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The phenotypic spectrum of germline YARS2 variants: from isolated sideroblastic anemia to mitochondrial myopathy, lactic acidosis and sideroblastic anemia 2

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Contributions: G.A.H., L.M.H., J.A.K., J.L.K., C.A.S., D.P.S. R.A., E.B., M.A., R.F., S.S.B, L.A. and R.T. ascertained and phenotyped patients. M.M.H. and R.Z. collated and analyzed the clinical data. M.M.H and M.D.F. recruited and phenotyped families 1-3, and 5-9, which were genotyped by D.R.C. L.G.R performed in silico analysis of variants, cloning of missense variants and wrote the initial draft of the manuscript. J.R.T. and M.F. performed amino-acylation assays and protein modeling. J.F. and M.S. performed genotyping and/or data analysis for patient 10. A.R., A.S., N.S., R.J.R., T.K. and D.W.S. ascertained, phenotyped, and genotyped patient 4. M.D.F and J.C coordinated all aspects of the work and wrote the final manuscript. All authors contributed to the preparation of the manuscript.