RESEARCH PAPER

Tarsal Tunnel Syndrome in Children with Mucopolysaccharidosis: An Egyptian Center Study

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ABSTRACT

Background: Tarsal tunnel syndrome (TTS) is compression of the posterior tibial nerve as it passes inferiorly to the flexor-retinaculum in its fibro-osseous tunnel. We aimed to assess early detection of TTS in children with mucopolysaccharidosis (MPS) to help intervention at the proper time.

Methods: This cross-sectional study was performed on 40 pediatric patients with MPS patients (aged 1-18 years), who were receiving regular enzyme replacement therapy (ERT), were exposed to systemic clinical examination, appropriate laboratory and radiological evaluation, electrophysiological studies, and posterior tibial nerve motor conduction study.

Results: Among 40 children with MPS (median age 7 years; 62.5% male), MPS type I was most common (42.5%). All patients had joint stiffness and coarse facies; corneal clouding and hepatosplenomegaly were observed in 67.5% and ~45%, respectively. Growth impairment worsened with age, particularly in weight and height, while head circumference remained relatively preserved. Electrophysiological studies showed normal posterior tibial nerve conduction with no TTS observed in any of the patients.

Conclusions: TTS was a very rare entrapment of neuropathy in the general population and in MPS patients.

Keywords: *Tarsal tunnel Syndrome; Mucopolysaccharidosis; Posterior tibial nerve; Enzyme replacement therapy; Electrophysiological study*

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INTRODUCTION

A class of lysosomal storage diseases (LSDs) known as mucopolysaccharidosis (MPS) is caused by deficiencies in the enzymatic catabolism of glycosaminoglycan (GAG), a group of extracellular heteropolysaccharides that support various tissues structurally and also play more intricate roles in neurodevelopment, inflammation, and tumor progression [1, 2]. It can be categorized into seven key diseases, which are produced by deficits in 1 of 11 diverse enzymes, containing MPS I (Hurler–Scheie, Hurler, Scheie syndrome), MPS II (Hunter syndrome), MPS III (Sanfilippo syndrome), MPS IV (Morquio syndrome), MPS VI (Maroteaux-Lamy syndrome), MPS VII (Sly syndrome), and MPS IX (Natowicz syndrome, Hyaluronidase deficiency) [3]. Enzyme deficiency causes the progressive accumulation of GAGs, including dermatan sulfate (DS), chondroitin sulfate (CS), and heparan sulfate (HS), in lysosomes across numerous cell types, thereby affecting the functions of various organs and tissues [3]. Patients with MPS are classically characterized by coarse facies, hepatosplenomegaly, skeletal abnormalities (dysostosis multiplex), recurrent infections such as sinusitis or otitis media, respiratory issues, cardiac involvement, thick skin, decreased life expectancy, and intellectual disability [4]. Immediate diagnosis of MPS in the asymptomatic stage may be valuable in preserving organic function and improving outcomes. Definitive diagnosis depends on molecular tests, as the detection of the type of GAG, genetic testing, and specific enzyme assay [5]. Enzyme replacement therapy (ERT) and hematopoietic stem cell transplantation (HSCT) are two primary methods of managing MPS. Gene therapy (GT) is a promising approach to treating MPSs, as it could provide a way to permanently replace the deficient enzymes [6, 7]. Entrapment neuropathies are one of the most common musculoskeletal manifestations of MPS. Tarsal tunnel syndrome (TTS) is entrapment of the posterior tibial nerve as it arises below the flexor-retinaculum in its fibro-osseous tunnel, an anatomically narrow fibro-osseous canal located posterior to the medial malleolus [8]. Although it is considered relatively uncommon in the general pediatric population, its occurrence may be underrecognized in children with metabolic and skeletal disorders such as MPS due to

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overlapping musculoskeletal manifestations and communication limitations in younger patients. Progressive deposition of GAGs in connective tissues, tendonsheaths, and peri-neural structures may theoretically predispose affected individuals to nerve compression syndromes. Early identification of TTS is clinically important because delayed diagnosis can lead to chronic neuropathic pain, sensory deficits, gait abnormalities, and irreversible nerve damage. Therefore, electrophysiological screening may play a valuable role in detecting subclinical neuropathy before overt clinical manifestations appear [8].

The purpose of this study was early recognition of TTS in children with MPS to help intervention at the proper time.

METHODS

This cross-sectional study was performed on 40 pediatric patients identified with MPS who were attending regular follow-up visits and receiving ERT. Eligible patients were children aged 1–18 years with an approved diagnosis of MPS established by enzymatic assay and/or molecular genetic examining.

Patients were evaluated as a single study cohort and were subsequently categorized during analysis according to the presence and electrophysiological severity based on nerve conduction study findings and standardized grading criteria.

Exclusion criteria were Patients were excluded if they had MPS associated with diabetes mellitus, malignancy, or vertebral deformities causing spinal cord compression, as these conditions could interfere with neurological assessment and electrophysiological measurements.

Clinical, laboratory, and imaging assessment:

All enrolled patients endured a consistent assessment containing complete medical history, comprehensive general and systemic clinical examination, and anthropometric assessment (weight, height, and head circumference) interpreted using Egyptian /CDC, and WHO growth charts. Routine investigations included MRI of the brain and spine, echocardiography, skeletal survey, auditory brainstem response and audiometry, pelvi-abdominal ultrasonography, and laboratory investigations, including HbA1c to exclude diabetes mellitus. Diagnostic confirmation included urinary GAG analysis, enzyme assay, and molecular studies using dried blood spot testing.

Specialized electrophysiological assessment:

Electrophysiological evaluation was the principal method of this study and was performed using a Nihon Kohden Neuropack 2 electromyography system. Motor nerve conduction studies were conducted using surface electrodes to assess the posterior tibial nerve by evaluating compound muscle action potential amplitude, motor conduction velocity, and distal motor latency. All examinations were performed under standardized environmental and patient-related conditions, including limb temperature control, room temperature regulation, and proper patient positioning.

Statistical analysis

SPSS v27 (IBM Corporation, Armonk, NY, USA) was used for statistical analysis. The normality of the data distribution was assessed using the Shapiro-Wilks test and histograms. The unpaired student t-test was used to analyze quantitative parametric data, which were displayed as mean and standard deviation (SD). The Mann Whitney test was used to evaluate quantitative non-parametric data, which were displayed as the median and interquartile range (IQR). When appropriate, the Chi-square test or Fisher's exact test were used to examine the qualitative variables, which were displayed as frequency and percentage (%). Statistical significance was defined as a two-tailed P value of less than 0.05.

RESULTS

A total of 40 MPS patients were included, with a median age of seven years (IQR 3.75–11); males predominated (62.5 percent, M: F ratio 1.6:1). Consanguinity was common (77.5%), and 47.5% had a similar condition in the family. Type I (severe) was the most frequent subtype (42.5%), followed by Type IV (27.5%), Attenuated Type I (15%), and Types II and VI (7.5% each). Age at diagnosis and disease duration varied by subtype. Most patients (95%) received ERT, starting at a median age of 4 years (IQR 2–7); none underwent HSCT. No cases showed symptoms or signs of TTS.

The anthropometric measures (weight, height, and head circumference) across different age groups of patients with MPS. Regarding the weight Z score, there was a significant decline and worsening with age, from 0.043 ± 1.314 in the youngest group to -3.450 ± 1.700 in the oldest group ($P < 0.001$). Regarding the height Z score, there was a significant decline, from -2.087 ± 1.978 in the age group < 6 years to -6.100 ± 3.154 in the age group > 12 years ($P = 0.002$). The progressive decline in both weight and height Z-scores with age reflects progressive growth impairment in older MPS patients. Regarding H.C percentiles, there was a sharp decline with age, dropping from 93.800 ± 9.980 in the <6 years group to 50.000 ± 20.529 in the >12 years group ($P < 0.001$). Table 2

There were no significant differences in electrophysiological parameters (latency, amplitude, or velocity) between the right and left sides across posterior tibial motor nerve. Table 3

Table 1: Demographic characteristics and nerve entrapment manifestations among studied MPS patients

		N = 40
Age (years)		7 (3.75–11)
Sex	**Male**	25 (62.5%)
	Female	15 (37.5%)
	Male: Female ratio	1.6: 1

Family history	**Positive consanguinity**	31 (77.5%)
	Similar condition in family	19 (47.5%)
MPS Type	**Type I – Severe**	17 (42.5%)
	Type I – Attenuated	6 (15%)
	Type II	3 (7.5%)
	Type IV	11 (27.5%)
	Type VI	3 (7.5%)
Age at diagnosis (years)	**Type I – Severe**	1.83 ± 1.41
	Type I – Attenuated	6.42 ± 2.80
	Type II	2.33 ± 0.58
	Type IV	7.35 ± 3.88
	Type VI	5.00
Disease duration (years)	**Type I – Severe**	3.35 ± 1.93
	Type I – Attenuated	8.50 ± 3.15
	Type II	3.00 ± 3.46
	Type IV	8.64 ± 4.18
	Type VI	10.00 ± 2.65
Treatment type	**ERT**	38 (95%)
	HSCT	0 (0%)
	No treatment	2 (5%)
Age at starting ERT (years)		4 (2–7)
Nerve entrapment manifestations	Tarsal tunnel syndrome	0 (0%)

Data are stated as mean ± SD, median (IQR), or frequency (%). ERT: Enzyme Replacement Therapy, MPS: Mucopolysaccharidosis, HSCT: Hematopoietic Stem Cell Replacement

Table 2: Anthropometric measurements of MPS patients across different age groups:

	< 6 Years	**6–12 Years**	**> 12 Years**	**P**
Weight (kg)	15.07 ± 2.94	21.47 ± 5.91	29.75 ± 8.05	**<0.001***
Weight (Z score)	0.04 ± 1.31	-1.75 ± 1.40	-3.45 ± 1.70	**<0.001***
Height (cm)	89.07 ± 7.23	112.06 ± 14.51	118.88 ± 21.45	**<0.001***
Height (Z score)	-2.09 ± 1.98	-3.08 ± 2.44	-6.10 ± 3.15	**0.002***
Head circumference (cm)	52.93 ± 3.08	53.44 ± 1.94	53.56 ± 0.78	0.764
Head circumference percentile	93.80 ± 9.98	51.47 ± 21.85	50.00 ± 20.53	**<0.001***

Data are showed as mean ± SD. *Significant at $P < 0.05$.
MPS: Mucopolysaccharidoses

Table 3: Electrophysiological study of studied patients

	Right	**Left**	**Mean Difference**	**P-value**
Tibial motor				
Latency (ms)	2.804 ± 0.656	2.831 ± 0.688	-0.027 ± 0.632	0.784
Amplitude (mV)	9.133 ± 3.619	9.745 ± 4.049	-0.612 ± 2.881	0.187
Velocity (m/s)	49.630 ± 6.433	49.893 ± 6.367	-0.263 ± 3.860	0.670

Data are presented as mean ± SD. T: paired t-test; P-value: significance level. Nerve conduction parameters include latency (ms), amplitude (mV), and conduction velocity (m/s).

DISCUSSION

MPS is a group of progressive, inherited, multi-system lysosomal storage disorders with 11 enzyme deficits resulting in 7 MPS types [9].

Coarse facial features, skeletal abnormalities (dysostosis multiplex), hepatosplenomegaly, frequent infections like sinusitis or otitis media, thick skin, respiratory issues, cardiac involvement, mental retardation, and a shorter life expectancy were common clinical characteristics of patients with MPS [3].

The diagnosis of MPS has recently been evaluated in light of evaluating urinary GAGs, measuring enzyme activities, and identifying genetic variants [10]. ERT or hematopoietic stem cell transplant prolongs lifespan and improves the quality of life for certain MPS patients [9].

In our study, MPS type I was the most prevalent, accounting for the largest proportion of cases at 57.5%. This was followed by MPS type IV at 27.5%, while MPS types II and VI had the lowest proportions, each at 7.5%.

This aligned with the findings of Fateen et al. [11], Two Egyptian studies conducted over 18 years to provide an accurate overview of the division of every type of MPS amongst the Egyptian population. Both studies concluded that MPS I was the most common type, followed by MPS IV, MPS VI, and MPS II.

Also, our findings were in agreement with those of El Tellawy et al. [12], a study conducted in Upper Egypt. The results indicated that the most frequent type of MPS was Type I, which accounts for 55% of cases. This was followed by type IV at 26.66%, type II at 11.66%, and type VI at 3.3%.

In Tunisia, a study carried out above thirty five years from 1970-2005 found that MPS I was the most frequent MPS type detected, which aligns with our results, but the rest of the MPS types revealed a changed dispersal [13].

A study from the 1970s–2000s in Denmark and Norway informed that MPS I was the most common prevalent type. In America, MPS I came first, followed by MPS II, shadowed by MPS IV and MPS VI. This matches our results, as MPS I came first in the distribution [14, 15].

The results reported in other Arab nations were not consistent with the current findings. According to a survey conducted between 1983 and 2008, MPS VI was the most prevalent form in Saudi Arabia, followed by MPS I and IV. MPS IV and VI were the most prevalent forms of mucopolysaccharidoses in Emiratis [16, 17].

In Asia, the regularity of MPS types were MPS II, MPS I, then MPS IV and MPS VI regarding the studies performed in Japan and China. In Canada, the ranking was MPS IV, MPS VI, then MPS II [18-20].

In our study, the mean age of diagnosis varied widely. The severe subtype of MPS I had the earliest mean age of diagnosis, 1.8 ± 1.4 years, while the mean age of analysis of the attenuated subtype of MPS I was 6.4 ± 2.8 years. The highest mean age of finding was in type IV (7.3 ± 3.8 years). The mean age of diagnosis of type II was 2.3 ± 0.5 years and 5 years for type VI.

Similar results were informed by Fateen et al. [11], and Sestito et al. [21]. This can be explained as the severely affected patients may begin to show signs and symptoms within the first year of life. While the attenuated form typically develops during childhood or adolescence. These milder cases were often underdiagnosed and were usually only recognized when their signs and symptoms become more pronounced [22].

In the current study, all patients underwent enzyme assays for MPS using dried blood spots, which were confirmed by molecular testing in most cases. Molecular diagnostics was necessary to verify an enzyme deficiency and offers diagnostic certainty for conditions for which the use of current medications necessitates hospitalization and was very costly [23].

In this study, 95% of the enrolled patients obtained regular ERT. The availability of ERT at our genetics and inborn errors of metabolism unit since December 2019 encouraged affected families to seek a diagnosis, resulting in more individuals attending clinics. This increased patient attendance motivated many physicians to imply their patients for evaluation. Over time, this process causes greater awareness among affected families and enhanced expertise among physicians.

In all categories of MPS, growth deficiency containing inequality of growth and short stature was a common characteristic. Stunted growth becomes very noticeable with age during the early stages of the disease. This could be explained by the gathering of GAGs that impair the function of bone and cartilage cells, causing constant and advanced damage to cartilage and bone [24, 25].

Furthermore, ERT was less efficient at thoroughly cartilage and bone, which in part describes the reduced clinical improvement in the skeletal symptoms detected in MPS patients, even after long-term ERT.

In our study, there was a progressive decline in both weight and height Z-scores with age, with less affected head circumference suggesting a disproportionate growth pattern and some patients experienced hydrocephalus as a complication of MPS I and II. It highlights the value of early diagnosis and management to support optimal growth and minimize long-term growth deficits.

Tylki-Szymanska et al. [26], stated that the anthropometric traits of patients with MPS I significantly vary from healthy population, and children with MPS I grew greatly slower, and alterations among involved and healthy children were enhanced with age. Also, Różdżyńska-Świątkowska et al. [25], had similar findings in different MPS types.

In this study, all enrolled patients (100%) exhibited no symptoms or signs of TTS with negative NCV of the posterior tibial nerves, indicating the rarity of TTS in this population. Currently, there were no announced scientific articles detailing TTS in patients with MPS.

Only Williams et al. [8], studied nineteen MPS patients, 4 patients with a supposed clinical diagnosis of TTS were identified, 3 of them with inconclusive NCV results. Overall, published evidence suggests that tarsal tunnel involvement was uncommon compared to other entrapment neuropathies in MPS, such as carpal tunnel syndrome.

Limitations: Small sample size constrains the ability to generalize the data with recommended further multicenter studies, lack of screening protocols for screening and follow-up of nerve entrapment syndromes in MPS, and early surgical intervention was challenging due to associated MPS serious co-morbidities and anesthesia-related issues.

CONCLUSIONS

TSS was a very rare entrapment neuropathy in general population and also in MPS patients.

LIST OF ABBREVIATIONS

TTS: Tarsal Tunnel Syndrome

MPS: Mucopolysaccharidosis

GAG: Glycosaminoglycan

LSD: Lysosomal Storage Disease

ERT: Enzyme Replacement Therapy

HSCT: Hematopoietic Stem Cell Transplantation

GT: Gene Therapy

DS: Dermatan Sulfate

CS: Chondroitin Sulfate

HS: Heparan Sulfate

MRI: Magnetic Resonance Imaging

HbA1c: Glycated Hemoglobin

SD: Standard Deviation

IQR: Interquartile Range

NCV: Nerve Conduction Velocity

CMAP: Compound Muscle Action Potential

SPSS: Statistical Package for the Social Sciences

DECLARATIONS

Human Ethics and Consent to Participate declarations:
The study was carried out at Tanta University Hospitals over a period extending from June 2022 to August 2024. Ethical approval was attained from the Ethical Committee of the Faculty of Medicine, Tanta University (approval number: 35390/3/22). Written informed consent was acquired from parents or legal guardians before acceptance.

Consent for publication:
Not applicable

Availability of data and material:
Data is available upon reasonable request from corresponding author.

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AUTHORS' CONTRIBUTIONS
All authors contributed to the study conception and design. Material preparation, data collection and analysis were performed by [M.R.E.], [H.H.A.], [H.M.E.] and [H.E.D.]. The first draft of the manuscript was written by [A.L.Y] and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript

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