

# Newly discovered genetic mutation predisposes osteoporosis patients to femur fracture

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Researchers at the Hospital del Mar Medical Research Institute (IMIM) and the University of Barcelona have uncovered a genetic mutation that makes bone vulnerable to bisphosphonates, drugs used to combat osteoporosis. Instead of strengthening bone and preventing fractures, these medicines induce a critical problem that makes the femur more prone to breaks. This discovery has been published in the *New England Journal of Medicine*.

Osteoporosis causes fractures that affect up to 40 percent of [people](#) over the age of 50. Bisphosphonates are efficient and cheap, making them the first line of treatment for this condition. Nevertheless, they have been associated with atypical fracturing of the femur. "Despite the rarity of this complication and the fact that many more fractures are prevented than induced, fear of this complication has led to inhibited prescription of these drugs, especially for long-term treatment," explains study leader Dr. Adolf Díez, emeritus head of [internal medicine](#) at Hospital del Mar and a researcher in the musculoskeletal research group at the IMIM. The consequence is that the majority of people at high risk of fracture due to osteoporosis (for example, those who have already suffered fractures) do not receive treatment.

The infrequency of this problem made the researchers suspicious that a genetic predisposition makes some people more likely to present atypical fracturing. "The opportunity offered by three cases of atypical fracture

in three sisters treated with bisphosphonates over several years, gave us an opportunity to study a genetic basis that, otherwise, would have been almost impossible to detect," says Dr. Xavier Nogués, head of internal [medicine](#) at Hospital del Mar and coordinator of IMIM's musculoskeletal research group.

## An exhaustive genome study

An exhaustive study of their genome, using the whole exome sequencing technique produced a mutation common to the three sisters that could explain why they presented this unusual fracturing. The mutation damages a protein (GGPPS) that is part of a metabolic chain essential for bone health, known as the mevalonate pathway. It is believed that this mutation makes bone vulnerable to the drug, and instead of strengthening it and preventing fractures, it makes it more prone to [fractures](#).

Given this finding, broader studies are needed for patient care applications, allowing clinicians to detect people prone to this atypical fracture and who, therefore, should not receive biophosphonates. This would be the first step in confidently prescribing a [treatment](#) received by millions of people around the world.

**More information:** "GGPS1 Mutation and Atypical Femoral Fractures with Bisphosphonates" *The New England Journal of Medicine* 2017.

Provided by IMIM (Hospital del Mar Medical Research Institute)

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