

Revisiting the implications of positive germline testing results using multi-gene panels in a cohort of individuals with personal and/or family history of Breast cancer

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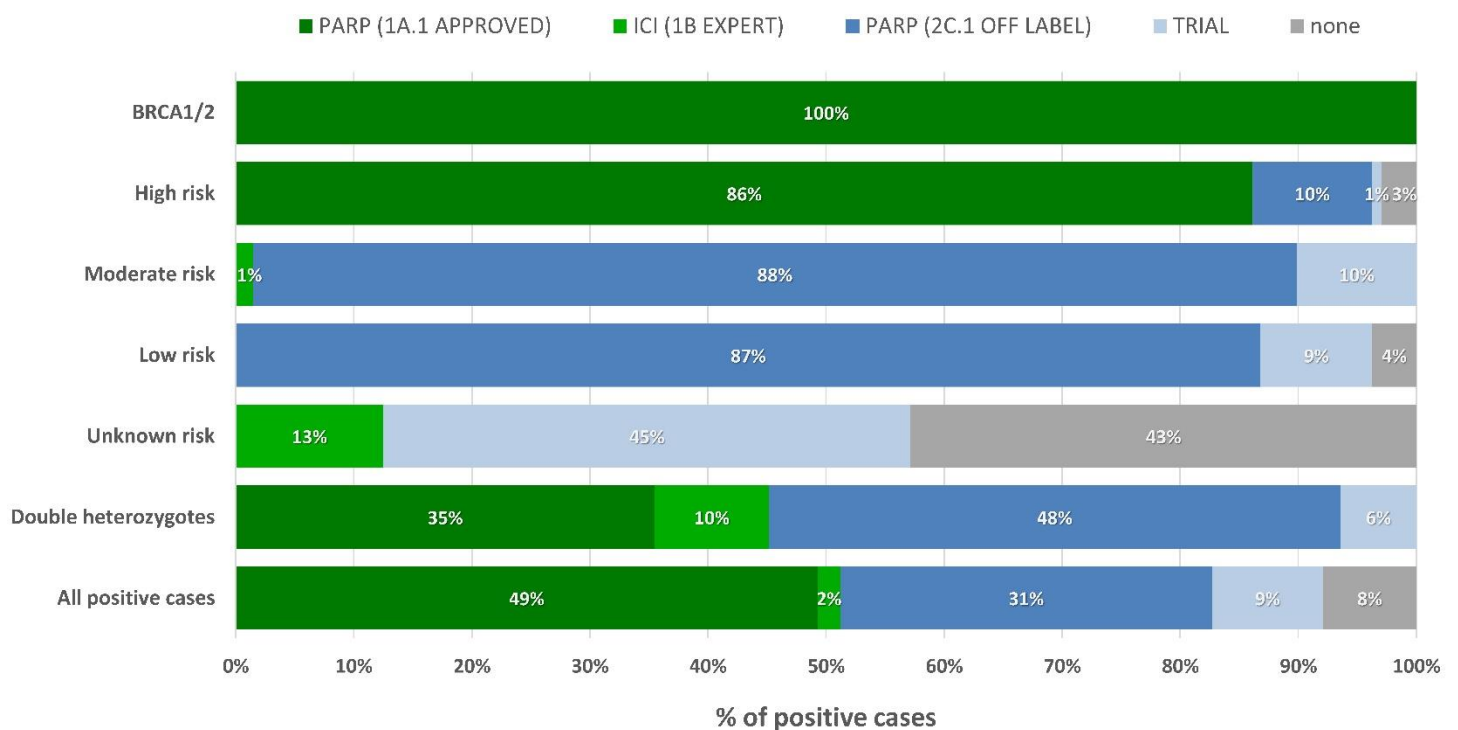


Figure S1. Treatment implications of germline findings in the 415 individuals with P/LP variants grouped by the level of breast cancer risk.

Table S1. Clinical utility and different levels of information associated with pathogenic/likely pathogenic variants in genes tested positive in our cohort.

Breast cancer risk					Other cancer risk				Treatment actionability	
Gene	Cases	Risk	Evidence	Management	Type	Risk	Evidence	Management	Category	Evidence
<i>BRCA1</i>	136	High (>60%)	Very strong	RRS, Surveillance	Ovarian	High (>39-58%)	Very strong	RRS, Surveillance	PARPi	Approved
					Pancreatic	Moderate (<5%)	Strong	Surveillance	PARPi	Approved
					Prostate	Increased	Limited	Surveillance	PARPi	Approved
									PARPi	Approved
<i>BRCA2</i>	69	High (>60%)	Very strong	RRS, Surveillance	Ovarian	High (>39-58%)	Very strong	RRS, Surveillance	PARPi	Approved
					Pancreatic	Moderate (5-10%)	Strong	Surveillance	PARPi	Approved
					Prostate	Low	Strong	Surveillance	PARPi	Approved
					Melanoma	Increased	Limited	Surveillance	PARPi	Off label/Clinical trials
<i>PALB2</i>	24	High (41-60%)	Strong	RRS, Surveillance	Ovarian	Increased (3-5%)	Strong	RRS, Surveillance	PARPi	Off label/Clinical trials
					Pancreatic	Moderate (5-10%)	Limited	Surveillance	PARPi	Off label/Clinical trials
<i>PTEN</i>	2	High (>60%)	Strong	RRS, Surveillance	Colorectal	Increased	Strong	Surveillance	-	-
					Endometrial	High (19-28%)	Strong	RRS, Surveillance	-	-
					Melanoma	Increased	Limited	Surveillance	-	-
					Renal cell	Increased	Limited	Surveillance	-	-
					Thyroid	Increased	Limited	Surveillance	-	-
<i>TP53</i>	7	High (>60%)	Strong	RRS, Surveillance	Pancreatic	Moderate (5-10%)	Limited	Surveillance	-	-
					Colorectal	Increased	Strong	Surveillance	-	-
					Melanoma	Increased	Limited	Surveillance	-	-
					Brain	Increased	Limited	Surveillance	-	-
					Multiple	Increased	Limited	Surveillance	-	-
<i>ATM</i>	27	Moderate (15-40%)	Strong	Surveillance	Ovarian	Low (<3%)	Strong	Surveillance	PARPi	Off label/Clinical trials
					Pancreatic	Moderate (5-10%)	Strong	Surveillance	PARPi	Off label/Clinical trials
					Prostate	Unknown	Insufficient	-	PARPi	Approved
					Colorectal	Unknown	Limited	-	PARPi	Off label/Clinical trials
<i>CHEK2</i> *	39	Moderate (15-40%)	Strong	Surveillance	Colorectal	Increased	Limited	Surveillance	PARPi	Off label/Clinical trials
					Ovarian	Unknown	Insufficient	-	PARPi	Off label/Clinical trials
					Prostate	Increased	Limited	-	PARPi	Approved
<i>NBN</i> **	7	Moderate	Limited	based on FH	Ovarian	Unknown	Insufficient	-	PARPi	Clinical trials
					Prostate	Increased	Limited	-	PARPi	Off label/Clinical trials
<i>BARD1</i>	7	Low	Limited	Surveillance	Ovarian	Potentially Increased	Limited	-	PARPi	Off label/Clinical trials
									PARPi	Off label/Clinical trials
<i>BRIP1</i>	7	Low	Limited	based on FH	Ovarian	High (>10%)	Strong	RRS, Surveillance	PARPi	Off label/Clinical trials
					Prostate	Potentially Increased	Limited	-	PARPi	Approved

Table S1. Clinical utility and different levels of information associated with pathogenic /likely pathogenic variants in genes tested positive in our cohort (continued).

Gene	Breast cancer risk				Other cancer risk				Treatment actionability	
	Cases	Risk	Evidence	Management	Type	Risk	Evidence	Management	Category	Evidence
<i>MLH1</i>	1	Low (<15%)	Limited	based on FH	Ovarian	High (4-20%)	Strong	RRS, Surveillance	ICI	Well-powered studies
					Colorectal	High (46-61%)	Strong	Surveillance	ICI	Well-powered studies
					Endometrial	High (34-54%)	Strong	RRS, Surveillance	ICI	Well-powered studies
					Gastric	High (5-7%)	Strong	Surveillance	ICI	Well-powered studies
					Pancreatic	Low (5%)	Limited	Surveillance	ICI	Well-powered studies
					Prostate	Low (4-12%)	Limited	-	ICI	Well-powered studies
<i>MSH2</i>	1	Low (<15%)	Limited	based on FH	Ovarian	High (>10%)	Strong	RRS, Surveillance	ICI	Well-powered studies
					Colorectal	High (33-52%)	Strong	Surveillance	ICI	Well-powered studies
					Endometrial	High (21-57%)	Strong	RRS, Surveillance	ICI	Well-powered studies
					Gastric	High (<9%)	Strong	Surveillance	ICI	Well-powered studies
					Pancreatic	Low (1-2%)	Limited	Surveillance	ICI	Well-powered studies
					Prostate	Low (4-16%)	Limited	-	ICI	Well-powered studies
<i>MSH6</i>	3	Low (<15%)	Limited	based on FH	Ovarian	High (>10%)	Strong	RRS, Surveillance	ICI	Well-powered studies
					Colorectal	High (10-44%)	Strong	Surveillance	ICI	Well-powered studies
					Endometrial	High (16-49%)	Strong	RRS, Surveillance	ICI	Well-powered studies
					Gastric	High (1-8%)	Strong	Surveillance	ICI	Well-powered studies
					Pancreatic	Low (1-2%)	Limited	Surveillance	ICI	Well-powered studies
					Prostate	Low (3-12%)	Limited	-	ICI	Well-powered studies
<i>NF1</i>	2	Low (15-40%)	Strong	Surveillance	Brain, MPNST	High (14-22%)	Limited	Surveillance	-	-
<i>RAD51C</i>	3	Low (15-40%)	Limited	based on FH	Ovarian	High (>10%)	Strong	RRS, Surveillance	PARPi	Off label/Clinical trials
<i>BLM</i>	2	Unknown	Insufficient	-					PARPi	Clinical trials
					Colorectal	Unknown	Limited	-	PARPi	Clinical trials
<i>ABRAXAS1 (FAM175A)</i>	1	Unknown	Insufficient	-					-	-
<i>FANCA</i>	4	Unknown	Insufficient	-	Ovarian	Unknown	Insufficient	-	PARPi	Off label/Clinical trials
					Prostate	Unknown	Insufficient	-	PARPi	Clinical trials
									PARPi	Off label/Clinical trials
<i>FANCM</i>	4	Unknown	Insufficient	-	Ovarian	Unknown	Insufficient	-	PARPi	Off label/Clinical trials
					Prostate	Unknown	Insufficient	-	PARPi	Clinical trials
<i>MRE11</i>	1	Unknown	Insufficient	based on FH					PARPi	Off label/Clinical trials
					Prostate	Unknown	Insufficient	-	PARPi	Clinical trials
<i>MUTYH</i> monoallelic	30	Unknown	Insufficient	based on FH	Colorectal	Increased	Limited	Surveillance	-	-
					Endometrial	Potentially Increased	Limited	-	-	-
					Gastric	Increased	Limited	-	-	-

Table S1. Clinical utility and different levels of information associated with pathogenic /likely pathogenic variants in genes tested positive in our cohort (continued).

Gene	Breast cancer risk				Other cancer risk				Treatment actionability	
	Cases	Risk	Evidence	Management	Type	Risk	Evidence	Management	Category	Evidence
<i>PMS2</i>	9	Unknown/Low(<12%)	Insufficient	based on FH	Ovarian	Low (1-2%)	Limited	RRS, Surveillance	ICI	1B
					Colorectal	High (9-20%)	Strong	Surveillance	ICI	1B
					Endometrial	High (13-26%)	Strong	RRS, Surveillance	ICI	1B
					Gastric	Unknown	Insufficient	-	ICI	1B
					Pancreatic	Unknown	Insufficient	-	ICI	1B
					Prostate	Unknown	Insufficient	-	ICI	1B
<i>RAD50</i>	18	Unknown	Insufficient	based on FH				PARP	Clinical trials	
<i>RAD51B</i>	1	Unknown	Insufficient	-				-	-	
<i>XRCC2</i>	1	Unknown	Insufficient	-				-	-	

RRS: Risk-reducing surgery, FH: Family history, PARPi: poly (ADP-ribose) polymerase inhibitors, ICI: Immune checkpoint inhibitors

* Risk estimates are based on only on frameshift pathogenic/likely pathogenic variants. For the c.470T>C(p.Ile157Thr) variant the risk for breast cancer appears to be lower.

** Breast cancer risks refer only to the c.657del5 variant.

Table S2. Information for the 415 individuals with personal and/or family history of breast cancer (BC) tested positive with a multi-gene panel for pathogenic/likely pathogenic variants. Results for double heterozygotes are indicated with genes and variants separated with a '//

Patient ID	Gene	Variant	Sex	Patient diagnosis	BC risk	Meeting NCCN Selection criteria	Treatment implications
GTBC386	ATM	c.(-30+1_-29-1)_(331+1_332-1)del	F	breast cancer, thyroid cancer	moderate	YES	PARPi (Off-Label)
GTBC116	ATM	c.1564_1565delGA p.Glu522Ilefs*43	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC320	ATM	c.2125-1G>T	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC072	ATM	c.2250G>A p.=	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC183	ATM	c.2250G>A p.=	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC285	ATM	c.3327dupA (p.Ala1110Serfs*12)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC223	ATM	c.3576G>A (p.=)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC247	ATM	c.3576G>A (p.=)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC016	ATM	c.3577-1G>C	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC209	ATM	c.5932G>T, p.(Glu1978*)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC140	ATM	c.5979_5983delTAAAG p.Ser1993Argfs*23	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC293	ATM	c.6628delC (p.Gln2210Serfs*25)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC311	ATM	c.6679C>T (p.Arg2227Cys)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC173	ATM	c.6679C>T p.Arg2227Cys	F	breast cancer	moderate	NO	PARPi (Off-Label)
GTBC069	ATM	c.7630-2A>C	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC305	ATM	c.8122G>A (p.Asp2708Asn)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC278	ATM	c.8147T>C (p.Val2716Ala)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC382	ATM	c.8585-2A>C	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC316	ATM	c.8766dupT (p.Val2923Cysfs*2)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC161	ATM	c.8988-1G>C	F	breast cancer	moderate	NO	PARPi (Off-Label)
GTBC336	ATM/CHEK2	c.4002_4004delinsTTGCAGATTG (p.Leu1335Cysfs*6) / c.1100delC (p.Thr367Metfs*15)	F	breast cancer	moderate+moderate	YES	PARPi (Off-Label)
GTBC379	ATM/CHEK2	c.5932G>T (p.Glu1978Ter) / c.1100delC (p.Thr367Metfs*15)	F	breast cancer	moderate+moderate	NO	PARPi (Off-Label)
GTBC252	ATM/CHEK2	c.9007_9034del (p.Asn3003Aspfs*6) / c.(792+1_793-1)_(846+1_847-1)	F	bilateral breast cancer	moderate+moderate	YES	PARPi (Off-Label)
GTBC358	ATM/FANCA	c.5980A>T (p.Lys1994Ter) / c.1A>G (p.Ser2_Met116del)	F	breast cancer	moderate+unknown	NO	PARPi (Off-Label)
GTBC373	ATM/FANCA	c.6095G>A (p.Arg2032Lys) / c.(893+1_894-1)_(1359+1_1360-1)del	F	breast cancer	moderate+unknown	NO	PARPi (Off-Label)
GTBC219	ATM/MUTYH	c.5318delA (p.Lys1773Serfs*3) / c.721C>T (p.Arg241Trp)	F	breast cancer	moderate+unknown	YES	PARPi (Off-Label)
GTBC310	ATM/RAD51C	c.7542T>A (p.Tyr2514Ter) / c.181_182delCT (p.Leu61Alafs*11)	F	breast cancer	moderate+low	YES	PARPi (Off-Label)

GTBC284	<i>BARD1</i>	c.1690C>T (p.Gln564Ter)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC044	<i>BARD1</i>	c.1690C>T p.Gln564*	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC172	<i>BARD1</i>	c.1690C>T p.Gln564*	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC193	<i>BARD1</i>	c.1690C>T p.Gln564*/	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC206	<i>BARD1</i>	c.1690C>T, p.(Gln564*)	F	unaffected	low	YES	PARPi (Off-Label)
GTBC288	<i>BARD1</i>	c.632T>A (p.Leu211Ter)	F	uterus cancer, breast cancer	low	NO	PARPi (Off-Label)
GTBC043	<i>BARD1</i>	c.632T>A p.Leu211Ter	F	unaffected	low	YES	PARPi (Off-Label)
GTBC055	<i>BLM/BRCA1</i>	c.1642C>T p.Gln548*/c.3700_3704delGTAAA p.Val1234_Asn1235?fs	F	breast cancer	high+unknown	YES	PARPi (Approved)
GTBC048	<i>BLM/CHEK2</i>	c.1642C>T p.Gln548*/c.1283C>T p.Ser428Phe	F	breast cancer	moderate+unknown	YES	PARPi (Off-Label)
GTBC130	<i>BRCA1</i>	c.(?-19)_(80+1_81-1)del	F	bilateral breast cancer	high	YES	PARPi (Approved)
GTBC154	<i>BRCA1</i>	c.(?-19)_(80+1_81-1)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC210	<i>BRCA1</i>	c.(212+1_213-1)_(670+1_671-1)	F	breast cancer, ovarian cancer	high	YES	PARPi (Approved)
GTBC070	<i>BRCA1</i>	c.(441+1_442-1)_(547+1_548-1)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC037	<i>BRCA1</i>	c.(5075-?_5193+?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC274	<i>BRCA1</i>	c.(5193+1_5194-1)_(5277+1_5278-1)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC235	<i>BRCA1</i>	c.(5406+1_5407-1)_(?*1_?)del	F	unaffected	high	YES	PARPi (Approved)
GTBC324	<i>BRCA1</i>	c.(5406+1_5407-1)_(?*1_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC370	<i>BRCA1</i>	c.(5406+1_5407-1)_(?*1_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC390	<i>BRCA1</i>	c.(5406+1_5407-1)_(?*1_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC395	<i>BRCA1</i>	c.(5406+1_5407-1)_(?*1_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC074	<i>BRCA1</i>	c.(5467+1_5468-1)_(?*1_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC255	<i>BRCA1</i>	c.(5467+1_5468-1)_(?*1_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC299	<i>BRCA1</i>	c.(5467+1_5468-1)_(?*1_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC368	<i>BRCA1</i>	c.(5467+1_5468-1)_(?*1_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC139	<i>BRCA1</i>	c.(80+1_81-1)_(134+1_135-1)del	F	bilateral breast cancer	high	YES	PARPi (Approved)
GTBC269	<i>BRCA1</i>	c.(80+1_81-1)_(441+1_442-1)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC071	<i>BRCA1</i>	c.1504_1508delTTAAA p.Leu502Alafs*2	F	breast cancer	high	YES	PARPi (Approved)
GTBC360	<i>BRCA1</i>	c.1612C>T (p.Gln538Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC077	<i>BRCA1</i>	c.1714G>T p.Glu572Ter	F	breast cancer	high	YES	PARPi (Approved)
GTBC053	<i>BRCA1</i>	c.1789G>T p.Glu597*	F	ovarian cancer+breast cancer	high	YES	PARPi (Approved)
GTBC218	<i>BRCA1</i>	c.181T>G (p.Cys61Gly)	F	breast cancer, ovarian cancer	high	YES	PARPi (Approved)
GTBC276	<i>BRCA1</i>	c.181T>G (p.Cys61Gly)	F	breast cancer	high	NO	PARPi (Approved)
GTBC325	<i>BRCA1</i>	c.181T>G (p.Cys61Gly)	F	breast cancer	high	YES	PARPi (Approved)

GTBC363	BRCA1	c.181T>G (p.Cys61Gly)	F	breast cancer	high	YES	PARPi (Approved)
GTBC042	BRCA1	c.181T>G p.Cys61Gly	F	breast cancer	high	YES	PARPi (Approved)
GTBC049	BRCA1	c.181T>G p.Cys61Gly	F	breast cancer	high	YES	PARPi (Approved)
GTBC063	BRCA1	c.181T>G p.Cys61Gly	F	breast cancer	high	YES	PARPi (Approved)
GTBC266	BRCA1	c.2019del (p.Glu673fs)	F	breast cancer	high	YES	PARPi (Approved)
GTBC066	BRCA1	c.2241dupC p.Lys748Glnfs*14	F	breast cancer	high	YES	PARPi (Approved)
GTBC137	BRCA1	c.2504dupA p.His835Glnfs*2	F	unaffected	high	YES	PARPi (Approved)
GTBC163	BRCA1	c.2681_2682delAA p.Lys894Thrfs*8	F	Breast cancer, Ovarian cancer, Lung cancer	high	YES	PARPi (Approved)
GTBC176	BRCA1	c.2761C>T p.Gln921*	F	breast cancer	high	YES	PARPi (Approved)
GTBC367	BRCA1	c.2921+1G>A	F	breast cancer	high	YES	PARPi (Approved)
GTBC004	BRCA1	c.3607C>T (p.Arg1203*)	F	breast cancer	high	YES	PARPi (Approved)
GTBC019	BRCA1	c.3607C>T (p.Arg1203*)	F	breast cancer	high	YES	PARPi (Approved)
GTBC008	BRCA1	c.3607C>T (p.Arg1203*)	F	unaffected	high	YES	PARPi (Approved)
GTBC340	BRCA1	c.3607C>T (p.Arg1203Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC350	BRCA1	c.3607C>T (p.Arg1203Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC415	BRCA1	c.3607C>T (p.Arg1203Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC047	BRCA1	c.3607C>T p.Arg1203*	F	breast cancer	high	YES	PARPi (Approved)
GTBC061	BRCA1	c.3607C>T p.Arg1203*	F	breast cancer	high	YES	PARPi (Approved)
GTBC081	BRCA1	c.3607C>T p.Arg1203*	F	breast cancer	high	YES	PARPi (Approved)
GTBC099	BRCA1	c.3700_3704delGTAAA p.Val1234_Asn1235?fs	F	breast cancer	high	YES	PARPi (Approved)
GTBC101	BRCA1	c.3756_3759delGTCT p.Ser1253Argfs*10	F	breast cancer	high	YES	PARPi (Approved)
GTBC244	BRCA1	c.3770_3771delAG (p.Glu1257Glyfs*9)	F	breast cancer	high	YES	PARPi (Approved)
GTBC040	BRCA1	c.4035delA p.Glu1346Lysfs*20	F	breast cancer	high	NO	PARPi (Approved)
GTBC088	BRCA1	c.4035delA p.Glu1346Lysfs*20	F	breast cancer	high	YES	PARPi (Approved)
GTBC017	BRCA1	c.4372_4373delCA p.Gln1458Glufs*3	F	breast cancer	high	YES	PARPi (Approved)
GTBC408	BRCA1	c.4391_4393delinsTT (p.Pro1464Leufs*2)	F	breast cancer	high	YES	PARPi (Approved)
GTBC036	BRCA1	c.4391_4393delinsTT p.Pro1464Leufs*2	F	breast cancer	high	YES	PARPi (Approved)
GTBC175	BRCA1	c.4391_4393delinsTT p.Pro1464Leufs*2	F	ovarian cancer, breast cancer	high	NO	PARPi (Approved)
GTBC052	BRCA1	c.4484+1G>A	F	breast cancer	high	YES	PARPi (Approved)
GTBC062	BRCA1	c.4487C>A p.Ser1496*	F	breast cancer	high	YES	PARPi (Approved)
GTBC409	BRCA1	c.4576G>T (p.Glu1526Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC337	BRCA1	c.4689C>G (p.Tyr1563Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC120	BRCA1	c.4818delA p.Val1607Leufs*26	F	breast cancer	high	YES	PARPi (Approved)
GTBC313	BRCA1	c.493_494delCT (p.Leu165Glufs*16)	F	breast cancer	high	YES	PARPi (Approved)
GTBC132	BRCA1	c.4986+6T>C	F	breast cancer	high	YES	PARPi (Approved)

GTBC289	BRCA1	c.5030_5033delCTAA (p.Thr1677Ilefs*2)	F	breast cancer	high	YES	PARPi (Approved)
GTBC127	BRCA1	c.5075-1G>A	F	breast cancer	high	YES	PARPi (Approved)
GTBC398	BRCA1	c.5123C>A (p.Ala1708Glu)	F	breast cancer	high	YES	PARPi (Approved)
GTBC082	BRCA1	c.5194-10_5236dup, p.(His1746Leufs*37)	F	unaffected	high	YES	PARPi (Approved)
GTBC231	BRCA1	c.5212G>A (p.Gly1738Arg)	F	breast cancer	high	YES	PARPi (Approved)
GTBC307	BRCA1	c.5212G>A (p.Gly1738Arg)	F	breast cancer	high	YES	PARPi (Approved)
GTBC366	BRCA1	c.5212G>A (p.Gly1738Arg)	F	breast cancer	high	YES	PARPi (Approved)
GTBC399	BRCA1	c.5212G>A (p.Gly1738Arg)	F	breast cancer	high	YES	PARPi (Approved)
GTBC107	BRCA1	c.5212G>A p.Gly1738Arg	F	endometrial cancer+ breast cancer	high	YES	PARPi (Approved)
GTBC135	BRCA1	c.5212G>A p.Gly1738Arg	F	breast cancer	high	YES	PARPi (Approved)
GTBC136	BRCA1	c.5212G>A p.Gly1738Arg	F	breast cancer	high	YES	PARPi (Approved)
GTBC151	BRCA1	c.5212G>A p.Gly1738Arg	F	bilateral breast cancer	high	YES	PARPi (Approved)
GTBC304	BRCA1	c.5251C>T (p.Arg1751Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC345	BRCA1	c.5251C>T (p.Arg1751Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC357	BRCA1	c.5251C>T (p.Arg1751Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC383	BRCA1	c.5251C>T (p.Arg1751Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC384	BRCA1	c.5251C>T (p.Arg1751Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC396	BRCA1	c.5251C>T (p.Arg1751Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC009	BRCA1	c.5260G>T (p.Glu1754*)	F	breast cancer	high	NO	PARPi (Approved)
GTBC003	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC018	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC045	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC050	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	ovarian cancer+breast cancer	high	YES	PARPi (Approved)
GTBC051	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC064	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC091	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC092	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC093	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC095	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC098	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC103	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC105	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC110	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC128	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC129	BRCA1	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)

GTBC143	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	unaffected	high	YES	PARPi (Approved)
GTBC166	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC184	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC190	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC199	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC208	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC212	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC215	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC225	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC257	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC265	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC287	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer, colon cancer	high	YES	PARPi (Approved)
GTBC290	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC302	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC319	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC326	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC380	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC393	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC410	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC413	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC006	<i>BRCA1</i>	c.5266dupC (p.Gln1756Profs*74)	F	bilateral breast cancer	high	YES	PARPi (Approved)
GTBC201	<i>BRCA1</i>	c.5266dupC p.(Gln1756Profs*74)	F	breast cancer	high	YES	PARPi (Approved)
GTBC260	<i>BRCA1</i>	c.5382delG (p.Glu1794Aspfs*40)	F	breast cancer	high	YES	PARPi (Approved)
GTBC054	<i>BRCA1</i>	c.5468 ?_*?	F	bilateral breast cancer	high	YES	PARPi (Approved)
GTBC033	<i>BRCA1</i>	c.5468 ?_*?del	F	breast cancer	high	YES	PARPi (Approved)
GTBC195	<i>BRCA1</i>	c.68_69delAG (p.Glu23Valfs*17), Exon 2	F	breast cancer	high	YES	PARPi (Approved)
GTBC067	<i>BRCA1</i>	c.68_69delAG p.Glu23Valfs*17	F	breast cancer	high	YES	PARPi (Approved)
GTBC100	<i>BRCA1</i>	c.68_69delAG p.Glu23Valfs*17	F	breast cancer	high	YES	PARPi (Approved)
GTBC239	<i>BRCA1</i>	c.843_846del (p.Ser282fs)	F	breast cancer	high	YES	PARPi (Approved)
GTBC267	<i>BRCA1</i>	c.843_846delCTCA (p.Ser282Tyrfs*15)	F	breast cancer	high	YES	PARPi (Approved)
GTBC152	<i>BRCA1</i>	g.(?_43057046)_(43057141_?)del	F	breast cancer	high	NO	PARPi (Approved)
GTBC224	<i>BRCA1</i>	g.(?_43057046)_(43057141_?)del	F	breast cancer	high	YES	PARPi (Approved)
GTBC280	<i>BRCA1/CHEK2</i>	c.5266dupC (p.Gln1756Profs*74) / c.470T>C (p.Ile157Thr)	F	breast cancer	high+low	YES	PARPi (Approved)
GTBC150	<i>BRCA1/BRIP1</i>	c.5212G>A (p.Gly1738Arg) / c.2947delA (p.Ile983Leufs*2)	F	breast cancer	high+low	YES	PARPi (Approved)

GTBC010	<i>BRCA1/CHEK2</i>	c.181T>G (p.Cys61Gly) /c.470T>C (p.Ile157Thr)	F	breast cancer	high+low	YES	PARPi (Approved)
GTBC349	<i>BRCA1/CHEK2</i>	c.181T>G (p.Cys61Gly) /c.470T>C (p.Ile157Thr)	F	breast cancer	high+low	YES	PARPi (Approved)
GTBC309	<i>BRCA1/CHEK2</i>	c.5266dupC (p.Gln1756Profs*74) / c.470T>C (p.Ile157Thr)	F	breast cancer	high+low	YES	PARPi (Approved)
GTBC030	<i>BRCA1/CHEK2</i>	c.737delT p.Leu246*/c.470T>C (p.Ile157Thr)	F	breast cancer	high+low	YES	PARPi (Approved)
GTBC102	<i>BRCA1/CHEK2</i>	c.843_846delCTCA p.Ser282Tyrfs*15 / c.470T>C (p.Ile157Thr)	F	breast cancer	high+low	YES	PARPi (Approved)
GTBC115	<i>BRCA2</i>	c.10G>T p.Gly4*	F	breast cancer	high	YES	PARPi (Approved)
GTBC179	<i>BRCA2</i>	c.1117C>T p.Gln373*	F	breast cancer	high	NO	PARPi (Approved)
GTBC094	<i>BRCA2</i>	c.1189C>T p.Gln397*	F	breast cancer	high	YES	PARPi (Approved)
GTBC270	<i>BRCA2</i>	c.1310_1313delAAGA (p.Lys437Ilefs*22)	F	breast cancer	high	YES	PARPi (Approved)
GTBC065	<i>BRCA2</i>	c.1389_1390delAG p.Val464Glyfs*3	F	breast cancer	high	NO	PARPi (Approved)
GTBC029	<i>BRCA2</i>	c.1405_1406delGA p.Asp469*	F	breast cancer	high	YES	PARPi (Approved)
GTBC122	<i>BRCA2</i>	c.2095_2096delCA p.Gln699Valfs*8	F	breast cancer	high	YES	PARPi (Approved)
GTBC279	<i>BRCA2</i>	c.2095C>T (p.Gln699Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC321	<i>BRCA2</i>	c.2490_2491insT (p.Val831Cysfs*2)	F	breast cancer	high	YES	PARPi (Approved)
GTBC164	<i>BRCA2</i>	c.2490_2491insT p.Val831Cysfs*2	F	unaffected	high	YES	PARPi (Approved)
GTBC317	<i>BRCA2</i>	c.2808_2811delACAA (p.Ala938Profs*21)	F	breast cancer	high	YES	PARPi (Approved)
GTBC344	<i>BRCA2</i>	c.2808_2811delACAA (p.Ala938Profs*21)	F	breast cancer	high	YES	PARPi (Approved)
GTBC352	<i>BRCA2</i>	c.2808_2811delACAA (p.Ala938Profs*21)	F	breast cancer	high	YES	PARPi (Approved)
GTBC041	<i>BRCA2</i>	c.3975_3978dupTGCT p.Ala1327Cysfs*4	F	breast cancer	high	YES	PARPi (Approved)
GTBC263	<i>BRCA2</i>	c.4140delT (p.Glu1382Lysfs*6)	F	breast cancer	high	YES	PARPi (Approved)
GTBC080	<i>BRCA2</i>	c.4769delA p.Lys1590Serfs*27	F	breast cancer	high	YES	PARPi (Approved)
GTBC339	<i>BRCA2</i>	c.4889C>G (p.Ser1630Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC181	<i>BRCA2</i>	c.4936_4939delGAAA p.Glu1646Glnfs*23	F	unaffected	high	YES	PARPi (Approved)
GTBC236	<i>BRCA2</i>	c.5213_5216delCTTA (p.Thr1738Ilefs*2)	F	breast cancer	high	YES	PARPi (Approved)
GTBC281	<i>BRCA2</i>	c.537dupT (p.Ile180Tyrfs*3)	F	breast cancer	high	YES	PARPi (Approved)
GTBC178	<i>BRCA2</i>	c.537dupT p.Ile180Tyrfs*3	F	unaffected	high	YES	PARPi (Approved)
GTBC189	<i>BRCA2</i>	c.5557dupT p.Cys1853Leufs*5	F	breast cancer	high	YES	PARPi (Approved)
GTBC356	<i>BRCA2</i>	c.5645C>A (p.Ser1882Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC341	<i>BRCA2</i>	c.5796_5797delTA (p.His1932Glnfs*12)	F	breast cancer	high	YES	PARPi (Approved)
GTBC169	<i>BRCA2</i>	c.5925delT p.Cys1975Trpfs*29	F	unaffected	high	YES	PARPi (Approved)
GTBC377	<i>BRCA2</i>	c.6267_6269delinsC (p.Glu2089Aspfs*2)	F	breast cancer	high	NO	PARPi (Approved)
GTBC400	<i>BRCA2</i>	c.6389_6390delTT (p.Phe2130Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC402	<i>BRCA2</i>	c.6466_6469delTCTC (p.Ser2156_Gln2157Asnfs)	F	bilateral breast cancer	high	YES	PARPi (Approved)
GTBC242	<i>BRCA2</i>	c.6490C>T (p.Gln2164Ter)	F	breast cancer	high	YES	PARPi (Approved)
GTBC372	<i>BRCA2</i>	c.6490delC (p.Gln2164Serfs*4)	F	breast cancer	high	YES	PARPi (Approved)

GTBC203	BRCA2	c.6771delC, p.Glu2258Lysfs*22	F	breast cancer	high	YES	PARPi (Approved)
GTBC002	BRCA2	c.6842-?_c.7007+?	F	breast cancer	high	YES	PARPi (Approved)
GTBC282	BRCA2	c.6941delC (p.Thr2314Lysfs)	F	breast cancer	high	YES	PARPi (Approved)
GTBC200	BRCA2	c.7007G>C p.Arg2336Pro	F	breast cancer	high	YES	PARPi (Approved)
GTBC119	BRCA2	c.7209_7212delinsGG p.Lys2404Glyfs*7	F	breast cancer	high	YES	PARPi (Approved)
GTBC023	BRCA2	c.7806-2A>T	F	breast cancer	high	YES	PARPi (Approved)
GTBC391	BRCA2	c.7878G>C (p.Trp2626Cys)	F	breast cancer	high	YES	PARPi (Approved)
GTBC097	BRCA2	c.793+1G>A splicing	F	breast cancer	high	NO	PARPi (Approved)
GTBC158	BRCA2	c.8002A>T p.Arg2668Ter	F	breast cancer	high	YES	PARPi (Approved)
GTBC194	BRCA2	c.8023A>G (p.Ile2675Val), Exon 18	F	breast cancer	high	YES	PARPi (Approved)
GTBC141	BRCA2	c.8087T>A p.Leu2696*	F	breast cancer	high	YES	PARPi (Approved)
GTBC298	BRCA2	c.8164dupA (p.Thr2722Asnfs*8)	M	breast cancer	high	YES	PARPi (Approved)
GTBC412	BRCA2	c.8168A>G (p.Asp2723Gly)	F	breast cancer	high	YES	PARPi (Approved)
GTBC407	BRCA2	c.8331+1G>A	F	breast cancer	high	YES	PARPi (Approved)
GTBC148	BRCA2	c.8548delG p.Glu2850Lysfs*13	M	breast cancer	high	YES	PARPi (Approved)
GTBC291	BRCA2	c.8655dupA (p.Pro2886Thrfs*21)	F	breast cancer	high	YES	PARPi (Approved)
GTBC111	BRCA2	c.8655dupA p.Pro2886Thrfs*21	F	breast cancer	high	YES	PARPi (Approved)
GTBC228	BRCA2	c.8800delC (p.Gln2934Lysfs*3)	F	breast cancer	high	YES	PARPi (Approved)
GTBC273	BRCA2	c.8800delC (p.Gln2934Lysfs*3)	F	breast cancer	high	YES	PARPi (Approved)
GTBC197	BRCA2	c.9097dupA (p.Thr3033Asnfs*11)	F	breast cancer	high	YES	PARPi (Approved)
GTBC312	BRCA2	c.9097dupA (p.Thr3033Asnfs*11)	F	breast cancer	high	YES	PARPi (Approved)
GTBC394	BRCA2	c.9117G>A (p.Pro3039=)	F	breast cancer	high	YES	PARPi (Approved)
GTBC160	BRCA2	c.9154C>T p.Arg3052Trp	F	breast cancer	high	YES	PARPi (Approved)
GTBC214	BRCA2	c.9253delA (p.Thr3085Glnfs*19)	F	breast cancer	high	YES	PARPi (Approved)
GTBC059	BRCA2	c.9317G>A p.Trp3106*	F	breast cancer	high	YES	PARPi (Approved)
GTBC211	BRCA2	c.9371A>T (p.Asn3124Ile)	F	breast cancer	high	YES	PARPi (Approved)
GTBC227	BRCA2	c.9371A>T (p.Asn3124Ile)	F	unaffected	high	YES	PARPi (Approved)
GTBC334	BRCA2	c.9371A>T (p.Asn3124Ile)	F	breast cancer	high	YES	PARPi (Approved)
GTBC348	BRCA2	c.9371A>T (p.Asn3124Ile)	F	breast cancer	high	YES	PARPi (Approved)
GTBC031	BRCA2	c.9371A>T p.Asn3124Ile	F	breast cancer	high	YES	PARPi (Approved)
GTBC108	BRCA2	c.9371A>T p.Asn3124Ile	F	breast cancer	high	YES	PARPi (Approved)
GTBC146	BRCA2	c.9371A>T p.Asn3124Ile	F	breast cancer	high	YES	PARPi (Approved)
GTBC180	BRCA2	c.9371A>T p.Asn3124Ile	F	breast cancer	high	YES	PARPi (Approved)
GTBC221	BRCA2	c.9376C>T (p.Gln3126Ter)	F	unaffected	high	YES	PARPi (Approved)
GTBC277	BRCA2	c.966_970delAGTAA (p.Val323Asnfs*2)	F	breast cancer	high	YES	PARPi (Approved)
GTBC114	BRCA2	c.9682delA p.Ser3228Valfs*21	F	unaffected	high	YES	PARPi (Approved)

GTBC331	<i>BRCA2/CHEK2</i>	c.5946delT (p.Ser1982Argfs*22) / c.(908+1_909-1)_(1095+1_1096-1)del	F	breast cancer	high+moderate	YES	PARPi (Approved)
GTBC159	<i>BRCA2/CHEK2</i>	c.9371A>T p.Asn3124Ile / c.902delT p.Leu301Trpfs*3	F	breast cancer	high+moderate	YES	PARPi (Approved)
GTBC073	<i>BRCA2/MUTYH</i>	c.9154C>T p.Arg3052Trp / c.1187G>A p.Gly396Asp	F	breast cancer	high+unknown	YES	PARPi (Approved)
GTBC365	<i>BRIP1</i>	c.2392C>T (p.Arg798Ter)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC106	<i>BRIP1</i>	c.2392C>T p.Arg798*	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC104	<i>BRIP1</i>	c.298delA p.Met100*	F	unaffected	low	YES	PARPi (Off-Label)
GTBC165	<i>BRIP1</i>	c.747delG p.Thr250Hisfs*24	F	bilateral breast cancer	low	YES	PARPi (Off-Label)
GTBC359	<i>BRIP1</i>	c.761_764delAGCA (p.Lys254Argfs*19)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC338	<i>BRIP1/ MUTYH</i>	c.751C>T (p.Arg251Cys) / c.536A>G (p.Tyr179Cys)	F	breast cancer	low+unknown	YES	PARPi (Off-Label)
GTBC205	<i>CHEK2</i>	c.(908+1_909-1)_(1095+1_1096-1)del	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC246	<i>CHEK2</i>	c.1100delC (p.Thr367Metfs*15)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC262	<i>CHEK2</i>	c.1100delC (p.Thr367Metfs*15)	F	unaffected	moderate	YES	PARPi (Off-Label)
GTBC375	<i>CHEK2</i>	c.1100delC (p.Thr367Metfs*15)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC038	<i>CHEK2</i>	c.1100delC p.Thr367Metfs*15	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC087	<i>CHEK2</i>	c.1164delC p.Thr389Profs*25	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC406	<i>CHEK2</i>	c.1188delT (p.Val397Phefs*17)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC232	<i>CHEK2</i>	c.1232G>A (p.Trp411Ter)	M	unaffected	moderate	YES	PARPi (Off-Label)
GTBC261	<i>CHEK2</i>	c.1232G>A (p.Trp411Ter)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC058	<i>CHEK2</i>	c.1232G>A p.Trp411*	F	bilateral breast cancer	moderate	YES	PARPi (Off-Label)
GTBC157	<i>CHEK2</i>	c.1232G>A p.Trp411*	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC123	<i>CHEK2</i>	c.1427C>T p.Thr476Met	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC124	<i>CHEK2</i>	c.1427C>T p.Thr476Met	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC177	<i>CHEK2</i>	c.1427C>T p.Thr476Met	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC057	<i>CHEK2</i>	c.1461+2T>C	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC078	<i>CHEK2</i>	c.444+1G>A	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC414	<i>CHEK2</i>	c.444+1G>A	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC021	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC024	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	breast cancer	low	NO	PARPi (Off-Label)
GTBC034	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC090	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	unaffected	low	YES	PARPi (Off-Label)
GTBC162	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC167	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC185	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	breast cancer	low	NO	PARPi (Off-Label)
GTBC186	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	bilateral breast cancer	low	YES	PARPi (Off-Label)
GTBC192	<i>CHEK2</i>	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)

GTBC213	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC238	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC241	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC243	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	NO	PARPi (Off-Label)
GTBC254	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC264	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC271	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC275	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC196	CHEK2	c.470T>C (p.Ile157Thr), Exon 4	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC376	CHEK2	c.470T>C, p.(Ile157Thr) L.P	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC389	CHEK2	c.499G>A (p.Gly167Arg)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC112	CHEK2	c.499G>A p.Gly167Arg	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC388	CHEK2	c.507delT (p.Phe169Leufs*2)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC237	CHEK2	c.793-1G>A	F	unaffected	moderate	YES	PARPi (Off-Label)
GTBC001	CHEK2	c.(908+1_909-1)_(1095+1_1096-1)del	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC286	CHEK2	c.(792+1_793-1)_(846+1_847-1)	F	breast cancer	moderate	NO	PARPi (Off-Label)
GTBC355	CHEK2	c.(908+1_909-1)_(1095+1_1096-1)del	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC351	CHEK2	c.(908+1_909-1)_(1095+1_1096-1)del	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC330	CHEK2	c.1188delT (p.Val397Phefs*17)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC329	CHEK2	c.1427C>T (p.Thr476Met)	F	breast cancer	moderate	NO	PARPi (Off-Label)
GTBC294	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC303	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC315	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC327	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	NO	PARPi (Off-Label)
GTBC332	CHEK2	c.470T>C (p.Ile157Thr)	F	bilateral breast cancer	low	YES	PARPi (Off-Label)
GTBC342	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC346	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC283	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC292	CHEK2	c.470T>C (p.Ile157Thr)	F	breast cancer	low	NO	PARPi (Off-Label)
GTBC204	CHEK2	c.470T>C (p.Ile157Thr) Homozygous	F	breast cancer	low*	YES	PARPi (Off-Label)
GTBC306	CHEK2	c.499G>A (p.Gly167Arg)	F	breast cancer, thyroid cancer	moderate	YES	PARPi (Off-Label)
GTBC308	CHEK2	c.499G>A (p.Gly167Arg)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC343	CHEK2	c.499G>A (p.Gly167Arg)	F	breast cancer	moderate	YES	PARPi (Off-Label)
GTBC118	CHEK2/CHEK2	c.470T>C p.Ile157Thr / c.499G>A p.Gly167Arg	F	breast cancer	moderate+low	YES	PARPi (Off-Label)
GTBC013	CHEK2/PMS2	c.1232G>A (p.Trp411*) / c.(?-87)_(?160?)del	F	breast cancer	moderate+low	YES	PARPi (Off-Label) / ICI (Well-poered)

							studies)
GTBC392	<i>CHEK2/RAD50</i>	c.499G>A (p.Gly167Arg) / c.326_329delCAGA (p.Thr109Asnfs*20)	F	breast cancer	moderate+unknown	YES	PARPi (Off-Label)
GTBC075	<i>FAM175A/RAD50</i>	c.1106dupG p.Ser370Ilefs*2 / c.3779_3791dupGTAACITCCAGCT p.Leu1265*	F	breast cancer	unknown+unknown	YES	Clinical Trials
GTBC364	<i>FANCA</i>	c.(1626+1_1627-1)_(2852+1_2853-1)del	F	breast cancer, thyroid cancer	unknown	NO	Clinical Trials
GTBC314	<i>FANCA</i>	c.82G>T (p.Gly28Ter)	F	breast cancer	unknown	YES	Clinical Trials
GTBC347	<i>FANCM</i>	c.4368_4369dupCA (p.Arg1457Thrfs*5)	F	breast cancer	unknown	YES	Clinical Trials
GTBC323	<i>FANCM</i>	c.5791C>T (p.Arg1931Ter)	F	breast cancer	unknown	YES	Clinical Trials
GTBC362	<i>FANCM</i>	c.5791C>T (p.Arg1931Ter)	F	breast cancer	unknown	YES	Clinical Trials
GTBC353	<i>FANCM/NBN</i>	c.5791C>T (p.Arg1931Ter) / c.657_661delACAAA (p.Lys219Asnfs*16)	F	breast cancer	moderate+unknown	YES	Clinical Trials
GTBC027	<i>MLH1</i>	c.199G>A (p.Gly67Arg)	F	breast cancer	LOW	YES	ICI (Well-powered studies)
GTBC378	<i>MRE11</i>	c.1714C>T (p.Arg572Ter)	F	breast cancer	unknown	YES	Clinical Trials
GTBC248	<i>MSH2</i>	c.(1076+1_1077-1)_(1386+1_1387-1)del	F	Colon, Breast	low	YES	ICI (Well-powered studies)
GTBC216	<i>MSH6</i>	c.2764C>T (p.Arg922Ter)	F	unaffected	low	NO	ICI (Well-powered studies)
GTBC171	<i>MSH6</i>	c.3261dupC p.Phe1088Leufs*5	F	breast cancer	low	YES	ICI (Well-powered studies)
GTBC011	<i>MSH6/MUTYH</i>	c.2136delG (p.Asp713Ilefs*23)/c.1187G>A (p.Gly396Asp)	F	breast cancer	low+unknown	YES	ICI (Well-powered studies)
GTBC233	<i>MUTYH</i>	c.1187G>A (p.Gly396Asp)	F	breast cancer	unknown	YES	None
GTBC251	<i>MUTYH</i>	c.1187G>A (p.Gly396Asp)	F	breast cancer	unknown	YES	None
GTBC256	<i>MUTYH</i>	c.1187G>A (p.Gly396Asp)	F	breast cancer	unknown	NO	None
GTBC268	<i>MUTYH</i>	c.1187G>A (p.Gly396Asp)	F	breast cancer	unknown	NO	None
GTBC297	<i>MUTYH</i>	c.1187G>A (p.Gly396Asp)	F	breast cancer	unknown	NO	None
GTBC322	<i>MUTYH</i>	c.1187G>A (p.Gly396Asp)	F	breast cancer	unknown	YES	None
GTBC403	<i>MUTYH</i>	c.1187G>A (p.Gly396Asp)	F	breast cancer	unknown	YES	None
GTBC015	<i>MUTYH</i>	c.1187G>A p.Gly396Asp	F	breast cancer	unknown	YES	None
GTBC230	<i>MUTYH</i>	c.1227_1228dupGG (p.Glu410Glyfs*43)	F	breast cancer	unknown	NO	None
GTBC138	<i>MUTYH</i>	c.1437_1439delGGA	F	breast cancer	unknown	YES	None
GTBC234	<i>MUTYH</i>	c.1437_1439delGGA (p.Glu479_Glu480delinsGlu)	F	Breast, kidney	unknown	YES	None
GTBC300	<i>MUTYH</i>	c.1437_1439delGGA (p.Glu479_Glu480delinsGlu)	F	breast cancer	unknown	YES	None
GTBC354	<i>MUTYH</i>	c.1437_1439delGGA (p.Glu479_Glu480delinsGlu)	F	breast cancer	unknown	YES	None
GTBC220	<i>MUTYH</i>	c.536A>G (p.Tyr179Cys)	F	breast cancer	unknown	YES	None
GTBC385	<i>MUTYH</i>	c.536A>G (p.Tyr179Cys)	F	breast cancer	unknown	YES	None
GTBC401	<i>MUTYH</i>	c.536A>G (p.Tyr179Cys)	F	breast cancer	unknown	NO	None

GTBC039	<i>MUTYH</i>	c.536A>G p.Tyr179Cys	F	breast cancer	unknown	YES	None
GTBC089	<i>MUTYH</i>	c.536A>G p.Tyr179Cys	F	breast cancer	unknown	YES	None
GTBC133	<i>MUTYH</i>	c.536A>G p.Tyr179Cys	F	unaffected	unknown	YES	None
GTBC153	<i>MUTYH</i>	c.536A>G p.Tyr179Cys	F	breast cancer	unknown	YES	None
GTBC191	<i>MUTYH</i>	c.536A>G p.Tyr179Cys	F	breast cancer	unknown	NO	None
GTBC258	<i>MUTYH</i>	c.734G>A (p.Arg245His)	F	breast cancer	unknown	YES	None
GTBC253	<i>MUTYH</i>	c.884C>T (p.Pro295Leu)	F	breast cancer	unknown	YES	None
GTBC207	<i>MUTYH</i>	c.884C>T p.(Pro295Leu)	F	breast cancer	unknown	YES	None
GTBC126	<i>MUTYH/PALB2</i>	c.734G>A p.Arg245His / c.2257C>T p.Arg753*	F	breast cancer	high+unknown	YES	PARPi (Off-Label)
GTBC147	<i>MUTYH/RAD51C</i>	c.536A>G p.Tyr179Cys / c.905-2A>G	F	breast cancer	low+unknown	YES	PARPi (Off-Label)
GTBC084	<i>NBN</i>	c.2140C>T p.Arg714*	F	breast cancer	unknown	YES	Clinical Trials
GTBC245	<i>NBN</i>	c.657_661delACAAA (p.Lys219Asnfs*16)	F	breast cancer	moderate	YES	Clinical Trials
GTBC113	<i>NBN</i>	c.657_661delACAAA (p.Lys219Asnfs*16)	F	breast cancer	moderate	NO	Clinical Trials
GTBC117	<i>NBN</i>	c.657_661delACAAA (p.Lys219Asnfs*16)	F	breast cancer	moderate	YES	Clinical Trials
GTBC125	<i>NBN</i>	c.657_661delACAAA (p.Lys219Asnfs*16)	F	breast cancer	moderate	YES	Clinical Trials
GTBC374	<i>NBN</i>	c.657_661delACAAA (p.Lys219Asnfs*16)	F	breast cancer	moderate	NO	Clinical Trials
GTBC096	<i>NBN/PMS2</i>	c.657_661delACAAA (p.Lys219Asnfs*16) / c.1261C>T p.Arg421*	F	breast cancer	moderate+unknown	YES	ICI (Well-powered studies)
GTBC381	<i>NF1</i>	c.2033dupC (p.Ile679Aspfs*21)	F	breast cancer, ovarian cancer	low	NO	None
GTBC170	<i>NF1</i>	c.2455delC p.His819Metfs*2	F	Breast cancer, NF1	low	YES	None
GTBC226	<i>PALB2</i>	c.1037_1041delAAGAA (p.Lys346Thrfs*13)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC397	<i>PALB2</i>	c.1317dupG (p.Phe440Valfs*2)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC240	<i>PALB2</i>	c.172_175delTTGT (p.Gln60Argfs*7)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC335	<i>PALB2</i>	c.172_175delTTGT (p.Gln60Argfs*7)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC387	<i>PALB2</i>	c.172_175delTTGT (p.Gln60Argfs*7)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC079	<i>PALB2</i>	c.172_175delTTGT p.Gln60Argfs*7	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC121	<i>PALB2</i>	c.2257C>T (p.Arg753Ter)	F	bilateral breast cancer	high	YES	PARPi (Off-Label)
GTBC145	<i>PALB2</i>	c.2257C>T (p.Arg753Ter)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC301	<i>PALB2</i>	c.2257C>T (p.Arg753Ter)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC411	<i>PALB2</i>	c.2257C>T (p.Arg753Ter)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC022	<i>PALB2</i>	c.2747_2748+4delAGGTAA	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC168	<i>PALB2</i>	c.3271C>T p.Gln1091*	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC371	<i>PALB2</i>	c.3340C>T (p.Gln1114Ter)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC028	<i>PALB2</i>	c.3549C>G p.Tyr1183*	F	unaffected	high	YES	PARPi (Off-Label)
GTBC155	<i>PALB2</i>	c.49-1G>A	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC014	<i>PALB2</i>	c.509_510delGA p.Arg170Ilefs*14	F	breast cancer	high	YES	PARPi (Off-Label)

GTBC056	<i>PALB2</i>	c.509_510delGA p.Arg170Ilefs*14	F	breast cancer	high	NO	PARPi (Off-Label)
GTBC026	<i>PALB2</i>	c.79G>T p.Glu29*	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC202	<i>PALB2</i>	c.868G>T p.Glu290*	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC369	<i>PALB2</i>	c.93dupA (p.Leu32Thrfs*11)	F	breast cancer, colon cancer	high	NO	PARPi (Off-Label)
GTBC007	<i>PALB2</i>	c.93dupA (p.Leu32Thrfs*11)	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC046	<i>PALB2</i>	c.93dupA p.Leu32Thrfs*11	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC068	<i>PALB2</i>	c.93dupA p.Leu32Thrfs*11	F	breast cancer	high	YES	PARPi (Off-Label)
GTBC405	<i>PMS2</i>	c.(705+1_706-1)_(2006+1_2007-1)del	F	breast cancer	unknown	NO	ICI (Well-powered studies)
GTBC328	<i>PMS2</i>	c.(903+1_904-1)_(988+1_989-1)del	F	breast cancer, colon cancer	unknown	NO	ICI (Well-powered studies)
GTBC333	<i>PMS2</i>	c.1239dupA (p.Asp414Argfs*44)	F	breast cancer	unknown	YES	ICI (Well-powered studies)
GTBC144	<i>PMS2</i>	c.1239dupA p.Asp414Argfs*44	F	breast cancer	unknown	YES	ICI (Well-powered studies)
GTBC188	<i>PMS2</i>	c.1239dupA p.Asp414Argfs*44	F	breast cancer	unknown	YES	ICI (Well-powered studies)
GTBC005	<i>PMS2</i>	c.1297A>T (p.Lys433*)	F	breast cancer	unknown	YES	ICI (Well-powered studies)
GTBC086	<i>PMS2</i>	c.1414A>T p.Lys472*	F	breast cancer	unknown	YES	ICI (Well-powered studies)
GTBC249	<i>PTEN</i>	c.1117G>T (p.Glu373Ter)	F	bilateral breast cancer	high	YES	Clinical Trials
GTBC109	<i>PTEN</i>	c.389G>T p.Arg130Leu	F	breast cancer	high	YES	Clinical Trials
GTBC187	<i>RAD50</i>	c.1245+1G>A	F	breast cancer	unknown	YES	Clinical Trials
GTBC361	<i>RAD50</i>	c.1636-2A>G	F	breast cancer	unknown	NO	Clinical Trials
GTBC182	<i>RAD50</i>	c.1875C>G p.Tyr625*	F	breast cancer	unknown	YES	Clinical Trials
GTBC012	<i>RAD50</i>	c.2260_2266del (p.Leu754Metfs*9)	F	breast cancer	unknown	NO	Clinical Trials
GTBC174	<i>RAD50</i>	c.3103G>T p.Glu1035*	F	unaffected	unknown	YES	Clinical Trials
GTBC198	<i>RAD50</i>	c.326_329delCAGA (p.Thr109Asnfs*20)	F	breast cancer	unknown	YES	Clinical Trials
GTBC222	<i>RAD50</i>	c.326_329delCAGA (p.Thr109Asnfs*20)	F	breast cancer	unknown	NO	Clinical Trials
GTBC250	<i>RAD50</i>	c.326_329delCAGA (p.Thr109Asnfs*20)	F	unaffected	unknown	YES	Clinical Trials
GTBC272	<i>RAD50</i>	c.326_329delCAGA (p.Thr109Asnfs*20)	F	breast cancer	unknown	YES	Clinical Trials
GTBC296	<i>RAD50</i>	c.326_329delCAGA (p.Thr109Asnfs*20)	F	breast cancer	unknown	YES	Clinical Trials
GTBC318	<i>RAD50</i>	c.326_329delCAGA (p.Thr109Asnfs*20)	F	breast cancer	unknown	YES	Clinical Trials
GTBC035	<i>RAD50</i>	c.326_329delCAGA p.Thr109Asnfs*20	F	breast cancer	unknown	YES	Clinical Trials
GTBC060	<i>RAD50</i>	c.326_329delCAGA p.Thr109Asnfs*20	F	breast cancer	unknown	YES	Clinical Trials
GTBC404	<i>RAD50</i>	c.3553C>T (p.Arg1185Ter)	F	breast cancer	unknown	YES	Clinical Trials
GTBC131	<i>RAD50</i>	c.8756G>A p.Gly2919Asp/c.326_329delCAGA p.Thr109Asnfs*20	F	breast cancer	unknown	YES	Clinical Trials

GTBC295	<i>RAD50/CHEK2</i>	c.470T>C (p.Ile157Thr) / c.326_329delCAGA (p.Thr109Asnfs*20)	F	breast cancer	low+unknown	YES	PARPi (Off-Label)
GTBC156	<i>RAD51B</i>	c.84G>A p.=	F	breast cancer+Kidney cancer+Thyroid cancer	unknown	YES	Clinical Trials
GTBC229	<i>RAD51C</i>	c.706-2A>G	F	breast cancer, ovarian cancer	low	YES	PARPi (Off-Label)
GTBC142	<i>RAD51C</i>	c.904+5G>T	F	breast cancer	low	YES	PARPi (Off-Label)
GTBC032	<i>TP53</i>	c.1025G>C p.Arg342Pro	F	breast cancer	high	YES	None
GTBC025	<i>TP53</i>	c.469G>T p.Val157Phe	F	breast cancer	high	YES	None
GTBC259	<i>TP53</i>	c.586C>T (p.Arg196Ter)	F	breast cancer	high	YES	None
GTBC149	<i>TP53</i>	c.673-1G>A	F	breast cancer	high	YES	None
GTBC217	<i>TP53</i>	c.736A>G (p.Met246Val)	F	breast cancer	high	YES	None
GTBC076	<i>TP53</i>	c.743G>A p.Arg248Gln	F	breast cancer	high	YES	None
GTBC020	<i>TP53</i>	c.818G>A p.Arg273His	F	breast cancer	high	YES	None
GTBC085	<i>XRCC2</i>	c.190C>T p.Arg64*	F	breast cancer	unknown	NO	Clinical Trials