

GEN	HGVSc	HGVSp	dbSNP
ACADM	NM_000016.4:c.799G>A	NP_000007.1:p.Gly267Arg	rs121434274
ACADM	NM_000016.5:c.447G>A	NP_000007.1:p.Met149Ile	rs121434277
ACADM	NM_000016.4:c.583G>A	NP_000007.1:p.Gly195Arg	rs121434278
ACADM	NM_000016.4:c.199T>C	NP_000007.1:p.Tyr67His	rs121434280
ACADM	NM_000016.5:c.734C>T	NP_000007.1:p.Ser245Leu	rs121434281
ACADM	NM_000016.5:c.362C>T	NP_000007.1:p.Thr121Ile	rs121434283
ACADM	NM_000016.4:c.999_1011dup	NP_000007.1:p.Gln338Ter	rs1225471006
ACADM	NM_000016.4:c.468+2T>G	-	rs1647214301
ACADM	NM_000016.5:c.617G>A	NP_000007.1:p.Arg206His	rs200724875
ACADM	NM_000016.4:c.797A>G	NP_000007.1:p.Asp266Gly	rs201375579
ACADM	NM_000016.4:c.600-18G>A		rs370523609
ACADM	NM_000016.5:c.616C>T	NP_000007.1:p.Arg206Cys	rs373715782
ACADM	NM_000016.5:c.599+3A>G	NP_000007.1:c.599+3A>G	rs375921211
ACADM	NM_000016.4:c.1102_1105delTTA	NP_000007.1:p.Ala369Leufs	rs387906297
ACADM	NM_000016.5:c.233T>C	NP_000007.1:p.Ile78Thr	rs398123074
ACADM	NM_000016.5:c.250C>T	NP_000007.1:p.Leu84Phe	rs762114560
ACADM	NM_000016.5:c.698T>C	NP_000007.1:p.Ile233Thr	rs770273135
ACADM	NM_000016.5:c.443G>A	NP_000007.1:p.Arg148Lys	rs778906552
ACADM	NM_000016.4:c.985A>G	NP_000007.1:p.Lys329Glu	rs77931234
ACADM	NM_000016.5:c.387+1del		rs786204424
ACADM	NM_000016.4:c.244dup	NP_000007.1:p.Trp82LeufsTer23	rs786204566
ACADM	NM_000016.5:c.347G>A	NP_000007.1:p.Cys116Tyr	rs875989859
ACADM	NM_000016.4:c.948+1G>A		rs878853106
ACADS	NM_000017.3:c.1164_1165del	NP_000008.1:p.Glu389AspfsTer101	rs1057516421
ACADS	NM_000017.3:c.136C>T	NP_000008.1:p.Arg46Trp	rs121908003
ACADS	NM_000017.3:c.973C>T	NP_000008.1:p.Arg325Trp	rs121908006
ACADS	NM_000017.3:c.c.988C>T	NP_000008.1:p.Arg330Cys	rs140853839
ACADS	NM_000017.2:c.164C>T	NP_000008.1:p.Pro55Leu	rs147442301
ACADS	NM_000017.2:c.1147C>T	NP_000008.1:p.Arg383Cys	rs28940872
ACADS	NM_000017.3:c.1138C>T	NP_000008.1:p.Arg380Trp	rs28940875
ACADS	NM_000017.2:c.1058C>T	NP_000008.1:p.Ser353Leu	rs28941773
ACADS	NM_000017.3:c.310_312delGAG	NP_000008.1:p.Glu104del	rs387906308
ACADS	NM_000017.2:c.529T>C	NP_000008.1:p.Trp177Arg	rs57443665
ACADS	NM_000017.2:c.319C>T	NP_000008.1:p.Arg107Cys	rs61732144
ACADS	NM_000017.3:c.682_683delGA	NP_000008.1:p.Glu228Argfs	rs786204691
ACADVL	NM_000018.3:c.497_498del	NP_000009.1:p.Ile166SerfsTer5	rs1057516369
ACADVL	NM_000018.3:c.1606-1G>A		rs1057517386
ACADVL	NM_000018.3:c.1182+1G>A		rs113690956
ACADVL	NM_000018.3:c.848T>C	NP_000009.1:p.Val283Ala	rs113994167
ACADVL	NM_000018.3:c.779C>T	NP_000009.1:p.Thr260Met	rs113994168
ACADVL	NM_000018.3:c.1405C>T	NP_000009.1:p.Arg469Trp	rs113994170
ACADVL	NM_000018.3:c.1679-6G>A		rs113994171
ACADVL	NM_000018.3:c.1837C>T	NP_000009.1:p.Arg613Trp	rs118204014
ACADVL	NM_000018.3:c.1349G>A	NP_000009.1:p.Arg450His	rs118204016
ACADVL	NM_000018.3:c.1372T>C	NP_000009.1:p.Phe458Leu	rs118204017
ACADVL	NM_000018.3:c.1322G>A	NP_000009.1:p.Gly441Asp	rs2309689
ACADVL	NM_000018.3:c.194C>T	NP_000009.1:p.Pro65Leu	rs28934585
ACADVL	NM_000018.3:c.520G>A	NP_000009.1:p.Val174Met	rs369560930
ACADVL	NM_000018.3:c.343delG	NP_000009.1:p.Glu115Lysfs	rs387906249
ACADVL	NM_000018.3:c.388_390delGAG	NP_000009.1:p.Glu130del	rs387906251
ACADVL	NM_000018.3:c.1406G>A	NP_000009.1:p.Arg469Gln	rs398123083
ACADVL	NM_000018.3:c.538G>A	NP_000009.1:p.Ala180Thr	rs727503791

GEN	HGVSc	HGVSp	dbSNP
ACADVL	NM_000018.3:c.298_299delCA	NP_000009.1:p.Gln100Valfs	rs786204713
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ADGRV1	NM_032119.3:c.6901C>T	NP_115495.3:p.Gln2301Ter	rs121909762
ADGRV1	NM_032119.3:c.18131A>G	NP_115495.3:p.Tyr6044Cys	rs121909763
ADGRV1	NM_032119.3:c.14973-2A>G		rs371981035
ADGRV1	NM_032119.3:c.2398C>T	NP_115495.3:p.Arg800Ter	rs373780305
ADGRV1	NM_032119.3:c.11253C>G	NP_115495.3:p.Tyr3751Ter	rs376689763
ADGRV1	NM_032119.3:c.17662delT	NP_115495.3:p.Ser5888Hisfs	rs397517426
ADGRV1	NM_032119.3:c.2870dupA	NP_115495.3:p.Asn957Lysfs	rs397517429
ADGRV1	NM_032119.3:c.7374_7375delTG	NP_115495.3:p.Glu2459Glyfs	rs397517435
ADGRV1	NM_032119.3:c.7406G>A	NP_115495.3:p.Trp2469Ter	rs397517436
ADGRV1	NM_032119.3:c.8737delG	NP_115495.3:p.Val2913Tyrfs	rs397517441
ADGRV1	NM_032119.3:c.5643delG	NP_115495.3:p.Tyr1882Ilefs	rs727503076
ADGRV1	NM_032119.3:c.12631C>T	NP_115495.3:p.Arg4211Ter	rs727504777
ADGRV1	NM_032119.3:c.4702delA	NP_115495.3:p.Ser1568Valfs	rs794727347
ADGRV1	NM_032119.3:c.8204delA	NP_115495.3:p.Asn2735Metfs	rs794727584
ADGRV1	NM_032119.3:c.8790delC	NP_115495.3:p.Met2931Trpfs	rs796051864
AGL	NM_000642.2:c.16C>T	NP_000633.2:p.Gln6Ter	rs113994126
AGL	NM_000642.2:c.17_18delAG	NP_000633.2:p.Gln6Hisfs	rs113994127
AGL	NM_000642.2:c.1222C>T	NP_000633.2:p.Arg408Ter	rs113994128
AGL	NM_000642.2:c.2039G>A	NP_000633.2:p.Trp680Ter	rs113994129
AGL	NM_000642.2:c.2590C>T	NP_000633.2:p.Arg864Ter	rs113994130
AGL	NM_000642.2:c.3682C>T	NP_000633.2:p.Arg1228Ter	rs113994131
AGL	NM_000642.2:c.3965delT	NP_000633.2:p.Val1322Alafs	rs113994132
AGL	NM_000642.2:c.4456delT	NP_000633.2:p.Ser1486Profs	rs113994134
AGL	NM_000642.2:c.500dup	NP_000633.2:p.Leu168ThrfsTer3	rs1443902661
AGL	NM_000642.2:c.1735+1G>T		rs199922945
AGL	NM_000642.2:c.3980G>A	NP_000633.2:p.Trp1327Ter	rs267606640
AGL	NM_000642.2:c.4260-12A>G		rs369973784
AGL	NM_000642.2:c.4529dupA	NP_000633.2:p.Tyr1510Terfs	rs387906244
AGL	NM_000642.2:c.3911del	NP_000633.2:p.Asn1304IlefsTer10	rs745757264
AGL	NM_000642.2:c.753_756del	NP_000633.2:p.Asp251GluTer23	rs748789700
AGL	NM_000642.2:c.1571G>A	NP_000633.2:p.Arg524His	rs758182700
ALPL	NM_000478.4:c.535G>A	NP_000469.3:p.Ala179Thr	rs121918000
ALPL	NM_000478.4:c.881A>C	NP_000469.3:p.Asp294Ala	rs121918002
ALPL	NM_000478.4:c.571G>A	NP_000469.3:p.Glu191Lys	rs121918007
ALPL	NM_000478.4:c.1133A>T	NP_000469.3:p.Asp378Val	rs121918008
ALPL	NM_000478.4:c.979T>C	NP_000469.3:p.Phe327Leu	rs121918010
ALPL	NM_000478.4:c.407G>A	NP_000469.3:p.Arg136His	rs121918011
ALPL	NM_000478.4:c.346G>A	NP_000469.3:p.Ala116Thr	rs121918013
ALPL	NM_000478.4:c.1250A>G	NP_000469.3:p.Asn417Ser	rs121918014
ALPL	NM_000478.4:c.526G>A	NP_000469.3:p.Ala176Thr	rs121918019
ALPL	NM_000478.4:c.668G>A	NP_000469.3:p.Arg223Gln	rs199665722
ALPL	NM_000478.4:c.1559delT	NP_000469.3:p.Leu520Argfs	rs387906525
ALPL	NM_000478.4:c.667C>T	NP_000469.3:p.Arg223Trp	rs766076920
ALPL	NM_000478.4:c.119C>T	NP_000469.3:p.Ala40Val	rs770093969
ALPL	NM_000478.4:c.871G>A	NP_000469.3:p.Glu291Lys	rs786204473
ANXA5	NM_001154.3:c.-229G>A (v rámci M2 haplotypu)		rs112782763
ANXA5	NM_001154.3:c.-135G>A (v rámci M2 haplotypu)		rs113588187
ANXA5	NM_001154.3:c.-184T>C (v rámci M2 haplotypu)		rs28651243
ANXA5	NM_001154.3:c.-210A>C (v rámci M2 haplotypu)		rs28717001
AR	NM_000044.3:c.4G>A	NP_000035.2:p.Glu2Lys	rs104894742

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AR	NM_000044.3:c.2157G>A	NP_000035.2:p.Trp719Ter	rs137852563
AR	NM_000044.3:c.2391G>A	NP_000035.2:p.Trp797Ter	rs137852565
AR	NM_000044.3:c.1771A>T	NP_000035.2:p.Lys591Ter	rs137852566
AR	NM_000044.3:c.2291A>G	NP_000035.2:p.Tyr764Cys	rs137852567
AR	NM_000044.3:c.2650A>T	NP_000035.2:p.Lys884Ter	rs137852568
AR	NM_000044.3:c.2362A>G	NP_000035.2:p.Met788Val	rs137852570
AR	NM_000044.3:c.2191G>A	NP_000035.2:p.Val731Met	rs137852571
AR	NM_000044.3:c.2610T>G	NP_000035.2:p.Ile870Met	rs137852574
AR	NM_000044.3:c.178C>T	NP_000035.2:p.Gln60Ter	rs137852575
AR	NM_000044.3:c.1826G>A	NP_000035.2:p.Arg609Lys	rs137852576
AR	NM_000044.3:c.2623C>T	NP_000035.2:p.His875Tyr	rs137852581
AR	NM_000044.3:c.2708A>G	NP_000035.2:p.Gln903Arg	rs137852582
AR	NM_000044.3:c.1943G>A	NP_000035.2:p.Ser648Asn	rs137852584
AR	NM_000044.3:c.2123T>G	NP_000035.2:p.Leu708Arg	rs137852585
AR	NM_000044.3:c.1739G>T	NP_000035.2:p.Cys580Phe	rs137852586
AR	NM_000044.3:c.1748T>A	NP_000035.2:p.Phe583Tyr	rs137852587
AR	NM_000044.3:c.1645C>T	NP_000035.2:p.Pro549Ser	rs137852588
AR	NM_000044.3:c.2343G>T	NP_000035.2:p.Met781Ile	rs137852589
AR	NM_000044.3:c.521T>G	NP_000035.2:p.Leu174Ter	rs137852590
AR	NM_000044.3:c.2423T>C	NP_000035.2:p.Met808Thr	rs137852592
AR	NM_000044.3:c.2180G>T	NP_000035.2:p.Arg727Leu	rs137852593
AR	NM_000044.3:c.2137C>T	NP_000035.2:p.Leu713Phe	rs137852595
AR	NM_000044.3:c.1732G>A	NP_000035.2:p.Gly578Arg	rs137852596
AR	NM_000044.3:c.2596T>C	NP_000035.2:p.Ser866Pro	rs137852597
AR	NM_000044.3:c.2069A>C	NP_000035.2:p.His690Pro	rs137852599
AR	NM_000044.3:c.2231G>T	NP_000035.2:p.Gly744Val	rs137852600
AR	NM_000044.3:c.2222C>G	NP_000035.2:p.Ser741Cys	rs137852601
AR	NM_000044.3:c.2567G>A	NP_000035.2:p.Arg856His	rs9332971
ARSA	NM_000487.5:c.684+1G>A		rs146371968
ARSA	NM_000487.5:c.979G>A	NP_000478.3:p.Gly327Ser	rs148092995
ARSA	NM_000487.5:c.256C>T	NP_000478.3:p.Arg86Trp	rs199476352
ARSA	NM_000487.5:c.869G>A	NP_000478.3:p.Arg290His	rs199476355
ARSA	NM_000487.5:c.1283C>T	NP_000478.3:p.Pro428Leu	rs28940893
ARSA	NM_000487.5:c.862A>C	NP_000478.3:p.Thr288Pro	rs28940894
ARSA	NM_000487.5:c.302G>A	NP_000478.3:p.Gly101Asp	rs74315455
ARSA	NM_000487.5:c.293C>T	NP_000478.3:p.Ser98Phe	rs74315456
ARSA	NM_000487.5:c.542T>G	NP_000478.3:p.Ile181Ser	rs74315457
ARSA	NM_000487.5:c.931G>A	NP_000478.3:p.Gly311Ser	rs74315459
ARSA	NM_000487.5:c.641C>T	NP_000478.3:p.Ala214Val	rs74315467
ARSA	NM_000487.5:c.739G>A	NP_000478.3:p.Gly247Arg	rs74315471
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ARSA	NM_000487.5:c.1114C>T	NP_000478.3:p.Arg372Trp	rs74315476
ARSA	NM_000487.5:c.1136C>T	NP_000478.3:p.Pro379Leu	rs74315478
ARSA	NM_000487.5:c.1150G>A	NP_000478.3:p.Glu384Lys	rs74315479
ARSA	NM_000487.5:c.1232C>T	NP_000478.3:p.Thr411Ile	rs74315481
ARSA	NM_000487.5:c.418dup	NP_000478.3:p.His140ProfsTer36	rs745884435
ARSA	NM_000487.5:c.418del	NP_000478.3:p.His140IlefsTer8	rs745884435
ARSA	NM_000487.5:c.302del	NP_000478.3:p.Gly101AlafsTer7	rs761606317
ARSA	NM_000487.5:c.302dup	NP_000478.3:p.Leu102ProfsTer32	rs761606317
ARSA	NM_000487.5:c.465+1G>A		rs80338815

GEN	HGVSc	HGVSp	dbSNP
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ARSA	NM_000487.5:c.1210+1G>A		rs80338820
ASL	NM_000048.3:c.446+1G>A		rs142637046
ASL	NM_000048.3:c.35G>A	NP_000039.2:p.Arg12Gln	rs145138923
ASL	NM_000048.3:c.283C>T	NP_000039.2:p.Arg95Cys	rs28940585
ASL	NM_000048.3:c.857A>G	NP_000039.2:p.Gln286Arg	rs28941472
ASL	NM_000048.3:c.532G>A	NP_000039.2:p.Val178Met	rs28941473
ASL	NM_000048.3:c.649C>T	NP_000039.2:p.Arg217Ter	rs369879957
ASL	NM_000048.3:c.578G>A	NP_000039.2:p.Arg193Gln	rs373697663
ASL	NM_000048.3:c.566A>G	NP_000039.2:p.Glu189Gly	rs756363516
ASL	NM_000048.3:c.467C>T	NP_000039.2:p.Pro156Leu	rs769017508
ASL	NM_000048.3:c.175G>A	NP_000039.2:p.Glu59Lys	rs869312985
ASL	NM_000048.3:c.762C>A	NP_000039.2:p.Ser254Arg	rs869312991
ASPA	NM_000049.2:c.454T>C	NP_000040.1:p.Cys152Arg	rs104894548
ASPA	NM_000049.2:c.654C>A	NP_000040.1:p.Cys218=	rs104894549
ASPA	NM_000049.2:c.71A>G	NP_000040.1:p.Glu24Gly	rs104894551
ASPA	NM_000049.2:c.746A>T	NP_000040.1:p.Asp249Val	rs104894552
ASPA	NM_000049.2:c.212G>A	NP_000040.1:p.Arg71His	rs104894553
ASPA	NM_000049.2:c.693C>A	NP_000040.1:p.Tyr231Ter	rs12948217
ASPA	NM_000049.2:c.854A>C	NP_000040.1:p.Glu285Ala	rs28940279
ASPA	NM_000049.2:c.914C>A	NP_000040.1:p.Ala305Glu	rs28940574
ASPA	NM_000049.2:c.433-2A>G		rs63751297
ASPA	NM_000049.2:c.79G>A	NP_000040.1:p.Gly27Arg	rs766328537
ASPA	NM_000049.2:c.503G>A	NP_000040.1:p.Arg168His	rs770706390
ASS1	NM_000050.4:c.470G>A	NP_000041.2:p.Arg157His	rs121908637
ASS1	NM_000050.4:c.1168G>A	NP_000041.2:p.Gly390Arg	rs121908641
ASS1	NM_000050.4:c.256C>T	NP_000041.2:p.Arg86Cys	rs121908644
ASS1	NM_000050.4:c.835C>T	NP_000041.2:p.Arg279Ter	rs121908645
ASS1	NM_000050.4:c.535T>C	NP_000041.2:p.Trp179Arg	rs121908646
ASS1	NM_000050.4:c.793C>T	NP_000041.2:p.Arg265Cys	rs148918985
ASS1	NM_000050.4:c.787G>A	NP_000041.2:p.Val263Met	rs192838388
ASS1	NM_000050.4:c.805G>A	NP_000041.2:p.Val269Met	rs370595480
ASS1	NM_000050.4:c.836G>A	NP_000041.2:p.Arg279Gln	rs371265106
ASS1	NM_000050.4: c.970+5G-A		rs372128852
ASS1	NM_000050.4:c.814C>T	NP_000041.2:p.Arg272Cys	rs762387914
ASS1	NM_000050.4:c.469C>T	NP_000041.2:p.Arg157Cys	rs770585183
ASS1	NM_000050.4:c.349G>A	NP_000041.2:p.Gly117Ser	rs770944877
ASS1	NM_000050.4:c.352G>A	NP_000041.2:p.Ala118Thr	rs775305020
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ATM	NM_000051.3:c.6096-13_6096-9delTCTTT		c.6096-13_6096-
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ATM	NM_000051.3:c.5044G>C	NP_000042.3:p.Asp1682His	rs121434217
ATM	NM_000051.3:c.7327C>T	NP_000042.3:p.Arg2443Ter	rs121434220
ATM	NM_000051.3:c.5309C>G	NP_000042.3:p.Ser1770Ter	rs121434223
ATM	NM_000051.3:c.6095G>A	NP_000042.3:p.Arg2032Lys	rs139770721
ATM	NM_000051.3:c.1235+3A>G		rs1417190414
ATM	NM_000051.3:c.3304G>T	NP_000042.3:p.Gly1102Ter	rs147557621
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ATM	NM_000051.3:c.3993+1G>A		rs200196781
ATM	NM_000051.3:c.3931C>T	NP_000042.3:p.Gln1311Ter	rs200976093
ATM	NM_000051.3:c.7875_7876delTGinsGC	NP_000042.3:p.AspAla2625GluPro	rs267606668
ATM	NM_000051.3:c.7271T>G	NP_000042.3:p.Val2424Gly	rs28904921

GEN	HGVSc	HGVSp	dbSNP
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ATM	NM_000051.3:c.1110C>G	NP_000042.3:p.Tyr370Ter	rs376170600
ATM	NM_000051.3:c.6100C>T	NP_000042.3:p.Arg2034Ter	rs532480170
ATM	NM_000051.3:c.802C>T	NP_000042.3:p.Gln268Ter	rs557012154
ATM	NM_000051.3:c.103C>T	NP_000042.3:p.Arg35Ter	rs55861249
ATM	NM_000051.3:c.6679C>T	NP_000042.3:p.Arg2227Cys	rs564652222
ATM	NM_000051.3:c.3576G>A	NP_000042.3:p.Lys1192=	rs587776551
ATM	NM_000051.3:c.1235G>A	NP_000042.3:p.Trp412Ter	rs587779813
ATM	NM_000051.3:c.1564_1565delGA	NP_000042.3:p.Glu522IlefsTer	rs587779817
ATM	NM_000051.3:c.170G>A	NP_000042.3:p.Trp57Ter	rs587779818
ATM	NM_000051.3:c.2502dupA	NP_000042.3:p.Val835Serfs	rs587779822
ATM	NM_000051.3:c.3372C>G	NP_000042.3:p.Tyr1124Ter	rs587779833
ATM	NM_000051.3:c.3802delG	NP_000042.3:p.Val1268Terfs	rs587779834
ATM	NM_000051.3:c.3836G>A	NP_000042.3:p.Trp1279Ter	rs587779836
ATM	NM_000051.3:c.5932G>T	NP_000042.3:p.Glu1978Ter	rs587779852
ATM	NM_000051.3:c.7456C>T	NP_000042.3:p.Arg2486Ter	rs587779865
ATM	NM_000051.3:c.7630-2A>C		rs587779866
ATM	NM_000051.3:c.8494C>T	NP_000042.3:p.Arg2832Cys	rs587779872
ATM	NM_000051.3:c.283C>T	NP_000042.3:p.Gln95Ter	rs587781545
ATM	NM_000051.3:c.2921+1G>A		rs587781558
ATM	NM_000051.3:c.7096G>T	NP_000042.3:p.Glu2366Ter	rs587781672
ATM	NM_000051.3:c.5908C>T	NP_000042.3:p.Gln1970Ter	rs587781722
ATM	NM_000051.3:c.381delA	NP_000042.3:p.Val128Terfs	rs587781831
ATM	NM_000051.3:c.3388G>T	NP_000042.3:p.Gly1130Ter	rs587781911
ATM	NM_000051.3:c.4198A>T	NP_000042.3:p.Lys1400Ter	rs587781950
ATM	NM_000051.3:c.8185C>T	NP_000042.3:p.Gln2729*	rs587781967
ATM	NM_000051.3:c.1158delG	NP_000042.3:p.Lys387Argfs	rs587782085
ATM	NM_000051.3:c.3077G>A	NP_000042.3:p.Trp1026Ter	rs587782103
ATM	NM_000051.3:c.6326G>A	NP_000042.3:p.Trp2109Ter	rs587782114
ATM	NM_000051.3:c.1898+2T>G		rs587782124
ATM	NM_000051.3:c.3980T>G	NP_000042.3:p.Leu1327Ter	rs587782192
ATM	NM_000051.3:c.3994-2A>G		rs587782276
ATM	NM_000051.3:c.2548G>T	NP_000042.3:p.Glu850Ter	rs587782280
ATM	NM_000051.3:c.8147T>C	NP_000042.3:p.Val2716Ala	rs587782652
ATM	NM_000051.3:c.2880delC	NP_000042.3:p.Leu961Cysfs	rs730881300
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ATM	NM_000051.3:c.2251-10T>G		rs730881346
ATM	NM_000051.3:c.2376+1G>T		rs730881347
ATM	NM_000051.3:c.2426C>A	NP_000042.3:p.Ser809Ter	rs730881348
ATM	NM_000051.3:c.1A>G	NP_000042.3:p.Met1Val	rs730881359
ATM	NM_000051.3:c.154G>T	NP_000042.3:p.Gly52Ter	rs730881362
ATM	NM_000051.3:c.3049C>T	NP_000042.3:p.Gln1017Ter	rs730881388
ATM	NM_000051.3:c.5177+5G>A		rs759373136
ATM	NM_000051.3:c.4852C>T	NP_000042.3:p.Arg1618Ter	rs762083530
ATM	NM_000051.3:c.5188C>T	NP_000042.3:p.Arg1730Ter	rs764389018
ATM	NM_000051.3:c.5763-1050A>G		rs774925473
ATM	NM_000051.3:c.1737G>A	NP_000042.3:p.Trp579Ter	rs786201689
ATM	NM_000051.3:c.513C>G	NP_000042.3:p.Tyr171Ter	rs786201693
ATM	NM_000051.3:c.3349C>T	NP_000042.3:p.Gln1117Ter	rs786201957
ATM	NM_000051.3:c.4052delT	NP_000042.3:p.Leu1351Tyrfs	rs786202350
ATM	NM_000051.3:c.1880dupT	NP_000042.3:p.Gln628Profs	rs786202474
ATM	NM_000051.3:c.2754delT	NP_000042.3:p.Phe918Leufs	rs786202608

GEN	HGVSc	HGVSp	dbSNP
ATM	NM_000051.3:c.2098C>T	NP_000042.3:p.Gln700Ter	rs786202743
ATM	NM_000051.3:c.2789T>G	NP_000042.3:p.Leu930Ter	rs786203309
ATM	NM_000051.3:c.1424C>G	NP_000042.3:p.Ser475Ter	rs786203550
ATM	NM_000051.3:c.2129delC	NP_000042.3:p.Thr710Lysfs	rs786203807
ATM	NM_000051.3:c.151C>T	NP_000042.3:p.Gln51Ter	rs786203888
ATM	NM_000051.3:c.5644C>T	NP_000042.3:p.Arg1882Ter	rs786204433
ATM	NM_000051.3:c.5515C>T	NP_000042.3:p.Gln1839Ter	rs786204751
ATM	NM_000051.3:c.9021dup	NP_000042.3:p.Arg3008ThrfsTer55	rs876660235
ATM	NM_000051.3:c.3850delA	NP_000042.3:p.Thr1284fs	rs876660865
ATM	NM_000051.3:c.5573G>A	NP_000042.3:p.Trp1858Ter	rs879254076
ATM	NM_000051.3:c.6069-9_6069-5delTTCTT		rs879254095
ATM	NM_000051.3:c.601C>T	NP_000042.3:p.Gln201Ter	rs886039666
ATP7B	NM_000053.3:c.1340_1343del	NP_000044.2:p.Gln447LeufsTer50	rs1057517444
ATP7B	NM_000053.3:c.3809A>G	NP_000044.2:p.Asn1270Ser	rs121907990
ATP7B	NM_000053.3:c.3796G>A	NP_000044.2:p.Gly1266Arg	rs121907992
ATP7B	NM_000053.3:c.2755C>T	NP_000044.2:p.Arg919Trp	rs121907993
ATP7B	NM_000053.3:c.2621C>T	NP_000044.2:p.Ala874Val	rs121907994
ATP7B	NM_000053.3:c.2906G>A	NP_000044.2:p.Arg969Gln	rs121907996
ATP7B	NM_000053.3:c.1934T>G	NP_000044.2:p.Met645Arg	rs121907998
ATP7B	NM_000053.3:c.3402del	NP_000044.2:p.Ala1135Glnfs	rs137853281
ATP7B	NM_000053.3:c.2336G>A	NP_000044.2:p.Trp779Ter	rs137853283
ATP7B	NM_000053.3:c.2332C>G	NP_000044.2:p.Arg778Gly	rs137853284
ATP7B	NM_000053.3:c.2128G>A	NP_000044.2:p.Gly710Ser	rs137853285
ATP7B	NM_000053.3:c.2304del	NP_000044.2:p.Met769CysfsTer38	rs137853287
ATP7B	NM_000053.3:c.2304dup	NP_000044.2:p.Met769HisfsTer26	rs137853287
ATP7B	NM_000053.3:c.2605G>A	NP_000044.2:p.Gly869Arg	rs191312027
ATP7B	NM_000053.3:c.2305A>G	NP_000044.2:p.Met769Val	rs193922103
ATP7B	NM_000053.3:c.3955C>T	NP_000044.2:p.Arg1319Ter	rs193922109
ATP7B	NM_000053.3:c.2962G>C	NP_000044.2:p.Gly988Arg	rs199623434
ATP7B	NM_000053.3:c.3688A>G	NP_000044.2:p.Ile1230Val	rs200911496
ATP7B	NM_000053.3:c.3007G>A	NP_000044.2:p.Ala1003Thr	rs201497300
ATP7B	NM_000053.3:c.2333G>T	NP_000044.2:p.Arg778Leu	rs28942074
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ATP7B	NM_000053.3:c.1285+5G>T		rs370579582
ATP7B	NM_000053.3:c.3053C>T	NP_000044.2:p.Ala1018Val	rs371840514
ATP7B	NM_000053.3:c.1969A>C	NP_000044.2:p.Ser657Arg	rs372436901
ATP7B	NM_000053.3:c.2930C>T	NP_000044.2:p.Thr977Met	rs72552255
ATP7B	NM_000053.3:c.3121C>T	NP_000044.2:p.Arg1041Trp	rs746485916
ATP7B	NM_000053.3:c.3207C>A	NP_000044.2:p.His1069Gln	rs76151636
ATP7B	NM_000053.3:c.3295G>A	NP_000044.2:p.Gly1099Ser	rs761632029
ATP7B	NM_000053.3:c.3784G>T	NP_000044.2:p.Val1262Phe	rs769484789
ATP7B	NM_000053.3:c.3649_3654del	NP_000044.2:p.Val1217_Leu1218del	rs781266802
AZFa	sY86		
AZFa	sY84		
AZFb	sY127		
AZFb	sY134		
AZFb/c	sY1191_G73809		
AZFb/c	sY254		
AZFb/c	sY1291_G72340		
AZFc	sY255		

GEN	HGVSc	HGVSp	dbSNP
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BLM	NM_000057.2:c.461G>A	NP_000048.1:p.Trp154Ter	c.461G>A
BLM	NM_000057.2:c.2207_2212delATC	NP_000048.1:p.Tyr736Leufs	rs113993962
BLM	NM_000057.3:c.3558+1G>A		rs148969222
BLM	NM_000057.3:c.3875-2A>G		rs150421256
BLM	NM_000057.2:c.1642C>T	NP_000048.1:p.Gln548Ter	rs200389141
BLM	NM_000057.2:c.1544del	NP_000048.1:p.Asn515MetfsTer16	rs367543043
BLM	NM_000057.2:c.1544dup	NP_000048.1:p.Asn515LysfsTer2	rs367543043
BLM	NM_000057.2:c.2695C>T	NP_000048.1:p.Arg899Ter	rs587779884
BLM	NM_000057.2:c.98+1G>T		rs750293380
BLM	NM_000057.2:c.2206_2207insT	NP_000048.1:p.Tyr736Leufs	rs886051551
BTD	NM_000060.2:c.1207T>G	NP_000051.1:p.Phe403Val	rs104893686
BTD	NM_000060.2:c.235C>T	NP_000051.1:p.Arg79Cys	rs104893687
BTD	NM_000060.2:c.1595C>T	NP_000051.1:p.Thr532Met	rs104893688
BTD	NM_000060.2:c.1466A>C	NP_000051.1:p.Asn489Thr	rs104893692
BTD	NM_000060.2:c.100G>A	NP_000051.1:p.Gly34Ser	rs119103232
BTD	NM_000060.2:c.1334G>T	NP_000051.1:p.Gly445Val	rs1223614917
BTD	NM_000060.2:c.41_44del	NP_000051.1:p.Gly14ValfsTer35	rs1249246307
BTD	NM_000060.2:c.511G>A	NP_000051.1:p.Ala171Thr	rs13073139
BTD	NM_000060.2:c.1330G>C	NP_000051.1:p.Asp444His	rs13078881
BTD	NM_000060.2:c.1489C>T	NP_000051.1:p.Pro497Ser	rs138818907
BTD	NM_000060.2:c.470G>A	NP_000051.1:p.Arg157His	rs146015592
BTD	NM_000060.2:c.1369G>A	NP_000051.1:p.Val457Met	rs146600671
BTD	NM_000060.2:c.262C>T	NP_000051.1:p.Gln88Ter	rs151071780
BTD	NM_000060.2:c.250-1G>T		rs1553653053
BTD	NM_000060.2:c.1432G>C	NP_000051.1:p.Ala478Pro	rs181396238
BTD	NM_000060.2:c.643C>T	NP_000051.1:p.Leu215Phe	rs190386869
BTD	NM_000060.2:c.1046A>C	NP_000051.1:p.Asn349Thr	rs200327983
BTD	NM_000060.2:c.664G>C	NP_000051.1:p.Asp222His	rs200337373
BTD	NM_000060.2:c.1455C>G	NP_000051.1:p.His485Gln	rs201604102
BTD	NM_000060.2:c.755A>G	NP_000051.1:p.Asp252Gly	rs28934601
BTD	NM_000060.2:c.1171C>T	NP_000051.1:p.Pro391Ser	rs35034250
BTD	NM_000060.2:c.1284C>A	NP_000051.1:p.Tyr428Ter	rs35145938
BTD	NM_000060.2:c.820A>G	NP_000051.1:p.Ile274Val	rs35976361
BTD	NM_000060.2:c.1237G>A	NP_000051.1:p.Gly413Ser	rs374141881
BTD	NM_000060.2:c.454A>C	NP_000051.1:p.Thr152Pro	rs374681173
BTD	NM_000060.2:c.341G>T	NP_000051.1:p.Gly114Val	rs375712490
BTD	NM_000060.2:c.935G>A	NP_000051.1:p.Gly312Asp	rs377651057
BTD	NM_000060.2:c.124G>A	NP_000051.1:p.Val42Met	rs397507170
BTD	NM_000060.2:c.194A>G	NP_000051.1:p.His65Arg	rs397514341
BTD	NM_000060.2:c.409C>T	NP_000051.1:p.Arg137Cys	rs397514363
BTD	NM_000060.2:c.528G>T	NP_000051.1:p.Lys176Asn	rs397514367
BTD	NM_000060.2:c.933delT	NP_000051.1:p.Ser311Argfs	rs397514395
BTD	NM_000060.2:c.626G>A	NP_000051.1:p.Arg209His	rs398123139
BTD	NM_000060.2:c.98_104delGCGGCT	NP_000051.1:p.Cys33_Cys35?fs	rs80338684
BTD	NM_000060.2:c.1368A>C	NP_000051.1:p.Gln456His	rs80338685
BTD	NM_000060.2:c.1612C>T	NP_000051.1:p.Arg538Cys	rs80338686
CBS	NM_000071.2:c.954+1G>A		rs1057517373
CBS	NM_000071.2:c.1126G>A	NP_000062.1:p.Asp376Asn	rs1170128038
CBS	NM_000071.2:c.919G>A	NP_000062.1:p.Gly307Ser	rs121964962
CBS	NM_000071.2:c.341C>T	NP_000062.1:p.Ala114Val	rs121964964
CBS	NM_000071.2:c.415G>A	NP_000062.1:p.Gly139Arg	rs121964965

GEN	HGVSc	HGVSp	dbSNP
CBS	NM_000071.2:c.430G>A	NP_000062.1:p.Glu144Lys	rs121964966
CBS	NM_000071.2:c.1616T>C	NP_000062.1:p.Leu539Ser	rs121964968
CBS	NM_000071.2:c.797G>A	NP_000062.1:p.Arg266Lys	rs121964969
CBS	NM_000071.2:c.1058C>T	NP_000062.1:p.Thr353Met	rs121964972
CBS	NM_000071.2:c.572C>T	NP_000062.1:p.Thr191Met	rs121964973
CBS	NM_000071.2:c.146C>T	NP_000062.1:p.Pro49Leu	rs148865119
CBS	NM_000071.2:c.1330G>A	NP_000062.1:p.Asp444Asn	rs28934891
CBS	NM_000071.2:c.1111G>A	NP_000062.1:p.Val371Met	rs372010465
CBS	NM_000071.2:c.1224-2A>C		rs375846341
CBS	NM_000071.2:c.1006C>T	NP_000062.1:p.Arg336Cys	rs398123151
CBS	NM_000071.2:c.816T>A	NP_000062.1:p.Cys272Ter	rs528689432
CBS	NM_000071.2:c.833T>C	NP_000062.1:p.Ile278Thr	rs5742905
CBS	NM_000071.2:c.19dup	NP_000062.1:p.Gln7ProfsTer30	rs748695461
CBS	NM_000071.2:c.19del	NP_000062.1:p.Gln7ProfsTer30	rs748695461
CBS	NM_000071.2:c.828+1G>A		rs763290176
CBS	NM_000071.2:c.1135C>T	NP_000062.1:p.Arg379Trp	rs769080151
CBS	NM_000071.2:c.361C>T	NP_000062.1:p.Arg121Cys	rs775992753
CBS	NM_000071.2:c.374G>A	NP_000062.1:p.Arg125Gln	rs781444670
CBS	NM_000071.2:c.959T>C	NP_000062.1:p.Val320Ala	rs781567152
CDH23	NM_022124.5:c.6049+1G>A		rs111033247
CDH23	NM_022124.5:c.5237G>A	NP_071407.4:p.Arg1746Gln	rs111033270
CDH23	NM_022124.5:c.6442G>A	NP_071407.4:p.Asp2148Asn	rs111033271
CDH23	NM_022124.5:c.4488G>C	NP_071407.4:p.Gln1496His	rs121908347
CDH23	NM_022124.5:c.6133G>A	NP_071407.4:p.Asp2045Asn	rs121908348
CDH23	NM_022124.5:c.6604G>A	NP_071407.4:p.Asp2202Asn	rs121908349
CDH23	NM_022124.5:c.4021G>A	NP_071407.4:p.Asp1341Asn	rs121908351
CDH23	NM_022124.5:c.5663T>C	NP_071407.4:p.Phe1888Ser	rs121908352
CDH23	NM_022124.5:c.719C>T	NP_071407.4:p.Pro240Leu	rs121908354
CDH23	NM_022124.5:c.902G>A	NP_071407.4:p.Arg301Gln	rs121908355
CDH23	NM_022124.5:c.6049G>A	NP_071407.4:p.Gly2017Ser	rs183431253
CDH23	NM_022124.5:c.6050-9G>A		rs367928692
CDH23	NM_022124.5:c.2012delT	NP_071407.4:p.Phe671Serfs	rs397517313
CDH23	NM_022124.5:c.3481C>T	NP_071407.4:p.Arg1161Ter	rs397517323
CDH23	NM_022124.5:c.3628C>T	NP_071407.4:p.Gln1210Ter	rs397517326
CDH23	NM_022124.5:c.3706C>T	NP_071407.4:p.Arg1236Ter	rs397517327
CDH23	NM_022124.5:c.4309C>T	NP_071407.4:p.Arg1437Ter	rs397517329
CDH23	NM_022124.5:c.46delG	NP_071407.4:p.Val16Cysfs	rs397517331
CDH23	NM_022124.5:c.5272C>T	NP_071407.4:p.Gln1758Ter	rs397517337
CDH23	NM_022124.5:c.5712+1G>A		rs397517341
CDH23	NM_022124.5:c.5712G>A	NP_071407.4:p.Thr1904=	rs397517342
CDH23	NM_022124.5:c.5923+1G>A		rs397517346
CDH23	c.6614C>T	NP_071407.4:p.Pro2205Leu	rs397517349
CDH23	NM_022124.5:c.6968delC	NP_071407.4:p.Pro2323Leufs	rs397517350
CDH23	NM_022124.5:c.7776G>A	NP_071407.4:p.Trp2592Ter	rs397517353
CDH23	NM_022124.5:c.8781C>A	NP_071407.4:p.Tyr2927Ter	rs397517362
CDH23	NM_022124.5:c.9629_9632delTCA	NP_071407.4:p.Ile3210_Lys3211d	rs397517367
CDH23	NM_022124.5:c.6085C>T	NP_071407.4:p.Arg2029Trp	rs750880909
CDH23	NM_022124.5:c.193delC	NP_071407.4:p.Leu65Trpfs	rs796051861
CFTR	dele2,3(21 kb)		
CFTR	NM_000492.3:c.4056G>C	NP_000483.3:p.Gln1352His	rs113857788
CFTR	NM_000492.3:c.328G>C	NP_000483.3:p.Asp110His	rs113993958
CFTR	NM_000492.3:c.1624G>T	NP_000483.3:p.Gly542Ter	rs113993959

Seznam vyšetřovaných variant - CarrierTest



GEN	HGVSc	HGVSp	dbSNP
CFTR	NM_000492.3:c.1521_1523delCTT	NP_000483.3:p.Phe508delPhe	rs113993960
CFTR	NM_000492.3:c.220C>T	NP_000483.3:p.Arg74Trp	rs115545701
CFTR	NM_000492.3:c.1519_1521delATC	NP_000483.3:p.Ile507del	rs121908745
CFTR	NM_000492.3:c.2052_2053insA (2052dupA)	NP_000483.3:p.Gln685Thrfs	rs121908746
CFTR	NM_000492.3:c.2052delA	NP_000483.3:p.Lys684Asnfs	rs121908746
CFTR	NM_000492.3:c.1766+1G>C		rs121908748
CFTR	NM_000492.3:c.1766+1G>A		rs121908748
CFTR	NM_000492.3:c.1766+1G>T		rs121908748
CFTR	NM_000492.3:c.223C>T	NP_000483.3:p.Arg75Ter	rs121908749
CFTR	NM_000492.3:c.271G>A	NP_000483.3:p.Gly91Arg	rs121908750
CFTR	NM_000492.3:c.274G>A	NP_000483.3:p.Glu92Lys	rs121908751
CFTR	NM_000492.3:c.274G>T	NP_000483.3:p.Glu92Ter	rs121908751
CFTR	NM_000492.3:c.617T>G	NP_000483.3:p.Leu206Trp	rs121908752
CFTR	NM_000492.3:c.1055G>A	NP_000483.3:p.Arg352Gln	rs121908753
CFTR	NM_000492.3:c.1572C>A	NP_000483.3:p.Cys524Ter	rs121908754
CFTR	NM_000492.3:c.1646G>A	NP_000483.3:p.Ser549Asn	rs121908755
CFTR	NM_000492.3:c.1645A>C	NP_000483.3:p.Ser549Arg	rs121908757
CFTR	NM_000492.3:c.1721C>A	NP_000483.3:p.Pro574His	rs121908758
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CFTR	NM_000492.3:c.3587C>G	NP_000483.3:p.Ser1196Ter	rs121908763
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CFTR	NM_000492.3:c.1477_1478del	NP_000483.3:p.Gln493ValfsTer10	rs121908775
CFTR	NM_000492.3:c.1545_1546delTA	NP_000483.3:p.Tyr515_Arg516del	rs121908776
CFTR	NM_000492.3:c.1923_1931delinsA	NP_000483.3:p.Ser641ArgfsTer5	rs121908779
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CFTR	NM_000492.3:c.2737_2738insG	NP_000483.3:p.Tyr913Ter	rs121908788
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CFTR	NM_000492.3:c.1680-1G>A		rs121908794
CFTR	NM_000492.3:c.1766+5G>T		rs121908796
CFTR	NM_000492.3:c.2988G>A	NP_000483.3:p.Gln996%3D	rs121908797
CFTR	NM_000492.3:c.325_327delinsG	NP_000483.3:p.Tyr109GlyfsTer4	rs121908798
CFTR	NM_000492.3:c.2051_2052delinsG	NP_000483.3:p.Lys684SerfsTer38	rs121908799
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CFTR	NM_000492.3:c.1397C>G	NP_000483.3:p.Ser466Ter	rs121908805
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CFTR	NM_000492.3:c.2290C>T	NP_000483.3:p.Arg764Ter	rs121908810
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CFTR	NM_000492.3:c.1687T>G	NP_000483.3:p.Tyr563Asp	rs121909006
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CFTR	NM_000492.3:c.2551C>T	NP_000483.3:p.Arg851Ter	rs121909012
CFTR	NM_000492.3:c.1651G>A	NP_000483.3:p.Gly551Ser	rs121909013
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CFTR	NM_000492.3:c.1475C>T	NP_000483.3:p.Ser492Phe	rs121909017
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CFTR	NM_000492.3:c.171G>A	NP_000483.3:p.Trp57Ter	rs121909025
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CFTR	NM_000492.3:c.3468G>A	NP_000483.3:p.Leu1156%3D	rs139729994
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CFTR	NM_000492.3:c.2856G>C	p.Met952Ile	rs151048781
CFTR	NM_000492.3:c.4127_4131delTGGAT	NP_000483.3:p.Leu1376Serfs	rs1554397527
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CFTR	NM_000492.3:c.2900T>C	NP_000483.3:p.Leu967Ser	rs1800110
CFTR	NM_000492.3:c.1210-7_1210-6delTT		rs1805177
CFTR	NM_000492.3:c.772A>G	NP_000483.3:p.Arg258Gly	rs191456345
CFTR	NM_000492.3:c.1367T>C	NP_000483.3:p.Val456Ala	rs193922500
CFTR	NM_000492.3:c.1585-8G>A		rs193922503
CFTR	NM_000492.3:c.1673T>C	NP_000483.3:p.Leu558Ser	rs193922504
CFTR	NM_000492.3:c.1692del	NP_000483.3:p.Asp565MetfsTer7	rs193922505
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CFTR	NM_000492.3:c.825C>G	NP_000483.3:p.Tyr275Ter	rs193922532
CFTR	NM_000492.3:c.1521_1523delCTT	NP_000483.3:p.Phe508delPhe	rs199826652
CFTR	NM_000492.3:c.3205G>A	NP_000483.3:p.Gly1069Arg	rs200321110
CFTR	NM_000492.3:c.165-3C>T		rs200337193
CFTR	NM_000492.3:c.1837G>A	NP_000483.3:p.Ala613Thr	rs201978662
CFTR	NM_000492.3:c.2538G>A	NP_000483.3:p.Trp846Ter	rs267606722
CFTR	NM_000492.3:c.3731G>T	NP_000483.3:p.Gly1244Val	rs267606723
CFTR	NM_000492.3:c.3731G>A	NP_000483.3:p.Gly1244Glu	rs267606723
CFTR	NM_000492.3:c.3302T>A	NP_000483.3:p.Met1101Lys	rs36210737
CFTR	NM_000492.3:c.200C>T	NP_000483.3:p.Pro67Leu	rs368505753
CFTR	NM_000492.3:c.2723C>A	NP_000483.3:p.Thr908Asn	rs369521395
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CFTR	NM_000492.3:c.4242+1G>T		rs372227120
CFTR	NM_000492.3:c.4426C>T	NP_000483.3:p.Gln1476Ter	rs374705585
CFTR	NM_000492.3:c.2353C>T	NP_000483.3:p.Arg785Ter	rs374946172
CFTR	NM_000492.3:c.3988C>T	NP_000483.3:p.Gln1330Ter	rs375661578
CFTR	NM_000492.3:c.3988C>G	NP_000483.3:p.Gln1330Glu	rs375661578
CFTR	NM_000492.3:c.2909G>A	NP_000483.3:p.Gly970Asp	rs386134230
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CFTR	NM_000492.3:c.3718-1G>A		rs387906369
CFTR	NM_000492.3:c.3883_3886del	NP_000483.3:p.Ile1295PhefsTer32	rs387906373
CFTR	NM_000492.3:c.3532_3535dup	NP_000483.3:p.Thr1179IlefsTer17	rs387906378
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CFTR	NM_000492.3:c.1007T>A	NP_000483.3:p.Ile336Lys	rs397508139
CFTR	NM_000492.3:c.1021T>C	NP_000483.3:p.Ser341Pro	rs397508144
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CFTR	NM_000492.3:c.11C>A	NP_000483.3:p.Ser4Ter	rs397508173
CFTR	NM_000492.3:c.1202G>A	NP_000483.3:p.Trp401Ter	rs397508174
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CFTR	NM_000492.3:c.1393-2A>G		rs397508201
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CFTR	NM_000492.3:c.1466C>A	NP_000483.3:p.Ser489Ter	rs397508211
CFTR	NM_000492.3:c.1487G>A	NP_000483.3:p.Trp496Ter	rs397508216
CFTR	NM_000492.3:c.1505T>C	NP_000483.3:p.Ile502Thr	rs397508222
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CFTR	NM_000492.3:c.1648G>T	NP_000483.3:p.Gly550Ter	rs397508247
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CFTR	NM_000492.3:c.1650del	NP_000483.3:p.Gly551ValfsTer8	rs397508251
CFTR	NM_000492.3:c.166G>A	NP_000483.3:p.Glu56Lys	rs397508256
CFTR	NM_000492.3:c.1670del	NP_000483.3:p.Ser557PhefsTer2	rs397508257
CFTR	NM_000492.3:c.1680-877G>T		rs397508261
CFTR	NM_000492.3:c.1679+1G>C		rs397508263
CFTR	NM_000492.3:c.1679+1G>A		rs397508263
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CFTR	NM_000492.3:c.1680A>C	NP_000483.3:p.Arg560Ser	rs397508267
CFTR	NM_000492.3:c.169T>G	NP_000483.3:p.Trp57Gly	rs397508272
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CFTR	NM_000492.3:c.2017G>T	NP_000483.3:p.Gly673Ter	rs397508331
CFTR	NM_000492.3:c.2053C>T	NP_000483.3:p.Gln685Ter	rs397508336
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CFTR	NM_000492.3:c.2491G>T	NP_000483.3:p.Glu831Ter	rs397508387
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CFTR	NM_000492.3:c.263T>G	NP_000483.3:p.Leu88Ter	rs397508412
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CFTR	NM_000492.3:c.2859_2890del	NP_000483.3:p.Leu953PhefsTer11	rs397508445
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CFTR	NM_000492.3:c.296C>T	NP_000483.3:p.Pro99Leu	rs397508467
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CFTR	NM_000492.3:c.3017C>A	NP_000483.3:p.Ala1006Glu	rs397508480
CFTR	NM_000492.3:c.305T>G	NP_000483.3:p.Leu102Arg	rs397508490
CFTR	NM_000492.3:c.310del	NP_000483.3:p.Arg104GlufsTer3	rs397508499
CFTR	NM_000492.3:c.3124C>T	NP_000483.3:p.Gln1042Ter	rs397508500
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CFTR	NM_000492.3:c.3160C>G	NP_000483.3:p.His1054Asp	rs397508510
CFTR	NM_000492.3:c.3293G>A	NP_000483.3:p.Trp1098Ter	rs397508532
CFTR	NM_000492.3:c.3294G>A	NP_000483.3:p.Trp1098Ter	rs397508533
CFTR	NM_000492.3:c.3294G>C	NP_000483.3:p.Trp1098Cys	rs397508533
CFTR	NM_000492.3:c.3304A>T	NP_000483.3:p.Arg1102Ter	rs397508536
CFTR	NM_000492.3:c.3310G>T	NP_000483.3:p.Glu1104Ter	rs397508538
CFTR	NM_000492.3:c.3415A>G	NP_000483.3:p.Ile1139Val	rs397508556
CFTR	NM_000492.3:c.3430C>T	NP_000483.3:p.Gln1144Ter	rs397508560
CFTR	NM_000492.3:c.3435G>A	NP_000483.3:p.Trp1145Ter	rs397508561
CFTR	NM_000492.3:c.3475T>C	NP_000483.3:p.Ser1159Pro	rs397508572
CFTR	NM_000492.3:c.3476C>T	NP_000483.3:p.Ser1159Phe	rs397508573
CFTR	NM_000492.3:c.3605del	NP_000483.3:p.Asp1202AlafsTer9	rs397508587
CFTR	NM_000492.3:c.3718-3T>G		rs397508596
CFTR	NM_000492.3:c.3719T>G	NP_000483.3:p.Val1240Gly	rs397508598
CFTR	NM_000492.3:c.3761T>G	NP_000483.3:p.Leu1254Ter	rs397508604
CFTR	NM_000492.3:c.3883del	NP_000483.3:p.Ile1295PhefsTer33	rs397508630
CFTR	NM_000492.3:c.38C>T	NP_000483.3:p.Ser13Phe	rs397508635
CFTR	NM_000492.3:c.3908del	NP_000483.3:p.Asn1303ThrfsTer25	rs397508637
CFTR	NM_000492.3:c.3971T>C	NP_000483.3:p.Leu1324Pro	rs397508653
CFTR	NM_000492.3:c.4004T>C	NP_000483.3:p.Leu1335Pro	rs397508658
CFTR	NM_000492.3:c.4036_4042del	NP_000483.3:p.Leu1346MetfsTer6	rs397508662
CFTR	NM_000492.3:c.4077_4080delinsAA	NP_000483.3:p.Val1360ThrfsTer3	rs397508668
CFTR	NM_000492.3:c.4086dup	NP_000483.3:p.Lys1363Ter	rs397508669
CFTR	NM_000492.3:c.4111G>T	NP_000483.3:p.Glu1371Ter	rs397508675
CFTR	NM_000492.3:c.4124A>C	NP_000483.3:p.His1375Pro	rs397508678
CFTR	NM_000492.3:c.412_413insACT	NP_000483.3:p.Leu137_Leu138insHis	rs397508679
CFTR	NM_000492.3:c.4144C>T	NP_000483.3:p.Gln1382Ter	rs397508684
CFTR	NM_000492.3:c.4147dup	NP_000483.3:p.Ile1383AsnfsTer3	rs397508685
CFTR	NM_000492.3:c.413_415dup	NP_000483.3:p.Leu138dup	rs397508686
CFTR	NM_000492.3:c.4197_4198del	NP_000483.3:p.Cys1400Ter	rs397508693
CFTR	NM_000492.3:c.4231C>T	NP_000483.3:p.Gln1411Ter	rs397508701

GEN	HGVSc	HGVSp	dbSNP
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CFTR	NM_000492.3:c.4251del	NP_000483.3:p.Glu1418ArgfsTer14	rs397508706
CFTR	NM_000492.3:c.4300_4301dup	NP_000483.3:p.Ser1435GlyfsTer14	rs397508709
CFTR	NM_000492.3:c.50del	NP_000483.3:p.Phe17SerfsTer8	rs397508714
CFTR	NM_000492.3:c.481T>G	NP_000483.3:p.Tyr161Asp	rs397508729
CFTR	NM_000492.3:c.494T>C	NP_000483.3:p.Leu165Ser	rs397508736
CFTR	NM_000492.3:c.4C>T	NP_000483.3:p.Gln2Ter	rs397508740
CFTR	NM_000492.3:c.53+1G>T		rs397508746
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CFTR	NM_000492.3:c.577G>T	NP_000483.3:p.Glu193Ter	rs397508759
CFTR	NM_000492.3:c.579+3A>G		rs397508761
CFTR	NM_000492.3:c.57G>T	NP_000483.3:p.Trp19Cys	rs397508762
CFTR	NM_000492.3:c.647G>A	NP_000483.3:p.Trp216Ter	rs397508775
CFTR	NM_000492.3:c.658C>T	NP_000483.3:p.Gln220Ter	rs397508778
CFTR	NM_000492.3:c.680T>G	NP_000483.3:p.Leu227Arg	rs397508782
CFTR	NM_000492.3:c.695T>A	NP_000483.3:p.Val232Asp	rs397508783
CFTR	NM_000492.3:c.79G>A	NP_000483.3:p.Gly27Arg	rs397508796
CFTR	NM_000492.3:c.79G>T	NP_000483.3:p.Gly27Ter	rs397508796
CFTR	NM_000492.3:c.79G>C	NP_000483.3:p.Gly27Arg	rs397508796
CFTR	NM_000492.3:c.828C>A	NP_000483.3:p.Cys276Ter	rs397508799
CFTR	NM_000492.3:c.861_865del	NP_000483.3:p.Asn287LysfsTer19	rs397508805
CFTR	NM_000492.3:c.874G>T	NP_000483.3:p.Glu292Ter	rs397508811
CFTR	NM_000492.3:c.88C>T	NP_000483.3:p.Gln30Ter	rs397508815
CFTR	NM_000492.3:c.987del	NP_000483.3:p.Gly330GluTer39	rs397508824
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CFTR	NM_000492.3:c.273+3A>C		rs74467662
CFTR	NM_000492.3:c.3752G>A	NP_000483.3:p.Ser1251Asn	rs74503330
CFTR	NM_000492.3:c.1364C>T	NP_000483.3:p.Ala455Val	rs74551128
CFTR	NM_000492.3:c.1364C>A	NP_000483.3:p.Ala455Glu	rs74551128
CFTR	NM_000492.3:c.1523T>G	NP_000483.3:p.Phe508Cys	rs74571530
CFTR	NM_000492.3:c.1657C>T	NP_000483.3:p.Arg553Ter	rs74597325
CFTR	NM_000492.3:c.2052dup	NP_000483.3:p.Gln685ThrfsTer4	rs746460279
CFTR	NM_000492.3:c.3484C>T	NP_000483.3:p.Arg1162Ter	rs74767530
CFTR	NM_000492.3:c.3717+12191C>T		rs75039782
CFTR	NM_000492.3:c.1079C>A	NP_000483.3:p.Thr360Lys	rs75053309
CFTR	NM_000492.3:c.2988+1G>A		rs75096551
CFTR	NM_000492.3:c.2128A>T	NP_000483.3:p.Lys710Ter	rs75115087
CFTR	NM_000492.3:c.3700A>G	NP_000483.3:p.Ile1234Val	rs75389940
CFTR	NM_000492.3:c.1652G>A	NP_000483.3:p.Gly551Asp	rs75527207
CFTR	NM_000492.3:c.948delT	NP_000483.3:p.Phe316Leufs	rs75528968
CFTR	NM_000492.3:c.3368-2A>G		rs755416052
CFTR	NM_000492.3:c.3454G>C	NP_000483.3:p.Asp1152His	rs75541969
CFTR	NM_000492.3:c.1675G>A	NP_000483.3:p.Ala559Thr	rs75549581
CFTR	NM_000492.3:c.254G>A	NP_000483.3:p.Gly85Glu	rs75961395
CFTR	NM_000492.3:c.3140-26A>G		rs76151804
CFTR	NM_000492.3:c.416A>G	NP_000483.3:p.His139Arg	rs76371115
CFTR	NM_000492.3:c.1654C>T	NP_000483.3:p.Gln552Ter	rs76554633
CFTR	NM_000492.3:c.3764C>A	NP_000483.3:p.Ser1255Ter	rs76649725
CFTR	NM_000492.3:c.1585-1G>A		rs76713772
CFTR	NM_000492.3:c.3217dup	NP_000483.3:p.Tyr1073Leufs	rs768963919
CFTR	NM_000492.3:c.3846G>A	NP_000483.3:p.Trp1282Ter	rs77010898
CFTR	NM_000492.3:c.3691del	NP_000483.3:p.Ser1231ProfsTer4	rs77035409

GEN	HGVSc	HGVSp	dbSNP
CFTR	NM_000492.3:c.1477C>T	NP_000483.3:p.Gln493Ter	rs77101217
CFTR	NM_000492.3:c.579+1G>T		rs77188391
CFTR	NM_000492.3:c.178G>T	NP_000483.3:p.Glu60Ter	rs77284892
CFTR	NM_000492.3:c.1013C>T	NP_000483.3:p.Thr338Ile	rs77409459
CFTR	NM_000492.3:c.1558G>T	NP_000483.3:p.Val520Phe	rs77646904
CFTR	NM_000492.3:c.349C>T	NP_000483.3:p.Arg117Cys	rs77834169
CFTR	NM_000492.3:c.3848G>T	NP_000483.3:p.Arg1283Met	rs77902683
CFTR	NM_000492.3:c.1040G>A	NP_000483.3:p.Arg347His	rs77932196
CFTR	NM_000492.3:c.1040G>C	NP_000483.3:p.Arg347Pro	rs77932196
CFTR	NM_000492.3:c.3196C>T	NP_000483.3:p.Arg1066Cys	rs78194216
CFTR	NM_000492.3:c.579+5G>A		rs78440224
CFTR	NM_000492.3:c.350G>C	NP_000483.3:p.Arg117Pro	rs78655421
CFTR	NM_000492.3:c.350G>A	NP_000483.3:p.Arg117His	rs78655421
CFTR	NM_000492.3:c.489+1G>T		rs78756941
CFTR	NM_000492.3:c.3266G>A	NP_000483.3:p.Trp1089Ter	rs78802634
CFTR	NM_000492.3:c.3528delC	NP_000483.3:p.Thr1176Thr=fs	rs78984783
CFTR	NM_000492.3:c.988G>T	NP_000483.3:p.Gly330Ter	rs79031340
CFTR	NM_000492.3:c.2668C>T	NP_000483.3:p.Gln890Ter	rs79633941
CFTR	NM_000492.3:c.366T>A	NP_000483.3:p.Tyr122Ter	rs79660178
CFTR	NM_000492.3:c.2463_2464delTG	NP_000483.3:p.Ser821ArgfsTer4	rs797045156
CFTR	NM_000492.3:c.1117-1G>A		rs797045160
CFTR	NM_000492.3:c.1365_1366delGG	NP_000483.3:p.Val456CysfsTer25	rs797045161
CFTR	NM_000492.3:c.2053dup	NP_000483.3:p.Gln685ProfsTer84	rs797045162
CFTR	NM_000492.3:c.3472C>T	NP_000483.3:p.Arg1158Ter	rs79850223
CFTR	NM_000492.3:c.3909C>G	NP_000483.3:p.Asn1303Lys	rs80034486
CFTR	NM_000492.3:c.1679G>A	NP_000483.3:p.Arg560Lys	rs80055610
CFTR	NM_000492.3:c.1679G>C	NP_000483.3:p.Arg560Thr	rs80055610
CFTR	NM_000492.3:c.2657+5G>A		rs80224560
CFTR	NM_000492.3:c.532G>A	NP_000483.3:p.Gly178Arg	rs80282562
CLRN1	NM_001195794.1:c.144T>G	NP_001182723.1:p.Asn48Lys	rs111033258
CLRN1	NM_001195794.1:c.567T>G	NP_001182723.1:p.Tyr189Ter	rs121908140
CLRN1	NM_001195794.1:c.118T>G	NP_001182723.1:p.Cys40Gly	rs121908143
CLRN1	NM_001195794.1:c.92C>T	NP_001182723.1:p.Pro31Leu	rs374390376
CLRN1	NM_001195794.1:c.301_305delGT	NP_001182723.1:p.Val101_Ile102	rs397517932
COL4A5	NM_000495.4:c.446del	NP_000486.1:p.Pro149LeufsTer6	rs104886054
COL4A5	NM_000495.4:c.1871G>A	NP_000486.1:p.Gly624Asp	rs104886142
COL4A5	NM_000495.4:c.2215C>T	NP_000486.1:p.Pro739Ser	rs104886164
COL4A5	NM_000495.4:c.2057del	NP_000486.1:p.Pro686GlnfsTer50	rs104886167
COL4A5	NM_000495.4:c.41_42insTCTT	NP_000486.1:p.Leu14?fs	rs104886408
COL4A5	NM_000495.4:c.687del	NP_000486.1:p.Gly230ValfsTer24	rs1556405926
CTNS	NM_001031681.2:c.414G>A	NP_001026851.2:p.Trp138Ter	rs113994205
CTNS	NM_001031681.2:c.473T>C	NP_001026851.2:p.Leu158Pro	rs113994206
CTNS	NM_001031681.2:c.1015G>A	NP_001026851.2:p.Gly339Arg	rs121908127
CTNS	NM_001031681.2:c.140+1G>T		rs1567701415
CTNS	NM_001031681.2:c.433C>T	NP_001026851.2:p.Gln145Ter	rs2076101632
CYP21A2	NM_000500.7:c.293-13C>G		rs6467
CYP21A2	NM_000500.7:c.844G>T	NP_000491.4:p.Val282Leu	rs6471
CYP21A2	NM_000500.7:c.844G>C	NP_000491.4:p.Val282Leu	rs6471
CYP21A2	NM_000500.7:c.1069C>T	NP_000491.4:p.Arg357Trp	rs7769409

GEN	HGVSc	HGVSp	dbSNP
CarrierTest je pouze screeningovou metodou detekující časté varianty genu CYP21A2: c.293-13C>G; p.Arg357Trp a p.Val282Leu, která je asociována s mírnějším fenotypem. Z důvodu výskytu vysoce homologního pseudogenu a genových přestaveb v příslušné genomické oblasti tato metoda nedetekuje výskyt chimerního genu (vyskytuje se u 30 % pacientů), změny v počtu kopií částí CYP21A2 či celého genu a další patogenní mutace genu CYP21A2.			
CYP27A1	NM_000784.3:c.1183C>T	NP_000775.1:p.Arg395Cys	rs121908096
CYP27A1	NM_000784.3:c.1420C>T	NP_000775.1:p.Arg474Trp	rs121908098
CYP27A1	NM_000784.3:c.1214G>A	NP_000775.1:p.Arg405Gln	rs121908099
CYP27A1	NM_000784.3:c.379C>T	NP_000775.1:p.Arg127Trp	rs201114717
CYP27A1	NM_000784.3:c.646G>C	NP_000775.1:p.Ala216Pro	rs201346271
CYP27A1	NM_000784.3:c.1184+1G>A		rs587778777
CYP27A1	NM_000784.3:c.1184G>A	NP_000775.1:p.Arg395His	rs587778778
CYP27A1	NM_000784.3:c.410G>A	NP_000775.1:p.Arg137Gln	rs587778818
CYP27A1	NM_000784.3:c.808C>T	NP_000775.1:p.Arg270Ter	rs72551318
CYP27A1	NM_000784.3:c.1435C>T	NP_000775.1:p.Arg479Cys	rs72551322
CYP27A1	NM_000784.3:c.1381C>T	NP_000775.1:p.Gln461Ter	rs771819245
DHCR7	NM_001360.2:c.452G>A	NP_001351.2:p.Trp151Ter	rs11555217
DHCR7	NM_001360.2:c.866C>T	NP_001351.2:p.Thr289Ile	rs121909765
DHCR7	NM_001360.2:c.3G>A	NP_001351.2:p.Met1Ile	rs121909767
DHCR7	NM_001360.2:c.1055G>A	NP_001351.2:p.Arg352Gln	rs121909768
DHCR7	NM_001360.2:c.326T>C	NP_001351.2:p.Leu109Pro	rs121912195
DHCR7	NM_001360.2:c.964-1G>T		rs138659167
DHCR7	NM_001360.2:c.964-1G>C		rs138659167
DHCR7	NM_001360.2:c.322-1G>A		rs1565588719
DHCR7	NM_001360.2:c.841G>A	NP_001351.2:p.Val281Met	rs398123607
DHCR7	NM_001360.2:c.1210C>T	NP_001351.2:p.Arg404Cys	rs61757582
DHCR7	NM_001360.2:c.385_412+5del		rs746482788
DHCR7	NM_001360.2:c.355del	NP_001351.2:p.His119IlefsTer8	rs747827699
DHCR7	NM_001360.2:c.1337G>A	NP_001351.2:p.Arg446Gln	rs751604696
DHCR7	NM_001360.2:c.470T>C	NP_001351.2:p.Leu157Pro	rs753960624
DHCR7	NM_001360.2:c.1190C>T	NP_001351.2:p.Ser397Leu	rs773134475
DHCR7	NM_001360.2:c.278C>T	NP_001351.2:p.Thr93Met	rs80338853
DHCR7	NM_001360.2:c.506C>T	NP_001351.2:p.Ser169Leu	rs80338855
DHCR7	NM_001360.2:c.724C>T	NP_001351.2:p.Arg242Cys	rs80338856
DHCR7	NM_001360.2:c.725G>A	NP_001351.2:p.Arg242His	rs80338857
DHCR7	NM_001360.2:c.976G>T	NP_001351.2:p.Val326Leu	rs80338859
DHCR7	NM_001360.2:c.1054C>T	NP_001351.2:p.Arg352Trp	rs80338860
DHCR7	NM_001360.2:c.1228G>A	NP_001351.2:p.Gly410Ser	rs80338862
DHCR7	NM_001360.2:c.832-1G>C		rs80338863
DHCR7	NM_001360.2:c.1342G>A	NP_001351.2:p.Glu448Lys	rs80338864
DHCR7	NM_001360.2:c.740C>T	NP_001351.2:p.Ala247Val	rs886041354
DMD	NM_004006.2:c.6439-10_8217+32103dup		none
DMD	NM_004006.2:c.7309+12789_7490del		none
DMD	NM_004006.2:c.6251_6290+12981del		none
DMD	NM_004006.2:c.650-1827_771del		none
DMD	NM_004006.2:c.32-?_93+?dup62		none
DMD	NM_004006.2:c.4231C>T	NP_003997.2:p.Gln1411Ter	rs1010666282
DMD	NM_004006.3:c.2626G>T	NP_003997.2:p.Glu876Ter	rs1040401556
DMD	NM_004006.2:c.10108C>T	NP_003997.2:p.Arg3370Ter	rs104894787

GEN	HGVSc	HGVSp	dbSNP
DMD	NM_004006.3:c.10019G>A	NP_003997.2:p.Cys3340Tyr	rs104894788
DMD	NM_004006.2:c.9928C>T	NP_003997.2:p.Gln3310Ter	rs104894789
DMD	NM_004006.2:c.10141C>T	NP_003997.2:p.Arg3381Ter	rs104894790
DMD	NM_004006.2:c.9568C>T	NP_003997.2:p.Arg3190Ter	rs104894797
DMD	NM_004006.2:c.2776C>T	NP_003997.2:p.Gln926Ter	rs1055371114
DMD	NM_004006.3:c.1249G>T	NP_003997.2:p.Glu417Ter	rs1057515877
DMD	NM_004006.3:c.6139C>T	NP_003997.2:p.Gln2047Ter	rs1057516028
DMD	NM_004006.3:c.1602+1G>T		rs1057517960
DMD	NM_004006.2:c.1602+1G>A		rs1057517960
DMD	NM_004006.2:c.2168+1G>A		rs1057518207
DMD	NM_004006.3:c.3365_3366del	NP_003997.2:p.Glu1122ValfsTer6	rs1057518284
DMD	NM_004006.3:c.3366_3367insAG	NP_003997.2:p.Phe1123SerfsTer31	rs1057518284
DMD	NM_004006.3:c.5652del	NP_003997.2:p.Arg1884SerfsTer7	rs1057518692
DMD	NM_004006.3:c.177del	NP_003997.2:p.Gln60LysfsTer15	rs1057518834
DMD	NM_004006.2:c.5699T>G	NP_003997.2:p.Leu1900Ter	rs1057518866
DMD	NM_004006.2:c.1637G>A	NP_003997.2:p.Trp546Ter	rs1057518962
DMD	NM_004006.3:c.79dup	NP_003997.2:p.Ala27GlyfsTer5	rs1057519180
DMD	NM_004006.2:c.5758C>T	NP_003997.2:p.Gln1920Ter	rs1057520764
DMD	NM_004006.2:c.1006G>T	NP_003997.2:p.Glu336Ter	rs1057522454
DMD	NM_004006.2:c.9621T>A	NP_003997.2:p.Cys3207Ter	rs1057524037
DMD	NM_004006.2:c.4222C>T	NP_003997.2:p.Gln1408Ter	rs1057524291
DMD	NM_004006.2:c.5872G>T	NP_003997.2:p.Glu1958Ter	rs1057524825
DMD	NM_004006.2:c.3603+3A>T		rs1060502615
DMD	NM_004006.3:c.8391-1_8391delinsAA		rs1060502619
DMD	NM_004006.3:c.6662del	NP_003997.2:p.Asn2221MetfsTer26	rs1060502620
DMD	NM_004006.2:c.8209C>T	NP_003997.2:p.Gln2737Ter	rs1060502621
DMD	NM_004006.2:c.5647A>T	NP_003997.2:p.Lys1883Ter	rs1060502623
DMD	NM_004006.3:c.9238_9241del	NP_003997.2:p.Thr3080LeufsTer8	rs1060502624
DMD	NM_004006.3:c.10725del	NP_003997.2:p.Met3576CysfsTer18	rs1060502625
DMD	NM_004006.2:c.10797+1G>A		rs1060502626
DMD	NM_004006.2:c.2276T>A	NP_003997.2:p.Leu759Ter	rs1060502627
DMD	NM_004006.2:c.1653G>A	NP_003997.2:p.Trp551Ter	rs1060502629
DMD	NM_004006.2:c.10406_10407insAA	NP_003997.2:p.His3469fs	rs1060502630
DMD	NM_004006.3:c.4934_4937del	NP_003997.2:p.Lys1645ArgfsTer11	rs1060502632
DMD	NM_004006.2:c.8914C>T	NP_003997.2:p.Gln2972Ter	rs1060502633
DMD	NM_004006.2:c.1149+1G>A		rs1060502634
DMD	NM_004006.3:c.2076_2077insA	NP_003997.2:p.Gln693ThrfsTer27	rs1060502635
DMD	NM_004006.2:c.2202G>A	NP_003997.2:p.Trp734Ter	rs1060502636
DMD	NM_004006.2:c.4072-1G>T		rs1060502637
DMD	NM_004006.2:c.2665C>T	p.Arg889Ter	rs1060502639
DMD	NM_004006.3:c.6205del	NP_003997.2:p.Val2069TrpfsTer4	rs1060502640
DMD	NM_004006.2:c.6614+2T>C		rs1060502641
DMD	NM_004006.2:c.4071+1G>A		rs1060502643
DMD	NM_004006.2:c.1061G>A	NP_003997.2:p.Trp354Ter	rs1060502644
DMD	NM_004006.2:c.4858G>T	NP_003997.2:p.Glu1620Ter	rs1060502645
DMD	NM_004006.3:c.5884_5891del	NP_003997.2:p.Lys1962SerfsTer15	rs1060502646
DMD	NM_004006.3:c.5100_5101del	NP_003997.2:p.Leu1701fs	rs1060502647
DMD	NM_004006.2:c.9434del	NP_003997.2:p.Met3145fs	rs1060502648
DMD	NM_004006.3:c.38_39GA[1]	NP_003997.2:p.Glu14fs	rs1060502652
DMD	NM_004006.3:c.9650-4_9655del		rs1060502653
DMD	NM_004006.2:c.6118-1G>A		rs1060502656
DMD	NM_004006.3:c.7068_7069del	NP_003997.2:p.Gln2356HisfsTer8	rs1060502659

GEN	HGVSc	HGVSp	dbSNP
DMD	NM_004006.2:c.2077C>T	NP_003997.2:p.Gln693Ter	rs1060502661
DMD	NM_004006.3:c.5530C>T		rs1064325
DMD	NM_004006.3:c.7241_7243delinsGTTT	NP_003997.2:p.Asn2414SerfsTer13	rs1064792967
DMD	NM_004006.2:c.9287-27_9287-2del		rs1064792968
DMD	NM_004006.3:c.4538_4541del	NP_003997.2:p.Ser1513LysfsTer2	rs1064793479
DMD	NM_004006.3:c.9370_9371del	NP_003997.2:p.Leu3124GlufsTer7	rs1064793644
DMD	NM_004006.2:c.10651C>T	NP_003997.2:p.Gln3551Ter	rs1064793742
DMD	NM_004006.2:c.2555G>A	NP_003997.2:p.Trp852Ter	rs1064793964
DMD	NM_004006.2:c.3487C>T	NP_003997.2:p.Gln1163Ter	rs1064793986
DMD	NM_004006.3:c.8390+1G>C		rs1064794569
DMD	NM_004006.2:c.8390+1G>A		rs1064794569
DMD	NM_004006.2:c.5448+1G>A		rs1064796764
DMD	NM_004006.3:c.5448+1G>T		rs1064796764
DMD	NM_004006.3:c.10211del	NP_003997.2:p.Asp3404fs	rs1114167437
DMD	NM_004006.3:c.4071+1del		rs1114167439
DMD	NM_004006.3:c.5985T>G	NP_003997.2:p.Tyr1995Ter	rs1131691292
DMD	NM_004006.2:c.9958C>A	NP_003997.2:p.Pro3320Thr	rs1131691677
DMD	NM_004006.3:c.6124A>T	NP_003997.2:p.Lys2042Ter	rs1178785582
DMD	NM_004006.3:c.10077T>G	NP_003997.2:p.Tyr3359Ter	rs1181271457
DMD	NM_004006.3:c.4660G>T	NP_003997.2:p.Glu1554Ter	rs1216000876
DMD	NM_004006.3:c.1254_1255dup	NP_003997.2:p.Glu419ValfsTer7	rs1220963408
DMD	NM_004006.3:c.2968C>T	NP_003997.2:p.Gln990Ter	rs1233380601
DMD	NM_004006.2:c.831+1G>C		rs1239018406
DMD	NM_004006.2:c.831+1G>T		rs1239018406
DMD	NM_004006.2:c.831+1G>A		rs1239018406
DMD	NM_004006.2:c.7400T>A	NP_003997.2:p.Leu2467Ter	rs1254776844
DMD	NM_004006.3:c.3469G>T	NP_003997.2:p.Glu1157Ter	rs128625226
DMD	NM_004006.2:c.2791G>T	NP_003997.2:p.Glu931Ter	rs128625227
DMD	NM_004006.2:c.5551C>T	NP_003997.2:p.Gln1851Ter	rs128625228
DMD	NM_004006.2:c.8944C>T	NP_003997.2:p.Arg2982Ter	rs128625229
DMD	NM_004006.2:c.6955C>T	NP_003997.2:p.Gln2319Ter	rs128625230
DMD	NM_004006.3:c.161T>G	NP_003997.2:p.Leu54Arg	rs128626231
DMD	NM_004006.3:c.2017C>T	NP_003997.2:p.Gln673Ter	rs128626232
DMD	NM_004006.2:c.178C>T	NP_003997.2:p.Gln60Ter	rs128626233
DMD	NM_004006.3:c.253C>T	NP_003997.2:p.Gln85Ter	rs128626234
DMD	NM_004006.2:c.433C>T	NP_003997.2:p.Arg145Ter	rs128626235
DMD	NM_004006.3:c.503C>A	NP_003997.2:p.Ala168Asp	rs128626236
DMD	NM_004006.3:c.691T>A	NP_003997.2:p.Tyr231Asn	rs128626237
DMD	NM_004006.3:c.724C>T	NP_003997.2:p.Gln242Ter	rs128626238
DMD	NM_004006.3:c.748G>T	NP_003997.2:p.Glu250Ter	rs128626239
DMD	NM_004006.3:c.1438G>T	NP_003997.2:p.Gly480Ter	rs128626240
DMD	NM_004006.3:c.1489C>T	NP_003997.2:p.Gln497Ter	rs128626241
DMD	NM_004006.3:c.1952G>A	NP_003997.2:p.Trp651Ter	rs128626242
DMD	NM_004006.3:c.2308A>T	NP_003997.2:p.Lys770Ter	rs128626243
DMD	NM_004006.3:c.2317A>G	NP_003997.2:p.Lys773Glu	rs128626244
DMD	NM_004006.2:c.3121C>T	NP_003997.2:p.Gln1041Ter	rs128626245
DMD	NM_004006.2:c.3188G>A	NP_003997.2:p.Trp1063Ter	rs128626246
DMD	NM_004006.3:c.4213C>T	NP_003997.2:p.Gln1405Ter	rs128626247
DMD	NM_004006.2:c.4414C>T	NP_003997.2:p.Gln1472Ter	rs128626248
DMD	NM_004006.2:c.5899C>T	NP_003997.2:p.Arg1967Ter	rs128626249
DMD	NM_004006.2:c.6292C>T	NP_003997.2:p.Arg2098Ter	rs128626250
DMD	NM_004006.2:c.6373C>T	NP_003997.2:p.Gln2125Ter	rs128626251

GEN	HGVSc	HGVSp	dbSNP
DMD	NM_004006.3:c.6790C>T	NP_003997.2:p.Gln2264Ter	rs128626252
DMD	NM_004006.3:c.7402G>T	NP_003997.2:p.Glu2468Ter	rs128626253
DMD	NM_004006.2:c.9197C>A	NP_003997.2:p.Ser3066Ter	rs128626254
DMD	NM_004006.3:c.835A>G	NP_003997.2:p.Thr279Ala	rs128627255
DMD	NM_004006.2:c.8713C>T	NP_003997.2:p.Arg2905Ter	rs128627256
DMD	NM_004006.3:c.5984del	NP_003997.2:p.Tyr1995LeufsTer7	rs128627257
DMD	NM_004006.2:c.5082G>A	NP_003997.2:p.Trp1694Ter	rs1295394996
DMD	NM_004006.3:c.5653C>T	NP_003997.2:p.Gln1885Ter	rs1295935628
DMD	NM_004006.3:c.1837A>T	NP_003997.2:p.Lys613Ter	rs1302764635
DMD	NM_004006.3:c.6580G>T	NP_003997.2:p.Glu2194Ter	rs1304218104
DMD	NM_004006.3:c.3162+1G>A		rs1335666832
DMD	NM_004006.3:c.3697C>T	NP_003997.2:p.Gln1233Ter	rs1339088514
DMD	NM_004006.2:c.589G>T	NP_003997.2:p.Glu197Ter	rs1340365803
DMD	NM_004006.2:c.5533G>T	NP_003997.2:p.Glu1845Ter	rs1354256883
DMD	NM_004006.2:c.10783C>T	NP_003997.2:p.Gln3595Ter	rs1385794215
DMD	NM_004006.3:c.31+5G>A		rs1403865104
DMD	NM_004006.3:c.2440G>T	NP_003997.2:p.Glu814Ter	rs1408435603
DMD	NM_004006.2:c.7454G>A	NP_003997.2:p.Trp2485Ter	rs142500746
DMD	NM_004006.2:c.6423C>A	NP_003997.2:p.Tyr2141Ter	rs1425702864
DMD	NM_004006.3:c.6423C>G	NP_003997.2:p.Tyr2141Ter	rs1425702864
DMD	NM_004006.3:c.2556G>A	NP_003997.2:p.Trp852Ter	rs1429329309
DMD	NM_004006.2:c.3020C>A	NP_003997.2:p.Ser1007Ter	rs144732570
DMD	NM_004006.3:c.3603+2T>G		rs146071084
DMD	NM_004006.3:c.3603+2T>A		rs146071084
DMD	NM_004006.3:c.3603+2T>C		rs146071084
DMD	c.4000G>A	NP_003997.2:p.Gly1334Arg	rs146880270
DMD	NM_004006.2:c.4000G>T	NP_003997.2:p.Gly1334Ter	rs146880270
DMD	NM_004006.2:c.918T>A	NP_003997.2:p.Tyr306Ter	rs149106712
DMD	NM_004006.3:c.10554_10555insG		rs1555998436
DMD	NM_004006.3:c.10519G>T	NP_003997.2:p.Glu3507Ter	rs1556024095
DMD	NM_004006.3:c.10504del	NP_003997.2:p.Glu3502LysfsTer5	rs1556024147
DMD	NM_004006.3:c.10367del	NP_003997.2:p.Asn3456MetfsTer10	rs1556028034
DMD	NM_004006.2:c.10238del	NP_003997.2:p.Ile3413fs	rs1556035795
DMD	NM_004006.2:c.10224-175_10230del		rs1556035817
DMD	NM_004006.2:c.10121del	NP_003997.2:p.Lys3374fs	rs1556037429
DMD	NM_004006.2:c.10109G>C	NP_003997.2:p.Arg3370Pro	rs1556037455
DMD	NM_004006.2:c.9975-1G>A		rs1556040444
DMD	NM_004006.3:c.9881_9890del	NP_003997.2:p.Trp3294SerfsTer33	rs1556046080
DMD	NM_004006.2:c.9691C>T	NP_003997.2:p.Gln3231Ter	rs1556235825
DMD	NM_004006.2:c.9649+1G>A		rs1556256329
DMD	NM_004006.3:c.9560A>G	NP_003997.2:p.Asp3187Gly	rs1556280638
DMD	NM_004006.2:c.9183G>A	NP_003997.2:p.Trp3061Ter	rs1556503937
DMD	NM_004006.2:c.9163+2T>G		rs1556539065
DMD	NM_004006.3:c.9040_9041insC	NP_003997.2:p.Leu3014ProfsTer27	rs1556641290
DMD	NM_004006.3:c.8937+1G>T		rs1556656077
DMD	NM_004006.3:c.8937+1G>C		rs1556656077
DMD	NM_004006.3:c.8880G>A	NP_003997.2:p.Trp2960Ter	rs1556656184
DMD	NM_004006.3:c.8782A>T	NP_003997.2:p.Lys2928Ter	rs1556656298
DMD	NM_004006.3:c.8683_8684insG	NP_003997.2:p.Glu2895GlyfsTer15	rs1556656456
DMD	NM_004006.3:c.8675del	NP_003997.2:p.Pro2892LeufsTer27	rs1556656487
DMD	NM_004006.3:c.8647A>T	NP_003997.2:p.Lys2883Ter	rs1556656767
DMD	NM_004006.3:c.8604dup	NP_003997.2:p.Val2869CysfsTer26	rs1556656818

Seznam vyšetřovaných variant - CarrierTest



GEN	HGVSc	HGVSp	dbSNP
DMD	NM_004006.3:c.8579del	NP_003997.2:p.Pro2860LeufsTer2	rs1556656847
DMD	NM_004006.2:c.8575G>T	NP_003997.2:p.Glu2859Ter	rs1556656851
DMD	NM_004006.3:c.8549_8550insAGGC		rs1556656895
DMD	NM_004006.2:c.8547+1G>A		rs1556665052
DMD	NM_004006.3:c.8391-2A>G		rs1556665303
DMD	NM_004006.3:c.8263del	NP_003997.2:p.Leu2755TrpfsTer9	rs1556669914
DMD	NM_004006.3:c.8197G>T	NP_003997.2:p.Glu2733Ter	rs1556764743
DMD	NM_004006.2:c.8196del	NP_003997.2:p.Glu2733fs	rs1556764753
DMD	NM_004006.3:c.8096_8112del	NP_003997.2:p.Glu2699AlafsTer5	rs1556764880
DMD	NM_004006.2:c.8077C>T	NP_003997.2:p.Gln2693Ter	rs1556764922
DMD	NM_004006.3:c.8066_8070del	NP_003997.2:p.Arg2689ThrfsTer19	rs1556764934
DMD	NM_004006.3:c.2132del	NP_003997.2:p.Lys711ArgfsTer18	rs1556780579
DMD	NM_004006.3:c.2101G>T	NP_003997.2:p.Glu701Ter	rs1556780711
DMD	NM_004006.3:c.8027+1G>T		rs1556789913
DMD	NM_004006.2:c.8027+1G>A		rs1556789913
DMD	NM_004006.3:c.7899G>A	NP_003997.2:p.Trp2633Ter	rs1556790316
DMD	NM_004006.2:c.1992+1G>T		rs1556802319
DMD	NM_004006.3:c.1977_1981del	NP_003997.2:p.Lys660TyrfsTer58	rs1556802365
DMD	NM_004006.3:c.7661-1G>C		rs1556806346
DMD	NM_004006.3:c.7661-1G>A		rs1556806346
DMD	NM_004006.2:c.7661-2A>G		rs1556806356
DMD	NM_004006.3:c.1812+2T>A		rs1556809625
DMD	NM_004006.3:c.1795del	NP_003997.2:p.Ser599ValfsTer8	rs1556809704
DMD	NM_004006.3:c.1659_1660insGTAA	NP_003997.2:p.Leu554ValfsTer14	rs1556810362
DMD	NM_004006.3:c.7660+1G>C		rs1556852444
DMD	NM_004006.3:c.1481del	NP_003997.2:p.Lys494ArgfsTer7	rs1556853022
DMD	NM_004006.3:c.1476del	NP_003997.2:p.Gln492HisfsTer9	rs1556853039
DMD	NM_004006.2:c.1366A>T	NP_003997.2:p.Lys456Ter	rs1556853298
DMD	NM_004006.2:c.1304_1331+10del		rs1556875089
DMD	NM_004006.2:c.1292G>A	NP_003997.2:p.Trp431Ter	rs1556875224
DMD	NM_004006.2:c.1075G>T	NP_003997.2:p.Glu359Ter	rs1556876346
DMD	NM_004006.3:c.1048G>T	NP_003997.2:p.Glu350Ter	rs1556876421
DMD	NM_004006.3:c.7319dup	NP_003997.2:p.Thr2441AspfsTer12	rs1556880327
DMD	NM_004006.2:c.7310-1G>A		rs1556880354
DMD	NM_004006.3:c.7268del	NP_003997.2:p.Lys2423SerfsTer5	rs1556917057
DMD	NM_004006.3:c.857dup	NP_003997.2:p.Tyr286Ter	rs1556929259
DMD	NM_004006.3:c.7159C>T	NP_003997.2:p.Gln2387Ter	rs1556930579
DMD	NM_004006.2:c.686T>G	NP_003997.2:p.Leu229Ter	rs1556930769
DMD	NM_004006.3:c.656dup	NP_003997.2:p.Asp219GluTer7	rs1556930839
DMD	NM_004006.3:c.7098+1G>A		rs1556962223
DMD	NM_004006.2:c.7098+1G>T		rs1556962223
DMD	NM_004006.2:c.7085_7088dup	NP_003997.2:p.Asp2364fs	rs1556962271
DMD	NM_004006.3:c.7052_7053del	NP_003997.2:p.Leu2351Ter	rs1556962326
DMD	NM_004006.3:c.6913-11_6918del		rs1556962571
DMD	NM_004006.3:c.6665_6668del	NP_003997.2:p.Glu2222ValfsTer24	rs1557011872
DMD	NM_004006.3:c.6651_6652del	NP_003997.2:p.Asp2219PhefsTer3	rs1557011929
DMD	NM_004006.2:c.6615-2A>T		rs1557011973
DMD	NM_004006.3:c.6615-2A>G		rs1557011973
DMD	NM_004006.3:c.6544C>T	NP_003997.2:p.Gln2182Ter	rs1557038061
DMD	NM_004006.3:c.6472_6473del	NP_003997.2:p.Val2158CysfsTer64	rs1557038198
DMD	NM_004006.3:c.649+5G>T		rs1557047808
DMD	NM_004006.3:c.630del	NP_003997.2:p.Lys211AsnfsTer19	rs1557047827

GEN	HGVSc	HGVSp	dbSNP
DMD	NM_004006.3:c.598_599insGCAT	NP_003997.2:p.Phe200CysfsTer18	rs1557047875
DMD	NM_004006.3:c.478_479insAC	NP_003997.2:p.Thr160AsnfsTer11	rs1557052560
DMD	NM_004006.3:c.468delinsGG	NP_003997.2:p.Ile157AspfsTer9	rs1557052599
DMD	NM_004006.3:c.365del	NP_003997.2:p.Asn122MetfsTer2	rs1557052797
DMD	NM_004006.2:c.357+1G>A		rs1557058294
DMD	NM_004006.3:c.356del	NP_003997.2:p.Gln119ArgfsTer5	rs1557058308
DMD	NM_004006.3:c.319dup	NP_003997.2:p.Thr107AsnfsTer31	rs1557058353
DMD	NM_004006.3:c.283G>T	NP_003997.2:p.Gly95Ter	rs1557058415
DMD	NM_004006.3:c.238_239insG	NP_003997.2:p.Ala80GlyfsTer9	rs1557079469
DMD	NM_004006.2:c.186+1G>A		rs1557084067
DMD	NM_004006.3:c.153dup	NP_003997.2:p.Asp52ArgfsTer37	rs1557084128
DMD	NM_004006.2:c.100A>T	NP_003997.2:p.Lys34Ter	rs1557084280
DMD	NM_004006.2:c.6424A>T	NP_003997.2:p.Lys2142Ter	rs1557218076
DMD	NM_004006.3:c.6423del	NP_003997.2:p.Tyr2141Ter	rs1557218093
DMD	NM_004006.2:c.6408G>A	NP_003997.2:p.Trp2136Ter	rs1557218131
DMD	NM_004006.2:c.6317G>A	NP_003997.2:p.Trp2106Ter	rs1557218269
DMD	NM_004006.3:c.6025_6026del	NP_003997.2:p.Gln2009SerfsTer13	rs1557272125
DMD	NM_004006.3:c.5910del	NP_003997.2:p.Phe1971LeufsTer12	rs1557290181
DMD	NM_004006.3:c.5800G>T	NP_003997.2:p.Glu1934Ter	rs1557290355
DMD	NM_004006.3:c.5713G>T	NP_003997.2:p.Glu1905Ter	rs1557291003
DMD	NM_004006.3:c.5613_5640del	NP_003997.2:p.Lys1871AsnfsTer11	rs1557291170
DMD	NM_004006.2:c.5602A>T	NP_003997.2:p.Arg1868Ter	rs1557291229
DMD	NM_004006.3:c.5595del	NP_003997.2:p.Arg1865SerfsTer9	rs1557291242
DMD	NM_004006.2:c.5586+1G>A		rs1557292599
DMD	NM_004006.3:c.5548_5552del	NP_003997.2:p.Lys1850AlafsTer8	rs1557292678
DMD	NM_004006.3:c.5407C>T	NP_003997.2:p.Gln1803Ter	rs1557294228
DMD	NM_004006.3:c.5360_5361insA	NP_003997.2:p.Asn1787LysfsTer9	rs1557294296
DMD	NM_004006.3:c.5334del	NP_003997.2:p.Pro1779LeufsTer2	rs1557294341
DMD	NM_004006.3:c.19del	NP_003997.2:p.Val7Ter	rs1557300135
DMD	NM_004006.2:c.5266C>T	NP_003997.2:p.Gln1756Ter	rs1557303544
DMD	NM_004006.3:c.5139del	NP_003997.2:p.Glu1714LysfsTer7	rs1557304860
DMD	NM_004006.2:c.5024T>A	NP_003997.2:p.Leu1675Ter	rs1557305418
DMD	NM_004006.3:c.4918_4919delinsTG	NP_003997.2:p.Thr1640Ter	rs1557305645
DMD	NM_004006.3:c.4845+2T>G		rs1557315881
DMD	NM_004006.2:c.4838G>A	NP_003997.2:p.Trp1613Ter	rs1557315928
DMD	NM_004006.2:c.4618G>T	NP_003997.2:p.Glu1540Ter	rs1557320151
DMD	NM_004006.2:c.4591C>T	NP_003997.2:p.Gln1531Ter	rs1557320194
DMD	NM_004006.2:c.4501C>T	NP_003997.2:p.Gln1501Ter	rs1557322201
DMD	NM_004006.2:c.4295del	NP_003997.2:p.Gln1432fs	rs1557322834
DMD	NM_004006.3:c.4290_4291del	NP_003997.2:p.His1430GlnfsTer15	rs1557322838
DMD	NM_004006.3:c.4120G>T	NP_003997.2:p.Glu1374Ter	rs1557340452
DMD	NM_004006.3:c.4060del	NP_003997.2:p.Leu1354TyrfsTer6	rs1557357445
DMD	NM_004006.3:c.3885del	NP_003997.2:p.Pro1296LeufsTer11	rs1557359155
DMD	NM_004006.3:c.3817del	NP_003997.2:p.Ser1273HisfsTer10	rs1557359217
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DMD	NM_004006.3:c.3389_3390insGA	NP_003997.2:p.Leu1131AsnfsTer23	rs1557369413
DMD	NM_004006.3:c.3309_3313del	NP_003997.2:p.Ser1104LysfsTer5	rs1557369493
DMD	NM_004006.3:c.3276+2del		rs1557369964
DMD	NM_004006.2:c.3268C>T	NP_003997.2:p.Gln1090Ter	rs1557369991
DMD	NM_004006.2:c.2949+1G>A		rs1557374482
DMD	NM_004006.3:c.2804-1del		rs1557374667

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.2669T>A	NP_003997.2:p.Leu890Ter	rs1557380616
DMD	NM_004006.3:c.2644del	NP_003997.2:p.Asp882IlefsTer9	rs1557380670
DMD	NM_004006.3:c.2638del	NP_003997.2:p.Leu880TyrfsTer11	rs1557380685
DMD	NM_004006.2:c.2423_2424insA	NP_003997.2:p.Asn809fs	rs1557383751
DMD	NM_004006.3:c.2416G>T	NP_003997.2:p.Glu806Ter	rs1557383761
DMD	NM_004006.2:c.2292+2T>G		rs1557396600
DMD	NM_004006.2:c.2257G>T	NP_003997.2:p.Glu753Ter	rs1557396632
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DMD	NM_004006.2:c.2111del	NP_003997.2:p.Pro704fs	rs1569169736
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DMD	NM_004006.2:c.1652G>A	NP_003997.2:p.Trp551Ter	rs1569230215
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DMD	NM_004006.2:c.7252C>T	NP_003997.2:p.Gln2418Ter	rs1569451994
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DMD	NM_004006.2:c.9634G>T	NP_003997.2:p.Glu3212Ter	rs1569463953
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DMD	NM_004006.3:c.703_704insATCAC	NP_003997.2:p.Leu235HisfsTer11	rs1569469298
DMD	NM_004006.2:c.6874G>T	NP_003997.2:p.Glu2292Ter	rs1569515486
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DMD	NM_004006.2:c.9100C>T	NP_003997.2:p.Arg3034Ter	rs1569530432
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DMD	NM_004006.3:c.6025C>T	NP_003997.2:p.Gln2009Ter	rs1569556943
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DMD	NM_004006.2:c.5867G>A	NP_003997.2:p.Trp1956Ter	rs1569558906
DMD	NM_004006.3:c.5729_5733del	NP_003997.2:p.Arg1910AsnfsTer5	rs1569558953
DMD	NM_004006.3:c.5680del	NP_003997.2:p.Asp1894MetfsTer7	rs1569558964

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.5459_5460insGACAA	NP_003997.2:p.Asn1820LysfsTer7	rs1569559110
DMD	NM_004006.3:c.5390_5411del	NP_003997.2:p.Leu1797ArgfsTer2	rs1569559198
DMD	NM_004006.2:c.5371C>T	NP_003997.2:p.Gln1791Ter	rs1569559204
DMD	NM_004006.2:c.5154+2T>C		rs1569559822
DMD	NM_004006.2:c.5026-2A>G		rs1569559849
DMD	NM_004006.3:c.4786_4787del	NP_003997.2:p.Lys1596GluTer5	rs1569560475
DMD	NM_004006.2:c.4758G>A	NP_003997.2:p.Trp1586Ter	rs1569560477
DMD	NM_004006.2:c.4546A>T	NP_003997.2:p.Lys1516Ter	rs1569560626
DMD	NM_004006.2:c.4486G>T	NP_003997.2:p.Glu1496Ter	rs1569560739
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DMD	NM_004006.2:c.3856G>T	NP_003997.2:p.Glu1286Ter	rs1569562570
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DMD	NM_004006.2:c.3742C>T	NP_003997.2:p.Gln1248Ter	rs1569562936
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DMD	NM_004006.3:c.1323dup	NP_003997.2:p.Gln442fs	rs1602459252
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DMD	NM_004006.3:c.1255G>T	NP_003997.2:p.Glu419Ter	rs1602460062
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DMD	NM_004006.3:c.1144del	NP_003997.2:p.His382fs	rs1602467590
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DMD	NM_004006.3:c.998C>A	NP_003997.2:p.Ser333Ter	rs1602468968
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GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.31+1del		rs1603419074
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DMD	NM_004006.3:c.635_636dup	NP_003997.2:p.Leu213fs	rs1603432059
DMD	NM_004006.2:c.616C>T	NP_003997.2:p.Gln206Ter	rs1603432076
DMD	NM_004006.3:c.453T>G	NP_003997.2:p.Tyr151Ter	rs1603437254
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DMD	NM_004006.3:c.7858dup	NP_003997.2:p.Ile2620fs	rs1603445283
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DMD	NM_004006.3:c.263_264dup	NP_003997.2:p.Val89fs	rs1603447057
DMD	NM_004006.3:c.196A>T	NP_003997.2:p.Lys66Ter	rs1603447072
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DMD	NM_004006.3:c.7580T>A		rs1603452200
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DMD	NM_004006.3:c.7048_7057del		rs1603513697
DMD	NM_004006.3:c.7029G>A	NP_003997.2:p.Trp2343Ter	rs1603513803
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DMD	NM_004006.3:c.6754C>T	NP_003997.2:p.Gln2252Ter	rs1603615883
DMD	NM_004006.3:c.6407G>A	NP_003997.2:p.Trp2136Ter	rs1603628479
DMD	NM_004006.3:c.6364_6365del	NP_003997.2:p.Glu2122fs	rs1603628482
DMD	NM_004006.3:c.6357G>A	NP_003997.2:p.Trp2119Ter	rs1603628483
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DMD	NM_004006.3:c.6300del	NP_003997.2:p.Asp2100fs	rs1603628487
DMD	NM_004006.3:c.6151del	NP_003997.2:p.Arg2051fs	rs1603629988
DMD	NM_004006.3:c.6023C>G	NP_003997.2:p.Ser2008Ter	rs1603630431
DMD	NM_004006.3:c.5922+4A>G		rs1603631218
DMD	NM_004006.3:c.5922+1G>T		rs1603631219
DMD	NM_004006.3:c.5821dup	NP_003997.2:p.Ala1941fs	rs1603631225
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DMD	NM_004006.3:c.5689dup	NP_003997.2:p.Glu1897fs	rs1603631241
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DMD	NM_004006.3:c.5387dup	NP_003997.2:p.Leu1797fs	rs1603631365
DMD	NM_004006.3:c.5376del	NP_003997.2:p.Lys1792fs	rs1603631367
DMD	NM_004006.3:c.5358del	NP_003997.2:p.Phe1786fs	rs1603631368
DMD	NM_004006.3:c.5167G>T	NP_003997.2:p.Glu1723Ter	rs1603631705
DMD	NM_004006.3:c.5056C>T	NP_003997.2:p.Gln1686Ter	rs1603631744
DMD	NM_004006.3:c.4980G>A	NP_003997.2:p.Trp1660Ter	rs1603631752

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.4846-1G>T		rs1603631757
DMD	NM_004006.3:c.4693C>T	NP_003997.2:p.Gln1565Ter	rs1603632117
DMD	NM_004006.3:c.4682_4685AAAG[1]	NP_003997.2:p.Arg1562fs	rs1603632119
DMD	NM_004006.3:c.4610_4611insA		rs1603632245
DMD	NM_004006.3:c.4568_4569dup	NP_003997.2:p.Lys1524Ter	rs1603632246
DMD	NM_004006.3:c.4536_4540del	NP_003997.2:p.Val1515_Lys1516insTer	rs1603632248
DMD	NM_004006.3:c.4477_4493del	NP_003997.2:p.Val1493fs	rs1603632309
DMD	NM_004006.3:c.4438G>T	NP_003997.2:p.Glu1480Ter	rs1603632311
DMD	NM_004006.2:c.4433T>G	NP_003997.2:p.Leu1478Ter	rs1603632312
DMD	NM_004006.3:c.4423A>T	NP_003997.2:p.Lys1475Ter	rs1603632314
DMD	NM_004006.3:c.4352T>G	NP_003997.2:p.Leu1451Ter	rs1603632316
DMD	NM_004006.3:c.4327C>T	NP_003997.2:p.Gln1443Ter	rs1603632322
DMD	NM_004006.3:c.4306_4309del	NP_003997.2:p.Ala1436fs	rs1603632324
DMD	NM_004006.3:c.4294C>T	NP_003997.2:p.Gln1432Ter	rs1603632326
DMD	NM_004006.3:c.3980del	NP_003997.2:p.Ala1327fs	rs1603633486
DMD	NM_004006.3:c.3843G>A	NP_003997.2:p.Trp1281Ter	rs1603633541
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DMD	NM_004006.3:c.3533_3536del	NP_003997.2:p.Glu1178fs	rs1603633864
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DMD	NM_004006.3:c.3013G>T	NP_003997.2:p.Glu1005Ter	rs1603634206
DMD	NM_004006.3:c.2960del	NP_003997.2:p.Ser987fs	rs1603634207
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DMD	NM_004006.3:c.2790_2791AG[1]	NP_003997.2:p.Glu931fs	rs1603634604
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DMD	NM_004006.3:c.2461_2462GA[2]	NP_003997.2:p.Arg822fs	rs1603634744
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DMD	NM_004006.3:c.2461G>T	NP_003997.2:p.Glu821Ter	rs1603634745
DMD	NM_004006.3:c.2448C>A	NP_003997.2:p.Cys816Ter	rs1603634746
DMD	NM_004006.3:c.2442del	NP_003997.2:p.Glu814fs	rs1603634747
DMD	NM_004006.3:c.2407del	NP_003997.2:p.Gln803fs	rs1603634748
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DMD	NM_004006.3:c.2114del	NP_003997.2:p.Pro705fs	rs1603635949
DMD	NM_004006.3:c.2101del	NP_003997.2:p.Glu701fs	rs1603635950
DMD	NM_004006.3:c.2064dup	NP_003997.2:p.Thr689fs	rs1603635953
DMD	NM_004006.3:c.1973_1992+966del		rs1603636515
DMD	NM_004006.3:c.1966C>T	NP_003997.2:p.Gln656Ter	rs1603636537
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DMD	NM_004006.3:c.5363C>G		rs199774535

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.8656C>T	NP_003997.2:p.Gln2886Ter	rs201361100
DMD	NM_004006.3:c.2302C>T		rs201366610
DMD	NM_004006.3:c.6760A>T	NP_003997.2:p.Lys2254Ter	rs201818335
DMD	NM_004006.3:c.10813A>T	NP_003997.2:p.Lys3605Ter	rs2036700718
DMD	NM_004006.3:c.10797+2T>A		rs2036834025
DMD	NM_004006.3:c.10699C>T	NP_003997.2:p.Gln3567Ter	rs2036842297
DMD	NM_004006.3:c.10409del	NP_003997.2:p.Leu3470fs	rs2039782181
DMD	NM_004006.3:c.10297dup	NP_003997.2:p.Thr3433fs	rs2040217313
DMD	NM_004006.3:c.3008_3009del	NP_003997.2:p.Val1003fs	rs2040303197
DMD	NM_004006.3:c.2949+2T>C		rs2040709857
DMD	NM_004006.3:c.2945T>A	NP_003997.2:p.Leu982Ter	rs2040710629
DMD	NM_004006.3:c.10224-175_10238del		rs2040711393
DMD	NM_004006.3:c.10227del	NP_003997.2:p.Val3410fs	rs2040714340
DMD	NM_004006.3:c.10210dup	NP_003997.2:p.Asp3404fs	rs2040821014
DMD	NM_004006.3:c.9980_9981insTAG	NP_003997.2:p.Arg3327_Ser3328insSer	rs2041040360
DMD	NM_004006.3:c.9897_9898del	NP_003997.2:p.His3299fs	rs2041303734
DMD	NM_004006.3:c.9868C>T	NP_003997.2:p.Gln3290Ter	rs2041306808
DMD	NM_004006.3:c.9852G>A	NP_003997.2:p.Trp3284Ter	rs2041309291
DMD	NM_004006.3:c.2617dup	NP_003997.2:p.Cys873fs	rs2042972321
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DMD	NM_004006.3:c.2508dup	NP_003997.2:p.Asn837Ter	rs2042983140
DMD	NM_004006.3:c.2439del	NP_003997.2:p.Ile813fs	rs2042990064
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DMD	NM_004006.3:c.2253del	NP_003997.2:p.Lys752fs	rs2046034052
DMD	NM_004006.3:c.2227C>T	NP_003997.2:p.Gln743Ter	rs2046036327
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DMD	NM_004006.3:c.1357C>T	NP_003997.2:p.Gln453Ter	rs2057404615
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DMD	NM_004006.3:c.1283del	NP_003997.2:p.Asn428fs	rs2059643357
DMD	NM_004006.3:c.1238C>A	NP_003997.2:p.Ser413Ter	rs2059646164
DMD	NM_004006.3:c.1087C>T	NP_003997.2:p.Gln363Ter	rs2059695805
DMD	NM_004006.3:c.976G>T	NP_003997.2:p.Glu326Ter	rs2059702093
DMD	NM_004006.3:c.832-2A>C		rs2063781031
DMD	NM_004006.3:c.811C>T	NP_003997.2:p.Gln271Ter	rs2063890662
DMD	NM_004006.3:c.803del	NP_003997.2:p.Leu268fs	rs2063891560
DMD	NM_004006.3:c.650-1G>A		rs2063903818
DMD	NM_004006.3:c.9014T>A	NP_003997.2:p.Leu3005Ter	rs2065150560
DMD	NM_004006.3:c.8821G>T	NP_003997.2:p.Glu2941Ter	rs2067953453
DMD	NM_004006.3:c.8756_8763del	NP_003997.2:p.Asn2919fs	rs2067963703
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GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.8596_8600del	NP_003997.2:p.Leu2866fs	rs2068038543
DMD	NM_004006.3:c.8483T>A	NP_003997.2:p.Leu2828Ter	rs2069915167
DMD	NM_004006.3:c.8395del	NP_003997.2:p.His2799fs	rs2069923072
DMD	NM_004006.3:c.8329A>T	NP_003997.2:p.Arg2777Ter	rs2071051447
DMD	NM_004006.3:c.488G>A	NP_003997.2:p.Trp163Ter	rs2077774135
DMD	NM_004006.3:c.402_408dup	NP_003997.2:p.Glu137delinsGlnGlnTer	rs2077781516
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DMD	NM_004006.3:c.187-2A>G		rs2080514041
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DMD	NM_004006.3:c.183dup	NP_003997.2:p.Leu62fs	rs2080978755
DMD	NM_004006.3:c.178del	NP_003997.2:p.Gln60fs	rs2080979223
DMD	NM_004006.3:c.131dup	NP_003997.2:p.Gln45fs	rs2080984178
DMD	NM_004006.3:c.7852A>T	NP_003997.2:p.Lys2618Ter	rs2082253437
DMD	NM_004006.3:c.7833T>A	NP_003997.2:p.Tyr2611Ter	rs2082254429
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DMD	NM_004006.3:c.7736T>A	NP_003997.2:p.Leu2579Ter	rs2082260036
DMD	NM_004006.3:c.7720C>T	NP_003997.2:p.Gln2574Ter	rs2082260996
DMD	NM_004006.3:c.7618_7619del	NP_003997.2:p.Lys2540fs	rs2086354048
DMD	NM_004006.3:c.7577dup	NP_003997.2:p.Leu2527fs	rs2086356460
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DMD	NM_004006.3:c.88del	NP_003997.2:p.Ser30fs	rs2093885666
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DMD	NM_004006.3:c.6253del	NP_003997.2:p.Trp2085fs	rs2097447453
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DMD	NM_004006.3:c.6175A>T	NP_003997.2:p.Lys2059Ter	rs2097447879
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DMD	NM_004006.3:c.5812G>T	NP_003997.2:p.Glu1938Ter	rs2097747311
DMD	NM_004006.3:c.5794C>T	NP_003997.2:p.Gln1932Ter	rs2097747388
DMD	NM_004006.3:c.5704_5707del	NP_003997.2:p.Ser1902fs	rs2097752909
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DMD	NM_004006.3:c.5521G>T	NP_003997.2:p.Glu1841Ter	rs2097762588
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DMD	NM_004006.3:c.4914_4915del	NP_003997.2:p.Lys1639fs	rs2097850041
DMD	NM_004006.3:c.4903G>T	NP_003997.2:p.Gly1635Ter	rs2097850128
DMD	NM_004006.3:c.4840G>T	NP_003997.2:p.Gly1614Ter	rs2097920506
DMD	NM_004006.3:c.4675-1G>A		rs2097921115

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.4649T>A	NP_003997.2:p.Leu1550Ter	rs2097958158
DMD	NM_004006.3:c.4600C>T	NP_003997.2:p.Gln1534Ter	rs2097958423
DMD	NM_004006.3:c.4519-2A>C		rs2097958830
DMD	NM_004006.3:c.4454T>A	NP_003997.2:p.Leu1485Ter	rs2097986340
DMD	NM_004006.3:c.4402G>T	NP_003997.2:p.Glu1468Ter	rs2097986793
DMD	NM_004006.3:c.4375del	NP_003997.2:p.Arg1459fs	rs2097987064
DMD	NM_004006.3:c.4351_4352insA	NP_003997.2:p.Leu1451fs	rs2097987198
DMD	NM_004006.3:c.4345-3C>G		rs2097987224
DMD	NM_004006.3:c.4342C>T	NP_003997.2:p.Gln1448Ter	rs2097990454
DMD	NM_004006.3:c.4271T>A	NP_003997.2:p.Leu1424Ter	rs2097991272
DMD	NM_004006.3:c.4003G>C	NP_003997.2:p.Val1335Leu	rs2098269145
DMD	NM_004006.3:c.3921+1_3921+2dup		rs2098280263
DMD	NM_004006.3:c.3898G>T	NP_003997.2:p.Glu1300Ter	rs2098280321
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DMD	NM_004006.3:c.3490A>T	NP_003997.2:p.Lys1164Ter	rs2098349160
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DMD	NM_004006.3:c.5255T>G	NP_003997.2:p.Leu1752Ter	rs373428963
DMD	NM_004006.3:c.6072T>A		rs373804251
DMD	NM_004006.3:c.693C>G	NP_003997.2:p.Tyr231Ter	rs374222301
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DMD	NM_004006.2:c.9G>A	NP_003997.2:p.Trp3Ter	rs398122853
DMD	NM_004006.2:c.10033C>T	NP_003997.2:p.Arg3345Ter	rs398123827
DMD	NM_004006.3:c.10086+1G>T		rs398123828
DMD	NM_004006.2:c.10086+1G>A		rs398123828
DMD	NM_004006.3:c.1012G>T		rs398123830
DMD	NM_004006.2:c.10171C>T	NP_003997.2:p.Arg3391Ter	rs398123832
DMD	NM_004006.3:c.10223+1G>C		rs398123834
DMD	NM_004006.2:c.10223+1G>A		rs398123834
DMD	NM_004006.3:c.10258_10259insT		rs398123835
DMD	NM_004006.3:c.10447_10448del		rs398123837
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DMD	NM_004006.2:c.1261C>T	NP_003997.2:p.Gln421Ter	rs398123852
DMD	NM_004006.3:c.1286C>G		rs398123853
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DMD	NM_004006.3:c.1341_1342dup	NP_003997.2:p.Val448fs	rs398123856
DMD	NM_004006.3:c.1371del		rs398123857
DMD	NM_004006.3:c.137_138dup		rs398123859
DMD	NM_004006.3:c.1465C>T		rs398123861
DMD	NM_004006.2:c.1482+1G>T		rs398123862
DMD	NM_004006.3:c.1529_1530del	NP_003997.2:p.Leu510fs	rs398123863

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.1734dup	NP_003997.2:p.Glu579fs	rs398123866
DMD	NM_004006.2:c.1734del	NP_003997.2:p.Glu579fs	rs398123866
DMD	NM_004006.3:c.1886C>A		rs398123867
DMD	NM_004006.2:c.1990C>T	NP_003997.2:p.Gln664Ter	rs398123870
DMD	NM_004006.3:c.2032C>T		rs398123872
DMD	NM_004006.3:c.2125del		rs398123875
DMD	NM_004006.2:c.2281_2285del	NP_003997.2:p.Glu761fs	rs398123881
DMD	NM_004006.3:c.2294_2297del		rs398123882
DMD	NM_004006.2:c.2332C>T	NP_003997.2:p.Gln778Ter	rs398123883
DMD	NM_004006.3:c.2380+1G>C		rs398123884
DMD	NM_004006.3:c.2380+1G>A		rs398123884
DMD	NM_004006.3:c.2381-1G>T		rs398123887
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DMD	NM_004006.3:c.2436G>A		rs398123889
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DMD	NM_004006.3:c.2547del		rs398123895
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DMD	NM_004006.3:c.264+5G>A		rs398123902
DMD	NM_004006.2:c.2650C>T	NP_003997.2:p.Gln884Ter	rs398123903
DMD	NM_004006.2:c.2758C>T	NP_003997.2:p.Gln920Ter	rs398123905
DMD	NM_004006.3:c.2803+1G>T		rs398123908
DMD	NM_004006.2:c.2804-1G>T		rs398123909
DMD	NM_004006.2:c.2804-1G>A		rs398123909
DMD	NM_004006.3:c.2804-1G>C		rs398123909
DMD	NM_004006.3:c.2816T>A		rs398123910
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DMD	NM_004006.3:c.3076G>T		rs398123920
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DMD	NM_004006.3:c.3276+1G>A		rs398123934
DMD	NM_004006.3:c.3276+1G>C		rs398123934
DMD	NM_004006.2:c.3295C>T	NP_003997.2:p.Gln1099Ter	rs398123935
DMD	NM_004006.3:c.336G>A		rs398123936
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DMD	NM_004006.3:c.412_413del		rs398123949
DMD	NM_004006.3:c.4315del	NP_003997.2:p.Arg1439fs	rs398123950
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DMD	NM_004006.2:c.434G>C	NP_003997.2:p.Arg145Pro	rs398123952

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.4405C>T		rs398123954
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DMD	NM_004006.2:c.4518+5G>A		rs398123960
DMD	NM_004006.3:c.4534_4535del		rs398123961
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DMD	NM_004006.3:c.4843A>T		rs398123969
DMD	NM_004006.2:c.4902_4908del	NP_003997.2:p.Gly1635fs	rs398123971
DMD	NM_004006.2:c.4996C>T	NP_003997.2:p.Arg1666Ter	rs398123973
DMD	ATTCATT		rs398123978
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DMD	NM_004006.2:c.5134C>T	NP_003997.2:p.Gln1712Ter	rs398123980
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DMD	NM_004006.2:c.531-2A>G		rs398123985
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DMD	NM_004006.2:c.5773G>T	NP_003997.2:p.Glu1925Ter	rs398123997
DMD	NM_004006.2:c.583C>T	NP_003997.2:p.Arg195Ter	rs398123999
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DMD	NM_004006.2:c.6283C>T	NP_003997.2:p.Arg2095Ter	rs398124008
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DMD	NM_004006.3:c.676_678del		rs398124034
DMD	NM_004006.3:c.6906G>A		rs398124036
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DMD	NM_004006.3:c.6986_6987insA		rs398124040
DMD	NM_004006.3:c.7189C>T		rs398124042
DMD	NM_004006.3:c.7309+1G>A		rs398124044
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DMD	NM_004006.2:c.7657C>T	NP_003997.2:p.Arg2553Ter	rs398124050
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DMD	NM_004006.3:c.7682G>A		rs398124052
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DMD	NM_004006.3:c.7894C>T		rs398124058
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DMD	NM_004006.3:c.8069T>G		rs398124061
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DMD	NM_004006.2:c.8443C>T	NP_003997.2:p.Gln2815Ter	rs398124072
DMD	NM_004006.2:c.8608C>T	NP_003997.2:p.Arg2870Ter	rs398124074
DMD	NM_004006.3:c.8652_8653del		rs398124075
DMD	NM_004006.3:c.8912_8913dup	NP_003997.2:p.Gln2972fs	rs398124078
DMD	NM_004006.3:c.8908_8909TC[2]	NP_003997.2:p.Leu2971fs	rs398124078
DMD	NM_004006.3:c.9125del		rs398124080

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.9337C>T	NP_003997.2:p.Arg3113Ter	rs398124092
DMD	NM_004006.3:c.9361+1G>A		rs398124094
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DMD	NM_004006.3:c.9564-1G>A		rs398124096
DMD	NM_004006.3:c.9650-2A>G		rs398124100
DMD	NM_004006.3:c.9767_9768insG		rs398124103
DMD	NM_004006.3:c.9854_9863del		rs398124105
DMD	NM_004006.2:c.9862G>T	NP_003997.2:p.Glu3288Ter	rs398124106
DMD	NM_004006.3:c.3940C>T	NP_003997.2:p.Arg1314Ter	rs5030730
DMD	NM_004006.2:c.9094G>T	NP_003997.2:p.Glu3032Ter	rs72466562
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DMD	NM_004006.3:c.4806A>T		rs727503830
DMD	NM_004006.3:c.3532G>T		rs727503836
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DMD	NM_004006.3:c.2755A>T		rs727503850
DMD	NM_004006.3:c.2484T>G		rs727503858
DMD	NM_004006.2:c.883C>T	NP_003997.2:p.Arg295Ter	rs727503864
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DMD	NM_004006.3:c.2642C>A	NP_003997.2:p.Ser881Ter	rs747937552
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DMD	NM_004006.3:c.3892G>T	NP_003997.2:p.Gly1298Ter	rs750349613
DMD	NM_004006.3:c.2485C>T		rs753662330
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DMD	NM_004006.3:c.6982A>T		rs754896795
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DMD	NM_004006.3:c.4495C>T		rs755686224
DMD	NM_004006.2:c.2797C>T	NP_003997.2:p.Gln933Ter	rs756949497
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DMD	NM_004006.2:c.2368C>T	NP_003997.2:p.Gln790Ter	rs762860653
DMD	NM_004006.3:c.1594C>T		rs763936813
DMD	NM_004006.3:c.9563+5G>C		rs764119686
DMD	NM_004006.2:c.5110G>T	NP_003997.2:p.Glu1704Ter	rs765445866
DMD	NM_004006.3:c.2926G>T	NP_003997.2:p.Glu976Ter	rs767387334
DMD	NM_004006.3:c.1819A>T	NP_003997.2:p.Lys607Ter	rs767698069
DMD	NM_004006.3:c.2530C>T	NP_003997.2:p.Gln844Ter	rs768892830
DMD	NM_004006.3:c.7170C>G		rs770183212
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DMD	NM_004006.3:c.3824T>A	NP_003997.2:p.Leu1275Ter	rs770934386
DMD	NM_004006.3:c.4684A>T	NP_003997.2:p.Arg1562Ter	rs771425504
DMD	NM_004006.3:c.4350dup	NP_003997.2:p.Leu1451fs	rs772220893
DMD	NM_004006.3:c.6217A>T	NP_003997.2:p.Lys2073Ter	rs773372553
DMD	NM_004006.2:c.5851C>T	NP_003997.2:p.Gln1951Ter	rs773643220
DMD	NM_004006.3:c.1603-2A>C		rs773779441
DMD	NM_004006.3:c.1405G>T	NP_003997.2:p.Glu469Ter	rs774163062
DMD	NM_004006.3:c.5350G>T		rs777864641
DMD	NM_004006.2:c.6502G>T	NP_003997.2:p.Glu2168Ter	rs779739455
DMD	NM_004006.2:c.1093C>T	NP_003997.2:p.Gln365Ter	rs794726993

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.2479G>T		rs794727323
DMD	NM_004006.3:c.2804-2A>T		rs794727357
DMD	NM_004006.2:c.2804-2A>C		rs794727357
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DMD	NM_004006.3:c.3838A>T		rs794727463
DMD	NM_004006.3:c.133C>T		rs794727499
DMD	NM_004006.3:c.4315A>T		rs794727550
DMD	NM_004006.3:c.4523del		rs794727567
DMD	NM_004006.3:c.4675-2A>G		rs794727575
DMD	NM_004006.3:c.5697dup	NP_003997.2:p.Leu1900fs	rs794727661
DMD	NM_004006.3:c.5697del		rs794727661
DMD	NM_004006.3:c.5868G>A		rs794727666
DMD	NM_004006.3:c.6238del		rs794727672
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DMD	NM_004006.3:c.10554-2A>G		rs794727890
DMD	NM_004006.2:c.2623-1G>T		rs794729005
DMD	NM_004006.3:c.5383G>T	NP_003997.2:p.Glu1795Ter	rs79541196
DMD	:c.14_15delinsT		rs796065325
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DMD	NM_004006.3:c.3603+2dup		rs796523999
DMD	NM_004006.3:c.6673_6674insGTTT		rs797044743
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DMD	NM_004006.2:c.7247dup	NP_003997.2:p.Leu2416fs	rs797044764
DMD	NM_004006.2:c.10133del	NP_003997.2:p.Asn3378fs	rs863224975
DMD	NM_004006.2:c.10412T>G	NP_003997.2:p.Leu3471Ter	rs863224976
DMD	NM_004006.2:c.10504G>T	NP_003997.2:p.Glu3502Ter	rs863224977
DMD	NM_004006.2:c.1150-2del		rs863224978
DMD	NM_004006.2:c.1324C>T	NP_003997.2:p.Gln442Ter	rs863224979
DMD	NM_004006.2:c.1331+1G>A		rs863224980
DMD	NM_004006.2:c.1388G>A	NP_003997.2:p.Trp463Ter	rs863224981
DMD	NM_004006.2:c.1483-1G>C		rs863224982
DMD	NM_004006.2:c.1663C>T	NP_003997.2:p.Gln555Ter	rs863224983
DMD	NM_004006.2:c.1683G>A	NP_003997.2:p.Trp561Ter	rs863224984

GEN	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.2407C>T	NP_003997.2:p.Gln803Ter	rs863224986
DMD	NM_004006.2:c.2611A>T	NP_003997.2:p.Lys871Ter	rs863224987
DMD	NM_004006.2:c.2623-3C>G		rs863224988
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DMD	NM_004006.2:c.280del	NP_003997.2:p.Ile94fs	rs863224990
DMD	NM_004006.2:c.282dup	NP_003997.2:p.Gly95fs	rs863224991
DMD	NM_004006.2:c.2991C>G	NP_003997.2:p.Tyr997Ter	rs863224992
DMD	NM_004006.2:c.3427C>T	NP_003997.2:p.Gln1143Ter	rs863224993
DMD	NM_004006.2:c.3433-5_3434del		rs863224994
DMD	NM_004006.2:c.355C>T	NP_003997.2:p.Gln119Ter	rs863224995
DMD	NM_004006.2:c.358-2A>G		rs863224996
DMD	NM_004006.2:c.4240C>T	NP_003997.2:p.Gln1414Ter	rs863224997
DMD	NM_004006.2:c.4606G>T	NP_003997.2:p.Glu1536Ter	rs863224998
DMD	NM_004006.2:c.4729C>T	NP_003997.2:p.Arg1577Ter	rs863224999
DMD	NM_004006.2:c.4918del	NP_003997.2:p.Thr1640fs	rs863225000
DMD	NM_004006.2:c.5131C>T	NP_003997.2:p.Gln1711Ter	rs863225001
DMD	NM_004006.2:c.5461G>T	NP_003997.2:p.Glu1821Ter	rs863225002
DMD	NM_004006.2:c.5602_5605del	NP_003997.2:p.Arg1868fs	rs863225003
DMD	NM_004006.2:c.5641C>T	NP_003997.2:p.Gln1881Ter	rs863225004
DMD	NM_004006.2:c.5917C>T	NP_003997.2:p.Gln1973Ter	rs863225005
DMD	NM_004006.2:c.6128_6131del	NP_003997.2:p.Asp2043fs	rs863225006
DMD	c.6611dup	NP_003997.2:p.Arg2205GluTer18	rs863225007
DMD	NM_004006.3:c.6610_6611del		rs863225007
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DMD	NM_004006.2:c.8027+2T>A		rs863225010
DMD	NM_004006.2:c.8038C>T	NP_003997.2:p.Arg2680Ter	rs863225011
DMD	NM_004006.2:c.8357G>A	NP_003997.2:p.Trp2786Ter	rs863225012
DMD	NM_004006.2:c.8390+2T>C		rs863225013
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DMD	NM_004006.2:c.9551dup	NP_003997.2:p.Asn3184fs	rs863225017
DMD	NM_004006.2:c.9938_9941dup	NP_003997.2:p.Asn3314delinsLysTer	rs863225018
DMD	NM_004006.3:c.5026G>T	NP_003997.2:p.Glu1676Ter	rs868267519
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DMD	NM_004006.3:c.10509_10510del		rs878854366
DMD	NM_004006.2:c.1504C>T	NP_003997.2:p.Gln502Ter	rs878854618
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DMD	NM_004006.3:c.9471_9474del		rs886039532
DMD	NM_004006.2:c.9148C>T	NP_003997.2:p.Gln3050Ter	rs886039533
DMD	NM_004006.2:c.6762+1G>T		rs886039534
DMD	NM_004006.2:c.5314A>T	NP_003997.2:p.Lys1772Ter	rs886039535
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DMD	NM_004006.3:c.9503del		rs886039681
DMD	NM_004006.2:c.8459G>A	NP_003997.2:p.Trp2820Ter	rs886039785
DMD	NM_004006.2:c.1865C>A	NP_003997.2:p.Ser622Ter	rs886041344

GEN	HGVSc	HGVSp	dbSNP
DMD	NM_004006.2:c.1865C>G	NP_003997.2:p.Ser622Ter	rs886041344
DMD	NM_004006.2:c.6290+1G>C		rs886041527
DMD	GTAGGTGTAAGAAAACCTTTAAAGAACTTTAAA		rs886041629
DMD	NM_004006.3:c.5276_5277del		rs886041653
DMD	NM_004006.2:c.1288A>T	NP_003997.2:p.Arg430Ter	rs886041802
DMD	NM_004006.3:c.5032C>T		rs886042154
DMD	NM_004006.3:c.5089C>T		rs886042347
DMD	NM_004006.3:c.2603del		rs886042348
DMD	NM_004006.3:c.2512C>T		rs886042351
DMD	NM_004006.3:c.8105_8111dup		rs886042373
DMD	NM_004006.3:c.2316_2317del		rs886042437
DMD	NM_004006.2:c.10477C>T	NP_003997.2:p.Gln3493Ter	rs886042495
DMD	NM_004006.3:c.1417A>T		rs886042499
DMD	NM_004006.3:c.93+1G>T		rs886042604
DMD	NM_004006.2:c.93+1G>C		rs886042604
DMD	NM_004006.2:c.93+1G>A		rs886042604
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DMD	NM_004006.3:c.2168+2T>G		rs886042618
DMD	NM_004006.3:c.10279C>T		rs886042691
DMD	NM_004006.2:c.5140G>T	NP_003997.2:p.Glu1714Ter	rs886042747
DMD	NM_004006.3:c.10098AGA[1]	NP_003997.2:p.Glu3367del	rs886042840
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DMD	NM_004006.3:c.457C>T		rs886042983
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DMD	NM_004006.3:c.3238del		rs886043052
DMD	NM_004006.2:c.10361del	NP_003997.2:p.Tyr3454fs	rs886043084
DMD	NM_004006.3:c.6393_6394insCA		rs886043157
DMD	NM_004006.2:c.1150-1G>A		rs886043251
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DMD	NM_004006.3:c.4550_4556del		rs886043348
DMD	NM_004006.2:c.10453dup	NP_003997.2:p.Leu3485Profs	rs886043375
DMD	NM_004006.3:c.10453del		rs886043375
DMD	NM_004006.3:c.1530dup		rs886043376
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DMD	NM_004006.3:c.4174C>T		rs886043496
DMD	NM_004006.2:c.9938G>T	NP_003997.2:p.Cys3313Phe	rs886043597
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DMD	NM_004006.3:c.2052_2053del		rs886043699
DMD	NM_004006.3:c.10572T>A		rs886043817
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DMD	NM_004006.3:c.10651_10652insCC		rs886044325
DMD	NM_004006.3:c.4312C>T		rs886044402
DMD	NM_004006.3:c.3413G>A		rs886044406
DMD	NM_004006.2:c.170T>G	NP_003997.2:p.Leu57Arg	rs886044431
DMD	NM_004006.3:c.1534_1535insTC		rs886044440

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DMD	NM_004006.2:c.5344G>T	NP_003997.2:p.Glu1782Ter	rs886044455
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DMD	NM_004006.3:c.4845+1G>T		rs886044501
DMD	NM_004006.2:c.9975-2A>C		rs886044502
DMD	NM_004006.2:c.358-1G>A		rs886044582
DMD	NM_004006.3:c.1128_1129insA		rs886044624
DMD	NM_004006.3:c.9527A>G	NP_003997.2:p.Asp3176Gly	rs886044916
DMD	NM_004006.3:c.10429C>T	NP_003997.2:p.Gln3477Ter	rs895755247
DMD	NM_004006.3:c.4057G>T	NP_003997.2:p.Glu1353Ter	rs907237595
F2	NM_000506.3:c.1139T>C	NP_000497.1:p.Met380Thr	rs121918481
F2	NM_000506.3:c.*97G>A		rs1799963
F5	NM_000130.4:c.1000A>G	NP_000121.2:p.Arg334Gly	rs118203905
F5	NM_000130.4:c.1001G>C	NP_000121.2:p.Arg334Thr	rs118203906
F5	NM_000130.4:c.1601G>A	NP_000121.2:p.Arg534Gln	rs6025
FAH	NM_000137.2:c.615del	NP_000128.1:p.Phe205LeufsTer2	rs1057517084
FAH	NM_000137.2:c.615dup	NP_000128.1:p.Val206CysfsTer18	rs1057517084
FAH	NM_000137.2:c.456G>A	NP_000128.1:p.Trp152Ter	rs370686447
FAH	NM_000137.2:c.554-1G>T		rs80338895
FAH	NM_000137.2:c.782C>T	NP_000128.1:p.Pro261Leu	rs80338898
FAH	NM_000137.2:c.1009G>A	NP_000128.1:p.Gly337Ser	rs80338900
FAH	NM_000137.2:c.1062+5G>A		rs80338901
FSHR	NM_000145.3:c.2039G>A	NP_000136.2:p.Ser680Asn	rs6166
G6PC1	NM_000151.2:c.247C>T	NP_000142.1:p.Arg83Cys	rs1801175
G6PC1	NM_000151.2:c.248G>A	NP_000142.1:p.Arg83His	rs1801176
G6PC1	NM_000151.2:c.508C>T	NP_000142.1:p.Arg170Ter	rs373345919
G6PC1	NM_000151.2:c.230+4A>G		rs587776757
G6PC1	NM_000151.2:c.189G>A	NP_000142.1:p.Trp63Ter	rs764920787
G6PC1	NM_000151.2:c.79delC	NP_000142.1:p.Gln27Argfs	rs80356479
G6PC1	NM_000151.2:c.562G>A	NP_000142.1:p.Gly188Ser	rs80356482
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GALT	NM_000155.3:c.197C>T	NP_000146.2:p.Pro66Leu	rs111033656
GALT	NM_000155.3:c.413C>T	NP_000146.2:p.Thr138Met	rs111033686
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GALT	NM_000155.3:c.442C>T	NP_000146.2:p.Arg148Trp	rs111033693
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GALT	NM_000155.3:c.602G>A	NP_000146.2:p.Arg201His	rs111033735
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GALT	NM_000155.3:c.601C>T	NP_000146.2:p.Arg201Cys	rs111033739
GALT	NM_000155.3:c.667C>T	NP_000146.2:p.Arg223Cys	rs111033750
GALT	NM_000155.3:c.667C>A	NP_000146.2:p.Arg223Ser	rs111033750
GALT	NM_000155.3:c.692G>A	NP_000146.2:p.Arg231His	rs111033754
GALT	NM_000155.3:c.821-2A>G		rs111033767
GALT	NM_000155.3:c.855G>T	NP_000146.2:p.Lys285Asn	rs111033773
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GALT	NM_000155.3:c.957C>A	NP_000146.2:p.His319Gln	rs111033792
GALT	NM_000155.3:c.997C>G	NP_000146.2:p.Arg333Gly	rs111033800
GALT	NM_000155.3:c.997C>T	NP_000146.2:p.Arg333Trp	rs111033800
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GALT	NM_000155.3:c.1006A>T	NP_000146.2:p.Met336Leu	rs111033810
GALT	NM_000155.3:c.1030C>A	NP_000146.2:p.Gln344Lys	rs111033814
GALT	NM_000155.3:c.1138T>C	NP_000146.2:p.Ter380ArgextTer49	rs111033824
GALT	NM_000155.3:c.652C>G	NP_000146.2:p.Leu218Val	rs2070075
GALT	NM_000155.3:c.563A>G	NP_000146.2:p.Gln188Arg	rs75391579
GALT	NM_000155.3:c.776G>A	NP_000146.2:p.Arg259Gln	rs886042070
GBA	NM_001005741.2:c.115+1G>A		rs104886460
GBA	NM_001005741.2:c.1342G>C	NP_001005741.1:p.Asp448His	rs1064651
GBA	NM_001005741.2:c.1361C>G	NP_001005741.1:p.Pro454Arg	rs121908295
GBA	NM_001005741.2:c.535G>C	NP_001005741.1:p.Asp179His	rs147138516
GBA	NM_001005741.2:c.1296G>A	NP_001005741.1:p.Trp432Ter	rs1484043383
GBA	NM_001005741.2:c.886C>T	NP_001005741.1:p.Arg296Ter	rs1553217626
GBA	NM_001005741.2:c.1093G>A	NP_001005741.1:p.Glu365Lys	rs2230288
GBA	NM_001005741.2:c.680A>G	NP_001005741.1:p.Asn227Ser	rs364897
GBA	NM_001005741.2:c.882T>G	NP_001005741.1:p.His294Gln	rs367968666
GBA	NM_001005741.2:c.681T>G	NP_001005741.1:p.Asn227Lys	rs381418
GBA	NM_001005741.2:c.84dup	NP_001005741.1:p.Leu29Alafs	rs387906315
GBA	NM_001005741.2:c.721G>A	NP_001005741.1:p.Gly241Arg	rs409652
GBA	NM_001005741.2:c.1448T>C	NP_001005741.1:p.Leu483Pro	rs421016
GBA	NM_001005741.2:c.764T>A	NP_001005741.1:p.Phe255Tyr	rs74500255
GBA	NM_001005741.2:c.1444G>A	NP_001005741.1:p.Asp482Asn	rs75671029
GBA	NM_001005741.2:c.1604G>A	NP_001005741.1:p.Arg535His	rs75822236
GBA	NM_001005741.2:c.1085C>T	NP_001005741.1:p.Thr362Ile	rs76539814
GBA	NM_001005741.2:c.1226A>G	NP_001005741.1:p.Asn409Ser	rs76763715
GBA	NM_001005741.2:c.1226A>C	NP_001005741.1:p.Asn409Thr	rs76763715
GBA	NM_001005741.2:c.521A>G	NP_001005741.1:p.Tyr174Cys	rs781152868
GBA	NM_001005741.2:c.509G>A	NP_001005741.1:p.Arg170His	rs80356763
GBA	NM_001005741.2:c.1297G>T	NP_001005741.1:p.Val433Leu	rs80356769
GBA	NM_001005741.2:c.1504C>T	NP_001005741.1:p.Arg502Cys	rs80356771
GCDH	NM_000159.3:c.1262C>T	NP_000150.1:p.Ala421Val	rs121434367
GCDH	NM_000159.3:c.1204C>T	NP_000150.1:p.Arg402Trp	rs121434369
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GCDH	NM_000159.3:c.680G>C	NP_000150.1:p.Arg227Pro	rs121434373
GCDH	NM_000159.3:c.463T>C	NP_000150.1:p.Tyr155His	rs1482995062
GCDH	NM_000159.3:c.770G>A	NP_000150.1:p.Arg257Gln	rs751583656
GCDH	NM_000159.3:c.1156C>T	NP_000150.1:p.Arg386Ter	rs752127949
GCDH	NM_000159.3:c.737C>T	NP_000150.1:p.Ser246Leu	rs754312389
GCDH	NM_000159.3:c.764C>T	NP_000150.1:p.Ser255Leu	rs758503371
GJB2	NM_004004.5:c.230G>A	NP_003995.2:p.Trp77Ter	rs104894395
GJB2	NM_004004.5:c.71G>A	NP_003995.2:p.Trp24Ter	rs104894396
GJB2	NM_004004.5:c.229T>C	NP_003995.2:p.Trp77Arg	rs104894397
GJB2	NM_004004.5:c.139G>T	NP_003995.2:p.Glu47Ter	rs104894398
GJB2	NM_004004.5:c.34G>C	NP_003995.2:p.Gly12Arg	rs104894408
GJB2	NM_004004.5:c.95G>A	NP_003995.2:p.Arg32His	rs111033190
GJB2	NM_004004.5:c.299_300del	NP_003995.2:p.His100ArgfsTer14	rs111033204
GJB2	NM_004004.5:c.313_326del	NP_003995.2:p.Lys105_Gly109del	rs111033253
GJB2	NM_004004.5:c.1A>G	NP_003995.2:p.Met1?	rs111033293
GJB2	NM_004004.5:c.617A>G	NP_003995.2:p.Asn206Ser	rs111033294
GJB2	NM_004004.5:c.169C>T	NP_003995.2:p.Gln57Ter	rs111033297
GJB2	NM_004004.5:c.94C>A	NP_003995.2:p.Arg32Ser	rs371024165
GJB2	NM_004004.5:c.370C>T	NP_003995.2:p.Gln124Ter	rs397516874

GEN	HGVSc	HGVSp	dbSNP
GJB2	NM_004004.5:c.334_335del	NP_003995.2:p.Lys112GlufsTer2	rs756484720
GJB2	NM_004004.5:c.416G>A	NP_003995.2:p.Ser139Asn	rs76434661
GJB2	NM_004004.5:c.439G>A	NP_003995.2:p.Glu147Lys	rs767178508
GJB2	NM_004004.5:c.35delG	NP_003995.2:p.Gly12Valfs	rs80338939
GJB2	NM_004004.5:c.-23+1G>A		rs80338940
GJB2	NM_004004.5:c.56G>C	NP_003995.2:p.Ser19Thr	rs80338941
GJB2	NM_004004.5:c.167delT	NP_003995.2:p.Leu56Argfs	rs80338942
GJB2	NM_004004.5:c.231G>A	NP_003995.2:p.Trp77Ter	rs80338944
GJB2	NM_004004.5:c.269T>C	NP_003995.2:p.Leu90Pro	rs80338945
GJB2	NM_004004.5:c.358_360delGAG	NP_003995.2:p.Glu120del	rs80338947
GJB2	NM_004004.5:c.427C>T	NP_003995.2:p.Arg143Trp	rs80338948
GJB2	NM_004004.5:c.551G>C	NP_003995.2:p.Arg184Pro	rs80338950
GJB2	NM_004004.5:c.551G>A	NP_003995.2:p.Arg184Gln	rs80338950
GLA	NM_000169.2:c.988C>T	NP_000160.1:p.Gln330Ter	
GLA	NM_000169.2:c.1078G>A	NP_000160.1:p.Gly360Ser	
GLA	NM_000169.2:c.838C>A		
GLA	NM_000169.2:c.511G>C		
GLA	NM_000169.2:c.427G>A	NP_000160.1:p.Ala143Thr	rs104894845
GLA	NM_000169.2:c.281G>A	NP_000160.1:p.Cys94Tyr	rs113173389
GLA	NM_000169.2:c.463G>C	NP_000160.1:p.Asp155His	rs113419388
GLA	NM_000169.2:c.865A>T	NP_000160.1:p.Ile289Phe	rs140329381
GLA	NM_000169.2:c.404C>T	NP_000160.1:p.Ala135Val	rs1569304221
GLA	NM_000169.2:c.644A>G	NP_000160.1:p.Asn215Ser	rs28935197
GLA	NM_000169.2:c.1025G>A	NP_000160.1:p.Arg342Gln	rs28935493
GLA	NM_000169.2:c.1019_1020insA	NP_000160.1:p.Trp340Terfs	rs398123197
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GLA	NM_000169.2:c.277G>A	NP_000160.1:p.Asp93Asn	rs869312270
GLA	NM_000169.2:c.717_718delAA	NP_000160.1:p.Lys240fs	rs869312389
GLB1	NM_000404.3:c.602G>A	NP_000395.2:p.Arg201His	rs189115557
GLB1	NM_000404.2:c.442C>T	NP_000395.2:p.Arg148Cys	rs192732174
GLB1	NM_000404.2:c.808T>G	NP_000395.2:p.Tyr270Asp	rs376663785
GLB1	NM_000404.2:c.464T>G	NP_000395.2:p.Leu155Arg	rs376710410
GLB1	NM_000404.2:c.75+2dup		rs587776525
GLB1	NM_000404.2:c.1369C>T	NP_000395.2:p.Arg457Ter	rs72555359
GLB1	NM_000404.2:c.601C>T	NP_000395.2:p.Arg201Cys	rs72555360
GLB1	NM_000404.2:c.622C>T	NP_000395.2:p.Arg208Cys	rs72555366
GLB1	NM_000404.2:c.202C>T	NP_000395.2:p.Arg68Trp	rs72555370
GLB1	NM_000404.2:c.1771T>A	NP_000395.2:p.Tyr591Asn	rs72555373
GLB1	NM_000404.2:c.176G>A	NP_000395.2:p.Arg59His	rs72555392
GLB1	NM_000404.2:c.716C>T	NP_000395.2:p.Thr239Met	rs746766232
GNPTAB	NM_024312.4:c.2864C>T	NP_077288.2:p.Ala955Val	rs138390866
GNPTAB	NM_024312.4:c.1090C>T	NP_077288.2:p.Arg364Ter	rs200646278
GNPTAB	NM_024312.4:c.1000C>T	NP_077288.2:p.Arg334Ter	rs281864969
GNPTAB	NM_024312.4:c.1514G>A	NP_077288.2:p.Cys505Tyr	rs281864980
GNPTAB	NM_024312.4:c.2866C>T	NP_077288.2:p.His956Tyr	rs281865004
GNPTAB	NM_024312.4:c.2715+1G>A		rs281865031
GNPTAB	NM_024312.4:c.3503_3504delTC	NP_077288.2:p.Leu1168Glnfs	rs34002892
GNPTAB	NM_024312.4:c.10A>C	NP_077288.2:p.Lys4Gln	rs34159654
GNPTAB	NM_024312.4:c.3335+6T>G		rs34788341
GNPTAB	NM_024312.4:c.569A>T	NP_077288.2:p.Asp190Val	rs34946266
GNPTAB	NM_024312.4:c.1580delC	NP_077288.2:p.Ser527Ser=fs	rs36007394
GNPTAB	NM_024312.4:c.1399del	NP_077288.2:p.Asp467IlefsTer33	rs397507448

GEN	HGVSc	HGVSp	dbSNP
GNPTAB	NM_024312.4:c.136C>T	NP_077288.2:p.Arg46Ter	rs78347057
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HADHA	NM_000182.4:c.1132C>T	NP_000173.2:p.Gln378Ter	rs137852770
HADHA	NM_000182.4:c.1678C>T	NP_000173.2:p.Arg560Ter	rs137852771
HADHA	NM_000182.4:c.1025T>C	NP_000173.2:p.Leu342Pro	rs137852772
HADHA	NM_000182.4:c.845T>A	NP_000173.2:p.Val282Asp	rs137852773
HADHA	NM_000182.4:c.914T>A	NP_000173.2:p.Ile305Asn	rs137852774
HADHA	NM_000182.4:c.871C>T	NP_000173.2:p.Arg291Ter	rs137852775
HADHA	NM_000182.4:c.157C>T	NP_000173.2:p.Arg53Ter	rs147103714
HADHA	NM_000182.4:c.919-2A>G		rs200017313
HADHA	NM_000182.4:c.180+3A>G		rs781222705
HADHA	NM_000182.4:c.180+1G>A		rs786205088
HBB	NM_000518.4:c.93G>T	NP_000509.1:p.Arg31Ser	rs1135071
HBB	NM_000518.4:c.118C>T	NP_000509.1:p.Gln40Ter	rs11549407
HBB	NM_000518.4:c.20A>T	NP_000509.1:p.Glu7Val	rs334
HBB	NM_000518.4:c.316-3C>T		rs33913413
HBB	NM_000518.4:c.316-3C>A		rs33913413
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HBB	NM_000518.4:c.19G>A	NP_000509.1:p.Glu7Lys	rs33930165
HBB	NM_000518.4:c.315+1G>A		rs33945777
HBB	NM_000518.4:c.364G>A	NP_000509.1:p.Glu122Lys	rs33946267
HBB	NM_000518.4:c.364G>T	NP_000509.1:p.Glu122Ter	rs33946267
HBB	NM_000518.4:c.79G>A	NP_000509.1:p.Glu27Lys	rs33950507
HBB	NM_000518.4:c.92+1G>A		rs33971440
HBB	NM_000518.4:c.93-21G>A		rs35004220
HBB	NM_000518.4:c.82G>T	NP_000509.1:p.Ala28Ser	rs35424040
HBB	NM_000518.4:c.c.92+6T>C		rs35724775
HBB	NM_000518.4:c.61G>A	NP_000509.1:p.Val21Met	rs35890959
HEXA	NM_000520.4:c.196C>T	p.Gln66Ter	rs1057516957
HEXA	NM_000520.4:c.805G>A	NP_000511.2:p.Gly269Ser	rs121907954
HEXA	NM_000520.4:c.1511G>A	NP_000511.2:p.Arg504His	rs121907955
HEXA	NM_000520.4:c.409C>T	NP_000511.2:p.Arg137Ter	rs121907962
HEXA	NM_000520.4:c.1A>C	p.Met1?	rs121907965
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HEXA	NM_000520.4:c.590A>C	NP_000511.2:p.Lys197Thr	rs121907973
HEXA	NM_000520.4:c.745C>T	NP_000511.2:p.Arg249Trp	rs138058578
HEXA	NM_000520.4:c.1421+1G>C		rs147324677
HEXA	NM_000520.4:c.709C>T	NP_000511.2:p.Gln237Ter	rs150675340
HEXA	NM_000520.4:c.533G>A	NP_000511.2:p.Arg178His	rs28941770
HEXA	NM_000520.4:c.1510C>T	NP_000511.2:p.Arg504Cys	rs28942071
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HEXA	NM_000520.4:c.1073+1G>A		rs76173977
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HEXA	NM_000520.4:c.2T>C	NP_000511.2:p.Met1?	rs786204721
HFE	NM_000410.3:c.845G>A	NP_000401.1:p.Cys282Tyr	rs1800562
CHRNE	NM_000080.3:c.1090del	NP_000071.1:p.Arg364GlyfsTer21	rs1156634884
CHRNE	NM_000080.3:c.488C>T	NP_000511.2:p.Ser163Leu	rs121909516
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CHRNE	NM_000080.3:c.103T>C	NP_000511.2:p.Tyr35His	rs144169073
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CHRNE	NM_000080.3:c.130dup	NP_000071.1:p.Glu44GlyfsTer3	rs762368691
CHRNE	NM_000080.3:c.1327delG	NP_000071.1:p.Glu443Lysfs	rs763258280
CHRNE	NM_000080.3:c.1353dup	NP_000071.1:p.Asn452GlufsTer4	rs773526895
CHRNE	NM_000080.3:c.971del	NP_000071.1:p.Ile324ThrfsTer61	rs879255562
IDUA	NM_000203.3:c.1205G>A	NP_000194.2:p.Trp402Ter	rs121965019
IDUA	NM_000203.3:c.208C>T	NP_000194.2:p.Gln70Ter	rs121965020
IDUA	NM_000203.4:c.192C>A	NP_000194.2:p.Tyr64Ter	rs121965022
IDUA	NM_000203.3:c.1861C>T	NP_000194.2:p.Arg621Ter	rs121965025
IDUA	NM_000203.3:c.1855C>T	NP_000194.2:p.Arg619Ter	rs121965031
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IDUA	c.46_57dup	NP_000194.2:p.Ser16_Ala19dup	rs398123260
IDUA	NM_000203.3:c.1874A>G	NP_000194.2:p.Tyr625Cys	rs587779401
IDUA	NM_000203.3:c.1882C>T	NP_000194.2:p.Arg628Ter	rs756572099
IDUA	NM_000203.3:c.223G>A	NP_000194.2:p.Ala75Thr	rs758452450
IDUA	NM_000203.3:c.1139A>G	NP_000194.2:p.Gln380Arg	rs762903007
IDUA	NM_000203.3:c.1029C>G	NP_000194.2:p.Tyr343Ter	rs764196171
IDUA	NM_000203.3:c.1898C>T	NP_000194.2:p.Ser633Leu	rs886043347
IKBKAP (ELP1)	NM_003640.3:c.2204+6T>C		rs111033171
IKBKAP (ELP1)	NM_003640.3:c.2087G>C	NP_003631.2:p.Arg696Pro	rs137853022
IKBKAP (ELP1)	NM_003640.3:c.2741C>T	NP_003631.2:p.Pro914Leu	rs28939712
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IL2RG	NM_000206.2:c.854G>A	NP_000197.1:p.Arg285Gln	rs111033617
IL2RG	NM_000206.2:c.664C>T	NP_000197.1:p.Arg222Cys	rs111033618
IL2RG	NM_000206.2:c.186T>A	NP_000197.1:p.Cys62Ter	rs111033619
IL2RG	NM_000206.2:c.341G>A	NP_000197.1:p.Gly114Asp	rs111033620
IL2RG	NM_000206.2:c.458T>A	NP_000197.1:p.Ile153Asn	rs111033621
IL2RG	NM_000206.2:c.343T>C	NP_000197.1:p.Cys115Arg	rs111033622
IL2RG	NM_000206.2:c.355A>T	NP_000197.1:p.Lys119Ter	rs137852507
IL2RG	NM_000206.2:c.865C>T	NP_000197.1:p.Arg289Ter	rs137852508
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IL2RG	NM_000206.2:c.878T>A	NP_000197.1:p.Leu293Gln	rs137852510
IL2RG	NM_000206.2:c.452T>C	NP_000197.1:p.Leu151Pro	rs137852511
IL2RG	NM_000206.2:c.270-1G>T		rs193922346
IL2RG	NM_000206.2:c.314A>G	NP_000197.1:p.Tyr105Cys	rs193922347
IL2RG	NM_000206.2:c.455T>C	NP_000197.1:p.Val152Ala	rs193922348
IL2RG	NM_000206.2:c.662T>C	NP_000197.1:p.Phe221Ser	rs193922349
IL2RG	NM_000206.2:c.710G>A	NP_000197.1:p.Trp237Ter	rs193922350
MCCC1	NM_020166.3:c.974T>G	NP_064551.3:p.Met325Arg	rs119103212
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MCCC1	NM_020166.4:c.1114C>T	NP_064551.3:p.Gln372Ter	rs544349961
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MCCC2	NM_022132.4:c.1015G>A	NP_071415.1:p.Val339Met	rs150591260
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MEFV	NM_000243.2:c.501G>C	NP_000234.1:p.Glu167Asp	rs104895079
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MEFV	NM_000243.2:c.2082G>A	NP_000234.1:p.Met694Ile	rs28940578
MEFV	NM_000243.2:c.2177T>C	NP_000234.1:p.Val726Ala	rs28940579
MEFV	NM_000243.2:c.2040G>A	NP_000234.1:p.Met680Ile	rs28940580
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MEFV	NM_000243.2:c.2040G>T	NP_000234.1:p.Met680Ile	rs28940580
MEFV	NM_000243.2:c.2080A>G	NP_000234.1:p.Met694Val	rs61752717
MTM1	NM_000252.2:c.566A>G	NP_000243.1:p.Asn189Ser	rs132630302
MTM1	NM_000252.2:c.1190A>G	NP_000243.1:p.Tyr397Cys	rs132630303
MTM1	NM_000252.2:c.721C>T	NP_000243.1:p.Arg241Cys	rs132630305
MTM1	NM_000252.2:c.670C>T	NP_000243.1:p.Arg224Ter	rs132630306
MTM1	NM_000252.2:c.469G>A	NP_000243.1:p.Glu157Lys	rs132630307
MTM1	NM_000252.2:c.1261-10A>G		rs397518445
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MTM1	NM_000252.2:c.1054-5G>C		rs398123265
MTM1	NM_000252.2:c.122A>G	NP_000243.1:p.Glu41Gly	rs398123266
MTM1	NM_000252.2:c.1306_1310dupCCT	NP_000243.1:p.Phe438Leufs	rs398123267
MTM1	NM_000252.2:c.1357_1358delCC	NP_000243.1:p.Pro453Tyrfs	rs398123268
MTM1	NM_000252.2:c.1369G>A	NP_000243.1:p.Glu457Lys	rs398123269
MTM1	NM_000252.2:c.142_143delGA	NP_000243.1:p.Glu48Serfs	rs398123270
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MTM1	NM_000252.2:c.1644+1G>T		rs398123272
MTM1	NM_000252.2:c.595_599delCCTGC	NP_000243.1:p.Pro199Serfs	rs398123273
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MTM1	NM_000252.2:c.70C>T	NP_000243.1:p.Arg24Ter	rs398123275
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MYO7A	NM_000260.3:c.1556G>A	NP_000251.3:p.Gly519Asp	rs111033206
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MYO7A	NM_000260.3:c.3764delA	NP_000251.3:p.Lys1255Argfs	rs111033347
MYO7A	NM_000260.3:c.448C>T	NP_000251.3:p.Arg150Ter	rs121965079
MYO7A	NM_000260.3:c.634C>T	NP_000251.3:p.Arg212Cys	rs121965080
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MYO7A	NM_000260.3:c.652G>A	NP_000251.3:p.Asp218Asn	rs201539845
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MYO7A	NM_000260.3:c.93C>A	NP_000251.3:p.Cys31Ter	rs35689081
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MYO7A	NM_000260.3:c.5573T>C	NP_000251.3:p.Leu1858Pro	rs368657015
MYO7A	NM_000260.3:c.3728_3729insC	NP_000251.3:p.Pro1244Alafs	rs397516304
MYO7A	NM_000260.3:c.5618G>A	NP_000251.3:p.Arg1873Gln	rs397516322
MYO7A	NM_000260.3:c.700C>T	NP_000251.3:p.Gln234Ter	rs41298133
MYO7A	NM_000260.3:c.3862G>C	NP_000251.3:p.Ala1288Pro	rs749747871
MYO7A	NM_000260.3:c.2683C>T	NP_000251.3:p.Arg895Cys	rs781916427
MYO7A	NM_000260.3:c.284A>T	NP_000251.3:p.Tyr95Phe	rs797044489

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NBN	NM_002485.4:c.127C>T	NP_002476.2:p.Arg43Ter	rs200287925
NBN	NM_002485.4:c.37+1G>A		rs574673404
NBN	NM_002485.4:c.657_661delACAAA	NP_002476.2:p.Lys219fsTer16	rs587776650
NBN	NM_002485.4:c.188delT	NP_002476.2:p.Ala32fs	rs864622253
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NPC1	NM_000271.4:c.2665G>A	NP_000262.2:p.Val889Met	rs120074130
NPC1	NM_000271.4:c.3639G>C	NP_000262.2:p.Leu1213Phe	rs120074131
NPC1	NM_000271.4:c.2873G>A	NP_000262.2:p.Arg958Gln	rs120074132
NPC1	NM_000271.4:c.1133T>C	NP_000262.2:p.Val378Ala	rs120074134
NPC1	NM_000271.4:c.2848G>A	NP_000262.2:p.Val950Met	rs120074135
NPC1	NM_000271.4:c.337T>C	NP_000262.2:p.Cys113Arg	rs120074136
NPC1	NM_000271.4:c.1114C>T	NP_000262.2:p.Arg372Trp	rs1346436537
NPC1	NM_000271.4:c.3557G>A	NP_000262.2:p.Arg1186His	rs200444084
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NPC1	NM_000271.4:c.3265G>A	NP_000262.2:p.Glu1089Lys	rs374526072
NPC1	NM_000271.4:c.2196dupT	NP_000262.2:p.Pro733Serfs	rs398123284
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NPC1	NM_000271.4:c.3019C>G	NP_000262.2:p.Pro1007Ala	rs80358257
NPC1	NM_000271.4:c.3160G>A	NP_000262.2:p.Ala1054Thr	rs80358258
NPC1	NM_000271.4:c.3182T>C	NP_000262.2:p.Ile1061Thr	rs80358259
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NPC2	NM_006432.3:c.436C>T	NP_006423.1:p.Gln146Ter	rs104894457
NPC2	NM_006432.3:c.199T>C	NP_006423.1:p.Ser67Pro	rs11694
NPC2	NM_006432.3:c.3G>T	p.Met1?	rs483352893
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NPC2	NM_006432.3:c.115G>A	NP_006423.1:p.Val39Met	rs80358261
NPC2	NM_006432.3:c.133C>T	NP_006423.1:p.Gln45Ter	rs80358262
NPC2	NM_006432.3:c.141C>A	NP_006423.1:p.Cys47Ter	rs80358263
NPC2	NM_006432.3:c.295T>C	NP_006423.1:p.Cys99Arg	rs80358264
NPC2	NM_006432.3:c.332delA	NP_006423.1:p.Asn111Ilefs	rs80358265
NPC2	NM_006432.3:c.352G>T	NP_006423.1:p.Glu118Ter	rs80358266
NPC2	NM_006432.3:c.27delG	NP_006423.1:p.Leu10Serfs	rs80358267
OTC	NM_000531.5:c.809A>C	NP_000522.3:p.Gln270Pro	rs1800328
OTC	NM_000531.5:c.386G>A	NP_000522.3:p.Arg129His	rs66656800
OTC	NM_000531.5:c.274C>T	NP_000522.3:p.Arg92Ter	rs67418243
OTC	NM_000531.5:c.140A>T	NP_000522.3:p.Asn47Ile	rs67939655
OTC	NM_000531.5:c.374C>T	NP_000522.3:p.Thr125Met	rs72554356
OTC	NM_000531.5:c.491C>G	NP_000522.3:p.Ser164Ter	rs72556274
OTC	NM_000531.5:c.548A>G	NP_000522.3:p.Tyr183Cys	rs72556293
OTC	NM_000531.5:c.626C>T	NP_000522.3:p.Ala209Val	rs72558417
OTC	NM_000531.5:c.626C>A	NP_000522.3:p.Ala209Glu	rs72558417
OTC	NM_000531.5:c.808C>T	NP_000522.3:p.Gln270Ter	rs72558451
OTC	NM_000531.5:c.829C>T	NP_000522.3:p.Arg277Trp	rs72558454
PAH	NM_000277.1:c.1161C>A	NP_000522.3:p.Tyr387Ter	rs149595475
PAH	NM_000277.1:c.1161C>G	NP_000522.3:p.Tyr387Ter	rs149595475
PAH	NM_000277.1:c.516G>T	NP_000268.1:p.Gln172His	rs192592111
PAH	NM_000277.1:c.165del	NP_000268.1:p.Phe55LeufsTer6	rs199475566
PAH	NM_000277.1:c.822_832del	NP_000268.1:p.Lys274AsnfsTer5	rs199475581
PAH	NM_000277.1:c.673C>A	NP_000268.1:p.Pro225Thr	rs199475589
PAH	NM_000277.1:c.165T>G	NP_000268.1:p.Phe55Leu	rs199475598
PAH	NM_000277.1:c.529G>C	NP_000268.1:p.Val177Leu	rs199475602
PAH	NM_000277.1:c.529G>A	NP_000268.1:p.Val177Met	rs199475602
PAH	NM_000277.2:c.960G>C	NP_000268.1:p.Lys320Asn	rs199475615
PAH	NM_000277.1:c.520A>G	NP_000268.1:p.Ile174Val	rs199475632
PAH	NM_000277.1:c.498C>A	NP_000268.1:p.Tyr166Ter	rs199475645
PAH	NM_000277.1:c.464G>A	NP_000268.1:p.Arg155His	rs199475663
PAH	NM_000277.1:c.506G>A	NP_000268.1:p.Arg169His	rs199475679
PAH	NM_000277.1:c.143T>C	NP_000268.1:p.Leu48Ser	rs5030841
PAH	NM_000277.1:c.473G>A	NP_000268.1:p.Arg158Gln	rs5030843
PAH	NM_000277.1:c.727C>T	NP_000268.1:p.Arg243Ter	rs5030846
PAH	NM_000277.1:c.754C>T	NP_000268.1:p.Arg252Trp	rs5030847
PAH	NM_000277.1:c.782G>A	NP_000268.1:p.Arg261Gln	rs5030849
PAH	NM_000277.1:c.842C>T	NP_000268.1:p.Pro281Leu	rs5030851
PAH	NM_000277.1:c.842+1G>A		rs5030852
PAH	NM_000277.1:c.898G>T	NP_000268.1:p.Ala300Ser	rs5030853
PAH	NM_000277.1:c.1066-11G>A		rs5030855
PAH	NM_000277.1:c.1169A>G	NP_000268.1:p.Glu390Gly	rs5030856
PAH	NM_000277.1:c.1208C>T	NP_000268.1:p.Ala403Val	rs5030857
PAH	NM_000277.1:c.1222C>T	NP_000268.1:p.Arg408Trp	rs5030858
PAH	NM_000277.1:c.1241A>G	NP_000268.1:p.Tyr414Cys	rs5030860
PAH	NM_000277.1:c.1315+1G>A		rs5030861
PAH	NM_000277.1:c.441+5G>T		rs62507321
PAH	NM_000277.1:c.1200-1G>A		rs62507322
PAH	NM_000277.1:c.1066-3C>T		rs62507344
PAH	NM_000277.1:c.974A>G	NP_000268.1:p.Tyr325Cys	rs62508578

GEN	HGVSc	HGVSp	dbSNP
PAH	NM_000277.2:c.969+1G>A		rs62508584
PAH	NM_000277.1:c.728G>T	NP_000268.1:p.Arg243Leu	rs62508588
PAH	NM_000277.1:c.728G>A	NP_000268.1:p.Arg243Gln	rs62508588
PAH	NM_000277.3:c.1045T>C	NP_001341233.1:p.Ser349Pro	rs62508646
PAH	NM_000277.1:c.283A>T	NP_000268.1:p.Ile95Phe	rs62508682
PAH	NM_000277.1:c.805A>C	NP_000268.1:p.Ile269Leu	rs62508692
PAH	NM_000277.1:c.838G>A	NP_000268.1:p.Glu280Lys	rs62508698
PAH	NM_000277.1:c.284_286delTCA	NP_000268.1:p.Ile95_Lys96delin	rs62508727
PAH	NM_000277.1:c.722G>A	NP_000268.1:p.Arg241His	rs62508730
PAH	NM_000277.1:c.1184C>G	NP_000268.1:p.Ala395Gly	rs62508736
PAH	NM_000277.1:c.442-5C>G		rs62514909
PAH	NM_000277.1:c.611A>G	NP_001341233.1:p.Tyr204Cys	rs62514927
PAH	NM_000277.1:c.809G>A	NP_000268.1:p.Arg270Lys	rs62514950
PAH	NM_000277.2:c.814G>T	NP_000268.1:p.Gly272Ter	rs62514952
PAH	NM_000277.1:c.912+1G>A		rs62514956
PAH	NM_000277.1:c.1042C>G	NP_000268.1:p.Leu348Val	rs62516092
PAH	NM_000277.1:c.1162G>A	NP_000268.1:p.Val388Met	rs62516101
PAH	NM_000277.1:c.1183G>C	NP_000268.1:p.Ala395Pro	rs62516103
PAH	NM_000277.1:c.638T>C	NP_000268.1:p.Leu213Pro	rs62516109
PAH	NM_000277.1:c.1180G>C	NP_000268.1:p.Asp394His	rs62516142
PAH	NM_000277.1:c.261C>A	NP_000268.1:p.Ser87Arg	rs62516151
PAH	NM_000277.1:c.688G>A	NP_000268.1:p.Val230Ile	rs62516152
PAH	NM_000277.1:c.441+1G>A		rs62517166
PAH	NM_000277.1:c.847A>T	NP_000268.1:p.Ile283Phe	rs62517168
PAH	NM_000277.1:c.601C>T	NP_000268.1:p.His201Tyr	rs62517205
PAH	NM_000277.1:c.117C>G	NP_000268.1:p.Phe39Leu	rs62642926
PAH	NM_000277.1:c.311C>A	NP_000268.1:p.Ala104Asp	rs62642929
PAH	NM_000277.1:c.896T>G	NP_000268.1:p.Phe299Cys	rs62642933
PAH	NM_000277.1:c.916A>G	NP_000268.1:p.Ile306Val	rs62642934
PAH	NM_000277.1:c.926C>T	NP_000268.1:p.Ala309Val	rs62642935
PAH	NM_000277.1:c.1139C>T	NP_000268.1:p.Thr380Met	rs62642937
PAH	NM_000277.1:c.890G>A	NP_000268.1:p.Arg297His	rs62642939
PAH	NM_000277.1:c.1243G>A	NP_000268.1:p.Asp415Asn	rs62644499
PAH	NM_000277.1:c.509+1G>A		rs63102461
PAH	NM_000277.1:c.527G>T	NP_000268.1:p.Arg176Leu	rs74486803
PAH	NM_000277.1:c.472C>T	NP_000268.1:p.Arg158Trp	rs75166491
PAH	NM_000277.1:c.194T>C	NP_000268.1:p.Ile65Thr	rs75193786
PAH	NM_000277.1:c.734T>C	NP_000268.1:p.Val245Ala	rs76212747
PAH	NM_000277.1:c.1218A>G	NP_000268.1:p.Ile406Met	rs773526027
PAH	NM_000277.1:c.533A>G	NP_000268.1:p.Glu178Gly	rs77958223
PCDH15	NM_033056.3:c.733C>T	NP_149045.3:p.Arg245Ter	rs111033260
PCDH15	NM_033056.3:c.3233-2A>G		rs1554872194
PCDH15	NM_033056.3:c.1086delT	NP_149045.3:p.Asp362Asp=fs	rs199469706
PCDH15	NM_033056.3:c.3316C>T	NP_149045.3:p.Arg1106Ter	rs202033121
PCDH15	NM_033056.3:c.3983+1G>T		rs758921360
PEX1	NM_000466.2:c.2479C>T	NP_000457.1:p.Arg827Ter	rs1232449804
PEX1	NM_000466.2:c.2071+1G>T		rs267608177
PEX1	NM_000466.2:c.2230C>T	NP_000457.1:p.Gln744Ter	rs398123409
PEX1	NM_000466.2:c.1108del	NP_000457.1:p.Ile370LeufsTer17	rs61750406
PEX1	NM_000466.2:c.1108dup	NP_000457.1:p.Ile370AsnfsTer2	rs61750406
PEX1	NM_000466.2:c.2097dupT	NP_000457.1:p.Ile700TyrfS	rs61750415
PEX1	NM_000466.2:c.2528G>A	NP_000457.1:p.Gly843Asp	rs61750420

GEN	HGVSc	HGVSp	dbSNP
PEX1	NM_000466.2:c.2916delA	NP_000457.1:p.Gly973Alafs	rs61750426
PEX1	NM_000466.2:c.3379dupC	NP_000457.1:p.Arg1127Profs	rs794729652
PEX10	NM_002617.3:c.776+2T>C		rs1335685844
PEX10	NM_002617.3:c.600+1G>A		rs267608183
PEX10	NM_002617.3:c.373C>T	NP_002608.1:p.Arg125Ter	rs61750434
PEX10	NM_002617.3:c.704dupA	NP_002608.1:p.Leu236Alafs	rs61750435
PEX10	NM_002617.3:c.730C>T	NP_002608.1:p.Arg244Ter	rs61752092
PEX10	NM_002617.3:c.814_815del	NP_002608.1:p.Leu272ValfsTer66	rs61752093
PEX10	NM_002617.3:c.870C>G	NP_002608.1:p.His290Gln	rs61752095
PEX10	NM_002617.3:c.337delC	NP_002608.1:p.Leu113Trpfs	rs724159999
PEX12	NM_000286.2:c.691A>T	NP_000277.1:p.Lys231Ter	rs104894616
PEX12	NM_000286.2:c.126+1G>T		rs144259891
PEX12	NM_000286.2:c.681-2A>C		rs187526749
PEX12	NM_000286.2:c.888_889delCT	NP_000277.1:p.Leu297Thrfs	rs398123301
PEX12	NM_000286.2:c.538C>T	NP_000277.1:p.Arg180Ter	rs61752103
PEX12	NM_000286.2:c.604C>T	NP_000277.1:p.Arg202Ter	rs61752105
PEX12	NM_000286.2:c.744dup	NP_000277.1:p.Thr249TyrfTer14	rs61752108
PEX13	NM_002618.3:c.702G>A	NP_002609.1:p.Trp234Ter	rs104893661
PEX14	NM_004565.2:c.553C>T	NP_004556.1:p.Gln185Ter	rs61752116
PEX16	NM_057174.2:c.526C>T	NP_476515.1:p.Arg176Ter	rs61752117
PEX2	NM_000318.2:c.355C>T	NP_000309.1:p.Arg119Ter	rs61752123
PEX2	NM_000318.2:c.669G>A	NP_000309.1:p.Trp223Ter	rs61752127
PEX2	NM_000318.2:c.865dupA	NP_000309.1:p.Ser289Lysfs	rs724160029
PEX6	NM_000287.3:c.1314_1321del	NP_000278.3:p.Glu439GlyfsTer3	rs267608216
PEX6	NM_000287.3:c.2440C>T	NP_000278.3:p.Arg814Ter	rs267608241
PEX6	NM_000287.3:c.1802G>A	NP_000278.3:p.Arg601Gln	rs34324426
PEX6	NM_000287.3:c.1601T>C	NP_000278.3:p.Leu534Pro	rs387906809
PEX6	NM_000287.3:c.1338_1339delTG	NP_000278.3:p.Ala447Cysfs	rs398123303
PEX6	NM_000287.3:c.689_690dupAG	NP_000278.3:p.Ser232Hisfs	rs398123305
PEX7	NM_000288.3:c.653C>T	NP_000279.1:p.Ala218Val	rs121909151
PEX7	NM_000288.3:c.649G>A	NP_000279.1:p.Gly217Arg	rs121909152
PEX7	NM_000288.3:c.694C>T	NP_000279.1:p.Arg232Ter	rs121909153
PEX7	NM_000288.3:c.345T>G	NP_000279.1:p.Tyr115Ter	rs121909154
PEX7	NM_000288.3:c.903+1G>C		rs148591292
PEX7	NM_000288.3:c.875T>A	NP_000279.1:p.Leu292Ter	rs1805137
PEX7	NM_000288.3:c.340-10A>G		rs267608255
PEX7	NM_000288.3:c.854A>G	NP_000279.1:p.His285Arg	rs62653611
PMM2	NM_000303.3:c.484C>T	NP_000294.1:p.Arg162Trp	rs104894526
PMM2	NM_000303.2:c.442G>A	NP_000294.1:p.Asp148Asn	rs148032587
PMM2	NM_000303.2:c.470T>C	NP_000294.1:p.Phe157Ser	rs190521996
PMM2	NM_000303.2:c.422G>A	NP_000294.1:p.Arg141His	rs28936415
PMM2	NM_000303.2:c.640-9T>G		rs370160676
PMM2	NM_000303.2:c.109C>T	NP_000294.1:p.Gln37Ter	rs764353860
PMM2	NM_000303.2:c.24del	NP_000294.1:p.Cys9AlafsTer27	rs768021123
PMM2	NM_000303.2:c.647A>T	NP_000294.1:p.Asn216Ile	rs78290141
PMM2	NM_000303.2:c.338C>T	NP_000294.1:p.Pro113Leu	rs80338700
PMM2	NM_000303.2:c.691G>A	NP_000294.1:p.Val231Met	rs80338707
PMM2	NM_000303.2:c.710C>G	NP_000294.1:p.Thr237Arg	rs80338708
PMM2	NM_000303.2:c.722G>C	NP_000294.1:p.Cys241Ser	rs80338709
PRORY	n.392T>C		rs747573166
RPS4Y2			rs544793767
SERPINA1	NM_001127701.1:c.1096G>A	NP_001121173.1:p.Glu366Lys	rs28929474

GEN	HGVSc	HGVSp	dbSNP
SGSH	NM_000199.3:c.734G>A	NP_000190.1:p.Arg245His	rs104894635
SGSH	NM_000199.3:c.220C>T	NP_000190.1:p.Arg74Cys	rs104894636
SGSH	NM_000199.3:c.197C>G	NP_000190.1:p.Ser66Trp	rs104894637
SGSH	NM_000199.3:c.449G>A	NP_000190.1:p.Arg150Gln	rs104894638
SGSH	NM_000199.3:c.383C>T	NP_000190.1:p.Pro128Leu	rs104894642
SGSH	NM_000199.3:c.617G>C	NP_000190.1:p.Arg206Pro	rs104894643
SGSH	NM_000199.3:c.1A>G	p.Met1?	rs1250300189
SGSH	NM_000199.3:c.1A>C	p.Met1?	rs1250300189
SGSH	NM_000199.3:c.892T>C	NP_000190.1:p.Ser298Pro	rs138504221
SGSH	NM_000199.3:c.697C>T	NP_000190.1:p.Arg233Ter	rs374621913
SGSH	NM_000199.3:c.364G>A	NP_000190.1:p.Gly122Arg	rs761607612
SGSH	NM_000199.3:c.1167C>A	NP_000190.1:p.Asn389Lys	rs764057581
SGSH	NM_000199.3:c.1135del	NP_000190.1:p.Val379CysfsTer34	rs777956287
SLC26A4	NM_000441.1:c.412G>T	NP_000432.1:p.Val138Phe	rs111033199
SLC26A4	NM_000441.1:c.85G>C	NP_000432.1:p.Glu29Gln	rs111033205
SLC26A4	NM_000441.1:c.1003T>C	NP_000432.1:p.Phe335Leu	rs111033212
SLC26A4	NM_000441.1:c.1229C>T	NP_000432.1:p.Thr410Met	rs111033220
SLC26A4	NM_000441.1:c.1151A>G	NP_000432.1:p.Glu384Gly	rs111033244
SLC26A4	NM_000441.1:c.1588T>C	NP_000432.1:p.Tyr530His	rs111033254
SLC26A4	NM_000441.1:c.2T>C	NP_000432.1:p.Met1Thr	rs111033302
SLC26A4	NM_000441.1:c.626G>T	NP_000432.1:p.Gly209Val	rs111033303
SLC26A4	NM_000441.1:c.1226G>A	NP_000432.1:p.Arg409His	rs111033305
SLC26A4	NM_000441.1:c.1334T>G	NP_000432.1:p.Leu445Trp	rs111033307
SLC26A4	NM_000441.1:c.2015G>A	NP_000432.1:p.Gly672Glu	rs111033309
SLC26A4	NM_000441.1:c.1264-1G>C		rs111033311
SLC26A4	NM_000441.1:c.1614+1G>A		rs111033312
SLC26A4	NM_000441.1:c.919-2A>G		rs111033313
SLC26A4	NM_000441.1:c.1149+3A>G		rs111033314
SLC26A4	NM_000441.1:c.2027T>A	NP_000432.1:p.Leu676Gln	rs111033318
SLC26A4	NM_000441.1:c.578C>T	NP_000432.1:p.Thr193Ile	rs111033348
SLC26A4	NM_000441.1:c.2000T>G	NP_000432.1:p.Phe667Cys	rs121908360
SLC26A4	NM_000441.1:c.1105A>G	NP_000432.1:p.Lys369Glu	rs121908361
SLC26A4	NM_000441.1:c.2168A>G	NP_000432.1:p.His723Arg	rs121908362
SLC26A4	NM_000441.1:c.2162C>T	NP_000432.1:p.Thr721Met	rs121908363
SLC26A4	NM_000441.1:c.1115C>T	NP_000432.1:p.Ala372Val	rs121908364
SLC26A4	NM_000441.1:c.397T>A	NP_000432.1:p.Ser133Thr	rs121908365
SLC26A4	NM_000441.1:c.1540C>A	NP_000432.1:p.Gln514Lys	rs121908366
SLC26A4	NM_000441.1:c.349C>T	NP_000432.1:p.Leu117Phe	rs145254330
SLC26A4	NM_000441.1:c.1468A>C	NP_000432.1:p.Ile490Leu	rs200511789
SLC26A4	NM_000441.1:c.1246A>C	NP_000432.1:p.Thr416Pro	rs28939086
SLC26A4	NM_000441.1:c.-3-2A>G		rs397516411
SLC26A4	NM_000441.1:c.1198delT	NP_000432.1:p.Cys400Valfs	rs397516413
SLC26A4	NM_000441.1:c.1336C>T	NP_000432.1:p.Gln446Ter	rs397516416
SLC26A4	NM_000441.1:c.1437+2T>G		rs397516418
SLC26A4	NM_000441.1:c.164+2T>C		rs397516420
SLC26A4	NM_000441.1:c.1708G>A	NP_000432.1:p.Val570Ile	rs397516421
SLC26A4	NM_000441.1:c.1963A>G	NP_000432.1:p.Ile655Val	rs397516424
SLC26A4	NM_000441.1:c.2188C>T	NP_000432.1:p.Gln730Ter	rs397516428
SLC26A4	NM_000441.1:c.68C>A	NP_000432.1:p.Ser23Ter	rs397516430
SLC26A4	NM_000441.1:c.997_998insA	NP_000432.1:p.Arg333Lysfs	rs431905486

Seznam vyšetřovaných variant - CarrierTest



GEN	HGVSc	HGVSp	dbSNP
SLC26A4	NM_000441.1:c.554G>C	NP_000432.1:p.Arg185Thr	rs542620119
SLC26A4	NM_000441.1:c.1554G>A	NP_000432.1:p.Trp518Ter	rs727503428
SLC26A4	NM_000441.1:c.3G>C	NP_000432.1:p.Met1Ile	rs786204426
SLC26A4	NM_000441.1:c.890del	NP_000432.1:p.Pro297GlnfsTer6	rs786204600
SLC26A4	NM_000441.1:c.707T>C	NP_000432.1:p.Leu236Pro	rs80338848
SLC26A4	NM_000441.1:c.1001+1G>A		rs80338849
SMN1	NM_000344	NP_000335	EX8del
SMPD1	NM_000543.4:c.1493G>A	NP_000534.3:p.Arg498His	rs120074117
SMPD1	NM_000543.4:c.1493G>T	NP_000534.3:p.Arg498Leu	rs120074117
SMPD1	NM_000543.4:c.788T>A	NP_000534.3:p.Leu263Ter	rs120074120
SMPD1	NM_000543.4:c.730G>A	NP_000534.3:p.Gly244Arg	rs120074122
SMPD1	NM_000543.4:c.911T>C	NP_000534.3:p.Leu304Pro	rs120074124
SMPD1	NM_000543.4:c.1267C>T	NP_000534.3:p.His423Tyr	rs120074126
SMPD1	NM_000543.4:c.1327C>T	NP_000534.3:p.Arg443Ter	rs120074127
SMPD1	NM_000543.4:c.880C>A	NP_000534.3:p.Gln294Lys	rs120074128
SMPD1	NM_000543.4:c.1426C>T	NP_000534.3:p.Arg476Trp	rs182812968
SMPD1	NM_000543.4:c.1406A>C	NP_000534.3:p.Tyr469Ser	rs267607074
SMPD1	NM_000543.4:c.996delC	NP_000534.3:p.Phe333Serfs	rs387906289
SMPD1	NM_000543.4:c.1420_1421delCT	NP_000534.3:p.Leu474Glnfs	rs398123476
SMPD1	NM_000543.4:c.1624C>T	NP_000534.3:p.Arg542Ter	rs398123478
SMPD1	NM_000543.4:c.1133G>A	NP_000534.3:p.Arg378His	rs559088058
SMPD1	NM_000543.4:c.354delC	NP_000534.3:p.Ile119Serfs	rs727504165
SMPD1	NM_000543.4:c.581del	NP_000534.3:p.Pro194GlnfsTer63	rs748165078
SMPD1	NM_000543.4:c.748A>C	NP_000534.3:p.Ser250Arg	rs750779804
SMPD1	NM_000543.4:c.564dup	NP_000534.3:p.Lys189GlnfsTer4	rs756366019
SMPD1	NM_000543.4:c.518dup	NP_000534.3:p.Ser174LeufsTer19	rs786204733
SRY			sY14
SRY			sY14
TGM1	NM_000359.2:c.968G>A	NP_000350.1:p.Arg323Gln	rs121918717
TGM1	NM_000359.2:c.1135G>C	NP_000350.1:p.Val379Leu	rs121918720
TGM1	NM_000359.2:c.1187G>T	NP_000350.1:p.Arg396Leu	rs121918721
TGM1	NM_000359.2:c.1166G>C	NP_000350.1:p.Arg389Pro	rs121918723
TGM1	NM_000359.2:c.1166G>A	NP_000350.1:p.Arg389His	rs121918723
TGM1	NM_000359.2:c.857G>A	NP_000350.1:p.Arg286Gln	rs121918727
TGM1	NM_000359.2:c.281G>A	NP_000350.1:p.Gly94Asp	rs121918729
TGM1	NM_000359.2:c.866A>C	NP_000350.1:p.Asn289Thr	rs121918730
TGM1	NM_000359.2:c.919C>T	NP_000350.1:p.Arg307Trp	rs121918731
TGM1	NM_000359.2:c.919C>G	NP_000350.1:p.Arg307Gly	rs121918731
TGM1	NM_000359.2:c.877-2A>G		rs142634031
TGM1	NM_000359.2:c.944G>A	NP_000350.1:p.Arg315His	rs143473912
TGM1	NM_000359.2:c.876+2T>C		rs151054393
TGM1	NM_000359.2:c.1075G>A	NP_000350.1:p.Val359Met	rs202037016
TGM1	NM_000359.2:c.788G>A	NP_000350.1:p.Trp263Ter	rs367699137
TGM1	NM_000359.2:c.943C>T	NP_000350.1:p.Arg315Cys	rs397514525
TPP1	NM_000391.3:c.1094G>A	NP_000382.3:p.Cys365Tyr	rs119455954
TPP1	NM_000391.3:c.622C>T	NP_000382.3:p.Arg208Ter	rs119455955
TPP1	NM_000391.3:c.617G>A	NP_000382.3:p.Arg206His	rs121908209
TPP1	NM_000391.3:c.237C>G	NP_000382.3:p.Tyr79Ter	rs1564855860

GEN	HGVSc	HGVSp	dbSNP
TPP1	NM_000391.3:c.509-1G>A		rs56144125
TPP1	NM_000391.3:c.509-1G>C		rs56144125
TPP1	NM_000391.3:c.509-1G>T		rs56144125
TPP1	NM_000391.3:c.833A>G	NP_000382.3:p.Gln278Arg	rs796053439
TTY15			rs773105453
USH1C			1220delG
USH1C			36+1G>T
USH1C	NM_005709.3:c.91C>T	NP_005700.2:p.Arg31Ter	rs121908370
USH1C	NM_005709.3:c.496+1G>T		rs138138689
USH1C	NM_005709.3:c.496+1G>A		rs138138689
USH1C	NM_005709.3:c.216G>A	NP_005700.2:p.Val72=	rs151045328
USH1C	NM_005709.3:c.308G>A	NP_005700.2:p.Arg103His	rs397514500
USH1C	NM_005709.3:c.238dupC	NP_005700.2:p.Arg80Profs	rs397515359
USH1C	NM_005709.3:c.388G>A	NP_005700.2:p.Val130Ile	rs55843567
USH2A	NM_206933.2:c.10561T>C	NP_996816.2:p.Trp3521Arg	rs111033264
USH2A	NM_206933.2:c.1606T>C	NP_996816.2:p.Cys536Arg	rs111033273
USH2A	NM_206933.2:c.2209C>T	NP_996816.2:p.Arg737Ter	rs111033334
USH2A	NM_206933.2:c.11864G>A	NP_996816.2:p.Trp3955Ter	rs111033364
USH2A	NM_206933.2:c.6224G>A	NP_996816.2:p.Trp2075Ter	rs111033386
USH2A	NM_206933.2:c.13335_13343del	NP_996816.2:p.Glu4445_Met4447del	rs111033408
USH2A	NM_206933.2:c.11048-1G>A		rs111033414
USH2A	NM_206933.2:c.3902G>T	NP_996816.2:p.Gly1301Val	rs111033524
USH2A	NM_206933.2:c.779T>G	NP_996816.2:p.Leu260Ter	rs121912598
USH2A	NM_206933.2:c.1256G>T	NP_996816.2:p.Cys419Phe	rs121912600
USH2A	NM_206933.2:c.13339A>G	NP_996816.2:p.Met4447Val	rs139474806
USH2A	NM_206933.2:c.14803C>T	NP_996816.2:p.Arg4935Ter	rs146733615
USH2A	NM_206933.2:c.10073G>A	NP_996816.2:p.Cys3358Tyr	rs148660051
USH2A	NM_206933.2:c.14402_14403del	NP_996816.2:p.Tyr4801PhefsTer20	rs1553250192
USH2A	NM_206933.2:c.9191del	NP_996816.2:p.Tyr3064Metfs	rs1553268435
USH2A	NM_206933.2:c.12575G>A	NP_996816.2:p.Arg4192His	rs199605265
USH2A	NM_206933.2:c.6937G>T	NP_996816.2:p.Gly2313Cys	rs199840367
USH2A	NM_206933.2:c.11048-2A>G		rs200871041
USH2A	NM_206933.2:c.2802T>G	NP_996816.2:p.Cys934Trp	rs201527662
USH2A	NM_206933.2:c.8682-9A>G		rs372347027
USH2A	NM_206933.2:c.1143+1G>A		rs397517974
USH2A	NM_206933.2:c.11954G>A	NP_996816.2:p.Trp3985Ter	rs397517976
USH2A	NM_206933.2:c.12714T>G	NP_996816.2:p.Tyr4238Ter	rs397517981
USH2A	NM_206933.2:c.12868C>T	NP_996816.2:p.Gln4290Ter	rs397517983
USH2A	NM_206933.2:c.14180G>A	NP_996816.2:p.Trp4727Ter	rs397517989
USH2A	NM_206933.2:c.14911C>T	NP_996816.2:p.Arg4971Ter	rs397517994
USH2A	NM_206933.2:c.3309C>A	NP_996816.2:p.Tyr1103Ter	rs397518011
USH2A	NM_206933.2:c.3435delA	NP_996816.2:p.Val1147Serfs	rs397518012
USH2A	NM_206933.2:c.3547_3548delAT	NP_996816.2:p.Ile1183Phefs	rs397518013
USH2A	NM_206933.2:c.3558delT	NP_996816.2:p.Cys1186Trpfs	rs397518014
USH2A	NM_206933.2:c.5858-1G>A		rs397518023
USH2A	NM_206933.2:c.653T>A	NP_996816.2:p.Val218Glu	rs397518026
USH2A	NM_206933.2:c.7244C>G	NP_996816.2:p.Ser2415Ter	rs397518029
USH2A	NM_206933.2:c.820C>T	NP_996816.2:p.Arg274Ter	rs397518036

GEN	HGVSc	HGVSp	dbSNP
USH2A	NM_206933.2:c.8559-2A>G		rs397518039
USH2A	NM_206933.2:c.8981G>A	NP_996816.2:p.Trp2994Ter	rs397518041
USH2A	NM_206933.2:c.9159T>G	NP_996816.2:p.Tyr3053Ter	rs397518042
USH2A	NM_206933.2:c.9304C>T	NP_996816.2:p.Gln3102Ter	rs397518046
USH2A	NM_206933.2:c.9424G>T	NP_996816.2:p.Gly3142Ter	rs397518048
USH2A	NM_206933.2:c.6862G>T	NP_996816.2:p.Glu2288Ter	rs398124619
USH2A	NM_206933.2:c.11065C>T	NP_996816.2:p.Arg3689Ter	rs41314534
USH2A	NM_206933.2:c.2983C>T	NP_996816.2:p.Gln995Ter	rs527236135
USH2A	NM_206933.2:c.1876C>T	NP_996816.2:p.Arg626Ter	rs534534437
USH2A	NM_206933.2:c.13374delA	NP_996816.2:p.Glu4458Aspfs	rs727503715
USH2A	NM_206933.2:c.6398G>A	NP_996816.2:p.Trp2133Ter	rs727503725
USH2A	NM_206933.2:c.2541C>A	NP_996816.2:p.Cys847Ter	rs727503736
USH2A	NM_206933.2:c.14248C>T	NP_996816.2:p.Gln4750Ter	rs727504867
USH2A	NM_206933.2:c.632G>A	NP_996816.2:p.Trp211Ter	rs727504893
USH2A	NM_206933.2:c.5877delT	NP_996816.2:p.Ser1961Glnfs	rs727505343
USH2A	NM_206933.2:c.9459C>A	NP_996816.2:p.Cys3153Ter	rs73090721
USH2A	NM_206933.2:c.1214delA	NP_996816.2:p.Asn405Ilefs	rs750228923
USH2A	NM_206933.2:c.12574C>T	NP_996816.2:p.Arg4192Cys	rs750396156
USH2A	NM_206933.2:c.7524del	NP_996816.2:p.Arg2509GlyfsTer19	rs751176116
USH2A	NM_206933.2:c.13316C>T	NP_996816.2:p.Thr4439Ile	rs753330544
USH2A	NM_206933.3:c.2610C>A	NP_996816.2:p.Cys870Ter	rs767078782
USH2A	NM_206933.2:c.9270C>A	NP_996816.2:p.Cys3090Ter	rs779572631
USH2A	NM_206933.2:c.1055C>T	NP_996816.2:p.Thr352Ile	rs780308389
USH2A	NM_206933.2:c.12268C>A	NP_996816.2:p.Pro4090Thr	rs780893919
USH2A	NM_206933.2:c.2276G>T	NP_996816.2:p.Cys759Phe	rs80338902
USH2A	NM_206933.2:c.2299delG	NP_996816.2:p.Glu767Serfs	rs80338903
USH2A	NM_206933.2:c.14020A>G	NP_996816.2:p.Arg4674Gly	rs80338904
USH2A	NM_206933.2:c.11585C>G	NP_996816.2:p.Thr3862Arg	rs886044126