

Conclusions Evobrutinib is the first BTK inhibitor to demonstrate disease activity reduction in RMS. The observed benefit-risk profile supports further clinical development.

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A RARE PRESENTATION OF A RARE DISEASE: ADULT ONSET KRABBE'S DISEASE PRESENTING WITH PROGRESSIVE HYPERTROPHIC POLYRADICULONEUROPATHY

Alycia MacDonald*, Phillipa Lamont, Wai Leong. *Department of Neurology, Royal Perth Hospital, Perth, WA, Australia*

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Introduction Globoid Cell Leukodystrophy (Krabbe's disease) is a rare autosomal recessive condition caused by defects in the lysosomal enzyme galactosylceramidase. The disease typically occurs in infants and is rapidly progressive. Fewer than 5% of cases develop symptoms in adulthood, the majority of which present with spastic paraparesis.

Case A 40-year-old Italian man presented with a 20 year history of gradually progressive asymmetrical hand paraesthesias and weakness. EMG demonstrated demyelinating features and absent sensory responses. CSF examination revealed elevated protein with no cells and negative oligoclonal bands. Treatment with intravenous immunoglobulin (IVIg) was commenced and continued for 2 years for presumed CIDP without clinical response. Upper limb wasting and weakness progressed over the following 10 years with associated loss of reflexes, prompting consideration of an underlying genetic neuropathy. DNA testing for common CMT mutations was negative. MRI cervical spine and brachial plexus demonstrated diffuse bilateral non-enhancing enlargement of cervical nerve roots. IVIg was recommenced over the next 10 years with continued clinical deterioration to the point of bilateral flail arms with relatively preserved lower limb motor function, in addition to bulbar and respiratory dysfunction by age 64. MRI brain demonstrated thickening of a number of cranial nerves without contrast enhancement, and extensive T2-hyperintensity of periventricular white matter. Next generation sequencing revealed two novel mutations in the galactosylceramidase (*GALC*) gene, and beta-galactocerebrosidase activity was reduced, confirming the diagnosis of Krabbe's disease.

Conclusion This case highlights an unusual presentation of Krabbe's disease with progressive hypertrophic polyradiculoneuropathy, associated with a novel genetic mutation.

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TAKOTSUBO CARDIOMYOPATHY AND MYASTHENIC CRISES: A CASE SERIES

¹Matthew Katz*, ²Stephen Walsh, ³Benjamin Tsang, ¹Pamela McCombe, ²Arman Sabet. *¹Royal Brisbane and Women's Hospital, Brisbane, QLD, Australia; ²Gold Coast University Hospital, Gold Coast, QLD, Australia; ³Sunshine Coast University Hospital, Sunshine Coast, QLD, Australia*

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Introduction Takotsubo cardiomyopathy (TCM) is an acute, reversible cardiomyopathy that can mimic acute coronary syndrome.¹ It is characterised by left ventricular dysfunction, electrocardiogram (ECG) changes and transient apical ballooning in the absence of significant coronary artery disease.¹ It is usually triggered by acute stress with catecholamine surge but the exact pathogenesis is not known.¹ Takotsubo cardiomyopathy

has been described in patients with myasthenic crisis. We present the first and largest case series of four patients with TCM in the setting of myasthenic crisis and discuss possible causes.

Methods Two patients from each tertiary neurologic centre were identified by their treating neurologist for inclusion in the series. We performed a review of their case notes with respect to history, examination, investigations and management. A brief literature review was also completed.

Results The mean age was 78 with a 1:1 female to male ratio. Three of the patients were newly diagnosed with myasthenia gravis (MG) at the time of their TCM. All patients were AChRab positive. One patient had a previous thymectomy but the others had no evidence of thymoma.

On review of the literature most cases of TCM in myasthenic crisis occurred in older females. Abnormalities of the ECG were universal. Most cases did not have a thymoma or history of thymectomy.

Conclusion Takotsubo cardiomyopathy may be easily overlooked in those presenting with myasthenic crises as they share overlapping clinical features. Rigorous attention to the cardiac status of these patients, especially the ECG, may help to avoid missing this important diagnosis.

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IMPAIRED COLOR DISCRIMINATION IS ASSOCIATED WITH HALLUCINATIONS IN DEMENTIA WITH LEWY BODIES

¹Elie Matar*, ²Joseph R Phillips, ¹Kaylena A Ehgoetz Martens, ¹Glenda M Halliday, ¹Simon JG Lewis. *¹Brain and Mind Centre, University of Sydney, Camperdown, NSW, Australia; ²MARCS Institute for Brain, Behaviour and Development, Western Sydney University, Sydney, NSW, Australia*

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Introduction Emerging evidence indicates that color discrimination impairments can predict the development of dementia across a range of prodromal Lewy body conditions. However, color vision deficits are not seen uniformly in patients with Dementia with Lewy Bodies (DLB), suggesting a more nuanced association. Visual hallucinations (VH) represent a discriminating feature of DLB, and recent evidence implicates visual pathway dysfunction as a significant contributor to this phenomenon. We therefore hypothesized that color impairment will more closely associate with VH in DLB rather than general cognition.

Methods In this study, we examined the relationship between color vision impairment and VH, along with other clinical and neuropsychological features in 24 patients with DLB alongside 25 age-matched controls. Color discrimination was assessed using the Farnsworth-Munsell-100 Hue (FM-100) test.

Results Color discrimination impairment was seen in 16/24 DLB participants (67%) with a higher error score relative to controls ($p=0.001$). We demonstrate for the first time a strong association between color discrimination errors and both the presence and severity of VH in DLB based on clinician-derived ($p=0.008$) and questionnaire-derived ($p=0.03$) measures. Correlation with clinical and neuropsychological variables revealed that color discrimination is significantly

related to visuospatial impairment ($p=0.02$) but not to global measures of cognition, motor severity, age or disease duration. Factor analysis confirmed a unique relationship between color discrimination, visual hallucinations and visuospatial function.

Conclusion Our results suggest that color impairments may be a specific biomarker of visual hallucinations and associated visuo-perceptual deficits in evolving Lewy body disorders rather than dementia per se and thus providing insight into a shared pathophysiological substrate.

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TRAINING IN HEADACHE IN AUSTRALIA, NEW ZEALAND AND ASIA

^{2,1}Lin Zhang*, ³Bronwyn Jenkins, ^{2,1}Richard Stark, ^{2,1}Elspeth Hutton. ¹Neurosciences, The Alfred Hospital, Melbourne, VIC, Australia; ²Faculty of medicine nursing and health sciences, Monash University, Melbourne, VIC, Australia; ³Neurology Department, Royal North Shore Hospital, Sydney, NSW, Australia

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Introduction Migraine is the leading cause of age-adjusted neurological disability in Australia, but little is known about headache training in our region. We aimed to assess the quantity of teaching in headache subjects during undergraduate and postgraduate years.

Method This is a cross-sectional survey study where questionnaires were sent to 137 delegates from Australia, New Zealand and Asia, prior to the Headache Master School in Sydney in August 2018. The main outcome measured are recalled number of hours of teaching in undergraduate year and postgraduate years in: 1) Migraine; 2) Trigeminal autonomic cephalalgias (TACs); 3) Asthma; 4) Myasthenia gravis (MG).

Results The questionnaire response rate was 73% (100 of 137), of which 29 delegates were within 10 years of completing their undergraduate degree and 98 were neurologists. In undergraduate training, there was much greater quantity of teaching in asthma than migraine ($Z=5.007$, $p<0.000$) despite both being high-prevalent (asthma 11%, migraine 15–20%) conditions. Similarly, for diseases of medium-to-low prevalence, there was less training in TACs (1/1000), compared to MG (1.2/10,000) ($Z=6.196$, $p<0.000$). These major differences in training were also seen in postgraduate years even though overall headache teaching was greater in postgraduate than undergraduate training ($p<0.000$).

Conclusions Despite the high prevalence and morbidity of headache disorders, they receive less attention in training than conditions with similar prevalence. We propose that headache training opportunities should be improved in our region, particularly in the undergraduate course and preceptorships or fellowships in postgraduate years.

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ALCOHOL ASSOCIATED TOXIC MYELOPATHY WITH NORMAL HEPATIC FUNCTION

Srimathy Vijayan*, Scott Davies, Wai Leong. *Neurology, Royal Perth Hospital, Perth, WA, Australia*

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Introduction Alcohol related neurological dysfunction affects the central and peripheral nervous system. We present a rare consequence of excessive alcohol consumption. A 42-year-old female consuming 2 bottles of wine daily for the preceding 5 years presented with 12 months of progressive gait disturbance. Examination revealed a spastic paraparesis with prominent dorsal column signs. Extensive work up found no alternate cause for her presentation. The final diagnosis was direct alcohol mediated toxic myelopathy. We reviewed the current literature on this rare condition.

Methods We searched PubMed/OVID databases with the terms ‘alcohol AND myelopathy’.

Results There is a paucity of research on this clinical entity. Sage *et al.*¹ described 5 well-nourished alcoholics presenting with a progressive myelopathy. As with our patient, routine laboratory tests, including nutritional screen, were normal. CT Myelogram revealed no structural cause. CSF was normal. In contrast, a myelopathic syndrome can occur in alcoholic hepatic failure, attributed to portosystemic shunting of blood, with resultant hyper-ammonaemia and demyelination of corticospinal tracts.^{2–4}

Conclusion Alcohol associated toxic myelopathy is a rare entity, distinct from hepatic myelopathy. It is thought to result from direct toxic effects of ethanol on corticospinal and large myelinated sensory tracts. The sensory involvement is unique for this entity, which distinguishes it from hepatic myelopathy, where corticospinal involvement predominates. It is prudent to look for other potentially treatable secondary causes of myelopathy. Abstinence from alcohol results in modest improvement in symptoms. Minimal literature exists on this clinical entity and further research is required.

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HEADACHE WITH PERIORBITAL ECCHYMOSIS; A RARE CLINICAL ENTITY

Srimathy Vijayan*, Carolyn Orr, Catherine Franconi. *Neurology, Royal Perth Hospital, Perth, WA, Australia*

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Objectives Primary headache disorders are common with migraine and tension headache accounting for the vast majority of cases. A smaller proportion suffer from trigeminal autonomic cephalgia (TAC). We present a 23-year-old Caucasian female who described characteristic, episodic headaches starting with a dull retro-orbital/bi-frontal pressure evolving, over the course of 1 minute, to experience florid periorbital ecchymosis. While this phenomenon has been described in the literature, the characteristics of our case are unique and noteworthy of reporting.