

# Confirmed Versus Suspected: The Social Significance of a Genetic or Non-Genetic Diagnosis of Mitochondrial Disease

Elizabeth Krieg, Laurel Calderwood, MaryAnn W. Campion, Katherine E. Krepkovich  
Boston University School of Medicine, Boston, Massachusetts

**Objective:** To investigate how the attitudes and beliefs regarding the importance of a genetic versus non-genetic diagnosis impact an individual's support needs and interactions within the mitochondrial disease community. This study explored the impact of diagnosis type on various aspects of the patient experience by identifying and comparing patient sub-groups within the mitochondrial disease population.

**Methods:** The study population (n=201) was surveyed via an online survey. Respondents were categorized into two groups, those with a genetic diagnosis of mitochondrial disease and those with a non-genetic diagnosis. Survey responses were compared between the two groups using methods of quantitative and qualitative analysis.

**Results:** The two patient groups have different support needs but are both fairly significantly affected by mitochondrial disease and experience similar problems relating to medical providers, costs of care, lack of appropriate support resources, and concerns regarding available treatment. The non-genetic group faces additional challenges related to not having a confirmed genetic diagnosis.

**Conclusion:** There are significant problems with the perceived quality of care for both groups but overall, individuals with a non-genetic diagnosis experience more problems and unmet needs in areas related to both medical and social support due to their lack of a definitive diagnosis. Support resources for the non-genetic diagnosis group must be designed to address the unique challenges associated with not having a confirmed genetic diagnosis.

**Practice Implications:** It is important to understand the efficacy of existing patient support resources for different sub-groups within the mitochondrial disease patient population so that appropriate services and resources can be developed or improved to address specific needs of that sub-group and provide effective patient care and support.