

SNPs of the FADS gene cluster are associated with polyunsaturated fatty acids in a cohort of patients with cardiovascular disease.

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Polymorphisms of the human Delta-5 (FADS1) and Delta-6 (FADS2) desaturase genes have been recently described to be associated with the level of several long-chain n-3 and n-6 polyunsaturated fatty acids (PUFAs) in serum phospholipids. We have genotyped 13 single nucleotide polymorphisms (SNPs) located on the FADS1-FADS2-FADS3 gene cluster (chromosome 11q12-13.1) in 658 Italian adults (78% males; mean age 59.7 +/- 11.1 years) participating in the Verona Heart Project. Polymorphisms and statistically inferred haplotypes showed a strong association with arachidonic acid (C20:4n-6) levels in serum phospholipids and in erythrocyte cell membranes (rs174545 adjusted P value for multiple tests, $P < 0.0001$ and $P < 0.0001$, respectively). Other significant associations were observed for linoleic (C18:2n-6), alpha-linolenic (C18:3n-3) and eicosadienoic (C20:2n-6) acids. Minor allele homozygotes and heterozygotes were associated to higher levels of linoleic, alpha-linolenic, eicosadienoic and lower levels of arachidonic acid. No significant association was observed for stearidonic (C18:4n-3), eicosapentaenoic (C20:5n-3) and docosahexaenoic (C22:6n-3) acids levels. The observed strong association of FADS gene polymorphisms with the levels of arachidonic acid, which is a precursor of molecules involved in inflammation and immunity processes, suggests that SNPs of the FADS1 and FADS2 gene region are worth studying in diseases related to inflammatory conditions or alterations in the concentration of PUFAs.

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