Reviewer's report

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

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Reviewer: Maria Marluce S Vilela

Reviewer's report:

The article writes about a missense mutation type (p.P116L), which occurs when one amino acid is substituted for another. Being a missense mutation and not yet published in the literature, I think the authors should do additional testing to prove that it is not polymorphism but a new mutation in BTK.

Authors can seek nucleotide exchange (c.347C> T) in healthy controls by enzyme restriction or direct sequencing. If this same exchange occurs in healthy controls, it is polymorphism. I cannot say what would be the ideal number of controls to be analyzed, but we suggest evaluating 100 controls. But some investigators analyze around 800 controls. To complement the analysis there are also computer programs (Mutation taster, Polyphen 2) to predict the pathogenic potential of mutation.

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 have no conflict of interesse.