

Claire's Story



Costello syndrome is a very rare spontaneous genetic disorder which affects many areas of the body including the heart, lymphatic system, metabolic system, cognitive development, mobility, eye sight, and hearing, as well as increases their risk of childhood cancer by 15%. The clinical description that best describes an individual with Costello syndrome is that they have "warm and sociable personalities." They are truly adorable. Approximately 500 to 600 individuals have been diagnosed with Costello syndrome in the world to date, and my daughter, Claire, is one of them.

The amazing Claire is almost 4 years old. Claire's life from the beginning was not easy. She was born with ectopic atrial tachycardia requiring a 9-day NICU stay and came home with heart medication to control her arrhythmia. Initially, everyone thought Claire simply had a crazy heart rhythm, but deep down I knew, from the beginning, something was not right with Claire.

Claire faced numerous medical problems throughout the first couple of years. The two major problems were her heart condition and macrocephaly, later diagnosed as hydrocephalus. It was at the time of her macrocephaly diagnosis that I broke down and admitted to her pediatrician that I have always felt something was wrong with Claire. I couldn't put my finger on it but since she was born, I had felt something was not right. Something was causing her problems, but I didn't know what. Claire's pediatrician also had concerns with her development and other aspects of her health. So, we started genetic testing when Claire was around 6 months old.

In the meantime, life continued. Claire was diagnosed with global developmental delay. She started physical and occupational therapy at 6 months old to help her work towards her developmental milestones that were not being met. Claire underwent 2 major brain surgeries to correct the hydrocephalus. She had her first endoscopic third ventriculostomy at 17 months old. Little did we know the surgery would fail just 6 weeks later, and she had a second brain surgery at 18.5 months old.



Claire has 9 different specialists who watch her grow and develop. She has had 7 surgeries ranging from PE tubes to brain surgery to tethered cord release and a growth removed from her bladder. She recently graduated from physical and occupational therapy, but does speech therapy twice a week. She is in a regular PreK 3 classroom with her peers and doing amazingly well. Claire recently found her passion for dance class which frequently results in impromptu living room performances. She brings so much joy and laughter into our lives.



After over a year of genetic testing, we finally got Claire's diagnosis of Costello syndrome when she was 20 months old. Many emotions went through my head that day. That night as I sat researching my daughter's new diagnosis, I found the Costello Syndrome Family Network Facebook page. I remember reaching out to one of the mothers on the page. As scared as I was that night, she shed hope on the situation. She was so comforting and knew just what I was feeling and going through at that time. Since that day, CSFN has been so supportive and always there to provide answers and suggestions when doctors have no idea how to treat our kids. Without CSFN, Claire would not be the happy thriving 4-year-old that she is today. She is excelling in all areas of life, and her joyous personality shows she is loving every minute of it.