Molecular genetics of atopic eczema.

Abstract:
In both clinical experience and molecular genetic studies, atopic diseases (asthma, rhinoconjunctivitis and eczema) have been found to be strongly genetically influenced. The epidermal protein filaggrin seems to play a crucial role and is encoded within the so-called epidermal differentiation complex on chromosome 1. Filaggrin mutations predispose significantly to an increased risk to develop atopic eczema. Apart from filaggrin, other proteins involved in skin barrier functions such as lipid synthesis and metabolism, protease or protease inhibitor functions also seem to play a role. Besides skin barrier function, immune deviation versus a Th2 dominance and increased IgE production is also genetically determined. Polymorphisms have been found in genes encoding IL-4, IL-13 and STAT-6, and recently a polymorphism on the high-affinity IgE receptor gene has been found. Using genome-wide association studies, new genes with yet unknown functions have been determined to be associated with atopy and atopic eczema.
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