KLLN gene

killin, p53 regulated DNA replication inhibitor

Normal Function

The *KLLN* gene provides instructions for making a protein called killin. The activity of the *KLLN* gene is controlled by a protein called p53 (which is produced from the *TP53* gene). Little is known about the function of killin, although it is thought to trigger cells to self-destruct (undergo apoptosis) when they are damaged or no longer needed. In this way, killin helps to prevent abnormal cells from growing and dividing unchecked to form tumors. Through its role in regulating cell division, killin helps maintain the stability of a cell's genetic information. Based on these roles, killin is thought to be a tumor suppressor.

Health Conditions Related to Genetic Changes

Cowden syndrome

Some cases of Cowden syndrome and a similar condition called Cowden-like syndrome result from a change involving the *KLLN* gene. These conditions are characterized by multiple tumor-like growths called hamartomas and an increased risk of developing certain cancers. When Cowden syndrome and Cowden-like syndrome are caused by *KLLN* gene mutations, the conditions are associated with a particularly high risk of developing breast and kidney cancers.

The genetic change associated with these conditions is known as promoter hypermethylation. The promoter is a region of DNA near the gene that controls gene activity (expression). Hypermethylation occurs when too many small molecules called methyl groups are attached to the promoter region. The extra methyl groups reduce the expression of the *KLLN* gene, which means that less killin is produced. A reduced amount of killin may allow abnormal cells to survive and proliferate inappropriately, which can lead to the formation of tumors.

The promoter region of the *KLLN* gene is shared with another gene, *PTEN*. The single promoter controls the expression of both genes. However, specific hypermethylation of the *KLLN* gene promoter only affects the expression of the *KLLN* gene; people with this type of genetic change have normal expression of the *PTEN* gene. Other types of mutations in the *PTEN* gene can cause Cowden syndrome and Cowden-like syndrome.

Other Names for This Gene

- KILIN_HUMAN
- KILLIN
- killin
- killin, p53-regulated DNA replication inhibitor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KLLN (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=100144748[geneid])

Scientific Articles on PubMed

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

Catalog of Genes and Diseases from OMIM

- KILLIN; KLLN (https://omim.org/entry/612105)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/100144748)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=KLLN[gene])

References

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- Jelovac D, Park BH. PTEN promoter silencing and Cowden syndrome: the role ofepigenetic regulation of KILLIN. JAMA. 2010 Dec 22;304(24):2744-5. doi:10.1001/jama.2010.1863. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21177512)
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- Sankunny M, Eng C. Identification of nuclear export signal in KLLN suggestspotential role in proteasomal degradation in cancer cells. Oncotarget. 2020 Dec15;11(50):4625-4636. doi: 10.18632/oncotarget.27833. eCollection 2020 Dec 15. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/33400740)
- Sankunny M, Eng C. KLLN-mediated DNA damage-induced apoptosis is associatedwith regulation of p53 phosphorylation and acetylation in breast cancer cells.Cell Death Discov. 2018 Sep 11;4:31. doi: 10.1038/s41420-018-0094-x. eCollection2018. Erratum In: Cell Death Discov. 2019 Jul 10;5:116. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/30245854)

Genomic Location

The *KLLN* gene is found on chromosome 10 (https://medlineplus.gov/genetics/chromosome/10/).

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