Clinical Manifestations and Disease Burden of Primary Mitochondrial Myopathies (PMM): Results from a Patient Journey Analysis Shows Substantial Healthcare Resource Utilization

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SCAN ME

BACKGROUND

- Primary mitochondrial myopathies (PMM) are a group of disabling and underdiagnosed rare genetic disorders characterized by a range of clinical presentations and multisystemic impact[1]
- Diagnosis and management of PMM can be challenging due to the heterogeneity of clinical manifestations, including age of onset[1-3]
- Patients may engage a variety of healthcare professionals to manage their accumulating symptoms and can receive their first clinical diagnosis from a wide spectrum of specialists[3-5]
- Diagnosis with genetic testing is recommended by the Mitochondrial Medicine Society,[1] but utilization remains low
- With no approved treatments for PMM, current treatment involves symptom management[2,6]
- This approach is not optimal for treating the underlying cause or enabling patients to improve their physical and social functioning[7]
- To better understand the path to PMM diagnosis and management, a patient journey analysis was conducted using Komodo closed-claims data—one of the richest longitudinal data sets available for patient-level analyses

METHODS

- The Komodo closed-claims database was analyzed for US patients with suspected PMM between 2016-2021
- Due to the absence of a PMM-specific ICD-10 diagnosis code, patients with suspected PMM were identified via a stepwise approach as follows: mitochondrial disorder identified → myopathy presentation confirmed → secondary mitochondrial disorders excluded
- Claims data for patients aged ≥16 years with suspected PMM and continuous enrollment in 2018-2019 were segmented based on procedure codes indicating myopathy and inpatient or emergency room admission
- In patients with suspected PMM, claims were categorized by organ system at 3 years (24-36 months) and 1 year (0-12 months) before their first mitochondrial diagnosis

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ACKNOWLEDGEMENTS

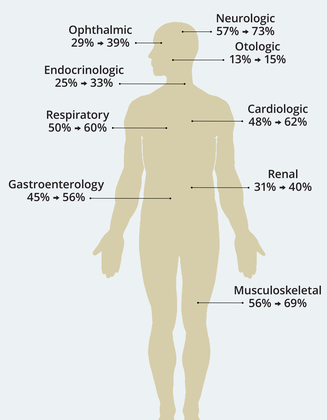
The authors thank Akshay Mehta and Ayesha Bhatia from Trinity Life Sciences for their contributions to these analyses.

RESULTS & KEY POINTS

Multisystem manifestations accumulate in the lead-up to diagnosis

Analyses by organ system indicate that manifestations in these regions increased from the 3-year period (24-36 months) to the 1-year period (0-12 months) before patients' first mitochondrial diagnosis

Accumulation of manifestations by organ system involvement **from 3 years → 1 year** before diagnosis (% patients)*



97% of patients had multisystem manifestation, with symptoms affecting an average of **~6 organ systems**

*Based on n=3.7k patients with suspected PMM and continuous enrollment in medical coverage for 3 years prior to first mitochondrial diagnosis

The variety, volume and frequency of myopathy-related claims observed in the data reflect the high comorbidity burden and substantial healthcare resource utilization in patients with suspected PMM

Increasing burdens of myopathy

% Patients	No claims for any myopathy 34%	≥1 claim for muscle spasm, fatigue, or muscle weakness 30%	1-3 claims for ptosis, myalgia, abnormalities of gait, or myopathy 17%	>3 claims for ptosis, myalgia, abnormalities of gait, or myopathy 11%	≥1 claim for paralytic gait, critical myopathy, ataxia, rhabdomyolysis, sarcopenia, and/or ER or inpatient visit 8%
Comorbid Signs/Symptoms and Conditions[a]: % of Patients with Suspected PMM in Segment					
Nervous system	13%	23%	40%	50%	69%
Respiratory	38%	52%	55%	58%	64%
Abdominal/GI	35%	49%	53%	55%	58%
Circulatory	23%	36%	37%	41%	50%
Hypertension	39%	44%	46%	49%	46%
Depression	24%	35%	36%	40%	44%
Esophageal	25%	34%	36%	44%	41%
Kidney disease	21%	21%	22%	24%	26%
Diabetes	21%	24%	25%	23%	26%
Pain	62%	79%	83%	86%	78%[b]
Healthcare Visits: % of Patients with Suspected PMM in Segment (Average # Visits over 2 Years)					
Setting of Patient Encounters					
Physician office	16% (2.9)	19% (3.7)	30% (3.3)	37% (3.9)	46% (7.1)
Hospital outpatient	6% (2.9)	6% (2.4)	13% (3.2)	21% (3.7)	34% (5.5)
Hospital inpatient	0	0	0	0	33% (2.0)
Emergency room	0	0	0	0	13% (1.7)
Specialist Visits[c]					
Any specialist	64% (8)	73% (10)	84% (11)	90% (15)	91% (19)
Neurologist	22% (3.9)	28% (4.4)	44% (4.8)	58% (6.1)	66% (7.4)
Cardiologist	31% (5.3)	40% (5.6)	44% (5.3)	50% (5.4)	61% (7.1)
Gastroenterologist	21% (3.3)	29% (3.9)	31% (3.8)	36% (4.8)	36% (6.5)
Ophthalmologist	24% (3.8)	26% (4.0)	37% (4.2)	44% (5.3)	35% (4.0)
Pulmonologist	10% (3.7)	14% (4.1)	15% (4.3)	20% (4.9)	26% (7.1)

[a]Groupings derived based on Chronic Comorbidity Warehouse (CCW) and Clinical Classifications Software Refined (CCSR) recommendations.
[b]Data for patients with listed myopathies only. Of patients with ER or inpatient visit, 88% reported pain.
[c]Patients may have visited one or more specialists during the 2-year study period.
ER, emergency room; GI, gastrointestinal.

The accumulation of multisystem manifestations during a patient's long journey to a PMM diagnosis leads to substantial healthcare resource utilization

NP-MAV-00027-0923

Presented at the Academy of Managed Care Pharmacy Nexus Annual Meeting; October 16-19, 2023.