Reviewer’s report

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

**Version:** 2  **Date:** 30 July 2007

**Reviewer:** Isabelle Rapin

I am familiar with the literature and believe that this case meets one of the 7 criteria for evaluation in the journal: Presentations, diagnoses and/or management of new and emerging diseases

**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?** Yes

**Does the case report have diagnostic value?** Yes

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

**Comments to authors:**

**General**

Revisions necessary for publication

Review by Isabelle Rapin <rapin@aeom.yu.edu>

This is a well written paper with adequate documentation of the clinical evidence for autism which is essentially ready for publication. I would suggest softening the conclusion: in my opinion it would be very nice to be able to do metabolic studies on all cases of autism, but this is unrealistic, except in the context of a research study. Today the yield of routine metabolic and genetic testing in unselected individuals on the autism spectrum is extremely small. As is true in general, the more severely handicapped the child, i.e., the lower the IQ and the clearer other evidence for severe brain dysfunction, the higher the yield of this type of study. So it seems to me that you might want to say that, in routine clinical practice, metabolic/genetic/imaging investigations are indicated in selected children based on the clinical evidence, or something to this effect. You might also want to say
that abnormalities uncovered by such studies rarely dictate any change in the management of the child but may provide potentially important information for family members and, in an occasional case, novel evidence on brain mechanisms that may advance our understanding of the brain dysfunction responsible for this phenotype. I am not qualified to comment on the metabolic aspects of the paper.

**What next?:** Accept after minor revisions

**Quality of written English:** Acceptable