Print ISSN: 2321-6379
Online ISSN: 2321-595X

Original Article

Ocular Findings in Patients with Oculocutaneous Albinism: A Retrospective Study

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Abstract

Background: Oculocutaneous albinism (OCA) is a group of autosomal recessive disorders characterized by a reduction in melanin production in the skin and eye tissues. It is associated with a variable number of ocular morbidities which include foveal hypoplasia, nystagmus, refractive errors, iris transillumination defects, and strabismus. The purpose of this study was to determine ocular findings and visual status of patients with oculocutaneous albinism.

Materials and Methods: In the present retrospective study, we analyzed 30 case sheets of patients with OCA who had presented to our clinic over the past 2 years. Variables noted were best-corrected visual acuity (BCVA), type of refractive error, presence or absence of foveal hypoplasia, nystagmus, strabismus, iris transillumination defects, or any other ocular morbidity.

Results: Mean age of our study group was 21.66 years (range 5–45 years) with a male:female ratio of 1.3:1. We noted a high percentage of consanguineous marriages (73.33%). All patients had foveal hypoplasia, nystagmus (horizontal), and iris transillumination defects. Eighty percent of the patients had BCVA between 6/24 and 6/60. Astigmatism (with the rule) was the most common refractive error seen in 56.66% of the cases, followed by hypermetropia (33.33%) and myopia in 10% of cases. Strabismus was noted in 33.33% of the cases, the most common type being esotropia (70%).

Conclusion: OCA is associated with varied ocular manifestations, so it is important to screen these patients early in their lives so that appropriate rehabilitative measures are taken early improving the quality of life and better social and economic integration of these individuals. Moreover, given the genetic nature of the disease, consanguineous marriages in families with a history of OCA should only be done after proper genetic counseling.

Key words: Oculocutaneous albinism, Foveal hypoplasia, Refractive errors

INTRODUCTION

The term albinism is derived from the latin word "albus " meaning white.[1] Oculocutaneous albinism (OCA) is a group of autosomal recessive disorder characterized by a reduction in the production of melanin pigment in the structures of skin and eyes [Figure 1].[2] The deficiency of tyrosinase enzyme is responsible for the variable production of melanin in these patients. OCA is associated with variable number of ocular morbidities which include foveal hypoplasia, fundus hypopigmentation [Figure 2], iris transillumination defects, misrouting of the optic nerve fibers at the chiasm, nystagmus, reduced visual acuity, strabismus, photophobia, and refractive errors.[3,4]

The purpose of this study was to determine ocular findings and visual status of patients with OCA. Despite recent advances in medical care, there is no cure for OCA, hence making it even more important to detect various ocular abnormalities, especially refractive errors so that they can be treated at the appropriate time. Early interventions in this group can prevent visual disability later on in their life.

Access this article online

www.ijss-sn.com

Month of Submission : 09-2020
Month of Peer Review : 09-2020
Month of Acceptance : 10-2020
Month of Publishing : 11-2020

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MATERIALS AND METHODS

The present retrospective study was done at Vision Center (A complete Eye clinic) at Lal Bazar Srinagar, which is a registered eye clinic with the directorate of health services Srinagar J&K. We analyzed the clinical case sheets of patients with OCA who had presented to the clinic over the past 2 years. The diagnosis of OCA was based on clinical findings of hypopigmentation of skin and hair in addition to characteristic ocular finding.[5] Variables noted were visual acuity, type of refractive error, presence or absence of foveal hypoplasia, nystagmus, strabismus, and iris transillumination defects. The presence of any other ocular anomaly was also noted. Consanguinity of marriages was also noted in our study as OCA is a heritable disorder. Patients of <5 years age were excluded as it is difficult to obtain measurements of various variables from them.

The study adhered to the tenets of the Declaration of Helsinki and consents were obtained from patients/ parents. Data were analyzed using the Statistical Package for the Social Sciences (SPSS). A $P < 0.5$ was considered statistically significant.

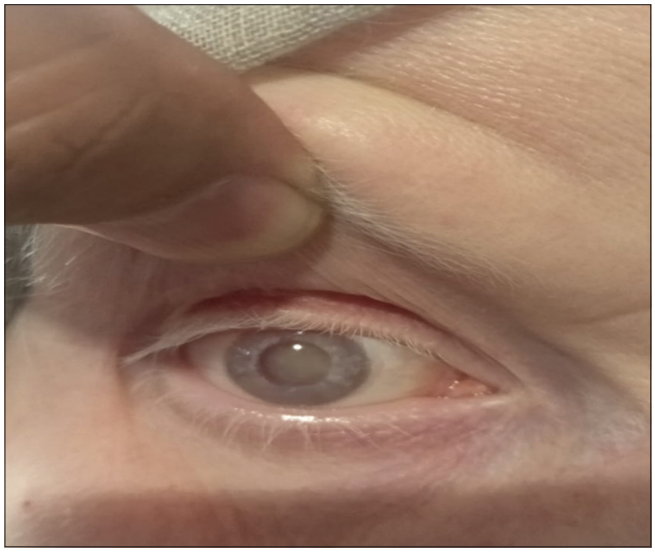

Figure 1: A case of OCA with mature cataract

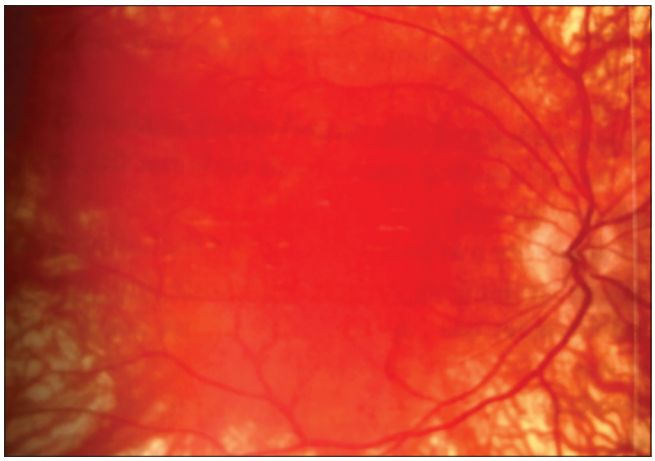

Figure 2: Fundus hypopigmentation and foveal hypoplasia

RESULTS

Data comprising 30 patients (60 eyes) were analyzed in our study. The mean age of our study group was 21.66 years with a range of 5 to 45 years. Out of the total 30 patients, 17 were male and 13 were female with a male:female ratio of 1.3:1. We recorded a high percentage (73.33%) of consanguineous marriages in our patients. All of our patients had typical characteristics of OCA which includes foveal hypoplasia, fundus hypopigmentation, iris transillumination defects, and nystagmus.

The visual status of the patients is depicted in Figure 3. Eighty percent of the patients had vision between 6/24 to 6/60. The distribution of refractive errors is shown in Figure 4. The most common refractive error in our study was astigmatism 56.66% (with the rule), followed by hypermetropia (33.33%) and myopia (10%).

Three of our patients had cataracts (one nuclear cataract, one mature cataract, and one had posterior subcapsular).

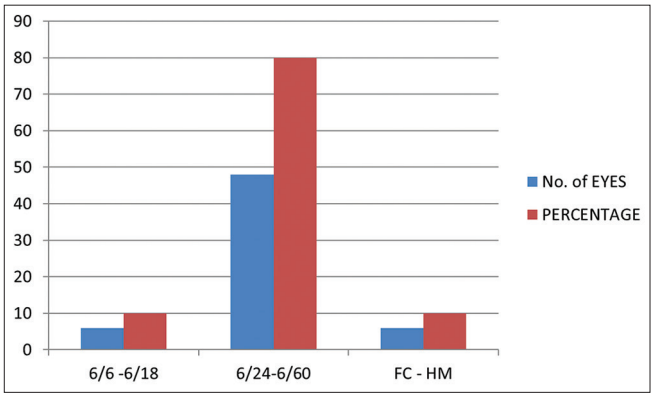

Figure 3: Visual status of patients with OCA

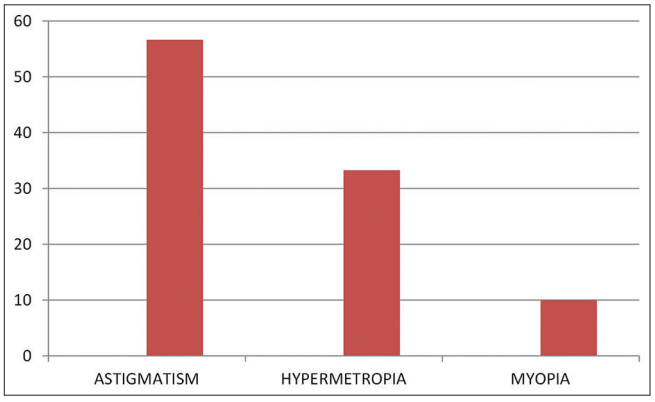

Figure 4: Astigmatism (56.66%) to be the most common refractive error

All of them underwent phacoemulsification with good post-operative results. One patient had keratoconus.

DISCUSSION

A total of 30 patients (60 eyes) of OCA were analyzed in our study, we found that the mean age of our study subjects was 21.66 years which is more than reported by some other studies.[6,7] This late presentation can be attributed to a lack of awareness of the disease and its associated ocular morbidities. The male:female ratio in our study was 1.3:1, which is comparable to other studies.[8]

One of the striking observations of our study was the high rate of consanguinity. Out of the total 30 patients, 22 patients had a history of consanguineous marriage (73.33%). This is close to the study done by Mohammed *et al.*,[9] in which 66.37% of patients had a history of consanguineous marriages. Another study done by Gamella *et al.*[10] also reported role of consanguineous marriages in OCA. All types of OCA are inherited as autosomal recessive disorders.[3,11] Thus, the presence of various types of carriers within different generations of a family is the cause of high prevalence of OCA in them. Consanguineous marriages markedly increase the chance of children developing OCA among parents who are themselves asymptomatic carriers, hence the need for carrier detection and genetic counseling in such families.

In our study, we found that 80% (48 eyes) had best-corrected visual acuity between 6/24 and 6/60 of Snellens chart [Figure 3]. The cause of this low vision was foveal hypoplasia, nystagmus, iris transillumination defects, and refractive errors. These findings were present in all of our patients. Foveal hypoplasia is one of the most significant vision limiting factors found in albinism.[1] The cause of foveal hypoplasia in OCA is thought to be related to decreased amounts of melanin in retinal pigment epithelial cells.[12] All of our subjects had horizontal nystagmus, the cause of which is foveal hypoplasia and misrouting of the optic nerve fibers.[4] Horizontal nystagmus has been reported by other studies[13] to be the predominant type of nystagmus in patients with OCA. Iris transillumination defects were seen in all subjects, cause of which is reduced melanin production in the iris as a result of which light reflected from retina is not filtered by iris due to which the iris appears pink in pigment deficient areas.[5] Iris transillumination defects are also the cause of photophobia which is the most common symptom of OCA[5,14] and is also the cause of disability glare resulting in decreased visual acuity. Strabismus was present in 33.33% of the cases, the most common type being esotropia accounting for 70% of the cases. The high prevalence of strabismus in OCA cases has been supported by other studies.[5,15] Misrouting of the optic nerve fibers due to incomplete pigmentation is one of the causes, the abnormalities of decussation result in a monocular representation of the visual field in each occipital cortex.[3] Abnormal visual cortex development has also been reported by some studies as a cause of strabismus in these cases.[16]

Refractive errors are common in OCA. We found astigmatism (with the rule) to be the most common refractive error in 56.66% of the cases. This is in agreement with various other studies.[4,6,8,14,17-19] Hypermetropia was present in 33.33% of the cases and myopia in 10%. Hence, it is very important that these refractive errors are detected and treated early in their life so that visual morbidities like amblyopia are prevented. The mean age of presentation in our study was 21.66 years, hence the need for awareness among people with OCA to get screened for any kind of refractive error early in their life. This will not only improve quality of life of these patients but also help in better social and economic integration in to the society.

CONCLUSION

OCA is a condition that is associated with various ocular manifestations. Given the fact there is no cure for the disease at present, it is imperative to screen these patients for refractive errors early in their lives. Moreover, given the genetic nature of the disease, consanguineous marriages in families with OCA should be avoided or done only after proper genetic counseling. Further, an effort should be made to increase the awareness of the disease which will help in better management of the ocular morbidities associated with OCA.

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How to cite this article: Rashid W, Lone IA, Shaheen N, Zargar S. Ocular Findings in Patients with Oculocutaneous Albinism: A Retrospective Study. Int J Sci Stud 2020;8(9):93-96.

Source of Support: Nil, **Conflicts of Interest:** None declared.