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Case Report / Olgu Sunumu

Oculocutaneous Albinism and Autism: A Case Report and Review of Literature

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ABSTRACT

Oculocutaneous albinism and autism: a case report and review of literature

Autistic disorder is a highly heritable disorder characterized by impaired communication, social interaction, and repetitive behaviors. Several inherited medical and psychological disorders have been reported in association with childhood autism and many of these disorders shed light on the role of genetics in the etiology of childhood autism. Association of autism and oculocutaneous albinism is rarely reported in the literature. Herein, we report the joint occurrence of autistic disorder (AD) and oculocutaneous albinism (OCA) in a two-year and six-month old boy. This association was documented in a few previous reports about the affected individuals and families of individuals with childhood autism. All these reports and this case connote whether childhood autism has any genetic and clinical relationship with oculocutaneous albinism.

Key words: Autism, child, oculocutaneous albinism

ÖZET

Okülokütenöz albinizm ve otizm: Olgu sunumu ve literatür derlemesi

Otistik bozukluk; iletişimde ve sosyal etkileşimde bozulmalara neden olan, ayrıca, tekrarlayıcı davranışlarla giden genetik yönü güçlü olan bir bozukluktur. Çok sayıda kalıtsal tıbbi ve psikolojik bozuklukla çocukluk çağı otizmi arasında ilişki olduğu bildirilmiş, bu bozuklukların çoğu, otizmin etiyolojisinde genetiğin rolü konusunda ışık tutmuştur. Literatürde, otizm ve okülokütanöz albinizm birlikteliği nadir olarak bildirilmiştir. Bu yazıda, otizm ve okülokütanöz albinizmi olan 2 yaş 6 aylık bir olgu sunulmuştur. Bu ilişki ve etkilenen kişilerin aile bireyleri ile ilgili daha önce yayınlanmış az sayıda bildiri mevcuttur. Tüm bu bildirimler ve sunulan olgu, çocukluk çağı otizmi ve okülokütanöz albinizm arasında genetik bir ilişkinin olup olmadığı düşüncesini akla getirmektedir.

Anahtar kelimeler: Otizm, çocuk, okulokutanöz albinizm

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INTRODUCTION

Autism, a neurodevelopmental disorder included in the category of pervasive developmental disorders, has a high heritability, and is characterized by severe and pervasive impairment in reciprocal socialization, qualitative impairment in communication, and repetitive or unusual behaviors (1,2). A variety of genetic mechanisms may be involved in the etiology of autism, e.g., single gene disorders, copy number variations and polygenic mechanisms (3). Clinical studies show that organic causes are involved in 11-37% of the autistic cases (4). Many disorders have been reported to be associated with childhood autism providing valuable information on its genetic etiology (5-7). Sometimes the autistic phenotype is a part of a broader genetic condition called syndromic autism (3,8-10). Hypomelanotic skin disorders with multiple systemic manifestations like tuberous sclerosis and hypomelanosis of Ito have been previously reported in association with childhood autism (11).

Oculocutaneous albinism is a group of autosomal recessive disorders of melanin biosynthesis that is characterized by a generalized reduction in pigmentation of hair, skin and eyes (12). It is also a hypomelanotic skin disorder that may present with multiple systemic manifestations like photophobia, nystagmus and decreased visual acuity. It is seldom reported in association with childhood autism as opposed to tuberous sclerosis and hypomelanosis of Ito (11). Herein we report the joint occurrence of an autistic disorder and oculocutaneous albinism. When the words 'oculocutaneous albinism' and 'autism' searched in Pubmed, we saw that this association was documented in three previous reports (11,13,14).

CASE REPORT

The patient was thirty months old when he was referred to child and adolescent psychiatry outpatient clinic with disinterest to others, lack of eye-to-eye contact and lack of speech. When he was fifteen months old, he began to count and call his mother's name. He began to walk when he was 20 months. At that time he would not play with his peers. He avoided eye-to-eye contact and did not turn around when his name was called. Entourages were asking his mother if he is deaf or not. When he was twenty months of age his mother had a psychological trauma, so he experienced a period of time during which his father and other relatives took care of him. After this period, he stopped talking and saying the words he used to say. He was doing repetitive movements with his hands and spending too much time with colored objects. At the time of the examination he still had no meaningful words. He failed to reciprocate any social gestures. His family was not complaining of his behaviors as he was not shouting or screaming.

He had no serious medical disorder other than oculocutaneous albinism. Developmental impairments were restricted to the areas of communication and social interaction.

His mother and father were close relatives (offsprings of sister and brother). There were six more relatives of his father with oculocutaneous albinism. But none of them had any impairment in socialization and communication.

Physical, Ophthalmologic, Psychiatric Examination and Diagnostic Tests

His hair and skin were white, and his eyes were red. Other than bilateral horizontal nistagmus his neurologic examination was normal. The patient had photophobia but no strabismus. His biomicroscopic examination was also in normal limits. Some pathological findings were revealed by fundus examination which was performed under general anesthesia. Bilateral optic discs were hypoplastic and the epithelium of the retina was atrophic. He had decreased visual acuity.

He was disinterested to others. He wasn't answering the questions asked and often appeared to be staring into an unseen object. His affect was dull. He was standing by his father and wasn't paying attention during the interview. He didn't show any interest to the toys in the policlinic. Sometimes he was joining his hands and shaking his fingers. Denver Developmental Screening Test II (15) was carried out. He was 3 years and 2 months old at the time of the evaluation. His personal–social development was at the fifteen and a half months level, fine motor-adaptive development at the twenty and a half months level, language development at the eleven and a half months level and gross motor development at the nineteen and a half months level. He had developmental delay in all developmental steps.

His sleep EEG showed that cerebral bioelectric activity was in normal limits. There was no pathological finding in cranial MRI. His hearing was determined to be normal by evoked auditory potentials.

Diagnosis and Treatment

Based on the history taken from his family, physical examination and clinical interaction with the patient, diagnoses of oculocutaneous albinism and childhood autism were made based on ICD-10 (16). There was severe impairment in his social interaction and communication. He began to be followed at child and adolescent psychiatry, child neurology and ophthalmology outpatient clinics. Special education was offered for him. There was not any need for the initiation of a psychopharmacologic agent since he displayed no problematic behavior.

DISCUSSION

When the presented case was twenty month-old, after his mother's psychological trauma he had lost his mother's care for a time. After this period he stopped talking and saying the words he used to say. This clinic may be seen in reactive attachment disorder also. But presences of disinterest to others before his mother's illness make us not to think this diagnosis.

Epidemiological studies of autistic twins showed

that concordance rate is higher in monozygotic twins (69-95%) than in dizygotic twins (0-24%). These studies estimated the heritability of autism to be more than 90% (17-19). Hypomelanotic skin disorders with multiple systemic manifestations have been reported in association with childhood autism like tuberous sclerosis (7,20,21) and hypomelanosis of Ito (5,22). In order to gain a better understanding of the mechanism of these associations, Bakare and Ikegwuonu (11) emphasized the need for further studies on dysfunctional maturation and differentiation of ectodermal and mesodermal cell precursors during embryogenesis.

The case had decreased visual acuity. Bilateral optic discs were hypoplastic. It is shown that children with optic nerve hypoplasia are at risk of social, communication, and repetitive/restrictive difficulties and clinical ASD (23).

Association of autism and oculocutaneous albinism is rarely reported in the literature. Rogawski et al. (13) reported such an association in two boys. Delong (14) described four families of individuals with childhood autism and noted the additional feature of oculocutaneous albinism in some families in addition to major affective disorder or psychotic disorder and special talents or intellectual abilities. He reported two OCA and autism cases in his study. Furthermore, he also reported two other autism cases with a possible but undiagnosed OCA: the first of these had abnormal eye movements and pale iris, and the second was a pale-looking individual with cotton-white hair and nystagmus. DeLong et al. (24) described a strong correlation among autism, bipolar disorder and special abilities in families of children with autism.

Bakare and Ikegwuonu (11) recently reported a 13 year-old Nigerian boy who had oculocutaneous albinism and autism. He was unable to develop speech and was incapable of verbal communication. He was only screaming when in distress or in need of attention (11).

To the knowledge of the author, there is no other report except the three mentioned reports in the literature about this association. All these reports and this case may indicate a genetic and clinical relationship with oculocutaneous albinism and autism. These cases may provide useful clues to the genetic mechanism of a part of the autism spectrum disorders and may also help explain some of the eye movements and gaze abnormalities in individuals with autism, possibly through misdirection of optic nerve pathways (14). The frequency of autism-like symptoms in individuals with OCA has not been studied yet. Genetic studies may be carried out in OCA individuals with autism-like symptoms which may provide a better understanding of the etiology of autism.

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