I SUPPORT
RARE DISEASE DAY
FEBRUARY 28TH

The KAT6 Foundation proudly supports Rare Disease Day (RDD) the globally coordinated movement on rare diseases. RDD works towards equity in social opportunity, healthcare, and access to diagnosis and therapies for people living with a rare disease. A disease is rare when it affects fewer than 1 in 2,000 people.

KAT6A and KAT6B gene mutations have been diagnosed in approximately 600 individuals worldwide, we are ULTRA rare!

What is KAT6 syndrome?

KAT6 syndrome results from a genetic mutation in the KAT6A or KAT6B gene that occurs early in embryonic development. Typically, KAT6 mutations are de novo, which means they are not inherited from a parent but are rather completely new to the family's genealogy.

How can I find out if my child has a rare disorder, such as KAT6A or KAT6B?

Until recently, KAT6A and KAT6B gene mutations could only be found using a DNA test called Whole Exome Sequencing. Today, there is a Non-Specific Intellectual Disability panel that can detect these variants. Because KAT6 syndrome can be classified under larger umbrella diagnoses, such as autism, cerebral palsy, or global developmental delay, it is possible people with these gene mutations are being overlooked.
Discuss these options with a geneticist and genetic counselor. Testing may give you the answers to your child's developmental delays and/or health conditions.

"Before receiving my son's diagnosis of KAT6A, I thought we were alone in this journey. The sleepless nights, the GI issues, the surgeries, the unknowns were overwhelming, to say the least. After joining the KAT6 community and Facebook support group, I finally felt like there was hope, relief, and comfort in knowing we were certainly not in this by ourselves."

KAT6 Foundation

KAT6 Syndrome

KAT6A & KAT6B GENETIC MUTATIONS

COMMON TRAITS

- global developmental delay (physical and/or cognitive)
- intellectual disability
- significant speech and language delay
- congenital heart defects
- distinct facial features
- feeding difficulties
- vision problems
- gastrointestinal problems including reflux and constipation
- hypotonia
- microcephaly
- sleep disturbances
- problems with hips or knees
- growth problems
- genital anomalies
- autism spectrum disorder

LESS COMMON TRAITS

affecting fewer than 50% of diagnosed individuals

- endocrine conditions such as hypothyroidism
- respiratory problems
- skull abnormalities
- cortical visual impairment
- sensory processing disorder
- seizure disorders
- kidney problems
- dental anomalies
- hearing impairment
- ADHD
- frequent infections
- fractures

LEARN MORE AT KAT6FOUNDATION.ORG