Alkaptonuria: New hope for treatment of rare genetic disease

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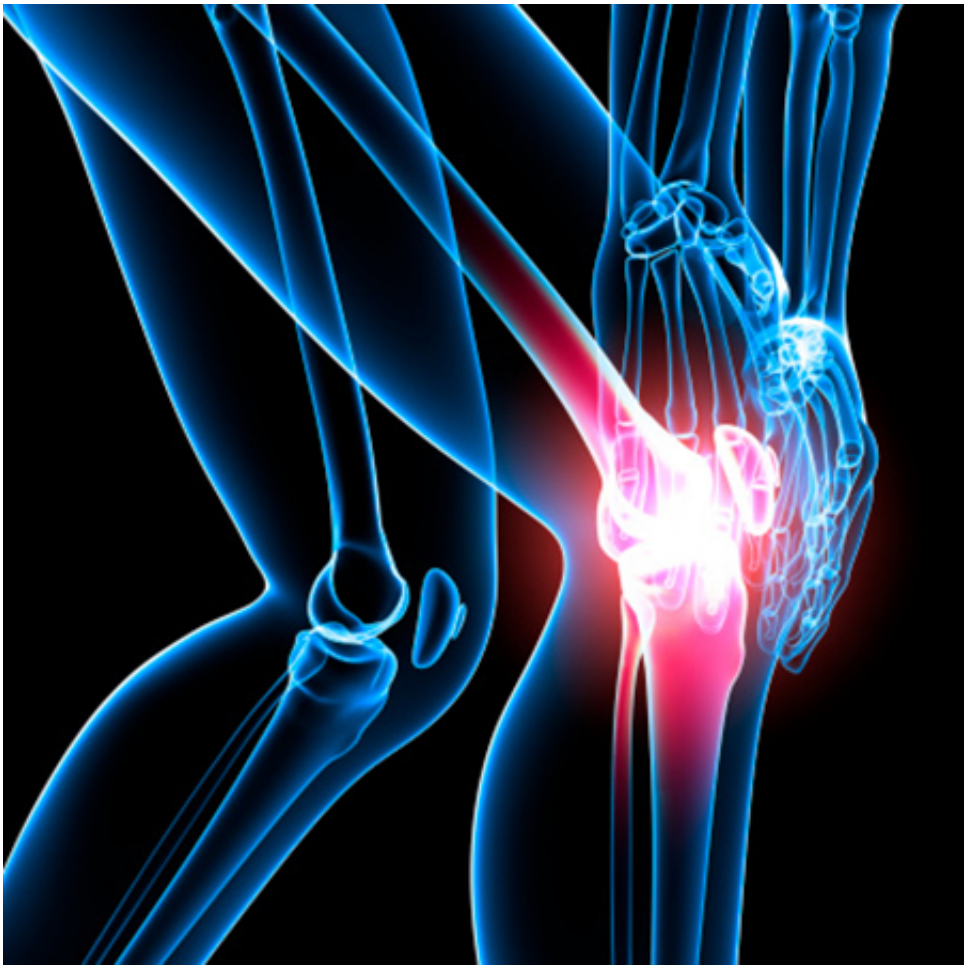

(Medical Xpress)—Scientists at the University of Liverpool have found that a drug treatment administered at the earliest signs of a rare genetic

disease could prevent the condition from developing in later life.

The disease, called alkaptonuria (AKU), was the first human disorder that was recognised as a genetic disease 100 years ago, but is often mistaken for severe arthritis. Patients with the condition do not have enough of a particular enzyme, leading to a build up of a chemical in the body, which is eliminated in the urine and deposited in the body tissue where it is toxic.

Erosion of cartilage

A black pigment, called ochronosis, forms and binds to bone, cartilage and skin, causing the eventual erosion of cartilage and the onset of osteoarthritis. As part of a new research programme, Professor Jim Gallagher, Dr Jonathan Jarvis, and Dr Lakshminarayan Ranganath, have improved animal models of how the disease works and found that signs of the condition can appear very early on in life.

The team showed that a drug treatment, called nitisinone, is effective in preventing the onset of the disease if it is used at the earliest signs of the condition, which could appear in the younger years of a patient's life. They found that if the drug is administered throughout a patient's lifespan it has the potential to completely prevent joint disease.

Professor Jim Gallagher, from the University's Institute of Ageing and Chronic Disease, said: "These findings are not only significant for patients with AKU, but could go some way to understanding the driving processes of ageing and more common degenerative diseases such as osteoarthritis.

Potential therapies

"The availability of a model that shows the early stages of joint pathology can be studied to allow us to screen other potential therapies for AKU. We have already begun work with international partners to test gene and protein replacement and we are working on the possibilities for novel anti-sense therapies."

The research is published in the journal *Annals of the Rheumatic Diseases*.

Provided by University of Liverpool