

079

HALLUCINOGENIC PERSISTING PERCEPTION DISORDER: A CASE SERIES AND REVIEW OF THE LITERATURE

¹Hannah HF Ford, ²Clare CF Fraser, ¹Emma ES Solly, ^{1,3}Joanne JF Fielding, ^{3,4}Owen OW White, ^{4,5,6}Anneke AWWVan der Walt. ¹Department of Neurosciences, The Alfred Hospital, Melbourne, VIC, Australia; ²Neuro-Ophthalmology, Sydney University, Sydney, NSW, Victoria; ³Ocular Motor Research Laboratory, Monash University, Clayton, VIC, Australia; ⁴Neuro-ophthalmology Service, The Alfred Hospital, Melbourne, VIC, Australia; ⁵Multiple Sclerosis and Neuroimmunology (MSNI), The Alfred Hospital, Melbourne, VIC, Australia; ⁶Multiple Sclerosis and Neuro-ophthalmology Group, Monash University, Clayton, VIC, Australia

10.1136/bmjno-2021-ANZAN.79

Objectives To report the clinical characteristics and investigation findings of a series of Hallucinogenic Persisting Perception Disorder (HPPD) cases and review previous HPPD case reports from the literature.

Methods Case studies were collected from consultant neuro-ophthalmologists between 2019 and 2020. PubMed and MEDLINE databases were searched for case reports between 2000 and 2020 using the terms 'hallucinogenic persisting perception disorder' and 'case report'

Results Thirteen case studies were reviewed. Lysergic acid diethylamide (LSD), 3,4-Methyl enedioxy methamphetamine (MDMA) and cannabinoids were the most common drugs used prior to HPPD onset. Twenty-two different visual symptoms were described. The most commonly reported were visual snow, floaters, palinopsia, photophobia and photopsia. Ophthalmic and neurologic investigations were normal. Two patients fully recovered after benzodiazepine treatment or no treatment. Twenty-four literature case reports were identified. LSD, MDMA and cannabinoids were the most frequent drugs used. Seventeen different visual symptoms were described. Ophthalmic and neurologic investigations showed no clinically significant findings in the majority of cases. 25% of cases fully recovered after treatment with benzodiazepines, eye movement desensitisation and reprocessing therapy, anti-epileptic drugs or no treatment.

Conclusions A wide variety of hallucinogenic and non-hallucinogenic recreational substances are implicated in HPPD. Clinical presentation includes a diverse range of positive visual phenomena and overlaps with Visual Snow Syndrome (VSS). Neurologic and ophthalmic investigations are typically normal. Management is complicated due to a lack of high-quality evidence. Controlled trials are needed to better understand the pathophysiology and optimize treatment for HPPD.

080

THE DIAGNOSTIC JOURNEY OF MITOCHONDRIAL DISEASE PATIENTS

^{1,2}Laura I Rudaks, ¹Eloise Watson, ¹Michal Lubomski, ¹Fabienne Edema-Hildebrand, ¹Kate Ahmad, ¹Christina Liang, ^{1,3}Ryan Davis, ^{1,3}Carolyn Sue. ¹Royal North Shore Hospital, St Leonards, NSW, Australia; ²Neurology Unit, Concord Repatriation General Hospital, Concord, NSW, Australia; ³Kolling Institute, St Leonards, NSW, Australia

10.1136/bmjno-2021-ANZAN.80

Introduction Mitochondrial disorders can often be challenging to diagnose and patients may undergo protracted investigative odysseys before reaching a diagnosis.¹ This study reviewed the diagnostic journey for genetically confirmed Mitochondrial Disease patients.

Methods Patients with a genetic diagnosis of mitochondrial disease seen at the Department of Neurogenetics, Royal

North Shore Hospital, were invited to complete an online survey at their appointment or via telephone. Participant clinical records were reviewed for additional data, including genetic diagnosis.

Results Between October 2018 and April 2020, survey results were obtained from 68 patients. The most common presenting symptoms were fatigue (39%), weakness (31%), and droopy eyelids (31%). The most frequently completed investigations were MRI (55%), neurophysiologic testing (45%) and EEG (44%). 33% of participants had consulted five or more doctors with an overall mean time to diagnosis of 6.2 years. 41% of patients received a diagnosis within two years of symptom onset, 31% between 5 and 15 years, and 11% after 15 years or more. 38% of participants received at least one alternative diagnosis prior to their definitive genetic mitochondrial disease diagnosis. Following diagnosis, 34% of patients joined a support group and 87% felt that this was beneficial.

Conclusions Our results demonstrate that many patients experience long delays, undergo many investigations and see multiple doctors before a diagnosis of mitochondrial disease is reached. It is hoped that advances in diagnostic pathways and access to earlier genetic testing may streamline the process.²

REFERENCES

- Grier J, Hirano M, Karaa A, et al. Diagnostic odyssey of patients with mitochondrial disease. Results of a survey. *Neurol Genet* 2018;4: e230. doi:10.1212/NXG.0000000000000230
- Watson E, Davis R, Sue C. New diagnostic pathways for mitochondrial disease. *J Transl Genet Genom* 2020;4:188–202.

081

IGLON5 AUTOIMMUNITY IN TWO CASES WITH PERIPHERAL NERVOUS SYSTEM FEATURES

^{1,2}Laura I Rudaks, ^{3,4}Victor SC Fung, ⁵Jean-Pierre Halpern, ^{2,5}Omar Ahmad. ¹Concord Repatriation General Hospital, Concord, NSW, Australia; ²Royal North Shore Hospital, St Leonards, NSW, Australia; ³Neurology Department, Westmead Hospital, Westmead, NSW, Australia; ⁴Sydney Medical School, University of Sydney, Sydney, NSW, Australia; ⁵Neurology Department, Sydney Adventist Hospital, Wahroonga, NSW, Australia

10.1136/bmjno-2021-ANZAN.81

Introduction IgLON5 autoimmunity has four main clinical patterns; a sleep disorder, bulbar syndrome, PSP-like pattern and predominant cognitive impairment. Other manifestations include movement disorders, gait instability, dysautonomia and neuropsychiatric features [1-3]. Peripheral nervous system involvement has been occasionally reported [3,4]. We describe two cases of IgLON5 autoimmunity presenting with peripheral neuropathy.

Cases A 72-year-old lady presented with progressive distal lower limb numbness, paraesthesia, incoordination and gait disturbance. Associated features included fluctuating facial numbness, limb and trunk fasciculations, upper limb tremor, anxiety, episodic hyperventilation, nocturnal myoclonus and vocalisations in sleep. Nerve conduction studies (NCS) demonstrated demyelinating features in the lower limbs. Anti-IgLON5 antibodies were detected in cerebrospinal fluid. She was treated with IVIg, oral prednisolone, azathioprine and plasma exchange.

A 73-year-old man presented with worsening tremor. Evolving features included facial paraesthesia, imbalance, head 'fog-giness', visual agnosia, constipation, insomnia, sleep utterances, somnambulism, nocturnal tremor and myoclonus. NCS showed a generalised demyelinating sensorimotor polyneuropathy. Neuropathy screen demonstrated anti-IgLON5 antibodies and IgG

kappa paraprotein, leading to a new diagnosis of monoclonal gammopathy of undetermined significance (MGUS). IgLON5 autoimmunity was considered the likely explanation for the peripheral neuropathy, as sural nerve biopsy findings were not typical for MGUS-related neuropathy. He received IVIg, oral prednisolone, plasma exchange and Rituximab. During follow-up, he progressed to multiple myeloma and commenced lenalidomide and dexamethasone.

Conclusion Our two cases and the few published reports suggest an association of peripheral neuropathy and IgLON5 autoimmunity. We recommend cases of IgLON5 autoimmunity undergo routine neurophysiological studies.

REFERENCES

1. Sabater L, Gaig C, Gelpi E, et al. A novel NREM and REM parasomnia with sleep breathing disorder associated with antibodies against IgLON5: a case series, pathological features, and characterisation of the antigen. *Lancet Neurology* 2014;**13**:575–586.
2. Gaig C, Graus F, Compta Y, et al. Clinical manifestations of the anti-IgLON5 disease. *Neurology* 2017;**88**:1736–1743.
3. Honorat JA, Komorowski L, Josephs KA, et al. IgLON5 antibody: neurological accompaniments and outcomes in 20 patients. *Neurol Neuroimmunol Neuroinflamm* 2017;**4**:e385. doi: 10.1212/NXI.0000000000000385
4. Wenninger S. Expanding the clinical spectrum of IgLON5-syndrome. *Journal of Neuromuscular Diseases* 2017;**4**:337–339.

082

FULMINANT ADEM MIMICKING A GLIAL TUMOUR

¹Jessica Qiu, ²Elizabeth Thompson, ³Michael J Fulham, ⁴Mrudula Krishnaswamy, ⁴Michael E Buckland, ⁵Liane Khoo, ⁶Nicolás Uriola, ¹Kaitlyn Parratt. ¹Department of Neurology, Royal Prince Alfred Hospital, Camperdown, NSW, Australia; ²Department of Radiology, Royal Prince Alfred Hospital, Sydney, NSW, Australia; ³Department of Molecular Imaging, Royal Prince Alfred Hospital, Sydney, NSW, Australia; ⁴Department of Neuropathology, Royal Prince Alfred Hospital, Sydney, NSW, Australia; ⁵Department of Haematology, Royal Prince Alfred Hospital, Sydney, NSW, Australia; ⁶Department of Immunology, Royal Prince Alfred Hospital, Sydney, NSW, Australia

10.1136/bmjno-2021-ANZAN.82

Introduction We describe an atypical case of fulminant acute disseminated encephalomyelitis (ADEM).

Case A 47 year-old Southeast Asian lady presented after developing headache, aphasia and right hemiparesis over four hours, preceded by dry cough for one week and fevers for two days. CT brain noted vasogenic oedema without enhancement in the left frontoparietal lobe, midline shift and incidental upper lobe consolidation and calcified hilar lymph nodes on CT chest. A provisional diagnosis of cerebral tuberculosis was made. MRI brain noted gross mass effect and T2 hyperintensity localised to the white matter, crossing the midline and extending directly to the pons without significant restricted diffusion. Ill-defined enhancement was noted without tuberculomas or leptomeningeal enhancement. MRI spine was unremarkable, as were extensive tests for infectious aetiologies on serum, sputum and CSF. A glial tumour was suspected; FDG-PET-CT did not show regions of increased metabolism. As the patient rapidly deteriorated, empirical corticosteroids, plasmapheresis and IVIg were commenced just prior to decompressive craniectomy and biopsy four days post-presentation. The biopsy demonstrated reactive astrocytosis and perivascular macrophages localised to the white matter, as well as perivascular and perivenular demyelination consistent with ADEM. Absence of a significant lymphocytic infiltrate may have been influenced by the short time to biopsy. The

patient made a remarkable recovery following cyclophosphamide, achieving independence in mobility and driving within two months.

Conclusion Atypical features of fulminant ADEM highlight the need for a high index of clinical suspicion and early institution of aggressive immunosuppressive therapy for a favourable outcome.

083

AN ATYPICAL CASE OF IDIOPATHIC INTRACRANIAL HYPERTENSION

¹Yasser M Mansour, ²Nick Saad, ²Simon Liew, ¹Sameen Haque. ¹Nepean Hospital, Kingswood, NSW, Australia; ²Marsden Eye Clinic, Parramatta, NSW, Australia

10.1136/bmjno-2021-ANZAN.83

Idiopathic Intracranial Hypertension (IIH) is an increasingly common condition that usually presents with younger obese female patients. Studies report between 74–94% of patients with a BMI >30, 1–5–95% with ages under 50⁽⁷⁾ and over 87–91% of the patients being overwhelmingly females.^{8–9}

In this case report, we present a 66-year-old man with a BMI of 24.7 kg/m² who was referred by his ophthalmologist with bilateral papilledema on the ophthalmic examination and OCTs. The 30-2 Humphrey Visual Field testing showed significant loss of his inferior field on the right side. There were also early field losses noted on the left side. Lumbar puncture showed a borderline elevated CSF opening pressure of 25 cmH₂O. Initial and subsequent MRI brain and orbits have shown constellation of findings consistent with idiopathic intracranial hypertension. Extensive investigations were carried out to identify any secondary cause. These included CT venogram, CT neck, chest abdomen and pelvis, serum and CSF testing for inflammatory/autoimmune, paraneoplastic, infectious and metabolic causes. His non-compliance with Acetazolamide led to clinical deterioration and optic atrophy on the right side. After 2 years of the onset, the patient is clinically stable on 250 mg TDS of Acetazolamide with normal CSF opening pressure on repeat testing recently.

REFERENCES

1. Durcan F, Corbett J, Wall M. The incidence of pseudotumor cerebri. *Archives of Neurology* 1988;**45**(8):875.
2. Galvin J, van Stavem G. Clinical characterization of idiopathic intracranial hypertension at the detroit medical center. *Journal of the Neurological Sciences* 2004;**223**(2):157–160.
3. Kesler A, Gadoth N. Epidemiology of idiopathic intracranial hypertension in Israel. *Journal of Neuro-Ophthalmology* 2001;**21**(1):12–14.
4. Radhakrishnan K. Idiopathic intracranial hypertension (pseudotumor cerebri). *Archives of Neurology* 1993;**50**(1):78.
5. Radhakrishnan K, Ahlskog J, Garrity J, Kurland L. Idiopathic intracranial hypertension. *Mayo Clinic Proceedings* 1994;**69**(2):169–180.
6. Radhakrishnan K, Sridharan R, Ashok P, Mousa M. Pseudotumour cerebri: incidence and pattern in North-Eastern Libya. *European Neurology* 1986;**25**(2):117–124.
7. Bruce B, Kedar S, van Stavem G, Corbett J, Newman N, Biousse V. Atypical idiopathic intracranial hypertension: normal BMI and older patients. *Neurology* 2010;**74**(22):1827–1832.
8. McCluskey G, Doherty-Allan R, McCarron P, Loftus A, McCarron L, Mulholland D, et al. Meta-analysis and systematic review of population-based epidemiological studies in idiopathic intracranial hypertension. *European Journal of Neurology* 2018;**25**(10):1218–1227.
9. Fraser J, Bruce B, Rucker J, Fraser L, Atkins E, Newman N, et al. Risk factors for idiopathic intracranial hypertension in men: a case-control study. *Journal of the Neurological Sciences* 2010;**290**(1–2):86–89.