

**P.058****Neuromuscular neurologists' experience in recognizing, diagnosing, and treating Long-chain fatty acid disorders (LC-FAOD): a national survey**

*CD Kassardjian (Toronto)\* A Dyck (Calgary) S Andrews (Calgary) K Schellenberg (Saskatoon) H McMillan (Ottawa) V Hodgkinson (Calgary) L Korngut (Calgary) On behalf of the CNDR Investigator Network*

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**

*H Chiu (Ottawa) R Almarwani (Ottawa) H McMillan (Ottawa) J Richer (Ottawa) J Roth (Lucerne) A Yaworski (Ottawa)\**

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**

*CB Smith (Vancouver) K Beadon (Vancouver)\* E Ogalo (Vancouver) M Ashe (Vancouver) MM Mezei (Vancouver) KM Chapman (Vancouver)*

doi: 10.1017/cjn.2024.167

**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 canonical features of distal amyotrophy, acroparesthesia, and tight tendoachilles of either axonal (type 2) or demyelinating (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