



Variants in *CPT1A*, *FADS1*, and *FADS2* are associated with higher levels of estimated plasma and erythrocyte delta-5 desaturases in Alaskan Eskimos

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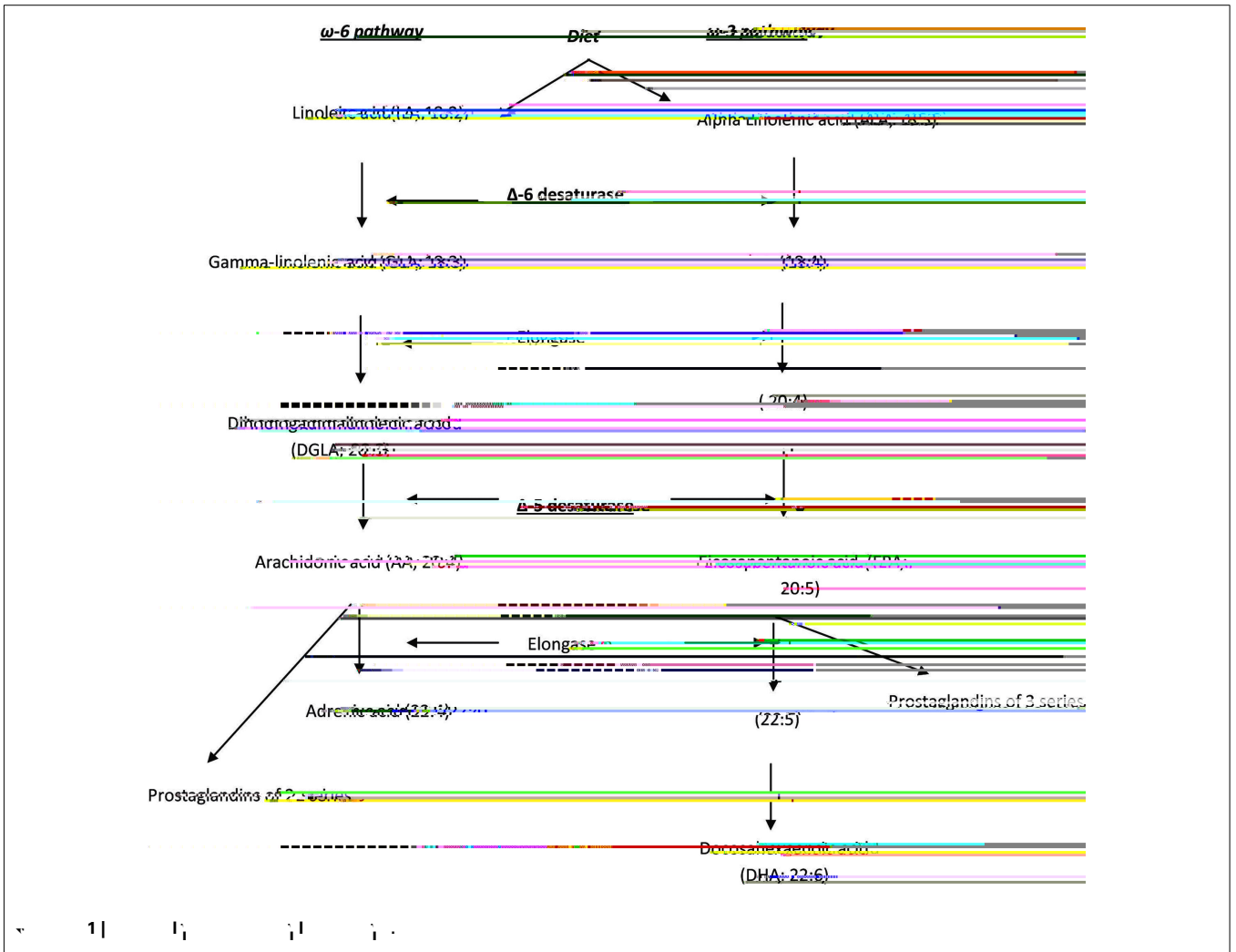
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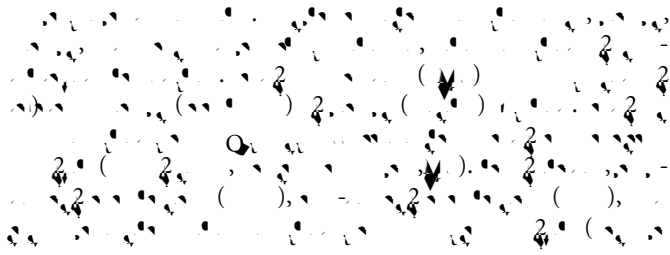
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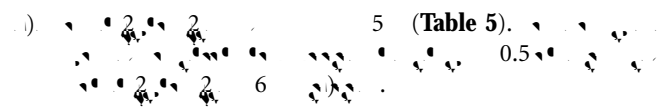
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The delta-5 and delta-6 desaturases (D5D and D6D), encoded by fatty acid desaturase 1 (*FADS1*) and 2 (*FADS2*) gene, respectively, are rate-limiting enzymes in the metabolism of omega-3 and omega-6 fatty acids. The objective of this study was to identify gene influencing variation in estimated D5D and D6D activities in plasma and erythrocytes in Alaskan Eskimo ($n=761$) participating in the genetic of common allele discovery in Alaska Native (GOCADAN) study. Desaturase activities were estimated by product specific ratio of polyunsaturated fatty acids. We found evidence of linkage for the estimated erythrocyte D5D (eD5D) on chromosome 11 (12-13) (logarithm of odds score = 3.5). The confidence interval contains candidate gene *FADS1*, *FADS2*, 7-dehydrocholesterol oxidase (*DHCR7*), and carnitine palmitoyltransferase 1A, like (*CPT1A*). Measured genotype analysis found association between *CPT1A*, *FADS1*, and *FADS2* single-nucleotide polymorphism (SNP) and estimated eD5D activity (p -value between 10^{-28} and 10^{-5}). A Bayesian analysis of linkage disequilibrium analysis identified rs3019594 in *CPT1A*

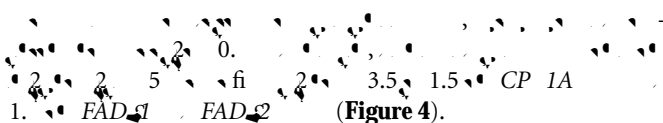


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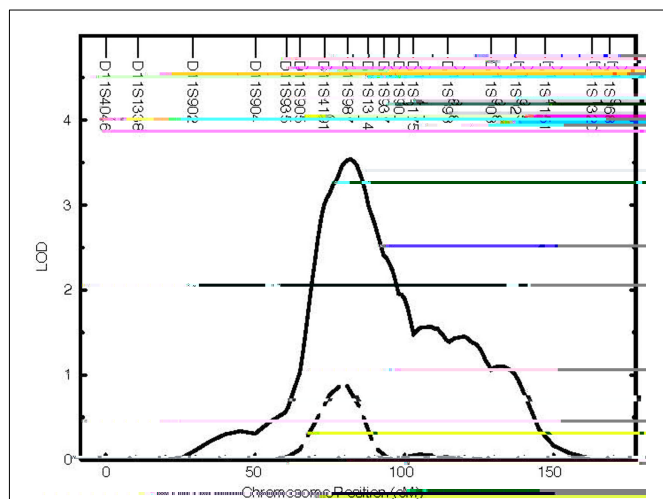


CONDITIONAL LINKAGE

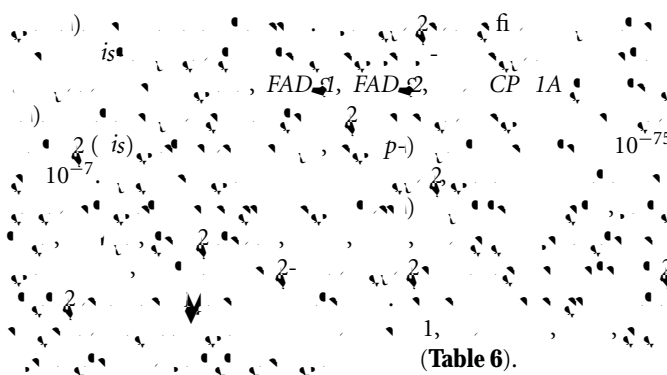


EXAMINATION OF CIS-REGULATED FADS1, FADS2, AND CPT1A TRANSCRIPTS IN MEXICAN AMERICANS

(2007),



11. The QTL for plasma and erythrocyte D5D on chromosome 11. Solid line, erythrocyte D5D (LOD = 3.5 a-.82 cM); dashed line, plasma D5D (LOD = 0.9 a-.81 cM).



DISCUSSION

fi FADS1, FADS2, CP 1A

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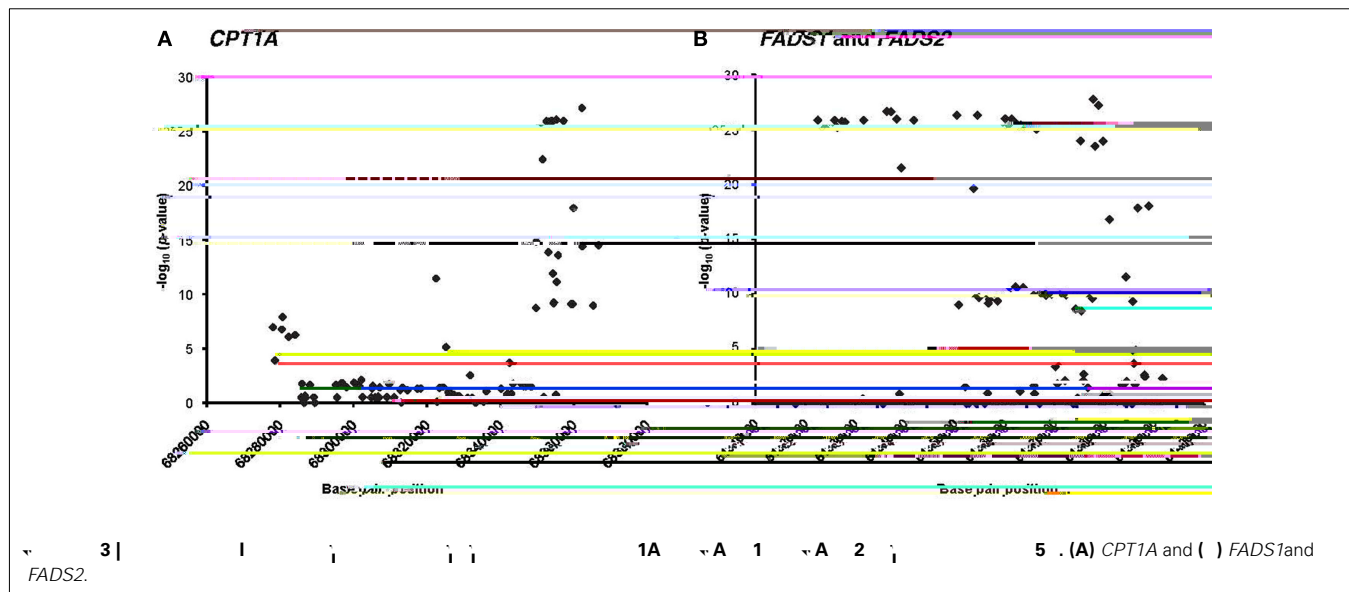
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11, 12, 13

FADS1, FADS2, CP 1A, DHC 7.



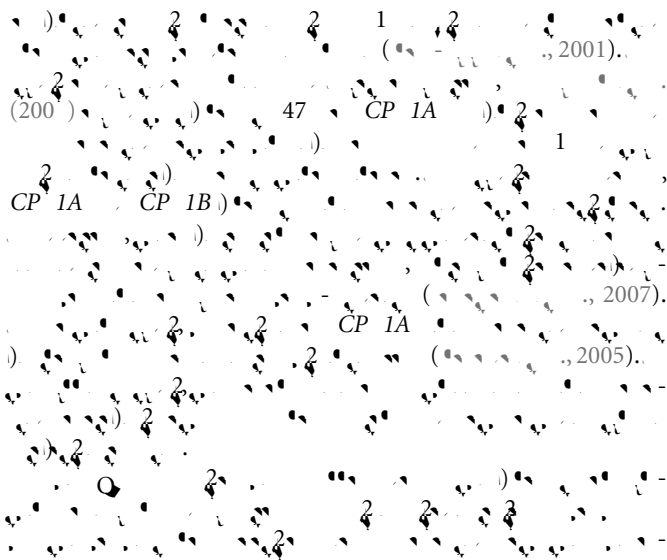
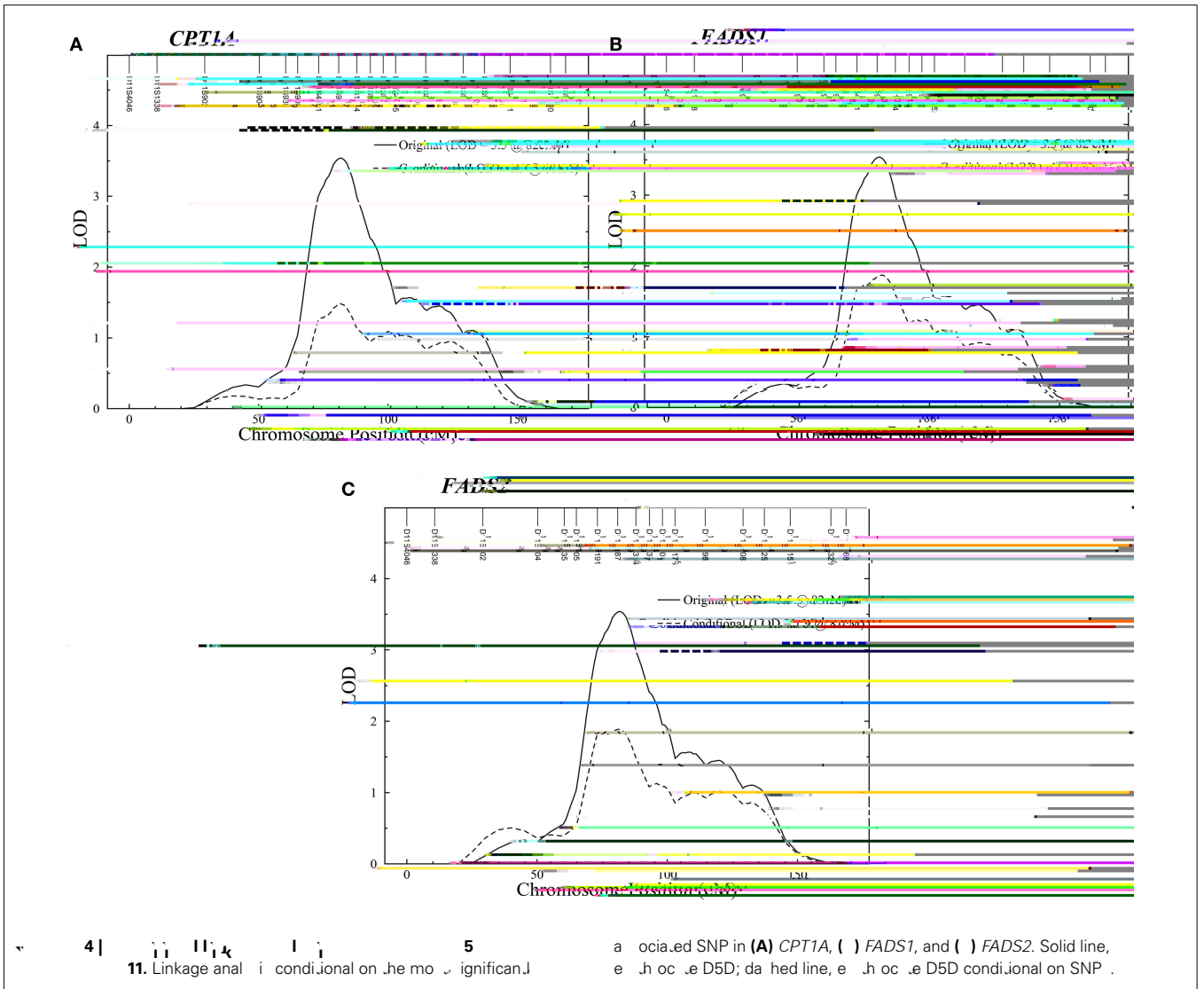
5 . (A) CPT1A and () FADS1 and FADS2.

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ii xi A | .

A (p-v |)*

