



KANSL1 gene

KAT8 regulatory NSL complex subunit 1

Normal Function

The *KANSL1* gene provides instructions for making a member (subunit) of a group of interacting proteins called the KAT8 regulatory NSL complex. This complex is categorized as a histone acetyltransferase (HAT) complex. It helps regulate gene activity (expression) by modifying chromatin, the complex of DNA and protein that packages DNA into chromosomes.

The protein produced from the *KANSL1* gene is found in most organs and tissues of the body before birth and throughout life. By its involvement in controlling the activity of other genes, this protein plays an important role in the development and function of many parts of the body.

Health Conditions Related to Genetic Changes

Koolen-de Vries syndrome

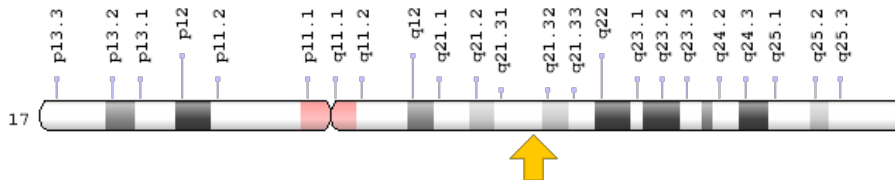
KANSL1 gene mutations or deletions of genetic material including this gene cause Koolen-de Vries syndrome. This disorder is characterized by developmental delay, intellectual disability, a cheerful and sociable disposition, and a variety of physical abnormalities.

Loss of one copy of the *KANSL1* gene in each cell impairs normal development and function of various organs and tissues of the body, but the relationship of *KANSL1* gene loss to the specific signs and symptoms of Koolen-de Vries syndrome is unclear.

Chromosomal Location

Cytogenetic Location: 17q21.31, which is the long (q) arm of chromosome 17 at position 21.31

Molecular Location: base pairs 46,027,335 to 46,225,374 on chromosome 17 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CENP-36
- centromere protein 36
- DKFZP727C091
- hMSL1v1
- KANL1_HUMAN
- KDVS
- KIAA1267
- male-specific lethal 1 homolog
- MLL1/MLL complex subunit KANSL1
- MSL1 homolog 1
- MSL1v1
- non-specific lethal 1 homolog
- NSL complex protein NSL1
- NSL1

Additional Information & Resources

Clinical Information from GeneReviews

- Koolen-de Vries Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK24676>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28KANSL1%5BTIAB%5D%29+OR+%28%28KDVS%5BTIAB%5D%29+OR+%28NSL1%5BTIAB%5D%29+OR+%28MSL1v1%5BTIAB%5D%29+OR+%28hMSL1v1%5BTIAB%5D%29+OR+%28KIAA1267%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- KAT8 REGULATORY NSL COMPLEX, SUBUNIT 1
<http://omim.org/entry/612452>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_KANSL1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=KANSL1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:24565
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:284058>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/284058>
- UniProt
<https://www.uniprot.org/uniprot/Q7Z3B3>

Sources for This Summary

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