Neither maternal nor fetal mutation (E474Q) in the alpha-subunit of the trifunctional protein is frequent in pregnancies complicated by HELLP syndrome.

OBJECTIVE: An association between maternal HELLP syndrome and fetal long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency has been proposed. LCHAD catalyzes the third step in the beta-oxidation of fatty acids in mitochondria. Whereas about 75% of LCHAD-deficient patients carry a G-to-C mutation at nucleotide position 1528 (Glu474Gln, E474Q) on both chromosomes, compound heterozygosity for E474Q on one chromosome and a second different LCHAD mutation on the other can be observed in up to 25% of LCHAD-deficiency cases; only very few patients carry two mutations different from E474Q. Genetic analysis of the mother alone is insufficient in case of compound heterozygosity. Since information on the fetal carrier status of the E474Q mutation in maternal HELLP syndrome is rare, we investigated the frequency of the E474Q mutation in families where the mother had HELLP syndrome.

METHODS: The occurrence of the E474Q mutation was analyzed by PCR and RFLP in 103 mothers with HELLP syndrome, in 82 children of affected pregnancies and in 21 fathers in families where fetal DNA was not available. In addition, 103 control women with only uncomplicated pregnancies were investigated. RESULTS: The mutation E474Q was not detected in the study
population. CONCLUSION: Neither maternal nor fetal heterozygosity for the E474Q mutation is a relevant factor of HELLP syndrome.