Osteoarthritis

Description

Osteoarthritis is a common disease of the joints that primarily occurs in older adults. This condition is characterized by the breakdown of cartilage, the tough but flexible tissue that covers the ends of the bones at the joints and allows smooth joint movements. One or more parts of the body can be affected, most often the hands, shoulders, spine, knees, or hips.

Osteoarthritis usually develops slowly, causing pain, stiffness, and restricted movement as the condition gets worse. Areas of bone no longer cushioned by cartilage rub against each other and start to break down. Further damage is caused as the body attempts to repair and rebuild these tissues. The immune system, which plays a role in healing injuries, targets these areas, and its response leads to inflammation of the joint tissues. Abnormal growths of bone (osteophytes) and other tissue can also occur, and may be visible as enlarged joints. Enlargement of the joints of the fingers is especially noticeable.

People with osteoarthritis typically experience stiffness following periods of inactivity such as upon awakening or rising from a chair; the stiffness usually improves as they move around. In some affected individuals, the condition never causes major problems. In others, severe osteoarthritis can impair mobility and the ability to perform daily tasks, affecting quality of life and increasing the risk of other health conditions such as cardiovascular disease.

Osteoarthritis is most common in middle age or late adulthood, because the cartilage at the joints naturally begins to thin as people age. However, it can occur earlier in life, especially after injuries affecting the joints such as a type of knee injury called an anterior cruciate ligament (ACL) tear. People who are overweight or whose activities are particularly stressful to the joints are also at increased risk of developing osteoarthritis.

Frequency

Osteoarthritis is a very common condition, affecting about 23 percent of adults in the United States. In middle age it affects more women than men, but by about age 70 most people of both sexes have some symptoms of the condition. Severe osteoarthritis is a major contributor to disability worldwide.

The prevalence of osteoarthritis has doubled in the United States since the 1940s, and

research indicates that longer lifespans and higher rates of obesity do not fully explain the increase. Scientists suggest that other, undetermined features of modern life are involved in the development of the condition.

Causes

Common variations that affect many genes, some of which are unidentified, contribute to the risk of developing osteoarthritis. The condition was once believed to be caused primarily by "wear and tear" damage to the joints over time. However, it is now thought to be mainly the result of the body's failed attempts to repair this damage. In healthy cartilage, there is a balance between buildup and breakdown of the tissue. This balance is lost in osteoarthritis, leading to cartilage damage and, over time, complete breakdown. Without the protection of the cartilage, bone damage occurs at the joint. In response, the body builds new bone, which leads to overgrowth and reduced mobility of the joints. In addition, the cartilage damage triggers an immune response, causing inflammation of other joint tissues and leading to further joint damage.

The majority of variations associated with osteoarthritis risk are thought to act by subtly changing the amount, timing, and location of gene activity (expression). The genes whose expression influences osteoarthritis risk are typically involved in the formation and maintenance of bone and cartilage. For example, some of these genes are involved in the development of cartilage; if the genes are not expressed in the right location, at the right time, or in the right amount due to genetic variations, the function of this tissue may be impaired and the risk of developing osteoarthritis may be increased.

In most cases, multiple genetic changes, each with a small effect, combine to increase the risk of developing the disorder. The genetic changes can also interact with environmental and lifestyle factors that are associated with osteoarthritis risk, such as obesity and activity that places excessive stress on the joints.

Learn more about the gene associated with Osteoarthritis

- COL11A1

Additional Information from NCBI Gene:

- ALDH1A2
- ASTN2
- DOT1L
- GDF5
- MCF2L
- NCOA3

Inheritance

People inherit an increased risk of developing osteoarthritis, not the condition itself. This predisposition can be passed through generations in families, but the inheritance pattern is unknown.

Other Names for This Condition

- Arthritis, degenerative
- Arthropathy
- Degenerative joint disease
- Degenerative polyarthritis
- Hypertrophic arthritis
- OA
- Osteoarthritis deformans
- Osteoarthrosis

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Osteoarthritis susceptibility 1 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C3887876/)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Osteoarthritis%22)

Catalog of Genes and Diseases from OMIM

- OSTEOARTHRITIS SUSCEPTIBILITY 2; OS2 (https://omim.org/entry/140600)
- OSTEOARTHRITIS SUSCEPTIBILITY 1; OS1 (https://omim.org/entry/165720)
- OSTEOARTHRITIS SUSCEPTIBILITY 3; OS3 (https://omim.org/entry/607850)
- OSTEOARTHRITIS SUSCEPTIBILITY 4; OS4 (https://omim.org/entry/610839)
- OSTEOARTHRITIS SUSCEPTIBILITY 5; OS5 (https://omim.org/entry/612400)
- OSTEOARTHRITIS SUSCEPTIBILITY 6; OS6 (https://omim.org/entry/612401)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Osteoarthritis%5BMAJR%5D%29+AND+%28osteoarthritis%5BTI%5D%29+AND+genetics%5Bmh%5D+AND+review%5Bpt%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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