

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)