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 6A

KAT6B

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

DIAGNOSING KAT6

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

>1,000

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

Most KAT6 variants are

DE NOVO

meaning they aren't inherited.

KAT6 Foundation was established in **2017**

We support the **international** KAT6 community.

COMMON SYMPTOMS

feeding difficulty

GI issues

dental anomalies

microcephaly

intellectual disability

ADHD

autism

hypotonia

sleep disturbance

frequent infections

motor/movement disorder

vision issues

distinctive facial features

cardiac malformation

LESSER KNOWN SYMPTOMS

hearing loss

growth delay

respiratory problems

fractures

kidney/urogenital abnormalities

spinal differences

seizures

thyroid issues

ULTRA RARE

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

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

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

GASTROINTESTINAL COMPLICATIONS

Individuals with KAT6 may develop **bowel obstructions**, (malrotation or volvulus), which require urgent treatment. This is a medical emergency — never ignore signs of abdominal pain, vomiting, or severe constipation.

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