

Genotype of Duchenne muscular dystrophy cell lines characterized by the GeT-RM

Characterization Method Used*

Coriell Cell Line	Sex	Consensus Genotype	Affymetrix 6.0	MLPA (Laboratory 1)	MLPA (Laboratory 2)	Targeted Multiplex [#]	DNA Sequence Analysis
GM07691	Male	del 5'end -18	del 5'end -18	del 5'end - 18	del 5'-18	del pb - 17	ND ^{^^}
GM07692	Female	del 5'end-18	del 5' - 18	del 5' - 18	del 5' - 18	ND	ND
GM03782	Male	del 3-17	del 3-17	del 3-17	del 3-17	del 3-17	ND
GM05089	Male	del 3-5	del 3-5	del 3-5	del 3-5	del 3-4	ND
GM05170	Male	del 4-43	del 4-43	del 4-43	del 4-43	del 4-43	ND
GM23094	female	del 35-43	del 35-43	del 35-43	del 35-43	ND	ND
GM04315	Female	del 44	del 44	del 44	del 44	ND	ND
GM05115	Male	del 45	del 45	del 45	del 45	del 45	ND
GM05117	Female	del 45	del 45	del 45	del 45	ND	ND
GM04981	Male	del 45-53	del 45-53	del 45-53	del 45-53	del 45-52	ND
GM03929	Male	del 46-50	del 46-50	del 46-50	del 46-50	del 46-50	ND
GM05159	Female	del 46-50	del 46-50	del 46-50	del 46-50	ND	ND
GM04100	Male	del 49-52	del 49-52	del 49-52	del 49-52	del 49-52	ND
GM04099	Female	del 49-52	del 49-52	del 49-52	del 49-52	ND	ND
GM04364	Male	del 51-55	del 51-55	del 51-55	del 51-55	del 51-52	ND
GM10283	Male	del 72-79	del 73-79**	del 72-79	del 72-79	ND	ND
GM23086	Male	dup 2-30	dup 2-30	dup 2-30	dup 2-30	ND	ND
GM23087	Female	dup 2-30	dup 2-30	dup 2-30	dup 2-30	ND	ND
GM09981	Male	dup 2-4	dup 2-4	dup 2-4	dup 2-4	ND	ND
GM04327	Male	dup 5-7	dup 5-7	dup 5-7	dup 5-7	ND	ND
GM23099	female	dup 8-17	dup 8-17	dup 8-17	dup 8-17	ND	ND
GM23159	Male	dup 17	dup 17	dup 17	dup 17	ND	ND
GM23127	Male	dup 27-28	dup 27**	dup 27-28	dup 27-28	ND	ND
GM05124	Male	dup 45-62	dup 45-62	dup 45-62	dup 45-62	ND	ND
GM05263	Male	ND	no del/dup	no del/dup	ND	ND	c.7893delC (p.Q2632Sfs6)

GM05127	Male	ND	no del/dup	no del/dup	ND	ND	c.5533G>T (p.E1845X)
GM02298	Male	ND	no del/dup	no del/dup	ND	ND	No Mutation
GM04569	Male	ND	no del/dup	no del/dup	ND	ND	No Mutation
GM04619	Male	ND	no del/dup	no del/dup	ND	ND	c.8713C>T (p.R2905X)
GM04978	Male	ND	no del/dup	no del/dup	ND	ND	c.5893C>T (p.Q1965X)
GM05082	Male	ND	no del/dup	no del/dup	ND	ND	No Mutation

*A complete description of this project will be provided in: Kalman et al, Quality Assurance for Duchenne and Becker Muscular Dystrophy Genetic Testing: Development of a Genomic DNA Reference Material Panel, Journal of Molecular Diagnostics, 2011, in press.

**The Affy SNP 6.0 array is unable to detect mutations in exons 28 and 72 due to the lack of probes in these regions. A duplication was identified in GM23127 using the Affy Cytogenetics 2.7M array, which is also unable to identify mutations in exon 28 due to a lack of probe coverage.

#This assay does not detect all deleted exons

^ND- Not Determined