Reviewer’s report

Title: West syndrome followed by juvenile myoclonic epilepsy: a coincidental occurrence?

Version: 1 Date: 28 December 2012

Reviewer: ville dorothee

Reviewer’s report:

methods
You should define more accurately the concept of idiopathic epilepsies and the characteristic of JME, because large part of the discussion is based on these points.

What is new and interest of this manuscript, aim of the study
- First observation of association of West Syndrome and myoclonic juvenile epilepsy
- Discussion of link between WS and JME (coincidence or age-related expression of epilepsy)
- Discussion of the concept of idiopathic west syndrome debated by some authors

introduction
- Define more accurately concept of idiopathic epilepsy
- “one third of the patients were seizure-free, another third had seizures daily or monthly” (do you mean to monthly? Because there is a large difference between daily and monthly)

Case presentation
- « Chromosome analysis revealed a XY karyotype without mosaicism”.: do you mean Karyotype was normal? (detection of mosaicism with skin biopsy performed?)
- You should complete EEG data at 12 years of age (background pattern, reaction with hyperpnea, light stimulation…)
- age of the patient when MRI was performed?

discussion
Discussion has to be more consistent:
- It’s not possible to conclude about link or coincidence between these to age-dependant epileptic syndrome: this observation is clearly “syndromic”, evocating genetic aetiology; so diagnosis of idiopathic epilepsy could not be retained
- EEG pattern at 12 years of age should be discussed on the light of characteristics reported in JME.

It may be useful to

- You should discuss aetiological explorations: MRI control and especially new genetic screening on the light of advances in genetic investigations (array CGH, exome sequencing…) in order to identify a candidate gene of epilepsies associated to cerebral maturation or presence of a deletion which could explain this “syndromic” presentation. This point lacks strongly in your observation; you may also confront your observation with already reported genetic data in WS and JME in order to find a common genetic abnormality?

- Concept of idiopathic spasm widely debated in the literature has to be discussed.

Table and figure

Age of the children and sequence should be mentioned on the MRI figure legend.

**Level of interest:** An article whose findings are important to those with closely related research interests.

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

**Statistical review:** No, the manuscript does not need to be seen by a statistician.