

Rett syndrome genotypes

Coriell Cell Line #	Consensus Genotype Determined by Study
GM11299	c.316C>T; p.R106W heterozygous
GM11310	c.502C>T; p.R168* heterozygous
GM16270	c.763C>T; p.R255* heterozygous
GM16271	c.473 C>T; p.T158M heterozygous
GM16480	c.916C>T; p.R306C heterozygous
GM16546	c.880C>T; p.R294* heterozygous
GM17540	c.401C>G p.S134C heterozygous
GM11313	gross deletion of exons 3 and 4 in the MECP2 gene (EX3_4del) heterozygous
NA23459	del 5' exon 4 heterozygous
NA23461	c.455C>G; p.P152R heterozygous
NA23462	c.808C>T; p.R270* heterozygous
NA23463	c.1173_1207del p.Glu392* heterozygous

Characterization study of these samples is described in:

Lisa Kalman; Jack Tarleton; Alan Percy; Swaroop Aradhya; Sherri Bale; Pinar Bayrak-Toydemir; Arlene Buller-Burckle; Soma Das; Ram Iyer; Timothy Vo; Val Zvereff; Lorraine Toji.
 Development of a genomic DNA reference material panel for Rett Syndrome Genetic Testing.
 Journal of Molecular Diagnostics 2014 16:273-279

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NA23503	c.806delG; p.Gly269Alafs*20 heterozygous
NA23598	c.808C>T; p.R270* heterozygous
NA23599	del exon 3-4 heterozygous
NA23605	c.421C>G; p.Y141* heterozygous
NA23606	c.27-8C>G (Intron 2 IVS8C>G) heterozygous
NA23607	c.1157_1200del44; p.L386Gfs*4 heterozygous
NA23624	c.398G>C; p.R133P heterozygous
NA23625	c.808C>T; p.R270* heterozygous
NA23626	c.455C>G; p.P152R heterozygous
NA23634	c.397C>T; p.R133C heterozygous
NA23635	del exon 3 and part of exon 4, heterozygous
NA23647	c.378-3C>G (IVS3-3) heterozygous

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NA23648	del exon 4 into <i>IRAK1</i> , heterozygous
NA23654	del exon 3 and part of exon 4, heterozygous
NA23659	c.397C>T; p.R133C heterozygous
NA23663	c.397C>T; p.R133C heterozygous
NA23675	Male with complete <i>MeCP2</i> duplication plus duplication of flanking <i>IRAK1</i> (Exons 3, 4)
NA23676	Female with complete <i>MeCP2</i> duplication plus duplication of flanking <i>IRAK1</i> , heterozygous
NA23733	Female with complete <i>MeCP2</i> duplication plus duplication of flanking genes (<i>FLNA-MeCP2-IRAK1-L1CAM</i>), heterozygous

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NA23734	Male with complete <i>MeCP2</i> duplication plus duplication of flanking genes (<i>FLNA-MeCP2-IRAK1-L1CAM</i>)
NA23747	c.806delG; p.Gly269Alafs*20 heterozygous
NA23748	c.806delG; p.Gly269Alafs*20 heterozygous
NA23846	c.1163_1188del26; p.Pro388Argfs*7 heterozygous