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TITLE:

CHILDHOOD AUTISM IN A 13 YEAR OLD BOY WITH OCULOCUTANEOUS ALBINISM: CASE REPORT

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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. Oculocutaneous albinism is another hypomelanotic skin disorder that rarely present with multiple systemic manifestations, it is infrequently reported in association with childhood autism when compared to tuberous sclerosis and hypomelanosis of Ito.

However, the observation in this case report and the report of two previous literatures that have documented association between oculocutaneous albinism and childhood autism both in the affected individuals and families of individuals with childhood autism raises the question of possible genetic and clinical association between oculocutaneous albinism and childhood autism. More family and genetic studies into relationship between oculocutaneous albinism and childhood autism is desirable. This may provide useful clues into etiology, prevention and management of childhood autism vis-à-vis oculocutaneous albinism.

Key Words – Oculocutaneous Albinism, Hypomelanosis, Childhood Autism
CASE PRESENTATION

CHILDHOOD AUTISM IN A 13 YEAR OLD BOY WITH OCULOCUTANEOUS ALBINISM: CASE REPORT

Introduction

The patient is a 13 year old boy who had been an inmate of a destitute home managed by a Catholic Reverend Sister in Enugu, Nigeria. The destitute home provides habitation for mainly abandoned children and vagrant psychotic patients. The first psychiatric service contact with the patient had been during a community mental health service sponsored by the Rotary Club in which the authors volunteered. The patient was abandoned in a refuse dump few days after delivery and he had grown up with other abandoned children who found habitation in the destitute home because his parents could not be traced.

Medical and Psychiatric History

The patient was born with oculocutaneous albinism and on growing up he had been a child in a world of his own. He rarely played with other children in the destitute home and failed to develop like other children of his age. He was unable to develop speech and incapable of verbal communication, he only screamed sharply if in distress or in need of attention. He avoids eye to eye contact and he often appeared to be looking into space focusing on an unseen object. He failed to reciprocate any social gestures extended to him by his multiple care-givers over the years. Do not turn around if his name was called and he was most time preoccupied with playing with his fingers. He often snatched away other children’s meal and snacks after finishing his own. Presently at the age of 13 years the patient could not utter a word, he only shout and scream sometimes without
apparent reason. He does not respond to instructions and appeared distant away when interacted with. He however often respond to the word ‘take’ especially if the individual interacting with him was holding a biscuit or any other snacks which he usually snatched away forcefully and ate voraciously. Associated behavioral problems include destructive tendencies, screaming without apparent reason and running around the destitute home in a circle which gives him delight because he almost always resists any attempt to stop his running trip and often require forceful intervention to get him to stop.

**Developmental and Social History**

Gross motor development was said to have been normal when compared to other children in the destitute home and that the developmental impairments were restricted to the areas of communication, cognition and social interaction. The patient has not been exposed to any form of schooling and he has had no social interaction outside the destitute home where he had lived for thirteen years.

**Family History**

No information is available as regards the family history.

**Mental State Evaluation**

He appeared to be oblivious of his environment and was found to be staring into space, focusing on an unseen object. He did not respond to any question or instruction during interaction and made no speech of his own. His attention was however drawn with the word ‘take’ to which he snatched away
forcefully the biscuit from the examiner’s hand and ate voraciously as if the biscuit would be taken away from him if he was not fast at it. After this he was once again in his own world.

**Physical Examination and Psychological Investigation**

Physical examination revealed a young boy with features of oculocutaneous albinism, small in stature for his age. There are no other specific skin lesions, examination of the Central Nervous System (CNS) revealed no gross motor abnormality or sensory deficit. Examinations of other systems were essentially normal.

Formal Intelligent Quotient (I.Q) test was not carried out on this patient because of the confounding variation that socio-cultural influences have on standardized I.Q tests. However, empirical judgment based on interaction with the patient in the milieu of his socio-cultural environment showed that he was severely retarded with mental age estimated at about 3 to 4 years.

**Diagnosis and Treatment**

Based on the history, physical examination and clinical interaction with the patient, co-morbid diagnoses of oculocutaneous albinism (E 70.3) and childhood autism (F 84.0) with severe mental retardation (F 72) were made based on World Health Organization (W.H.O) International Classification of Diseases, 10th Edition (ICD – 10) (1). Identified problems in the patient were, communication impairment, poor social interaction, behavioral problems characterized by unwarranted screaming, unruly behavior of snatching away other children’s snacks, hyperactivity which is often displayed by running around the destitute home in a circle and destructive tendencies. He was referred to the Child and Adolescent Psychiatry unit of Federal Neuro-Psychiatric Hospital, Enugu, Nigeria for further evaluation and management where the patient was seen. Because of the
multi-disciplinary approach to the management of children with autism, there are limited facilities for children with autism in a third world country like Nigeria.

However, the behavioral problems were managed with oral Haloperidol 2.5mg daily on which the patient had been on for three weeks with reduction in hyperactivity and unruly behavior towards other children.

DISCUSSION

Several inherited medical and psychological disorders have been reported in association with childhood autism and many of these disorders are providing valuable information on the role played by genetics in etiology of childhood autism (2, 3, 4).

Inherited Hypomelanotic Skin Disorders and Childhood Autism

Some inherited hypomelanotic skin disorders that present with multiple systemic manifestations have been reported in association with childhood autism. These include tuberous sclerosis (4, 5, 6) and hypomelanosis of Ito (2, 7). The fact that these two hypomelanotic skin disorders associated with childhood autism showed neuro-cutaneous and multi-systemic manifestations would point to possible dysfunctional migration of ectodermal and mesodermal cell precursors during embryogenesis in etiology of these conditions and possibly leading to the neuro-developmental problems that characterized childhood autism. The pattern of chromosomal aberrations found in hypomelanosis of Ito and the polymorphic nature of the condition have led to the belief that hypomelanosis of Ito syndrome is a descriptive term rather than a true syndrome (8). If this belief is anything to go by, then the genetic basis and etiological process of the dysfunctional migration in
ectodermal and mesodermal cell precursors in this condition during embryogenesis leading to possible neuro-developmental problems observed in childhood autism could be heterogeneous. Therefore, further studies on dysfunctional maturation and differentiation of ectodermal and mesodermal cell precursors during embryogenesis in hypomelanotic skin disorders in general may be needed to unravel the patho-physiology of childhood autism.

Oculocutaneous Albinism and Childhood Autism

Oculocutaneous albinism is another hypomelanotic skin disorder. It is inherited in an autosomal recessive process and it rarely presents with multiple systemic manifestations that have been found in other inherited hypomelanotic skin disorders like tuberous sclerosis and hypomelanosis of Ito associated with childhood autism. Oculocutaneous albinism is infrequently reported in association with childhood autism when compared to tuberous sclerosis and hypomelanosis of Ito. However, Rogawski et al (9) had reported co-morbidity of oculocutaneous albinism and childhood autism in two boys and Delong (10) in a recent description of families of individuals with childhood autism had noted additional feature of oculocutaneous albinism in some families in addition to major affective disorder and special talents. Going by the observation of this present case report and the report of these two previous literatures (9, 10), the question arises whether childhood autism has any genetic and clinical relationship with oculocutaneous albinism.
CONCLUSIONS

Further studies on dysfunctional maturation and differentiation of ectodermal and mesodermal cell precursors during embryogenesis in inherited hypomelanotic skin disorders that have been associated with childhood autism are needed, more family and genetic studies into exact relationship that could be existing between oculocutaneous albinism and childhood autism are also desirable. These may provide useful clues into etiology, prevention and management of childhood autism vis-à-vis oculocutaneous albinism.

Competing Interest: The authors have declared no competing interest.

Authors Contributions: Both authors contributed equally to the management of the patient and preparation of this manuscript.

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REFERENCES


