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Breakthrough offers hope to thousands with arthritis;   
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**BYLINE:** Nina Lakhani  
  
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A genetic breakthrough into the causes of an incurable spine condition could help tens of thousands of young people avoid pain and disability.

Scientists from the University of Bristol have discovered seven **genes** involved in **ankylosing** **spondylitis** (AS), an autoimmune arthritis that causes debilitating joint pain for an estimated 200,000 Britons.

The findings pave the way for better diagnostic tests and new treatments for AS sufferers, but could prove important for other conditions, according to research in Nature Genetics.

Two of the seven genetic mutations are also involved in other common autoimmune diseases including Crohn's and coeliac disease.

AS affects one in every 200 Europeans, and is three times more common in men than women. Common symptoms, which tend to manifest themselves in the late teens or early 20s, include episodes of severe stiffness and pain in the lower back, although the pelvis, neck, hip and knees can also be affected.

Painful joints are accompanied by extreme fatigue, but the symptoms are frequently mistaken for sports injuries.

Delays in diagnosis are also caused by the fact doctors have, until now, relied upon X-rays and a complicated, expensive genetic test.

But the Wellcome Trust-funded project has uncovered a connection between two genetic mutations in AS which will make it much easier, and cheaper, to diagnose patients at the early stages of the disease.

Dr David Evans, lead author and senior lecturer in statistical genetics at the University of Bristol said:"Patients who are diagnosed earlier have a much better prognosis because they get the right drugs and physiotherapy."

Debbie Cook, director of the National **Ankylosing** **Spondylitis** Society, said the findings were a "major step forward" which would improve the understanding of a "very painful, invisible condition that mainly affects young people".

The Times (London)

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Genetic study could produce a drug for cruel back condition  
  
**BYLINE:** Mark Henderson  
  
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A set of **genes** that influence a painful back and joint disorder affecting 200,000 Britons has been identified, offering hope of new treatments.

A study of more than 5,000 people with **ankylosing** **spondylitis** has linked eight new DNA variants to the autoimmune disease, which affects up to one in 200 men and one in 500 women.

One of the **genes**, called ERAP1, points to a biological mechanism that may explain many cases of the condition.

This insight could lead to drugs that calm down the over-active immune system that causes **ankylosing** **spondylitis** and control the disease.

"As we understand better how these genetic factors operate, we may be able to use that understanding to develop new therapies," said Professor Peter Donnelly, director of the Wellcome Trust Centre for Human Genetics at the University of Oxford, who led the international study.

**Ankylosing** **spondylitis** principally affects the spine, causing stiffness, inflammation, curvature and back pain.

It can also affect other joints, tendons and ligaments, and in rarer cases the eyes, lungs, bowel and heart.

The condition generally begins between the ages of 15 and 35.

While the inflammation and pain can be managed, there is no cure.

The results of the study, published in the journal Nature Genetics, linked three genetic variants conclusively to a raised risk of **ankylosing** **spondylitis**, and found four more that were strongly associated with the disease.

The study also found an interaction between the ERAP1 **gene** and a **gene** called HLA-B27, which has been known for almost 40 years to be a significant factor in **ankylosing** **spondylitis**.

People with the HLA-B27 variant have up to 80 times the normal risk of developing it.

But those with HLA-B27 who also have a certain version of ERAP1 have a risk four times lower than normal, suggesting that ERAP1 has a powerful protective effect.

"It seems to slow down the immune system, so it can't work too hard and cause problems.

That naturally leads to the suggestion of possible drug therapies that do the same thing," Professor Donnelly said.