Curr Opin Lipidol. 2010 Feb;21(1):64-9. doi: 10.1097/MOL.0b013e3283327ca8.

Genetic variants of the FADS1 FADS2 gene cluster as related to essential fatty acid metabolism.

Lattka E[1], Illig T, Koletzko B, Heinrich J.

Author information

1 Institute of Epidemiology, Helmholtz Zentrum München, German Research Center for Environmental Health, Neuherberg, Germany.

Abstract

PURPOSE OF REVIEW: The delta-5 and delta-6 desaturases have long been known to be important enzymes in the endogenous formation of long-chain polyunsaturated fatty acids (LC-PUFAs). Cloning of the coding sequences and chromosomal localization of the desaturase encoding genes fatty acid desaturase 1 and 2 (FADS1 and FADS2) opened the way for analyses of genetic factors as regulators of desaturase activity and LC-PUFA homeostasis. The present review summarizes the recent association studies on FADS genotypes and LC-PUFA levels and suggests ideas how FADS genotypes can be integrated in future research.

RECENT FINDINGS: An initial candidate gene study reported highly significant associations between FADS gene cluster polymorphisms and fatty acid levels in serum phospholipids with an extraordinary high genetically explained variance for arachidonic acid levels of 28.5%. Carriers of the minor alleles had enhanced levels of desaturase substrates and decreased levels of desaturase products, suggesting a decline in desaturase expression or activity because of the polymorphisms. These results were replicated in several association studies additionally showing an effect in different human tissues as well as in a recent genome-wide association study on LC-PUFA levels.

SUMMARY: The validated strong association between FADS genotypes and fatty acid levels in diverse human tissues shows that FADS gene cluster polymorphisms are, in addition to nutritional regulation of fatty acid synthesis, a very important regulator of LC-PUFA synthesis.

PMID: 19809313 DOI: 10.1097/MOL.0b013e3283327ca8