

## **Atypical Variant of Oculocutaneous Albinism Associated with Deafness and Mental Retardation: A Case Report**

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**Abstract:** 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 oculocutaneous albinism varies considerably worldwide and has been estimated to be about 1/17,000. Diagnosis is based on clinical findings of hypopigmentation of the skin and hair, in addition to the characteristic ocular symptoms. Usually affected patients have normal lifespan, neurological development, intelligence and fertility. Treatment is usually by correction of refractive errors with spectacles; strabismus surgery can be considered. Hats with brims and dark glasses. Protection from sun exposure with appropriate skin-covering clothing and sunscreens prevent the risk of skin cancer. . Differential diagnosis includes Mental Retardation, Chediak-Higashi syndrome. Full Gene Sequencing panel of tests to identify the genes involved can be done to identify the mutation. Disadvantage is its cost. Diagnosis is by clinical evaluation. Correction of refractive errors, protection from sun exposure, educating the parents about the care of the patient, will have better prognosis.

**Keywords:** Oculocutaneous albinism, Atypical Variant, Mental Retardation, Mental Retardation, Chediak-Higashi syndrome

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### **INTRODUCTION**

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 oculocutaneous albinism varies considerably worldwide and has been estimated to be about 1/17,000. Basically 4 types of OCA are seen. Type 1A being the most severe type with a complete lack of melanin production throughout life, while the milder forms Type 1B, Type 2, Type 3 and Type 4 show some pigment accumulation over time. Clinical features include various degrees of congenital nystagmus, iris hypopigmentation and translucency, reduced pigmentation of the retinal pigment epithelium foveal hypoplasia, reduced visual acuity and refractive errors, colour vision impairment and prominent photophobia. Diagnosis is based on clinical findings of hypopigmentation of the skin and hair, in addition to the characteristic ocular symptoms. Usually affected patients have normal lifespan, neurological development, intelligence and fertility. Treatment is usually by correction of refractive errors with spectacles; strabismus surgery can be considered. Hats with brims and dark glasses. Protection from sun exposure with appropriate skin-covering clothing and sunscreens prevent the risk of skin cancer. Differential

diagnosis includes Hermansky-Pudlak syndrome, Chediak-Higashi syndrome.

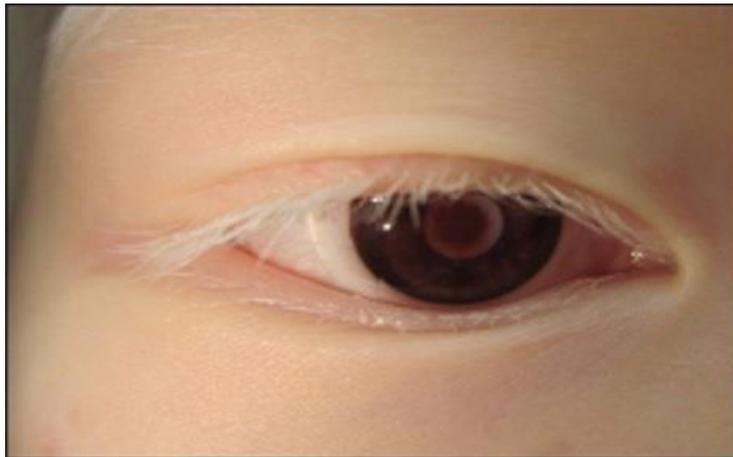
### **CASE HISTORY**

#### **History**

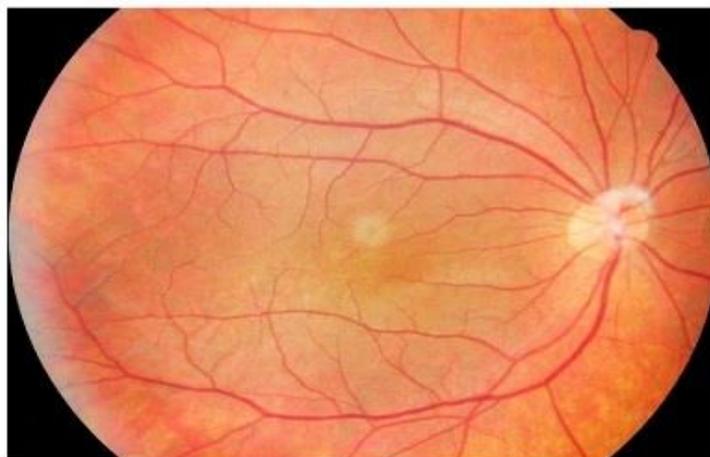
A 25 year old female patient came with a history of generalised weakness since 1 month and abdominal distension and pedal edema since 1 month. She had generalised hypopigmentation of skin, hair and iris since birth. Patient is also mentally retarded and is unable to listen and talk. She also had photophobia and significant diminished vision.

#### **Physical Examination**

On examination, she had generalised hypopigmentation with severe pallor of palpebral conjunctiva, palmar creases and nails. She also had bilateral pedal edema and koilonychia. Her eyelashes, eyebrows and scalp hair are hypopigmented. Abdominal examination revealed moderate splenomegaly. On ocular examination she had hypopigmented iris with iris transillumination positive. Fundoscopy revealed hypopigmented retina with foveal hypoplasia. Her visual acuity was significantly decreased. She had periodic alternating nystagmus which was less marked during rest and relaxed state.



**Fig-1: Depigmented eyelashes and eyebrows with hypopigmented iris**



**Fig-2: Fundus examination showing pale background**

### Laboratory Evaluation and Imaging

Complete blood count showed hemoglobin of 6.3gm% and total count 10400cells/cumm with platelet count of 1.2lakhs/cumm and RDW was 14.0, PDW and MPV was normal. ESR was 50mm at the end of 1<sup>st</sup> hour. Urine routine was normal. Peripheral smear showed microcytic hypochromic blood picture with normal platelets. Her liver function tests and renal function tests were normal. Stool occult blood was negative. Bone marrow showed erythroid hyperplasia.

### DISCUSSION

OCA Type 1 is caused by mutations in the tyrosinase gene on chromosome 11. Mutations in the OCA Type 2 causes the OCA Type 2 phenotype. OCA Type 3 is caused by mutations in tyrosinase-related protein (TRP-1). Mutations in the membrane-associated transporter protein gene (MATP) causes OCA Type 4. All four types of OCA are inherited as autosomal recessive disorders. The parents of the affected child are obligate carriers. Offspring of an affected person are obligate carriers. Carriers are asymptomatic. In most cases, there is no previous family history of albinism but the condition does occur in individual of two generations of family, so called pseudodominance, and is

due to an affected person having a children with a person who is a carrier [1]. Among the disorders where albinism is a part of a bigger syndrome are Hermansky-Pudlak Syndrome (HPS), Chediak-Higashi Syndrome(CHS), Griscelli Syndrome and Waardenburg Syndrome type 2(Ws2). The Hermansky-Pudlak Syndrome is characterized by hypopigmentation and accumulation of a material called ceroid in tissues throughout the body [2]. Further, patients exhibit severe immunologic deficiency with neutropenia and lack of killer cells[3]. HPS is very rare, except in Puerto Rico where it affects approximately 1 in 1,800 individuals [4]. The most important medical problems in HPS are related to interstitial lung fibrosis, granulomatous colitis and mild bleeding problems due to a deficiency of granules in platelets [5]. The Chediak-Higashi Syndrome is a rare condition that includes an increased susceptibility to bacterial infections, hypopigmentation, prolonged bleeding time, easy bruisability and peripheral neuropathy. The skin, hair and eye pigmentation is reduced or diluted in CHS [6, 7]. The Griscelli syndrome is a rare disorder with immune impairment an neurological deficit and hypopigmentation of skin and hair and presence of large clumps of pigment in hair shafts [8]. A syndrome of

sensory deafness and partial albinism is referred to as the albinism-deafness syndrome or the Waardenburg Syndrome[9]. Angelman Syndrome(AS) is a neurodevelopmental disorder characterized by mental retardation, speech impairment, ataxia and happy disposition with frequent smiling. Patients are often seen with hypopigmentation of skin, eye and hair correlating with deletion of P gene localized in the distal part of Prader-Willi (PWS)/ AS region [10,11].

#### ACKNOWLEDGEMENT

We are thankful to the Department of Ophthalmology and Dermatology, BLDE University's Shri B. M. Patil Medical College, Vijayapur for their contribution to this work.

#### KEY MESSAGES

Full Gene Sequencing panel of tests to identify the genes involved can be done to identify the mutation. Disadvantage is its cost. Diagnosis is by clinical evaluation. Correction of refractive errors, protection from sun exposure, educating the parents about the care of the patient, will have better prognosis.

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