

MEDIA RELEASE

BREAKTHROUGH RESEARCH HOLDS CLUES ABOUT MS CAUSE

STRICT EMBARGO: 3AM THURSDAY 11 AUGUST 2011

In one of the largest human genetic studies ever undertaken, scientists have identified the major common genetic variants that contribute to the cause of the devastating neurologic disease, multiple sclerosis (MS).

The results of the study are published today in the prestigious scientific journal, *Nature*. They represent years of work by the International Multiple Sclerosis Genetics Consortium (IMSGC) involving more than 250 researchers in 15 countries. Australian scientists have played a significant role and more than 1000 Australians with MS contributed DNA samples.

The study confirmed the presence of up to 57 MS genes with a remarkable pattern that shows that the reason some people get MS and others don't is largely due to subtle, inherited differences in immune function. It points to a pivotal role for T cells – the 'orchestra leaders' of the immune system and makes it clear that MS is primarily an immunologic disease.

The Australian and New Zealand contribution was led by Prof Graeme Stewart, a Clinical Immunologist in the Westmead Millennium Institute, University of Sydney . It involved a consortium of 18 researchers from 5 states and New Zealand (in a group called ANZgene). Prof Stewart is one of five governance members of the IMSGC (with colleagues from Cambridge, Harvard, Yale and UCSF) and a member of the 11 person Project Direction Committee for the *Nature* study.

"Discovering so many new leads is an enormous step towards understanding the cause of MS," Professor Stewart said. "Most importantly, for people with MS, these genes also strengthen the case for immunologic treatments currently in clinical trials and point to new therapeutic approaches." A/Prof David Booth, the Australian representative on IMSGC Strategy Group, added "It is amazing that we have over 50 MS genes when as recently as 2006, we had only one. Such is the enormous power of the new research tools coupled to a massive international collaborative effort"

Previous Australian research has suggested a link between Vitamin D deficiency and an increased risk of multiple sclerosis and the ANZgene consortium identified a vitamin D gene on chromosome 12. The international study has now identified a second vitamin D gene and provides insight into a link between genetic and environmental risk factors.

Multiple Sclerosis Research Australia (MSRA), together with the Australian government, has funded MS genetic research over the past ten years.

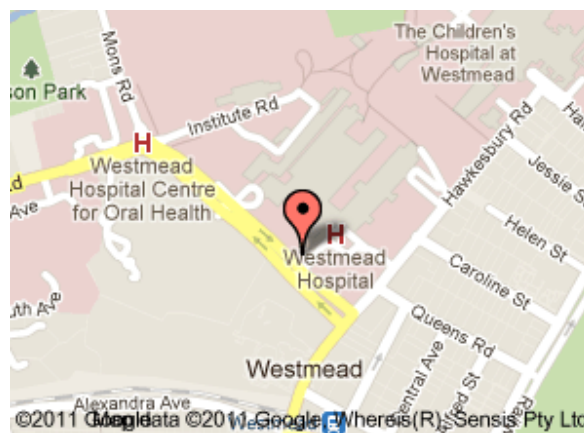
MSRA's Executive Director Jeremy Wright welcomed the breakthrough announcement. "This is a terrific milestone which brings welcome new hope to people with MS and great credit to the researchers. The Westmead Millennium Institute and ANZgene groups played a significant role in this

international effort and have put Australia in the front line of potential new findings in both the diagnosis and treatment of MS,” Mr Wright said.

The genetic testing and statistical analyses for the *Nature* paper were funded by the Wellcome Trust in the UK and the study was led from Cambridge and Oxford Universities.

Ends

MEDIA CONFERENCE: Thursday 11th August, 10:30am, Westmead Millennium Institute, Darcy Road, Westmead



Interviews available with:

- Prof Stewart, Clinical Immunologist, Westmead Millennium Institute – will outline the research and its significance
- Mr Wright, MS Research Australia Executive Director will outline the significance of this breakthrough to Australia's 20,000 MS sufferers
- Liz Taylor - person with MS, will outline what this means to her

Photo opportunities available of:

- Prof Stewart in the lab where the work was done.
- Person with MS
- Jeremy Wright, Executive Director of MS Research Australia

For more information please contact:

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BACKGROUND

About multiple sclerosis (MS)

Multiple sclerosis is one of the most common neurological conditions among young adults, affecting around 20,000 Australians and 2.5 million individuals worldwide. It is most commonly diagnosed between the ages of 20 and 40, and 75% of those diagnosed are women. The disease results from damage to nerve fibres and their protective insulation, the myelin sheath, in the brain and spinal cord. The affected pathways - responsible in health for everyday activities such as seeing, walking, feeling, thinking and controlling the bowel and bladder – are prevented from 'firing' properly and eventually are destroyed.

The findings announced today focus attention on the pivotal role of the immune system in causing the damage and help to explain the nature of the immune attack on the brain and spinal cord.

The path to discovery of the “MS genes”

The first MS gene, HLA was discovered in 1972. It took until 2007 to confirm the second MS gene, Interleukin 7 receptor (IL7R) following work by Prof Stewart's team at the Westmead Millennium Institute and at the Karolinska Institute, Stockholm.

In 2009 the ANZgene consortium reported 2 new MS genes, a Vitamin D pathway gene and CD40.

The rapid rise to 57 confirmed MS genes in 2011 results from remarkable advances in science over the past 10 years : (i) knowledge of the human genome (ii) extraordinary automated technologies allowing rapid genetic typing of thousands of DNA samples (iii) plummeting costs/test (iv) computer capacity to handle the statistical analyses and (v) national and international consortia amassing large DNA banks. These have allowed whole genome screening of thousands of patients compared to healthy controls.

The IMSGC whole genome screen involved over 27,000 people: 9,772 MS patients and 17,376 healthy controls.

The study is reminiscent of Winston Churchill's letter to President Roosevelt in Feb 1941 when he wrote "...give us the tools and we will finish the job"

There are likely to be other rare genetic variants yet to be discovered using new technologies but the remarkable pattern of immune system genes reported in the Nature paper will remain as will the insights it brings to understanding the cause of MS.

About Westmead Millennium Institute

The Westmead Millennium Institute is one of the largest medical research institutes in Australia, conducting research into a wide range of important human disorders affecting both adults and children, including MS. Because of our close affiliations with both Westmead Hospital and the University of Sydney, our research is able to extend from the laboratory to the patient.

About MSRA

MSRA is the research arm of MS Australia and facilitates MS research at Universities and medical research institutes around Australia. MSRA's objective is to accelerate research into finding the cause and a cure for MS.

This research is a key part of MSRA's goal to accelerate progress towards new treatments and a cure for MS. Our investment in MS genetics in Australia so far is over \$2.6 million thanks to contributions from John T Reid Charitable Trusts, the Trish MS Research Foundation, the Australian Research Council and the National Health & Medical Research Council.