Additional information from samples characterized by previous GeT-RM study (data from one laboratory)								
Coriell Catalog Info.		**Confirmed Genotype[1,2]**						
Cell Line Number	**DNA Number**	***F5***	***HFE***	***MTHFR***	***SERPINA1***	***SERPINE1***	***TPMT***	***VKORC1***
GM20730	NA20730	R506Q[3] heterozygous	H63D[3] heterozygous	677C>T[3] heterozygous 1298A>C[3] homozygous	SM[4]	(-)675 5G/4G[3]		(-)1639G>A[3] heterozygous
GM20835	NA20835			1298A>C[3] heterozygous	SM[4]	(-)675 4G/4G[3]		(-)1639G>A[3] heterozygous
GM20918	NA20918			677C>T[3] heterozygous 1298A>C[3] heterozygous	ZS[4]	(-)675 5G/4G[3]		(-)1639G>A[3] heterozygous
GM03578	NA03578			677C>T[3] heterozygous	ZZ[4]	(-)675 4G/4G[3]	A719G[5] heterozygous	(-)1639G>A[3] heterozygous
GM03579	NA03579		C282Y[3] heterozygous		ZM[4]	(-)675 4G/4G[3]	A719G[5] heterozygous	(-)1639G>A[3] heterozygous

[1]Allele names given here are colloquial. For proper mutation nomenclature, please refer to the Human Genome Variation Society (HGVS) website: www.hgvs.org/mutnomen/

[2]The complete mutation panel for each test are as follows: *F5* (R506Q only), *HFE* (C282Y, H63D, and S65C), *MTHFR* (677C>T and 1298A>C only), *SERPINA1* (S and Z alleles), *SERPINE1* (-675 4G/5G only), *TPMT* (G238C, G460A, A719G), *VKORC1* (-1639G>A only). All five samples were also tested for mutations in the *F2* (Prothrombin) gene (20210G>A only), the *CYP2C9* allele (430C>T and 1075A>C), and the *DPD* gene (IVS14+1G>A only) with no mutations detected.

[3]HGVS nomenclature: R506Q mutation = *F5* NM_000130.3:c.1601G>A (p.Arg534Gln); H63D mutation = *HFE* NM_000410.3:c.187C>G (p.His63Asp); C282Y mutation = *HFE* NM_000410.3:c.845G>A (p.Cys282Tyr); 677C>T variant = *MTHFR* NM_005957.3:c.665C>T (p.Ala222Val); 1298A>C variant = *MTHFR* NM_005957.3:c.1286A>C (p.Glu429Ala); (-)675 4G or 5G = *SERPINE1* NM_000602.2:c.-1968G(4_5); (-)1639G>A = *VKORC1* NM_024006.4:c.-1639G>A.

[4]The letters M, S, and Z were originally used to designate the protein, anode to cathode, in isoelectric focusing (See Science 149:986-7, 1965). M represents a wildtype allele, S represents the mutation *SERPINA1* NM_00295:c.863A>T (p.Glu264Val), and Z represents the mutation *SERPINA1* NM_00295:c.1096G>A (p.Glu342Lys).

[5]The A719G *TPMT* mutation is also known as *3C (*Hum Mutat* 12:177, 1998); and HGVS nomenclature for this mutation is *TPMT* NM_000367.2:c.719A>G (p.Tyr240Cys).