A RARE KEL17/KEL(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 know. Among them Kell(*KEL*1), Kp^a(*KEL*3), and Js^a(*KEL*6) are well known. The antithetic antigens KEL11/17 further contribute to this list. However, KEL17 is considered as very rare, with an approximte frequency of one *KEL*17 homozygote among 30'000 Europeans only (Daniels G, Human Blood Groups, 2002). Therefore, anti-KEL11 immunization is rarely observed and may be caused by unusual *KEL* genotypes, as exemplified here.
Methods: 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, Kronberg i. T., Germany) and in house *KEL*11/17 PCR-using Sequence Specific Priming technique (SSP) and *KEL* gene sequencing.
Results: After standard serological investigation, a 73 year old female presented anti-KEL11 in her serum. Reasoned by the rarity of this observation, molecular confirmation was intended. An in house *KEL*11/17 PCR-SSP was performed, but resulted in an inexplicable heterozygosity for *KEL*11/17. Therefore "*KEL*plus" typing was performed and delivered *KEL*-1,2,-3,4,-6,7 (K, Kp^a, Js^a negative), and surprisingly *KEL*(IVS3+1g>a), for the investigated DNA. Finally, *KEL* gene sequencing of exons 3 and 8 and adjacent intron sequences confirmed the unusual *KEL* genotype of the patient: Compound heterozygosity for an expressed *KEL*-1,2,-3,4,-6,7,**-11,17** and an unexpressed *KEL*-1,2,-3,4,-6,7,**11,-17,(IVS3+1g>a)** allele (relevant specificities are displayed in bold and underlined).
Conclusions: *KEL*(IVS3+1g>a) is the most frequent unexpressed *KEL* allele, encoding a further exeedingly rare, so called $Kell_0$ phenotype, when present in homozygous, or compound heterozygous form, together with other unexpressed *KEL* alleles (Koermoeczi G et al, Transfusion, 2007). Inherited hemizygously however, unexpressed *KEL* alleles will allow the second inherited *KEL* allele to behave as seemingly homozygous, when expressed. Thus explaining the reported phenotypical behaviour in the observed *KEL*17/(IVS3+1g>a) heterozygous case. Such individuals might be expected at a frequency of one among 520'000 Europeans, only. Indeed, this is the second report on an anti-KEL11 immunization from the area of Zurich, a truly *KEL*17 homozygote at that time, which may indicate a pronounced elevated frequency for KEL17 in this part of Switzerland compared to other European countries.

Allel-1:	*ABO* *A(261G, 802G, 803G, 1061C)
Allel-2:	*ABO* *B(261G, 802G, 803G>C, 1061C)
serologische Ableitung:	AB

Allel-1:	*JK* *A
Allel-2:	*JK* *B
serologische Ableitung:	Jk(a+b+)
	Jk^a pos, Jk^b pos

Allel-1:	*KEL* *1
Allel-2:	*KEL* 2
serologische Ableitung:	Kk

Allel-1:	*KEL* *2,4,7,11,(IVS3+1g>a)null
Allel-2:	*KEL* *2,4,7,17
serologische Ableitung:	Kk, Kp(a-b+), Js(a-b+), Kell11/17

Allel-1:	*KEL* 2,3,7,11
Allel-2:	*KEL* 2,4,7,11
serologische Ableitung:	Kk, Kp(a-b+), Js(a-b+), Kell11/17