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A GENETIC WEAKNESS—PHOENICIAN LEGACY OR CELTIC HERITAGE?

Yi Ng, Charlotte Alston, Rita Horvath, Maria Farrugia, Patrick Chinnery, Douglass Turnbull, Robert Taylor, Robert McFarland, Andrew Schaefer, Grainne Gorman. *Newcastle University; Southern General Hospital, Glasgow*

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Mitochondrial diseases are increasingly recognised as common genetic disorders exhibiting prodigious genotypic and phenotypic heterogeneity. Given that all mammalian cells contain mitochondrial DNA except mature red blood cells, it is not surprising that manifestations of the disorder are frequently multi systemic. Whilst brain, skeletal muscle, and cardiac symptoms are recognised prominent manifestations of mitochondrial disorders, other non-mitotic tissues maybe similarly affected. MLASA is a rare but distinct clinical entity characterised by myopathy, lactic acidosis and sideroblastic anaemia; originally attributed to mutations in the PUS1 (pseudouridylate synthase 1) gene. Mutations in the YAR2 (tyrosyl-tRNA synthetase 2; mitochondrial) gene, have recently been implicated in eight patients with MLASA (7 Lebanese and one of French ethnic origin). We present three unrelated adults, with novel mutations in YAR2 gene that challenge the 'Lebanese founder effect' and acronym, synonymous with this form of mitochondrial disease.