

Article

Ultra-Deep Sequencing of Plasma-Circulating DNA for the Detection of Tumor-Derived Mutations in Patients with Early-Stage Colorectal Cancer

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Supplementary Materials

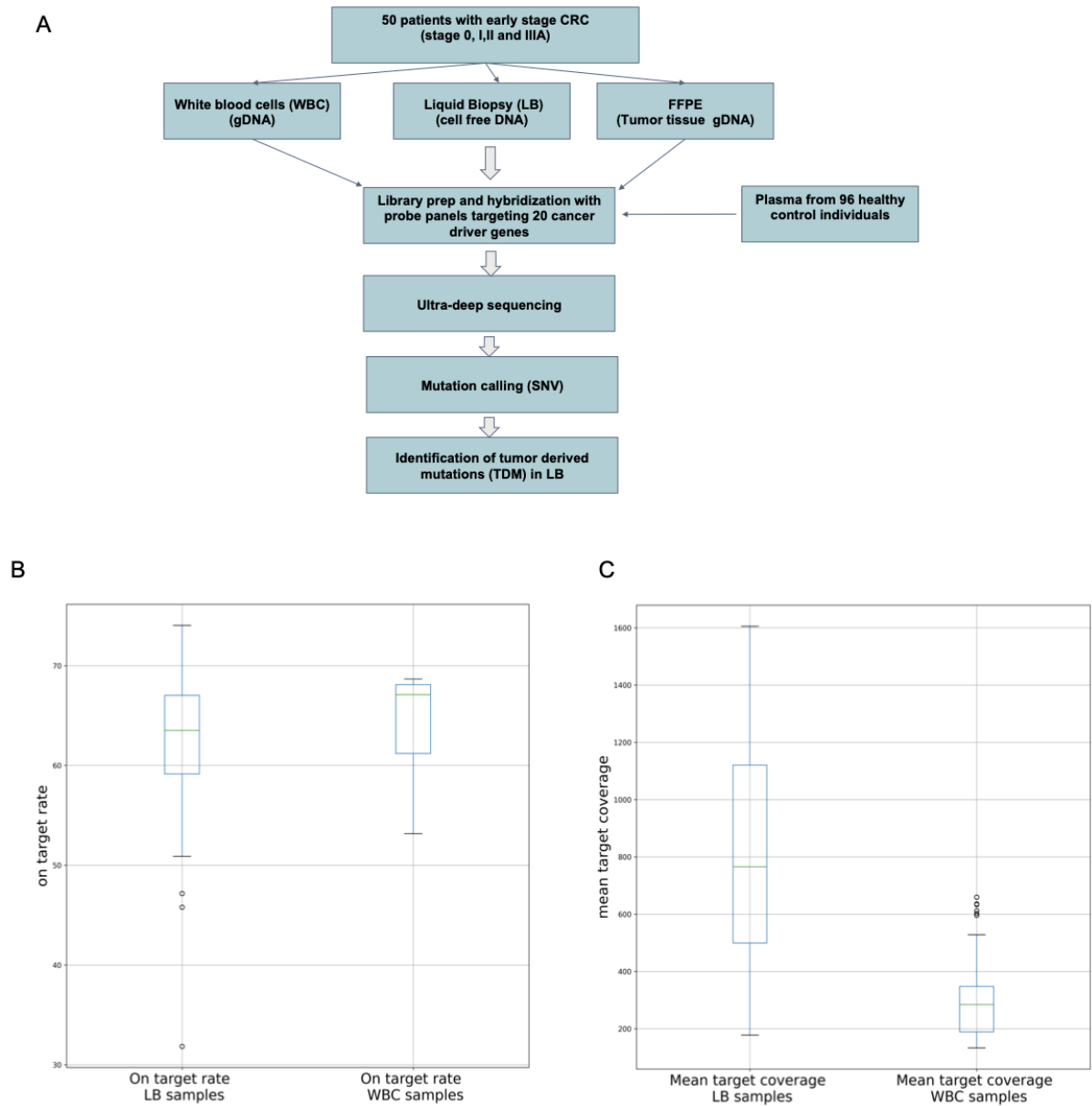


Figure S1: Study design and performance of target sequencing using unique molecular identifiers

A: study design

B and C: On-target rate (B) and mean depth coverage (C) of all 50 paired liquid biopsy cfDNA and WBC gDNA samples

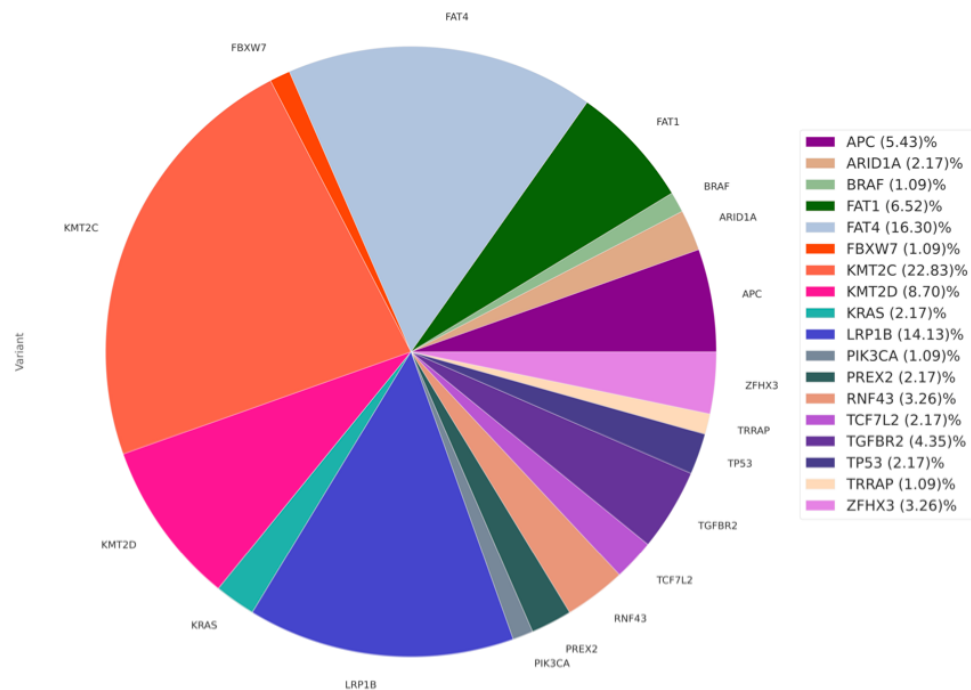


Figure S2: Gene distribution of white blood cell-derived mutations (WDMs) in liquid biopsies (LBMs).

Table S1 Patients' clinical characteristics

Sample ID	Patient ID	Age	Gender	Stage	Tumor location	Tumor size (cm)
1	CRCB2	52	F	I	Rectum	3.5
2	CRCB3	68	F	II	colon	7
3	CRCB4	59	M	0	colon	3
4	CRCB5	64	M	N/A	colon	2
5	CRCB6	60	M	N/A	Rectum	6
6	CRCB9	66	M	III	Rectum	7
7	CRCB11	78	F	I	Rectum	3
8	CRCB14	57	F	III	colon	9
9	CRCB15	79	F	II	colon	4
10	CRCB16	45	M	III	colon	4
11	CRCB17	59	M	III	colon	7
12	CRCB18	58	F	II	colon	4
13	CRCB19	47	F	III	colon	4
14	CRCB20	66	M	II	colon	3
15	CRCB22	60	M	II	colon	4
16	CRCB23	44	M	III	Anacanal	4
17	CRCB25	70	M	III	Rectum	1.5
18	CRCB27	85	M	III	Colon	4

19	CRCB28	81	M	III	colon	6
20	CRCB29	76	F	II	colon	5
21	CRCB30	63	M	II	Colorectum	7
22	CRCB31	66	M	II	Rectum	4.5
23	CRCB32	80	F	III	Colon	3
24	CRCB34	61	M	I	Rectum	1.5
25	CRCB35	68	M	I	Colon	NA
26	CRCB36	52	M	II	Colon	5
27	CRCB37	63	F	I	Colon	2.5
28	CRCB38	45	F	III	Anacanal	2
29	CRCB40	47	M	III	Rectum	3.5
30	CRCB41	63	M	III	Colon	6
31	CRCB42	48	M	II	Colon	4
32	CRCB43	61	M	II	Colon	5
33	CRCB45	55	M	III	Colon	4
34	CRCB46	74	M	II	Colon	5
35	CRCB47	77	M	III	Colon	2
36	CRCB48	61	F	III	Colon	3
37	CRCB49	70	M	II	Colon	4.5
38	CRCB50	57	M	II	Colon	4
39	CRCB51	73	F	II	Colon	3.5
40	CRCB52	63	M	II	Colon	5.5
41	CRCB53	82	F	II	Colon	6
42	CRCB54	66	M	II	Colon	6
43	CRCB55	48	M	II	Colon	10
44	CRCB56	57	F	III	Cecum	6
45	CRCB58	59	M	III	Colon	3
46	CRCB59	80	F	III	Rectum	3
47	CRCB60	70	M	0	Colon	2.5
48	CRCB61	52	F	III	Colon	4
49	CRCB62	61	M	II	Rectum	4
50	CRCB63	27	F	III	Cecum	6.5

Table S2 Gene panel

No .	Gene Symbol	Gene ID	Chromosome	start	end	Size	strand
1	<i>APC</i>	NG_008481.4	5	112737888	112846239	108351	positive strand
2	<i>TP53</i>	NG_017013.2	17	7668402	7687538	19136	negative strand
3	<i>FAT4</i>	NG_033865.1	4	125316399	125492932	176533	positive strand
4	<i>LRP1B</i>	NG_051023.1	2	140231423	142131701	1900278	negative strand

5	<i>TGFB2</i>	NG_007490.1	3	30606601	30694142	87541	positive strand
6	<i>FAT1</i>	NG_046994.1	4	186587783	186723855	136072	negative strand
7	<i>BRAF</i>	NG_007873.3	7	140730665	140924928	194263	negative strand
8	<i>KMT2C</i>	NC_000007.14	7	152134925	152436005	301080	negative strand
9	<i>KMT2D</i>	NG_027827.1	12	49018975	49055324	36349	negative strand
10	<i>ARID1A</i>	NG_029965.1	1	26696033	26782110	86077	positive strand
11	<i>TRRAP</i>	NG_030010.1	7	98878490	99013243	134753	positive strand
12	<i>ZFHX3</i>	NG_013211.2	16	72782885	73891871	1108986	negative strand
13	<i>TCF7L2</i>	NG_012631.1	10	112950407	113167493	217086	positive strand
14	<i>KRAS</i>	NG_007524.2	12	25209431	25250803	41372	negative strand
15	<i>PIK3CA</i>	NG_012113.2	3	179148523	179240093	91570	positive strand
16	<i>ACVR2A</i>	NC_000002.12	2	147844517	147930824	86307	positive strand
17	<i>FBXW7</i>	NG_029466.2	4	152320544	152536063	215519	negative strand
18	<i>RNF43</i>	NG_042894.1	17	58353676	58417533	63857	negative strand
19	<i>SMAD4</i>	NG_013013.2	18	51030213	51085039	54826	positive strand
20	<i>PREX2</i>	NG_047022.1	8	67952118	68237030	284912	positive strand

Table S3 Characteristics of TDMs

	Variant	#-occurrences-LB	VAFLB	Clinical Significance
1	chr7:152163781-G>A	1	0.0136	-
2	chr12:25245328-C>A	1	0.0054	likely_pathogenic
3	chr4:152324239-A>C	1	0.0061	-
4	chr5:112821953-C>A	1	0.012	Pathogenic
5	chr12:49033896-C>T	15	0.003673333	-
6	chr5:112819074-C>T	1	0.0014	Pathogenic
7	chr12:49032701-G>A	1	0.0009	-
8	chr16:72959263-G>A	1	0.011	-

9	chr12:49026 677-G>A	1	0.0009	-
10	chr3:179218 307-A>G	1	0.0241	pathogenic,likely_pathogenic
11	chr17:76748 94-G>A	2	0.0358	Pathogenic
12	chr5:112839 714-G>T	1	0.0155	-
13	chr5:112839 196-C>A	1	0.0099	likely_pathogenic
14	chr5:112839 954-A>G	1	0.0015	benign/likely_benign,likely_benign,benign,uncertain_s ignificance
15	chr5:112838 086-T>C	1	0.0028	uncertain_significance
16	chr17:76748 85-C>T	1	0.0225	likely_pathogenic, pathogenic/likely_pathogenic,uncertain_significance,p athogenic
17	chr17:76751 39-C>T	1	0.004	pathogenic,pathogenic/likely_pathogenic
18	chr16:72794 653-C>T	1	0.0096	-
19	chr18:51048 839-C>T	1	0.0088	Pathogenic
20	chr7:152235 864-C>A	1	0.0045	-
21	chr5:112827 194-C>T	1	0.0087	Pathogenic
22	chr4:152328 233-G>A	2	0.0104	likely_pathogenic
23	chr3:179199 088-G>A	1	0.0176	pathogenic,likely_pathogenic
24	chr5:112840 311-G>T	1	0.0072	-
25	chr4:152326 214-C>T	1	0.0053	likely_pathogenic
26	chr5:112839 444-G>T	1	0.0099	-
27	chr17:76750 88-C>T	2	0.0152 5	likely_pathogenic,not_provided,pathogenic
28	chr5:112839 558-G>T	1	0.0202	-
29	chr5:112839 510-G>T	1	0.0125	likely_pathogenic,pathogenic
30	chr7:152139 719-G>A	1	0.0223	-
31	chr18:51065 518-G>A	1	0.0052	pathogenic
32	chr5:112828 889-C>T	1	0.0062	pathogenic
33	chr3:179210 192-T>C	1	0.0191	pathogenic,likely_pathogenic