Reviewer's report

Title: 2-methylbutyryl-CoA dehydrogenase deficiency associated with autism and mental retardation: a case report

Version: 2 Date: 3 August 2007

Reviewer: Carolyn Ellaway

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?: Yes

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?: No

Does the case report have diagnostic value?: Yes

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

Comments to authors:

General

The authors report a case of 2 methylbutyryl CoA dehydrogenase (SBCAD) deficiency in a 4 year old Somali boy with developmental delay, autism and a past history of seizures. This patient is the first patient with the disorder to be reported with autism.

The boy was found to be homozygous for the c.303+3A>G change in the SBCAD gene.

Interestingly the authors also report other asymptomatic cases with the identical genotype. However environmental and other factors are also important for some inborn errors of metabolism to lead to clinical symptoms.

The association of SBCAD with autism may be causative or possibly a chance association, given the frequency of autism in the general population. However with rare disorders such as this, case reports are important in helping to define the clinical phenotype.

The case reports also highlights the importance of considering an underlying
inborn error of metabolism in children presenting with autism and other neurological problems.

Revisions necessary for publication

No revisions necessary

**What next?:** Accept

**Quality of written English:** Acceptable