Genotypes of patients with Vitamin K dependent factors deficiency

Gene	Mutation	Location	Type	Genotype	Origin	VKDF** Activity %	VKDF** Antigen %	Comments	Reference
GGCX	IVS1 del 14 bp	Intron 1	Undefined	Hom		9-30			1
GGCX	IVS2+1 G>T IVS11+3 A>G	Intron 2 Intron 11	Splicing Splicing	Comp het	Hispanic	2-4			2
GGCX	IVS2-1 G>T Arg485Pro*	Intron 2 Exon 11	Splicing Missense	Comp het	Germany	21-42			3
GGCX	Trp157Arg Thr591Lys and Asp31Asn	Exon 4 Exon 13	Missense Missense	Comp het	Tunis	5-9	60-73	Expression Expression Expression indicated polymorphism	4
GGCX	Leu394Arg	Exon 9	Missense	Hom	Arab	2-8	10-57	Expression	5,6,7
GGCX	His 404Pro Arg485Pro*	Exon 9 Exon 11	Missense Missense	Comp het	Germany	13-54			8
GGCX	Trp501Ser*	Exon 11	Missense	Hom	Lebanon	1-6		Expression	9,10
GGCX	Trp501Ser*	Exon 11	Missense	Hom	Lebanon	9-35			11
VKORC1	Arg98Trp*	Exon 3	Missense	Hom	Lebanon	20-60			12,13
VKORC1	Arg98Trp*	Exon 3	Missense	Hom	Germany	20-60			12,13
VKORC1	Arg98Trp*	Exon 3	Missense	Hom	Italy	8-26	35-100		14

Nucleotide numbers of *GGCX* are based on the Genebank file NM_000821 using the A (nt 29) of the ATG initiation methionine as +1.
Nucleotide numbers of *VKORC1* are based on the Genebank file AY423044 using the A (nt 1) of the ATG initiation methionine as +1.
*A mutation that was identified in more than 1 family.
**VKDF are prothrombin and factors VII, IX and X

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