myositis panel. Needle EMG did not show myopathic features and MRI of the quadriceps was normal. FDG-PET scan showed increased uptake in lower limb muscles. A muscle biopsy demonstrated a necrotizing myopathy with dystrophic features. Subsequent genetic panel revealed a FKRP gene mutation.

The diagnoses of LGMD Type 2I with necrotising myopathy was made. IVIG and prednisolone was initiated with a reduction in CK to 2500. Weaning of prednisolone however resulted in a rise in CK. Azathioprine was added to the inmunotherapy with subsequent improvement in clinical signs and CK to <1000.

Conclusion The current case illustrates that FKRP gene mutations causing a genetic myopathy, can be associated with an inflammatory component that may be amenable to immunotherapy treatment.

REFERENCE

1. Svahn J, Streichenberger N, Benveniste O, Menassa R, Michel L, Fayolle H, Petiot P. Significant response to immune therapies in a case of subacute necrotizing myopathy and FKRP mutations. *Neuromuscul Disord*. 2015 Nov;**25**(11):865–8

2835 SPONTANEOUS LATE ONSET NEMALINE MYOPATHY (SLONM) AND ITS DIAGNOSTIC CHALLENGE

Andrew Clarke*, Antony Winkel. *Sunshine Coast University Hospital, Caloundra, QLD, Australia*

10.1136/bmjno-2023-ANZAN.170

SLONM is an exceedingly rare disease, with often long delays in diagnosis and severe functional disability. Approximately half of patients have an associated monoclonal gammopathy of unknown significance (MGUS). Immunotherapy can be restorative for some patients but often requires second or third line therapies.

A 68 years old lady presented with a 6 month history of progressive hip girdle pain, altered gait and difficulty climbing stairs. On examination she had a lower limb isolated proximal myopathy. She had no toxic exposures nor relevant family history. Her extensive blood work up was remarkable only for a low-level IgG lambda paraprotein of 3g/l in keeping with MGUS.

Her neurophysiology demonstrated myopathic changes of the iliopsoas and quadriceps. Her MRI demonstrated oedema with some fatty atrophy of hip girdles bilaterally. The muscle biopsy of left iliopsoas demonstrated non-specific changes of denervation, myopathy and a small degree of inflammatory infiltrate. She underwent Facio-Scapulo-Humeral-Dystrophy (FSHD) testing which revealed a false positive due to genetic rearrangement which delayed diagnosis. A muscular panel was non-contributory. She trialled immunoglobulin, corticosteroids and mycophenolate without benefit.

She continued to clinically deteriorate and at the 24 month mark now had head drop, camptocormia, bulbar and upper limb weakness. She underwent repeat muscle biopsy which revealed myopathic changes with nemaline bodies suggestive of SLONM. She is currently undergoing escalation in immunotherapy after myeloma was diagnosed on bone marrow aspirate.

This case highlights the challenges in making a diagnosis of SLONM, the need for repeat investigations, associated hematological conditions and immunotherapy considerations.

2837 A NOVEL PATHOGENIC MUTATION ASSOCIATED WITH CEREBROTENDINOUS XANTHOMATOSIS

[1]Aaron Gaekwad*, [2,3]Matthew Kiernan, [1,2]William Huynh. *[1]Prince of Wales Hospital, Sydney, Sydney, NSW, Australia*; *[2]Brain and Mind Centre, University of Sydney, Sydney, NSW, Australia*; *[3]Royal Prince Alfred Hospital, Sydney, NSW, Australia*

10.1136/bmjno-2023-ANZAN.171

Objectives Cerebrotendinous Xanthomatosis (CTX) is a rare autosomal recessive disorder of bile acid synthesis and is associated with an abnormally high concentration of cholestanol in the blood, neural tissues and tendons that lead to infiltrative pathology.[1]

Methods A case report.

Results We present a case of a 57-year-old female with a 25-year history of slowly progressive spastic paraparesis. MRI of the neural axis was unremarkable with CSF studies and peripheral neurophysiological studies normal. Transcranial magnetic stimulation showed inexcitable motor corticies bilaterally. An initial clinical diagnosis of primary lateral sclerosis (PLS) or gene-negative hereditary spastic paraparesis was made and treated symptomatically. At 5 years follow-up, the patient had minimal neurological progression and developed right ankle pain with swelling. MRI of the ankle revealed xanthoma infiltration into the Achilles tendon. CTX was considered and genetic testing revealed a homozygous missense variant in exon 5 of the CYP27A1 gene that was deemed novel but considered a variant of uncertain significance (VUS). Phenotyping of this variant with further urine and serum studies demonstrated elevation in cholestanol levels.

Conclusion The diagnosis was thus revised to spinal CTX from a novel pathogenic mutation and the patient was commenced on oral chenodeoxycholic acid with subsequent improvements in biochemical profile.

REFERENCE

1. Wong JC, Walsh K, Hayden D, Eichler FS. Natural history of neurological abnormalities in cerebrotendinous xanthomatosis. *J Inherit Metab Dis.* 2018 Jul;**41**(4):647–656

2838 GFAP ASTROCYTOPATHY MIMICKING AN INFECTIVE MENINGOMYELOENCEPHALITIS

Aaron Gaekwad*, Mark Taylor, Michal Lubomski. *Prince of Wales Hospital, Sydney, Sydney, NSW, Australia*

10.1136/bmjno-2023-ANZAN.172

Objectives To highlight a rare mimic of an infective meningomyeloencephalitis.

Methods A case report.

Results A 44-year-old male presented with intractable fevers and headache. There is no significant past medical or drug history or regular medication use. He reported nausea, fevers and malaise over 2 days. Severe bifrontal headaches persisted for 5 days, whilst urinary hesitancy and difficulty urinating coupled with progressive lower limb weakness developed over the proceeding days of admission. He remained febrile for multiple days despite regular antipyretic use and subsequently became confused and somnolent followed by developing hallucinations, acute urinary retention and increasing pyramidal lower limb signs. Investigations revealed a CRP of 4 with a normal urinalysis and normal CT Brain. CSF studies showed a raised protein of 1.94g/L and a lymphocytic predominant