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 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" Medina et al described the cases of two young children with intermittent thrombocytopenia. These children after work up were found to have X linked thrombocytopenia with, novel mutation in the gene coding for the Wiskott Aldrich syndrome protein. Their take home massage is that when there is thrombocytopenia is noted X linked thrombocytopenia should be in the differential diagnosis.

I fully agree with the authors that when there is thrombocytopenia, WAS / XLT should be in the differential diagnosis, I do differ from them in the approach what point needs to be emphasized to the readers specially people in general pediatric practice.

1. Thrombo-cytopenia is not a common blood finding which general pediatrician will see often and if they do have a patient with thrombocytopenia intermittent (repeated episodes) or persistent, that child needs further investigations. Thrombocytopenia is rare enough and serious enough that it should always investigated till you know exact cause for it, as that may require specific treatment depending upon the diagnosis. So massage to the readers should be that if thrombocytopenia is noted, all potential differential diagnosis should be taken in account while considering the work up., especially when patient does not have classical phenotyping of the disease.

2. The table with various causes for thrombocytopenia is OK but should be linked with the work up. Step wise. What findings of phenotyping suggest a given diagnosis and what tests will need to be done to rule out or rule in the diagnosis and progress to the level of establishing genotypic diagnosis. And then they can talk about their novel gene mutation and missense mutation in discussion that in some cases it will be genotypic analysis which will establish and confirm the diagnosis. Again emphasis on like their two atypical patients that though they had evidence of low platelet volume it was their high index of suspicion which lead to gene mutation studies and which confirmed the diagnosis and identification of novel gene mutation for Wiskott Aldrich syndrome protein.
Are the methods appropriate and well described?
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