Non-familial case of relapsing neuralgic amyotrophy with paediatric onset

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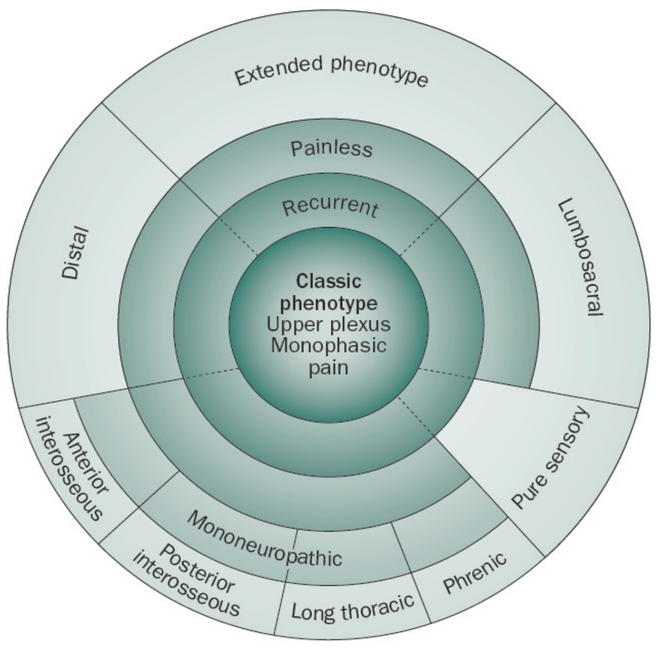
Introduction

Neuralgic amyotrophy is a peripheral neuropathy involving the brachial plexus and, less often, other peripheral nerves (phrenic nerve, lumbosacral plexus). The etiopathogenesis of Neuralgic amyotrophy is not completely understood, three main factors are involved: mechanical vulnerability, autoimmunity, and genetic predisposition. Neuralgic amyotrophy mainly involves the upper trunk of the brachial plexus, and each attack is characterized by extreme pain at the onset followed by paresis and atrophy; hypestesia may be present. Painless attacks have also been described, and they are frequent in hereditary Neuralgic amyotrophy. Neuralgic amyotrophy can have monophasic or relapsing course, and may be idiopathic or hereditary (autosomal dominant trait), linked to point mutation or duplication in the SEPT9 gene on chromosome 17q25. Hereditary Neuralgic amyotrophy shows more often paediatric onset, familial history, more relapses and nerve involvement outside the

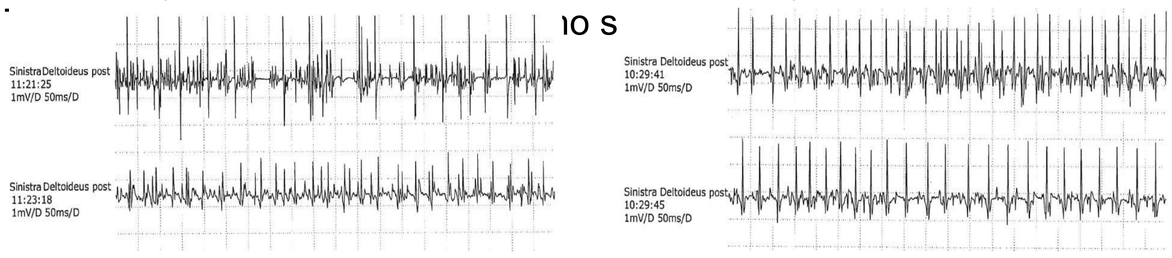
Case description

We report the case of a 50 years old man, with no familial history, affected by an alternating relapsing Neuralgic Amyotrophy.
The first relapse took place when he was 7 years old, and was characterized by left deltoid muscle palsy. The EMG study showed prolonged polyphasic potentials in the deltoid muscle. At the age of 20, he suffered a sudden painful palsy of left arm after physical exercise, which spontaneously improved without treatment. No diagnosis was made at the time.
He came to our attention in 1998 for a subacute onset of right deltoid and fingers' extensor palsy. The EMG study at rest showed spontaneous activity (fibrillation), and no voluntary response in right deltoid and in the extensor pollicis longus muscles. The axillary nerve motor conduction velocity was undeterminable. The cervical spine MRI showed no disc herniations.
A DNA analysis for CMT/HNPP gene mutation was performed to rule out a Hereditary Neuropathy with Liability to Pressure Palsies and resulted negative for 17p11.212 deletions.
He was diagnosed Neuralgic Amyotrophy and was treated with oral corticosteroids.
A clinical and neurophysiological improvement was observed, with evidence of mild voluntary muscle activity and motor axillary nerve velocity reduction.

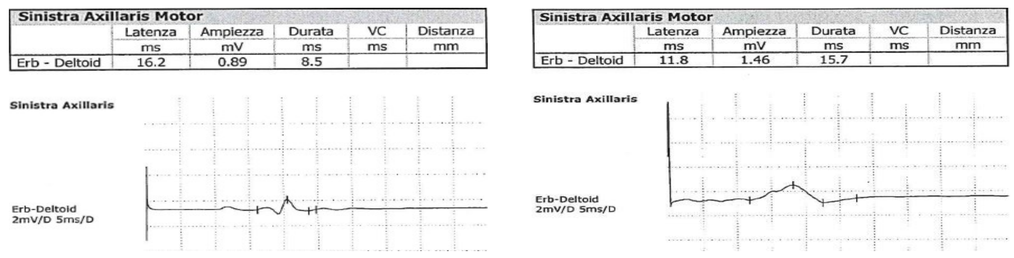


Van Alfen, Nat Rev Neurol, 2011

In 2016 the patient presented a subacute pain in the left deltoid area, followed by muscle atrophy. The EMG study showed bilateral axillary nerve involvement. The ENG study showed an improvement of the axillary nerve amplitude on the right side.



EMG study before (left) and after (right) steroid treatment



Sinistra Axillaris Motor	Latenza	Ampiezza	Durata	VC	Distanza
	ms	mV	ms	ms	mm
Erb - Deltoid	16.2	0.89	8.5		

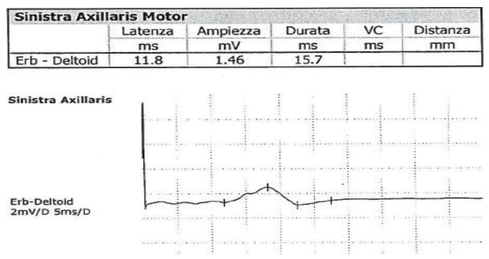
Sinistra Axillaris
Erb-Deltoid
2mV/D 5ms/D

Sinistra Axillaris Motor	Latenza	Ampiezza	Durata	VC	Distanza
	ms	mV	ms	ms	mm
Erb - Deltoid	11.8	1.46	15.7		

Sinistra Axillaris
Erb-Deltoid
2mV/D 5ms/D

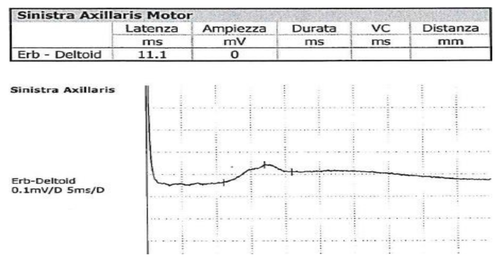
ENG study before (left) and after (right) steroid treatment

The following year, the patient suffered a sudden pain in the left shoulder, which was similar to the previous relapses, but without consequent motor symptoms. The ENG study showed a worsening of left axillary nerve amplitude, while a cervical MRI scan did not find any cervical compression: a pure sensory relapse was hence supposed.
The patient spontaneously improved over few weeks without treatment.



Sinistra Axillaris Motor	Latenza	Ampiezza	Durata	VC	Distanza
	ms	mV	ms	ms	mm
Erb - Deltoid	11.8	1.46	15.7		

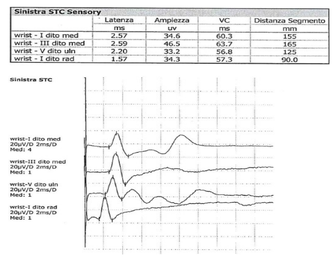
Sinistra Axillaris
Erb-Deltoid
2mV/D 5ms/D



Sinistra Axillaris Motor	Latenza	Ampiezza	Durata	VC	Distanza
	ms	mV	ms	ms	mm
Erb - Deltoid	11.1	0			

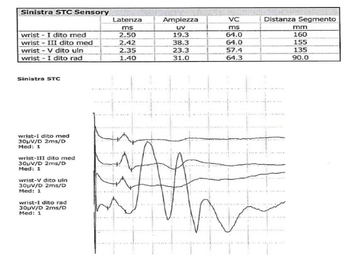
Sinistra Axillaris
Erb-Deltoid
0.1mV/D 5ms/D

ENG study before (left) and after (right) the sensory relapse



Sinistra STC Sensory	Latenza	Ampiezza	VC	Distanza Segmento
	ms	uV	ms	mm
wrist - I dito med	2.57	34.6	60.3	155
wrist - III dito med	2.59	46.5	63.7	165
wrist - V dito uln	2.20	33.2	56.8	125
wrist - I dito rad	1.57	34.3	57.3	90.0

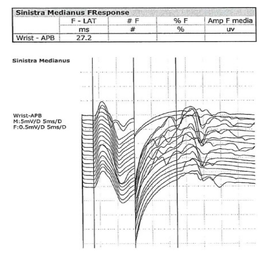
Sinistra STC



Sinistra STC Sensory	Latenza	Ampiezza	VC	Distanza Segmento
	ms	uV	ms	mm
wrist - I dito med	2.50	19.3	64.0	160
wrist - III dito med	2.42	38.3	64.0	155
wrist - V dito uln	2.35	23.3	57.4	135
wrist - I dito rad	1.40	31.0	64.3	90.0

Sinistra STC

Sensory study before (left) and after (right) the sensory relapse



Sinistra Medianus FResponse	F - LAT	# F	% F	Amp F media
	ms	#	%	uV
Wrist - APB	27.2			

Sinistra Medianus

F – wave

Discussion

We report a case of relapsing Neuralgic Amyotrophy with paediatric onset and multiple, bilateral relapses which are frequently linked with Hereditary Neuralgic Amyotrophy. However our patient did not have a positive familial history of disease. The genetic testing for HNPP mutations excluded a possible differential diagnosis of relapsing Neuralgic Amyotrophy. Our patient experienced many relapses and he was treated with oral corticosteroids, which are known to hasten the recovery. Corticosteroids, however, do not prevent further relapses. No preventive treatment is available for Neuralgic Amyotrophy.

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