

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 after major revision

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

Reviewer's ID: JPR-237

Detailed Reviewer's Report

Present case is unusual and rare combination of bronchiectasis, severe malnutrition, neurological manifestations, and vasculitic skin disease. This study would be benefited for further study, diagnosis and treatment and control in future. This manuscript required revision and correction before consideration for publication. These following points need to be revised or correction:

- 1. What is the criteria for diagnosis. The authors should provide evidence of reduced/absent MHC class I (HLA-A, -B, -C) surface expression, preferably by flow cytometry, and relevant immunological investigations such as CD8+ T-cell numbers/function, NK-cell profile, immunoglobulins, and lymphocyte subsets.***
- 2. Provide detailed characterization of the TAP2 variant. The manuscript reports c.1272+1del, classified as "likely pathogenic," but the evidence supporting this classification is insufficient***
- 3. The nomenclature of the variant should be carefully verified***
- 4. Please provide absolute lymphocyte count, CD3/CD4/CD8 counts,***

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CD4/CD8 ratio, B-cell and NK-cell populations, serum IgG/IgA/IgM, vaccine-specific antibody responses where available, and relevant functional immune assays.

- 5. The complex interaction between MHC-I deficiency, NK-cell function, chronic inflammation, and infection susceptibility should be discussed.*
- 6. The authors should report HRCT findings, including distribution, severity, and associated structural abnormalities. If available, pulmonary function testing should also be included. A baseline comparison with previous imaging would strengthen the case.*
- 7. The authors should explain the indication and evidence supporting HSCT in TAP2-associated BLS I. If HSCT is being considered because of severe disease, this should be explicitly stated. The potential benefits, limitations, and reported outcomes of HSCT in BLS I should be discussed.*
- 8. The differential diagnosis should be expanded.*
- 9. Recent literature on TAP deficiency, MHC-I deficiency, BLS I, and HSCT should be incorporated.*
- 10. Define all abbreviations at first use, including BLS, MHC-I, TAP, WES, HRCT, mMRC, HSCT, and HLA.*
- 11. The conclusion should avoid implying that genetic testing alone provides "targeted" treatment. In this disorder, molecular diagnosis primarily facilitates confirmation, genetic counselling, family screening, prognostication, and consideration of definitive management.*