

Supplementary Material: Utility of Circulating Tumor DNA for Detection and Monitoring of Endometrial Cancer Recurrence and Progression

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Table S1. Genomic coordinates for personalised ctDNA panel and target mutations derived from whole exome sequencing of the primary tumor.

Chromosome	Start	End	Target
chr1	37984113	37984293	SF3A3 p.C92S_1,SF3A3 p.C92S_2
chr2	47414258	47414558	BAT-26_1,BAT-26_2,BAT-26_3,BAT-26_4
chr2	95183472	95183653	NR-24_1, NR-24_2, NR-24_3, NR-24_4
chr3	195783832	195784012	MUC4 p.P2562S_MUC4 p.S2544P_1,MUC4 p.P2562S_MUC4 p.S2544P_2
chr3	195784551	195784731	MUC4 p.A2313V_1,MUC4 p.A2313V_2
chr4	54731896	54732196	BAT-25_1,BAT-25_2,BAT-25_3,BAT-25_4
chr6	32221013	32221193	NOTCH4 p.G225R_1,NOTCH4 p.G225R_2
chr7	100952460	100952640	MUC3A p.P258S_1,MUC3A p.P258S_2
chr7	100953384	100953564	MUC3A p.H563Q_MUC3Ap.S568Y_1,MUC3A p.H563Q_MUC3Ap.S568Y_2
chr9	8375930	8376110	PTPRD p.E1119K_1,PTPRD p.E1119K_2
chr10	87933057	87933237	PTEN p.R130Q_PTEN p.R130L_1,PTEN p.R130Q_PTEN p.R130L_2
chr10	121520072	121520252	FGFR2 p.S252W_1,FGFR2 p.S252W_2
chr11	1017234	1017414	MUC6 p.Y1826D_1,MUC6 p.Y1826D_2
chr11	1017562	1017764	MUC6 p.P1716S_1,MUC6 p.P1716S_2,MUC6 p.P1716S_3
chr11	14358713	14358893	RRAS p.G23C_1,RRAS p.G23C_2
chr11	46898858	46899038	LRP4 p.D211H_1,LRP4 p.D211H_2
chr11	102322679	102322919	NR-27_1, NR-27_2, NR-27_3
chr13	48459617	48459797	RB1 p.R661W_1, RB1 p.R661W_2
chr14	23182997	23183297	NR-21_1, NR-21_2, NR-21_3, NR-21_4
chr17	43091512	43091692	BRCA1 p.T1263S_1, BRCA1 p.T1263S_2
chr19	8888681	8888861	MUC16 p.S13577G_MUC16 p.H13576N_1,MUC16 p.S13577G_MUC16 p.H13576N_2
chr19	52212627	52212807	PPP2R1A p.P179R_1,PPP2R1A p.P179R_2

Table S2. The somatic mutations identified in the matched tumour-normal patient samples analysed (*n* = 13).

Patient ID	ctDNA Detected?	Mutations Detected in Tumor	%VAF in Tumour	Mutation(s) detected in ctDNA (Any Timepoint)	Timepoint Detected (%VAF)	Total Timepoints
1 ^Δ	Y *	<i>PTEN</i> p.R130Q	14.3	<i>MUC4</i> p.H2306P	3 (9.57%), 5 (7.34%)	
		<i>MUC16</i> p.H13577G	14.6	MSI markers	3 (1/5 markers), 4 (1/5 markers), 6 (3/5 MSI markers)	
		<i>MUC4</i> p.S2544P	26.2			
		MSI markers	5/5 detected			6
2 ^Δ	Y	<i>MUC4</i> p.A2313V	16	<i>MUC4</i> p.A2313V	2 (42.3%), 3 (54.35%)	
		<i>PTEN</i> p.R130L	19.3	MSI markers	3 (2/5 MSI markers)	
		MSI markers	None			3
3 ^Δ	Y *	<i>PTPRD</i> p.E1119K	20.3	<i>MUC4</i> p.H2290L	1 (9.48%)	
		<i>MUC6</i> p.P1716S	20.7	<i>MUC4</i> p.A2313V	1 (19.48%)	
		<i>SF3A3</i> p.C92S	21.2			
		<i>MUC4</i> p.P2562S	24.8			
		<i>PPP2R1A</i> p.P179R	27.3			
		<i>MUC6</i> p.Y1826D	37.5			
		<i>LRP4</i> p.D211H	55.2			
		<i>RRAS</i> p.G23C	58.3			
		<i>PIK3CA</i> p.H1047L	18.53	<i>PIK3CA</i> p.E545A	1 (0.16%)	
		<i>KRAS</i> p.G12A	20.67	<i>PIK3CA</i> p.H1047L	1 (2.19%)	
4	Y [±]			<i>KRAS</i> p.G12A	1 (2.12%)	
				<i>CTNNB1</i> p.T41A	1 (2.29%)	
		<i>TP53</i> p.Y220C (primary tumor and 2 metastatic sites)	91.59 (primary tumour), 80.91 (ovary), 69.63 (omentum)	<i>TP53</i> p.Y220C	1 (19.05%), 4 (2.72%), 5 (4.3%), 6 (6.76%), 7 (14.3%)	
				<i>AKT</i> p.E17K	4 (3.05%)	7
6	Y [±]	<i>PIK3CA</i> p.H1047R	59.65	<i>PIK3CA</i> p.H1047R	1 (1.18%)	
		<i>PTEN</i> p.Y155C	33.58	<i>ESR1</i> p.D538G	1 (2.75%)	1
7	Y	<i>TP53</i> p.R273C	85.39	<i>TP53</i> p.R273C	1 (1.53%)	
		<i>BRCA1</i> p.T1263S	14.9	<i>MUC4</i> p.A2313V	1 (10.56%)	
8 ^Δ	Y [±]	<i>MUC16</i> p.H13576N	15.7	<i>NOTCH4</i> p.G225R	1 (50.59%)	
		<i>MUC3A</i> p.H563Q	20.2			
		<i>RB1</i> p.R661W	21.4			
		<i>NOTCH4</i> p.G225R	21.5			
						1

			<i>PIK3CA</i> p.H1047R (primary and relapse)	27.94 (primary tumour), 35.33 (relapse biopsy)	<i>PIK3CA</i> p.H1047R	1 (3.03%)	
9	Y [‡]				<i>MET</i> p.T1010I	1 (16.71%)	3
					<i>FGFR2</i> p.S252W	1 (2.15%)	
					<i>KRAS</i> p.G12A	1 (1.89%)	
10	Y *		<i>KRAS</i> p.G12A	22.78	<i>PIK3CA</i> p.E545A	1 (0.43%)	1
			<i>PIK3CA</i> p.G1049R	12.6			
11	N		<i>PIK3CA</i> p.G106V	35.14	None detected	-	3
12	N		<i>PIK3CA</i> p.R88Q	18.32	None detected (<i>KRAS</i> p.G12D)	-	1
			<i>KRAS</i> p.G12D	23.99			
13	Y *		<i>PIK3CA</i> p.E545A	38.09	<i>TP53</i> p.S241F	1 (0.28%)	1
			<i>TP53</i> p.C242F	40			

[‡] Patient's whose tumor underwent whole exome sequencing. * ctDNA positive patient where mutation in ctDNA did not correlate with tumor. [‡] Additional mutations detected in ctDNA. Note that mutations in bold were validated by ddPCR. For patient 1, mutation was also detected at timepoint 2 but not called due to poor sequencing over the locus.