HCP EDUCATION

Impact of Continuing Medical Education on Health Care Providers' Ability to Improve The Recognition and Genetic Diagnosis of Primary Mitochondrial Diseases

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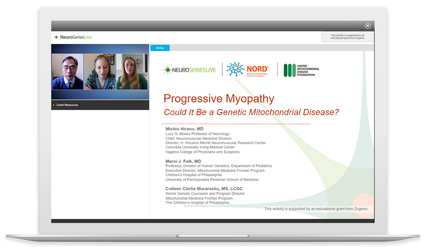
INTRODUCTION

While the burden caused by primary mitochondrial diseases (PMDs) on patients and their caregivers is extensive, these conditions are often missed because of their diverse clinical manifestations and genetic etiologies.

We assessed the impact of online continuing medical education (CME) for health care professionals' (HCPs') competence related to the recognition and diagnosis of PMD, as well as the place of genetic testing in the diagnostic process.



METHODOLOGY

Educational Program and Evaluation Details

Partners

Advocacy groups: UMDF, NORD

Education: PlatformQ Heath, Postgraduate Institute for Medicine

Data collected

Changes in knowledge, competence, reported behavior, engagement, and identification of continuing gaps.

Interventions

One 60-minute online CME activity was launched live on 3/19/21 and remained on-demand for 1 year, covering themes relative to awareness of PMD burden, the most common disease manifestations, and the appropriate place and interpretation of genetic testing.

Measurements

Questions asked before and immediately after the activity. Two-month follow-up survey was sent to learners to evaluate change in practice. Chi Square tests used for statistical analysis.

Title:

Progressive Myopathy: Could it be a Genetic Mitochondrial Disease?

Learning Objectives:

- Demonstrate an enhanced index of suspicion for the diagnosis of a genetic mitochondrial disease in infants, children, and adults who present with progressive muscle weakness
- Describe the diagnostic work-up for a patient with mitochondrial disease and how to utilize genetic testing as part of the diagnostic approach
- Describe key manifestations of thymidine kinase 2 (TK2) deficiency across the spectrum of disease severity and age as an example of mitochondrial DNA replication deficiency syndrome (MDS)

Faculty:

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Activity featured downloadable slides, panel discussions, live polling, and pre-program and live Q&A.

RESULTS

Learner Demographics

622 total learners

118 identified themselves as treaters

61% target audience learners*

6.56 visits of progressive myopathy patients impacted monthly

Learner Engagement

292 certificates awarded

750 total polling responses

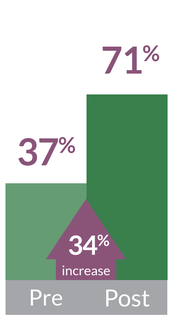
187 slide downloads

23 learner questions

Target Audience: Pediatric Neurology, Neurology, Pediatrics, Neuromuscular Specialists, Primary Care, Pulmonology, Gastroenterology, Physical Therapy, Occupational Therapy, Nutrition, and Genetics

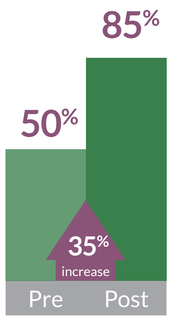
Changes in Knowledge/Competence

In addition to muscle weakness, which of the following symptoms may lead you to suspect the presence of a mitochondrial disease for a young child, based on their documented frequency in the published literature? (*Answer:* Difficulty swallowing, exercise intolerance, and developmental delay)



n = 622 pre, n = 303 post
P<0.05

You suspect a mitochondrial disorder in your patient. Which of the following laboratory and genetic-testing results do you expect will be abnormal? (*Answer:* Presence of ragged red fibers and COX-deficient fibers on muscle biopsy)



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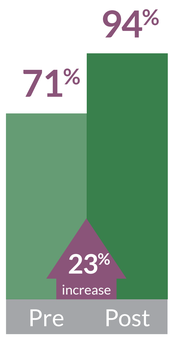
Based on clinical evaluation and biochemical labs, you suspect a mitochondrial disease and order whole exome sequencing with mtDNA genome sequencing from a blood sample. The results come back as inconclusive. What would be an appropriate next step? (*Answer:* Order a muscle biopsy to evaluate mtDNA sequencing and deletion/duplication studies via next-generation sequencing)



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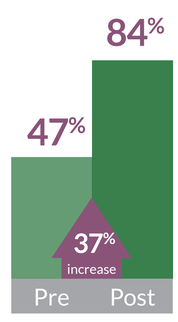
Gary, a married adult and biological father to two 6-year-old twins, presents with muscle weakness, ptosis, and unexplained dysphagia. Lab tests reveal elevated creatine kinase, lactate, and transaminases. You suspect the mitochondria disorder, "thymidine kinase 2 deficiency" (TK2d). What would you tell/ask your patient to confirm your diagnosis? (*Answer:* Whole exome sequencing of blood cells will give us some information regarding your underlying genetic cause)



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P<0.05

Positive Impact on Patient Outcomes and Clinical Practice

Among Those Who Responded to the 2 Month Follow-Up Survey (n=61)

89% reported the activity positively impacted patient experiences/outcomes

n=35

86% reported the activity positively impacted clinical practice

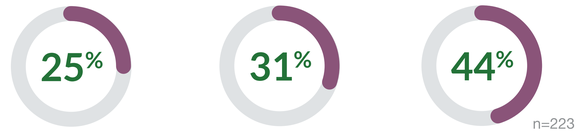
n=37

58 write-in examples were shared

- Two patients were referred for further work-ups and gave me feedback that they felt like they were close to getting a diagnosis.
- Increasing diagnostic testing in mitochondrial suspected patients.
- I am more confident in my therapeutic differentiation of mitochondrial DNA clinical syndromes (e.g., point mutations, deletion syndromes).
- I will address diagnostic suspicions based on clinical evolution and laboratory studies suggesting myopathies and subsequently confirm with genetic studies.

Attitudes of Clinicians Before the Activity (Collected with In-Activity Polling Questions):

When you see a patient with progressive myopathy, do you consider a mitochondrial disorder...?



n=223

What is a mitochondrial disorder? | All the time | When neurological or muscular motor symptoms are the only manifestations

Can you diagnose mitochondrial myopathy on physical exam alone?

n=179

Yes | No (*Correct*)

All patients must have a muscle biopsy to diagnosis mitochondrial myopathy?

n=171

Yes (*Correct*) | No

All mitochondrial disease is inherited from the mother?

n=177

Yes | No (*Correct*)

CONCLUSION

Post-test and HCP surveys completed 2 months after a one-time CME education activity suggested a positive impact resulted from both live and on-demand online CME education on HCPs' awareness of PMD burden, the most common disease manifestations, and the appropriate place and interpretation of genetic testing. Low baseline knowledge in multiple areas point to a continuing need for CME activities for HCPs related to improving their ability to recognize and diagnose PMD.