



POSTER PRESENTATION

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Association between filaggrin family member genes, asthma, atopy and atopic asthma with atopic dermatitis history in the subjects from the Saguenay-Lac-Saint-Jean founder population

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Objective/purpose

To perform an association study between the filaggrin (*FLG*) and the filaggrin family member 2 (*FLG2*) tagging single nucleotide polymorphisms (tagSNPs) and asthma, atopy, atopic asthma, as well as these affections in the presence of atopic dermatitis (AD).

Methods

Five tagSNPs covering *FLG* have been genotyped in 237 trios from the Saguenay-Lac-Saint-Jean population using a Sequenom panel. In addition, a genome-wide association study (GWAS) has also been done for the same trios in the large-scale GABRIEL project <http://www.gabriel-fp6.org/>.

The polymorphisms (SNPs) included in *FLG* and *FLG2* as well as those in the 3' and 5' UTR regions were extracted. Six SNPs were extracted for *FLG* (for a total of 11 SNPs when including the Sequenom panel) and 2 SNPs for *FLG2*. The association study for all the affections was done using a family-based association test (FBAT). The results were corrected using the Li and Ji method [1].

Findings

Positive associations were found between a haplotype block formed by *FLG* rs2184951 and rs12730241 (H1) and asthma and related phenotypes (see results in Table 1).

Table 1 Association of *FLG* haplotype and tagSNPs with asthma and atopy (A) and with asthma and atopy that co-occur with the presence of a personal history of atopic dermatitis (B)

A										
SNP	Allele	Asthma			Atopy			Atopic asthma		
		S:E(S)	Z	p	S:E(S)	Z	p	S:E(S)	Z	p
rs3126085	A	32.0:50.2	-3.48	0.0005	35.0:49.8	-2.94	0.0033	25.0:37.2	-2.74	0.0061
	G	148.0:129.8	3.48		141.0:126.2	2.94		109.0:96.8	2.74	
B										
SNP/Haplotype	Allele	Asthma and AD			Atopy and AD			Atopic asthma and AD		
		S:E(S)	Z	p	S:E(S)	Z	P	S:E(S)	Z	p
rs3126085	A	14.0:24.0	-2.94	0.0033	16.0:24.5	-2.59	0.0097	10.0:18.0	-2.74	0.0062
	G	66.0:56.0	2.94		62.0:53.5	2.59		52.0:44.0	2.74	
H1	T G	75.0:66.0	2.61	0.009	75.0:64.0	3.33	0.0009	60.0:51.0	2.94	0.0033

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Deliverables

To conclude, *FLG* and *FLG2* are genes associated with asthma. Functional studies will be necessary to document the molecular structure (sequence) and role of these genes in asthma and the impact of the genetic variants.

Relevance

Identification of associated genes is fundamental to document the molecular nature of asthma in order to increase knowledge of the pathophysiology of this complex trait.

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Reference

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