VCAN gene

versican

Normal Function

The *VCAN* gene provides instructions for making a protein called versican. Versican is a type of protein known as a proteoglycan, which means it has several sugar molecules attached to it. Versican is found in the extracellular matrix of many different tissues and organs. The extracellular matrix is the intricate lattice of proteins and other molecules that forms in the spaces between cells. Versican interacts with many proteins and molecules to facilitate the assembly of the extracellular matrix and ensure its stability. Within the eye, versican interacts with other proteins to maintain the structure and gel-like consistency of the thick clear fluid that fills the eyeball (the vitreous).

Researchers have proposed several additional functions for versican. This protein likely helps regulate cell growth and division, the attachment of cells to one another (cell adhesion), and cell movement (migration). Studies suggest that versican plays a role in forming new blood vessels (angiogenesis), wound healing, inflammation, and preventing the growth of cancerous tumors. Versican also regulates the activity of several growth factors, which control a diverse range of processes important for cell growth.

Four different versions (isoforms) of the versican protein are produced from the *VCAN* gene. These isoforms (called V0, V1, V2, and V3) vary by size and by their location within the body.

Health Conditions Related to Genetic Changes

Wagner syndrome

At least 11 mutations in the *VCAN* gene have been found to cause Wagner syndrome, a condition that leads to progressive vision loss starting in childhood or early adulthood. The *VCAN* gene mutations that cause Wagner syndrome disrupt the way the gene's instructions are used to make versican. These mutations occur in two areas of the gene called intron 7 and exon 8; mutations in these regions lead to a decrease in the production of versican isoforms V0 and V1 and an increase in the production of isoforms V2 and V3. Researchers believe that this imbalance of versican isoforms in the vitreous impairs versican's interaction with other proteins, causing the vitreous to become unstable. This lack of stability in the vitreous affects other areas of the eye and

contributes to the vision problems that occur in people with Wagner syndrome. It is unknown why *VCAN* gene mutations seem solely to affect vision.

Other Names for This Gene

- chondroitin sulfate proteoglycan 2
- CSPG2
- CSPG2_HUMAN
- glial hyaluronate-binding protein
- versican proteoglycan

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of VCAN (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1462[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28VCAN%5BTIAB%5D%29+OR+%28versican%5BTI%5D%29+OR+%28CSPG2%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+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- VERSICAN; VCAN (https://omim.org/entry/118661)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1462)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=VCAN[gene])

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Genomic Location

The *VCAN* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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