



Meet Kirkland Kilbride

Born March 6, 2001, Kirkland was a healthy 8 lb. 9 oz. bouncing baby boy. He was cherished by his two older brothers and parents right from the very start. In fact, this sweet, gentle little boy won the hearts of all who met him. Newborn life for Kirkland was typical as his personality flourished and he began to reach expected developmental milestones.

At two months of age however, Kirkland began to suffer from Infantile Spasms- a rare type of seizure that proved difficult to control. The epilepsy worsened and side effects from medication resulted in the first of many hospitalizations for Kirk. At five months old, his family was informed that initial laboratory tests indicated low activity in the mitochondria of his muscle cells. At that time, it was felt probable that Kirkland suffered from a rare, progressive mitochondrial disease called Complex One Deficiency.

Kirkland did not however, exhibit some of the key symptoms that occur in most children with mitochondrial (*mito*) disease- mainly high levels of lactic acid in his blood, brain, or muscles or typical changes on his brain MRI. His scan was initially abnormal but then improved with time. For this reason and because DNA analysis for Complex One Deficiency was not yet (and remains unavailable as) a diagnostic tool, his metabolic doctor had never been convinced that Kirkland's defect of complex one was the sole cause of his severe health problems. In hopes of identifying a different disease that might provide the missing link, other metabolic tests were conducted on Kirkland's tissue samples over the years.

In December 2006, results from such a test came back with suspicions that Kirkland's cell activity was indicating Niemann-Pick Disease Type C (*NPC*) - a different, fatal genetic disease. NPC is characterized by the inability of cells to properly metabolize cholesterol and if confirmed, would place Kirkland among one of only 500 cases diagnosed worldwide. This abnormal accumulation of cholesterol and lipids is known to eventually smother cells and result in cell death in the brain, central nervous system, and other vital organs in the body.

This new discovery might explain why Kirkland's case of Complex One Deficiency had always appeared 'atypical' and never fully fit the mold. Doctors hypothesized that Kirkland's unique features of mitochondrial disease were actually secondary to an equally unusual presentation of Niemann-Pick Type C - which was the underlying genetic defect. As of April 2008, a definite diagnosis remains inconclusive because Kirkland's genes are still under the microscope.

Clinically, patients with different metabolic diseases often show similar symptoms. When one patient does not present with the 'typical' symptoms, as was the case with Kirkland, an accurate diagnosis can prove problematic. Where metabolic diseases *do* differ, is at the cellular level. Testing for the NPC gene is available; this makes its diagnosis more accurate. Scientists however, have yet to identify the genes for many of the other rare metabolic diseases such as those affecting the mitochondria. This challenge along with inadequate research funding, can frustrate the efforts of metabolic doctors and researchers who often struggle to offer diagnoses and treatments to their patients.

In Kirkland's case, it is thought that the cholesterol accumulation resulting from NPC affected his mitochondrial physiology. Mitochondria are found in all cells and convert food into energy. Hence, this could explain at least some of the reasons for his degenerative health. In most cases, the brain, lungs, muscles, central nervous system and other vital organs that require a great deal of energy to function are affected.

When such an energy crisis happens on a large scale basis throughout the body, whole systems begin to fail. Lives are compromised, changed, and for many...even prematurely ended. Mito diseases are very complex- often severe, unpredictable and difficult to diagnose. Because Kirkland's *mito* involvement was being connected to NPC, we knew that his early death was inevitable.

There continue to be no proven treatments or cures!

Symptoms resulting from defects of the mitochondria might include:

- poor growth
- visual* and/or hearing problems
- loss of motor control *
- gastro-intestinal disorders*
- muscle weakness*
- developmental delays* learning disabilities*
- Diabetes
- Seizures and neurological problems*
- cardiac disease
- susceptibility to infection*
- respiratory disorders*
- liver disease
- autonomic system dysfunction*

(*) Kirkland experienced health problems in these areas.

More information about mitochondrial disease can be found at The United Mitochondrial Disease Foundation's website: www.umdf.org.

More information about NPC can be found at The National Niemann-Pick Disease Foundation's website: www.nnpdf.org.

Treatment of Kirkland's condition required extensive medical intervention. During his short life, his complex health issues were overseen by fifteen paediatricians, four doctors, nine home nurses, four dieticians, a chiropractor, a Reiki practitioner, three social workers, five occupational/physical therapists, a developmental counsellor, an intervenor and a specialist for the visually impaired. Kirkland travelled to over one hundred and fifty medical appointments.

Drug treatment was a necessity. Throughout Kirkland's lifetime, a total of thirteen different anti-convulsants plus the ketogenic diet were administered in an attempt to control his intractable seizures. At six years of age, Kirkland's daily drug regime consisted of six anti-epileptic medications and nine other prescription drugs taken twice daily.

Despite these efforts, Kirkland's fight against NPD and the resulting mitochondrial defects was a challenge, taking its toll on both him and his family. Over two hundred days of his life were spent in hospital. Gastro-intestinal and respiratory problems resulted in thirteen cases of pneumonia and the need for a G-J feeding tube from the age of two. Kirkland was continuously fed a complete formula via pump and required frequent suctioning.

Twenty-four hour oxygen, pulse oximetry monitoring, and nebulizer inhalation treatments were needed at home to minimize hospital stays. In 2005, a bi-pap machine to assist with breathing was added to Kirkland's equipment cart beside his hospital bed in the family living room. Intermittent catheterization reduced pain and infection caused by urinary retention due to autonomic system malfunctions and daily enemas were a necessary part of his bowel routine.

Due to constant seizure activity in his brain, Kirkland's development was severely affected. Motor abilities were minimal and a seating system/wheelchair for proper posturing and transportation was required. A stander helped apply weight to his brittle, osteopenic bones. Kirkland was unable to support his head, sit independently, crawl, or walk. Although he was non-verbal, Kirkland understood several words, tactile cues, and signs.

Remarkably, Kirkland proved able to learn! Acute illness tended to hinder and cause losses in motor development, but his ability to listen, see, and communicate through sounds remained intact. A specialized developmental program was initiated by his intervenor and implemented by his caregivers when Kirkland was well enough.

Kirkland enjoyed music therapy, outdoor walks, simple cause and effect activities, visual stimulation, water play and interaction with friends and family. He was admired by all for his calm, soothing and complaisant demeanor. He silently tolerated the pain and discomfort of his symptoms without complaint, rarely crying or expressing frustration with his inability to communicate his needs. Kirkland was admired by all for the heroic manner with which he fought this debilitating disease and has been missed dearly since December 2007, when he finally succumbed to it.

Despite the challenges he faced each day of his life, Kirkland's impact was measurable and celebrated by many. His eyes exuded love and compassion, tolerance and faith-cherished gifts

that have enriched the lives of all who were fortunate enough to benefit from his unspoken wisdom.

The magnitude of Kirkland's legacy continues to unfold as he inspires many to work tirelessly on his behalf to raise awareness of mitochondrial disease and fund research at SickKids Hospital in Toronto.

We are all so proud of his accomplishments and feel blessed to have had him as our teacher.

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