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: Neena Kapoor

Reviewer's report:

In the paper entitled "Intermittent low platelet counts hampering differential diagnosis of thrombocytopenia in Children: report of two unrelated cases and a novel mutation in the gene of Wiskott-Aldrich Syndrome Protein" Medina et al. report two unrelated cases with low expression of Wiskott-Aldrich syndrome protein. Molecular analysis identified a mutation in exon 2 of the Wiskott-Aldrich syndrome protein gene, leading to a p.Thr45Met amino acid change in one patient and the presence of a novel missense mutation in exon 2 of the Wiskott-Aldrich syndrome protein gene, resulting in p.Pro58Leu amino acid change in other patient.

In the paper authors emphasized that intermittent thrombocytopenia is a common presentation and how pediatrician mistake XLT for immune thrombocytopenia and pediatrician should be aware of this disease's presentation. Novel mutation identification, is important, for the expansion of the disease related knowledge and for subsequent testing in the sib ship and for counseling.

I fully agree with the authors that pediatrician should be aware and educated about this clinical presentation. And any time patient has there is thrombocytopenia, physician should examine the smear to rule out platelet abnormalities, Need some clarification ………. incidence of ITP in pediatric is somewhere 1-5/100,000 children. So, the statement it is a very common condition seen by pediatricians, is not really true. The article gives a sense that when patient is thrombocytopenic, physician label it as immune thrombocytopenia without any further investigation. I am sure, if pediatrician have a case of thrombocytopenia, they will be investigating for all the possible causes of thrombocytopenia even if it is intermittent, or will be consulting with the specialist to establish the diagnosis.

I would recommend to change the text emphasizing, how some of these children have atypical presentation and because of that diagnosis could be difficult to make. Diagnostic measures are essential to rule out all other potential diagnosis and molecular studies may be necessary to confirm the diagnosis. Fig 1 and 2 are not needed. Patient info is described in the method and is not needed in the abstract in that detail. In abstract authors should only talk about novel gene mutation, which helped in confirming the diagnosis.
It is a case report, should be short precise and to the point.

**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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Unable to assess

**Are the conclusions drawn adequately supported by the data shown?**
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Yes

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Not relevant to this manuscript

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