Author's response to reviews

Title: Childhood autism in a 13 year old boy with oculocutaneous albinism: a case report

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Author's response to reviews: see over
The Editor
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Dear Sir,

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

Many thanks for the comments of the Reviewers.

Adjustments have been made to the manuscript based on the comments of the Reviewers. The following changes were effected:

Reviewer 1 (Suad Kapetanovic);

- The ethnicity of the patient has been stated under Introduction sub-heading.

- Childhood autism core-symptoms and prevalence have been included under ‘Discussion’ heading. This has led to inclusion of two additional literatures in the Reference section. The numerical order of the References list has also changed following the introduction of the two additional literatures in the References.

- Examination of the patient’s Central Nervous System (CNS) revealed no vision or hearing impairment. This has been clarified under ‘Physical Examination and Psychological Investigation’ sub-heading.

- To the best of knowledge of the authors, we are not aware of any documented evidence that parents with autism or other pervasive developmental disorders are more likely to abandon their children than non-autistic parents. This would worth a further investigation. However, there was no other child with autistic disorder in the destitute home aside the patient.
Reviewer 2 (Ann E. Maloney);

- Clarifications have been made to the mental status examination of the patient on attention and motor abnormality.
- Other corrections pointed out concerning phrasing of sentences and punctuations have been effected on a line-by-line basis.

Reviewer 3 (Karen Harman)

- Reviewer’s queries on evidence of co-morbid diagnoses of Oculo-cutaneous Albinism and Childhood Autism made in this patient:

**Oculo-cutaneous Albinism:** Though the diagnosis and specific type of oculo-cutaneous albinism might need to be confirmed by genetic testing, especially among people of white skin color. Obviously there are no facilities for genetic testing to classify or confirm oculo-cutaneous albinism in this environment. However, the diagnosis of oculo-cutaneous albinism can be made based on just physical appearance in a clinical setting among black Africans of Sub-Saharan African descent. Among black Africans, variations in coloration of hairs and eyes are not common as observed among people of white skin color. In Sub-Saharan Africa, it is not difficult for even a lay person to recognize individuals with oculo-cutaneous albinism whom are often termed “Albino”. It might be interesting to state that the recognition is so classical that individuals with oculo-cutaneous albinism in some rural communities in this environment are often idolized and worshiped as “gods”. The diagnosis of Oculo-cutaneous Albinism made in this particular patient was based on physical appearance of his skin, hairs and eyes colors which are classical in an individual of black African descent. I think this is one of the reasons the First Reviewer commented on given the specific detail of the patient’s race or ethnicity.

**Childhood Autism:** Most diagnoses made in the field of Psychiatry today including Childhood Autism are syndromes (i.e. constellation of signs and symptoms) highlighted as criteria in the two most widely used method of classification: International Classification of Diseases, tenth Edition (ICD-10) compiled by World Health Organization (W.H.O) and Diagnostic and Statistical Manual of Mental Disorder, fourth Edition (DSM-IV) compiled by the American Psychiatric Association (APA). The diagnosis of Childhood Autism made in this patient was based on the history, mental state examination and physical examination of which symptoms and signs elicited fulfilled the criteria specified for childhood autism in ICD-10. The Reviewer may want to look through these criteria in ICD-10.
• The suggestion, to affirm that the boy had not suffered any head injury or other medical conditions during the process of development that can influence his cognitive and developmental impairment is well appreciated. Any medical condition that occurred in the mother during pregnancy and before the age of three years in this patient would be more important to us. It can be inferred that the pregnancy and birth history of the patient is not known to us. Therefore, the detail of maternal medical condition in pregnancy, perinatal and postnatal injuries, if any in this patient is obscured to us.

• On the assessment of patient’s mental age: It had already been stated that there was no impairment in other areas of development apart from speech development (communication), cognition, and social interaction. Therefore, the estimation of mental age in this patient in the context of his socio-cultural milieu was based on the areas of speech, social interaction, cognition and adaptive functioning deemed appropriate for specific age in the patient’s socio-cultural milieu. Estimation of mental age in this patient was not based on gross and/or fine motor skills which were assessed as appropriate for patient’s biological age. That the environment in which the patient had grown up could be less stimulating to the patient’s intellectual development is a well taken point. This however can not rule out the fact that core symptoms of childhood autism are present in this patient. In addition, it is also a known fact that mental retardation and childhood autism co-exist in majority of patients with autism.

• In as much as it is agreed that hearing and vision impairments in a child can mimic symptoms of childhood autism. These impairments, if present could be detected during general physical examination. For example, standing behind the child and making some noise with a bunch of keys to draw the child’s attention could serve as a gross test for hearing. So also is standing at some angle and distance away from the patient in the examining room and trying to draw his attention with the word ‘take’. That the child responded to the word ‘take’ and was able to find the examiner’s location within the examination room to snatch away the snacks was a gross test of hearing and vision. This however, is not to excuse the need for a detail examination of the child by an ophthalmologist, audiologist or a geneticist. However, in a case where the specialists are not available at all as in the case of geneticist and audiologist or are available at an unaffordable cost to the patient and the careers in a peculiar environment where modality of payment for health care services is still largely out of pocket pay. We think there is a need for adaptive practice in such environment like ours. In recognizing the need for this adaptive practice, we had done a self criticism that this multi-disciplinary approach might not be feasible in our environment for now because of the limited availability of specialists in various fields to work collaboratively in management of children with autism.
• The corresponding author, whose area of interest is in child and adolescent psychiatry did a detail assessment of the patient at the Child and Adolescent Unit of Federal Neuro-Psychiatric Hospital, Enugu, Nigeria.

• Lastly, it is important to state that this case report is not concluding that the patient had a co-morbid diagnosis of childhood autism and oculo-cutaneous albinism due to an underlying genetic predisposition as the Reviewer possibly presumed. The manuscript is actually reporting another co-existence of childhood autism and oculo-cutaneous albinism. The two conditions may be co-existing in the patient independently and totally unrelated or alternatively have some connections clinically or genetically (Evidence for either is not presently available). However, building on the reports 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 (Rogawski et al, 1978 and Delong, 2007) and the observation in this present case report, a search light is being pointed at possible or likely clinical or genetic association between the two disorders. Only future studies can provide concluding evidence.

• Changes made on the manuscript based on this Reviewer’s comments:

  Permission sought and consent obtained to include patient’s photographs alongside the Manuscript for the purpose of publication if the Manuscript is eventually accepted for publication.

Thank you.

Yours faithfully,

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