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Joint press release of the University Medical Center Schleswig-Holstein (UKSH), Campus Kiel, of Kiel University (CAU), the Cluster of Excellence Inflammation at Interfaces, the Max Delbrück Center for Molecular Medicine (MDC) Berlin-Buch and the Experimental and Clinical Research Center (ECRC) of the Charité and the MDC

Scientists from Kiel and Berlin Identify New Genetic Risk Loci for Atopic Dermatitis

In collaboration with researchers from England, Ireland and Switzerland as well as the U.S., Japan and China, scientists in Kiel and Berlin have identified variants in four gene regions which strongly increase the risk for atopic dermatitis. The results of the study conducted by the Department of Dermatology, Venerology and Allergology, the Institute of Clinical Molecular Biology (IKMB) of the University Medical Center Schleswig-Holstein (UKSH), Campus Kiel, Kiel University (CAU), the Cluster of Excellence Inflammation at Interfaces, the Max Delbrück Center for Molecular Medicine (MDC) Berlin-Buch, and the Department of Pediatric Allergology of the Experimental and Clinical Research Center (ECRC) of the Charité and the MDC have now been published in the journal *Nature Genetics* (<http://www.nature.com/ng/journal/vaop/ncurrent/full/ng.2642.html>).

With a disease risk of 15 percent, atopic dermatitis is one of the most common chronic skin diseases. In 60 percent of all those affected, the disease appears in early childhood, and some of the patients suffer from the disease during their entire lives. Often the patients go on to develop hay fever and asthma. Atopic dermatitis is also associated with a number of other diseases. A large part of the risk of developing atopic dermatitis and allergies is heritable. In combination with environmental influences this leads to the development of disease.

Under the direction of Professor Stephan Weidinger, Professor Andre Franke, Cluster of Excellence Inflammation at Interfaces (Department of Dermatology and the Institute of Clinical Molecular Biology, UKSH/CAU), and Professor Young-Ae Lee of the Max Delbrück Center for Molecular Medicine (MDC) and the Charité in Berlin, and with funding of the National Genome Research Network (NGFN), the so far world's largest genetic study on atopic dermatitis was conducted. In the study, researchers compared the DNA profiles of almost 10,000 patients with those of more than 20,000 healthy individuals, focusing particularly on those gene loci which are already known to play a role in inflammatory diseases.

The researchers identified four new gene loci in which individuals suffering from the disease significantly differ from healthy individuals. These loci contain the blueprints for proteins which are especially active in immune cells, as could be shown in the framework of these studies. So far, 16 risk loci were known for atopic dermatitis, in particular the filaggrin gene, in which mutations lead to dry skin and an impaired skin barrier.

“In this study we have identified four additional gene loci and confirmed two further loci for patients from Europe and North America, which were initially only known for patients from Asia. We could thus significantly increase the number of known risk loci. Gradually, our understanding of the genetic basis of allergic disease is becoming more complete“, said Dr. David Ellinghaus of the

Institute of Clinical Molecular Biology of Kiel University, who conducted large parts of the study. The results of the current study show that patients with atopic dermatitis not only exhibit inherited disturbances of the skin barrier function, but also abnormalities of the immune system. “These findings will accelerate the development of targeted treatment approaches,” said Professor Weidinger and Professor Lee.

Link to the publication Ellinghaus *et al.* in Nature Genetics:

“High density genotyping study identifies four new susceptibility loci for atopic dermatitis”
<http://www.nature.com/ng/journal/vaop/ncurrent/full/ng.2642.html>

National Genome Research Network (NGFN):

The Federal Ministry of Education and Research (BMBF) has been funding the study of inflammatory diseases since 2001 in the National Genome Research Network. Since 2008 the funding has been continued in the program of Medical Genome Research within the scope of NGFN-Plus. The studies presented here have been performed in the Integrated Genome Network “Environment-Related Diseases”.

www.ngfn.de/en/start.html

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