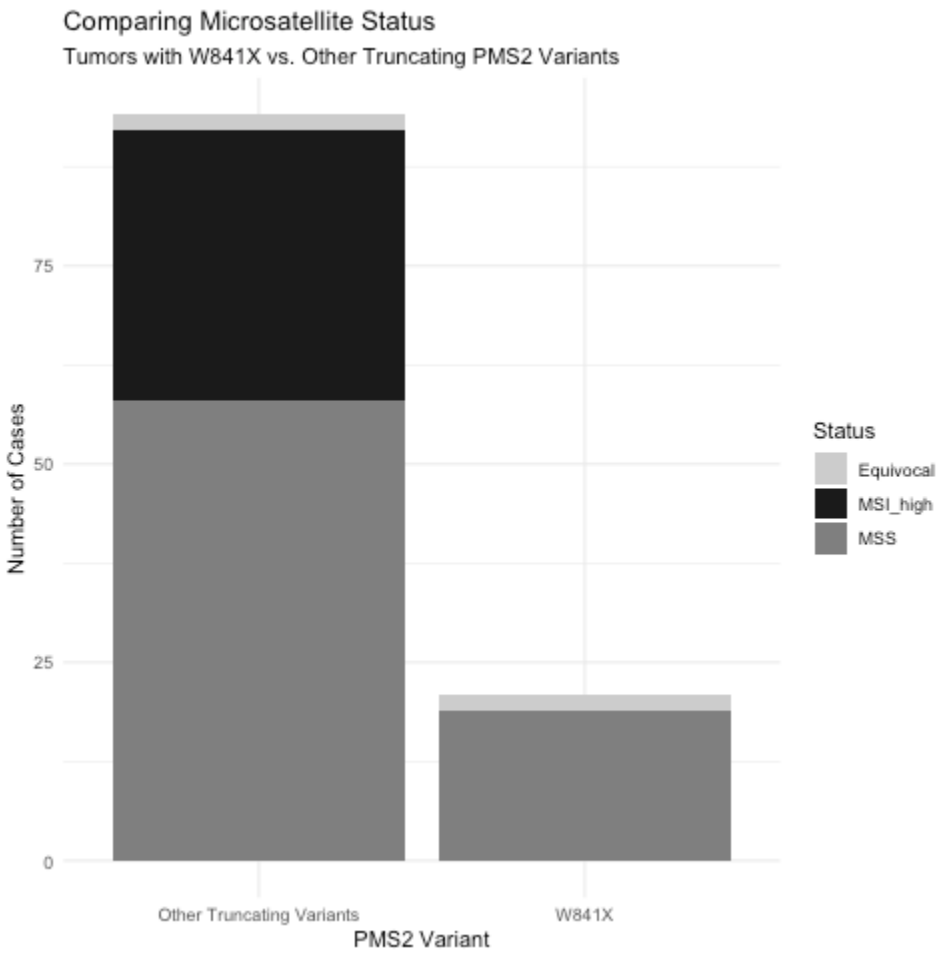
Supplementary Materials



Supplemental Figure 1: Comparing MS status for tumors containing the W841* variant versus tumors containing other truncating PMS2 variants. 19 of 21 tumors with W841* were MSS, none were MSI-high and two were equivocal. Of the 94 tumors with other truncating PMS2 variants, 58 were MSS, 34 were MSI-high, and two were equivocal.

Supplementary Table 1: *PMS2* Somatic Variants in tumors with data on microsatellite status

HGVS Transcript	Gene	HGVS c.	HGVS p.	ACMG Classification	ClinVar ID
NM_000535.7	PMS2	c.12dup	p.(E5*)	Pathogenic	1406959
NM_000535.7	PMS2	c.23+1G>T	p.?	Pathogeinc/Likely Pathogenic	141871
NM_000535.7	PMS2	c.24-1G>A	p.?	Pathogenic/Likely pathogenic	924601
NM_000535.7	PMS2	c.164-1G>C	p.?	Pathogenic/Likely Pathogenic	186061
NM_000535.7	PMS2	c.241G>T	p.(E81*)	Pathogenic/Likely Pathogenic	439243
NM_000535.7	PMS2	c.248T>G	p.(L83*)	Pathogenic/Likely Pathogenic	419752
NM_000535.7	PMS2	c.251-2A>T	p.?	Pathogenic/Likely Pathogenic	183893
NM_000535.7	PMS2	c.325del	p.(E109fs*3)	Pathogenic	141395
NM_000535.7	PMS2	c.354-1G>A	p.?	Pathogenic/Likely Pathogenic	187726
NM_000535.7	PMS2	c.354-2A>G	p.?	Pathogenic/Likely Pathogenic	185340
NM_000535.7	PMS2	c.400C>T	p.(R134*)	Pathogenic	9234
NM_000535.7	PMS2	c.433C>T	p.(Q145*)	Pathogenic	1739846
NM_000535.7	PMS2	c.538-1G>A	p.?	Pathogenic/Likely Pathogenic	948334
NM_000535.7	PMS2	c.538-1G>C	p.?	Pathogenic/Likely Pathogenic	434027
NM_000535.7	PMS2	c.538G>T	p.(E180fs*21)	Pathogenic/Likely Pathogenic	1747225
NM_000535.7	PMS2	c.631C>T	p.(R211*)	Pathogenic	234508
NM_000535.7	PMS2	c.697C>T	p.(Q233*)	Pathogenic	91362
NM_000535.7	PMS2	c.703C>T	p.(Q235*)	Pathogenic	91363
NM_000535.7	PMS2	c.706-1G>T	p.?	Likely Pathogenic	1756960
NM_000535.7	PMS2	c.730C>T	p.(Q244*)	Pathogenic	620098
NM_000535.7	PMS2	c.735dup	p.(P246fs*3)	Pathogenic	1074077
NM_000535.7	PMS2	c.750del	p.(V251fs*7)	Pathogenic	2674259
NM_000535.7	PMS2	c.765C>A	p.(Y255*)	Pathogenic	183716
NM_000535.7	PMS2	c.79del	p.(C27fs*7)	Pathogenic/Likely Pathogenic	1761572
NM_000535.7	PMS2	c.802del	p.(Y268fs*31)	Pathogenic/Likely Pathogenic	922836
NM_000535.7	PMS2	c.802dup	p.(Y268fs*39)	Pathogenic	91369
NM_000535.7	PMS2	c.804-1G>A	p.?	Pathogenic(1)/Likely Pathogenic(3)/VUS(1)	573060
NM_000535.7	PMS2	c.823C>T	p.(Q275*)	Pathogenic	127796
NM_000535.7	PMS2	c.828C>A	p.(C276*)	Pathogenic	421403
NM_000535.7	PMS2	c.861_864del	p.(R287fs*19)	Pathogenic	91375
NM_000535.7	PMS2	c.862_863del	p.(Q288fs*10)	Pathogenic	91376
NM_000535.7	PMS2	c.903G>A	p.?	Pathogenic/Likely Pathogenic	237932
NM_000535.7	PMS2	c.1145-2A>G	p.?	Likely Pathogenic	822245
NM_000535.7	PMS2	c.1239del	p.(D414fs*34)	Pathogenic	1171824
NM_000535.7	PMS2	c.1239dup	p.(D414fs*44)	Pathogenic	216072
NM_000535.7	PMS2	c.1261C>T	p.(R421*)	Pathogenic	91299
NM_000535.7	PMS2	c.1264G>T	p.(E422*)	Pathogenic	525843
NM_000535.7	PMS2	c.1266_1281del	p.(E44fs*13)	Pathogenic	2674271
NM_000535.7	PMS2	c.1351del	p.(R451fs*7)	Likely Pathogenic	633373
NM_000535.7	PMS2	c.139_146del	p.(L47fs*4)	Pathogenic/Likely Pathogenic	265547
NM_000535.7	PMS2	c.1471G>T	p.(E491*)	Pathogenic/Likely Pathogenic	486935
NM_000535.7	PMS2	c.1591G>T	p.(E531*)	Pathogenic	237890
NM_000535.7	PMS2	c.1606C>T	p.(Q536*)	Pathogenic	486937
NM_000535.7	PMS2	c.1687C>T	p.(R563*)	Pathogenic	135067
NM_000535.7	PMS2	c.1743del	p.(E582fs*8)	Pathogenic	372470
NM_000535.7	PMS2	c.1810C>T	p.(Q604*)	Pathogenic/Likely Pathogenic	858883
NM_000535.7	PMS2	c.1831del	p.(I611fs*2)	Pathogenic	1780965
NM_000535.7	PMS2	c.1924del	p.(E642*)	Pathogenic/Likely Pathogenic	1404626
NM_000535.7	PMS2	c.1927C>T	p.(Q643*)	Pathogenic	91320
NM_000535.7	PMS2	c.1939A>T	p.(K647*)	Pathogenic	91321
NM_000535.7	PMS2	c.1972C>T	p.(Q658*)	Pathogenic	1451259
NM_000535.7	PMS2	c.1981G>T	p.(E661*)	Pathogenic	185743
NM_000535.7	PMS2	c.2156del	p.(Q719fs*6)	Pathogenic	183755
NM_000535.7	PMS2	c.2331dup	p.(F778fs*8)	Pathogenic	2674214
NM_000535.7	PMS2	loss exons 2-11	p.?	Likely Pathogenic	Not in Clinvar
NM_000535.7	PMS2	loss exons 13-14	p.?	Likely Pathogenic	Not in Clinvar
NM_000535.7	PMS2	c.2006+2T>C	p.?	Likely Pathogenic	Not in Clinvar
NM_000535.7	PMS2	Not provided(1)	p.(C73fs*6)	Likely Pathogenic	Not in Clinvar
NM_000535.7	PMS2	Not provided(1)	p.(G466fs*3)	Likely Pathogenic	Not in Clinvar
NM_000535.7	PMS2	Not provided(1)	p.(I688fs*5)	Likely Pathogenic	Not in Clinvar
NM_000535.7	PMS2	Not provided(1)	p.(Q161*)	Likely Pathogenic	Not in Clinvar
NM_000535.7	PMS2	Not provided(1)	p.(S478fs*4)	Likely Pathogenic	Not in Clinvar

1: cDNA position not provided