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

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**2) Authors who participated in the conception of the study: Amanda Walne, Tanya Bhagat, Tom Vulliamy, Inderjeet Dokal**

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

- Amanda Walne and Tanya Bhagat were responsible for the genetic analysis
- Michael Kirwan and Tom Vulliamy were responsible for the telomere length measurements
- Inderjeet Dokal, Cyril Gitiaux, Isabelle Desguerre, Norma Leonard, Elena Nogales /were responsible for collecting patient samples and providing clinical data
- Tom Vulliamy was responsible for the co-ordination of the research

**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):**

- Amanda Walne and Tanya Bhagat were responsible for the identification of CTC1 mutations and the generation of figure 1
- Michael Kirwan and Tom Vulliamy were responsible for telomere length measurements and the generation of figure 2
- Inderjeet Dokal was responsible for table 1

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

- Amanda Walne, Tom Vulliamy and Inderjeet Dokal were responsible for the writing of 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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