

KLKB1 gene

kallikrein B1

Normal Function

The *KLKB1* gene provides instructions for making a protein called prekallikrein. Prekallikrein is produced in the liver and circulates in the blood. A molecule called factor XII converts prekallikrein to another protein called plasma kallikrein, and plasma kallikrein helps turn on (activate) more factor XII. Plasma kallikrein and factor XII are involved in the early stages of blood clotting as part of a process called the intrinsic coagulation pathway (also called the contact activation pathway). Blood clots protect the body after an injury by sealing off damaged blood vessels and preventing further blood loss.

The interaction between plasma kallikrein and factor XII also initiates a series of chemical reactions resulting in the release of a protein called bradykinin. Bradykinin promotes inflammation by increasing the permeability of blood vessel walls, allowing more fluids to leak into body tissues. This leakage causes the swelling that accompanies inflammation.

Health Conditions Related to Genetic Changes

Prekallikrein deficiency

At least nine *KLKB1* gene mutations have been identified in people with a blood condition called prekallikrein deficiency, which does not generally cause any health problems. The condition is usually discovered when blood tests are done for other reasons.

The *KLKB1* gene mutations that cause this condition reduce or eliminate functional plasma kallikrein in the blood of affected individuals and likely impair the intrinsic coagulation pathway. Researchers suggest that this lack (deficiency) of functional plasma kallikrein protein does not generally cause any symptoms because another process called the extrinsic coagulation pathway (also known as the tissue factor pathway) can compensate for the impaired intrinsic coagulation pathway. Either pathway can activate proteins that are needed later in the clotting process.

Other Names for This Gene

- Fletcher factor
- kallikrein B, plasma (Fletcher factor) 1
- kininogenin
- plasma kallikrein
- plasma kallikrein preproprotein
- plasma prekallikrein
- PPK

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of KLKB1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3818\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3818[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28KLKB1%5BTIAB%5D%29+OR+%28%28PPK%5BTIAB%5D%29+OR+%28plasma+kallikrein%5BTIAB%5D%29+OR+%28kininogenin%5BTIAB%5D%29+OR+%28Fletcher+factor%5BTIAB%5D%29+OR+%28plasma+prekallikrein%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+720+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- KALLIKREIN B, PLASMA, 1; KLKB1 (<https://omim.org/entry/229000>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3818>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=KLKB1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=KLKB1[gene]))

References

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Genomic Location

The *KLKB1* gene is found on chromosome 4 (<https://medlineplus.gov/genetics/chromosome/4/>).

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