Reviewer's report

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

Version: 3 Date: 29 August 2007

Reviewer: Ann E. Maloney

I am familiar with the literature and believe that this case meets one of the 7 criteria for evaluation in the journal: New associations or variations in disease processes

Has the case been reported coherently?: No

Is the case report authentic?: Yes

Is this case worth reporting?: Yes

Is the case report persuasive?: Yes

Does the case report have explanatory value?: Yes

Does the case report have diagnostic value?: Yes

Will the case report make a difference to clinical practice?: Yes

Comments to authors:

General

I believe this adds to the literature about neurodevelopmental disorders and cutaneous manifestations. It may have missed some key literature, like Thiele E, Danforth, N et al. Most TS patients, for instance, do not have autism. There may be a link between cutaneous lesions, but it is unclear when these started (since there was little physical exam at birth--abandoned child--so hard to know when the lesions appeared). The mental status examination should include more about attention and movements. I believe that reorganizing and revising would be helpful to the reader.

Revisions necessary for publication

LOCATION: Page 2, Line 9

TEXT: "However, the observation in this case report and in two previous reports which 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."

He does not respond to instructions and appeared distant 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 included destructive tendencies, screaming without apparent reason and running around the destitute home in a circle which gives him delight because he almost always resisted any attempt to stop him and often required forceful intervention to get him to stop.

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screaming without any apparent reason and running around the destitute home in a circle which gives him delight because he almost always resisted any attempt to stop him and often required forceful intervention to get him to stop."

ERROR: "get" is an informal word.
SUGGESTION: redirect him, etc--this makes me wonder about his hearing, too

LOCATION: Page 4, Line 15

TEXT: "Family History No information is available as regards the family history."
ERROR: "as regards" is a verbose phrase.
SUGGESTION: about

LOCATION: Page 4, Line 20

TEXT: "He appeared to be oblivious of his environment and was found to be staring into space, focusing on an unseen object."
ERROR: "was found to be" is a verbose phrase.
SUGGESTION: was

LOCATION: Page 4, Line 24

TEXT: "After this he was once again in his own world."
ERROR: Comma needed after introductory clause.
SUGGESTION: Insert comma after clause: "After....."

LOCATION: Page 5, Line 11

TEXT: "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."
ERROR: "estimated at about" is a redundant phrase.
SUGGESTION: "estimated at"

LOCATION: Page 5, Line 11

TEXT: "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."
ERROR: Use tilde (~) symbol
SUGGESTION: ~3 and could elaborate here about movements, gait, balance, gross motor, etc

LOCATION: Page 5, Line 21

TEXT: "Identified problems in the patient were, th 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."
ERROR: Comma needed
SUGGESTION: Insert comma before "which" or change "which" to "that"
LOCATION: Page 6, Line 4
TEXT: "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."
ERROR: "towards" is not a preferred word choice.
SUGGESTION: toward

LOCATION: Page 6, Line 15
TEXT: "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."
ERROR: "the fact that" is a verbose/expendable phrase.
SUGGESTION: None

Page 8, line 2
Vis-a-vis is not generally used in medical correlations of syndromes, could use word like etiology or clustering of symptoms, or another linking term

What next?: Revise and resubmit

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