

Table S1 BRCA1 Variants

Amino acid change	HG19 Genomic Location	Category in manuscript	Notes	Reference
L30F	41267786G>T	cosmic		Forbes, S. A. et al. COSMIC: mining complete cancer genomes in the Catalogue of Somatic Mutations in Cancer. <i>Nucl. Acids Res.</i> 39 , D945-50 (2011).
L49M	41258539C>A	cosmic		Forbes, S. A. et al. COSMIC: mining complete cancer genomes in the Catalogue of Somatic Mutations in Cancer. <i>Nucl. Acids Res.</i> 39 , D945-50 (2011).
C47W	41258543C>G	cosmic		Forbes, S. A. et al. COSMIC: mining complete cancer genomes in the Catalogue of Somatic Mutations in Cancer. <i>Nucl. Acids Res.</i> 39 , D945-50 (2011).
C47R	41258545T>C	cosmic		Forbes, S. A. et al. COSMIC: mining complete cancer genomes in the Catalogue of Somatic Mutations in Cancer. <i>Nucl. Acids Res.</i> 39 , D945-50 (2011).
E9Q	41276088G>C	cosmic		Forbes, S. A. et al. COSMIC: mining complete cancer genomes in the Catalogue of Somatic Mutations in Cancer. <i>Nucl. Acids Res.</i> 39 , D945-50 (2011).
R7H	41276094C>T	evs	Less than 1% Allele Frequency	Exome Variant Server, NHLBI GO Exome Sequencing Project (ESP), Seattle, WA (URL: http://evs.gs.washington.edu/EVS/) [(October, 2014)].
G98C	41256894C>A	evs	Less than 1% Allele Frequency	Exome Variant Server, NHLBI GO Exome Sequencing Project (ESP), Seattle, WA (URL: http://evs.gs.washington.edu/EVS/) [(October, 2014)].
E23Q	41276047C>G	evs	Less than 1% Allele Frequency	Exome Variant Server, NHLBI GO Exome Sequencing Project (ESP), Seattle, WA (URL: http://evs.gs.washington.edu/EVS/) [(October, 2014)].
Q60R	41258506T>C	evs	Less than 1% Allele Frequency	Exome Variant Server, NHLBI GO Exome Sequencing Project (ESP), Seattle, WA (URL: http://evs.gs.washington.edu/EVS/) [(October, 2014)].
D67Y	41258486C>A	benign	IARC Class 1- Neutral	Easton, D. F. et al. A systematic genetic assessment of 1,433 sequence variants of unknown clinical significance in the BRCA1 and BRCA2 breast cancer-predisposition genes. <i>Amer. J. Hum. Gen.</i> 81 , 873-883 (2007).
R7C	41276095G>A	benign	benign	ClinVar – Sharing Clinical Reports Project
K45Q	41243060A>G	benign	IARC Class 1 – Not pathogenic	Easton, D. F. et al. A systematic genetic assessment of 1,433 sequence variants of unknown clinical significance in the BRCA1 and BRCA2 breast cancer-predisposition genes. <i>Am. J. Hum. Gen.</i> 81 , 873-883 (2007).
I68R	41258482A>C	VUS		ClinVar

Table S1 BRCA1 Variants

I68K	41258482A>T	VUS		ClinVar
D67E	41258484A>C	VUS		ClinVar
I89T	41256920A>G	VUS		ClinVar
I89M	41256919G>C	VUS		ClinVar
C39S	41267762A>T	pathogenic	Pathogenic (Clinical classification)	
K38N	41267763C>A	VUS		ClinVar
T69N	41258479G>T	VUS		ClinVar
T77M	41256956G>A	VUS		ClinVar
M18K	41276061A>T	VUS		ClinVar
M18T	41276061A>G	VUS	IARC Class 4 –likely pathogenic	Easton, D. F. et al. A systematic genetic assessment of 1,433 sequence variants of unknown clinical significance in the BRCA1 and BRCA2 breast cancer-predisposition genes. <i>Am. J. Hum. Gen.</i> 81 , 873-883 (2007).
L28P	41267794A>G	VUS		ClinVar
L49R	41258539A>C	VUS		ClinVar
I31M	41267784G>C	VUS		ClinVar
I21V	41276053T>C	VUS		ClinVar
C47G	41258546A>C	VUS		ClinVar
I42V	41267753T>C	VUS		ClinVar
I15L	41276071T>G	VUS		ClinVar
I15T	41276070A>G	VUS		ClinVar
E33Q	41267780C>G	VUS		ClinVar
L52F	41258531G>A	VUS		ClinVar
V11A	41276082A>G	VUS		ClinVar
T37R	41267767G>C	VUS		ClinVar
C24R	41276044A>G	VUS		ClinVar
C24Y	41276043C>T	VUS		ClinVar
D96N	41256900C>T	VUS		ClinVar
L63F	41258496T>A	VUS		ClinVar
K45N	41258550T>A	VUS		ClinVar
K45T	41267743T>G	VUS		ClinVar
G98R	41256894C>G	VUS		ClinVar
L87V	41256927A>C	VUS		ClinVar
S72R	41256970G>T	VUS		ClinVar
I90T	41256917A>G	VUS		ClinVar
H41R	41267755T>C	pathogenic	Pathogenic	Whiley, P. J. et al. Multifactorial likelihood assessment of BRCA1 and BRCA2 missense variants confirms that BRCA1:c.122A>G(p.His41Arg) is a pathogenic mutation. <i>PLoS ONE</i> 9 , e86836 (2014)
C39R	41267762A>G	pathogenic	Pathogenic	ClinVar – Sharing Clinical Reports Project
C39Y	41267761C>T	pathogenic	IARC Class 5 – Definitely pathogenic	Sweet, K., Senter, L., Pilarski, R., Wei, L. & Toland, A. E. Characterization of BRCA1 ring finger variants of uncertain significance. <i>Br. Ca. Res. Treat.</i> 119 , 737-743 (2010).
R71G	41258474T>C	pathogenic	Pathogenic (causes aberrant splicing)	Vega, A. et al. The R71G BRCA1 is a founder Spanish mutation and leads to aberrant

Table S1 BRCA1 Variants

				splicing of the transcript. <i>Human Mutat.</i> 17 , 520-521 (2001).
R71K	41258473C>T	pathogenic	Pathogenic (causes aberrant splicing)	Zhang, L. et al. BRCA1 R71K missense mutation contributes to cancer predisposition by increasing alternative transcript levels. <i>Br. Ca. Res. Treat.</i> 130 , 1051-1056 (2011).
C61R	41258504A>G	pathogenic	Pathogenic	
C47F	41258545C>A	pathogenic	Pathogenic	
C64R	41258495A>G	pathogenic	Pathogenic	ClinVar – Sharing Clinical Reports Project
C64Y	41258494C>T	pathogenic	Pathogenic	ClinVar – Sharing Clinical Reports Project
C64G	41258495A>C	pathogenic	Pathogenic	ClinVar – Sharing Clinical Reports Project
L22S	41276049A>G	pathogenic	IARC Class 5 – Definitely	Sweet, K., Senter, L., Pilarski, R., Wei, L. & Toland, A. E. Characterization of BRCA1 ring finger variants of uncertain significance. <i>Br. Ca. Res. Treat.</i> 119 , 737-743 (2010).
T37K	41267767G>T	pathogenic	IARC Class 5 – Definitely	Sweet, K., Senter, L., Pilarski, R., Wei, L. & Toland, A. E. Characterization of BRCA1 ring finger variants of uncertain significance. <i>Br. Ca. Res. Treat.</i> 119 , 737-743 (2010).
C44S	41267747A>T	pathogenic	IARC Class 5 – Definitely	Sweet, K., Senter, L., Pilarski, R., Wei, L. & Toland, A. E. Characterization of BRCA1 ring finger variants of uncertain significance. <i>Br. Ca. Res. Treat.</i> 119 , 737-743 (2010).
C44Y	41267746C>T	pathogenic	IARC Class 5 – Definitely	Sweet, K., Senter, L., Pilarski, R., Wei, L. & Toland, A. E. Characterization of BRCA1 ring finger variants of uncertain significance. <i>Br. Ca. Res. Treat.</i> 119 , 737-743 (2010).
C44F	41267746C>A	pathogenic	Pathogenic	ClinVar – Sharing Clinical Reports Project
C61Y	41258503C>T	pathogenic	Pathogenic	ClinVar – Sharing Clinical Reports Project
C61G	41258504A>C	pathogenic	IARC Class 5 – Definitely	Spearman, A. D. et al. Clinically applicable models to characterize BRCA1 and BRCA2 variants of uncertain significance. <i>J. Clin. Oncol.</i> 26 , 5393-5400 (2008).

Table S2 is available for download as an Excel file at www.genetics.org/lookup/suppl/doi:10.1534/genetics.115.175802/-/DC1