therapies. Methods: We describe a case of a 75-year-old female who presented with a rapidly progressive lower motor neuron syndrome leading to flaccid quadriparesis and complete loss of independence over a five month period. Results: Genetic testing demonstrated a heterozygous variant of uncertain significance in the SOD1 gene with a g > c point mutation at position 382 that has been described in one other patient in available literature. MR of the lumbar spine demonstrated abnormal smooth nerve root enhancement. Conclusions: This novel mutation in the SOD1 gene may be associated with a rapidly progressive phenotype of sporadic ALS. Ventral nerve root enhancement should not exclude a diagnosis of ALS especially in the absence of nodularity or nerve enlargement.

P.043

Plasmapheresis for treatment of light chain amyloidosis related myopathy

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Background: Light chain (AL) amyloidosis is a plasma cell disorder characterized by abnormal fibrillary light chain deposition causing cardiac, renal, hepatic, gastrointestinal and peripheral nervous system dysfunction. Muscle disease occurs in 1.5% of individuals causing progressive proximal weakness thus far considered untreatable. Methods: We reviewed two cases of AL amyloidosis associated myopathy at our institution who had robust response to plasmapheresis. Both were at stringent clinical response following CyBorME therapy during peak severity of their myopathy. Results: In case 1, a 70-year-old male with recently diagnosed kappa light chain multiple myeloma and cardiac/renal amyloidosis developed severe subacute proximal weakness preventing ambulation. CK was normal and electromyography was consistent with irritable myopathy. Deltoid biopsy showed perimysial and endomysial amyloidosis. A trial of plasmapheresis in a tapering schedule resulted in robust recovery of strength. In case 2, a 67-year-old female with recently diagnosed kappa light chain multiple myeloma with amyloidosis on fat pad aspirate developed severe subacute proximal weakness requiring prolonged hospital admission. CK was normal and electromyography demonstrated non-irritable myopathy. Bicep biopsy showed perivascular amyloidosis. A trial of plasmapheresis in a tapering schedule resulted in robust recovery of strength. Conclusions: Plasmapheresis is a novel and potentially effective treatment for patients with AL amyloidosis associated myopathy.

P.044

GMPPB mutation causes a muscular dystrophy-myasthenic spectrum

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Background: Mutations in GDP-Mannose Pyrophosphorylase B (GMPPB) cause a spectrum of disease ranging from muscular dystrophy to congenital myasthenic syndrome (CMS). Recognition of neuromuscular junction dysfunction has important treatment implications. Methods: We describe a person with GMPPB mutation causing an overlapping limb girdle muscular dystrophy - myasthenic syndrome with robust response to acetylcholinesterase inhibitors. We review the literature on the muscular dystrophy - CMS and explore the phenotypic features that aid in recognizing neuromuscular junction dysfunction. Results: A 35-year-old woman presented with a 10-year history of debilitating myalgias, symmetrical limb girdle and neck weakness, and chronic CK elevation. Electromyography showed a non-irritable myopathy. Biopsies were consistent with muscular dystrophy. Whole genome sequencing revealed two heterozygous pathogenic mutations in the GMPPB gene, giving a diagnosis of genetically confirmed limb girdle muscular dystrophy. Subsequently, repetitive nerve stimulation revealed decrement in the trapezius muscle suggestive of an overlap myasthenic syndrome. She was started on pyridostigmine resulting in recovery of full motor power with significant functional improvement. Conclusions: Identification and treatment of neuromuscular junction dysfunction caused by GMPPB mutations can significantly improve motor power and function. Early onset of progressive fatigable proximal weakness, spared ocular and facial muscles, and pyridostigmine responsiveness are important features of GMPPB-related CMS.

P.045

Biopsies of the transverse carpal ligament and tenosynovium for tissue confirmation of transthyretin amyloidosis

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Background: Transthyretin Amyloidosis (ATTR) is a common cause of both cardiomyopathy and carpal tunnel syndrome, with many patients needing carpal tunnel release (CTR). Although tafamidis is now an approved treatment of ATTR cardiomyopathy, insurers in most provinces require biopsy confirmation of amyloidosis. Endomyocardial biopsy is often the chosen approach due to optimal sensitivity, albeit with risk of serious adverse events such as stroke, cardiac tamponade, and arrhythmias. CTR may present an ideal opportunity for obtaining amyloidosis biopsy confirmation. Methods: ATTR patients undergoing CTR had biopsy of their transverse carpal ligament (TCL) and/or flexor tenosynovium to assess the sensitivity of both sites for biopsy confirmation of amyloidosis. Results: Twelve patients consecutively underwent biopsies during CTR, with 4 (33%) having bilateral CTR and biopsy. Among 16 TCL biopsies and 14 tenosynovium biopsies, 100% demonstrated amyloid deposition. Another patient had isolated tenosynovium biopsy without CTR and also demonstrated amyloidosis. There were no serious adverse events, and 1/13 (8%) had wound dehiscence requiring repeat suturing. Conclusions: Biopsy of the TCL and/or tenosynovium during CTR is safe, cost-effective, and sensitive, and may represent an alternative to endomyocardial biopsy in patients requiring tissue confirmation for tafamidis approval. ATTR patients eligible for tafamidis may benefit from early neurology assessment.