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Hereditary Neuropathy with Liability to Pressure Palsies: A Case Report

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Abstract **Background**: Hereditary neuropathy with liability to pressure palsies (HNPP) is an uncommon diagnosis that should be considered in patients with multiple compressive neuropathies. This case reports highlights a relatively rare neuropathy with genetic and occupational predisposition, probably first to be reported in the united arab emirates. **Case Presentation**: We present the case of a 41 years old male who presented with right hand numbness and weakness. Electrodiagnostic testing revealed multifocal neuropathy. Subsequent genetic testing identified heterogeneous deletion of the PMP22 gene to confirm the diagnosis of HNPP. **Conclusions**: Hereditary neuropathy with liability to pressure palsies can present as a multifocal neuropathy. Occupational history along with genetic testing plays a pivotal role to make the diagnosis.

Keywords: *hereditary neuropathy with liability to pressure palsies, compressive neuropathies, genetic testing*

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1. Background

Hereditary neuropathy with liability to pressure palsies is a demyelinating neuropathy that affects peripheral nerves. It is estimated to occur in 2 to 5 per 100,000 individuals [1]. In people with this disorder, the peripheral nerves are over sensitive to pressure, such as leaning on an elbow, or sitting without changing position for a prolonged period of time. These activities would not normally cause any sensory defects in individuals without this condition [2].

Hereditary neuropathy with liability to pressure palsies is characterized by episodic, tingling sensation and occasional impairment of muscle motor function (palsy) in the region associated with the nerve affected most frequently affected nerves include the axillary, median, radial, ulnar, peroneal, or brachial plexus nerves. An episode can last from days to several months, but recovery is usually complete [3]. Repeated insults however, can cause permanent muscle weakness or loss of sensation, it can also be associated with pain in the limbs.

2. Case Report

This is a 41 Year old male, a right handed, mechanic, who presented to the neurology clinic with complains of numbness and weakness of his right arm since three months, he reports sustaining continuous pressure during his work on his arm around half an hour each day, he felt that when he is extending his fingers, he would feel some tightness of the forearm. When he was seen, he mentioned that his symptoms were improving

On examination of the right arm, he has mild weakness of the extension of the right wrist only, rest of the examination was within normal.

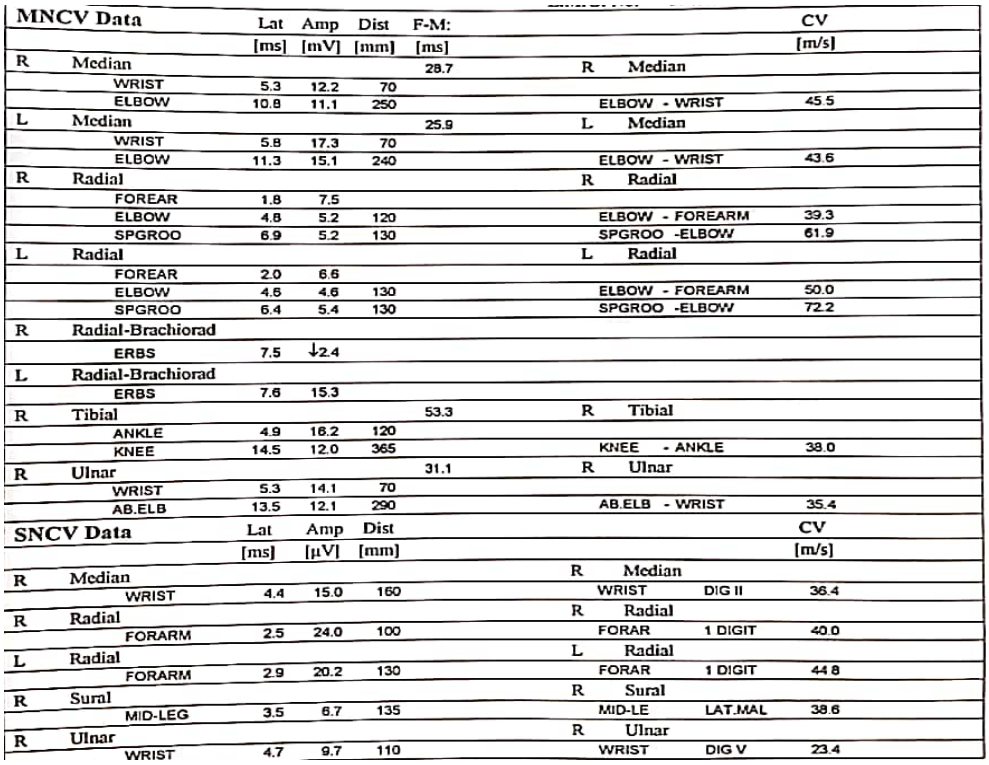
Clinical diagnosis was right radial palsy, nerve conduction study was done (Figure 1) supporting the diagnosis, a follow up nerve conduction was done (Figure 2) within a month that showed evidence of demyelinating neuropathy involving the median nerve, ulnar and peroneal nerve aswell. So a working diagnosis of multifocal neuropathy was raised.

He was admitted to the hospital for a course of IV immunoglobin at 0.4g/kg for five days, he could not tolerate the immunoglobin as he was having severe headache after three days, hence was aborted.

A vasculitic screening was negative including P-ANCA, C-ANCA, double –stranded DNA and brucella serology, CSF examination was negative for oligoclonal bands and his serum for anti-GM1 antibodies. His CBC and biochemistry where unremarkable.

Subsequently genetic testing was done for the possibility of hereditary neuropathy with liability to pressure palsy. His molecular analysis of PMP22 gene came heterogeneous deletion of the PMP22 gene supporting the diagnosis of the hereditary neuropathy with liability to pressure palsy.

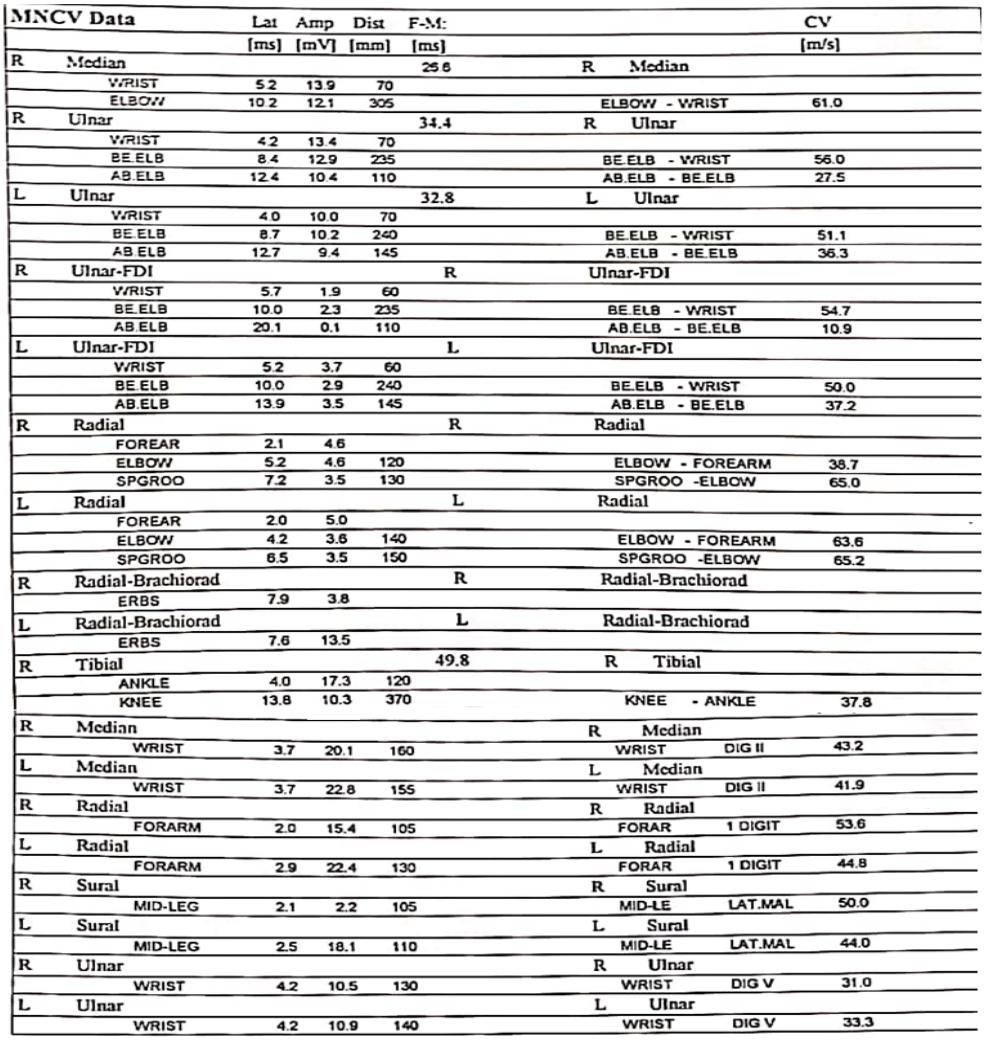
There was no family history of similar condition. No history of drug use or medications.



MNCV Data		Lat [ms]	Amp [mV]	Dist [mm]	F-M: [ms]		CV [m/s]
R	Median				28.7	R Median	
	WRIST	5.3	12.2	70			
	ELBOW	10.8	11.1	250		ELBOW - WRIST	45.5
L	Median				25.9	L Median	
	WRIST	5.8	17.3	70			
	ELBOW	11.3	15.1	240		ELBOW - WRIST	43.6
R	Radial					R Radial	
	FOREAR	1.8	7.5				
	ELBOW	4.8	5.2	120		ELBOW - FOREARM	39.3
	SPGROO	6.9	5.2	130		SPGROO -ELBOW	61.9
L	Radial					L Radial	
	FOREAR	2.0	6.6				
	ELBOW	4.6	4.6	130		ELBOW - FOREARM	50.0
	SPGROO	6.4	5.4	130		SPGROO -ELBOW	72.2
R	Radial-Brachiorad						
	ERBS	7.5	↓2.4				
L	Radial-Brachiorad						
	ERBS	7.6	15.3				
R	Tibial				53.3	R Tibial	
	ANKLE	4.9	16.2	120			
	KNEE	14.5	12.0	365		KNEE - ANKLE	38.0
R	Ulnar				31.1	R Ulnar	
	WRIST	5.3	14.1	70			
	AB.ELB	13.5	12.1	290		AB.ELB - WRIST	35.4

SNCV Data		Lat [ms]	Amp [μV]	Dist [mm]		CV [m/s]
R	Median				R Median	
	WRIST	4.4	15.0	160	WRIST DIG II	36.4
R	Radial				R Radial	
	FORARM	2.5	24.0	100	FORAR 1 DIGIT	40.0
L	Radial				L Radial	
	FORARM	2.9	20.2	130	FORAR 1 DIGIT	44.8
R	Sural				R Sural	
	MID-LEG	3.5	6.7	135	MID-LE LAT.MAL	38.6
R	Ulnar				R Ulnar	
	WRIST	4.7	9.7	110	WRIST DIG V	23.4

Figure 1. NCS showing evidence of conduction velocity delay in the right radial nerve (elbow – forearm) indicative of right radial nerve palsy



MNCV Data		Lat [ms]	Amp [mV]	Dist [mm]	F-M: [ms]		CV [m/s]
R	Median				25.6	R Median	
	WRIST	5.2	13.9	70			
	ELBOW	10.2	12.1	305		ELBOW - WRIST	61.0
R	Ulnar				34.4	R Ulnar	
	WRIST	4.2	13.4	70			
	BE.ELB	8.4	12.9	235		BE.ELB - WRIST	56.0
	AB.ELB	12.4	10.4	110		AB.ELB - BE.ELB	27.5
L	Ulnar				32.8	L Ulnar	
	WRIST	4.0	10.0	70			
	BE.ELB	8.7	10.2	240		BE.ELB - WRIST	51.1
	AB.ELB	12.7	9.4	145		AB.ELB - BE.ELB	36.3
R	Ulnar-FDI				R	Ulnar-FDI	
	WRIST	5.7	1.9	60			
	BE.ELB	10.0	2.3	235		BE.ELB - WRIST	54.7
	AB.ELB	20.1	0.1	110		AB.ELB - BE.ELB	10.9
L	Ulnar-FDI				L	Ulnar-FDI	
	WRIST	5.2	3.7	60			
	BE.ELB	10.0	2.9	240		BE.ELB - WRIST	50.0
	AB.ELB	13.9	3.5	145		AB.ELB - BE.ELB	37.2
R	Radial				R	Radial	
	FOREAR	2.1	4.6				
	ELBOW	5.2	4.6	120		ELBOW - FOREARM	38.7
	SPGROO	7.2	3.5	130		SPGROO -ELBOW	65.0
L	Radial				L	Radial	
	FOREAR	2.0	5.0				
	ELBOW	4.2	3.6	140		ELBOW - FOREARM	63.6
	SPGROO	6.5	3.5	150		SPGROO -ELBOW	65.2
R	Radial-Brachiorad				R	Radial-Brachiorad	
	ERBS	7.9	3.8				
L	Radial-Brachiorad				L	Radial-Brachiorad	
	ERBS	7.6	13.5				
R	Tibial				49.8	R Tibial	
	ANKLE	4.0	17.3	120			
	KNEE	13.8	10.3	370		KNEE - ANKLE	37.8
R	Median					R Median	
	WRIST	3.7	20.1	160		WRIST DIG II	43.2
L	Median					L Median	
	WRIST	3.7	22.8	155		WRIST DIG II	41.9
R	Radial					R Radial	
	FORARM	2.0	15.4	105		FORAR 1 DIGIT	53.6
L	Radial					L Radial	
	FORARM	2.9	22.4	130		FORAR 1 DIGIT	44.8
R	Sural					R Sural	
	MID-LEG	2.1	2.2	105		MID-LE LAT.MAL	50.0
L	Sural					L Sural	
	MID-LEG	2.5	18.1	110		MID-LE LAT.MAL	44.0
R	Ulnar					R Ulnar	
	WRIST	4.2	10.5	130		WRIST DIG V	31.0
L	Ulnar					L Ulnar	
	WRIST	4.2	10.9	140		WRIST DIG V	33.3

Figure 2. NCS showing evidence of demyelinating neuropathy involving the median nerve , ulnar and peroneal nerve

3. Discussion

HNPP is an autosomal dominant disorder. In 80 percent of HNPP cases, there is a 1.5 Mb deletion in chromosome 17p11.2 that results in reduced expression of the PMP22 gene this deletion corresponds to the duplicated region of PMP22. Approximately 20 percent of patients with HNPP have point mutations or small deletions in the PMP22 gene, and sporadic cases with de novo deletions have been described.

The PMP22 protein is a component of myelin, which is a protective coating that envelopes the nerves and promote the transmission of impulses in a timely manner. Studies suggest that the PMP22 protein is particularly important in protecting nerves from physical pressure, helping them recover their structure when exposed to compression, in patient with HNPP the myelin coating is more liable to damage [4].

Here we presented a case of a gentleman who works as a mechanic with prolonged periods of hand compression who developed HNPP, thus it must be taken into account that certain individuals might be at risk for this condition and at higher predisposition given their work occupation where there is an element of compression or prolonged pressure on certain anatomical parts, awareness should be raised in order to prevent disability for patients as a consequence of their occupation or certain hobbies they might be engaged at, such as minimizing the duration of exposure to such activities or adjusting the setting in away to reduce damage, furthermore screening family members for this condition is quite crucial.

This condition by far haven't been reported in the united arab emirates, and this might be the first case report to be addressed, few such examples have been reported in the existing peer-reviewed medical literature only.

4. Conclusions

Hereditary neuropathy with liability to pressure palsies can present as a multifocal neuropathy. Occupational history along with genetic testing plays a pivotal role to make the diagnosis.

List of Abbreviations

HNPP: Hereditary neuropathy with liability to pressure palsies

P-ANCA: Perinuclear anti-neutrophil cytoplasmic antibodies

C-ANCA: Cytoplasmic antineutrophil cytoplasmic antibodies

Anti-GM1: Antiganglioside antibodies

CBC: Complete Blood Count.

Availability of Data and Materials

All data are within the article.

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Authors Contribution

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Ethics Approval and Consent to Participate

Not applicable.

Consent for Publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing Interests

The authors declare that they have no competing interests.

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