

GENE	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	rs122547100
ACADM	NM_000016.4:c.468+2T>G	-	rs164721430
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	rs105751642
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	rs105751636
ACADVL	NM_000018.3:c.1606-1G>A		rs105751738
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

GENE	HGVSc	HGVSp	dbSNP
ACADVL	NM_000018.3:c.298_299delCA	NP_000009.1:p.Gln100Valfs	rs786204713
ADGRV1	NM_032119.3:c.8495C>A	NP_115495.3:p.Ser2832Ter	rs121909761
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	rs144390266:
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.Asp251GlufsTer23	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.-135G>A (within M2 haplotype)		rs113588187
ANXA5	NM_001154.3:c.-184T>C (within M2 haplotype)		rs28651243
ANXA5	NM_001154.3:c.-210A>C (within M2 haplotype)		rs28717001
ANXA5	NM_001154.3:c.-229G>A (within M2 haplotype)		rs112782763
AR	NM_000044.3:c.4G>A	NP_000035.2:p.Glu2Lys	rs104894742

GENE	HGVSc	HGVSp	dbSNP
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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
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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
ARSA	NM_000487.5:c.827C>T	NP_000478.3:p.Thr276Met	rs74315472
ARSA	NM_000487.5:c.868C>T	NP_000478.3:p.Arg290Cys	rs74315473
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
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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

GENE	HGVSc	HGVSp	dbSNP
ARSA	NM_000487.5:c.769G>C	NP_000478.3:p.Asp257His	rs80338819
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.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_7876delTGins	NP_000042.3:p.AspAla2625GluPro	rs267606668
ATM	NM_000051.3:c.7271T>G	NP_000042.3:p.Val2424Gly	rs28904921

GENE	HGVSc	HGVSp	dbSNP
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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
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ATM	NM_000051.3:c.2502dupA	NP_000042.3:p.Val835Serfs	rs587779822
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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
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ATM	NM_000051.3:c.3388G>T	NP_000042.3:p.Gly1130Ter	rs587781911
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ATM	NM_000051.3:c.2754delT	NP_000042.3:p.Phe918Leufs	rs786202608

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ATM	NM_000051.3:c.151C>T	NP_000042.3:p.Gln51Ter	rs786203888
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ATM	NM_000051.3:c.3850delA	NP_000042.3:p.Thr1284fs	rs876660865
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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
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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
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BLM	NM_000057.2:c.1642C>T	NP_000048.1:p.Gln548Ter	rs200389141
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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
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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
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GENE	HGVSc	HGVSp	dbSNP
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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
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CBS	NM_000071.2:c.374G>A	NP_000062.1:p.Arg125Gln	rs781444670
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CDH23	NM_022124.5:c.6133G>A	NP_071407.4:p.Asp2045Asn	rs121908348
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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
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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
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CFTR	NM_000492.3:c.2125C>T	NP_000483.3:p.Arg709Ter	rs121908760
CFTR	NM_000492.3:c.3276C>A	NP_000483.3:p.Tyr1092Ter	rs121908761
CFTR	NM_000492.3:c.3587C>G	NP_000483.3:p.Ser1196Ter	rs121908763
CFTR	NM_000492.3:c.3611G>A	NP_000483.3:p.Trp1204Ter	rs121908764
CFTR	NM_000492.3:c.3612G>A	NP_000483.3:p.Trp1204Ter	rs121908765
CFTR	NM_000492.3:c.935_937delTCT	NP_000483.3:p.Phe312del	rs121908768
CFTR	NM_000492.3:c.262_263delTT	NP_000483.3:p.Leu88Ilefs	rs121908769
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CFTR	NM_000492.3:c.531del	NP_000483.3:p.Ile177MetfsTer12	rs121908771
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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.3039del	NP_000483.3:p.Tyr1014ThrfsTer9	rs121908781
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CFTR	NM_000492.3:c.3744delA	NP_000483.3:p.Lys1250ArgfsTer9	rs121908784
CFTR	NM_000492.3:c.1155_1156dup	NP_000483.3:p.Asn386IlefsTer3	rs121908785
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CFTR	NM_000492.3:c.2737_2738insG	NP_000483.3:p.Tyr913Ter	rs121908788
CFTR	NM_000492.3:c.3773_3774insT	NP_000483.3:p.Leu1258delinsPhe	rs121908789
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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
CFTR	NM_000492.3:c.164+2T>C		rs121908800
CFTR	NM_000492.3:c.313delA	NP_000483.3:p.Ile105Serfs	rs121908801
CFTR	NM_000492.3:c.595C>T	NP_000483.3:p.His199Tyr	rs121908802

GENE	HGVSc	HGVSp	dbSNP
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CFTR	NM_000492.3:c.1397C>G	NP_000483.3:p.Ser466Ter	rs121908805
CFTR	NM_000492.3:c.3889dup	NP_000483.3:p.Ser1297PhefsTer5	rs121908808
CFTR	NM_000492.3:c.2290C>T	NP_000483.3:p.Arg764Ter	rs121908810
CFTR	NM_000492.3:c.3659delC	NP_000483.3:p.Thr1220Lysfs	rs121908811
CFTR	NM_000492.3:c.2012delT	NP_000483.3:p.Leu671Terfs	rs121908812
CFTR	NM_000492.3:c.1647T>G	NP_000483.3:p.Ser549Arg	rs121909005
CFTR	NM_000492.3:c.1687T>A	NP_000483.3:p.Tyr563Asn	rs121909006
CFTR	NM_000492.3:c.1687T>G	NP_000483.3:p.Tyr563Asp	rs121909006
CFTR	NM_000492.3:c.1000C>T	NP_000483.3:p.Arg334Trp	rs121909011
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
CFTR	NM_000492.3:c.933C>G	NP_000483.3:p.Phe311Leu	rs121909016
CFTR	NM_000492.3:c.1475C>T	NP_000483.3:p.Ser492Phe	rs121909017
CFTR	NM_000492.3:c.3197G>A	NP_000483.3:p.Arg1066His	rs121909019
CFTR	NM_000492.3:c.3199G>A	NP_000483.3:p.Ala1067Thr	rs121909020
CFTR	NM_000492.3:c.171G>A	NP_000483.3:p.Trp57Ter	rs121909025
CFTR	NM_000492.3:c.3937C>T	NP_000483.3:p.Gln1313Ter	rs121909026
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CFTR	NM_000492.3:c.3194T>C	NP_000483.3:p.Leu1065Pro	rs121909036
CFTR	NM_000492.3:c.3763T>C	NP_000483.3:p.Ser1255Pro	rs121909041
CFTR	NM_000492.3:c.4364C>G	NP_000483.3:p.Ser1455Ter	rs121909043
CFTR	NM_000492.3:c.1682C>A	NP_000483.3:p.Ala561Glu	rs121909047
CFTR	NM_000492.3:c.3230T>C	NP_000483.3:p.Leu1077Pro	rs139304906
CFTR	NM_000492.3:c.1400T>C	NP_000483.3:p.Leu467Pro	rs139573311
CFTR	NM_000492.3:c.3468G>A	NP_000483.3:p.Leu1156%3D	rs139729994
CFTR	NM_000492.3:c.2249C>T	NP_000483.3:p.Pro750Leu	rs140455771
CFTR	NM_000492.3:c.2490+1G>A		rs141158996
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CFTR	NM_000492.3:c.3717G>A	NP_000483.3:p.Arg1239%3D	rs144781064
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CFTR	NM_000492.3:c.1327G>T	NP_000483.3:p.Asp443Tyr	rs147422190
CFTR	NM_000492.3:c.2739T>A	NP_000483.3:p.Tyr913Ter	rs149790377
CFTR	NM_000492.3:c.137C>A	NP_000483.3:p.Ala46Asp	rs151020603
CFTR	NM_000492.3:c.2856G>C	p.Met952Ile	rs151048781
CFTR	NM_000492.3:c.4127_4131delTGAT	NP_000483.3:p.Leu1376Serfs	rs1554397527
CFTR	NM_000492.3:c.3011_3019del	NP_000483.3:p.Ala1004_Ala1006del	rs1562914072
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
CFTR	NM_000492.3:c.2810dup	NP_000483.3:p.Val938GlyfsTer37	rs193922510
CFTR	NM_000492.3:c.2989-2A>G		rs193922515

GENE	HGVSc	HGVSp	dbSNP
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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
CFTR	NM_000492.3:c.4242+1G>A		rs372227120
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
CFTR	NM_000492.3:c.2423_2424dup	NP_000483.3:p.Ser809IlefsTer13	rs387906359
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CFTR	NM_000492.3:c.1081del	NP_000483.3:p.Trp361GlyfsTer8	rs387906361
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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.115C>T	NP_000483.3:p.Gln39Ter	rs397508168
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.1373del	NP_000483.3:p.Gly458AspfsTer11	rs397508196
CFTR	NM_000492.3:c.1392G>T	NP_000483.3:p.Lys464Asn	rs397508198
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CFTR	NM_000492.3:c.1393-2A>G		rs397508201
CFTR	NM_000492.3:c.1418del	NP_000483.3:p.Gly473GlufsTer54	rs397508205
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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GENE	HGVSc	HGVSp	dbSNP
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CFTR	NM_000492.3:c.164+4dup		rs397508244
CFTR	NM_000492.3:c.1648G>T	NP_000483.3:p.Gly550Ter	rs397508247
CFTR	NM_000492.3:c.165-1G>A		rs397508249
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
CFTR	NM_000492.3:c.1680-886A>G		rs397508266
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
CFTR	NM_000492.3:c.1703del	NP_000483.3:p.Leu568CysfsTer4	rs397508274
CFTR	NM_000492.3:c.1705T>G	NP_000483.3:p.Tyr569Asp	rs397508276
CFTR	NM_000492.3:c.170G>A	NP_000483.3:p.Trp57Ter	rs397508279
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CFTR	NM_000492.3:c.1A>G	NP_000483.3:p.Met1?	rs397508328
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
CFTR	NM_000492.3:c.2126G>A	NP_000483.3:p.Arg709Gln	rs397508342
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CFTR	NM_000492.3:c.2464G>T	NP_000483.3:p.Glu822Ter	rs397508378
CFTR	NM_000492.3:c.2491G>T	NP_000483.3:p.Glu831Ter	rs397508387
CFTR	NM_000492.3:c.2502del	NP_000483.3:p.Phe834LeufsTer10	rs397508389
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CFTR	NM_000492.3:c.2537G>A	NP_000483.3:p.Trp846Ter	rs397508393
CFTR	NM_000492.3:c.2547C>A	NP_000483.3:p.Tyr849Ter	rs397508394
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CFTR	NM_000492.3:c.2589_2599del	NP_000483.3:p.Ile864SerfsTer28	rs397508400
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CFTR	NM_000492.3:c.263T>A	NP_000483.3:p.Leu88Ter	rs397508412
CFTR	NM_000492.3:c.263T>G	NP_000483.3:p.Leu88Ter	rs397508412
CFTR	NM_000492.3:c.2645G>A	NP_000483.3:p.Trp882Ter	rs397508413
CFTR	NM_000492.3:c.2658-1G>C		rs397508416

GENE	HGVSc	HGVSp	dbSNP
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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.2936A>T	NP_000483.3:p.Asp979Val	rs397508462
CFTR	NM_000492.3:c.293A>G	NP_000483.3:p.Gln98Arg	rs397508464
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
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CFTR	NM_000492.3:c.3139_3139+1del		rs397508505
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

GENE	HGVSc	HGVSp	dbSNP
CFTR	NM_000492.3:c.4234C>T	NP_000483.3:p.Gln1412Ter	rs397508702
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
CFTR	NM_000492.3:c.543_546del	NP_000483.3:p.Leu183PhefsTer5	rs397508750
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.Gly330GlufsTer39	rs397508824
CFTR	NM_000492.3:c.2453del	NP_000483.3:p.Leu818TrpfsTer3	rs397515498
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

GENE	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

GENE	HGVSc	HGVSp	dbSNP
CarrierTest is only a screening method detecting frequent variants in CYP21A2 gene: c.293-13C>G; p.Arg357Trp and p.Val282Leu, which is associated with milder phenotype. Because of presence of highly homologous pseudogene and frequent genomic rearrangements in given chromosomal region this method does not detect chimeric gene (present in 30 % patients), copy number variations of parts or whole CYP21A2 gene, frequent mutation p.Ile173Asn (in 11% patients) and other pathogenic CYP21A2 mutations.			
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
DMD	NM_004006.3:c.10019G>A	NP_003997.2:p.Cys3340Tyr	rs104894788

GENE	HGVSc	HGVSp	dbSNP
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
DMD	NM_004006.2:c.2077C>T	NP_003997.2:p.Gln693Ter	rs1060502661

GENE	HGVSc	HGVSp	dbSNP
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
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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
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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
DMD	NM_004006.3:c.6790C>T	NP_003997.2:p.Gln2264Ter	rs128626252

GENE	HGVSc	HGVSp	dbSNP
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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
DMD	NM_004006.3:c.8579del	NP_003997.2:p.Pro2860LeufsTer2	rs1556656847

GENE	HGVSc	HGVSp	dbSNP
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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
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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
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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.Asp219GlufsTer7	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
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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
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GENE	HGVSc	HGVSp	dbSNP
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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
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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
DMD	NM_004006.3:c.3713del	NP_003997.2:p.Lys1238ArgfsTer6	rs1557362198
DMD	NM_004006.3:c.3433-2_3433-1delinsCC		rs1557365503
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
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DMD	NM_004006.2:c.2949+1G>A		rs1557374482
DMD	NM_004006.3:c.2804-1del		rs1557374667
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GENE	HGVSc	HGVSp	dbSNP
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.Leu880TyrfTer11	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
DMD	NM_004006.3:c.8061del	NP_003997.2:p.His2688IlefsTer38	rs1569140330
DMD	NM_004006.3:c.2129del	NP_003997.2:p.Lys710ArgfsTer19	rs1569169560
DMD	NM_004006.2:c.2111del	NP_003997.2:p.Pro704fs	rs1569169736
DMD	NM_004006.2:c.8010G>A	NP_003997.2:p.Trp2670Ter	rs1569186252
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DMD	NM_004006.2:c.1652G>A	NP_003997.2:p.Trp551Ter	rs1569230215
DMD	NM_004006.2:c.1638G>A	NP_003997.2:p.Trp546Ter	rs1569230293
DMD	NM_004006.3:c.1620G>A	NP_003997.2:p.Trp540Ter	rs1569230389
DMD	NM_004006.2:c.1619G>A	NP_003997.2:p.Trp540Ter	rs1569230406
DMD	NM_004006.3:c.1577del	NP_003997.2:p.Thr526MetfsTer57	rs1569273905
DMD	NM_004006.3:c.1062G>A	NP_003997.2:p.Trp354Ter	rs1569376447
DMD	NM_004006.2:c.10546G>T	NP_003997.2:p.Glu3516Ter	rs1569401103
DMD	NM_004006.3:c.10087-2A>G		rs1569420861
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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.9807+5G>A		rs1569460607
DMD	NM_004006.3:c.9807+1G>T		rs1569460610
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DMD	NM_004006.2:c.9634G>T	NP_003997.2:p.Glu3212Ter	rs1569463953
DMD	NM_004006.3:c.776_777del	NP_003997.2:p.Lys259SerfsTer3	rs1569469138
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
DMD	NM_004006.3:c.6649C>T	NP_003997.2:p.Gln2217Ter	rs1569516166
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DMD	NM_004006.3:c.377_378insA	NP_003997.2:p.Asn126LysfsTer12	rs1569528138
DMD	NM_004006.2:c.9100C>T	NP_003997.2:p.Arg3034Ter	rs1569530432
DMD	NM_004006.3:c.264+1G>A		rs1569533147
DMD	NM_004006.3:c.141_142insG	NP_003997.2:p.Arg48GluTer41	rs1569533965
DMD	NM_004006.2:c.8655C>A	NP_003997.2:p.Tyr2885Ter	rs1569546174
DMD	NM_004006.3:c.8548-1G>C		rs1569546198
DMD	NM_004006.2:c.8524C>T	NP_003997.2:p.Gln2842Ter	rs1569547229
DMD	NM_004006.2:c.6276C>G	NP_003997.2:p.Tyr2092Ter	rs1569555987
DMD	NM_004006.3:c.6025C>T	NP_003997.2:p.Gln2009Ter	rs1569556943
DMD	NM_004006.3:c.5972del	NP_003997.2:p.Leu1991TrpfsTer11	rs1569556948
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
DMD	NM_004006.3:c.5563C>T	NP_003997.2:p.Gln1855Ter	rs1569559097

GENE	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.Lys1596GlufsTer5	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
DMD	NM_004006.3:c.4151del	NP_003997.2:p.Glu1384GlyfsTer34	rs1569561687
DMD	NM_004006.2:c.3923C>A	NP_003997.2:p.Ser1308Ter	rs1569562466
DMD	NM_004006.2:c.3856G>T	NP_003997.2:p.Glu1286Ter	rs1569562570
DMD	NM_004006.2:c.3804G>A	NP_003997.2:p.Trp1268Ter	rs1569562573
DMD	NM_004006.2:c.3742C>T	NP_003997.2:p.Gln1248Ter	rs1569562936
DMD	NM_004006.2:c.3679C>T	NP_003997.2:p.Gln1227Ter	rs1569562941
DMD	NM_004006.2:c.3606_3607del		rs1569562951
DMD	NM_004006.2:c.3604-1G>C		rs1569562952
DMD	NM_004006.3:c.3500C>G	NP_003997.2:p.Ser1167Ter	rs1569563278
DMD	NM_004006.2:c.3358G>T	NP_003997.2:p.Glu1120Ter	rs1569563678
DMD	NM_004006.3:c.3257dup	p.Gln1087AlafsTer11	rs1569563740
DMD	NM_004006.3:c.3257del	NP_003997.2:p.Lys1086SerfsTer3	rs1569563740
DMD	NM_004006.3:c.3220G>T	NP_003997.2:p.Glu1074Ter	rs1569563751
DMD	NM_004006.3:c.2914del	NP_003997.2:p.Tyr972MetfsTer32	rs1569564281
DMD	NM_004006.3:c.2380+3A>C		rs1569564916
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DMD	NM_004006.3:c.1774_1804del	NP_003997.2:p.Asp592fs	rs1601808175
DMD	NM_004006.3:c.1791dup	NP_003997.2:p.Ser598fs	rs1601808288
DMD	NM_004006.3:c.1788del	NP_003997.2:p.Met596fs	rs1601808296
DMD	NM_004006.3:c.1777C>T	NP_003997.2:p.Gln593Ter	rs1601808326
DMD	NM_004006.3:c.1769_1772dup	NP_003997.2:p.Lys591AsnfsTer2	rs1601808412
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DMD	NM_004006.3:c.1713dup	NP_003997.2:p.Ser572Ter	rs1601808932
DMD	NM_004006.2:c.1705-2A>G		rs1601809011
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DMD	NM_004006.3:c.1664_1667dup	NP_003997.2:p.Asp556fs	rs1601810667
DMD	NM_004006.3:c.9185_9186AG[2]	NP_003997.2:p.Arg3063fs	rs1601864055
DMD	NM_004006.3:c.9085_9086insA	NP_003997.2:p.Val3029fs	rs1602056443
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DMD	NM_004006.3:c.10494_10495AG[2]	p.Glu3499_Ser3500insTer	rs1602337330
DMD	NM_004006.3:c.10395-1G>A		rs1602338283
DMD	NM_004006.3:c.10333_10334insGCTAG		rs1602360475
DMD	NM_004006.3:c.10304C>G	NP_003997.2:p.Ser3435Ter	rs1602369798
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DMD	NM_004006.3:c.10087-1G>A		rs1602417503
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DMD	NM_004006.3:c.9950_9951del	NP_003997.2:p.Lys3317fs	rs1602454747
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GENE	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.1323dup	NP_003997.2:p.Gln442fs	rs1602459252
DMD	NM_004006.3:c.1270del	p.Gln423_Met424insTer	rs1602459861
DMD	NM_004006.3:c.1259dup	NP_003997.2:p.Gln421fs	rs1602459951
DMD	NM_004006.3:c.1255G>T	NP_003997.2:p.Glu419Ter	rs1602460062
DMD	NM_004006.3:c.1147G>T	NP_003997.2:p.Glu383Ter	rs1602467530
DMD	NM_004006.3:c.1144del	NP_003997.2:p.His382fs	rs1602467590
DMD	NM_004006.3:c.1129del	NP_003997.2:p.Asp377fs	rs1602467787
DMD	NM_004006.3:c.998C>A	NP_003997.2:p.Ser333Ter	rs1602468968
DMD	NM_004006.3:c.971del	NP_003997.2:p.Ala324fs	rs1602469251
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DMD	NM_004006.3:c.9649+5G>C		rs1602695527
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DMD	NM_004006.3:c.93_93+14del		rs1602765633
DMD	NM_004006.3:c.93+2T>A		rs1602765856
DMD	NM_004006.3:c.58del	NP_003997.2:p.Thr20fs	rs1602766499
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DMD	NM_004006.3:c.9033_9040del	NP_003997.2:p.Tyr3012fs	rs1602937725
DMD	NM_004006.3:c.9022C>T	NP_003997.2:p.Gln3008Ter	rs1602937974
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DMD	NM_004006.3:c.822T>A	p.Tyr274Ter	rs1603149954
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DMD	NM_004006.3:c.709del	NP_003997.2:p.Gln237fs	rs1603150993
DMD	NM_004006.3:c.666T>G	NP_003997.2:p.Tyr222Ter	rs1603151699
DMD	NM_004006.3:c.8923del	NP_003997.2:p.Leu2975fs	rs1603221854
DMD	NM_004006.2:c.8881C>T	NP_003997.2:p.Gln2961Ter	rs1603222095
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DMD	NM_004006.3:c.8451del	NP_003997.2:p.Leu2818fs	rs1603264659
DMD	NM_004006.3:c.8417del	NP_003997.2:p.Gln2806fs	rs1603264705
DMD	NM_004006.3:c.8391-1G>A		rs1603264731
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DMD	NM_004006.3:c.31+1del		rs1603419074
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DMD	NM_004006.3:c.8146C>T	NP_003997.2:p.Gln2716Ter	rs1603430178
DMD	NM_004006.3:c.8145dup	NP_003997.2:p.Gln2716fs	rs1603430181
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.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.7564C>T	NP_003997.2:p.Gln2522Ter	rs1603452207
DMD	NM_004006.3:c.7068del	NP_003997.2:p.Gln2356fs	rs1603513680
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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.7027del	NP_003997.2:p.Trp2343fs	rs1603513828
DMD	NM_004006.3:c.6913-1G>T		rs1603514271
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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
DMD	NM_004006.3:c.5689G>T	NP_003997.2:p.Glu1897Ter	rs1603631240
DMD	NM_004006.3:c.5689dup	NP_003997.2:p.Glu1897fs	rs1603631241
DMD	NM_004006.3:c.5636G>A	NP_003997.2:p.Trp1879Ter	rs1603631244
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
DMD	NM_004006.3:c.4858_4859GA[1]	NP_003997.2:p.Glu1620fs	rs1603631756

GENE	HGVSc	HGVSp	dbSNP
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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
DMD	NM_004006.3:c.3795G>A	NP_003997.2:p.Trp1265Ter	rs1603633545
DMD	NM_004006.3:c.3533_3536del	NP_003997.2:p.Glu1178fs	rs1603633864
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DMD	NM_004006.3:c.3460G>T	NP_003997.2:p.Gly1154Ter	rs1603633868
DMD	NM_004006.3:c.3295_3296del	NP_003997.2:p.Gln1099fs	rs1603634079
DMD	NM_004006.3:c.3193del	NP_003997.2:p.Ala1065fs	rs1603634105
DMD	NM_004006.3:c.3157dup	NP_003997.2:p.Ile1053fs	rs1603634199
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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
DMD	NM_004006.2:c.2869C>T	NP_003997.2:p.Gln957Ter	rs1603634298
DMD	NM_004006.3:c.2853del	NP_003997.2:p.Arg952fs	rs1603634299
DMD	NM_004006.3:c.2803+5G>C		rs1603634603
DMD	NM_004006.3:c.2790_2791AG[1]	NP_003997.2:p.Glu931fs	rs1603634604
DMD	NM_004006.3:c.2708del	NP_003997.2:p.Gly903fs	rs1603634607
DMD	NM_004006.2:c.2704C>T	NP_003997.2:p.Gln902Ter	rs1603634608
DMD	NM_004006.3:c.2694dup	NP_003997.2:p.Glu899fs	rs1603634609
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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
DMD	NM_004006.3:c.2381-2A>C		rs1603634749
DMD	NM_004006.3:c.2287_2292+12del		rs1603635326
DMD	NM_004006.3:c.2169-1G>A		rs1603635331
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
DMD	NM_004006.3:c.1894A>T	NP_003997.2:p.Lys632Ter	rs1603636541
DMD	NM_004006.3:c.5363C>G		rs199774535
DMD	NM_004006.2:c.10247G>A	NP_003997.2:p.Trp3416Ter	rs201217593

GENE	HGVSc	HGVSp	dbSNP
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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
DMD	NM_004006.3:c.2546del	NP_003997.2:p.Ala849fs	rs2042979418
DMD	NM_004006.3:c.2508dup	NP_003997.2:p.Asn837Ter	rs2042983140
DMD	NM_004006.3:c.2439del	NP_003997.2:p.Ile813fs	rs2042990064
DMD	NM_004006.3:c.9807+2T>G		rs2043792360
DMD	NM_004006.3:c.9743T>A	NP_003997.2:p.Leu3248Ter	rs2043796525
DMD	NM_004006.3:c.2348C>G	NP_003997.2:p.Ser783Ter	rs2044079791
DMD	NM_004006.3:c.9461T>A	NP_003997.2:p.Leu3154Ter	rs2044442536
DMD	NM_004006.3:c.9432del	NP_003997.2:p.Met3145fs	rs2044445392
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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
DMD	NM_004006.3:c.1855A>T	NP_003997.2:p.Lys619Ter	rs2051636517
DMD	NM_004006.3:c.29_30del	NP_003997.2:p.Cys10fs	rs2051898232
DMD	NM_004006.3:c.1703del	NP_003997.2:p.Gln568fs	rs2052700227
DMD	NM_004006.3:c.1672del	NP_003997.2:p.Leu558fs	rs2052703269
DMD	NM_004006.3:c.1509del	NP_003997.2:p.Glu503fs	rs2055460202
DMD	NM_004006.3:c.1501G>T	NP_003997.2:p.Glu501Ter	rs2055461260
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DMD	NM_004006.3:c.1294_1295dup	NP_003997.2:p.Cys433fs	rs2059642653
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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
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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
DMD	NM_004006.3:c.8666_8668+3del		rs2068033029
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GENE	HGVSc	HGVSp	dbSNP
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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
DMD	NM_004006.3:c.321dup	NP_003997.2:p.Leu108fs	rs2078432433
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DMD	NM_004006.3:c.187-2A>G		rs2080514041
DMD	NM_004006.3:c.8023A>T	NP_003997.2:p.Lys2675Ter	rs2080887929
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
DMD	NM_004006.3:c.7768G>T	NP_003997.2:p.Glu2590Ter	rs2082258116
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
DMD	NM_004006.3:c.7378G>T	NP_003997.2:p.Glu2460Ter	rs2090513299
DMD	NM_004006.3:c.7179dup	NP_003997.2:p.Pro2394fs	rs2093207581
DMD	NM_004006.3:c.88del	NP_003997.2:p.Ser30fs	rs2093885666
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DMD	NM_004006.3:c.6901C>T	NP_003997.2:p.Gln2301Ter	rs2094827204
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DMD	NM_004006.3:c.6290+2T>A		rs2097447247
DMD	NM_004006.3:c.6253del	NP_003997.2:p.Trp2085fs	rs2097447453
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DMD	NM_004006.3:c.5860A>T	NP_003997.2:p.Lys1954Ter	rs2097746958
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
DMD	NM_004006.3:c.5530del	NP_003997.2:p.Arg1844fs	rs2097762566
DMD	NM_004006.3:c.5521G>T	NP_003997.2:p.Glu1841Ter	rs2097762588
DMD	NM_004006.3:c.5452G>T	NP_003997.2:p.Glu1818Ter	rs2097762928
DMD	NM_004006.3:c.5114C>G	NP_003997.2:p.Ser1705Ter	rs2097848034
DMD	NM_004006.3:c.5012T>A	NP_003997.2:p.Leu1671Ter	rs2097849676
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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
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GENE	HGVSc	HGVSp	dbSNP
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
DMD	NM_004006.3:c.3630del	NP_003997.2:p.Glu1211fs	rs2098315232
DMD	NM_004006.3:c.3490A>T	NP_003997.2:p.Lys1164Ter	rs2098349160
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DMD	NM_004006.3:c.2314G>T		rs267606770
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DMD	NM_004006.3:c.4897G>T	NP_003997.2:p.Glu1633Ter	rs370800101
DMD	NM_004006.3:c.1812+1G>A		rs373286166
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
DMD	NM_004006.3:c.3374C>A		rs374744331
DMD	NM_004006.3:c.10248G>A	NP_003997.2:p.Trp3416Ter	rs376745644
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
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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
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DMD	NM_004006.3:c.10258_10259insT		rs398123835
DMD	NM_004006.3:c.10447_10448del		rs398123837
DMD	NM_004006.3:c.10454del		rs398123839
DMD	NM_004006.3:c.1047_1048insA		rs398123840
DMD	NM_004006.3:c.10625del		rs398123844
DMD	NM_004006.2:c.10797+5G>A		rs398123846
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.1306_1307insG		rs398123854
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
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GENE	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.1734del	NP_003997.2:p.Glu579fs	rs398123866
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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
DMD	NM_004006.2:c.2419C>T	NP_003997.2:p.Gln807Ter	rs398123888
DMD	NM_004006.3:c.2436G>A		rs398123889
DMD	NM_004006.2:c.251del	NP_003997.2:p.Leu84fs	rs398123893
DMD	NM_004006.3:c.2547del		rs398123895
DMD	NM_004006.3:c.2622+1G>T		rs398123901
DMD	NM_004006.2:c.2622+1G>A		rs398123901
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
DMD	NM_004006.3:c.28del		rs398123913
DMD	NM_004006.3:c.2950-2A>T		rs398123915
DMD	NM_004006.3:c.2950-2A>G		rs398123915
DMD	NM_004006.3:c.3059C>G		rs398123919
DMD	NM_004006.3:c.3059C>A	NP_003997.2:p.Ser1020Ter	rs398123919
DMD	NM_004006.3:c.3076G>T		rs398123920
DMD	NM_004006.2:c.31+1G>C		rs398123923
DMD	NM_004006.2:c.31+1G>T		rs398123923
DMD	NM_004006.2:c.3151C>T	NP_003997.2:p.Arg1051Ter	rs398123929
DMD	NM_004006.2:c.3217G>T	NP_003997.2:p.Glu1073Ter	rs398123931
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
DMD	NM_004006.2:c.3432+1G>A		rs398123937
DMD	NM_004006.3:c.3432+3A>G		rs398123938
DMD	NM_004006.2:c.3580C>T	NP_003997.2:p.Gln1194Ter	rs398123942
DMD	NM_004006.3:c.3639del		rs398123943
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DMD	NM_004006.3:c.4117C>T		rs398123948
DMD	NM_004006.3:c.412_413del		rs398123949
DMD	NM_004006.3:c.4315del	NP_003997.2:p.Arg1439fs	rs398123950
DMD	NM_004006.2:c.4314_4315del	NP_003997.2:p.Arg1439fs	rs398123950
DMD	NM_004006.2:c.434G>C	NP_003997.2:p.Arg145Pro	rs398123952
DMD	NM_004006.2:c.4375C>T	NP_003997.2:p.Arg1459Ter	rs398123953

GENE	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.4518+5G>A		rs398123960
DMD	NM_004006.3:c.4534_4535del		rs398123961
DMD	NM_004006.3:c.4545_4549del		rs398123962
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
DMD	NM_004006.3:c.5124_5127del		rs398123979
DMD	NM_004006.2:c.5134C>T	NP_003997.2:p.Gln1712Ter	rs398123980
DMD	NM_004006.2:c.5287C>T	NP_003997.2:p.Arg1763Ter	rs398123981
DMD	NM_004006.2:c.531-2A>G		rs398123985
DMD	NM_004006.2:c.5353C>T	NP_003997.2:p.Gln1785Ter	rs398123990
DMD	NM_004006.3:c.5554C>T		rs398123991
DMD	NM_004006.3:c.5640T>A		rs398123993
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
DMD	NM_004006.3:c.5938G>T		rs398124001
DMD	NM_004006.3:c.6000T>A		rs398124002
DMD	NM_004006.3:c.615T>A		rs398124004
DMD	NM_004006.3:c.6182del		rs398124005
DMD	NM_004006.2:c.6283C>T	NP_003997.2:p.Arg2095Ter	rs398124008
DMD	NM_004006.3:c.6391_6392del		rs398124012
DMD	NM_004006.3:c.6392_6393insCA		rs398124012
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DMD	NM_004006.3:c.676_678del		rs398124034
DMD	NM_004006.3:c.6906G>A		rs398124036
DMD	NM_004006.3:c.6986del		rs398124040
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
DMD	NM_004006.3:c.7309+1G>T		rs398124044
DMD	NM_004006.3:c.7309+1G>C		rs398124044
DMD	NM_004006.2:c.7657C>T	NP_003997.2:p.Arg2553Ter	rs398124050
DMD	NM_004006.2:c.7672C>T	NP_003997.2:p.Gln2558Ter	rs398124051
DMD	NM_004006.3:c.7682G>A		rs398124052
DMD	NM_004006.3:c.7683G>A		rs398124053
DMD	NM_004006.2:c.7771G>T	NP_003997.2:p.Glu2591Ter	rs398124055
DMD	NM_004006.3:c.7894C>T		rs398124058
DMD	NM_004006.3:c.7922del		rs398124059
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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
DMD	NM_004006.3:c.9158_9161del		rs398124081

GENE	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.9361+1G>C		rs398124094
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
DMD	NM_004006.3:c.1332-9A>G		rs72468700
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DMD	NM_004006.3:c.8680G>T		rs727503802
DMD	NM_004006.3:c.4806A>T		rs727503830
DMD	NM_004006.3:c.3532G>T		rs727503836
DMD	NM_004006.2:c.2956C>T	NP_003997.2:p.Gln986Ter	rs727503844
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
DMD	NM_004006.3:c.3982C>T	NP_003997.2:p.Gln1328Ter	rs745640786
DMD	NM_004006.3:c.2642C>A	NP_003997.2:p.Ser881Ter	rs747937552
DMD	NM_004006.2:c.4483C>T	NP_003997.2:p.Gln1495Ter	rs748769566
DMD	NM_004006.3:c.3892G>T	NP_003997.2:p.Gly1298Ter	rs750349613
DMD	NM_004006.3:c.2485C>T		rs753662330
DMD	NM_004006.3:c.10807G>T	NP_003997.2:p.Glu3603Ter	rs754333675
DMD	NM_004006.3:c.6982A>T		rs754896795
DMD	NM_004006.2:c.511G>C	NP_003997.2:p.Ala171Pro	rs755452188
DMD	NM_004006.3:c.4495C>T		rs755686224
DMD	NM_004006.2:c.2797C>T	NP_003997.2:p.Gln933Ter	rs756949497
DMD	NM_004006.2:c.4870C>T	NP_003997.2:p.Gln1624Ter	rs762250680
DMD	NM_004006.2:c.7755G>A	NP_003997.2:p.Trp2585Ter	rs762394978
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
DMD	NM_004006.2:c.5506C>T	NP_003997.2:p.Gln1836Ter	rs770845480
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
DMD	NM_004006.3:c.1070del		rs794726994

GENE	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.3:c.2601_2602del		rs794727322
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
DMD	NM_004006.3:c.2815_2816del		rs794727358
DMD	NM_004006.3:c.2933_2934del		rs794727359
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DMD	NM_004006.3:c.3535G>T		rs794727422
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.565C>T		rs794727861
DMD	NM_004006.3:c.627del		rs794727862
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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
DMD	NM_004006.2:c.547dup	NP_003997.2:p.Trp183fs	rs796065333
DMD	NM_004006.3:c.3603+2dup		rs796523999
DMD	NM_004006.3:c.6673_6674insGTTT		rs797044743
DMD	NM_004006.3:c.291dup		rs797044756
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
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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
DMD	NM_004006.2:c.2215G>T	NP_003997.2:p.Glu739Ter	rs863224985

GENE	HGVSc	HGVSp	dbSNP
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DMD	NM_004006.2:c.2611A>T	NP_003997.2:p.Lys871Ter	rs863224987
DMD	NM_004006.2:c.2623-3C>G		rs863224988
DMD	NM_004006.2:c.265-2A>G		rs863224989
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
DMD	NM_004006.3:c.6611del	NP_003997.2:p.Lys2204fs	rs863225007
DMD	NM_004006.2:c.7105G>T	NP_003997.2:p.Glu2369Ter	rs863225008
DMD	NM_004006.2:c.7817G>A	NP_003997.2:p.Trp2606Ter	rs863225009
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
DMD	NM_004006.3:c.8970_8971del		rs863225014
DMD	NM_004006.3:c.9204_9207del		rs863225015
DMD	NM_004006.2:c.94-1G>A		rs863225016
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_004006.2:c.1504C>T	NP_003997.2:p.Gln502Ter	rs878854618
DMD	NM_004006.2:c.1705-1G>T		rs878854619
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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
DMD	NM_004006.2:c.3259C>T	NP_003997.2:p.Gln1087Ter	rs886039536
DMD	NM_004006.3:c.10150del		rs886039663
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
DMD	NM_004006.2:c.1865C>G	NP_003997.2:p.Ser622Ter	rs886041344

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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
DMD	NM_004006.3:c.1387_1408del		rs886042616
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
DMD	NM_004006.3:c.10062T>A		rs886042875
DMD	NM_004006.3:c.457C>T		rs886042983
DMD	NM_004006.3:c.5739+1G>T		rs886043041
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
DMD	NM_004006.3:c.179_180insGGGCA		rs886043288
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
DMD	NM_004006.3:c.6223C>T		rs886043428
DMD	NM_004006.3:c.4174C>T		rs886043496
DMD	NM_004006.2:c.9938G>T	NP_003997.2:p.Cys3313Phe	rs886043597
DMD	NM_004006.3:c.3747del		rs886043635
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DMD	NM_004006.3:c.10126del		rs886043676
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.9563+1G>T		rs886043989
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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
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
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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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G6PC1	NM_000151.2:c.1039C>T	NP_000142.1:p.Gln347Ter	rs80356487
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
GALT	NM_000155.3:c.404C>T	NP_000146.2:p.Ser135Leu	rs111033690
GALT	NM_000155.3:c.442C>T	NP_000146.2:p.Arg148Trp	rs111033693
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GALT	NM_000155.3:c.505C>A	NP_000146.2:p.Gln169Lys	rs111033709
GALT	NM_000155.3:c.584T>C	NP_000146.2:p.Leu195Pro	rs111033728
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GALT	NM_000155.3:c.601C>T	NP_000146.2:p.Arg201Cys	rs111033739
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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
GALT	NM_000155.3:c.833T>A	NP_000146.2:p.Ile278Asn	rs111033778
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
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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
GCDH	NM_000159.3:c.1093G>A	NP_000150.1:p.Glu365Lys	rs121434370
GCDH	NM_000159.4:c.1198G>A	NP_000150.1:p.Val400Met	rs121434372
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
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GJB2	NM_004004.5:c.230G>A	NP_003995.2:p.Trp77Ter	rs104894395
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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
GJB2	NM_004004.5:c.334_335del	NP_003995.2:p.Lys112GlufsTer2	rs756484720

GENE	HGVSc	HGVSp	dbSNP
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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
GLA	NM_000169.2:c.901C>T	NP_000160.1:p.Arg301Ter	rs398123224
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
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GENE	HGVSc	HGVSp	dbSNP
HADHA	NM_000182.4:c.1528G>C	NP_000173.2:p.Glu510Gln	rs137852769
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.1A>T	p.Met1?	rs121907965
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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
HEXA	NM_000520.4:c.1330+1G>A		rs767041069
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
CHRNE	NM_000080.3:c.803-2A>G		rs1320610655
CHRNE	NM_000080.3:c.103T>C	NP_000511.2:p.Tyr35His	rs144169073
CHRNE	NM_000080.3:c.442T>A	NP_000511.2:p.Cys148Ser	rs1597621396
CHRNE	NM_000080.3:c.130dup	NP_000071.1:p.Glu44GlyfsTer3	rs762368691

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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
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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
IKBKAP (ELP1)	NM_003640.3:c.1814A>G	NP_003631.2:p.Tyr605Cys	rs756919296
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
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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
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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.872C>T	NP_064551.3:p.Ala291Val	rs201041864
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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.295G>C	NP_071415.1:p.Glu99Gln	rs119103219
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MCCC2	NM_022132.4:c.1309A>G	NP_071415.1:p.Ile437Val	rs119103224
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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.2040G>A	NP_000234.1:p.Met680Ile	rs28940580
MEFV	NM_000243.2:c.2040G>C	NP_000234.1:p.Met680Ile	rs28940580
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
MTM1	NM_000252.2:c.1040T>G	NP_000243.1:p.Leu347Ter	rs398123264
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
MTM1	NM_000252.2:c.1537_1564del28i	NP_000243.1:p.Phe513_Leu522del	rs398123271
MTM1	NM_000252.2:c.1644+1G>T		rs398123272
MTM1	NM_000252.2:c.595_599delCCTGC	NP_000243.1:p.Pro199Serfs	rs398123273
MTM1	NM_000252.2:c.688T>C	NP_000243.1:p.Trp230Arg	rs398123274
MTM1	NM_000252.2:c.70C>T	NP_000243.1:p.Arg24Ter	rs398123275
MTM1	NM_000252.2:c.605delT	NP_000243.1:p.Leu202Trpfs	rs672601325
MYO7A	NM_000260.3:c.494C>T	NP_000251.3:p.Thr165Met	rs111033174
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MYO7A	NM_000260.3:c.6070C>T	NP_000251.3:p.Arg2024Ter	rs111033198
MYO7A	NM_000260.3:c.2005C>T	NP_000251.3:p.Arg669Ter	rs111033201
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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.1884C>A	NP_000251.3:p.Cys628Ter	rs121965083
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MYO7A	NM_000260.3:c.635G>A	NP_000251.3:p.Arg212His	rs28934610
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
MYO7A	NM_000260.3:c.3503G>A	NP_000251.3:p.Arg1168Gln	rs797044516

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NBN	NM_002485.4:c.127C>T	NP_002476.2:p.Arg43Ter	rs200287925
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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.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
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NPC1	NM_000271.4:c.2783A>C	NP_000262.2:p.Gln928Pro	rs28940897
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NPC1	NM_000271.4:c.3265G>A	NP_000262.2:p.Glu1089Lys	rs374526072
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NPC1	NM_000271.4:c.2230_2231delGT	NP_000262.2:p.Val744Serfs	rs483352882
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NPC1	NM_000271.4:c.2972_2973del	NP_000262.2:p.Gln991ArgfsTer15	rs756815030
NPC1	NM_000271.4:c.3591+1G>A		rs786200877
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NPC1	NM_000271.4:c.2324A>C	NP_000262.2:p.Gln775Pro	rs80358253
NPC1	NM_000271.4:c.2974G>T	NP_000262.2:p.Gly992Trp	rs80358254
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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
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NPC2	NM_006432.3:c.3G>T	p.Met1?	rs483352893
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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
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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
PAH	NM_000277.2:c.969+1G>A		rs62508584

GENE	HGVSc	HGVSp	dbSNP
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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
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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
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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.Ile700Tyrf	rs61750415
PEX1	NM_000466.2:c.2528G>A	NP_000457.1:p.Gly843Asp	rs61750420
PEX1	NM_000466.2:c.2916delA	NP_000457.1:p.Gly973Alafs	rs61750426

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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.Thr249TyrfsTer14	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
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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
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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
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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
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SLC26A4	NM_000441.1:c.1614+1G>A		rs111033312
SLC26A4	NM_000441.1:c.919-2A>G		rs111033313
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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
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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
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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
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GENE	HGVSc	HGVSp	dbSNP
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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
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SMPD1	NM_000543.4:c.1420_1421delCT	NP_000534.3:p.Leu474Glufs	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
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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
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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
TPP1	NM_000391.3:c.509-1G>A		rs56144125

GENE	HGVSc	HGVSp	dbSNP
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
USH2A	NM_206933.2:c.8559-2A>G		rs397518039

GENE	HGVSc	HGVSp	dbSNP
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