Reviewer's report

Title: Atypical X-linked Agammaglobulinemia caused by a Novel BTK Mutation in a selective IgM Deficiency Patient.

Version: 3 Date: 9 June 2013

Reviewer: Esko Wiltshire

Reviewer's report:

Lim et al proved an interesting case report suggesting the phenotype of XLA may be broader than previously suspected. The background and case report are generally well written, and well referenced.

Case report: How was the haematuria/proteinuria originally detected? Were there symptoms? Routine screening? Other reasons?

Most importantly, what evidence do the authors have that the c.347C>T (p.P116L) mutation is pathogenic and not a polymorphism? They need to provide this to justify their conclusions, and without this information it is difficult to determine whether the case report adds new information.

The discussion is well written and appropriate

Minor Points:
1. Page 5 line 9 [pleksin
2. Page 5 line 12 “defective B cell developments” + development
3. Page 6 fourth to last line – Kidney Ultrasound?
4. Page 7 final lie should be” diagnoses” not diagnosis
5. Page 10 3 last line “maintained” should read “remained”

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 I have no competing interests in relation to this paper