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Information about the contributions of each person named as having participated in the study

- 1) **Guarantor(s)**, i.e., person(s) who is (are) responsible for the integrity of the work as a whole:
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2) Authors who participated in the **conception of the study**: Francisco Vidal, Irene Corrales, Rafael Parra and Carme Altisent.

3) **Design & Methods**. The following authors were responsible for specific investigations (please detail):

- Francisco Vidal, was the principal investigator, coordinated the research, created the database and takes primary responsibility for the paper.
- Irene Corrales, performed the complete sequencing of patients and the STR analysis, designed the family trees and compiled all data available in the database.
- Lorena Ramírez, participated in the laboratory work for this study.
- Júlia Ayats, Carme Altisent and Rafael Parra diagnosed and recruited the patients and clinical data.

4) **Results**. The following authors were responsible for specific portions of the results, including figures and tables (please indicate the person responsible for each figure and each table):

- Francisco Vidal and Irene Corrales were responsible for all figures and tables.
- Lorena Ramírez, Júlia Ayats, Carme Altisent and Rafael Parra were responsible for the diagnosis and laboratory test results of the patients (Table 1.)

5) **Writing the manuscript**. The following authors were responsible for writing the manuscript:

- Francisco Vidal and Irene Corrales were responsible for writing the manuscript.
- Lorena Ramírez, Júlia Ayats, Carme Altisent and Rafael Parra were responsible for the revision of the drafting and the approval of the final version of the manuscript.

6) **Contributors Listed in Acknowledgments**:

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