

Table S1 HRR genes analyzed by whole exome sequencing

Gene	Alias	Category	Process
BABAM1	NBA1, Merit40	core	HR
BARD1		core	HR
BLM		core	HR
BRCA1		core	HR
BRCA2	FANCD1	core	HR, FA
BRCC3	BRCC36	core	HR, Deubiquitylation
BRIP1	FANCI, BACH1	core	HR, FA
EME1	MMS4L	core	HR
EME2		core	HR
EXO1		core	HR
ABRAXAS1	AbraXas, ABRA1, FAM175A	core	HR
GEN1		core	HR
H2AFX	H2AX	core	DSB Signaling
MDC1		core	DSB Signaling
MRE11	MRE11A	core	DSB Signaling, HR
MUS81		core	HR
NBN	NBS	core	DSB Signaling, HR
PALB2	FANCD1	core	HR, FA
RAD50		core	DSB Signaling, HR
RAD51		core	HR
RAD51AP1		core	HR
RAD51B	RAD51L1	core	HR
RAD51C	FANCD1, RAD51L2	core	HR, FA
RAD51D	RAD51L3	core	HR
RAD52		core	HR
RAD54B		core	HR
RAD54L		core	HR
RBBP8	CTIP	core	HR
REV1	REV1L	core	HR
RPA1		core	HR, NER
RPA2		core	HR, NER
RTEL1		core	HR
SEM1	DSS1; SHFM1	core	HR
SLX1A	SLX1; GIYD1	core	HR
SLX1B	GIYD2	core	HR
SLX4	FANCD1	core	HR, FA
TP53BP1	53BP1	core	DSB Signaling
UIMC1	RAP80	core	HR
XRCC2		core	HR
XRCC3		core	HR
ABL1		related	DDR
ATM		related	DSB Signaling
ATR		related	DSB Signaling
ATRIP		related	DDR

BAP1		related	DDR
BRE	BRCC4	related	DDR
FAAP100	C17orf70	related	FA
FAAP24	C19orf40	related	HR, FA
FAAP20	C1orf86	related	FA
CDK12		related	DDR
CHEK1		related	DSB Signaling
CHEK2		related	DSB Signaling
DMC1		related	HR
DNA2		related	HR
DNMT3A		related	
ERCC4	XPF, FANCO	related	FA, SSA, NER
FAN1	MTMR15	related	FA
FANCA		related	FA
FANCB		related	FA
FANCC		related	FA
FANCD2		related	FA
FANCE		related	FA
FANCF		related	FA
FANCG	XRCC9	related	FA
FANCI	KIAA1794	related	FA
FANCL		related	FA
FANCM		related	FA
FEN1		related	DDR
FIGNL1		related	HR
HELQ		related	HR
KAT5	tip60	related	chromatin: HAT
NONO	NRB54; p54	related	HR
PARP1	ADPRT	related	SSBR
PIAS1		related	chromatin: sumoylation
PIAS4		related	chromatin: sumoylation
POLN	POLP4	related	Polymerase
POLQ		related	Polymerase
RAD1		related	DDR
RAD17	RAD24	related	DDR
RAD9A		related	DDR
RECQL	RECQ1	related	DDR
RECQL4		related	DDR
RIF1		related	HR
RMI1		related	HR
RMI2		related	HR
RNF168		related	DSB Signaling
RNF169		related	DSB Signaling
RNF4		related	Ubiquitination
RPA3		related	NER
SETMAR	Metnase	related	
SFPQ	PSF	related	

SFR1		related	HR
SMC1A	SMC1	related	cohesin
SMC3		related	cohesin
SMC5		related	cohesin
SPDR	KIAA0146	related	HR
TIMELESS		related	
TIPIN		related	DDR
TOPBP1		related	DDR
USP1		related	FA
USP11		related	HR, ubiquitination
WRN		related	DDR

FA: Fanconi Anemia; DSB Signaling: Double-strand break signaling; DDR: DNA Damage Response;
NER: nucleotide excision repair; SSBR: single-strand break repair

Table S2 Germline HRR gene mutations

ID	Chr	Start	End	Ref	Alt	Gene	Variant type	CLNSIG	Serum		Tumor			
									AD	AF	AD	AF	HRD	LOH
Patient1	chr17	43099817	43099817	G	A	BRCA1	stopgain	Pathogenic	41,51	0.554	31,66	0.680	83	+
Patient2	chr17	43063369	43063370	CA	-	BRCA1	frameshift deletion	Pathogenic	14,9	0.391	13,11	0.458	85	+
Patient3	chr13	32379885	32379885	-	A	BRCA2	frameshift insertion	Pathogenic	10,34	0.773	19,67	0.779	75	+
Patient4	chr13	32339764	32339765	TG	-	BRCA2	frameshift deletion	Pathogenic	17,26	0.605	18,38	0.679	49	+
Patient5	chr13	32331004	32331008	CAAAT	-	BRCA2	frameshift deletion	Pathogenic	8,4	0.333	5,6	0.545	62	+
Patient5	chr8	94399540	94399540	C	A	RAD54B	nonsynonymous SNV	Pathogenic	26,24	0.480	71,69	0.493	62	-
Patient6	chr13	32339929	32339932	AAT	-	BRCA2	frameshift deletion	Pathogenic	21,26	0.553	17,56	0.767	67	+
Patient7	chr13	32346841	32346841	C	T	BRCA2	stopgain	Pathogenic	12,3	0.200	12,19	0.613	47	+
Patient8	chr13	32346841	32346841	C	T	BRCA2	stopgain	Pathogenic	15,18	0.545	7,9	0.563	51	+
Patient9	chr13	32346841	32346841	C	T	BRCA2	stopgain	Pathogenic	14,17	0.548	23,4	0.148	29	+
Patient100	chr13	32333284	32333284	A	-	BRCA2	frameshift deletion	Pathogenic	7,10	0.588	20,20	0.500	28	-
Patient11	chr16	23621408	23621408	G	A	PALB2	stopgain	Pathogenic	17,26	0.605	47,51	0.520	39	+
Patient12	chr8	94399540	94399540	C	A	RAD54B	nonsynonymous SNV	Pathogenic	38,21	0.356	32,45	0.584	26	+

CLNSIG, clinical significance; AD, allele depth; AF, allele frequency; DP, Depth; GQ, Quality of the assigned genotype

Table S3: Somatic HRR gene mutations

ID	Chr	Start	End	Ref	Alt	Gene	Variant type	CLNSIG	AD	AF	HRD	LOH
Patient13	chr11	108321293	108321302	GCTACTAGAG	-	ATM	frameshift deletion		59,9	0.130	3	-
Patient14	chr11	108316019	108316019		C T	ATM	nonsynonymous SNV		81,6	0.078	12	-
Patient15	chr11	108251029	108251029		G C	ATM	nonsynonymous SNV		41,22	0.354	39	+
Patient16	chr3	142469454	142469454		C G	ATR	nonsynonymous SNV		23,8	0.273	59	+
Patient16	chr17	61683819	61683819		G A	BRIP1	nonsynonymous SNV	US	77,11	0.133	59	+
Patient16	chr17	81551048	81551048		C T	FAAP100	nonsynonymous SNV		407,156	0.278	59	+
Patient16	chrX	14850589	14850589		G C	FANCB	stopgain		49,16	0.254	59	-
Patient16	chr8	144515821	144515821		C G	RECQL4	nonsynonymous SNV		160,63	0.284	59	-
Patient16	chr3	133608923	133608923		G A	TOPBP1	stopgain		45,14	0.246	59	-
Patient17	chr3	142493248	142493248		C A	ATR	stopgain		57,6	0.107	50	-
Patient18	chr3	48447042	48447042		T A	ATRIP	nonsynonymous SNV		67,8	0.117	77	+
Patient19	chr17	43093458	43093458		T A	BRCA1	nonsynonymous SNV		77,22	0.228	22	+
Patient20	chr17	39462456	39462456		G A	CDK12	nonsynonymous SNV		75,7	0.089	5	+
Patient20	chr9	35079468	35079468		C G	FANCG	nonsynonymous SNV		215,21	0.092	5	-
Patient20	chr3	4313693	4313693		G C	SETMAR	nonsynonymous SNV		135,13	0.093	5	-
Patient21	chr22	28734714	28734714		C A	CHEK2	nonsynonymous SNV	US	89,6	0.072	30	-
Patient21	chr1	35192490	35192490		G T	SFPQ	nonsynonymous SNV		46,5	0.101	30	+
Patient22	chr16	13948099	13948116	TCTGAAACCC TTCCCGAG	-	ERCC4	nonframeshift deletion		143,11	0.077	15	-
Patient23	chr1	241866872	241866872		C T	EXO1	stopgain		98,7	0.074	17	-
Patient24	chr9	35075493	35075502	GGGCACCCAG	-	FANCG	frameshift deletion		285,17	0.059	16	-
Patient25	chr14	45167019	45167019		C A	FANCM	nonsynonymous SNV		33,6	0.170	36	-
Patient25	chr8	144512230	144512230		C G	RECQL4	nonsynonymous SNV		302,31	0.095	36	-
Patient25	chr2	151463251	151463251		C T	RIF1	nonsynonymous SNV		59,8	0.130	36	-
Patient25	chrX	47247887	47247887		C T	USP11	nonsynonymous SNV		58,27	0.322	36	-
Patient26	chr11	65860989	65860989		G C	MUS81	nonsynonymous SNV		187,12	0.065	2	-
Patient27	chr11	65860885	65860885		G C	MUS81	nonsynonymous SNV		157,18	0.107	44	+
Patient27	chr5	132640744	132640744		G C	RAD50	nonsynonymous SNV		90,9	0.099	44	+
Patient28	chrX	71291848	71291848		G A	NONO	nonsynonymous SNV		61,8	0.127	37	-
Patient29	chr16	23636244	23636244		T C	PALB2	nonsynonymous SNV		106,20	0.164	32	-

Patient30	chr3	121440114	121440114	T	C	POLQ	nonsynonymous SNV		26,6	0.205	11	-
Patient31	chr3	121487393	121487402	GCACCGCCAC	-	POLQ	frameshift deletion		87,7	0.083	29	-
Patient32	chr14	67865080	67865080	G	A	RAD51B	nonsynonymous SNV		54,9	0.153	32	-
Patient33	chr8	144511946	144511946	C	T	RECQL4	nonsynonymous SNV	US	191,51	0.209	22	-
Patient33	chrX	53405309	53405309	C	T	SMC1A	nonsynonymous SNV		175,59	0.254	22	-
Patient34	chr8	144517455	144517455	C	T	RECQL4	nonsynonymous SNV		153,13	0.083	58	-
Patient35	chr8	144514071	144514071	C	A	RECQL4	nonsynonymous SNV		216,36	0.146	15	-
Patient36	chr16	3597489	3597489	G	A	SLX4	unknown		86,17	0.171	44	-
Patient37	chr3	133656762	133656770	GCTACCAAC	-	TOPBP1	nonframeshift deletion		113,6	0.053	5	-
Patient38	chr15	43477599	43477599	T	C	TP53BP1	nonsynonymous SNV		61,15	0.206	34	-
Patient39	chr8	31067090	31067090	C	G	WRN	nonsynonymous SNV		59,5	0.090	12	-

CLNSIG, clinical significance; AD, allele depth; AF, allele frequency; DP, Depth; AMBQ, median base quality of alternate alleles; US, Uncertain significance