World Academy of Science, Engineering and Technology
International Journal of Medical and Health Sciences
Vol:17, No:03, 2023

# LYRM7-Associated Mitochondrial Complex III Deficiency with Non-Cavitating Leukoencephalopathy and Stroke-Like Episodes

**Authors :** Rita Alfattal, Maryam Alfarhan, Adeeb M. Algaith, Buthaina Albash, Reem M. Elshafie, Asma Alshammari, Ahmad Alahmad, Fatima Dashti, Rasha Alsafi, Hind Alsharhan

**Abstract :** Defects of respiratory chain complex III (CIII) result in characteristic but rare mitochondrial disorders associated with distinct neuroradiological findings. The underlying molecular defects affecting mitochondrial CIII assembly factors are few and yet to be identified. LYRM7 assembly factor is required for proper CIII assembly where it acts as a chaperone for the Rieske iron-sulfur (UQCRFS1) protein in the mitochondrial matrix and stabilizing it. We present here the seventeenth individual with LYRM7-associated mitochondrial leukoencephalopathy harboring a previously reported rare pathogenic homozygous LYRM 7 variant, c.2T>C, (p.Met1?). Like previously reported individuals, our 4-year-old male proband presented with recurrent metabolic and lactic acidosis, encephalopathy, and myopathy. Further, he has additional, previously unreported features, including an acute stroke like episode with bilateral central blindness and optic neuropathy, recurrent hyperglycemia and hypertension associated with metabolic crisis. However, he has no signs of psychomotor regression. He has been stable clinically with residual left-sided reduced visual acuity and amblyopia, and no more metabolic crises for 2-year-period while on the mitochondrial cocktail. Although the reported brain MRI findings in other affected individuals are homogenous, it is slightly different in our index, revealing evidence of bilateral almost symmetric multifocal periventricular T2 hyperintensities with hyperintensities of the optic nerves, optic chiasm, and corona radiata but with no cavitation or cystic changes. This report describes new clinical and radiological findings of LYRM7-associated disease. The report also summarizes the clinical and molecular data of previously reported individuals describing the full phenotypic spectrum.

**Keywords :** LYRM7 gene defect, mitochondrial disease, ,lactic acidosis, ,genetic disorder
**Conference Title :** ICP 2023 : International Conference on Pediatrics
**Conference Location :** London, United Kingdom
**Conference Dates :** March 16-17, 2023