Other Inherited Disorder Reference Materials Characterized by the GeT-RM							
		Confirmed Genotype		Coriell Catalog Info.		Characterization Methods:	
Disorder	Gene	Allele 1[1]	Allele 2[1]	Cell line number	DNA number	Sequencing	Other Methods[2] (# labs)
Hyperhomocysteinemia	***MTHFR***	677C>T[3] 1298A>C[3]	1298A>C[3]	GM20730	NA20730	1 lab	A(1), B(2), C(2), D(1), E(1)
AATD	***SERPINA1***	MS[4]		GM20835	NA20835	1 lab	A(1), B(2), E(2), F(1), G(1)
		SZ[4]		GM20918	NA20918	1 lab	A(1), B(2), E(2), F(1), G(1)
		ZZ[4]		GM03578	NA03578	1 lab	A(1), B(2), E(2), F(1), G(1)
		MZ[4]		GM03579	NA03579	1 lab	A(1), B(2), E(2), F(1), G(1)
MEN2A	***RET***	C620F[5]		GM16658	NA16658	5 labs	
		C618S[5]		GM16660	NA16660	5 labs	
Various Cancers	***BRCA1***	5382insC[6]		GM13715	NA13715	3 labs	H(1), I(1)
		185delAG[6]		GM14090	NA14090	3 labs	H(1), I(1)
Various Cancers	***BRCA2***	6174delT[6]		GM14170	NA14170	3 labs	H(1), I(1)

[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]Methods: A) PCR/Luminex; B) PCR with Hybridization Probes and Melting Curve Analysis; C) Third Wave Technologies Invader® Analyte Specific Reagent (ASR); D) Autogenomics INFINITI™ assay; E) PCR/Restriction enzyme digest/Electrophoresis; F) PCR/Restriction enzyme digest/ABI3100; G) Multiplex Allele-Specific PCR/Electrophoresis; H) Allele-Specific Oligonucleotide Hybridization; I) Heteroduplex Mobility Assay

[3]HGVS nomenclature for the 677C>T and 1298A>C variants are *MTHFR* NM_005957.3:c.665C>T (p.Ala222Val) and *MTHFR* NM_005957.3:c.1286A>C (p.Glu429Ala), respectively

[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 NM_00295:c.863A>T (p.Glu288Val), and Z represents the mutation NM_00295:c.1096G>A (p.Glu366Lys).

[5]The HGVS nomenclature for C620F and C618S is NM_020975.4:c.2049G>T (or p.Cys620Phe) and NM_020975.4:c.2043G>C (or p.Cys618Ser), respectively.

[6]185delAG and 5382insC are also known as 187delAG and 5385insC respectively, and both names are colloquial. The HGVS nomenclature for each is NC_000017.9:g.38529572_38529571delAG and NC_000017.9:g.38462606dupC, respectively. The HGVS nomeclature for 6174delT is NC_000013.9:c.24822delT.