

Lavinia Paternoster, et al. Nature Genetics



# STUDY SUMMARY

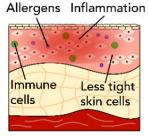
Discovery of 10 new genomic regions associated with atopic dermatitis, also known as eczema.

## YOUR RESULT

## STUDY DESCRIPTION



The skin is the largest organ in the body, serving as a barrier to the outside world. The immune system helps support this barrier function, attacking "foreign" substances that come in contact with the skin. When this happens, the skin can become inflamed, red, and itchy. Atopic dermatitis, also known as eozema, is a skin inflammation triggered by environmental factors that are actually not harmful to the body. Among many others, these factors can include temperature, soap, and clothing. Atopic dermatitis is highly heritable, with genetics determining up to 90% of an individual's susceptibility to the condition. To identify genetic variants associated with atopic dermatitis, this genome-wide association study examined over 116,000 individuals of European, African, Japanese, and Latino ancestries. The study found 21 regions of the genome associated with atopic dermatitis, 10 of which are



Atopic dermatitis is caused by immune cells in the skin that respond to contact with allergens.

novel. Altogether, these variants may explain around 15% of the variance in susceptibility to atopic dermatitis. The newly-identified genomic regions include genes involved in the immune system, particularly the function of <u>I cells</u> that play an important role in many autoimmune diseases.

## DID YOU KNOW?

Atopic dermatitis can be a persistent condition, with flare-ups occurring periodically. To help reduce the severity and frequency of symptoms, good skin care is critical. This includes regular application of a moisturizer, using a "scent-free" laundry detergent, and avoiding scratching of itchy skin.

#### YOUR DETAILED RESULTS

To calculate your genetic predisposition to atopic dermatitis we summed up the effects of genetic variants that were linked to atopic dermatitis in the study that this report is based on. These variants can be found in the table below. The variants highlighted in green have positive effect sizes and increase your genetic predisposition to atopic dermatitis. The variants highlighted in blue have negative effects sizes and decrease your genetic predisposition to atopic dermatitis. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to atopic dermatitis. By adding up the effect sizes of the highlighted variants we calculated your polygenic score for atopic dermatitis to be 0.34. To determine whether your score is high or low, we compared it to the scores of 5,000 other Nebula Genomics users. We found that your polygenic score for atopic dermatitis is in the 81st percentile. This means that it is higher than the polygenic scores 81% of people. We consider this to be an above average genetic predisposition to atopic dermatitis. However, please note that genetic predispositions do not account for important non-genetic factors like lifestyle. Furthermore, the genetics of most traits has not been fully understood yet and many associations between traits and genetic variants remain unknown. For additional explanations, click on the column titles in the table below and visit our Nebula Library tutorial.

VARIANT <sup>©</sup>	YOUR GENOTYPE <sup>©</sup>	EFFECT SIZE <sup>®</sup>	VARIANT FREQUENCY <sup>™</sup>	SIGNIFICANCE <sup>®</sup>
rs61813875_G	NA	0.48 (-)	2%	5.60 × 10 <sup>-29</sup>
rs10791824_G	A / G	0.11 (1)	57%	2.10 x 10 <sup>-19</sup>
rs12188917_C	T/C	0.13 (1)	21%	4.00 x 10 <sup>-17</sup>
rs6419573_T	C / C	0.10 (-)	26%	1.50 × 10 <sup>-13</sup>
rs2212434_T	C / C	0.09 (-)	45%	$4.60 \times 10^{-13}$
rs4809219_C	A / A	-0.11 (-)	27%	7.00 x 10 <sup>-13</sup>
rs2918307_G	A / A	0.11 (-)	16%	$4.60 \times 10^{-12}$
rs2041733_C	T/T	-0.08 (-)	55%	2.50 x 10 <sup>-11</sup>
rs12730935_A	A / A	0.08 (1)	39%	6.10 x 10 <sup>-11</sup>
rs2038255_T 🌞	C/C	0.10 (-)	18%	$1.80 \times 10^{-10}$
rs7127307_C 🌼	T/T	-0.07 (-)	47%	3.90 × 10 <sup>-10</sup>
rs7512552_T 🌼	C/C	-0.07 (-)	49%	9.10 × 10 <sup>-10</sup>
rs6473227_A 🌼	C / A	-0.07 (↓)	61%	1.40 x 10 <sup>-9</sup>
rs6602364_G 🖐	C/C	0.08 (-)	45%	1.50 × 10 <sup>-9</sup>
rs4713555_T	G / T	-0.09 (↓)	27%	5.40 x 10 <sup>-9</sup>
rs10214237_C 💮	T/C	-0.07 (↓)	27%	2.90 x 10 <sup>-8</sup>
rs10199605_A 🖐	G / A	-0.07 (↓)	30%	3.40 x 10 <sup>-8</sup>
rs4643526_A	G / G	0.09 (-)	19%	3.50 x 10 <sup>-8</sup>
rs12951971_G	T / G	0.12 (1)	9%	4.10 x 10 <sup>-8</sup>
rs7625909_T 💮	T/T	0.07 (↑)	32%	4.90 × 10 <sup>-8</sup>

N/A indicates variants that could not be imputed using the 1000 genomes project datasets and variants that have a frequency of < 5%. Your genome was sequenced at 30x/100x coverage and is not imputed. However, to calculate percentiles, we need to compare your data with other users imputed data. To make the data comparable, we need to exclude some of the variants from your data.