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

1Hannah HF Ford, 2Clare CF Fraser, 3Emma ES Solly, 1,3Jeanne JF Fielding, 4Owen OW White, 5,6Anreke AVW van der Walt. 1Department of Neurosciences, The Alfred Hospital, Melbourne, VIC, Australia; 2Neuro-Ophthalmology, Sydney University, Sydney, 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; 6Multiple Sclerosis and Neuroimmunology (MSNI), The Alfred Hospital, Melbourne, 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-Methylenedioxy 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.

THE DIAGNOSTIC JOURNEY OF MITOCHONDRIAL DISEASE PATIENTS

11Laura I Rudaks, 12Eloise Watson, 1Michal Lubomski, 1Fabienne Edema-Hildebrand, 1Kate Ahmad, 1Christina Liang, 1Ryan Davis, 1Carolyn Sue. 1Department of Neurosciences, The Alfred Hospital, Melbourne, VIC, Australia; 2Neurology Department, Westmead Hospital, Westmead, NSW, Australia; 3Neurology Department, The Alfred Hospital, Melbourne, VIC, Australia; 4Neurology Department, Westmead Hospital, Westmead, NSW, Australia; 5Multiple Sclerosis and Neuroimmunology (MSNI), The Alfred Hospital, Melbourne, VIC, Australia; 6Multiple Sclerosis and Neuroimmunology Group, Monash University, Clayton, VIC, 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
1. 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
2. Watson E, Davis R, Sue C. New diagnostic pathways for mitochondrial disease. J Transl Genom Genom 2020;4:188–202.

IGLONS AUTOIMMUNITY IN TWO CASES WITH PERIPHERAL NERVOUS SYSTEM FEATURES

1Laura I Rudaks, 1,2Victor SC Fung, 1Jean-Pierre Halpern, 1Omar 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 dysphagia, glottis glossoptosis, unsteady gait, ataxia, 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 IVlg, oral prednisolone, azathioprine and plasma exchange.

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

BMJ Neurol Open 2021;3(Suppl 1):A1–A45

A29
kappa paraprotein, leading to a new diagnosis of monoclonal gammopathy of undetermined significance (MGUS). IgLONs autoimmunity was considered the likely explanation for the peripheral neuropathy, as sural nerve biopsy findings were not typical for MGUS-related neuropathy. He received IVlg, 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 IgLONs autoimmunity. We recommend cases of IgLONs autoimmunity undergo routine neuropsychological 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 IgLONs: a case series, pathological features, and characterisation of the antigen. Lancet Neurology 2014;13:575-586.

2. Gaig C, Graus F, Compta V, et al. Clinical manifestations of the anti-IgLONs disease. Neurology 2017;88:1736–1743.

3. Honorat JA, Komorowski L, Josephs KA, et al. IgLON5 antibody: neurological accompaniments and outcomes in 20 patients. Neurology Neuroimmunol Neuroinflamm 2017;4:e285. doi: 10.1212/NXI.0000000000000285

4. Wenninger S. Expanding the clinical spectrum of IgLON5-syndrome. Journal of Neuromuscular Diseases 2017;4:337–339.

O82 FULMINANT ADEM MIMICKING A GLIAL TUMOUR

1Jessica Qiu, 2Elizabeth Thompson, 3Michael J Fulham, 4Mrunula Krishnaswamy, 5Michael E Buckland, 1Lane Kito, 4Nicolas Umiola, 1Kathryn 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 Neurology, 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 and incidental upper lobe consolidation and calcified hilar lymph nodes were noted on the right 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 Acetzolamide with normal CSF opening pressure on repeat testing recently.

REFERENCES

1. Duncan F, Corbett J, Wall M. The incidence of pseudotumor cerebri. Archives of Neurology 1988;45(8):875.

2. Galvin J, van Stavern 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, Gadot H. 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, Ahlkog J, Garrant J, Kurland L. Idiopathic intracranial hypertension. Mayo Clinic Proceedings 1988;63(2):169–180.

6. Radhakrishnan K, Srisharan 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 Stavern G, Corbett J, Newman N, Biousses V. Atypical idiopathic intracranial hypertension: normal BMI and older patients. Neurology 2010;74(22):1827–1832.

8. McCluskey G, Doherty-Allan R, McCarron P, Lofus 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 2015;22(10):1218–1227.

9. Fraser I, Bruce B, Buckler I, Fraser I, 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.