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NATIONAL ORGANIZATION FOR RARE DISORDERS

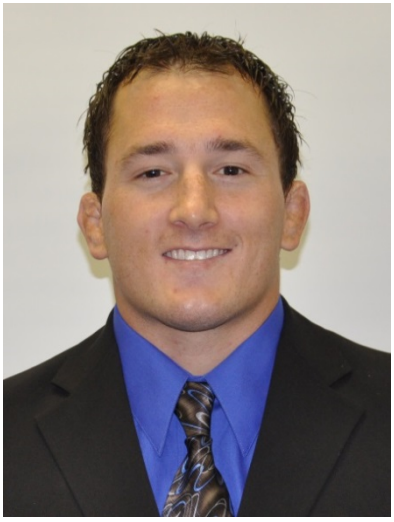
MAINTAINING INCENTIVES FOR RARE DISEASE DRUG DEVELOPMENT: A SNAPSHOT OF PATIENT EXPERIENCES

INTRODUCTION

The Orphan Drug Act (ODA) is an issue of huge importance to the rare disease community, patients, and families. Its impact on their lives is beyond measure. The incentives the ODA established for rare disease drug development—including the Orphan Drug Tax Credit—have successfully spurred innovation in treatment options for rare diseases (i.e., those that impact fewer than 200,000 people in the U.S.). Since the ODA was enacted in 1983, the Food and Drug Administration (FDA) has approved 486 orphan products, granted 3,280 orphan drug designations, and received 4,378 orphan drug designation requests.[1,2] Yet even these numbers do not reflect the full impact of the ODA on rare disease drug development since some of these innovative products have been approved for more than one disease or condition. For the rare disease community, these orphan products may not only significantly improve quality of life, but may be life-saving. The patient stories included below show the impact of orphan drugs on these individual patients, and serve as examples of the potential impact of orphan drugs on rare disease patients' lives.

SNAPSHOTS OF PATIENTS' EXPERIENCES

Joe Ellenberger, Diagnosed with a Rare Blood Disease

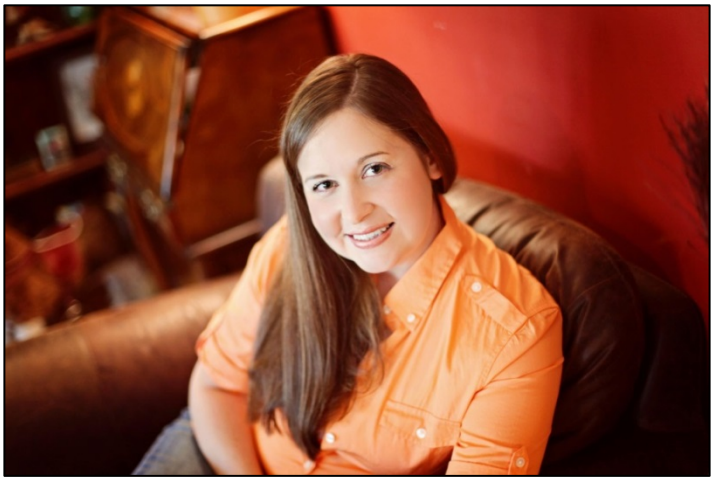


Joe Ellenberger is a former high school state wrestling champion and two-time NCAA All-American wrestler from Nebraska. In 2009, at the age of 24, he was diagnosed with Paroxysmal Nocturnal Hemoglobinuria (PNH), the rare blood disease that destroys red blood cells at an alarming rate and can lead to chronic kidney disease, blood clots, and other severe conditions. Research predicted he would not live beyond the age of 30. Joe was prescribed and took an orphan therapy that is intended to help mitigate some of the symptoms of the disease. Today, Joe is back in the cage pursuing his dreams to compete as a professional mixed martial artist in the Ultimate Fighting Championship. He is also a husband and father of two.

[1] FDA (2015, January 28). Search Orphan Drug Designations and Approvals, available at: http://www.accessdata.fda.gov/scripts/opdlisting/oopd/.

[2] Karst, K. (2015, February 15). The 2014 Numbers Are In: FDA's Orphan Drug Program Shatters Record [Web log post], available at: http://www.fdalawblog.net/fda_law_blog_hyman_phelps/2015/02/the-2014-numbers-are-in-fdas-orphan-drug-program-shatters-records.html.

Jill Ziegler, Diagnosed with a Life-Threatening Disease Causing Abnormal Blood Clots



At the age of 28, Jill Ziegler was the picture of health, living a busy life as a mom to a 3-year-old girl, wife, and pediatric occupational therapist. In December 2008, she became suddenly ill with renal failure although doctors did not know why. After tests and hospitalizations, Jill was diagnosed with Atypical Hemolytic Uremic Syndrome (aHUS), the life-threatening disease that causes abnormal blood clots to form in small blood vessels in the kidneys and can progressively damage vital organs.
Over the next two years, Jill faced an onslaught of dialysis, nausea, exhaustion, dangerously high blood pressure, seizures, and a mini-stroke. She tried to maintain a somewhat normal life despite facing end-stage kidney failure with no hope for a kidney transplant or effective treatment options. By June 2011, Jill began exploratory treatment with a therapy that, at the time it was first prescribed for her, was FDA approved to treat another condition—though it has been subsequently approved to mitigate the symptoms of aHUS—and also received a kidney transplant from a friend. In fact, she continues to receive that orphan therapy today and her life is restored: Jill is back to work and enjoys being an active mother and wife once again.

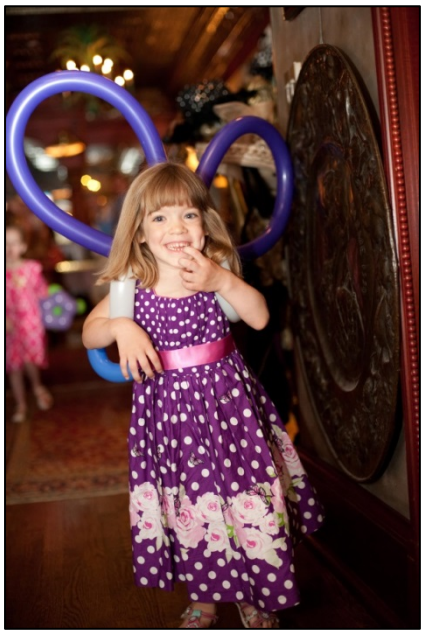
Bill Erickson, Diagnosed with a Rare Blood Disease

For Bill Erickson, the diagnosis of primary myelofibrosis (MF), one of the three rare blood diseases known as myeloproliferative neopl asms (MPNs) that can cause severe and debilitating symptoms, came as a huge surprise in May 2012. MF causes scar tissue to build up in the bone marrow preventing normal blood cells from being produced. While outwardly he had shown no symptoms, his spleen had already enlarged to twice its normal size. Bill began treatment with an orphan drug and his doctors found that his symptoms began to improve.

As part of his treatment, Bill also underwent a blood transfusion and stem cell transplant. Adopted at birth and with no knowledge of biological siblings, Bill relied on strangers in the bone marrow registry. Knowing that some people lose their lives waiting for a donor, Bill was surprised to learn in July 2012, six weeks after joining the registry, that he had six potential matches. Eventually, though he struggled through a series of complications, Bill's blood counts returned to normal levels. Today, he is feeling well and is focused on helping other MF patients through the MPN Research Foundation.

Joe, Jill and Bill's stories provide examples of rare diseases that may present later in life and may strike previously healthy individuals. Many rare diseases, however, are present at birth or in infancy. In fact, two thirds of America's 30 million rare disease patients are children.

Annabelle Bozarth, Diagnosed with an Inherited Metabolic Disorder



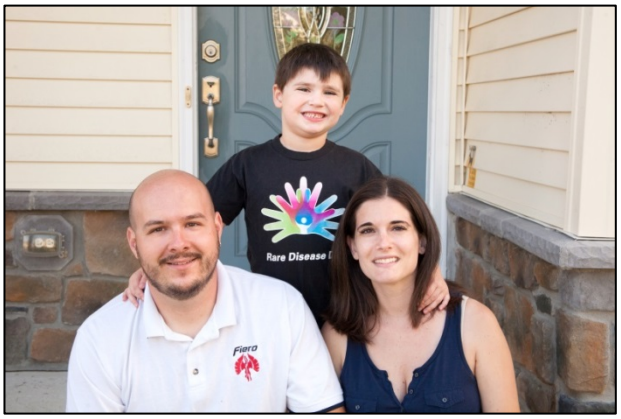
Annabelle Bozarth is a little girl living with Morquio A Syndrome (MPS IVA), which is caused by the lack of an enzyme needed to break down long chains of sugar molecules. Without treatment, it can lead to problems with bone development, growth, mobility, and cognitive function, and life expectancy is short.

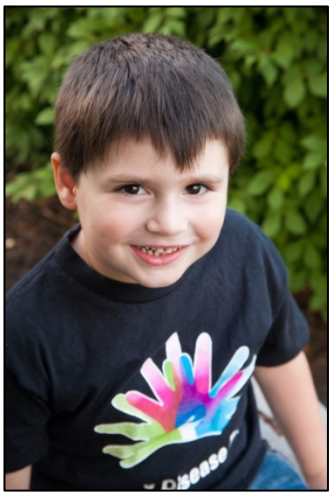
"Upon Annabelle's diagnosis at 6 months, our lives were turned upside down," says her mother, Stephanie. "Up until 5 years, Annabelle was in a downward spiral of fatigue, pain, and surgeries. In 2011, Annabelle began treatment and now, rather than running for the pain medicine, I am chasing my child through parks." Annabelle was prescribed and took a first-in-class FDA-approved orphan therapy intended to help mitigate the symptoms of Morquio. Her family has seen vast improvements. Today, she has the opportunity to be a kid again and the chance to keep fighting this disease.

Evan Miller, Diagnosed with a Potentially Life-Threatening Inherited Disorder

After a false negative as part of a standard newborn screening test, Evan Miller suffered a medical crisis at 12 weeks old that nearly took his life. He was diagnosed with Hereditary Tyrosinemia Type 1 (HT-1), the potentially life-threatening condition caused by a lack of the FAH enzyme than can lead to failure to thrive, jaundice, an enlarged liver, and other symptoms that may progress to more serious complications.

Following his diagnosis, Evan began treatment with an FDA-approved therapy that is intended to help mitigate the symptoms of HT-1. Today he is a lively, healthy 6-year old boy.





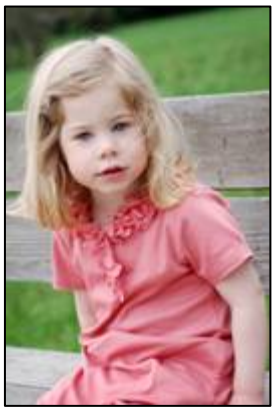
Melissa Landau, Diagnosed with a Lysosomal Storage Disorder

Some rare diseases have more than one approved treatment, such as Gaucher Disease. Melissa Landau Steinman is a Type 1 Gaucher patient who first learned of her diagnosis when she was five months pregnant with her first child. Her liver and spleen were enlarged, leading to concerns about expanding her family in the future. Melissa was prescribed and took an FDA-approved therapy to mitigate the symptoms of Type 1 Gaucher. One of her children, now 13, also has Gaucher although he is not yet symptomatic. Over the years, Melissa has tried nearly every treatment available, participating in FDA studies for several because she "likes to see as many drugs on the market as possible because some day my son will likely need treatment, too. It's good for me, good for him, and good for the world."

She calls the treatments that FDA has approved to mitigate Gaucher symptoms essential: "I'm a law firm partner, I work very hard, and I have a successful practice. Without treatment there's the potential to be very sick. The treatments are pretty essential… It is a matter of getting at the source of the problem until there is a cure."

These patients and their families are living proof of the life-changing and potentially life-saving impact that orphan drugs can have. Yet despite the fact that more than 400 orphan products have been approved since 1983, there are millions of Americans living with rare diseases who still have no FDA-approved treatment.

Noah and Laine VanHoutan, Diagnosed with a Rare Neurodegenerative Disease



Laine before diagnosis

These include Noah and Laine VanHoutan, who have a devastating rare condition known as Batten Disease. Their parents are among the many who are in a race against time to encourage research and the development of treatments to save their children's lives. "The hardest part about this disease [is] knowing that once upon a time, they were regular kids," says Jennifer VanHoutan, mom to Noah and Laine, who have Batten disease, a devastating and progressive neurological condition.

Noah before diagnosis

Noah and Laine began their lives as healthy, energetic kids who loved to jump, giggle, and run. They had endless things to say and were always looking for their next big adventures. They appeared perfectly healthy until they turned 3 years old, when they began suffering seizures and gradually lost motor skills. The family loses more of Noah, 11, and Laine, 9, with each month. At this time, Batten disease is always terminal, usually between the ages of 8 and 12.

The VanHoutan Family

Noah and Laine are among the millions who suffer from a rare disease that does not yet have an approved treatment. While the incentives established by the ODA have spurred progress, there is much more work to be done: there are thousands of identified rare diseases, and for the majority of these, there is not yet an approved treatment.[3] Thus, a commitment to maintaining the incentives in the ODA is crucial to continuing to make progress to find treatments and cures for these patients.

[3] NIH. National Center for Advancing Translational Sciences (NCATS), Office of Rare Disease Research (ORDR). (2015). Frequently Asked Questions, available at: https://rarediseases.info.nih.gov/about-ordr/pages/31/frequently-asked-questions.