

## Variability of clinical manifestation of Factor VII-deficiency in homozygous and heterozygous subjects of the European F7 gene mutation A294V

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**Supplementary Table S2.** Clinical manifestations in patients heterozygous for A294V patients (Index patient of the family is underlined). G –Germany, Sw-Sweden, SI-Slovakia).

Family number (origin)	Sex (age)	FVII:C	FVII:Ag	Presentation symptoms	Bleeding symptoms	Symptomatic heterozygous patient	Asymptomatic heterozygous subjects in the family	Other Index-patients in the family (Genotype)
<b>A. Heterozygous subjects FVII deficient families</b>								
f.1 (G)	M (18)	31	–	epistaxis	epistaxis, hematoma	<u>son</u>	father	–
f.2 (G)	M (30)	26	–	None	gum bleed	brother	mother	sister (294A>V/97G>S)
f.3 (G)	F (20)	33	–	None	easy bruising, gum bleed, menorrhagia	<u>daughter</u>	mother	–
f.4 (SI)	M (22)	30	50	epistaxis	epistaxis	son	son	mother (A294V/A294V; 404delC)
f.5 (SI)	M (30)	50	44	epistaxis	epistaxis	brother	father	brother (A294V/V362F)
f.6 (G)	F (51)	70	87	epistaxis	epistaxis, hematoma	mother	son	–
f.6 (G)	M (19)	55	67	epistaxis	epistaxis	<u>son</u>	–	–
f.7 (G)	F (18)	35	–	epistaxis	epistaxis	sister	–	–
f.7 (G)	M (13)	38	–	hematoma	easy bruising, hematoma	brother	–	–
f.8 (G)	F (38)	43	–	preop. diagn.	hematoma	mother	–	father (A294V/S111F)
f.8 (G)	F (3)	72	64	multiple	hematoma	easy bruising, epistaxis, hematoma	daughter	–
f.9 (G)	F (9)	39-55	–	epistaxis	epistaxis	sister	–	–
f.10 (G)	M (19)	34	–	easy bruising, hematoma	easy bruising, hematoma	<u>brother</u>	sister	sister (A294V hz (+vWD))
f.11 (G)	F (10)	30	–	epistaxis	epistaxis	<u>sister</u>	1 sister	–
ID-Number (origin)	Sex (age)	FVII:C	FVII:Ag	Presentation symptoms	Bleeding symptoms			
<b>B. Heterozygous Isolated/single cases</b>								
1 (G)	M (15)	36	–	postop. bleeding	hematoma			
2 (Sw)	M (42)	42	100	epistaxis	epistaxis			
3 (G)	M (70)	28	–	postop. bleeding	epistaxis			
4 (Sw)	F (53)	35	49	epistaxis	easy bruising, epistaxis, menorrhagia			
5 (Sw)	F (36)	37	–	menorrhagia	easy bruising, menorrhagia			
6 (G)	F (36)	27	–	easy bruising, hematomas	hematoma			
7 (G)	F (32)	32	–	menorrhagia	epistaxis, menorrhagia			
8 (G)	F (39)	39	–	gum bleed	epistaxis, gum bleed, menorrhagia			
9 (SI)	M (26)	32	66	epistaxis	easy bruising, epistaxis, gum bleed			

### Appendix

Members of the International Study Group of Greifswald Registry FVII D, who have enrolled A294V subjects: Germany: F.H. Herrmann, K. Wulff, H. Affeldt, O. Anders, M. Arlt, G. Auerswald, V. Aumann, S. Becker, F. Bergmann, K. Bergmann, E. Bratanoff, P. Bruhn, J. Buderer, G. Caspari, St. Demuth, B. Eifrig, R. Eisert, D. Franke, Funk, H.-D. Gabriel, A. Ganser, U. Geisen, E. Gilberg, M. Gnad, U. Göbel, N. Graf, Hähnlein, A. Hänel, M. Heinrichs, Ch. Heller, S. Horneff, R. Kätzel, K. Kentouche, R. Klamroth, D. Klarmann, U. Kluba, W. Kreuz, P. Küpferling, K. Kurnik, H. Lenk,

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