

Supp. Table S1. Deleterious *BRCA1* and *BRCA2* mutations identified in 705 contralateral (CBC) and 1398 unilateral breast cancer patients (UBC).

Gene	Exon	NT <sup>1</sup>	Base Change	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA1	2	185	del AG	Stop 39	185delAG	c.68_69delAG	p.Glu23fs	F	16	12	4
BRCA1	2	188	ins AG	Stop 31	188insAG	c.68_69dupAG	p.Cys24fs	F	2	2	
BRCA1	3	202	del TG	Stop 39	202delTG	c.83_84delTG	p.Leu28fs	F	1	1	
BRCA1	3	249	T to A	Cys to Ser	C44S	c.130T>A	p.Cys44Ser	M	2	2	
BRCA1	5	297	del CA	Stop 64	297delCA	c.178_179delCA	p.Gln60fs	F	1		1
BRCA1	5	300	T to G	Cys to Gly	C61G	c.181T>G	p.Cys61Gly	M	10	8	2
BRCA1	5	330	A to G	Arg to Gly	R71G	c.211A>G	p.Arg71Gly	S	1	1	
BRCA1	7	421-2	del A	-	IVS6-2delA	c.302-2delA		S	1	1	
BRCA1	7	546	G to T	Glu to Stop	E143X	c.427G>T	p.Glu143X	N	1		1
BRCA1	8	604	del TG	Stop 180	604delTG	c.485_486delTG	p.Val162fs	F	1	1	
BRCA1	10	713-2	A to C	-	IVS9-2A>C	c.594-2A>C		S	1	1	
BRCA1	11	1201	del 11	Stop 361	1201del11	c.1082_1092del11	p.Ser361X	F	1		1
BRCA1	11	1240	del C	Stop 375	1240delC	c.1121delC	p.Thr374fs	F	1	1	
BRCA1	11	1623	del TTAAA	Stop 503	1623del5	c.1504_1508del5	p.Leu502fs	F	2	1	1
BRCA1	11	1648	C to A	Ser to Stop	S510X	c.1529C>A	p.Ser510X	N	1		1
BRCA1	11	1675	del A	Stop 531	1675delA	c.1556delA	p.Lys519fs	F	2	2	
BRCA1	11	1806	C to T	Gln to Stop	Q563X	c.1687C>T	p.Gln563X	N	1	1	
BRCA1	11	1921	del 248	Stop 603	1921del248	c.1802_2049del248	p.His601fs	F	1	1	
BRCA1	11	2154	A to T	Lys to Stop	K679X	c.2035A>T	p.Lys679X	N	1	1	
BRCA1	11	2433	del G	Stop 791	2433delG	c.2314delG	p.Val772fs	F	1		1
BRCA1	11	2508	del G	Stop 802	2508delG	c.2389delG	p.Glu797fs	F	1	1	
BRCA1	11	2552	del C	Stop 814	2552delC	c.2433delC	p.Lys812fs	F	1		1
BRCA1	11	2594	del C	Stop 845	2594delC	c.2475delC	p.Asp825fs	F	3	1	2
BRCA1	11	2634	del C	Stop 845	2634delC	c.2515delC	p.His839fs	F	1	1	
BRCA1	11	2754	G to T	Glu to Stop	E879X	c.2635G>T	p.Glu879X	N	1	1	
BRCA1	11	2798	del GAAA	Stop 998	2798del4	c.2679_2682delGAAA	p.Lys893fs	F	1	1	
BRCA1	11	2841	G to T	Glu to Stop	E908X	c.2722G>T	p.Glu908X	N	1	1	
BRCA1	11	2983	C to A	Ser to Stop	S955X	c.2864C>A	p.Ser955X	N	1		1
BRCA1	11	3124	del A	Stop 1023	3124delA	c.3005delA	p.Asn1002fs	F	1		1
BRCA1	11	3148	del CT	Stop 1016	3148delCT	c.3029_3030delCT	p.Pro1010fs	F	1		1
BRCA1	11	3171	ins TGAGA	Stop 1025	3171ins5	c.3048_3052dup5	p.Asn1018fs	F	1	1	
BRCA1	11	3307	del CC ins G	Stop 1063	3307delCCinsG	c.3188_3189delCCinsG	p.Ser1063X	F	1		1
BRCA1	11	3347	del AG	Stop 1084	3347delAG	c.3228_3229delAG	p.Gly1077fs	F	1	1	
BRCA1	11	3438	G to T	Glu to Stop	E1107X	c.3319G>T	p.Glu1107fs	N	2	2	
BRCA1	11	3450	del CAAG	Stop 1115	3450del4	c.3331_3334delCAAG	p.Gln1111fs	F	1	1	

Gene	Exon	NT <sup>1</sup>	Base Change	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA1	11	3508	C to G	Ser to Stop	S1130X	c.3389C>G	p.Ser1130X	N	1		1
BRCA1	11	3600	del 11	Stop 1163	3600del11	c.3481_3491del11	p.Glu1161fs	F	2	2	
BRCA1	11	3726	C to T	Arg to Stop	R1203X	c.3607C>T	p.Arg1203X	N	3	1	2
BRCA1	11	3829	del T	Stop 1263	3829delT	c.3710delT	p.Ile1237fs	F	1	1	
BRCA1	11	3867	G to T	Glu to Stop	E1250X	c.3748G>T	p.Glu1250X	N	4	2	2
BRCA1	11	3875	del GTCT	Stop 1262	3875del4	c.3756_3759delGTCT	p.Ser1253fs	F	4	4	
BRCA1	11	4184	del TCAA	Stop 1364	4184del4	c.4065_4068delTCAA	p.Asn1355fs	F	2		2
BRCA1	12	4302	C to T	Gln to Stop	Q1395X	c.4183C>T	p.Gln1395X	N	1	1	
BRCA1	13	4341	C to T	Gln to Stop	Q1408X	c.4222C>T	p.Gln1408X	N	1		1
BRCA1	13	4370	del GT	Stop 1426	4370delGT	c.4251_4252delGT	p.Leu1418fs	F	1	1	
BRCA1	13	4446	C to T	Arg to Stop	R1443X	c.4327C>T	p.Arg1443X	N	2	1	1
BRCA1	13	4476+1	G to T	-	IVS13+1G>T	c.4357+1G>T		S	2	1	1
BRCA1	16	5105+2	T to C	-	IVS16+2T>C	c.4986+2T>C		S	1	1	
BRCA1	18	5214	C>T	Arg to Trp	R1699W	c.5095C>T	p.Arg1699Trp	M	1		1
BRCA1	18	5242	C>A	Ala to Glu	A1708E	c.5123C>A	p.Ala1708Glu	M	1	1	
BRCA1	20	5332	G to A	Gly to Glu	G1738E	c.5213G>A	p.Gly1738Glu	M	1		1
BRCA1	20	5349	del A	Stop 1764	5349delA	c.5230delA	p.Arg1744fs	F	1	1	
BRCA1	20	5382	ins C	Stop 1829	5382insC	c.5266dupC	p.Gln1756fs	F	10	2	8
BRCA1	21	5443	T to G	Met to Arg	M1775R	c.5324T>G	p.Met1775Arg	M	1		1
BRCA1	23	5563	G to A	Trp to Stop	W1815X	c.5444G>A	p.Trp1815X	N	1		1
BRCA1	24	5622	C to T	Arg to Stop	R1835X	c.5503C>T	p.Arg1835X	N	1		1
BRCA1	24	5677	ins A	Stop 1853	5677insA	c.5558dupA	p.Tyr1853X	F	1	1	
BRCA1	Total								109	67	42
BRCA2	2	231	G to A	Met to Ile	M11	c.3G>A	p.Met1Ile	M	1		1
BRCA2	2	279	del AC	Stop 29	279delAC	c.51_52delAC	p.Arg18fs	F	1		1
BRCA2	4	592	del A	Stop 135	592delA	c.364delA	p.Thr122fs	F	1	1	
BRCA2	5	697	del AA	Stop 181	697delAA	c.469_470delAA	p.Lys157fs	F	1		1
BRCA2	7	838	del C	Stop 210	838delC	c.610delC	p.Leu204fs	F	1	1	
BRCA2	9	983	del ACAG	Stop 275	983del4	c.755_758delACAG	p.Asp252fs	F	2	1	1
BRCA2	10	1461	ins A	Stop 420	1461insA	c.1233dupA	p.Pro412fs	F	1		1
BRCA2	10	1538	del AAGA	Stop 458	1538del4	c.1310_1313delAAGA	p.Lys437fs	F	3	3	
BRCA2	10	1898	del 14	Stop 557	1898del14	c.1670_1683del14	p.Leu557X	F	1	1	
BRCA2	10	2034	del A	Stop 613	2034delA	c.1813delA	p.Ile605fs	F	1	1	
BRCA2	10	2041	ins A	Stop 615	2041insA	c.1813dupA	p.Ile605fs	F	3	2	1
BRCA2	11	2482	del GACT	Stop 770	2482del4	c.2254_2257delGACT	p.Asp752fs	F	1	1	
BRCA2	11	2600	C to A	Ser to Stop	S791X	c.2372C>A	p.Ser791X	N	1	1	

Gene	Exon	NT <sup>1</sup>	Base Change	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA2	11	3036	del ACAA	Stop 958	3036del4	c.2808_2811delACAA	p.Ala938fs	F	5	4	1
BRCA2	11	3058	A to T	Lys to Stop	K944X	c.2830A>T	p.Lys944X	N	1		1
BRCA2	11	3337	C to T	Gln to Stop	Q1037X	c.3109C>T	p.Gln1037X	N	1	1	
BRCA2	11	3492	ins T	Stop 1098	3492insT	c.3264dupT	p.Gln1089fs	F	1		1
BRCA2	11	3773	del TT	Stop 1182	3773delTT	c.3545_3546delTT	p.Phe1182X	F	1		1
BRCA2	11	3827	del GT	Stop 1200	3827delGT	c.3599_3600delGT	p.Cys1200X	F	1	1	
BRCA2	11	4075	del GT	Stop 1284	4075delGT	c.3847_3848delGT	p.Val1283fs	F	1	1	
BRCA2	11	4093	del AAAT	Stop 1291	4093del4	c.3865_3868delAAAT	p.Lys1289fs	F	1		1
BRCA2	11	4206	ins TGCT	Stop 1330	4206ins4	c.3975_3978dupTTGC	p.Ala1327fs	F	1	1	
BRCA2	11	4677	del A	Stop 1485	4677delA	c.4449delA	p.Asp1484fs	F	1	1	
BRCA2	11	5164	del GAAA	Stop 1668	5164del4	c.4936_4939delGAAA	p.Glu1646fs	F	1	1	
BRCA2	11	5804	del TTAA	Stop 1861	5804del4	c.5576_5579delTTAA	p.Ile1859fs	F	1		1
BRCA2	11	5844	del AGTAA	Stop 1873	5844del5	c.5616_5620del5	p.Lys1872fs	F	1		1
BRCA2	11	5873	C to A	Ser to Stop	S1882X	c.5645C>A	p.Ser1882X	N	1	1	
BRCA2	11	5910	C to G	Tyr to Stop	Y1894X	c.5682C>G	p.Tyr1894X	N	1	1	
BRCA2	11	5950	del CT	Stop 1909	5950delCT	c.5722_5723delCT	p.Leu1908fs	F	2	1	1
BRCA2	11	6174	del T	Stop 2003	6174delT	c.5946delT	p.Ser1982fs	F	5	2	3
BRCA2	11	6286	G to T	Glu to Stop	E2020X	c.6058G>T	p.Glu2020X	N	1		1
BRCA2	11	6503	del TT	Stop 2098	6503delTT	c.6275_6276delTT	p.Leu2092fs	F	1		1
BRCA2	11	6633	del CTTAA	Stop 2137	6633del5	c.6405_6409del5	p.Asn2135fs	F	1		1
BRCA2	11	6671	del CT	Stop 2149	6671delCT	c.6443_6444delCT	p.Ser2148fs	F	1		1
BRCA2	11	6714	del ACAA	Stop 2166	6714del4	c.6486_6489delACAA	p.Lys2162fs	F	1		1
BRCA2	11	6884	C to G	Ser to Stop	S2219X	c.6656C>G	p.Ser2219X	N	1		1
BRCA2	13	7169	del C	Stop 2315	7169delC	c.6941delC	p.Thr2314fs	F	1		1
BRCA2	14	7253	del AA	Stop 2358	7253delAA	c.7025_7026delAA	p.Gln2342fs	F	1	1	
BRCA2	14	7295	del TT	Stop 2358	7295delTT	c.7067_7068delTT	p.Phe2356fs	F	1		1
BRCA2	14	7297	del CT	Stop 2358	7297delCT	c.7069_7070delCT	p.Leu2357fs	F	2	1	1
BRCA2	14	7438	del 7 insTG	Stop 2410	7438del7ins2	c.7210_7216del7insTG	p.Lys2404fs	F	1		1
BRCA2	15	7786	C to T	Arg to Stop	R2520X	c.7558C>T	p.Arg2520X	N	1		1
BRCA2	16	7990	del ATA ins TT	Stop 2647	7990del3ins2	c.7762_7764delATAinsTT	p.Ile2588fs	F	1	1	
BRCA2	17	8141	del TTCCT	Stop 2638	8141del5	c.7913_7917del5	p.Phe2638X	F	1	1	
BRCA2	18	8550	ins T	Stop 2781	8550insT	c.8322dupT	p.Met2775fs	F	1	1	
BRCA2	18	8558	del A	Stop 2820	8558delA	c.8330delA	p.Lys2777fs	F	1	1	
BRCA2	20	8803	del C	Stop 2862	8803delC	c.8575delC	p.Gln2859fs	F	1	1	
BRCA2	20	8823	ins T	Stop 2868	8823insT	c.8594dupT	p.Leu2865fs	F	1	1	
BRCA2	21	8958	del T	Stop 2926	8958delT	c.8730delT	p.Asn2910fs	F	1	1	
BRCA2	22	9181+1	G to T	-	IVS22+1G>T	c.8953+1G>T		S	1	1	

Gene	Exon	NT <sup>1</sup>	Base Change	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC	
BRCA2	23	9197	G to A	Trp to Stop	W2990X	c.8969G>A	p.Trp2990fs	N	1		1	
BRCA2	23	9326	ins A	Stop 3043	9326insA	c.9097dupA	p.Thr3033fs	F	1	1		
BRCA2	24	9481	ins A	Stop 3110	9481insA	c.9253dupA	p.Thr3085fs	F	1	1		
BRCA2	25	9610	C to T	Arg to Stop	R3128X	c.9382C>T	p.Arg3128X	N	1	1		
BRCA2	25	9729+3	A to T	-	IVS25+3A>T	c.9501+3A>T		S	2		2	
BRCA2	26	9769	del 14	Stop 3183	9769del14	c.9541_9554del14	p.Met3181fs	F	1	1		
BRCA2	Total									72	41	31

<sup>1</sup> BRCA1: nucleotide number according to GenBank U14680.1. <http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?db=nucleotide&val=555931>

<sup>1</sup> BRCA2: nucleotide number according to GenBank NM\_000059.1. <http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?db=nucleotide&val=4502450>

<sup>2</sup> BIC (Breast Cancer Information Core). <http://research.nhgri.nih.gov/bic/>

<sup>3</sup> HGVS (Human Genome Variation Society). <http://www.hgvs.org/mutnomen/recs.html>

<sup>4</sup> Type: F=frame shift, M=missense, S=splice site, N=nonsense

<sup>5</sup> Number of observations

mutations not earlier described in BIC (by November 2008)

Supp. Table S2. *BRCA1* and *BRCA2* unclassified variants identified in 705 contralateral (CBC) and 1398 unilateral breast cancer patients (UBC).

Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA1	2	101-10	T to C	-	IVS1-10T>C	c.-29T>C		IVS	1	1	0
BRCA1	3	200-129	G to A	-	IVS2-129G>A	c.81-129G>A		IVS	1	0	1
BRCA1	3	200-55	T to G	-	IVS2-55T>G	c.81-55T>G		IVS	1	0	1
BRCA1	3	233	G to A	Lys to Lys	K38K	c.114T>A	p.=	Syn	3	3	0
BRCA1	3	253	A to C	Lys to Thr	K45T	c.134A>C	p.Lys45Thr	M	1	0	1
BRCA1	5	254-53	T to C	-	IVS4-53T>C	c.135-53T>C		IVS	1	0	1
BRCA1	5	254-23	C to T	-	IVS4-23C>T	c.135-23C>T		IVS	1	0	1
BRCA1	5	331+23	T to A	-	IVS5+23T>A	c.212+23T>A		IVS	4	1	3
BRCA1	7	421-70	G to A	-	IVS6-70G>A	c.302-70G>A		IVS	1	0	1
BRCA1	7	433	A to G	Tyr to Cys	Y105C	c.314A>G	p.Tyr105Cys	M	1	0	1
BRCA1	7	544	C to A	Pro to His	P142H	c.425C>A	p.Pro142His	M	4	2	2
BRCA1	7	560+36	del 14	-	IVS7+36del14	c.441+36del14		IVS	1154	377	777
BRCA1	8	561-34	C to T	-	IVS7-34C>T	c.442-34C>T		IVS	779	254	525
BRCA1	8	628	G to A	Arg to Gln	R170Q	c.509G>A	p.Arg170Gln	M	2	1	1
BRCA1	8	655	A to G	Tyr to Cys	Y179C	c.536A>G	p.Tyr179Cys	M	2	0	2
BRCA1	9	667-80	T>C	-	IVS8-80T>C	c.548-80T>C		IVS	5	3	2
BRCA1	9	667-58	delT	-	IVS8-58delT	c.548-58delT		IVS	1044	342	702
BRCA1	9	667-17	G to T	-	IVS8-17G>T	c.548-17G>T		IVS	2	1	1
BRCA1	9	667-3	del T	-	IVS8-3delT	c.548-3delT		IVS	1	0	1
BRCA1	9	710	C to T	Cys to Cys	C197C	c.591C>T	p.=	Syn	3	1	2
BRCA1	10	713-72	A>G	-	IVS9-72A>G	c.594-72A>G		IVS	1	1	0
BRCA1	10	713-34	T to C	-	IVS9-34T>C	c.594-34T>C		IVS	1	0	1
BRCA1	10	760	A to G	Asp to Gly	D214G	c.641A>G	p.Asp214Gly	M	1	1	0
BRCA1	11	812	G to A	Thr to Thr	T231T	c.693G>A	p.=	Syn	1	0	1
BRCA1	11	855	T to G	Leu to Val	L246V	c.736T>G	p.Leu246Val	M	3	2	1
BRCA1	11	944	C to T	Gly to Gly	G275G	c.825C>T	p.=	Syn	1	0	1
BRCA1	11	1067	C to G	Ser to Arg	S316R	c.948C>G	p.Ser316Arg	M	1	0	1
BRCA1	11	1100	A to G	Thr to Thr	T327T	c.981A>G	p.=	Syn	4	0	4
BRCA1	11	1152	G to T	Asp to Tyr	D345Y	c.1033G>T	p.Asp345Tyr	M	1	1	0
BRCA1	11	1184	G to A	Lys to Lys	K355K	c.1065G>A	p.=	Syn	1	0	1
BRCA1	11	1186	A to G	Gln to Arg	Q356R	c.1067A>G	p.Gln356Arg	M	251	81	170
BRCA1	11	1224	del GAT	del Asp	D369del	c.1105delGAT	p.Asp369del	IFD	1	0	1
BRCA1	11	1256	T to G	Ile to Met	I379M	c.1137T>G	p.Ile379Met	M	1	0	1
BRCA1	11	1575	T to C	Phe to Leu	F486L	c.1456T>C	p.Phe486Leu	M	4	1	3
BRCA1	11	1605	C to T	Arg to Cys	R496C	c.1486C>T	p.Arg496Cys	M	2	1	1

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BRCA1	11	1606	G to A	Arg to His	R496H	c.1487G>A	p.Arg496His	M	4	0	4
BRCA1	11	1728	A to G	Asn to Asp	N537D	c.1609A>G	p.Asn537Asp	M	1	0	1
BRCA1	11	1767	A to C	Asn to His	N550H	c.1648A>C	p.Asn550His	M	3	1	2
BRCA1	11	1831	T to C	Ile to Thr	I571T	c.1712T>C	p.Ile571Thr	M	1	0	1
BRCA1	11	1908	G to A	Glu to Lys	E597K	c.1789G>A	p.Glu597Lys	M	3	0	3
BRCA1	11	1965	del TCT	del Ser	S616del	c.1846delTCT	p.Ser616del	IFD	1	0	1
BRCA1	11	1984	C to T	Ala to Val	A622V	c.1865C>T	p.Ala622Val	M	1	1	0
BRCA1	11	2030	T to C	Thr to Thr	T637T	c.1911T>C	p.=	Syn	1	0	1
BRCA1	11	2090	A to G	Gln to Gln	Q657Q	c.1971A>G	p.=	Syn	3	0	3
BRCA1	11	2196	G to A	Asp to Asn	D693N	c.2077G>A	p.Asp693Asn	M	296	106	190
BRCA1	11	2201	C to T	Ser to Ser	S694S	c.2082C>T	p.=	Syn	1040	337	703
BRCA1	11	2418	A to G	Ser to Gly	S767G	c.2299A>G	p.Ser767Gly	M	1	0	1
BRCA1	11	2430	T to C	Leu to Leu	L771L	c.2311T>C	p.=	Syn	987	321	666
BRCA1	11	2471	G to A	Ser to Ser	S784S	c.2352G>A	p.=	Syn	1	1	0
BRCA1	11	2535	G to A	Ala to Thr	A806T	c.2416G>A	p.Ala806Thr	M	1	1	0
BRCA1	11	2577	A to G	Lys to Glu	K820E	c.2458A>G	p.Lys820Glu	M	5	1	4
BRCA1	11	2592	G to T	Asp to Tyr	D825Y	c.2473G>T	p.Asp825Tyr	M	1	1	0
BRCA1	11	2596	C to A	Thr to Lys	T826K	c.2477C>A	p.Thr826Lys	M	1	0	1
BRCA1	11	2602	del GCT	del Gly 828	G828del	c.2483delGCT	p.Gly828del	IFD	1	0	1
BRCA1	11	2637	A to G	Ser to Gly	S840G	c.2518A>G	p.Ser840Gly	M	1	0	1
BRCA1	11	2640	C to T	Arg to Trp	R841W	c.2521C>T	p.Arg841Trp	M	8	2	6
BRCA1	11	2641	G to A	Arg to Gln	R841Q	c.2522G>A	p.Arg841Gln	M	1	0	1
BRCA1	11	2685	T to C	Tyr to His	Y856H	c.2566T>C	p.Tyr856His	M	1	1	0
BRCA1	11	2715	C to T	Arg to Cys	R866C	c.2596C>T	p.Arg866Cys	M	1	0	1
BRCA1	11	2727	G to C	Ala to Pro	A870P	c.2608G>C	p.Ala870Pro	M	1	1	0
BRCA1	11	2731	C to T	Pro to Leu	P871L	c.2612C>T	p.Pro871Leu	M	1029	333	696
BRCA1	11	2847	C to G	Gln to Glu	Q910E	c.2728C>G	p.Gln910Glu	M	1	0	1
BRCA1	11	2852	A to G	Gly to Gly	G911G	c.2733A>G	p.=	Syn	5	0	5
BRCA1	11	2933	A to G	Pro to Pro	P938P	c.2814A>G	p.=	Syn	2	0	2
BRCA1	11	3003	G to A	Glu to Lys	E962K	c.2884G>A	p.Glu962Lys	M	1	0	1
BRCA1	11	3119	G to A	Glu to Glu	E1000E	c.3000G>A	p.=	Syn	1	1	0
BRCA1	11	3143	G to A	Met to Ile	M1008I	c.3024G>A	p.Met1008Ile	M	4	0	4
BRCA1	11	3232	A to G	Glu to Gly	E1038G	c.3113A>G	p.Glu1038Gly	M	996	324	672
BRCA1	11	3238	G to A	Ser to Asn	S1040N	c.3119G>A	p.Ser1040Asn	M	73	28	45
BRCA1	11	3340	G to C	Arg to Thr	R1074T	c.3221G>C	p.Arg1074Thr	M	1	1	0
BRCA1	11	3415	C to T	Pro to Leu	P1099L	c.3296C>T	p.Pro1099Leu	M	1	0	1
BRCA1	11	3421	G to A	Ser to Asn	S1101N	c.3302G>A	p.Ser1101Asn	M	2	0	2
BRCA1	11	3535	G to T	Ser to Ile	S1139I	c.3416G>T	p.Ser1139Ile	M	1	1	0

Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA1	11	3537	A to G	Ser to Gly	S1140G	c.3418A>G	p.Ser1140Gly	M	2	0	2
BRCA1	11	3573	G to A	Asp to Asn	D1152N	c.3454G>A	p.Asp1152Asn	M	1	0	1
BRCA1	11	3667	A to G	Lys to Arg	K1183R	c.3548A>G	p.Lys1183Arg	M	998	328	670
BRCA1	11	3719	G to C	Gln to His	Q1200H	c.3600G>C	p.Gln1200His	M	2	2	0
BRCA1	11	3727	G to A	Arg to Gln	R1203Q	c.3608G>A	p.Arg1203Gln	M	1	0	1
BRCA1	11	3827	T to G	Asn to Lys	N1236K	c.3708T>G	p.Asn1236Lys	M	1	0	1
BRCA1	11	3836	T to A	Ser to Ser	S1239S	c.3717T>A	p.=	Syn	1	1	0
BRCA1	11	3864	A to C	Thr to Pro	T1249P	c.3745A>C	p.Thr1249Pro	M	1	0	1
BRCA1	11	3916	G to C	Ser to Thr	S1266T	c.3797G>C	p.Ser1266Thr	M	2	1	1
BRCA1	11	3942	A to G	Ile to Val	I1275V	c.3823A>G	p.Ile1275Val	M	1	0	1
BRCA1	11	4158	A to G	Arg to Gly	R1347G	c.4039A>G	p.Arg1347Gly	M	16	5	11
BRCA1	11	4166	G to T	Thr to Thr	T1349T	c.4047G>T	p.=	Syn	1	0	1
BRCA1	11	4215+3	A to G	-	IVS11+3A>G	c.4096+3A>G		IVS	1	1	0
BRCA1	11	4215+39	T to C	-	IVS11+39T>C	c.4096+39T>C		IVS	1	0	1
BRCA1	12	4216-28	A to G	-	IVS11-28A>G	c.4097-28A>G		IVS	1	1	0
BRCA1	12	4216-11	T to C	-	IVS11-11T>C	c.4097-11T>C		IVS	1	0	1
BRCA1	12	4230	G to C	Gly to Arg	G1371R	c.4111G>C	p.Gly1371Arg	M	1	1	0
BRCA1	12	4291	T to C	Ile to Thr	I1391T	c.4172T>C	p.Ile1391Thr	M	1	1	0
BRCA1	12	4304+20	del 18	-	IVS12+20del18	c.4185+20del18		IVS	1	0	1
BRCA1	12	4304+21	del TG	-	IVS12+21delTG	c.4185+21delTG		IVS	1	1	0
BRCA1	12	4304+37	del TG	-	IVS12+37delTG	c.4185+37delTG		IVS	2	2	0
BRCA1	13	4427	C to T	Ser to Ser	S1436S	c.4308C>T	p.=	Syn	1005	323	682
BRCA1	13	4476+17	A to G	-	IVS13+17A>G	c.4357+17A>G		IVS	1	0	1
BRCA1	14	4477-37	G to A	-	IVS13-37G>A	c.4358-37G>A		IVS	1	1	0
BRCA1	14	4477-28	T to C	-	IVS13-28T>C	c.4358-28T>C		IVS	1	1	0
BRCA1	14	4477-10	C to T	-	IVS13-10C>T	c.4358-10C>T		IVS	2	1	1
BRCA1	14	4529	A to T	Glu to Asp	E1470D	c.4410A>T	p.Glu1470Asp	M	1	0	1
BRCA1	14	4603+14	A to G	-	IVS14+14A>G	c.4484+14A>G		IVS	2	0	2
BRCA1	14	4603+61	G to T	-	IVS14+61G>T	c.4484+61G>T		IVS	1	0	1
BRCA1	15	4654	G to T	Ser to Ile	S1512I	c.4535G>T	p.Ser1512Ile	M	18	3	15
BRCA1	15	4755	G to A	Asp to Asn	D1546N	c.4636G>A	p.Asp1546Asn	M	2	0	2
BRCA1	16	4869	G to T	Ala to Ser	A1584S	c.4750G>T	p.Ala1584Ser	M	1	1	0
BRCA1	16	4890	G to A	Gly to Ser	G1591S	c.4771G>A	p.Gly1591Ser	M	1	1	0
BRCA1	16	4931	A to G	Gln to Gln	Q1604Q	c.4812A>G	p.=	Syn	7	2	5
BRCA1	16	4956	A to G	Ser to Gly	S1613G	c.4837A>G	p.Ser1613Gly	M	1069	348	721
BRCA1	16	5002	T to C	Met to Thr	M1628T	c.4883T>C	p.Met1628Thr	M	1	0	1
BRCA1	16	5075	G to A	Met to Ile	M1652I	c.4956G>A	p.Met1652Ile	M	61	18	43
BRCA1	17	5106-20	A to G	-	IVS16-20A>G	c.4987-20A>G		IVS	1	0	1



Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA1	17	5124	G to T	Ala to Ser	A1669S	c.5005G>T	p.Ala1669Ser	M	1	0	1
BRCA1	18	5194-53	C to T	-	IVS17-53C>T	c.5075C>T		IVS	114	31	83
BRCA1	20	5313-43	T to C	-	IVS19-43T>C	c.5194-43T>C		IVS	1	0	1
BRCA1	20	5396+60	ins 12	-	IVS20+60ins12	c.5277+60ins12		IVS	11	2	9
BRCA1	20	5396+78	G to A	-	IVS20+78G>A	c.5277+78G>A		IVS	8	4	4
BRCA1	20	5396+102	A to G	-	IVS20+102A>G	c.5277+102A>G		IVS	1	0	1
BRCA1	21	5451+78	C to T	-	IVS21+78C>T	c.5332+78C>T		IVS	2	1	1
BRCA1	21	5451+81	T to C	-	IVS21+81T>C	c.5332+81T>C		IVS	1	1	0
BRCA1	22	5520	G to T	Gly to Cys	G1801C	c.5401G>T	p.Gly1801Cys	M	1	0	1
BRCA1	22	5525+33	A to T	-	IVS22+33A>T	c.5406+33A>T		IVS	2	0	2
BRCA1	22	5525+68	T to C	-	IVS22+68T>C	c.5406+68T>C		IVS	6	2	4
BRCA1	23	5530	T to A	Val to Asp	V1804D	c.5411T>A	p.Val1804Asp	M	1	0	1
BRCA1	24	.5587-10	C to A	-	IVS23-10C>A	c.5468-10C>A		IVS	1	0	1
BRCA1	24	.5587-8	G to T	-	IVS23-8G>T	c.5468-8G>T		IVS	1	0	1
BRCA1	24	5688	C to G	Gln to Glu	Q1857E	c.5569C>G	p.Gln1857Glu	M	1	1	0
BRCA1	24	5747	C to G	-	UGA+36C>G	c.*36C>G		UTR	5	1	4
BRCA1	24	5807	C to T	-	UGA+96C>T	c.*96C>T		UTR	1	1	0
BRCA2	2	203	G to A	-	203G>A	c.-26G>A		UTR	797	269	528
BRCA2	2	218	C to T	-	218C>T	c.-11C>T		UTR	1	0	1
BRCA2	2	295+62	T to G	-	IVS2+62T>G	c.67+62T>G		IVS	13	6	7
BRCA2	3	296-31	A to G	-	IVS2-31A>G	c.68-31A>G		IVS	1	0	1
BRCA2	3	296-7	T to A	-	IVS2-7T>A	c.68-7T>A		IVS	10	1	9
BRCA2	3	353	A to G	Tyr to Cys	Y42C	c.125A>G	p.Tyr42Cys	M	11	3	8
BRCA2	3	407	A to G	Asn to Ser	N60S	c.179A>G	p.Asn60Ser	M	1	1	0
BRCA2	3	451	G to C	Ala to Pro	A75P	c.223G>C	p.Ala75Pro	M	2	2	0
BRCA2	3	495	G to A	Pro to Pro	P89P	c.267G>A	p.=	Syn	1	1	0
BRCA2	4	545-37	insTATG	-	IVS3-37insTATG	c.317-37insTATG		IVS	1	1	0
BRCA2	4	653+33	A to G	-	IVS4+33A>G	c.425+33A>G		IVS	1	1	0
BRCA2	5	654-37	T to A	-	IVS4-37T>A	c.426-37T>A		IVS	1	0	1
BRCA2	5	661	del GTT	del Val	V145del	c.433delGTT	p.Val145del	IFD	1	1	0
BRCA2	5	683	C to A	Thr to Lys	T152K	c.455C>A	p.Thr152Lys	M	1	0	1
BRCA2	5	695	A to G	Asp to Gly	D156G	c.467A>G	p.Asp156Gly	M	1	1	0
BRCA2	6	744+21	A to T	-	IVS6+21A>T	c.516+21A>T		IVS	4	1	3
BRCA2	6	744+54	T to C	-	IVS6+54T>C	c.516+54T>C		IVS	3	2	1
BRCA2	6	744+58	G to A	-	IVS6+58G>A	c.516+58G>A		IVS	1	0	1
BRCA2	7	745-19	C to T	-	IVS6-19C>T	c.517-19C>T		IVS	1	1	0
BRCA2	7	823	G to A	Ala to Thr	A199T	c.595G>A	p.Ala199Thr	M	2	1	1



Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA2	7	859+25	C to T	-	IVS7+25C>T	c.631+25C>T		IVS	4	2	2
BRCA2	8	860-69	T to C	-	IVS7-69T>C	c.632-69T>C		IVS	6	2	4
BRCA2	8	860-64	G to A	-	IVS7-64G>A	c.632-64G>A		IVS	1	0	1
BRCA2	8	860-27	C to G	-	IVS7-27C>G	c.632-27C>G		IVS	1	0	1
BRCA2	9	910-32	A to G	-	IVS8-32A>G	c.682-32A>G		IVS	2	1	1
BRCA2	9	910-12	del TA	-	IVS8-12delTA	c.682-12delTA		IVS	1	1	0
BRCA2	9	997	A to C	Asn to His	N257H	c.769A>C	p.Asn257His	M	1	1	0
BRCA2	10	1093	A to C	Asn to His	N289H	c.865A>C	p.Asn289His	M	148	51	97
BRCA2	10	1184	A to G	Asn to Ser	N319S	c.956A>G	p.Asn319Ser	M	1	0	1
BRCA2	10	1192	A to C	Lys to Gln	K322Q	c.964A>C	p.Lys322Gln	M	1	0	1
BRCA2	10	1206	C to A	Ser to Arg	S326R	c.978C>A	p.Ser326Arg	M	5	1	4
BRCA2	10	1261	A to G	Lys to Glu	K345E	c.1033A>G	p.Lys345Glu	M	1	0	1
BRCA2	10	1268	A to G	Gln to Arg	Q347R	c.1040A>G	p.Gln347Arg	M	1	1	0
BRCA2	10	1342	A to C	Asn to His	N372H	c.1114A>C	p.Asn372His	M	1022	360	662
BRCA2	10	1379	C to T	Ser to Phe	S384F	c.1151C>T	p.Ser384Phe	M	5	2	3
BRCA2	10	1395	G to A	Pro to Pro	P389P	c.1167G>A	p.=	Syn	1	1	0
BRCA2	10	1460	T to C	Ile to Thr	I411T	c.1232T>C	p.Ile411Thr	M	1	0	1
BRCA2	10	1503	A to G	Glu to Glu	E425E	c.1275A>G	p.=	Syn	1	1	0
BRCA2	10	1590	A to G	Lys to Lys	K454K	c.1362A>G	p.=	Syn	3	0	3
BRCA2	10	1593	A to G	Ser to Ser	S455S	c.1365A>G	p.=	Syn	146	51	95
BRCA2	10	1613	A to G	Glu to Gly	E462G	c.1385A>G	p.Glu462Gly	M	1	0	1
BRCA2	10	1675	G to C	Ala to Pro	A483P	c.1447G>C	p.Ala483Pro	M	1	0	1
BRCA2	10	1688	C to A	Ala to Glu	A487E	c.1460C>A	p.Ala487Glu	M	3	0	3
BRCA2	10	1694	C to G	Ser to Cys	S489C	c.1466C>G	p.Ser489Cys	M	3	0	3
BRCA2	10	1700	C to G	Thr to Ser	T491S	c.1472C>G	p.Thr491Ser	M	1	0	1
BRCA2	10	1742	T to C	Ile to Thr	I505T	c.1514T>C	p.Ile505Thr	M	2	0	2
BRCA2	10	2014	G to C	Asp to His	D596H	c.1786G>C	p.Asp596His	M	3	2	1
BRCA2	10	2016	T to C	Asp to Asp	D596D	c.1788T>C	p.=	Syn	4	3	1
BRCA2	10	2020	A to G	Thr to Ala	T598A	c.1792A>G	p.Thr598Ala	M	11	5	6
BRCA2	10	2032	G to A	Gly to Arg	G602R	c.1804G>A	p.Gly602Arg	M	1	1	0
BRCA2	10	2046	G to A	Pro to Pro	P606P	c.1818G>A	p.=	Syn	1	0	1
BRCA2	10	2117	C to T	Thr to Ile	T630I	c.1889C>T	p.Thr630Ile	M	1	0	1
BRCA2	10	2137+23	ins T	-	IVS10+23insT	c.1909+23insT		IVS	5	0	5
BRCA2	11	2139	T to C	Gly to Gly	G637G	c.1911T>C	p.=	Syn	2	1	1
BRCA2	11	2166	C to T	Ser to Ser	S646S	c.1938C>T	p.=	Syn	6	1	5
BRCA2	11	2192	C to G	Pro to Arg	P655R	c.1964C>G	p.Pro655Arg	M	7	2	5
BRCA2	11	2310	T to C	Asn to Asn	N694N	c.2082T>C	p.=	Syn	1	1	0
BRCA2	11	2457	T to C	His to His	H743H	c.2229T>C	p.=	Syn	144	49	95

Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA2	11	2578	A to G	Met to Val	M784V	c.2350A>G	p.Met784Val	M	2	0	2
BRCA2	11	2640	A to G	Glu to Glu	E804E	c.2412A>G	p.=	Syn	1	1	0
BRCA2	11	2778	A to G	Gln to Gln	Q850Q	c.2550A>G	p.=	Syn	1	0	1
BRCA2	11	2937	A to G	Leu to Leu	L903L	c.2709A>G	p.=	Syn	1	0	1
BRCA2	11	3031	G to A	Asp to Asn	D935N	c.2803G>A	p.Asp935Asn	M	1	1	0
BRCA2	11	3069	G to T	Leu to Phe	L947F	c.2841G>T	p.Leu947Phe	M	1	1	0
BRCA2	11	3111	G to A	Gln to Gln	Q961Q	c.2883G>A	p.=	Syn	4	1	3
BRCA2	11	3147	G to A	Ser to Ser	S973S	c.2919G>A	p.=	Syn	1	0	1
BRCA2	11	3185	A to G	Asn to Ser	N986S	c.2957A>G	p.Asn986Ser	M	1	1	0
BRCA2	11	3191	A to C	Asp to Ala	D988A	c.2963A>C	p.Asp988Ala	M	1	1	0
BRCA2	11	3199	A to G	Asn to Asp	N991D	c.2971A>G	p.Asn991Asp	M	148	51	97
BRCA2	11	3283	C to G	Leu to Val	L1019V	c.3055C>G	p.Leu1019Val	M	1	0	1
BRCA2	11	3414	T to C	Pro to Pro	P1062P	c.3186T>C	p.=	Syn	1	0	1
BRCA2	11	3444	A to G	Leu to Leu	L1072L	c.3216A>G	p.=	Syn	1	0	1
BRCA2	11	3492	T to C	Pro to Pro	P1088P	c.3264T>C	p.=	Syn	4	0	4
BRCA2	11	3624	A to G	Lys to Lys	K1132K	c.3396A>G	p.=	Syn	926	318	608
BRCA2	11	3645	G to A	Lys to Lys	K1139K	c.3417G>A	p.=	Syn	1	0	1
BRCA2	11	3648	T to C	Ser to Ser	S1140S	c.3420T>C	p.=	Syn	2	0	2
BRCA2	11	3744	G to A	Ser to Ser	S1172S	c.3516G>A	p.=	Syn	4	0	4
BRCA2	11	3945	A to G	Lys to Lys	K1239K	c.3717A>G	p.=	Syn	1	1	0
BRCA2	11	4035	T to C	Val to Val	V1269V	c.3807T>C	p.=	Syn	651	212	439
BRCA2	11	4042	A to G	Met to Val	M1272V	c.3814A>G	p.Met1272Val	M	1	0	1
BRCA2	11	4062	T to C	His to His	H1278H	c.3834T>C	p.=	Syn	1	0	1
BRCA2	11	4076	T to C	Val to Ala	V1283A	c.3848T>C	p.Val1283Ala	M	1	1	0
BRCA2	11	4097	G to A	Cys to Tyr	C1290Y	c.3869G>A	p.Cys1290Tyr	M	4	1	3
BRCA2	11	4144	G to A	Val to Ile	V1306I	c.3916G>A	p.Val1306Ile	M	1	1	0
BRCA2	11	4289	C to T	Thr to Met	T1354M	c.4061C>T	p.Thr1354Met	M	2	1	1
BRCA2	11	4296	G to A	Leu to Leu	L1356L	c.4068G>A	p.=	Syn	15	6	9
BRCA2	11	4318	A to C	Ile to Leu	I1364L	c.4090A>C	p.Ile1364Leu	M	2	0	2
BRCA2	11	4415	A to G	Gln to Arg	Q1396R	c.4187A>G	p.Gln1396Arg	M	1	1	0
BRCA2	11	4452	G to A	Gln to Gln	Q1408Q	c.4224G>A	p.=	Syn	1	1	0
BRCA2	11	4469	C to T	Thr to Met	T1414M	c.4241C>T	p.Thr1414Met	M	1	0	1
BRCA2	11	4486	G to T	Asp to Tyr	D1420Y	c.4258G>T	p.Asp1420Tyr	M	31	6	25
BRCA2	11	4587	A to G	Lys to Lys	K1453K	c.4359A>G	p.=	Syn	1	0	1
BRCA2	11	4664	G to C	Ser to Thr	S1479T	c.4436G>C	p.Ser1479Thr	M	2	0	2
BRCA2	11	4789	C to T	Leu to Leu	L1521L	c.4561C>T	p.=	Syn	1	1	0
BRCA2	11	4791	G to A	Leu to Leu	L1521L	c.4563G>A	p.=	Syn	5	2	3
BRCA2	11	4813	G to A	Gly to Arg	G1529R	c.4585G>A	p.Gly1529Arg	M	2	2	0

Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA2	11	4842	T to C	Ser to Ser	S1538S	c.4614T>C	p.=	Syn	1	0	1
BRCA2	11	4909	C to A	His to Asn	H1561N	c.4681C>A	p.His1561Asn	M	2	1	1
BRCA2	11	4995	A to G	Pro to Pro	P1589P	c.4767A>G	p.=	Syn	1	1	0
BRCA2	11	5042	T to C	Val to Ala	V1605A	c.4814T>C	p.Val1605Ala	M	1	0	1
BRCA2	11	5058	G to A	Val to Val	V1610V	c.4830G>A	p.=	Syn	1	1	0
BRCA2	11	5093	G to C	Arg to Thr	R1622T	c.4865G>C	p.Arg1622Thr	M	1	0	1
BRCA2	11	5155	G to A	Val to Ile	V1643I	c.4927G>A	p.Val1643Ile	M	1	1	0
BRCA2	11	5248	A to G	Ser to Gly	S1674G	c.5020A>G	p.Ser1674Gly	M	1	1	0
BRCA2	11	5279	C to G	Thr to Ser	T1684S	c.5051C>G	p.Thr1684Ser	M	1	0	1
BRCA2	11	5419	C to T	His to Tyr	H1731Y	c.5191C>T	p.His1731Tyr	M	1	0	1
BRCA2	11	5426	C to T	Ser to Phe	S1733F	c.5198C>T	p.Ser1733Phe	M	1	1	0
BRCA2	11	5427	C to T	Ser to Ser	S1733S	c.5199C>T	p.=	Syn	18	5	13
BRCA2	11	5547	G to A	Glu to Glu	E1773E	c.5319G>A	p.=	Syn	1	0	1
BRCA2	11	5606	A to G	Asn to Ser	N1793S	c.5378A>G	p.Asn1793Ser	M	1	0	1
BRCA2	11	5646	A to G	Glu to Glu	E1806E	c.5418A>G	p.=	Syn	3	0	3
BRCA2	11	5723	C to A	Ser to Tyr	S1832Y	c.5495C>A	p.Ser1832Tyr	M	1	0	1
BRCA2	11	5806	A to G	Lys to Glu	K1860E	c.5578A>G	p.Lys1860Glu	M	1	0	1
BRCA2	11	5862	C to G	Asn to Lys	N1878K	c.5634C>G	p.Asn1878Lys	M	2	2	0
BRCA2	11	5863	G to A	Glu to Lys	E1879K	c.5635G>A	p.Glu1879Lys	M	1	1	0
BRCA2	11	5868	T to G	Asn to Lys	N1880K	c.5640T>G	p.Asn1880Lys	M	3	2	1
BRCA2	11	5887	A to G	Thr to Ala	T1887A	c.5659A>G	p.Thr1887Ala	M	1	0	1
BRCA2	11	5896	A to G	Met to Val	M1890V	c.5668A>G	p.Met1890Val	M	1	0	1
BRCA2	11	5911	G to A	Glu to Lys	E1895K	c.5683G>A	p.Glu1895Lys	M	1	0	1
BRCA2	11	5932	G to A	Asp to Asn	D1902N	c.5704G>A	p.Asp1902Asn	M	1	0	1
BRCA2	11	5972	C to T	Thr to Met	T1915M	c.5744C>T	p.Thr1915Met	M	108	30	78
BRCA2	11	5996	A to C	Asp to Ala	D1923A	c.5768A>C	p.Asp1923Ala	M	1	0	1
BRCA2	11	6013	A to G	Ile to Val	I1929V	c.5785A>G	p.Ile1929Val	M	1	0	1
BRCA2	11	6107	G to A	Cys to Tyr	C1960Y	c.5879G>A	p.Cys1960Tyr	M	1	1	0
BRCA2	11	6172	A to G	Ser to Gly	S1982G	c.5944A>G	p.Ser1982Gly	M	1	0	1
BRCA2	11	6328	C to T	Arg to Cys	R2034C	c.6100C>T	p.Arg2034Cys	M	21	6	15
BRCA2	11	6347	T to A	Ile to Lys	I2040K	c.6119T>A	p.Ile2040Lys	M	1	1	0
BRCA2	11	6545	T to C	Leu to Pro	L2106P	c.6317T>C	p.Leu2106Pro	M	1	0	1
BRCA2	11	6550	C to T	Arg to Cys	R2108C	c.6322C>T	p.Arg2108Cys	M	2	1	1
BRCA2	11	6551	G to A	Arg to His	R2108H	c.6323G>A	p.Arg2108His	M	7	2	5
BRCA2	11	6575	A to G	His to Arg	H2116R	c.6347A>G	p.His2116Arg	M	2	1	1
BRCA2	11	6640	G to T	Val to Phe	V2138F	c.6412G>T	p.Val2138Phe	M	2	1	1
BRCA2	11	6683	C to A	Ser to Tyr	S2152Y	c.6455C>A	p.Ser2152Tyr	M	1	0	1
BRCA2	11	6693	C to T	Leu to Leu	L2155L	c.6465C>T	p.=	Syn	1	1	0

Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA2	11	6717	A to G	Gln to Gln	Q2163Q	c.6489A>G	p.=	Syn	1	1	0
BRCA2	11	6741	C to G	Val to Val	V2171V	c.6513C>G	p.=	Syn	4	2	2
BRCA2	11	6841	G to A	Val to Met	V2205M	c.6613G>A	p.Val2205Met	M	1	0	1
BRCA2	11	6976	A to G	Thr to Ala	T2250A	c.6748A>G	p.Thr2250Ala	M	2	1	1
BRCA2	11	7049	G to T	Gly to Val	G2274V	c.6821G>T	p.Gly2274Val	M	1	0	1
BRCA2	11	7069+11	A to G	-	IVS11+11A>G	c.6841+11A>G		IVS	1	0	1
BRCA2	11	7069+33	A to G	-	IVS11+33A>G	c.6841+33A>G		IVS	1	1	0
BRCA2	11	7069+36	A to G	-	IVS11+36A>G	c.6841+36A>G		IVS	1	1	0
BRCA2	12	7070-73	T to A	-	IVS11-73T>A	c.6842-73T>A		IVS	1	1	0
BRCA2	12	7070-22	T to A	-	IVS11-22T>A	c.6842-22T>A		IVS	1	1	0
BRCA2	12	7081	A to G	Ile to Val	I2285V	c.6853A>G	p.Ile2285Val	M	4	0	4
BRCA2	12	7149	A to G	Ser to Ser	S2307S	c.6921A>G	p.=	Syn	1	0	1
BRCA2	13	7171	A to C	Ile to Leu	I2315L	c.6943A>C	p.Ile2315Leu	M	1	0	1
BRCA2	13	7235+4	A to G	-	IVS13+4A>G	c.7007+4A>G		IVS	1	0	1
BRCA2	13	7235+52	C to A	-	IVS13+52C>A	c.7007+52C>A		IVS	1	0	1
BRCA2	14	7236-62	A to G	-	IVS13-62A>G	c.7008-62A>G		IVS	1	1	0
BRCA2	14	7245	G to C	Lys to Asn	K2339N	c.7017G>C	p.Lys2339Asn	M	2	0	2
BRCA2	14	7275	T to A	Phe to Leu	F2349L	c.7047T>A	p.Phe2349Leu	M	1	1	0
BRCA2	14	7280	C to G	Ala to Gly	A2351G	c.7052C>G	p.Ala2351Gly	M	1	1	0
BRCA2	14	7285	G to C	Gly to Arg	G2353R	c.7057G>C	p.Gly2353Arg	M	1	1	0
BRCA2	14	7378	C to A	Gln to Lys	Q2384K	c.7150C>A	p.Gln2384Lys	M	1	0	1
BRCA2	14	7460	A to C	Lys to Thr	K2411T	c.7232A>C	p.Lys2411Thr	M	1	0	1
BRCA2	14	7470	A to G	Ser to Ser	S2414S	c.7242A>G	p.=	Syn	743	245	498
BRCA2	14	7547	A to G	His to Arg	H2440R	c.7319A>G	p.His2440Arg	M	3	1	2
BRCA2	14	7625	C to T	Ala to Val	A2466V	c.7397C>T	p.Ala2466Val	M	3	1	2
BRCA2	14	7641	A to G	Thr to Thr	T2471T	c.7413A>G	p.=	Syn	1	0	1
BRCA2	14	7663+5	T to C	-	IVS14+5T>C	c.7435+5T>C		IVS	1	0	1
BRCA2	14	7663+23	A to G	-	IVS14+23A>G	c.7435+23A>G		IVS	1	0	1
BRCA2	14	7663+53	C to T	-	IVS14+53C>T	c.7435+53C>T		IVS	2	2	0
BRCA2	15	7697	T to C	Ile to Thr	I2490T	c.7469T>C	p.Ile2490Thr	M	11	5	6
BRCA2	15	7778	C to G	Thr to Ser	T2517S	c.7550C>G	p.Thr2517Ser	M	1	0	1
BRCA2	15	7845+15	A to G	-	IVS15+15A>G	c.7617+15A>G		IVS	1	0	1
BRCA2	15	7845+17	C to T	-	IVS15+17C>T	c.7617+17C>T		IVS	1	0	1
BRCA2	16	7854	G to A	Thr to Thr	T2542T	c.7626G>A	p.=	Syn	2	0	2
BRCA2	16	8033+6	C to G	-	IVS16+6C>G	c.7805+6C>G		IVS	2	1	1
BRCA2	17	8034-40	A to G	-	IVS16-40A>G	c.7806-40A>G		IVS	4	1	3
BRCA2	17	8034-39	A to G	-	IVS16-39A>G	c.7806-39A>G		IVS	1	1	0
BRCA2	17	8034-14	T to C	-	IVS16-14T>C	c.7806-14T>C		IVS	1122	391	731

Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA2	17	8106	G to C	Trp to Cys	W2626C	c.7878G>C	p.Trp2626Cys	M	1	1	0
BRCA2	17	8107	A to T	Ile to Phe	I2627F	c.7879A>T	p.Ile2627Phe	M	1	0	1
BRCA2	17	8142	T to G	Phe to Leu	F2638L	c.7914T>G	p.Phe2638Leu	M	1	0	1
BRCA2	17	8204+35	C to A	-	IVS17+35C>A	c.7976+35C>A		IVS	2	0	2
BRCA2	17	8204+36	G to A	-	IVS17+36G>A	c.7976+36G>A		IVS	1	0	1
BRCA2	18	8216	A to T	Glu to Val	E2663V	c.7988A>T	p.Glu2663Val	M	1	0	1
BRCA2	18	8222	A to G	Asp to Gly	D2665G	c.7994A>G	p.Asp2665Gly	M	2	1	1
BRCA2	18	8255	T to C	Met to Thr	M2676T	c.8027T>C	p.Met2676Thr	M	1	0	1
BRCA2	18	8267	A to G	Asp to Gly	D2680G	c.8039A>G	p.Asp2680Gly	M	1	0	1
BRCA2	18	8318	G to A	Ser to Asn	S2697N	c.8090G>A	p.Ser2697Asn	M	1	1	0
BRCA2	18	8362	G to A	Asp to Asn	D2712V	c.8134G>A	p.Asp2712Asn	M	1	1	0
BRCA2	18	8377	G to T	Ala to Ser	A2717S	c.8149G>T	p.Ala2717Ser	M	6	3	3
BRCA2	18	8410	G to A	Val to Ile	V2728I	c.8182G>A	p.Val2728Ile	M	10	2	8
BRCA2	19	8688	A to C	Val to Val	V2820V	c.8460A>C	p.=	Syn	7	3	4
BRCA2	20	8795	A to C	Glu to Ala	E2856A	c.8567A>C	p.Glu2856Ala	M	15	4	11
BRCA2	20	8797	G to A	Ala to Thr	A2857T	c.8569G>A	p.Ala2857Thr	M	1	0	1
BRCA2	20	8860+14	T to C	-	IVS20+14T>C	c.8632+14T>C		IVS	1	0	1
BRCA2	20	8860+143	insC	-	IVS20+143insC	c.8632+143insC		IVS	1	1	0
BRCA2	21	8861-16	C to G	-	IVS20-16C>G	c.8633-16C>G		IVS	2	0	2
BRCA2	21	8875	C to T	Pro to Ser	P2883S	c.8647C>T	p.Pro2883Ser	M	1	0	1
BRCA2	21	8982+75	A to G	-	IVS21+75A>G	c.8754+75A>G		IVS	1	0	1
BRCA2	22	9058	A to T	Ile to Phe	I2944F	c.8830A>T	p.Ile2944Phe	M	2	1	1
BRCA2	22	9078	G to T	Lys to Asn	K2950N	c.8850G>T	p.Lys2950Asn	M	3	3	0
BRCA2	22	9079	G to A	Ala to Thr	A2951T	c.8851G>A	p.Ala2951Thr	M	22	10	12
BRCA2	22	9130	A to C	Thr to Pro	T2968P	c.8902A>C	p.Thr2968Pro	M	2	1	1
BRCA2	22	9133	G to A	Val to Met	V2969M	c.8905G>A	p.Val2969Met	M	1	0	1
BRCA2	23	9332	A to G	Tyr to Cys	Y3035C	c.9104A>G	p.Tyr3035Cys	M	2	1	1
BRCA2	24	9382	C to T	Arg to Trp	R3052W	c.9154C>T	p.Arg3052Trp	M	1	0	1
BRCA2	24	9463	G to A	Val to Ile	V3079I	c.9235G>A	p.Val3079Ile	M	1	0	1
BRCA2	24	9484+28	del T	-	IVS24+28delT	c.9256+28delT		IVS	2	2	0
BRCA2	25	9485-113	T to G	-	IVS24-113T>G	c.9257-113T>G		IVS	5	0	5
BRCA2	25	9485-110	A to G	-	IVS24-110A>G	c.9257-110A>G		IVS	1	0	1
BRCA2	25	9485-83	G to A	-	IVS24-83G>A	c.9257-83G>A		IVS	18	8	10
BRCA2	25	9485-49	T to C	-	IVS24-49T>C	c.9257-49T>C		IVS	1	1	0
BRCA2	25	9485-16	T to C	-	IVS24-16T>C	c.9257-16T>C		IVS	34	8	26
BRCA2	25	9520	T to C	Tyr to His	Y3098H	c.9292T>C	p.Tyr3098His	M	1	0	1
BRCA2	25	9529	C to T	Leu to Leu	L3101L	c.9301C>T	p.=	Syn	1	0	1
BRCA2	25	9594	A to G	Ala to Ala	A3122A	c.9366A>G	p.=	Syn	1	0	1

Gene	Exon	NT <sup>1</sup>	Base Ch	AA Change	BIC <sup>2</sup>	HGV <sup>3</sup> DNA	HGV protein	Type <sup>4</sup>	Total <sup>5</sup>	CBC	UBC
BRCA2	25	9639	T to G	Thr to Thr	T3137T	c.9411T>G	p.=	Syn	1	1	0
BRCA2	26	9741	A to G	Ile to Met	I3171M	c.9513A>G	p.Ile3171Met	M	1	1	0
BRCA2	26	9834	G to C	Pro to Pro	P3202P	c.9606G>C	p.=	Syn	1	1	0
BRCA2	26	9876+54	G to A	-	IVS26+54G>A	c.9648+54G>A		IVS	2	1	1
BRCA2	26	9876+84	G to A	-	IVS26+84G>A	c.9648+84G>A		IVS	12	3	9
BRCA2	26	9876+106	del T	-	IVS26+106delT	c.9648+106delT		IVS	5	2	3
BRCA2	27	9877-67	del TTAC	-	IVS26-67del4	c.9649-67delTTAC		IVS	1	0	1
BRCA2	27	9877-19	G to A	-	IVS26-19G>A	c.9649-19G>A		IVS	2	0	2
BRCA2	27	9958	G to A	Val to Ile	V3244I	c.9730G>A	p.Val3244Ile	M	2	1	1
BRCA2	27	10103	C to T	Pro to Leu	P3292L	c.9875C>T	p.Pro3292Leu	M	1	0	1
BRCA2	27	10204	A to T	Lys to stop	K3326X	c.9976A>T	p.Lys3326X	N	40	9	31
BRCA2	27	10273	A to G	Thr to Ala	T3349A	c.10045A>G	p.Thr3349Ala	M	1	0	1
			del C ins								
BRCA2	27	10323	11	Stop 3369	10323delCins11	c.10095delCins11	p.Ser3366fs	F	2	1	1
BRCA2	27	10338	G to A	Arg to Arg	R3370R	c.10110G>A	p.=	Syn	8	3	5
BRCA2	27	10339	A to G	Thr to Ala	T3371A	c.10111A>G	p.Thr3371Ala	M	3	0	3
BRCA2	27	10378	C to T	Arg to Stop	R3384X	c.10150C>T	p.Arg3384X	N	1	1	0
BRCA2	27	10431	G to A	Thr to Thr	T3401T	c.10203G>A	p.=	Syn	1	0	1
BRCA2	27	10462	A to G	Ile to Val	I3412V	c.10234A>G	p.Ile3412Val	M	28	10	18
BRCA2	27	10516	C to T	3' UTR	UAA+31C>T	c.*31C>T		UTR	1	0	1

<sup>1</sup> BRCA1: nucleotide number according to GenBank U14680.1. <http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?db=nucleotide&val=555931>

<sup>1</sup> BRCA2: nucleotide number according to GenBank NM\_000059.1. <http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?db=nucleotide&val=4502450>

<sup>2</sup> BIC (Breast Cancer Information Core). <http://research.nhgri.nih.gov/bic/>

<sup>3</sup> HGVS (Human Genome Variation Society). <http://www.hgvs.org/mutnomen/recs.html>

<sup>4</sup> Type: F=frame shift, M=missense, S=splice site, N=nonsense

<sup>5</sup> Number of observations

mutations not earlier described in BIC (by November 2008)





Gene	Exon	NT <sup>1</sup>	Base Ch	BIC <sup>2</sup>	Type <sup>3</sup>	AGVGD <sup>4</sup> Sea urchin			AGVGD Pufferfish			AGVGD Xenopus		
						Class <sup>5</sup>	GV	GD	Class	GV	GD	Class	GV	GD
BRCA1	11	1256	T to G	I379M	M	C0	120.48	0.00	C0	92.35	0.00	C0	89.28	0.00
BRCA1	11	1575	T to C	F486L	M	C0	225.33	0.00	C0	212.92	0.00	C0	212.92	0.00
BRCA1	11	1605	C to T	R496C	M	C0	244.95	56.64	C0	244.95	56.64	C15	139.47	112.77
BRCA1	11	1606	G to A	R496H	M	C0	244.95	0.00	C0	244.95	0.00	C0	139.47	0.00
BRCA1	11	1728	A to G	N537D	M	C0	96.20	0.00	C0	96.20	0.00	C0	96.20	0.00
BRCA1	11	1767	A to C	N550H	M	C0	151.68	0.00	C0	118.33	4.81	C0	79.79	26.81
BRCA1	11	1831	T to C	I571T	M	C0	171.12	0.00	C0	142.37	0.00	C0	142.37	0.00
BRCA1	11	1908	G to A	E597K	M	C0	152.38	0.00	C0	152.38	0.00	C0	128.69	0.00
BRCA1	11	1965	del TCT	S616del	IFD									
BRCA1	11	1984	C to T	A622V	M	C0	224.84	2.03	C0	224.84	2.03	C0	224.84	2.03
BRCA1	11	2030	T to C	T637T	Syn									
BRCA1	11	2090	A to G	Q657Q	Syn									
BRCA1	11	2196	G to A	D693N	M	C0	176.25	0.00	C0	176.25	0.00	C0	170.55	0.00
BRCA1	11	2201	C to T	S694S	Syn									
BRCA1	11	2418	A to G	S767G	M	C0	167.35	29.38	C0	167.35	29.38	C0	167.35	29.38
BRCA1	11	2430	T to C	L771L	Syn									
BRCA1	11	2471	G to A	S784S	Syn									
BRCA1	11	2535	G to A	A806T	M	C0	254.20	0.00	C0	254.20	0.00	C0	155.03	0.00
BRCA1	11	2577	A to G	K820E	M	C0	353.86	0.00	C0	353.86	0.00	C0	262.85	0.00
BRCA1	11	2592	G to T	D825Y	M	C0	353.86	0.00	C0	353.86	0.00	C0	251.13	51.67
BRCA1	11	2596	C to A	T826K	M	C0	353.86	0.00	C0	353.86	0.00	C0	215.31	73.23
BRCA1	11	2602	del GCT	G828del	IFD									
BRCA1	11	2637	A to G	S840G	M	C0	144.64	28.37	C0	144.64	28.37	C0	123.92	28.37
BRCA1	11	2640	C to T	R841W	M	C15	111.49	78.29	C15	111.49	78.29	C15	109.21	85.03
BRCA1	11	2641	G to A	R841Q	M	C0	111.49	0.00	C0	111.49	0.00	C0	109.21	0.00
BRCA1	11	2685	T to C	Y856H	M	C0	209.64	0.00	C0	209.64	0.00	C0	209.02	0.00
BRCA1	11	2715	C to T	R866C	M	C65	0.00	179.53	C65	0.00	179.53	C65	0.00	179.53
BRCA1	11	2727	G to C	A870P	M	C0	152.19	0.00	C0	152.19	0.00	C0	142.37	0.00
BRCA1	11	2731	C to T	P871L	M	C0	97.78	0.00	C0	97.78	0.00	C0	97.78	0.00
BRCA1	11	2847	C to G	Q910E	M	C0	170.63	16.18	C0	145.16	16.18	C0	124.29	16.18
BRCA1	11	2852	A to G	G911G	Syn									
BRCA1	11	2933	A to G	P938P	Syn									
BRCA1	11	3003	G to A	E962K	M	C0	353.86	0.00	C0	353.86	0.00	C0	251.53	0.00
BRCA1	11	3119	G to A	E1000E	Syn									
BRCA1	11	3143	G to A	M1008I	M	C0	232.73	0.00	C0	167.35	0.00	C0	144.08	0.00
BRCA1	11	3232	A to G	E1038G	M	C0	262.60	0.00	C0	262.60	0.00	C0	262.60	0.00

Gene	Exon	NT <sup>1</sup>	Base Ch	BIC <sup>2</sup>	Type <sup>3</sup>	AGVGD <sup>4</sup> Sea urchin			AGVGD Pufferfish			AGVGD Xenopus		
						Class <sup>5</sup>	GV	GD	Class	GV	GD	Class	GV	GD
BRCA1	11	3238	G to A	S1040N	M	C0	247.33	0.00	C0	247.33	0.00	C0	222.77	2.03
BRCA1	11	3340	G to C	R1074T	M	C0	353.86	0.00	C0	353.86	0.00	C0	353.86	0.00
BRCA1	11	3415	C to T	P1099L	M	C0	244.61	50.17	C0	244.61	50.17	C0	209.54	94.04
BRCA1	11	3421	G to A	S1101N	M	C0	276.16	0.00	C0	276.16	0.00	C0	276.16	0.00
BRCA1	11	3535	G to T	S1139I	M	C0	220.50	74.79	C0	220.50	74.79	C15	90.49	89.28
BRCA1	11	3537	A to G	S1140G	M	C0	353.86	0.00	C0	353.86	0.00	C0	251.59	0.00
BRCA1	11	3573	G to A	D1152N	M	C0	202.97	0.00	C0	195.29	0.00	C0	195.29	0.00
BRCA1	11	3667	A to G	K1183R	M	C0	165.50	0.00	C0	165.50	0.00	C0	145.74	0.00
BRCA1	11	3719	G to C	Q1200H	M	C0	174.30	0.00	C0	147.42	0.00	C0	147.42	0.00
BRCA1	11	3727	G to A	R1203Q	M	C0	126.59	0.00	C0	57.03	0.00	C0	57.03	0.00
BRCA1	11	3827	T to G	N1236K	M	C0	241.85	0.00	C0	217.19	63.83	C25	46.24	93.76
BRCA1	11	3836	T to A	S1239S	Syn									
BRCA1	11	3864	A to C	T1249P	M	C0	255.91	0.00	C0	254.20	0.00	C0	134.03	0.00
BRCA1	11	3916	G to C	S1266T	M	C0	227.23	6.47	C0	217.79	8.22	C0	82.67	8.47
BRCA1	11	3942	A to G	I1275V	M	C0	261.33	0.00	C0	261.33	0.00	C0	153.36	0.00
BRCA1	11	4158	A to G	R1347G	M	C0	353.86	0.00	C0	353.86	0.00	C0	253.50	0.00
BRCA1	11	4166	G to T	T1349T	Syn									
BRCA1	11	4215+3	A to G	IVS11+3A>G	IVS									
BRCA1	11	4215+39	T to C	IVS11+39T>C	IVS									
BRCA1	12	4216-28	A to G	IVS11-28A>G	IVS									
BRCA1	12	4216-11	T to C	IVS11-11T>C	IVS									
BRCA1	12	4230	G to C	G1371R	M	C0	231.75	41.54	C0	231.75	41.54	C0	112.66	42.00
BRCA1	12	4291	T to C	I1391T	M	C25	30.92	69.84	C25	30.92	69.84	C65	4.86	89.28
BRCA1	12	4304+20	del 18	IVS12+20del18	IVS									
BRCA1	12	4304+21	del TG	IVS12+21delTG	IVS									
BRCA1	12	4304+37	del TG	IVS12+37delTG	IVS									
BRCA1	13	4427	C to T	S1436S	Syn									
BRCA1	13	4476+17	A to G	IVS13+17A>G	IVS									
BRCA1	14	4477-37	G to A	IVS13-37G>A	IVS									
BRCA1	14	4477-28	T to C	IVS13-28T>C	IVS									
BRCA1	14	4477-10	C to T	IVS13-10C>T	IVS									
BRCA1	14	4529	A to T	E1470D	M	C0	105.70	0.00	C0	61.81	44.60	C0	61.81	44.60
BRCA1	14	4603+14	A to G	IVS14+14A>G	IVS									
BRCA1	14	4603+61	G to T	IVS14+61G>T	IVS									
BRCA1	15	4654	G to T	S1512I	M	C0	180.63	62.93	C0	180.63	62.93	C0	180.63	62.93
BRCA1	15	4755	G to A	D1546N	M	C0	353.86	0.00	C0	353.86	0.00	C0	353.86	0.00



Gene	Exon	NT <sup>1</sup>	Base Ch	BIC <sup>2</sup>	Type <sup>3</sup>	AGVGD <sup>4</sup> Sea urchin			AGVGD Pufferfish			AGVGD Xenopus		
						Class <sup>5</sup>	GV	GD	Class	GV	GD	Class	GV	GD
BRCA2	5	654-37	T to A	IVS4-37T>A	IVS									
BRCA2	5	661	del GTT	V145del	IFD									
BRCA2	5	683	C to A	T152K	M	C0	241.08	44.44	C0	109.43	44.44	C15	58.02	73.23
BRCA2	5	695	A to G	D156G	M	C0	204.26	0.00	C0	148.38	0.00	C0	123.56	29.56
BRCA2	6	744+21	A to T	IVS6+21A>T	IVS									
BRCA2	6	744+54	T to C	IVS6+54T>C	IVS									
BRCA2	6	744+58	G to A	IVS6+58G>A	IVS									
BRCA2	7	745-19	C to T	IVS6-19C>T	IVS									
BRCA2	7	823	G to A	A199T	M	C0	110.42	5.07	C0	110.42	5.07	C55	0.00	58.02
BRCA2	7	859+25	C to T	IVS7+25C>T	IVS									
BRCA2	8	860-69	T to C	IVS7-69T>C	IVS									
BRCA2	8	860-64	G to A	IVS7-64G>A	IVS									
BRCA2	8	860-27	C to G	IVS7-27C>G	IVS									
BRCA2	9	910-32	A to G	IVS8-32A>G	IVS									
BRCA2	9	910-12	del TA	IVS8-12delTA	IVS									
BRCA2	9	997	A to C	N257H	M	C0	241.85	0.00	C0	231.11	0.00	C0	95.08	0.00
BRCA2	10	1093	A to C	N289H	M	C0	164.97	0.00	C0	146.96	0.00	C0	85.11	5.07
BRCA2	10	1184	A to G	N319S	M	C0	181.34	6.18	C0	181.34	6.18	C0	181.34	6.18
BRCA2	10	1192	A to C	K322Q	M	C0	56.87	12.95	C0	56.87	12.95	C0	56.87	12.95
BRCA2	10	1206	C to A	S326R	M	C0	184.79	19.25	C0	135.90	70.92	C0	135.90	70.92
BRCA2	10	1261	A to G	K345E	M	C0	184.70	0.00	C0	184.70	0.00	C0	167.48	11.33
BRCA2	10	1268	A to G	Q347R	M	C0	199.95	5.07	C0	199.95	5.07	C0	199.95	5.07
BRCA2	10	1342	A to C	N372H	M	C0	246.50	0.00	C0	242.39	0.00	C0	106.33	0.00
BRCA2	10	1379	C to T	S384F	M	C0	353.86	0.00	C0	244.82	21.28	C0	144.64	21.28
BRCA2	10	1395	G to A	P389P	Syn									
BRCA2	10	1460	T to C	I411T	M	C0	353.86	0.00	C0	243.48	0.00	C0	141.80	0.00
BRCA2	10	1503	A to G	E425E	Syn									
BRCA2	10	1590	A to G	K454K	Syn									
BRCA2	10	1593	A to G	S455S	Syn									
BRCA2	10	1613	A to G	E462G	M	C0	353.86	0.00	C0	353.86	0.00	C0	222.81	53.41
BRCA2	10	1675	G to C	A483P	M	C0	353.86	0.00	C0	244.82	0.00	C0	144.64	0.00
BRCA2	10	1688	C to A	A487E	M	C0	353.86	0.00	C0	242.64	53.41	C0	135.42	54.82
BRCA2	10	1694	C to G	S489C	M	C0	353.86	0.00	C0	266.79	11.33	C0	186.88	92.04
BRCA2	10	1700	C to G	T491S	M	C0	353.86	0.00	C0	243.48	0.00	C0	142.37	0.00
BRCA2	10	1742	T to C	I505T	M	C0	353.86	0.00	C0	353.86	0.00	C0	258.32	0.00
BRCA2	10	2014	G to C	D596H	M	C0	152.22	11.15	C0	85.08	36.57	C0	85.08	36.57

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						Class <sup>5</sup>	GV	GD	Class	GV	GD	Class	GV	GD
BRCA2	10	2016	T to C	D596D	Syn									
BRCA2	10	2020	A to G	T598A	M	C0	170.63	0.00	C0	167.89	0.00	C0	144.64	0.00
BRCA2	10	2032	G to A	G602R	M	C0	174.09	13.17	C0	174.09	13.17	C0	170.79	13.17
BRCA2	10	2046	G to A	P606P	Syn									
BRCA2	10	2117	C to T	T630I	M	C0	353.86	0.00	C0	248.25	28.78	C0	202.47	39.32
BRCA2	10	2137+23	ins T	IVS10+23insT	IVS									
BRCA2	11	2139	T to C	G637G	Syn									
BRCA2	11	2166	C to T	S646S	Syn									
BRCA2	11	2192	C to G	P655R	M	C0	353.86	0.00	C0	353.86	0.00	C0	261.33	13.17
BRCA2	11	2310	T to C	N694N	Syn									
BRCA2	11	2457	T to C	H743H	Syn									
BRCA2	11	2578	A to G	M784V	M	C0	353.86	0.00	C0	353.86	0.00	C0	258.32	0.00
BRCA2	11	2640	A to G	E804E	Syn									
BRCA2	11	2778	A to G	Q850Q	Syn									
BRCA2	11	2937	A to G	L903L	Syn									
BRCA2	11	3031	G to A	D935N	M	C0	353.86	0.00	C0	353.86	0.00	C0	271.19	0.00
BRCA2	11	3069	G to T	L947F	M	C0	353.86	0.00	C0	353.86	0.00	C0	353.86	0.00
BRCA2	11	3111	G to A	Q961Q	Syn									
BRCA2	11	3147	G to A	S973S	Syn									
BRCA2	11	3185	A to G	N986S	M	C0	353.86	0.00	C0	217.19	0.00	C0	217.19	0.00
BRCA2	11	3191	A to C	D988A	M	C0	353.86	0.00	C0	276.16	0.00	C0	276.16	0.00
BRCA2	11	3199	A to G	N991D	M	C0	353.86	0.00	C0	268.84	0.00	C0	266.51	0.00
BRCA2	11	3283	C to G	L1019V	M	C0	246.16	0.00	C0	246.16	0.00	C0	245.67	11.33
BRCA2	11	3414	T to C	P1062P	Syn									
BRCA2	11	3444	A to G	L1072L	Syn									
BRCA2	11	3492	T to C	P1088P	Syn									
BRCA2	11	3624	A to G	K1132K	Syn									
BRCA2	11	3645	G to A	K1139K	Syn									
BRCA2	11	3648	T to C	S1140S	Syn									
BRCA2	11	3744	G to A	S1172S	Syn									
BRCA2	11	3945	A to G	K1239K	Syn									
BRCA2	11	4035	T to C	V1269V	Syn									
BRCA2	11	4042	A to G	M1272V	M	C0	353.86	0.00	C0	353.86	0.00	C0	235.10	0.00
BRCA2	11	4062	T to C	H1278H	Syn									
BRCA2	11	4076	T to C	V1283A	M	C0	353.86	0.00	C0	353.86	0.00	C0	246.16	35.60
BRCA2	11	4097	G to A	C1290Y	M	C0	353.86	0.00	C0	353.86	0.00	C0	248.25	53.70

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						Class <sup>5</sup>	GV	GD	Class	GV	GD	Class	GV	GD
BRCA2	11	4144	G to A	V1306I	M	C0	353.86	0.00	C0	240.36	0.00	C0	96.19	0.00
BRCA2	11	4289	C to T	T1354M	M	C0	353.86	0.00	C0	353.86	0.00	C0	246.14	46.93
BRCA2	11	4296	G to A	L1356L	Syn									
BRCA2	11	4318	A to C	I1364L	M	C0	353.86	0.00	C0	353.86	0.00	C0	260.21	4.86
BRCA2	11	4415	A to G	Q1396R	M	C0	353.86	0.00	C0	233.41	39.51	C0	107.73	39.51
BRCA2	11	4452	G to A	Q1408Q	Syn									
BRCA2	11	4469	C to T	T1414M	M	C0	261.51	0.00	C0	261.51	0.00	C0	261.51	0.00
BRCA2	11	4486	G to T	D1420Y	M	C0	119.25	62.47	C0	119.25	62.47	C15	87.36	87.67
BRCA2	11	4587	A to G	K1453K	Syn									
BRCA2	11	4664	G to C	S1479T	M	C0	254.15	0.00	C0	254.15	0.00	C0	244.14	0.00
BRCA2	11	4789	C to T	L1521L	Syn									
BRCA2	11	4791	G to A	L1521L	Syn									
BRCA2	11	4813	G to A	G1529R	M	C65	0.00	125.13	C65	0.00	125.13	C65	0.00	125.13
BRCA2	11	4842	T to C	S1538S	Syn									
BRCA2	11	4909	C to A	H1561N	M	C0	181.60	0.00	C0	165.75	0.00	C0	165.75	0.00
BRCA2	11	4995	A to G	P1589P	Syn									
BRCA2	11	5042	T to C	V1605A	M	C0	244.82	0.00	C0	144.08	1.01	C0	144.08	1.01
BRCA2	11	5058	G to A	V1610V	Syn									
BRCA2	11	5093	G to C	R1622T	M	C0	353.86	0.00	C0	353.86	0.00	C0	353.86	0.00
BRCA2	11	5155	G to A	V1643I	M	C0	353.86	0.00	C0	353.86	0.00	C0	242.23	0.00
BRCA2	11	5248	A to G	S1674G	M	C0	138.80	0.00	C0	138.80	0.00	C0	138.80	0.00
BRCA2	11	5279	C to G	T1684S	M	C0	171.12	0.00	C0	142.37	0.00	C0	103.68	0.00
BRCA2	11	5419	C to T	H1731Y	M	C0	353.86	0.00	C0	353.86	0.00	C0	231.11	63.24
BRCA2	11	5426	C to T	S1733F	M	C0	353.86	0.00	C15	209.54	110.53	C15	99.44	110.53
BRCA2	11	5427	C to T	S1733S	Syn									
BRCA2	11	5547	G to A	E1773E	Syn									
BRCA2	11	5606	A to G	N1793S	M	C0	353.86	0.00	C0	353.86	0.00	C0	353.86	0.00
BRCA2	11	5646	A to G	E1806E	Syn									
BRCA2	11	5723	C to A	S1832Y	M	C0	353.86	0.00	C0	353.86	0.00	C0	257.76	12.16
BRCA2	11	5806	A to G	K1860E	M	C0	246.80	16.18	C0	244.26	16.18	C0	244.26	16.18
BRCA2	11	5862	C to G	N1878K	M	C0	262.60	0.00	C0	170.08	0.00	C0	170.08	0.00
BRCA2	11	5863	G to A	E1879K	M	C0	98.18	0.00	C0	61.81	0.00	C0	61.81	0.00
BRCA2	11	5868	T to G	N1880K	M	C0	112.83	34.69	C0	112.83	34.69	C0	111.88	36.71
BRCA2	11	5887	A to G	T1887A	M	C0	353.86	0.00	C0	255.29	0.00	C0	144.99	0.00
BRCA2	11	5896	A to G	M1890V	M	C0	353.86	0.00	C0	353.86	0.00	C0	236.57	0.00
BRCA2	11	5911	G to A	E1895K	M	C0	353.86	0.00	C0	276.16	0.00	C0	176.53	0.00

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						Class <sup>5</sup>	GV	GD	Class	GV	GD	Class	GV	GD
BRCA2	11	5932	G to A	D1902N	M	C0	353.86	0.00	C0	353.86	0.00	C0	213.16	22.75
BRCA2	11	5972	C to T	T1915M	M	C0	353.86	0.00	C0	258.55	0.00	C0	162.56	0.00
BRCA2	11	5996	A to C	D1923A	M	C0	353.86	0.00	C0	251.13	0.00	C0	158.16	1.52
BRCA2	11	6013	A to G	I1929V	M	C0	353.86	0.00	C0	353.86	0.00	C0	242.23	0.00
BRCA2	11	6107	G to A	C1960Y	M	C0	353.86	0.00	C0	255.29	4.05	C0	225.33	4.05
BRCA2	11	6172	A to G	S1982G	M	C0	46.24	50.15	C0	46.24	50.15	C0	46.24	50.15
BRCA2	11	6328	C to T	R2034C	M	C0	353.86	0.00	C0	353.86	0.00	C0	262.81	11.33
BRCA2	11	6347	T to A	I2040K	M	C0	353.86	0.00	C0	353.86	0.00	C0	353.86	0.00
BRCA2	11	6545	T to C	L2106P	M	C0	353.86	0.00	C0	353.86	0.00	C0	251.03	0.00
BRCA2	11	6550	C to T	R2108C	M	C0	353.86	0.00	C0	353.86	0.00	C0	267.27	0.00
BRCA2	11	6551	G to A	R2108H	M	C0	353.86	0.00	C0	353.86	0.00	C0	267.27	0.00
BRCA2	11	6575	A to G	H2116R	M	C0	353.86	0.00	C0	258.33	1.62	C0	116.00	1.62
BRCA2	11	6640	G to T	V2138F	M	C0	261.33	21.28	C0	261.33	21.28	C0	261.33	21.28
BRCA2	11	6683	C to A	S2152Y	M	C0	226.57	85.34	C0	215.31	85.34	C15	76.97	92.25
BRCA2	11	6693	C to T	L2155L	Syn									
BRCA2	11	6717	A to G	Q2163Q	Syn									
BRCA2	11	6741	C to G	V2171V	Syn									
BRCA2	11	6841	G to A	V2205M	M	C0	353.86	0.00	C0	238.68	0.00	C0	134.86	0.00
BRCA2	11	6976	A to G	T2250A	M	C0	152.38	0.00	C0	114.06	0.00	C0	114.06	0.00
BRCA2	11	7049	G to T	G2274V	M	C0	257.76	0.00	C0	160.70	0.00	C0	160.70	0.00
BRCA2	11	7069+11	A to G	IVS11+11A>G	IVS									
BRCA2	11	7069+33	A to G	IVS11+33A>G	IVS									
BRCA2	11	7069+36	A to G	IVS11+36A>G	IVS									
BRCA2	12	7070-73	T to A	IVS11-73T>A	IVS									
BRCA2	12	7070-22	T to A	IVS11-22T>A	IVS									
BRCA2	12	7081	A to G	I2285V	M	C0	35.68	26.34	C15	4.86	28.68	C25	0.00	28.68
BRCA2	12	7149	A to G	S2307S	Syn									
BRCA2	13	7171	A to C	I2315L	M	C0	109.50	0.00	C0	109.50	0.00	C0	109.50	0.00
BRCA2	13	7235+4	A to G	IVS13+4A>G	IVS									
BRCA2	13	7235+52	C to A	IVS13+52C>A	IVS									
BRCA2	14	7236-62	A to G	IVS13-62A>G	IVS									
BRCA2	14	7245	G to C	K2339N	M	C0	248.62	0.00	C0	96.44	28.61	C0	96.44	28.61
BRCA2	14	7275	T to A	F2349L	M	C0	261.25	0.00	C0	124.40	0.00	C0	108.52	0.00
BRCA2	14	7280	C to G	A2351G	M	C0	251.03	0.00	C0	154.93	28.37	C0	154.93	28.37
BRCA2	14	7285	G to C	G2353R	M	C0	251.53	5.07	C0	170.54	5.07	C0	170.54	5.07
BRCA2	14	7378	C to A	Q2384K	M	C0	353.86	0.00	C0	266.79	0.00	C0	263.90	0.00







Gene	Exon	NT <sup>1</sup>	Base Ch	BIC <sup>2</sup>	Type <sup>3</sup>	AGVGD <sup>4</sup> Sea urchin			AGVGD Pufferfish			AGVGD Xenopus		
						Class <sup>5</sup>	GV	GD	Class	GV	GD	Class	GV	GD
BRCA2	27	10462	A to G	I3412V	M	C0	353.86	0.00	C0	353.86	0.00	C0	244.82	0.00
BRCA2	27	10516	C to T	UTR	UTR									

<sup>1</sup> BRCA1: nucleotide number according to GenBank U14680.1. <http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?db=nucleotide&val=555931>







<sup>1</sup> BRCA2: nucleotide number according to GenBank NM\_000059.1. <http://www.ncbi.nlm.nih.gov/entrez/viewer.fcgi?db=nucleotide&val=4502450>

<sup>2</sup> BIC (Breast Cancer Information Core). <http://research.nhgri.nih.gov/bic/>. HGVS nomenclature in Supp. Table S2.

<sup>3</sup> Type: F=frame shift, M=missense, N=nonsense, Syn=Synonymous, IFD=in frame deletion, IVS=intervening sequence, UTR=untranslated region

<sup>4</sup> Align Grantham Variation and Grantham Deviation (AGVGD) scores (of October 2008) <http://agvgd.iarc.fr>, aligned down to sea urchin (*Strongylocentrotus purpuratus*), pufferfish (*Tetraodon nigroviridis*) or frog (*Xenopus laevis*)

<sup>5</sup> A-GVGD graded classifiers, ordered from most likely to interfere with function to least likely:

	$GD \geq 65 + \tan(10) \times (GV^{2.5}) \Rightarrow$ Class C65 $\Leftrightarrow$ most likely
	$GD \geq 55 + \tan(10) \times (GV^{2.0}) \Rightarrow$ Class C55
	$GD \geq 45 + \tan(15) \times (GV^{1.7}) \Rightarrow$ Class C45
	$GD \geq 35 + \tan(50) \times (GV^{1.1}) \Rightarrow$ Class C35
	$GD \geq 25 + \tan(55) \times (GV^{0.95}) \Rightarrow$ Class C25
	$GD \geq 15 + \tan(75) \times (GV^{0.6}) \Rightarrow$ Class C15
	Else ( $GD < 15 + \tan(75) \times (GV^{0.6}) \Rightarrow$ Class C0 $\Leftrightarrow$ less likely