Abstracts

111 RECURRENT HEADACHES WITH PSYCHOSIS, CSF LYMPHOCYTOSES, VESSEL BEADING AND PAPILLOEDEMA-AUTOIMMUNE/VIRAL ENCEPHALITIS WITH VASCULAR ABNORMALITY OR UNUSUAL PRESENTATION OF REVERSIBLE CEREBRAL VASOCONSTRICTION SYNDROME (RCVS)?

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Introduction Headache is a common Neurology presentation in both outpatient and ED settings. We present a challenging headache in a previously asymptomatic young female patient.

Case A 27 year old female patient presented to ED with severe headache, vomiting and photophobia, after multiple presentations elsewhere, diagnosed as migraine, with normal neurological examination and MRI. Episodes of excruciating headache were associated with writhing and vomiting but spontaneously resolved with residual background pain. Indomethinac helped but she re-presented with headache and psychosis requiring intubation and ICU admission.

Investigations included CSF - protein 0.85, leucocytosis 58 cells (lymphocytic) and normal cerebral venography. Ayclovir was commenced but viral PCR results were negative and she was extubated. MRI was suspicious for sub posterir sulcal hyperintensity and beading in occipital and posterior cerebral arteries.

Another episode occurred ten days later requiring intubation, examination showed papilloedema. CSF examination showed raised pressure, normal protein and 48 lymphocytes; flow cytometry and cytology were unrevealing. NMDA receptor antibody returned positive in serum and CSF. Subtle beading in right PCA branches remained.

Conclusion We present an interesting case of recurrent acute headaches with intracranial hypertension, psychosis and CSF lymphocytosis. The recurrent headaches and vessel beading suggest RCVS, perhaps triggered by viral or autoimmune encephalitis. The NMDA receptor antibody result should be interpreted with caution given the absence of antibody in CSF and dramatic recovery.

112 AXONAL POLYNEUROPATHY WITH ONSET IN YOUNG ADULTHOOD DUE TO TUBB3 MUTATION

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Introduction The TUBB3 gene encodes the protein Beta-tubulin isotype III, a component of the microtubule cytoskeleton. Mutations in this gene have been associated with axonal polyneuropathy, however usually associated with congenital fibrosis of the extraocular muscles (CFOEM) and other abnormalities of cerebral development. 1 2 We report a case of isolated neuropathy associated with a TUBB3 mutation.

Methods Case report - clinical information and next generation sequencing results were obtained.

Results A 64 year old man presented with a severe, progressive, length dependent sensorimotor polyneuropathy which commenced in his late twenties. There was no clinical involvement of the extraocular muscles and cognition was normal. Family history was limited, but there were no other members affected.

The patient had previously been extensively investigated including sural nerve biopsy, which confirmed axonal neuropathy without a specific diagnosis. Intravenous immunoglobulins and steroids had been trialled without benefit.

A neuromuscular gene panel utilising next generation sequencing was performed and demonstrated heterozygosity for a variant of the TUBB3 gene (D417N substitution).

Case series describing TUBB3 mutations show a large heterogeneity in phenotypic expression depending on the amino acid substitution. 2 3 4 There is also heterogeneity in patients with D417N mutations, although a small number have been reported to develop a polyneuropathy without CFOEM. 5

Conclusions This case strengthens previous reports that TUBB3 mutation can be associated with a pure, axonal, sensorimotor polyneuropathy and highlights the use of next generation sequencing in streamlining the diagnostic process.

REFERENCE


113 A CASE OF SUSPECTED AUTOIMMUNE ENCEPHALITIS SECONDARY TO NIVOLUMAB

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Objective To describe a case of suspected immune encephalitis following nivolumab for metastatic melanoma.

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