Reviewer’s report

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

Version: 3 Date: 9 August 2014

Reviewer: SHYH-DAR SHYUR

Reviewer’s report:

This is a paper entitled “A novel Bruton’s tyrosine kinase gene (BTK) missense mutation in a Chinese family with X-linked agammaglobulinemia”.

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

Major Compulsory Revisions

1. In P6, L150-4 – about the sentences “It is not necessary for every patient to carry out a genetic analysis for the high cost of the test if patients had a typical presentation with hypogammaglobulinemia and family history. Only suspected patients need the evaluation of BTK gene analysis to make an early diagnosis and receive the future IVIG treatment.” should be rewrite, because the author must know that patients with typical presentation with hypogammaglobulinemia and low level of peripheral B cells can have other mutated gene defects different from BTK gene.

Minor Essential Revisions

2. In P2, L52 - the sentence “anti-infective and therapies” should be corrected to “anti-infective therapies”.
3. In P2, L55 - the sentence “staphylococcus aureus and streptococcus pneumonia” should be corrected to “staphylococcus aureus and streptococcus pneumoniae”.
4. In P2, L152 - the world “patitents” should be corrected to “patients”.
5. In P7, L186 - the abbreviation “SH1,Scr Homology1;” should be deleted.

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

Quality of written English: Acceptable

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