

New osteoarthritis genes discovered

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In the largest study of its kind, nine novel genes for osteoarthritis have been discovered by scientists from the Wellcome Sanger Institute and their collaborators. Results of the study, published today (19 March) in *Nature Genetics*, could open the door to new targeted therapies for this debilitating disease in the future.

Almost nine million people in the UK suffer from [osteoarthritis](#), a

degenerative joint [disease](#) in which a person's joints become damaged, stop moving freely and become painful. Osteoarthritis is the most prevalent musculoskeletal disease and a leading cause of disability worldwide.

There is no treatment for osteoarthritis. The disease is managed with pain relief and culminates in joint replacement surgery, which has variable outcomes.

In the largest study of its kind, scientists from the Wellcome Sanger Institute and their collaborators investigated the genetics behind osteoarthritis, as well as the diseases and traits that are linked to it.

To understand more about the genetic basis of osteoarthritis, the team studied 16.5 million DNA variations from the UK Biobank resource. Following combined analysis in up to 30,727 people with osteoarthritis and nearly 300,000 people without osteoarthritis in total—the controls—scientists discovered nine new [genes](#) that were associated with osteoarthritis, a significant result for this disease.

Professor Eleftheria Zeggini, senior author from the Wellcome Sanger Institute, said: "Osteoarthritis is challenging to study because the disease can vary among people, and also between the different joints affected, for example knee, hip, hand and spine. Using data from the UK Biobank resource, we have undertaken the largest genetic study of osteoarthritis to date and uncovered nine new genes associated with the disease."

Researchers then investigated the role of the nine new genes in osteoarthritis, by studying both normal cartilage and diseased cartilage from individuals who had a [joint replacement](#).

The team looked for genes that were active in the progression of the disease by extracting the relevant cells from healthy and diseased tissue,

studying the levels of proteins in the tissue and sequencing the RNA—the messenger that carries instructions from DNA for controlling the production of proteins.

Of the nine genes associated with osteoarthritis, researchers identified five genes in particular that differed significantly in their expression in healthy and diseased tissue. The five genes present novel targets for future research into therapies.

Ms Eleni Zengini, joint first author from the University of Sheffield and Dromokaiteio Psychiatric Hospital in Athens, said: "These results are an important step towards understanding the genetic causes of osteoarthritis and take us closer to uncovering the mechanism behind the disease. Once we know that, it opens the door to developing new therapies for this debilitating disease."

The team also explored genetic correlations between osteoarthritis and obesity, bone mineral density, type 2 diabetes, and raised blood lipid levels.

Researchers applied a statistical technique known as causal inference analysis to uncover which traits and diseases cause osteoarthritis, and which do not.

Within the limits of their study, scientists discovered that type 2 diabetes and high levels of lipids in the blood do not have causal effects on osteoarthritis, but reaffirm that obesity does.

Dr Konstantinos Hatzikotoulas, joint first author from the Wellcome Sanger Institute, said: "Using genetic data, we have shown that type 2 diabetes and increased [blood lipid levels](#) do not appear to be on the causal path to osteoarthritis. We also reconfirmed that obesity is on the causal path to osteoarthritis."

Dr Natalie Carter, Head of research liaison & evaluation at Arthritis Research UK, who did not fund the study, said: "The discovery of these genes is positive news for the 8.5 million people in the UK living with osteoarthritis. People living with this debilitating condition currently have limited treatment options. Meanwhile, they can struggle to do the day-to-day things most of us take for granted, like going to work or getting dressed independently. By revealing how these genes contribute to osteoarthritis, this research could open the door for new treatments to help millions of people live the pain free life they deserve."

More information: Genome-wide analyses using UK Biobank data provide insights into the genetic architecture of osteoarthritis, *Nature Genetics* (2018). [nature.com/articles/doi:10.1038/s41588-018-0079-y](https://doi.org/10.1038/s41588-018-0079-y)

Provided by Wellcome Trust Sanger Institute

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