KAT6 Foundation

KAT6A & KAT6B Caregiver Handbook

WWW.KAT6.ORG

Hello there!

WELCOME VIDEO

Learning that your child has KAT6 syndrome can bring many emotions all at once—questions, uncertainty, and hopes for what the future will look like for them. It's completely natural to feel overwhelmed by the unknown, but know that you're not alone as you take the next steps forward.

The first weeks and months after diagnosis can feel confusing and isolating. But as you learn more and meet other families on a similar journey, you'll see that you're part of a strong, compassionate community that lifts one another up. There is real hope ahead. Children with KAT6 continue to grow, learn, and surprise their families every day.

As time goes on, your child has more in common with other children than you first expected. Your child's needs are the same: love, learning, friendship, and connection, though they just might reach milestones in their own way and in their own time. KAT6 is a medical condition, it does not define your child's abilities, spirit, or potential.

While KAT6 syndrome brings unique challenges, it also reveals extraordinary lessons in resilience, advocacy, and unconditional love. Although it is one of the rarest genetic conditions in the world, affecting fewer than 1 in 10 million people, you are far from alone. The KAT6 community is filled with families who have walked this path before you and are eager to share guidance, encouragement, and support. Together with them, the KAT6 Foundation is committed to helping your family navigate every stage of this journey.

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What is KAT6 syndrome?

KAT6 syndrome is an ultra-rare genetic condition caused by variants (changes) in the KAT6A or KAT6B genes. These genes act as **epigenetic regulators**—they help "open up" our DNA so the right genes can be used at the right time for healthy growth and development.

Think of DNA like a thread tightly wound on a spool. For our bodies to work properly, that thread needs to be carefully unwrapped at just the right moment so each gene can do its job. KAT6 genes don't control just one function; they influence the activity of many different genes across our chromosomes.

Because of this, studying KAT6A and KAT6B gives scientists important insights into how different parts of the body grow and work together. What we learn from KAT6 research has the potential to improve health well beyond these rare syndromes.

Children with KAT6 syndrome grow and develop in their own unique ways, and their needs can vary widely. Some children benefit from more medical or developmental support, while others may have milder differences. No matter where your child falls on that spectrum, there are tools, therapies, and caring professionals who can help them learn, grow, and thrive at their own pace.

Scientists are still learning why this variability occurs, but evidence suggests that the type of variant and its location within the gene can influence both the type and severity of symptoms. This relationship between a specific gene change and the traits it affects is known as a **genotype–phenotype** correlation.

Diagnosing KAT6

KAT6 syndrome happens when a genetic variant arises very early in fetal development. In most cases, these changes are ***de novo*** (a Latin term meaning “from the beginning”), which means the variant is not inherited from a parent, but rather occurs spontaneously in the affected person.

Until recently, identifying KAT6 variants required comprehensive genetic testing that was often expensive and not widely available. Today, these tests are more commonly covered by insurance, and KAT6 variants are included in more targeted testing panels, making genetic testing faster and more accessible for families.

The main testing options include:

- **Whole Exome Sequencing (WES)**: Sequences all coding regions (exons) of the genome, which contain most disease-causing variants.
- **Whole Genome Sequencing (WGS)**: Sequences the entire genome, including coding and non-coding regions, making it the most comprehensive option.
- **Intellectual Disability Next-Generation Sequencing (ID NGS) Panel**: A targeted panel that includes KAT6 variants, often faster, more accessible, and more likely to be approved by insurance as an initial step.

Availability of testing may vary by country, so families should check local options.

Even with these advances, many children and adults with KAT6 syndrome are still overlooked. Because the features of KAT6 can resemble other conditions, such as autism, cerebral palsy, or global developmental delay, families may receive a general diagnosis without ever learning the underlying genetic cause. In addition, not all healthcare providers are familiar with rare genetic syndromes, so the right testing might not be offered.

The KAT6 Foundation is dedicated to raising awareness among families, clinicians, and researchers. Our goal is to help more people access genetic testing, so children and adults can receive an earlier and accurate diagnosis —and the support that comes with it.

As of 2026, fewer than 1,000 individuals worldwide have been formally diagnosed with KAT6 syndrome. But as testing becomes more common and awareness grows, we expect these numbers to rise, giving families and doctors a deeper understanding of this rare condition.

KAT6 Variants

KAT6 VARIANTS: WHAT WE KNOW AND WHAT WE ARE STILL LEARNING

Much remains to be learned about how specific KAT6A and KAT6B gene variants affect individuals. Most current findings are based on small research studies, and scientists are still working to understand exactly how different variants influence development and health. As research continues, what we know today may change.

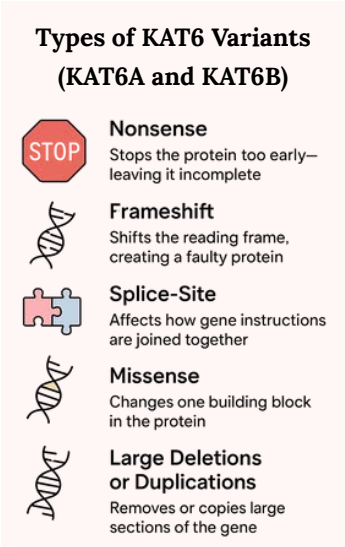
Genetic information can feel overwhelming, but you do not need to be an expert in genetics to understand your child. In most cases, a gene variant can help explain general patterns, but it cannot predict a child's future, abilities, personality, or potential. Your child's development is shaped by many factors, including therapies, medical care, family support, and their own unique strengths.

UNDERSTANDING KAT6 VARIANTS

Genes are like instructions written in a book. These instructions tell the body how to make proteins, which help cells do their jobs. Variants (or changes) in the KAT6A and KAT6B genes can affect how well these instructions are read and followed. Understanding your child's specific variant may help you better understand their symptoms.

TYPES OF KAT6 VARIANTS:

- **Nonsense or frameshift variants**– These cause the instructions to stop too early, so the protein is incomplete. This is like a sentence in a book that ends halfway through and does not make sense.
- **Splice-site variants**– These affect how the gene's instructions are cut and pieced together. Important parts may be left out or rearranged. This is like removing the prefix of a word, which can change its meaning or make it difficult to understand.
  For example, *remember → member*
- **Missense variants**– a single "letter" in the gene is changed, which swaps one building block of the protein for another. These are usually linked to milder symptoms. This is like a misspelled word: sometimes you you can still understand it, and other times the meaning changes.
  For example, sit → sat (here the content is still understood)
  sat → cat (here the meaning changes)
- **Large deletions or duplications** – These occur when part of a gene, or sometimes the whole gene, is missing (deleted) or repeated (duplicated). This is like leaving out entire sentences or having the same sentence repeated multiple times in a paragraph.



Learn more about genetic variants.

HOW VARIANT LOCATION RELATES TO SYMPTOMS

Researchers study where a variant occurs in the KAT6A or KAT6B gene because location can sometimes influence how the gene's instructions are read and how the resulting protein functions. This type of research helps identify general patterns across groups of individuals, but it is not used to predict outcomes for any one child. Genes are like long instruction manuals. A change in one part of the instructions may affect the final result differently than a change in another part. By studying variant location across many individuals, researchers can better understand how KAT6 genes function overall. Importantly, these patterns are not absolute, and children with the same variant location can have very different abilities, challenges, and developmental paths.

WHAT RESEARCH HAS OBSERVED SO FAR

Current research suggests that variant location may influence symptoms, although findings are based on a limited number of individuals and continue to evolve. In the largest published study of individuals with KAT6A variants, researchers found that variants occurring later in the gene were more often associated with more significant developmental challenges, while variants earlier in the gene were more commonly linked to milder speech and learning differences (Kennedy et al., 2019).

For KAT6B, clinical research has shown that variants tend to cluster in specific regions of the gene and are associated with different clinical patterns. Variants at the beginning of exon 18 are more often linked to Genitopatellar syndrome (GPS), which typically includes more significant motor, skeletal, and brain findings, while variants at the end of exon 18 or in exons 3, 7, 11, and 14-17 are more commonly associated with Say-Barber-Biesecker-Young-Simpson syndrome (SBBYS) (Johnston et al., 2018; GeneReviews, 2020). These findings suggest a spectrum of effects based on where the variant occurs, although every child's experience remains unique.

AN IMPORTANT MESSAGE FOR FAMILIES

These observations describe group trends, not individual outcomes. A child with a later variant may develop skills that exceed expectations, while a child with an earlier variant may face challenges that were not predicted. Variant location is only one part of a much larger picture that includes medical care, therapies, family support, and each child's unique strengths and needs. This information is meant to support understanding, not to limit hope or define what a child can achieve. Every child with KAT6 is unique, and their journey cannot be determined by genetics alone.

Features of KAT6

Gene	Syndrome Name(s)	Other Names for the Gene
KAT6A	Arboleda-Tham syndrome (ARTHS)	Lysine acetyltransferase 6A MOZ (Monocytic Leukemia Zinc finger protein) MYST3
KAT6B	Say-Barber-Biesecker-Young-Simpson syndrome (SBBYSS) Genitopatellar syndrome (GPS) Ohdo syndrome (OHDOS)	Lysine acetyltransferase 6B MORF (MOZ-related factor) MYST4

Children with KAT6A and KAT6B share many similarities. These two genes belong to the same family of **epigenetic regulators**, which means they help control how other genes work. When there's a change in one of these genes, it can affect many parts of development. While each syndrome has its own unique traits, there's a great deal of overlap between them.

Most children with KAT6 experience some degree of speech and motor delay, low muscle tone, and feeding or gastrointestinal challenges, particularly in early childhood. Sleep challenges and vision issues are also common. Children with KAT6B more often have distinct skeletal or joint differences, including joint contractures, limb differences, or kneecap abnormalities.

Some children with KAT6 may also experience neurological findings, such as seizures or differences in brain structure. Musculoskeletal concerns, including joint laxity, scoliosis, contractures, or muscle tightness, may become more noticeable as children grow. Despite these potential medical findings, data from the KAT6A/KAT6B Patient Registry (2025) show that **the majority of parents report their children are in good overall health.**

It's important to remember that every child is unique. The specific features, and how much they affect your child, can vary widely, even among children with the same genetic change. Your child will have their own combination of strengths, challenges, and personality. Families often describe their children as social, affectionate, and wonderfully resilient.

The chart on the next two pages gives a detailed overview of features seen in KAT6A and KAT6B, along with notes on when they tend to appear. Your family may find it helpful to print the chart and share it with your child's medical team to guide care and track progress.

REPORTED FEATURES IN KAT6 SYNDROMES

Very common: observed in 50% or more of individuals
Common: observed in 25–49% of individuals
Less common: observed in 5–24% of individuals
Rare: observed in fewer than 5% of individuals

Category	Feature	KAT6A	KAT6B	Onset
Development	Global developmental delay / intellectual disability	Very Common [5]	Very Common [5]	Recognized in infancy/toddler years
	Speech and language disorders (limited speech, apraxia)	Very Common [5]	Very Common [5]	Speech delay apparent in early childhood
	Hypotonia (low muscle tone)	Common [5]	Very Common [8]	Typically noted in infancy
	Behavioral disorders (includes: autism, sensory processing, ADHD)	Common [7]	Less Common [8]	Observed across childhood
	Motor skill delays (gross/ fine motor, movement disorder)	Common [5]	Very Common [5]	Evident in infancy/ toddler years and can persist
Neurology	Epilepsy / seizures	Less Common [5]	Common [5]	May appear in childhood
	Brain structure differences (Chiari malformation, corpus callosum abnormalities, ventriculomegaly, agenesis, lissencephaly, tethered cord, delayed myelination)	Rare [4]	Very Common [8]	Identified on MRI, usually in childhood
Feeding & GI	Feeding difficulties (swallowing issues, tube-feeding)	Very Common [7]	Very Common [8]	Very evident in infancy; some require long-term tube feeding
	Gastrointestinal issues (constipation, reflux)	Very Common [7]	Very Common [8]	Persist throughout childhood/adulthood and often requires ongoing intervention
Vision	Strabismus, cortical visual impairment (CVI), refractive errors	Very Common [7]	Very Common [8]	Typically recognized in infancy/early childhood
Hearing	Hearing loss (conductive and sensorineural), perforated ear drums	Less Common [7]	Common [8]	Typically recognized in infancy/early childhood

Commonality was based on data from the following sources: 1. Arboleda et al., 2015 4. Kennedy et al., 2019 5. NORD Patient Registry, 2025 6. National Organization for Rare Disorders, 2023 7. Tripathi et al., 2025 8. Yabumoto et al., 2021

REPORTED FEATURES IN KAT6 SYNDROMES

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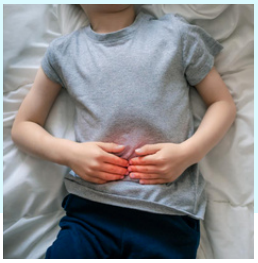
Category	Feature	KAT6A	KAT6B	Onset
Congenital / Organ	Distinctive facial features (microcephaly, dysmorphism)	Very Common [7]	Very Common [8]	Recognized from infancy
	Congenital heart defects (Atrial Septal Defect (ASD), Ventricular Septal Defect (VSD), Patent Ductus Arteriosus (PDA), and valve anomalies)	Very Common [7]	Very Common [8]	Present at birth, if present
	Microcephaly	Common [7]	Common [8]	Recognized from infancy
	Kidney / urinary tract / genital anomalies (hydroenphrosis, reflux, structural anomalies)	Less common [7]	Very Common [8]	Detected in infancy/childhood
	Endocrine issues (thyroid, growth hormone, puberty)	Less Common [7]	Common [8]	Onset is variable
Musculoskeletal	Joint laxity / hypermobility	Less Common [1]	Very Common [8]	Seen in infancy/early childhood
	Spinal differences	Less Common [1]	Less Common [8]	Develops later in childhood/adolescence
	Abnormal patella (agenesis/hypoplasia)	Not Reported	Very Common [8]	Seen in infancy/early childhood
	Contractures / stiff joints	Not Reported	Very Common [8]	Present at birth but may develop with age and mobility limitations
Other Health Concerns	Growth concerns (short stature, weight)	Less Common [7]	Less Common [8]	Usually noted in infancy/early years
	Frequent infections (respiratory, ear)	Common [5]	Common [5]	Start in infancy/toddler years
	Sleep issues	Common [7]	Less Common [8]	Often apparent in infancy and may persist
	Dental abnormalities	Common [6]	Common [8]	Seen in infancy/early childhood

Commonality was based on data from the following sources: 1. Arboleda et al., 2015 4. Kennedy et al., 2019 5. NORD Patient Registry, 2025 6.National Organization for Rare Disorders, 2023 7. Tripathi et al., 2025 8. Yabumoto et al., 2021

Gastrointestinal (GI) Issues in KAT6



GI issues are common in individuals with KAT6 due to low muscle tone and differences in gut motility. These can lead to constipation, slowed digestion, acid reflux, or more serious complications such as bowel obstructions. This may sound overwhelming, but treatments and feeding supports can make a big difference. Although rare, conditions like **malrotation** or **volvulus** can be life-threatening if not treated promptly. Because communication challenges and a high tolerance for pain are common in KAT6, you should monitor your child's bowel habits closely, watch for changes in behavior, and seek medical attention immediately if something seems wrong. Early recognition and intervention are key to preventing serious complications.

GI Terms to Know

Malrotation: abnormal positioning of the intestines during fetal development

Volvulus: Twisting of intestine causing bowel obstruction

MUST READ

BOWEL OBSTRUCTION – QUICK GUIDE

Children and adults with KAT6 may experience bowel obstructions, which can become life-threatening if not treated quickly.

SIGNS TO WATCH FOR:

- Changes in bowel movements or constipation
- Vomiting, diarrhea, bloating, or swelling
- Unusual irritability, withdrawal, lethargy, or restlessness
- If your child cannot reliably communicate pain, changes in behavior may be the first sign.

WHY IT MATTERS:

Some intestinal problems, like malrotation or volvulus need fast medical care. Even if your child is still passing gas or stool, a blockage can still be serious. If something seems wrong, seek medical attention immediately.

YOUR ROLE:

You know your child better than anyone. If you ever notice something that feels different for them or doesn't seem quite right, it's okay to reach out to your healthcare team. Trusting your instincts and speaking up when you have concerns can make a big difference in keeping your child healthy and supported.

Caregiver Checklist for KAT6 Syndromes

MEDICAL FOLLOW UP

- ◯ Meet with your child's pediatrician or geneticist to review the diagnosis.
- ◯ Confirm which specialists to see and schedule initial appointments.
- ◯ Keep copies of reports, test results, imaging, and recommendations.

EARLY INTERVENTION AND SCHOOL SERVICES

Ages 0–3:

- ◯ Request an Early Intervention evaluation.
- ◯ Track therapy recommendations and progress.

School age:

- ◯ Request a school-based evaluation.
- ◯ Set up an Individualized Education Plan (IEP) or the equivalent used in your country.

REGISTER

- ◯ Join the KAT6A/KAT6B Patient Registry. Register Here
- ◯ Complete the KAT6 Community Survey. Take Survey

CAREGIVER SUPPORT

- ◯ Join the KAT6 Support Group on Facebook to connect with others for shared experiences and advice. Join Group
- ◯ Watch the Foundation's welcome video for new families. Watch Video
- ◯ Lean on your friends and family—they can be a vital source of emotional support.

ORGANIZE YOUR RECORDS

- ◯ Create a binder or digital folder for:
  - Medical summaries and evaluations
  - Therapy and school plans and reports
  - Specialist contact information
  - Appointment notes
  - Development logs

STAY UPDATED

- ◯ Follow the KAT6 Foundation on Facebook, Instagram, LinkedIn, X, and YouTube.

RESEARCH OPPORTUNITIES

- ◯ Review current research studies and data collection projects at KAT6.org.
- ◯ Sign up to contribute to the KAT6 iPSC Bank. Learn more.
- ◯ Save any consent documents for your records.

Medical Specialists

Because KAT6 affects many areas of development and health, most children work with a team of specialists who coordinate care together. Families usually start with a pediatrician or geneticist who oversees early needs and makes referrals. If available, a multidisciplinary clinic can also simplify care by bringing multiple specialists into one setting.

Common specialists may include:

- **Geneticist** – explains the diagnosis, reviews your child's genetic report, and guides long-term care.
- **Developmental pediatrician** – monitors milestones and coordinates referrals.
- **Neurologist** – evaluates muscle tone, seizures, and other nervous system concerns.
- **Gastroenterologist (GI)** – manages feeding issues, reflux, and constipation.
- **Cardiologist** – checks for heart differences.
- **Endocrinologist** – monitors growth, thyroid, and hormone development.
- **Ophthalmologist and audiologist** – assess vision and hearing.
- **Orthopedist and orthotist** – evaluate bones and joints and provides supportive devices if needed.
- **Ear, nose, and throat specialist (ENT)** – addresses airway concerns, ear infections, hearing, and swallowing or breathing difficulties.
- **Nutritionist, feeding therapist, or dietitian** – support feeding and healthy growth.
- **Dentist** – monitors oral health, tooth development, and dental issues.

Your child's care team may change over time. Some specialists, like behavioral or mental health providers, may be added as new needs arise, while others may no longer be needed as your child grows. Because every child with KAT6 is unique, it's important for their care to adapt to who they are and what they need at each stage.

KAT6 Clinic Multi-Disciplinary Clinic at Boston Children's Hospital

Led by Dr. Olaf Bodamer and Dr. William Brucker, a multidisciplinary clinical program opened in February 2026, offering a comprehensive medical home for patients with KAT6A or KAT6B syndromes.

To schedule an appointment or for further information, please contact the KAT6 clinic coordinator at rarediseases@childrens.harvard.edu.

Boston Children's Hospital
Where the world comes for answers

Medical Checklist – KAT6A/KAT6B Syndrome

Tips for Families:

*Baseline assessments for heart, kidney, brain, and endocrine function are especially important.
*Your child may not need every test; follow your pediatrician or geneticist's guidance.
*Keep a copy of all results to track progress over time.

CARDIOLOGY
- ◯ Echocardiogram (to detect congenital heart defects: ASD, VSD, PDA, valve anomalies)

DEVELOPMENT & THERAPY
- ◯ Developmental evaluation by a developmental pediatrician or neurodevelopmental specialist
- ◯ Early intervention referral (PT, OT, Speech-Language Therapy)
- ◯ Feeding/swallowing evaluation by a speech-language pathologist or feeding specialist

NEUROLOGY
- ◯ Pediatric neurology evaluation (tone, reflexes, seizures)
- ◯ EEG (if seizures suspected or as baseline)
- ◯ Brain MRI (assess for Chiari malformation, ventriculomegaly, delayed myelination, tethered cord, agenesis of the corpus callosum) Your doctor may time the MRI for when anesthesia is already planned, such as during another procedure.

RENAL / URINARY TRACT
- ◯ Renal ultrasound (to detect structural anomalies or hydronephrosis)

ENDOCRINE
- ◯ Thyroid function tests
- ◯ Growth monitoring (height, weight, growth curves)
- ◯ Puberty evaluation (as child approaches adolescence)

GASTROINTESTINAL & NUTRITION
- ◯ GI evaluation (reflux, constipation, other issues)
- ◯ Nutrition assessment / dietitian consult (if growth or feeding concerns exist)

VISION & HEARING
- ◯ Ophthalmology exam (strabismus, cortical visual impairment, refractive errors)
- ◯ Hearing assessment

MUSCULOSKELETAL / ORTHOPEDIC
- ◯ Orthopedic evaluation (joint laxity, scoliosis, contractures, agenesis, hypoplasia)

SLEEP
- ◯ Sleep assessment (if trouble falling/staying asleep/sleep apnea suspected)

Therapies

Early intervention and ongoing therapy can make a remarkable difference for your child with KAT6 and are often available at no cost to families living in the U.S. Therapy referrals often come from your child's medical providers or educational team, and families may also choose to pursue therapies privately. Your child will progress at their own pace, but with the right supports and plenty of encouragement, you will see new skills emerge in beautiful and unexpected ways.

SPEECH THERAPY

Speech therapy addresses communication delays and can help your child develop verbal and non-verbal communication skills. Since children with KAT6 often have significant speech delays, your child's therapist can include augmentative and alternative communication (AAC), sign language, or other strategies to support expression and understanding. PROMPT therapy, which uses tactile cues to guide speech production, is often recommended for children with KAT6 to improve articulation and intelligibility.

FEEDING THERAPY

Feeding therapy addresses oral motor skills, swallowing difficulties, and sensory feeding challenges. Some children with KAT6 experience reflux, poor oral intake, or are tube-fed. Therapists work on safe feeding strategies, transitioning to oral feeding when possible, and improving nutritional intake.

PHYSICAL THERAPY (PT)

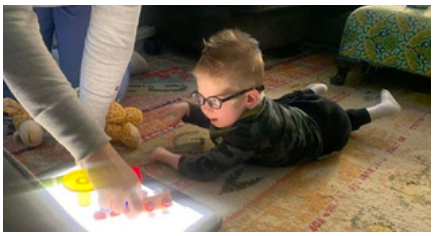
Physical therapy will help your child build strength, coordination, and confidence in their movements. Many children with KAT6 have low muscle tone (hypotonia), loose joints, or delayed motor milestones, which can make walking, standing, and other activities more challenging. PT supports your child in moving safely, improving balance, and reaching new milestones. Therapists can work with you to find the right equipment, like gait trainers, orthotics, or wheelchairs, so your child can participate fully and thrive.

OCCUPATIONAL THERAPY (OT)

Occupational therapy can help your child develop fine motor skills, self-care abilities, and adaptive strategies. OT can assist with feeding, dressing, writing, and using technology, as well as addressing sensory processing challenges, which are common in KAT6. Your child's OT may recommend adaptive equipment to support independence at home and school.

VISION THERAPY

Vision therapy may help your child if they have cortical visual impairment (CVI), strabismus, or other vision challenges. Therapy can improve tracking, focus, eye coordination, and visual processing, which are skills that support learning, safety, and everyday activities. Early evaluation by an ophthalmologist or vision therapist can help your child make the most of their visual development.



APPLIED-BEHAVIOR ANALYSIS (ABA)

ABA therapy focuses on building behavioral skills, social communication, and daily living abilities. For children with KAT6 syndromes, it can support language development, social interaction, and help reduce challenging behaviors. Programs are tailored to each child's strengths and needs, using positive reinforcement and skill-building to encourage growth.

AQUATIC THERAPY

Aquatic therapy leverages the buoyancy and resistance of water to support muscle strengthening, mobility, and coordination. Water provides a safe environment for children with hypotonia, limited endurance, or joint issues, and can make therapy enjoyable for your child while promoting functional movement skills.

HIPPOTHERAPY

Hippotherapy uses the gentle, rhythmic movement of a horse to improve balance, posture, coordination, and core strength. Beyond physical gains, many children enjoy riding, which boosts confidence, engagement, and participation in daily activities and therapy. While most insurance plans do not cover hippotherapy, many communities have nonprofits or riding programs that offer therapeutic horse services at low cost or through financial assistance.

INTENSIVE THERAPY APPROACH

Intensive therapy, typically lasting about three weeks, involves daily PT, OT, or speech sessions using specialized tools and techniques such as DMI (dynamic movement intervention). This intensive approach can lead to greater developmental gains in strength, mobility, communication, or functional skills compared to standard weekly sessions. Many KAT6 families notice significant progress during and after an intensive session.

Connect with families in your local disability community to learn from their experiences. Beyond traditional therapies, some families have found benefit in approaches such as music or art therapy, play therapy, biofeedback, reflex integration therapy, pet therapy, and respiratory therapy.

FAQ

Will my child walk?

Many children with KAT6 do learn to walk, though it often happens later than their peers. Others may move differently by using assistive equipment, wheelchairs, or alternative ways of getting around. Your child may need physical therapy or other supports to build strength and balance. Progress may come slowly, but with consistent therapy, encouragement, and patience, your child will continue to gain new skills and confidence in their own time.

What will learning and school look like for my child?

Children with KAT6 often experience developmental delays and learning differences. With early intervention, individualized education plans (IEPs), and therapy, your child can make meaningful academic progress. Every child's strengths and challenges are unique, so learning approaches and supports should be tailored to your child's individual needs. School supports may include classroom accommodations, a 1:1 aide, smaller learning environments, inclusive settings with supports in place, or specialized schools.

Will my child learn to talk?

Most individuals with KAT6 syndromes experience language delays, but abilities vary widely. Some children may be nonspeaking and communicate using signs, gestures, or adaptive technology, while others may become verbal teens or adults despite early delays. Many parents report that their children understand more than they can express. It's important to support your child's communication in all forms and maintain high expectations for their potential.

How can I support my child's communication if they're nonspeaking?

Many children with KAT6 are nonspeaking or have limited verbal language, but that doesn't mean they can't communicate. Speech therapy and alternative communication methods can help your child express their thoughts and connect with others. Tools such as sign language, picture boards, or speech-generating devices (known as augmentative and alternative communication, or AAC) give children a voice in their own way. It's important to model and encourage all forms of communication: gestures, facial expressions, or technology, and to celebrate every success.

How many individuals are diagnosed with KAT6?

As of 2026, fewer than 1,000 people worldwide have been diagnosed with a KAT6A or KAT6B variant. Based on a global population of more than 8 billion, this places KAT6 among the ultra-rare conditions, with a prevalence of roughly 1 in 10 million people.
The true number is likely higher, since genetic testing systems and medical institutions do not share data across countries. The most reliable way we track our community is through the KAT6 Patient Registry, which includes more than 500 individuals and continues to grow as awareness and testing improve.

What is the likelihood that I could have another child with KAT6?

Parents should consult with a geneticist to understand their individual likelihood and any considerations regarding future pregnancies. Most KAT6 gene changes occur for the first time in the affected child and are not inherited from either parent. In these cases, the chance of having another child with the same genetic change is low, estimated at about 2%. In rare situations, a parent may carry the genetic change in a small number of their reproductive cells, a condition known as germline mosaicism, which can increase the chance of recurrence. Very rarely, a parent may have the KAT6 variant in all of their cells and be able to pass it on in an inherited pattern. In those cases, each child has a 50% chance of inheriting the condition.

Where are KAT6 families located in the US and internationally?

Individuals with KAT6 have been identified in at least 40 countries. You can communicate with others through Facebook support groups and international WhatsApp groups. The KAT6 Foundation offers translated materials and can provide additional resources on request. You can also join the KAT6 Foundation's family map to connect with others nearby.

Join the map.

What treatments are available?

There are currently no approved therapies specifically for treating KAT6 syndrome, however some treatments may help specific symptoms. Doctors treat symptoms individually, such as constipation, acid reflux, behavioral issues, seizures, or sleep disturbances. Some parents report benefits from mitochondrial supplements like l-carnitine, pantothenic acid (B5), coenzyme Q10, Vitamin E, Vitamin C, or Cytra-3. Always consult your child's physician before starting any new supplement. More information on mitochondrial treatment can be found in Richard I Kelley's GI Webinar and in his 2023 conference presentation, Treatment of Metabolic Abnormalities in KAT6A and KAT6B Syndromes.

What is the life expectancy for individuals diagnosed with KAT6?

The KAT6 Foundation is actively collecting data through the KAT6 Patient Registry to study long-term outcomes. As of 2025, the oldest known individual with KAT6A is in their 50s, and the oldest with KAT6B is in their 40s. Because KAT6 is very rare and only recently recognized, most diagnosed individuals are children, so only a small number of adults have been identified. Ongoing research will continue to improve our understanding of health, longevity, and how the syndromes progress over time.

How can my family participate in research?

The best way to support KAT6 research is by joining the KAT6 Patient Registry, which collects valuable information from KAT6 caregivers to help scientists better understand KAT6A and KAT6B syndromes. You can also contribute biospecimens to the KAT6A and KAT6B iPSC Bank at Boston University. Stay up to date and find information on current research studies by visiting www.kat6.org.

Is our family eligible for financial assistance?

Your child may be eligible for government-supported healthcare or financial assistance related to disability, even if your family doesn't meet income requirements. Many countries offer programs that help cover medical care, therapy, equipment, and support services for children with genetic or developmental conditions. To learn what's available where you live, you can contact your child's healthcare team, a hospital social worker, or your local government or disability services office. They can guide you to benefits your child may qualify for and help you apply.

Information Resources

KAT6 FOUNDATION

- KAT6 Foundation Welcome Video
- KAT6 Biobank status report
- KAT6 Documentary: *Genetic Defekt* by Niko Mylonas
- KAT6 YouTube Channel: Research presentations and webinars on KAT6
- Of special concern: Bowel obstructions in the KAT6 population
- Archive of KAT6A and KAT6B Published research

KAT6A

- KAT6A fact sheet by Centre for Genetics Education
- KAT6A syndrome report by NORD
- KAT6A syndrome: Genotype–phenotype correlation in 76 patients with pathogenic KAT6A variants
- Speech and language development and genotype–phenotype correlation in 49 Individuals with KAT6A syndrome

KAT6B

- KAT6B disorders
- KAT6B-related disorders report by NORD
- Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms
- Say-Barber Biesecker Syndrome (SBBS) by Unique

INTERNATIONAL KAT6 CONTACTS

- Australia
- Austria
- France
- Italy
- Japan
- Netherlands
- New Zealand
- Norway
- Portugal
- Spain
- Switzerland
- Turkey
- United Kingdom

RARE DISEASE ORGANIZATIONS

- Asia Pacific Alliance of Rare Disease Organizations
- Global Genes
- Eurordis: Rare Diseases Europe
- International Rare Disease Consortium
- NORD (National Organization for Rare Disorders)
- Rare Disease Day
- Rare Diseases International
- Unique: Understanding Genes and Chromosomes

MAJOR FACEBOOK GROUPS

- KAT6 Foundation: KAT6A and KAT6B Research and Raising Awareness Group
- KAT6 Support Group
- KAT6A and KAT6B Gene Families

Visit the **Family Resources** page at KAT6.org for a more extensive library.

KAT6 Foundation

OUR MISSION

We support individuals and families living with KAT6 syndrome, caused by KAT6A and KAT6B variants, around the world. Our goal is to advance scientific research that may lead to treatments, and to raise awareness of KAT6 so it can be more easily recognized, understood, and studied.

KAT6 ADVOCACY

Our advocacy team is here to support your family every step of the way. We can help you navigate educational and medical systems, make informed decisions, and access the services and care your child needs to thrive.

We're also helpful for families transitioning from school to adult services. Our team can guide you through the process, explain your child's rights under an Individual Education Plan (IEP), and help you understand evaluation reports. You're not alone. Our goal is to make these sometimes confusing steps easier and less overwhelming.

.

We're here for you.

"You can't find a better resource than the actual community itself.... They're probably going to have more useful information than your pediatrician. It has been a huge support to have access to medically savvy parents, who know the condition, and those who have older kids and have been through this as well."

- Lindsey Blanch -

FUNDING RESEARCH

Thanks to family participation, researchers are uncovering new insights every year. These efforts have already led to better understanding of how KAT6 affects development, and are paving the way toward future treatments. The KAT6 Foundation proudly funds international research aimed at improving the lives of children and families affected by KAT6 syndrome. Our latest projects explore potential medicines, study brain and behavior models, identify important biomarkers, and develop clinical programs, laying the groundwork for future treatments and clinical trials. Every study we support brings us closer to better care and brighter futures for our community.
Learn more.

KAT6 PATIENT REGISTRY

Launched in 2019 through the National Organization for Rare Disorders (NORD), the KAT6A/KAT6B Patient Registry is the first long-term study of KAT6 syndrome. By collecting information from families like yours, the registry helps researchers better understand the full range of KAT6 characteristics and identify areas for new studies.

The KAT6 Foundation owns the de-identifiable data, which allows us to guide research and share findings responsibly. We also analyze the information and share insights with families, helping parents and caregivers better understand and support their loved ones. By participating, you're not only helping your own child, you're making a difference for the entire KAT6 community.

JOIN PATIENT REGISTRY

KAT6 IPSC BANK

In 2022, the KAT6 Foundation and collaborators created the first patient-derived induced pluripotent stem cell (iPSC) bank for KAT6A and KAT6B. These are cells made from patient samples, like skin or blood, that help researchers study KAT6 and test potential therapies. Families can choose to contribute samples from their child to the iPSC bank, giving your child a way to help advance research and support treatments for the KAT6 community. Tour Serrano Lab

JOIN THE IPSC BANK

"Working side by side with the KAT6 Foundation is one of the most rewarding parts of the work we do in the lab."

- Dr. Angie Serrano -

Empowered Grants

Supporting KAT6 families is at the heart of our mission. Through our Empowered Grants program, we provide funding to help families access assistive equipment, devices, technology, and therapies that can make a meaningful difference in daily life.

These resources can empower individuals with KAT6 syndromes to participate more fully in their communities, achieve educational goals, improve communication, strengthen connections, and foster social and emotional growth.

Since launching this initiative in 2020, the KAT6 Foundation has awarded more than 120 Empowered Grants to individuals and families in the U.S. and around the world. Each grant provides financial support to meet practical and life-enriching needs.

DOWNLOAD APPLICATION

WHAT YOU'LL NEED:

- ✓ Proof of diagnosis, such as a genetics report
- ✓ Completed application with contact information
- ✓ Estimated cost of item or therapy requested for reimbursement




Empowered Grants: List of Eligible Items

The following items are eligible for reimbursement through the **Empowered Grant Program**. This list is meant to serve as a guide rather than a limitation. If your child has a specific need that is not listed here but can be justified as beneficial to their development, health, or daily functioning, please include it in your application.

PT

- therapy balls
- matts, wedges
- foam rollers
- weights, resistance bands
- balance beams
- mobility aids, walkers, gait trainers, strollers, standers
- balance boards
- weighted vests
- adaptive seating
- portable wheelchair ramps
- scooter boards
- climbing walls, ladders
- obstacle course equipment
- orthotics
- pulley systems
- pedal exercisers
- trampolines
- crawling tunnels
- adaptive bikes and trikes

SENSORY

- swings (various types)
- chew tubes, chewable jewelry
- brushing systems
- Air Toobz
- weighted blanket/vests
- vibration tools
- fidget tools, stress balls, spinners
- light boxes/light tables
- lava lamps
- cooling vest
- mobiles
- headphones (noise reducing)

FEEDING/DAILY LIVING

- toileting aids
- eating utensils and cups (adaptive)
- reachers and grabbers
- adaptive toothbrush
- dressing aids
- eye glasses, hearing aids
- potty watches and timers
- adaptive high chairs

OT

- shape sorters
- tweezers and tongs
- handwriting tools
- construction toys such as blocks, legos
- craft supplies
- Squigz
- puzzles
- Play Doh, Theraputty, slime
- pegboards
- kinetic sand
- lacing cards
- games that require fine motor skills (Jenga, Operation, Marble Run, etc)
- easels, slant boards

SPEECH/COMMUNICATION

- AAC devices, tablets and cases (iPads), speech generating devices
- communication software and apps
- mirrors
- picture cards
- microphones and headphones
- whistles, bubbles, horns (blow toys)
- visual schedules, social stories
- games that promote turn taking and language development

EDUCATIONAL

- learning website subscriptions, e-books
- workbooks
- keyboards
- adaptive learning tools (stylus pens)

IN-PERSON THERAPIES

- physical therapy
- occupational therapy
- hippotherapy
- music therapy
- ABA therapy
- aquatic therapy
- vision therapy
- speech, feeding and AAC therapy
- social skills group

Community Events

KATWALK

The KATwalk is our fun, family-friendly annual event where supporters come together to raise awareness and funds for KAT6 research. It's a day full of smiles, activities, and community spirit, and a chance to celebrate our amazing KAT6 families! Together, we walk for a brighter tomorrow!

RARE DISEASE DAY

Rare Disease Day is a special day to shine a spotlight on rare conditions like KAT6 syndrome. It always takes place on the last day in February. We join together with families and supporters around the world to share stories, raise awareness, and show that no one needs to face a rare disease alone.

INTERNATIONAL KAT6 CONFERENCE

The annual conference is a unique opportunity for families, doctors, and researchers to come together. It's a place to learn the latest about KAT6, meet others who truly understand the journey, and even participate in research.

FIND AN EVENT

"I come because it makes me feel good to be here. I love that people love my son. I love that they cheer him on. I love that people tell me they see his progress. . . being a part of our community is really unique and special in a lot of different ways."

- Amy Young -

Get Involved

The KAT6 Foundation is powered by parents, friends, and supporters like you. Every effort makes a difference!

OFFER YOUR TIME

We're always looking for volunteers to support the Foundation's work. If you have special skills or talents to share, we'd love to hear from you! For those interested in a larger role, there are also opportunities to serve on the KAT6 Foundation Board of Directors.

VOLUNTEER

RAISE AWARENESS

You can help spread the word about KAT6 in your community and online.

- Join us each February for our Rare Disease Day Awareness Campaign.
- Encourage your friends and followers to connect with the Foundation on Facebook, Instagram, X (Twitter), LinkedIn, and YouTube.
- Share photos and stories about your child—we love highlighting our amazing KAT6 families and showing the world the faces behind our mission.

SHOP FOR KAT6

Our Bonfire shop has top quality clothing and merch for the whole family.

VISIT KAT6 SHOP

FUNDRAISE FOR KAT6

Our programs and research initiatives rely on the generosity of our community.

The KATwalk

Held each September, the KATwalk is our largest community fundraising and awareness event. You can support the cause by starting a fundraising team to host an in-person walk or by joining us virtually through KATwalk Anywhere. However you participate, every step helps us move closer to a brighter tomorrow.

Annual Appeal

Our Year-End Appeal invites major donors, corporations, and supporters to make an impactful contribution to our mission.

"I Care for Rare" Campaign

The "I Care for Rare" campaign is a flexible, ongoing fundraiser that you can start anytime. Partner with a local business, host an event, or dedicate a day of giving to raise awareness and funds for KAT6 families in your community.

WRITE FOR OUR BLOG

We'd love to hear your story!
Because KAT6 is so rare, many families can feel isolated. If you'd like to share your perspective, please reach out! Your words can help educate, inspire, and support families across the world. Read our blog.

Testimonial

Before receiving my son's diagnosis of KAT6, 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. The commonalities of our children blew me away and the insight and advice from parents have helped to guide many difficult decisions and have led us in the right direction for others. The work of the KAT6 Foundation is by far the most important research and education of our lives. There is no better way for us to support the foundation than through fundraising efforts that directly impact our son.

- Katie Bator -

JOIN OUR SUPPORT GROUP

Our Team

KAT6 Foundation Board Members

Dr. Jordan Muller
Board Chair

Karen Ginsburg
Fundraising

Dr. Katie LaRow Brown
Science

Maureen Martini
Board Member

Dr. Andrew Rankin
Science

David Woodbury
Secretary

Rachel Worden
Fundraising & Grants

Kevin Young
Finance

KAT6 Foundation Team Members

Aimee Reitzen
Marketing & Communications

Megan Stetts
Social Media

Susan Hartung
Advocacy & Patient Registry

Katie Bator
KATwalk

Dr. Tanya Tripathi
Research Coordinator

Dr. Typhaine Lejeune
Science Advisor

Jessica Wiemann
Empowered Grants

Lindsey Blanch
Grants and Sponsors

Parent volunteers support our board and team members in a range of roles. If you're interested in supporting our team or exploring a Board position, we'd love to connect.

The Foundation welcomes volunteers who want to help advance our work, especially those with skills or experience they'd like to share.

Contact our Board Chair, Jordan Muller, at support@kat6.org.

Let's Connect!

We'd love to stay in touch! By filling out the brief survey below, you help us share important research updates, community events, and support services. Your information stays within our KAT6 community and will never be shared outside of it.

Providing these details is a simple but powerful way to help us advance our mission and support you and your family. [View our privacy policy.]

COMMUNITY SURVEY

KAT6 Foundation

8 Leland Court
Chevy Chase, MD, US 20815

EIN # 82-3118535

Email: support@kat6.org

WWW.KAT6.ORG

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We are deeply grateful to the volunteers and professional advisors who shared their time and expertise throughout the development of this handbook, as well as to the members of the KAT6 Board who provided thoughtful feedback and review during the final stages. We also extend our sincere thanks to the KAT6 caregivers for their contributions to research within our ultra rare community and for their tireless efforts in advocating for and supporting their loved ones.

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