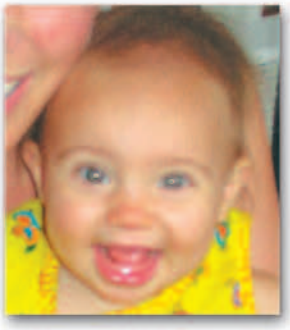
the Olive Branch fund:
a Thisbe and Noah Scott legacy



Thisbe Scott

Noah Scott

Elenna "Thisbe" Scott would have been three on July 11th, 2007. She was healthy the first 16 months of her life and sick beyond all comprehension the last 17 months.

It took five very long months to get a diagnosis. Nearly every pediatric neurologist from UCLA to Stanford and Boston Children's to the NIH could only tell us, "I've never heard of it." Even the diagnosing physician had never heard of it but found it by Googling Thisbe's symptoms. Eventually, we found one doctor who had seen a case like hers: 30 years earlier.

The medical term is progressive bulbar palsy (PBP), which is not a specific disease but more like a constellation of symptoms. These include the loss of swallowing, breathing, speaking, communicating, seeing, laughing, smiling, and eating. When this syndrome also involves hearing loss, it has been named Brown-Vialetto-Van Laere (BVVL).

This syndrome falls into a category of diseases known as pediatric motor neuron diseases (MND). Simply stated, motor neurons are tiny nerve cells that send signals to muscles to contract or move. When the motor nerve cells die, they stop sending signals to the muscles, and the muscles atrophy. While different motor neuron diseases can target other muscles, PBP and BVVL specifically target motor neurons within the cranial nerves and the muscles that are responsible for the basic functions of life. In Thisbe's case, BVVL began by paralyzing her vocal cords, and it kept on taking until there was nothing left - except her mind. As if this disease were not torture enough for a small child, it spared her mind so that she also cognitively suffered the ravaging of her little girl body.

It began on Thanksgiving Day of 2005, with something seemingly as simple as a wheeze. But within five weeks, we learned it was by no means something simple. Her vocal cords were paralyzed in a near-closed position, and she was having a hard time breathing. The motor neurons that controlled the muscles for speaking and breathing had been dying over the course of that month. As more neurons died, the muscles received less and less stimulation to move, and eventually, they just stopped moving altogether.

The only way to keep her from suffocating was to give her a tracheostomy: a tube in her neck that connected her lungs to the outside air that she breathed. Due to this unnatural mechanism for breathing, she developed repeated pneumonias and tiny "plugs" of mucus in the tube over time. These plugs repeatedly cut off her airway, sometimes as often as several times a day. In essence, each time she "plugged up" she was suffocating again and again – over and over, and we would rush to try to open her airway through the emergency procedures we'd learned in the hospital...and others we'd learned "in the field."

Eventually, because of a slow descending paralysis, Thisbe lost her ability to walk, hold up her neck, pick up her arms, move her fingers, and so much more. We felt as if we were watching our daughter being buried alive in her own skin. Her mind was perfect. She knew what was happening to her, but she couldn't understand why. In the beginning, she learned to communicate using sign language, but as she began to lose her arm, hand, and finger function, this became more and more difficult. And like any two-year-old, she would get frustrated. After all, she had swapped her beautiful, healthy life for a life where machines kept her alive and connected to a wall.

On Thanksgiving Day of 2006, exactly one year after her symptoms began, Thisbe developed a mucus plug that suffocated her...to death. She died in our arms, only to be resurrected by our crude emergency procedures and brought back to her hell to survive another five months. On April 30th, 2007, Thisbe died. She was two months shy of her 3rd birthday. Our little Thisbe could fight no longer.

One month after Thisbe's death, the other unimaginable happened. Our son, Noah, just ten-months-old at the time, began showing one of the initial symptoms of the same disease: a droopy eyelid. Two

months later, a test confirmed hearing loss. Two weeks later, his vocal cords were paralyzed, although in a slightly more open position than Thisbe's had been. Within five months, our son had lost all facial expression, and his walking became clumsy. His hand grasp was weakening, but he could still shoot hoops with the big boys. We had no idea how close we were to losing him.

Six months after the onset of his disease, on April 9, 2008, Noah died. He was five months shy of his 2nd birthday. Just like his sister, Noah died in our arms. He fought, just like his sister. He fought for every breath – for one long hour, every few minutes we thought he'd gone, but he took another horrible gasp. And even after all the oxygen in his body had run out, his strong little heart was still beating – barely – just barely – until he was gone. We buried our son exactly three weeks before the year-mark of Thisbe's funeral.

There are many forms of pediatric motor neuron diseases. Specifically, Thisbe and Noah suffered from the Brown-Vialetto-Van Laere type, which is rare, because while affecting a host of cranial nerves, it also affects the eighth cranial nerve, causing hearing loss. Though their minds were unaffected, their disease caused breathing and speaking difficulties, and a progressive descending paralysis resulting in the inability to communicate, laugh, eat, and even smile, before taking their lives. More common and well-known MNDs include ALS/Lou Gehrig's disease, progressive bulbar palsy (PBP), primary lateral sclerosis (PLS), progressive muscular atrophy (PMA), and spinal muscular atrophy (SMA). These affect both children and adults, who suffer through these diseases with normal cognition or thinking, even as they become trapped in a body that is wasting away. In the world of pediatrics, there are more than 25,000 Americans with spinal muscular atrophy (SMA), which is the most common pediatric MND. Although there are different types of SMA, together they make up the leading genetic cause of death of infants and toddlers.

It is important to know that 1 in every 35-40 people carries a gene that causes a motor neuron disease, which translates into approximately 7 million people. Thus, the child of 2 people with MND genes has a 1 in 4 chance of developing the disease. In other words, you and your spouse could each be carriers, and yet you could have four healthy children, all of whom had a 25% chance of being affected by the disease. If each of your children is healthy, then they hit the 75% jackpot; if this is the case, you will never know that you carry this gene or that you have passed the gene on to your children - that is, unless a grandchild or great-grandchild develops the disease. That, in turn, is how approximately 1 in 6,000 infants are born annually worldwide with a motor neuron disease.

There is no cure or standard treatment for MNDs. Most forms are fatal, and all forms are severely disabling due to paralysis of muscles. Sadly, many Americans have never heard of these diseases. Because of the intense national focus and fundraising efforts for pediatric cancer research in the past 20 years, the survival rate for a large number of pediatric cancers has increased dramatically, from as little as 4% in 1962 to 94% today. With cystic fibrosis, where in 1955 most children didn't make it to elementary school, today the median survival age is 37 years. The same ***can*** and ***must*** happen in the world of pediatric motor neuron diseases, a world where not one single disease has a viable treatment, even though we've known about them since the late 1800s.

We need to get to a place where everyone knows the name: "motor neuron disease." Just as there are many types of cancers, there are many types of motor neuron diseases. Although each cancer may have a different origin, they all have a common "front line" of treatment by way of chemotherapy and radiation. Similarly, although each MND may have a different genetic or environmental predisposition, there is likely a common thread of treatment for them all.

Researchers need funding to identify the genetic and environmental factors responsible for MNDs. If we can solve the mystery of how and why motor neurons die in these diseases, we can better understand how to reverse this process; regenerate the neurons themselves; and even create new neurons from other types of cells to forge new pathways to the atrophied muscles. The possibilities for treatments and a cure are endless and very much within our grasp.

Our names are John and Laurian Scott, and we are Thisbe and Noah's parents. We will never get over the loss of our daughter and our son, and their suffering is something we will equally never be able to reconcile. The first step toward finding a cure is finding the causes of motor neuron degeneration. This research is already underway, including research for the gene causing Brown-Vialetto-Van Laere. Once the BVVL gene is isolated, *The Olive Branch Fund* will initiate the next great leap: hiring a researcher for the cure. It is the mission of *The Olive Branch Fund: A Thisbe and Noah Scott Legacy* to promote research, awareness, and support for families of all pediatric motor neuron diseases, including Brown-Vialetto-Van Laere. As Thisbe and Noah's parents, we can think of no better way to honor their lives than to assure that a motor neuron disease will never again take the life or health of another child.

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The Olive Branch Fund is a component fund of
The Community Foundation of Middle Tennessee

THE COMMUNITY FOUNDATION OF MIDDLE TENNESSEE

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