Scientists identify single-gene mutations that lead to atopic dermatitis
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The scientists next set out to understand how the newly discovered CARD11 mutations contribute to atopic dermatitis. Each of the four families had a distinct mutation that affected a different region of the CARD11 protein, but all the mutations had similar effects on T-cell signaling. With cell culture and other laboratory experiments, the researchers determined that the mutations led to defective activation of two cell-signaling pathways, one of which typically is activated in part by glutamine.

Growing cultured T cells from patients with CARD11 mutations with excess glutamine boosted mTORC1 activation, a key part of one of the affected pathways, suggesting the potential to partially correct the cell-signaling defects that may contribute to atopic dermatitis. The scientists now are planning a study to assess the effect of supplemental glutamine and leucine, another amino acid that activates mTORC1, in people with atopic dermatitis with and without CARD11 mutations.

More information: Chi A Ma et al, Germline hypomorphic CARD11 mutations in severe atopic disease, Nature Genetics (2017). DOI: 10.1038/ng.3898

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