Media

Contact the Press Office
Science blog
News releases for journalists
Find an expert
Filming in Oxford
For University members

11 JAN Genes associated with common type of arthritis identified

11 Jan 10

Health

Six genetic regions associated with a common type of arthritis called ankylosing spondylitis have been identified by an international consortium jointly led by Oxford University.

Six genetic regions associated with a common type of arthritis have been identified

The results of their large-scale study are published in the journal Nature Genetics.

Two genes in particular show a strong connection with the disease. They are both involved in inflammatory processes in the body, processes which can now be investigated as targets for developing better treatments for ankylosing spondylitis.

Currently little is known about the causes of ankylosing spondylitis and the best available treatment is expensive and only provides symptom relief rather than being a cure.

‘ We have identified regions of our DNA that are strongly associated with susceptibility to ankylosing spondylitis,’ says Professor Paul Wordsworth of Oxford University’s Nuffield Department of Orthopaedics, Rheumatology and Musculoskeletal Sciences, and one of the lead researchers of the study. ‘We knew there was a strong genetic component to this disease, and we now have the foundation we need for future research to pin down the genetic causes of this condition.’

Ankylosing spondylitis is a common cause of inflammatory

Further information

- Nature Genetics
- Nuffield Department of Orthopaedics, Rheumatology and Musculoskeletal Sciences
- National Ankylosing Spondylitis Society

arthritis affecting up to 200,000 people in the UK alone and is a significant cause of work-related disability. It typically first arises in young people around the age of 20. It affects the spine, resulting in progressive stiffness, loss of movement and pain as the disease develops. In the most severe cases, it can lead to fusion of the spine through new bone formation, with people becoming fixed in a bent position looking at their feet.

Treatment typically involves physiotherapy and painkillers. Potent new antibody (anti-TNF) therapies are effective in ankylosing spondylitis, but they only suppress symptoms of the disease rather than curing it and are needed life-long at a cost of around £10,000 per year.

> Cheaper or curative alternatives are a pressing need for this debilitating disease.
>
> Professor Paul Wordsworth

‘ Cheaper or curative alternatives are a pressing need for this debilitating disease,’ says Professor Wordsworth.

There is known to be a strong genetic component to ankylosing spondylitis. If a family member has the condition, your chances of having ankylosing spondylitis rise to 1 in 10. The gene HLA-B27, a gene involved in immune responses to infection, has been known for three decades to be associated with the disease, but there is convincing evidence that other genes are involved.

An international consortium of researchers, led by the University of Queensland (Professor Matthew Brown), Oxford University and the University of Texas Health Science Center at Houston, compared the genetic profiles of over 2,000 people with ankylosing spondylitis with a control group of over 5,000 people without the disease. They scanned their entire genomes for any of 300,000 genetic markers that repeatedly turned up in those with the disease and could reliably be associated with susceptibility to ankylosing spondylitis.

They found six regions of the genome that were associated with ankylosing spondylitis outside the large set of immune genes of which HLA-B27 is a member. The two strongest associations were found in genes called ERAP1 and IL23R.

ERAP1 is involved in important pathways in the body that are thought to have roles in controlling inflammation. IL23R has also been implicated in psoriasis and inflammatory bowel disease, other conditions that also involve inappropriate inflammatory responses.

‘ We know something of the inflammatory processes in the body in which these two genes are involved,’ says Professor Wordsworth of Oxford University. ‘The hope is now that we can look to develop drugs that inhibit these processes as potential new treatments for ankylosing spondylitis.’‘None of this work could have happened without the enthusiastic support of the members of the UK National Ankylosing Spondylitis Society,’ adds Professor Wordsworth.