

S1 Table. A list of all variants identified in clinical samples

Gene	Coding variant	Protein	dbSNP	BIC/ Clinical importance	BIC/ Clinical classification	HGMD
<i>BRCA1</i>	c.114G>A	p.=	rs1800062	No	Class 1	NA
<i>BRCA1</i>	c.154C>T	p.Leu52Phe	rs80357084	Unknown	Pending	DM?
<i>BRCA1</i>	c.390C>A	p.Tyr130*	rs80356888	Unknown	Class 5	DM
<i>BRCA1</i>	c.922_924delinsT	p.Ser308*	rs80357729	NA	NA	DM
<i>BRCA1</i>	c.1322T>C	p.Ile441Thr		NA	NA	NA
<i>BRCA1</i>	c.2082C>T	p.=	rs1799949	No	Class 1/ Pending	NA
<i>BRCA1</i>	c.2311T>C	p.=	rs16940	No	Class 1/ Pending	NA
<i>BRCA1</i>	c.2566T>C	p.Tyr856His	rs80356892	Unknown	Pending	DM?
<i>BRCA1</i>	c.2612C>T	p.Pro871Leu	rs799917	No	Class 1	DFP
<i>BRCA1</i>	c.3113A>G	p.Glu1038Gly	rs16941	No	Pending	DP
<i>BRCA1</i>	c.3540C>T	p.=		None	None	NA
<i>BRCA1</i>	c.3548A>G	p.Lys1183Arg	rs16942	No	Pending	DP
<i>BRCA1</i>	c.3627dupA	p.Glu1210Argfs*9	rs80357729	Yes	Class 5	DM
<i>BRCA1</i>	c.4308T>C	p.=	rs1060915	No	Class 1	NA
<i>BRCA1</i>	c.4347A>G	p.=	rs80356840	No	Pending	NA
<i>BRCA1</i>	c.4422T>C	p.=	rs756281673	None	None	NA
<i>BRCA1</i>	c.4484+14A>G	p.?	rs80358022	No	Class 1	DM?
<i>BRCA1</i>	c.4837A>G	p.Ser1613Gly	rs1799966	No	Pending	NA
<i>BRCA1</i>	c.4883T>C	p.Met1628Thr	rs4986854	Unknown	Pending	DM?
<i>BRCA2</i>	c.276dupA	p.Ser93fs*8	rs80359345	Yes	Class 5	DM
<i>BRCA2</i>	c.865A>C	p.Asn289Asp	rs766173	No	Pending	DP
<i>BRCA2</i>	c.943T>A	p.Cys315Ser	rs80359065	Unknown	Pending	DM?
<i>BRCA2</i>	c.994delA	p.Ile332Phefs*7	rs80359778	Yes	Class 5	DM

<i>BRCA2</i>	c.1114A>C	p.Asn372His	rs144848	None	None	DFP
<i>BRCA2</i>	c.1365A>G	p.=	rs1801439	No	Class 1	NA
<i>BRCA2</i>	c.1644G>A	p.=	rs55986646	No	Pending	NA
<i>BRCA2</i>	c.2229T>C	p.=	rs1801499	No	Class 1	NA
<i>BRCA2</i>	c.2350A>G	p.Met784Val	rs11571653	Unknown	Pending	DM?
<i>BRCA2</i>	c.2435delA	p.Asn812Ilefs*13	rs80359329	Yes	Class 5	DM
<i>BRCA2</i>	c.2798_2799delCA	p.Thr933Argfs	rs80359348	Yes	Class 5	DM
<i>BRCA2</i>	c.2971A>G	p.Asn991Asp	rs1799944	No	Class 1	NA
<i>BRCA2</i>	c.3195_3198het_delTAAT	p.Asn1066Leufs*10	rs80359375	Yes	Class 5	DM
<i>BRCA2</i>	c.3396A>G	p.=	rs1801406	No	Class 1	NA
<i>BRCA2</i>	c.3462C>G	p.=	rs4986856	NA	NA	NA
<i>BRCA2</i>	c.3744_3747delTGAG	p.Ser1248Argfs*10	rs80359403	Yes	Class 5	DM
<i>BRCA2</i>	c.3807T>C	p.=	rs543304	No	Class 1	NA
<i>BRCA2</i>	c.4563A>G	p.=	rs206075	No	Class 1	NA
<i>BRCA2</i>	c.5270A>G	p.Tyr1757Cys	rs58776466	NA	NA	NA
<i>BRCA2</i>	c.5575_5578delATTA	p.Ile1859Lysfs	rs80359520	Yes	Class 5	NA
<i>BRCA2</i>	c.5785A>G	p.Ile1929Val	rs79538375	No	Pending	DM?
<i>BRCA2</i>	c.5969A>C	p.Asp1990Ala	rs148618542	None	None	DM?
<i>BRCA2</i>	c.6322C>T	p.Arg2108Cys	rs55794205	Unknown	Pending	DM?
<i>BRCA2</i>	c.6325G>A	p.Val2109Ile	rs79456940	Unknown	Pending	DM?
<i>BRCA2</i>	c.6513G>C	p.=	rs206076	NA	NA	NA
<i>BRCA2</i>	c.7188G>T	p.=	rs587780871	NA	NA	NA
<i>BRCA2</i>	c.7242A>G	p.=	rs1799955	No	Class 1	NA
<i>BRCA2</i>	c.7397T>C	p.Val2466Ala	rs169547	NA	NA	DM?
<i>BRCA2</i>	c.7806-14T>C	p.?	rs9534262	No	Pending	FP
<i>BRCA2</i>	c.8187G>T	p.Lys2729Asn	rs80359065	Unknown	Pending	DM?

<i>BRCA2</i>	c.10234A>G	p.Ile3412Val	rs1801426	No	Class 1	DM?
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dbSNP, Short Genetic Variations database; BIC, Breast Cancer Information Core; HGMD, Human Genetic Mutation Database; NA, not available; DM, disease causing mutation; DM?, likely disease causing mutation; DFP, disease associated polymorphism with supporting functional evidence; DP, disease associated polymorphism; FP, *in vitro*/laboratory or *in vivo* functional polymorphism.