

Table 2

GGCX mutation	Age	Genotype	Non-haemostatic Phenotype/s	γ -carboxylated GRP ([%] at 10 μ M K ₁)	γ -carboxylated MGP ([%] at 10 μ M K ₁)	γ -carboxylated BGLAP ([%] at 10 μ M K ₁)	Comments	VKORC1 c.-1639	Ref.
R83P R83P	3 year	HMZ	Facial Dysmorphism + Septal defect	12.6 % 12.6 %	26.5 % 26.5 %	34 % 34 %		AA	Watzka et al., 2014
D153G M174R+ (R325Q)	4 months	CH	Keutel syndrome like phenotype	93.5 % 0.8 %	38.3 % 1.2 %	81.5 % 0 %			Tie et al., 2016
S284P W315X	13 year	CH	Midfacial hypoplasia + Septal defect	80.1% -	100.2% -	63.2 % -	Mother had hyperemesis gravidarum	AA	Watzka et al., 2014
G558R F299S	40 year	CH	PXE-like phenotype	31.7 % 0 %	18.2 % 1 %	65.8 % 0 %	No skeletal phenotype although low level of γ -carboxylated MGP		Vanakker et al., 2007
V255M S300F	16 year	CH	PXE-like phenotype + Cardiac abnormality	30.7 % 1.5 %	10.2 % 2.9 %	57.3 % 0 %	No skeletal phenotype although low level of γ -carboxylated MGP		Li, Grange et al., 2009
R83W Q374X	48 year	CH	PXE-like phenotype	26.2 % -	31.7 % -	15.8 % -	No skeletal phenotype although low level of γ -carboxylated MGP		Li, Schurgers et al., 2009