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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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