Case Competition

Fri 8 May, 09:00 - 10:00

Hall 1

1. When the Cord Tells a Tera-able Tale: **Callum Wood**
2. When Leptomeningeal Enhancement Defies Diagnosis – A rare case with diagnostic challenges: **Indrachapa Ranasinghe**
3. Novel neuromuscular hyperexcitability (clinical percussion myotonia) and hypophysitis associated with immune-checkpoint inhibitor therapy: **Shweta Hota**
4. A Rare Cause of Exertional Headaches in a Young Man Under Pressure: **Sai Ambati**
5. Sudden Silence: **Bhavana Atanasova**
6. Acute Respiratory Failure with Rapidly Progressive Muscle Contractures - A Complex Multisystem Case: **Alayna Batson**

When the cord tells a tera-able tale

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A 35-year-old woman with no significant past medical history presented with a 3-day history of viral symptoms including sore throat, coryza, headache, photophobia and vomiting. She was treated with intravenous fluids and discharged after 48 hours. Four days later, she re-presented with upper abdominal pain followed by progressive lower limb weakness, constipation and urinary retention. Her neurological status rapidly deteriorated with reduced consciousness, involuntary facial grimacing, tongue protrusion and myoclonus of the head, trunk and upper limbs, culminating in a generalised tonic–clonic seizure. Blood tests demonstrated an inflammatory response (white cell count 14×10^9/L, CRP 51mg/L). Cerebrospinal fluid analysis revealed marked lymphocytic pleocytosis (259 cells/µL, 100% lymphocytes) and elevated protein (1.74 g/L). CT brain imaging was unremarkable. MRI brain and spinal cord with contrast on day 21 showed normal brain appearances but extensive longitudinally extensive T2 hyperintensity from C7 to the conus with cord swelling and diffuse leptomeningeal enhancement. Cerebrospinal fluid demonstrated glial fibrillary acidic protein (GFAP) antibodies. Pelvic ultrasound later identified a left ovarian teratoma, which was subsequently surgically excised. B cells cultured from teratoma sections produced GFAP antibodies. High-dose steroids, followed by an oral prednisolone taper, was associated with gradual but incomplete recovery in cognition and limb strength.

When Leptomeningeal enhancement defies diagnosis – A rare case with diagnostic challenges

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Background: Primary diffuse leptomeningeal gliomatosis (PDLG) is a rare, aggressive neoplasm characterized by diffuse leptomeningeal infiltration of the subarachnoid space by glial tumor cells in the absence of intra-axial glioma. Although exceedingly rare, it represents an important differential diagnosis for extensive leptomeningeal disease.

History and examination: A 50-year-old man presented with a three-month history of neck pain, progressive upper-limb weakness, and gait unsteadiness. He later developed diplopia, headache, and focal seizures.

Examination revealed ophthalmoplegia, mild left proximal arm and leg weakness, brisk reflexes, and extensor plantar responses.

Investigations: MRI of the spine and brain demonstrated florid, diffuse leptomeningeal enhancement with nodularity.

CSF opening pressure was 29 cmH_2O, protein 8.6 g/L, 55 lymphocytes, and 19 polymorphs. Cytology was negative on three separate occasions. A left frontal dural and parenchymal biopsy demonstrated chronic inflammation with reactive gliosis, without neoplastic cells and was reported as suggestive of neurosarcoidosis. Extensive investigations for infectious and inflammatory leptomeningitis were negative.

Despite extensive immunotherapy for a presumed inflammatory condition, the patient deteriorated and died. Post-mortem examination revealed PDLG.

Conclusion: In PDLG, CSF cytology is frequently non-diagnostic, and meningeal biopsy may miss pathology, highlighting the importance of repeat or multisite biopsies from enhancing regions in diagnostically challenging cases.

Novel neuromuscular hyperexcitability (clinical percussion myotonia) and hypophysitis associated with immune-checkpoint inhibitor therapy

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A 54-year-old right-handed gentleman presented with 4-months of muscle stiffness with visible muscle 'rippling' triggered by touch but not occurring spontaneously, nor at rest. He reported difficulty standing from crouching, exiting bed and climbing stairs. He denied problems with vision, speech, swallowing, or breathing. He had hereditary haemochromatosis, and left renal cell carcinoma was diagnosed 1 year prior, treated with nephrectomy and monthly pembrolizumab. 5 months into treatment, he reported fatigue and depression, and was diagnosed with a pituitary adenoma following blood tests and MRI.

Cranial nerve examination was normal. Tone, power, co-ordination, reflexes and sensation were normal but there was marked percussion myotonia in the quadriceps where symptomatic (video), without eyelid or thenar myotonia. Serum anti-GAD, anti-ampiphysin, anti-glycine, anti-LGI1, anti-CASPR2 and paraneoplastic antibodies were negative. Quadriceps and biceps MRI were normal. Serial MRI brain review by neuroradiology showed reduction in pituitary volume from previous swelling, in keeping with hypophysitis (not tumour). EMG showed no electrophysiological features of (neuro)myotonia, despite reproducible signs. Repeating EMG for more sensitive nerve hyperexcitability using provocation and examining exteroceptive reflexes are planned.

Whilst immune phenomena are well recognised with immune checkpoint inhibitor therapy, this represents the first case of clinical percussion myotonia with associated hypophsitis.

A rare cause of exertional headaches in a young man under pressure

Ambati S , Edwards M

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A 29-year-old male presented with 2 years of headaches - initially these episodes were present with exercise only but now occurred at rest. He reported a crushing occipital pain associated with palpitations and vomiting. He has a background of neurofibromatosis 1 and had received chemotherapy for optic pathway glioma in childhood. He was significantly visually impaired. On examination, reflexes were brisker throughout the left upper and lower limb with normal power. There was persistent clonus in his left ankle. Fundoscopy revealed pale optic discs. Annual MRI brain imaging showed stable findings of the optic chiasm glioma, a cystic enhancing mass lesion in the right basal ganglia and several small cysts in the splenium of the corpus callosum. 24-hour urinary metadrenaline profile was requested.

Normetadrenalines were raised at 60.34umol/24hr and 3-Methoxytyramine abnormal at 5.00umol/24hr. Plasma metanephrine & normetanephrine levels were raised at 15400 pmol/L & 48200 pmol/L respectively. Abdominal imaging showed a 7.5 cm right adrenal mass compatible with a phaeochromocytoma. Other findings included further neurofibromas and a D1 gastrointestinal stromal tumour. Phenoxybenzamine was started with no further headaches suffered prior to surgery. Laparoscopic right adrenalectomy was performed with histopathology confirming phaeochromocytoma. Urinary metadrenaline levels normalised 1-month post-operatively.

Sudden Silence

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Paraneoplastic syndromes often have strong associations with specific cancers and antibodies. However, when antibodies are negative or the presentation is unusual, the diagnosis can be more challenging. We report a 70-year-old female who first presented with bilateral sequential severe sensorineural hearing loss with no tinnitus or vertigo, followed by peripheral length-dependent asymmetrical sensory disturbance, then cognitive dysfunction.

Initial workup was inconclusive with normal MRI head, inconclusive neurophysiological studies and negative paraneoplastic antibodies. However, her symptoms progressed and repeat nerve conduction studies demonstrated severe sensory neuronopathy, follow-up cranial imaging showed new bilateral internal capsule white matter signal change, electroencephalogram demonstrated diffuse encephalopathy, and a CT body scan demonstrated an ovarian mass. Biopsy confirmed a high grade serous ovarian carcinoma with a raised CA125. Palliative chemotherapy with carboplatin and high-dose steroid treatment were given with a biochemical response and stabilisation but no improvement in neurological symptoms to date.

This case describes an unusual paraneoplastic association between ovarian serous carcinoma and sensorineural hearing loss, encephalitis and sensory neuronopathy with negative paraneoplastic antibodies. It also underscores the need to consider paraneoplastic syndromes if a patient presents with unexplained and progressive sensorineural hearing loss.

Acute respiratory failure with rapidly progressive muscle contractures - A complex multisystem case

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Idiopathic Inflammatory Myopathy (IIM) and Myasthenia Gravis (MG) are rare autoimmune disorders that do not typically occur simultaneously. Muscle contractures occur occasionally in myositis but usually develop slowly and most often in the presence of anti-NXP2 antibodies.

We report a woman in her mid-sixties who presented acutely with painful upper-limb swelling, dysphagia, dysphonia, rapidly progressive fixed flexion contractures and respiratory failure. Extensive investigations including serology, imaging, neurophysiology and muscle biopsy supported the presence of both IIM and MG, although myositis antibodies were negative. Thymectomy ultimately confirmed a WHO type B2 thymoma, supporting a diagnosis of thymoma associated autoimmune myopathy–myasthenia overlap.

The patient received a combination of immunomodulatory therapy targeting both conditions, followed by thymectomy. Bulbar and respiratory symptoms rapidly improved, with more modest improvement seen in the contractures.

This case demonstrates a rare and severe presentation of thymoma and showcases the importance of multidisciplinary involvement and early, aggressive treatment.