

## Connexin Genes Variants Associated with Non-Syndromic Hearing Impairment: A Systematic Review of the Global Burden

**Table S1.** Connexin genes primers sets used for targeted sequencing.

Gene	Forward/Reverse Primer	Frequency
GJB2	TTGGTGTTCGCTCAGGAAGA/GGCCTACAGGGGTTTCAAAT	8
GJB2	TCTTTCCAGAGCAAACCGC/GACACGAAGATCAGCTGCAG	7
GJB2	CCTATGACAAACTAAGTTGGTTC/GACAGCTGAGCACGGGTTGCCTC	4
GJB2	GCTTACCCAGACTCAGAGAAG/CTACAGGGGTTCAAATGGTTGC	4
GJB2	TCCGTAACTTTCCCAGTCTCCGAGGGAAGAGG/CCCAAGGACGTGTGTTGGTCCAGCCCC	4
GJB2	CCAGGCTGCAAGAACGTGTG/GGGCAATGCGTAAACTGGC	3
GJB2	CTCCCTGTTCTGTCCTAGCT/CTCATCCCTCTCATGTCTC	3
GJB2	TCTTTCCAGAGCAAACCGC/GGGCAATGCGTAAACTGGC	3
GJB2	AGTGGCCATGCACGTGGCCTA/TGATCTCCTCGATGTCCTTAA	2
GJB2	CCAGGCTGCAAGAACGTGT/ACAGCTGAGCACGGGTTG	2
GJB2	CCGGGAAGCTCTGAGGAC/GCAACCGCTCTGGGTCTC	2
GJB2	CTCCCTGTTCTGTCCTAGCT/AGCTGAGCACGGGTTGCCTCA	2
GJB2	CTGTCCTAGCTATGTTCC/TCAGCACGGGTTGCCTCAATC	2
GJB2	GAGGTGTGTAAGAGTTGGTGT/TCTTCTCATGTCTCCGGTAG	2
GJB2	GCCGCCCCCTCCGTAACCTTC/CGTGTGTTGGTCCAGCCCCC	2
GJB2	GCTTACCCAGACTCAGAGAAG/CTTAATCTAACAACCTGGGCAATGC	2
GJB2	GTCTCCCTGTTCTGTCCTA/TCTAACAACCTGGGCAATG	2
GJB2	TCAAAGGAACTAGGAGATCGG/CAAGGACGTGTGTTGGTCCAG	2
GJB2	TCTTTCCAGAGCAAACCGC/CTGGGCAATGCGTAAACTGG	2
GJB2	TCTTTCCAGAGCAAACCGC/TGAGCACGGGTTGCCTCATC	2
GJB2	TGCTTACCCAGACTCAGAGAA/GACTGAGCCTTGACAGCTGAG	2
GJB2	AAGGGGTGGCTCTTCA/GCACTTGGCCTGGGTAA	1
GJB2	AAGTCTCCCTGTTCTGTCCTAG/CCTCATCCCTCTCATGTCTC	1
GJB2	ACACGTTCAAGAGGGTTGG/CCTCATCCCTCTCATGCT	1
GJB2	ACACGTTCAAGAGGGTTGG/GGGAAATGCTAGCGACTGAG	1
GJB2	ACTCATGGGGGCTCAAAGGA/GCAACCGCTCTGGGTCTCGCGGTCCCT	1
GJB2	AGAAGAAGAGGAAGTTCATCA/CCTCTCATGCTGTCTATTTT	1
GJB2	AGAAGAAGAGGAAGTTCATCAAGG/CTGAGGCCTACAGGGGTTT	1
GJB2	AGACTCAGAGAAGTCTCCCTG/GACACGAAGATCAGCTGCAG	1
GJB2	AGACTCAGAGAAGTCTCCCTG/GCCAGTTAAACGCATTGCC	1
GJB2	AGAGTGGTGTTCGCTCAGGA/GACTGAGCCTTGACAGCTGA	1



GJB2	AGCAAACCGCCAGAGTAGAAG/TGATCTTCGTGCCACGCCAG	1
GJB2	AGGCCGACTTTGTCTGCAACA/GTGGGCCGGGACACAAAG	1
GJB2	AGTCTCCCTGTTCTGTCCTA/TGAGCACGGGTTGCCTCATC	1
GJB2	AGTGCCITTCAGCTAACGA/GTGGCATCTGGAGTTTCACC	1
GJB2	CACGCTGCAGACGATCC/GGTGGAGTGTTTGTTAC	1
GJB2	CACTTCCCAAGGCCTCTCCAC/GTACGTCCACCACAGCGAC	1
GJB2	CAGTCTCCGAGGGAAGAGG/AAGGACGTGTGTTGGTCCAG	1
GJB2	CAGTCTCCGAGGGAAGAGG/GCAACCGCTCTGGGTCTC	1
GJB2	CCACGCCAGCGCTCCTAGTG/GAAGATGCTGCTGTGTAGG	1
GJB2	CCAGCATGACCTTACCAG/ATCACTTGAATAAGAAGCCATTTG	1
GJB2	CCAGGCTGCAAGAACGTGTG/GACACGAAGATCAGCTGCAG	1
GJB2	CCAGGCTGCAAGAACGTGTG/RTGAGCACGGGTTGCCTCATC	1
GJB2	CCAGGCTGCAAGAACGTGTG/TGAGCACGGGTTGCCTCATC	1
GJB2	CCCTCCGTAACCTTCCCAGT/CCAAGGACGTGTGTTGGTC	1
GJB2	CCCTCCGTAACCTTCCCAGT/GCAACCGCTCTGGGTCTC	1
GJB2	CCTATGACAAACTAAGTTGGTTC/GCCTCATCCCTCTCATGCTGTC	1
GJB2	CCTCCGTAACCTTCCCAGT/AAGGACGTGTGTTGGTCCAG	1
GJB2	CCTGTGTTGTGTGCATTCGTC/CTCATCCCTCTCATGCTGTC	1
GJB2	CGAAGCCGCCTTCATGTACG/TTAGGGGAGCAGAGCTCCAT	1
GJB2	CGCACTATGCGGAGTACAGA/GGTGGCAGTGGGTCAAGTAG	1
GJB2	CGCGCTCCTCTCCCGACT/TCCTTTGCAGCCACAACGAGGAT	1
GJB2	CGGAGACATGAGAAGAAGAGG/GATCTCCTCGATGTCTTAA	1
GJB2	CGTAACTTCCCAGTCTCCGAGGGAAGAGG/CCCAAGGACGTGTGTTGGTCCAGCC	1
GJB2	CGTAGCACACGTTCTTGCAGCCTG/CGATGCGGACCTTCTGGGTTTG	1
GJB2	CGTCTTTCCAGAGCAAACCG/AGCTCCATTGTGGCATCTGG	1
GJB2	CGTTCGTTCCGATTGGTGTG/CAGAAACGCCCGCTCCAGAA	1
GJB2	CTCATGGGGGCTCAAAGGAACTAGGAGATCGG/GGGGCTGGACCAACACACGTCCTTGGG	1
GJB2	CTCATGTCTCCGGTAGGCCAC/GCAGCATCTTCTCCGGGT	1
GJB2	CTGGTGCTACGATCACTAC/TTCCAGACACTGCAATCATG	1
GJB2	CTTTTCCAGAGCAAACCGC/GGGCAATGCGTTAAACTGGC	1
GJB2	FTCTTTTCCAGAGCAAACCGCC/GACACGAAGATCAGCTGCAG	1
GJB2	GAAGTAGTGATCGTAGCACACGTTCTTGCA	1
GJB2	GAAGTCTCCCTGTTCTGTCTT/TCTAACAACCTGGGCAATGC	1
GJB2	GAGCCTTCGATGCGGACCTT/TCATCCCTCTCATGCTGTC	1
GJB2	GCATTCGTCTTTTCCAGAGC/GGCCTACAGGGGTTTCAAAT	1
GJB2	GCATTCGTCTTTTCCAGAGCA/GAGCCTTCGATGCGGACCTT	1
GJB2	GCCAGGCTGCAAGAACGTGT/GGAGAAGCCGTCGTACATGA	1
GJB2	GCGCAAGCTTTATGGATTGGGGCAGCT/GCGCGGATCCCTAACTGGCTTTTTGAC	1
GJB2	GCTTACCAGACTCAGAGAAG/TGAGCACGGGTTGCCTCATC	1



GJB2	GGGCAATGCGTTAAACTGGC/CCAGGCTGCAAGAACGTGTG	1
GJB2	GGGGAGATGAGCAGGCCGAC/CGGCTGGTGAAGTGCAACGC	1
GJB2	GGGGCTCAAAGGAACTAGGA/AAGGACGTGTGTGGTCC	1
GJB2	GGGGGCACTGGGGAACTCA/GCAGAAAACGCCCGCTCCAGAA	1
GJB2	GGTGAGGTTFTTAAGAGTTGG/CTACGGTGTACTCGAGACGA	1
GJB2	GGTGAGGTTGTGTAAGAGTTGG/TAGCGACTGAGCCTTGACAG	1
GJB2	GTCTCCCTGTTCTGTCTAG/CTTCGAAGATGACCCGGAAG	1
GJB2	GTGCATTTCGTCTTTCCAGAGCA/TTGACAGCTGAGCACGGGTTG	1
GJB2	GTGGCCTACCGGAGACAT/CACTCTTTATCTCCCCCTTG	1
GJB2	GTGTGCATTTCGTCTTTCCAG/GCGACTGAGCCTTGACA	1
GJB2	GTGTTGTGTGCATTTCGTCTTTTC/ACCTTCTGGGTTTTGATCTCCIC	1
GJB2	TACGATGGTTTTTCCTTAATTCT/TTGCATAACTTAGTGAACCTCAGAG	1
GJB2	TATGTCATGTACGACGGCT/TCTAACAACTGGGCAATGC	1
GJB2	TATGTTCTGTGTTGTGTGC/CCTTCTGGGTTTTGATCTCC	1
GJB2	TCAAGGGGGAGATAAAGAGT/TGAGCACGGGTTGCCTCATC	1
GJB2	TCAAGGGGGAGATAAAGAGTG/TGAGCACGGGTTGCCTCATC	1
GJB2	TCAGAGAAGTCTCCCTGTTCTGTCC/TGAGGCCTACAGGGGTTTCAA	1
GJB2	TCCGTAACCTCCCAGTCTCCGAGGGAAGAGG/CCCAAGGACGTGTGTTGGTCCAGCCCC	1
GJB2	TCCGTAACCTTCCCAGTCTCCGAGGGAAGAG/CCCAAGGACGTGTGTTGGTCCAGCCCC	1
GJB2	TCCGTAACCTTCCCAGTCTCCGAGGGAAGAGG/CCCAAGGACGTGTGTTGGTCCAGCCCC	1
GJB2	TCCTGGGGGTGTGA/CCTGGGGGGTGTG	1
GJB2	TCGGCCCCAGTGGTACAG/CTGGGCAATGCGTTAAACTGG	1
GJB2	TCTTTTCCAGAGCAAACCGC/GTTGGGAAATGCTAGCGACT	1
GJB2	TCTTTTCCAGAGCAAACCGC/TGGGCAATGCGTTAAACTGGC	1
GJB2	TCTTTTCCAGAGCAAACCGC/TTGCCTCATCCCTCTCATGTGT	1
GJB2	TCTTTTCCAGAGCAAACCGC3/CTGGGCAATGCGTTAAACTGG	1
GJB2	TCTTTTCCAGAGCAAACCGCC/CACGTGCATGGCCAGTAG	1
GJB2	TGCATCACCTCACATAGGTTA/TATTGGATACTGAATCTGCTG	1
GJB2	TGCTTACCCAGACTCAGAGAA/CGACTGAGCCTTGACAGCTGA	1
GJB2	TGCTTGCTTACCCAGACTCA/CCTCATCCCTCTCATGTGT	1
GJB2	TGGGGAACCTCATGGGGGCTCAAAG/AGGTTCTGGCCGGCAGTCC	1
GJB2	TGGTGCCATGCATGTGCCTAC/CAGAAGTCTCCTTATGACGCAGC	1
GJB2	TGGTGTGTTGCTCAGGAAGAG/TTGTGTAGGTCCACCACAGG	1
GJB2	TGTGCATTTCGTCTTTTCC/CAGATCTTTCCAATGCTGGT	1
GJB2	TGTGCATTTCGTCTTTTCCAG/CAGATCTTTCCAATGCTGGT	1
GJB2	TGTGCATTTCGTCTTTTCCAG/GGGAAATGCTAGCGACTGAG	1
GJB2	TTGGGGCACCTGCAGACGATCCTGGGGAG	1
GJB2	TTGGTGTGTTGCTCAGGAAGA/GGCATCTGGAGTTTACCTG	1
GJB2	TTTGCTCAGGAAGAGATTTAAGCA/GGGTTTTGATCTCCTCGATGTCC	1



GJB6	AGACTAGCAGGGCAGGGAGT/GGTTGGTATTGCCTTCTGGA	1
GJB6	GACTAGCAGGGCAGGGAGT/CTCTTCTCTCCTCGCTGAA	1
GJB6	TCCGTAACCTTCCAGTCTCCGAGGGA AGAGG/CCCAAGGACGTGTGTTGGTCCAGCCCC	1
GJB6	GTCTGTAATATCACCGTGTAC/CTCTCAGGCTACAGAAGGAAC	1
GJB6	GGCAGGGAGTTGAAGTTG/ACTCTCAGGCTACAGAAGGAAC	1
GJB6	CTTGGCCACTTTTGTCTGT/GACCCCTCTATCCGAACCTT	1
GJB6	TTTAGGGCATGATTGGGGTGATT/CACCATGCCGTAGCCTTAACCATTTT	1
GJB6	CTCCTTAGGGCATGATTGG/CCATGCCGTAGCCTTAACCAT	1
GJB6	GACCACTTTTCCCGGTGT/AGGTTGGTATTGCCTTCTGG	1
GJB6	CCTCCAGCTGATCTTCGTCT/GCAGCAGGTAGCACAACTCT	1
GJB6	TGCCACCCCCCAAGTAGAG/TTTCGGTTTCATTCTTTCCCTATT	1
GJB6	GCCCCAACCTTGTGACTGC/GTTGGTATTGCCTTCTGGAG	1
GJB6	GGCAGGGAGTTGAAGTTGTA/ACGTTGTGTATGAATGGAGCA	1
GJB6	GAAAGGAAGTTCGGGCAAGG/CACAATCAAACCTCACTGCCATCTT	1
GJB6	GTTGCTTGTGCTTTTGGTGTAT/AGCCAGAAACAAACCTTACATA	1
GJB3	GCAGGTAGGCAAGCCCCACCAG/GCCACACTGCCCTGCATTCC	1
GJB3	ATTCTCTCAGGTAGGCACGG/TGGTGTGCAGTCAAAGTCC	1
GJB3	CTATTCTCATAACGATGG/TCACTCAGCCCTGTAGGAC	1
GJB3	CCCAGTCCCAGTGTCAAA/TACGACAACGCAGGCAAGA	1
GJB3	TACGATGGTTTTTCCCTAATTCT/TGCATAACTTAGTGAACCTCAGAG	1
GJB3	TATACGTGGTGGCTGCAGAG/CTGCGTTGTCGTACAGCTTG	1
GJB3	TAGGTCGGGCAATGTAGCA/GAACTCAGAACACTGCCTGGT	1
GJB3	CTCGCTGCTGGTCATCCT/CATATGAAGCCATGCCAGA	1
GJB3	TTCTCTACCTGCTGCACAC/GGCAGATGAGGTAGCAGAGC	1
GJB3	AGAGGGTCTGTGTAGTATTG/AGAGGCGGATGTTGGAGATG	1
GJB3	TGCAGCTTGGGAGGAATAAC/CCCCTGTAGGACCTCTCCAC	1
GJB3	TGCTACGACAACACTTCCCC/GGCGCCACCATGAAGTAG	1
GJA1	GAAATACGTGAAACCGTT GG/ CCT GGT GCA CTT TCT ACA GC	1
GJA1	TGCGGTCTACACCTGCAAGA/ACCAAGGACACCACCAGCAT	1
GJA1	CGTGAAACCGTTGGTAGTATTT/CCTCCACCGGATCAAAATTA	1
GJA1	GGAGTTCAATCACCTTGGCGT/TCTTGTGCTTTCAAGCCTGT	1
GJA1	TTGCAATCTGTGATCCTTGA/CCTGGTGCACCTTCTACAGC	1
GJB4	TCAATCGCACCCAGCATTAAAG/GGGGGACCTGTGATCTTATC	1
GJB4	GCATTAAGGGTGCCCATCTC/TTTTCTGGGTGGCCTCAT	1
GJC3	GCTCCCTCTGAAGGACAGTG /GGGAGGAGATCATCAGGACA	1
GJC3	TGGGTACGCACTGTGAAAAA/AGCTCCTCCTGGACAGGAT	1



**Table S2.** *GJB2* variants with no clinical significance data from the three databases used.

<b>Variant</b>	<b>Allele frequency [Reference]</b>
IVS1+12G>A (NM_004004.6:c.+12G>A)	4/126 [1]; 1/1220 [2]
c.IVS1+27G>C	1/152 [3]
c.-3179G>A	1/218 [2]
c.23+G>A	8/90 [4]
51del12insA	2/72 [5]; 2/150 [6]
c.684C>A	1/300 [7]; 1/546 [8]; 1/209 [9]; 1/1220 [2]
p.His100ArgfsTer14/+ (c.299-300delAT)	1/200 [10]
delE119 (c.355-357delGAG)	2/67 [11]; 1/40 [12]; 3/150 [13]; 2/32 [14]
682C>T	1/114 [11]; 1/1220 [2]
c.964C>T	5/36 [15]
c.-31T>C	1/418 [9]
c.765C>T	3/113 [16]
p.P87del	1/112 [17]

**Table S3.** Eight commonly reported connexin 26 gene (*GJB2*) variants.

Country	p.Gly12 ValfsTer2 (c.35delG) rs80338939	p.M34T (c.101T> C) rs3588762 2	p.L79Cfs (c.235delC) rs80338943	p.V37I (c.109G>A) rs72474224	p.H100RfsTer1 4 (c.299_300delA T) rs111033204	p.W24* (c.71G>A) rs10489439 6	p.L56Rfs (c.167del T) rs8033894 2	p.R143W (c.167delT) rs80338948	Referenc e
Japan								2/2	[18]
Japan			10/70	1/70				4/70	[19]
Morocco	61/162			3/162					[20]
Ghana								64/442	[21]
Israel	2/8								[22]
Malaysia				2/66		4/66			[23]
Syria	9/70								[24]
Syria	36/100	2/100					5/100		[25]
Saudi Arabia	14/118			1/118		2/118			[26]
Kuwait	21/200								[27]
Spain	4/4								[28]
Spain	5/29					23/29			[29]
Algeria	55/62								[30]
Algeria	36/50			3/50			2/50		[31]
Algeria	27/232								[32]
Venezuela	5/84								[33]
Greece	7/58								[34]
Turkey	11/58				2/58				[35]
Iran	12/90				2/90				[4]
Bangladesh						19/134			[36]
Morocco	107/304			10/304					[37]
Turkey	25/62								[38]
India						2/116			[5]
Yakutia	51/786			14/786			1/786		[39]
Turkey	96/470								[40]
USA	6/6								[41]
Brazil	74/600	8/600	1/600	1/600		1/600	2/600		[7]
Brazil	3/4								[42]
Slovenia	15/138								[43]
Turkey	2/40				2/40				[44]
Iran	547/4644		1/4644	2/4644	8/4644	31/4644	12/4644	7/4644	[45]
Germany	110/138	1/138		2/138					[46]



Tunisia	44/204								[47]
Brazil	14/66			1/66					[48]
Brazil	14/66			1/66					[49]
Tunisia	6/6								[50]
India	27/116					10/116			[51]
Russia	527/1410	12/1410	14/1410	4/1410		3/1410	12/1410		[52]
Russia	86/222	2/222	1/111						[53]
Germany	8/50								[54]
Iran	7/348		1/348						[55]
Iran	10/1016		2/1016	2/1016	1/1016	4/1016	2/1016	1/1016	[56]
Hungary	2/326								[57]
Romania	3/692								[57]
Ashkenazi							9/372		[57]
Turkey	8/154								[58]
Ghana								44/58	[59]
Pakistan	2/140					2/140			[60]
Germany	13/1012	1/1012					1/1012		[61]
Italy	164/752	5/752					2/752		[62]
Sweden	44/158	1/158						2/158	[63]
São Tomé and Príncipe		2/272							[64]
Guatemala		1/266							[65]
Brazil	8/154	1/154							[66]
China			27/228	84/228	3/228				[67]
China			7/1238						[68]
Iran	10/68		2/68			1/68			[69]
USA	28/146		3/146	47/146	2/146	2/146	1/146	2/146	[70]
USA	20/542	7/542	2/542	40/542	2/542	3/542	2/542	2/542	[71]
China			42/230	6/230	3/230				[72]
China			41/230						[73]
China						1/70			[74]
China			1/58						[75]
China				2/200					[10]
China			118/1402	101/1402					[76]
China	2/11600		356/11600	597/11600	45/11600				[77]
China			6/50	11/50	4/50				[78]
China	9/160		47/160		7/160				[79]
Italy	9/258	1/258				1/258	1/258		[80]
Korea			10/102	1/102					[81]



Brazil	24/98			2/98					[82]
Chile	28/226								[83]
Italy	310/12395	54/12395							[84]
USA	76/98						11		[85]
Brazil	12/154								[86]
Belgium	67/554			5/554			11/554	3/554	[87]
Australia	50/104	5/104		8/104		3/104	1/104	1/104	[88]
China	12/4126		510/4126	200/4126	97/4126			4/4126	[89]
China			721/6008						[90]
China			278/2348	88/2348	19/2348				[91]
Argentina	16/92	1/92							[92]
Argentina	85/952	13/952		7/952			11/952	6/952	[93]
Argentina	46/504	7/504		2/504			2/504	1/504	[94]
Belarus	407/782		5/782				6/782		[95]
Iran	1/2								[96]
Portugal	75/234			1/234		2/234			[97]
Netherlands	8/18								[98]
Brazil	12/154								[99]
Mexico	12/152						2/152		[100]
Brazil	84/414	6/414		4/414					2006
Romania	11/250					8/250			[101]
Romania	11/700								[102]
Turkey	2/2								[103]
Egypt	11/102								[104]
Egypt	13/72								[105]
USA	27/136	1/136	2/136	2/136			3/136		[106]
Iran	49/266								[107]
Brazil	8/202								[108]
Italy	83/120								[109]
Spain	18/120								[109]
Kuwait	18/200								[110]
France	10/376	12/376							[111]
Peru	16/266		1/266	2/266			3/266	21/266	[112]
USA	29								2004
Austria	2/90								[113]
Austria	50/204					3/204			[11]
Austria	20/86					2/86			[12]
Germany	38/268				2/268				[114]
China				15/1300					[115]









Iran	15/228		1/228		1/228			[191]
Iran	17/154							[192]
India	1/90				1/90		1/90	[193]
China		23/270						[194]
India			2/1060		174/1060		1/1060	[195]
Brazil	41/162							[196]
France	287/414	1/414			4/414	6/414		[197]
France	62/192					2/192		[198]
Mexico	4/4							[199]
Tunisia	10/70							[200]
Portugal					2/12			[201]
Croatia	37/126	1/126					1/126	[202]
Croatia	3/18							[203]
Jordan	22/136							[204]
Gado Bravo	5/88				3/88			[205]
Queimadas	21/152							[205]
USA			2/16					[206]
Rhode Island New	9/84					2/84		[207]
Slovakia	9/108		1/108		25/108			[208]
Slovakia	122/546	6/546	6/546		15/546	3/546		[8]
India	1/30				6/30			[209]
Mauritania	5/278							[210]
Egypt	27/194							[211]
Brazil	6/142							[212]
Italy	40/106							[213]
Iran	25/262	4/262						[214]
Iran	11/336				1/336			[215]
India					7/54			[216]
Cyprus	49/60					1/60		[217]
Japan		96/2454	61/2454	8/2454			8/2454	[218]
Brazil	43/72		3/72					[219]
Iran	1/2							[220]
Iran	1/2							[221]
Italy	136/200	2/200				3/200	1/200	[222]
UK	4/4							[223]
France	3/10							[224]
USA		11/1042	1/1042	5/1042	1/102	24/1042	4/1042	[225]
Korea	1/294		15/294		1/294		1/294	[226]
Ecuador	2/222						3/222	[227]



China			52/180		6/180			[228]
Athens	1/2							[229]
Athens	4/6							[230]
Iran	8/84							[231]
Brazil	14/66			1/66				[48]
Italy	32/308	2/308		1/308		1/308		[232]
Poland	0	17/466	3/466	17/466		7/466		[233]
Bulgaria	45/102					1/102		[234]
Russia	18/152		6/152					[3]
USA	86/418	5/418		2/418				[235]
Italy				1/8				[236]
China	2/2328		290/2328	210/2328	55/2328			[237]
Romania	32/90				1/90	2/90		[238]
Austria	26/42							[239]
Italy	3/78	1/78		1/78				[240]
Iran	39/126							[241]
Tunisia	2/4							[242]
Tunisia	82/262		2/262	2/262				[243]
Tunisia				2/2				[244]
UK	3/102					2/102		[245]
USA	2/288	2/288		3/288				[246]
Colombia	1/10							[247]
France	31/60	5/60	1/60	3/60		1/60	1/60	[16]
Iran	10/106					1/10/6		[248]
Iran	7/80					1/80		[249]
Pakistan						43/340		[250]
Italy	147/204	3/204					12/204	[251]
Sicily	35/128							[252]
USA	1/218	2/218		12/218				[253]
Pakistan						9/392		[254]
Netherlands	24/444	1/444		1/444			1/444	[255]
USA	28/95	2/95	2/95	10/95		1/95		[256]
Czech	10/13							[257]
Czech	113/312					13/312		[258]
Pakistan	19/125					52/125		[259]
Palestin	11/96		2/96			7/96		[260]
USA	13/418	1/418		2/418		3/418	1/418	[9]
China			9/20		1/20			[261]
Korea			31/842		6/842		4/842	[262]



Norway	82/201				2/201			[263]
Austria	56/500	9/500	5/500	10/500	6/500	4/500	1/500	[264]
India	6/632			26/632	38/632			[265,266]
UK	22/48							[266]
Hungary					9/1982			[267]
Indonesia				4/240				[268]
Egypt	24/222			3/222				[269]
Israel	31/150					21/150		[6]
Brazil	47/360	3/360		2/360	2/360	1/360		[270]
Colombia	38/224	3/224				9/224		[271]
USA	120/1220	19/1220	1/1220	38/1220		6/1220	1/1220	[2]
Japan			8/1018	9/1018			/10181	[272]
Turkey	22/188					1/188		[273]
Turkey	33/42					1/42		[274]
Turkey	141/742				1/742	2/742	1/742	[275]
Mongolia			16/1068	6/1068	4/1068		1/1068	[276]
China			27/200					[277]
China	1/256		28/256		2/256			[278]
United Arab Emirates	14/100							[279]
Hungary	104/NA							2002
Japan	2/2686		142/2686	47/2686	11/2686		18/2686	[280]
Turkey	28/120					3/120		[281]
China			8/34		1/34			[282]
China			22/338		12/338			[283]
China			64/672		18/672			[284]
Thailand	0		10/332	37/332		3/332		[17]
China	2/1316		154/1316	23/1316	33/1316			[285]
USA	6/20							[286]
Austria	11/36	2/36		3/36		1/36		[287]
USA	71/648	28/648	2/648	11/648	1/648	24/648	1/648	[288]
Taiwan			38/840	129/840	8/840		1/840	[289]
Taiwan			25/2034	184/2034				[290]
China			1/4					[291]
China			47/470		6/470			[292]
Taiwan			12/506					[293]
UK						10/354		[294]
China	5/424		164/424		24/424			[295]
China			1/228	2/228	1/228			[296]



Malaysia		1/64		5/64		3/64		[297]
Iran	105/836		1/836	2/836			2/386	[298]
China	4/2134		298/2134	189/2134	27/2134		2/2134	[299]
China			188/2402	34/2402	35/2402			[300]
Azerbaijan	14/258		2/258					[301]
Ghana							37/40	[302]
India						30/430		[303]
Algeria	27/232							[304]
USA	54/238			1/238			3/238	[305]
Turkey	57/346							[306]
Malaysia				1/62		3/62		[307]
USA	301/434	31/434		18/434			22/434	[308]
Turkey	1/14	1/14	1/14				1/14	[309]
Turkey	4/24							[310]
Germany	102/670			4/670	1/670	3/670	3/670	[311]
Slovenia	115/436			5/436		4/436		[312]
Turkey	13/296							[313]
Italy	199/277	15/277		1/277		3/277	3/277	[314]
Russia	527/1410	12/1410	9/1410	4/1410		3/1410		[315]
Turkey	76/542		4/542			3/542	1/542	[13]
Balkan Gypsies						28/1206		[316]
Russia	29/54		1/54				1/54	[14]
Iran							3/76	[317]
Iran	15/200							[318]
USA	178/386	11/386	3/386	11/386		1/386	23/386	[319]
Korea			2/290		2/290			[320]
Brazil	12/77							[321]
USA	2/26	14/26						[322]
Brazil	43/370							[323]
Brazil	40/1200	2/1200		2/1200		1/1200		[324]
Iran	26/332				1/332	1/332		[325]
Iran	18/100				2/100			[326]
China			10/30					[327]
USA	7/38	2/38						[328]
Finland	14/184	3/184					2/184	[329]
Portugal	3/13	11/13						[330]
Portugal	19/178	6/178						[331]
China	2/1076		73/1076	24/1076	46/1076			[332]
China	24/4796		292/4796		57/4796		4/4796	[333]



China	1/566		29/566	10/566	6/566			[334]
China	2/968		46/968	10/968	20/968		2/968	[335]
Turkey	2/2							[336]
Egypt	24/102							[337]
Mongolia	6/378		8/378	4/378	4/378			[338]
Turkey	15/112					2/112		[339]
Iran	27/100						5	[340]
Brazil	16/100							[341]
Spain	3/76							[342]
Spain	5/8							[343]
Toronto (Canada)						4/4	8	[344]
Vietnam			2/174	54/174	1/174			[345]
China	2/142		5/142		2/142			[346]
Korea			26/4144	28/4144	3/4144			[347]
China	3/12		3/12					[348]
Iran							2/4	[349]
Iran	10/158							[350]
China	24/2114		340/2114		40/2114		3/2114	[351]
Mexico	7/156	1/156					1/156	[352]
China					3/60			[353]
Greece	111							[354]
Slovak Republic	13					1		[355]
China	2		105	29	25			[356]
Japan			1		1			[357]
USA	33	2					9	[358]
Russia	51							[359]
Korea			8	10				[360]
USA	32			1			2	[361]
USA	58						1	[362]
Japan			16/38	4/38			4/38	[363]
China			2/6		2/6			[364]
Romania	98/358					13/358		[365]
Romania	50/150					8/150		[366]
China	4/324		76/324	13/324	12/324			[367]
Slovenia	3/64							[368]
China			104/1268		10/1268			[369]
USA	47/64			2/64				[370]
China			26/171		2/171			[371]
China	1/420		67/420	1/420	1/420			[372]



China			10/86		2/86				[373]
Finland		19/30		2/30				1/30	[374]
France	14/512								[375]
China			108/1070	12/1070	27/1070			3/1070	[376]
Portugal	84/528	7/528		5/528		5/528	1/528		[377]
Mexico	3/22								[378]
Slovakia	9/108			1/108		25/108			[379]
Morocco	13/278								[380]
India						7/27			[381]
Italy	130/178								[382]
Iran						10/200	2/200		[383]
Hungary	69/1892								[384]
Iran	167/1328					11/1328	3/1328	1/1328	[385]
Cyprus	19/60								[386]
Italy	34/2080	1/2080		2/2080			8/2080		[387]
Poland	2/6								[388]
Japan	3/30	1/30							[389]
India			2/912			52/912	1/912		[390]
India						2/2			[391]
France	14/74	1/74		2/74					[392]
Greece	139/510					2/510			[393]
China	6/760		55/760	10/760	26/760				[394]
Brazil	15/82								[395]
Russia			5/440	2/440	2/440				[396]
Germany	313/NA								[397]
Italy	73/1468	11/1468		4/1468	1/1468		6/1468		[398]
USA	1001/14802	256/14802	36/14802	183/14802	3/14802	33/14802	83/14802	16/14802	[399]
China	1/358		57/358						[400]
India						14/26			[401]
Morocco	24/50								[402]
Croatia	28/54					1/54			[403]
Croatia	41/116			2/116		1/116			[404]
Iran	2/448					4/448			[405]
Czech Republic	113/312	2/312				13/312			[406]
Pakistan	4/300		2/300			9/300			[407]
Israel	2/2								[408]
Turkey	13/70								[409]
16 countries	2103/3062	70/3062	1/3062	56/3062		37/3062	79/3062	10/3062	[410]
Israel	31/150						21/150		[411]





Macedonia	15/66			2/66		4/66		[412]
Latvia	69/130	3/130	1/130					[413]
USA	48/204						9/204	[414]
Belgium	21/32							[415]
Japan			19/106				1/106	[416]
Iran	6/12							[417]
Algeria	34/160							[418]
Algeria	8/22							[419]
USA	1/20						1/20	[420]
Hungary	104/NA							[421]
Romania	17/174							[422]
Tunisia	45/190			1/190				[423]
Venezuela	13/80							[424]
Germany	2/46							[425]
China	24/194		4/194					[426]
Poland	73/204	1/204						[427]
USA			16/428	93/428				[428]
China	2/764		111/764		20/764		8/764	[429]
China			80/465	86/465	6/465			[430]
China			10/180	20/180				[431]
China			2/2					[432]
China			148/1012	33/1012	13/1012		2/1012	[433]
China	2/138		20/138					[434]
China	13/236		25/236					[435]
China			3/42					[436]
China	35/202						4/202	[437]
Australia			31/520		7/520			[438]
Turkey	51/302							[439]
China			222/2380		62/2380			[440]
China			140/678	17/678	38/678		2/678	[441]
China			6/472	12/472	1/472		1/472	[442]
China	1/636		50/636		13/636			[443]
Germany	54/456	9/456						[444]
France	69/280							[445]
USA	304/1474	11/1474		5/1474	4/1474			[225]
USA	33/768			2/768			1/768	[361]

The numerators in this column represent the number of mutated alleles, and the denominators the total number of screened alleles.)

NA, not applicable (the authors were not clear on the total number of alleles they have screened)



**Table S4.** Known but rare PLP *GJB2* variants common in isolated populations.

Variant		(c.IVS1+1G>A)		p.W172* (c.516G>A)		p.W172C (c.516G>C)		p.W172R (c.514T>A)		p.W44* (c.131G>A)	
Variant reference number		rs1302739538		rs1302739538		rs1302739538		rs770330002		rs104894407	
Country	Continent	n/N	%	n/N	%	n/N	%	n/N	%	n/N	%
Algeria	Africa	2/118	1.7%								
Ghana	Africa									2/162	1.2%
Egypt	Africa	1/222	0.5%								
Bangladesh	Asia	10/106	9.4%								
China	Asia	12/2502	0.5%								
India	Asia	8/1062	0.8%					4/1060	0.4%		
Iran	Asia	134/7842	1.7%								
Mongolia	Asia	42/1068	3.9%	1/1068	0.1%						
Palestin	Asia	1/96	1.0%								
Qatar	Asia	10/252	4.0%								
Russia	Asia	13/140	9.3%			52/592	8.8%				
Syria	Asia	14/140	10.0%								
Turkey	Asia	55/4814	1.1%	1/3062	0.03%						
Yakutia	Asia	382/702	54.4%								
Austria	Europe	3/398	0.8%								
Belgium	Europe	2/116	1.7%								
Czech	Europe	9/312	2.9%								
France	Europe	7/830	0.8%							1/318	0.3%
Italy	Europe	18/1338	1.3%								
Netherlands	Europe	3/488	0.6%								
Poland	Europe	24/466	5.2%								
Portugal	Europe	2/529	0.4%	3/528	0.6%						
Slovakia	Europe	3/546	0.5%								
USA	North America	14/4736	0.3%	2/15090	0.0%					6/2918	0.2%
Argentina	South America	1/188	0.5%								
Brazil	South America	5/1570	0.3%	3/632	0.5%						
Guatemala	North America									21/266	7.9%

n/N = number of affected alleles out of total alleles tested, % = allele frequency.

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