

First genetic mutations linked to atopic dermatitis identified in African-American children

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Two specific genetic variations in people of African descent are responsible for persistent atopic dermatitis (AD), an itchy, inflammatory form of the skin disorder eczema. A new report by researchers in the Perelman School of Medicine at the University of Pennsylvania found that loss-of-function mutations to Filaggrin-2 (FLG2), a gene that creates a protein responsible for retaining moisture and protecting the skin from environmental irritants, were associated with atopic dermatitis in African American children. The study, the first report to deduce the mechanism responsible for the persistent form of the condition in African American children, was published in the *Journal of Allergy and Clinical Immunology*.

Nearly half of people with [atopic dermatitis](#) in the United States are African-American [children](#). Previous studies have shown that those of African descent do not usually carry a mutation to the filaggrin gene (FLG) that has been associated with the risk of onset and persistence of AD in those of European and Asian ancestry.

"This finding helps confirm that [skin](#) barrier proteins are important in Atopic Dermatitis for people of all ancestries," said lead study author David Margolis, MD, PhD, professor of Dermatology and Epidemiology. "It could also lead to a way to determine which children are most likely to have persistent flare ups throughout their lives."

The team evaluated DNA from 299 African American children, none of whom had experienced skin free of symptoms of AD while not on medication in the previous 6 months. Within the group, researchers discovered that children with either one of two FLG2 mutations - rs12568784 or rs16833974 - were more than 50 percent more likely to have persistent AD than those without the mutations.

Future research will work to better understand [mutations](#) of FLG2 and determine if they result in functional changes to the FLG2 protein. In addition, the team is continuing research into mechanisms that may turn off the immune response to irritants that pass through the dysfunctional skin barrier and incite the inflammatory response seen in AD.

Provided by University of Pennsylvania School of Medicine

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