Conclusion Although useful as diagnostic biomarkers, further studies are required to evaluate these potential biomarkers longitudinally and to assess their reliability as surrogates for underlying neurodegenerative changes and clinical disease severity. DTI showed the most promise as a biomarker for disease severity in HSP.

047 ACUTE LUMBOSACRAL PLEXOPATHY AFTER BLOOD LOSS IN A PATIENT WITH BILATERAL COMMON ILIAC ARTERY OCCLUSION

Matthew Katz*, Pamela McCombe. *Royal Brisbane and Women's Hospital, Brisbane, QLD, Australia*

10.1136/jnnp-2019-anzan.42

Introduction We present a case of a 56 year old male with bilateral, asymmetric leg weakness from ischemic lumbosacral plexopathy. This followed acute blood loss from an episode of self-harm, which occurred in the setting of chronic thrombotic occlusion of bilateral common iliac artery (CIA) stents.

Methods A detailed history, examination and neurophysiology study were performed in addition to reviewing case notes and results of other investigations. A brief literature review was also completed.

Results The patient presented with acute blood loss following self-inflicted lacerations to his anterior neck and both wrists. He had asymmetric flaccid, areflexic weakness of his legs with decreased sensation over the right foot and absent distal leg pulses. Magnetic resonance imaging (MRI) of brain and whole spine was normal. Computed tomography angiogram (CTA) revealed significant distal aortic atherosclerotic disease with complete occlusion of bilateral CIA stents. The patient underwent endovascular re-stenting of both common iliac arteries with a good angiographic result. An electromyogram performed one month later was supportive of a lumbosacral plexopathy, mainly on the right.

Only three cases of acute ischemic lumbosacral plexopathy have been reported in the setting of aorto-iliac occlusive disease.¹⁻³

Conclusion Ischemic injury to the lumbosacral plexus is rare, especially outside the surgical setting as seen in this case. It is important to consider this cause in any patient presenting with rapid onset asymmetric, bilateral leg weakness as early recognition and reperfusion may prevent further damage and reduce long-term disability.

REFERENCES

- Schreuder AH, Fennis TF, Teijink JA, Koehler PJ. Lumbosacral plexopathy associated with aortoiliac occlusive disease. *J Neurol*. 2007;254(6):803-5.
- Chhetri SK, Lekwuwa G, Seriki D, Majeed T. Acute flaccid paraparesis secondary to bilateral ischaemic lumbosacral plexopathy. *QJM*. 2013;106(5):463-5.
- Yun S. Ischaemic lumbosacral plexopathy following an acute thrombosed abdominal aortic aneurysm mimicking stroke: A case report. *Hong Kong Journal of Emergency Medicine*. 2016;23(2):52-6.

048 HARDING'S DISEASE: AN IMPORTANT MS MIMIC

¹Stuti Joshi*, ²Allan Kermode. ¹Sir Charles Gairdner Hospital, Stirling, WA, Australia; ²Perron Institute, Sir Charles Gairdner Hospital, Nedlands, WA, Australia

10.1136/jnnp-2019-anzan.43

Introduction Leber's hereditary optic neuropathy is a mitochondrially-inherited disorder characterized by bilateral,

painless visual loss, which leads to severe optic atrophy.¹ LHON can be associated with an MS-like illness referred to as Harding's disease.²

We report two siblings, who both harbour the 11778 mtDNA mutation, but manifest markedly different clinical phenotypes; a male with classical LHON and a female with Harding's disease.

Methods and Results A 61-year-old female, who was diagnosed with MS 22 years ago was referred to our service for a second opinion. She developed unilateral painless visual loss in her 20's, was diagnosed with optic neuritis and treated with corticosteroids with some recovery. A second episode of more severe visual loss at age 39 left her with visual impairment to less than finger counting. 4 years later, she had an episode of dysarthria and gait ataxia. MRI showed multifocal white matter lesions involving the juxta-cortical and periventricular regions, cerebellar peduncle and cervical cord. Targeted views of the optic pathways showed hyperintensity of the left optic nerve, with involvement extending into the optic canal.

The patient has one brother who was diagnosed with LHON at age 37 after presenting with severe painless bilateral sequential visual loss. Genetic testing of the index patient confirmed the presence of the same mutation identified in her brother.

Conclusion LHON and Harding's disease demonstrate a great degree of variability in clinical phenotype and penetrance between males and females as well as individuals within the same family.³ While there is no evidence for screening MS cohorts for the LHON, consider genetic testing in patients with severe and persistent bilateral visual loss or with a suggestive family history.⁴

REFERENCES

- Harding AE, Sweeney MG, Miller DH, Mumford CJ, Kellar-Wood H, Menard D, McDonald WI, Compston DA. Occurrence of a multiple sclerosis-like illness in women who have a Leber's hereditary optic neuropathy mitochondrial DNA mutation. *Brain*. 1992 August;115 (Pt 4):979-89.
- Palace J. Multiple sclerosis associated with Leber's Hereditary Optic Neuropathy. *J Neurol Sci*. 2009 November 15;286(1-2):24-7. Review.
- Pfeffer G, Burke A, Yu-Wai-Man P, Compston DAS, Chinnery PF. Clinical features of MS associated with Leber hereditary optic neuropathy mtDNA mutations. *Neurology*. 2013;81(24):2073-2081.
- Yu-Wai-Man P, Chinnery PF. Leber hereditary optic neuropathy. In: Pagon RA MP, Adam Ardinger HH eds. *GeneReviews*. Seattle, WA: University of Washington, Seattle; 2013. Accessed May 7, 2018.

049 REAL WORLD EVIDENCE (RWE) ON IMPACT OF AGE ON LONG-TERM PERSISTENCE TO DISEASE MODIFYING THERAPIES (DMTs) IN RELAPSING-REMITTING MULTIPLE SCLEROSIS (RRMS) IN AUSTRALIA

¹Andrew Kornberg*, ²Tim Spelman, ³Mark Schulz, ³Birendra Arora, ⁴Eric Chung, ⁵Prabjot Juneja, ³Robert Walker, ⁶Anneke van der Walt, ⁶Helmut Butzkueven. ¹Royal Children's Hospital, Parkville, VIC, Australia; ²Department of Medicine, University of Melbourne, Melbourne, VIC, Australia; ³Novartis, North Ryde, NSW, Australia; ⁴Prospection Pty Ltd, Australian Technology Park, Eveleigh, NSW, Australia; ⁵Prospection Pty Ltd, Australian Technology Park, Eveleigh, NSW, Australia; ⁶MS and Neuroimmunology Group, Central Clinical School, Monash University and Alfred Hospital, Melbourne, VIC, Australia

10.1136/jnnp-2019-anzan.44

Background Age has been independently associated with higher rates of treatment discontinuation in multiple sclerosis.

Objective The current study examines the impact of age on persistence for all reimbursed DMTs for RRMS in Australia.

Methods The Pharmaceutical Benefits Scheme (PBS) 10% sample supplied by the Department of Human Services was used

in this study. Eligible patients must have received a script for a reimbursed DMT for RRMS between September 2011 and February 2016. Patients were classified into five age-groups (ages 18–30; 31–40; 41–50; 51–60; 61+) and defined as persistent if their DMT script was filled within 4 months. Persistence was derived using the Kaplan-Meier method and hazard ratios (HR) to represent the relative rate of drop-off of different age groups.

Results Patients aged 18–30 (n=250) had a 44% increased risk of discontinuation (HR 1.44 (95%CI: 1.22–1.72) compared to the ‘all ages’ cohort (n=1,866); no significant difference was observed for any other age group (HRs between 1.08 and 0.92). Patients in this 18–30 age-group had a significantly higher risk of discontinuation on injectable therapy (glatiramer acetate, interferon beta-1a, interferon beta-1b) compared to those on non-injectable therapy (dimethyl fumarate, fingolimod, natalizumab, teriflunomide) (HR 2.42 (95% CI: 1.63–3.61).

Conclusions Patients aged 18–30 were the least persistent age group in this study. Patients aged 18–30 were less persistent on injectable than non-injectable DMTs.

051 OVERLAPPING SYNDROME OF GIANT CELL ARTERITIS AND ANCA-ASSOCIATED VASCULITIS COMPLICATED BY SEVERE AXONAL NEUROPATHY: A CASE REPORT

¹Laura Perju-Dumbrava*, ¹Fariha Islam, ¹Rohitha Makonahalli, ²Catrina McLean, ³Hedley Griffiths, ¹Abhishek Malhotra. ¹Neurology, Barwon Health, Geelong, VIC, Australia; ²Anatomical Pathology, Alfred Hospital, Melbourne, VIC, Australia; ³Rheumatology, Barwon Health, Geelong, VIC, Australia

10.1136/jnnp-2019-anzan.45

Case A 61-year-old woman presented with what was thought to be a refractory, temporal artery biopsy-proven giant cell arteritis (GCA). After 4 months of therapy she was unable to get the prednisolone dose below 40 mg/day, so weekly subcutaneous Tocilizumab was introduced but with minimal response after 8 weeks. She was then admitted to hospital with a severe rapidly progressive length dependent sensorimotor peripheral neuropathy. Nerve conduction studies showed predominantly an axonal neuropathy with multiple pseudo-conduction blocks.

Sural nerve and gastrocnemius biopsies revealed a necrotising vasculitis with numerous giant cells and active axonal degeneration. She proved to be ANCA-PR3 positive, without other systemic manifestations. ANCA-associated vasculitic neuropathy was diagnosed and she was treated with two 1g Rituximab infusions, 2 weeks apart.

On follow-up after a month, she had regained some strength but still required a wheelchair for mobility. Further Rituximab infusion after 6 months is planned; prednisolone had been successfully weaned to 10 mg/day.

Conclusion Peripheral nerve involvement, which is relatively common in ANCA-associated vasculitis, has also been reported in 14% of GCA cases. But ANCA-PR3 positivity is rare in biopsy-proven GGA, with only a handful of well-documented cases.

Heightened suspicion of an alternative diagnosis in the face of an unusual clinical course (lack of steroid response and appearance of small vessel vasculitic symptoms for what is accepted to be a large vessel vasculitis) is critical and our experience highlights the important fact that a diagnosis of one of these disorders does not preclude the subsequent diagnosis of the other.

053 GLUT-1 DEFICIENCY PRESENTING AS DOPA-RESPONSIVE DYSTONIA: AN ATYPICAL PHENOTYPE OF A RARE DISEASE

¹Sophie E Waller*, ¹Jeffrey Blackie, ²Himanshu Goel. ¹Department of Neurology, John Hunter Hospital, New Lambton Heights, NSW, Australia; ²John Hunter Hospital, New Lambton Heights, NSW, Australia

10.1136/jnnp-2019-anzan.46

Introduction The *SLC2A1* gene encodes the glucose transporter GLUT1, responsible for normal glucose transport across the blood-brain barrier. Mutations in this gene have classically been associated with an epileptic encephalopathy referred to as GLUT-1 Deficiency Syndrome, which typically presents with early-onset refractory epilepsy, developmental delay and complex movement disorders.^{1–2} More recently, *SLC2A1* variants have been identified in patients with paroxysmal exercise-induced dyskinesia (PED), with or without a history of epilepsy.^{3–4} Response to medication is typically poor; however, both seizures and dyskinesia may improve following implementation of a ketogenic diet.⁵ A single case of levodopa responsiveness has previously been described in a subject with *SLC2A1* mutation and PED.⁵

Methods and results We describe a 47-year-old female with mild intellectual disability since childhood but no history of epilepsy, who developed episodic dystonia affecting the lower limbs in her early 20's. A clinical diagnosis of dopa-responsive dystonia was made following a marked, sustained response to levodopa. There was no significant family history. In her 40's she developed breakthrough dystonia with exertion and choreiform movements affecting the fingers and face. A subsequent dystonia panel identified a heterozygous variant c.[1199G>T]; [=] (p.Arg400Leu) in the *SLC2A1* gene. Cerebrospinal fluid glucose concentration was low (2.0 mmol/L). She declined a trial of a ketogenic diet.

SLC2A1 variants are associated with PED; however, response to levodopa has not been widely reported.

Conclusion It is becoming increasingly evident that the phenotypic presentations of GLUT1 deficiency are diverse, and *SLC2A1* testing should be considered in a broader range of patients.

REFERENCES

- Wang, D, Kranz-Eble P, De Vivo DK. Mutational analysis of GLUT1 (SLC2A1) in Glut-1 Deficiency Syndrome. *Human Mutation* 2001;**16**:224–231.
- Klepper J, Leiendecker B. GLUT1 deficiency syndrome – 2007 update. *Developmental Medicine & Child Neurology* 2007;**49**:707–716.
- Schneider SA, Paisan-Ruiz C, Garcia-Gorostiza I, Quinn NP, Weber YG, et al. GLUT1 gene mutations cause sporadic paroxysmal exercise-induced dyskinesias. *Mov Disord* 2009;**24**(11):1684–1696.
- Suls A, Dedeken P, Goffin K, Van Esch H, Dupont P. Paroxysmal exercise-induced dyskinesia and epilepsy is due to mutations in *SLC2A1*, encoding the glucose transporter GLUT1. *Brain* 2008;**131**:1831–1844.
- Baschieri F, Batla A, Erro R, Ganos C, Cordivari C, Bhatia KP. Paroxysmal exercise-induced dystonia due to GLUT-1 mutation can be responsive to levodopa: a case report. *J Neurol* 2014;**261**:615–616.

054 IMPACT OF ONLINE ADULT HEADACHE GUIDELINE ON HEADACHE REFERRAL TO NEUROLOGY CLINIC

Leon Huang, David Bourke*, Anna Ranta. Capital and Coast District Health Board, Wellington, Wellington, New Zealand

10.1136/jnnp-2019-anzan.47

Introduction Headache is a common problem in primary care and is one of the main reasons for general practitioners to