MedicalXpress

Genetic research increases understanding of autoimmune disease risk

May 22 2007

Geneticists have identified a link between the number of copies of a specific gene an individual has and their susceptibility to autoimmune diseases like lupus. Research using DNA has revealed that people who have a below average number of copies of a gene, known as FCGR3B, have an increased risk of developing diseases caused when the body's immune system attacks its own tissue.

The research by Professor Tim Aitman of the Medical Research Council Clinical Sciences Centre at Imperial College London, and colleagues, is published in *Nature Genetics*.

Professor Aitman explains the team's research discovery: "The variations in DNA that people carry contribute to observable characteristics like height, weight and skin colour. Genetic variations have similar effects on individual susceptibility to disease. In this research our team focused on structural differences in the genome and set out to determine whether the number of copies of a particular gene a person has influences their chances of developing an autoimmune disease. We discovered that not only does the number of copies of a gene you have influence your chances of disease but that this kind of structural variation in the genome could be driving evolution of human weaknesses for infection and inflammation."

The team studied DNA from two groups of people living in the UK and France. They discovered that people who have a comparatively low number of FCGR3B genes are more likely to suffer from autoimmune

diseases like lupus (systemic lupus erythematosus) that affect the whole body. The same link was not found to autoimmune conditions affecting just one organ such as Addisons' disease, which damages the adrenal gland, or Graves' disease, which attacks the thyroid.

Human genome research increasingly provides evidence that individuals vary in the number of copies of genes present in each of their genomes. Professor Aitman concludes: "Our discovery highlights the importance of gene copy number variation, that is differences in the number of copies of a specific gene a person carries, in genetic predisposition to common human diseases. The next step is to find out whether genes that are closely related to this susceptibility gene, FCGR3B, also vary in copy number and predispose to similar diseases."

The research team hopes to achieve these aims by studying the genomes of individual people to find out if there is any correlation between gene copy number and patterns of disease presentation or responses to specific treatments.

Source: Imperial College London

Citation: Genetic research increases understanding of autoimmune disease risk (2007, May 22) retrieved 27 April 2024 from https://medicalxpress.com/news/2007-05-genetic-autoimmune-disease.html

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