KHDC3L gene

KH domain containing 3 like, subcortical maternal complex member

Normal Function

The *KHDC3L* gene provides instructions for making a protein whose role is not known. The KHDC3L protein is thought to be involved in regulating gene activity (expression) through a phenomenon known as genomic imprinting. Through genomic imprinting, certain genes are turned off (inactivated) based on which parent the copy of the gene came from. For most genes, both copies of the gene (one copy inherited from each parent) are active in all cells. However, for a small subset of genes, only one of the two copies is active and the other is turned off. For some of these genes, the copy from the father is normally active, while for others, the copy from the mother is normally active.

It is likely that the KHDC3L protein has additional roles in egg cell (oocyte) and embryonic development; however, its exact functions are unclear.

Health Conditions Related to Genetic Changes

Recurrent hydatidiform mole

At least six mutations in the *KHDC3L* gene have been found to cause a pregnancy-related condition known as recurrent hydatidiform mole. A hydatidiform mole is a mass that forms early in pregnancy and is made up of cells from an abnormally developed embryo and placenta. The placenta, a structure in the uterus that normally provides nutrients to a growing fetus, is dysfunctional and appears as numerous small sacs, often described as resembling a bunch of grapes. When a hydatidiform mole develops more than once, the condition is known as recurrent hydatidiform mole. *KHDC3L* gene mutations account for recurrent hydatidiform mole in about 5 percent of women with this condition.

KHDC3L gene mutations result in the production of a protein with reduced function. As a result, oocytes do not develop normally. A pregnancy that results from an abnormal oocyte cannot develop properly, resulting in recurrent hydatidiform mole. *KHDC3L* gene mutations can also prevent proper imprinting of multiple genes that contribute to a developing embryo, leading to abnormal gene activity (expression). It is not clear if problems with imprinting also contribute to the development of a hydatidiform mole. In women with *KHDC3L* gene mutations, a hydatidiform mole will develop in every pregnancy that occurs with her egg cells.

Other Names for This Gene

- C6orf221
- ECAT1
- ES cell-associated transcript 1 protein
- HYDM2
- KH domain containing 3-like, subcortical maternal complex member
- KHDC3-like protein

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KHDC3L (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=154288[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28KHDC3L%5BTIAB%5D%29+OR+%28C6orf221%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

- KHDC3-LIKE PROTEIN, SUBCORTICAL MATERNAL COMPLEX MEMBER; KHDC3L (https://omim.org/entry/611687)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/154288)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KHDC3L[gene])

References

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Genomic Location

The *KHDC3L* gene is found on chromosome 6 (https://medlineplus.gov/genetics/chromosome/6/).

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