INTRODUCTION

The likelihood of developing significant primary mitochondrial disease (PMD) in the overall population is estimated to be 1 in 5,000,000. Worldwide statistics may vary because some patients with primary mitochondrial myopathies (PMMs) develop mild, undetectable symptoms. The overall awareness of PMD has increased over the past 25 years, although clinician knowledge remains suboptimal.

Evidence-based clinical protocols are the preferred method for developing diagnostic and medical management recommendations; however, they are often not currently available or used in PMD. Patients with PMM are not diagnosed in a timely manner and have difficulty reaching the appropriate specialist to manage their mitochondrial symptoms.

Patients with PMM often see multiple clinicians and initially consult a primary care physician; however, 55% of these patients receive their diagnosis from a neurologist.

OBJECTIVE

To understand how the lives of PMM patients with neuromuscular disease manifestations can be positively impacted by improvements in their management and journey through the healthcare system by increasing understanding of the neurologist’s practice type/focus, knowledge base, and experience with PMMs.

METHODS

Respondents were recruited and screened via criteria designed to determine if the study was relevant to their clinical practice.

Qualitative Research included:

- Up-front questionnaire completed prior to interviews (Figure 1)
- One-on-one highly qualitative interviews were 1 hour in duration
- Web-assisted telephone interviews were conducted

RESULTS

Neurologists’ Practice Type and Focus

- Qualitative Research included interviews of neurologists (N=18) representing various areas of training and specialization, practice settings, and number of patients seen per month (Table 1)

Table 1. Neurologist Practice Characteristics (N=19)

<table>
<thead>
<tr>
<th>Type of in-person interview</th>
<th>Sample of Neurologists</th>
<th>Clinical Practice Setting</th>
<th>Number of Patients Seen Per Month</th>
</tr>
</thead>
<tbody>
<tr>
<td>One-on-one in-person interview</td>
<td>10-8 neurologists (various areas of training and specialization)</td>
<td>4 in university hospital setting</td>
<td>~230 (range 150 to 400)</td>
</tr>
<tr>
<td>One-on-one web-assisted telephone interview</td>
<td>11-1 neurologists (8 in neuromuscular specialists and 3 in neurology specialists)</td>
<td>4 in neuromuscular in hospital setting</td>
<td>~250 (range 50 to 350)</td>
</tr>
</tbody>
</table>

- Neurologists reported seeing a greater number of neuromuscular patients per month, whether in a hospital or private practice setting

- In the in-person interviews, neurologists reported an average of 30% of the patients in their practice had a diagnosis of neuromuscular disorder (range 15% to 80%)

- In the telephone interviews, neurologists in both hospital and private practice reported seeing ~100 neuromuscular disorder patients per month (range 75 to 140)

- Neurologists identified the specific neuromuscular and neurodegenerative conditions they manage in clinical practice (Table 2)

- Amyotrophic lateral sclerosis, myasthenia gravis, muscular dystrophy, neuropathies, and myopathies were reported as being the most common

Neurodegenerative and Neuromuscular Conditions Treated by Neurologists in Clinical Practice

- Alzheimer’s disease
- Cognitive decline (for example, broader/dementia, vascular, cognitive, epileptic)
- Parkinson’s disease
- Huntington’s disease
- Amyotrophic lateral sclerosis
- Charcot Marie Tooth disease
- Multiple sclerosis
- Muscular dystrophy
- Myasthenia gravis
- Myopathy
- Myositis, including polymyositis and dermatomyositis
- Peripheral neuropathy

- Neurologists reported the most commonly seen conditions in clinical practice, which included muscle weakness, chronic fatigue, balance problems, and exercise intolerance (Figure 3)

- Estimates of the commonness of symptoms were similar for practice type and for general and neuromuscular neurologists

Diagnosing Neuromuscular Disease

- Three of the 8 (37.5%) neurologists in the in-person interviews reported treating patients diagnosed with PMD

- These patients with PMD are slow, rigid, have loss of balance, and other concomitant conditions, such as insomnia, constipation, gastrointestinal issues, ataxia, cardiac disease, and hearing difficulty

- One patient was diagnosed through muscle biopsy

- Two patients were diagnosed with both genetic testing and a muscle biopsy

Neurologists’ Review of the Primer

- The primer was reviewed by 18 neurologists
- Neurologists self-rated at 4 or above regarding their awareness and knowledge of PMD

- Tests (3) general neurologists in telephone interviews rated knowledge as an average of 4

- Six (6) neurologists, larger study with neurologists, and the in-one-on-one interviews rated knowledge as 5 or higher

- Eight (8) neurologists in web-based interviews rated knowledge as an average of 6

- Neurologists reported that overall knowledge of PMD was not very detailed or specific

- In the telephone interviews, while all indicated they had some awareness of PMD, most stated that they only knew general information

- Typically, the source of information was medical training or exposure via the patients that were ultimately diagnosed with PMD in clinical practice

- After reviewing the PMM primer information, the neurologists identified key points and potential gaps in their knowledge (Figure 4)

CONCLUSIONS

- This small qualitative research study was highly iterative and serves as a preliminary step to a more focused study, and while the findings are qualitative in nature and should be considered directional rather than conclusive

- Neurologists see an average of ~230 to 250 patients per month with numerous neuromuscle and neuromuscular conditions

- The symptoms most commonly seen in clinical practice by neurologists include the 5 most common symptoms reported by patients with PMM, which are muscle weakness, fatigue, exercise intolerance, gastrointestinal problems, and balance problems

- Muscle biopsy and genetic testing is rarely ordered

- Neurologists frequently used one specific type of PMD rather than commonly of the presentation

- General neurologists and subspecialists indicated that neuromuscular subspecialists would be the primary specialty to manage adult patients with PMM

REFERENCES


Acknowledgements:

This study was sponsored by Stealth BioTherapeutics. Medical writing assistance was provided by James A. Shiffer, RPh, Write On Time Medical Communications, LLC.