

Supplementary

Whole-Exome Sequencing of Rare Site Endometriosis-Associated Cancer

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Supplementary Table 1. Candidate actionable variants in case 1.

Function	Gene variants	VAF (%)	CN
OG	<i>CTNNB1</i> V358A	22.2	2.4
OG	<i>MNX1</i> L34F	24.8	2.4
OG	<i>TFG</i> G241D	25	1.7
OG	<i>ZCCHC8</i> A47P	36	2.5
OG	<i>MUC4</i> T3718A	47.2	3
TSG	<i>SMC1A</i> R771Q	35.7	2.8
TSG	<i>FBXO11</i> L560F	21.1	2.5

VAF: Variant allele frequency

CN: Copy number

OG: Oncogene

TSG: Tumor suppressor gene

Supplementary Table 2. Candidate actionable variants in case 2.

Function	Gene variants	VAF (%)	CN
OG	<i>USP</i> R763W	8	1.8
OG	<i>MUC16</i> P563T	40.7	2.1
OG	<i>USP8</i> N764K	8	1.8
TSG	<i>PRF1</i> C407Y	40.8	1.9

VAF: Variant allele frequency

CN: Copy number

OG: Oncogene

TSG: Tumor suppressor gene

Supplementary Table 3. Gene list of copy number alteration in case 2

Function	Gene name	CNA	Copy number
dMMR	<i>PMS2</i>	LOH	1.2
HRD	<i>ARID1A</i>	LOH	1.1
HRD	<i>BRCA2</i>	LOH	1
HRD	<i>CHEK2</i>	LOH	1.1
HRD	<i>FANCA</i>	LOH	1.1
HRD	<i>FANCC</i>	LOH	1
HRD	<i>RAD50</i>	LOH	0.9
HRD	<i>RAD51</i>	LOH	1
HRD	<i>WRN</i>	LOH	1.2
OG	<i>CRLF2</i>	Amp	4.4
OG	<i>ELF4</i>	Amp	4.4
OG	<i>MTCP1</i>	Amp	4.8
OG	<i>MYC</i>	Amp	4.2
OG	<i>P2RY8</i>	Amp	4.4
OG	<i>PRDM16</i>	Amp	6
TSG	<i>BUB1B</i>	LOH	1
TSG	<i>CDKN1B</i>	LOH	0.8
TSG	<i>STK11</i>	LOH	1.2
TSG	<i>ERCC5</i>	LOH	1
TSG	<i>B2M</i>	LOH	1
TSG	<i>NCOR1</i>	LOH	1
TSG	<i>TSC1</i>	LOH	1
TSG	<i>RB1</i>	LOH	1
TSG	<i>RBM10</i>	LOH	1.1
TSG	<i>KDM6A</i>	LOH	1.1
TSG	<i>NF2</i>	LOH	1.1
TSG	<i>NF1</i>	LOH	1
TSG	<i>CNOT3</i>	LOH	1.1
TSG	<i>FLCN</i>	LOH	1
TSG	<i>CDKN2A</i>	LOH	1
TSG	<i>SMARCB1</i>	LOH	1.1
TSG	<i>PIK3R1</i>	LOH	0.9
TSG	<i>KMT2D</i>	LOH	0.8
TSG	<i>ARID2</i>	LOH	0.8
TSG	<i>XPA</i>	LOH	1
TSG	<i>CDH1</i>	LOH	1.1
TSG	<i>BCOR</i>	LOH	1.1
TSG	<i>EP300</i>	LOH	1.1
TSG	<i>SMARCD1</i>	LOH	0.8
TSG	<i>SDHB</i>	LOH	1.1

TSG	<i>ARID1B</i>	LOH	1
TSG	<i>TNFAIP3</i>	LOH	1
TSG	<i>APOBEC3B</i>	LOH	1.1
TSG	<i>PRDM1</i>	LOH	1

dMMR: Deficient mismatch repair

HRD: Homologous recombination deficiency

OG: Oncogene

TSG: Tumor suppressor gene