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Neuromuscular neurologists' experience in recognizing, diagnosing, and treating Long-chain fatty acid disorders (LC-FAOD): a national survey

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doi: 10.1017/cjn.2024.165

Background: LC-FAOD may be missed in neuromuscular (NM) clinics due to its rarity and absence from common NM genetic panels. The Canadian Neuromuscular Disease Registry (CNDR) collects real-world patient data and includes a network of clinician-investigators. Our objective was to inform future registry work by evaluating diagnosis pathways for LC-FAOD patients and estimating the number followed at Canadian NM clinics. Methods: A questionnaire was developed with an expert committee and circulated to 111 CNDR-affiliated NM neurologists. Results: 12 neurologists in 5 provinces, primarily adult-treating (n=8) completed the survey (10.8% response rate). Eleven (91.7%) practiced for >10 years. Agreement trends existed between definition of, and tests to evaluate, rhabdomyolysis. Four clinics routinely follow LC-FAOD patients. In the last 1-2 years, respondents diagnosed approximately 91 patients with LC-FAOD (mean=7.5 per clinic). 83.3% never received continuing education on LC-FAOD, though 75% indicated interest in expert-led webinars. Further data will be presented. Conclusions: Low sample size limits conclusions about LC-FAOD clinical trends. Results suggest LC-FAOD may be under-diagnosed or not routinely followed by NM specialists, limiting viability of an LC-FAOD registry. Practitioners may be interested in LC-FAOD-specific education. Future work could include collaboration with metabolic geneticists on education initiatives to raise awareness and improve care for these patients.

P.059

Weill-Marchesani Syndrome – a rare etiology for bilateral carpal tunnel syndrome in children

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doi: 10.1017/cjn.2024.166

Background: Carpal tunnel syndrome (CTS) is less common in children but can be associated with significant disability. Pediatric CTS can be associated with an underlying disorder most commonly storage disorders including mucopolysaccharidoses (MPS). We report a patient with bilateral severe CTS secondary to Weill-Marchesani Syndrome (WMS). Methods: A retrospective chart review was completed. Results: A five-year-old female presented with a three-year history of bilateral thumb weakness and insensate digits two and three. Nerve conduction studies (NCS) revealed severe bilateral CTS. She underwent bilateral carpal tunnel release (CTR). Unfortunately, post-operative NCS was unchanged. Ultrasound showed significant median nerve compression with flexor tendon thickening. Metabolic investigations showed no evidence of a storage disorder. Trio whole exome sequencing showed two de novo likely pathogenic variants in ADAMTS10: c.1174delC, p.H392TfsX9 and a deletion of exons 3-8. Her exam was also noted to show bilateral camptodactyly and brachydactyly, and bilateral cataracts characteristic of WMS. Conclusions: Identifying the etiology of CTS is important for management and prognosis. WMS is a genetic connective tissue disorder that can cause brachydactyly and abnormal tendon thickening, which can have implications on surgical outcomes. Awareness of this diagnosis prior to surgery would allow for better patient counseling and management decisions.

P.060

Provider and patient perspectives on outcome measure use in clinical care for chronic inflammatory neuropathy

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Background: The use of patient reported and functional outcome measures in routine practice enhances shared decision making and supports patient-centred care. This study compared the perspectives of Chronic Inflammatory Neuropathy (CIN) patients and providers regarding their experience using an outcome measure panel. Methods: A one year study was conducted to evaluate a nine measure outcome set in routine clinical practice for CIN. The panel included patient-reported outcome measures (e.g., I-RODS and EQ-5D-5L) and functional measures (e.g., grip strength). At the conclusion of the study, participants and providers completed an online questionnaire on their experience. Results: 25 patients and five providers completed the questionnaire. Both patients and providers reported benefit in tracking disease progression, supporting treatment-related decisions, and broadening views of health. Both groups agreed patient involvement in care was enhanced. Preference for specific measures, frequency, and data presentation differed. Providers emphasized integration into electronic medical records and streamlining processes. 100% of providers and 80% of patients wanted to continue completing outcome measures. Conclusions: CIN patients and providers recognize the value of integrating outcome measures into routine care. To effectively implement these measures in clinical settings, it is important to understand the patient and provider perspective and prevent unnecessary burdens to ensure sustainability of use.

P.062

Normal NCS in 42-year-old man with PMP22 duplication

*S Baker (Hamilton)**

doi: 10.1017/cjn.2024.168

Background: Charcot Marie Tooth disease is a polygenic disorder with cannonical features of distal amyotrophy, acrohypesthesia, and tight tendoachilles of either axonal (type 2) or demeylinating (type 1) varieties. Type 1 CMT patients are required to possess conduction slowing of a sufficient degree to qualify as demyelinating. Presented is a middle-aged man with an

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unremarkable neurologic exam and normal electrophysiology. Methods: Standard electrophysiological techniques were employed to obtain the nerve conduction data (Natus Nicolet EDX AT2; Middleton, WI, USA). Repeated next generation sequencing and deletion/duplication analyses were performed. Results: The nerve conduction studies showed no evidence of demyelination in the upper or lower extremeties. The duplication error was confirmed with repeat testing. The heterozygous PMP22 gene duplication encompassed the entire coding sequence involving exons 1-5. Conclusions: CMT1A accounts for the vast majority of dysmyelinating hereditary neuropathies. Phenotypic variability is well described. Presentations include (a) classic conduction slowing, (b) intermediate slowing, (c) conduction block, (d) HNPP-like, (e) absent CMAPs, and (f) normal NCSs in young infants. This is the first case of a neurologically intact adult with CMT1A. Cryptogenic genetic modifier-effect(s) are posited as a possible explanation of the lack of penetrance. Identifying the nature of this modification may prove instructive for future therapies.

P.063

Refractory pediatric CIDP converting to full recovery with rituximab

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Background: Chronic inflammatory demyelinating polyradiculoneuropathies (CIDP) is a rare, acquired polyneuropathy, especially in children, affecting the peripheral nervous system. It most commonly presents in a symmetric, proximal and distal, sensorimotor fashion. Immunosuppression and immunomanipulation are treatment modalities. We present a case of a 14 year old male with severe progressive CIDP who became refractory to steroid and IVIg but responded to Rituximab. Methods: Case presentation: A 14-year-old male with a history of asymmetric quadriparesis was diagnosed with CIDP. He had an initial partial response to IVIG and prednisone but then rapidly became refractory to even weekly IVIG and prednisone. Rituximab was therefore started. Results: Within 12 weeks his strength improved from quadriplegia to walker-assisted gait. By 22 weeks he achieved independent ambulation. His JAMAR hand grip increased from 0 to 28 kg. His worst recordable median conduction velocity (CV) improved from a nadir (MRC 0/5) of 14% of normal to 52% at full recovery (MRC 5/5). Conclusions: This case highlights several important clinical points. Dramatic improvement is possible in cases of quadriplegic CIDP. Strength recovery is not linearly related to CV recovery. There appears to be a role for polytherapy.

P.064

A case of late onset Pompe Disease presenting in 6th decade

*A Opala (Hamilton)**

doi: 10.1017/cjn.2024.170

Background: Late onset Pompe disease (LOPD), rare autosomal recessive lysosomal storage disease, resulting from mutation in alpha glucosidase enzyme (GAA) can present even in 6th decade of life. Slowly progressive, subtle, limb girdle pattern of weakness (LGPW), with auxiliary features such as ptosis, enlarged tongue, axial rigidity, facial diplegia, variable degree of respiratory weakness is not uncommon. Hypertrophic and electrical cardiac abnormalities are well described in LOPD. Methods: We present a case of 67-year-old male presenting with proximal weakness, subtle ptosis, bilateral quadriceps and shoulder girdle atrophy, and left toe numbness. PMHx: CABG, NSTEMI. Statin use. FMHx: noncontributory. Results: EMG: L5 radiculopathy, with unexpected myopathic units in hip/pelvic/shoulder girdle muscles with active denervation and muscle irritability. CK, CRP, SPEP, ANA, LFTs, HMG-CoA reductase: normal. GAA enzymatic activity=0.96μmol/L/hr (low), genetics: pathogenic variants in GAA gene: c.-32-13T>G and c.1194 +3G>C. ECHO: severe diastolic dysfunction, restrictive left ventricular filling. PFTs: normal. Diagnosed with LOPD, started on therapy. Conclusions: LOPD remains a differential for LGPW especially in older patient population with history of cardiopulmonary features. Age-appropriate conconminant pathologies may confound the diagnostic process.Symptoms may preceed diagnosis for years.GAA enzymatic activity followed by genetic testing remains readily available and can confirm diagnosis, preventing delay of approved therapy.

Other Adult Neurology

P.065

Understanding treatment barriers and adherence among people living with amyotrophic lateral sclerosis

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Background: Amyotrophic lateral sclerosis (ALS) is a fatal neurodegenerative disease with only four approved treatments in North America - sodium phenylbutyrate (PB) and ursodoxicoltaurine (TURSO, also known as taurursodiol), riluzole, edaravone, and tofersen. Poor treatment adherence reduces clinical effectiveness which can adversely impact disease progression and mortality rates. Understanding barriers and adherence to treatment in clinical practice is essential to address these issues. Methods: A scoping review was conducted in PubMed, Medline, Embase, and Web of Science. Retained studies were, (1) published in English, (2) included adults with ALS, (3) explored treatment non-adherence and/or identified barriers associated with non-adherence in ALS in real world clinical practice, (4) focused on ≥1 of the four approved ALS medications, and (5) used a measurement of adherence. Observational studies, real-world data, and case reports were included. Quality assessment was performed. Results: The review illustrated several knowledge gaps, including limited data on the incidence of non-adherence to ALS treatment in clinical practice, a lack of understanding regarding barriers to treatment adherence in ALS, and an absence of studies outside of western societies. Conclusions: We demonstrate a dearth of real-world data on treatment adherence in ALS and highlight opportunities for advancing research into this important area.