

unable to manage at home. He had frequent falls, shuffling gait, slow movements, right sided limb tremor along with difficulty with eye movements, dysphagia and several collapses. On examination, he had gait and other features of parkinsonism (tremor, bradykinesia, rigidity and postural instability). He also had ophthalmoparesis, suggestive of PSP.

However, there were some incongruencies that prompted for a more detailed examination. This revealed subtle but definite features in keeping with functional parkinsonism. The patient was referred to physiotherapy and cognitive behavioural therapy. He is also awaiting a dopamine transporter imaging.

During my oral presentation, I will be showing videos of this patient's examination findings and present a literature review. This case demonstrates how easy it is to miss the signs if they are not carefully and actively looked for. It also highlights the importance of challenging and digging deeper when things do not quite fit.

2408

THE FOREST NOT THE TREES: A PRESENTATION OF A CHALLENGING CASE

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A 37-years-old NZ European man presented with a history of five years of progressive neurological deficits. These included dysarthria, hyper-salivation, vivid dreams, sleep disordered breathing, elevated hemi-diaphragm, left upper limb paraesthesia, syncopal episodes, lethargy, irregular bowels, difficulty passing urine, difficulty with temperature regulation, locked and painful jaw, anxiety, reduced sleep and headaches. On examination, he had mild dysarthria, wasted tongue with deviation to left and weakness on the left. The reflexes were brisk in the lower limbs with an up-going plantar response on the left. MRI revealed an ill-defined heterogeneous enhancement of the medulla extending to the right cerebellar peduncle. The patient underwent extensive work up and treatment trial with steroids with a working diagnosis of Neuro-sarcoidosis. He developed several further symptoms of tremors and further paraesthesia over the next three months. The patient had a sudden death and the final diagnosis of Alexander disease was revealed in his autopsy.

Retrospectively, the symptoms collectively clearly point to a neurological disorder, but during the five years of the disease progression, his complaints were approached individually as separate issues by multiple specialities. Due to this, despite the numerous red flags, these were unrecognised and the patient presented to Neurology with an advanced illness. This is a valuable case for learning and it reminds us how an eye for detail and careful observation in history and examination is critical, especially for patients presenting as a diagnostic challenge. This very fact is also what makes Neurology such a fascinating and intriguing specialty.

2409

AN ATYPICAL PRESENTATION OF AUTOIMMUNE GLIAL FIBRILLARY ACIDIC PROTEIN (GFAP) ASTROCYTOPATHY WITH EXTENSIVE SPINAL CORD DISEASE WITHOUT BRAIN INVOLVEMENT

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Autoimmune glial fibrillary acidic protein (GFAP) astrocytopathy is a novel central nervous system disorder that presents with one or more of meningitis, encephalitis and myelitis. 95% of patients present with either meningoencephalitis or meningo-encephalo-myelitis. Myelitis without cerebral involvement is rare and thought to represent only about 5% or less of cases.

We report, along with review of literature, a rare presentation of autoimmune GFAP astrocytopathy, who presented with myelitis without encephalitis and experienced initial misdiagnosis and a delay in the diagnosis.

A 25-year-old male, kiwi packer, migrant from India presented with meningism (fevers, headache, neck stiffness, photophobia, nausea and vomiting) with subsequent development of urinary retention and progressive weakness and sensory change in the limbs. CSF examination revealed the GFAP-IgG with significantly elevated lymphocytes and protein. Magnetic resonance imaging revealed a rare finding of longitudinally extensive myelitis extending from the C2 to T11 level without any brain lesions. He had significantly elevated lymphocytes and protein in the CSF with the presence of GFAP-IgG. Interestingly, He was initially diagnosed with viral meningitis and had multiple re-presentations to the hospital with ongoing deterioration in clinical status despite antibiotic and antiviral therapy. This led to further investigations and immunotherapy (IV steroids and plasma exchange) with good recovery.

This is a valuable case for learning, which reports an uncommon presentation of a rare disorder. It highlights the importance of detailed history and examination, having broad differentials in mind and early re-evaluation of diagnosis when things do not go as planned.

2413

DOES SERUM NEUROFILAMENT LIGHT CHAIN LEVEL CONTRIBUTE TO THE PREDICTION OF TREATMENT RESPONSE IN MULTIPLE SCLEROSIS?

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Objectives The Crystal Ball model is a previously published and externally validated algorithm.¹ that helps predict the individual response to Multiple Sclerosis (MS) therapies. In this study, we investigated the contribution of Neurofilament Light (NfL) chain levels to the prediction of treatment response.

Methods Data from 3 participating centers were collated to include patients with relapsing MS, recorded Expanded Disability Status Scale and NfL at baseline, while being treated with or commencing a disease modifying therapy. We used principal component analysis to reduce the dimensionality of the demographic and clinical data. Cox proportional hazards models with three principal components and treatment duration with vs. without NfL at baseline were used to model the risk of relapses, 6-month confirmed disability worsening and 9-month confirmed disability improvement. The accuracy of the NfL and non-NfL models was compared using 10-fold cross-validation and Harrell's C-index.

Results 1716 patients across 12 MS therapies were included (68% female, mean age 38±11). The accuracy of the prediction of treatment response in the models without NfL was comparable to the original study. Addition of NfL did not further increase the predictive accuracy (C-indices: relapses 0.59, disability worsening 0.64, disability improvement 0.82).

Conclusion Serum NfL, an emerging prognostic marker, does not substantially contribute to differentiating individual response to MS therapies among patients with various clinical and demographic characteristics. Clinical and demographic information remains the most useful indicator of future individual response to MS therapies.

REFERENCE

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2415 COLPOCEPHALY WITH REMOTE RIGHT PARIETAL INFARCTION AND CONGENITAL RUBELLA SYNDROME (CRS)

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Objective Describe a rare and complicated case.

A 46 y/o female with CRS presented, post head-stroke, with imaging showing remote right parietal infarct, colpocephaly and thinning of the corpus callosum.

Colpocephaly is a rare condition, reported in only a dozen adults. It occurs in 1:700,000 live births and is usually diagnosed before birth. It is an imaging diagnosis of the lateral ventricles: when the widest portion of posterior horn is greater than three times that of the anterior horn. It may be unilateral, bilateral, or asymmetric.

Pathophysiologically, it appears to reflect the retention of the early fetal configuration of enlarged ventricles that, usually, are slowly filled by the radial out-migration of cells from the subventricular zone (SVZ), to the Subplate (Sp) with the formation of white matter being the remnant of the subplate.

First reported in 1941, it has been associated with: dysgenesis of the corpus callosum, pachygyria, lissencephaly, schizencephaly, microcephaly, meningomyelocoele. Aetiologically it has been associated with intra-uterine infection, Intra-Uterine Growth Retardation, perinatal anoxic-hypoxic injury, maternal

drugs, genetics, migration disorder. It may affect: cognition, cerebral palsy, gait, speech, epilepsy, vision.

Conclusion Remarkably, neurologically normal (or at least asymptomatic) children and adults have been reported although few have had extensive, appropriate, neuropsychological assessments. Neuropsychological assessment is marred by the lack of normalized data for such a unique group. Cerebral connectivity in this condition remains unexplored.

2416 CONGENITAL RUBELLA SYNDROME (CRS) WITH RARE COLPOCEPHALY

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Objective First description of association of Colpocephaly with CRS

A 46 y/o female with Congenital Rubella Syndrome (CRS) presented, post head-stroke, with imaging showing a remote prenatal right parietal infarct, and colpocephaly. She exhibited a smaller, spastic, right upper limb, bilateral congenital deafness. Cognitively, she had worsening behavioural and memory issues recently.

CRS was first flagged by Australian ophthalmologist Norman Gregg (1941). With effective vaccination, Rubella cases in Australia fell markedly, from 4,000 to less than 40 per year (92% now immune). CRS cases concomitantly fell from 300 to around 1 per year; to women born overseas (AusInstHealthWelfare). CRS outcomes reflect virus interaction s with the neurodevelopment epoch: early infection results in high embryo-fetal wastage. If contracted after week 20, little deficit results as neurogenesis, path-finding migration and synaptogenesis have progressed.

Brain involvement may result in mixtures of grey and white matter disease and infarction leading to blindness, deafness, intellectual impairment, shortened limbs. The virus may persist for decades. Neurodevelopment involves many simultaneous processes, in many different parts of the neuraxis that have delimited time-lines – both for immediate and consequential outcomes: these colour the final clinical picture. Other organ involvement includes cardiac outlet pathology.

Conclusion Colpocephaly (Ventriculomegaly of the posterior horns of the lateral ventricles) has not previously been a reported association. It's rarity, variation when it is noted, and associated lesions, have conspired to mean only case reports are available and no definitive study of neurologic dysfunction is available.

2420 NEUROPSYCHOLOGICAL DEFICITS POST-STROKE MAY INVALIDATE CONSENT

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Objective Review pre-requisites of valid consent.

According to NSW Health Guidelines, consent, to be valid, entails four attributes: it be freely given; the patient must have capacity; it must be 'informed'; it be specified to a particular act or therapy. There are two unvoiced assumptions that underpin these requirements: that the mind is relatively 'fixed' over time and that awareness can be reliably measured.