

# Supporting Information

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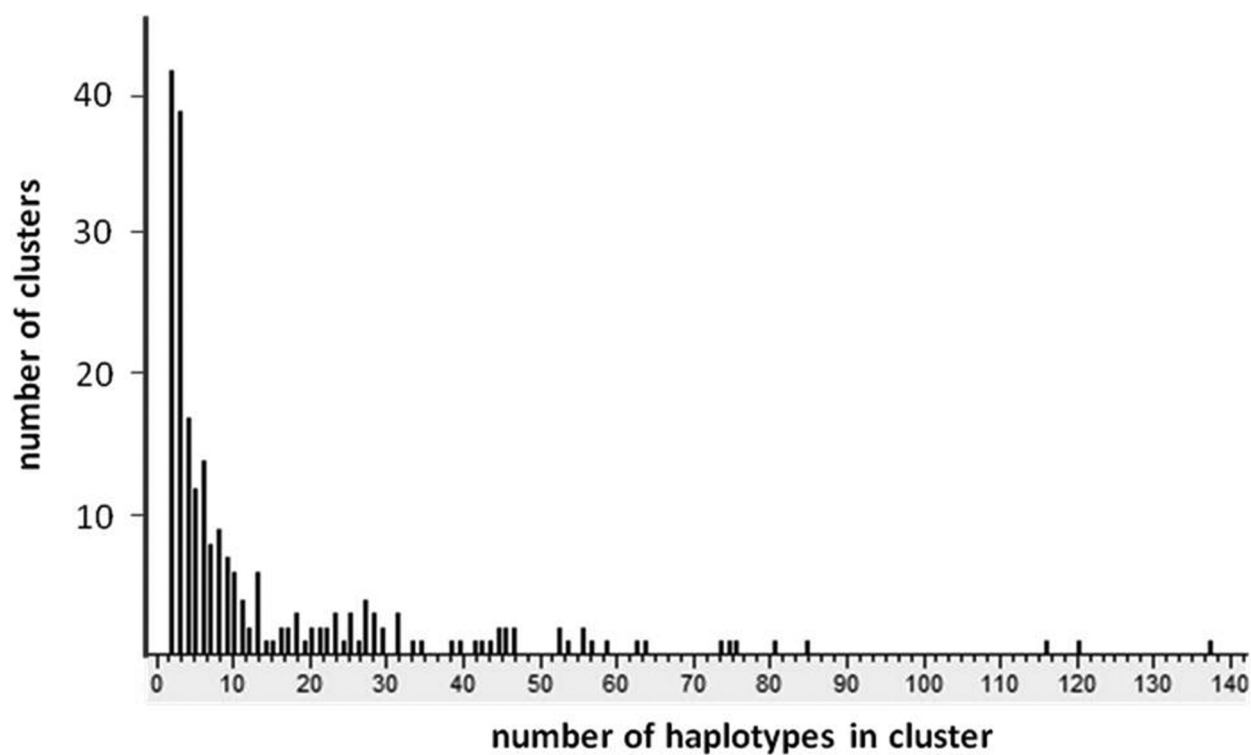


Fig. S1. Histogram of haplotype cluster sizes. The x-axis shows the haplotypes binned by the number of haplotypes in each cluster and the number of clusters for each bin is shown on the y-axis.





**Table S1. Linkage equilibrium between the ABCG8 nonsense mutation and rs12185607/G allele**

		rs12185607	
		GG	GT
ABCG8 nonsense mutation	GG	2512	64
	GC	321	2
	CC	5	0

**Table S2. Lathosterol:cholesterol and other plasma lipid levels. ANOVA analysis of lathosterol:cholesterol and plasma lipid levels comparing carriers of the ABCG8 mutation and 526-kb haplotype and individuals who are wild type for both mutations**

	Total Population	Noncarriers	ABCG8 mutation carriers	PPS haplotype carriers	Non- carriers vs. ABCG8 mutation carriers	Non- carriers vs. PPS haplotype carriers	Non- carriers vs ABCG8 carriers vs. PPS carriers
Lath:Chol	1.50 ± 0.66 (1,900)	1.52 ± 0.67 (1,687)	1.33 ± 0.55 (213)	0.84 ± 0.38 (44)	1.08 × 10 <sup>-4</sup>	3.68 × 10 <sup>-11</sup>	<0.0001
TC, mg/dL	166.09 ± 34.43 (2,819)	165.48 ± 34.35 (2,452)	170.2 ± 34.48 (315)	167.55 ± 34.92 (52)	0.022	N.S.	N.S
LDL-c, mg/dL	102.1 ± 29.54 (1,899)	101.63 ± 29.3 (1,645)	105.42 ± 30.63 (210)	100.84 ± 30.73 (44)	N.S.	N.S.	N.S
ApoB, mg/dL	86.97 ± 21.32 (1,884)	86.65 ± 21.31 (1,621)	88.53 ± 21.64 (219)	89.7 ± 19.8 (35)	N.S.	N.S.	N.S
HDL-c, mg/dL	38.28 ± 11.04 (1,899)	38.09 ± 10.95 (1,645)	38.77 ± 11.36 (210)	42.00 ± 9.93 (44)	N.S.	N.S.	N.S
ApoA1, mg/dL	116.87 ± 24.39 (1,875)	116.64 ± 24.07 (1,621)	118.07 ± 26.5 (219)	122.21 ± 25.3 (35)	N.S.	N.S.	N.S
TG, mg/dL	99.94 ± 46.16 (2,817)	100.94 ± 45.91 (2,450)	95.58 ± 39.02 (315)	93.26 ± 36.32 (52)	0.05	N.S.	N.S

*N*, number of individuals; Lath:Chol, lathosterol/cholesterol ratio; TC, total cholesterol; LDL-c, low density lipoprotein cholesterol; HDL-c, high density lipoprotein cholesterol; TG, triglycerides. All numbers are averages ± standard deviation and the sample size is shown in parentheses.