Supplementary material

Single Nucleotide Variants Associated With Polygenic Hypercholesterolemia in Families Diagnosed Clinically With Familial Hypercholesterolemia

Table 1 of the supplementary material

Allele Frequency of Genetic Variants in NFHGH Blood Family Members Comparing Participants with LDL-C < 90th Percentile and Participants with LDL-C ≥ 90th Percentile.

Gene	SNV	Nucleotide change	Risk allele	Allelic risk frequency		*P*
				LDL-C ≥ 90th percentile (n = 129)	LDL-C < 90th percentile (n = 139)	
SORT1	rs629301	c.*1635G>T	T	0.833	0.828	.876
APOB	rs1367117	c.293G>A	A	0.323	0.316	.870
ABCG8	rs6544713	c.322+431T>C	C	0.405	0.371	.418
LDLR	rs6511720	c.321+711 G>T	G	0.855	0.815	.212
APOE	rs429358	c.388T>C	C	0.136	0.137	.9912
	rs7412	c.526C>T	C	0.035	0.061	.157
Gene score		Median cholesterol gene score				*P*

	LDL-C ≥ 90th percentile (N = 129)	LDL-C < 90th percentile (N = 139)	
	40.2	37.4	.048

A, adenine; C, cytosine; G, guanine; LDL-C, low-density lipoprotein cholesterol; NFHGH, nonfamilial hypercholesterolemia genetic hypercholesterolemia; SNV, single nucleotide variant; T, thymine.

The *P* vale was calculated by the Mann-Whitney *U* or chi-square tests, by comparing mutant vs wild-type allelic frequencies, as appropriate.

Table 2 of the supplementary material

Linear Regression Analysis of Clinical, Biochemical and Single Nucleotide Variants with Low-density Lipoprotein Concentration in NFHGH Blood Family Members

Variable	B Coefficient	95%CI	*P*	Corrected R^2
Age	0.544	1.434-2.259	< .001	0.278
Waist circumference	-0.282	-2.169 to-0.385	.005	
APOE (c.526C>T)	-0.131	-42.451 to -4.602	.015	
*APOB (*c.293G>A)	0.106	0.086-17.610	.048	

95%CI, 95% confidence interval, NFHGH, nonfamilial hypercholesterolemia genetic hypercholesterolemia.

Linear regression model adjusted for body mass index, age, sex, and waist circumference.