

Scientists find cause of Job's syndrome

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U.S. and Japanese scientists have identified a specific genetic mutation as the cause of a rare immunodeficiency disorder known as Job's syndrome.

In independent studies, researchers from the National Institutes of Health and the Tokyo Medical and Dental University determined the genetic mutation both overstimulates and understimulates the human immune system, leading to harmful bacterial and fungal infections and the physical features characteristic of the syndrome.

"Understanding the genetic cause of this rare immunological disorder is a signal accomplishment, revealing information that has been sought for decades," said Dr. Anthony Fauci, director of the National Institute of Allergy and Infectious Diseases. "The immunological insights from this study ... also provide new leads for treating other immunodeficiency diseases."

Only about 250 cases of Job's syndrome have been reported since its discovery in 1966. Although afflicted people often live long lives, they are subject to various infections and bone fractures, as well as spinal, facial and dental developmental difficulties. There is no cure for the disease.

The NIH team's report is available online in The New England Journal of Medicine. The Tokyo group published its findings last month in the journal Nature.

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