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3) **Design & Methods**. The following authors were responsible for specific investigations
• Marc Fouassier, Catherine Ternisien, Armelle Lefrancois were responsible for the several patients consultations to inform them about the pathology and its heredity, pick their consents for molecular studies and for publication, carry out the biological phenotypes of the young girl and her parents,.
• Pierre Boisseau, Mathilde Giraud, Stéphane Bezieau were responsible for the molecular genetics studies : sequencing of the coding region of the gene and the haplotype study
• Agnès Veyradier, Edith Fressinaud were responsible for specific analyses of the patients phenotype

4) **Results**. The following authors were responsible for specific portions of the results, including figures and tables:
• Marc Fouassier was responsible for table 1
• Pierre Boisseau was responsible for figure 1

5) **Writing the manuscript**. The following authors were responsible for writing the manuscript:
• Marc Fouassier, Catherine Ternisien, Armelle Lefrancois were responsible for the introduction, description on biological phenotype and conclusion
• Pierre Boisseau, Mathilde Giraud, Stéphane Bezieau were responsible for molecular results and conclusion
• Agnès Veyradier and Edith Fressinaud were responsible for the title, notices and conclusion

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