Conclusion Acute causes of bilateral ophthalmoplegia include Wernicke's encephalopathy, Miller-Fisher Syndrome (MFS) variant of Guillain Barre Syndrome, brainstem stroke, myasthenia gravis and botulism. Of these, only MFS and botulism will cause both internal and external ophthalmoplegia.

087

AN INTEGRATED NEUROGENOMICS CLINIC – 28-MONTHS EXPERIENCE AND OUTCOME OF A TERTIARY REFERRAL CENTRE

^{1,2}Alison McLean, ^{3,4}Michel Tchan, ¹Sophie Devery, ¹Renee Smyth, ^{1,5,6}Kishore Kumar, ^{1,4,7}Susan Tomlinson, ^{1,7,2}Stephen Tisch, ^{1,2,4,7}Kathy Wu. ¹St Vincent's Hospital, Sydney, Darlinghurst, NSW, Australia; ²Faculty of Medicine, University of New South Wales, Sydney, NSW, Australia; ³Westmead Hospital, Sydney, NSW, Australia; ⁴The University of Sydney, Sydney, NSW, Australia; ⁵The Garvan Institute of Medical Research, Sydney, NSW, Australia; ⁶Molecular Medicine in Neurology, Concord Repatriation General Hospital, Sydney, NSW, Australia; ⁷School of Medicine, University of Notre Dame, Sydney, NSW, Australia

10.1136/bmjno-2021-ANZAN.87

Objectives To retrospectively review attendance and outcomes of a single centre, integrated multi-disciplinary (MDT) neurogenomics clinic at St Vincent's Hospital, Sydney.

Methods An audit of patients who attended the neurogenomics clinic was conducted over a 28-month period from 2017 to 2020. The clinic comprises of neurologists, clinical geneticists and genetic counsellors assessing each of the patients concurrently during the consultation.

Results In the audit period 99 patients were referred spanning 45 different clinical diagnoses. Following MDT assessment, 23% (23/99) of referring diagnoses were revised. Seventy-nine patients (80%) underwent genetic testing. The type of genetic tests ordered includes 41 exome-based panels, 14 whole genome sequencing, 13 single gene tests, 30 repeat expansion disorders and 2 chromosomal microarrays. Molecular confirmation was achieved in 22 patients following testing a yield of 28% (22/79); of which, 2% had their clinical diagnosis revised following testing. Overall, a diagnosis was achieved in 29/99 patients (29%), of whom 7 patients' diagnosis was achieved without genetic testing. From referral to the results of genetic testing, 25% (25/99) of patients had their diagnosis revised as a result of MDT input.

Conclusions Provision of an integrated multidisciplinary neurogenomics clinic in a tertiary setting provides an invaluable service with a diagnostic yield of 28%. This model provides a gold standard for diagnostic evaluation of patients with suspected neurogenetic disorders. Psychosocial benefits for patients, such as convenience/satisfaction of an MDT clinic, psychological closure for patients/families, and reproductive options enabled by achieving a genetic diagnosis, will be audited via patient survey.

088

ATYPICAL PRESENTATIONS AND COURSE OF JC VIRUS INFECTION

^{1,2}Sophie Chatterton, ³Liam Dwyer, ³Claire Thomson, ^{1,4,5,6,7}Bruce J Brew. ¹Neurology, St Vincent's Hospital, Sydney, NSW, Australia; ²University of New South Wales, Sydney, NSW, Australia; ³Lung Transplantation, St Vincent's Hospital, Sydney, NSW, Australia; ⁴Royal North Shore Hospital, Brighton-Le-Sands, NSW, Australia; ⁵University of Notre Dame, Sydney, NSW, Australia; ⁶Neurosciences Program and Peter Duncan Neurosciences Unit, St Vincent's Centre for Applied Medical Research, Sydney, NSW, Australia; ⁷Department of Neuroscience, International Society for NeuroVirology, Temple University School of Medicine, Philadelphia, USA

10.1136/bmjno-2021-ANZAN.88

Objective(s) There is increasing evidence that the spectrum of JC virus (JCV) CNS disease includes novel syndromes other than Progressive Multifocal Leukoencephalopathy (PML), the appreciation of which is increasingly important in the context of MS therapies and immunodeficiency states. Our objective is to describe unusual presentations of JCV infection to heighten clinician awareness.

Method Three case reports.

Results A 56 year-old male HIV+ with decades of viral suppression and normal immune function presented with 1 month of non-specific headache that spontaneously resolved despite an MRI showing a new area of PML and CSF being JCV DNA+. He had had two similar episodes in 2003 and 2014 with MRI scans consistent with PML, CSF JCV PCR positivity once and brain biopsy positive twice. Another 61 year-old male presented with subacute binocular vision loss and was found to have newly diagnosed HIV and JCV DNA detected in CSF. MRI brain only demonstrated symmetrical chiasmohypothalamic enhancement. There has been some improvement after cART and steroids for IRIS. Thirdly, a 65 year-old presented with subacute progressive confusion and behavioural disturbance, one year post bilateral lung transplantation. MRI brain demonstrated no evidence of PML but CSF on three occasions demonstrated a progressively increasing JCV DNA load. Despite reduction in his immunosuppression the patient developed profound encephalopathy without localising features leading to death two months later.

Conclusion These cases emphasize the atypical presentations of JCV: chronic relapsing, unusual symmetrical visual pathway disease, and non-localizing encephalopathy without MRI evidence of PML.

089

CLINICAL VARIATION IN THE USE OF DISEASE-MODIFYING THERAPIES FOR MULTIPLE SCLEROSIS BETWEEN DIFFERENT STATES AND TERRITORIES IN AUSTRALIA, 2019–20

^{1,2}Kieren Po, ^{2,3}Michael H Barnett. ¹Sydney Medical School, The University of Sydney, Sydney, NSW, Australia; ²Department of Neurology, Royal Prince Alfred Hospital, Camperdown, NSW, Australia; ³Brain and Mind Centre, The University of Sydney, Camperdown, NSW, Australia

10.1136/bmjno-2021-ANZAN.89

Objectives In Australia, patient costs for multiple sclerosis disease-modifying therapies (DMTs) are government-subsidised under the Pharmaceutical Benefits Scheme (PBS). Unlike many countries, any DMT can be selected for a patient who fulfils treatment criteria. We aimed to determine whether clinical variation exists in the use of DMTs in different Australian states and territories.

Methods DMT usage was determined from PBS prescription dispensing statistics for all DMTs available in Australia from July 2019 to June 2020.¹ Pa

Results There were 176,163 prescriptions for DMTs dispensed for an estimated 18,596 patients, with a total drug cost of AU\$344.1 million and mean drug cost per patient of AU\$18,501. Most patients were treated with oral (41.7%) or infusion (33.8%) maintenance DMTs. Fewer received platform injectable DMTs (15.3%) or immune reconstitution DMTs (9.1%) in the study period. The most common DMTs were fingolimod (23.5%), ocrelizumab (21.8%), natalizumab (12.1%), and dimethyl fumarate (10.8%). Platform injectable DMTs were most common in ACT (20.1%) and least common

in Victoria (13.0%). Oral DMTs were most common in NSW (44.0%) and least common in Tasmania (29.3%). Infusion DMTs were most common in Tasmania (47.4%) and least common in ACT (27.7%).

Conclusion We have demonstrated marked variation in DMT use in different Australian states and territories during the study period. The use of DMTs in Australia has evolved over time,³ but differences between states/territories remain unexplained. We plan to investigate the observed clinical variation in future studies.

REFERENCES

1. Department of Health. Pharmaceutical Benefits Scheme. Canberra: Australian Government; 2021. <https://www.pbs.gov.au/> (accessed 19 Feb 2021).
2. WHO Collaborating Centre for Drug Statistics Methodology. ATC-DDD Toolkit: Defined Daily Dose (DDD). Geneva: World Health Organization; 2021. <https://www.who.int/tools/atc-ddd-toolkit/about-ddd> (accessed 19 Feb 2021).
3. Goudarzi MH, et al. Disease modifying therapies for relapsing-remitting multiple sclerosis: use and costs in Australia (1996-2019). *Mult Scler Relat Disord* 2021;**50**:102835.

090 PAINLESS PROGRESSIVE MONONEURITIS MULTIPLEX SECONDARY TO AML ASSOCIATED NEUROLEUKAEMIOSIS

¹Nimalan Harinesan, ¹Rajiv Wijesinghe, ^{2,3}Nicole Wong-Doo, ^{1,3}Katrina Morris, ¹James D Triplett. ¹Neurology and Neurophysiology Department, Concord Repatriation General Hospital, Sydney, NSW, Australia; ²Haematology Department, Concord Repatriation General Hospital, Sydney, NSW, Australia; ³Concord Clinical School, The University of Sydney, Sydney, NSW, Australia

10.1136/bmjno-2021-ANZAN.90

Objectives To report the clinical history, imaging and neurophysiology findings of a case of mononeuritis multiplex caused by acute myeloid leukemia (AML) neuroleukaemiosis

Case A 58-year-old male presented with painless and progressive mononeuropathies after completing high dose cytarabine consolidation treatment for AML. Nine days following chemotherapy a right third nerve palsy developed followed by right 5th, right 7th, left 3rd and left radial, ulnar and peroneal neuropathies.

Serial MRI and PET imaging was unremarkable and 2 cerebrospinal fluid (CSF) were normal. Nerve conduction studies demonstrated abnormal right blink responses, a peroneal neuropathy and evidence of conduction block at a non-compressible site within the left ulnar nerve, however nerve ultrasound did not demonstrate any causative lesion.

Differentials considered included opportunistic fungal infections and a paraneoplastic neuropathy. A third CSF sample performed on day 24 demonstrated myeloblasts, consistent with central nervous system leukaemic infiltration. A diagnosis of neuroleukemiosis was made and intrathecal chemotherapy (initial methotrexate [MTX] and cytarabine, followed by alternating MTX/cytarabine twice a week) plus systemic chemotherapy (fludarabine, cytarabine, idarubicin) and granulocyte-colony stimulating factor was commenced resulting in partial resolution of pre-treatment symptoms.

Conclusions Progressive neuropathies in patients with leukemia are rarely reported and can be diagnostically challenging. Mononeuritis multiplex associated with AML may be painless and focused imaging may fail to demonstrate significant abnormalities. A high index of clinical suspicion is required as the differential diagnoses of neuroleukaemiosis is broad including paraneoplastic syndromes, infection and inflammatory

conditions. As in this instance multiple CSF examinations maybe required to confirm its diagnosis.

091 THE IMPACT OF DEVICE-ASSISTED THERAPY INITIATION ON THE GUT MICROBIOME IN PARKINSON'S DISEASE

^{1,2}Michal Lubomski, ³Xiangnan Xu, ⁴Andrew J Holmes, ³Jean Yang, ^{1,2}Carolyn M Sue, ¹Ryan L Davis. ¹Department of Neurogenetics, Kolling Institute, University of Sydney and Royal North Shore Hospital, Sydney, NSW, Australia; ²Neurology Department, Royal North Shore Hospital, St Leonards, NSW, Australia; ³School of Mathematics and Statistics, Sydney Precision Bioinformatics, University of Sydney, Camperdown, NSW, Australia; ⁴School of Life and Environmental Sciences, The Charles Perkins Centre, University of Sydney, Sydney, NSW, Australia

10.1136/bmjno-2021-ANZAN.91

Objectives Several studies have evaluated the impact of oral medication on the gut microbiome (GM) in Parkinson's disease (PD). However, the impact of PD device-assisted therapies (DAT) on the GM remains to be investigated. We profiled acute temporal GM stability around the initiation of PD DAT.

Methods The GM of 21 PD patients initiating either Deep Brain Stimulation (DBS) or levodopa-carbidopa intestinal gel (LCIG) were compared to 10 spousal healthy control (HC) subjects. 16S amplicon sequencing of the V3-V4 region of stool bacterial DNA was used to compare temporal GM stability between groups and with clinical outcome measures, including disease alternations relative to therapy initiation. GM response to therapy in the PD group was assessed by comparing pre-therapy (-2 and 0 weeks) with post-therapy initiation timepoints (+2 and +4 weeks) and HCs at baseline (0 weeks).

Results Altered GM compositions were noted between the PD and HC groups at various taxonomic levels, including specific differences for DBS and LCIG therapies. Beta diversity changes were also identified across the 4 week post-treatment initiation period, implying a therapy-effect on the GM.

Conclusions We present the first acute longitudinal assessment of GM response to PD DAT. The pre-treatment PD-specific GM (consistent with previous studies) was altered following DAT initiation, indicating DATs have a modulatory impact on the GM in PD.

092 CRYOLIPOLYSIS-INDUCED RADIAL MONONEUROPATHY

Nimalan Harinesan, Rajiv Wijesinghe, Michael Hayes, James D Triplett. *Neurology and Neurophysiology Department, Concord Repatriation General Hospital, Sydney, NSW, Australia*

10.1136/bmjno-2021-ANZAN.92

Objectives To report an association between cryolipolysis or 'fat-freezing' in the upper arm and development of an acute radial neuropathy likely secondary to a combination of thermal and pressure effects.

Results A 31-year-old female presented with an acute right wrist drop that occurred following a cryolipolysis procedure to the upper right arm. Paraesthesia and numbness occurred towards the end of a 40-minute cryolipolysis procedure, with weakness reported within 24 hours. Examination of the arm 1 week following symptom onset revealed a significant ecchymosis at application site of the fat freezing device. In addition,