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

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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

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