GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
ACADM	c.199T>C (p.Tyr67His)	rs121434280
ACADM	c.233T>C (p.Ile78Thr)	rs398123074
ACADM	c.347G>A (p.Cys116Tyr)	rs875989859
ACADM	c.362C>T (p.Thr121Ile)	rs121434283
ACADM	c.387+1del	rs786204424
ACADM	c.447G>A (p.Met149Ile)	rs121434277
ACADM	c.455G>A (p.Arg152Lys) [c.443G>A (p.Arg148Lys)]	rs778906552
ACADM	c.583G>A (p.Gly195Arg)	rs121434278
ACADM	c.616C>T (p.Arg206Cys)	rs373715782
ACADM	c.617G>A (p.Arg206His)	rs200724875
ACADM	c.734C>T (p.Ser245Leu)	rs121434281
ACADM	c.799G>A (p.Gly267Arg)	rs121434274
ACADM	c.985A>G (p.Lys329Glu)	rs77931234
ACADM	c.1102_1105delTTAG (p.Ala369LeufsTer18)	rs387906297
ACADS	c.136C>T (p.Arg46Trp)	rs121908003
ACADS	c.164C>T (p.Pro55Leu)	rs147442301
ACADS	c.310_312delGAG (p.Glu104del)	rs387906308
ACADS	c.319C>T (p.Arg107Cys)	rs61732144
ACADS	c.529T>C (p.Trp177Arg)	rs57443665
ACADS	c.682_683delGA (p.Glu228ArgfsTer16)	rs786204691
ACADS	c.973C>T (p.Arg325Trp)	rs121908006
ACADS	c.988C>T (p.Arg330Cys)	rs140853839
ACADS	c.1058C>T (p.Ser353Leu)	rs28941773
ACADS	c.1138C>T (p.Arg380Trp)	rs28940875
ACADS	c.1147C>T (p.Arg383Cys)	rs28940872
ACADVL	c.194C>T (p.Pro65Leu)	rs28934585
ACADVL	c.298_299delCA (p.Gln100ValfsTer3)	rs786204713
ACADVL	c.343delG (p.Glu115LysfsTer2)	rs387906249
ACADVL	c.388_390delGAG (p.Glu130del)	rs387906251
ACADVL	c.520G>A (p.Val174Met)	rs369560930
ACADVL	c.538G>A (p.Ala180Thr)	rs727503791
ACADVL	c.779C>T (p.Thr260Met)	rs113994168
ACADVL	c.848T>C (p.Val283Ala)	rs113994167
ACADVL	c.1182+1G>A	rs113690956
ACADVL	c.1322G>A (p.Gly441Asp)	rs2309689
ACADVL	c.1349G>A (p.Arg450His)	rs118204016
ACADVL	c.1405C>T (p.Arg469Trp)	rs113994170
ACADVL	c.1406G>A (p.Arg469Gln)	rs398123083
ACADVL	c.1679-6G>A	rs113994171
ACADVL	c.1837C>T (p.Arg613Trp)	rs118204014
ADGRV1	c.2398C>T (p.Arg800Ter)	rs373780305
ADGRV1	c.2870dupA (p.Asn957Lysfs)	rs397517429
ADGRV1	c.4702delA (p.Ser1568ValfsTer78)	rs794727347
ADGRV1	c.5643delG (p.Tyr1882IlefsTer6)	rs727503076
ADGRV1	c.6901C>T (p.Gln2301Ter)	rs121909762

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
ADGRV1	c.7374_7375delTG (p.Glu2459GlyfsTer35)	rs397517435
ADGRV1	c.7406G>A (p.Trp2469Ter)	rs397517436
ADGRV1	c.8204delA (p.Asn2735MetfsTer13)	rs794727584
ADGRV1	c.8495C>A (p.Ser2832Ter)	rs121909761
ADGRV1	c.8737delG (p.Val2913TyrfsTer4)	rs397517441
ADGRV1	c.8790delC (p.Met2931TrpfsTer11)	rs796051864
ADGRV1	c.11253C>G (p.Tyr3751Ter)	rs376689763
ADGRV1	c.12631C>T (p.Arg4211Ter)	rs727504777
ADGRV1	c.14973-2A>G	rs371981035
ADGRV1	c.17662delT (p.Ser5888HisfsTer54)	rs397517426
ADGRV1	c.18131A>G (p.Tyr6044Cys)	rs121909763
AGL	c.16C>T (p.Gln6Ter)	rs113994126
AGL	c.18_19delGA (p.Gln6HisfsTer20)	rs113994127
AGL	c.1222C>T (p.Arg408Ter)	rs113994128
AGL	c.1735+1G>T	rs199922945
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.4260-12A>G	rs369973784
AGL	c.4456delT (p.Ser1486ProfsTer18)	rs113994134
AGL	c.4529dupA (p.Tyr1510Ter)	rs387906244
ALPL	c.346G>A (p.Ala116Thr)	rs121918013
ALPL	c.407G>A (p.Arg136His)	rs121918011
ALPL	c.526G>A (p.Ala176Thr)	rs121918019
ALPL	c.535G>A (p.Ala179Thr)	rs121918000
ALPL	c.571G>A (p.Glu191Lys)	rs121918007
ALPL	c.881A>C (p.Asp294Ala)	rs121918002
ALPL	c.979T>C (p.Phe327Leu)	rs121918010
ALPL	c.1133A>T (p.Asp378Val)	rs121918008
ALPL	c.1250A>G (p.Asn417Ser)	rs121918014
ALPL	c.1559delT (p.Leu520ArgfsTer86)	rs387906525
ANXA5	c.-135G>A (v rámci M2 haplotypu)	rs113588187
ANXA5	c.-184T>C (v rámci M2 haplotypu)	rs28651243
ANXA5	c.-210A>C (v rámci M2 haplotypu)	rs28717001
ANXA5	c.-229G>A (v rámci M2 haplotypu)	rs112782763
AR	c.4G>A (p.Glu2Lys)	rs104894742
AR	c.178C>T (p.Gln60Ter)	rs137852575
AR	c.521T>G (p.Leu174Ter)	rs137852590
AR	c.1645C>T (p.Pro549Ser)	rs137852588
AR	c.1732G>A (p.Gly578Arg)	rs137852596
AR	c.1739G>T (p.Cys580Phe)	rs137852586
AR	c.1748T>A (p.Phe583Tyr)	rs137852587
AR	c.1771A>T (p.Lys591Ter)	rs137852566
AR	c.1826G>A (p.Arg609Lys)	rs137852576

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
AR	c.1943G>A (p.Ser648Asn)	rs137852584
AR	c.2069A>C (p.His690Pro)	rs137852599
AR	c.2123T>G (p.Leu708Arg)	rs137852585
AR	c.2137C>T (p.Leu713Phe)	rs137852595
AR	c.2157G>A (p.Trp719Ter)	rs137852563
AR	c.2191G>A (p.Val731Met)	rs137852571
AR	c.2222C>G (p.Ser741Cys)	rs137852601
AR	c.2231G>T (p.Gly744Val)	rs137852600
AR	c.2291A>G (p.Tyr764Cys)	rs137852567
AR	c.2323C>T (p.Arg775Cys)	rs137852562
AR	c.2343G>T (p.Met781Ile)	rs137852589
AR	c.2362A>G (p.Met788Val)	rs137852570
AR	c.2391G>A (p.Trp797Ter)	rs137852565
AR	c.2395C>G (p.Gln799Glu)	rs137852591
AR	c.2423T>C (p.Met808Thr)	rs137852592
AR	c.2567G>A (p.Arg856His)	rs9332971
AR	c.2596T>C (p.Ser866Pro)	rs137852597
AR	c.2610T>G (p.Ile870Met)	rs137852574
AR	c.2623C>T (p.His875Tyr)	rs137852581
AR	c.2650A>T (p.Lys884Ter)	rs137852568
AR	c.2708A>G (p.Gln903Arg)	rs137852582
ARSA	c.256C>T (p.Arg86Trp)	rs199476352
ARSA	c.293C>T (p.Ser98Phe)	rs74315456
ARSA	c.302G>A (p.Gly101Asp)	rs74315455
ARSA	c.465+1G>A	rs80338815
ARSA	c.542T>G (p.Ile181Ser)	rs74315457
ARSA	c.641C>T (p.Ala214Val)	rs74315467
ARSA	c.739G>A (p.Gly247Arg)	rs74315471
ARSA	c.769G>C (p.Asp257His)	rs80338819
ARSA	c.827C>T (p.Thr276Met)	rs74315472
ARSA	c.862A>C (p.Thr288Pro)	rs28940894
ARSA	c.1114C>T (p.Arg372Trp)	rs74315476
ARSA	c.1136C>T (p.Pro379Leu)	rs74315478
ARSA	c.1232C>T (p.Thr411Ile)	rs74315481
ARSA	c.1283C>T (p.Pro428Leu)	rs28940893
ASL	c.35G>A (p.Arg12Gln)	rs145138923
ASL	c.283C>T (p.Arg95Cys)	rs28940585
ASL	c.446+1G>A	rs142637046
ASL	c.532G>A (p.Val178Met)	rs28941473
ASL	c.578G>A (p.Arg193Gln)	rs373697663
ASL	c.762C>A (p.Ser254Arg)	rs869312991
ASPA	c.71A>G (p.Glu24Gly)	rs104894551
ASPA	c.212G>A(p.Arg71His)	rs104894553
ASPA	c.433-2A>G	rs63751297
ASPA	c.454T>C (p.Cys152Arg)	rs104894548

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
ASPA	c.654C>A (p.Cys218Ter)	rs104894549
ASPA	c.693C>A (p.Tyr231Ter)	rs12948217
ASPA	c.746A>T (p.Asp249Val)	rs104894552
ASPA	c.854A>C (p.Glu285Ala)	rs28940279
ASPA	c.914C>A (p.Ala305Glu)	rs28940574
ASS1	c.256C>T (p.Arg86Cys)	rs121908644
ASS1	c.470G>A (p.Arg157His)	rs121908637
ASS1	c.535T>C (p.Trp179Arg)	rs121908646
ASS1	c.787G>A (p.Val263Met)	rs192838388
ASS1	c.814C>T (p.Arg272Cys)	rs762387914
ASS1	c.835C>T (p.Arg279Ter)	rs121908645
ASS1	c.836G>A (p.Arg279Gln)	rs371265106
ASS1	c.970+5G-A	rs372128852
ASS1	c.1168G>A (p.Gly390Arg)	rs121908641
ATM	c.1A>G (p.Met1Val) [c.1A>C (p.Met1Val)]	rs730881359
ATM	c.103C>T (p.Arg35Ter)	rs55861249
ATM	c.151C>T (p.Gln51Ter)	rs786203888
ATM	c.154G>T (p.Gly52Ter)	rs730881362
ATM	c.170G>A (p.Trp57Ter)	rs587779818
ATM	c.237delA (p.Lys79AsnfsTer37)	rs730881303
ATM	c.283C>T (p.Gln95Ter)	rs587781545
ATM	c.381delA (p.Val128Terfs)	rs587781831
ATM	c.513C>G (p.Tyr171Ter)	rs786201693
ATM	c.802C>T (p.Gln268Ter)	rs557012154
ATM	c.1110C>G (p.Tyr370Ter)	rs376170600
ATM	c.1158delG (p.Lys387ArgfsTer3)	rs587782085
ATM	c.1235G>A (p.Trp412Ter)	rs587779813
ATM	c.1424C>G (p.Ser475Ter)	rs786203550
ATM	c.1447_1448delCT (p.Trp484fs)	
ATM	c.1564_1565delGA (p.Glu522IlefsTer)	rs587779817
ATM	c.1737G>A (p.Trp579Ter)	rs786201689
ATM	c.1880dupT (p.Gln628ProfsTer7)	rs786202474
ATM	c.1898+2T>G	rs587782124
ATM	c.2098C>T (p.Gln700Ter)	rs786202743
ATM	c.2129delC (p.Thr710LysfsTer25)	rs786203807
ATM	c.2251-10T>G	rs730881346
ATM	c.2376+1G>T	rs730881347
ATM	c.2426C>A (p.Ser809Ter)	rs730881348
ATM	c.2502dupA (p.Val835SerfsTer7)	rs587779822
ATM	c.2548G>T (p.Glu850Ter)	rs587782280
ATM	c.2754delT (p.Phe918LeufsTer11)	rs786202608
ATM	c.2789T>G (p.Leu930Ter)	rs786203309
ATM	c.2880delC (p.Leu961CysfsTer10)	rs730881300
ATM	c.2921+1G>A	rs587781558
ATM	c.3049C>T (p.Gln1017Ter)	rs730881388

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
ATM	c.3077G>A (p.Trp1026Ter)	rs587782103
ATM	c.3304G>T (p.Gly1102Ter)	rs147557621
ATM	c.3349C>T (p.Gln1117Ter)	rs786201957
ATM	c.3372C>G (p.Tyr1124Ter)	rs587779833
ATM	c.3388G>T (p.Gly1130Ter)	rs587781911
ATM	c.3576G>A (p.Lys1192=) [c.3576G>A (p.Lys1192%3D)]	rs587776551
ATM	c.3802delG (p.Val1268Ter)	rs587779834
ATM	c.3836G>A (p.Trp1279Ter)	rs587779836
ATM	c.3850delA (p.Thr1284fs)	rs876660865
ATM	c.3931C>T (p.Gln1311Ter)	rs200976093
ATM	c.3980T>G (p.Leu1327Ter)	rs587782192
ATM	c.3993+1G>A	rs200196781
ATM	c.3994-2A>G	rs587782276
ATM	c.4052delT (p.Leu1351TyrfsTer35)	rs786202350
ATM	c.4198A>T (p.Lys1400Ter)	rs587781950
ATM	c.4370T>G (p.Leu1457Ter)	rs373226793
ATM	c.4852C>T (p.Arg1618Ter)	rs762083530
ATM	c.5044G>C (p.Asp1682His)	rs121434217
ATM	c.5188C>T (p.Arg1730Ter)	rs764389018
ATM	c.5309C>G (p.Ser1770Ter)	rs121434223
ATM	c.5515C>T (p.Gln1839Ter)	rs786204751
ATM	c.5644C>T (p.Arg1882Ter)	rs786204433
ATM	c.5908C>T (p.Gln1970Ter)	rs587781722
ATM	c.5932G>T (p.Glu1978Ter)	rs587779852
ATM	c.6069-9_6069-5delTTCTT	rs879254095
ATM	c.6095G>A (p.Arg2032Lys)	rs139770721
ATM	c.6096-13_6096-9delTCTTT	
ATM	c.6100C>T (p.Arg2034Ter)	rs532480170
ATM	c.6326G>A (p.Trp2109Ter)	rs587782114
ATM	c.6679C>T (p.Arg2227Cys)	rs564652222
ATM	c.7096G>T (p.Glu2366Ter)	rs587781672
ATM	c.7271T>G (p.Val2424Gly)	rs28904921
ATM	c.7327C>T (p.Arg2443Ter)	rs121434220
ATM	c.7456C>T (p.Arg2486Ter)	rs587779865
ATM	c.7630-2A>C	rs587779866
ATM	c.7875_7876delTGinsGC (p.AspAla2625GluPro)	rs267606668
ATM	c.8147T>C (p.Val2716Ala)	rs587782652
ATM	c.8185C>T (p.Gln2729*)	rs587781967
ATM	c.8494C>T (p.Arg2832Cys)	rs587779872
ATM	c.9046A>G (p.Lys3016Glu)	
ATM	g.91279A [c.5763-1050A>G]	rs774925473
ATP7B	c.1934T>G (p.Met645Arg)	rs121907998
ATP7B	c.1969A>C (p.Ser657Arg)	rs372436901
ATP7B	c.2128G>A (p.Gly710Ser)	rs137853285
ATP7B	c.2305A>G (p.Met769Val)	rs193922103

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
ATP7B	c.2605G>A (p.Gly869Arg)	rs191312027
ATP7B	c.2906G>A (p.Arg969Gln)	rs121907996
ATP7B	c.2930C>T (p.Thr977Met)	rs72552255
ATP7B	c.3207C>A (p.His1069Gln)	rs76151636
ATP7B	c.3402del (p.Ala1135Glnfs)	rs137853281
ATP7B	c.3400delC (p.Ala1135GlnfsTer13)	rs137853286
ATP7B	c.3796G>A (p.Gly1266Arg)	rs121907992
ATP7B	c.3809A>G (p.Asn1270Ser)	rs121907990
ATP7B	c.3955C>T (p.Arg1319Ter)	rs193922109
AZFa	sY84	
AZFa	sY86	
AZFb	sY127	
AZFb	sY134	
AZFb/c	sY254	
AZFc	sY255	
BLM	c.253del (p.Arg85GlyfsTer44)	
BLM	c.461G>A (p.Trp154Ter)	
BLM	c.1642C>T (p.Gln548Ter)	rs200389141
BLM	c.2206_2207insT (p.Tyr736Leufs)	rs886051551
BLM	c.2207_2212delATCTGAins7 [c.2207_2212delATCTGAinsTAGATTC (p.Tyr736LeufsTer5)]	rs113993962
BLM	c.2695C>T (p.Arg899Ter)	rs587779884
BLM	c.3558+1G>A	rs148969222
BTD	c.98_104delGCGGCTGinsTCC (p.Cys33PhefsTer36)	rs80338684
BTD	c.100G>A (p.Gly34Ser)	rs119103232
BTD	c.235C>T (p.Arg79Cys)	rs104893687
BTD	c.262C>T (p.Gln88Ter)	rs151071780
BTD	c.341G>T (p.Gly114Val)	rs375712490
BTD	c.454A>C (p.Thr152Pro)	rs374681173
BTD	c.470G>A (p.Arg157His)	rs146015592
BTD	c.511G>A (p.Ala171Thr) HAPLOTYP[p.Ala171Thr;Asp444His]	rs13073139
BTD	c.528G>T (p.Lys176Asn)	rs397514367
BTD	c.643C>T (p.Leu215Phe)	rs190386869
BTD	c.664G>C (p.Asp222His)	rs200337373
BTD	c.755A>G (p.Asp252Gly)	rs28934601
BTD	c.880A>G (p.Ile294Val)	rs35976361
BTD	c.933delT (p.Ser311ArgfsTer23)	rs397514395
BTD	c.935G>A (p.Gly312Asp)	rs377651057
BTD	c.1046A>C (p.Asn349Thr)	rs200327983
BTD	c.1171C>T (p.Pro391Ser)	rs35034250
BTD	c.1207T>G (p.Phe403Val)	rs104893686
BTD	c.1237G>A (p.Gly413Ser)	rs374141881
BTD	c.1284C>A (p.Tyr428Ter)	rs35145938
BTD	c.1334G>T (p.Gly445Val)	rs35034250
BTD	c.1368A>C (p.Gln456His)	rs80338685

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
BTD	c.1369G>A (p.Val457Met)	rs146600671
BTD	c.1432G>C (p.Ala478Pro)	rs181396238
BTD	c.1455C>G (p.His485Gln)	rs201604102
BTD	c.1466A>C (p.Asn489Thr)	rs104893692
BTD	c.1489C>T (p.Pro497Ser)	rs138818907
BTD	c.1595C>T (p.Thr532Met)	rs104893688
BTD	c.1612C>T (p.Arg538Cys)	rs80338686
CBS	c.341C>T (p.Ala114Val)	rs121964964
CBS	c.374G>A (p.Arg125Gln)	rs781444670
CBS	c.415G>A (p.Gly139Arg)	rs121964965
CBS	c.430G>A (p.Glu144Lys)	rs121964966
CBS	c.572C>T (p.Thr191Met)	rs121964973
CBS	c.797G>A (p.Arg266Lys)	rs121964969
CBS	c.833T>C (p.Ile278Thr)	rs5742905
CBS	c.919G>A (p.Gly307Ser)	rs121964962
CBS	c.954+1G>A	rs1057517373
CBS	c.1006C>T (p.Arg336Cys)	rs398123151
CBS	c.1058C>T (p.Thr353Met)	rs121964972
CBS	c.1330G>A (p.Asp444Asn)	rs28934891
CBS	c.1616T>C (p.Leu539Ser)	rs121964968
CDH23	c.46delG (p.Val16CysfsTer2)	rs397517331
CDH23	c.193del (p.Leu65TrpfsTer49)	rs796051861
CDH23	c.719C>T (p.Pro240Leu)	rs121908354
CDH23	c.902G>A (p.Arg301Gln)	rs121908355
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.4021G>A (p.Asp1341Asn) [c.4036G>A (p.Asp1346Asn)]	rs121908351
CDH23	c.4309C>T (p.Arg1437Ter) [c.4324C>T (p.Arg1442Ter)]	rs397517329
CDH23	c.4488G>C (p.Gln1496His) [c.4503G>C (p.Gln1501His)]	rs121908347
CDH23	c.5237G>A (p.Arg1746Gln) [c.5252G>A (p.Arg1751Gln)]	rs111033270
CDH23	c.5272C>T (p.Gln1758Ter) [c.5287C>T (p.Gln1763Ter)]	rs397517337
CDH23	c.5663T>C (p.Phe1888Ser) [c.5678T>C (p.Phe1893Ser)]	rs121908352
CDH23	c.5712G>A (p.Thr1904=) [c.5727G>A (p.Thr1909%3D)]	rs397517342
CDH23	c.5712+1G>A [c.5727+1G>A]	rs397517341
CDH23	c.5923+1G>A [c.5938+1G>A]	rs397517346
CDH23	c.6049+1G>A	rs111033247
CDH23	c.6050-9G>A	rs367928692
CDH23	c.6133G>A (p.Asp2045Asn) [c.6148G>A (p.Asp2050Asn)]	rs121908348
CDH23	c.6442G>A (p.Asp2148Asn) [c.6457G>A (p.Asp2153Asn)]	rs111033271
CDH23	c.6604G>A (p.Asp2202Asn) [c.6619G>A (p.Asp2207Asn)]	rs121908349
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

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CDH23	c.9629_9632delTCAA (p.Ile3210ArgfsTer5) [c.9644_9647delTCAA (p.Ile3215ArgfsTer5)]	rs397517367
CFTR	c.1A>G (p.Met1Val)	rs397508328
CFTR	c.4C>T (p.Gln2Ter)	rs397508740
CFTR	c.11C>A (p.Ser4Ter)	rs397508173
CFTR	c.38C>T (p.Ser13Phe)	rs397508635
CFTR	c.50del (p.Phe17SerfsTer8)	rs397508714
CFTR	c.53+1G>T	rs397508746
CFTR	c.57G>T (p.Trp19Cys)	rs397508762
CFTR	c.79G>A (p.Gly27Arg)	rs397508796
CFTR	c.79G>C (p.Gly27Arg)	rs397508796
CFTR	c.79G>T (p.Gly27Ter)	rs397508796
CFTR	c.88C>T (p.Gln30Ter)	rs397508815
CFTR	c.115C>T (p.Gln39Ter)	rs397508168
CFTR	c.137C>A (p.Ala46Asp)	rs151020603
CFTR	c.164+1G>A	rs397508243
CFTR	c.164+1G>T	rs397508243
CFTR	c.164+2T>C	rs121908800
CFTR	c.164+4dup	rs397508244
CFTR	c.165-1G>A	rs397508249
CFTR	c.165-3C>T	rs200337193
CFTR	c.166G>A (p.Glu56Lys)	rs397508256
CFTR	c.169T>G (p.Trp57Gly)	rs397508272
CFTR	c.170G>A (p.Trp57Ter)	rs397508279
CFTR	c.171G>A (p.Trp57Ter)	rs121909025
CFTR	c.174_177del (p.Asp58GlufsTer32)	rs397508295
CFTR	c.175dup (p.Arg59LysfsTer10)	rs397508294
CFTR	c.178G>T (p.Glu60Ter)	rs77284892
CFTR	c.200C>T (p.Pro67Leu)	rs368505753
CFTR	c.223C>T (p.Arg75Ter)	rs121908749
CFTR	c.233dup (p.Trp79LeufsTer32)	rs397508366
CFTR	c.254G>A (p.Gly85Glu)	rs75961395
CFTR	c.262_263delTT (p.Leu88IlefsTer22)	rs121908769
CFTR	c.263T>A (p.Leu88Ter)	rs397508412
CFTR	c.263T>G (p.Leu88Ter)	rs397508412
CFTR	c.271G>A (p.Gly91Arg)	rs121908750
CFTR	c.273+1G>A	rs121908791
CFTR	c.273+3A>C	rs74467662
CFTR	c.274G>A (p.Glu92Lys)	rs121908751
CFTR	c.274G>T (p.Glu92Ter)	rs121908751
CFTR	c.274-1G>A	rs121908792
CFTR	c.274-2A>G	rs397508426
CFTR	c.292C>T (p.Gln98Ter)	rs397508461
CFTR	c.293A>G (p.Gln98Arg)	rs397508464
CFTR	c.296C>T (p.Pro99Leu)	rs397508467

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CFTR	c.305T>G (p.Leu102Arg)	rs397508490
CFTR	c.310del (p.Arg104GlufsTer3)	rs397508499
CFTR	c.313delA (p.Ile105SerfsTer2) [c.444delA (p.Ile105SerfsTer2)]	rs121908801
CFTR	c.325_327delinsG (p.Tyr109GlyfsTer4)	rs121908798
CFTR	c.328G>C (p.Asp110His)	rs113993958
CFTR	c.349C>T (p.Arg117Cys)	rs77834169
CFTR	c.350G>A (p.Arg117His)	rs78655421
CFTR	c.350G>C (p.Arg117Pro)	rs78655421
CFTR	c.366T>A (p.Tyr122Ter)	rs79660178
CFTR	c.412_413insACT (p.Leu137_Leu138insHis)	rs397508679
CFTR	c.416A>G (p.His139Arg)	rs76371115
CFTR	c.442del (p.Ile148LeufsTer5)	rs121908770
CFTR	c.481T>G (p.Tyr161Asp)	rs397508729
CFTR	c.489+1G>T	rs78756941
CFTR	c.494T>C (p.Leu165Ser)	rs397508736
CFTR	c.531del (p.Ile177MetfsTer12)	rs121908771
CFTR	c.532G>A (p.Gly178Arg)	rs80282562
CFTR	c.543_546del (p.Leu183PhefsTer5)	rs397508750
CFTR	c.577G>T (p.Glu193Ter)	rs397508759
CFTR	c.579+1G>T (711+1 G>T)	rs77188391
CFTR	c.579+3A>G	rs397508761
CFTR	c.579+5G>A	rs78440224
CFTR	c.580-1G>T	rs121908793
CFTR	c.595C>T (p.His199Tyr)	rs121908802
CFTR	c.613C>T (p.Pro205Ser)	rs121908803
CFTR	c.617T>G (p.Leu206Trp)	rs121908752
CFTR	c.647G>A (p.Trp216Ter)	rs397508775
CFTR	c.658C>T (p.Gln220Ter)	rs397508778
CFTR	c.680T>G (p.Leu227Arg)	rs397508782
CFTR	c.695T>A (p.Val232Asp)	rs397508783
CFTR	c.723_743+1del	rs121908804
CFTR	c.803del (p.Asn268IlefsTer17)	rs121908772
CFTR	c.825C>G (p.Tyr275Ter)	rs193922532
CFTR	c.828C>A (p.Cys276Ter)	rs397508799
CFTR	c.861_865del (p.Asn287LysfsTer19)	rs397508805
CFTR	c.933C>G (p.Phe311Leu)	rs121909016
CFTR	c.935_937delTCT (p.Phe312del)	rs121908768
CFTR	c.948delT (p.Phe316LeufsTer12)	rs75528968
CFTR	c.987del (p.Gly330GlufsTer39)	rs397508824
CFTR	c.988G>T (p.Gly330Ter)	rs79031340
CFTR	c.1000C>T (p.Arg334Trp)	rs121909011
CFTR	c.1006_1007insG (p.Ile336SerfsTer28)	rs397508138
CFTR	c.1007T>A (p.Ile336Lys)	rs397508139
CFTR	c.1013C>T (p.Thr338Ile)	rs77409459
CFTR	c.1021T>C (p.Ser341Pro)	rs397508144

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CFTR	c.1021_1022dup (p.Phe342HisfsTer28)	rs387906360
CFTR	c.1029del (p.Cys343Ter)	rs121908774
CFTR	c.1040G>A (p.Arg347His)	rs77932196
CFTR	c.1040G>C (p.Arg347Pro)	rs77932196
CFTR	c.1055G>A (p.Arg352Gln)	rs121908753
CFTR	c.1075_1079delinsAAAAA (p.Gln359_Thr360delinsLysLys)	rs397508152
CFTR	c.1079C>A (p.Thr360Lys)	rs75053309
CFTR	c.1081del (p.Trp361GlyfsTer8)	rs387906361
CFTR	c.1116+1G>A	rs397508158
CFTR	c.1117-1G>A	rs797045160
CFTR	c.1130dup (p.Gln378AlafsTer4)	rs397508163
CFTR	c.1155_1156dup (p.Asn386IlefsTer3)	rs121908785
CFTR	c.1202G>A (p.Trp401Ter)	rs397508174
CFTR	c.1203G>A (p.Trp401Ter)	rs397508175
CFTR	c.1209+1G>A	rs397508176
CFTR	c.1210-7_1210-6del (+12/13TG); 5T_12/13GT	rs1805177
CFTR	c.1240C>T (p.Gln414Ter)	rs397508183
CFTR	c.1301_1307del (p.Ser434LeufsTer6)	rs397508186
CFTR	c.1301_1307delCACTTCT (p.Ser434LeufsTer6)	rs397508186
CFTR	c.1327G>T (p.Asp443Tyr)	rs147422190
CFTR	c.1327_1330dup (p.Ile444ArgfsTer3)	rs397508189
CFTR	c.1340del (p.Lys447ArgfsTer2)	rs397508192
CFTR	c.1364C>A (p.Ala455Glu)	rs74551128
CFTR	c.1365_1366del (p.Val456CysfsTer25)	rs797045161
CFTR	c.1365_1366delGG (p.Val456CysfsTer25)	rs797045161
CFTR	c.1367T>C (p.Val456Ala)	rs193922500
CFTR	c.1373del (p.Gly458AspfsTer11)	rs397508196
CFTR	c.1393-1G>A	rs397508200
CFTR	c.1393-2A>G	rs397508201
CFTR	c.1397C>A (p.Ser466Ter)	rs121908805
CFTR	c.1397C>G (p.Ser466Ter)	rs121908805
CFTR	c.1400T>C (p.Leu467Pro)	rs139573311
CFTR	c.1418del (p.Gly473GlufsTer54)	rs397508205
CFTR	c.1466C>A (p.Ser489Ter)	rs397508211
CFTR	c.1475C>T (p.Ser492Phe)	rs121909017
CFTR	c.1477C>T (p.Gln493Ter)	rs77101217
CFTR	c.1477_1478del (p.Gln493ValfsTer10)	rs121908775
CFTR	c.1487G>A (p.Trp496Ter)	rs397508216
CFTR	c.1505T>C (p.Ile502Thr)	rs397508222
CFTR	c.1519_1521del (p.I507del)	rs121908745
CFTR	c.1519_1521delATC (p.Ile507del)	rs121908745
CFTR	c.1521_1523delCTT (p.Phe508del)	rs113993960
CFTR	c.1538A>G (p.Asp513Gly)	rs397508225
CFTR	c.1545_1546delTA (p.Tyr515_Arg516delins) [c.1545_1546delTA (p.Tyr515Ter)]	rs121908776

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CFTR	c.1558G>T (p.Val520Phe)	rs77646904
CFTR	c.1572C>A (p.Cys524Ter)	rs121908754
CFTR	c.1573C>T (p.Gln525Ter)	rs397508227
CFTR	c.1584+1G>A	rs397508230
CFTR	c.1585-1G>A [1717-1G>A]	rs76713772
CFTR	c.1585-8G>A	rs193922503
CFTR	c.1624G>T (p.Gly542Ter)	rs113993959
CFTR	c.1645A>C (p.Ser549Arg)	rs121908757
CFTR	c.1646G>A (p.Ser549Asn)	rs121908755
CFTR	c.1647T>G (p.Ser549Arg)	rs121909005
CFTR	c.1648G>T (p.Gly550Ter)	rs397508247
CFTR	c.1650del (p.Gly551ValfsTer8)	rs397508251
CFTR	c.1651G>A (p.Gly551Ser)	rs121909013
CFTR	c.1652G>A (p.Gly551Asp)	rs75527207
CFTR	c.1654C>T (p.Gln552Ter)	rs76554633
CFTR	c.1657C>T (p.Arg553Ter)	rs74597325
CFTR	c.1670del (p.Ser557PhefsTer2)	rs397508257
CFTR	c.1673T>C (p.Leu558Ser)	rs193922504
CFTR	c.1675G>A (p.Ala559Thr)	rs75549581
CFTR	c.1679G>A (p.Arg560Lys)	rs80055610
CFTR	c.1679G>C (p.Arg560Thr)	rs80055610
CFTR	c.1679+1G>A	rs397508263
CFTR	c.1680A>C (p.Arg560Ser)	rs397508267
CFTR	c.1680-1G>A	rs121908794
CFTR	c.1680-877G>T	rs397508261
CFTR	c.1680-886A>G	rs397508266
CFTR	c.1680-886A>G	rs397508266
CFTR	c.1682C>A (p.Ala561Glu)	rs121909047
CFTR	c.1687T>A (p.Tyr563Asn)	rs121909006
CFTR	c.1687T>G (p.Tyr563Asp)	rs121909006
CFTR	c.1692del (p.Asp565MetfsTer7)	rs193922505
CFTR	c.1703del (p.Leu568CysfsTer4)	rs397508274
CFTR	c.1705T>G (p.Tyr569Asp)	rs397508276
CFTR	c.1721C>A (p.Pro574His)	rs121908758
CFTR	c.1753G>T (p.Glu585Ter)	rs397508296
CFTR	c.1766+1G>A	rs121908748
CFTR	c.1766+3A>G	rs397508298
CFTR	c.1766+5G>T	rs121908796
CFTR	c.1792_1798del (p.Lys598GlyfsTer11)	rs397508303
CFTR	c.1826A>G (p.His609Arg)	rs397508310
CFTR	c.1837G>A (p.Ala613Thr)	rs201978662
CFTR	c.1923_1931delinsA (p.Ser641ArgfsTer5)	rs121908779
CFTR	c.1973_1985delinsAGAAA (p.Arg658LysfsTer4)	rs121908780
CFTR	c.1986_1989del (p.Thr663ArgfsTer8)	rs397508325
CFTR	c.2012delT (p.Leu671Ter)	rs121908812

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CFTR	c.2017G>T (p.Gly673Ter)	rs397508331
CFTR	c.2051_2052delinsG (p.Lys684SerfsTer38)	rs121908799
CFTR	c.2052delA (p.Lys684AsnfsTer38)	rs121908746
CFTR	c.2052dup (p.Gln685ThrfsTer4)	rs121908786
CFTR	c.2053C>T (p.Gln685Ter)	rs397508336
CFTR	c.2053dup (p.Gln685ProfsTer84)	rs797045162
CFTR	c.2125C>T (p.Arg709Ter)	rs121908760
CFTR	c.2126G>A (p.Arg709Gln)	rs397508342
CFTR	c.2128A>T (p.Lys710Ter)	rs75115087
CFTR	c.2143C>T (p.Gln715Ter)	rs397508343
CFTR	c.2158C>T (p.Gln720Ter)	rs397508346
CFTR	c.2175dup (p.Glu726ArgfsTer4)	rs121908787
CFTR	c.2195T>G (p.Leu732Ter)	rs397508350
CFTR	c.2215delG (p.Val739TyrfsTer16)	rs397508353
CFTR	c.2241_2248del (p.Ile748SerfsTer28)	rs397508355
CFTR	c.2290C>T (p.Arg764Ter)	rs121908810
CFTR	c.2353C>T (p.Arg785Ter)	rs374946172
CFTR	c.2374C>T (p.Arg792Ter)	rs145449046
CFTR	c.2423_2424dup (p.Ser809IlefsTer13)	rs387906359
CFTR	c.2453del (p.Leu818TrpfsTer3)	rs397515498
CFTR	c.2463_2464del (p.Ser821ArgfsTer4)	rs797045156
CFTR	c.2463_2464delTG (p.Ser821ArgfsTer4)	rs797045156
CFTR	c.2464G>T (p.Glu822Ter)	rs397508378
CFTR	c.2490+1G>A	rs141158996
CFTR	c.2491G>T (p.Glu831Ter)	rs397508387
CFTR	c.2502dup(p.Asp835Ter)	rs397508389
CFTR	c.2537G>A (p.Trp846Ter)	rs397508393
CFTR	c.2538G>A (p. Trp846Ter)	rs267606722
CFTR	c.2547C>A (p.Tyr849Ter)	rs397508394
CFTR	c.2551C>T (p.Arg851Ter)	rs121909012
CFTR	c.2583del (p.Phe861LeufsTer3)	rs397508399
CFTR	c.2589_2599del (p.Ile864SerfsTer28)	rs397508400
CFTR	c.2601dup (p.Val868SerfsTer28)	rs397508405
CFTR	c.2645G>A (p.Trp882Ter)	rs397508413
CFTR	c.2657+5G>A	rs80224560
CFTR	c.2658-1G>C	rs397508416
CFTR	c.2658-1G>T	rs397508416
CFTR	c.2668C>T (p.Gln890Ter)	rs79633941
CFTR	c.2735C>T (p.Ser912Leu)	rs121909034
CFTR	c.2737_2738insG (p.Tyr913Ter)	rs121908788
CFTR	c.2739T>A (p.Tyr913Ter)	rs149790377
CFTR	c.2763_2764dup (p.Val922GlufsTer2)	rs397508431
CFTR	c.2780T>C (p.Leu927Pro)	rs397508435
CFTR	c.2810dup (p.Val938GlyfsTer37)	rs193922510
CFTR	c.2825del (p.Ile942ThrfsTer26)	rs397508441

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CFTR	c.2834C>T (p.Ser945Leu)	rs397508442
CFTR	c.2859_2890del (p.Leu953PhefsTer11)	rs397508445
CFTR	c.2875del (p.Ala959HisfsTer9)	rs397508447
CFTR	c.2896del (p.Thr966ArgfsTer2)	rs397508451
CFTR	c.2908G>C (p.Gly970Arg)	rs397508453
CFTR	c.2909G>A (p.Gly970Asp)	rs386134230
CFTR	c.2936A>T (p.Asp979Val)	rs397508462
CFTR	c.2988G>A (p.Gln996%3D)	rs121908797
CFTR	c.2988+1G>A	rs75096551
CFTR	c.2989-1G>A	rs397508470
CFTR	c.2989-2A>G	rs193922515
CFTR	c.3002_3003del (p.Val1001AspfsTer45)	rs397508477
CFTR	c.3009_3017delAGCTATAGC (p.Ala1004_Ala1006del)	
CFTR	c.3011_3019delCTATAGCAG (p.Ala1004_Ala1006del)	
CFTR	c.3017C>A (p.Ala1006Glu)	rs397508480
CFTR	c.3039del (p.Tyr1014ThrfsTer9)	rs121908781
CFTR	c.3039dup (p.Tyr1014LeufsTer33)	rs121908781
CFTR	c.3124C>T (p.Gln1042Ter)	rs397508500
CFTR	c.3139_3139+1del	rs397508505
CFTR	c.3140-26A>G	rs76151804
CFTR	c.3160C>G (p.His1054Asp)	rs397508510
CFTR	c.3181G>C (p.Gly1061Arg)	rs142394380
CFTR	c.3194T>C (p.Leu1065Pro)	rs121909036
CFTR	c.3196C>T (p.Arg1066Cys)	rs78194216
CFTR	c.3197G>A (p.Arg1066His)	rs121909019
CFTR	c.3199G>A (p.Ala1067Thr)	rs121909020
CFTR	c.3217dup (p.Tyr1073Leufs)	
CFTR	c.3230T>C (p.Leu1077Pro)	rs139304906
CFTR	c.3266G>A (p.Trp1089Ter)	rs78802634
CFTR	c.3276C>A (p.Tyr1092Ter)	rs121908761
CFTR	c.3293G>A (p.Trp1098Ter)	rs397508532
CFTR	c.3294G>A (p.Trp1098Ter)	rs397508533
CFTR	c.3294G>C (p.Trp1098Cys)	rs397508533
CFTR	c.3302T>A (p.Met1101Lys)	rs36210737
CFTR	c.3304A>T (p.Arg1102Ter)	rs397508536
CFTR	c.3310G>T (p.Glu1104Ter)	rs397508538
CFTR	c.3353C>T (p.Ser1118Phe)	rs146521846
CFTR	c.3368-2A>G	rs755416052
CFTR	c.3435G>A (p.Trp1145Ter)	rs397508561
CFTR	c.3454G>C (p.Asp1152His)	rs75541969
CFTR	c.3468G>A (p.Leu1156%3D)	rs139729994
CFTR	c.3472C>T (p.Arg1158Ter)	rs79850223
CFTR	c.3475T>C (p.Ser1159Pro)	rs397508572
CFTR	c.3476C>T (p.Ser1159Phe)	rs397508573
CFTR	c.3484C>T (p.Arg1162Ter)	rs74767530

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CFTR	c.3528delC (p.Thr1176Thr=fs) [c.3528delC (p.Lys1177SerfsTer15)]	rs78984783
CFTR	c.3532_3535dup (p.Thr1179IlefsTer17)	rs387906378
CFTR	c.3587C>G (p.Ser1196Ter)	rs121908763
CFTR	c.3605del (p.Asp1202AlafsTer9)	rs397508587
CFTR	c.3611G>A (p.Trp1204Ter)	rs121908764
CFTR	c.3612G>A (p.Trp1204Ter)	rs121908765
CFTR	c.3659delC (p.Thr1220Lysfs)	rs121908811
CFTR	c.3691del (p.Ser1231ProfsTer4)	rs77035409
CFTR	c.3700A>G (p.Ile1234Val)	rs75389940
CFTR	c.3717G>A (p.Arg1239%3D)	rs144781064
CFTR	c.3717+4A>G	rs387906362
CFTR	c.3717+5G>A	rs193922520
CFTR	c.3717+12191C>T (3849+10kbC>T) [c.3718-2477C>T]	rs75039782
CFTR	c.3718-1G>A	rs387906369
CFTR	c.3718-3T>G	rs397508596
CFTR	c.3719T>G (p.Val1240Gly)	rs397508598
CFTR	c.3731G>A (p.Gly1244Glu)	rs267606723
CFTR	c.3744del (p.Lys1250ArgfsTer9)	rs121908784
CFTR	c.3744delA (p.Lys1250ArgfsTer9)	rs121908784
CFTR	c.3752G>A (p.Ser1251Asn)	rs74503330
CFTR	c.3761T>G (p.Leu1254Ter)	rs397508604
CFTR	c.3763T>C (p.Ser1255Pro)	rs121909041
CFTR	c.3764C>A (p.Ser1255Ter)	rs76649725
CFTR	c.3773_3774insT (p.Leu1258delinsPheGlu) [c.3773dupT (p.Leu1258PhefsTer7)]	rs121908789
CFTR	c.3846G>A (p.Trp1282Ter)	rs77010898
CFTR	c.3848G>T (p.Arg1283Met)	rs77902683
CFTR	c.3873+1G>A	rs143570767
CFTR	c.3873+2T>C	rs146795445
CFTR	c.3883del (p.Ile1295PhefsTer33)	rs397508630
CFTR	c.3883_3886del (p.Ile1295PhefsTer32)	rs387906373
CFTR	c.3889dup (p.Ser1297PhefsTer5)	rs121908808
CFTR	c.3908del (p.Asn1303ThrfsTer25)	rs397508637
CFTR	c.3909C>G (p.Asn1303Lys)	rs80034486
CFTR	c.3937C>T (p.Gln1313Ter)	rs121909026
CFTR	c.3971T>C (p.Leu1324Pro)	rs397508653
CFTR	c.3988C>G (p.Gln1330Glu)	rs375661578
CFTR	c.3988C>T (p.Gln1330Ter)	rs375661578
CFTR	c.4004T>C (p.Leu1335Pro)	rs397508658
CFTR	c.4036_4042del (p.Leu1346MetfsTer6)	rs397508662
CFTR	c.4046G>A (p.Gly1349Asp)	rs193922525
CFTR	c.4056G>C (p.Gln1352His)	rs113857788
CFTR	c.4077_4080delinsAA (p.Val1360ThrfsTer3)	rs397508668
CFTR	c.4086dup (p.Lys1363Ter)	rs397508669
CFTR	c.4111G>T (p.Glu1371Ter)	rs397508675

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CFTR	c.4124A>C (p.His1375Pro)	rs397508678
CFTR	c.4127_4131delTGGAT (p.Leu1376Serfs)	
CFTR	c.4144C>T (p.Gln1382Ter)	rs397508684
CFTR	c.4147dup (p.Ile1383AsnfsTer3)	rs397508685
CFTR	c.4197_4198del (p.Cys1400Ter)	rs397508693
CFTR	c.4231C>T (p.Gln1411Ter)	rs397508701
CFTR	c.4234C>T (p.Gln1412Ter)	rs397508702
CFTR	c.4242+1G>A	rs372227120
CFTR	c.4242+1G>T	rs372227120
CFTR	c.4251del (p.Glu1418ArgfsTer14)	rs397508706
CFTR	c.4300_4301dup (p.Ser1435GlyfsTer14)	rs397508709
CFTR	c.4426C>T (p.Gln1476Ter)	rs374705585
CFTR	c.-9_14del	rs397508136
CFTR	c.54-5940_273+10250del21kb [CFTR exon 2-3 delece; c.54-5940_273+10250del21kb (p.Ser18ArgfsX16)]	
CHRNE	c.1267G delG (c.1327delG) [c.1327delG (p.Glu443LysfsTer64)]	rs763258280
CLRN1	c.92C>T (p.Pro31Leu)	rs374390376
CLRN1	c.118T>G (p.Cys40Gly)	rs121908143
CLRN1	c.301_305delGTCAT (p.Val101_Ile102) [c.301_305delGTCAT (p.Val101SerfsTer27)]	rs397517932
CLRN1	c.567T>G (p.Tyr189Ter) [c.528T>G (p.Tyr176Ter)]	rs121908140
CLRN1	c.144T>G (p.Asn48Lys)	rs111033258
COL4A5	c.41_42insTCTT (p.Leu14?fs) [c.38_41dupTCTT (p.Leu14PhefsTer27)]	rs104886408
COL4A5	c.1871G>A (p.Gly624Asp)	rs104886142
COL4A5	c.2215C>T (p.Pro739Ser)	rs104886164
CTNS	c.124G>A (p.Val42Ile)	rs35086888
CTNS	c.414G>A (p.Trp138Ter)	rs113994205
CTNS	c.473T>C (p.Leu158Pro)	rs113994206
CTNS	c.1015G>A (p.Gly339Arg)	rs121908127
CYP21A2	c.293-13C>G	rs6467
CYP21A2	c.844G>C/T (p.Val282Leu)	rs6471
CYP21A2	c.1069C>T (p.Arg357Trp)	rs7769409
	CarrierTest is only a screening method detecting frequent variants of *CYP21A2* gene: c.293-13C>G; p.Arg357Trp and p.Val282Leu. Variant p.Val282Leu is associated with milder phenotype. Due to the existence of highly homologue pseudogene and gene rearrangements in respective genomic region, this method does not detect presence of chimeric genes (present in 30 % of patients), copy number variations of parts of *CYP21A2* or whole gene, frequent mutation p.Ile173Asn (in 11 % of patients) and other pathogenic mutations of *CYP21A2* gene.	
CYP27A1	c.1183C>T (p.Arg395Cys)	rs121908096
CYP27A1	c.1184G>A (p.Arg395His)	rs587778778
CYP27A1	c.1184+1G>A	rs587778777

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
CYP27A1	c.1214G>A (p.Arg405Gln)	rs121908099
CYP27A1	c.1381C>T (p.Gln461Ter)	rs771819245
CYP27A1	c.1435C>T (p.Arg479Cys)	rs72551322
DHCR7	c.3G>A (p.Met1Ile)	rs121909767
DHCR7	c.278C>T (p.Thr93Met)	rs80338853
DHCR7	c.326T>C (p.Leu109Pro)	rs121912195
DHCR7	c.452G>A (p.Trp151Ter)	rs11555217
DHCR7	c.506C>T (p.Ser169Leu)	rs80338855
DHCR7	c.724C>T (p.Arg242Cys)	rs80338856
DHCR7	c.725G>A (p.Arg242His)	rs80338857
DHCR7	c.740C>T (p.Ala247Val)	rs886041354
DHCR7	c.832-1G>C	rs80338863
DHCR7	c.841G>A (p.Val281Met)	rs398123607
DHCR7	c.964-1G>C	rs138659167
DHCR7	c.964-1G>T	rs138659167
DHCR7	c.976G>T (p.Val326Leu)	rs80338859
DHCR7	c.1054C>T (p.Arg352Trp)	rs80338860
DHCR7	c.1190C>T (p.Ser397Leu)	rs773134475
DHCR7	c.1210C>T (p.Arg404Cys)	rs61757582
DHCR7	c.1228G>A (p.Gly410Ser)	rs80338862
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.79delC (p.Gln27ArgfsTer9)	rs80356479
G6PC	c.247C>T (p.Arg83Cys)	rs1801175
G6PC	c.562G>C (p.Gly188Arg)	rs80356482
G6PC	c.1039C>T (p.Gln347Ter)	rs80356487
GALT	c.197C>T (p.Pro66Leu)	rs111033656
GALT	c.404C>T (p.Ser135Leu)	rs111033690
GALT	c.413C>T (p.Thr138Met)	rs111033686
GALT	c.428C>T (p.Ser143Leu)	rs111033697
GALT	c.563A>G (p.Gln188Arg)	rs75391579
GALT	c.584T>C (p.Leu195Pro)	rs111033728
GALT	c.607G>A (p.Glu203Lys)	rs111033736
GALT	c.652C>G (p.Leu218Val)	rs2070075
GALT	c.667C>A (p.Arg223Ser)	rs111033750
GALT	c.776G>A (p.Arg259Gln)	rs886042070
GALT	c.855G>T (p.Lys285Asn)	rs111033773

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
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
GBA	c.84_85insG (p.Leu29AlafsTer18)	rs387906315
GBA	c.115+1G>A	rs104886460
GBA	c.535G>C (p.Asp179His)	rs147138516
GBA	c.680A>G (p.Asn227Ser)	rs364897
GBA	c.745C>T (p.Arg249Trp)	rs138058578
GBA	c.882T>G (p.His294Gln)	rs367968666
GBA	c.1085C>T (p.Thr362Ile)	rs76539814
GBA	c.1093G>A (p.Glu365Lys)	rs2230288
GBA	c.1226A>C (p.Asn409Thr)	rs76763715
GBA	c.1297G>T (p.Val433Leu)	rs80356769
GBA	c.1342G>C (p.Asp448His)	rs1064651
GBA	c.1361C>G (p.Pro454Arg)	rs121908295
GBA	c.1444G>A (p.Asp482Asn)	rs75671029
GBA	c.1448T>C (p.Leu483Pro)	rs421016
GBA	c.1504C>T (p.Arg502Cys)	rs80356771
GBA	c.1604G>A (p.Arg535His)	rs75822236
GCDH	c.680G>C (p.Arg227Pro)	rs121434373
GCDH	c.1093G>A (p.Glu365Lys)	rs121434370
GCDH	c.1198G>A (p.Val400Met)	rs121434372
GCDH	c.1204C>T (p.Arg402Trp)	rs121434369
GCDH	c.1262C>T (p.Ala421Val)	rs121434367
GJB2	c.1A>G (p.Met1Val)	rs111033293
GJB2	c.34G>C (p.Gly12Arg)	rs104894408
GJB2	c.35delG (p.Gly12ValfsTer2)	rs80338939
GJB2	c.71G>A (p.Trp24Ter)	rs104894396
GJB2	c.109G>A (p.Val37Ile)	rs72474224
GJB2	c.139G>T (p.Glu47Ter)	rs104894398
GJB2	c.167delT (p.Leu56ArgfsTer26)	rs80338942
GJB2	c.169C>T (p.Gln57Ter)	rs111033297
GJB2	c.229T>C (p.Trp77Arg)	rs104894397
GJB2	c.231G>A (p.Trp77Ter)	rs80338944
GJB2	c.269T>C (p.Leu90Pro)	rs80338945
GJB2	c.313_326del (p.Lys105_Gly109del) [c.313_326delAAGTTCATCAAGGG (p.Lys105GlyfsTer5)]	rs111033253
GJB2	c.358_360delGAG (p.Glu120del)	rs80338947
GJB2	c.416G>A (p.Ser139Asn)	rs76434661
GJB2	c.427C>T (p.Arg143Trp)	rs80338948
GJB2	c.551G>A (p.Arg184Gln)	rs80338950
GJB2	c.617A>G (p.Asn206Ser)	rs111033294
GLA	c.277G>A (p.Asp93Asn)	
GLA	c.281G>A (p.Cys94Tyr)	rs113173389

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
GLA	c.404C>T (p.Ala134Val)	
GLA	c.427G>A (p.Ala143Thr)	rs104894845
GLA	c.463G>C (p.Asp155His)	rs113419388
GLA	c.511G>C (p.Gly171Arg)	
GLA	c.644A>G (p.Asn215Ser)	rs28935197
GLA	c.717_718delAA	
GLA	c.838C>A (p.Gln280Lys)	
GLA	c.865A>T (p.Ile289Phe)	rs140329381
GLA	c.901C>T (p.Arg301Ter)	rs398123224
GLA	c.988C>T (p.Gln330Ter)	
GLA	c.1019_1020insA (p.Trp340Ter)	rs398123197
GLA	c.1025G>A (p.Arg342Gln)	rs28935493
GLA	c.1078G>A (p.Gly360Ser)	
GLB1	c.176G>A (p.Arg59His)	rs72555392
GLB1	c.442C>T (p.Arg148Cys)	rs192732174
GLB1	c.601C>T (p.Arg201Cys)	rs72555360
GLB1	c.602G>A (p.Arg201His)	rs189115557
GLB1	c.622C>T (p.Arg208Cys)	rs72555366
GLB1	c.1771T>A (p.Tyr591Asn)	rs72555373
GNPTAB	c.10A>C (p.Lys4Gln)	rs34159654
GNPTAB	c.136C>T (p.Arg46Ter)	rs78347057
GNPTAB	c.1514G>A (p.Cys505Tyr)	rs281864980
GNPTAB	c.1580delC (p.Ser527Ser=fs) [c.1581delC (p.Cys528ValfsTer19)]	rs36007394
GNPTAB	c.2715+1G>A	rs281865031
GNPTAB	c.2864C>T (p.Ala955Val)	rs138390866
GNPTAB	c.3335+6T>G	rs34788341
GNPTAB	c.3503_3504delTC (p.Leu1168GlnfsTer5)	rs34002892
HADHA	c.157C>T (p.Arg53Ter)	rs147103714
HADHA	c.180+1G>A	rs786205088
HADHA	c.180+3A>G	rs781222705
HADHA	c.845T>A (p.Val282Asp)	rs137852773
HADHA	c.871C>T (p.Arg291Ter)	rs137852775
HADHA	c.914T>A (p.Ile305Asn)	rs137852774
HADHA	c.919-2A>G	rs200017313
HADHA	c.1025T>C (p.Leu342Pro)	rs137852772
HADHA	c.1132C>T (p.Gln378Ter)	rs137852770
HADHA	c.1528G>C (p.Glu510Gln)	rs137852769
HBB	c.19G>A(p.Glu7Lys)	rs33930165
HBB	c.20A>T (p.Glu7Val)	rs334
HBB	c.79G>A (p.Glu27Lys)	rs33950507
HBB	c.92+1G>A	rs33971440
HBB	c.93G>T (p.Arg31Ser)	rs1135071
HBB	c.93-21G>A	rs35004220
HBB	c.92+6T>C	rs35724775
HBB	c.118C>T (p.Gln40Ter)	rs11549407

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
HBB	c.315+1G>A	rs33945777
HBB	c.316-2A>G	rs33914668
HBB	c.316-3C>A	rs33913413
HBB	c.364G>T (p.Glu122Ter)	rs33946267
HEXA	c.590A>C (p.Lys197Thr)	rs121907973
HEXA	c.805G>A (p.Gly269Ser)	rs121907954
HEXA	c.1073+1G>A	rs76173977
HEXA	c.1274_1277dupTATC (p.Tyr427IlefsTer5)	rs387906309
HEXA	c.1421+1G>C	rs147324677
HEXA	c.1510C>T (p.Arg504Cys)	rs28942071
HFE	c.845G>A (p.Cys282Tyr)	rs1800562
IDUA	c.192C>A (p.Tyr64Ter)	rs121965022
IDUA	c.208C>T (p.Gln70Ter)	rs121965020
IDUA	c.1205G>A (p.Trp402Ter)	rs121965019
IDUA	c.1861C>T (p.Arg621Ter)	rs121965025
IDUA	c.1962A>T (p.Ter654Cys) [c.1962A>T (p.Ter654Cys)]	rs199794428
IKBKAP	c.1814A>G (p.Tyr605Cys)	rs756919296
IKBKAP	c.2087G>C (p.Arg696Pro)	rs137853022
IKBKAP	c.2204+6T>C	rs111033171
IKBKAP	c.2741C>T (p.Pro914Leu)	rs28939712
IL2RG	c.186T>A (p.Cys62Ter)	rs111033619
IL2RG	c.270-1G>T	rs193922346
IL2RG	c.314A>G (p.Tyr105Cys)	rs193922347
IL2RG	c.341G>A (p.Gly114Asp)	rs111033620
IL2RG	c.343T>C (p.Cys115Arg)	rs111033622
IL2RG	c.355A>T (p.Lys119Ter)	rs137852507
IL2RG	c.452T>C (p.Leu151Pro)	rs137852511
IL2RG	c.455T>C (p.Val152Ala)	rs193922348
IL2RG	c.458T>A (p.Ile153Asn)	rs111033621
IL2RG	c.662T>C (p.Phe221Ser)	rs193922349
IL2RG	c.664C>T (p.Arg222Cys)	rs111033618
IL2RG	c.710G>A (p.Trp237Ter)	rs193922350
IL2RG	c.854G>A (p.Arg285Gln)	rs111033617
IL2RG	c.865C>T (p.Arg289Ter)	rs137852508
IL2RG	c.878T>A (p.Leu293Gln)	rs137852510
IL2RG	c.923C>A (p.Ser308Ter)	rs137852509
MCCC1	c.974T>G (p.Met325Arg)	rs119103212
MCCC1	c.1114C>T (p.Gln372Ter)	rs544349961
MCCC1	c.1155A>C (p.Arg385Ser)	rs119103213
MCCC2	c.295G>C (p.Glu99Gln)	rs119103219
MCCC2	c.568C>T (p.His190Tyr)	rs773774134
MCCC2	c.838G>T (p.Asp280Tyr)	rs119103226
MCCC2	c.929C>G (p.Pro310Arg)	rs119103221
MCCC2	c.1015G>A (p.Val339Met)	rs150591260
MCCC2	c.1309A>G (p.Ile437Val)	rs119103224

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
MEFV	c.442G>C (p.Glu148Gln)	rs3743930
MEFV	c.2040G>C (p.Met680Ile)	rs28940580
MEFV	c.2080A>G (p.Met694Val)	rs61752717
MEFV	c.2082G>A p.(Met694Ile)	rs28940578
MEFV	c.2084A>G (p.Lys695Arg)	rs104895094
MEFV	c.2177T>C (p.Val726Ala)	rs28940579
MEFV	c.2230G>T (p.Ala744Ser)	rs61732874
MTHFR	c.665C>T (p.Ala222Val)	rs1801133
MTHFR	c.1129C>T (p.Arg377Cys)	rs121434296
MTHFR	c.1743G>A (p.Met581Ile)	rs45590836
MTM1	c.70C>T (p.Arg24Ter)	rs398123275
MTM1	c.122A>G (p.Glu41Gly)	rs398123266
MTM1	c.142_143delGA (p.Glu48SerfsTer12)	rs398123270
MTM1	c.469G>A (p.Glu157Lys)	rs132630307
MTM1	c.566A>G (p.Asn189Ser)	rs132630302
MTM1	c.595_599delCCTGC (p.Pro199SerfsTer11)	rs398123273
MTM1	c.605delT [c.605delT (p.Leu202Trpfs)]	rs672601325
MTM1	c.670C>T (p.Arg224Ter)	rs132630306
MTM1	c.688T>C (p.Trp230Arg)	rs398123274
MTM1	c.721C>T (p.Arg241Cys)	rs132630305
MTM1	c.1040T>G (p.Leu347Ter)	rs398123264
MTM1	c.1054-5G>C	rs398123265
MTM1	c.1190A>G (p.Tyr397Cys)	rs132630303
MTM1	c.1261-10A>G	rs397518445
MTM1	c.1306_1310dupCCTAT (p.Phe438LeufsTer28)	rs398123267
MTM1	c.1357_1358delCC (p.Pro453TyrfsTer4)	rs398123268
MTM1	c.1369G>A (p.Glu457Lys)	rs398123269
MTM1	c.1537_1564del28insAACTGGA [c.1537_1564delTTCTATACTAAAGAAATCAATCGAGTTTinsAACTGGA (p.Phe513_Leu522delinsAsnTrpIle)]	rs398123271
MTM1	c.1644+1G>T	rs398123272
MYO7A	c.93C>A (p.Cys31Ter)	rs35689081
MYO7A	c.448C>T (p.Arg150Ter)	rs121965079
MYO7A	c.634C>T (p.Arg212Cys)	rs121965080
MYO7A	c.635G>A (p.Arg212His)	rs28934610
MYO7A	c.652G>A (p.Asp218Asn)	rs201539845
MYO7A	c.700C>T (p.Gln234Ter)	rs41298133
MYO7A	c.731G>C (p.Arg244Pro)	rs121965081
MYO7A	c.999T>G (p.Tyr333Ter)	rs111033285
MYO7A	c.1373A>T (p.Asn458Ile)	rs121965084
MYO7A	c.1797G>A (p.Met599Ile)	rs121965082
MYO7A	c.1884C>A (p.Cys628Ter)	rs121965083
MYO7A	c.1996C>T (p.Arg666Ter)	rs121965085
MYO7A	c.2005C>T (p.Arg669Ter)	rs111033201
MYO7A	c.3476G>T (p.Gly1159Val)	rs199897298

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
MYO7A	c.3719G>A (p.Arg1240Gln)	rs111033178
MYO7A	c.3728_3729insC (p.Pro1244AlafsTer64)	rs397516304
MYO7A	c.3764delA (p.Lys1255ArgfsTer8)	rs111033347
MYO7A	c.5573T>C (p.Leu1858Pro)	rs368657015
NBN	c.5G>A (p.Trp2*)	
NBN	c.37+1G>A	rs574673404
NBN	c.93_94delTG (p.Ala32fs)	rs864622253
NBN	c.188delT (p.Ile63fs)	
NBN	c.511A>G (p.Ile171Val)	rs61754966
NBN	c.643C>T [c.643C>T (p.Arg215Trp)]	rs34767364
NBN	c.657_661delACAAA [c.657_661delACAAA (p.Lys137AsnfsTer16)]	rs587776650
NBN	c.1393A>T (p.Lys465*)	
NPC1	c.337T>C (p.Cys113Arg)	rs120074136
NPC1	c.416dupC (p.Asn140LysfsTer30)	rs483352880
NPC1	c.530G>A (p.Cys177Tyr)	rs80358252
NPC1	c.1030delT (p.Ser344LeufsTer105)	rs483352883
NPC1	c.1133T>C (p.Val378Ala)	rs120074134
NPC1	c.1142G>A (p.Trp381Ter)	rs794727897
NPC1	c.1800delC (p.Ile601PhefsTer13)	rs483352879
NPC1	c.2128C>T (p.Gln710Ter)	rs483352889
NPC1	c.2196dupT (p.Pro733SerfsTer10)	rs398123284
NPC1	c.2230_2231delGT (p.Val744SerfsTer27)	rs483352882
NPC1	c.2302dupG (p.Val768GlyfsTer4)	rs483352881
NPC1	c.2324A>C (p.Gln775Pro)	rs80358253
NPC1	c.2621A>T (p.Asp874Val)	rs372030650
NPC1	c.2665G>A (p.Val889Met)	rs120074130
NPC1	c.2777C>T (p.Ala926Val)	rs730880963
NPC1	c.2783A>C (p.Gln928Pro)	rs28940897
NPC1	c.2795dupA (p.Tyr932Ter)	rs483352884
NPC1	c.2848G>A (p.Val950Met)	rs120074135
NPC1	c.2873G>A (p.Arg958Gln)	rs120074132
NPC1	c.2932C>T (p.Arg978Cys)	rs28942108
NPC1	c.2974G>T (p.Gly992Trp)	rs80358254
NPC1	c.3019C>G (p.Pro1007Ala)	rs80358257
NPC1	c.3107C>T (p.Thr1036Met)	rs28942104
NPC1	c.3160G>A (p.Ala1054Thr)	rs80358258
NPC1	c.3182T>C (p.Ile1061Thr)	rs80358259
NPC1	c.3263A>G (p.Tyr1088Cys)	rs28942106
NPC1	c.3265G>A (p.Glu1089Lys)	rs374526072
NPC1	c.3467A>G (p.Asn1156Ser)	rs28942105
NPC1	c.3639G>C (p.Leu1213Phe)	rs120074131
NPC1	c.3662delT (p.Phe1221Serfs)	rs786200878
NPC2	c.3G>C/A (p.Met1Ile) [c.3G>C/A (p.Met1?)]	rs483352893
NPC2	c.27delG (p.Leu10SerfsTer25)	rs80358267
NPC2	c.58G>T (p.Glu20Ter)	rs80358260

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
NPC2	c.115G>A (p.Val39Met)	rs80358261
NPC2	c.133C>T (p.Gln45Ter)	rs80358262
NPC2	c.141C>A (p.Cys47Ter)	rs80358263
NPC2	c.199T>C (p.Ser67Pro)	rs11694
NPC2	c.295T>C (p.Cys99Arg)	rs80358264
NPC2	c.332delA (p.Asn111IlefsTer5)	rs80358265
NPC2	c.352G>T (p.Glu118Ter)	rs80358266
NPC2	c.436C>T (p.Gln146Ter)	rs104894457
OTC	c.140A>C (p.Asn47Thr)	rs67939655
OTC	c.140A>T (p.Asn47Ile)	rs67939655
OTC	c.274C>T (p.Arg92Ter)	rs67418243
OTC	c.374C>T (p.Thr125Met)	rs72554356
OTC	c.386G>A (p.Arg129His)	rs66656800
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.809A>C (p.Gln270Pro)	rs1800328
OTC	c.829C>T (p.Arg277Trp)	rs72558454
PAH	c.117C>G (p.Phe39Leu)	rs62642926
PAH	c.143T>C (p.Leu48Ser)	rs5030841
PAH	c.194T>C (p.Ile65Thr)	rs75193786
PAH	c.283A>T (p.Ile95Phe)	rs62508682
PAH	c.284_286delTCA (p.Ile95_Lys96delins) [c.284_286delTCA (p.Ile95del)]	rs62508727
PAH	c.311C>A (p.Ala104Asp)	rs62508727
PAH	c.441+5G>T	rs62507321
PAH	c.473G>A (p.Arg158Gln)	rs5030843
PAH	c.472C>T (p.Arg158Trp)	rs75166491
PAH	c.506G>A (p.Arg169His)	rs199475679
PAH	c.527G>T (p.Arg176Leu)	rs74486803
PAH	c.611A>G (p.Tyr204Cys)	rs62514927
PAH	c.722G>A (p.Arg241His)	rs62508730
PAH	c.727C>T (p.Arg243Ter)	rs5030846
PAH	c.734T>C (p.Val245Ala)	rs76212747
PAH	c.754C>T (p.Arg252Trp)	rs5030847
PAH	c.782G>A (p.Arg261Gln)	rs5030849
PAH	c.814G>T (p.Gly272Ter)	rs62514952
PAH	c.838G>A (p.Glu280Lys)	rs62508698
PAH	c.842C>T (p.Pro281Leu)	rs5030851
PAH	c.896T>G (p.Phe299Cys)	rs62642933
PAH	c.898G>T (p.Ala300Ser)	rs5030853
PAH	c.912+1G>A	rs62514956
PAH	c.926C>T (p.Ala309Val)	rs62642935
PAH	c.960G>C (p.Lys320Asn)	rs199475615

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PAH	c.969+1G>A	rs62508584
PAH	c.1042C>G (p.Leu348Val)	rs62516092
PAH	c.1045T>C(p.Ser349Pro)	rs62508646
PAH	c.1066-11G>A	rs5030855
PAH	c.1139C>T (p.Thr380Met)	rs62642937
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
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
PCDH15	c.3983+1G>T	rs758921360
PEX1	c.2097dupT (p.Ile700TyrfsTer42)	rs61750415
PEX1	c.2230C>T (p.Gln744Ter)	rs398123409
PEX1	c.2528G>A (p.Gly843Asp)	rs61750420
PEX1	c.2916delA (p.Gly973AlafsTer16)	rs61750426
PEX1	c.3379dupC (p.Arg1127ProfsTer19)	rs794729652
PEX2	c.355C>T (p.Arg119Ter)	rs61752123
PEX2	c.669G>A (p.Trp223Ter)	rs61752127
PEX2	c.865dupA (p.Ser289LysfsTer36)	rs724160029
PEX6	c.689_690dupAG (p.Ser232HisfsTer15)	rs398123305
PEX6	c.1338_1339delTG (p.Ala447CysfsTer17)	rs398123303
PEX6	c.1601T>C (p.Leu534Pro)	rs387906809
PEX6	c.2440C>T (p.Arg814Ter)	rs267608241
PEX7	c.12_18dupGTGCGGT (p.Val4_Gly6) [c.13_19dupTGCGGTG (p.Gly7ValfsTer51)]	rs62636519
PEX7	c.40A>C (p.Thr14Pro)	rs61753233
PEX7	c.45_52dupGGGACGCC [c.45_52dupGGGACGCC (p.His18ArgfsTer35)]	rs63535662
PEX7	c.120C>G (p.Tyr40Ter)	rs61753238
PEX7	c.340-10A>G	rs267608255
PEX7	c.345T>G (p.Tyr115Ter)	rs121909154
PEX7	c.649G>A (p.Gly217Arg)	rs121909152
PEX7	c.653C>T (p.Ala218Val)	rs121909151
PEX7	c.694C>T (p.Arg232Ter)	rs121909153
PEX7	c.854A>G (p.His285Arg)	rs62653611
PEX7	c.875T>A (p.Leu292Ter)	rs1805137
PEX7	c.903+1G>C	rs148591292
PEX7	c.-45C>T	rs267608252
PEX10	c.1A>G (p.Met1Val) [c. 1A>G (p.Met1?)]	rs886041314
PEX10	c.2T>C (p.Met1Thr) [c.2T>C (p.Met1?)]	rs724160002
PEX10	c.337delC (p.Leu113TrpfsTer40)	rs724159999
PEX10	c.373C>T (p.Arg125Ter)	rs61750434

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
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
PEX12	c.126+1G>T	rs144259891
PEX12	c.538C>T (p.Arg180Ter)	rs61752103
PEX12	c.691A>T (p.Lys231Ter)	rs104894616
PEX12	c.888_889delCT (p.Leu297ThrfsTer12)	rs398123301
PEX13	c.702G>A (p.Trp234Ter)	rs104893661
PEX14	c.553C>T (p.Gln185Ter)	rs61752116
PEX16	c.526C>T (p.Arg176Ter)	rs61752117
PMM2	c.338C>T (p.Pro113Leu)	rs80338700
PMM2	c.422G>A (p.Arg141His)	rs28936415
PMM2	c.647A>T (p.Asn216Ile)	rs78290141
PMM2	c.691G>A (p.Val231Met)	rs80338707
PMM2	c.710C>G (p.Thr237Arg)	rs80338708
SERPINA1	c.1096G>A(p.Glu366Lys)	rs28929474
SGSH	c.197C>G (p.Ser66Trp)	rs104894637
SGSH	c.220C>T (p.Arg74Cys)	rs104894636
SGSH	c.383C>T (p.Pro128Leu)	rs104894642
SGSH	c.449G>A (p.Arg150Gln)	rs104894638
SGSH	c.617G>C (p.Arg206Pro)	rs104894643
SGSH	c.734G>A (p.Arg245His)	rs104894635
SGSH	c.892T>C (p.Ser298Pro)	rs138504221
SLC26A4	c.2T>C (p.Met1Thr)	rs111033302
SLC26A4	c.68C>A (p.Ser23Ter)	rs397516430
SLC26A4	c.85G>C (p.Glu29Gln)	rs111033205
SLC26A4	c.164+2T>C	rs397516420
SLC26A4	c.349C>T (p.Leu117Phe)	rs145254330
SLC26A4	c.397T>A (p.Ser133Thr)	rs121908365
SLC26A4	c.412G>T (p.Val138Phe)	rs111033199
SLC26A4	c.554G>C (.p.Arg185Thr)	rs542620119
SLC26A4	c.578C>T(p.Thr193Ile)	rs111033348
SLC26A4	c.626G>T (p.Gly209Val)	rs111033303
SLC26A4	c.707T>C (p.Leu236Pro)	rs80338848
SLC26A4	c.997_998insA (p.Arg333Lysfs)	rs431905486
SLC26A4	c.1001+1G>A	rs80338849
SLC26A4	c.1105A>G (p.Lys369Glu)	rs121908361
SLC26A4	c.1115C>T (p.Ala372Val)	rs121908364
SLC26A4	c.1149+3A>G	rs111033314
SLC26A4	c.1151A>G (p.Glu384Gly)	rs111033244
SLC26A4	c.1198delT (p.Cys400ValfsTer7)	rs397516413
SLC26A4	c.1226G>A (p.Arg409His)	rs111033305
SLC26A4	c.1229C>T (p.Thr410Met)	rs111033220
SLC26A4	c.1246A>C (p.Thr416Pro)	rs28939086
SLC26A4	c.1264-1G>C	rs111033311

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
SLC26A4	c.1334T>G (p.Leu445Trp)	rs111033307
SLC26A4	c.1336C>T (p.Gln446Ter)	rs397516416
SLC26A4	c.1468A>C (p.Ile490Leu)	rs200511789
SLC26A4	c.1540C>A (p.Gln514Lys)	rs121908366
SLC26A4	c.1554G>A (pTrp518Ter)	rs727503428
SLC26A4	c.1588T>C (p.Tyr530His)	rs111033254
SLC26A4	c.1614+1G>A	rs111033312
SLC26A4	c.1963A>G (p.Ile655Val)	rs397516424
SLC26A4	c.2000T>G (p.Phe667Cys)	rs121908360
SLC26A4	c.2015G>A (p.Gly672Glu)	rs111033309
SLC26A4	c.2027T>A (p.Leu676Gln)	rs111033318
SLC26A4	c.2162C>T (p.Thr721Met)	rs121908363
SLC26A4	c.2168A>G (p.His723Arg)	rs121908362
SLC26A4	c.2188C>T (p.Gln730Ter)	rs397516428
SLC26A4	c.-3-2A>G	rs397516411
SMN1	EX8DEL	
SMPD1	c.354delC (p.Ile119SerfsTer7)	rs727504165
SMPD1	c.730G>A (p.Gly244Arg)	rs120074122
SMPD1	c.788T>A (p.Leu263Ter)	rs120074120
SMPD1	c.880C>A (p.Gln294Lys)	rs120074128
SMPD1	c.911T>C (p.Leu304Pro)	rs120074124
SMPD1	c.996delC (p.Phe333SerfsTer52)	rs387906289
SMPD1	c.1267C>T (p.His423Tyr)	rs120074126
SMPD1	c.1327C>T (p.Arg443Ter)	rs120074127
SMPD1	c.1406A>C (p.Tyr469Ser)	rs267607074
SMPD1	c.1420_1421delCT (p.Leu474GlufsTer20)	rs398123476
SMPD1	c.1426C>T (p.Arg476Trp)	rs182812968
SMPD1	c.1493G>T (p.Arg498Leu)	rs120074117
SMPD1	c.1624C>T (p.Arg542Ter)	rs398123478
TGM1	c.281G>A (p.Gly94Asp)	rs121918729
TGM1	c.866A>C (p.Asn289Thr)	rs121918730
TGM1	c.877-2A>G	rs142634031
TGM1	c.919C>G (p.Arg307Gly)	rs121918731
TGM1	c.919C>T (p.Arg307Trp)	rs121918731
TGM1	c.968G>A (p.Arg323Gln)	rs121918717
TGM1	c.1075G>A (p.Val359Met)	rs202037016
TGM1	c.1135G>C (p.Val379Leu)	rs121918720
TGM1	c.1166G>A (p.Arg389His)	rs121918723
TGM1	c.1187G>T (p.Arg396Leu)	rs121918721
TPP1	c.509-1G>C	rs56144125
TPP1	c.622C>T (p.Arg208Ter)	rs119455955
TPP1	c.1094G>A (p.Cys365Tyr)	rs119455954
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

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
USH1C	c.238dup (p.Arg80ProfsTer69)	rs397515359
USH1C	c.308G>A (p.Arg103His)	rs397514500
USH1C	c.388G>A (p.Val130Ile)	rs55843567
USH1C	c.496+1G>A/T	
USH1C	c.1220delG	
USH2A	c.632G>A (p.Trp211Ter)	rs727504893
USH2A	c.779T>G (p.Leu260Ter)	rs121912598
USH2A	c.820C>T (p.Arg274Ter)	rs397518036
USH2A	c.1143+1G>A	rs397517974
USH2A	c.1214delA (p.Asn405IlefsTer3)	rs750228923
USH2A	c.1256G>T (p.Cys419Phe)	rs121912600
USH2A	c.1606T>C (p.Cys536Arg)	rs111033273
USH2A	c.2209C>T (p.Arg737Ter)	rs111033334
USH2A	c.2299delG (p.Glu767SerfsTer21)	rs80338903
USH2A	c.2541C>A (p.Cys847Ter)	rs727503736
USH2A	c.2610C>A (p.Cys870Ter)	rs767078782
USH2A	c.2802T>G (p.Cys934Trp)	rs201527662
USH2A	c.2983C>T (p.Gln995Ter)	rs527236135
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.5877delT (p.Ser1961GlnfsTer6)	rs727505343
USH2A	c.6224G>A (p.Trp2075Ter)	rs111033386
USH2A	c.6398G>A (p.Trp2133Ter)	rs727503725
USH2A	c.6862G>T (p.Glu2288Ter)	rs398124619
USH2A	c.7244C>G (p.Ser2415Ter)	rs397518029
USH2A	c.8559-2A>G	rs397518039
USH2A	c.8981G>A (p.Trp2994Ter)	rs397518041
USH2A	c.9159T>G (p.Tyr3053Ter)	rs397518042
USH2A	c.9191del (p.Thr3064MetfsTer3)	rs1553268435
USH2A	c.9304C>T (p.Gln3102Ter)	rs397518046
USH2A	c.9424G>T (p.Gly3142Ter)	rs397518048
USH2A	c.9459C>A (p.Cys3153Ter)	rs73090721
USH2A	c.10073G>A (p.Cys3358Tyr)	rs148660051
USH2A	c.10561T>C (p.Trp3521Arg)	rs111033264
USH2A	c.11048-1G>A	rs111033414
USH2A	c.11864G>A (p.Trp3955Ter)	rs111033364
USH2A	c.11954G>A (p.Trp3985Ter)	rs397517976
USH2A	c.12714T>G (p.Tyr4238Ter)	rs397517981
USH2A	c.12868C>T (p.Gln4290Ter)	rs397517983
USH2A	c.13316C>T (p.Thr4439Ile)	rs753330544
USH2A	c.13374delA (p.Glu4458AspfsTer3)	rs727503715
USH2A	c.14020A>G (p.Arg4674Gly)	rs80338904

GENE	HGVSc (HGVSp) [Ensembl nomenclature]	dbSNP
USH2A	c.14180G>A (p.Trp4727Ter)	rs397517989
USH2A	c.14248C>T (p.Gln4750Ter)	rs727504867
USH2A	c.14803C>T (p.Arg4935Ter)	rs146733615
USH2A	c.14911C>T (p.Arg4971Ter)	rs397517994