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Recurrent heteroplasmy for the MT-ATP6 p.Ser148Asn (m.8969G>A) mutation in patients with syndromic congenital sideroblastic anemia of variable clinical severity

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Contributions: V.R., J.H., and E.C. ascertained patients and together with M.M.P., D.L.R., S.S.B., M.D.F., M.M.H. performed in-depth phenotyping. R.W.A.P. performed urine organic acid assays. D.R.C. performed and analyzed MT-DNA Sanger sequencing. J.F.T., E.J.W., and T.R. performed MT-DNA heteroplasmy analysis. S.B. assembled and analyzed all the data and wrote the first draft of the manuscript. M.D.F. oversaw all the work and completed the manuscript with the review of the other authors.