

Koolen-de Vries Syndrome

KdVS can affect many different body systems. Look out for and consider referrals to specialists for the following:

Ophthalmology:
hypermetropia, myopia,
strabismus, congenital
cataract, and optic atrophy

Brain Structural differences:
ventriculomegaly, corpus
callosum anomalies, Arnold-
Chiari type I malformation

**Speech and Language
Delay:** childhood apraxia of
speech, expressive and/or
receptive speech delay

Seizures: 50% of
individuals with KdVS
have epilepsy; ESES
may also be seen

ENT: recurrent ear
infections, hearing
impairment, stridor,
laryngeomalacia,
tracheomalacia, GERD

Congenital heart defects
may be present including:
VSD, ASD, bicuspid aortic
valve, cardiomyopathy,
aortic root dilatation

Genitourinary differences:
Cryptorchidism,
hypospadias,
hydronephrosis/VUR, renal
duplication

Musculoskeletal:
scoliosis, joint laxity,
flat feet, hip dysplasia

Hypotonia: feeding
difficulties, motor delay

Endocrine:
hypothyroidism and
neutropenia, Addison's
disease

Koolen-de Vries Syndrome

Specialist Checklist

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Ophthalmologist (eyes)

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ENT (ear, nose, throat)

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Speech Therapist

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Occupational Therapist (fine motor development)

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Physical Therapist (motor development)

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Cardiologist (heart)

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Neurologist (brain)

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Endocrinologist (hormones)

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Orthopedist (spine and bones)

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Urologist (urinary tract, kidneys, bladder)

This is a list of commonly seen specialists. Your child may have different medical needs.

For more information, visit <https://kdvsfoundation.org>