E-poster 74 : Urodynamic findings in Charcot-Marie Tooth patients with Lower Urinary Tract Symptoms (LUTS)

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Hypothesis/Aim of study :

- The aim of the present study is to determine the clinical and urodynamic findings in Charcot- Marie-Tootth (CMT) patients referred to our Urology Department

Study design, materials and methods

- Retrospective study of those patients with CMT disease diagnosed at the Neurology Department of our Tertiary Hospital and referred to our Urology Department since 2008 due to lower urinary tract symptoms (LUTS).
- We reviewed their clinical charts regarding the age at CMT disease diagnosis, type of CMT disease and the presence of other comorbidities which could cause LUTS. We collected data on urinary symptoms, urodynamic and ultrasonographic findings, physical and neurological examination and therapeutic approach.

Results.

- Seven patients were referred to our department due to the presence of LUTS. They were 3 male and 4 female, with median age at the moment of LUTS onset of 55 (29-67) years and median time from the diagnosis of the neuropathy to the onset of LUTS was 14 (1-37) years.

Patients	Type CMT	Neurological Findings	LUTS	USD Findings	Terapeutic approach
1 (♂)	Low penetrancy	• Hypotrophy posterior muscles LE. • ↓ tibial&perineos reflex 3/5. • Deep sensitivity of hands & feet abolished.	Acute Urinary retention rUTI	Neurogenic acontractile detrusor	Indwelling bladder catheter
2 (♂)	Type 1 b	• Atrophy distal muscles LE. • Weel chair	Urgency Urinary Incontinence	Detrusor Overactivity	Botulinum Toxine + CIC
3 (♂)	Type 3	• Atrophy peroneal muscles. • ↓ strength LE	Acute Urinary Retention	Neurogenic acontractile detrusor	CIC
4 (♂)	Type 1	• ↓ superficial and vibratory sensitivity in feet+hands • No rotulian/aquileus reflex	Voiding symptoms SUI rUTI	Urodinamic SUI Detrusor underactivity	Scheduled micturition
5 (♂)	Unknown	• Hypotrophy distal muscles LE.	SUI	Normal USD	Pelvic Floor Rehabilitation
6 (♂)	Unknown	• Atrophy distal muscles LE. • Atrophy hand intrinsec muscles	Acute Urinary Retention	High PVR Low urinary flow curve	
7 (♂)	Type 1	• Atrophy distal muscles LE. • Needs walker	Hesitancy	Delay in initiating micturition due to delay relaxation of urethral sphincter + Detrusor underactivity	Alphablockers

LE:Lower extremities /UTIr: Recurrent Urinary tract Infections/CIC : Clean Intermittent Catheterization/SUI: Stress Urinary Incontinence/ PVR : Post voided residual

- Mean follow-up was 45.9 months (10.5-75.5). At this time, no changes at the initial treatment strategies were needed.
- In the ultrasound exam, there was no deterioration of the upper urinary tract in any patient.

Interpretation of results

Charcot-Marie-Tooth Syndrome (CMT) is the most prevalent peripheral sensory-motor inherited neuropathy (1). Despite having no effect on the autonomic system, Krhut et al demonstrated higher occurrence of LUTS in CMT patients using validated questionnaires (2). To our knowledge, urodynamic findings in these patients have been hardly reported (3).

So far, our study constitutes the longest series of CMT patients with LUTS studied with urodynamics.

LUTS are rare in CMT patients but voiding symptoms are the most prevalent.

Voiding symptoms in CMT patients correlate with impairment of detrusor contractility in urodynamics studies.

There is no deterioration of the upper urinary tract due to high bladder pressure in CMT patients

Concluding message

Most of CMT patients with LUTS complained of voiding symptoms presenting, mainly, impairment of detrusor contractility in functional studies. The onset of urological symptoms is delayed in time. We strongly recommend urodinamic evaluation in CMT patients reporting voiding symptoms

1.Braathen GJ et al. Genetic epidemiology of Charcot-Marie-Tooth in the general population. Eur J Neurol. 2011;18(1):39-48. 2.Krhut J et al. Lower urinary tract functions in a series of Charcot-Marie-Tooth neuropathy patients. Acta Neurol Scand. 2014;129(5):319-24. 3.Stojkovic T et al Autonomic and respiratory dysfunction in Charcot–Marie–Tooth disease due to Thr124Met mutation in the myelin protein zero gene. Clin Neurophysiol. 2003;114(9):1609-14.