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REVIEWER'S REPORT

Manuscript No.: IJAR-58818

Title: Bare Lymphocytic Syndrome Type I Due to TAP2 Mutation in an 18 Year Old Male: A Case Report

Recommendation:

Accept as it is .....
✓ Accept after minor revision.....
Accept after major revision .....
Do not accept (*Reasons below*) .....

Rating	Excel.	Good	Fair	Poor
Originality		✓		
Techn. Quality		✓		
Clarity		✓		
Significance	✓			

Reviewer Name: Dr S. K. Nath

Detailed Reviewer's Report

Strength of the study:

- The case describes a rare primary immunodeficiency disorder
- The patient has a clinically interesting multisystem presentation
- Whole exome sequencing confirmed a homozygous TAP2 mutation
- The clinical history and diagnostic progression are well described
- Bronchiectasis, vasculitis and neurological manifestations are documented
- The clinical timeline improves understanding of disease progression
- Genetic and HLA reports provide useful supporting evidence

Weakness of the study:

- Ethical clearance and written consent for publication are not clearly mentioned
- The relationship between vasculitis and TAP2 deficiency is not firmly established
- Immunological investigations supporting MHC class I deficiency are limited
- Long term clinical outcome is not available
- The patient was discharged for future transplantation evaluation without further follow up
- Some discussion points extend beyond the evidence presented in the case
- Grammar, formatting and sentence construction need careful editing

Reviewers Comments:

This case report describes a rare and clinically important presentation of Bare Lymphocyte Syndrome Type I associated with a homozygous TAP2 splice site mutation. The combination of recurrent respiratory infections, bilateral bronchiectasis, vasculitic ulcers, severe malnutrition and previous neurological involvement makes the case interesting and potentially valuable for clinicians. The genetic finding provides strong support for the diagnosis, and the clinical timeline and supplementary reports add useful documentation. However, the manuscript has some important limitations. Ethical clearance and specific written consent from the patient or guardian for publication of clinical details and genetic reports are not clearly stated and should be documented. The manuscript also provides limited immunological data directly demonstrating MHC class I deficiency. The proposed association between TAP2 mutation and vasculitic manifestations should be presented more cautiously. Long term follow up and treatment outcome are unavailable. Grammar, punctuation, spacing and sentence construction require editing. Overall, the case is worthwhile, but ethical clarification and moderate editorial and scientific revision are needed.

Previously Published anywhere/Plagiarism check:

No clear evidence of previous publication or duplicate reporting is apparent from the submitted case report. However, plagiarism cannot be confirmed from manuscript review alone. A formal similarity check should be performed before publication. The authors should also confirm that the genetic reports, clinical timeline and supplementary documents are original and have not been published previously.