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## Press information

### Identification of a gene predisposing to multiple sclerosis

Researchers at the Toulouse Purpan Hospital Physiopathology Centre (Inserm Unit 563, Université Paul Sabatier Toulouse III, Immunology and Infectious Pathologies), working in cooperation with a Swedish team, recently identified a gene that predisposes to multiple sclerosis. This discovery opens up new avenues of research into the disease and other pathologies that affect the immune system in general. The results were published in *Science Translational Medicine*.

Multiple sclerosis is a multigene disorder, implying that several genes are involved in its development. The role played by genes belonging to the Human Leucocyte Antigens (HLA) system was established over thirty years ago.

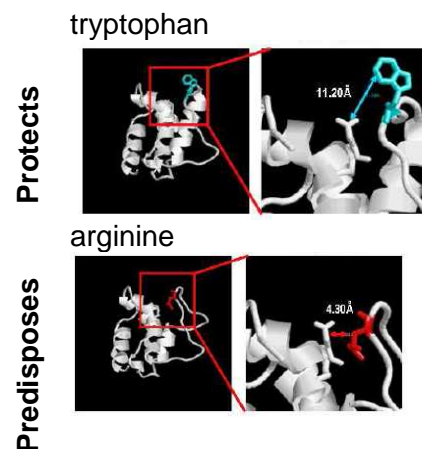
But it is only recently that the role of other genes was discovered.

A new gene that predisposes to multiple sclerosis, *VAV1*, has just been identified in rats by the teams working with Gilbert Fournié and Abdelhadi Saoudi.

The role of this gene in multiple sclerosis was initially discovered using experimental models in work conducted with a Swedish team. Through this discovery, the French and Swedish researchers demonstrated that this same gene is involved in human pathology. This was accomplished by conducting a large-scale study on over 12,000 patients from different countries, including France.

The *VAV1* gene plays a key role in the development and activation of T lymphocytes, major players in the immune system that are usually called on to fight viruses and other pathogens.

The researchers who made this discovery are now investigating the mechanisms used by this gene to influence the predisposition to multiple sclerosis. In time, these studies may lead to the development of new approaches in the treatment of multiple sclerosis and other diseases that call into play the same type of dysfunction in the immune system.



Mutation of the gene *Vav1*, discovered in experimental pathology, modifies the structure of *Vav1* protein. The variant of the coding gene for a protein with one tryptophan protects against multiple sclerosis, while the coding gene variant for a protein with one arginine implies predisposition.

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## What is multiple sclerosis?

Multiple sclerosis is an autoimmune neurological disease of the central nervous system. It is characterised by demyelination caused by immune system cells which attack the myelin sheath that insulates and protects nerve fibres. Damage to the myelin keeps electrical impulses from being conducted correctly, leading to "short circuits" that cause the various symptoms of the disease (such as motor, sensory and visual disorders).

## For further information

### Source:

### **"A role for VAV1 in experimental autoimmune encephalomyelitis and multiple sclerosis"**

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<http://stm.sciencemag.org/content/1/10/10ra21.abstract>

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