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CASE REPORT

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Shining Light on Albinism: A Family Case Series

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Abstract

Albinism is a common hereditary condition characterized by the absence or reduction of melanin production in melanocytes, resulting in the distinctive white appearance of the skin, hair, and eyes. The complex process of melanin production involves the tyrosinase enzyme, copper, tyrosine (phenylalanine), and is under genetic control. Ocular albinism, primarily affecting the eyes, is X-linked and presents with varying amounts of melanin in the hair and skin. However, some individuals exhibit a complete absence of melanin in the skin, eyes, and hair, which is transmitted as an autosomal recessive trait. The incidence of albinism varies across populations, with a higher prevalence observed in regions with darker skin tones, such as Africa, where the incidence is approximately 1 in 200 individuals. In contrast, in regions with predominantly light-colored skin, like Europe, albinism is less prevalent, affecting approximately 1 in 2000 individuals. India, with its diverse population, presents a unique context, making individuals with albinism easily noticeable. Exposure to sunlight can lead to sunburn and photophobia, further limiting their daily activities and livelihoods. In this case series, we present five cases of albinism within a single family, highlighting the challenges they face in their daily lives, including the avoidance of outdoor activities due to sun sensitivity. The psychological schemas of experiencing feelings of shyness, embarrassment, and social withdrawal due to the heightened visibility of their condition were discussed in focused group discussion. This case series underscores the importance of understanding and addressing the unique social and healthcare needs of individuals and families affected by albinism in diverse global contexts.

Keywords: Albinism; Melanocyte; Melanin; Tyrosinase; OA (ocular albinism); OCA (Oculocutaneous Albinism)

History of albinism in epics and in nature

In the epics, Lord Indra (King of all gods), had a divine elephant, which was main vehicle of Indra, moving from one place to another place. The name of this divine elephant is "Airavata", and is snow white by birth [1,2].

Uchchaihshravas is a divine flying horse. Suras and Auras were churning the milk of ocean the time when Uchchaihshravas rose out of ocean along with other divine treasures like goddess Lakshmi. Uchchaihshravas is snow white in color [3].

Animals and birds are colorful naturally and appear attractive to anyone who sees and make people enjoy with their wings, mane, etc. Absence of melanin pigment result in white skin, white scales, white hairs and pink eyes (pink color is due to transparency). These animals will have vision abnormality. Without Melanin development of iris, retina, eye muscles, optic nerve is disturbed and hence they cannot focus eyes properly (suffer from Nystagmus & photophobia), and skin cancers. Examples of albino animals in nature are 1) albino rats and mice, seen in most of the laboratories, 2) white peacock, 3) albino frog, 4) white koala bear, 5) white squirrel, 6) arctic fox, 7) albino rabbits, 8) white lions, tigers, bears and elephants, 9) white snakes and 10) white monkeys and chimpanzees [4]

Clinical history

30 year female patient presented to OPD with the complaints of the intolerance to sunlight. She had white skin, white hairs, and light colored eyes. She told four more relatives in the family are having similar type of skin, and they are born with the same color of skin. All five of them are scared to work in sunlight. Because of poverty she has to go to field and work in sunlight. After working for some time in sunlight, she will have discomfort in the skin, she start scratching the skin, developing excoriations and ulcers. Repeated scratching of the forearm skin resulted in thickening of the skin and skin markings are enhanced (lichenification). Over the right cheek, slowly growing papule since eight months has attained the size of 3X3 cm nodule. This nodule move over the underlying tissue. Smaller two papules of 0.5X0.5 cm present on right cheek, near big nodule. Nodule of 2X2 cm present on left flank. Oral hygiene is poor.

Hairs

Hairs all over the body are completely depigmented. Scalp hairs, eyebrows, eyelashes, hairs in the nose, axilla, pubic region, and on both extremities hairs are depigmented.

Eyes

Patient complaints of diminution of vision in both eyes, patient feels discomfort to walk in sunlight. Examination of eyes with torch light, show red glow (red reflex). Slit lamp examination confirms the red glow. On examination the patient had pendular horizontal nystagmus and uncorrected visual acuity of 6/60 in both eyes. Anterior segment evaluation showed Iris depigmentation in and normal 3 mm pupils reacting to direct and consensual light and bright red fundal glow. On indirect ophthalmoscopy, both eyes showed severe fundus hypopigmentation, prominent large choroidal vessels and foveal hypoplasia (Courtesy: Dr Sangeeta T, Associate Professor, Ophthalmology department, SDUMC, Tamaka, Kolar).

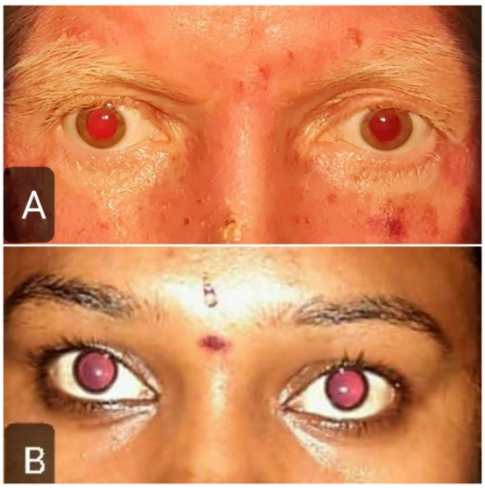

Fig 1. A- Bright red reflex from this OCA patient with red fundal glow and partial iris translucency. B- Normal red reflex in a normal person. Both eyes show normal light pink colour and no iris translucency. Courtesy: Dr Sangeeta T, Associate Professor, Ophthalmology department, SDUMC, Tamaka, Kolar.

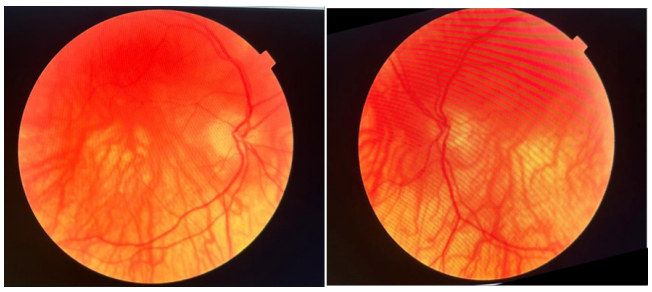

Fig 2. Severe fundus hypopigmentation and foveal aplasia of both eyes. Courtesy: Dr. Sangeeta T, Associate Professor, Ophthalmology department, SDUMC, Tamaka, Kolar.

Smaller 2 lesions present above the 1st swelling, below the right lateral angle of the eye, size 1X1 cm.

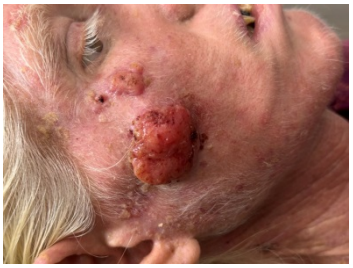

Fig 3. 3 X 3 cm nodulo-ulcero proliferative growth present over the right cheek, with everted edges, it is painless, and moves over the underlying structures and does not bleed on touch.

Hairs of the scalp, eyebrows and eyelashes are white.

Courtesy: Dr Ravikiran HR, Associate Professor, Surgery Department, SDUMC, Kolar.

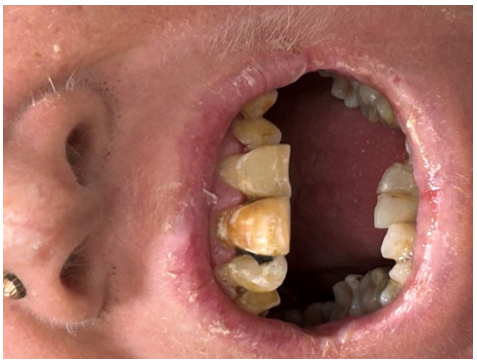

Fig 4. Hairs in the nostrils, over the alae of the nose, upper lips are white.

Oral hygiene is poor.

Investigations

Majority of the routine investigations proved negative, except Vitamin B12 and Vitamin D which were 212 pg/ml and 37.7 mg/ml respectively. As vitamin B12 is involved with the one carbon metabolism and methionine metabolism, methionine or its metabolites viz homocysteine may be directly or indirectly involved with our observations.

The observed lower Vitamin D may be due to the hypopigmentation leading to decreased synthesis from the sunlight. However the deficiency manifestations of vitamin D could not be documented in this case or in their family members

Further the serum and urine fluoride also we estimated as this area is endemic for fluorosis, but we could not observe any significant elevation in the case.

Family tree (Informant is patient 6):

1. Grandmother (1) 60 years, having normal skin and hairs, is healthy. They have four female children (3, 5, 6 and 7) (Grandmother has married mother's brother, 1^0 consanguinity).
2. Grandfather (2) died at the age of 50 years (died due respiratory illness 20 years back, and had normal skin and hairs).
3. Eldest daughter (3), 40 years having normal skin and hairs, and is healthy. 2^{nd} daughter (5) had normal skin and hairs, died at the age of 30, due to fever, 5 years back. Both of them marry to their own uncle (4 – mother's brother, 1^0 consanguinity). Eldest daughter is having four children. Two female children and two male children. Two female children 30 years, 16 years and one male child aged 20 year, are suffering from albinism (9, 10, and 12). One male child of 18 years is having normal skin and hairs (11). Second daughter had one son; aged 15 year is having normal skin and hairs (8). Second daughter died of fever, five years back.
4. Third daughter (6) is forty year old, and fourth daughter (7) is suffering from albinism. Fourth daughter is the present patient.

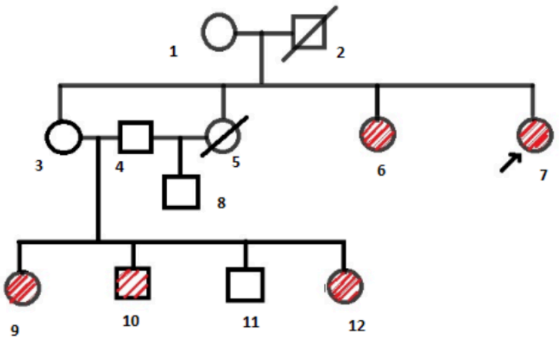

Fig 5. Photo courtesy: Dr. Karthik S, Assistant Professor, Department of Paediatrics, SDUMC, Kolar.

Discussion

OCA has to be treated by explaining the genetic involvement in pathogenesis of the disease. Counselling has to be done to the patient and also to all the relatives of the patient. Patient has to protect herself from sunlight and bright artificial light also. Patient has to wear the clothes covering entire body, broad brimmed hat, goggle, hand gloves while going out of house, and shoes to protect from sun exposure. She has to modify her working patterns, so that, she can work in early morning hours and evening hours and to avoid afternoon hours. There is no curative medicine. Nitisinone has shown increase in tyrosine levels and improved pigmentation in few patients[5].

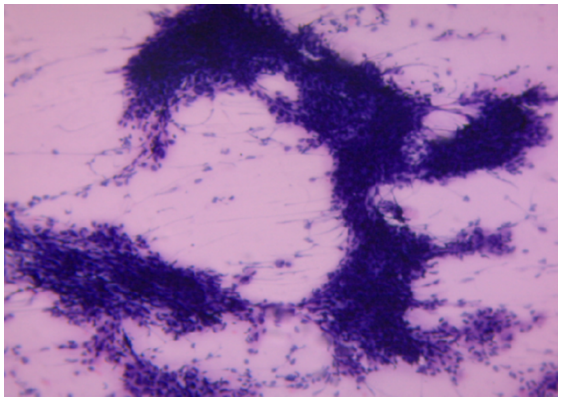

Fig 6. Microphotograph of FNAC smear shows cellular aspirate of cells arranged in clusters with peripheral palisading pattern (H and E, X100), Curtesy: Pathology department, SDUMC, Kolar.

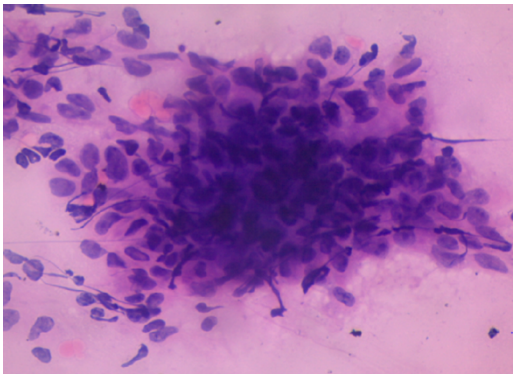

Fig 7. Microphotograph of FNAC smear shows cluster of tumour cells arranged in loose cohesive clusters, showing peripheral palisading pattern. (H and E,X 400X Curtesy: Pathology department, SDUMC, Kolar

Our female patient is having white skin, white hairs, and white eyes since birth. Patient is 40 years old as on today, and minimal pigmentation has not taken in skin, hairs and eyes. Hence she can be grouped under oculocutaneous albinism 1A (OCA 1A). There is total loss of melanin pigment in skin, hairs, and eyes; hence it can be grouped under OCA 1 A. She has also developed skin malignancy. She is having eye symptoms, still she manages to work. Tyrosinase is a copper containing enzyme present in plants and animal tissue, help in production of melanin and other pigments from tyrosine by oxidation. TYR gene encodes the enzyme[6]. Tyrosinase is a single membrane spanning transmembrane protein[7]. Tyrosinase is present in melanosomes of melanocytes, and retinal pigment epithelium and help in synthesis of both eumelanin and pheomelanin[8]. Absence of melanin pigment in this patient resulted in skin malignancy, and absence melanin in retinal pigment epithelium has resulted in poor eye development.

Conclusion

The presented case series underscores the complex clinical manifestations and social challenges faced by individuals with albinism, particularly in resource-constrained settings where sun exposure is unavoidable due to economic circumstances. This study not only underscores the importance of comprehensive care and education for individuals and families affected by albinism to mitigate both dermatological and social repercussions but also serves as a poignant reminder of the multifaceted challenges confronting individuals with albinism and their families, especially in regions with limited resources. It underscores the urgent need for policy-makers and healthcare authorities to consider implementing reforms that include comprehensive genetic counseling initiatives aimed at raising awareness and discouraging consanguineous marriages within affected communities. Such measures hold the potential to mitigate the prevalence of albinism and its associated social and dermatological complications in vulnerable populations.

References

1) Dalal R. Hinduism: An Alphabetical Guide. 2010.
2) Williams GM. Handbook of Hindu Mythology. New York. Oxford University Press. 2008.
3) Wilson HH. The Vishnu Purana. 2006.
4) Creel D. Inappropriate use of albino animals as models in research. *Pharmacology Biochemistry and Behavior*. 1980;12(6):969–977. Available from: https://doi.org/10.1016/0091-3057(80)90461-x.
5) Onojafe IF, Adams DR, Simeonov DR, Zhang J, Chan CCC, Bernardini IM, et al. Nitisinone improves eye and skin pigmentation defects in a mouse model of oculocutaneous albinism. *Journal of Clinical Investigation*. 2011;121(10):3914–3923. Available from: https://doi.org/10.1172/jci59372.
6) Barton DE, Kwon BS, Francke U. Human tyrosinase gene, mapped to chromosome 11 (q14 → q21), defines second region of homology with mouse chromosome 7. *Genomics*. 1988;3(1):17–24. Available from: https://doi.org/10.1016/0888-7543(88)90153-x.
7) Kwon BS, Haq AK, Pomerantz SH, Halaban R. Isolation and sequence of a cDNA clone for human tyrosinase that maps at the mouse c-albino locus. *Proceedings of the National Academy of Sciences*. 1987;84(21):7473–7477. Available from: https://doi.org/10.1073/pnas.84.21.7473.
8) Kondo T, Hearing VJ. Update on the regulation of mammalian melanocyte function and skin pigmentation. *Expert Review of Dermatology*. 2011;6(1):97–108. Available from: https://doi.org/10.1586/edm.10.70.