UNDERSTANDING KAT6 SYNDROME

Variants in **KAT6A** and **KAT6B** genes can lead to a neurodevelopmental disorder.

KAT6A *AKA*: **Arboleda-Tham syndrome**, MOZ, MYST3, Lysine (K) acetyltransferase 6 A

KAT6B *AKA*: Genitopatellar syndrome (**GPS**), Say-Barber-Biesecker-Young-Simpson Syndrome **(SBBYSS)**, Ohdo syndrome, MORF, MYST4, Lysine (K) acetyltransferase 6 B

COMMON SYMPTOMS

- feeding difficulty
- GI issues
- motor/movement disorder
- intellectual disability
- ADHD
- autism
- sleep disturbance
- vision issues
- hypotonia
- craniofacial dysmorphism
- cardiac malformation

LESSER KNOWN SYMPTOMS

- frequent infections
- hearing loss
- fractures
- CVI
- dental anomalies
- respiratory problems
- renal/urogenital abnormalities
- thyroid issues
- chiari
- epilepsy
- growth delay

ULTRA RARE

A disease is considered **rare** if it affects fewer than **1 in 2,000** people.

DIAGNOSING KAT6

Ask your medical team about **Whole Exome Sequencing** and the **Intellectual Disability NGS Panel**. Both tests can identify KAT6 variants.

SPEECH DELAY

is the most common feature — ranging from mild to severe.

>1,000

Today, fewer than 1,000 individuals worldwide have been diagnosed with KAT6A or KAT6B genetic variants.

YOU CAN ACCELERATE **TREATMENTS** FOR KAT6 BY **SIGNING UP** TO PARTICIPATE IN RESEARCH AND JOINING OUR PATIENT REGISTRY.

Most KAT6 variants are

DE NOVO

meaning they aren't inherited.

GASTROINTESTINAL COMPLICATIONS

Individuals with KAT6 may develop **bowel obstructions**, (malrotation or volvulus), which require urgent treatment.

Never ignore signs of abdominal pain, vomiting, or severe constipation.

KAT6 Foundation was established in **2017**

We support the **international** KAT6 community.

THROUGH EVERY RESEARCH STUDY AND EVERY DONATION, WE'RE BUILDING A BRIGHTER, STRONGER FUTURE FOR KAT6 FAMILIES — TOGETHER

KAT6 Foundation

LEARN MORE AT KAT6.ORG