KIF1A Associated Neurological Disorder (KAND)

KIF1A-associated neurological disorder (KAND) is a neurodegenerative genetic disorder caused by variants in the KIF1A gene, a motor protein that transports cargo in neurons. There are many KIF1A variants, leading to a broad range of symptoms.

Neurological & Behavioral

- Developmental delay/ intellectual disability: 92%
- Abnormal MRI: 56%
- Requires seizure medication: 61%
- Increased prevalence of autism, obsessive compulsive behavior, anxiety

Stomach & Digestion

- Reflux: 34%
- Constipation: 35%
- Requires gastrostomy tube: 6%

Muscles & Bones

- Hypotonia: 83%
- Hypertonia: 75%
- Scoliosis: 14%

Vision & Eyesight

- Overall vision/eye conditions: 83%
- Optic nerve atrophy: 43%
- Cortical visual impairment: 16%
- Strabismus: 23%

Other Symptoms

- Kidney problems: 23%
- Cerebellar atrophy: 7%
- Peripheral neuropathy: 27%
- Increased pain tolerance: 65%,
- Difficulty regulating temperature, cold feet/hands: 46%

Clinical Importance of KAND Diagnosis

KAND has a wide spectrum of symptoms and severity, depending on the specific mutation in the KIF1A gene. Patients may receive other initial diagnoses based on overlapping symptoms:

Children: Cerebral Palsy, Lennox-Gastaut, Rett Syndrome, Ataxia, Spastic Paraplegia.

Adults: Amyotrophic Lateral Sclerosis, Multiple Sclerosis.

There is currently no ICD-10 code for KAND, but diagnosis is crucial for future clinical trials and access to KAND-specific expertise and resources.

Lifelong Patient and Caregiver Support

As a progressive disorder, patient needs and may change: Puberty and transition into adulthood are both important milestones for symptom management and support. It is crucial to monitor for slow and subtle progression of symptoms and update care/treatment accordingly.

Referral to local community resources and social services is recommended.

Who can help me learn more?

Dr. Wendy Chung - Wendy.Chung@childrens.harvard.edu

- Chair of Pediatrics Boston Children's Hospital, oversees the KIF1A Natural History Study.

KIF1A.ORG - impact@kif1a.org

- Science-informed patient advocacy group with daily interactions with KAND families and the world's largest KIF1A registry.

KIF1A
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KIF1A Management Checklist

	Presentation	Management	Patient/Family observations: How does this affect this patient?
☐	Seizures	Refer to neurology Baseline 24-hr EEG followed by annual EEG (24-hr preferred) Epilepsy in KAND fact sheet	
☐	Neuropathy	Refer to neurology Hands and feet monitoring is critical Patients may be insensitive to injury-related pain while experiencing neuropathic tingling or pain in their extremities.	
☐	Neurodevelopment and Behavior	Developmental and behavioral assessments Early intervention and/or support services may be necessary (includes in-school, IEP) Refer for relevant therapies (speech, physical, occupational, ABA)	
☐	Movement	Refer to physiatry (if accessible, otherwise refer to PT)	
☐	Musculoskeletal	Yearly monitoring until growth is complete; adolescence/ growth spurts may need heightened monitoring Consider referral to orthopedic surgery, and consider orthotist	
☐	Vision	Refer to ophthalmology (or neuro-ophthalmology, if available). Consider referral to low vision services KAND Ophthalmologist Exams	
☐	Speech and Language	Refer to speech pathology Speech in KAND fact sheet	
☐	Digestive and Feeding	At least annual screening Consider referral to gastroenterology	
☐	Genitourinary	Refer to urology Urinary retention issues or genital irregularities may worsen in adolescence as sensation/muscle control decreases	