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

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

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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.

059 TAKOTSUBO CARDIOMYOPATHY AND MYASTHENIC CRISES: A CASE SERIES

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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.

REFERENCE

1. Akashi YJ, Goldstein DS, Barbaro G, Ueyama T. Takotsubo cardiomyopathy: a new form of acute, reversible heart failure. *Circulation* 2008;**118**:2754–2762.

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

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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