

Vasculitis related genes cause inflammation of blood vessels

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(Medical Xpress) -- Medical Scientists at Trinity College Dublin in a pan-European collaborative study have discovered genes that contribute to the condition vasculitis, causing the inflammation of blood vessels. The findings have been recently published in the leading international publication *The New England Journal of Medicine*.

Vasculitis can affect all sizes of [blood vessels](#), including tiny vessels only visible with the aid of a [microscope](#). When a blood vessel becomes inflamed, damage to the tissue to which it supplies ensues. These conditions may be difficult to diagnose and the events that lead to vasculitis are poorly understood. Genetic factors are known to be involved and details of these genes were revealed in the international study to which the School of Medicine's Department of Immunology was a contributing centre.

The study focused on one type of vasculitis called 'granulomatosis with polyangiitis' (GPA), an auto-immune disease where cells and other components of the immune system are responsible for the inflammation. GPA is potentially a life threatening condition and its diagnosis has been revolutionised by the development of an antibody test, called the C-ANCA antibody. This test was introduced in Trinity's Department of Immunology 15 years ago through which it was established that this condition was much more common than previously known. More than 250 patients with samples analysed in the Department of Immunology laboratory have been diagnosed over this 15 year time period.

Through the use of the C-ANCA test, GPA can be diagnosed very specifically and this is a critical detail in entering patients for genetic screening studies. In this recent study, a powerful method of identifying these [genes](#) examining single nucleotide polymorphisms, was applied to a large group of patients with GPA. In the first part of the study, 1,223 patients from the UK were investigated and the findings were then confirmed and replicated in 1,454 Northern European patients, including a cohort from Department of Immunology in St James's Hospital. The patients with the C-ANCA antibody possessed a distinct immune-related gene profile which differentiated them from GPA patients with a separate type of auto-antibody, referred to as P-ANCA.

Commenting on these findings, Professor Conleth Feighery, Emeritus Professor of Immunology and Consultant Immunologist at St James's Hospital says: "These results not only identify key genetic factors contributing to this inflammatory disease but this in turn could lead to more specific therapies targeting components of the immune system."

The recently appointed Professor of Nephrology in TCD, Professor Mark Little was also a co-author of this study and has been awarded a research grant to focus on disease mechanisms involved in GPA.

The lead author of the study was Professor Kenneth GC Smith at the Cambridge Institute for Medical Research at Addenbrooke's Hospital. Other prominent centres included the University of Birmingham, the University of Groningen and Maastricht University and the Karolinska University Hospital in Sweden and many more.

Provided by Trinity College Dublin

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