The Barth Syndrome Foundation Delivers Petition to FDA Advocating for a Fair, Equitable and Appropriate Review of the Only Potential Treatment for Barth Syndrome

In 2 Months, Petition Garnered Nearly 20,000 Signatures from Individuals in All 50 U.S. States

Boston (PR WEB) – December 21, 2023 – The Barth Syndrome Foundation (BSF), the only patient advocacy organization dedicated to Barth syndrome and saving lives around the world through education, advances in treatment and finding a cure, today announced that it is petitioning the U.S. Food and Drug Administration (FDA) to review the New Drug Application (NDA) for elamipretide, the only potential treatment for Barth syndrome, in a fair, equitable and appropriate manner. Despite compelling evidence that elamipretide has been well-tolerated and has demonstrated clinical benefit in Barth syndrome, and despite ongoing efforts by the Barth Syndrome Foundation and other advocates to engage with regulators, the FDA has refused to review an NDA to date. The petition ran from mid-September to mid-November, and in just two months, garnered nearly 20,000 signatures from individuals across all 50 U.S. states, plus Washington, DC, Puerto Rico, the U.S. Virgin Islands and deployed military personnel.

Barth syndrome is an ultra-rare, life-threatening, genetic disease primarily affecting males. Eighty-five percent of deaths due to Barth syndrome occur by age 5. There are currently no FDA-approved treatments for Barth syndrome, and there are no other potential therapies in clinical development.

"For many Barth syndrome families, elamipretide represents their only hope for an affected child to have an increased chance to live into adulthood and to enjoy a life with reduced debilitating weakness and fatigue that precludes them from going to school, playing with friends, or holding a job," **said Kate McCurdy.** As co-founder and Chair of the Board of BSF, she hand-delivered the petition to Dr. Norman Stockbridge, Director of the Division of Cardiology and Nephrology, and Dr. Hylton Joffe, Director of the Office of Cardiology, Hematology, Endocrinology and Hematology at FDA, which refused to review the elamipretide NDA in 2021.

McCurdy continued: "We lost our son, Will, to this devastating disease in 2014, the same year BSF approached Stealth to ask that they initiate development efforts. During the 10 years, 2 clinical trials, and 2 dozen FDA meetings since then, over 10% of our global patient population have died as the result of this disease. Meanwhile, elamipretide has been shown to be well-tolerated and has demonstrated clear, statistically significant improvements relative to the natural history of the disease. Patients who have no approved treatment options deserve access to this medicine. It is unconscionable that the FDA refuses to even fully review the data. We

implore them to be fair and equitable and to utilize the appropriate flexibility that has been granted to them by the US Congress for cases just like ours."

The Barth Syndrome Foundation and leading scientific thought leaders lobbied for the development of elamipretide for Barth syndrome beginning in 2014. After completing promising nonclinical studies, and publication of extensive natural history studies characterizing the disease in more than 50% of the U.S. Barth syndrome patient population, the sponsor, Stealth BioTherapeutics, agreed to start development efforts in 2016. Since Barth Syndrome Foundation and the sponsor met with FDA to discuss the clinical data in 2019, FDA has moved the program through four different FDA review divisions, rejected multiple proposed new trial protocols, and most recently this summer said there is no feasible path forward for development because there are too few patients to enroll in clinical trials.

"People with Barth syndrome have experienced greater than 40% improvement in heart function, greater than 25% improvement in exercise tolerance, and greater than 40% improvement in muscle function, none of which is expected in the natural course of Barth syndrome," **said Emily Milligan, Executive Director of BSF. "**This has helped make transformational and meaningful changes in their lives. There simply aren't enough people with Barth syndrome, though, to meet the impossible statistical requirements of the FDA to give elamipretide a full drug review. In our many meetings with Congress, we have been assured that the FDA has the regulatory flexibility to review these data, yet they have not used it. This is a systemic problem affecting all who seek treatments for ultra-rare diseases. Every patient – no matter how rare their disease – deserves a chance at fulfilling, longer life. We are not too rare for care."

The patient community is also concerned that the boys and young men currently receiving elamipretide under an expanded access protocol will lose access to the drug if the FDA fails to act. For Jamie Dubuque, a young mother whose one-year-old son Declan experienced an unprecedented heart recovery while receiving elamipretide treatment through emergency access, the prospect of losing access is incredibly frightening.

"At 11 months old, my son Declan experienced life-threatening acute heart failure. He was admitted to a pediatric intensive care unit, placed on a ventricular assist device that helped pump blood for his failing heart, and was listed for a heart transplant. His doctors indicated his native heart would not recover sufficiently and a transplant was the only course of action," **said Jamie Dubuque.**

Dubuque continued: "To help him survive until the transplant could take place, Declan was placed on emergency access elamipretide. Less than three months after starting elamipretide, Declan's heart structure and function improved more than doctors ever expected, and he regained strength and energy. After nearly eight months on elamipretide, Declan's ventricular assist device was removed, and he was discharged from the hospital without requiring a heart transplant. His doctors credit elamipretide with making the difference. For Declan, now nearly 2 years old, and other clinical trial patients who are still on elamipretide, we need the FDA to act now and see the inevitable pain we'd face otherwise."

In addition to delivering the petition to FDA, BSF is spearheading a grassroots advocacy campaign called "Not Too Rare to Care" to raise awareness about Barth syndrome and the community's advocacy for a fair review of the only potential treatment for this rare condition.

Barth syndrome is a rare, X-linked genetic disorder with an estimated incidence of only 1 in 1 million live births. Caused by a mutation in the *TAFAZZIN* gene, also called *G4.5*, that results in an inborn error of phospholipid metabolism, Barth syndrome affects many systems of the body and is characterized by cardiac abnormalities often leading to heart failure and reduced life expectancy, recurrent infections, muscle weakness and delayed growth. Barth syndrome can be fatal in childhood due to heart failure or uncontrollable infection, with approximately 50 percent of deaths due to Barth syndrome occurring within the first year of life and 85 percent before the fifth year of life. Those who survive to adulthood have a severely reduced life expectancy and typically experience compromised physical health with medical conditions that can become life-threatening with little or no warning.

About Barth Syndrome Foundation (BSF)
Barth Syndrome Foundation and our international affiliates comprise the only global network of families, healthcare providers, and researchers solely driven by the mission to save lives through education, advances in treatment and finding a cure for Barth syndrome. Considered a role model in rare disease advocacy, BSF has funded $6.2M USD and catalyzed over $33M USD in funding from other sources to advance global scientific discoveries to end the suffering and loss of life from Barth syndrome. Additionally, BSF provides a lifeline to families and individuals living with Barth syndrome around the world, offering individualized support, educational conferences, a patient registry and collaborations with specialist healthcare providers to define standards of care, treatment and rapid diagnosis.