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Cardiac involvement in mitochondrial myopathy, encephalopathy, lactic acidosis and stroke

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Mitochondrial disorders are genetic disorders that result in dysfunction in the energy metabolism pathway. The prevalence of mitochondrial disease is estimated at 9-16 per 100,000 with the most common type being Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis and Stroke (MELAS). A 61-year-old female has presented to our facility on numerous occasions over the last 10-year period, with thorough inpatient and outpatient investigations. She was diagnosed with mitochondrial myopathy, encephalopathy, lactic acidosis and stroke syndrome in 2015 after 6-7 years of a constellation of symptoms including muscle weakness, ataxia, fatigue, headaches and sensorineural hearing loss. Her other past medical history includes an initial diagnosis of hypertrophic cardiomyopathy (with an IVDd of 19 mm), chronic kidney disease and recurrent bowel obstruction. During the last few years there were concerns that her heart function was worsening and hence the patient was arranged for a right ventricular cardiac biopsy. The tissue histopathology slides showed hypertrophied myocytes with diffuse vacuolization but no evidence of myocyte disarray. This is most consistent with mitochondrial cardiomyopathy. This case presents a patient with mitochondrial cardiomyopathy that has possibly presented itself initially as a phenocopy of HCM. Studies have also shown that cardiac involvement possibly stems from mitochondrial dysfunction with hypertrophy present in the early remodeling stage, with progression to dilated cardiomyopathy. This link has already been hypothesized in our patient and she is currently being monitored for further progression of her potential mitochondrial cardiomyopathy.

Biography

Shaun Khanna has completed his MBBS from Bond University and is currently pursuing his Master of Medicine at University of Sydney, Australia. He is also working at Blacktown Hospital as a Medical Officer and as a Conjoint Lecturer with the University of Western Sydney. His research interest is in arrhythmias and cardiovascular imaging.

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