

S1 Table. Classification of meSNPs from the sequence ontology (SO term) annotation provided by Ensembl

Categories	Variables that make up each category (SO term)
Proximal promoter	Upstream gene variant
Intronic region	Splice acceptor variant Splice donor variant Intron variant
Coding region	Stop gained Frameshift variant Stop lost Start lost Inframe insertion Inframe deletion Missense variant Protein altering variant Incomplete terminal codon variant Stop retained variant Synonymous variant Coding sequence variant NMD transcript variant
Non-coding RNA region	Mature miRNA variant Non-coding transcript exon variant Non coding transcript variant
Intergenic region	Downstream gene variant Intergenic variant
Regulatory region	TFBS ablation TFBS amplification TF binding site variant Regulatory region ablation Regulatory region amplification Regulatory region variant
5'UTR	5 prime UTR variant
3'UTR	3 prime UTR variant
Transcript deletion region	Transcript ablation
Transcription amplification region	Transcript amplification
Splice region	Splice region variant
Elongation region	Feature elongation
Feature truncation	Feature truncation