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Autism spectrum disorder in a child with Oculocutaneous Albinism

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ABSTRACT

Oculocutaneous albinism (OCA) is a collection of autosomal recessive conditions of melanin biosynthesis that rarely presents with multiple systemic signs. Autistic spectrum disorder (ASD) is characterized by impaired communication, social interaction, and repetitive behaviours. ASD has been reported in association with several inherited medical and psychological disorders. The association of ASD and OCA is rarely reported in the literature. Herein, we report the joint occurrence of these conditions in a three year old boy. This rare association might offer pointers about the genetic relationship between ASD and OCA.

Key Words:

Autism spectrum disorder, Oculocutaneous albinism, melanin synthesis, hypopigmentation, nystagmus, genetic testing.

Introduction:

Oculocutaneous albinism (OCA) is a group of rare inherited disorders characterized by a reduction or complete lack of melanin pigment in the skin, hair and eyes.[1]Autism spectrum disorder (ASD) is a neurodevelopmental disorder where the child has social communication difficulties and repetitive behaviours.[2] There are very few case reports of OCA and ASD presenting as co morbidities. We present a 3 year old child diagnosed with albinism who presented with symptoms of ASD.

Case Report:

History

A 3-year-old boy was referred to Sethu Centre for Child Development and Family Guidance, Goa with complaints of decreased response to name call and poor eye contact. Parents shared that their son never initiated conversations or shared his interests. He would communicate by leading them by their hand, screaming or occasionally using single meaningful words. He did not use gestures to communicate. The child spent a lot of time playing alone, being in a world of his own and ignoring his peers.

When excited, he would repeat words and flap his hands. He had rigid thinking and would throw tantrums if things were not done according to his liking. He was fascinated with colors, observed objects closely and liked to arrange incense sticks in the shape of alphabets. He also had tactile defensiveness and did not allow haircutting or toothbrushing.

The child was born full term by Caesarean section, cried immediately at birth and had a birth weight of 2.5 kgs. As the child was born with hypopigmented skin, blond hair, and blue eyes he was examined by the pediatrician at the government hospital during a routine well baby visit and was diagnosed as Albinism but was not evaluated further. He had no previous history of hospitalization.

Gross motor and fine motor milestones were

attained age appropriately. He started reciting alphabets by the age of 18 months and could name multiple body parts/fruits and vegetables by the age of 2 years. He can presently use 2-word phrases occasionally (need based). He is dependent on his parents for his eating, toileting, and dressing needs.

The child is first born to a non-consanguineous marriage. There is no family history of language delay, developmental regression, seizures or albinism.

Examination:

The child was active alert with stable vitals during examination. His weight was 13 kgs (0-25th centiles), height was 94 cms (25th-50th centile) and head circumference was 50 cms (50th centile). His hair, eyebrows and eyelashes were blond (Fig 1). Iris was hypopigmented with blue color. Nystagmus was noticed with a fast component towards the right side. His skin was hypopigmented over his entire body (Fig 2). Neurological examination done was normal with no cerebellar or meningeal signs. The systemic examination was also normal.

A developmental assessment revealed that his gross motor and fine motor milestones were age appropriate. His language was limited to occasional 2-word phrases, grunts, and screams. He did not follow any verbal instructions or visual schedules cues during assessment. Name response, joint attention and eye contact were absent. He was engaged in solitary play, was in a world of his own and approached parents only if he needed something. He exhibited hand flapping when he was excited, and frequently used scripted, repetitive phrases like "Oh No". He was interested in playing with the wheels of the toy cars and would squint and look at them closely. An assessment for Autism Spectrum Disorder based on the DSM V criteria was fulfilled. The Indian Scale for Assessment of Autism (ISAA) score was 112 which indicated moderate autism.

Diagnosis And Treatment:

Based on history and physical examination and developmental assessments the child was diagnosed to have Oculocutaneous albinism and Autism spectrum disorder. The parents were counselled about both the diagnosis. They were recommended genetic testing for OCA along with regular ophthalmology reviews and skin care protocols. Early intervention behavioral and communication therapy were advised for ASD.

Discussion:

Though hypomelanotic skin disorders like tuberous sclerosis and hypomelanosis of Ito have been reported in association with childhood autism[3,4], an association between oculocutaneous albinism with autism has been reported rarely. Rogawski et al in 1978 first reported such an association in two boys.[5] Four families of individuals who had childhood autism and the additional feature of oculocutaneous albinism in addition to major affective disorder were described by Delong.[6] A 13 year-old Nigerian boy who had oculocutaneous albinism and autism was described by Bakare and Ikegwuonu in 2008.[7] In the Indian scenario, Sandhya and Aravinda HR described a four year old female child, born of a consanguineous marriage who presented with delayed speech and social skills and clinical features of OCA.[8] Genetic evaluation was normal but visual evoked potentials showed decreased visual acuity. She was diagnosed with autism along with OCA type 1. Raj G et al described a three year old female child with low weight, neonatal ICU stay at birth, delay in fine motor, social cognition and language, and hyperactivity, along with inappropriate behaviours, poor eye contact and solitary play.[9] Ophthalmology consultation showed foveal hypoplasia and she was diagnosed to have oculocutaneous albinism. A thorough literature search has revealed that this report is the first of its kind for a male child and the third report of combined OCA and ASD from India.

Conclusion:

A variety of genetic mechanisms like single gene disorders, copy number variations and polygenic mechanisms may be involved in the etiology of autism.[10] Genetic variation in the GABA(A) receptor alpha 5 subunit gene and GPR143 gene have been found to occur mainly in Ocular and oculocutaneous albinism and to a smaller extent to the development of autism like features [6,11]. Vitamin-D level which is involvedwith melanin production and is also being studied as a risk factorfor autism may further explain the association between autism and hypomelanotic skin disorders [12]. Further research into the role of a common etiology between the two could help with early detection and management of both childhood autism and OCA.

Consent:

A written consent from the parents was obtained for the publication of this case report.

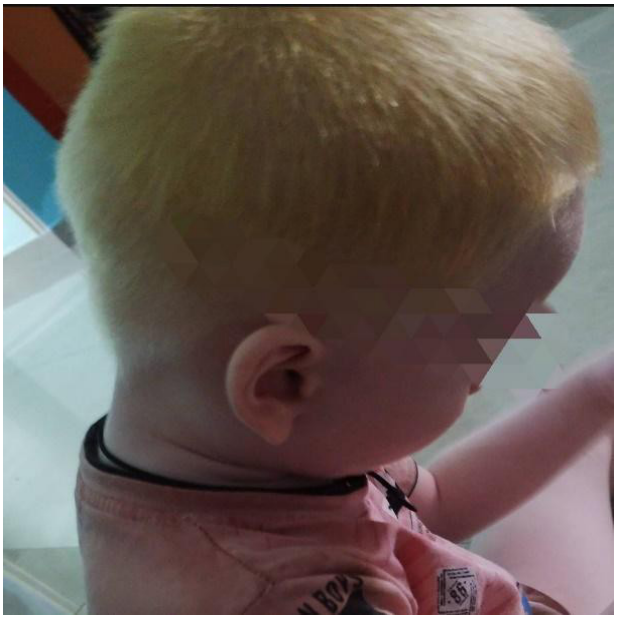

Figure 1

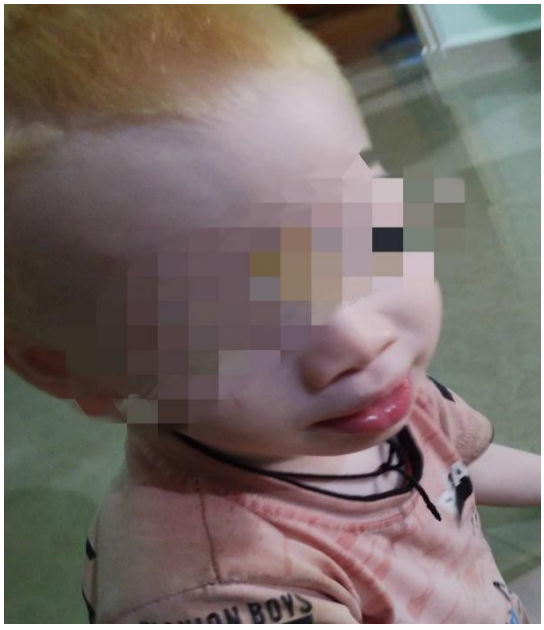

Figure 2

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