Neuralgic Amyotrophy: a rare pediatric case associated with Claude Bernard Horner syndrome

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1 INTRODUCTION

Neuralgic Amyotrophy (NA) is a likely inflammatory neurological condition whose typical presentation is sudden onset of unbearable pain, followed by weakness and atrophy in the brachial plexus distribution as the pain subsides. The onset is more frequent in adult males and uncommon in childhood. Rare cases associated with Claude Bernard Horner (CBH) syndrome have been described.

2 MATERIALS AND METHODS

We report a pediatric NA associated with CBH syndrome in a Caucasian 3-year-old boy.

The patient underwent extensive investigations including lumbar puncture (LP), laboratory and genetic tests, neurophysiological/EMG evaluation, brain and spine MRI, and PET/MRI.

3 RESULTS

A previously healthy 3-year-old boy presented with sudden onset of right shoulder pain with nighttime awakenings followed by right arm weakness and scapular winging. A few days later, he also complained of left distal radial nerve palsy. Two weeks before, the child had developed left upper eyelid drooping and miosis consistent with a CBH syndrome. Examination also showed skin creases at four limbs.
Cervical plexus MRI revealed multifocal inflammation of the brachial plexus, involving bilateral post-ganglion roots and radial nerves (Figure 1).
EMG showed denervation of the right infraspinatus and deltoid muscles, and of the left triceps brachii and common extensor muscles. Nerve ultrasound showed thickening of the right C5-C6-C7 roots and left radial nerve (figure 2). Cerebrospinal fluid analysis and a whole-body PET-MRI were unremarkable. Genetic panel study including PMP22 and SEPT9 mutations turned out negative.
The pain subsided quickly and stably with gabapentin. Soon after the boy was started on three monthly courses of intravenous immunoglobulin with no benefit. After intensive physical therapy, the deficits slowly and gradually improved in the following 12 months. Two years later, the boy experienced a clinical relapse of left shoulder pain soon treated with corticosteroid. At last follow up, 3 years after the onset, neurological examination exhibited left scapular winging, left trapezius atrophy and marked improvement of anisocoria with complete recovery of the motility at four limbs. He continues a rehabilitation program focused on regaining motor control.

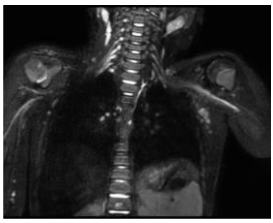

Figure.1 MRI of the brachial plexus. *T2/STIR hyperintensity of the roots from C5 to T1*

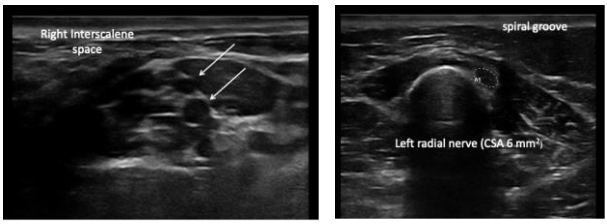

Figure.2 Nerve US. *Enlargement of the right C5-C6-C7 roots in the interscalene space and of the left radial nerve.*

4 CONCLUSION

NA is rare in childhood and even rarer is its association with CBH syndrome. Unusual skin fold and creases have been described in hereditary neuralgic amyotrophy even though in our patient SEPT9 mutation was excluded pointing to a likely broader phenotypic spectrum of the idiopathic form or to a novel mutation not yet identified. To date, this is the first case of NA associated with CBH syndrome in a pediatric patient.

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