

raises questions about possible immune or toxin-mediated mechanisms.

### 2308 CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY IN A PATIENT WITH MULTIPLE SCLEROSIS

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Inflammatory demyelinating diseases, resulting in immune mediated destruction of myelin sheaths surrounding axons, can affect both the central (CNS) and peripheral (PNS) nervous systems. Their clinical course can be acute and monophasic such as in Guilliane Barre syndrome and acute disseminated encephalomyelitis, or chronic and relapsing as in chronic inflammatory demyelinating polyneuropathy and multiple sclerosis, depending on localisation to the PNS or CNS. Despite a common pathophysiological outcome – i.e. destruction of myelin – co-occurrence of peripheral and central demyelinating disorders in a single individual is rare. Herein we present a patient diagnosed with both MS and CIDP. The case raises the possibility of a common immunopathogenic process, which may have important implications for treatment.

### 2309 ACBD5-RELATED RETINAL DYSTROPHY WITH LEUKODYSTROPHY DUE TO NOVEL MUTATIONS IN ACBD5 AND WITH ADDITIONAL FEATURES INCLUDING OVARIAN INSUFFICIENCY

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**Introduction** Acyl-CoA-binding domain (ACBD) proteins function in the peroxisomal sequestration, transport, and distribution of long-chain acyl-CoAs. *ACBD5* mutations result in retinal dystrophy with leukodystrophy (OMIM 616618) with accumulation of very long chain fatty acids (VLCFA). Only five cases of *ACBD5*-related retinal dystrophy are reported. We add an adult diagnosis of retinal dystrophy with leukodystrophy secondary to novel compound heterozygous *ACBD5* mutations.

**Case A** 29-year-old female, born to non-consanguineous parents developed multidirectional nystagmus at two months of age, followed by photophobia, leading to the diagnosis of rod monochromatism at 18 months. Electroretinogram suggested a rod-cone dystrophy. From age 2, progressive lower limb predominant weakness, spasticity, and ataxia developed, with superimposed reversible deterioration during fevers. She required a wheelchair by age 5. Additional features included progressive dysarthria, dysphagia and urinary dysfunction accompanied by gradual cognitive decline, and prominent emotional lability from adolescence. Other medical issues

included primary premature ovarian failure, osteoporosis and recurrent renal calculi.

Plasma VLCFAs were normal. Serial MRI scans of the brain demonstrated progressive hypomyelination involving frontal, occipital and parietal lobes, progressive widespread atrophy and abnormally small bony orbits. Spinal MRI demonstrated progressive spinal and conus atrophy. Brainstem auditory evoked potentials revealed symmetrical delay.

Trio exome sequencing demonstrated compound heterozygous truncating variants in *ACBD5*, c.979G>T (p.Gly327\*) and c.399del (p.Ile134Leufs\*6), both classified as pathogenic by ACMG criteria. Both parents were shown to be carriers of one variant, confirming variants were in trans.

**Conclusion** *ACBD5*-related retinal dystrophy with leukodystrophy, secondary to *ACBD5* mutations, is a rare multi-system slowly progressive peroxisomal disorder.

### 2310 BARORECEPTOR REFLEX FAILURE AS A LONG-TERM SEQUELA OF HEAD AND NECK IRRADIATION: A LITERATURE REVIEW AND CASE SERIES

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**Objectives** We present the largest case series to date of patients with long term follow up who developed clinically significant autonomic dysfunction due to baroreflex failure following neck irradiation for head and neck cancer (HNC) and Hodgkin's lymphoma.

**Methods** We retrospectively collated clinical, radiological, and autonomic testing data on nine patients who were referred to a quaternary institution in Sydney, Australia for evaluation of autonomic function following head and neck irradiation for malignancy.

**Results** All patients demonstrated baroreflex failure and had symptomatic orthostatic hypotension. The average time to symptom onset was 9.1 years (range 0 to 26 years). One patient demonstrated isolated baroreflex failure and two patients displayed baroreflex failure and segmental hypohidrosis, suggestive of local nerve damage involving efferent baroreceptor pathways. Five patients demonstrated significant cardiovagal and adrenergic dysfunction and one patient had severe cardiovagal and adrenergic dysfunction with additional features of a length-dependent neuropathy. Five of the six patients with severe pan-autonomic failure received concomitant chemotherapy. The features of baroreflex failure occurred independently of radiologically significant carotid atherosclerotic disease.

**Conclusion** This case series highlights the phenomenon of radiotherapy-induced damage to blood pressure regulatory structures and provides further commentary on the pathophysiology of baroreflex failure. The appearance of urinary frequency, gastrointestinal disturbance, or abnormal sweating in addition to postural symptoms was more common in patients with pan-autonomic failure. The constellation of presenting symptoms, as well as the presence of paroxysmal hypertension, can in turn guide pharmacotherapeutic management in these cases.