S1 Table. Ninety-eight pathogenic and likely pathogenic variants of 39 genes in 77 patients

Pat ID	Gene	Chromosome	Position	HGVSc	HGVSp	Mutation type	mRNA	Ref	Alt	Gene ID	QUAL	Depth	Allele freq	Pathogenicity	Somatic/ Germline	Methods for somatic calls
Pat12	ATR	chr3	142274783	c.2277delA	p.Ala760Leufs*11	Frameshift deletion	NM_001184.3	T	-	545	157	90	0.16	Novel	Somatic	Matched normal
Pat53	ATR	chr3	142217557	c.5440delA	p.Arg1814Glufs*10	Frameshift deletion	NM_001184.3	T	-	545	233	62	0.29	Novel	Somatic	Matched normal
Pat11	BRCA1	chr17	41256190	c.390C>A	p.Tyr130*	Stopgain	NM_007294.3	G	T	672	1395	60	0.95	Pathogenic	Germline	Matched normal
Pat46	BRCA1	chr17	41256190	c.390C>A	p.Tyr130*	Stopgain	NM_007294.3	G	T	672	2134	105	0.83	Pathogenic	Germline	Matched normal & BRCA sanger
Pat49	BRCA1	chr17	41256190	c.390C>A	p.Tyr130*	Stopgain	NM_007294.3	G	T	672	787	39	0.77	Pathogenic	Germline	Matched normal
Pat45	BRCA1	chr17	41246624	c.923_924delGC	p.Ser308Lysfs*11	Frameshift deletion	NM_007294.3	GC	-	672	3067	89	0.86	Pathogenic	Germline	Matched normal & BRCA sanger
OCA02	BRCA1	chr17	41246620	c.928C>T	p.Gln310*	Stopgain	NM_007294.3	G	A	672	4000	2533	0.65	Pathogenic	Germline	BRCA sanger
OCA16	BRCA1	chr17	41246578	c.969_970delAA	p.Ser324*	Stopgain	NM_007294.3	TT	-	672	4000	4217	0.67	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Somatic	BRCA sanger
Pat42	BRCA1	chr17	41246436	c.1112delC	p.Pro371Leufs*3	Frameshift deletion	NM_007294.3	G	-	672	1754	107	0.55	Pathogenic	Somatic	Matched normal & BRCA sanger
Pat10	BRCA1	chr17	41245832	c.1716dupA	p.Ser573Ilefs*13	Frameshift insertion	NM_007294.3	-	T	672	437	21	0.75	Pathogenic	Germline	Matched normal
OCA19	BRCA1	chr17	41245735	c.1813delG	p.Ala605Hisfs*7	Frameshift deletion	NM_007294.3	C	-	672	4000	2456	0.84	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Somatic	BRCA sanger
OCA07	BRCA1	chr17	41245500	c.2048delA	p.Lys683Serfs*18	Frameshift deletion	NM_007294.3	T	-	672	4000	2930	0.86	Pathogenic	Germline	BRCA sanger
Pat55	BRCA1	chr17	41245189	c.2359delG	p.Glu787Lysfs*5	Frameshift deletion	NM_007294.3	C	-	672	4669	169	0.85	Pathogenic	Germline	Matched normal
Pat60	BRCA1	chr17	41244528	c.3020delC	p.Ser1007*	Frameshift deletion	NM_007294.3	G	-	672	1459	41	0.95	Pathogenic	Germline	Matched normal

Pat48	BRCA1	chr17	41244106	c.3442G>T	p.Glu1148*	Stopgain	NM_007294.3	C	A	672	1269	57	0.88	Pathogenic	Germline	Matched normal & BRCA sanger
Pat20	BRCA1	chr17	41243844	c.3700_3704delTAAA	p.Val1234Glnfs*8	Frameshift deletion	NM_007294.3	TTTAC	-	672	4034	137	0.78	Pathogenic	Germline	Matched normal & BRCA sanger
OCA13	BRCA1	chr17	41243735	c.3813dupT	p.Asn1272*	Stopgain	NM_007294.3	-	A	672	4000	6841	0.79	Pathogenic	Germline	BRCA sanger
OCA18	BRCA1	chr17	41243653	c.3895C>T	p.Gln1299*	Stopgain	NM_007294.3	G	A	672	4000	2566	0.89	Pathogenic	Germline	BRCA sanger
Pat36	BRCA1	chr17	41243557	c.3991C>T	p.Glu1331*	Stopgain	NM_007294.3	G	A	672	4649	210	0.90	Pathogenic	Do not know	
Pat47	BRCA1	chr17	41234491	c.4287C>A	p.Tyr1429*	Stopgain	NM_007294.3	G	T	672	832	40	0.75	Pathogenic	Somatic	Matched normal
Pat21	BRCA1	chr17	41223130	c.4801A>T	p.Lys1601*	Stopgain	NM_007294.3	T	A	672	3486	132	0.96	Pathogenic	Germline	Matched normal & BRCA sanger
Pat23	BRCA1	chr17	41201205	c.5339T>C	p.Leu1780Pro	missense	NM_007294.3	A	G	672	2068	102	0.81	Likely pathogenic	Germline	Matched normal
Pat28	BRCA1	chr17	41201205	c.5339T>C	p.Leu1780Pro	missense	NM_007294.3	A	G	672	2123	129	0.70	Likely pathogenic	Germline	Matched normal
Pat41	BRCA1	chr17	41197781	c.5496_5506delGGTGACCCGAGinsA	p.Val1833Serfs*7	in-frame deletion	NM_007294.3	CTCGGGTCACC	T	672	1386	47	0.74	Pathogenic	Germline	Matched normal
OCA14	BRCA2	chr13	32900664	c.547delA	p.Ser183Valfs*2	Frameshift deletion	NM_000059.3	A	-	675	4000	6675	0.59	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Somatic	BRCA sanger
Pat54	BRCA2	chr13	32911057	c.2567delA	p.Asn856Ilefs*2	Frameshift deletion	NM_000059.3	A	-	675	934	47	0.69	Novel	Germline	Matched normal
Pat05	BRCA2	chr13	32912346	c.3860delA	p.Asn1287Ilefs*6	Frameshift deletion	NM_000059.3	A	-	675	433	27	0.82	Pathogenic	Germline	BRCA sanger
Pat08	BRCA2	chr13	32914066	c.5576_5579delTTAA	p.Ile1859Lysfs*3	Frameshift deletion	NM_000059.3	AATT	-	675	783	23	0.87	Pathogenic	Germline	Matched normal & BRCA sanger
Pat17	FANCA	chr16	89809249	c.3720_3724delAAACA	p.Glu1240Aspfs*36	Frameshift deletion	NM_000135.2	TGTTT	-	2175	680	92	0.25	pathogenic	Germline	Matched normal
Pat53	MSH2	chr2	47637247	c.387_388delTC	p.Gln130Valfs*2	Frameshift deletion	NM_000251.2	TC	-	4436	2101	71	0.83	Pathogenic	Germline	Matched normal
Pat07	MSH2	chr2	47708005	c.2633_2634delAG	p.Glu878Alafs*3	Frameshift deletion	NM_000251.2	AG	-	4436	1323	83	0.49	Pathogenic	Germline	Matched normal

Pat14	MSH6	chr2	48033753	c.3964G>T	p.Glu1322*	Stopgain	NM_000179.2	G	T	2956	86	105	0.15	Pathogenic	Somatic	Matched normal
Pat21	RAD50	chr5	131925407	c.1330G>T	p.Glu444*	Stopgain	NM_005732.3	G	T	10111	189	26	0.35	Novel	Somatic	Matched normal
Pat53	RAD50	chr5	131931451	c.2165delA	p.Lys722Argfs*14	Frameshift deletion	NM_005732.3	A	-	10111	1886	146	0.70	Pathogenic	Somatic	Matched normal
Pat14	RAD50	chr5	131953844	c.3247G>T	p.Glu1083*	Stopgain	NM_005732.3	G	T	10111	71	45	0.18	Novel	Somatic	Matched normal
OCA01	TP53	chr17	7579722	c.97-1G>T	-	Splicing	NM_000546.5	C	T	7157	4000	2969	0.93	Pathogenic	Do not know	
Pat41	TP53	chr17	7579573	c.114delA	p.Ala39Glnfs*5	Frameshift deletion	NM_000546.5	T	-	7157	1111	38	0.92	Novel	Somatic	Matched normal
Pat17	TP53	chr17	7579513	c.153_174delACAATGGTTCACTGAAGACCCA	p.Gln52Valfs*64	Frameshift deletion	NM_000546.5	TGGGTCTTCAGTGAACCATTGT	-	7157	416	30	0.41	Novel	Somatic	Matched normal
Pat36	TP53	chr17	7579414	c.273G>A	p.Trp91*	Stopgain	NM_000546.5	C	T	7157	392	22	0.77	Pathogenic	Do not know	
OCA15	TP53	chr17	7578539	c.391A>T	p.Asn131Tyr	Missense	NM_000546.5	T	A	7157	4000	4377	0.69	Pathogenic	Do not know	
OCA13	TP53	chr17	7578535	c.395A>G	p.Lys132Arg	Missense	NM_000546.5	T	C	7157	4000	4395	0.43	Likely pathogenic	Do not know	
Pat44	TP53	chr17	7578535	c.395A>G	p.Lys132Arg	Missense	NM_000546.5	T	C	7157	347	51	0.31	Likely pathogenic	Somatic	Matched normal
Pat11	TP53	chr17	7578535	c.395A>G	p.Lys132Arg	Missense	NM_000546.5	T	C	7157	1718	82	0.76	Likely pathogenic	Somatic	Matched normal
OCA07	TP53	chr17	7578525	c.405C>G	p.Cys135Trp	Missense	NM_000546.5	G	C	7157	4000	8223	0.78	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Do not know	
OCA05	TP53	chr17	7578524	c.406C>T	p.Gln136*	Stopgain	NM_000546.5	G	A	7157	4000	3612	0.80	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Do not know	
Pat22	TP53	chr17	7578469	c.450_460delACCCCCGCCCG	p.Pro151Hisfs*26	Frameshift deletion	NM_000546.5	CGGGCGGGGGT	-	7157	1124	49	0.60	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Somatic	Matched normal

OCA12	TP53	chr17	7578478	c.452C>G	p.Pro151Arg	Missense	NM_000546.5	G	C	7157	4000	4106	0.76	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Do not know	
Pat24	TP53	chr17	7578442	c.488A>G	p.Tyr163Cys	Missense	NM_000546.5	T	C	7157	798	54	0.59	Pathogenic	Somatic	Matched normal
Pat61	TP53	chr17	7578407	c.523C>G	p.Arg175Gly	Missense	NM_000546.5	G	C	7157	1743	76	0.83	Pathogenic	Somatic	Matched normal
OCA09	TP53	chr17	7578235	c.497A>G	p.Tyr166Cys	Missense	NM_001126118.1	T	C	7157	4000	3405	0.58	Likely pathogenic	Do not know	
Pat10	TP53	chr17	7578406	c.524G>A	p.Arg175His	Missense	NM_000546.5	C	T	7157	1076	48	0.81	Pathogenic	Somatic	Matched normal
Pat18	TP53	chr17	7578406	c.524G>A	p.Arg175His	Missense	NM_000546.5	C	T	7157	867	40	0.78	Pathogenic	Somatic	Matched normal
Pat25	TP53	chr17	7578403	c.527G>T	p.Cys176Phe	Missense	NM_000546.5	C	A	7157	722	38	0.74	Likely pathogenic	Do not know	
OCA06	TP53	chr17	7578394	c.536A>G	p.His179Arg	Missense	NM_000546.5	T	C	7157	4000	3302	0.70	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Do not know	
Pat52	TP53	chr17	7578392	c.538G>A	p.Glu180Lys	Missense	NM_000546.5	C	T	7157	212	37	0.30	Likely pathogenic	Do not know	
OCA18	TP53	chr17	7578283	c.566C>T	p.Ala189Val	Missense	NM_000546.5	G	A	7157	4000	3393	0.93	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Do not know	
Pat58	TP53	chr17	7578271	c.578A>G	p.His193Arg	Missense	NM_000546.5	T	C	7157	608	64	0.44	Likely pathogenic	Do not know	
Pat55	TP53	chr17	7578271	c.578A>G	p.His193Arg	Missense	NM_000546.5	T	C	7157	1319	77	0.70	Likely pathogenic	Somatic	Matched normal
Pat21	TP53	chr17	7578269	c.580C>T	p.Leu194Phe	Missense	NM_000546.5	G	A	7157	1507	71	0.85	Likely pathogenic	Somatic	Matched normal
OCA03	TP53	chr17	7578265	c.584T>C	p.Ile195Thr	Missense	NM_000546.5	A	G	7157	4000	5700	0.52	Pathogenic	Do not know	
Pat20	TP53	chr17	7578265	c.584T>C	p.Ile195Thr	Missense	NM_000546.5	A	G	7157	886	71	0.52	Pathogenic	Somatic	Matched normal
Pat45	TP53	chr17	7578265	c.584T>C	p.Ile195Thr	Missense	NM_000546.5	A	G	7157	1088	71	0.61	Pathogenic	Somatic	Matched normal
Pat57	TP53	chr17	7578262	c.587G>C	p.Arg196Pro	Missense	NM_000546.5	C	G	7157	1140	108	0.46	Likely pathogenic	Somatic	Matched normal
Pat42	TP53	chr17	7578249	c.599delA	p.Asn200Ilefs*47	Frameshift deletion	NM_000546.5	T	-	7157	1150	75	0.56	pathogenic	Somatic	Matched normal

Pat16	TP53	chr17	7578219	c.629delA	p.Asn210Thrfs*37	Frameshift deletion	NM_000546.5	T	-	7157	1107	93	0.43	Novel	Somatic	Matched normal
Pat53	TP53	chr17	7578212	c.637C>T	p.Arg213*	Stopgain	NM_000546.5	G	A	7157	1711	81	0.83	Pathogenic	Somatic	Matched normal
Pat12	TP53	chr17	7578190	c.659A>G	p.Tyr220Cys	Missense	NM_000546.5	T	C	7157	109	56	0.18	Pathogenic	Somatic	Matched normal
Pat15	TP53	chr17	7578190	c.659A>G	p.Tyr220Cys	Missense	NM_000546.5	T	C	7157	899	47	0.72	Pathogenic	Somatic	Matched normal
Pat35	TP53	chr17	7578190	c.659A>G	p.Tyr220Cys	Missense	NM_000546.5	T	C	7157	432	69	0.28	Pathogenic	Do not know	
OCA02	TP53	chr17	7577610	c.673-2A>T	-	splicing	NM_000546.5	T	C	7157	4000	6469	0.49	Pathogenic	Do not know	
OCA10	TP53	chr17	7577609	c.673-1G>A	-	splicing	NM_000546.5	C	T	7157	4000	5122	0.48	Pathogenic	Do not know	
Pat43	TP53	chr17	7577607	c.673dupG	p.Val225Glyfs*4	Frameshift insertion	NM_000546.5	-	C	7157	1587	74	0.73	Novel	Somatic	Matched normal
OCA08	TP53	chr17	7577593	c.688_689insTGTA	p.Thr230Metfs*11	Frameshift insertion	NM_000546.5	-	TACA	7157	4000	3255	0.95	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Do not know	
Pat29	TP53	chr17	7577581	c.700T>C	p.Tyr234His	Missense	NM_000546.5	A	G	7157	1915	115	0.70	Pathogenic	Somatic	Matched normal
Pat54	TP53	chr17	7577580	c.701A>G	p.Tyr234Cys	Missense	NM_000546.5	T	C	7157	872	61	0.62	Pathogenic	Somatic	Matched normal
Pat31	TP53	chr17	7577570	c.711G>C	p.Met237Ile	Missense	NM_000546.5	C	G	7157	976	41	0.80	pathogenic	Do not know	
Pat49	TP53	chr17	7577570	c.711G>A	p.Met237Ile	Missense	NM_000546.5	C	T	7157	856	44	0.84	Likely pathogenic	Somatic	Matched normal
Pat09	TP53	chr17	7577547	c.734G>A	p.Gly245Asp	Missense	NM_000546.5	C	T	7157	1038	85	0.54	Pathogenic	Somatic	Matched normal
Pat27	TP53	chr17	7577539	c.742C>T	p.Arg248Trp	Missense	NM_000546.5	G	A	7157	470	40	0.51	Pathogenic	Somatic	Matched normal
OCA18	TP53	chr17	7577538	c.743G>A	p.Arg248Gln	Missense	NM_000546.5	C	T	7157	4000	13331	0.89	Pathogenic	Do not know	
Pat28	TP53	chr17	7577536	c.745A>G	p.Arg249Gly	Missense	NM_000546.5	T	C	7157	649	60	0.47	Somatic: pathogenic	Somatic	Matched normal
Pat33	TP53	chr17	7577511	c.770T>G	p.Leu257Arg	Missense	NM_000546.5	A	C	7157	594	33	0.72	Likely pathogenic	Somatic	Matched normal
Pat02	TP53	chr17	7577507	c.774A>C	p.Glu258Asp	Missense	NM_000546.5	T	G	7157	424	28	0.67	Pathogenic	Somatic	Matched normal
OCA11	TP53	chr17	7577153	c.785G>T	p.Gly262Val	Missense	NM_000546.5	C	A	7157	4000	3643	0.57	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Do not know	

OCA16	TP53	chr17	7577153	c.785G>T	p.Gly262Val	Missense	NM_000546.5	C	A	7157	4000	3194	0.58	Pathogenic (Loss-of-Function in Oncomine Knowledge base)	Do not know	
Pat05	TP53	chr17	7577124	c.814G>C	p.Val272Leu	Missense	NM_000546.5	C	G	7157	679	21	1.00	Pathogenic	Somatic	Matched normal
Pat38	TP53	chr17	7577120	c.818G>A	p.Arg273His	Missense	NM_000546.5	C	T	7157	60	24	0.75	Pathogenic	Do not know	
Pat14	TP53	chr17	7577117	c.821T>G	p.Val274Gly	Missense	NM_000546.5	A	C	7157	36	29	0.83	Pathogenic	Somatic	Matched normal
Pat39	TP53	chr17	7577108	c.830G>T	p.Cys277Phe	Missense	NM_000546.5	C	A	7157	459	36	0.50	Pathogenic	Somatic	Matched normal
Pat13	TP53	chr17	7577106	c.832C>A	p.Pro278Thr	Missense	NM_000546.5	G	T	7157	1031	44	0.89	Pathogenic	Somatic	Matched normal
Pat06	TP53	chr17	7577046	c.892G>T	p.Glu298*	Stopgain	NM_000546.5	C	A	7157	3071	117	0.93	Pathogenic	Somatic	Matched normal
Pat23	TP53	chr17	7577022	c.916C>T	p.Arg306*	Stopgain	NM_000546.5	G	A	7157	3269	161	0.86	Pathogenic	Somatic	Matched normal
Pat40	TP53	chr17	7577022	c.916C>T	p.Arg306*	Stopgain	NM_000546.5	G	A	7157	923	68	0.60	Pathogenic	Somatic	Matched normal
Pat08	TP53	chr17	7577022	c.916C>T	p.Arg306*	Stopgain	NM_000546.5	G	A	7157	1235	63	0.78	Pathogenic	Somatic	Matched normal
Pat47	TP53	chr17	7576928	c.920-2A>G		splicing	NM_000546.5	T	C	7157	1030	45	0.80	Likely pathogenic	Somatic	Matched normal
OCA04	TP53	chr17	7574018	c.1009C>T	p.Arg337Cys	Missense	NM_000546.5	G	A	7157	4000	3729	0.63	Pathogenic	Do not know	
Pat26	TP53	chr17	7574000	c.1027G>T	p.Glu343*	Stopgain	NM_000546.5	C	A	7157	224	39	0.31	Novel	Somatic	Matched normal
Pat30	TP53	chr17	7573991	c.1035dupT	p.Glu346*	Stopgain	NM_000546.5	-	A	7157	593	64	0.35	Novel	Somatic	Matched normal