Neuromuscular Ultrasound with EMG/NCS to evaluate rapidly progressing symptoms in a 17-year-old female with CMT 4J due to homozygous FIG4 mutation.: A Case Report

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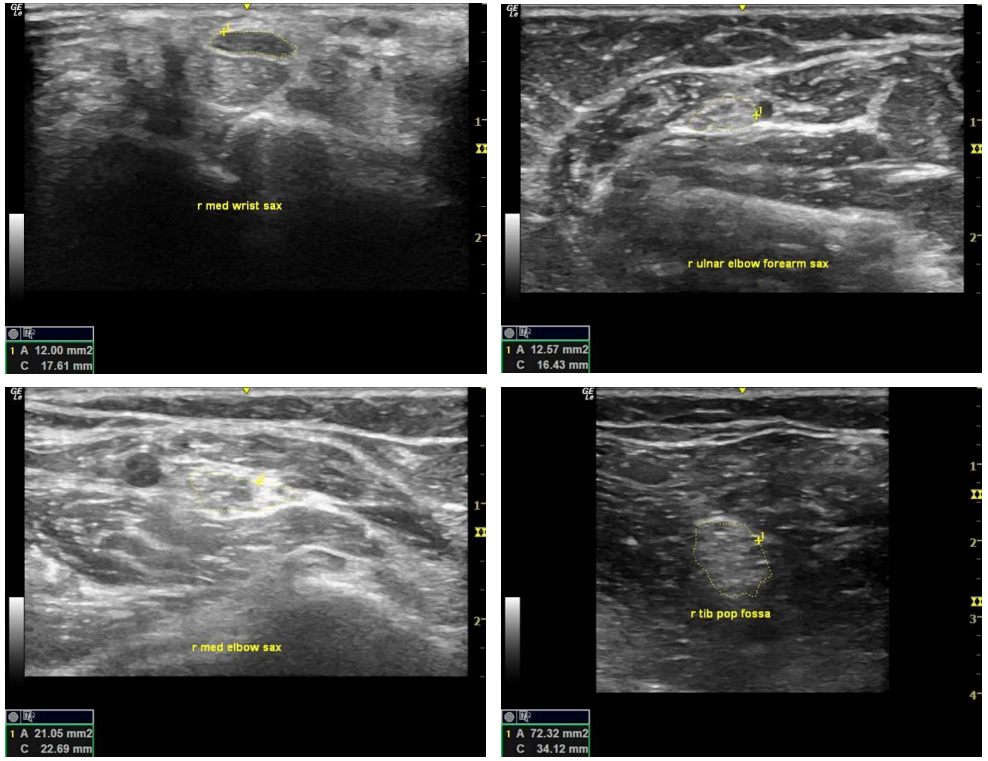
Case Description

Patient presented with acute worsening of lower extremity weakness and new onset hand weakness after discontinuing immunotherapy for 1 month. She had been diagnosed with CIDP by EMG at the age of 15 and started on immunotherapy with temporary improvement in her weakness. However, the weakness continued to progress and further investigation revealed homozygous FIG4 (exon 2, c. 112T>C (p.Ile41Thr)) mutation. Neuromuscular ultrasound was performed in conjunction with EMG/NCS demonstrating defuse enlargement of nerves with asymmetric segmental enlargement in the Tibal nerve at the popliteal fossa and ankle as well as in the median nerve at the wrist and antecubital fossa, the ulnar nerve at the forearm, the brachial plexus trunk, and C5 nerve root. NCV & EMG demonstrated significant progression in the decreased motor response with slowed conduction velocities, conduction block, and decreased amplitudes as compared to her study a year and a half prior.

Figures



Nerve and Location	Right CSA (mm2)	Left CSA (mm2)	Normal CSA (mm2)	Normal side to side difference (mm2)
Median Wrist	12.0	8.3	<14.6	<3.4
Median Antecubital	21.95	18.39	<13.2	<4.3
Ulnar Wrist	6.86	6.81	<8.1	<2.6
Ulnar Forearm	12.57	9.4	<8.3	<2.0
Ulnar Medial Epicondyle	8.65	12.9	<8.8	<2.2
Brachial Plexus Trunk	15.79	9.84	<11.1	<4.5
C5	11.1	17.66		
Fibular Pop Fossa	16.22	17.4	<20.9	<9.5
Tibial Pop Fossa	72.32	40.38	<55.9	<15.7
Tibial Ankle	22.95	26.01	<22.3	<5.7
Sural	BiFid 1.86/1.99	6.42	<8.9	<2.6

Conclusion

Neuromuscular Ultrasound can be a valuable addition to electro-diagnostic evaluation in rare genetic disorders and future studies are needed to evaluate whether it can be used to assess response to treatment

Discussion

Homogenous FIG4 mutations are a rare cause of Charcot-Marie-Tooth disease which causes motor and sensory neuropathies. The autosomal recessive neuropathy is categorized as a demyelinating process, axonal loss, or an intermediate form with both axonal loss and demyelinating processes present. Symptoms typically present in the first and second decades of life with a slow onset of symmetrical distal muscle weakness with sensory loss, decreased proprioception, loss of reflexes, and impaired temperature discrimination. Diagnosis is made through history and physical exam, nerve conduction studies, and verified by genetic testing. Neuromuscular ultrasound has previously demonstrated segmental nerve enlargement in CMT4J patients in distinct areas not susceptible to nerve entrapment. The unique asymmetrical presentation of nerve enlargement is observed in this patient and supports its possible use as a diagnostic measure.

References

1. Bo Hu et al. “Myelin Abnormality in Charcot–Marie– Tooth type 4J Recapitulates Features of Acquired Demyelination.” ANN NEUROL 2018;83:756–770
2. Castoro R, Crisp J, Caress JB, Li J, Cartwright MS. Segmental nerve enlargement in CMT4J. Muscle Nerve. 2020 Jun;61(6):E44-E46. doi: 10.1002/mus.26873. Epub 2020 Apr 4. PMID: 32239724.
3. Walker, Francis and Michael Cartwright. *Neuromuscular Ultrasound.* Elsevier Inc, 2011