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ORIGINAL ARTICLE

Visual deficits in Nepalese patients with oculocutaneous albinism

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Abstract
Background: Albinism poses a significant threat to visual functions and causes remarkable ocular morbidity often resulting in visual disabilities. The study aimed at describing the visual status in patients with diagnosed cases of complete oculocutaneous albinism (OCA) attending to a tertiary eye hospital in Nepal.
Methods: This was a cross-sectional descriptive hospital-based study of all diagnosed oculocutaneous albinotic cases (16 males and 9 females; mean age of 16 years) who visited the Department of Ophthalmology at the Institute of Medicine, for ocular consultation between September 1, 2011 and December 1, 2013.
Results: Twenty-five cases (50 eyes) with OCA were enrolled in the study. All the participants had maximally reduced visual acuity (mean: 1.24 ± 0.50 logMAR). Myopic astigmatism was the most common refractive error ($n = 17$; 34%). 58% of all participants had with-the-rule astigmatism.

Considering the spherical equivalent power, most of the eyes ($n = 30$; 60%) had myopia, with overall mean SE refractive error of -1.59 ± 5.39 D. Visual acuity improved significantly with refractive correction in place (paired sample t-test, $p < 0.05$). Horizontal pendular nystagmus was the most common nystagmus ($n = 34$ eyes; 68%). Alternating esotropia and alternating exotropia each were observed in 16% of participants who had strabismus (40% of all cases). The diaphanous iris, foveal hypoplasia and poliosis were the most consistent clinical features.
Conclusion: Patients with OCA present with a broad spectrum of visual deficits that impair the visual functions. Significant improvement in visual acuity following optical correction serves as an impetus to the reduction of visual disabilities in individuals with albinism.

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PALABRAS CLAVE
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Error refractivo

Déficit visual en pacientes nepalíes con albinismo oculocutáneo

Resumen
Antecedentes: El albinismo constituye una amenaza significativa para las funciones visuales, y causa una morbilidad ocular considerable que deriva a menudo en discapacidades visuales. El objetivo del estudio fue la descripción de la condición visual de los pacientes con diagnóstico de albinismo oculocutáneo completo (OCA), en un hospital ocular terciario de Nepal.
Métodos: Este es un estudio transversal y descriptivo llevado a cabo en ámbito hospitalario, que incluyó a todos los casos diagnosticados de albinismo oculocutáneo (16 varones y 9 mujeres; edad media, 16 años) que visitaron el Departamento de Oftalmología del Instituto de Medicina para realizar una revisión ocular entre el 1 de septiembre de 2011 y el 1 de diciembre de 2013.
Resultados: Se incluyó en el estudio a veinticinco casos (50 ojos) con OCA. Todos los participantes tenían agudeza visual máximamente reducida (media: 1,24±0,50 logMAR). El astigmatismo miópico constituyó el error refractivo más común (n=17; 34%). El 58% de los participantes tenía astigmatismo a favor de la regla.

Considerando el equivalente esférico, la mayoría de los ojos (n=30; 60%) tenían miopía, con un valor medio de -1,59±5,39 D. La agudeza visual mejoró considerablemente con la realización de la corrección refractiva (prueba de t de la muestra pareada, p<0,05). El nistagmo pendular horizontal fue el nistagmo más común (n=34 ojos; 68%). La esotropía alternante y la exotropía alternante se observaron cada una en el 16% de los casos con estrabismo (el 40% de los casos). Iris diáfano, hipoplasia foveal y poliosis fueron la características clínicas más consistentes.
Conclusión: Los pacientes con OCA presentan un amplio espectro de déficits visuales que afectan a la función visual. La mejora considerable de la agudeza visual tras la corrección óptica sirve de estímulo para reducir las discapacidades visuales en los individuos con albinismo.

Introduction

Albinism is a genetic disorder involving defective melanin biosynthesis, attributed to an inborn error of metabolism. A defective synthesis of melanin from tyrosine or its absence resulting from the mutation of one or more associated genes causes albinism.[1] The consequence of such mutations is a reduction or absence of melanin in various parts of the body, particularly hair, skin and eyes. Albinism is universal, though unevenly distributed.[2] Its frequency is estimated to be approximately one in 20,000.[2] On account of its autosomal recessive inheritance pattern in most cases, males and females are often equally affected.[3,4]

Varying degrees of hypopigmentation occur due to different mutation types, which give rise to phenotypic heterogeneity. Traditionally the phenotypic classification of albinism hinges on dichotomous foundation – oculocutaneous albinism (OCA) and ocular albinism (OA). Oculocutaneous albinism succinctly implies reduction (incomplete OCA) or absence (complete OCA) of melanin in the eyes, skin and hair, whereas ocular albinism entails the reduction or absence of melanin in the eyes only, with the melanocytic system offering normal pigmentation to the rest of the body.[5]

Oculocutaneous albinism is associated with a number of visual disorders. Abnormal refractive profiles in patients with OCA have been previously established, with high refractive errors, including high with-the rule astigmatism being frequently encountered.[6] There has been a bias towards both hyperopia[2,15,16] and myopia[6,17] in various studies regarding the distribution of refractive errors.[2] Furthermore, OCA has significant anatomical defects, such as large corneal astigmatism, foveal hypoplasia and abnormal decussation of optic nerve fibers. The abnormal decussation is due to reduced or absent melanin, which is thought to determine the neuronal target specificity in the brain. This occurs secondary to misrouting of retinogeniculate projections resulting in strabismus and reduced stereoscopic vision.[3,4,7] Consequently, visual acuity is generally reduced, leading to visual impairment, and cases tend to have severe photophobia as a consequence of ocular hypopigmentation. In addition, nystagmus and color vision impairment tend to occur frequently in patients with OCA.[4]

Although the condition is rare, it is potentially blinding because of its strong association with higher amounts of refractive error, especially corneal astigmatism, intraocular light scatter, light-induced retinal damage, and a partially or un-differentiated fovea.[5] It is highly important that refractive errors and poorer visual acuity presenting from an early age be detected and managed appropriately. In the Asian literature, there is a paucity of data on the vision and refractive status of patients with OCA. Optometric care can help albinotic cases enhance their visual performance by improving their retinal image or training them to use their residual vision in the best possible way. As the first of its kind in Nepal, the study documents the visual deficits present in a clinical population with OCA and compares them with those observed in previous studies abroad.

Methodology

This was a cross-sectional descriptive hospital-based study. Twenty-five consecutive cases (50 eyes) diagnosed with