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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.** | Authors presented a detailed diagnosis of the condition which involved genetic testing and other examinations based on medical history and presentations. Outcome from this case study would inform best approach to include some genetic testing as well as promote genetic counseling for would-be couples with a greater likelihood of having children with FD. |  |
| **Is the title of the article suitable?****(If not please suggest an alternative title)** | Yes |  |
| 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. | Abstract is comprehensive |  |
| **Is the manuscript scientifically, correct? Please write here.**  | Yes |  |
| **Are the references sufficient and recent? If you have suggestions of additional references, please mention them in the review form.****-** | Yes |  |
| Is the language/English quality of the article suitable for scholarly communications? | Yes |  |
| Optional/General comments | * Abstract

Farber disease - Capitalize “d”* Case presentation

Heterozygous variant of uncertain significance (VUS) – Capitalize each word* Background

Farber disease (Capitalize “d”)FD-Write in fullClassifications of Farber disease Farber disease (Capitalize “d”)Farber disease (FD),we present an atypical case of FD (FD with a comma(FD,)Case presentationMRI- write in fullFarber disease(Capitalize “d”)DiscussionThe atypical presentation of Farber disease (FD)that the diagnosis of Farber disease (FD)- (Capitalize “d”)loss of function (LOF)- Capitalize each wordRT-PCR studies performed on (Write in full)juvenile idiopathic arthritis (JIA)- Capitalize each wordASAH1: c.1042-2A > C (Intron 12)variant of uncertain significance (VUS) - Capitalize each wordConclusionFarber disease- (Capitalize “d”)* **Abbreviations**

FD Farber disease (Capitalize “d”)VUS Variants of uncertain significance – Capitalize each wordESR Erythrocyte sedimentation rate- you may have to delete, cannot be found in your write-up |  |

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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:**

**Richcane Amankwa, Ghana**