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

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Background: Late onset Pompe disease (LOPD), rare autosomal recessive lysosomal storage disease, resulting from mutation in alpha glucosidase enzyme (GAA) can present even in 6<sup>th</sup>

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 concomminant 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, edarayone. 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.

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