Available online at www.sciencedirect.com

ScienceDirect

Neuromuscular Disorders 29 (2019) 398–400

www.elsevier.com/locate/nmd

Case report

Wrist drop in an arcade dancing game: Unusual sudden bilateral radial palsy

Gabriel Cea[a,b,*], Juan Pablo Contreras[a], Shirley Aguilar[b], Julia Vera[a]

[a] Departamento de Ciencias Neurológicas, Facultad de Medicina, Universidad de Chile, Avda Infante 553, Providencia, Santiago, Chile
[b] Servicio de Neurología, Hospital del Salvador, Santiago, Chile

Received 26 November 2018; accepted 12 February 2019

Abstract

Bilateral simultaneous radial palsy is uncommon, and the few cases reported in the literature are due to compressive injuries, such as in the use of axillary crutch or birthing bar during labor. We present a patient who developed a severe bilateral palsy after playing in a dancing simulator machine. The patient's position during the game was a combination of wrist extension, elbow flexion, retroversion of arms and a degree of minor torsion of both upper limbs. This mechanism has not been reported as a cause of neuropathic damage. An underlying neuropathy was suspected, and most acquired causes of neuropathy were excluded. A sequence analysis showed a novel point mutation in NM_000304.3(PMP22):c.83G>A (p.Trp28Ter), an heterozygous pathogenic variant. Hereditary neuropathy with liability to pressure palsies is an autosomal dominant disorder characterized by recurrent painless entrapment neuropathies; no case of bilateral simultaneous radial paralysis has been reported previously.

Keywords: Hereditary neuropathy with liability to pressure palsies; Radial nerve lesion; PMP22; Hereditary neuropathy; HNPP; Entrapment neuropathies.

1. Introduction

Bilateral radial palsy is an uncommon clinical presentation, and the few cases reported in the literature are due to compressive injuries, such as in the use of axillary crutch [1] or birthing bar during labor [2]. Non-compressive demyelinated inflammatory mononeuropathy has been reported [3] but it is not of sudden onset nor does it present bilaterally simultaneously. One of the largest series of radial palsy describes 103 cases of which only 3 were bilateral: one was a bilateral sleep palsy, one was a mononeuropathy multiplex associated to HIV infection and the third was entrapment in the supinator channel associated with neuropathy of unclear origin [4].

No cases have been reported of hereditary neuropathy with susceptibility to pressure palsy (HNPP), as a cause of bilateral simultaneous radial paralysis. Hereditary neuropathy with liability to pressure palsies (HNPP) is an autosomal dominant disorder characterized by recurrent painless entrapment neuropathies. Focal neuropathies mainly occur at common entrapment sites: carpal tunnel at the wrist, ulnar groove at the elbow, or head of the fibula at the knee. Brachial plexus palsies rarely occur and are usually painless [5].

We present a patient with bilateral wrist drop due to a mechanism that, to our knowledge, has not been reported in the literature. We discuss a hypothetical mechanism and report a new genetic defect for HNPP.

2. Case report

A 20-year-old man was playing on a dancing simulator machine for one or two minutes when he noticed numbness in the dorsal aspect of his left thumb. He carried on for another couple of minutes in the same position when he noticed his

* Corresponding author at: Departamento de Ciencias Neurológicas, Facultad de Medicina, Universidad de Chile, Avda Infante 553, Providencia, Santiago, Chile.
E-mail address: jcea@med.uchile.cl (G. Cea).

https://doi.org/10.1016/j.nmd.2019.02.005
0960-8966/

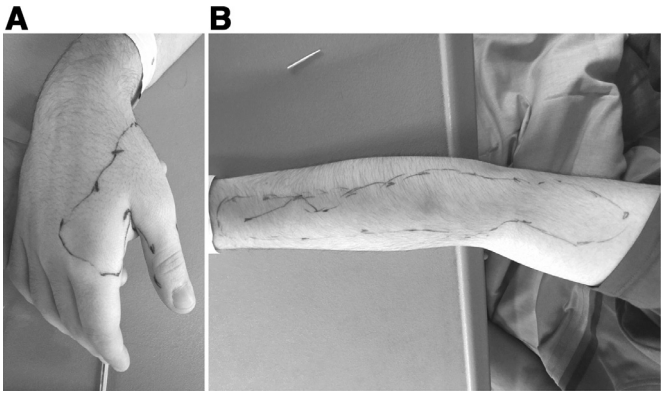

Fig. 1. (A) Right hand with wrist and finger drop. Note the zone of sensory loss over the lateral dorsum of the hand and part of the thumb corresponding to the area innervated by the superficial radial sensory nerve, and (B) Right forearm and arm with sensory loss in the distribution of the posterior cutaneous nerve of the forearm and lower lateral cutaneous nerve of the arm.

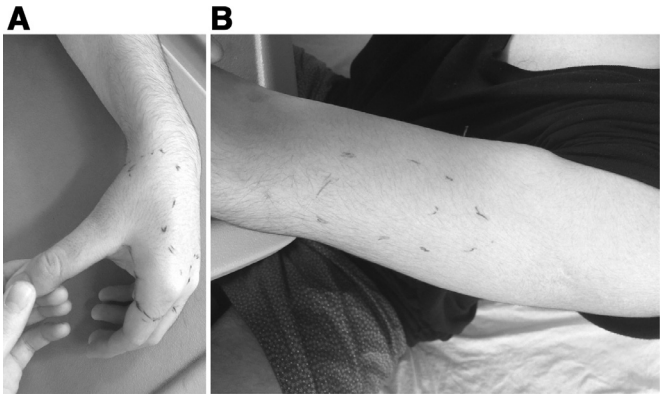

Fig. 2. (A) Left hand with wrist and finger drop. Note the zone of sensory loss over the lateral dorsum of the hand, and part of the thumb and index finger corresponding to the distribution of the superficial radial sensory nerve, and (B) Left arm showing a patch of sensory loss in the distribution of the lower lateral cutaneous nerve of the arm and no sensory loss in the area innervated by the posterior cutaneous nerve of the forearm.

right thumb went numb too, and then the numbness spread to the dorsal aspect of his right forearm and lateral aspects of both arms. He also noticed hand weakness and that he was unable to extend both wrists and all fingers. As is usual in dancing simulator machines he was holding a bar behind his back with both hands to keep himself steady and he was supporting some of his weight with his arms while he moved his feet to the rhythm of the music. During this manoeuvre, his arms were in retroversion, his forearms flexed and his wrists in extension.

He consulted in two private clinics without receiving much help, and so two days later, he came to the Emergency Department of our hospital. Clinical examination revealed severe bilateral weakness grade 1/5 in the Medical Research Council (MRC) scale, in the extension of both wrists and all fingers; hypoesthesia to pin-prick and cotton touch in the dorsal aspect of both thumbs, the lateral third of the dorsal side of both hands, the dorsal side of his right forearm and the inferior two thirds of the lateral aspect of both arms (see Figs. 1 and 2). The rest of his neurological examination was unremarkable. He stated no family history of genetic or neurologic disorders, although he had no contact with his father since infancy.

The following laboratory studies were all within normal range: full blood count, erythrocyte sedimentation rate, C-reactive protein, serum creatinine, electrolytes, creatine kinase, cerebrospinal fluid analysis, antinuclear antibody panel, antineutrophil cytoplasmic antibodies panel, complement components 3 and 4, serum levels of immunoglobulin (IgA, IgG, IgM), serum and urine protein electrophoresis, serum free light chains (Kappa, Lambda), HIV test, antibodies against hepatitis C virus, hepatitis B surface antigen and serum VDRL. An MRI of the cervical spinal cord and brachial plexus was unremarkable.

Bilateral radial nerve conduction studies performed six days after the event revealed signs suggestive of motor conduction block when stimulated in the axilla and there was a drop in the potential amplitude in the right radial sensory nerve. The electromyography did not show signs of denervation at any time, but there was no activity in the *extensor digitorum communis*, *extensor carpi radialis* and only minimal activity in *the brachioradilis* in both forearms. The motor conduction velocity in his legs was normal, with a prolonged latency and low amplitude potential in the left superficial peroneal nerve. Multiplex ligation-dependent probe amplification (MLPA) assay did not reveal deletions or duplications in the PMP22 gene.

His condition did not change for about a month. Then, he started a slow recovery, and six months later all his sensory symptoms had faded away. However, his muscle power did not recover entirely: finger and carpal extension in both upper limbs was MRC grade 4/5, slightly better on the left. Despite the negative result on MLPA, we still thought a genetic cause was most likely. We arranged a sequence analysis, at the Clinical Laboratory Invitae, San Francisco, USA, that revealed a novel point mutation in NM_000304.3 (PMP22):c.83G>A (p.Trp28Ter), an heterozygous pathogenic variant.

3. Discussion

This patient's presentation poses several challenges to understand the mechanism of injury and to localize the lesion. The patient's position during the game was a combination of wrist extension, elbow flexion, retroversion of arms and perhaps a degree of minor torsion of all segments in both upper limbs including the scapula, which could result in traction and compressive injury of the radial nerve or the posterior cord of the brachial plexus. The windmill pitcher's radial neuropathy, due to the "windmill" pitching motion of softball, proposes a similar mechanism but with a much strongest force [6]. The most likely location of our patient's radial nerve lesions is just above the spiral groove before it gives off the lower lateral cutaneous nerve of the arm. However, the sparing of the posterior antebrachial cutaneous nerve on the left is puzzling but could be explained by

a selective fascicular involvement. This has been proposed in radial nerve injuries where there is often a significant variability of the motor and sensory deficits [7]. We cannot exclude a torsion and stretch further up in the brachial plexus as signs of a block in the radial nerve are always difficult to establish [8]. We found no signs of denervation with any increase in recruitment during effort, as in demyelinated lesions.

The force applied during the game that he was playing did not seem to merit a severe radial motor deficit, unlike the tremendous force of the "windmill" pitching motion of softball. This provoked us to continue the search for an underlying cause. However, tests for an acquired underlying neuropathy were all negative, and the MLPA assay of the PMP22 gene did not find any deletion. A genetic neuropathy was still highly likely, and this was finally found on sequence analysis. Loss-of-function variants in PMP22 are known to be pathogenic [9], and this sequence change creates a premature translational stop signal in the gene with an expected absent or disrupted protein product. Point mutations that result in a premature stop codon, as found in this case, have been previously described in other HNPP cases [10–14]. However, this variant has not been reported in individuals with a PMP22-related disease in the literature or in population databases.

Bilateral radial palsy is uncommon and when presenting in a young patient without a reasonable precipitating factor, an underlying neuropathy should always be suspected. To our knowledge, this is the first case of HNPP presenting with bilateral radial nerve palsy and the described pathogenic variant has not been reported before.

Acknowledgments

We would like to acknowledge our patient and his family. We acknowledge Invitae, in Santiago, Chile for arranging the sequence analysis of the genetic study.

References

[1] Poddar SB, Gitelis S, Heydemann PT, Piasecki P. Bilateral predominant radial nerve crutch palsy. A case report. Clin Orthop Relat Res 1993;297:245–6.
[2] Roubal PJ, Chavinson AH, LaGrandeur RM. Bilateral radial nerve palsies from use of the standard birthing bar. Obstet Gynecol 1996;87:820–1.
[3] Garnero M, Martinoli C, Tagliafico A, et al. Dysimmune mononeuropathies: a diagnosis not to be missed. Muscle Nerve 2016;54:1145–6.
[4] Sturzenegger M, Rutz M. Bilateral radial nerve paralysis. Diagnostic and differential diagnostic aspects. Schweiz Med Wochenschr 1990;120:1325–34.
[5] Stogbauer F, Young P, Kuhlenbaumer G, De Jonghe P, Timmerman V. Hereditary recurrent focal neuropathies: clinical and molecular features. Neurology 2000;54:546–51.
[6] Sinson G, Zager EL, Kline DG. Windmill pitcher's radial neuropathy. Neurosurgery 1994;34:1087–9 discussion 1089-90.
[7] Stewart JD. Peripheral nerve fascicles: anatomy and clinical relevance. Muscle Nerve 2003;28:525–41.
[8] Kimura J. Electrodiagnosis in diseases of nerve and muscle: principles and practice. 4th ed. New York; Oxford: Oxford University Press; 2013. ed.
[9] Li J, Parker B, Martyn C, Natarajan C, Guo J. The PMP22 gene and its related diseases. Mol Neurobiol 2013;47:673–98.
[10] Luigetti M, Conte A, Madia F, et al. A new single-nucleotide deletion of PMP22 in an HNPP family without recurrent palsies. Muscle Nerve 2008;38:1060–4.
[11] Muglia M, Patitucci A, Rizzi R, et al. A novel point mutation in PMP22 gene in an Italian family with hereditary neuropathy with liability to pressure palsies. J Neurol Sci 2007;263:194–7.
[12] 3rd workshop of the European CMT consortium: 54th ENMC international workshop on genotype/phenotype correlations in Charcot-Marie-Tooth type 1 and hereditary neuropathy with liability to pressure palsies 28-30 November 1997, Naarden, The Netherlands. Neuromuscul Disord 1998;8:591–603.
[13] Pareyson D, Taroni F. Deletion of the PMP22 gene and hereditary neuropathy with liability to pressure palsies. Curr Opin Neurol 1996;9:348–54.
[14] Pegoraro E, Gavassini BF, Benedetti S, et al. Co-segregation of LMNA and PMP22 gene mutations in the same family. Neuromuscul Disord 2005;15:858–62.