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

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PAM designed the study, performed part of the genetic study, gathered data information and wrote the paper. BG, SC and EC carried out part of the molecular studies and approved the final version of the manuscript. FB, MN, KL and JFS examined patients, gathered clinical information, were in charge of the follow-up and approved the final version of the manuscript. JR performed part of the molecular studies, carried out the bioinformatics analysis and wrote the paper.

2) Authors who participated in the conception of the study: P. Aguilar Martinez and J. Rochette were responsible for the conception of the study.

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

- P Aguilar Martinez was responsible for design and method of the study and for carrying out part of the molecular studies.
- B Grandchamp was responsible for carrying out part of the molecular studies and approved the final version of the manuscript.
- S Cunat was responsible for carrying out part of the molecular studies and approved the final version of the manuscript.
- E Cadet was responsible for carrying out part of the molecular studies and approved the final version of the manuscript.
- F Blanc was responsible for the examination of patients, for gathering clinical information, was in charge of the follow-up and approved the final version of the manuscript.
- M Nourrit was responsible for the examination of patients, for gathering clinical information, was in charge of the follow-up and approved the final version of the manuscript.
- K Lassoued was responsible for the examination of patients, for gathering clinical information, was in charge of the follow-up and approved the final version of the manuscript.
- JF Schved was responsible for the examination of patients, for gathering clinical information, was in charge of the follow-up and approved the final version of the manuscript.

- J Rochette was responsible for performing part of the molecular studies and for carrying out the bioinformatics analysis.

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):

- P Aguilar Martinez and J Rochette were responsible for gathering and synthesizing the results.
- P Aguilar Martinez was responsible for drafting tables 1, 2 and 3, which were subsequently completed by J Rochette, E Cadet and B Grandchamp.
- S Cunat and P Aguilar Martinez were responsible for drawing figure 1.

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

- P Aguilar Martinez and J Rochette were responsible for writing the manuscript.

6) **Contributors Listed in Acknowledgments:** (Examples of those who might be acknowledged include a person who provided purely technical help, writing assistance, or a department chair who provided only general support. Corresponding authors must declare whether they had assistance with the study design, data collection, data analysis, or manuscript preparation. If such assistance was available, the authors must disclose the identity of the individuals who provided this assistance and the entity that supported it. Financial and material support should also be acknowledged.)

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