Additional information from samples characterized by previous GeT-RM study (data from one lab)

Coriell Catalog Info.		Confirmed Genotype[1,2,3]						
Cell Line Number	DNA Number	*F5*	*HFE*	*MTHFR*	*SERPINA1*	*SERPINE1*	*TPMT*	*VKORC1*
GM20730	NA20730	R506Q[4] heterozygous	H63D[4] heterozygous	677C>T[4] heterozygous 1298A>C[4] homozygous	SM[5]	(-)675 5G/4G		(-)1639G>A heterozygous
GM20835	NA20835			1298A>C[4] heterozygous	SM[5]	(-)675 4G/4G		(-)1639G>A heterozygous
GM20918	NA20918			677C>T[4] heterozygous 1298A>C[4] heterozygous	ZS[5]	(-)675 5G/4G		(-)1639G>A heterozygous
GM03578	NA03578			677C>T[4] heterozygous	ZZ[5]	(-)675 4G/4G	A719G[6] heterozygous	(-)1639G>A heterozygous
GM03579	NA03579		C282Y[4] heterozygous		ZM[5]	(-)675 4G/4G	A719G[6] heterozygous	(-)1639G>A homozygous

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

[2]Results are extra data acquired from one laboratory during a GeT-Rm characterization project for MTHFR, SERPINA1, RET, BRCA1, and BRCA2

[3]The complete mutation panel for each Specialty lab 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.

[4]Standard nomenclature for the R606Q mutation is NM_000130.3:c.1601G>A (p.Arg534Gln); standard nomenclature for H63D is NM_000410.3:c.187C>G (p.His63Asp); standard nomenclature for the C282Y is NM_000410.3:c.845G>A (p.Cys282Tyr); standard nomenclature for 677C>T variant is NM_005957.3:c.665C>T (p.Ala222Val); standard nomenclature for 1298A>C is c.1298A>C;

[5]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 p.Glu264Val, and Z represents the mutation p.Glu342Lys.

[6]The A719G *TPMT* mutation is also known as *3C (*Hum Mutat* 12:177, 1998).