

A genetic accelerator hits the gas on autoimmune diseases

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A "genetic accelerator" is responsible for the most severe cases of Lupus (systemic lupus erythematosus), an autoimmune disease: the accelerator, called enhancer HS1.2, speeds up the activity of some critical genes of the immune system involved in the disease.

A team of Italian researchers at the Catholic University of Sacred Heart in Rome found that the enhancer HS1.2 is like the accelerator of the car and boosts the pathological immune response typical of the disease by enhancing the production of the pathological antibodies that attack the patient's body instead of defending it (autoantibodies).

Professor Gianfranco Ferraccioli, Head of the Rheumatology Unit of Rheumatology and Internal Medicine of the Catholic University led the research in collaboration with Professor Domenico Frezza at Tor Vergata University of Rome and Professor Raffaella Scorza at University of Milan and they published their results in the *Annals of the Rheumatic Diseases*.

The discovery could lead to more targeted and effective therapies against this complex disease, in particular against the most severe cases, Professor Ferraccioli explained.

[Systemic lupus erythematosus](#) is an autoimmune disease, that is a condition in which the patient's immune system goes haywire and begins to attack the body rather than defend it. [Lupus](#) affects about 60,000 people in Italy, with a major prevalence among females. Lupus affects

so several different organs and tissues and causes a variety of symptoms, including joint pain, fever, skin rashes, hair loss, Raynaud's disease, anemia, nephritis.

The therapies currently used are based on [cortisone](#), anti-malarial drugs and immunosuppressants ([azathioprine](#), mycophenolate, cyclophosphamide) and biologic drugs (rituximab, Belimumab).

But in many cases Lupus is more aggressive and so far the origin of this particular severity was quite unclear.

Italian researchers discovered that the cause of the most severe cases is the accelerator HS1.2 enhancer. Enhancers are DNA sequences that accelerate the activation of neighboring genes and enhance their functioning, hence the name.

HS1.2 leads to enhanced activation of the "transcription factor NF-KB" (a transcription factor is a molecule that "reads" the genes to make them work), which in turn dramatically increases the aggressiveness of the inflammatory processes underlying the disease.

Italian researchers have discovered that over 30 per cent of the patients has the enhancer HS1.2 in their Dna and that it causes a more severe form of Lupus.

The researchers reached this finding after demonstrating that the enhancer HS1.2 promotes also other autoimmune diseases such as rheumatoid arthritis and identified how the enhancer causes increased susceptibility to [autoimmune diseases](#).

"Our results suggest that new drugs that turn off the enhancer HS1.2, or inhibit its effect on NF-KB, can stop the disease without the need for immunosuppressive drugs or other therapies with many side effects,"

Ferraccioli said. "Moreover the discovery of the role of this enhancer allows us to better classify patients and formulate a precise prognosis for each one moving toward more personalized care."

Provided by Catholic University of Rome

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