From Clinical Manifestations of Primary Mitochondrial Myopathies (PMM) to Diagnosis: Results from a Patient Journey Analysis Shows Limited Utilization of Genetic Testing

Mai Sirimanne,[1] Joseph Kates,[1] Sri Saikumar,[2] Matthew Warner,[3] Saloni Shah,[2] Adrienne Lovink,[2] Yuqing Xue[2]

[1]Reneo Pharmaceuticals, Inc., Irvine, California, USA; [2]Trinity Life Sciences, LLC, Waltham, Massachusetts, USA; [3]Commercial Rx, Inc., Corona del Mar, California, USA

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 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 in payer-complete closed claims via a stepwise approach as follows: mitochondrial disorder identified ➔ myopathy presentation confirmed ➔ secondary mitochondrial disorders excluded
- In patients with suspected PMM, healthcare resource utilization (HCRU) claims were categorized by physician visits before and after the first mitochondrial diagnosis
- Patients included in the analysis were continuously enrolled in medical and pharmacy coverage through the duration of study, ensuring complete longitudinal visibility across all healthcare encounters

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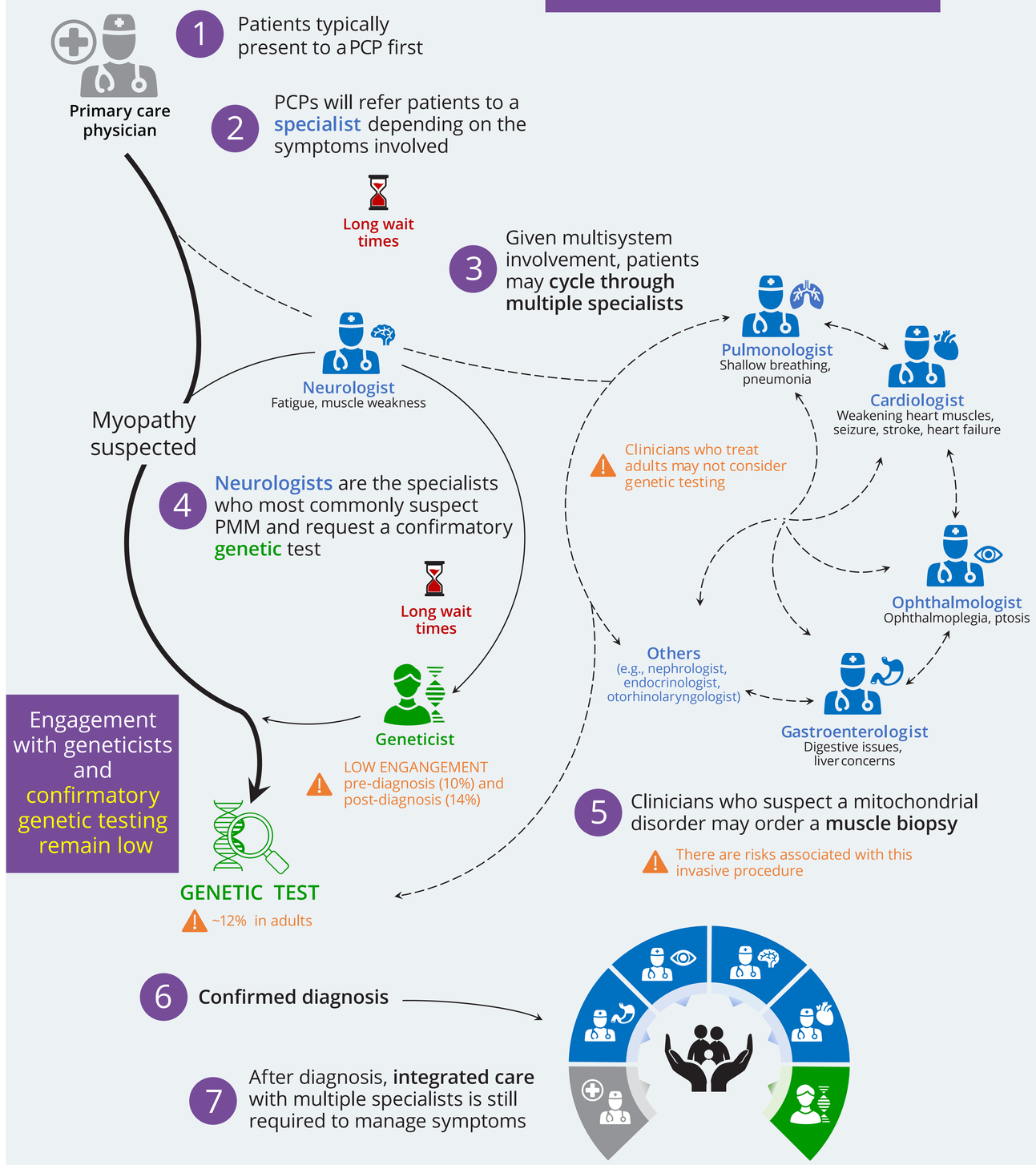
The diagnostic journey for patients with PMM is often long and convoluted; low engagement with geneticists and genetic testing represents a significant gap that creates a suboptimal and burdensome diagnostic approach

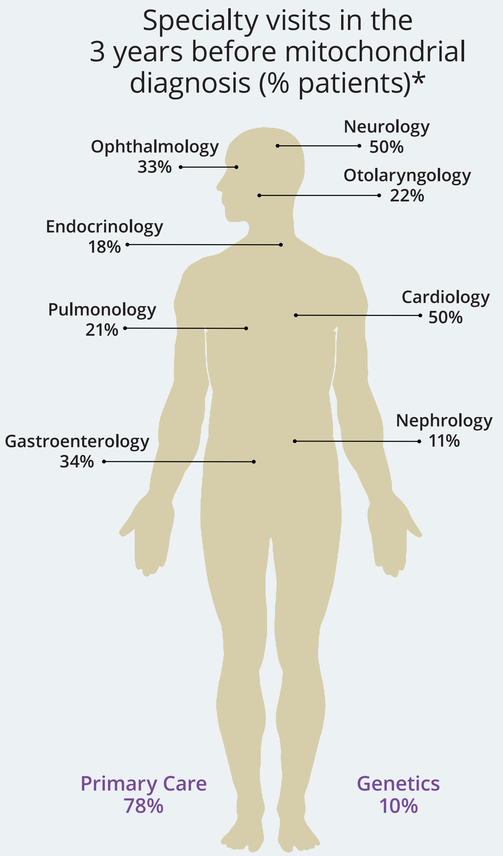
Ideally, the path to PMM diagnosis is streamlined with **early genetic testing** (bold arrows)

In reality, patients experience **long wait times** and **multiple referrals** before receiving a confirmatory genetic test for PMM (dotted arrows)

With **prolonged delays in getting a confirmatory diagnosis** with genetic testing, multisystem **manifestations accumulate** and **healthcare visits increase**



The percentage of patients with ≥3 specialist visits increased from 28% in the 3-year period before mitochondrial diagnosis to 44% in the 1-year period before diagnosis



Primary care visits are high; engagement with geneticists is low

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

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