United States Senate Health, Education, Labor and Pensions (HELP) Committee, Children and Families Subcommittee Hearing:
Rare Diseases: Expediting Treatment for Patients
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Chairman Paul, Ranking Member Casey and distinguished members of the Senate Health, Education, Labor and Pensions Committee, I am privileged to be here today to present my perspective as the parent of a child with a rare disease, and to represent the 1 in 10 Americans affected by the more than 7,000 known rare diseases. I serve as Chairman of the Board of 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.

When my son Ryan was 3 years old, he was diagnosed with MPS 1 – a rare lysosomal storage disorder. The cells in Ryan's body lacked a crucial enzyme they needed to break down sugar. The geneticist at Dallas Children's Hospital told us that children with MPS 1 almost never lived past their teens, 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. My wife Jeanne and I were told there were no options other than to take Ryan home and love him for as long as he lived.

I refused to accept the doctor's recommendations. Despite working the night shift after recently being promoted to Lieutenant in the Carrollton, Texas Police Department, I spent all my off-duty time trying to understand Ryan's condition. After a year of lying on the floor next to our son's bed at night and crying, my wife Jeanne and I founded a non-profit called the Ryan Foundation to raise money to find a treatment for MPS in time for our son. Our first fundraiser was a bake sale that netted $342. After several years of going door-to-door asking for donations and a series of conversations with leading scientists across the United States and Europe, I was told about a pediatric researcher who was working to find treatments for children with MPS 1: Dr. Emil Kakkis at Harbor UCLA. It was late 1994, 11 years after the passage of the Orphan Drug Act, yet Dr. Kakkis had no funding for his research and was working out of a one-story World War II era bungalow behind the county hospital in Torrance, California in a lab he'd constructed with the help of his own family members.

Over the course of the next several years, the all-volunteer Ryan Foundation managed to raise more than $1 million for Dr. Kakkis' work on MPS 1, which culminated in a new drug therapy. This therapy would never have come to fruition in enough time for Ryan without the formation of a small biotech company, which pulled Ryan's drug through the pipeline in time for him to survive. Rare disease absolutely needs biotech partners. Family organizations simply do not have the capital necessary to bring treatments to approval. There are simply not enough companies to bring science already available to approved therapies.

In 2003, the FDA approved Aldurazyme for the treatment of MPS I – five years after Ryan and nine other children began the trial at UCLA. Enzyme Replacement Therapy later turned out to be instrumental in treating several other previously untreated and devastating disorders, proving again that biotech involvement in one disorder leads to not one, but countless more rare disease treatments.

Ryan is now 30 years old and the longest treated MPS I person in the world. Unfortunately, Ryan's story is the exception. So many parents hope to be able to find the right experts and raise enough money in time to save their children, but most of them will not be as lucky as we were.

We are now 35 years since the Orphan Drug Act was signed into law, yet fewer than 400 of the 7,000 plus known rare diseases have FDA-approved treatments. We know from our work on Aldurazyme that it is possible to generate the commitments needed to bring rare disease drugs through the development process. It is often even faster and simpler to repurpose existing therapies for rare disease indications. We must incentivize industry to invest in rare disease therapies and to repurpose existing therapies for rare disease indications.

We call on Congress 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; including my own son Ryan. Even rare disease patients who are fortunate enough to be treated with an FDA-approved therapy have multiple unmet needs that continue to alter their ability to live life without the pain and disability typically associated with their rare disease. 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), 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. I urge Congress pass the OPEN ACT before the end of this year.

I 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, I ask Congress to seek policy solutions to alleviate the devastating diagnostic odyssey for our community. 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 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.

I ask all of you gathered here today – Republicans, Democrats, Independents – please put your politics aside and join the ***rare*** party. I have spoken to countless rare disease families like my own across the

country and their message is the same: Drug companies are not the enemy, nor is the FDA. Our enemies are the rare diseases that steal livelihoods, mobility, vision, minds, and in the most devastating cases – lives.

I work with many parents who have raised the money to develop the science, yet no drug company is interested in developing the treatment. My advice is for them is to start their own drug company. However, I ask you: Should that also be their burden? We need Congress to incentivize drug companies and innovators to partner with us to bring lifesaving treatments to patients before it's too late.

I have personally felt the pain of finding no hope because a rare disease has stolen the promise of our tomorrows. I have attended countless funerals of children who lost their battle to a rare disease and witnessed the pain in their parents as they say goodbye. We must work together to change our system to increase the speed of safe and effective treatments from the scientific bench to the bedside by removing the barriers to novel trial designs. We must consider the heterogeneity of ultra-rare diseases and understand the true value of "all comer trials" so that our small patient populations are no longer overlooked, and the value of their data understood. Treatments come from the partnership of patients, science, industry, and the FDA.

Our children's lives depend on it.