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An atlas of genetic influences on osteoporosis in humans and mice

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This ground-breaking study has succeeded in compiling an atlas of genetic factors associated with estimated bone mineral density (BMD), one of the most clinically relevant factors in diagnosing osteoporosis. The paper identifies 518 genome-wide loci, of which 301 are newly discovered, that explain 20% of the genetic variance associated with osteoporosis. Having identified so many genetic factors, this research offers great promise for the development of novel targeted therapeutics to treat the disease and reduce the risk of fracture.

This was the largest study ever undertaken of the genetic determinants of osteoporosis, assessing more than 426,000 individuals in the UK Biobank. After analyzing the data, the researchers further refined their findings to isolate a set of genes that are very strongly enriched for known drug targets. This smaller set of target genes will allow drug developers to narrow their search for a solution to the clinical problem of preventing fractures in those people who are predisposed to osteoporotic fractures. Animal models have already proven the validity of some of these genes.

There are currently few treatment options for osteoporosis and many patients who are at high risk of fractures do not take existing medications because of fear of side effects. As a result, the number of people who should be treated, but are not, is high. Having found many genetic factors associated with BMD, the kind of precision medicine that genetics offers should allow researchers to hone in on those factors that can have the greatest effect on improving bone density and lessening the risk of fracture.

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