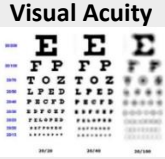




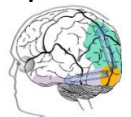


Visiting your Ophthalmologist: KAND Vision Exam List

KIF1A Associated Neurological Disorder (KAND) is a progressive neurodegenerative developmental disorder. 83% of patients with KAND experience visual impairment, but symptoms may be subtle¹. Dr. Aliaa Abdelhakim, MD, lead ophthalmologist in the KOALA clinical endpoint study, has compiled a list of potential vision symptoms in KAND, and relevant ophthalmic tests.

The 2024 KAND Natural History Study recommends *“regular assessments with a neuro-ophthalmologist to assess for optic nerve atrophy and cataracts that we have observed in some older individuals.”*¹ If KAND patients have reduced vision, a Low Vision Specialist may help address visual disability using directed functional adjustments and low vision technologies.

¹ [Sudhawa et al. 2024, Genetics in Medicine](#)

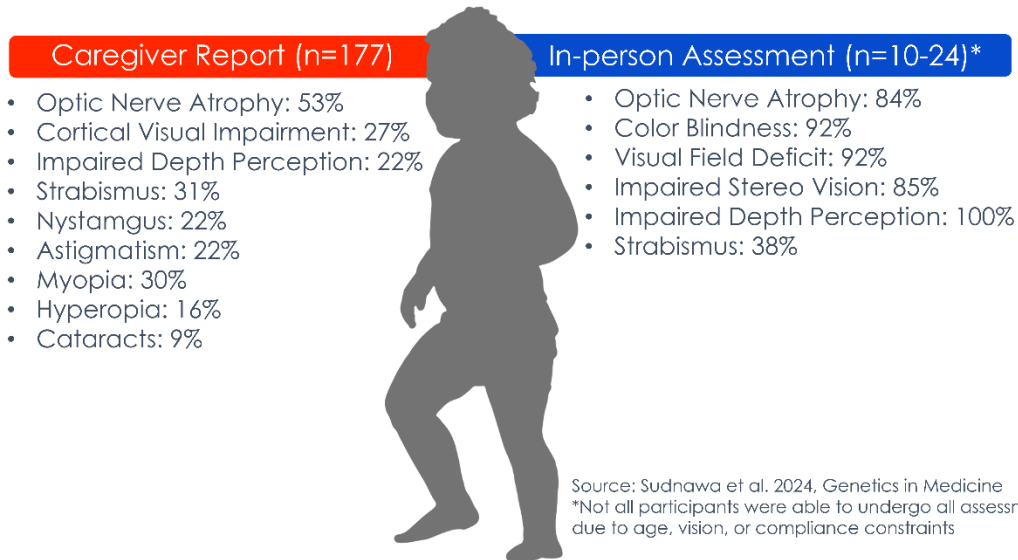
Clinical Feature	Signs	Tests and Considerations
Visual Acuity 	<ul style="list-style-type: none"> • Can't recognize people at a distance • Can't focus on nearby objects • Squints at light sources 	<p>Patients with KAND should be checked for Refractive Errors such as hyperopia (“far-sightedness”), myopia (“near-sightedness”), or astigmatism. These can be corrected with a glasses prescription.</p> <p>Children tend to want to take frames off, so it is important to find comfortable frames. Resilient frames and lenses are recommended for fall-prone patients.</p>
Field of Vision 	<ul style="list-style-type: none"> • Doesn't react to objects from side or center • Night blindness 	<p>Field of vision is how much you see in your peripheral vision or “side vision”. Your doctor can perform a test called Visual Field Testing. Visual field testing can be accomplished in different ways. Not all patients will be able to do this test as it requires focusing on the examiners face while reporting whether they can see objects in their peripheral vision.</p>
Strabismus/Nystagmus 	<ul style="list-style-type: none"> • Eyes don't align when looking at objects (may close one eye often) • Misjudges distances • Struggles to re-orient • Tilts or turns neck to direct small visual adjustments • Pain from eye strain or headaches 	<p>Many patients with KAND have strabismus, which means the two eyes are not working together the way they should, or nystagmus, which means that the eyes are reacting improperly to balance input from the inner ear. These conditions can be assessed with an Ocular Motility Test or other tests for vestibulo-ocular reflexes. Follow up frequency will depend on the degree of strabismus/nystagmus and their impact on vision.</p>
Optic Nerve Atrophy 	<ul style="list-style-type: none"> • Blind spots • Blurry vision 	<p>The majority of KAND patients have optic nerve atrophy¹, including thinned retinal nerve and ganglion cell layers. Your doctor will likely check for optic atrophy by doing a dilated eye exam, but this may not detect subtle nerve atrophy alone.</p> <p>To best track exam-to-exam changes, an optical coherence tomography (OCT) scan measures the thickness of the optic nerve and health of retinal cells. Not all patients can perform OCT.</p> <p>Optic nerve function may be compromised even when the structure appears intact. Functional testing with a Visual Evoked Potentials (VEPs) test checks electrical communication between the eyes and the brain. Not all ophthalmologists can test VEPs, but it may be worth asking if it is available.</p>
Color Vision 	<ul style="list-style-type: none"> • Uses the wrong colors when drawing • Lumps different colors together 	<p>10/11 children tested in the Natural History Study had color vision deficits. Your doctor can check for this using a color plates booklet, but if the patient is too young or if the vision is too severely affected, they may not be able to participate in this testing.</p>
Cerebral Visual Impairment 	<ul style="list-style-type: none"> • Struggles to recall names of objects • Overwhelmed in cluttered environments • Doesn't recognize faces • Delayed reactions to visual stimuli • Preference for stationary objects 	<p>Visual processing goes beyond the eye; problems with the brain's visual systems may manifest as blind spots or blurriness, or affect the way a person interacts with visual stimuli in more subtle ways. About 25% of patients with KAND have been diagnosed with cerebral visual impairment¹, which is likely underdiagnosed due to a lack of testing.</p>

For your Ophthalmologist: Vision in KAND Overview

In 2023, vision was established as a major symptom in KIF1A Neurological Disorder, and assessed as part of the KAND Natural History Study. Visit www.kif1a.org/Vision-in-KAND for more information.

As a fast and long-range neuronal motor protein, KIF1A is particularly important for cargo transport in white matter projection neurons: Optic nerve atrophy may be subtle and should be monitored closely.

Vision Symptoms in KAND



Quotes from caregivers on vision in KAND patients:

“Optic nerve atrophy was the first sign that something was not right.”

“He was also later diagnosed with optic nerve atrophy which has now declined to the point the ophthalmologist said it is the lowest reading the machine calculates so it is going to be difficult to track any more decline.”

“He is completely night blind... the combination of mobility difficulties with the blindness makes things extremely difficult for any independence safely.”

“He can see the moon... but he cannot identify us farther than 3-4 meters.”

“He struggles to find an object, even when it is right in front of him.”

“...she would look towards the TV when it was dark in the room but not look towards her sister playing next to her... I genuinely thought she had some form of autism and it was a form of avoidance rather than not actually being able to see.”

“[Vision loss] played a huge role in her slow development because of the lack of motivation due to not being able to see.”

Sudnawa et al. 2024. Heterogeneity of comprehensive clinical phenotype and longitudinal adaptive function and correlation with computational predictions of severity of missense genotypes in KIF1A-associated neurological disorder. *Genetics in Medicine*, <https://doi.org/10.1016/j.gim.2024.101169>