DPY19L2 gene

dpy-19 like 2

Normal Function

The *DPY19L2* gene provides instructions for making a protein that is found in developing sperm cells. The DPY19L2 protein plays a role in the development of the acrosome, a cap-like structure in the head of sperm cells. The acrosome contains enzymes that break down the outer membrane of egg cells, allowing the sperm to fertilize an egg.

The developing acrosome is attached to the nucleus of the cell. The DPY19L2 protein, which is found within the membrane of the nucleus, helps attach the forming acrosome to the nuclear membrane. As the acrosome develops and the sperm cell matures, the acrosome moves to the tip of the head of the sperm, which helps the head elongate into an oval shape.

Health Conditions Related to Genetic Changes

Globozoospermia

At least 17 *DPY19L2* gene mutations have been found in men with globozoospermia, a condition characterized by abnormal sperm cells that have a round head and no acrosome. Approximately 70 percent of men with this condition have mutations in *DPY19L2*. Most of these mutations delete large regions of the gene or the whole gene. Others change single protein building blocks (amino acids) in the DPY19L2 protein. These mutations lead to a loss of functional DPY19L2 protein. Without this protein, the forming acrosome is not attached to the nucleus and is removed from the cell. As a result, sperm cells have no acrosome and the head of the sperm does not elongate. The abnormal sperm are unable to get through the outer membrane of an egg cell to fertilize it, leading to an inability to father biological children (infertility) in affected men. Researchers have described other characteristics of the abnormal sperm cells that make fertilization of an egg cell difficult, although it is not clear how changes in the *DPY19L2* gene are involved in development of these characteristics.

Other Names for This Gene

- D19L2_HUMAN
- dpy-19 like 2 (C. elegans)

- dpy-19-like 2 (C. elegans)
- FLJ32949
- probable C-mannosyltransferase DPY19L2
- protein dpy-19 homolog 2
- SPATA34
- spermatogenesis associated 34
- SPGF9

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DPY19L2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=283417[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28DPY19L2%5BTIAB%5D%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

- DPY19-LIKE 2; DPY19L2 (https://omim.org/entry/613893)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/283417)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=DPY19L2[gene])

References

- Coutton C, Abada F, Karaouzene T, Sanlaville D, Satre V, Lunardi J, Jouk PS, Arnoult C, Thierry-Mieg N, Ray PF. Fine characterisation of a recombinationhotspot at the DPY19L2 locus and resolution of the paradoxical excess ofduplications over deletions in the general population. PLoS Genet. 2013Mar;9(3):e1003363. doi: 10.1371/journal.pgen.1003363. Epub 2013 Mar 21. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23555282) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3605140/)
- Coutton C, Zouari R, Abada F, Ben Khelifa M, Merdassi G, Triki C, Escalier D,

Hesters L, Mitchell V, Levy R, Sermondade N, Boitrelle F, Vialard F, Satre V,Hennebicq S, Jouk PS, Arnoult C, Lunardi J, Ray PF. MLPA and sequence analysis ofDPY19L2 reveals point mutations causing globozoospermia. Hum Reprod. 2012Aug;27(8): 2549-58. doi: 10.1093/humrep/des160. Epub 2012 May 24. Citation on PubMed (htt ps://pubmed.ncbi.nlm.nih.gov/22627659)

- Escoffier J, Yassine S, Lee HC, Martinez G, Delaroche J, Coutton C, KaraouzeneT, Zouari R, Metzler-Guillemain C, Pernet-Gallay K, Hennebicq S, Ray PF, FissoreR, Arnoult C. Subcellular localization of phospholipase Czeta in human sperm and itsabsence in DPY19L2-deficient sperm are consistent with its role in oocyteactivation. Mol Hum Reprod. 2015 Feb;21(2):157-68. doi: 10.1093/molehr/ gau098.Epub 2014 Oct 29. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25 354701) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/article s/PMC4311148/)
- Harbuz R, Zouari R, Pierre V, Ben Khelifa M, Kharouf M, Coutton C, Merdassi G, Abada F, Escoffier J, Nikas Y, Vialard F, Koscinski I, Triki C, Sermondade N, Schweitzer T, Zhioua A, Zhioua F, Latrous H, Halouani L, Ouafi M, Makni M, JoukPS, Sele B, Hennebicq S, Satre V, Viville S, Arnoult C, Lunardi J, Ray PF. Arecurrent deletion of DPY19L2 causes infertility in man by blocking sperm headelongation and acrosome formation. Am J Hum Genet. 2011 Mar 11;88(3):351-61. doi:10.1016/j. ajhg.2011.02.007. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21397064) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3 059422/)
- Koscinski I, Elinati E, Fossard C, Redin C, Muller J, Velez de la Calle J,Schmitt F, Ben Khelifa M, Ray PF, Kilani Z, Barratt CL, Viville S. DPY19L2deletion as a major cause of globozoospermia. Am J Hum Genet. 2011 Mar11;88(3):344-50. doi: 10. 1016/j.ajhg.2011.01.018. Erratum In: Am J Hum Genet.2011 Apr 8;88(4):517. Ray, Pierre [corrected to Ray, Pierre F]. Citation on PubMed (https://pubmed.ncbi.nlm.nih. gov/21397063) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc /articles/PMC3059416/)
- Perrin A, Coat C, Nguyen MH, Talagas M, Morel F, Amice J, De Braekeleer M. Molecular cytogenetic and genetic aspects of globozoospermia: a review.Andrologia. 2013 Feb;45(1):1-9. doi: 10.1111/j.1439-0272.2012.01308.x. Epub 2012May 10. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22571172)
- Pierre V, Martinez G, Coutton C, Delaroche J, Yassine S, Novella C,Pernet-Gallay K, Hennebicq S, Ray PF, Arnoult C. Absence of Dpy19l2, a new innernuclear membrane protein, causes globozoospermia in mice by preventing theanchoring of the acrosome to the nucleus. Development. 2012 Aug;139(16):2955-65.doi: 10.1242/ dev.077982. Epub 2012 Jul 4. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/ 22764053)
- Yassine S, Escoffier J, Martinez G, Coutton C, Karaouzene T, Zouari R, RavanatJL, Metzler-Guillemain C, Lee HC, Fissore R, Hennebicq S, Ray PF, Arnoult C.Dpy19l2-deficient globozoospermic sperm display altered genome packaging and DNAdamage that compromises the initiation of embryo development. Mol Hum Reprod.2015 Feb;21(2):169-85. doi: 10.1093/molehr/gau099. Epub 2014 Oct 29. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25354700) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4311149/)

Genomic Location

The *DPY19L2* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

Last updated April 1, 2015