Recently discovered genetic mutation could be behind some cases of severe osteoporosis

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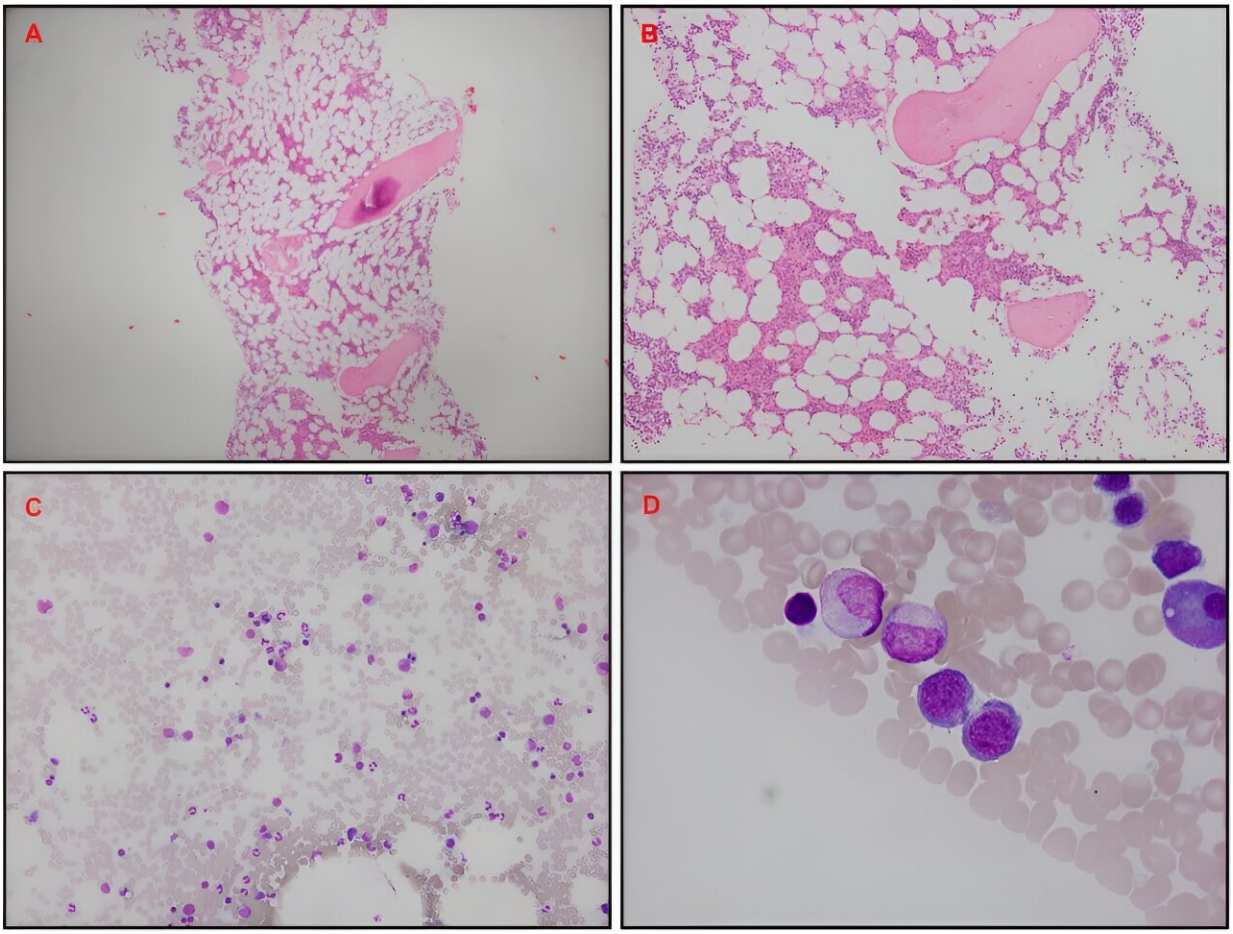

Bone marrow biopsy. (A and B) Hematoxylin and eosin trephine stain demonstrating mild to moderate hypocellular marrow (20%–30%) with moderately reduced erythropoiesis. (C and D) Aspirate demonstrating mild dyserythropoiesis with normal granulocytic and megakaryocytic lineages. Credit: *JBMR Plus* (2023). DOI: 10.1002/jbm4.10791

A recently discovered genetic mutation could be the cause of some severe and baffling cases of osteoporosis—including cases in young people.

The condition called idiopathic osteoporosis (IOP) occurs in younger adults and often involves bone fractures, even in patients with no history of physical trauma.

Head of Hudson Institute's Metabolic Bone Research Group, Associate Professor Frances Milat, says the condition is often challenging to diagnose and treat due to factors including a poor understanding of the underlying cause, a lack of management guidelines, and limited research in this area.

"IOP has been associated with abnormal bone structure. It is thought that IOP patients may have a yet undiscovered genetic mutation that is responsible for their severe osteoporosis and multiple fractures," said A/Prof Milat, who is also Deputy Director of Endocrinology at Monash Health.

"Our team of clinicians and scientists identified a novel RUNX1 genetic variant in a young male patient that may have caused changes in bone structure and severe osteoporosis. This patient had suffered from multiple fractures with little or no trauma.

"This genetic mutation may be the cause of some cases of unexplained severe osteoporosis in young adults," she said.

Finding genetic link opens care and therapy options

The research was published in the journal *JBMR Plus*. First author Dr. Tomasz Block, an Advanced Trainee in Endocrinology at Monash Health, said the successful identification of underlying genetic causes of

osteoporosis can lead to appropriate preventive care for patients, including surveillance and/or directed treatment to reduce the risk of a future fracture.

"This study gives insight into a potential genetic cause for unexplained severe osteoporosis and potential targeted therapy," he said.

"In the case described, bone formation is potentially impaired by this mutation and therefore, the optimal treatment for this patient would be medications that stimulate new bone to form."

More information: Tomasz J. Block et al, A Novel RUNX1 Genetic Variant Identified in a Young Male with Severe Osteoporosis, *JBMR Plus* (2023). DOI: 10.1002/jbm4.10791

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