

The Leukodystrophy Care Network

The Need

Imagine your child experiencing an unexplained sudden loss of basic functions – a toddler who once constantly walked and ran suddenly stumbling with each step; an infant with a healthy appetite who once smiled and cooed now crying inconsolably for every waking hour and only eating a few ounces of milk a day; a high school cheerleader suddenly losing all muscular control at a football game and shaking violently on the field... Each of these real life examples was the result of a disease group called, Leukodystrophies.

Leukodystrophies are an inherited group of over 40 disorders of the white matter in the brain, affecting every 1 in 7,000 individuals. Onset of symptoms can occur in the first months of life through adulthood, and cause a rapid loss of abilities such as voluntary movement and speaking, and result in severe morbidity and death. In spite of the thousands affected by these diseases, Leukodystrophies remain virtually unknown, even in the medical community. This often results in misdiagnosis and insufficient therapies and treatment options once a correct diagnosis is reached.

Although, currently there is no cure for Leukodystrophies, ***they are all treatable***. And, while all of them are terminal disorders, individuals affected by Leukodystrophies require and deserve expert medical care, essential to providing life-altering treatment and therapies to vastly improve health and overall quality of life.

One Family's Story – Hunter Kelly

Pro Football Hall of Fame Quarterback, Jim Kelly and his wife Jill started Hunter's Hope Foundation after their son, Hunter (2/14/97 – 8/05/05), was diagnosed with a fatal genetic disease called Krabbe Disease, a form of Leukodystrophy.

Born on his father's birthday, Hunter seemed perfectly healthy at birth. In his first weeks of life, however, he became increasingly more irritable, but doctors reassured his parents that it was only colic. With their son's crying becoming more severe and continuing for more than twenty hours a day, the family desperately sought to ease Hunter's pain. Upon further examination and a battery of tests by medical specialists, Jim and Jill were told Hunter had Cerebral Palsy. This proved to be another misdiagnosis. Hunter's symptoms continued to rapidly worsen and he became virtually inconsolable. Additional tests were performed...

Hunter Kelly was diagnosed with Krabbe Leukodystrophy.

Jim and Jill were devastated as the doctor described the prognosis for their beautiful four-month-old son –

Hunter has Krabbe Leukodystrophy...

He lacks an important enzyme, which is causing his body to destroy its myelin, the white matter of the brain...

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He will never talk, he will never walk, and if you choose, he will need a feeding tube...

There is no treatment, no cure...

Take him home, make him comfortable...

He likely won't live beyond his first birthday...

The Kellys were given **no answers, no medical plan of action** to ease the debilitating effects of Krabbe Disease, and, perhaps worst of all, they were given **no hope**.

Although Hunter defied all medical expectations his doctors placed on his life, living a valiant eight-and-a-half years, Jim and Jill did their best to manage Hunter's pain and the manifestation of this horrible disease. They tried to give him the best quality of life possible. But due to the lack of expert, comprehensive care for those affected by Leukodystrophies, most of what they learned was through trial and error, and reactive to the damaging and painful complications that could have been prevented.

These include but are not limited to:

- His lungs were severely damaged from chronic pneumonia, common to children with Leukodystrophies. This was caused by his inability to swallow, aspiration of formula and lack of movement. His bouts of pneumonia were significantly reduced with a feeding tube change bypassing his stomach eliminating aspiration of formula, along with an aggressive respiratory therapy care plan.
- Hunter broke his arm and leg within a year of each other, for no apparent reason. His bones were weakened due to lack of movement and medications. The Kellys were unaware, until after his bones started breaking, that he needed to supplement his diet with calcium, magnesium and potassium.
- Chronic eye infections due to his limited ability to blink, which resulted in scarring that impaired Hunter's vision.
- Both of his hips were dislocated, which restricted his mobility and caused extreme, unnecessary pain.
- He had painful kidney stones.

Most, if not all, of these agonizing complications could have been avoided and certainly reduced in severity had Hunter received *proactive* and *all-inclusive* expert care from the very start.

Shortly after Hunter's diagnosis, the Kellys founded the Hunter's Hope Foundation. From the beginning, the Foundation aimed to:

- Fund research for better treatments and cure for Krabbe and other Leukodystrophies
- Raise awareness of the widely unknown disorders
- Help those affected by Leukodystrophies and their families, as they cope with the devastation of these horrible diseases.

Since the inception of Hunter's Hope, much progress has been made. There is now a lifesaving treatment to halt disease progression for those with Krabbe and some of the other Leukodystrophies

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through cord blood transplantation, available if diagnosed early, ideally through state mandated newborn screening nationwide.

Although improvement in a correct and timely diagnosis is still urgently needed, individuals affected by Leukodystrophies *are* being diagnosed earlier, giving relief they so desperately need sooner than ever before. Perhaps most importantly, through family care programs offered by Leukodystrophy foundations, such as Hunter's Hope Foundation's Annual Family and Medical Symposium, together with the advent of social media, hundreds of families affected by Leukodystrophies have established a network of hope and support, one that did not exist eighteen years ago.

So much has been accomplished, but **families are still faced with a lack of the full range of expert care their children desperately need and deserve**. Even for those diagnosed too late for treatment, there are many proven methods to provide a child with optimal care to provide the best quality of life possible. Yet, most clinicians remain unaware of the extensive and comprehensive medical care Leukodystrophy patients require.

Hunter's Hope together with numerous members of the Leukodystrophy community have a vision to provide *every* individual affected by Leukodystrophies immediate and exceptional comprehensive care through the Leukodystrophy Care Network (*LCN*).