Medical Xpress

Scientists discover new genetic mutation linked to eczema

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Scientists collaborating on an international research project led by Trinity College Dublin and the University of Dundee have identified a new genetic mutation linked to the development of a type of eczema known as atopic dermatitis (AD).

They found that a mutation in the gene Matt/Tmem79 led to the development of spontaneous dermatitis in mice. The gene is involved in producing a protein, now called mattrin. However, protein expression was defective in individuals with the mutant gene, and this led to skin problems. In humans, mattrin is expressed within the cells that produce and maintain the skin's function as a barrier.

After identifying the relationship between the mutation and AD, the scientists looked for a similar pattern in people. They screened large cohorts of patients that suffered from AD, comparing them with unaffected control patients, and found that the equivalent human gene MATT/TMEM79 was similarly associated. The results of this study are published in the November issue of the leading peer-reviewed journal in allergy research, *The Journal of Allergy and Clinical Immunology*.

Professor Padraic Fallon, Chair of Translational Immunology in the School of Medicine at Trinity, who led the project, said: "We have identified a new gene mutation that leads to atopic dermatitis (AD) in mice, and have taken that work further to demonstrate that a variant of the human gene is associated with AD in patients."

Professor Fallon added: "This study highlights the value of research in which genetic patterns in animals provide a starting point to investigating human disease. This strategy enables us to identify new genes that are relevant in human disease and then examine the function of these genes during inflammation. This approach will ultimately help us to understand the factors leading to inflammatory diseases and assist in the development of new therapeutic strategies."

Professor Irwin McLean, Scientific Director of the Centre for Dermatology and Genetic Medicine at the University of Dundee, jointly led the research that involved collaboration between scientists in Ireland, the United Kingdom, USA, Germany and Singapore. The work was funded by grants from the Wellcome Trust, Science Foundation Ireland and the National Children's Research Centre.

Professor McLean added: "This study shows that disruption of the barrier function of the skin is a key driving force in the development of eczema. Without an intact skin barrier, foreign substances can enter the body and trigger inflammation and allergy."

AD is the most commonly diagnosed skin condition, affecting up to 20% of children. As well as having a genetic basis, the condition can also be triggered by environmental factors, such as pet fur, pollen and house dustmites. Dairy products, eggs, nuts and wheat have also been linked to the condition.

Provided by Trinity College Dublin