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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: Inderjeet Dokal, Tom Vulliamy, Amanda Walne

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

- Amanda Walne and Tom Vulliamy contributed to the design of all experimental work
- Richard Beswick, Vincent Plagnol, Michael Kirwan and Arran Dokal were responsible for contributing to the design of specific experimental details within the project

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):

- Arran Dokal and Amanda Walne were responsible for screening the patient samples and the results presented within figure 1
- Amanda Walne and Inderjeet Dokal were responsible for the presentation of the data in table 1
- Vincent Plagnol was responsible for the exome sequencing pipeline analysis
- Richard Beswick was responsible for figure 2
- Josu de la Fuente was responsible for acquisition of some of the patient material and clinical information

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

- Amanda Walne and Inderjeet Dokal had overall responsibility for drafting and editing the manuscript
- Vincent Plagnol, Arran Dokal, Josu de la Fuente and Richard Beswick were responsible for editing specific sections of the manuscript relating to their contribution and approval of the final version
- Tom Vulliamy and Michael Kirwan were responsible for critical review of the manuscript

6) Contributors Listed in Acknowledgments:

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