Reviewer’s report

Title: Intermittent low platelet counts hampering diagnosis of X-linked thrombocytopenia in children: report of two unrelated cases and a novel mutation in the gene coding for the Wiskott-Aldrich syndrome protein

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Reviewer: William Mitchell

Reviewer's report:

Medina et al present two case reports detailing X-linked thrombocytopenia patients who were misdiagnosed initially as having ITP. The manuscript is well written and complies with the clinical trials guidelines and checklist. The cases and their workup are described in adequate detail. The major point the authors are trying to make is that these two patients were not initially diagnosed with WAS because of their variable platelet counts. However, they do not present a complete review of presenting platelet counts at diagnosis of WAS. For example, the authors of one multicenter review (Sullivan KE, Mullen CA, Blaese RM, Winkelstein JA. A multiinstitutional survey of the Wiskott-Aldrich syndrome. J Pediatr. 1994 Dec. 125(6 Pt 1):876-85) reported that individual patients could have large variations in their platelet counts.

The use of the phrase "hampering differential diagnosis" does not seem completely accurate. The irregular symptoms make the final diagnosis unclear, but the differential diagnosis of childhood thrombocytopenia does not change - e.g. WAS is part of the differential diagnosis of any male child with thrombocytopenia, and this does not change. It is the final diagnosis that changes and that final diagnosis may be "hampered" by uncharacteristic symptoms.

A minor point is that the authors also state that the platelet size is always small in WAS, however there have been reports of normal platelet size in WAS patients (A unique presentation of Wiskott-Aldrich syndrome in relation to platelet size. Patel PD, Samanich JM, Mitchell WB, Manwani D. Pediatr Blood Cancer. 2011 Jul 1;56(7):1127-9).

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
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Yes

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