eyes. The cover test showed intermittent exotropia X(T) along with nystagmus in both eyes, while extraocular motility was full in both eyes. On slit-lamp examination, the cornea and lens appeared clear, the anterior chamber was normal, and the pupils were round, regular, and reactive with no relative afferent pupillary defect (RAPD) in both eyes. Horizontal nystagmus was present in both eyes. Transillumination was noted from the anterior iris to the posterior region. Fundus examination under mydriasis (FEUM) showed positive transillumination, and the cup-to-disc ratio could not be accurately assessed due to abnormal vascularization. Additionally, abnormal vascular tuft was also observed around the macula.

IMPRESSION

A diagnosis of Albinism X(T) with nystagmus with refractive error on both eye was made based on findings of fundus examination, orthoptic evaluation, positive transillumination test and physical appearance.

MANAGEMENT

We prescribed glasses for myopic astigmatism. Further we advised to go for tinted or photochromic lens to reduce light sensitivity and also prescribed lubricating for comfort. Patient was asked for follow

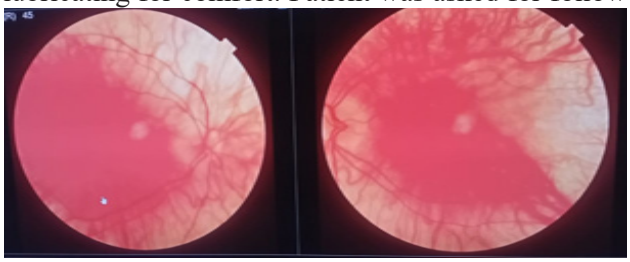

Figure 1. Fundus photo of BE

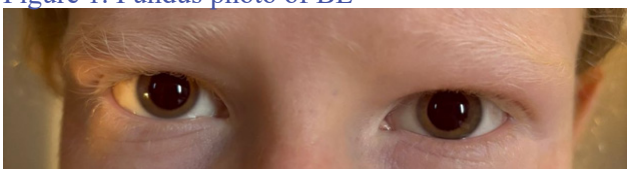

Figure 2. Eyebrows and eyelashes are blonde due to the decrease melanin on body

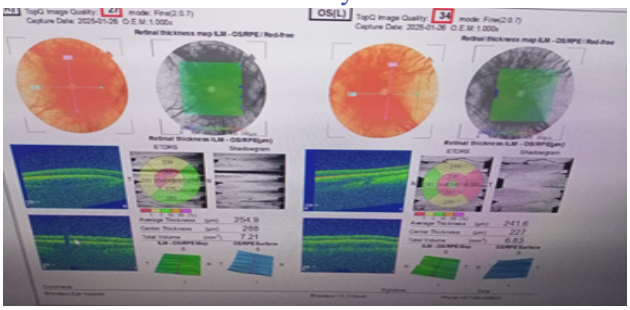

Figure 3. OCT macula of BE

up in every 3 months for evaluation of refractive error and any additional symptoms such as progressive vision loss or worsening nystagmus.

DISCUSSION

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.[5] While specific prevalence data for Nepal was limited. OCA1 has a prevalence of approximately 1 per 40,000[6] in most populations but is very uncommon among African-Americans. In contrast, OCA2 is the most common type of albinism in African Black OCA patients. The overall prevalence of OCA2 is estimated to be 1:36,000 in the USA, but is about 1:10,000 among African Americans.[7] It affects 1 in 3,900 of the population in some parts of the southern part of Africa.[8] OCA3 or Rufous oculocutaneous albinism has been reported to affect 1:8,500 individuals in Africa, whereas it is very rare in Caucasians and Asiatic populations.9 Among disorders where albinism is part of a larger syndrome are Hermansky-Pudlak syndrome (HPS), Chediak-Higashi syndrome (CHS), Griscelli Syndrome, and Waardenburg Syndrome type II (WS2).[10] Lifespan in patients with OCA is not limited, and medical problems are generally not increased compared to those in the general population. As mentioned, skin cancers may occur and regular skin checks should be offered. [11] Development and intelligence are normal. Persons with OCA have normal fertility.[12]

CONCLUSION

Oculocutaneous albinism (OCA) is a group of rare autosomal recessive disorders characterized by reduced or absent melanin production, leading to hypopigmentation of the hair, skin, and eyes and significant impact on visual acuity due to foveal hypoplasia, optic nerve misrouting, and associated complications such as nystagmus, photophobia, and refractive errors.

Conflict of interest: None

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