



Nerve ultrasound evaluation in patients with Friedreich's ataxia

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Background: Friedreich's ataxia (FRDA) is a rare autosomal recessive disorder due to GAA triplet expansion of the frataxin gene. Besides ataxia, FRDA is characterized also by peripheral nervous system manifestation, namely sensory axonal neuropathy.

Aim of the study. To assess nerve US in patients with FRDA and sensory axonal neuropathy.

Patients and method: 8 patients (5 women, 3 men; mean age 34.6 yrs, range 20-45) with FRDA (confirmed by GAA triplet expansion of the frataxin gene) were recruited from the Neurology Clinic of Naples and Padua. Extensive neurophysiological examination was performed in all the patients.

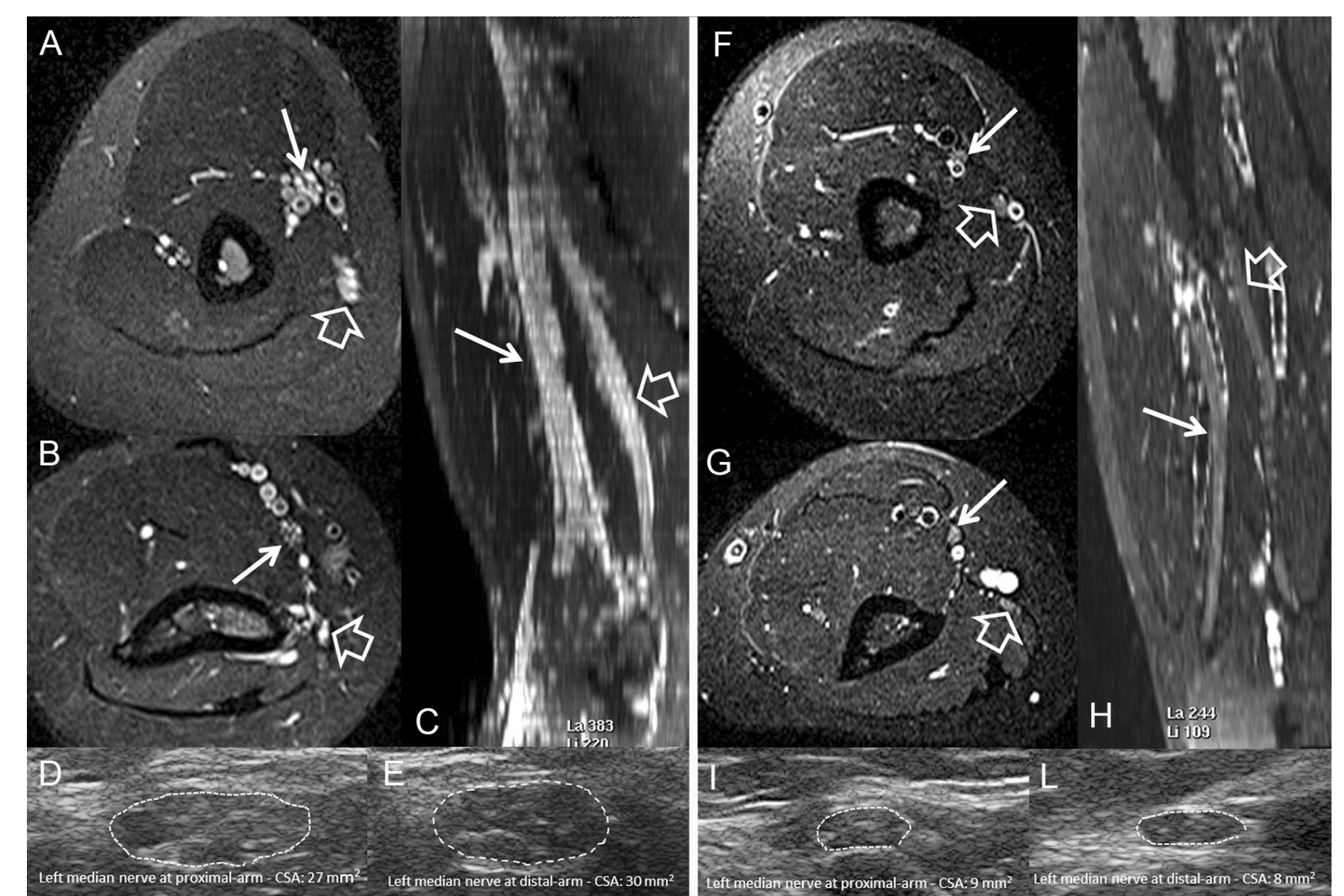
Ultrasound evaluation (18 MHz Esaote Mylab 70 Xvision, Genova, Italy) was performed at four limbs. CSA of median nerve at wrist, mid-forearm, elbow, mid-upper arm and axilla were performed. Measurement of cross-sectional-area (CSA) of ulnar nerve at wrist, cubital, mid-upper arm and axilla were performed. CSA of radial nerve at arm, posterior interosseus nerve, peroneal nerve at fibular head, tibial nerve at popliteal region and sural nerve was also measured. All the measurements were performed bilaterally. Echogenicity was also evaluated. Spearman test was performed to obtain correlation coefficient between CSA and triplet expansion, age and SAP. One patient underwent also MR Neurography.

Results: All patients presented with widespread SAP reduction (sensory axonal neuropathy). 7/8 patients (88%) presented with increased CSA of median and ulnar nerves at arm and axilla, while 2/8 (25%) and 0/8 presented with increased CSA of median and ulnar nerves at elbow. Mean median nerve CSA (right and left) at mid-upper arm were 21 mm² (normal values < 11 mm²), mean ulnar nerve CSA (right and left) at mid-upper arm were 14 mm² (normal values < 8 mm²). Mean median nerve CSA (right and left) at axilla were 14 mm² (normal values < 11 mm²), mean ulnar nerve CSA (right and left) at axilla were 10 mm² (normal values < 8 mm²). Mean CSA of median and ulnar nerve at wrist, mid-forearm, and elbow and mean CSA of radial nerve, posterior interosseus nerve, peroneal nerve, tibial nerve and sural nerve resulted within the normal values. The correlations between CSA and triplet expansion, age and SAP did not showed statistical significance except for the correlation between left ulnar nerve CSA at arm and age of onset (r:-0.77).

The patient who underwent MR Neurography showed diffuse swelling and signal hyperintensity of median and ulnar nerves at the arm.

Patient	Median Nerve										Ulnar Nerve							
	Right					Left					Right				Left			
	Wrist	Forearm	Elbow	Arm	Axilla	Wrist	Forearm	Elbow	Arm	Axilla	Wrist	Elbow	Arm	Axilla	Wrist	Elbow	Arm	Axilla
1	6	4	12	19	15	5	6	10	17	16	3	6	14	17	4	6	16	7
2	4	7	11	21	15	5	6	13	23	17	4	9	16	13	3	6	18	12
3	5	5	7	19	9	6	5	9	19	10	4	7	9	6	3	7	8	6
4	5	4	7	15	11	5	4	8	14	9	3	5	11	10	3	7	12	9
5	8	8	14	38	38	12	9	17	24	16	5	9	14	16	5	9	9	12
6	5	6	12	26	12	5	8	10	26	9	3	7	18	9	3	6	18	6
7	10			28	14	11	8	8	27	14			23			10	20	14
8	4	4	10	10	7	4	4	4	9	8	3	5	5	4	3	6	6	7

CSA values (mm²), abnormal values are red



MRN (A-C) and US (D,E) of the patient's left arm, compared to a healthy subject MRN (F-H) and US (I,L). Axial MRN at proximal (A,F) and distal (B,G) arm, reformatted coronal oblique view of the arm (C,H). US of the median nerve at proximal (D,I) and distal (E,L) arm. At MRN (A-C) the median (arrow) and ulnar (empty arrow) nerves of the patient appear significantly enlarged and markedly hyperintense with fascicular hypertrophy over a large segment (13 cm approximately), compared to the healthy subject (35-yo female) (F-H). Similar findings are detected with US, which shows increased CSA and heterogeneous fascicular hypertrophy of the patient's right median nerve (D,E), compared to the healthy subject (I,L).

Discussion and conclusions: FRDA patients presented with an axonal neuropathy characterized, at ultrasound, by marked nerve enlargement at the upper limbs. Nerve enlargement was almost always limited to nerves at mid-upper arms. This finding seems to represent a peculiar ultrasound feature in FRDA, but its pathophysiologic meaning remains unclear.