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| Book Name: | [**An Overview of Disease and Health Research**](https://bookstore.bookpi.org/product/an-overview-of-disease-and-health-research-vol-1/) |
| Manuscript Number: | **Ms\_BPR\_6029** |
| Title of the Manuscript: | **Case report: dual diagnosis of Mulibrey nanism and Jacobs syndrome in an Indian boy** |
| Type of the Article | **Book Chapter** |

**Special note:**

**A research paper already published in a journal can be published as a Book Chapter in an expanded form with proper copyright approval.**

**Source Article:**

**This chapter is an extended version of the article published by the same author(s) in the following journal.**

**Egyptian Journal of Medical Human Genetics, (2025) 26:59.**

**DOI: https://doi.org/10.1186/s43042-025-00694-8**

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| PART 1: Comments | | |
|  | Reviewer’s comment **Artificial Intelligence (AI) generated or assisted review comments are strictly prohibited during peer review.** | Author’s Feedback *(Please correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)* |
| **Please write a few sentences regarding the importance of this manuscript for the scientific community. A minimum of 3-4 sentences may be required for this part.** | The significance of this case report to the scientific and clinical community results from the following extreme rarity of co-occurrence of Mulibrey nanism and 47,XYY syndrome (Jacob's syndrome). The manuscript reports a rare dual diagnosis not reported earlier in the literature, providing information regarding phenotypic overlap, diagnostic difficulties, and possible clinical management options. Furthermore, this case contributes to broadening the database about syndromic growth disorders and chromosomal defects in children, especially in underrepresented regions such as India. |  |
| **Is the title of the article suitable?**  **(If not please suggest an alternative title)** | Yes, the title is accurate and descriptive. It clearly states the content and novelty of the case.  Suggestion: may like to refine it a little for clarity and flow:  Suggested alternative title (optional):  "Dual diagnosis of Mulibrey nanism and Jacob's syndrome: A rare case report from India" |  |
| Is the abstract of the article comprehensive? Do you suggest the addition (or deletion) of some points in this section? Please write your suggestions here. | The abstract is mostly complete and does a good job of describing the dual diagnosis and the key clinical features.  Suggestions:  • Briefly mention the diagnostic approach adopted (e.g., genetic investigation, imaging).  • Insert one or two words regarding clinical implications or significance for diagnosis and follow-up.  • Specify age at diagnosis and define follow-up period, if data are available. |  |
| **Is the manuscript scientifically, correct? Please write here.** | Yes. The article is scientifically sound and technically robust. The case is well presented and differential diagnosis appropriately considered. There is a logical account of pathophysiology and phenotype-genotype correlation. Some additional explanation on long-term follow-up and management strategy would enhance clinical utility. |  |
| **Are the references sufficient and recent? If you have suggestions of additional references, please mention them in the review form.**  **-** | References are in general good and relevant. A couple of additional recent references on Mulibrey nanism and 47,XYY syndrome (within the last 5 years) would put rarity and management of double diagnosis into perspective.  Recommended additional references:  • Karlberg N. et al. Mulibrey nanism: Clinical features and diagnostic criteria. Horm Res Paediatr. 2019.  • Tartaglia N. et al. 47,XYY syndrome: Updated clinical review. Am J Med Genet C Semin Med Genet. 2020. |  |
| Is the language/English quality of the article suitable for scholarly communications? | Generally, yes. The manuscript is readable and understandable, although there are certain areas where language could be slightly enhanced towards fluency and clarity.  Recommendation: Minor editing for grammar, punctuation, and flow (e.g., sentence structure in the discussion section and consistency of terminologies). There are a few sentences that are too long or redundant. |  |
| Optional/General comments | Might include diagram/table summaries of the phenotypic features of both conditions.  •Emphasize in the discussion if the patient has overlapping, additive, or divergent features of the two syndromes.  •A brief comment on counselling implications (genetic, fertility, psychosocial) would be an addition to the manuscript. |  |

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| **PART 2:** | | |
|  | **Reviewer’s comment** | **Author’s comment** *(if agreed with reviewer, correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)* |
| **Are there ethical issues in this manuscript?** | *(If yes, Kindly please write down the ethical issues here in details)* |  |

**Reviewer details:**

**Yerko P. Ivanovic-Barbeito, Trinity College, Tallaght University Hospital, Ireland**