



Genotyping takes us closer to an osteoporosis fingerprint

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For the first time ever, an extensive genome-wide search has been undertaken to find the genes linked to osteoporosis and fracture. Five regions of interest have been identified that appear to warrant further scientific investigation.

From the age of 60, 40% women and 25% men will sustain a fracture due to osteoporosis, with the risk being higher in people with a family history of fracture. There are 30,000 genes in the human genome, but until now few have been unequivocally linked to osteoporosis and fragility fractures.

The Garvan Institute for Medical Research collaborated with the Icelandic genetics company, deCode, in a project that looked at 1500 women from Garvan's Dubbo Osteoporosis Epidemiology Study as well as more than 12,000 women from Iceland and Denmark.

The results of this multi-nation study are reported in a paper appearing online today in the *New England Journal of Medicine*.

"Genome-wide genotyping, a very demanding and labour-intensive procedure, measures genetic variations called 'Snips' (SNPs or single nucleotide polymorphisms), within each of our 30,000 genes," said Garvan's Associate Professor Tuan Nguyen, who has been involved with the Dubbo project over a period of nearly 20 years. "The collaborative study examined more than 300,000 such markers and found 12 that were linked to bone mineral density and 6 linked to fragility fractures. Some of these Snips are close to genes that are already known to be associated with osteoporosis,"

Professor John Eisman, Head of Garvan's Bone Program, is very pleased with these findings. "This international study and the access to the information it brings is a positive example of the value of world-wide scientific collaborations in the area of human genetics. The study identified a number of regions in the human genome that are already known to be important in bone biology, while others are yet to be investigated," he said.

"The next step will be identifying what those genes are and how they might contribute to our understanding of osteoporosis and its prevention. This is an important example of Australian science participating in international science at the highest level."

The discovery of genes linked to osteoporosis will allow researchers to better develop prognostic models, and help clinicians identify individuals with high risk of fracture for intervention.

ABOUT GARVAN

The Garvan Institute of Medical Research was founded in 1963. Initially a research department of St Vincent's Hospital in Sydney, it is now one of Australia's largest medical research institutions with approximately 400 scientists, students and support staff. Garvan's main research programs are: Cancer, Diabetes & Obesity, Immunology and Inflammation, Bone, and Neuroscience. The Garvan's mission is to make significant contributions to medical science that will change the directions of science and medicine and have major impacts on human health. The outcome of Garvan's discoveries is the development of better methods of diagnosis, treatment, and ultimately, prevention of disease.

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