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Chromosome Disorder Outreach

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ABOUT US

Chromosome Disorder Outreach provides support and information to anyone diagnosed with a rare chromosome change, rearrangement or disorder. CDO actively promotes research and a positive community understanding of all chromosome disorders.

CDO is a 501c3 organization founded in 1992.

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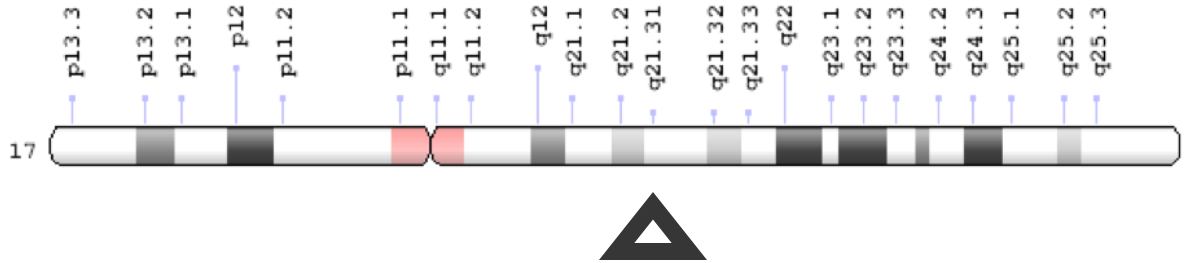
Koolen-de Vries Syndrome

17q21.31 microdeletion

Koolen-de Vries Syndrome 17q21.31 Microdeletion

Koolen-de Vries syndrome (KdVS) was first reported as a genetic disorder in 2006, with key characteristics including neonatal hypotonia (low muscle tone at infancy), intellectual disability, and distinctive facial features. In terms of the genetics of the syndrome, it includes patients with 17q21.31 microdeletions and patients with loss-of-function mutation in the KANSL1 gene. This syndrome occurs in about 1 in every 16,000 individuals.



The symptoms mentioned at left are the most common for KdVS. Other symptoms – though less common – include speech developmental delay, seizures, subtle brain abnormalities, and urogenital abnormalities. Individuals with KdVS have also been described as having friendly natures.

Intellectual disability varies among individuals, ranging from mild to severe. Speech development showed some delays in individuals, but the development of language comprehension and motor skills remains relatively intact. Most individuals gain some speech capabilities, and only about 10% of individuals are non-verbal.

About half of individuals with KdVS have seizures during their lifetime. The most common type are focal impaired awareness seizures, where the electrical activity occurs in one section of the brain, and the individual is only partially aware of its occurrence.

In terms of specifics, the distinct facial features include a prominent philtrum, bulbous nasal tip, everted lower lip and sparse eyebrows recurring in most cases. The subtle brain abnormalities mentioned above can include enlarged ventricles, corpus callosum hypoplasia/aplasia and subependymal heterotopia. Urogenital abnormalities can include hydronephrosis, vesicoureteric reflux, pyelectasis, hypospadias, and duplex renal system. Brain and urogenital abnormalities can be different among individuals with KdVS, and an individual will not display every symptom mentioned above.

Symptomatic and supportive treatments and early intervention therapies and other services are recommended as necessary.