Gene symbol	Gene name	PTC location	Phenotype	References
VWF	von Willebrand factor	5' PTC	Recessively inherited type 3 von Willebrand disease	50
		3' PTC	Dominantly inherited type 2A von Willebrand disease	
F10	Coagulation factor X	5' PTC	Bleeding tendency associated to factor X deficiency (recessive)	51
		3' PTC	Bleeding tendency associated to factor X deficiency (dominant)	
RHO	Rhodopsin	5' PTC	Retinitis pigmentosa (recessive)	122, 123
		3' PTC	Retinitis pigmentosa (dominant)	
CLCN1	Chloride channel 1	5' PTC	Becker disease	124
		3' PTC	Thomsen disease	
CRX	Cone-rod homeobox-containing gene	5' PTC	No homozygotes to date, normal heterozygotus	52
		3' PTC	Leber congenital amaurosis (dominant)	
ABCC6	ATP-binding cassette, subfamily C member 6	5' PTC	Pseudoxanthoma elasticum (recessive)	125
		3' PTC	Pseudoxanthoma elasticum (dominant)	

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