2772 NEW DIAGNOSIS OF MITOCHONDRIAL ENCEPHALOPATHY, LACTIC ACIDOSIS, AND STROKE-LIKE EPISODES (MELAS) PRESENTING AS BILATERAL CEREBRAL INFARCTS

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Background Mitochondrial disorders can manifest in stroke-like episodes which can pose several diagnostic and management challenges in the acute setting.

Presentation We present the case of a 45-year-old female, presenting with bilateral cerebral infarcts on a background of cerebral atrophy, cognitive decline, ataxia, sensorineural hearing loss, peripheral neuropathy, Type 1 Diabetes mellitus, and androgen insensitivity syndrome with investigations revealing a new diagnosis of Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes (MELAS).

Management and Outcomes Initial diagnostic investigations revealed an elevated serum and cerebrospinal fluid lactate, and MRI Brain revealed bilateral cerebral infarcts. Genetic testing on blood confirmed the presence of the m.3243A>G pathogenic variant in the mitochondrial gene *MT-TL1*, one of the common genetic mutations causing MELAS in the caucasian population. The heteroplasmy level on the sample was 24.6%. Further cascade testing identified several other affected family members. Ultimately, the patient was managed with oral arginine and subsequently oral taurine for secondary stroke prevention.

Discussion This case demonstrates the importance of recognising mitochondrial disorders in atypical stroke presentations as the acute and long-term treatments can vary from standard stroke care. While treatment with arginine therapy acutely is established in management of stroke-like presentations in MELAS, taurine therapy has also demonstrated potential reduction in stroke-like events in this patient group.[1 2] Making an accurate diagnosis is important in guiding long term management, family counselling and identifying relatives at risk who would benefit from early arginine treatment when presenting with stroke-like episodes, and screening for associated co-morbidities.

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2773 NOVEL *BAG3* VARIANT IDENTIFIED IN A CASE OF ADULT-ONSET MYOFIBRILLAR MYOPATHY

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Objectives Myofibrillary myopathies (MFM) are a heterogenous group of disorders characterised by myofibrillar dissolution and accumulation of protein degradation products in myofibres. *BCL2-associated anthanogene 3* (*BAG3*)-related MFM has been reported to present with sensorimotor neuropathy and cardiomyopathy. We report a case of a 54-year-old man with a progressive sensorimotor axonal neuropathy with respiratory involvement who was found to have a novel *BAG3* variant.

Case A 54-year-old man presented with a six-month history of increasing dyspnoea with severe restrictive ventilatory dysfunction evident on pulmonary function testing. He has a five-year history of progressive sensorimotor disturbance in his limbs with muscle cramps and gait impairment leaving him wheelchair-bound. Examination revealed marked wasting and florid fasciculations in the upper and lower limbs, distal greater than proximal weakness, absent reflexes and a stocking-and-glove distribution sensory disturbance. Electrodiagnostic studies confirmed a severe length-dependent sensorimotor axonal neuropathy with active and chronic denervation on electromyography. Singleton whole exome sequencing identified a heterozygous missense variant of unknown significance in *BAG3* [NM_004281.3:c.625C>G, p.(Pro209Ala)]. Other variants in *BAG3* at the same amino acid residue [p.(Pro209Leu) and p.(Pro209Gln)] have been reported to be associated with *BAG3*-related MFM. Subsequent muscle biopsy demonstrated chronic denervation and reinnervation of myofibres with pseudodystrophic changes, suggestive of an underlying myofibrillar myopathy. He was managed supportively with nocturnal bilevel positive airway pressure and discharged into supported accommodation.

Conclusion This is a case of adult-onset myofibrillary myopathy suspected to be due to a novel missense *BAG3* variant located in a previously reported *BAG3* mutation hotspot (Proline 209).

2774 PLANNED DOSE REDUCTION OF OCRELIZUMAB IN RELAPSING-REMITTING MULTIPLE SCLEROSIS: A SINGLE CENTRE OBSERVATIONAL STUDY

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