Show your stripes: One family's rare disease journey

If you hear hooves, think horses.

It's an old medical saying. If doctors observe a symptom, think about treating the most likely cause.

Chris Ostertag has heard the phrase, working as a director of mission services at OSF HealthCare. But he wants you to keep another animal in mind: zebras.

Zebras and their rare stripe pattern have become a symbol of rare disease awareness. It's a topic the Ostertag family knows well.

Hans' story

Chris and his wife Rachel had their second child, Hans, in June 2017. It was a typical pregnancy, Chris Ostertag says, but doctors were concerned after the birth. They noticed the soft spot on Hans' head was larger than normal. He had low muscle tone, dysmorphic features and failed a hearing screening.

"They told us any one of these things, we're not too worried about," Ostertag recalls. "But the fact that they saw *all* of these little things, they were suspicious there might be some sort of genetic diagnosis."

Several tests later yielded a diagnosis of Peroxisome Biogenesis Disorder – Zellweger Spectrum Disorder (PBD-ZSD), a disease that involves gene mutations which lead to a variety of symptoms and challenges like deafblindness, liver disease, seizures, feeding issues, adrenal insufficiency, low bone density and muscle tone, developmental delays and many others. Zellweger refers to Dr. Hans Zellweger, a leading researcher in this area of diseases. Ostertag says it's estimated that PBD-ZSD is seen in one in 50,000 births in the United States.

"Unfortunately, it's a terminal disorder. It's one that doesn't really have any effective treatment or cure," Ostertag says.

Hans passed away in June 2023, six days shy of his sixth birthday and one month away from a Make-A-Wish trip to Wisconsin. A sad time for the Ostertag family, but a time to amplify their message to other families facing this hardship: you are not alone.

Rare disease awareness

Ostertag says different countries define a rare disease in different ways. In the United States, the standard is 200,000 people or less impacted. Cystic fibrosis is a common example. While 200,000 or less may sound small, Ostertag says rare diseases impact one in 10 people worldwide.

Chris and his family continue to advocate for rare disease awareness. He has lobbied with state and federal lawmakers, spoken to college students and done countless media interviews. Since 2020, he has served on the board for the Global Foundation for Peroxisomal Disorders. In 2023, he became board chairperson.

"The foundation of what we do is connecting families who have this shared experience," Ostertag says. "We also do a lot of fundraising for research to find a potential treatment or cure."

While finding a cure may be tough, Ostertag says research could yield treatments to lessen the blow of certain symptoms, like blindness.

The last day in February is recognized as Rare Disease Day. In 2024 (a leap year), it falls on February 29th. A fitting match – rare disease awareness on a rare day.

What can you do? Ostertag says consider advocating about access to pediatric care and more comprehensive newborn screenings. Better screenings mean an earlier diagnosis and less wondering "What's next?"

Or, something simpler: show your stripes. Wear striped clothing on rare disease awareness day, akin to the zebra.

“Chances are, we know someone in our community who is impacted by a rare disease,” Ostertag says. “All of us can be advocates by supporting our local families. Get to know them and understand their disorder, their challenges and more importantly how to help.”