

REVIEWER'S REPORT

Manuscript No.: IJAR-57196**Title: Exploration du déficit immunitaire primitif : démarche diagnostique et état des lieux en Mauritanie,****Recommendation:****Accept after minor revision**

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

Reviewer Name: Abdul Haseeb Mir**Detailed Reviewer's Report**

The research article titled "Exploration of Primary Immunodeficiency: Diagnostic Approach and Current Status in Mauritania" provides a critical and timely examination of a group of inherited immune system disorders that, while individually rare, collectively represent a significant public health challenge in developing nations. The author establishes at the outset that primary immunodeficiencies (PIDs) are characterized by a high degree of clinical heterogeneity, ranging from common recurrent infections to complex, life-threatening multisystemic syndromes. By synthesizing international diagnostic standards with a pragmatic assessment of the medical landscape in Mauritania, the paper addresses the "invisible" burden of these diseases in low-resource settings. The narrative effectively argues that the perceived rarity of PIDs is often a byproduct of systemic underdiagnosis and a lack of clinical awareness rather than a true epidemiological absence.

The narrative begins by defining the immunological framework of PIDs, which arise from genetic defects affecting the development, maintenance, or function of immune cells such as T lymphocytes, B lymphocytes, phagocytes, and the complement system. A central theme of the article is that clinical suspicion remains the most vital tool in the physician's arsenal. The author underscores that early detection is paramount, as delayed diagnosis inevitably leads to cumulative organ damage—particularly in the lungs—or premature mortality. The paper provides a thorough review of the "warning signs" that should trigger an immunological investigation, emphasizing that infections that are unusually severe, persistent, or caused by opportunistic pathogens must be viewed as red flags. This clinical baseline is

REVIEWER'S REPORT

essential for shifting the medical perspective from treating isolated infections to identifying an underlying systemic vulnerability.

In terms of the diagnostic methodology, the study evaluates a tiered approach tailored to the economic and technical constraints of a country like Mauritania. The author describes a "first-line" battery of tests—including the complete blood count (CBC), HIV screening to rule out secondary deficiencies, and serum protein electrophoresis—as accessible and cost-effective measures that can detect a vast majority of humoral and major cellular defects. The analysis then transitions to more specialized investigations, such as lymphocyte phenotyping and functional assays, while candidly acknowledging that these resources are frequently centralized or entirely unavailable in local clinical practice. By documenting this gap, the author provides a rigorous audit of the technical infrastructure required to move beyond symptomatic treatment toward precision medicine.

The evaluation of the specific status of PIDs in Mauritania constitutes the most original and impactful section of the paper. The author identifies several structural barriers that perpetuate the cycle of underdiagnosis. Chief among these is the limited awareness among general practitioners and pediatricians, which often results in patients undergoing a "medical odyssey" through various specialties before an immunodeficiency is ever considered. Furthermore, the absence of a national registry prevents the collection of accurate epidemiological data, making it difficult to advocate for the necessary policy changes or resource allocation. The study also highlights the critical shortage of essential therapies, such as intravenous immunoglobulin (IVIG) and hematopoietic stem cell transplantation, which further compromises the survival rates of children diagnosed with the most severe forms of the disease.

Furthermore, the paper addresses the long-term socioeconomic implications of these health disparities. The author evaluates the cost of reactive care—treating the repeated hospitalizations and chronic complications of an undiagnosed PID—against the cost of proactive management and replacement therapy. The narrative effectively argues that investing in diagnostic capacity and treatment access is not only a humanitarian necessity but an economically sound strategy for the Mauritanian health system. By linking clinical findings with social pediatrics and health policy, the author elevates the discussion from a technical medical review to a compelling call for institutional reform and international cooperation.

In summary, this article offers a profound and necessary inquiry into the realities of managing primary immunodeficiency in a developing context. It successfully bridges the gap between high-level molecular

REVIEWER'S REPORT

immunology and the practical challenges of frontline healthcare. The author's ability to contextualize international guidelines within the Mauritanian setting makes this a significant contribution to the fields of clinical immunology and global health. It is an essential read for pediatricians, policymakers, and researchers dedicated to improving the health outcomes of vulnerable populations in the Global South.

Recommendations for Improving the Article

- The author should consider including specific "case vignettes" from the Mauritanian clinical experience to illustrate the typical diagnostic delays and the clinical diversity of PIDs encountered locally, which would add a more personal and illustrative dimension to the data.
- To enhance the academic rigor of the "Current Status" section, the paper would benefit from a more detailed comparison of Mauritania's diagnostic capabilities with neighboring Maghreb or West African countries, highlighting regional best practices that could be adapted.
- The discussion regarding "limited awareness" would be improved by proposing a specific curriculum or set of "Clinical Decision Support" tools that could be integrated into existing primary care training programs in Mauritania.
- The author should explicitly address the role of consanguinity in the Mauritanian context, as high rates of endogamy in certain populations can significantly increase the prevalence of autosomal recessive PIDs, a factor that warrants specific epidemiological attention.
- It is recommended that the author expand on the potential for "South-South cooperation," exploring how Mauritania might leverage diagnostic laboratories in North Africa to provide genetic testing and advanced phenotyping until local capacities are fully developed.

Recommendation: Recommend for publication with minor revision.