**Gene for osteoporosis disorder discovered**

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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.'

**Notes to editors**

A copy of the Nature Genetics paper is available on request.

The study was part-funded by the British Heart Foundation, and Cure Kids New Zealand.

1. The comprehensive Biomedical Research Centre at Guy's and St Thomas' NHS Foundation Trust and King's College London, is one of five National Institute for Health Research (NIHR) comprehensive Biomedical Research Centres in England. With its strong focus on ‘translational research’ across seven research themes and a number of cross-cutting disciplines, it aims to take advances in basic medical research out of the laboratory and into the clinical setting to benefit patients at the earliest opportunity. Access to the uniquely diverse patient population of London and the south east enables it to drive forward research into a wide range of diseases and medical conditions.  Website: [www.biomedicalresearchcentre.org](http://www.biomedicalresearchcentre.org/)

2. Guy's and St Thomas' provides around 900,000 patient contacts in acute and specialist hospital services every year. As one of the biggest NHS Trusts in the UK, it employs almost 11,000 staff. The Trust works in partnership with the Schools of Medicine, Dentistry, Nursing and Biomedical Sciences of King's College London and other Higher Education Institutes to deliver high quality education and research. Website: [www.guysandstthomas.nhs.uk](http://www.guysandstthomas.nhs.uk/).

3. King's College London is one of the top 25 universities in the world (2010 QS international world rankings), The Sunday Times 'University of the Year 2010/11' and the fourth oldest in England. A research-led university based in the heart of London, King's has nearly 23,000 students (of whom more than 8,600 are graduate students) from nearly 140 countries, and some 5,500 employees. King's is in the second phase of a £1 billion redevelopment programme which is transforming its estate.

4. Guy’s and St Thomas’ is part of King’s Health Partners Academic Health Sciences Centre (AHSC), a pioneering collaboration between King’s College London, and Guy’s and St Thomas’, King’s College Hospital and South London and Maudsley NHS Foundation Trusts. King’s Health Partners is one of only five AHSCs in the UK and brings together an unrivalled range and depth of clinical and research expertise, spanning both physical and mental health. Our combined strengths will drive improvements in care for patients, allowing them to benefit from breakthroughs in medical science and receive leading edge treatment at the earliest possible opportunity. For more information, visit [www.kingshealthpartners.org](http://www.kingshealthpartners.org/)

5. The National Institute for Health Research (NIHR) provides the framework through which the research staff and research infrastructure of the NHS in England is positioned, maintained and managed as a national research facility. The NIHR provides the NHS with the support and infrastructure it needs to conduct first-class research funded by the Government and its partners alongside high-quality patient care, education and training. Its aim is to support outstanding individuals (both leaders and collaborators), working in world-class facilities (both NHS and university), conducting leading-edge research focused on the needs of patients.

[www.nihr.ac.uk](http://www.nihr.ac.uk/)

**For further information please contact Emma Reynolds, Press Officer, King’s College London, on 0207 848 4334 or email** [**emma.reynolds@kcl.ac.uk**](mailto:emma.reynolds@kcl.ac.uk)