






CORRECTION



Correction: The frequency and pathogenicity of BRCA1 and BRCA2 variants in the general Japanese population

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Correct Table 2 is as follows.

In Table 2 of this article PDF, the data in the rows headed “c.7601_7602ins” and “c.7606_7607ins” were mistakenly listed as another rows.

Table 2. Pathogenic variants of *BRCA2* and their frequencies in the general Japanese population (ToMMo:54KJPN)

cDNA	Left aligned	HGVS	Amino acids	Exon No.	Type	dbSNP	ToMMo: 54KJPN		gnomAD Exomes v4.0		gnomAD Genomes v4.0		Pathogenicity
							Allele count	Allele frequency	Total	East Asian	Total	East Asian	
c.632-1G>A			Splice site	8	Splicing	rs81002820	1/108558	9.212 × 10 ⁻⁶				Pathogenic	
c.657_658del	c.658_659del	p.V220fs*4		8	Frameshift	rs80359604	1/108568	9.211 × 10 ⁻⁶	2.352 × 10 ⁻⁵	4.601 × 10 ⁻⁵		Pathogenic	
c.1023_1027del		p.C341*		10	Nonsense		1/108586	9.209 × 10 ⁻⁶				Pathogenic (E)	
c.1274del	c.1278del	p.D427fs*3		10	Frameshift	rs80359274	2/108574	1.842 × 10 ⁻⁵				Pathogenic	
c.1389_1390del		p.V464Gfs*3		10	Frameshift	rs80359283	1/108584	9.209 × 10 ⁻⁶	2.741 × 10 ⁻⁶			Pathogenic	
c.1399del	c.1400del	p.K467Rfs*18		10	Frameshift		1/108582	9.210 × 10 ⁻⁶				Pathogenic (E)	
c.1776T>G		p.Y592*		10	Nonsense		1/108602	9.208 × 10 ⁻⁶				Pathogenic (E)	
c.1794_1798del	c.1796_1800del	p.S599*		10	Nonsense	rs276174813	1/108604	9.208 × 10 ⁻⁶	2.750 × 10 ⁻⁶			Pathogenic	
c.1806del	c.1813del	p.I605Yfs*9		10	Frameshift	rs80359306	6/108604	5.525 × 10 ⁻⁵	9.657 × 10 ⁻⁶	2.524 × 10 ⁻⁵	1.932 × 10 ⁻⁴	Pathogenic	
c.1806_1807del	c.1812_1813del	p.K604Nfs*11		10	Frameshift		1/108604	9.208 × 10 ⁻⁶				Pathogenic (E)	
c.1888dup		p.T630Nfs*6		10	Frameshift	rs80359314	1/108604	9.208 × 10 ⁻⁶	6.860 × 10 ⁻⁷			Pathogenic	
c.2073del	c.2076del	p.K692Nfs*38		11	Frameshift		1/108604	9.208 × 10 ⁻⁶				Pathogenic (E)	
c.2582dup	c.2588dup	p.N863Kfs*18		11	Frameshift	rs80359335	1/108572	9.210 × 10 ⁻⁶	4.865 × 10 ⁻⁶	2.528 × 10 ⁻⁵	6.579 × 10 ⁻⁶	Pathogenic	
c.2622dup		p.V875Cfs*6		11	Frameshift	rs886038076	1/108578	9.210 × 10 ⁻⁶				Pathogenic	
c.2806_2809del	c.2808_2811del	p.A938Pfs*21		11	Frameshift	rs80359351	4/108574	3.684 × 10 ⁻⁵	2.121 × 10 ⁻⁵	2.523 × 10 ⁻⁵	1.314 × 10 ⁻⁵	Pathogenic	
c.2830A>T		p.K944*		11	Nonsense	rs80358533	1/108568	9.211 × 10 ⁻⁶	3.421 × 10 ⁻⁶		6.569 × 10 ⁻⁶	Pathogenic	
c.3086dup		p.M1029Ifs*7		11	Frameshift	rs2072472083	1/108568	9.211 × 10 ⁻⁶				Pathogenic/Likely pathogenic	
c.3846_3847del	c.3847_3848del	p.V1283Kfs*2		11	Frameshift	rs80359405	1/108470	9.219 × 10 ⁻⁶	3.722 × 10 ⁻⁵	7.239 × 10 ⁻⁵	1.925 × 10 ⁻⁴	Pathogenic	
c.3854_3855del	c.3859_3860del	p.N1287*		11	Nonsense	rs80359406	1/108470	9.219 × 10 ⁻⁶				Pathogenic	
c.4020del	c.4021del	p.S1341Qfs*33		11	Frameshift	rs397507702	1/108440	9.222 × 10 ⁻⁶				Pathogenic	
c.4339del		p.V1447*		11	Nonsense	rs80359443	6/108442	5.533 × 10 ⁻⁵				Pathogenic	
c.4461_4462del	c.4464_4465del	p.H1488Qfs*25		11	Frameshift	rs397507720	4/108402	3.690 × 10 ⁻⁵				Pathogenic	
c.4588dup	c.4593dup	p.V1532Sfs*2		11	Frameshift	rs397507731	3/108420	2.767 × 10 ⁻⁵	3.182 × 10 ⁻⁶			Pathogenic	
c.4770_4771del	c.4772_4773del	p.C1591*		11	Nonsense	rs2072512484	2/108396	1.845 × 10 ⁻⁵				Pathogenic	
c.4822G>T		p.E1608*		11	Nonsense	rs1566231194	1/108414	9.224 × 10 ⁻⁶	1.591 × 10 ⁻⁶	2.773 × 10 ⁻⁵		Likely pathogenic	
c.5000C>G		p.S1667*		11	Nonsense	rs397507346	1/108418	9.224 × 10 ⁻⁶				Pathogenic	
c.5066del		p.A1689Efs*17		11	Frameshift		1/108404	9.225 × 10 ⁻⁶				Pathogenic	
c.5067dup	c.5073dup	p.W1692Mfs*3		11	Frameshift	rs80359479	2/108400	1.845 × 10 ⁻⁵	3.895 × 10 ⁻⁵	2.537 × 10 ⁻⁵	1.979 × 10 ⁻⁵	Pathogenic	
c.5211_5214del	c.5213_5216del	p.T1738Ifs*2		11	Frameshift	rs80359493	1/108462	9.220 × 10 ⁻⁶	6.867 × 10 ⁻⁶			Pathogenic	
c.5241_5242insAGCT	c.5243_5246dup	p.Y1749*		11	Nonsense		1/108440	9.222 × 10 ⁻⁶				Pathogenic (E)	
c.5308_5309insCTGGTATTGAG	c.5310_5320dup	p.P1774Lfs*7		11	Frameshift		1/108444	9.221 × 10 ⁻⁶				Pathogenic (E)	
c.5327T>A		p.L1776*		11	Nonsense		1/108430	9.223 × 10 ⁻⁶				Pathogenic (E)	
c.5479_5483del	c.5482_5486del	p.K1828Vfs*4		11	Frameshift	rs80359516	1/108442	9.222 × 10 ⁻⁶				Pathogenic	
c.5574_5577del	c.5576_5579del	p.I1859Kfs*3		11	Frameshift	rs80359520	23/108464	2.121 × 10 ⁻⁴	1.927 × 10 ⁻⁵	1.515 × 10 ⁻⁴	1.316 × 10 ⁻⁵	Pathogenic	
c.5635G>T		p.E1879*		11	Nonsense	rs55996097	2/108466	1.844 × 10 ⁻⁵				Pathogenic	
c.5674del	c.5675del	p.G1892Vfs*17		11	Frameshift	rs1593906164	4/108480	3.687 × 10 ⁻⁵				Pathogenic	
c.5681dup		p.Y1894*		11	Nonsense	rs80359527	1/108478	9.218 × 10 ⁻⁶	2.053 × 10 ⁻⁶	2.521 × 10 ⁻⁵		Pathogenic	
c.5718_5719del	c.5722_5723del	p.L1908Rfs*2		11	Frameshift	rs80359530	1/108508	9.216 × 10 ⁻⁶	1.505 × 10 ⁻⁵	5.042 × 10 ⁻⁵		Pathogenic	
c.5728dup	c.5729dup	p.N1910Kfs*2		11	Frameshift		1/108502	9.216 × 10 ⁻⁶				Pathogenic (E)	
c.6085G>T		p.E2029*		11	Nonsense	rs397507828	1/108546	9.213 × 10 ⁻⁶				Pathogenic	
c.6140dup		p.Y2047*		11	Nonsense		1/108548	9.213 × 10 ⁻⁶				Pathogenic	
c.6402_6406del	c.6405_6409del	p.N2135Kfs*3		11	Frameshift	rs80359584	3/108558	2.763 × 10 ⁻⁵	2.065 × 10 ⁻⁵	7.577 × 10 ⁻⁵	1.314 × 10 ⁻⁵	Pathogenic	
c.6438_6441del	c.6440_6443del	p.H2147Lfs*20		11	Frameshift	rs1566234607	5/108562	4.606 × 10 ⁻⁵				Pathogenic (E)	
c.6482_6485del	c.6486_6489del	p.K2162Nfs*5		11	Frameshift	rs80359598	1/108576	9.210 × 10 ⁻⁶	9.720 × 10 ⁻⁶		1.972 × 10 ⁻⁵	Pathogenic	

Table 2. continued

cDNA	HGVS	Amino acids	Exon No.	Type	dbSNP	ToMMo: 54KJPN		gnomAD Exomes v4.0		gnomAD Genomes v4.0		Pathogenicity
						Allele count	Allele frequency	Total	East Asian	Total	East Asian	
c.6516del		p.V2174Lfs*17	11	Frameshift	rs2072558268	4/108576	3.684 × 10 ⁻⁵					Pathogenic/Likely pathogenic
c.6552del	c.6553del	p.A2185Lfs*6	11	Frameshift	rs80359603	1/108578	9.210 × 10 ⁻⁶	1.373 × 10 ⁻⁶	2.524 × 10 ⁻⁵			Pathogenic
c.6656C>A		p.S2219*	11	Nonsense		1/108588	9.209 × 10 ⁻⁶					Pathogenic
c.6666C>G		p.V2222*	11	Nonsense		1/108690	9.209 × 10 ⁻⁶					Pathogenic (E)
c.6715G>T		p.E2239*	11	Nonsense	rs276174876	1/108600	9.208 × 10 ⁻⁶					Pathogenic
c.6893dup	c.6896dup	p.N2299Kfs*41	12	Frameshift	rs886040933	1/108514	9.215 × 10 ⁻⁶					Conflicting
c.6922A>T		p.K2308*	12	Nonsense		3/108520	2.764 × 10 ⁻⁵					Conflicting
c.6952C>T		p.R2318*	13	Nonsense	rs80358920	37/108476	3.411 × 10 ⁻⁴	1.031 × 10 ⁻⁵	3.548 × 10 ⁻⁴			Pathogenic
c.7585del	c.7586del	p.G2529Afs*22	15	Frameshift	rs2072699136	1/108604	9.208 × 10 ⁻⁶					Pathogenic
c.7601_7602ins ^a		p.S2536_I3418delinsY	15	Deletion		1/108604	9.208 × 10 ⁻⁶					Pathogenic (E)
c.7605_7606insGGAGC		p.S2536Gfs*17	15	Frameshift		1/108604	9.208 × 10 ⁻⁶					Pathogenic (E)
c.7606_7607ins ^b		p.S2536_I3418delinsYPS5SP	15	Deletion		1/108604	9.208 × 10 ⁻⁶					Pathogenic (E)
c.7615C>T		p.Q2639*	15	Nonsense	rs886040720	3/108604	2.762 × 10 ⁻⁵	3.181 × 10 ⁻⁶	2.773 × 10 ⁻⁵			Pathogenic
c.7671_7672del	c.7673_7674del	p.E2558Vfs*7	16	Frameshift	rs80359672	1/108604	9.208 × 10 ⁻⁶	1.591 × 10 ⁻⁶				Pathogenic
c.7806-2A>T		Splice site	17	Splicing	rs81002836	1/108604	9.208 × 10 ⁻⁶					Pathogenic/Likely pathogenic
c.7806-2A>G		Splice site	17	Splicing	rs81002836	1/108604	9.208 × 10 ⁻⁶					Pathogenic
c.8023A>G		p.I2675V	18	Missense	rs397507954	5/108596	4.604 × 10 ⁻⁵	1.591 × 10 ⁻⁶	2.773 × 10 ⁻⁵			Pathogenic
c.8167G>C		p.D2723H	18	Missense	rs41293511	1/108590	9.209 × 10 ⁻⁶	1.368 × 10 ⁻⁵	6.571 × 10 ⁻⁶			Pathogenic
c.8243G>A		p.G2748D	18	Missense	rs80359071	3/108594	2.763 × 10 ⁻⁵	3.420 × 10 ⁻⁶				Pathogenic
c.8504C>A		p.S2835*	20	Nonsense	rs80359102	2/108604	1.842 × 10 ⁻⁵					Pathogenic
c.8629G>T		p.E2877*	20	Nonsense	rs80359121	2/108604	1.842 × 10 ⁻⁵					Pathogenic
c.8713_8716del		p.Y2905Kfs*3	21	Frameshift	rs80359726	1/108604	9.208 × 10 ⁻⁶					Pathogenic
c.9076C>T		p.Q3026*	23	Nonsense	rs80359159	1/108582	9.210 × 10 ⁻⁶	2.053 × 10 ⁻⁶	5.044 × 10 ⁻⁵			Pathogenic
c.9090del	c.9097del	p.T3033Lfs*29	23	Frameshift	rs397507419	2/108586	1.842 × 10 ⁻⁵					Pathogenic
c.9117G>A		p.P3039=	23	Splicing	rs28897756	3/108590	2.763 × 10 ⁻⁵	3.422 × 10 ⁻⁶	6.574 × 10 ⁻⁶			Pathogenic
c.9154C>T		p.R3052W	24	Missense	rs45580035	1/108600	9.208 × 10 ⁻⁶	5.473 × 10 ⁻⁶				Pathogenic
c.9501+1G>T		Splice site	25	Splicing	rs397508058	1/108570	9.211 × 10 ⁻⁶					Pathogenic/Likely pathogenic

^aInsert sequence: TTGCTATTGAGCAAGGCTGTTGATCAGTAGGATGCCGATACCGTAGCCTTTC

^bInsert sequence: ACCCGAGCTTTCACCGTGACCTCGCGGAGTATGAAGCGATGCA

In Table 3, the layout has been changed to identify four tables respectively.

Table 3. Number and position of *BRCA1/2* variants and frequencies based on pathogenic variants

Unique variant No.	<i>BRCA1</i>			<i>BRCA2</i>		
	Total	P/LP	Trunc.	Total	P/LP	Trunc.
Exonic	333	34	4	607	51	14
Splicing	4	2		4	4	
Intronic	7258	1		5575	0	
Total	7711	37	4	6320	55	14
	Average	P/LP		Average	P/LP	
Allele count	108587.4	135	5	108536.7	156	18
	<i>BRCA1</i>			<i>BRCA2</i>		
	ClinVar	+Trunc.		ClinVar	+Trunc.	
Allele frequency	1.243×10^{-3}	1.289×10^{-3}		1.473×10^{-3}	1.603×10^{-3}	
Carrier frequency	2.485×10^{-3}	2.577×10^{-3}		2.873×10^{-3}	3.204×10^{-3}	
	<i>BRCA1 or BRCA2</i>					
	ClinVar	+Trunc.				
Carrier frequency	5.350×10^{-3}	5.772×10^{-3}				

P/LP pathogenic or likely pathogenic in ClinVar, *Trunc.* truncating variants that are not reported as P/LP in ClinVar, but classified as Pathogenic according to ENIGMA criteria. (Frequencies were calculated under the assumption that each variant is independent of each other because allele frequencies are very low and the possibility that one person simultaneously has more than two variants can be ignored.)

The original article has been corrected.