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Supplemental Data

Using Somatic Mutations from Tumors

to Classify Variants in Mismatch Repair Genes

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Supplemental Tables:

Table S1: Z-score calculations for changes at canonical splice sites defined by MaxEnt score distributions for all MMR genes See Excel file.

Table S2: Variant counts used to derive likelihood ratios where loss of heterozygosity is observed and where no loss of heterozygosity is observed

Loss of heterozygosity present, variant read fraction cutoff = 0.8

All tumors	Below	Above
Known Pathogenic and Loss of Function	7	37
Intronic	244	8

Colorectal		
only	Below	Above
Known Pathogenic and Loss of Function	4	28
Intronic	155	6

Endometrial		
only	Below	Above
Known		
Pathogenic	2	0
and Loss of	2	9
Function		
Intronic	71	2

No loss of heterozygosity present, variant read fraction cutoff = 0.35

	Below	Above
Known		
Pathogenic		400
and Loss of	9	100
Function		
Intronic	209	299

	Below	Above
Known		
Pathogenic	2	44
and Loss of	2	41
Function		
Intronic	43	82

_	Below	Above
Known		
Pathogenic	6	
and Loss of	6	53
Function		
Intronic	164	215

Table S3: Multifactorial likelihood calculations for 165 variants in *MLH1*, *MSH2*, *MSH6*, and *PMS2* See Excel file.