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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**: Giovanna Graziadei and Maria Domenica Cappellini

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

- Giovanna Graziadei, Wilma Barcellini, Elena Cassinero were responsible for clinical diagnosis and patient follow up.
- Claudia Cesaretti was responsible for the genetic counseling.
- Chiara Refaldi was responsible for the molecular 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):

- Giovanna Graziadei, Khaled M. Musallam and Maria Domenica Cappellini prepared the Results text, tables and figures.

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

- Giovanna Graziadei and Khaled M. Musallam wrote the manuscript.
- Chiara Refaldi, Wilma Barcellini, Claudia Cesaretti, Elena Cassinero, Maria Domenica Cappellini revised the manuscript critically for intellectual content.

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