Gene find offers hope of screening test for bone disease

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Scientists have discovered three genes linked to the development of Paget's disease, a painful bone condition that affects up to one million people in the UK.

The international team of scientists, led by the University of Edinburgh, believes the genes are involved in regulating the rate at which bone is repaired, providing an explanation of why the disease might occur.

Paget's disease disrupts the body's normal process of breaking down old bone and replacing it. The condition leads to enlarged and malformed bones and patients can suffer from bone pain, brittle bones susceptible to fractures, and advanced arthritis. It affects more people in the UK than anywhere else in the world.

The scientists say that identifying the genes that predispose people to the bone disease could lead to the development of a screening test to identify those most at risk, and improve access to preventative treatment.

Researchers - funded by Arthritis Research UK and Paget Association UK - studied the genes of 1250 patients with Paget's disease to find the genes that could cause the condition.

The team - which included scientists from Spain, UK, New Zealand, and Australia - found that three genes that were faulty more frequently in patients with the bone disease than in healthy people.

Together, the faulty genes accounted for the development of Paget's disease in about 70 per cent of cases.

The results - published in the journal *Nature Genetics* - confirm that genes play a crucial role in the development of Paget's disease, which explains why many patients have a family history of the condition.

It is hoped that the discovery will allow early detection of the disease and allow doctors to give preventative treatment before bones have become damaged.

Dr Omar Albagha, who performed the study at the University of Edinburgh, said, "These findings represent a major advancement to our understanding of the disease since, until now, only one gene was known to cause about 10 per cent of cases with Paget's disease. The three genes identified from this study contribute to 70 per cent of the disease risk - quite unusual in common diseases. We are currently extending our studies to identify the genes responsible for the remaining 20 per cent of the disease risk."

Professor Stuart Ralston, Arthritis Research UK Professor of Rheumatology, who led the project at the University of Edinburgh, said: "Our work shows that these three genes together very strongly predict the development of Paget's disease. Their effects are so powerful that they could be of real value in screening for risk of the disease. This is important since we know that if treatment is left too late, then irreversible damage to the bones can occur. If we were able to intervene at an early stage with preventative therapy, guided by genetic profiling, this would be a major advance."

Provided by University of Edinburgh