Reviewer's report

Title: A syndrome of lethal familial hyperekplexia associated with brain malformation.

Version: 4 Date: 8 July 2012

Reviewer: Konstantinos Vasiliadis

Reviewer's report:

Reviewer report:
This paper could be considered for publication after major compulsory revision.

Comments for authors:
A syndrome of lethal familial hyperekplexia associated with brain malformation is potentially valuable to clinicians. However, this manuscript needs to be substantially reworked to make it meaningful.

1. First Problem: Structure. At present, it's a lump of long paragraphs and is hard to follow. I'd recommend using sections and subsections: Patient 1. Patient 2, also abstract, introduction, case report and discussion. And please consider separate paragraphs for initial clinical presentation, workup, subsequent clinical events and MRI findings for each patient.

Discussion:
CNS findings, MRI Findings, Neurophysiological Findings, EEG findings. Conclude with a few paragraphs about how your patients do or do not fit "classic" lethal familial hyperekplexia syndrome. Finally a paragraph about the literature review.

2. Second Problem: Content. This manuscript purports to focus on brain malformations but does not provide enough detail. More information about the epilepsy would have been most useful (what did the EEGs show? What types of seizures did the pts have? How well did the medications work?). Supportive information about the degree of motor + cognitive impairment would also have been helpful. The biggest problem is with the discussion of radiographic findings. The authors lump together what has been described radiographically with what has been found pathologically. I propose clearly description of MRI finding in small but understanding paragraph, also the same on figure legends.

3. Third problem: figures and genealogy. Please make them more understanding, especially the figure legends and mark them correctly in the paper.

I consider this paper for publication after major compulsory revision.

Level of interest
- An article of importance in its field

Quality of written English
- Acceptable

With best regards,

E. Vargiami, Child Neurologist, 1st Pediatric Clinic, Aristotle University of Thessaloniki

**Level of interest:** An article of importance in its field

**Quality of written English:** Acceptable

**Statistical review:** Yes, and I have assessed the statistics in my report.