

Supplementary methods

Statistical Analysis

Using logistic regression analysis, crude odds ratios (ORs), and 95% confidence intervals (CIs), associations between atopic dermatitis and the SNPs under investigation were calculated. Associations between *COL6A6* polymorphisms and AD were assessed using Fisher's exact test, as this test is most commonly used for this type of case-control study.

Supplementary data

Table S1. Clinical characteristics of the subjects

	Healthy controls	Early onset-AD
Total, <i>n</i>	61	112
Age, years, mean±SD	0.9 ± 0.7	1.8 ± 0.8
Male %	54.1%	63.4%
Total IgE, KU/L	-	159.5 ± 294.6
Eosinophils, mm ³	-	382.0 ± 422.9

Table S2. Odds ratios (ORs) and 95% confidence intervals (CIs) for atopic dermatitis associated with *COL6A6* polymorphisms in Korean patients.

SNP	Allele		Genotype (Dominant)				Genotype (Recessive)					
	Minor allele	Major allele	OR (95% CI)	<i>p</i> -value	Case	Control	OR (95% CI)	<i>p</i> -value	Case	Control	OR (95% CI)	<i>p</i> -value
rs16830494	A	G	0.7971 (0.4465 to 1.4231)	0.4431	AA+GA	GG	0.7181 (0.3694 to 1.3958)	0.3288	AA	GA+GG	1.6667 (0.1696 to 16.3780)	0.6613
rs59021909	T	C	0.9691(0.4837 to 1.9417)	0.9295	TT+TC	CC	0.8676 (0.4088 to 1.8413)	0.7114	TT	TC+CC	2.7828 (0.1315 to 58.9004)	0.5111
rs200963433	T	C	2.2000 (0.2431 to 19.9057)	0.4829	TT+TC	CC	2.2222 (0.2428 to 20.3374)	0.4796	TT	TC+CC	0.5020 (0.0098 to 25.6072)	0.7312

Table S3. Fliaggrin variants detected in three families

AD-associated variants				Functional prediction program									
Gene	#RS	Chr	POS	Amino acid change	Type	Family	SIFT	Polyphen2	PhyloP	PhastCons	Global	East Asian	Korean
<i>FLG</i>	.	chr1	152,277,578	A3262T	cSNP	A	1	0.995	0.628	0.009	.	.	.
<i>FLG</i>	80088153	chr1	152,276,803	Q3520P	cSNP	B	0.32	0.996	-0.086	.	0.01	0.05	.
<i>FLG2</i>	2282302	chr1	152,329,369	C298S	cSNP	C	0.97	0.965	-0.065	0.999	0.17	0.21	0.218

Figure S1. Pedigree diagrams of three families with atopic dermatitis. Squares indicate males, and circles indicate females. Solid symbols represent affected individuals with AD. Unshaded symbols represent unaffected individuals.

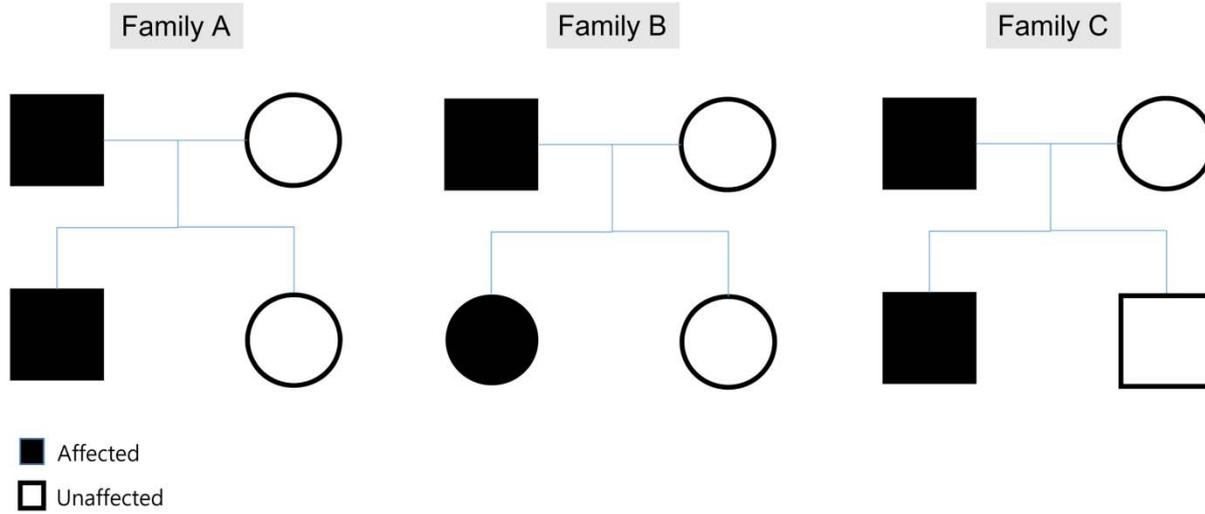


Figure S2. Flow chart of the process for filtering variants.

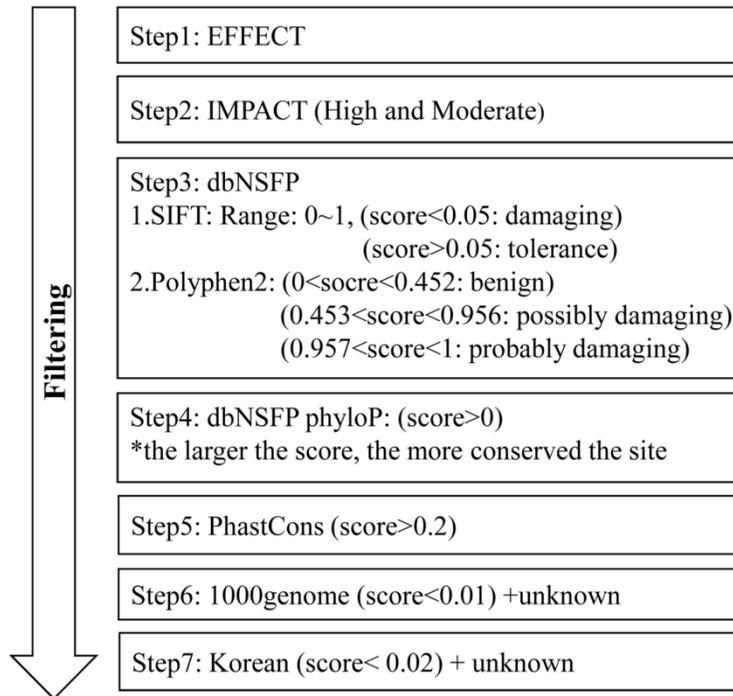


Figure S3. A Venn diagram showing the number of overlapping genes. A Venn diagram depicting the number of overlapping genes with common and rare variants among the three families.

