

Parents Share the Challenges of Caring for a Child with MMA

HemoShear is developing HST5040 for methylmalonic acidemia (MMA) and propionic acidemia (PA). These ultra-rare genetic metabolic diseases cause the build-up of harmful toxins that affect systems throughout the body and can lead to severe organ damage, seizures, developmental deficits, and even premature death.

Infants born with MMA and PA can become very sick within days after birth. Parents must cope with a potentially life-threatening illness, receive an unfamiliar diagnosis, quickly learn about these complex diseases, and begin to navigate the challenges of caring for their fragile babies.

We are working with online rare disease research platform AllStripes to conduct a natural history study called JUMP (Journey to Understand MMA and PA) to analyze the experience of patients. Every story is different as patients and families face the daunting journey of managing a complicated disease for life. The de-identified and aggregated data collected from patient records may help HemoShear and other researchers develop better treatments.

“We are grateful to the families who participate in our HERO clinical trial, and research projects like JUMP that help us to better understand the patient journey with these diseases,” says Brian Wamhoff, PhD, interim CEO. “The HemoShear team is motivated every day by the dedication of caregivers, courageous patients and the hope for families seeking better quality of life.”

Learn what families are going through by reading these two parent stories featured by AllStripes.



Two-year-old Callaway was born with MMA and spent much of her first year in the hospital before doctors ultimately recommended a life-saving liver transplant. MMA is now a smaller part of the picture of what her life looks like and she is a happy, fun, well-adjusted child.

The transplant has been life changing. I cannot imagine doing what we did for that first year for multiple more years while we wait for clinical trials. It was a brutal decision, because it's like trading one chronic disease for another. - Carina Farkas, Callaway's mom

Emma was born with MMA seven years ago. While her twin brother was healthy, she spent her first 23 days of life in the hospital. She was in and out of the hospital for short visits in her first few years. When she was four, she got sicker and eventually received a liver transplant at her doctor's recommendation.

We're still very diligent with illness. It's not that she's totally avoided the hospital, but she seems to bounce back. She played soccer and softball this year and did ballet. Her level of methylmalonic acid in her blood is still extremely high, but it's about 80% lower than it was before and it's become a bit more manageable. - Patrick, Emma's dad

