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

   • SC, EFW, KBi, MGS, and AB cared for the patients, had clinical, therapeutic, and/or diagnostic responsibilities
   • MGS arranged and E5, ESV, MS and KBo performed homozygosity mapping, whole exome sequencing and bioinformatics analyses in family A
   • SD, KBI, and MG performed exome sequencing and bioinformatic analyses in families B & C
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4) Results. The following authors were responsible for specific portions of the results, including figures and tables:

- MGS, SC, and KBi prepared the table
- ES and KBo prepared the figures 1, S4;
- ES, SD, KBi, and KBo prepared figures S1, S2, S3
- see also “3) Design and Methods” for specific portions of the experiments/results
- MGS, KBi, AB, and KBo coordinated the multicentre cooperation to finalize comprehensive data analyses and manuscript preparation

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

- MGS, KBo, ES, KBi, SC, and AB prepared the manuscript
- SC edited the manuscript

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