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.

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

Tests Listed in the Genetic Testing Registry

- Tests of KANSL1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=284058[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; KANSL1 (https://omim.org/entry/612452)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/284058)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KANSL1[gene])

References

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- Koolen DA, Kramer JM, Neveling K, Nillesen WM, Moore-Barton HL, Elmslie FV, Toutain A, Amiel J, Malan V, Tsai AC, Cheung SW, Gilissen C, Verwiel ET, MartensS, Feuth T, Bongers EM, de Vries P, Scheffer H, Vissers LE, de Brouwer AP, Brunner HG, Veltman JA, Schenck A, Yntema HG, de Vries BB. Mutations in thechromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. NatGenet. 2012 Apr 29;44(6):639-41. doi: 10.1038/ng.2262. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22544363)
- Zollino M, Orteschi D, Murdolo M, Lattante S, Battaglia D, Stefanini C,Mercuri E, Chiurazzi P, Neri G, Marangi G. Mutations in KANSL1 cause the 17q21.31microdeletion syndrome phenotype. Nat Genet. 2012 Apr 29;44(6):636-8. doi:10.1038/ng.2257. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22544367)

Genomic Location

The *KANSL1* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

Last updated March 1, 2013