Chairman of the EveryLife Foundation Board of Directors, Mark Dant will testify today before Senate Health, Education, Labor and Pensions Committee, Children and Families Subcommittee. Dant will represent the 1 in 10 Americans affected by the more than 7,000 known rare diseases.

Mark’s son Ryan was diagnosed in the 90s with MPS 1 — a rare lysosomal storage disorder. The cells in Ryan’s body lacked a crucial enzyme they needed to break down sugar. Mark and his wife were told Ryan would die before adolescence, as there was no treatment for his disorder. Because MPS I was so rare, affecting only a few thousand children around the world, drug companies were not interested in funding the research.

Mark refused to accept the doctor’s recommendations and eventually raised more than $1 million for Dr. Emil Kakkis’ work on MPS 1, which culminated in a new drug therapy. In 2003, the FDA approved Aldurazyme for the treatment of MPS I. Ryan is now 30 years old and the longest treated MPS I person in the world. Unfortunately, Ryan’s story is the exception.

Today, 35 years since the Orphan Drug Act was signed into law, fewer than 400 of the 7,000 plus known rare diseases have FDA-approved treatments. Dant will call on Congress today to help close the innovation gap for the 95 percent of rare diseases that have no treatment by incentivizing companies to repurpose already approved drugs for Rare Diseases. Many patients are using drugs off-label. Drugs used off-label to meet these needs do not have the appropriate safety, efficacy, and dosing information. They also often lack coverage for the cost of the drugs, as many insurers will not pay for off-label use.

The bipartisan OPEN ACT (S. 1509 and companion bill H.R. 1223), introduced by Senators Orrin Hatch (R-UT) and Robert Menendez (D-NJ), is a patient-driven legislative solution supported by more than 300 rare disease patient organizations. Modeled after the bipartisan Best Pharmaceuticals for Children Act of 2002, which resulted in over 600 labeling changes and provided substantial clinical data on drug safety and efficacy in pediatric populations, the OPEN ACT has the potential to double the number of FDA-approved therapies for rare disease patients at a lower average cost than current rare disease drugs.

Dant will also ask Congress to fund a Center of Excellence for Rare Diseases and more specialized review divisions at the Food and Drug Administration. The FDA must have specialized personnel who understand the complexity of rare disease drug development to allow more flexible clinical trial designs, such as an “all-comers” trial that will allow our very small, heterogeneous patient populations to participate. Additionally, rare diseases still do not have access to the Accelerated Approval Pathway as novel biomarkers for rare diseases are not accepted as endpoints. Allowing the use of a biomarker as a surrogate endpoint will lower the cost of rare disease drug development by 62 percent. Ensuring that the FDA has the expertise and understanding needed for rare disease trial design will help de-risk the regulatory process and encourage investment in ultra-rare diseases.

Finally, Dant will ask Congress to seek policy solutions to alleviate the devastating diagnostic odyssey for rare diseases. For a rare disease patient, the diagnostic odyssey, or the time it takes for an individual to be accurately diagnosed, is about 7 years. This is unacceptable. The devastating effects of many diseases are irreversible. Early diagnosis is critical to ensure patients have access to clinical trials and lifesaving
therapies. Congress must reauthorize the Newborn Screening Saves Lives Act before it expires on Sept. 30, 2019. Additionally, the Senate should introduce companion legislation to the House’s Precision Medicine Act (H.R. 5062) to help mitigate and eventually end the diagnostic odyssey so many patients and their families endure. 80 percent of rare diseases are genetically based so coverage for genomic sequencing is critical.

The EveryLife Foundation for Rare Diseases, a science-based advocacy organization that works to bring lifesaving treatments to the 30 million Americans with rare diseases. There are more Americans who live with a rare disease than those who have HIV, heart disease, or stroke combined. 50 percent of rare disease patients are children, many of whom will not live to see their fifth birthday. Only 5 percent of rare diseases have FDA-approved treatments.

Please contact Grant Kerber at ### with questions.