

**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

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

<sup>1,2</sup>Sophie Chatterton, <sup>3</sup>Liam Dwyer, <sup>3</sup>Claire Thomson, <sup>1,4,5,6,7</sup>Bruce J Brew. <sup>1</sup>Neurology, St Vincent's Hospital, Sydney, NSW, Australia; <sup>2</sup>University of New South Wales, Sydney, NSW, Australia; <sup>3</sup>Lung Transplantation, St Vincent's Hospital, Sydney, NSW, Australia; <sup>4</sup>Royal North Shore Hospital, Brighton-Le-Sands, NSW, Australia; <sup>5</sup>University of Notre Dame, Sydney, NSW, Australia; <sup>6</sup>Neurosciences Program and Peter Duncan Neurosciences Unit, St Vincent's Centre for Applied Medical Research, Sydney, NSW, Australia; <sup>7</sup>Department 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

<sup>1,2</sup>Kieren Po, <sup>2,3</sup>Michael H Barnett. <sup>1</sup>Sydney Medical School, The University of Sydney, Sydney, NSW, Australia; <sup>2</sup>Department of Neurology, Royal Prince Alfred Hospital, Camperdown, NSW, Australia; <sup>3</sup>Brain 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.<sup>1</sup> 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