

Targeted Genes and Methodology Details for MYOD1 Mutation Analysis

The following applies to MYODT / MYOD1 Mutation Analysis, Next-Generation Sequencing. Next-generation sequencing (NGS) is performed to test for the presence of single nucleotide variations, deletions and insertions in coding regions and intron/exon boundaries of the genes listed. When appropriate, alterations detected are confirmed by an independent reference method, such as Sanger sequencing. Default reportable range offset is +/-2 base pairs around each targeted exon region.

Genomic Build: GRCh37 (hg19) unless otherwise specified.

As a result of technical limitations of the assay (including regions of homology, high GC content, and repetitive sequences), there are regions of some genes that cannot be effectively evaluated. Refer to gene regions table below for complete gene coverage information. To verify if a specific region/exon/variant is covered by this assay, contact the laboratory at 800-533-1710.

Gene	Exon	Chromosome	Genomic Start	Genomic End	Transcript
MYOD1	Ex1	chr11	17741328	17741961	NM_002478
MYOD1	Ex2	chr11	17742447	17742529	NM_002478
MYOD1	Ex3	chr11	17742800	17743057	NM_002478