

Ankylosing spondylitis

Description

Ankylosing spondylitis is a form of painful, ongoing joint inflammation (chronic inflammatory arthritis) that primarily affects the spine. Early symptoms of ankylosing spondylitis typically begin between the ages of 15 and 30. Most commonly, affected individuals first experience chronic back pain and stiffness. This pain worsens with rest or inactivity, and tends to be relieved with physical activity or exercise.

Pain in ankylosing spondylitis results from inflammation of the joints between the pelvic bones (the ilia) and the base of the spine (the sacrum). These joints are called sacroiliac joints, and inflammation of these joints is known as sacroiliitis. The inflammation gradually spreads to the joints between the vertebrae, eventually involving the whole spine, causing a condition called spondylitis. Over time, back movement gradually becomes limited as the bones of the spine (vertebrae) fuse together. This progressive bony fusion is called ankylosis. These fused bones are prone to fracture.

Ankylosing spondylitis can involve other joints as well, including the shoulders, hips, and, less often, the knees. As the disease progresses, it can affect the joints between the spine and ribs, restricting movement of the chest and making it difficult to breathe deeply.

Ankylosing spondylitis affects the eyes in more than 30 percent of cases, leading to episodes of eye inflammation called acute iritis. Acute iritis typically affects one eye at a time and causes eye pain and increased sensitivity to light (photophobia). Rarely, ankylosing spondylitis can also cause serious complications involving the heart, lungs, and nervous system. Six to 10 percent of people with ankylosing spondylitis have additional inflammatory disorders such as psoriasis, which affects the skin, or ulcerative colitis or Crohn's disease, which both affect the digestive tract.

Frequency

Ankylosing spondylitis is part of a group of related diseases known as spondyloarthritis. In the United States, spondyloarthritis affect 3.5 to 13 per 1,000 people. Ankylosing spondylitis appears to be more common in certain indigenous populations in North America, Europe, and Asia.

Causes

Ankylosing spondylitis is likely caused by a combination of genetic and environmental factors, most of which have not been identified. However, researchers have found variations in several genes that influence the risk of developing this disorder.

The *HLA-B* gene provides instructions for making a protein that plays an important role in the immune system. The *HLA-B* gene is part of a family of genes called the human leukocyte antigen (HLA) complex. The HLA complex helps the immune system distinguish the body's own proteins from proteins made by foreign invaders (such as viruses and bacteria). The *HLA-B* gene has many different normal variations, allowing each person's immune system to react to a wide range of foreign proteins. A normal variant of the *HLA-B* gene called *HLA-B27* significantly increases the risk of developing ankylosing spondylitis. Although some people with ankylosing spondylitis have the *HLA-B27* variant, most people with this version of the *HLA-B* gene never develop the disorder. (Conversely, this condition can occur in people without the *HLA-B27* gene variant.) It is not fully known how *HLA-B27* increases the risk of developing ankylosing spondylitis.

Variations in several additional genes, including *ERAP1*, *IL1A*, and *IL23R*, have also been associated with ankylosing spondylitis. Although many of these genes play critical roles in the immune system, it is not fully known how variations in these genes affect a person's risk of developing ankylosing spondylitis. Changes in genes that have not yet been identified also likely affect the chances of developing ankylosing spondylitis and influence the progression of the disorder. Researchers are working to identify these genes and clarify their role in ankylosing spondylitis.

[Learn more about the genes associated with Ankylosing spondylitis](#)

- CARD9
- ERAP1
- HLA-B
- IL1A
- IL23R
- STAT3

Additional Information from NCBI Gene:

- ERAP2
- IL12B
- IL17A
- IL1R1
- IL1R2
- IL27
- IL6R

- PTGER4
- TYK2

Inheritance

Although ankylosing spondylitis can occur in more than one person in a family, it is not a purely genetic disease. Multiple genetic and environmental factors likely play a part in determining the risk of developing this disorder. As a result, inheriting a genetic variation linked with ankylosing spondylitis does not mean that a person will develop the condition, even in families in which more than one family member has the disorder. For example, studies show that about 75 percent of children who inherit *HLA-B27* from a parent with ankylosing spondylitis do not develop the disorder.

Other Names for This Condition

- axial spondylarthritis
- Bechterew disease
- Marie-Struempell disease
- SpA
- Spondylarthritis ankylopoietica
- Spondylitis ankylopoietica
- spondyloarthritis
- Spondyloarthritis ankylopoietica

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Spondyloarthropathy, susceptibility to, 1 (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1862852/>)

Genetic and Rare Diseases Information Center

- Ankylosing spondylitis (<https://rarediseases.info.nih.gov/diseases/9518/index>)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Ankylosing spondylitis %22](https://clinicaltrials.gov/search?cond=%22Ankylosing+spondylitis%22))

Catalog of Genes and Diseases from OMIM

- SPONDYLOARTHROPATHY, SUSCEPTIBILITY TO, 1; SPDA1 (<https://omim.org/entry/106300>)
- SPONDYLOARTHROPATHY, SUSCEPTIBILITY TO, 2; SPDA2 (<https://omim.org/entry/183840>)
- SPONDYLOARTHROPATHY, SUSCEPTIBILITY TO, 3; SPDA3 (<https://omim.org/entry/613238>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28Spondylitis,+Ankylosing%5BMAJR%5D%29+AND+%28ankylosing+spondylitis%5BTI%5D%29+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>)

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