# A RARE *KEL\*02.*17 | *KEL\*02N.06(*IVS3+1g>a) COMPOUND HETEROZYGOUS INDIVIDUAL, PRONE TO ANTI-KEL11 IMMUNIZATION

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#### Background

The Kell blood group system includes some of the most immunogenic antigens among blood groups. Beside the well-known antigens Kell(*KEL\*01*), Kp<sup>a</sup>(*KEL\*03*), and Js<sup>a</sup>(*KEL\*06*), the antithetic antigens KEL11/17 further contribute to this list. However, KEL17 is considered as very rare, with an approximate frequency of one *KEL\*02.17* homozygote among 30'000 Europeans only<sup>1</sup>. We recently observed an individual with a rare anti-K11 and describe here its unusual molecular mechanism of anti-K11 sensitization.

#### Methode

Standard serological methods for antigen- and antibody-detection and specification were used. *KEL* genotyping was performed using a commercially available test kit "*KEL*plus" (Inno-Train, Germany) and in house *KEL*11/17 PCR-using Sequence Specific Priming technique (SSP) and *KEL* gene sequencing.



#### Results

By standard serological investigation, a 73 year old female was found positive for anti-KEL11 in her serum. Reasoned by the rarity of this observation, molecular confirmation was intended. A *KEL*11/17 PCR-SSP was performed, but resulted in an unexpected heterozygosity for *KEL*11/17. Further "*KEL*plus" typing delivered *KEL*-1,2,-3,4,-6,7 (K, Kp<sup>a</sup>, Js<sup>a</sup> negative), and surprisingly *KEL\*02.06(IVS3+1g>a)*, for the investigated DNA. Finally, *KEL* gene sequencing of exon areas 3 and 8 confirmed the unusual *KEL* genotype of the patient:

Compound heterozygosity for an expressed KEL\*02.17 and an unexpressed KEL\*02N.06(IVS3+1g>a).



#### Conclusion

 $KEL^{*02.06}(IVS3+1g>a)$  is the most frequent unexpressed KEL allele, encoding  $Kell_0$ , when present in homozygous, or compound heterozygous form<sup>2</sup>. However, in inherited hemizygousity this  $KEL_0$  allele will allow the second KEL allele to behave as seemingly homozygous, when expressed, as observed in our *case*. Such individuals might be expected at a frequency of 1 among 520'000 Europeans, only. Since this is the second observation of an anti-KEL11, beside another with true homozygosity for KEL\*02.17, we assume elevated frequency of KEL17 in the Zurich area as compared to other European areas.

#### References

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45. Jahreskongress der Deutschen Gesellschaft für Transfusionsmedizin und Immunhämatologie (DGTI), 11. - 14. September 2012, Graz, Austria