VEL-NEGATIVE ALLELE FREQUENCY COMPARISON IN ONE AUSTRIAN AND THREE SWISS REGIONS FOR EFFICIENCY OPTIMIZATION OF DONOR SCREENING AND RECRUITMENT

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Background: The blood group Vel was discovered 60 years ago and individuals negative for this antigen are rare and are required for the safe transfusion of patients with antibodies to Vel. Very recently, the Vel-negative phenotype could be linked to *SMIM1*, located on chromosome 1p36. As reported three times timely in parallel, almost all Vel-negative individuals investigated so far (e.g. 164 of 169) were homozygous for a frameshift deletion of 17 bp in exon 3. An in house PCR using Sequence Specific Priming (PCR-SSP) was used for rapid Vel negative screening and genotyping.
Study design and methods: Austrian samples were from 422 blood donors of the Vienna region ("VI"), or were 855 DNAs collected in Switzerland in the course of the mandatory screening for the presence of *RHD* among RhD negative donors. Swiss regions were roughly represented by the cantons of Graubünden ("GR"), Central Switzerland ("CE": Luzern, Nidwalden, Obwalden, Schwyz and Zug), and Zurich and Schaffhausen ("ZH"). Using data published, two separate PCR-SSPs, one specific for the recognition of the Vel-negative allele with the 17 bp deletion (screening reaction) and another for the wildtype-allele were devised and positively validated using samples of known Vel negative phenotype.
Results: Of the Austrian and the three different Swiss regions of VI, GR, CE, and ZH, 422, 238, 196 and 421 individuals were screened and 13, 10, 8 and 10 were heterozygous for the Vel-negative allele, respectively. Allele-frequencies for this allele were calculated to be 0.0154, 0.0210, 0.0204 and 0.0119 in VI, GR, CE, and ZH, and Vel-negative homozygous donors may consequently be expected at a frequency of 2.4, 4.4, 4.2 and 1.4 per 10'000 donors, respectively.
Conclusion: Genetic linkage of the Vel locus *SMIM1* on chromosome 1 with the 25 Mbp distanced *RHD* may not be excluded a priori, and frequency data may therefore not be balanced among the investigated RhD negative Swiss donors, and additionally be incomparable to data obtained from RhD positive/negative Austrian donors. However, Vel-negative donor screening and recruitment would be more than three times more efficient in GR than in ZH, expecting 4.4 versus 1.4 Vel-negative donors per 10'000, respectively. Allocation of screening resources should therefore be done accordingly.