

Genetic breakthrough offers promise in tackling kidney tumors

January 10 2008

A new study has shown promising results in fighting a severe genetic disorder which can create tumours throughout the body.

Advances in genetic knowledge have transformed our understanding of disease in recent years. We now know which genes are linked to a wide variety of conditions. However, so far, very few cures and treatments have developed as a result of this knowledge.

Now a UK study led by researchers at Cardiff University suggests that a chemical first found on Easter Island could treat the genetic disorder tuberous sclerosis.

Tuberous sclerosis is an inherited disease affecting children and adults which causes tumours to grow in many of the organs of the body. Its consequences can include autism and epilepsy through its effects on the brain. A team from the Institute of Medical Genetics at Cardiff University's School of Medicine was the first to identify the genes linked with the disease – TSC1 and TSC2.

The Institute is now leading a UK-wide study on a new treatment involving the drug Sirolimus. This was first identified from a soil sample taken from Easter Island and has been shown in the past to control the activity of a protein, mTOR, which is normally controlled by the TSC1 and TSC2 genes. If the genes fail, mTOR is too active leading to tuberous sclerosis.

The UK study tested the effects of Sirolimus on kidney tumours in patients with tuberous sclerosis and a related condition, LAM, a lung and kidney disease affecting young women. The study is at the half-way stage but after one year of treatment the diameters of the tumours have shrunk by an average of 26 per cent. A parallel study in the United States has also reported similar results.

Professor Julian Sampson, director of the Institute of Medical Genetics, said: “This is a small-scale study and we will be treating patients for a further year before it is completed. However, what we have seen so far is very promising and already justifies progression to a larger study in many more patients. The findings offer new hope for what is a very serious and distressing genetic disease.”

The results of the two studies are published in the new edition of *The New England Journal of Medicine*, published on Thursday January 10.

Source: Cardiff University

Citation: Genetic breakthrough offers promise in tackling kidney tumors (2008, January 10) retrieved 6 May 2024 from

<https://medicalxpress.com/news/2008-01-genetic-breakthrough-tackling-kidney-tumors.html>

<p>This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.</p>
--