

Education's Caitlin Torcato heads to Washington, D.C. for Rare Disease Week

Caitlin Torcato, a Training and Development Specialist for the Oregon Child Support Program Education Team, was among the 600 rare disease advocates gathered in Washington, D.C., in March for the 12th Annual Rare Disease Week on Capitol Hill. Caitlin joined her sister, Meagan Moore, who is the Usher Syndrome Coalition USH Ambassador for Oregon. Meagan is also one of about 25,000 people living in the United States with Usher syndrome (USH), a genetic disorder that is the leading genetic cause of combined hearing and progressive vision loss.

Meagan's Story

Meagan's mother first noticed signs of her daughter's hearing loss when Meagan was about two years old. However, Meagan wasn't diagnosed with the rare genetic condition until age 24 when symptoms of retinitis pigmentosa developed. Retinitis pigmentosa, the cause of blindness in people with Usher syndrome, leads to progressive tunnel vision. Two years after graduating from college and beginning a nursing career, doctors told Meagan she would lose her vision due to Usher syndrome. Meagan and her family soon learned there were no FDA-approved treatments for the progressive vision loss. Although Meagan manages her hearing loss



Training and Development Specialist Caitlin Torcato (left) and her sister Meagan at the 12th Annual Rare Disease Week in Washington, D.C.

with hearing aids and by reading lips and body language, there is no course of treatment for her vision loss. Meagan chose to become a research patient for experimental treatments. Over seven years, she participated in various studies, attending countless appointments, which sometimes lasted for several days. While participating in a clinical trial investigating a

PROGRAM NEWS

new RNA therapy for the treatment of retinitis pigmentosa, Meagan's progressive loss of vision began to slow. Even more promising, the treatment had the potential to reverse some of her loss of vision. The company conducting the clinical trial reported that the treatment would be on the market in 2024 or 2025. Meagan and her family were hopeful and excited that a promising new treatment for retinitis pigmentosa would soon be available.

A year ago, however, researchers paused the clinical trial due to the standard FDA requirement that central vision improve with treatments for visual disorders. While Meagan's progressive tunnel vision loss was slowing, her central vision—the traditional measurement for improvement in ophthalmic treatments—was not improving. Central vision is unaffected by Usher syndrome. The trial therefore did not meet the FDA requirements to continue, which often happens in rare disease research. "This endpoint is impossible to achieve and makes the clinical trial have no way to be seen as successful by FDA standards," says Caitlin. "It is frustrating to know the science exists for many of these diseases to be treated, but they may never be developed because of roadblocks in the development process, one of which is a lack of funding."

Meagan is not alone. About 30 million Americans (1 in 10) are affected by a rare disease. Of more than 7,000 known rare diseases, 93% of them do not have an FDA-approved treatment. In 1983, Congress passed the Orphan Drug Act (ODA), which aims to increase the development of treatments for rare diseases. The Act recognizes a gap in research for rare diseases and provides incentives to conduct research into treatment, such as for treating visual impairment in people living with Usher syndrome, like Meagan.

Because Meagan's vision loss continues to progress, she made the difficult decision to



stop working as a nurse and to stop driving. She attended an orientation and mobility training to learn how to use a white cane and is learning to read braille. "She is adapting to her ever-changing world, all while being an awesome mom, advocating for herself and others, and educating those around her about the spectrum of blindness and deafness. Her husband and three adorable kids think she is the bee's knees, and I must agree!" says Caitlin.

Meagan continues to share her story, the importance of funding for rare disease research, and the value of achievable endpoints in clinical trials that reflect the nuances of rare diseases. Improvements in funding and research would increase the number of FDA-approved treatments for rare diseases. "Hearing stories, meeting people, and learning about other rare diseases made us feel less alone in this fight," Caitlin says. "Having a rare disease can make you and your family feel a bit alone, but Rare Disease Week showed us just how not alone we are and how strong we can be together."

For more information, visit the [EveryLife Foundation](#) and [Usher Syndrome Coalition](#) websites.

To learn more about Rare Disease Week on Capitol Hill, visit EveryLifeFoundation.org/rare-advocates/rare-disease-week/.