

Frequency of genetic variants involved in lipid metabolism and intrahepatic fat

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DOI: 10.31744/einstein_journal/2024CE1122

Dear Editor,

An alarming number of pandemics are affecting the world's population, including viral infections, overweight/obesity, and metabolic disorders. Genetic variants may be involved in underlying mechanisms of lipid metabolism and intrahepatic fat accumulation. The minor allele frequencies (MAFs) of variants in the *microsomal triglyceride transfer protein* (MTTP) gene, which unbalance the concentration of cholesterol, low-density lipoprotein and apolipoprotein B,⁽¹⁻³⁾ were investigated in 241 healthy Brazilians.

Data were examined in conjunction with results from the Allele Frequency Aggregator (ALFA) Project (Table 1), in which approximately one million individuals were included.⁽⁴⁾ The MAFs of H297Q and -493G/T variants were higher in the Brazilian population, possibly indicating a risk for metabolic disorders. These variations may be attributable to the fact that the ALFA Project was composed of a small Latin American population. Brazil has a unique population with evidence of high levels of genetic admixture,⁽⁵⁾ and we also considered epidemiological data on diversity, which is a challenging aspect of precision medicine.

The study was approved by the Ethics Committee of the *Hospital das Clínicas, Universidade de São Paulo* (CAAE: 57626816.3.0000.0068; #2.779.235).

How to cite this article:

Magri MC, Prata TV, Dantas BP, Manchiero C, Figueiredo GM, Tengan FM. Frequency of genetic variants involved in lipid metabolism and intrahepatic fat [letter]. *einstein* (São Paulo). 2024;22:eCE1122.

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Received on:

Mar 19, 2024

Accepted on:

Apr 15, 2024

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Table 1. Minor allele frequencies of genetic variants in the *microsomal triglyceride transfer protein* gene identified in the Brazilian population and in the NCBI SNP database (ALFA Project)

Variant (SNP)	Chr4 position	Allele	MAF			Brazilian population	HWE*
			NCBI Global	NCBI Latin American 1	NCBI Latin American 2		
-164T/C (rs1800804)	99574660	T/C	0.26	0.26	0.17	0.27	0.532
-400A/T (rs1800803)	99574424	A/T	0.37	0.46	0.26	0.37	0.212
H297Q (rs2306985)	99594865	C/G (H/Q)	0.39	0.53	0.27	0.50	0.220
-493G/T (rs1800591)	99574331	G/T	0.15	0.00†	0.00	0.33	0.723
I128T (rs3816873)	99583507	T/C (I/T)	0.26	0.28	0.17	0.26	0.277
Q95H (rs61733139)	99583409	G/C (Q/H)	0.05	0.04	0.02	0.09	0.098
Q244E (rs17599091)	99591762	C/G (Q/E)	0.03	0.04	0.01	0.06	0.302

* $p \geq 0.05$ indicates Hardy-Weinberg equilibrium in the Brazilian population (χ^2 test); † small sample size ($n=68$).

Chr4: chromosome 4; HWE: Hardy-Weinberg equilibrium; Latin American 1: Latin American individuals with Afro-Caribbean ancestry; Latin American 2: Latin American individuals with mostly European and Native American ancestry; MAF: minor allele frequency; NCBI: National Center for Biotechnology Information; SNP: single nucleotide polymorphism.

AUTHORS' CONTRIBUTION

Mariana Cavaleiro Magri: conceptualization, formal analysis, investigation, methodology, project administration and writing – original draft. Thamiris Vaz Gago Prata: formal analysis, investigation, methodology, and writing – original draft. Bianca Peixoto Dantas, Caroline Manchiero, Gerusa Maria Figueiredo and Fátima Mitiko Tengan: formal analysis, investigation, methodology, and writing – review & editing.

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