



## Osteoarthritis

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.

## **Genetic Changes**

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.

## **Inheritance Pattern**

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

## Diagnosis & Management

### Genetic Testing

- Genetic Testing Registry: Osteoarthritis  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0029408/>
- Genetic Testing Registry: Osteoarthritis susceptibility 3  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2675609/>
- Genetic Testing Registry: Osteoarthritis susceptibility 4  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1835815/>

### Other Diagnosis and Management Resources

- Agency for Healthcare Research and Quality: Managing Osteoarthritis Pain with Medicines  
[https://ahrq-ehc-application.s3.amazonaws.com/media/pdf/osteoarthritis-pain\\_consumer.pdf](https://ahrq-ehc-application.s3.amazonaws.com/media/pdf/osteoarthritis-pain_consumer.pdf)
- American Academy of Orthopaedic Surgeons: Osteoarthritis  
<https://orthoinfo.aaos.org/en/diseases--conditions/osteoarthritis>
- American College of Rheumatology: Osteoarthritis  
<https://www.rheumatology.org/I-Am-A/Patient-Caregiver/Diseases-Conditions/Osteoarthritis>
- American Occupational Therapy Association: Living with Arthritis  
<https://www.aota.org/~media/Corporate/Files/AboutOT/consumers/Adults/Arthritis/Arthritis%20tip%20sheet.ashx>
- Centers for Disease Control and Prevention: Physical Activity for Arthritis  
<https://www.cdc.gov/arthritis/basics/physical-activity-overview.html>
- MedlinePlus Health Topic: Hip Replacement  
<https://medlineplus.gov/hipreplacement.html>
- MedlinePlus Health Topic: Knee Replacement  
<https://medlineplus.gov/kneereplacement.html>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>

- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Encyclopedia: Osteoarthritis  
<https://medlineplus.gov/ency/article/000423.htm>
- Health Topic: Hip Replacement  
<https://medlineplus.gov/hipreplacement.html>
- Health Topic: Knee Replacement  
<https://medlineplus.gov/kneereplacement.html>
- Health Topic: Osteoarthritis  
<https://medlineplus.gov/osteoarthritis.html>

### Additional NIH Resources

- National Center for Complementary and Integrative Health (NCCIH): 6 Things You Should Know About Dietary Supplements for Osteoarthritis  
<https://nccih.nih.gov/health/tips/osteoarthritis-supplements>
- National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS): Osteoarthritis  
<https://www.niams.nih.gov/health-topics/osteoarthritis>

### Educational Resources

- American Academy of Family Physicians: Osteoarthritis  
<https://familydoctor.org/condition/osteoarthritis/?adfree=true>
- American Academy of Orthopaedic Surgeons: Osteoarthritis  
<https://orthoinfo.aaos.org/en/diseases--conditions/osteoarthritis>
- American College of Rheumatology: Osteoarthritis  
<https://www.rheumatology.org/I-Am-A/Patient-Caregiver/Diseases-Conditions/Osteoarthritis>
- Disease InfoSearch: Osteoarthritis  
<http://www.diseaseinfosearch.org/Osteoarthritis/9760>
- MalaCards: osteoarthritis  
<http://www.malacards.org/card/osteoarthritis>
- Merck Manual Consumer Version: Osteoarthritis  
<https://www.merckmanuals.com/home/bone,-joint,-and-muscle-disorders/joint-disorders/osteoarthritis-oa>

## Patient Support and Advocacy Resources

- Arthritis Research UK  
<https://www.arthritisresearchuk.org/arthritis-information/conditions/osteoarthritis.aspx>
- Arthritis Society (Canada)  
[http://www.arthritis.ca/about-arthritis/arthritis-types-\(a-z\)/types/osteoarthritis](http://www.arthritis.ca/about-arthritis/arthritis-types-(a-z)/types/osteoarthritis)
- Osteoarthritis Research Society International  
<https://www.oarsi.org/>

## ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22osteoarthritis%22>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?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>

## OMIM

- OSTEOARTHRITIS SUSCEPTIBILITY 1  
<http://omim.org/entry/165720>
- OSTEOARTHRITIS SUSCEPTIBILITY 2  
<http://omim.org/entry/140600>
- OSTEOARTHRITIS SUSCEPTIBILITY 3  
<http://omim.org/entry/607850>
- OSTEOARTHRITIS SUSCEPTIBILITY 4  
<http://omim.org/entry/610839>
- OSTEOARTHRITIS SUSCEPTIBILITY 5  
<http://omim.org/entry/612400>
- OSTEOARTHRITIS SUSCEPTIBILITY 6  
<http://omim.org/entry/612401>

## Sources for This Summary

- Chapman K, Valdes AM. Genetic factors in OA pathogenesis. *Bone*. 2012 Aug;51(2):258-64. doi: 10.1016/j.bone.2011.11.026. Epub 2011 Dec 8. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22178404>
- Cross M, Smith E, Hoy D, Nolte S, Ackerman I, Fransen M, Bridgett L, Williams S, Guillemin F, Hill CL, Laslett LL, Jones G, Cicuttini F, Osborne R, Vos T, Buchbinder R, Woolf A, March L. The global burden of hip and knee osteoarthritis: estimates from the global burden of disease 2010 study. *Ann Rheum Dis*. 2014 Jul;73(7):1323-30. doi: 10.1136/annrheumdis-2013-204763. Epub 2014 Feb 19.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24553908>
- Mahjoub M, Berenbaum F, Houard X. Why subchondral bone in osteoarthritis? The importance of the cartilage bone interface in osteoarthritis. *Osteoporos Int*. 2012 Dec;23 Suppl 8:S841-6. doi: 10.1007/s00198-012-2161-0. Epub 2012 Nov 22. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23179566>
- Reynard LN, Loughlin J. The genetics and functional analysis of primary osteoarthritis susceptibility. *Expert Rev Mol Med*. 2013 Feb 18;15:e2. doi: 10.1017/erm.2013.4. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23414786>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3575889/>
- Reynard LN. Analysis of genetics and DNA methylation in osteoarthritis: What have we learnt about the disease? *Semin Cell Dev Biol*. 2017 Feb;62:57-66. doi: 10.1016/j.semcdb.2016.04.017. Epub 2016 Apr 26. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/27130636>
- Rogers EL, Reynard LN, Loughlin J. The role of inflammation-related genes in osteoarthritis. *Osteoarthritis Cartilage*. 2015 Nov;23(11):1933-8. doi: 10.1016/j.joca.2015.01.003. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26521739>
- Ruiz-Romero C, Fernández-Puente P, Calamia V, Blanco FJ. Lessons from the proteomic study of osteoarthritis. *Expert Rev Proteomics*. 2015 Aug;12(4):433-43. doi: 10.1586/14789450.2015.1065182. Epub 2015 Jul 7. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26152498>
- Shen J, Abu-Amer Y, O'Keefe RJ, McAlinden A. Inflammation and epigenetic regulation in osteoarthritis. *Connect Tissue Res*. 2017 Jan;58(1):49-63. doi: 10.1080/03008207.2016.1208655. Epub 2016 Jul 7. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/27389927>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5266560/>
- Steinberg J, Zeggini E. Functional genomics in osteoarthritis: Past, present, and future. *J Orthop Res*. 2016 Jul;34(7):1105-10. doi: 10.1002/jor.23296. Epub 2016 May 30. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/27176659>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4980743/>
- Thysen S, Luyten FP, Lories RJ. Targets, models and challenges in osteoarthritis research. *Dis Model Mech*. 2015 Jan;8(1):17-30. doi: 10.1242/dmm.016881. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25561745>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4283647/>

- Wallace IJ, Worthington S, Felson DT, Jurmain RD, Wren KT, Maijanen H, Woods RJ, Lieberman DE. Knee osteoarthritis has doubled in prevalence since the mid-20th century. *Proc Natl Acad Sci U S A*. 2017 Aug 29;114(35):9332-9336. doi: 10.1073/pnas.1703856114. Epub 2017 Aug 14.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/28808025>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5584421/>
  - Zengini E, Finan C, Wilkinson JM. The Genetic Epidemiological Landscape of Hip and Knee Osteoarthritis: Where Are We Now and Where Are We Going? *J Rheumatol*. 2016 Feb;43(2):260-6. doi: 10.3899/jrheum.150710. Epub 2015 Dec 1. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/26628593>
- 

Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/condition/osteoarthritis>

Reviewed: October 2017

Published: June 19, 2018

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services