Reviewer’s report

Title: A novel Bruton’s tyrosine kinase gene (BTK) missense mutation in a Chinese family with X-linked agammaglobulinemia

Version: 2  Date: 7 July 2014

Reviewer: SHY-H-DAR SHYUR

Reviewer’s report:

The author must respond to these before a decision on publication can be reached. For example, additional necessary experiments or controls, statistical mistakes, errors in interpretation.

There are some comments regarding this paper that should be pointed out.

1. In P1, L16 - the sentence “X-linked agammaglobulinemia (XLA) is a rare inherited disease characterized by recurrent viral and bacterial infections” should be rewritten, because Viral infections are usually handled normally with the exceptions of hepatitis viruses and enteroviruses.

2. In P1, L23-24 – about the sentence “A genetic analysis of the family revealed an affected male sibling with a c.1117C>G mutation”, the author should detail the clinical manifestations and management of this affected male sibling.

3. P2, L42-43 – the sentence “Any mutation that that occurs in any site within one of these 5 domains” should be corrected to “Any mutation that occurs in any site within one of these 5 domains”.

4. P2, L51-53 – the sentence “In the subsequent 2 years, the boy experienced 1 episode of septicemia, 3 episodes of otitis media, 2 episodes of pneumonia, and 4 episodes of bronchitis.”, The author should detail the causative specific pathogens of each episodes.

5. P6, L142-147 – In patient with initial IgG < 0.34 g/L, he received IVIG 400 mg/kg/month for the first 3 months followed by subsequent infusions once every 2-3 months. The author should detail the follow-up serum IgG concentration.

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.

Declaration of competing interests:
I declare that I have no competing interests.