

Variants that were used to calculate carrier frequencies

AC	Chr	Position	Gene	Transcript	cDNA alteration	AA alteration
2	chr1	45966084	<i>MMACHC</i>	NM_015506.2	c.80A>G	p.Gln27Arg
1	chr1	45973932	<i>MMACHC</i>	NM_015506.2	c.328_331delAACC	p.Asn110fs
1	chr1	45974001	<i>MMACHC</i>	NM_015506.2	c.394C>T	p.Arg132X
1	chr1	45974004	<i>MMACHC</i>	NM_015506.2	c.398_399delAA	p.Gln133fs
2	chr1	45974034	<i>MMACHC</i>	NM_015506.2	c.427C>T	p.Gln143X
1	chr1	45974520	<i>MMACHC</i>	NM_015506.2	c.482G>A	p.Arg161Gln
2	chr1	45974604	<i>MMACHC</i>	NM_015506.2	c.567dupT	p.Ile190fs
1	chr1	45974647	<i>MMACHC</i>	NM_015506.2	c.609G>A	p.Trp203X
1	chr1	45974693	<i>MMACHC</i>	NM_015506.2	c.658_660delAAG	p.Lys220del
1	chr1	45974800	<i>MMACHC</i>	NM_015506.2	c.766_771delGCCCCC	p.Ala256_Pro257del
1	chr6	49399452	<i>MUT</i>	NM_000255.3	c.2242C>T	p.Gln748X
1	chr6	49409685	<i>MUT</i>	NM_000255.3	c.1677-1G>A	
2	chr6	49412365	<i>MUT</i>	NM_000255.3	c.1663G>A	p.Ala555Thr
1	chr6	49425530	<i>MUT</i>	NM_000255.3	c.626dupC	p.Lys210fs
1	chr6	49425733	<i>MUT</i>	NM_000255.3	c.424A>G	p.Thr142Ala
1	chr6	49426857	<i>MUT</i>	NM_000255.3	c.323G>A	p.Arg108His
1	chr6	49427179	<i>MUT</i>	NM_000255.3	c.1A>G	p.Met1?
1	chr11	112099388	<i>PTS</i>	NM_000317.2	c.155A>G	p.Asn52Ser
1	chr11	112100933	<i>PTS</i>	NM_000317.2	c.166G>A	p.Val56Met
1	chr11	112103901	<i>PTS</i>	NM_000317.2	c.259C>T	p.Pro87Ser
3	chr11	112103928	<i>PTS</i>	NM_000317.2	c.286G>A	p.Asp96Asn
1	chr12	103234172	<i>PAH</i>	NM_000277.1	c.1315+6T>A	
1	chr12	103234174	<i>PAH</i>	NM_000277.1	c.1315+4A>G	
1	chr12	103234237	<i>PAH</i>	NM_000277.1	c.1256A>G	p.Gln419Arg
1	chr12	103234270	<i>PAH</i>	NM_000277.1	c.1223G>A	p.Arg408Gln
1	chr12	103237451	<i>PAH</i>	NM_000277.1	c.1172G>C	p.Ser391Thr
1	chr12	103237461	<i>PAH</i>	NM_000277.1	c.1162G>A	p.Val388Met
1	chr12	103237555	<i>PAH</i>	NM_000277.1	c.1068C>A	p.Tyr356X
1	chr12	103240702	<i>PAH</i>	NM_000277.1	c.940C>A	p.Pro314Thr
2	chr12	103246707	<i>PAH</i>	NM_000277.1	c.728G>A	p.Arg243Gln
1	chr12	103246714	<i>PAH</i>	NM_000277.1	c.721C>T	p.Arg241Cys
2	chr12	103249009	<i>PAH</i>	NM_000277.1	c.611A>G	p.Tyr204Cys
1	chr12	103249088	<i>PAH</i>	NM_000277.1	c.532G>A	p.Glu178Lys
2	chr12	103249104	<i>PAH</i>	NM_000277.1	c.516G>T	p.Gln172His
1	chr12	103260378	<i>PAH</i>	NM_000277.1	c.505C>T	p.Arg169Cys
1	chr12	103260419	<i>PAH</i>	NM_000277.1	c.464G>A	p.Arg155His
1	chr12	103260442	<i>PAH</i>	NM_000277.1	c.442-1G>A	
1	chr12	103288534	<i>PAH</i>	NM_000277.1	c.331C>T	p.Arg111X
1	chr14	55310817	<i>GCH1</i>	NM_000161.2	c.671A>G	p.Lys224Arg
1	chr14	55369143	<i>GCH1</i>	NM_000161.2	c.239G>A	p.Ser80Asn
1	chr1	11854500	<i>MTHFR</i>	NM_005957.4	c.1262G>C	p.Trp421Ser
1	chr1	11856317	<i>MTHFR</i>	NM_005957.4	c.725delC	p.Ala242fs
1	chr1	11861356	<i>MTHFR</i>	NM_005957.4	c.337G>A	p.Ala113Thr
1	chr1	236979791	<i>MTR</i>	NM_000254.2	c.712G>T	p.Gly238X
1	chr2	73114672	<i>SPR</i>	NM_003124.4	c.112delG	p.Val38fs

1	chr2	211457661	<i>CPS1</i>	NM_001875.4	c.1145C>T	p.Pro382Leu
1	chr2	211476856	<i>CPS1</i>	NM_001875.4	c.2407C>G	p.Arg803Gly
1	chr2	211523339	<i>CPS1</i>	NM_001875.4	c.3683G>A	p.Arg1228Gln
1	chr2	211542707	<i>CPS1</i>	NM_001875.4	c.4501T>G	p.Ter1501GlueX?
1	chr3	49455047	<i>AMT</i>	NM_000481.3	c.1138G>A	p.Val380Met
1	chr3	49458998	<i>AMT</i>	NM_000481.3	c.264_265dupGA	p.Ile89fs
1	chr3	49459862	<i>AMT</i>	NM_000481.3	c.20_21delTG	p.Val7fs
1	chr5	7883293	<i>MTRR</i>	NM_002454.2	c.806C>A	p.Ser269X
1	chr6	80838876	<i>BCKDHB</i>	NM_183050.3	c.275-2A>G	
1	chr6	80878663	<i>BCKDHB</i>	NM_183050.3	c.550delT	p.Ser184fs
2	chr6	131900332	<i>ARG1</i>	NM_000045.3	c.212G>C	p.Arg71Thr
1	chr6	131900351	<i>ARG1</i>	NM_000045.3	c.232dupG	p.Glu78fs
2	chr6	131903857	<i>ARG1</i>	NM_000045.3	c.560+2T>C	
1	chr7	65546812	<i>ASL</i>	NM_000048.3	c.35G>A	p.Arg12Gln
1	chr7	65548162	<i>ASL</i>	NM_000048.3	c.446+1G>C	
2	chr7	95751240	<i>SLC25A13</i>	NM_014251.2	c.1638_1660dupGAGATTACA GGTGGCTGCCCGGG	p.Ala554fs
2	chr7	95751243	<i>SLC25A13</i>	NM_014251.2	c.1658G>A	p.Arg553Gln
1	chr7	95799356	<i>SLC25A13</i>	NM_014251.2	c.1311+1G>A	
1	chr7	95813609	<i>SLC25A13</i>	NM_014251.2	c.1157G>T	p.Gly386Val
1	chr7	95813718	<i>SLC25A13</i>	NM_014251.2	c.1048G>A	p.Asp350Asn
2	chr7	95822344	<i>SLC25A13</i>	NM_014251.2	c.615+5G>A	
1	chr7	95822452	<i>SLC25A13</i>	NM_014251.2	c.511dupG	p.Asp171fs
1	chr7	95822489	<i>SLC25A13</i>	NM_014251.2	c.475C>T	p.Gln159X
1	chr9	6533142	<i>GLDC</i>	NM_000170.2	c.2938A>G	p.Asn980Asp
1	chr9	6554768	<i>GLDC</i>	NM_000170.2	c.2216G>A	p.Arg739His
1	chr9	133333912	<i>ASS1</i>	NM_000050.4	c.299G>A	p.Arg100His
1	chr9	133342110	<i>ASS1</i>	NM_000050.4	c.421-2A>G	
1	chr9	133355188	<i>ASS1</i>	NM_000050.4	c.773+1G>A	
1	chr9	133355828	<i>ASS1</i>	NM_000050.4	c.830A>G	p.Lys277Arg
1	chr10	82034955	<i>MAT1A</i>	NM_000429.2	c.769G>A	p.Gly257Arg
1	chr10	82036212	<i>MAT1A</i>	NM_000429.2	c.688G>A	p.Val230Met
1	chr10	126086645	<i>OAT</i>	NM_000274.3	c.1186C>T	p.Arg396X
1	chr13	41383712	<i>SLC25A15</i>	NM_014252.3	c.815C>T	p.Thr272Ile
1	chr21	44478986	<i>CBS</i>	NM_000071.2	c.1316G>A	p.Arg439Gln
1	chr21	44483064	<i>CBS</i>	NM_000071.2	c.953C>T	p.Thr318Met
1	chr21	44486431	<i>CBS</i>	NM_000071.2	c.373C>T	p.Arg125Trp
1	chrX	38226614	<i>OTC</i>	NM_000531.5	c.148G>A	p.Gly50Arg
1	chr1	24140683	<i>HMGCL</i>	NM_000191.2	c.494G>A	p.Arg165Gln
1	chr1	24140684	<i>HMGCL</i>	NM_000191.2	c.493C>T	p.Arg165Trp
1	chr3	15643403	<i>BTD</i>	NM_000060.4	c.44+2T>C	
1	chr3	15676985	<i>BTD</i>	NM_000060.4	c.100_101delGG	p.Gly34fs
1	chr3	15676988	<i>BTD</i>	NM_000060.4	c.104_105delGT	p.Cys35fs
1	chr3	15677134	<i>BTD</i>	NM_000060.4	c.248T>C	p.Leu83Ser
1	chr3	15683511	<i>BTD</i>	NM_000060.4	c.406C>T	p.Gln136X
1	chr3	15683533	<i>BTD</i>	NM_000060.4	c.428G>T	p.Cys143Phe
1	chr3	15685994	<i>BTD</i>	NM_000060.4	c.631C>T	p.Arg211Cys

1	chr3	15686802	<i>BTD</i>	NM_000060.4	c.1439G>A	p.Gly480Glu
1	chr3	136047603	<i>PCCB</i>	NM_000532.4	c.1402G>A	p.Ala468Thr
1	chr3	182788996	<i>MCCC1</i>	NM_020166.4	c.639+2T>A	
1	chr3	182804555	<i>MCCC1</i>	NM_020166.4	c.295G>A	p.Gly99Ser
1	chr4	146563583	<i>MMAA</i>	NM_172250.2	c.508A>T	p.Arg170X
1	chr4	146572222	<i>MMAA</i>	NM_172250.2	c.742C>T	p.Gln248X
1	chr5	70900248	<i>MCCC2</i>	NM_022132.4	c.577C>T	p.Arg193Cys
1	chr5	70922494	<i>MCCC2</i>	NM_022132.4	c.652G>A	p.Ala218Thr
1	chr5	70948582	<i>MCCC2</i>	NM_022132.4	c.1574+1G>A	
2	chr7	40228131	<i>SUGCT</i>	NM_024728.2	c.286delG	p.Val96fs
1	chr10	124797335	<i>ACADSB</i>	NM_001609.3	c.275C>G	p.Ser92X
1	chr10	124803949	<i>ACADSB</i>	NM_001609.3	c.848A>G	p.Tyr283Cys
1	chr10	124803977	<i>ACADSB</i>	NM_001609.3	c.878dupG	p.Arg294fs
1	chr10	124812613	<i>ACADSB</i>	NM_001609.3	c.1165A>G	p.Met389Val
1	chr11	108016928	<i>ACAT1</i>	NM_000019.3	c.1006-1G>C	
1	chr11	134128412	<i>ACAD8</i>	NM_014384.2	c.384G>A	p.Met128Ile
1	chr11	134130962	<i>ACAD8</i>	NM_014384.2	c.730C>T	p.Arg244X
3	chr11	134131692	<i>ACAD8</i>	NM_014384.2	c.1000C>T	p.Arg334Cys
1	chr11	134131785	<i>ACAD8</i>	NM_014384.2	c.1092+1G>A	
1	chr12	109999224	<i>MMAB</i>	NM_052845.3	c.519+1G>A	
1	chr13	100962159	<i>PCCA</i>	NM_000282.3	c.1426C>T	p.Arg476X
1	chr13	101167783	<i>PCCA</i>	NM_000282.3	c.2002G>A	p.Gly668Arg
1	chr15	40710389	<i>IVD</i>	NM_002225.3	c.1208A>G	p.Tyr403Cys
1	chr16	83932808	<i>MLYCD</i>	NM_012213.2	c.60delG	p.Arg21fs
1	chr20	32868897	<i>AHCY</i>	NM_000687.3	c.1238_1241delAAGC	p.Gln413fs
1	chr20	32883275	<i>AHCY</i>	NM_000687.3	c.145C>T	p.Arg49Cys
1	chr21	38137471	<i>HLCS</i>	NM_000411.6	c.1522C>T	p.Arg508Trp
1	chr21	38309080	<i>HLCS</i>	NM_000411.6	c.663_664delCA	p.Arg222fs
1	chr21	38309463	<i>HLCS</i>	NM_000411.6	c.281dupG	p.Val96fs
1	chr1	53676781	<i>CPT2</i>	NM_000098.2	c.1438_1445delGGGCAGAC	p.Gly480fs
1	chr1	53676980	<i>CPT2</i>	NM_000098.2	c.1634A>C	p.Glu545Ala
1	chr1	76200511	<i>ACADM</i>	NM_000016.5	c.430_432delAAG	p.Lys144del
2	chr1	76200533	<i>ACADM</i>	NM_000016.5	c.449_452delCTGA	p.Thr150fs
1	chr1	76228411	<i>ACADM</i>	NM_000016.5	c.1229T>C	p.Ile410Thr
1	chr1	76228429	<i>ACADM</i>	NM_000016.5	c.1247T>C	p.Ile416Thr
1	chr1	120306794	<i>HMGCS2</i>	NM_005518.3	c.559+1G>A	
1	chr1	120306834	<i>HMGCS2</i>	NM_005518.3	c.520T>C	p.Phe174Leu
1	chr2	26501559	<i>HADHB</i>	NM_000183.2	c.520C>T	p.Arg174Cys
1	chr2	26502111	<i>HADHB</i>	NM_000183.2	c.739C>T	p.Arg247Cys
1	chr4	108940769	<i>HADH</i>	NM_005327.4	c.493C>T	p.Arg165X
1	chr5	131714104	<i>SLC22A5</i>	NM_003060.3	c.428C>T	p.Pro143Leu
1	chr5	131721047	<i>SLC22A5</i>	NM_003060.3	c.680G>A	p.Arg227His
3	chr5	131721127	<i>SLC22A5</i>	NM_003060.3	c.760C>T	p.Arg254X
2	chr5	131726524	<i>SLC22A5</i>	NM_003060.3	c.1195C>T	p.Arg399Trp
1	chr5	131728290	<i>SLC22A5</i>	NM_003060.3	c.1433C>T	p.Pro478Leu
1	chr6	24495387	<i>ALDH5A1</i>	NM_001080.3	c.164delC	p.Ser55fs
1	chr6	24505151	<i>ALDH5A1</i>	NM_001080.3	c.664G>A	p.Gly222Ser

2	chr6	24515432	<i>ALDH5A1</i>	NM_001080.3	c.764A>G	p.Asn255Ser
1	chr11	68530187	<i>CPT1A</i>	NM_001876.3	c.1783C>T	p.Arg595Trp
1	chr12	110017615	<i>MVK</i>	NM_000431.3	c.235G>T	p.Asp79Tyr
1	chr12	110034317	<i>MVK</i>	NM_000431.3	c.1126G>A	p.Gly376Ser
1	chr12	121164946	<i>ACADS</i>	NM_000017.3	c.164C>T	p.Pro55Leu
1	chr12	121176669	<i>ACADS</i>	NM_000017.3	c.981_983delGAC	p.Thr328del
1	chr12	121176678	<i>ACADS</i>	NM_000017.3	c.989G>A	p.Arg330His
2	chr12	121176944	<i>ACADS</i>	NM_000017.3	c.1031A>G	p.Glu344Gly
2	chr12	121177142	<i>ACADS</i>	NM_000017.3	c.1130C>T	p.Pro377Leu
1	chr17	7125268	<i>ACADVL</i>	NM_000018.3	c.623-2_623-1delAG	
1	chr17	7125572	<i>ACADVL</i>	NM_000018.3	c.833_835delAGA	p.Lys278del
1	chr17	7126184	<i>ACADVL</i>	NM_000018.3	c.1077G>A	p.Ala359Ala
1	chr17	7127154	<i>ACADVL</i>	NM_000018.3	c.1292A>G	p.Asp431Gly
1	chr17	7127303	<i>ACADVL</i>	NM_000018.3	c.1349G>A	p.Arg450His
1	chr17	7127311	<i>ACADVL</i>	NM_000018.3	c.1357C>T	p.Arg453X
1	chr17	7127674	<i>ACADVL</i>	NM_000018.3	c.1567G>A	p.Gly523Arg
1	chr4	159603400	<i>ETFDH</i>	NM_004453.3	c.229G>A	p.Gly77Ser
1	chr4	159603421	<i>ETFDH</i>	NM_004453.3	c.250G>A	p.Ala84Thr
1	chr4	159603466	<i>ETFDH</i>	NM_004453.3	c.295C>T	p.Arg99Cys
3	chr4	159603560	<i>ETFDH</i>	NM_004453.3	c.389A>T	p.Asp130Val
1	chr4	159611499	<i>ETFDH</i>	NM_004453.3	c.607-1G>T	
3	chr4	159616734	<i>ETFDH</i>	NM_004453.3	c.770A>G	p.Tyr257Cys
1	chr4	159624669	<i>ETFDH</i>	NM_004453.3	c.1211T>C	p.Met404Thr
1	chr4	159627450	<i>ETFDH</i>	NM_004453.3	c.1395T>G	p.Tyr465X
1	chr4	159627509	<i>ETFDH</i>	NM_004453.3	c.1454C>G	p.Thr485Ser
1	chr4	159629557	<i>ETFDH</i>	NM_004453.3	c.1732C>T	p.Arg578Trp
1	chr19	13004378	<i>GCDH</i>	NM_000159.3	c.416C>G	p.Ser139Trp
1	chr19	13004454	<i>GCDH</i>	NM_000159.3	c.492C>G	p.Tyr164X
1	chr19	13007743	<i>GCDH</i>	NM_000159.3	c.873delC	p.Asn291fs
3	chr19	13008224	<i>GCDH</i>	NM_000159.3	c.1064G>A	p.Arg355His
1	chr19	13008590	<i>GCDH</i>	NM_000159.3	c.1156C>T	p.Arg386X
1	chr19	13008632	<i>GCDH</i>	NM_000159.3	c.1198G>A	p.Val400Met
1	chr19	13010280	<i>GCDH</i>	NM_000159.3	c.1244-2A>C	