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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\_6030** |
| Title of the Manuscript: | **Unravelling variants in Farber disease: diagnostic and prenatal challenges in atypical presentations** |
| Type of the Article | **Book Chapter** |

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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.** | This manuscript describes a rare presentation of Farber disease involving three siblings with atypical clinical features and novel *ASAH1* variants. It contributes to the clinical and genetic understanding of the disease, including prenatal diagnostic relevance. The case highlights diagnostic challenges, expands the phenotypic spectrum, and has clear value for genetic counseling. It is a meaningful addition to the rare disease literature. |  |
| **Is the title of the article suitable?**  **(If not please suggest an alternative title)** | Suitable and relevant |  |
| 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 informative but does not follow proper case report structure. It currently includes: *Abstract, Case presentation, Conclusion*. Please revise to follow standard format: Background => Case Presentation => Conclusions  Also, please reduce background length and emphasize key clinical findings and genetic relevance more concisely. |  |
| **Is the manuscript scientifically, correct? Please write here.** | Scientifically correct in general, but the following major revisions are needed:  - Incomplete clinical details (e.g., enzyme activity, MRI, growth metrics)  - Genetic analysis lacks ACMG classification, population frequency, in silico scores  - Histopathology section weak (no immunostaining or EM images)  - Discussion lacks differential diagnosis, prognosis, management |  |
| **Are the references sufficient and recent? If you have suggestions of additional references, please mention them in the review form.** | Mostly recent and relevant. However, some important references on FD and variant classification are missing. Please update and ensure citation formatting is consistent. |  |
| Is the language/English quality of the article suitable for scholarly communications? | Understandable but requires professional editing. Grammatical errors (e.g., comma splices, unclear pronouns) and inconsistent nomenclature exist. Recommended to revise for clarity, consistency, and scholarly tone. |  |
| Optional/General comments | Please reformat the manuscript according to the CARE guidelines for case reports. Standard structure should be:   1. Introduction 2. Case Presentation 3. Discussion 4. Conclusion   Add missing figures: family pedigree, histopathology, timeline.  Improve figure quality (e.g., IGV resolution, image annotations). |  |

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| **PART 2:** | | |
|  | **Reviewer’s comment** | **Author’s Feedback** (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:**

**Nguyen Thi Hong Hanh, Vietnam**