

Partial Oculocutaneous Albinism: Two Siblings with Features of both Hermansky Pudlak and Waardenburg's Syndrome

Mazhar Ishaq, Muhammad Khizar Niazi, Muhammad Saim Khan and Yasser Nadeem

ABSTRACT

Albinism is an inherited abnormality of melanin synthesis with incidence of one per 20,000 births. Its clinical manifestations are related to the reduction or absence of pigmentation in the visual system and/or the skin and teguments. The clinical spectrum of Oculocutaneous Albinism (OCA) has four types ranging from OCA 1 - 4, of which OCA 1, A-1 is the most severe form. Partial cutaneous albinism which is a subtype of OCA is associated with systemic immunodeficiency disorders like Chediak Higashi (CHS), Griscelli (GS) and Hermansky-Pudlak (HPS) syndromes. A 7 years boy was labeled initially as a case of Hermansky Pudlak syndrome at the age of 01 year. He as well as his 4 years old younger brother when examined in detail along with audiological investigations were diagnosed as a rare presentation of both Hermansky Pudlak and Waardenburg's syndrome.

Key Words: Oculocutaneous albinism. Hermansky Pudlak syndrome. Waardenburg's syndrome.

INTRODUCTION

Oculocutaneous Albinism (OCA) is a group of conditions that affect pigmentation of the skin, hair and eyes. It is divided into various clinical types.¹⁻³ OCA type 1 is divided into partial and complete on the basis of tyrosine kinase activity. Hermansky Pudlak Syndrome (HPS), Waardenburg's Syndrome (WS) and Chediak Higashi syndromes are three rare systemic associations of partial oculocutaneous albinism with distinct clinical presentation.⁴⁻⁶

Two siblings of partial oculocutaneous albinism with features of both HPS and WS were identified.

The aim to report this case is to acquaint health professionals about the combined presentation of two rare systemic associations of OCA.

CASE REPORT

Case 1: A 7 years fair complexion boy presented with history of decreased vision and decreased hearing. There was history of prolonged bleeding after circumcision.

General physical examination revealed bruises over the elbow and shin bones. Ocular examination revealed best corrected visual acuity of 20/120 (6/36) in the right and 20/80 (6/24) in the left eye. Torch examination revealed broad nasal bridge and a scar of operated cleft lip and cleft palate. Ocular movements were of full

range. Slit lamp examination revealed iris transillumination, hypopigmented fundus, choroidal unmasking and loss of foveal reflex (Figure 1). Brainstem Evoked Response Audiometry (BERA) report revealed moderate hearing loss. Optical coherence tomography showed foveal hypoplasia (Figure 2). For confirmation of HPS, investigations like coagulation profile and released nucleotides were carried out. Coagulation profile was normal but there was no ADP release activity and very low ATP release activity.

All these clinical findings and the investigationus confirmed the diagnosis of partial oculocutaneous albinism with features of both HPS and WS. Patient was counselled regarding the nature and prognosis of disease, advised glasses for refractive error and referred to audiologist for hearing aids.

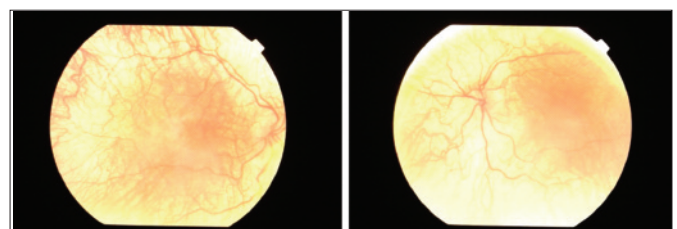


Figure 1: Hypopigmented fundus.

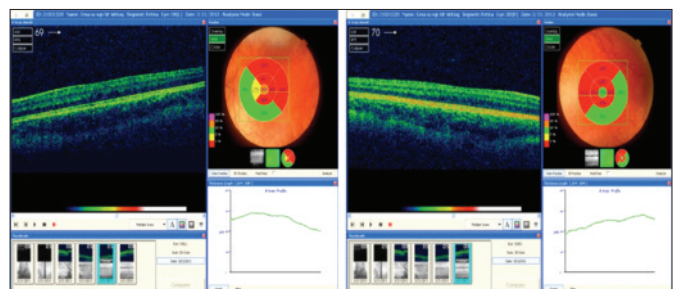


Figure 2: Optical coherence tomography.

Department of Ophthalmology, Armed Forces Institute of Ophthalmology, Rawalpindi.

Correspondence: Dr. Muhammad Saim Khan, Armed Forces Institute of Ophthalmology, Mall Road, Military Hospital, Rawalpindi.

E-mail: saim_amc@hotmail.com

Received: October 23, 2013; Accepted: August 27, 2014.

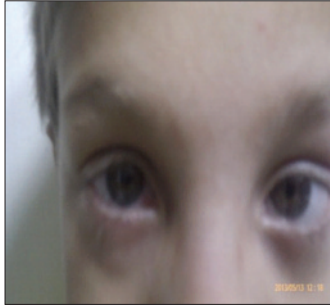


Figure 3: Left esotropia.

Case 2: A 5 years boy (brother of case 1), presented with same fair complexion, and history of decreased visual acuity since birth, left esotropia and decreased hearing which was noticed since 1 year of age. General physical examination revealed bruises over the elbow and shin bones.

Ocular examination revealed best corrected visual acuity of 20/100 (6/30) in right eye and 20/120 (6/36) in left eye. Torch examination revealed broad nasal bridge and a scar of operated cleft lip and cleft palate. There was 20° accommodative esotropia in left eye that was relieved with glasses (Figure 3). Ocular movements were of full range. Slit lamp examination revealed iris translumination, hypopigmented fundus, choroidal unmasking and loss of foveal reflex. On investigations, Brainstem Evoked Response Audiometry (BERA) revealed moderate hearing loss. Optical coherence tomography showed foveal hypoplasia.

Based on these findings he, like his elder brother, was diagnosed as case of partial oculocutaneous albinism with features of both HPS and WS. He was advised with glasses for refractive error and referred to audiologist for hearing aids.

DISCUSSION

Oculocutaneous albinism is a disorder affecting colouring (pigmentation) of the skin, hair, and eyes. Affected individuals typically have very fair skin and white or light-coloured hair. Hermansky-Pudlak Syndrome (HPS) is a multisystem disorder characterized by tyrosinase-positive oculocutaneous albinism; a bleeding diathesis resulting from a platelet storage pool deficiency; and in some cases, pulmonary fibrosis or granulomatous colitis.^{7,8} The albinism is characterized by hypopigmentation of the skin and hair; and ocular findings of reduced iris pigment with iris translumination, reduced retinal pigment, foveal hypoplasia with significant reduction in visual acuity (usually in the range of 20/50 to 20/400), nystagmus, and increased crossing of the optic nerve fibers.^{7,8}

Waardenburg's Syndrome (WS) is an inherited autosomal dominant genetic disorder which is characterized by partial albinism and hearing loss. Features of Waarden-

burg's syndrome include cleft lip, deafness, heterochromia, pale colour skin, hair and eyes (partial albinism), difficulty in completely straightening the joints, possible slight decrease in intellectual function, white patch of hair or early graying of the hair.⁹

Chediak-Higashi syndrome is characterized by varying degrees of oculocutaneous albinism, recurrent infections, bleeding disorders and variable neurological involvement. Bone marrow transplantation is done to correct the immunologic and hematologic defects. If not treated patients usually die of bacterial infections or develop accelerated phase lymphoproliferation.¹⁰

Both these siblings were product of a consanguineous marriage and their sisters were free of these disorders. They had features of both HPS and WS. Hence their parents were counselled and prescribed sight and hearing aids.

REFERENCES

1. Grankov K, Jacob EK, Brondum K. Oculocutaneous albinism. *Orphanet J Rare Dis* 2007; **2**:43.
2. Inagaki K, Suzuki T, Shimizu H, Ishii N, Umezawa Y, Tada J, et al. Oculocutaneous albinism type 4 is one of the most common types of albinism in Japan. *Am J Hum Genet* 2004; **74**: 466-71.
3. Wildsoet CF, Oswald PJ, Clark S. Albinism: its implications for refractive development. *Invest Ophthalmol Vis Sci* 2000; **41**: 1-7.
4. Dotta L, Parolini S, Prandini A, Tabellini G, Antolini M, Kingsmore SF, et al. Clinical, laboratory and molecular signs of immunodeficiency in patients with partial oculocutaneous albinism. *Orphanet J Rare Dis* 2013; **8**:168.
5. Kanski JJ, Bowling B, editors. Clinical ophthalmology. 7th ed. Edinburg: *Isevier Saunders*; 2011.
6. Pour-Jafari H, Zamanian A, Pour-Jafari B. Genetic analysis of oculocutaneous albinism type 1A in an Iranian family. *Iran J Public Health* 2010; **39**:100-4.
7. Rivera C, Golas G, Hess R, Nicholas D. Clinical, molecular and cellular features of non Puerto Rican hermansky - pudlak syndrome patients of Hispanic descent. *J Invest Dermatol* 2011; **131**:2394-400.
8. Schneier AJ, Fulton AB. The hermansky-pudlak syndrome: clinical features and imperatives from an ophthalmic perspective. *Semin Ophthalmol* 2013; **28**:387-91.
9. Kumar S, Rao K. Waardenburg's syndrome: a rare genetic disorder: a report of two cases. *Indian J Hum Genet* 2012; **18**: 254-5.
10. Fantinato GT, Cestari Sda C, Afonso JP, Sousa LS, Enokihara MM. Do you know this syndrome ? Chediak Higashi syndrome. *An Bras Dermatol* 2011; **86**:1029.

