

Genetic mutation causes lupus in mice

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Yale researchers have identified a genetic mutation that leads to lupus in mice. The discovery could open the way for development of therapies that target the mutation. The study appears in *Cell Reports*.

Systemic Lupus Erythematosus (SLE) is an autoimmune disease that causes widespread inflammation in internal organs as well as joints and the nervous system. It affects five million people around the world. The first sign of lupus may be severe joint pain or a butterfly-shaped rash on the face. There is no cure.

The research team focused on a gene known as POL B, which functions to repair breaks in DNA. Decreased POL B expression, which results in weakened DNA repair, has been linked to SLE.

To test whether this was true, researchers constructed mouse models with mutated POL B. They exhibited decreased expression and much slower DNA synthesis. The mice subsequently developed disease characteristics that strongly resemble SLE, including dermatitis and renal disease.

"This finding implicates abnormal DNA repair as one of the causes of lupus in people," said senior author Joann Sweasy, professor of therapeutic radiology and genetics at Yale School of Medicine and member of Yale Cancer Center. "This mouse model of SLE will be useful to study how abnormal DNA repair is linked to lupus and to identify new drugs to treat this disease."

Provided by Yale University

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