

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
ABCA4	c.52C>T (p.Arg18Trp)	rs121909205
ABCA4	c.5338C>G (p.Pro1780Ala)	rs121909207
ABCA4	c.5882G>A (p.Gly1961Glu)	rs1800553
ABCA4	c.5908C>T (p.Leu1970Phe)	rs28938473
ABCA4	c.1848delA (p.Glu616AspfsTer33)	rs387906386
ABCA4	c.6148G>C (p.Val2050Leu)	rs41292677
ABCA4	c.2791G>A (p.Val931Met)	rs58331765
ABCA4	c.3602T>G (p.Leu1201Arg)	rs61750126
ABCA4	c.4139C>T (p.Pro1380Leu)	rs61750130
ABCA4	c.634C>T (p.Arg212Cys)	rs61750200
ABCA4	c.3113C>T (p.Ala1038Val)	rs61751374
ABCA4	c.1622T>C (p.Leu541Pro)	rs61751392
ABCA4	c.6079C>T (p.Leu2027Phe)	rs61751408
ABCA4	c.2588G>C (p.Gly863Ala)	rs76157638
ACADM	c.799G>A (p.Gly267Arg)	rs121434274
ACADM	c.447G>A (p.Met149Ile)	rs121434277
ACADM	c.583G>A (p.Gly195Arg)	rs121434278
ACADM	c.199T>C (p.Tyr67His)	rs121434280
ACADM	c.734C>T (p.Ser245Leu)	rs121434281
ACADM	c.362C>T (p.Thr121Ile)	rs121434283
ACADM	c.127G>A (p.Glu43Lys)	rs147559466
ACADM	c.617G>A (p.Arg206His)	rs200724875
ACADM	c.616C>T (p.Arg206Cys)	rs373715782
ACADM	c.1102_1105delTTAG (p.Ala369LeufsTer18)	rs387906297
ACADM	c.233T>C (p.Ile78Thr)	rs398123074
ACADM	c.455G>A (p.Arg152Lys) [c.443G>A (p.Arg148Lys)]	rs778906552
ACADM	c.985A>G (p.Lys329Glu)	rs77931234
ACADS	c.136C>T (p.Arg46Trp)	rs121908003
ACADS	c.973C>T (p.Arg325Trp)	rs121908006
ACADS	c.164C>T (p.Pro55Leu)	rs147442301
ACADS	c.1147C>T (p.Arg383Cys)	rs28940872
ACADS	c.1138C>T (p.Arg380Trp)	rs28940875
ACADS	c.1058C>T (p.Ser353Leu)	rs28941773
ACADS	c.310_312delGAG (p.Glu104del)	rs387906308
ACADS	c.529T>C (p.Trp177Arg)	rs57443665
ACADS	c.319C>T (p.Arg107Cys)	rs61732144
ACADS	c.682_683delGA (p.Glu228ArgfsTer16)	rs786204691
ACADVL	c.1182+1G>A	rs113690956
ACADVL	c.848T>C (p.Val283Ala)	rs113994167
ACADVL	c.779C>T (p.Thr260Met)	rs113994168
ACADVL	c.1226C>T (p.Thr409Met)	rs113994169
ACADVL	c.1405C>T (p.Arg469Trp)	rs113994170
ACADVL	c.1679-6G>A	rs113994171
ACADVL	c.1837C>T (p.Arg613Trp)	rs118204014
ACADVL	c.1349G>A (p.Arg450His)	rs118204016

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
ACADVL	c.637G>A (p.Ala213Thr)	rs140629318
ACADVL	c.1844G>A (p.Arg615Gln)	rs148584617
ACADVL	c.1322G>A (p.Gly441Asp)	rs2309689
ACADVL	c.194C>T (p.Pro65Leu)	rs28934585
ACADVL	c.520G>A (p.Val174Met)	rs369560930
ACADVL	c.343delG (p.Glu115LysfsTer2)	rs387906249
ACADVL	c.388_390delGAG (p.Glu130del)	rs387906251
ACADVL	c.896_898delAGA (p.Lys299del)	rs387906252
ACADVL	c.1406G>A (p.Arg469Gln)	rs398123083
ACADVL	c.1700G>A (p.Arg567Gln)	rs398123084
ACADVL	c.538G>A (p.Ala180Thr)	rs727503791
ACADVL	c.298_299delCA (p.Gln100ValfsTer3)	rs786204713
ADGRV1	c.8790delC (p.Met2931TrpfsTer11)	rs796051864
ADGRV1	c.8495C>A (p.Ser2832Ter)	rs121909761
ADGRV1	c.6901C>T (p.Gln2301Ter)	rs121909762
ADGRV1	c.18131A>G (p.Tyr6044Cys)	rs121909763
ADGRV1	c.14973-2A>G	rs371981035
ADGRV1	c.2398C>T (p.Arg800Ter)	rs373780305
ADGRV1	c.11253C>G (p.Tyr3751Ter)	rs376689763
ADGRV1	c.17662delT (p.Ser5888HisfsTer54)	rs397517426
ADGRV1	c.2870dupA (p.Asn957Lysfs)	rs397517429
ADGRV1	c.7374_7375delTG (p.Glu2459GlyfsTer35)	rs397517435
ADGRV1	c.7406G>A (p.Trp2469Ter)	rs397517436
ADGRV1	c.8737delG (p.Val2913TyrfsTer4)	rs397517441
ADGRV1	c.5643delG (p.Tyr1882IlefsTer6)	rs727503076
ADGRV1	c.12631C>T (p.Arg4211Ter)	rs727504777
ADGRV1	c.4702delA (p.Ser1568ValfsTer78)	rs794727347
ADGRV1	c.8204delA (p.Asn2735MetfsTer13)	rs794727584
AGL	c.16C>T (p.Gln6Ter)	rs113994126
AGL	c.18_19delGA (p.Gln6HisfsTer20)	rs113994127
AGL	c.1222C>T (p.Arg408Ter)	rs113994128
AGL	c.2039G>A (p.Trp680Ter)	rs113994129
AGL	c.2590C>T (p.Arg864Ter)	rs113994130
AGL	c.3682C>T (p.Arg1228Ter)	rs113994131
AGL	c.3965delT (p.Val1322AlafsTer27)	rs113994132
AGL	c.1735+1G>T	rs199922945
AGL	c.4260-12A>G	rs369973784
AGL	c.4529dupA (p.Tyr1510Ter)	rs387906244
AGL	c.4456delT (p.Ser1486ProfsTer18)	rs387906245
ALPL	c.535G>A (p.Ala179Thr)	rs121918000
ALPL	c.881A>C (p.Asp294Ala)	rs121918002
ALPL	c.571G>A (p.Glu191Lys)	rs121918007
ALPL	c.1133A>T (p.Asp378Val)	rs121918008
ALPL	c.979T>C (p.Phe327Leu)	rs121918010
ALPL	c.407G>A (p.Arg136His)	rs121918011

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
ALPL	c.346G>A (p.Ala116Thr)	rs121918013
ALPL	c.1250A>G (p.Asn417Ser)	rs121918014
ALPL	c.526G>A (p.Ala176Thr)	rs121918019
ALPL	c.1559delT (p.Leu520ArgfsTer86)	rs387906525
ANXA5	c.-229G>A	rs112782763
ANXA5	c.-135G>A	rs113588187
ANXA5	c.-184T>C	rs28651243
ANXA5	c.-210A>C	rs28717001
AR	c.4G>A (p.Glu2Lys)	rs104894742
AR	c.2323C>T (p.Arg775Cys)	rs137852562
AR	c.2157G>A (p.Trp719Ter)	rs137852563
AR	c.2391G>A (p.Trp797Ter)	rs137852565
AR	c.1771A>T (p.Lys591Ter)	rs137852566
AR	c.2291A>G (p.Tyr764Cys)	rs137852567
AR	c.2650A>T (p.Lys884Ter)	rs137852568
AR	c.2362A>G (p.Met788Val)	rs137852570
AR	c.2191G>A (p.Val731Met)	rs137852571
AR	c.2610T>G (p.Ile870Met)	rs137852574
AR	c.178C>T (p.Gln60Ter)	rs137852575
AR	c.1826G>A (p.Arg609Lys)	rs137852576
AR	c.2623C>T (p.His875Tyr)	rs137852581
AR	c.2708A>G (p.Gln903Arg)	rs137852582
AR	c.1943G>A (p.Ser648Asn)	rs137852584
AR	c.2123T>G (p.Leu708Arg)	rs137852585
AR	c.1739G>T (p.Cys580Phe)	rs137852586
AR	c.1748T>A (p.Phe583Tyr)	rs137852587
AR	c.1645C>T (p.Pro549Ser)	rs137852588
AR	c.2343G>T (p.Met781Ile)	rs137852589
AR	c.521T>G (p.Leu174Ter)	rs137852590
AR	c.2395C>G (p.Gln799Glu)	rs137852591
AR	c.2423T>C (p.Met808Thr)	rs137852592
AR	c.2137C>T (p.Leu713Phe)	rs137852595
AR	c.1732G>A (p.Gly578Arg)	rs137852596
AR	c.2596T>C (p.Ser866Pro)	rs137852597
AR	c.2069A>C (p.His690Pro)	rs137852599
AR	c.2231G>T (p.Gly744Val)	rs137852600
AR	c.2222C>G (p.Ser741Cys)	rs137852601
AR	c.1937C>A (p.Ala646Asp)	rs1800053
AR	c.2567G>A (p.Arg856His)	rs9332971
ARSA	c.256C>T (p.Arg86Trp)	rs199476352
ARSA	c.1283C>T (p.Pro428Leu)	rs28940893
ARSA	c.862A>C (p.Thr288Pro)	rs28940894
ARSA	c.302G>A (p.Gly101Asp)	rs74315455
ARSA	c.293C>T (p.Ser98Phe)	rs74315456
ARSA	c.542T>G (p.Ile181Ser)	rs74315457

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ARSA	c.641C>T (p.Ala214Val)	rs74315467
ARSA	c.739G>A (p.Gly247Arg)	rs74315471
ARSA	c.827C>T (p.Thr276Met)	rs74315472
ARSA	c.1114C>T (p.Arg372Trp)	rs74315476
ARSA	c.1136C>T (p.Pro379Leu)	rs74315478
ARSA	c.1232C>T (p.Thr411Ile)	rs74315481
ARSA	c.465+1G>A	rs80338815
ARSA	c.769G>C (p.Asp257His)	rs80338819
ASL	c.446+1G>A	rs142637046
ASL	c.35G>A (p.Arg12Gln)	rs145138923
ASL	c.283C>T (p.Arg95Cys)	rs28940585
ASL	c.532G>A (p.Val178Met)	rs28941473
ASPA	c.454T>C (p.Cys152Arg)	rs104894548
ASPA	c.654C>A (p.Cys218Ter)	rs104894549
ASPA	c.71A>G (p.Glu24Gly)	rs104894551
ASPA	c.746A>T (p.Asp249Val)	rs104894552
ASPA	c.212G>A (p.Arg71His)	rs104894553
ASPA	c.693C>A (p.Tyr231Ter)	rs12948217
ASPA	c.854A>C (p.Glu285Ala)	rs28940279
ASPA	c.914C>A (p.Ala305Glu)	rs28940574
ASS1	c.470G>A (p.Arg157His)	rs121908637
ASS1	c.1168G>A (p.Gly390Arg)	rs121908641
ASS1	c.256C>T (p.Arg86Cys)	rs121908644
ASS1	c.835C>T (p.Arg279Ter)	rs121908645
ASS1	c.535T>C (p.Trp179Arg)	rs121908646
ASS1	c.787G>A (p.Val263Met)	rs192838388
ASS1	c.323G>T (p.Arg108Leu)	rs35269064
ATM	c.5044G>C (p.Asp1682His)	rs121434217
ATM	c.7327C>T (p.Arg2443Ter)	rs121434220
ATM	c.5309C>G (p.Ser1770Ter)	rs121434223
ATM	c.3304G>T (p.Gly1102Ter)	rs147557621
ATM	c.3993+1G>A	rs200196781
ATM	c.3931C>T (p.Gln1311Ter)	rs200976093
ATM	c.7875_7876delTGinsGC (p.AspAla2625GluPro)	rs267606668
ATM	c.7271T>G (p.Val2424Gly)	rs28904921
ATM	c.4370T>G (p.Leu1457Ter)	rs373226793
ATM	c.1110C>G (p.Tyr370Ter)	rs376170600
ATM	c.6100C>T (p.Arg2034Ter)	rs532480170
ATM	c.802C>T (p.Gln268Ter)	rs557012154
ATM	c.103C>T (p.Arg35Ter)	rs55861249
ATM	c.3576G>A (p.Lys1192=) [c.3576G>A (p.Lys1192%3D)]	rs587776551
ATM	c.1235G>A (p.Trp412Ter)	rs587779813
ATM	c.170G>A (p.Trp57Ter)	rs587779818
ATM	c.2502dupA (p.Val835SerfsTer7)	rs587779822
ATM	c.3372C>G (p.Tyr1124Ter)	rs587779833

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
ATM	c.3802delG (p.Val1268Ter)	rs587779834
ATM	c.3836G>A (p.Trp1279Ter)	rs587779836
ATM	c.5932G>T (p.Glu1978Ter)	rs587779852
ATM	c.283C>T (p.Gln95Ter)	rs587781545
ATM	c.7096G>T (p.Glu2366Ter)	rs587781672
ATM	c.5908C>T (p.Gln1970Ter)	rs587781722
ATM	c.381delA (p.Val128Terfs)	rs587781831
ATM	c.3388G>T (p.Gly1130Ter)	rs587781911
ATM	c.4198A>T (p.Lys1400Ter)	rs587781950
ATM	c.1158delG (p.Lys387ArgfsTer3)	rs587782085
ATM	c.3077G>A (p.Trp1026Ter)	rs587782103
ATM	c.6326G>A (p.Trp2109Ter)	rs587782114
ATM	c.1898+2T>G	rs587782124
ATM	c.3980T>G (p.Leu1327Ter)	rs587782192
ATM	c.3994-2A>G	rs587782276
ATM	c.2548G>T (p.Glu850Ter)	rs587782280
ATM	c.2880delC (p.Leu961CysfsTer10)	rs730881300
ATM	c.237delA (p.Lys79AsnfsTer37)	rs730881303
ATM	c.2251-10T>G	rs730881346
ATM	c.2376+1G>T	rs730881347
ATM	c.2426C>A (p.Ser809Ter)	rs730881348
ATM	c.1A>G (p.Met1Val) [c.1A>C (p.Met1?)]	rs730881359
ATM	c.154G>T (p.Gly52Ter)	rs730881362
ATM	c.3049C>T (p.Gln1017Ter)	rs730881388
ATM	c.4852C>T (p.Arg1618Ter)	rs762083530
ATM	c.5188C>T (p.Arg1730Ter)	rs764389018
ATM	g.91279A [c.5763-1050A>G]	rs774925473
ATM	c.1737G>A (p.Trp579Ter)	rs786201689
ATM	c.513C>G (p.Tyr171Ter)	rs786201693
ATM	c.3349C>T (p.Gln1117Ter)	rs786201957
ATM	c.4052delT (p.Leu1351TyrfsTer35)	rs786202350
ATM	c.1880dupT (p.Gln628ProfsTer7)	rs786202474
ATM	c.2754delT (p.Phe918LeufsTer11)	rs786202608
ATM	c.2098C>T (p.Gln700Ter)	rs786202743
ATM	c.2789T>G (p.Leu930Ter)	rs786203309
ATM	c.1424C>G (p.Ser475Ter)	rs786203550
ATM	c.2129delC (p.Thr710LysfsTer25)	rs786203807
ATM	c.151C>T (p.Gln51Ter)	rs786203888
ATM	c.5644C>T (p.Arg1882Ter)	rs786204433
ATM	c.5515C>T (p.Gln1839Ter)	rs786204751
ATP7B	c.3809A>G (p.Asn1270Ser)	rs121907990
ATP7B	c.3796G>A (p.Gly1266Arg)	rs121907992
ATP7B	c.2906G>A (p.Arg969Gln)	rs121907996
ATP7B	c.1934T>G (p.Met645Arg)	rs121907998

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ATP7B	c.2128G>A (p.Gly710Ser)	rs137853285
ATP7B	c.3400delC (p.Ala1135GlnfsTer13)	rs137853286
ATP7B	c.2305A>G (p.Met769Val)	rs193922103
ATP7B	c.3955C>T (p.Arg1319Ter)	rs193922109
ATP7B	c.1969A>C (p.Ser657Arg)	rs372436901
ATP7B	c.2930C>T (p.Thr977Met)	rs72552255
ATP7B	c.3207C>A (p.His1069Gln)	rs76151636
	c.2207_2212delATCTGAins7 [c.2207_2212delATCTGAinsTAGATTC	
BLM	(p.Tyr736LeufsTer5)]	rs113993962
BLM	c.3558+1G>A	rs148969222
BTD	c.1207T>G (p.Phe403Val)	rs104893686
BTD	c.235C>T (p.Arg79Cys)	rs104893687
BTD	c.1595C>T (p.Thr532Met)	rs104893688
BTD	c.1466A>C (p.Asn489Thr)	rs104893692
BTD	c.100G>A (p.Gly34Ser)	rs119103232
BTD	c.511G>A (p.Ala171Thr) HAPLOTYP[p.Ala171Thr;Asp444His]	rs13073139
BTD	c.1489C>T (p.Pro497Ser)	rs138818907
BTD	c.128A>G (p.His43Arg)	rs146011150
BTD	c.470G>A (p.Arg157His)	rs146015592
BTD	c.1369G>A (p.Val457Met)	rs146600671
BTD	c.262C>T (p.Gln88Ter)	rs151071780
BTD	c.1432G>C (p.Ala478Pro)	rs181396238
BTD	c.643C>T (p.Leu215Phe)	rs190386869
BTD	c.1046A>C (p.Asn349Thr)	rs200327983
BTD	c.664G>C (p.Asp222His)	rs200337373
BTD	c.1205A>G (p.Asn402Ser)	rs201023772
BTD	c.1455C>G (p.His485Gln)	rs201604102
BTD	c.755A>G (p.Asp252Gly)	rs28934601
BTD	c.133G>A (p.Gly45Arg)	rs34885143
BTD	c.1171C>T (p.Pro391Ser)	rs35034250
BTD	c.1284C>A (p.Tyr428Ter)	rs35145938
BTD	c.880A>G (p.Ile294Val)	rs35976361
BTD	c.1237G>A (p.Gly413Ser)	rs374141881
BTD	c.454A>C (p.Thr152Pro)	rs374681173
BTD	c.341G>T (p.Gly114Val)	rs375712490
BTD	c.935G>A (p.Gly312Asp)	rs377651057
BTD	c.528G>T (p.Lys176Asn)	rs397514367
BTD	c.933delT (p.Ser311ArgfsTer23)	rs397514395
BTD	c.98_104delGCGGCTGinsTCC (p.Cys33PhefsTer36)	rs80338684
BTD	c.1368A>C (p.Gln456His)	rs80338685
BTD	c.1612C>T (p.Arg538Cys)	rs80338686
CBS	c.919G>A (p.Gly307Ser)	rs121964962

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CBS	c.341C>T (p.Ala114Val)	rs121964964
CBS	c.415G>A (p.Gly139Arg)	rs121964965
CBS	c.430G>A (p.Glu144Lys)	rs121964966
CBS	c.1616T>C (p.Leu539Ser)	rs121964968
CBS	c.797G>A (p.Arg266Lys)	rs121964969
CBS	c.1058C>T (p.Thr353Met)	rs121964972
CBS	c.572C>T (p.Thr191Met)	rs121964973
CBS	c.1330G>A (p.Asp444Asn)	rs28934891
CBS	c.1006C>T (p.Arg336Cys)	rs398123151
CBS	c.833T>C (p.Ile278Thr)	rs5742905
CBS	c.374G>A (p.Arg125Gln)	rs781444670
CDH23	c.6049+1G>A	rs111033247
CDH23	c.5237G>A (p.Arg1746Gln) [c.5252G>A (p.Arg1751Gln)]	rs111033270
CDH23	c.6442G>A (p.Asp2148Asn) [c.6457G>A (p.Asp2153Asn)]	rs111033271
CDH23	c.4488G>C (p.Gln1496His) [c.4503G>C (p.Gln1501His)]	rs121908347
CDH23	c.6133G>A (p.Asp2045Asn) [c.6148G>A (p.Asp2050Asn)]	rs121908348
CDH23	c.6604G>A (p.Asp2202Asn) [c.6619G>A (p.Asp2207Asn)]	rs121908349
CDH23	c.4021G>A (p.Asp1341Asn) [c.4036G>A (p.Asp1346Asn)]	rs121908351
CDH23	c.5663T>C (p.Phe1888Ser) [c.5678T>C (p.Phe1893Ser)]	rs121908352
CDH23	c.719C>T (p.Pro240Leu)	rs121908354
CDH23	c.902G>A (p.Arg301Gln)	rs121908355
CDH23	c.6050-9G>A	rs367928692
CDH23	c.2012delT (p.Phe671SerfsTer23)	rs397517313
CDH23	c.3481C>T (p.Arg1161Ter) [c.3496C>T (p.Arg1166Ter)]	rs397517323
CDH23	c.3628C>T (p.Gln1210Ter) [c.3643C>T (p.Gln1215Ter)]	rs397517326
CDH23	c.3706C>T (p.Arg1236Ter) [c.3721C>T (p.Arg1241Ter)]	rs397517327
CDH23	c.4309C>T (p.Arg1437Ter) [c.4324C>T (p.Arg1442Ter)]	rs397517329
CDH23	c.46delG (p.Val16CysfsTer2)	rs397517331
CDH23	c.5272C>T (p.Gln1758Ter) [c.5287C>T (p.Gln1763Ter)]	rs397517337
CDH23	c.5712+1G>A [c.5727+1G>A]	rs397517341
CDH23	c.5712G>A (p.Thr1904=) [c.5727G>A (p.Thr1909%3D)]	rs397517342
CDH23	c.5923+1G>A [c.5938+1G>A]	rs397517346
CDH23	c.6968delC (p.Pro2323Leufs) [c.6983delC (p.Pro2328LeufsTer50)]	rs397517350
CDH23	c.7776G>A (p.Trp2592Ter) [c.7791G>A (p.Trp2597Ter)]	rs397517353
CDH23	c.8781C>A (p.Tyr2927Ter) [c.8796C>A (p.Tyr2932Ter)]	rs397517362
CDH23	c.9629_9632delTCAA (p.Ile3210ArgfsTer5) [c.9644_9647delTCAA (p.Ile3215ArgfsTer5)]	rs397517367
CFTR	c.4056G>C (p.Gln1352His)	rs113857788
CFTR	c.1624G>T (p.Gly542Ter)	rs113993959
CFTR	c.1521_1523delCTT (p.Phe508del)	rs113993960
CFTR	c.3808G>A (p.Asp1270Asn)	rs11971167
CFTR	c.948delT (p.Phe316LeufsTer12)	rs121908744
CFTR	c.1519_1521delATC (p.Ile507del)	rs121908745
CFTR	c.2052delA (p.Lys684AsnfsTer38)	rs121908746

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
CFTR	c.3528delC (p.Thr1176Thr=fs) [c.3528delC (p.Lys1177SerfsTer15)]	rs121908747
CFTR	c.1766+1G>A	rs121908748
CFTR	c.1646G>A (p.Ser549Asn)	rs121908755
CFTR	c.935_937delTCT (p.Phe312del)	rs121908768
CFTR	c.262_263delTT (p.Leu88IlefsTer22)	rs121908769
CFTR	c.1545_1546delTA (p.Tyr515_Arg516delins) [c.1545_1546delTA (p.Tyr515Ter)]	rs121908776
CFTR	c.3773_3774insT (p.Leu1258delinsPheGlu) [c.3773dupT (p.Leu1258PhefsTer7)]	rs121908789
CFTR	c.313delA (p.Ile105SerfsTer2) [c.444delA (p.Ile105SerfsTer2)]	rs121908801
CFTR	c.2012delT (p.Leu671Ter)	rs121908812
CFTR	c.1000C>T (p.Arg334Trp)	rs121909011
CFTR	c.3199G>A (p.Ala1067Thr)	rs121909020
CFTR	c.2735C>T (p.Ser912Leu)	rs121909034
CFTR	c.3230T>C (p.Leu1077Pro)	rs139304906
CFTR	c.1400T>C (p.Leu467Pro)	rs139573311
CFTR	c.1327G>T (p.Asp443Tyr)	rs147422190
CFTR	c.3659delC (p.Thr1220Lysfs)	rs121908811
CFTR	c.1585-8G>A	rs193922503
CFTR	c.3302T>A (p.Met1101Lys)	rs36210737
CFTR	c.200C>T (p.Pro67Leu)	rs368505753
CFTR	c.3469-20T>C	rs373002889
CFTR	c.4426C>T (p.Gln1476Ter)	rs374705585
CFTR	c.2215delG (p.Val739TyrfsTer16)	rs397508353
CFTR	c.2537G>A (p.Trp846Ter)	rs397508393
CFTR	c.3752G>A (p.Ser1251Asn)	rs74503330
CFTR	c.1364C>A (p.Ala455Glu)	rs74551128
CFTR	c.1657C>T (p.Arg553Ter)	rs74597325
CFTR	c.3484C>T (p.Arg1162Ter)	rs74767530
CFTR	c.3717+12191C>T (3849+10kbC>T) [c.3718-2477C>T]	rs75039782
CFTR	c.1079C>A (p.Thr360Lys)	rs75053309
CFTR	c.2988+1G>A	rs75096551
CFTR	c.1652G>A (p.Gly551Asp)	rs75527207
CFTR	c.3454G>C (p.Asp1152His)	rs75541969
CFTR	c.254G>A (p.Gly85Glu)	rs75961395
CFTR	c.3140-26A>G	rs76151804
CFTR	c.3764C>A (p.Ser1255Ter)	rs76649725
CFTR	c.3846G>A (p.Trp1282Ter)	rs77010898
CFTR	c.579+1G>T (711+1 G>T)	rs77188391
CFTR	c.178G>T (p.Glu60Ter)	rs77284892
CFTR	c.1558G>T (p.Val520Phe)	rs77646904
CFTR	c.349C>T (p.Arg117Cys)	rs77834169
CFTR	c.1040G>A (p.Arg347His)	rs77932196



GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
CFTR	c.3196C>T (p.Arg1066Cys)	rs78194216
CFTR	c.350G>A (p.Arg117His)	rs78655421
CFTR	c.489+1G>T	rs78756941
CFTR	c.2668C>T (p.Gln890Ter)	rs79633941
CFTR	c.366T>A (p.Tyr122Ter)	rs79660178
CFTR	c.3472C>T (p.Arg1158Ter)	rs79850223
CFTR	c.1679G>A (p.Arg560Lys)	rs80055610
CFTR	c.1519_1521del (p.I507del)	rs121908745
CFTR	c.2657+5G>A	rs80224560
CFTR	c.54-5940_273+10250del21kb [CFTRdele2,3 ; c.54-5940_273+10250del21kb (p.Ser18ArgfsX16)]	
CLRN1	c.567T>G (p.Tyr189Ter) [c.528T>G (p.Tyr176Ter)]	rs121908140
CLRN1	c.118T>G (p.Cys40Gly)	rs121908143
CLRN1	c.92C>T (p.Pro31Leu)	rs374390376
CLRN1	c.301_305delGTCAT (p.Val101_Ile102) [c.301_305delGTCAT (p.Val101SerfsTer27)]	rs397517932
COL4A5	c.1871G>A (p.Gly624Asp)	rs104886142
COL4A5	c.2215C>T (p.Pro739Ser)	rs104886164
COL4A5	c.2692A>G (p.Met898Val)	rs104886192
COL4A5	c.4228C>T (p.Arg1410Cys)	rs104886270
COL4A5	c.41_42insTCTT (p.Leu14?fs) [c.38_41dupTCTT (p.Leu14PhefsTer27)]	rs104886408
CTNS	c.414G>A (p.Trp138Ter)	rs113994205
CTNS	c.473T>C (p.Leu158Pro)	rs113994206
CTNS	c.1015G>A (p.Gly339Arg)	rs121908127
CTNS	c.124G>A (p.Val42Ile)	rs35086888
CYP21A2	c.710T>A (p.Ile237Asn)	rs111647200
CYP21A2	c.713T>A (p.Val238Glu)	rs12530380
CYP21A2	c.920_921insT (p.Phe307delinsPhefsTer6)	rs267606756
CYP21A2	c.923dupT (p.Leu308Phefs)	rs397515532
CYP21A2	c.844G>C (p.Val282Leu)	rs6471
CYP21A2	c.719T>A (p.Met240Lys)	rs6476
CYP21A2	c.955C>T (p.Gln319Ter)	rs7755898
CYP21A2	c.1069C>T (p.Arg357Trp)	rs7769409
CYP27A1	c.1183C>T (p.Arg395Cys)	rs121908096
CYP27A1	c.1214G>A (p.Arg405Gln)	rs121908099
CYP27A1	c.1435C>T (p.Arg479Cys)	rs72551322
DHCR7	c.452G>A (p.Trp151Ter)	rs11555217
DHCR7	c.3G>A (p.Met1Ile)	rs121909767
DHCR7	c.964-1G>C [c.964-1G>C]	rs138659167
DHCR7	c.841G>A (p.Val281Met)	rs398123607
DHCR7	c.1210C>T (p.Arg404Cys)	rs61757582
DHCR7	c.278C>T (p.Thr93Met)	rs80338853
DHCR7	c.506C>T (p.Ser169Leu)	rs80338855
DHCR7	c.724C>T (p.Arg242Cys)	rs80338856

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
DHCR7	c.725G>A (p.Arg242His)	rs80338857
DHCR7	c.976G>T (p.Val326Leu)	rs80338859
DHCR7	c.1054C>T (p.Arg352Trp)	rs80338860
DHCR7	c.1228G>A (p.Gly410Ser)	rs80338862
DHCR7	c.832-1G>C	rs80338863
DHCR7	c.1342G>A (p.Glu448Lys)	rs80338864
F2	c.*97G>A	rs1799963
F5	c.1000A>G (p.Arg334Gly)	rs118203905
F5	c.1001G>C (p.Arg334Thr)	rs118203906
F5	c.1601G>A( Arg534Gln)	rs6025
FAH	c.456G>A (p.Trp152Ter)	rs370686447
FAH	c.554-1G>T	rs80338895
FAH	c.782C>T (p.Pro261Leu)	rs80338898
FAH	c.1062+5G>A	rs80338901
FSHR	c.2039G>A (p.Ser680Asn)	rs6166
G6PC	c.247C>T (p.Arg83Cys)	rs1801175
G6PC	c.79delC (p.Gln27ArgfsTer9)	rs80356479
G6PC	c.562G>C (p.Gly188Arg)	rs80356482
G6PC	c.1039C>T (p.Gln347Ter)	rs80356487
GALT	c.197C>T (p.Pro66Leu)	rs111033656
GALT	c.413C>T (p.Thr138Met)	rs111033686
GALT	c.404C>T (p.Ser135Leu)	rs111033690
GALT	c.428C>T (p.Ser143Leu)	rs111033697
GALT	c.607G>A (p.Glu203Lys)	rs111033736
GALT	c.667C>A (p.Arg223Ser)	rs111033750
GALT	c.855G>T (p.Lys285Asn)	rs111033773
GALT	c.957C>A (p.His319Gln)	rs111033792
GALT	c.997C>T (p.Arg333Trp)	rs111033800
GALT	c.1006A>T (p.Met336Leu)	rs111033810
GALT	c.1030C>A (p.Gln344Lys)	rs111033814
GALT	c.1132A>G (p.Ile378Val)	rs111033819
GALT	c.652C>G (p.Leu218Val)	rs2070075
GALT	c.772C>T (p.Arg258Cys)	rs368166217
GALT	c.563A>G (p.Gln188Arg)	rs75391579
GBA	c.115+1G>A	rs104886460
GBA	c.1342G>C (p.Asp448His)	rs1064651
GBA	c.1361C>G (p.Pro454Arg)	rs121908295
GBA	c.535G>C (p.Asp179His)	rs147138516
GBA	c.1093G>A (p.Glu365Lys)	rs2230288
GBA	c.680A>G (p.Asn227Ser)	rs364897
GBA	c.882T>G (p.His294Gln)	rs367968666
GBA	c.84_85insG (p.Leu29AlafsTer18)	rs387906315
GBA	c.1448T>C (p.Leu483Pro)	rs421016
GBA	c.1444G>A (p.Asp482Asn)	rs75671029
GBA	c.1085C>T (p.Thr362Ile)	rs76539814

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
GBA	c.1226A>C (p.Asn409Thr)	rs76763715
GBA	c.1504C>T (p.Arg502Cys)	rs80356771
GBA	c.1604G>A (p.Arg535His)	rs80356773
GCDH	c.1204C>T (p.Arg402Trp)	rs121434369
GCDH	c.1093G>A (p.Glu365Lys)	rs121434370
GCDH	c.680G>C (p.Arg227Pro)	rs121434373
GJB2	c.71G>A (p.Trp24Ter)	rs104894396
GJB2	c.229T>C (p.Trp77Arg)	rs104894397
GJB2	c.139G>T (p.Glu47Ter)	rs104894398
GJB2	c.34G>C (p.Gly12Arg)	rs104894408
	c.313_326del (p.Lys105_Gly109del)	
GJB2	[c.313_326delAAGTTCATCAAGGG (p.Lys105GlyfsTer5)]	rs111033253
GJB2	c.617A>G (p.Asn206Ser)	rs111033294
GJB2	c.169C>T (p.Gln57Ter)	rs111033297
GJB2	c.101T>C (p.Met34Thr)	rs35887622
GJB2	c.109G>A (p.Val37Ile)	rs72474224
GJB2	c.416G>A (p.Ser139Asn)	rs76434661
GJB2	c.35delG (p.Gly12ValfsTer2)	rs80338939
GJB2	c.167delT (p.Leu56ArgfsTer26)	rs80338942
GJB2	c.231G>A (p.Trp77Ter)	rs80338944
GJB2	c.269T>C (p.Leu90Pro)	rs80338945
GJB2	c.358_360delGAG (p.Glu120del)	rs80338947
GJB2	c.427C>T (p.Arg143Trp)	rs80338948
GLA	c.1078G>A (p.Gly360Ser)	
GLA	c.838C>A (p.Gln280Lys)	
GLA	c.717_718delAA	
GLA	c.511G>C (p.Gly171Arg)	
GLA	c.404C>T (p.Ala134Val)	
GLA	c.277G>A (p.Asp93Asn)	
GLA	c.427G>A (p.Ala143Thr)	rs104894845
GLA	c.281G>A (p.Cys94Tyr)	rs113173389
GLA	c.463G>C (p.Asp155His)	rs113419388
GLA	c.865A>T (p.Ile289Phe)	rs140329381
GLA	c.352C>T (p.Arg118Cys)	rs148158093
GLA	c.644A>G (p.Asn215Ser)	rs28935197
GLA	c.1025G>A (p.Arg342Gln)	rs28935493
GLA	c.1019_1020insA (p.Trp340Ter)	rs398123197
GLA	c.901C>T (p.Arg301Ter)	rs398123224
GLB1	c.442C>T (p.Arg148Cys)	rs192732174
GLB1	c.601C>T (p.Arg201Cys)	rs72555360
GLB1	c.622C>T (p.Arg208Cys)	rs72555366
GLB1	c.1771T>A (p.Tyr591Asn)	rs72555373
GLB1	c.176G>A (p.Arg59His)	rs72555392
GNPTAB	c.2864C>T (p.Ala955Val)	rs138390866
GNPTAB	c.1514G>A (p.Cys505Tyr)	rs281864980

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
GNPTAB	c.2715+1G>A	rs281865031
GNPTAB	c.3503_3504delTC (p.Leu1168GlnfsTer5)	rs34002892
GNPTAB	c.3335+6T>G	rs34788341
GNPTAB	c.569A>T (p.Asp190Val)	rs34946266
GNPTAB	c.1580delC (p.Ser527Ser=fs) [c.1581delC (p.Cys528ValfsTer19)]	rs36007394
GNPTAB	c.136C>T (p.Arg46Ter)	rs78347057
HADHA	c.1528G>C (p.Glu510Gln)	rs137852769
HADHA	c.1132C>T (p.Gln378Ter)	rs137852770
HADHA	c.1025T>C (p.Leu342Pro)	rs137852772
HADHA	c.845T>A (p.Val282Asp)	rs137852773
HADHA	c.914T>A (p.Ile305Asn)	rs137852774
HADHA	c.871C>T (p.Arg291Ter)	rs137852775
HADHA	c.157C>T (p.Arg53Ter)	rs147103714
HADHA	c.919-2A>G	rs200017313
HADHA	c.180+3A>G	rs781222705
HADHA	c.180+1G>A	rs786205088
HBB	c.93G>T (p.Arg31Ser)	rs1135071
HBB	c.118C>T (p.Gln40Ter)	rs11549407
HBB	c.20A>T (p.Glu7Val)	rs334
HBB	c.316-3C>A	rs33913413
HBB	c.316-2A>G	rs33914668
HBB	c.19G>A(p.Glu7Lys)	rs33930165
HBB	c.315+1G>A	rs33945777
HBB	c.364G>T (p.Glu122Ter)	rs33946267
HBB	c.79G>A (p.Glu27Lys)	rs33950507
HBB	c.92+1G>A	rs33971440
HBB	c.93-21G>A	rs35004220
HEXA	c.805G>A (p.Gly269Ser)	rs121907954
HEXA	c.739C>T (p.Arg247Trp)	rs121907970
HEXA	c.590A>C (p.Lys197Thr)	rs121907973
HEXA	c.1421+1G>C	rs147324677
HEXA	c.1510C>T (p.Arg504Cys)	rs28942071
HEXA	c.1274_1277dupTATC (p.Tyr427IlefsTer5)	rs387906309
HFE	c.845G>A (p.Cys282Tyr)	rs1800562
CHRNE	c.1267G delG (c.1327delG) [c.1327delG (p.Glu443LysfsTer64) ]	rs763258280
IDUA	c.1225G>C (p.Gly409Arg)	rs11934801
IDUA	c.1205G>A (p.Trp402Ter)	rs121965019
IDUA	c.208C>T (p.Gln70Ter)	rs121965020
IDUA	c.1861C>T (p.Arg621Ter)	rs121965025
IDUA	c.1962A>T (p.Ter654Cys) [c.1962A>T (p.Ter654CysTer?)]	rs199794428
IKBKAP	c.2204+6T>C	rs111033171
IKBKAP	c.2087G>C (p.Arg696Pro)	rs137853022
IKBKAP	c.2741C>T (p.Pro914Leu)	rs28939712

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
IKBKAP	c.1814A>G (p.Tyr605Cys)	rs756919296
IL2RG	c.854G>A (p.Arg285Gln)	rs111033617
IL2RG	c.664C>T (p.Arg222Cys)	rs111033618
IL2RG	c.186T>A (p.Cys62Ter)	rs111033619
IL2RG	c.341G>A (p.Gly114Asp)	rs111033620
IL2RG	c.458T>A (p.Ile153Asn)	rs111033621
IL2RG	c.343T>C (p.Cys115Arg)	rs111033622
IL2RG	c.355A>T (p.Lys119Ter)	rs137852507
IL2RG	c.865C>T (p.Arg289Ter)	rs137852508
IL2RG	c.923C>A (p.Ser308Ter)	rs137852509
IL2RG	c.878T>A (p.Leu293Gln)	rs137852510
IL2RG	c.452T>C (p.Leu151Pro)	rs137852511
IL2RG	c.270-1G>T	rs193922346
IL2RG	c.314A>G (p.Tyr105Cys)	rs193922347
IL2RG	c.455T>C (p.Val152Ala)	rs193922348
IL2RG	c.662T>C (p.Phe221Ser)	rs193922349
IL2RG	c.710G>A (p.Trp237Ter)	rs193922350
MCCC1	c.974T>G (p.Met325Arg)	rs119103212
MCCC1	c.1155A>C (p.Arg385Ser)	rs119103213
MCCC1	c.1114C>T (p.Gln372Ter)	rs544349961
MCCC2	c.295G>C (p.Glu99Gln)	rs119103219
MCCC2	c.929C>G (p.Pro310Arg)	rs119103221
MCCC2	c.1309A>G (p.Ile437Val)	rs119103224
MCCC2	c.838G>T (p.Asp280Tyr)	rs119103226
MCCC2	c.568C>T (p.His190Tyr)	rs773774134
MEFV	c.800C>T (p.Thr267Ile)	rs104895081
MEFV	c.2084A>G (p.Lys695Arg)	rs104895094
MEFV	c.2177T>C (p.Val726Ala)	rs28940579
MEFV	c.2040G>C (p.Met680Ile)	rs28940580
MEFV	c.442G>C (p.Glu148Gln)	rs3743930
MEFV	c.2230G>T (p.Ala744Ser)	rs61732874
MEFV	c.2080A>G (p.Met694Val)	rs61752717
MTHFR	c.1129C>T (p.Arg377Cys)	rs121434296
MTHFR	c.665C>T (p.Ala222Val)	rs1801133
MTHFR	c.1743G>A (p.Met581Ile)	rs45590836
MTM1	c.605delT [c.605delT (p.Leu202Trpfs)]	rs672601325
MTM1	c.566A>G (p.Asn189Ser)	rs132630302
MTM1	c.1190A>G (p.Tyr397Cys)	rs132630303
MTM1	c.721C>T (p.Arg241Cys)	rs132630305
MTM1	c.670C>T (p.Arg224Ter)	rs132630306
MTM1	c.469G>A (p.Glu157Lys)	rs132630307
MTM1	c.1261-10A>G	rs397518445
MTM1	c.1040T>G (p.Leu347Ter)	rs398123264
MTM1	c.1054-5G>C	rs398123265
MTM1	c.122A>G (p.Glu41Gly)	rs398123266

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
MTM1	c.1306_1310dupCCTAT (p.Phe438LeufsTer28)	rs398123267
MTM1	c.1357_1358delCC (p.Pro453TyrfsTer4)	rs398123268
MTM1	c.1369G>A (p.Glu457Lys)	rs398123269
MTM1	c.142_143delGA (p.Glu48SerfsTer12)	rs398123270
	c.1537_1564del28insAACTGGA [c.1537_1564delTTCTATACTAAAGAAATCAATCGAGTTTinsAACTGGA (p.Phe513_Leu522delinsAsnTrpIle)]	rs398123271
MTM1	c.1644+1G>T	rs398123272
MTM1	c.595_599delCCTGC (p.Pro199SerfsTer11)	rs398123273
MTM1	c.688T>C (p.Trp230Arg)	rs398123274
MTM1	c.70C>T (p.Arg24Ter)	rs398123275
MYO7A	c.3719G>A (p.Arg1240Gln)	rs111033178
MYO7A	c.2005C>T (p.Arg669Ter)	rs111033201
MYO7A	c.999T>G (p.Tyr333Ter)	rs111033285
MYO7A	c.3764delA (p.Lys1255ArgfsTer8)	rs111033347
MYO7A	c.448C>T (p.Arg150Ter)	rs121965079
MYO7A	c.634C>T (p.Arg212Cys)	rs121965080
MYO7A	c.731G>C (p.Arg244Pro)	rs121965081
MYO7A	c.1797G>A (p.Met599Ile)	rs121965082
MYO7A	c.1884C>A (p.Cys628Ter)	rs121965083
MYO7A	c.1373A>T (p.Asn458Ile)	rs121965084
MYO7A	c.1996C>T (p.Arg666Ter)	rs121965085
MYO7A	c.652G>A (p.Asp218Asn)	rs201539845
MYO7A	c.635G>A (p.Arg212His)	rs28934610
MYO7A	c.93C>A (p.Cys31Ter)	rs35689081
MYO7A	c.5573T>C (p.Leu1858Pro)	rs368657015
MYO7A	c.3728_3729insC (p.Pro1244AlafsTer64)	rs397516304
MYO7A	c.700C>T (p.Gln234Ter)	rs41298133
NBN	c.657_661delACAAA [c.657_661delACAAA (p.Lys137AsnfsTer16) ]	rs587776650
NBN	c.643C>T [c.643C>T (p.Arg215Trp)]	rs34767364
NPC1	c.2665G>A (p.Val889Met)	rs120074130
NPC1	c.3639G>C (p.Leu1213Phe)	rs120074131
NPC1	c.2873G>A (p.Arg958Gln)	rs120074132
NPC1	c.1133T>C (p.Val378Ala)	rs120074134
NPC1	c.2848G>A (p.Val950Met)	rs120074135
NPC1	c.337T>C (p.Cys113Arg)	rs120074136
NPC1	c.2783A>C (p.Gln928Pro)	rs28940897
NPC1	c.3107C>T (p.Thr1036Met)	rs28942104
NPC1	c.3467A>G (p.Asn1156Ser)	rs28942105
NPC1	c.3263A>G (p.Tyr1088Cys)	rs28942106
NPC1	c.2932C>T (p.Arg978Cys)	rs28942108
NPC1	c.2621A>T (p.Asp874Val)	rs372030650
NPC1	c.3265G>A (p.Glu1089Lys)	rs374526072
NPC1	c.2196dupT (p.Pro733SerfsTer10)	rs398123284
NPC1	c.1800delC (p.Ile601PhefsTer13)	rs483352879

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
NPC1	c.416dupC (p.Asn140LysfsTer30)	rs483352880
NPC1	c.2302dupG (p.Val768GlyfsTer4)	rs483352881
NPC1	c.2230_2231delGT (p.Val744SerfsTer27)	rs483352882
NPC1	c.1030delT (p.Ser344LeufsTer105)	rs483352883
NPC1	c.2795dupA (p.Tyr932Ter)	rs483352884
NPC1	c.2128C>T (p.Gln710Ter)	rs483352889
NPC1	c.2777C>T (p.Ala926Val)	rs730880963
NPC1	c.3662delT (p.Phe1221Serfs)	rs786200878
NPC1	c.1142G>A (p.Trp381Ter)	rs794727897
NPC1	c.530G>A (p.Cys177Tyr)	rs80358252
NPC1	c.2324A>C (p.Gln775Pro)	rs80358253
NPC1	c.2974G>T (p.Gly992Trp)	rs80358254
NPC1	c.3019C>G (p.Pro1007Ala)	rs80358257
NPC1	c.3160G>A (p.Ala1054Thr)	rs80358258
NPC1	c.3182T>C (p.Ile1061Thr)	rs80358259
NPC2	c.436C>T (p.Gln146Ter)	rs104894457
NPC2	c.199T>C (p.Ser67Pro)	rs11694
NPC2	c.3G>C (p.Met1Ile) [c.3G>C (p.Met1?)]	rs483352893
NPC2	c.58G>T (p.Glu20Ter)	rs80358260
NPC2	c.115G>A (p.Val39Met)	rs80358261
NPC2	c.133C>T (p.Gln45Ter)	rs80358262
NPC2	c.141C>A (p.Cys47Ter)	rs80358263
NPC2	c.295T>C (p.Cys99Arg)	rs80358264
NPC2	c.332delA (p.Asn111IlefsTer5)	rs80358265
NPC2	c.352G>T (p.Glu118Ter)	rs80358266
NPC2	c.27delG (p.Leu10SerfsTer25)	rs80358267
OTC	c.809A>C (p.Gln270Pro)	rs1800328
OTC	c.386G>A (p.Arg129His)	rs66656800
OTC	c.274C>T (p.Arg92Ter)	rs67418243
OTC	c.140A>C (p.Asn47Thr)	rs67939655
OTC	c.298+5G>C	rs72554348
OTC	c.374C>T (p.Thr125Met)	rs72554356
OTC	c.491C>G (p.Ser164Ter)	rs72556274
OTC	c.548A>G (p.Tyr183Cys)	rs72556293
OTC	c.626C>T (p.Ala209Val)	rs72558417
OTC	c.808C>T (p.Gln270Ter)	rs72558451
OTC	c.829C>T (p.Arg277Trp)	rs72558454
PAH	c.727C>T (p.Arg243Ter)	rs5030846
PAH	c.143T>C (p.Leu48Ser)	rs5030841
PAH	c.473G>A (p.Arg158Gln)	rs5030843
PAH	c.754C>T (p.Arg252Trp)	rs5030847
PAH	c.782G>A (p.Arg261Gln)	rs5030849
PAH	c.842C>T (p.Pro281Leu)	rs5030851
PAH	c.898G>T (p.Ala300Ser)	rs5030853
PAH	c.1066-11G>A	rs5030855

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
PAH	c.1169A>G (p.Glu390Gly)	rs5030856
PAH	c.1208C>T (p.Ala403Val)	rs5030857
PAH	c.1222C>T (p.Arg408Trp)	rs5030858
PAH	c.1241A>G (p.Tyr414Cys)	rs5030860
PAH	c.1315+1G>A	rs5030861
PAH	c.441+5G>T	rs62507321
PAH	c.838G>A (p.Glu280Lys)	rs62508698
PAH	c.284_286delTCA (p.Ile95_Lys96delin) [c.284_286delTCA (p.Ile95del)]	rs62508727
PAH	c.912+1G>A	rs62514956
PAH	c.1042C>G (p.Leu348Val)	rs62516092
PAH	c.117C>G (p.Phe39Leu)	rs62642926
PAH	c.896T>G (p.Phe299Cys)	rs62642933
PAH	c.926C>T (p.Ala309Val)	rs62642935
PAH	c.1139C>T (p.Thr380Met)	rs62642937
PAH	c.527G>T (p.Arg176Leu)	rs74486803
PAH	c.194T>C (p.Ile65Thr)	rs75193786
PAH	c.734T>C (p.Val245Ala)	rs76212747
PCDH15	c.733C>T (p.Arg245Ter)	rs111033260
PCDH15	c.1086delT (p.Asp362Asp=fs) [c.1088delT (p.Leu363TrpfsTer58)]	rs199469706
PCDH15	c.3316C>T (p.Arg1106Ter)	rs202033121
PEX1	c.2230C>T (p.Gln744Ter)	rs398123409
PEX1	c.2097dupT (p.Ile700TyrfsTer42)	rs61750415
PEX1	c.2528G>A (p.Gly843Asp)	rs61750420
PEX1	c.2916delA (p.Gly973AlafsTer16)	rs61750426
PEX1	c.3379dupC (p.Arg1127ProfsTer19)	rs794729652
PEX10	c.373C>T (p.Arg125Ter)	rs61750434
PEX10	c.704dupA (p.Leu236AlafsTer103)	rs61750435
PEX10	c.790C>T (p.Arg264Ter) [c.730C>T (p.Arg244Ter)]	rs61752092
PEX10	c.930C>G (p.His310Gln) [c.870C>G (p.His290Gln)]	rs61752095
PEX10	c.337delC (p.Leu113TrpfsTer40)	rs724159999
PEX10	c.2T>C (p.Met1Thr) [c.2T>C (p.Met1?)]	rs724160002
PEX12	c.691A>T (p.Lys231Ter)	rs104894616
PEX12	c.888_889delCT (p.Leu297ThrfsTer12)	rs398123301
PEX12	c.538C>T (p.Arg180Ter)	rs61752103
PEX13	c.702G>A (p.Trp234Ter)	rs104893661
PEX14	c.553C>T (p.Gln185Ter)	rs61752116
PEX16	c.526C>T (p.Arg176Ter)	rs61752117
PEX2	c.355C>T (p.Arg119Ter)	rs61752123
PEX2	c.669G>A (p.Trp223Ter)	rs61752127
PEX2	c.865dupA (p.Ser289LysfsTer36)	rs724160029
PEX6	c.2440C>T (p.Arg814Ter)	rs267608241
PEX6	c.1601T>C (p.Leu534Pro)	rs387906809
PEX6	c.1338_1339delTG (p.Ala447CysfsTer17)	rs398123303
PEX6	c.689_690dupAG (p.Ser232HisfsTer15)	rs398123305



GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
PEX7	c.653C>T (p.Ala218Val)	rs121909151
PEX7	c.649G>A (p.Gly217Arg)	rs121909152
PEX7	c.694C>T (p.Arg232Ter)	rs121909153
PEX7	c.345T>G (p.Tyr115Ter)	rs121909154
PEX7	c.903+1G>C	rs148591292
PEX7	c.875T>A (p.Leu292Ter)	rs1805137
PEX7	c.45_52dupGGGACGCC [c.45_52dupGGGACGCC (p.His18ArgfsTer35)]	rs199470486
PEX7	c.-45C>T	rs267608252
PEX7	c.340-10A>G	rs267608255
PEX7	c.40A>C (p.Thr14Pro)	rs61753233
PEX7	c.120C>G (p.Tyr40Ter)	rs61753238
PEX7	c.12_18dupGTGCGGT (p.Val4_Gly6) [c.13_19dupTGCGGTG (p.Gly7ValfsTer51)]	rs62636519
PEX7	c.854A>G (p.His285Arg)	rs62653611
PMM2	c.422G>A (p.Arg141His)	rs28936415
PMM2	c.647A>T (p.Asn216Ile)	rs78290141
PMM2	c.338C>T (p.Pro113Leu)	rs80338700
PMM2	c.691G>A (p.Val231Met)	rs80338707
PMM2	c.710C>G (p.Thr237Arg)	rs80338708
SERPINA1	c.1096G>A(p.Glu366Lys )	rs28929474
SGSH	c.220C>T (p.Arg74Cys)	rs104894636
SGSH	c.197C>G (p.Ser66Trp)	rs104894637
SGSH	c.449G>A (p.Arg150Gln)	rs104894638
SGSH	c.383C>T (p.Pro128Leu)	rs104894642
SGSH	c.617G>C (p.Arg206Pro)	rs104894643
SGSH	c.892T>C (p.Ser298Pro)	rs138504221
SLC26A4	c.412G>T (p.Val138Phe)	rs111033199
SLC26A4	c.85G>C (p.Glu29Gln)	rs111033205
SLC26A4	c.1229C>T (p.Thr410Met)	rs111033220
SLC26A4	c.1151A>G (p.Glu384Gly)	rs111033244
SLC26A4	c.2T>C (p.Met1Thr)	rs111033302
SLC26A4	c.626G>T (p.Gly209Val)	rs111033303
SLC26A4	c.1226G>A (p.Arg409His)	rs111033305
SLC26A4	c.1334T>G (p.Leu445Trp)	rs111033307
SLC26A4	c.2015G>A (p.Gly672Glu)	rs111033309
SLC26A4	c.1264-1G>C	rs111033311
SLC26A4	c.1614+1G>A	rs111033312
SLC26A4	c.1149+3A>G	rs111033314
SLC26A4	c.2027T>A (p.Leu676Gln)	rs111033318
SLC26A4	c.578C>T(p.Thr193Ile)	rs111033348
SLC26A4	c.2000T>G (p.Phe667Cys)	rs121908360
SLC26A4	c.1105A>G (p.Lys369Glu)	rs121908361
SLC26A4	c.2168A>G (p.His723Arg)	rs121908362
SLC26A4	c.2162C>T (p.Thr721Met)	rs121908363

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl name]	dbSNP
SLC26A4	c.1115C>T (p.Ala372Val)	rs121908364
SLC26A4	c.397T>A (p.Ser133Thr)	rs121908365
SLC26A4	c.1540C>A (p.Gln514Lys)	rs121908366
SLC26A4	c.1468A>C (p.Ile490Leu)	rs200511789
SLC26A4	c.1246A>C (p.Thr416Pro)	rs28939086
SLC26A4	c.-3-2A>G	rs397516411
SLC26A4	c.1198delT (p.Cys400ValfsTer7)	rs397516413
SLC26A4	c.1336C>T (p.Gln446Ter)	rs397516416
SLC26A4	c.164+2T>C	rs397516420
SLC26A4	c.2188C>T (p.Gln730Ter)	rs397516428
SLC26A4	c.68C>A (p.Ser23Ter)	rs397516430
SLC26A4	c.997_998insA (p.Arg333Lysfs)	rs431905486
SLC26A4	c.554G>C (p.Arg185Thr)	rs542620119
SLC26A4	c.1554G>A (p.Trp518Ter)	rs727503428
SLC26A4	c.707T>C (p.Leu236Pro)	rs80338848
SLC26A4	c.1001+1G>A	rs80338849
SMN1	EX8DEL	
SMPD1	c.1493G>T (p.Arg498Leu)	rs120074117
SMPD1	c.788T>A (p.Leu263Ter)	rs120074120
SMPD1	c.730G>A (p.Gly244Arg)	rs120074122
SMPD1	c.911T>C (p.Leu304Pro)	rs120074124
SMPD1	c.1267C>T (p.His423Tyr)	rs120074126
SMPD1	c.1327C>T (p.Arg443Ter)	rs120074127
SMPD1	c.1426C>T (p.Arg476Trp)	rs182812968
SMPD1	c.1406A>C (p.Tyr469Ser)	rs267607074
SMPD1	c.996delC (p.Phe333SerfsTer52)	rs387906289
SMPD1	c.1420_1421delCT (p.Leu474GlufsTer20)	rs398123476
SMPD1	c.1624C>T (p.Arg542Ter)	rs398123478
SMPD1	c.354delC (p.Ile119SerfsTer7)	rs727504165
SMPD1	c.573delT (p.Ser192AlafsTer65)	rs727504167
TGM1	c.968G>A (p.Arg323Gln)	rs121918717
TGM1	c.1135G>C (p.Val379Leu)	rs121918720
TGM1	c.1187G>T (p.Arg396Leu)	rs121918721
TGM1	c.281G>A (p.Gly94Asp)	rs121918729
TGM1	c.866A>C (p.Asn289Thr)	rs121918730
TGM1	c.919C>T (p.Arg307Trp)	rs121918731
TGM1	c.1075G>A (p.Val359Met)	rs202037016
TPP1	c.1094G>A (p.Cys365Tyr)	rs119455954
TPP1	c.622C>T (p.Arg208Ter)	rs119455955
TPP1	c.509-1G>C	rs56144125
USH1C	c.496+1G>A	
USH1C	c.36+1G>T	
USH1C	c.91C>T (p.Arg31Ter)	rs121908370
USH1C	c.216G>A (p.Val72=) [c.216G>A (p.Val72%3D)]	rs151045328
USH1C	c.308G>A (p.Arg103His)	rs397514500

GEN	HGVSc (HGVS <sub>p</sub> ) [Ensembl nazev]	dbSNP
USH1C	c.388G>A (p.Val130Ile)	rs55843567
USH1C	c.238dupC ( p.Arg80Profs)	rs3975153
USH2A	c.10561T>C (p.Trp3521Arg)	rs111033264
USH2A	c.1606T>C (p.Cys536Arg)	rs111033273
USH2A	c.2209C>T (p.Arg737Ter)	rs111033334
USH2A	c.11864G>A (p.Trp3955Ter)	rs111033364
USH2A	c.6224G>A (p.Trp2075Ter)	rs111033386
USH2A	c.11048-1G>A	rs111033414
USH2A	c.779T>G (p.Leu260Ter)	rs121912598
USH2A	c.1256G>T (p.Cys419Phe)	rs121912600
USH2A	c.14803C>T (p.Arg4935Ter)	rs146733615
USH2A	c.10073G>A (p.Cys3358Tyr)	rs148660051
USH2A	c.2802T>G (p.Cys934Trp)	rs201527662
USH2A	c.11411delC (p.Pro3804LeufsTer13)	rs397517973
USH2A	c.1143+1G>A	rs397517974
USH2A	c.11954G>A (p.Trp3985Ter)	rs397517976
USH2A	c.12714T>G (p.Tyr4238Ter)	rs397517981
USH2A	c.12868C>T (p.Gln4290Ter)	rs397517983
USH2A	c.14180G>A (p.Trp4727Ter)	rs397517989
USH2A	c.14911C>T (p.Arg4971Ter)	rs397517994
USH2A	c.3309C>A (p.Tyr1103Ter)	rs397518011
USH2A	c.3435delA (p.Val1147SerfsTer6)	rs397518012
USH2A	c.3547_3548delAT (p.Ile1183PhefsTer19)	rs397518013
USH2A	c.3558delT (p.Cys1186TrpfsTer51)	rs397518014
USH2A	c.5858-1G>A	rs397518023
USH2A	c.7244C>G (p.Ser2415Ter)	rs397518029
USH2A	c.820C>T (p.Arg274Ter)	rs397518036
USH2A	c.8559-2A>G	rs397518039
USH2A	c.8981G>A (p.Trp2994Ter)	rs397518041
USH2A	c.9159T>G (p.Tyr3053Ter)	rs397518042
USH2A	c.9304C>T (p.Gln3102Ter)	rs397518046
USH2A	c.9424G>T (p.Gly3142Ter)	rs397518048
USH2A	c.6862G>T (p.Glu2288Ter)	rs398124619
USH2A	c.2983C>T (p.Gln995Ter)	rs527236135
USH2A	c.13374delA (p.Glu4458AspfsTer3)	rs727503715
USH2A	c.6398G>A (p.Trp2133Ter)	rs727503725
USH2A	c.2541C>A (p.Cys847Ter)	rs727503736
USH2A	c.14248C>T (p.Gln4750Ter)	rs727504867
USH2A	c.632G>A (p.Trp211Ter)	rs727504893
USH2A	c.11440G>T (p.Gly3814Ter)	rs727505337
USH2A	c.5877delT (p.Ser1961GlnfsTer6)	rs727505343
USH2A	c.9459C>A (p.Cys3153Ter)	rs73090721
USH2A	c.1214delA (p.Asn405IlefsTer3)	rs750228923
USH2A	c.13316C>T (p.Thr4439Ile)	rs753330544
USH2A	c.2299delG (p.Glu767SerfsTer21)	rs80338903

Seznam vyšetřovaných variant - CarrierTest



<b>GEN</b>	<b>HGVSc (HGVS<sub>p</sub>) [Ensembl nazev]</b>
USH2A	c.14020A>G (p.Arg4674Gly)

<b>dbSNP</b>
<a href="#">rs80338904</a>