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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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According to the International Committee of Medical Journal Editors (ICMJE) (http://www.icmje.org/ethical_1author.html): “*Authorship credit should be based on: 1) substantial contributions to conception and design, acquisition of data, or analysis and interpretation of data; 2) drafting the article or revising it critically for important intellectual content; and 3) final approval of the version to be published. Authors should meet conditions 1, 2, and 3 Acquisition of funding, collection of data, or general supervision of the research group alone does not constitute authorship*”.

The guarantor of this manuscript confirms that all persons designated as authors qualify for authorship, and that each author has participated sufficiently in the work to take public responsibility for appropriate portions of the content.

2) Authors who participated in the conception of the study: Carole Beaumont, Bernard Grandchamp, Mayka Sanchez and Alison May

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

- Carole Beaumont, Bernard Grandchamp, Mayka Sanchez and Alison May were responsible for methodologies design.
- Jose L Fuster Soler, Karim Maloum, Thomas Matthes, Caroline Oudot, Axelle Lascaux, Corinne Pondarré, Julian Sevilla Navarro, Sudharma Vidyatilake were involved in the clinical care of the patients and were responsible for making the provisional diagnosis, obtaining the required blood samples, providing relevant clinical information and for arranging appropriate family studies.
- Caroline Kannengiesser, Mayka Sanchez, Marion Sweeney, Gilles Hetet, Briedgeen Kerr, Erica Moran, Alison May were responsible for performing the molecular diagnosis and additional required haematological or biochemical tests.
- Caroline Kannengiesser, Mayka Sanchez, Marion Sweeney, Gilles Hetet, Briedgeen Kerr, Erica Moran, Bernard Grandchamp, Carole Beaumont and Alison May were responsible for interpretation of the results.
- Mayka Sanchez and Alison May were responsible for the protein 3-D structure modelling
- Bernard Grandchamp, Caroline Kannengiesser, Mayka Sanchez and Alison May were responsible for reviewing their individual patient databases for additional contextual information

4) Results. The following authors were responsible for specific portions of the results, including figures and tables:

- Mayka Sanchez and Erica Moran were responsible for Figure 1
- Alison May was responsible for Figure 2A
- Mayka Sanchez was responsible for Figure S2 in the on line supplementary appendix (Figure 2B in the original submission)
- Alison May was responsible for Figure 2B in the print version
- Alison May was responsible for Figure S1 in the on line supplementary appendix

- Caroline Kannengiesser, Bernard Grandchamp, Mayka Sanchez, Erica Moran and Alison May were responsible for Table S1 in the supplementary on line appendix.
- Caroline Kannengiesser, Mayka Sanchez, Jose L Fuster Soler, Karim Maloum, Thomas Matthes, Caroline Oudot, Axelle Lascaux, Corinne Pondarré, Julian Sevilla Navarro, Sudharma Vidyatilake, Bernard Grandchamp and Alison May were responsible for Table 1. Bernard Grandchamp was responsible the first draft and Alison May was responsible for the final version of Table 1.

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

- Bernard Grandchamp, Alison May and Mayka Sanchez drafted the manuscript.
- All authors (Caroline Kannengiesser, Mayka Sanchez, Marion Sweeney, Gilles Hetet, Briedgeen Kerr, Erica Moran, Jose L Fuster Soler, Karim Maloum, Thomas Matthes, Caroline Oudot, Axelle Lascaux, Corinne Pondarré, Julian Sevilla Navarro, Sudharma Vidyatilake, Carole Beaumont, Bernard Grandchamp, Alison May) were responsible for critical revision of the manuscript, completion of the manuscript and its approval.

6) **Contributors Listed in Acknowledgments:**

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