

Raising Awareness of Rare Diseases *with the West Family*

making a difference

By Content Coordinator Delia McMullen

Did you know that the month of February is officially designated as Rare Disease Awareness Month? While all families hope and pray that their lives won't be impacted by a rare disease diagnosis, for the West family, this became a reality when their youngest child was just four months old. Andrew West has an ultra-rare genetic form of epilepsy. We would like to tell their family's story here in our February issue to raise awareness for Andrew and the millions of people living with rare diseases.



Andrew at Shining Hope Farms

Lisa and Justin West met in medical school at Jefferson Medical College. Lisa is an ophthalmologist and Justin is a plastic and reconstructive surgeon. In November 2020, the Wests moved to Cornelius with their three young children (now 8, 6, and 4). Lisa's older sister, Amy Schaefer, has lived in Huntersville with her family since 2003. Lisa's parents, Margaret and Joseph Collea, moved to Cornelius in March 2020.

As physicians, Lisa and Justin are uniquely qualified to care for their son, Andrew, who has KCNT-1 epilepsy syndrome. In healthy individuals, the KCNT-1 gene plays an important role in our cells' ability to generate and transmit electrical signals. Mutations in this gene cause children to have medication-resistant seizures and developmental delay. Afflicted children start missing their normal milestones within weeks of the seizures starting, and the vast majority never learn to walk or talk. Lisa

explains, "Andrew had his first seizure at four months old, and up to that point he was developing normally. We didn't know his actual diagnosis until two months later when he was 6 months old. The doctors tested him for a genetic form of epilepsy after his seizures did not respond to medication. Andrew's disease is so rare, that the physicians could not give us a prognosis for him. He spent five months of his first year of life in the hospital." Andrew still suffers from daily seizures, but also cannot crawl, walk, talk or feed himself.

Andrew gets weekly occupational and physical therapy. He especially enjoys his Monday morning equine-assisted therapy sessions at Shining Hope Farms. He likes getting outside and being around the horses, and the movement of the horse will hopefully encourage him to walk on his own.

The West family enjoys spending time together outside in their beautiful neighborhood. They ride bikes, visit the Yacht Club playground, and walk their dog, Sawyer. Andrew's big sister, Carolyn, is an energetic and enthusiastic second grader who enjoys gymnastics, horseback riding, dance, chess club, and playing drums. Colin is an active and curious kindergartner who enjoys building Lego sets, learning about science, and playing guitar. They also both enjoy spending time with their older cousins who live in Huntersville. According to Justin, "I am proud of the people Carolyn and Colin



Justin and Andrew on Epilepsy Awareness Day 2021

are becoming. They are both happy, funny, loving and kind." Lisa adds, "We are raising our older children to be inclusive, and I believe having Andrew as their younger brother has made them extraordinarily empathetic at a

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young age. Their teachers and coaches have said both Carolyn and Colin are very caring with their classmates and are quick to help if one of their peers is having a hard time.”

The Wests continually strive to promote awareness of KCNT-1 Epilepsy for Andrew and other children like him. Justin is the Co-founder and Director of Clinical Medicine for the KCNT-1 Epilepsy Foundation, which supports research and drug development with a global network of doctors, research scientists, and pharmaceutical developers. Says Justin, “We are fortunate to have a world-class group collaborating with our foundation to help treat KCNT-1 children, help us better understand the disease, and develop a cure for this severe form of epilepsy.” In addition to supporting research and drug discovery efforts, the Foundation also provides much-needed support to families with newly diagnosed children. Lisa adds, “I think other rare-disease families have the same struggles we do with disease awareness. Justin and the other founding members of the KCNT-1 Foundation (all KCNT-1 parents and relatives) have worked diligently over the past few years to increase awareness with the end goal of finding a cure for our beautiful children. Andrew is so sweet and very brave, and we won’t ever stop seeking a cure for this terrible disease.”

How You Can Help

To learn more about the KCNT-1 Epilepsy Foundation or donate, please visit kcnt1epilepsy.org.

Information about Shining Hope Farms, an equine-assisted therapy program, can be found at shininghopefarms.org.



Lisa and Andrew



The West Family, Christmas Eve 2021



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