OCULOCUTANEOUS ALBINISM WITH CHILDHOOD AUTISM - A RARE ASSOCIATION
KAYATHRI
Department of Paediatrics, MADRAS MEDICAL COLLEGE AND GOVERNMENT GENERAL HOSPITAL

Abstract:
Hypomelanotic skin disorders like tuberous sclerosis and hypomelanosis of Ito that present with multiple systemic manifestations have been reported in association with childhood autism. We report a case of male child, 4 years old, a case of childhood autism with oculocutaneous albinism, is reported for its rarity.

Keyword: AUTISM, TUBEROUS SCLEROSIS, HYPOMELANOSIS OF ITO, ALBINISM.

INTRODUCTION:
A number of hypomelanotic skin disorders have been reported to occur co-morbidly with autism, and susceptibility genes for several of these hypomelanotic skin disorders have chromosomal loci that lie near the loci for several major susceptibility genes for autism. The hypomelanotic skin disorders that have been reported to occur co-morbidly with autism include oculocutaneous albinism, hypomelanosis of Ito, tuberous sclerosis, Angelman syndrome and Prader-Willi syndrome. Oculocutaneous albinism is infrequently reported in association with childhood autism when compared to tuberous sclerosis and hypomelanosis of Ito. (1)

CASE PRESENTATION:
A male child of 4 years of age, second born of 3rd degree consanguineous parents was born with oculocutaneous albinism and on growing up his parents noticed that he is not making eye contact and not responding to calls whenever called. On detailed history taking he does not take interest in other children and failure to orient to name, failure to use gestures to point, lack of interactive play, failure to smile, lack of sharing and seem totally aloof. He also had impairment in joint attention. He is often withdrawn and spend hours in solitary play. He had repetitive hand and finger movements. He moves to and fro across a room repetitively. He almost always chooses from a restricted range of repetitive activities when he is left to occupy himself. He had behavioral problems in the form of hyperactivity and aggressiveness. His mother had pregnancy induced hypertension during second trimester. Otherwise antenatal period was uneventful. It was a full term normal vaginal delivery, birth weight 2.8 Kg child not cried immediately after birth. Child was admitted in neonatal intensive care unit for about 17 days. He had neonatal seizures on day 3 of life and was treated with intravenous anticonvulsants and had neonatal jaundice which was treated with phototherapy. On discharge he was neurologically normal and not on any anticonvulsant. His gross motor development was said to be normal.

Developmental impairments were restricted to the areas of communication cognition and social interaction. He needs assistance for day to day activities. His elder sibling is seven years old female child studying second standard in regular school. There is no family history of albinism or similar illness in other family members.

Figure 1 & 2
Showing patient’s skin and hair color.
On general examination, he had features of oculocutaneous albinism. His height, weight and head circumference were within normal limits. His mental state evaluation revealed he did not make eye contact, not responding to any questions. He is not interested in surroundings and repetitively doing hand finger movements. His hearing and vision assessment were normal. There was no focal neurological deficit. Other system examinations were normal. Autistic disorder in the child is diagnosed by clinical examination.
Intelligent quotient test was not carried out on this patient because of qualitative impairment in social interaction and communication. Modified check list for autism in toddlers M-CHAT (2) diagnostic tool used and he failed in six critical items. A child fails the M-CHAT when two or more critical items are failed or when any three items are failed. He was referred to child guidance clinic for further evaluation and management. He was diagnosed as a case of autistic disorder by using diagnostic and statistical manual of mental disorders both edition. (3) Parents were counselled regarding the condition and the need for long term therapy and the diagnosis parents were counselled to avoid ultraviolet radiation by wearing protective long sleeved clothing and by using sunscreens. Child was started on risperidone for behavioral problem. Parents were counselled regarding speech therapy and occupational therapy and special schooling to maximize the child’s ultimate functional independence and quality of life. Follow up was planned in general medical, ophtalmology, ENT, dermatology and child guidance clinic OPD. The child is being in the regular follow up. This case of childhood autism is reported for its rare association with oculocutaneous albinism.

Discussion: The core features of autistic disorder (AD) include impairments in 3 symptom domains: social interaction; communication; and developmentally appropriate behavior, interest, or activities. Stereotypical body movements, a marked need for sameness, and a very narrow range of interests are also common. Aberrant development of social skills and impaired ability to engage in reciprocal social interactions are hallmark symptoms of AD. A current estimate of prevalence rate of AD is 20.6/10000 (3).

Hypomelanotic skin disorders and autism: A number of hypomelanotic skin disorders have been reported to occur co-morbidly with autism, and susceptibility genes for several of these hypomelanotic skin disorders have chromosomal loci that lie near the loci for several major susceptibility genes for autism. The hypomelanotic skin disorders that have been reported to occur co-morbidly with autism include oculocutaneous albinism, hypomelanosis of Ito, tuberous sclerosis, Angelman syndrome and Prader-Willi syndrome. Inconsistent findings on chromosomal abnormalities in hypomelanosis of Ito, together with its polymorphic nature, suggest that the disorder is not a single genetic syndrome, but rather a non-specific manifestation of hypopigmentation that is associated with a number of genetically heterogeneous disorders that often present with autistic features. An earlier report suggested that further embryo-genetic studies into the possible relationship between autism and associated hypomelanotic skin disorders may provide clues to the etiology of autism. It is important to note that these hypomelanotic skin disorders occur in about ten percent of individuals with autism and that autism in turn occurs in varied percentages, ranging between less than one percent and up to 60 percent, among individuals suffering from these hypomelanotic skin disorders. These variable rates of co-morbidity with hypomelanotic disorders in autistic patients may reflect the action of epigenetic factors that affect the expression of genes for these disorders.(4)

Oculocutaneous albinism and childhood autism: In Oculocutaneous albinism, lack of pigment is generalized affecting skin, hair, eyes. It is inherited in an autosomal recessive trait. It is rarely reported in association with childhood autism when compared to tuberous sclerosis and hypomelanosis of ito.

Rogawski et al had reported 2 tyrosinase-positive albino boys, 1 Caucasian and 1 American Negro, manifested moderate retardation and autistic behaviour. It is postulated that the as yet unidentified metabolic defect in albinism may be responsible for the CNS disorder in these 2 boys.

(5) Muideen O Bakare et al reported a case of childhood autism in a 13 year old boy with Oculocutaneous albinism. Based on the previous two case reports in literature the questions arises whether childhood autism has any genetic and clinical relationship with Oculocutaneous albinism. (1)

REFERENCES: