

**Table 4: Other Inherited Disorders Reference Materials Characterized by the GeT-RM**

Disorder	Gene	Allele 1 <sup>1</sup>	Allele 2 <sup>1</sup>	Transformed	Cell type	Cell Bank	Cell line number	DNA number	DNA sequencing	Characterization Methods <sup>2</sup> (# labs)
Hyperhomocysteinemia	MTHFR	677C>T heterozygous	1298A>C homozygous	Yes	B-Lymphocyte	Coriell	<a href="#">GM20730</a>	<a href="#">NA20730</a>	1 lab	A(1), B(2), C(2), D(1), E(1)
AATD	SERPINA1	MS		Yes	B-Lymphocyte	Coriell	<a href="#">GM20835</a>	<a href="#">NA20835</a>	1 lab	A(1), B(2), E(2), F(1), G(1)
		SZ		Yes	B-Lymphocyte	Coriell	<a href="#">GM20918</a>	<a href="#">NA20918</a>	1 lab	A(1), B(2), E(2), F(1), G(1)
		ZZ		Yes	B-Lymphocyte	Coriell	<a href="#">GM03578</a>	<a href="#">NA03578</a>	1 lab	A(1), B(2), E(2), F(1), G(1)
		MZ		Yes	B-Lymphocyte	Coriell	<a href="#">GM03579</a>	<a href="#">NA03579</a>	1 lab	A(1), B(2), E(2), F(1), G(1)
MEN2A	RET	C620F		Yes	B-Lymphocyte	Coriell	<a href="#">GM16658</a>	<a href="#">NA16658</a>	5 labs	
		C618S		Yes	B-Lymphocyte	Coriell	<a href="#">GM16660</a>	<a href="#">NA16660</a>	5 labs	
Various Cancers	BRCA1	5382insC		Yes	B-Lymphocyte	Coriell	<a href="#">GM13715</a>	<a href="#">NA13715</a>	4 labs	H(1), I(1)
		185delAG		Yes	B-Lymphocyte	Coriell	<a href="#">GM14090</a>	<a href="#">NA14090</a>	4 labs	H(1), I(1)
Various Cancers	BRCA2	6174delT		Yes	B-Lymphocyte	Coriell	<a href="#">GM14170</a>	<a href="#">NA14170</a>	4 labs	H(1), I(1)

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

<sup>2</sup>Methods: A) PCR/Luminex; B) PCR with Hybridization Probes and Melting Curve Analysis; C) Third Wave Technologies Invader® ASR; D) Autogenomics INFINITI™ assay; E) PCR/RE/Electrophoresis; F) PCR/RE/ABI3100; G) Multiplex Allele-Specific PCR/Electrophoresis; H) Allele-Specific Oligonucleotide Hybridization; I) Heteroduplex Mobility Assay