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*STAT*mutation

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Jonathan B Weitzman

Email: jonathanweitzman@hotmail.com

Susceptibility to [mycobacterial disease](#) leads to severe clinical infections and has been associated with mutations in proteins involved in interferon-gamma signalling. In the July 13 [Science](#), Dupuis *et al.*, from the [Hôpital Necker-Enfants Malades](#) in Paris, report the identification of a mutation in the human *STAT1* gene that affects susceptibility to mycobacterial but not viral disease (*Science* 2001, **293**:300-303). The heterozygous germline mutation, causing a leucine to serine substitution (L706S), affects STAT1 phosphorylation and nuclear accumulation upon stimulation by interferons. Transfection studies showed that the *L706S* allele exerts a dominant-negative affect on the wild-type *STAT1* allele in inducing gamma-activating factor (GAF) transcriptional activity. The clinical phenotype of the *STAT1*-mutant patients suggests that anti-mycobacterial immunity involves specific STAT1-dependent interferon-gamma signalling pathways.

References

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