Medical Xpress

Gene responsible for severe osteoporosis disorder discovered

March 6 2011

Scientists have identified a single mutated gene that causes Hajdu-Cheney syndrome, a disorder of the bones causing progressive bone loss and osteoporosis (fragile bones). The study, published in *Nature Genetics* today, gives vital insight into possible causes of osteoporosis and highlights the gene as a potential target for treating the condition.

There are only 50 reported cases of Hajdu-Cheney syndrome (HCS), of which severe osteoporosis is a main feature. Osteoporosis is a condition leading to reduction in bone strength and susceptibility to fractures. It is the most common bone disease, with one in two women and one in five men over 50 in the UK fracturing a bone because of the condition. This represents a major public health problem yet, until this study, possible genetic causes of osteoporosis were poorly understood.

The team of scientists, led by the National Institute for Health Research (NIHR) comprehensive Biomedical Research Centre (BRC) at King's College London and Guy's and St Thomas', set out to investigate the genetic cause of HCS in order to detect clues to the role genes might play in triggering osteoporosis.

Using a cutting edge technique for identifying disease-causing genes, known as exome sequencing, the team were able to identify NOTCH2 as the causative gene using DNA from just three unrelated HCS patients. The team then confirmed their findings in an additional 12 affected families, 11 of whom had an alteration in the identical portion of the same gene.

Professor Richard Trembath, Head of King's College London's Division of Genetics and Molecular Medicine and Medicine Director of the NIHR BRC, said: "Up until now, we knew very little about the genetic mechanisms of severe bone disease. But these findings add to our understanding of the uncommon condition of HCS and provide an important basis to develop future studies in more common forms of osteoporosis, including the development of potential new therapies."

Provided by King's College London

Citation: Gene responsible for severe osteoporosis disorder discovered (2011, March 6) retrieved 5 May 2024 from
https://medicalxpress.com/news/2011-03-gene-responsible-severe-osteoporosis-disorder.html