Reviewer's report

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

Version: 3 Date: 4 September 2007

Reviewer: Karen Harman

I am familiar with the literature and believe that this case meets one of the 7 criteria for evaluation in the journal: An unexpected association between diseases or symptoms

Has the case been reported coherently?: Yes

Is the case report authentic?: Yes

Is this case worth reporting?: No

Is the case report persuasive?: No

Does the case report have explanatory value?: No

Does the case report have diagnostic value?: No

Will the case report make a difference to clinical practice?: No

Comments to authors:

General

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Revisions necessary for publication

The case report entitled "Childhood Autism in a 13-Year-Old Boy With Oculocutaneous Albinism: A Case Report" is a case report that suggests an association between the two conditions of oculocutaneous albinism and autism. Given that the argument for further understanding of hypomelanotic skin disorders would assist in the understanding of the pathophysiology of childhood autism, the evidence of both of these diagnoses needs to be elaborated in the report.

Although the young boy in the case report is described as having features of oculocutaneous albinism, the details of his physical examination were not included. Recognizing the potential limited resources available for diagnostic evaluation, it would be important to include an explanation as to why other possible diagnoses on the differential list had been excluded. More detailed
description of the skin, hair and eyes and visual behaviour would be helpful. A comment of whether or not the child had ever had a vision examination or been seen by an ophthalmologist should be included. Additionally, if photographs had been taken of the child, these could be reviewed with a geneticist and ophthalmologist knowledgeable in oculocutaneous albinism. A functional assessment of what type of vision or hearing of the child would be important both in the description of the proposed diagnosis of oculocutaneous albinism, but also in the formulation of the developmental diagnostic profile. It would be important to make a comment that the child had not had other environmental or medical issues that might influence his cognitive and developmental impairments, such as head injury or an episode of encephalitis or meningitis. These pertinent negatives were not included.

From the developmental perspective a more detailed description of the acquisition of developmental milestones would have supported their judgment of the child's mental age. Not being able to complete a formal cognitive assessment in this situation is not unexpected or inappropriate. However, the information used to decide on the estimate of mental age needs to be included. More detailed observations of the child or a description of the child's adaptive abilities as observed by his care providers would give a better understanding of the child's developmental functioning level or abilities. One would expect that the description of the child's mental age be based on more than observation of his gross motor skills. Some attempt at eliciting his areas of strength, such as receptive language, self help, fine motor or visual-spatial skills, would be important to make such a judgment. It is important to provide sufficient evidence of the child’s’ cognitive functioning, in order to propose that the child's cognitive abilities were sufficiently high enough to make an additional diagnosis of autism rather than the overall diagnosis of a profound cognitive impairment along with a vision impairment. Additionally, an important aspect of the developmental assessment would include a description of his environment prior to age 6 years. It is indicated within the report that he was cared for by multiple people, lived with adult mental health patients and did not attend school. This would be a less than optimal stimulating environment for a child, particularly given the developmental and sensory concerns.

Given these limitations to describing the developmental profile (both the cognitive and qualitative social and communication aspects), there is insufficient evidence that the child had autism (due to an underlying genetic predisposition) rather than a presentation of autistic traits due the combined effects of his under-stimulating environment, vision impairments, and severe cognitive impairments.

The child was reportedly referred to the Child and Adolescent Psychiatry Unit of the Federal Neuro-psychiatric Hospital in Enugu, Nigeria for further evaluation and management. It would be important to include any evaluation completed at this site, particularly if there were consultations from a pediatrician, an ophthalmologist, an audiologist or a geneticist.

Without sufficient evidence for the oculocutaneous albinism diagnosis and the autism diagnosis, it is therefore hard to make the genetic hypothesis connection.
Significant modifications and additions of information would be necessary for this case report to be published.

**What next?:** Reject

**Quality of written English:** Needs some language corrections before being published