

Intervertebral disc disease

Description

Intervertebral disc disease is a common condition characterized by the breakdown (degeneration) of one or more of the discs that separate the bones of the spine (vertebrae), causing pain in the back or neck and frequently in the legs and arms. The intervertebral discs provide cushioning between vertebrae and absorb pressure put on the spine.

While the discs in the lower (lumbar) region of the spine are most often affected in intervertebral disc disease, any part of the spine can have disc degeneration. Depending on the location of the affected disc or discs, intervertebral disc disease can cause periodic or chronic pain in the back or neck. Pain is often worse when sitting, bending, twisting, or lifting objects.

Degenerated discs are prone to out-pouching (herniation); the protruding disc can press against one of the spinal nerves that run from the spinal cord to the rest of the body. This pressure causes pain, weakness, and numbness in the back and legs. Herniated discs often cause nerve pain called sciatica that travels along the sciatic nerve, which runs from the lower back down the length of each leg.

As a disc degenerates, small bony outgrowths (bone spurs) may form at the edges of the affected vertebrae. These bone spurs may pinch (compress) the spinal nerves, leading to weakness or numbness in the arms or legs. If the bone spurs compress the spinal cord, affected individuals can develop problems with walking and bladder and bowel control. Over time, a degenerating disc may break down completely and leave no space between vertebrae, which can result in impaired movement, pain, and nerve damage.

Frequency

Intervertebral disc disease is estimated to affect about 5 percent of the population in developed countries each year. Most individuals experience disc degeneration as they age; however, the severity of the degeneration and the pain associated with it varies.

Causes

Intervertebral disc disease results from a combination of genetic and environmental factors. Some of these factors have been identified, but many remain unknown.

Researchers have identified variations in several genes that may influence the risk of developing intervertebral disc disease. The most commonly associated genes provide instructions for producing proteins called collagens. Collagens are a family of proteins that strengthen and support connective tissues, such as skin, bone, cartilage, tendons, and ligaments. Collagens form a network of fibers that create structure and stability within the intervertebral discs. Specific variations in several collagen genes seem to affect the risk of developing intervertebral disc disease by impairing the ability of collagens to interact with each other, decreasing the stability of the disc and leading to its degeneration.

Normal variations in genes related to the body's immune function are also associated with an increased risk of developing intervertebral disc disease. These genes play a role in triggering an immune response when the body detects a foreign invader such as a virus. It is thought that these gene variants can lead to an immune response that results in inflammation and water loss (dehydration) of the discs, which causes their degeneration.

Variants in genes that play roles in the development and maintenance of the intervertebral discs and vertebrae have also been found to be associated with intervertebral disc disease. The associated variants can lead to disc degeneration and herniation. Researchers are working to identify and confirm other genetic changes associated with an increased risk of intervertebral disc disease.

Nongenetic factors that contribute to the risk of intervertebral disc disease are also being studied. These factors include aging, smoking, obesity, chronic inflammation, and driving for long periods of time (for example, as a long-haul trucker or taxi driver).

[Learn more about the genes associated with Intervertebral disc disease](#)

- ACAN
- COL11A1
- COL1A1
- COL9A2
- COL9A3
- IL1A
- MMP2
- VDR

Additional Information from NCBI Gene:

- ASPN
- CILP
- IGF1R
- MMP9
- THBS2

Inheritance

Intervertebral disc disease can run in families, but the inheritance pattern is usually unknown. People with a first-degree relative (such as a parent or sibling) with intervertebral disc disease have an increased risk of developing the disorder themselves. Individuals may inherit a gene variation that increases the risk of intervertebral disc disease, but do not inherit the condition itself. Not all people with intervertebral disc disease have an identified gene variation that increases the risk, and not all people with such a gene variation will develop the disorder.

Other Names for This Condition

- Discogenic disease
- Discogenic disorder
- Disorder of intervertebral disc
- IDD
- Intervertebral disc degeneration
- Intervertebral disc disorder
- Intervertebral disk degeneration

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Intervertebral disc disorder (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0158252/>)

Genetic and Rare Diseases Information Center

- Intervertebral disc disease (<https://rarediseases.info.nih.gov/diseases/8572/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Intervertebral disc disease%22](https://clinicaltrials.gov/search?cond=%22Intervertebral%20disc%20disease%22))

Catalog of Genes and Diseases from OMIM

- INTERVERTEBRAL DISC DISEASE; IDD (<https://omim.org/entry/603932>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28intervertebral+disc+disease%5BTIAB%5D%29+OR+%28intervertebral+disc+degeneration%5BTI%5D%29%29+AND+%28Intervertebral+Disc+Degeneration%5BMH%5D%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>)

References

- Feng Y, Egan B, Wang J. Genetic Factors in Intervertebral Disc Degeneration. *Genes Dis.* 2016 Sep;3(3):178-185. doi: 10.1016/j.gendis.2016.04.005. Epub 2016Apr 23. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/27617275>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5016799/>)
- Hanaei S, Abdollahzade S, Khoshnevisan A, Kepler CK, Rezaei N. Genetic aspects of intervertebral disc degeneration. *Rev Neurosci.* 2015;26(5):581-606. doi: 10.1515/revneuro-2014-0077. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/25996483>)
- Ikegawa S. The genetics of common degenerative skeletal disorders: osteoarthritis and degenerative disc disease. *Annu Rev Genomics Hum Genet.* 2013;14:245-56. doi: 10.1146/annurev-genom-091212-153427. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24003854>)
- Janeczko L, Janeczko M, Chrzanowski R, Zielinski G. The role of polymorphisms of genes encoding collagen IX and XI in lumbar disc disease. *Neurol Neurochir Pol.* 2014 Jan-Feb;48(1):60-2. doi: 10.1016/j.pjnns.2013.04.001. Epub 2014 Jan 23. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24636772>)
- Kalb S, Martirosyan NL, Kalani MY, Broc GG, Theodore N. Genetics of the degenerated intervertebral disc. *World Neurosurg.* 2012 Mar-Apr;77(3-4):491-501. doi: 10.1016/j.wneu.2011.07.014. Epub 2011 Nov 7. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/22120330>)
- Kepler CK, Ponnappan RK, Tannoury CA, Risbud MV, Anderson DG. The molecular basis of intervertebral disc degeneration. *Spine J.* 2013 Mar;13(3):318-30. doi: 10.1016/j.spinee.2012.12.003. Epub 2013 Feb 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/23537454>)
- Toktas ZO, Eksi MS, Yilmaz B, Demir MK, Ozgen S, Kilic T, Konya D. Association of collagen I, IX and vitamin D receptor gene polymorphisms with radiological severity of intervertebral disc degeneration in Southern European Ancestor. *Eur Spine J.* 2015 Nov;24(11):2432-41. doi: 10.1007/s00586-015-4206-5. Epub 2015 Sep 5. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/26342701>)
- Zhang Y, Sun Z, Liu J, Guo X. Advances in susceptibility genetics of intervertebral degenerative disc disease. *Int J Biol Sci.* 2008 Sep 2;4(5):283-90. doi: 10.7150/ijbs.4.283. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18781226>) or Free article

on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2532796/>)

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