

REVIEWER'S REPORT

Manuscript No.: IJAR-59148

Title: A CASE REPORT ON PHENYLKETONURIA: EARLY INTERVENTION AND TREATMENT.

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:

- Clinically important metabolic disorder
- Clear and relevant case presentation
- Diagnosis supported by biochemical and genetic findings
- Characteristic clinical features are well described
- Multidisciplinary management is presented
- Longitudinal biochemical and developmental follow up is included
- Treatment response is documented with objective measures
- Limitations are appropriately acknowledged

Weakness of the study:

- Exact PAH variant and zygosity are not reported
- Phenylalanine reference range contains an apparent error
- Detailed dietary management and adherence are not provided
- Follow up timing is incompletely described
- MRI findings need correlation with the original report
- Improvement cannot be attributed to diet alone
- Ethical approval or publication consent is not reported
- Grammar and formatting require substantial editing

Reviewers Comments:

The manuscript presents an interesting late diagnosed case of phenylketonuria with developmental delay, behavioural problems and characteristic clinical features. The biochemical and molecular findings support the diagnosis, and the multidisciplinary management with follow up is a strength. However, ethical clearance and written consent for publication are not reported and should be clearly stated for a case report. Important clinical information also requires clarification, particularly the exact PAH variant and zygosity, the incorrect or unclear phenylalanine reference range, dietary intake and adherence, and the timing of follow up assessments. The reduction in phenylalanine is encouraging, but the final level remains substantially above the recommended target, and clinical improvement cannot be attributed to dietary treatment alone because several rehabilitation interventions were provided. Grammar, sentence construction, spacing and formatting require careful editing. Overall, the case is clinically useful but requires substantial clarification and revision before publication.

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Previously Published anywhere/Plagiarism check:

An exact title search did not identify the same publication. However, several related published case reports and case series on late diagnosed PKU, developmental delay, hypopigmentation, brain MRI findings and PAH variants are available. The present case appears to have a different patient and presentation. A formal similarity check is recommended before publication.