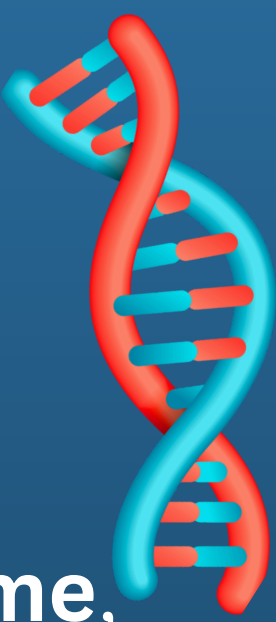


# UNDERSTANDING KAT6 SYNDROME

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



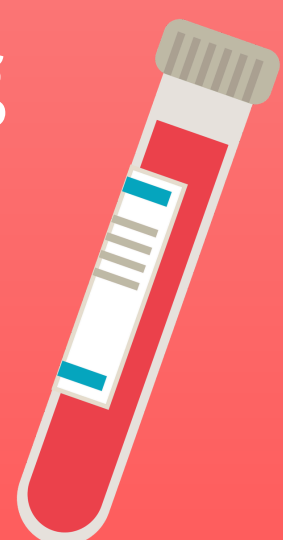
**KAT6A** AKA: **Arboleda-Tham syndrome**, MOZ, MYST3, Lysine (K) acetyltransferase 6A

**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.

feeding difficulty

hypotonia

GI issues

dental anomalies

microcephaly

intellectual disability

ADHD

autism

## COMMON SYMPTOMS

sleep disturbance

frequent infections

motor/movement disorder

distinctive facial features

vision issues

cardiac malformation

hearing loss

growth delay

respiratory problems

fractures

kidney/urogenital abnormalities

spinal differences

## LESSER KNOWN SYMPTOMS

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](http://KAT6.ORG)