15q24 microdeletion

Description

15q24 microdeletion is a chromosomal change in which a small piece of chromosome 15 is deleted in each cell. The deletion occurs on the long (q) arm of the chromosome at a position designated q24.

15q24 microdeletion is associated with mild to moderate intellectual disability and delayed speech development. Other common signs and symptoms include short stature, weak muscle tone (hypotonia), and skeletal abnormalities including loose (lax) joints. Affected males may have genital abnormalities, which can include an unusually small penis (micropenis) and the opening of the urethra on the underside of the penis (hypospadias). Affected individuals also have distinctive facial features such as a high front hairline, broad eyebrows, widely set eyes (hypertelorism), outside corners of the eyes that point downward (downslanting palpebral fissures), a broad nasal bridge, a full lower lip, and a long, smooth space between the upper lip and nose (philtrum).

Frequency

This condition is very rare; only a few dozen affected individuals have been identified.

Causes

People with a 15q24 microdeletion are missing between 1.7 million and 6.1 million DNA building blocks (base pairs), also written as 1.7-6.1 megabases (Mb), at position q24 on chromosome 15. The exact size of the deletion varies, but all individuals are missing the same 1.2 Mb region. This region contains several genes that are thought to be important for normal development.

The signs and symptoms that result from a 15q24 microdeletion are probably related to the loss of one or more genes in the deleted region. However, it is unclear which missing genes contribute to the specific features of the disorder.

Learn more about the chromosome associated with 15q24 microdeletion

- chromosome 15

Inheritance

The identified cases of 15q24 microdeletion have occurred in people with no history of the condition in their family. The chromosomal change likely occurs as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development.

Other Names for This Condition

- 15q24 deletion
- 15q24 microdeletion syndrome
- Interstitial deletion of chromosome 15q24

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: SIN3A-related intellectual disability syndrome due to a point mutation (https://www.ncbi.nlm.nih.gov/gtr/conditions/C4310804/)

Genetic and Rare Diseases Information Center

- 15q24 microdeletion syndrome (https://rarediseases.info.nih.gov/diseases/12219/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%2215q24 microdeletion%22)

Catalog of Genes and Diseases from OMIM

- WITTEVEEN-KOLK SYNDROME; WITKOS (https://omim.org/entry/613406)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%2815q24%5BTIAB%5D%29+AND+%28%28deletion*%5BTIAB%5D%29+OR+%28microdeletion*%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

References

- Andrieux J, Dubourg C, Rio M, Attie-Bitach T, Delaby E, Mathieu M, Journel H, Copin H, Blondeel E, Doco-Fenzy M, Landais E, Delobel B, Odent S, Manouvrier-HanuS, Holder-Espinasse M. Genotype-phenotype correlation in four 15q24 deletedpatients identified by array-CGH. Am J Med Genet A. 2009 Dec;149A(12): 2813-9.doi: 10.1002/ajmg.a.33097. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19921647) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2874573/)
- Cushman LJ, Torres-Martinez W, Cherry AM, Manning MA, Abdul-Rahman O, AndersonCE, Punnett HH, Thurston VC, Sweeney D, Vance GH. A report of three patients withan interstitial deletion of chromosome 15q24. Am J Med Genet A. 2005 Aug15;137(1):65-71. doi: 10.1002/ajmg.a.30836. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16007617)
- El-Hattab AW, Zhang F, Maxim R, Christensen KM, Ward JC, Hines-Dowell S, Scaglia F, Lupski JR, Cheung SW. Deletion and duplication of 15q24: molecularmechanisms and potential modification by additional copy number variants. GenetMed. 2010 Sep;12(9):573-86. doi: 10.1097/GIM.0b013e3181eb9b4a. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20860070)
- Klopocki E, Graul-Neumann LM, Grieben U, Tonnies H, Ropers HH, Horn D, MundlosS, Ullmann R. A further case of the recurrent 15q24 microdeletion syndrome, detected by array CGH. Eur J Pediatr. 2008 Aug;167(8):903-8. doi:10.1007/s00431-007-0616-7. Epub 2007 Oct 12. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17932688) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2757600/)
- McInnes LA, Nakamine A, Pilorge M, Brandt T, Jimenez Gonzalez P, Fallas M, Manghi ER, Edelmann L, Glessner J, Hakonarson H, Betancur C, Buxbaum JD. Alarge-scale survey of the novel 15q24 microdeletion syndrome in autism spectrumdisorders identifies an atypical deletion that narrows the critical region. MolAutism. 2010 Mar 19;1(1):5. doi: 10.1186/2040-2392-1-5. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20678247) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2907565/)
- Sharp AJ, Selzer RR, Veltman JA, Gimelli S, Gimelli G, Striano P, Coppola A, Regan R, Price SM, Knoers NV, Eis PS, Brunner HG, Hennekam RC, Knight SJ, deVries BB, Zuffardi O, Eichler EE. Characterization of a recurrent 15q24microdeletion syndrome. Hum Mol Genet. 2007 Mar 1;16(5):567-72. doi:10.1093/hmg/ddm016. Epub 2007 Mar 14. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17360722)

Last updated September 1, 2011