The SLK #RareKiddos

A Disease May Be **RARE** *But* **HOPE** *Should Not Be*

Sean is the reason SLK exists. His fight was the inspiration to start the journey of helping others with rare diseases and special needs. Sean has Mucopolysaccharidosis type II (MPS II) (Hunter Syndrome), a rare lysosomal inborn error of metabolism that affects every organ of the body. Age of onset, disease severity and the rate of progression of the disease vary significantly but normally become apparent in ages two to four in children.

Liana was diagnosed with Cerebral Palsy, a neurological movement disorder characterized by the lack of muscle control and impairment in the coordination of movement. This disorder is usually a result of injury to the brain during early development in the uterus, at birth or in the first two years of life and is not progressive.

Gracyn has received a partial diagnosis of Cerebral Palsy, unfortunately she has yet to receive a full diagnosis. Cerebral Palsy is a neurological movement disorder characterized by the lack of muscle control and impairment in the coordination of movement. This disorder is usually a result of injury to the brain during early development in the uterus, at birth or in the first two years of life and is not progressive.

Sabra was diagnosed with Leigh Syndrome, a rare genetic neurometabolic disorder. It is characterized by the degeneration of the central nervous system (i.e: brain, spinal cord and optic nerve). The symptoms of Leigh Syndrome usually begin between three months and two years, but some patients do not exhibit signs and symptoms until several years later. Symptoms may include loss of previously acquired motor skills, loss of appetite, vomiting, irritability and/or seizure activity.

Reed was diagnosed with Kleefstra Syndrome, a very rare genetic disorder in which only about 400 individuals worldwide have been diagnosed in the last 10-15 years. Reed has Hypotonia, developmental delays and other medical conditions. On average individuals with Kleefstra Syndrome do not walk until 2-3 years old and if verbal, do not start talking until about 5 years old.

Temperance and Eric were diagnosed with CHARGE Syndrome: rare disorder that arises during early fetal development and affects multiple organ systems. The CHARGE acronym comes from the first letter of some of the more common features seen in children:

C = coloboma (usually retinochoroidal) and cranial nerve defects (80-90%)
H = heart defects in 75-85% especially tetralogy of Fallot
A = atresia of the choanae (blocked nasal breathing passages) 50-60%,
R = retardation of growth 70-80% and development
G = genital underdevelopment due to hypogonadotropic hypogonadism
E = ear abnormalities and sensorineural hearing loss (<90%)