

C 8. AN INHERITED RARE CDE HAPLOTYPE.

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Background: The Rh system is one of the most complex human blood group system. The Rh antigens are encoded by a complex RH locus which has been located on the short arm of chromosome 1. Fisher-Race theory explains the transmission of the Rh characters in bloc, by a set of three closely linked genes, and Tippett's theory, validated by molecular genetic studies, shows that the RH loci of human genome consists of two closely linked homologous genes RHD and RHCE, which transmit the RhD and RhCCEe characters.

But for interpreting serological data, the Fisher-Race theory is still appropriate. The three pairs of alleles can comprise eight haplotype, which can form 36 different genotypes. From these genotypes only 18 different phenotypes can be recognized by serological tests with appropriate reagents.

Aim: Serological testing in a family for the evidence of the inherited rare CDE haplotype.

Subject, reagents, methods: A Caucasian male immunological tested for the target donation for his wife. The same character in AB0, Kk, MN Ss, Pp systems between wife and husband. In Rh system was discordance between the two subjects: wife's phenotype was CcDee (32% in caucasians) and husband's phenotype was CCDEe (0,2% in caucasians).

Serological investigations in husband family revealed CDE rare haplotype along 3 generations (grandfather, son and one of the nephews), and consequently, the CCDEe phenotype.

Conclusions: The CDE haplotype is rare haplotype (second category with under 1% frequency), resulting from "cross-over" between C and D factors of type 1 haplotypes. In our study it was transmitted along 3 generations resulting in three members CCDEe rare phenotype (0,20% frequency in caucasians). This situation for the subjects is very important in transfusion.