The Power of Patients — Harnessing Quality Registries to Understand Your Rare Disease.

Maximizing patient registry data to characterize a rare disease: A decade-long success story from Barth Syndrome Foundation

Barth syndrome is a rare, life-threatening, genetic mitochondrial disorder. Barth syndrome primarily affects boys due to an X-linked mutation in the *tafazzin* gene (*TAZ*, also called G4.5). As a multi-system disease, affected individuals may suffer from heart failure, muscle weakness, and infection (caused by neutropenia). Additional characteristics commonly include growth delay, impaired lipid metabolism, fatigue, and cardiolipin deficiency. In some cases, the symptoms can be very severe, sometimes resulting in heart transplant, infections requiring hospitalization, and even death.

Established in 2000, Barth Syndrome Foundation (BSF) is the only global network of families, healthcare providers, and researchers driven by the mission to save lives through education, advances in treatment, and finding a cure for Barth syndrome. Early on, BSF recognized a need to better characterize the disease. In 2006, BSF created its registry to gain critical insights into the disease by collecting physician-entered as well as patient-reported information from individuals living with Barth syndrome around the world.

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As a result of this strategic initiative, data compiled through BSF's registry has been the driving factor behind the collective understanding of the disease. In 2012, Dr. Amy Roberts, Boston Children's Hospital, and colleagues from the Royal Hospital for Children in the UK and the University of Florida used data from the BSF registry to define more completely the phenotype, genetic basis, and pathophysiology of Barth syndrome. From this registry analysis, an important finding emerged whereby researchers noted that cardiomyopathy and neutropenia were present in approximately 70% of individuals with Barth syndrome.

Building upon the findings from the 2012 registry analysis, Dr. Colin Steward from the Royal Hospital for Children in the UK and colleagues from around the world used BSF's registry data to characterize Barth syndrome associated neutropenia in greater depth, and to assess impact of common therapies employed by physicians treating individuals with Barth syndrome. Published in January 2019, the research registry findings not only continue to define the clinical nuances of Barth syndrome, but also suggest that Barth syndrome should be considered in the differential diagnosis of any boy with unexplained chronic or variable neutropenia.

In addition to informing the natural history of disease, BSF has used findings from the registry to inform organizational research priorities. Following the observation that 2/3 of individuals with Barth syndrome reported some degree of heart failure or cardiac insufficiency, BSF recently engaged with Dr. Brian Feingold from Children's Hospital of Pittsburg to pair information from the Pediatric Heart Transplant Society (PTHS) with patient-experience data from BSF's registry to better characterize transplantation in Barth syndrome.

BSF's registry is a testament to the power of patient registries. Insights into the characteristics of the disease extend far beyond that which could have been gained by a single clinician or even collection of case studies. Further, the insights gathered from cumulative years of physician-entered and patient-reported registry data have shaped the research focus and funding intention of BSF in ways that will be more impactful to affected individuals as well as current and future investigators.

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