Genetics Behind Koolen-de Vries Syndrome

What Genetic Changes Cause Koolen-de Vries Syndrome?

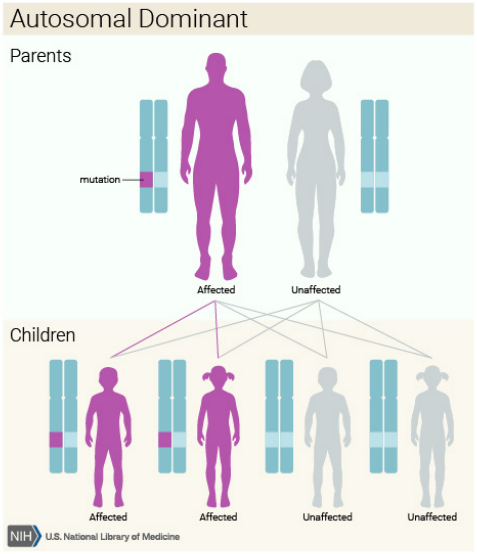
There are two types of genetic changes that can cause Koolen-de Vries Syndrome (KdVS). Chromosomes are structures in our cells that carry our genes and genetic information. People typically have 23 pairs of chromosomes, for a total of 46 chromosomes. KdVS occurs when a small portion of the 17th chromosome is missing (microdeletion) or when there is a genetic change in the *KANSL1* gene.

KANSL1 Gene Mutation:
Certain genetic mutations (variants) in the *KANSL1* gene can cause KdVS. Our genes help our bodies function by providing instructions to our cells. Our genetic material (DNA) has a specific sequence or spelling that tells our cells what proteins to make. When a genetic mutation is present, the typical spelling in our DNA is altered. These alterations change the way that the sequence is read and can lead cells to make the wrong proteins or make fewer proteins than they are supposed to.

17q21.31 Microdeletion:
When small parts of a chromosome are missing (microdeletion), cells no longer have the genes/instructions necessary to make certain proteins. KdVS can occur if the *KANSL1* gene is one of the genes on the part of the chromosome that is missing. Although the sizes of the microdeletion can vary amongst individuals with KdVS, the size of the microdeletion does not correlate with the severity of the syndrome but is rather likely due to other genetic factors.

How is Koolen-de Vries Syndrome Inherited?

Koolen-de Vries Syndrome is an autosomal dominant condition. Individuals typically have two copies of every gene. With autosomal dominant conditions, having one abnormal copy of a gene can cause the condition. This means that if an individual with KdVS has children, there is a 50% chance that their child will also inherit the genetic change for KdVS and have the syndrome.



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Although KdVS is a genetic syndrome, it is typically not a condition that is passed down or inherited. In most families, parents of an affected child do not have KdVS or a genetic change that would cause KdVS. Typically, an affected child is the first person in the family to have KdVS. Most genetic changes that cause KdVS are

de novo, or new in the affected individual. Parents did not do anything to cause the new genetic change in their child. These changes are due to random chance and errors during cell division and development.

Should Parents of a Child with Koolen-de Vries Syndrome Get Genetic Testing?

Parents of a child with KdVS may consider getting genetic testing to determine if they have the same genetic change that their child has. Some parents may also get genetic testing to help determine their chance of having another child with KdVS.

In rare cases, a parent may have a genetic change that causes KdVS in some but not all of their cells. Some individuals may have affected cells in a few different parts of their body, while others may only have affected cells in their egg or sperm cells that get passed onto their children. This is known as mosaicism. Parents who are mosaic for a genetic change that causes KdVS have an increased chance of having another child with KdVS.

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