

REVIEWER'S REPORT

Manuscript No.: IJAR-50649

Date: 15-03-2025

Title: Genetic Infertility: Exploring Six Rare Cases

Recommendation:

Accept as it is.....**YES**.....

Accept after minor revision.....

Accept after major revision

Do not accept (*Reasons below*)

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

Reviewer's Name: Dr Aamina

Reviewer's Decision about Paper: **Recommended for Publication.**

Comments (*Use additional pages, if required*)

Reviewer's Comment / Report

Title & Abstract: The title accurately reflects the content of the case report, emphasizing the rare nature of the genetic infertility cases. The abstract provides a concise and clear overview of the topic, presenting relevant statistical data from the WHO and introducing the key genetic syndromes discussed. The mention of specific syndromes provides insight into the scope of the report, ensuring clarity for the reader.

Introduction: The introduction effectively establishes the prevalence and complexity of genetic infertility, highlighting its contribution to reproductive challenges. The statement regarding the misconception that infertility cannot be transmitted, followed by evidence of genetic influences, strengthens the rationale for the study.

Case Reports:

- The structure of the case presentations is well-organized and allows for easy comparison between patients.
- The four Klinefelter syndrome cases are detailed systematically, presenting clinical examination findings, laboratory test results, and karyotype analysis. The descriptions are precise and provide a clear understanding of the phenotypic variations and diagnostic criteria.

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- The Jacob's syndrome case effectively describes characteristic features, including physical traits and sperm analysis, with clear confirmation from karyotyping.
- The Kallmann-de Morsier syndrome case is well-explained, particularly regarding the clinical presentation, imaging findings, and hormonal evaluations. The therapeutic approach leading to a successful pregnancy outcome adds valuable clinical insight.

Discussion:

- The discussion integrates relevant historical and scientific background, citing landmark discoveries and their impact on genetic infertility research.
- Klinefelter syndrome is thoroughly discussed, with references to its discovery, prevalence, and diagnostic markers.
- The explanation of Barr bodies and their role in sex determination is well-articulated and contributes to a deeper understanding of chromosomal analysis.
- The inclusion of WHO's definition of infertility reinforces the clinical significance of the topic.

Overall Quality:

- The report maintains scientific rigor while remaining accessible to a broad audience.
- Terminology and references are appropriate for the target readership.
- The logical flow from introduction to discussion ensures clarity and coherence.

Conclusion: The case report effectively illustrates the genetic underpinnings of infertility, providing valuable clinical insights into the diagnosis and management of Klinefelter syndrome, Jacob's syndrome, and Kallmann-de Morsier syndrome.