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Biallelic IARS2 mutations presenting as sideroblastic anemia

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Contributions: GB performed molecular researchs, data analysis and wrote the manuscript. AM supervised this study, performed clinical evaluation, and wrote the manuscript. BR performed data analysis. ZA and CB performed the molecular study. DP, AP, CO, ID performed clinical follow-up. CJR and NB performed neuro-imaging analysis. DRT, DB, JPB, AR, JS performed data analysis and supervised the study.