

JUNIOR AMBASSADOR ~ LUKAS, age 3, Pennsylvania

"Lukas (Luke) and his twin sister were born healthy at 38 weeks. When Luke turned 15 months, his cheek started swelling quickly and he could barely swallow. He was rushed to the hospital and his bloodwork showed a significant infection in his lymph nodes. It required a hospitalization and strong IV antibiotics. That was the beginning of his medical adventure. His follow up labs showed Luke's white blood cell count had dropped significantly and was not going back to normal the way it should. It was then that we were referred to Hematology. We ended up seeing a wonderful hematologist at Children's Hospital of Philadelphia who ordered more tests. They tested him for Autoimmune Neutropenia, but Luke's antibody test came back negative - it was at that time that he was diagnosed with Chronic Benign Neutropenia. After about a year of having low neutrophil counts, Luke's hematologist decided to do genetic testing. It was then that we found out that Luke has a mutation of the COH1 gene which is an indicator of Cohen's Syndrome. Luke does not have some of the typical features and characteristics of Cohen's syndrome, but intermittent congenital Neutropenia is common with these individuals, as well as eye issues which may possibly explain a situation with Luke's eyes called, optic nerve atrophy.

Luke's neutrophil count has been in the normal range once since being diagnosed in 2017. He usually averages in the low to mid hundreds but has dropped to 0. We choose to keep Luke and his sister home from daycare because of the risk of infection/illness. Even with many precautions in place, he usually ends up being hospitalized a few times a year with many Emergency Department visits. We do see a correlation between low ANC levels and lower energy/decreased appetite. Aside from having frequent episodes of petechia and skin staph infections, Luke is overall a really happy toddler."

Luke's family has become involved with fundraising for Neutropenia to raise money for the National Neutropenia Network and to help spread awareness and an understanding of blood disorders and their possible complications. Although Luke is only 3, he has played a huge active role in advocating and helping fundraise for the NNN. As a result, Luke has earned the title of one of our Junior Ambassadors for 2019-2020....

Luke has helped his mom custom design t-shirts for their fundraisers. He helps choose the color, type of shirts and designs they use for their t-shirt sales. Luke's family posts the fundraisers online on several Facebook pages as well as on Instagram. Through fundraising this past year, they have raised \$145 that was donated directly to the National Neutropenia Network with almost \$600 raised over the past 2 years!

Luke has also recently become an A.W.A.R.E. advocate for the Aware jewelry line that raises awareness for different causes-his being a "rare disease". His story has been shared and is helping to raise awareness about Neutropenia.

"Designing and wearing our Neutropenia Awareness shirts has brought a sense of pride and happiness. We are able to talk to others about Neutropenia and spread awareness. We can help represent the 'invisible' conditions that may otherwise go unknown."

