**Background**

In blood group determination, discrepancies between pheno- and genotypes are generally an indicator for the presence of unexpected or unknown, and usually very rare blood group alleles. In the course of routine screening RhD negative donors for the presence of RHD genes, a crosslink-ID-control PCR, specific for the B-allele of the ABO gene, tested positive in one case, although this donor had a record of blood group A at three independent blood donations.

**Method**

Standard serology was used. ABO genotyping was performed using a commercially available test kit “ABO” (Innotrain, Germany) and in house PCR-SSP technique. Allele-sequencing was accomplished by long range PCRs with generic amplifications of exons 1-3 and allele discriminative, nonO1- and O1-specific amplifications for exons 4 to 7, respectively.

**Results**

PCR-SSP ABO-genotyping at coding nucleotides 261, 802, 803 and 1’061, specific for alleles O1, O2, B, A2 and A, respectively, revealed a BO1 genotype of the investigated donor (Fig. 1). However, allele specific sequencing of exons 4-7 resulted in one “regular” A-allele as expected from the serological analysis (blood group A), and a second allele, carrying a G261del deletion, indicative of an O1 allele, and on the same allele the B-specific 803C (Gassner et al., 1996). The obtained sequences displayed identity to published ABO hybrid alleles O24 (O1v-B; www.ncbi.nlm.nih.gov) and O41 (O1v-B, tle13), respectively (Fig. 2). These alleles have been identified repeatedly in samples from Brazilian blacks and Akans from Ivory Coast (Olson et al., 1997; Roubinet et al., 2004).

**Conclusion**

The definitive name of our observed allele is uncertain, since the most similar O24 lacks sequence information of intron 6, while for O41, sequence of exons 1 to 5 is missing in the respective databases. Alternatively, a new allele, having originated from an independent crossing over event, may not be excluded at this time. However, the ethnic origin of our donor points to Brazilian ancestry and makes presence of one of the yet described alleles, O24 or O41 more likely. Although PCR-SSP technique is a well-accepted method for allele genotyping, the regular A allele in the investigated sample escaped correct identification, due to the pretended independent presence of O1 and B, but which were in fact encoded on one unexpressed allele simultaneously. Direct positive detection of A alleles would circumvent such errors, but is handicapped by the need for a 1’050 bp intron 6 crossing amplification, resulting in amplifications lengths, unusual for PCR-SSP.

**References**


14. Schweizerisches Symposium der Transfusionsmedizin, 06. - 07. September 2012, Basel, Switzerland