

XK gene

X-linked Kx blood group antigen, Kell and VPS13A binding protein

Normal Function

The *XK* gene provides instructions for producing a protein that is found in various tissues of the body, particularly the brain, muscle, and heart. This protein is also present on the surface of red blood cells and carries a molecule known as the Kx blood group antigen. Blood group antigens are found on the surface of red blood cells and determine a number of blood types. When blood cells with unfamiliar antigens enter the bloodstream, the body's immune system recognizes the cells as foreign and may trigger an immune reaction that destroys the foreign blood cells.

The function of the XK protein is unclear; researchers believe that it might play a role in transporting substances into and out of cells. On red blood cells, the XK protein attaches to another blood group protein, the Kell protein. The function of this blood group complex is unknown.

Health Conditions Related to Genetic Changes

McLeod neuroacanthocytosis syndrome

Approximately 30 mutations in the *XK* gene have been found to cause McLeod neuroacanthocytosis syndrome. Most of these mutations lead to the production of an abnormally short, nonfunctional XK protein or cause no XK protein to be produced at all. Missing or abnormal XK protein also affects another blood group antigen, the Kell protein, causing it to be less prevalent on the surface of red blood cells. How mutations in the *XK* gene cause the misshapen red blood cells and movement problems characteristic of McLeod neuroacanthocytosis syndrome is unknown.

Other Names for This Gene

- Kell blood group precursor (McLeod phenotype)
- KX
- Kx antigen
- membrane transport protein XK
- X-linked Kx blood group (McLeod syndrome)
- X1k

- XK, Kell blood group complex subunit (McLeod syndrome)
- XK-related protein 1
- XK_HUMAN
- XKR1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of XK ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7504\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=7504[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28XK%5BTIAB%5D%29+OR+%28KX+antigen%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+2520+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- KELL BLOOD GROUP PROTEIN, MCLEOD SYNDROME-ASSOCIATED; XK (<http://omim.org/entry/314850>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/7504>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=XK\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=XK[gene]))

References

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Genomic Location

The *XK* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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