CGT Essential - Variants List

Gene	Tested Variants
MMACHC NM_015506.2	c.1A>G; c.2T>C; c.2T>G; c.3G>A; c.72C>A; c.80A>G; c.81+1G>C; c.81+1G>T; c.81+1G>A; c.81+2T>C; c.81+2T>G; c.81G>A; c.82-1G>A; c.89G>A; c.90G>A; c.158T>C; c.178dupG; c.182G>C; c.187delC; c.202C>T; c.217C>T; c.270dupT; c.271dupA; c.273_276delAGAG; c.276G>A; c.276G>T; c.277-2A>G; c.285dupA; c.292C>T; c.310_313delGACT; c.315C>G; c.328_331delAACC; c.331C>T; c.347T>C; c.352delC; c.364dupC; c.384delC; c.388_390delTAC; c.388T>C; c.389A>G; c.391C>T; c.394C>T; c.398_399delAA; c.415_416delCCinsTA; c.420G>A; c.427C>T; c.429+1G>C; c.430-2A>G; c.433delA; c.434dupT; c.435_436delAT; c.439G>A; c.445_446delTG; c.440G>A; c.440G>C; c.452A>G; c.457C>T; c.464G>A; c.466G>A; c.467G>A; c.471G>A; c.471G>C; c.481C>T; c.482G>A; c.484G>T; c.497dupT; c.500delC; c.507_519delAGAGGTGCCAGAT; c.541_548delGACTGTGT; c.547_548delGT; c.551_554dupTACC; c.565C>A; c.565delC; c.567dupT; c.574delC; c.578T>C; c.600G>A; c.603_604delTG; c.608G>A; c.609G>A; c.615C>A; c.615C>G; c.616C>T; c.617G>A; c.617G>C; c.619dupG; c.626_627delTG; c.626dupT; c.648dupA; c.658_660delAAG; c.666C>A; c.675_679delCTCCA; c.685C>T; c.688C>T.
ACADM NM_001286043.1	c.708A>C; c.419T>C; c.826C>T; c.-6_6delGCCAACATGGCAinsACCCCGAAGG; c.-6_6delGCCAACATGGCAinsACCCCGAGTG; c.1012G>T; c.1025dupT; c.1026delC; c.1027G>A; c.1045-2A>C; c.1048C>T; c.1049A>T; c.104T>G; c.1056_1057delAT; c.1058C>A; c.107_113dupGATTTAG; c.1072dupG; c.1076T>C; c.1081A>G; c.1083delG; c.1084A>C; c.1084A>G; c.1098_1110dupTAGAATGAGTTAC; c.1111_1112insTAGAATGAGTTAC; c.1111C>T; c.1141C>T; c.1144C>T; c.1144delC; c.1151C>T; c.1154A>G; c.1172delA; c.118+1G>T; c.1184G>A; c.119-1G>C; c.1201_1204delTTAG; c.1213dupG; c.1223T>C; c.1249G>T; c.127G>A; c.1288dupT; c.1288T>A; c.1289A>C; c.1293+1G>A; c.1294-1G>C; c.1294-2A>T; c.1320_1321delAA; c.157C>T; c.165delT; c.173_174delAG; c.177A>C; c.199T>C; c.1A>G; c.201T>A; c.216+1G>T; c.216+2T>C; c.216+2T>G; c.217-1G>A; c.218A>G; c.224delT; c.233T>C; c.244dupT; c.250C>T; c.253G>T; c.270_271delCA; c.286+2T>G; c.30+2T>C; c.386-1G>C; c.386-2A>G; c.3G>C; c.421_424delATTA; c.432_444delATTGGCTTATGGA; c.442_447delGGATGT; c.446G>A; c.454dupG; c.461C>T; c.47delC; c.486+1delG; c.486+1G>A; c.486+1G>T; c.524delA; c.525delG; c.530_533delAGTA; c.536delT; c.542G>A; c.546G>A; c.546G>T; c.548_551delCTGA; c.553G>T; c.563T>C; c.567+2T>G; c.568-1G>A; c.568-2A>C; c.571T>C; c.602A>C; c.602A>G; c.640_641delGA; c.654T>G; c.676A>G; c.67C>T; c.682G>A; c.698+1G>A; c.698+2T>C; c.699-18G>A; c.700delT; c.707T>A; c.715C>T; c.716G>A; c.716G>T; c.782C>A; c.785delC; c.797T>C; c.807+1G>A; c.807+2T>G; c.807G>C; c.808-1G>A; c.808-2A>C; c.808G>T; c.816C>A; c.829T>C; c.833C>T; c.838A>G; c.841A>G; c.856C>A; c.85C>T; c.893_902delGTGACGGAGCinsTTTTAA; c.896A>G; c.898G>A; c.916_928delGCAATGGGAGCTT; c.925_927delGCTinsCC; c.941G>C; c.942A>T; c.948+1G>A; c.949-2A>G; c.980G>C; c.981_982delAG.
DHCR7 NM_001163817.1	c.862G>A; c.385_412+5delATCCAGGAGGGGGGCCGTGACTCCTGCAGGTAGC; c.89G>C; c.1214delA; c.1004delC; c.1054C>T; c.1055G>A; c.1057delG; c.1066delC; c.1076_1077dupAC; c.1080_1081delGT; c.1084C>T; c.1090A>G; c.1112G>A; c.111G>A; c.1138T>C; c.1139G>A; c.1146C>A; c.1190C>T; c.1199G>A; c.1210C>T; c.1219A>T; c.1222T>C; c.1228G>A; c.1290C>G; c.1325A>G; c.1327C>T; c.1328G>A; c.1337G>A; c.1342_1344delGAGinsC; c.1342G>A; c.1348delC; c.1349_1350delGCinsTG; c.1396G>A; c.1409T>A; c.1426T>C; c.151C>T; c.16C>T; c.1A>C; c.1A>G; c.278C>T; c.292C>T; c.296T>C; c.321G>C; c.322-2A>G; c.326T>C; c.355delC; c.356A>T; c.3G>A; c.413-1G>A; c.413-2A>G; c.429T>G; c.433A>C; c.440G>A; c.452G>A; c.453G>A; c.461C>G; c.461C>T; c.470T>C; c.501G>A; c.506C>T; c.546G>A; c.626+1G>T; c.627-1G>A; c.651C>A; c.655T>C; c.655T>G; c.696G>A; c.724C>T; c.725G>A; c.728C>G; c.730G>A; c.740C>T; c.744G>T; c.803delA; c.804delT; c.820_825delAACGTC; c.82C>T; c.831+2T>A; c.832-1G>C; c.839A>G; c.852C>A; c.858G>A; c.861C>A; c.866C>T; c.870G>A; c.874delC; c.889A>C; c.894C>A; c.895C>T; c.89delG; c.902A>G; c.906C>G; c.907G>A; c.915C>G; c.934_935delGT; c.939G>A; c.952delT; c.952T>A; c.963+1G>T; c.963+2T>G; c.964-1G>C; c.964-1G>T; c.970T>C; c.976G>T; c.98+2_98+6delTAAGG; c.981C>T; c.99-1G>C; c.991C>T; c.995delT; c.1138T>C; c.1139G>A; c.1146C>A.
HBB NM_000518.4	c.208G>A; c.179A>C; c.93G>T; c.93-17_93-1delTATTTTCCCACCCTTAG; c.-75G>C; c.316-7C>A; c.-29G>A; c.-18C>G; c.-136C>T; c.-136C>G; c.-121C>T; c.-137C>A; c.-137C>G; c.-137C>T; c.-138C>A; c.-138C>T; c.-140C>T; c.-151C>T; c.-50A>C; c.-78A>C; c.-78A>G; c.-79A>G; c.-80T>A; c.-81A>G; c.103G>T; c.107A>G; c.108C>A; c.108C>G; c.110C>A; c.110delC; c.111_118delTTGGACCC; c.112delT; c.113G>A; c.114_120delGACCCAG; c.114G>A; c.117_118delCC; c.118C>T; c.118delC; c.11dupT; c.123delG; c.125dupT; c.126_129delCTTT; c.126delC; c.127_129delTTT; c.127T>C; c.127T>G; c.128T>C; c.129delT; c.129dupT; c.130dupG; c.130G>T; c.134C>G; c.135delC; c.138delT; c.138dupT; c.142dupG; c.143_146dupATCT; c.143dupA; c.145_146insATCT; c.146delT; c.147delG; c.150delC; c.155delC; c.158delA; c.162delT; c.163dupG; c.164_168delTTATGinsGGCATCA; c.165_177delTATGGGCAACCCT; c.165delT; c.166delA; c.166dupA; c.17_18delCT; c.170_171insGCAACCCTAAGGTG; c.176C>G; c.176delC; c.176dupC; c.178A>T; c.179delA; c.18_19delTG; c.182T>A; c.184A>T; c.189_195delTCATGGC; c.19_20delGAinsAT; c.190C>T; c.194delG; c.199A>G; c.199A>T; c.19G>A; c.19G>T; c.1A>C; c.1A>G; c.20_32delAGGAGAAGTCTGC; c.201delA; c.203_204delTG; c.203T>A; c.206_212delTCGGTGC; c.206T>A; c.209delG; c.20A>T; c.20delA; c.210delT; c.216delT; c.216dupT; c.217_221delAGTGAinsT; c.217dupA; c.219_220delTG; c.219dupT; c.221_224dupATGG; c.226delC; c.229_230delGC; c.22G>T; c.230delC; c.232delC; c.235delC; c.235dupC; c.244delC; c.247A>G; c.248A>C; c.248A>T; c.24dupG; c.25_26delAA; c.251delG; c.253_260delACCTTTGC; c.253delA; c.255dupC; c.257T>C; c.258dupT; c.265_266delCT; c.266_267delTG; c.266dupT; c.268A>C; c.269G>A; c.27_28insAGAA; c.270_271delTG; c.271G>T; c.275dupT; c.275T>C; c.277C>A; c.277C>T; c.279_280insGTGG; c.27dupG; c.28_29insTA; c.282_283dupTG; c.283G>C; c.287dupA; c.292_295dupCACG; c.293A>T; c.294C>G; c.295G>A; c.298G>A; c.298G>C; c.298G>T; c.299A>C; c.299A>G; c.299A>T; c.2T>A; c.2T>C; c.2T>G; c.301_302delCCinsTCTGAGAACTT; c.302C>T; c.304G>A; c.305A>G; c.306G>C; c.30dupT; c.312C>G; c.315+1delG; c.315+1G>A; c.315+1G>C; c.315+1G>T; c.315+2delT; c.315+2T>C; c.315+2T>G; c.315+5G>C; c.316-14T>G; c.316-1delG; c.316-1G>A; c.316-1G>C; c.316-1G>T; c.316-2A>C; c.316-2A>G; c.316-3C>A; c.319C>G; c.320T>A; c.320T>G; c.323dupG; c.328delG; c.328G>A; c.332T>C; c.339_345dupTGTGCTG; c.339T>A; c.33delC; c.33dupC; c.341T>A; c.343_344delCTinsG; c.344T>C; c.347C>A; c.349_350insTGAT; c.354delC; c.364G>A; c.364G>C; c.364G>T; c.36delC; c.371_378delCCCCACCA; c.375dupA; c.378delA; c.380T>G; c.382C>T; c.383_385delAGG; c.383A>C; c.383A>G; c.385_388delGCTGinsCCACA; c.388G>C; c.393T>A; c.394C>T; c.395dupA; c.396_397delGA; c.397A>T; c.3G>A; c.3G>C; c.3G>T; c.417dupT; c.421G>A; c.428C>A; c.429_430delCC; c.431A>G; c.431A>C; c.435G>C; c.436T>A; c.436T>C; c.437A>G; c.438T>A; c.439C>G; c.440_441dupAC; c.440A>C; c.440A>T; c.*6C>G; c.44delT; c.44dupT; c.45dupG; c.46delT; c.47G>A; c.48G>A; c.4delG; c.4G>T; c.50delG; c.50dupG; c.51delC; c.52A>T; c.59A>G; c.61G>A; c.64dupG; c.67G>T; c.70G>T; c.68_74delAAGTTGG; c.71_73delTTG; c.74delG; c.74delGinsCAC; c.75T>A; c.78dupT; c.79_80insT; c.79G>A; c.79G>T; c.80A>G; c.81G>C; c.82G>T; c.85delC; c.85dupC; c.86T>A; c.89delG; c.8A>C; c.8delA; c.90C>T; c.91A>C; c.91A>G; c.91delA; c.92+1G>A; c.92+1G>C; c.92+1G>T; c.92+2T>A; c.92+2T>C; c.92+2T>G; c.92+5G>A; c.92+5G>C; c.92+5G>T; c.92+6T>C; c.92G>A; c.92G>C; c.93-15T>G; c.93-1G>A; c.93-1G>C; c.93-21G>A; c.93-2A>C; c.93-2A>G; c.93-3T>G; c.94delC; c.316-106C>G; c.316-146T>G; c.316-197C>T; c.316-202delT; c.-142C>T.

Gene	Variants
PAH NM_000277.1	c.1138dupA; c.1139C>A; c.1154T>A; c.360G>A; c.504C>G; c.693delT; c.898G>A; c.1004A>C; c.1012G>T; c.1024delG; c.1025C>A; c.1027T>A; c.1027T>G; c.1028A>G; c.1029T>A; c.1030G>A; c.1033G>A; c.1033G>T; c.1038delG; c.1042C>G; c.1043T>C; c.1045T>C; c.1046C>A; c.1046C>T; c.1049C>A; c.1055delG; c.1063C>T; c.1065+1G>A; c.1065+1G>T; c.1065+3A>G; c.1066-10_1070delGGGCCTACAGTACTG; c.1066-11G>A; c.1066-1G>A; c.1066-1G>C; c.1066-1G>T; c.1066-2A>T; c.1066-3C>T; c.1068C>A; c.1068C>G; c.1069T>G; c.1074A>T; c.1076C>G; c.1085C>G; c.1089delG; c.1092_1094delTCT; c.1097C>A; c.1099delC; c.1101G>A; c.1102G>A; c.1109A>G; c.110T>C; c.1114A>T; c.1118C>A; c.111dupG; c.1123C>G; c.1129delT; c.1129T>G; c.1139C>T; c.1146C>G; c.1147C>T; c.1149delG; c.1153delC; c.1157A>G; c.116_118delTCT; c.1161C>A; c.1161C>G; c.1162G>A; c.1162G>C; c.1163T>C; c.1166delC; c.1169A>G; c.1171_1172delAG; c.1171A>G; c.1172G>C; c.1174T>A; c.117C>G; c.1180delG; c.1180G>C; c.1180G>T; c.1183G>C; c.1184C>A; c.1184C>G; c.1187A>G; c.1196T>C; c.1197A>T; c.1198delA; c.1199+17G>A; c.1199+1G>A; c.1199+1G>C; c.1199+2T>G; c.1199G>A; c.1199G>C; c.1200-1G>A; c.1200-1G>C; c.1200-2A>C; c.1200-2A>G; c.1200-8G>A; c.1208C>T; c.1209delT; c.1215_1219delAATAC; c.1216A>G; c.1217T>C; c.1218A>G; c.1219C>T; c.121C>T; c.1220C>T; c.1222C>T; c.1223G>A; c.1228T>A; c.1238G>C; c.1240T>C; c.1241A>G; c.1243G>A; c.1249T>C; c.1252A>C; c.1256A>G; c.1259G>T; c.125A>T; c.127G>T; c.1282C>T; c.1285C>A; c.1293_1294delGA; c.1298dupT; c.1301C>A; c.1304A>T; c.1306delT; c.1314_1315+4delCAGTAA; c.1315+1G>A; c.1315+1G>T; c.1315+2T>C; c.1315+4A>G; c.1315+6T>A; c.1316-1G>A; c.1316-2A>C; c.1318G>T; c.1339G>A; c.1340C>A; c.1355dupA; c.1357_*2delTAAAG; c.136G>A; c.136G>C; c.137delG; c.13delG; c.140C>T; c.143T>C; c.155delT; c.164T>C; c.165delT; c.165T>G; c.168_168+1delGGinsAA; c.168+1G>A; c.168+1G>T; c.168+2T>C; c.168+5G>A; c.168+5G>C; c.169-13T>G; c.169-2A>G; c.169G>A; c.169G>T; c.171_172delGA; c.183C>A; c.183C>G; c.184_185insCTGA; c.184delC; c.185T>C; c.187_190delACCCinsCCCA; c.189_190dupCC; c.190_194delCACAT; c.193A>G; c.194T>A; c.194T>C; c.196G>T; c.199T>C; c.1A>G; c.1A>T; c.204A>T; c.206dupC; c.208_210delTCT; c.212G>A; c.223G>A; c.224A>G; c.224A>T; c.226G>T; c.227A>G; c.239dupT; c.241A>C; c.242C>A; c.250G>T; c.253A>T; c.261C>A; c.266_267insG; c.266dupC; c.283A>T; c.284_286delTCA; c.292T>G; c.293T>C; c.2T>C; c.2T>G; c.301G>A; c.30dupC; c.311C>A; c.320A>G; c.329delC; c.32T>A; c.331C>T; c.344_347delAAGA; c.346_347delGA; c.350delC; c.353-1G>A; c.353-2A>G; c.355C>T; c.357delC; c.359G>A; c.365delC; c.385G>T; c.386A>G; c.398_401delATCA; c.3G>A; c.3G>C; c.400C>T; c.428A>G; c.434A>T; c.439C>T; c.440C>T; c.441+1G>A; c.441+1G>C; c.441+3G>C; c.441+4A>G; c.441+5G>T; c.441+6T>A; c.442-1G>A; c.442-2A>C; c.442-5C>G; c.442G>A; c.443G>A; c.450dupA; c.460T>C; c.461A>T; c.464G>A; c.464G>C; c.47_48delCT; c.47_48insCT; c.470_471delGAinsAC; c.470G>C; c.472C>T; c.473G>A; c.482T>C; c.48dupT; c.490A>G; c.493G>A; c.494C>A; c.498C>A; c.498C>G; c.500A>T; c.503delA; c.504C>A; c.505C>A; c.505C>T; c.506G>A; c.506G>C; c.508C>G; c.509+1G>A; c.510-2A>G; c.510T>A; c.510T>G; c.511G>A; c.514C>T; c.516G>C; c.516G>T; c.520A>G; c.521T>C; c.523C>T; c.526C>T; c.527G>A; c.527G>T; c.529G>A; c.529G>C; c.533A>G; c.535T>A; c.545A>G; c.547_548delGAinsTT; c.547G>T; c.556delA; c.558_559delAT; c.559T>C; c.561G>A; c.563G>A; c.569T>C; c.580_581delCT; c.581T>C; c.58C>T; c.590T>A; c.591G>C; c.60+1G>C; c.60+4A>T; c.60+5G>C; c.60+5G>T; c.601C>T; c.602A>G; c.610dupT; c.611A>G; c.612T>G; c.613G>A; c.618C>A; c.618C>G; c.620A>G; c.631C>A; c.632C>T; c.632delC; c.635T>C; c.638T>C; c.648C>G; c.653G>T; c.659A>C; c.662A>G; c.664_665delGA; c.665A>G; c.671T>C; c.673C>A; c.673C>G; c.674C>T; c.676C>A; c.676C>T; c.677_686delAGCTGGAAGA; c.682G>A; c.686dupA; c.688G>A; c.689T>C; c.691T>C; c.694C>G; c.694C>T; c.697T>A; c.707-1G>A; c.707-2delA; c.712A>C; c.712A>G; c.716G>T; c.721C>T; c.722delG; c.722G>A; c.722G>T; c.724C>T; c.727C>T; c.728G>A; c.731C>T; c.733G>C; c.734_735delTGinsCA; c.734T>A; c.734T>C; c.737C>A; c.737delC; c.739G>A; c.739G>C; c.740G>T; c.745C>T; c.745delC; c.754C>G; c.754C>T; c.755G>A; c.755G>C; c.757G>A; c.764T>C; c.770G>T; c.773T>C; c.775G>A; c.776C>T; c.781C>G; c.781C>T; c.782G>A; c.782G>C; c.785T>G; c.788T>C; c.790delC; c.794G>A; c.795C>A; c.796A>C; c.799C>G; c.799C>T; c.800A>G; c.800A>T; c.801delG; c.801G>C; c.803A>G; c.805A>C; c.806delT; c.806T>A; c.809G>A; c.812A>G; c.812A>Tc.813T>G; c.814G>T; c.818C>T; c.822_832delGCCCATGTATA; c.823C>T; c.824C>G; c.824C>T; c.826A>G; c.827T>A; c.829T>G; c.830A>G; c.833C>A; c.833C>G; c.833C>T; c.835C>G; c.836C>T; c.837delC; c.838G>A; c.839A>G; c.841C>G; c.841C>T; c.842+1G>A; c.842+1G>T; c.842+2T>A; c.842+3G>C; c.842+4A>G; c.842+5G>A; c.842C>T; c.843-2A>T; c.844G>A; c.847A>T; c.848T>A; c.856G>A; c.859C>G; c.865G>A; c.865G>C; c.868C>T; c.869A>T; c.875C>T; c.884C>G; c.887A>G; c.889C>T; c.890G>A; c.890G>T; c.895T>C; c.896T>G; c.898G>T; c.901C>A; c.901C>T; c.902A>C; c.903G>T; c.907delT; c.907T>C; c.910C>T; c.912+1G>A; c.912+2T>C; c.912G>A; c.913-2A>C; c.913-2A>G; c.913-7A>G; c.916A>G; c.916delA; c.926C>A; c.926C>T; c.929C>T; c.930_940delTCTGGGTGCAC; c.931_932delCT; c.932T>C; c.934G>T; c.940C>A; c.940C>T; c.941C>A; c.955G>T; c.956A>G; c.960G>C; c.964G>A; c.965C>G; c.967_969delACA; c.969+1G>A; c.969+2T>G; c.969+5G>A; c.970-1G>A; c.970-1G>C; c.970-1G>T; c.970-2A>G; c.974A>G; c.975C>G; c.976delT; c.977G>A; c.978G>A; c.992T>C; c.997C>T.
PMM2 NM_000303.2	c.447+5G>A; c.179-1G>C; c.192C>G; c.435C>G; c.661T>G; c.72delT; c.104T>A; c.109C>T; c.127G>A; c.131T>C; c.157C>T; c.160dupG; c.178+1G>A; c.178+2T>G; c.178G>T; c.179-1G>T; c.189delA; c.190delT; c.193G>T; c.1A>G; c.205C>T; c.24delC; c.255+1G>A; c.255+2T>C; c.256-1G>C; c.256-2A>G; c.26G>A; c.27C>A; c.28C>T; c.310C>G; c.317A>T; c.323C>T; c.324delG; c.337C>A; c.338C>T; c.345dupG; c.347+1G>A; c.348-1G>C; c.348-2A>C; c.349G>C; c.349G>T; c.355T>C; c.357C>A; c.359T>C; c.367C>T; c.368G>A; c.385G>A; c.392delC; c.394A>T; c.395T>C; c.414delA; c.415G>A; c.421C>T; c.422G>A; c.430T>C; c.432C>A; c.442G>A; c.448-1_448delGA; c.44G>C; c.451_454delGAAA; c.454_455delAA; c.458T>C; c.470T>C; c.484C>T; c.488_491delAAGA; c.511dupA; c.524-2A>G; c.53C>G; c.548T>C; c.55delG; c.560G>A; c.561G>A; c.563A>G; c.580C>T; c.58C>T; c.620T>C; c.623G>C; c.639+1G>A; c.640-9T>G; c.640G>A; c.647A>G; c.647A>T; c.652C>G; c.653A>T; c.66+1delG; c.66+1G>T; c.669C>G; c.67-2A>T; c.677C>G; c.682G>T; c.687C>G; c.691G>A; c.710C>G; c.710C>T; c.713G>A; c.722G>C; c.95_96delTAinsGC; c.95T>G; c.97C>T.
GAA NM_000152.4	c.-32-13T>G; c.766_785delTATATCACAGGCCTCGCCGAinsC; c.-32-1G>C; c.-32-2A>G; c.1001G>A; c.1003G>A; c.1004G>A; c.1030_1031delGG; c.1051delG; c.1062C>A; c.1064T>C; c.1075T>G; c.1076-1G>A; c.1076-22T>G; c.1076-2A>G; c.1082C>T; c.1099delT; c.1100G>A; c.1115A>T; c.1124G>T; c.1128_1129delGGinsC; c.1129G>C; c.1130delG; c.1134C>G; c.1143delC; c.1153delC; c.1156C>T; c.1165delG; c.118C>T; c.1192delC; c.1192dupC; c.1193delT; c.1193T>C; c.1194+2T>C; c.1195-1G>A; c.1195-2A>C; c.1195-2A>G; c.1210G>A; c.1211A>G; c.1222A>G; c.1225dupG; c.1226_1227insG; c.1292_1295dupTGCA; c.1309C>T; c.1316T>A; c.1320_1322delGAT; c.1326+1G>A; c.1326+2T>C; c.1327-2A>G; c.1377dupC; c.1378G>A; c.1396delG; c.1408_1410delAAC; c.1411_1414delGAGA; c.1426_1429delCTGAinsGTTGG; c.1431delT; c.1432G>A; c.1437+1G>A; c.1437+2T>C; c.1437G>A; c.1438-1G>C; c.1438-1G>T; c.1441delT; c.1441T>C; c.1445C>T; c.1447G>A; c.1464dupC; c.1465G>A; c.1478C>T; c.148G>T; c.1496G>A; c.1501_1515delGACATGGTGGCTGAG; c.1519dupC; c.1538A>G; c.1548G>A; c.1551+1G>C; c.1551+1G>T; c.1552-2A>G; c.1552-3C>G; c.1556T>C; c.1561G>A; c.1564C>A; c.1564C>G; c.1564C>T; c.1567delT; c.1579delA; c.1585_1586delTCinsGT; c.1585T>C; c.1634C>T; c.1650dupG; c.1654delC; c.1655T>C; c.1657C>T; c.1669A>T; c.1687C>T; c.1692delT; c.169C>T; c.1716C>G; c.1725C>A; c.172C>T; c.1735G>A; c.1754+1_1754+7delGTGAGGG;

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	c.1754+1G>A; c.1755-1G>A; c.1761_1765delGGTGAinsT; c.1781G>A; c.1796C>A; c.1798C>T; c.1799G>A; c.1802C>A; c.1802C>G; c.1802C>T; c.1804A>G; c.1820G>A; c.1822C>T; c.1824_1828dupATACG; c.1826dupA; c.1827_1828insA; c.1827delC; c.1832G>A; c.1836C>G; c.1841C>A; c.1841C>T; c.1843G>A; c.1844G>A; c.1847dupA; c.1856G>A; c.1857C>G; c.1880C>T; c.1888+1G>A; c.1895T>G; c.1905C>A; c.1910_1918delTGGGGGTGC; c.1912G>T; c.1913G>T; c.1927G>A; c.1933G>A; c.1933G>C; c.1933G>T; c.1935C>A; c.1941C>A; c.1941C>G; c.1942G>A; c.1943G>A; c.1951_1952delGGinsT; c.1952delG; c.1961C>A; c.1962_1964delAGA; c.1978C>T; c.1979G>A; c.1A>G; c.2012T>G; c.2014C>T; c.2015G>A; c.2024_2026delACA; c.2040+1delG; c.2040+1G>T; c.2041-1G>A; c.2061delC; c.2066_2070dupAGCCG; c.2074C>T; c.2078dupA; c.2082delC; c.2104C>T; c.2105G>A; c.2105G>C; c.2105G>T; c.2136_2137delGT; c.2140delC; c.2161delG; c.2161dupG; c.2167_2179delGTGGCCCGGCCCinsTGCGACGTGG; c.2173C>T; c.2185delC; c.2188G>T; c.2189+1G>T; c.2210C>A; c.2213G>A; c.2214G>A; c.2219_2220delTG; c.2227C>T; c.2228A>G; c.2236T>C; c.2236T>G; c.2237G>A; c.2237G>C; c.2238G>A; c.2238G>C; c.2242delG; c.2242dupG; c.2242G>T; c.2269C>T; c.2281delGinsAT; c.2284G>T; c.2297A>C; c.2300delT; c.2331+2T>A; c.2332-1G>C; c.236_246delCCACACAGTGC; c.2367dupA; c.2383G>T; c.2407C>T; c.241C>T; c.2432C>T; c.2479C>T; c.2495_2496delCA; c.2501_2502delCA; c.2512C>T; c.2544delC; c.2560C>T; c.258delC; c.258dupC; c.2608C>T; c.2617dupT; c.2646+2T>A; c.2662G>T; c.266G>A; c.2704_2716dupCAGAAGGTGACTG; c.2704C>T; c.2706delG; c.2740C>T; c.276C>A; c.281_282delCT; c.2815_2816delGT; c.2846T>A; c.2853delG; c.307T>C; c.307T>G; c.342delG; c.343C>T; c.352C>T; c.365delT; c.376delT; c.377G>A; c.378G>A; c.379_380delTG; c.393delC; c.399C>A; c.3G>A; c.437delT; c.448dupG; c.461_469delGTACCACCC; c.461G>C; c.471delC; c.482_483delCC; c.4G>T; c.525_526delTG; c.525delT; c.546+2_546+5delTGGG; c.546G>A; c.546G>C; c.546G>T; c.55delG; c.569G>A; c.573C>A; c.643G>T; c.655G>A; c.670C>T; c.671G>A; c.671G>C; c.692+1G>A; c.692+1G>C; c.692+2T>C; c.698delA; c.711_712dupGC; c.716delT; c.722_723delTT; c.727G>A; c.736delC; c.743T>C; c.755dupT; c.759delC; c.761C>T; c.766_767insC; c.768dupT; c.784G>A; c.796C>T; c.799_803delCTGATinsA; c.836G>A; c.841C>T; c.853C>T; c.854C>G; c.858+2T>A; c.896T>C; c.896T>G; c.925G>A; c.934delC; c.953T>A; c.953T>C; c.955+1G>A; c.971C>T; c.989G>A; c.2647-1_2648delGAA; c.2647-7G>A; c.2655_2656delCG; c.875A>G; c.876C>G; c.877G>A.
AGXT NM_000030.2	c.1007T>A; c.1014C>G; c.1045G>A; c.1049G>A; c.106C>T; c.1071+1G>A; c.1076T>C; c.1079G>A; c.1079G>C; c.107G>A; c.1084G>A; c.1102G>A; c.1110_1111delCG; c.1125_1126delCG; c.1148C>A; c.1151T>C; c.116_117dupCA; c.121G>A; c.122G>A; c.122G>T; c.125G>A; c.126delG; c.126dupG; c.130C>T; c.139G>A; c.166-1G>A; c.166-2A>G; c.167T>A; c.175G>A; c.187G>C; c.198C>G; c.2_3delTGinsAT; c.205C>T; c.209C>A; c.215dupA; c.221_227dupTCACACT; c.242C>A; c.242C>T; c.244G>C; c.245G>A; c.248A>G; c.254C>A; c.276delT; c.283_285dupGAG; c.283G>A; c.28C>T; c.2T>C; c.302T>C; c.32_33delCC; c.322T>C; c.323G>A; c.324G>T; c.326G>T; c.327delG; c.32C>G; c.331C>T; c.332G>A; c.335C>A; c.33delC; c.33dupC; c.346G>A; c.349G>T; c.358+1G>T; c.358+2T>G; c.364C>T; c.374_381delCGATGACC; c.3G>T; c.406_410dupCTGCA; c.409C>T; c.416_418delTGG; c.423+1G>A; c.423G>T; c.424-2A>G; c.445delG; c.447_454del; c.449T>C; c.454T>A; c.457T>G; c.460delA; c.466G>A; c.466G>C; c.473C>A; c.473C>T; c.481G>A; c.481G>C; c.481G>T; c.497T>C; c.508G>A; c.518G>A; c.519C>A; c.524+2T>A; c.525-1G>A; c.533G>A; c.547G>A; c.557_562delCATCCCinsATCGGT; c.560C>T; c.568G>A; c.570delG; c.577delC; c.577dupC; c.583A>C; c.584T>G; c.595G>A; c.596-2A>G; c.603C>A; c.605T>A; c.612C>A; c.613T>C; c.614C>A; c.614C>T; c.628G>C; c.642_645delTCCA; c.646G>A; c.653C>T; c.661T>C; c.673_676delAAGG; c.679_680+2delAAGT; c.67C>T; c.680+1G>A; c.680+1G>C; c.680+2T>A; c.681-1G>T; c.686_688delAGA; c.697C>T; c.698G>A; c.698G>T; c.725dupT; c.727G>C; c.731T>C; c.737G>A; c.738delC; c.744delC; c.74T>G; c.751_752delTGinsAA; c.752G>A; c.753G>A; c.757T>C; c.776+1G>A; c.776+1G>C; c.776+2T>G; c.777-1G>C; c.777-2A>G; c.77T>C; c.783T>A; c.798_802delCATCAinsACAATCTCAG; c.806T>C; c.822G>C; c.823_824dupAG; c.823A>C; c.834delC; c.83delC; c.841G>T; c.844C>T; c.846+1G>A; c.846+1G>T; c.846G>C; c.847-1G>C; c.847-2_847-1delAG; c.847-3C>G; c.851T>C; c.853G>T; c.860_861delGCinsCG; c.891T>G; c.893T>C; c.907C>T; c.919delC; c.922C>T; c.942+1G>T; c.943-1G>A; c.943-1G>T; c.956C>T; c.959_960delCA; c.971_972delTG; c.973delG; c.976delG; c.983_988delCTGGCT; c.996G>A; c.997A>T; c.449T>C; c.454T>A; c.457T>G; c.460delA.
HADHA NM_000182.4	c.2114T>A; c.1981_1999delCTGCCTCCTAAGTCTGAAG; c.2020C>T; c.2200A>T; c.1025T>C; c.1052delA; c.1070delC; c.1086-3_1092delCAGGCATCTG; c.1132C>T; c.1195C>T; c.1202delA; c.1221-1G>C; c.1237A>T; c.129dupT; c.1344_1345delGT; c.1422dupT; c.1427_1428delCA; c.1479+1G>T; c.1528G>C; c.157C>T; c.1590delC; c.1620+2_1620+6delTAAGG; c.1637A>G; c.1644delC; c.1645A>G; c.1668_1674dupTGAAGTC; c.1678C>T; c.1689+2T>G; c.1690-2A>G; c.1720_1721delCT; c.1793_1794delAT; c.180_180+5delGGTATCinsAT; c.180+1G>A; c.180+3A>G; c.1811delG; c.1814_1815delAA; c.1885G>A; c.1915_1918delTATC; c.1916_1919dupATCA; c.1918C>T; c.1959dupT; c.1967delT; c.1A>G; c.2000+1G>C; c.2026delC; c.2027G>A; c.2059delA; c.2071G>T; c.2132dupC; c.2134_2138dupCCTTG; c.2146+1G>A; c.2146+2T>C; c.2220T>A; c.2222delG; c.2225_2228dupAACA; c.2227_2228delCA; c.240G>A; c.274_278delTCATC; c.315-1G>A; c.3G>A; c.403A>G; c.407C>G; c.499delA; c.515delC; c.539C>T; c.574-2A>G; c.612dupT; c.64C>T; c.676+1G>A; c.676+2T>C; c.677-1G>A; c.703C>T; c.72delT; c.800-1_801delGAA; c.800-1G>T; c.844_845insA; c.845T>A; c.871C>T; c.914T>A; c.914T>C; c.919-2A>G.
CBS NM_000071.2	c.536_553delACGTGCTGCGGGCACTGG; c.18_36delCCAGGCAGAAGTGGGGCCC; c.1005_1006delCCinsTT; c.1006C>T; c.1007G>A; c.1009_1012delATGC; c.1039G>A; c.1051G>C; c.1058C>T; c.1087delG; c.1109G>A; c.1111G>A; c.1126G>A; c.1135C>T; c.1136G>A; c.1150A>G; c.1152G>C; c.1218delG; c.1219_1223+8delCCCTGGTAAGACC; c.1221delC; c.1223+1G>T; c.1223G>A; c.1224-2A>C; c.1226G>A; c.1227G>A; c.1280C>T; c.1316G>A; c.1321A>T; c.1330G>A; c.1358+1G>A; c.1358+2T>C; c.1359-1G>C; c.1468-1G>T; c.146C>T; c.1498_1499delTC; c.153_165delGTGCACCTGGCAG; c.1545delG; c.1552+1G>A; c.156C>A; c.1603_1604delAC; c.162G>A; c.19delC; c.19dupC; c.208_209+8delCCGTAAGTCC; c.209+1G>A; c.209+1G>C; c.209+2T>C; c.233C>G; c.253G>A; c.284T>C; c.28delG; c.302T>C; c.306G>C; c.316+1G>A; c.325T>C; c.340G>A; c.341C>T; c.346G>A; c.361C>T; c.362G>A; c.371_374dupTGCG; c.373C>T; c.393G>C; c.402delG; c.403_404delACinsG; c.415G>A; c.429C>G; c.430G>A; c.434C>T; c.442G>A; c.451+1G>T; c.451G>A; c.457G>A; c.463G>A; c.467delT; c.476_477insCAGGGCCC; c.494G>A; c.502G>A; c.526G>A; c.526G>T; c.532-2A>G; c.572C>A; c.572C>T; c.667-14_667-7delCTCTTTCT; c.676G>A; c.684C>A; c.684C>G; c.689delT; c.700G>A; c.707_708delCCinsGGTG; c.736+2T>G; c.737-1G>C; c.738delG; c.770C>T; c.785C>T; c.797G>A; c.816T>A; c.828+1G>A; c.829-1G>C; c.833C>T; c.903C>G; c.904G>A; c.919G>A; c.941T>C; c.954+1G>A; c.954+2T>G; c.959T>C; c.969G>A; c.992C>A; c.1553-1G>C; c.1553-2A>C; c.1566delG; c.1576C>T.
ARSA NM_000487.5	c.257G>T; c.270C>A; c.774delA; c.1007T>C; c.1010A>T; c.1036delG; c.1079T>C; c.1087dupC; c.109_116delGACCTGGG; c.1107+1delG; c.1108-2A>G; c.1114C>T; c.1123_1126delCTCT; c.1125_1126delCT; c.1210+1G>A; c.1210+1G>T; c.1222_1232delAGTGATACCAC; c.1223_1231delGTGATACCA; c.1229C>T; c.1232C>T; c.1264delC; c.1269delT; c.1274A>G; c.1279C>A; c.1283C>T; c.1323delC; c.1337delG; c.1344dupC; c.1366C>T; c.1378C>T; c.1388delT; c.1408_1418delGCAGCTGTGAC; c.1447G>T; c.1462C>T; c.1489_1492dupCCCC; c.1492dupC; c.157C>T; c.179G>C;

	c.185_186dupCA; c.195delC; c.200delC; c.206_209delCTCT; c.211_212delTG; c.224+1G>A; c.225-2A>G; c.227_228insTA; c.229G>C; c.240dupC; c.244delC; c.245G>A; c.256C>T; c.257G>A; c.263G>A; c.292_293delTCinsCT; c.293C>T; c.295dupC; c.2T>C; c.302delG; c.302dupG; c.302G>A; c.302G>T; c.335T>A; c.346C>T; c.34delG; c.370G>A; c.371G>A; c.410T>C; c.418delC; c.418dupC; c.421C>T; c.433C>G; c.433C>T; c.442G>A; c.449C>G; c.449C>T; c.451dupT; c.465+1G>A; c.465+2T>A; c.466-2A>G; c.467delG; c.467G>A; c.470C>G; c.474C>A; c.494dupC; c.495_501delGCCGGCC; c.506C>G; c.526C>T; c.542dupT; c.542T>G; c.545C>G; c.545delC; c.582delC; c.582dupC; c.583delT; c.641C>T; c.674_675dupAT; c.677C>A; c.677C>T; c.684+1G>A; c.685-1G>A; c.697C>A; c.700C>T; c.712C>T; c.736C>T; c.737G>A; c.739G>A; c.746T>C; c.758dupT; c.763G>A; c.769G>C; c.827C>A; c.827C>T; c.836_837delTCinsAA; c.847G>T; c.852T>A; c.854+1G>A; c.854+1G>T; c.855-1G>A; c.862A>C; c.868C>T; c.869G>A; c.877C>T; c.883G>A; c.890C>A; c.891delC; c.899T>C; c.905G>A; c.905G>T; c.917C>T; c.925dupG; c.925G>A; c.925G>T; c.926A>T; c.929delG; c.929G>T; c.931G>A; c.931G>C; c.937C>T; c.938G>A; c.942G>T; c.956_958delTCT; c.960G>A; c.979_979+3delGGTC; c.979+1G>A; c.979G>A; c.980-2A>C; c.986C>T; c.98T>C; c.991G>T; c.1136C>T; c.1150G>A; c.1174C>T; c.1175G>A; c.1201_1203delTTC; c.608A>G; c.622delC.
BTD NM_001281723.2	c.1213T>G; c.1461C>G; c.1007_1008delTT; c.1007T>A; c.104_110delGCGGCTGinsTCC; c.1055delC; c.1058delC; c.106_107delGG; c.107_110delGCTG; c.1087T>G; c.1091T>A; c.110_111delGT; c.1102_1103dupTC; c.1102T>C; c.1132C>T; c.1135G>T; c.113dupA; c.1163G>A; c.1164G>A; c.1176_1177dupTC; c.1197_1198delGA; c.1220T>C; c.1233_1247delGGGAAAGGAAGGCTAinsTTCCAATGGCC; c.124_125delCT; c.1245delC; c.1247_1258delATCTCCACGTCT; c.1255G>T; c.1258T>C; c.1259G>C; c.1270dupC; c.1273T>C; c.1274G>C; c.1277G>A; c.1277G>C; c.1281T>G; c.1290C>A; c.1313_1314delAG; c.1319A>G; c.1320T>A; c.1330delG; c.1334T>C; c.1336G>C; c.1339G>A; c.1340G>A; c.1340G>T; c.1345C>T; c.1358_1359delGC; c.1358G>A; c.1364dupA; c.1367A>G; c.1374A>C; c.1375G>A; c.1378dupT; c.1390delA; c.1400dupG; c.1416dupC; c.142G>T; c.1453G>T; c.1464delG; c.1465delT; c.1466G>A; c.1467G>A; c.1485delC; c.1495C>T; c.1499dupT; c.1514_1518delGGATG; c.1517T>A; c.1531C>T; c.1532C>G; c.1561dupT; c.1563T>G; c.1601C>T; c.1607C>T; c.1616G>A; c.1618C>T; c.1619G>A; c.1622dupT; c.1633G>C; c.1634A>G; c.1634A>T; c.1635C>A; c.166G>T; c.177G>C; c.190G>A; c.190G>T; c.196G>A; c.198G>C; c.200A>G; c.208_211dupATCC; c.241C>T; c.242G>A; c.251C>A; c.251C>T; c.253_261delTTGGAGCTC; c.263T>G; c.268C>T; c.269delA; c.272delA; c.284A>G; c.289C>T; c.304G>A; c.305C>T; c.314delA; c.315+1G>A; c.315+1G>T; c.316-1G>T; c.316G>T; c.322C>T; c.327T>G; c.328A>G; c.329_330dupTA; c.332dupT; c.332T>G; c.340G>A; c.340G>C; c.347G>T; c.362A>G; c.378_381dupCATT; c.388T>G; c.399delC; c.412delC; c.413dupA; c.426G>A; c.430C>A; c.434G>T; c.43A>T; c.43C>T; c.44_44+3delGGTA; c.44+1delG; c.44+1G>A; c.44+1G>C; c.44+1G>T; c.448C>T; c.449G>A; c.451T>C; c.460A>C; c.461C>G; c.465G>A; c.466-2A>G; c.472C>T; c.475C>T; c.476G>A; c.485G>T; c.496_497delAG; c.517G>A; c.524T>G; c.534G>T; c.550delA; c.562T>G; c.563G>A; c.565C>T; c.588C>G; c.589A>G; c.590A>G; c.593C>G; c.593delC; c.594_608delAAATGTCGTGTTCAG; c.600_602delCGT; c.600delC; c.601G>A; c.611A>T; c.631C>T; c.632G>A; c.635A>G; c.637C>T; c.637delC; c.638G>A; c.643delC; c.649C>T; c.652T>A; c.655_657delTTTinsAGTA; c.658G>C; c.660G>C; c.670G>A; c.670G>C; c.688G>T; c.689A>G; c.699delC; c.701T>C; c.707C>T; c.715G>A; c.740G>A; c.749T>C; c.750A>G; c.761A>G; c.764C>T; c.771C>G; c.776T>A; c.789C>G; c.800A>T; c.820T>G; c.838C>G; c.842T>A; c.842T>G; c.862A>G; c.864delA; c.871C>G; c.872C>T; c.893T>G; c.901G>C; c.902C>T; c.904A>C; c.935G>A; c.938G>A; c.939delT; c.939T>G; c.998_1007delACCTTATAAT.
SLC26A2 NM_000112.3	c.117delC; c.1303dupA; c.1639C>T; c.166delAinsCTC; c.1714delC; c.223C>T; c.294G>A; c.2T>C; c.1800C>G; c.854C>A; c.909T>A; c.1020_1022delTGT; c.1060G>T; c.1157C>T; c.1242_1245delAAAC; c.1273A>G; c.1311dupT; c.1338delA; c.1343C>G; c.1361A>C; c.138dupT; c.1394delT; c.1421delT; c.1432delC; c.1441delA; c.1451G>A; c.145delA; c.15_19delTAAAG; c.1535C>A; c.1537_1541dupGTTAT; c.1565C>T; c.1596delT; c.1607_1610delGTTT; c.1649delA; c.1650delG; c.1655C>A; c.1707C>G; c.1724delA; c.1764C>A; c.1777G>T; c.1806_1809delAACT; c.1817delC; c.1831delG; c.1859delA; c.185C>G; c.1878delG; c.188delA; c.1926delT; c.1950delG; c.1955_1958delACTG; c.1956_1957dupCT; c.1957T>A; c.1976delT; c.1982delC; c.1983delA; c.1987A>G; c.1998_1999delAC; c.2017_2018delGA; c.2033G>T; c.2069_2070delTG; c.207delT; c.2120_2121delTT; c.2144C>T; c.235C>T; c.239_243dupGCAGT; c.255delC; c.299delC; c.325_326delTT; c.331G>T; c.374A>T; c.387T>G; c.391delC; c.398C>T; c.3G>T; c.403C>A; c.438delT; c.438dupT; c.451delT; c.47C>G; c.485_486delTG; c.496G>A; c.499delG; c.532C>T; c.541C>T; c.55G>T; c.578_581delCCTT; c.611T>G; c.63_64delCA; c.642_643delAA; c.697C>T; c.699+2T>C; c.700-1G>C; c.700-2A>G; c.705_711delGATGGGC; c.736_739delGTCT; c.746C>G; c.819delT; c.835C>T; c.870delG; c.906_907delCT; c.918delC; c.922delA; c.925delC; c.931T>C.
CFTR NM_000492.3	c.164+3_164+4insT; c.-9_14delCGAGAGACCATGCAGAGGTCGCC; c.459_476delAATAGCTATGTTTAGTTT; c.723_743+1delGAGAATGATGATGAAGTACAGG; c.1558G>A; c.1684G>C; c.2657+2_2657+3insA; c.2909-15T>G; c.2916_2952delTCTTAATAGATTCTCCAAAGATATAGCAATTTTGGAT; c.3038C>A; c.3718-2477C>T; c.1579_1584+11del; c.330C>A; c.2816A>G; c.2393dup; c.577G>A; c.1742dup; c.1652delG; c.2777delT; c.3000_3014del; c.926C>A; c.1713_1714delAG; c.1546A>G; c.358G>A; c.1000C>T; c.1001G>A; c.1001G>T; c.1006_1007insG; c.1007T>A; c.1008_1019dupATTCACCACCAT; c.1013C>T; c.1021_1022dupTC; c.1021T>C; c.1029delC; c.1037T>C; c.1040G>A; c.1040G>C; c.1040G>T; c.1045G>C; c.1046C>T; c.1054C>T; c.1055G>A; c.1057C>T; c.1075C>A; c.1079C>A; c.1081delT; c.1081T>C; c.1083delG; c.1084_1088dupTATGA; c.1093_1094delCT; c.1115delA; c.1116+1G>A; c.1116+1G>T; c.1116G>T; c.1117-1G>A; c.1117-2A>G; c.1117G>A; c.1126C>T; c.1130dupA; c.1135G>T; c.1141A>T; c.114C>G; c.1155_1156dupTA; c.115C>T; c.1162_1168delACGACTA; c.1197delC; c.11C>A; c.1202G>A; c.1203G>A; c.1209+1G>A; c.1209+1G>T; c.1209+2T>G; c.1209G>A; c.1209G>C; c.1210-2_1210-1delAG; c.1210-2A>C; c.1211delG; c.1219G>T; c.1234_1238delGCAAA; c.1235delC; c.1239dupA; c.1240C>T; c.1248dupT; c.1301_1307delCACTTCT; c.1301C>A; c.1301C>G; c.1315C>T; c.131A>G; c.1324A>T; c.1327_1330dupGATA; c.1327G>T; c.1330_1331delAT; c.1340delA; c.1347_1350delAAGA; c.1358T>C; c.1364C>A; c.1365_1366delGG; c.1367T>C; c.1373delG; c.1373G>T; c.1375_1383delTCCACTGGA; c.137C>A; c.137C>T; c.1390A>G; c.1391A>C; c.1392+1G>A; c.1392G>T; c.1393-1G>A; c.1393-2A>G; c.1394C>A; c.1397C>A; c.1397C>G; c.1397C>T; c.1399C>T; c.1400T>C; c.1405A>G; c.1409_1418delTGATTATGGG; c.1415dupT; c.1418delG; c.1420G>A; c.1420G>T; c.1438G>T; c.1466C>A; c.1469delT; c.147_150delATCT; c.1471T>C; c.1475C>T; c.1477_1478delCA; c.1477C>T; c.1486delT; c.1487G>A; c.148T>C; c.14C>T; c.1505T>A; c.1505T>C; c.1505T>G; c.1510G>T; c.1516A>C; c.1517T>C; c.1518C>G; c.1519_1521delATC; c.1519A>T; c.1521_1523delCTT; c.1525G>C; c.1530_1531delTT; c.1538A>G; c.1543_1555delTATAGATACAGAA; c.1545_1546delTA; c.1546_1548delAGAinsT; c.1550A>G; c.1558G>T; c.1559T>A; c.1572C>A; c.1573C>T; c.1580A>G; c.1584+1G>A; c.1584+2T>C; c.1585-1G>A; c.1585-2A>G; c.1585-8G>A; c.1616_1617dupTA; c.1624G>T; c.164+1G>A; c.164+1G>C; c.164+1G>T; c.164+2T>C; c.1642_1643delCT; c.1645A>C; c.1646G>A; c.1646G>T; c.1647T>A; c.1647T>G; c.1648G>T; c.165-1G>A; c.165-2A>C; c.165-2A>G; c.165-2delA; c.165-3C>T; c.1650delA; c.1651G>A; c.1652G>A; c.1654C>A; c.1654C>T; c.1656delA; c.1657C>T; c.166G>A; c.166G>T; c.1670delC; c.1673T>C; c.1674delA; c.1675G>A;

c.1679+1G>A; c.1679+1G>C; c.1679G>A; c.1679G>C; c.1680-1G>A; c.1680-1G>C; c.1680A>C; c.1682C>A; c.1687T>A; c.1687T>C; c.1687T>G; c.168delA; c.1692delA; c.1694A>G; c.169T>G; c.1700A>C; c.1703delT; c.1703T>A; c.1705T>C; c.1705T>G; c.1706A>G; c.1707T>A; c.170G>A; c.1712T>C; c.171G>A; c.1721C>A; c.1724T>A; c.1731C>A; c.1733_1734delTA; c.1736A>G; c.174_177delTAGA; c.1753G>T; c.175dupA; c.1760T>C; c.1763A>T; c.1766+1G>A; c.1766+1G>C; c.1766+1G>T; c.1766+2T>A; c.1766+2T>C; c.1766+3A>C; c.1766+3A>G; c.1766+5G>T; c.1766G>A; c.1766G>T; c.1767-1G>A; c.178G>A; c.178G>T; c.179_180delAG; c.1792_1798delAAAACTA; c.1801A>T; c.1801delA; c.1811C>T; c.1826A>G; c.1826A>T; c.1837G>A; c.1841A>G; c.1853T>C; c.1865G>A; c.1882G>A; c.1882G>C; c.1904delA; c.1911delG; c.1920_1921dupTA; c.1923_1931delCTCAAAACTinsA; c.1936G>T; c.1943A>T; c.1943delA; c.1966G>T; c.1973_1985delGAAATTCAATCCTinsAGAAA; c.1979C>G; c.1981delA; c.1986_1989delAACT; c.1990G>T; c.19G>T; c.1A>G; c.200C>T; c.2012delT; c.2017G>T; c.2036G>A; c.2044delA; c.2044dupA; c.2045delC; c.2045dupC; c.2051_2052delAA; c.2051_2052delAAinsG; c.2051_2052dupAA; c.2052delA; c.2052dupA; c.2053C>T; c.2053dupC; c.2065C>T; c.2083dupG; c.2089delA; c.2089dupA; c.2125C>T; c.2126G>A; c.2128A>T; c.2143C>T; c.2145_2146delAAinsGT; c.2146A>T; c.2158C>T; c.2173G>A; c.2175dupA; c.217delC; c.2184_2185insA; c.2195T>G; c.2203delA; c.220C>T; c.2215delG; c.2233G>T; c.223C>T; c.2241_2248delGATACTGC; c.2249_2256delCTCGCATC; c.2249C>T; c.2252G>T; c.2290C>T; c.2291delG; c.2299C>T; c.2324_2325delAC; c.2327C>G; c.233delT; c.233dupT; c.234dupC; c.2353C>T; c.236G>A; c.2373delA; c.2374C>T; c.2395C>T; c.2417A>G; c.2423_2424dupAT; c.2428A>T; c.2429delG; c.243delT; c.2443delG; c.2443G>T; c.2450delG; c.2453delT; c.2463_2464delTG; c.2464G>T; c.2475_2478dupCGAA; c.2476G>T; c.2479G>T; c.2483dupA; c.2488A>T; c.2490+1G>A; c.2490+1G>T; c.2490+2T>C; c.2491-2A>C; c.2491G>T; c.2496C>A; c.2502delT; c.2502dupT; c.2508delT; c.2537G>A; c.2538G>A; c.253G>T; c.2547C>A; c.254G>A; c.254G>T; c.2551C>T; c.2552G>T; c.2554dupT; c.2556T>G; c.2573delG; c.2583delT; c.2589_2599delAATTTGGTGCT; c.25dupG; c.2600T>A; c.2600T>G; c.2601dupA; c.2619+1G>A; c.262_263delTT; c.2620-2A>G; c.2639_2640dupTG; c.263T>A; c.263T>G; c.2644_2645dupTG; c.2645G>A; c.2657+1_2657+2insA; c.2657+5G>A; c.2658-1G>C; c.2658-1G>T; c.2658-2A>G; c.2668C>T; c.2700T>A; c.271G>A; c.2723C>A; c.273+1G>A; c.273+3A>C; c.2735C>A; c.2737_2738insG; c.2738A>G; c.2739T>A; c.273G>C; c.274-1G>A; c.274-1G>C; c.274-1G>T; c.274-2A>C; c.274-2A>Gc.274-5_274-2delTGTA; c.274G>A; c.274G>T; c.2757C>G; c.2758G>A; c.2763_2764dupAG; c.2768C>A; c.2780T>C; c.2797A>G; c.2805_2810delACCACTinsTCAGA; c.2810dupT; c.2812dupG; c.2813T>G; c.2825delT; c.2834C>T; c.2845C>T; c.2854delA; c.2855T>C; c.2856C>G; c.2869_2870insG; c.2875delG; c.2879_2882delCTAT; c.2896delA; c.2900T>C; c.2908+1G>A; c.2908G>C; c.2909-1G>C; c.2909delG; c.2909G>A; c.2924_2925delGA; c.292C>T; c.2930C>T; c.2932A>T; c.2936A>C; c.2936A>T; c.2939T>A; c.293A>G; c.296C>T; c.2977G>T; c.2982_2988+2delCATCCAGGT; c.2986dupC; c.2988+1G>A; c.2988+1G>C; c.2988+1G>T; c.2988+2T>C; c.2988G>A; c.2989-1G>A; c.2989-2A>G; c.2989-2A>T; c.2993delT; c.2997_3000delAATT; c.2T>A; c.2T>C; c.2T>G; c.3002_3003delTG; c.3011_3019delCTATAGCAG; c.3011delC; c.3014dupT; c.3014T>G; c.3017C>A; c.3022delG; c.3032T>G; c.3039delC; c.3039dupC; c.303dupA; c.3047T>C; c.305T>G; c.3067_3072delATAGTG; c.3068_3072delTAGTG; c.3068T>G; c.3095A>G; c.3103C>T; c.3107C>A; c.3107C>T; c.310delA; c.3110C>A; c.3124C>T; c.3134C>A; c.3139_3139+1delGG; c.3139+1delG; c.3139+1G>A; c.313delA; c.3140-1G>A; c.3140-26A>G; c.3154T>G; c.3160C>G; c.3161delA; c.3181G>C; c.3184_3188dupCTATG; c.3188G>A; c.3189G>A; c.319_326delGCTTCCTA; c.3194T>C; c.3196C>T; c.3197G>A; c.3197G>T; c.3200C>T; c.3205G>A; c.3209G>A; c.3211C>T; c.3212A>C; c.3217dupT; c.3229_3230delCT; c.3230T>C; c.3231_3232delGT; c.323C>T; c.325_327delTATinsG; c.3254A>G; c.325T>C; c.3263dupA; c.3264delC; c.326A>G; c.3266G>A; c.3276C>A; c.3276C>G; c.327T>A; c.328G>C; c.328G>T; c.3292T>C; c.3293G>A; c.3294G>A; c.3294G>C; c.3294G>T; c.3297C>A; c.3299A>C; c.3302T>A; c.3302T>G; c.3304A>T; c.3310G>T; c.331C>G; c.3324delC; c.332C>T; c.3343dupA; c.3353C>T; c.3367+1G>A; c.3367+2T>A; ; c.3368-2A>G; c.3368-2A>T; c.3400_3401delACinsGTA; c.3404T>A; c.3410T>G; c.3415A>G; c.3419_3421delTGA; c.3421_3424dupAGTA; c.3435G>A; c.3454G>C; c.3458T>A; c.3461A>G; c.3468+1G>A; c.3468+1G>T; c.3468+2dupT; c.3468+2T>C; c.3468+5G>A; c.3468G>A; c.3469-2A>G; c.346G>A; c.3472C>T; c.3475T>C; c.3476C>T; c.3484C>T; c.3492dupT; c.3486_3487delAG; c.3495delG; c.349C>G; c.349C>T; c.350G>A; c.350G>C; c.350G>T; c.3528delC; c.3529A>T; c.3530delA; c.3532_3535dupTCAA; c.3533C>A; c.3536_3539delCCAA; c.3540delA; c.3546C>G; c.3546delC; c.3569_3570delTT; c.3587C>G; c.3598_3599delAAinsG; c.3599_3600delAAinsG; c.3605delA; c.3611G>A; c.3612G>A; c.3617C>A; c.3617C>G; c.3618_3619delAG; c.3623delG; c.3659delC; c.366T>A; c.3691delT; c.3700A>G; c.370G>C; c.3712C>T; c.3717+1G>A; c.3717+4A>G; c.3717+5G>A; c.3717G>A; c.3718-1G>A; c.3718-3T>G; c.3719T>G; c.3728T>A; c.3731G>A; c.3737C>T; c.3739G>A; c.3741_3742insATCAG; c.3744delA; c.3745G>A; c.3745G>C; c.3746G>A; c.3747delG; c.3752G>A; c.3761T>G; c.3764C>A; c.3763T>C; c.3767dupC; c.376C>A; c.3773dupT; c.377G>A; c.3796G>T; c.380_382dupTAT; c.3806T>A; c.3808delG; c.3808G>A; c.3822G>A; c.3838C>T; c.3841C>T; c.3844T>G; c.3846G>A; c.3851_3852dupAA; c.3848G>T; c.3857T>C; c.3859G>T; c.3872A>G; c.3873+1G>T; c.3873+1G>A; c.3873+2T>C; c.3873G>C; c.3874-1G>A; c.3874-2A>G; c.3874-2A>C; c.3874-2A>G; c.3883_3884insG; c.3883_3886delATTT; c.3883delA; c.3889dupT; c.3891dupT; c.38C>T; c.3907A>C; c.3900dupT; c.3908A>T; c.3908delA; c.3909C>G; c.3922G>T; c.3925C>T; c.3929G>A; c.3937C>T; c.3944_3951delTATGGAAA; c.3947G>A; c.3957_3958insAGGG; c.3963+1G>A; c.3963+1G>C; c.3963+2T>A; c.3963+2T>G; c.3971T>C; c.3972delC; c.3988_3989delCA; c.3988C>T; c.3997G>T; c.3999delG; c.3G>A; c.4004T>C; c.4028delG; c.4035_4038dupCCTA; c.4036_4042delCTAAGCC; c.4046G>A; c.405_406dupAC; c.4051A>T; c.4056G>T; c.4058delT; c.4071delT; c.4077_4080delTGTTinsAA; c.4086dupT; c.4078delG; c.409_412delCTCC; c.4097T>C; c.409delC; c.40A>T; c.410_416delTCCTACA; c.410T>C; c.4111G>T; c.4124A>C; c.4127_4131delTGGAT; c.4129G>C; c.413_415dupTAC; c.4136+1G>A; c.4143C>G; c.4144C>T; c.4147dupA; c.4168C>T; c.416A>G; c.4197_4198delCT; c.4199delG; c.4200_4201delTG; c.4225G>T; c.422C>A; c.4231C>T; c.4234C>T; c.4242+1G>A; c.4242+1G>T; c.4243-2A>G; c.4242+2T>C; c.424delA; c.4251delA; c.4252delG; c.4252G>T; c.4272C>A; c.4272C>G; c.4276T>C; c.4280T>C; c.4297G>T; c.429delT; c.4300_4301dupAG; c.4303dupA; c.4307_4320delTCTTCCGGCCAAGCC; c.4329delC; c.4339delG; c.433delC; c.4364C>A; c.4364C>G; c.4389delG; c.4400delT; c.4417G>T; c.4426C>T; c.442delA; c.443T>A; c.445G>A; c.445G>T; c.44T>C; c.454A>G; c.470_483delTTAGTTTGATTTAT; c.476T>C; c.481T>G; c.483_484insCC; c.488delA; c.489+1G>T; c.489+2T>C; c.489+2T>G; c.489+3A>G; c.49_50dupTT; c.490-1G>A; c.490-1G>T; c.494T>C; c.4C>T; c.4delC; c.506dupG; c.509G>A; c.50delT; c.50dupT; c.51delC; c.523A>G; c.53+1G>T; c.531delT; c.531dupT; c.532G>A; c.543_546delTAGT; c.544dupA; c.560delA; c.567C>A; c.56G>A; c.571T>G; c.575A>G; c.577G>T; c.579+1G>A; c.579+1G>T; c.579+3A>G; c.579+5A>G; c.57G>A; c.57G>T; c.580-1G>T; c.580-2A>C; c.580-2A>G; c.581G>T; c.586dupG; c.592G>A; c.595C>T; c.596A>G; c.598T>A; c.601delG; c.613C>T; c.617T>G; c.619C>T; c.647G>A; c.653T>A; c.657dupA; c.658C>T; c.671delT; c.675T>A; c.680T>G; c.695T>A; c.708delT; c.717delG; c.739_742dupTACA; c.742A>G; c.743+1G>A; c.743+1G>C; c.743+2T>C; c.743G>C; c.744-2A>G; c.755delCinsGCTGGGAAGAT; c.772A>G; c.773delG; c.794T>G; c.79G>A; c.79G>T; c.803delA; c.805_806delAT; c.808C>T; c.811delT; c.825C>G; c.828C>A; c.830G>A; c.835G>T; c.848_860delAAATGATTGAAAAinsTG; c.850dupA; c.859A>T; c.860dupA; c.861_865del; c.863T>G; c.865_869delAGACA; c.865A>T; c.868C>T; c.869+1G>C; c.869+1G>T; c.870-1G>C; c.870-2A>G; c.874_875delGA; c.874G>T; c.888_889dupTC; c.88C>T; c.892A>T; c.912C>G; c.930delC; c.933C>G; c.935_937delTCT; c.941G>A;

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	c.948delT; c.950T>A; c.958T>G; c.959delT; c.959T>A; c.987delA; c.988G>T; c.997C>T; c.3080T>C; c.3208C>T; c.2998del; c.2859_2890del; c.1523T>G; c.4097T>A; c.4439T>C; c.709C>G; c.1366G>T; c.1689C>A; c.307G>T; c.3254A>C; c.3600del; c.461dup; c.761del; c.980del; c.1076A>G; c.1470_1471del; c.156_163dup; c.202A>T; c.3365del; c.3382A>T; c.580G>A; c.580G>T; c.1069del; c.1302del; c.1375_1385del; c.1446dup; c.1585-1G>T; c.1679+2T>C; c.1786_1787del; c.1954C>T; c.2062A>T; c.2156T>A; c.2188G>T; c.2261del; c.2335C>T; c.2341C>T; c.234del; c.2576_2588del; c.2440C>T; c.2619+2T>C; c.273+2T>G; c.2964dup; c.3007G>T; c.3008del; c.3106del; c.3275_3290del; c.3397del; c.3407_3422del; c.357del; c.3615del; c.3904A>T; c.3921T>A; c.393del; c.4028dup; c.3976del; c.489+1G>A; c.498del; c.522_526del; c.54-1G>A; c.543_556del; c.548del; c.550del; c.581_582del; c.708dup; c.764del; c.850del; c.1025_1026insTA; c.1053_1054del; c.1068G>A; c.1116_1116+1insATCAA; c.1118dup; c.112_113del; c.1130del; c.1132C>T; c.1157dup; c.1162_1163delinsTA; c.1209+1G>C; c.1210-1G>C; c.1210-1G>T; c.1221del; c.1243_1247del; c.1287del; c.1360_1387del; c.1366del; c.1392+1del; c.142_145del; c.1424del; c.1439del; c.1540G>T; c.154A>T; c.156del; c.1582G>T; c.164+2T>G; c.1680-2A>C; c.1682dup; c.1766+2del; c.177dup; c.1792A>T; c.1800del; c.1807del; c.1810_1826del; c.1846del; c.1846dup; c.185dup; c.1874_1875del; c.1900C>T; c.1972dup; c.1976del; c.1977_1986del; c.1987_2000del; c.2000_2001del; c.2057_2058delinsA; c.2074G>T; c.2083del; c.2091del; c.2148del; c.2277del; c.2303dup; c.2349dup; c.2380del; c.2425del; c.2454_2455insT; c.2489dup; c.2491-2A>G; c.2523_2548del; c.2562del; c.2592_2593del; c.2615del; c.2619+2T>A; c.2620-1G>T; c.2705del; c.2732_2733insA; c.2733_2734insA; c.2776_2777del; c.2803_2813del; c.2816del; c.2819_2823del; c.2819_2820insA; c.2836A>T; c.285dup; c.2883_2886dup; c.2897del; c.2899_2900delinsA; c.2986C>T; c.2989-1G>T; c.2989-2A>C; c.3033dup; c.3081dup; c.3136G>T; c.3139+2T>C; c.3176T>G; c.3227_3231del; c.3290_3305del; c.3294del; c.3315del; c.332del; c.3367+1G>T; c.3367+2T>C; c.3368-1G>A; c.3384_3388del; c.3389_3402del; c.340A>T; c.3430C>T; c.3469-1G>A; c.3493A>T; c.3497del; c.3539_3554del; c.3556C>T; c.3592del; c.3623dup; c.3658dup; c.367dup; c.3708del; c.3724del; c.3754dup; c.3758dup; c.3773del; c.3816_3817del; c.3827del; c.3829del; c.3876del; c.3917_3918dup; c.387del; c.3932_3933delinsAATATG; c.3957del; c.4046del; c.4033_4034del; c.4065_4066del; c.4090del; c.4139del; c.4170del; c.450dup; c.476dup; c.476T>A; c.490-2A>C; c.53+2T>G; c.53+2T>C; c.54-2A>G; c.579_579+3del; c.604_605del; c.606G>A; c.614dup; c.617del; c.748C>T; c.777del; c.905_906insA; c.87dup; c.908_911del; c.927del; c.982del; c.3222T>A; c.3222T>G; c.933C>A; c.2885C>A; c.2885C>G; c.1086T>A; c.1086T>G; c.49_50delTT; c.1082G>A; c.1083G>A; c.1551C>A; c.1551C>G; c.3533C>G; c.3743C>A; c.3743C>G.
GALT NM_000155.3	c.1014C>G; c.1001A>G; c.1006A>T; c.100T>A; c.1018G>A; c.1018G>T; c.1024C>A; c.1030C>A; c.1030C>T; c.1034C>A; c.1047delC; c.1048A>G; c.1048delA; c.1052delC; c.1057C>T; c.1059+1G>T; c.1060-1G>A; c.1072delC; c.1098C>A; c.1108C>T; c.1138T>C; c.1139G>C; c.113A>C; c.1140A>C; c.118G>A; c.118G>T; c.130G>A; c.130G>T; c.132delG; c.134C>A; c.136_140delGCTCA; c.14_23dupGAACCGATCC; c.152G>A; c.152G>T; c.158G>A; c.160C>T; c.163G>T; c.164delG; c.180delG; c.189dupG; c.18delC; c.197C>A; c.199C>T; c.1A>G; c.203A>C; c.207_214delCCCTCTCA; c.220_221delCT; c.220_221insG; c.221T>C; c.238C>T; c.252+1G>A; c.253-2A>G; c.25C>T; c.265T>C; c.265T>G; c.285T>G; c.289_291delAAC; c.289A>G; c.290A>G; c.290A>T; c.291C>G; c.292G>A; c.299C>G; c.2T>C; c.2T>G; c.307C>T; c.308A>G; c.328+2T>C; c.329-2A>C; c.329G>A; c.334dupA; c.337G>A; c.341A>T; c.350T>C; c.354A>C; c.359delA; c.367C>T; c.368G>A; c.374T>C; c.377+1G>T; c.378-2A>T; c.379A>G; c.385A>T; c.386T>C; c.392T>G; c.394C>T; c.396C>A; c.398_399dupCC; c.400delT; c.404C>G; c.404C>T; c.410dupT; c.413C>T; c.416T>C; c.41delCinsTT; c.424A>G; c.425T>A; c.425T>C; c.428C>T; c.442C>G; c.442C>T; c.443G>A; c.445dupG; c.448G>C; c.452T>C; c.460T>C; c.460T>G; c.462G>A; c.482T>C; c.490C>T; c.499T>C; c.502_504delGTG; c.502G>A; c.502G>T; c.505C>A; c.507+2T>C; c.508-1G>C; c.509T>A; c.509T>C; c.512T>C; c.513delT; c.513dupT; c.524G>A; c.528_529insG; c.536G>A; c.539G>T; c.541T>G; c.542C>T; c.547C>A; c.550C>G; c.552C>A; c.553C>T; c.554C>A; c.556C>A; c.556C>T; c.558C>A; c.561_564+9delCCAGGTAAGGGTG; c.562C>T; c.563A>G; c.564+1G>A; c.564+1G>T; c.565_578delGTATGGGCCAGCAG; c.565-2A>G; c.568T>C; c.572C>T; c.574A>G; c.575G>A; c.580T>C; c.584T>C; c.593T>C; c.594T>G; c.595G>A; c.598C>T; c.598delC; c.601C>T; c.602G>A; c.607G>A; c.610C>T; c.611G>C; c.616C>T; c.619C>T; c.626A>C; c.626A>G; c.627T>A; c.634C>T; c.635A>C; c.650T>C; c.652delC; c.658dupG; c.658G>A; c.667C>A; c.670delC; c.677T>C; c.680T>C; c.687+1G>T; c.687+2T>C; c.687G>T; c.688-1G>C; c.688-2A>C; c.691C>G; c.691C>T; c.692G>A; c.697G>C; c.71_72insA; c.719_728delTAGTACTGGT; c.745T>C; c.747G>A; c.748C>A; c.752_753delACinsCT; c.756G>T; c.761dupT; c.768_770delGCC; c.770C>T; c.772C>T; c.775C>T; c.776G>A; c.779_790delATGTGCGGCGGC; c.785G>C; c.790_792delCTAinsTAG; c.790delC; c.812A>G; c.814C>G; c.814C>T; c.815G>A; c.82+2delT; c.820+13A>G; c.821-2A>G; c.821-7A>G; c.824delT; c.82G>A; c.82G>C; c.82G>T; c.83-2A>G; c.833T>A; c.836T>G; c.844C>G; c.854A>G; c.855G>T; c.856_864delTATGACAAC; c.865C>T; c.866T>G; c.871G>A; c.872A>T; c.881T>A; c.882delT; c.888C>G; c.893T>C; c.895G>A; c.899G>A; c.900G>A; c.904+1G>A; c.904+1G>T; c.905-1G>A; c.905-2A>G; c.90G>C; c.912dupC; c.920C>A; c.922G>A; c.938G>A; c.939G>A; c.947G>A; c.948G>A; c.949delC; c.950A>G; c.951G>T; c.952delC; c.957C>A; c.958G>A; c.959C>T; c.95T>A; c.967T>C; c.967T>G; c.968A>G; c.974C>T; c.976delC; c.979_986delCTGCGCTC; c.979delC; c.980T>C; c.982C>T; c.983G>A; c.985T>C; c.986C>T; c.989C>T; c.98G>A; c.997C>G; c.997C>T; c.998G>A; c.998G>T.
GJB2 NM_004004.5	c.487A>C; c.176_191del16; c.-23+1G>A; c.-23G>T; c.290dupA; c.298C>T; c.365A>T; c.-22-2A>C; c.101T>C; c.101T>G; c.109G>A; c.109G>T; c.110T>C; c.119C>A; c.119C>G; c.11delG; c.125_127delAGG; c.130_131delTG; c.131G>A; c.131G>C; c.132G>A; c.132G>C; c.134delG; c.134G>A; c.136G>A; c.137_141delATGAG; c.139G>T; c.148G>A; c.148G>T; c.158G>A; c.158G>T; c.162C>A; c.164C>A; c.167delT; c.169C>T; c.172C>G; c.172C>T; c.175G>A; c.175G>C; c.176G>A; c.176G>C; c.193T>C; c.194A>G; c.195C>G; c.196G>C; c.196G>T; c.19C>T; c.1A>G; c.1A>T; c.205T>C; c.218A>C; c.223C>G; c.223C>T; c.224G>A; c.227T>C; c.229T>C; c.230G>A; c.231G>A; c.232dupG; c.235delC; c.238C>T; c.239A>C; c.23C>T; c.241C>G; c.244A>G; c.246C>G; c.250G>A; c.250G>C; c.250G>T; c.257C>G; c.257delC; c.269dupT; c.269T>C; c.279G>A; c.280_284dupCACGT; c.283G>A; c.2T>C; c.2T>G; c.32_45delGGGGTGTGAACAAA; c.34G>C; c.34G>T; c.35delG; c.35dupG; c.35G>T; c.370C>T; c.379C>T; c.37G>A; c.385G>T; c.389G>C; c.400T>C; c.402delG; c.408C>A; c.40A>G; c.416G>A; c.426C>A; c.427C>T; c.428G>A; c.439G>A; c.445G>A; c.44A>C; c.456C>A; c.465T>A; c.474C>G; c.49_50delTC; c.506G>A; c.508_511dupAACG; c.50C>T; c.51_62delCACCAGCATTGGinsA; c.514delT; c.516G>A; c.516G>C; c.517C>T; c.523_533delCCCAACACTGT; c.526A>G; c.533T>C; c.535G>A; c.550C>T; c.551G>A; c.551G>C; c.563A>G; c.564_565delGA; c.56G>C; c.575_576delCA; c.576delA; c.583A>G; c.592_600delGTGTCTGGAinsCAGTGTTCATGACATTC; c.596C>T; c.598G>A; c.598G>T; c.59T>C; c.605G>T; c.617A>G; c.632_633delGT; c.632G>A; c.645delT; c.647_650delGATA; c.668_671delAAAA; c.66G>T; c.71G>A; c.72G>A; c.85_87delTTC; c.94C>A; c.94C>T; c.95G>A; c.95G>T; c.9G>A; c.290dupA; c.298C>T; c.298delC; c.299_300delAT; c.299A>T; c.310_323delAGGAAGTTCATCAA; c.313_326delAAGTTCATCAAGGG; c.326G>T; c.327_328delGGinsA; c.334_335delAA; c.338_341dupGTGA; c.355G>A; c.355G>T; c.358_360delGAG; c.365A>T.

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Gene	Variants
GJB6 NM_001110219.3	c.485dupA; c.443delC; c.383_384delTA; c.263C>T; c.261dupA; c.169C>T; c.119C>T; c.110T>A; c.31G>A; c.14C>T; c.148G>C; c.487delC; c.346A>T; c.322C>T.
HEXA* NM_001318825.2	c.569A>G; c.254-1G>C; c.1121dupT; c.1283_1284delCT; c.1019+1G>A; c.1010delT; c.1019+3A>G; c.1020G>A; c.1076_1079delTCAA; c.1106+1G>A; c.1106+1G>T; c.1154A>C; c.1156delG; c.1173delA; c.1175_1179delTAAAG; c.1183C>T; c.1201C>T; c.1202_1204delAGGinsT; c.1209G>A; c.1210C>T; c.1211G>C; c.1216delG; c.1247_1248delAAinsG; c.1292G>A; c.1293G>C; c.1307_1310dupTATC; c.1308_1311dupATCC; c.1311_1312insTATC; c.1335C>G; c.1338C>T; c.1340_1341delTA; c.1363+1G>A; c.1382delC; c.139_140delCAinsT; c.1393G>A; c.1393G>C; c.139C>T; c.1418A>T; c.1452_1453delCT; c.1454+1G>C; c.1454+1G>T; c.1454G>A; c.1455-1G>T; c.1455G>C; c.1465G>A; c.1477G>A; c.1477G>T; c.1487G>A; c.1528C>T; c.1529G>A; c.1532delT; c.1535C>G; c.1543C>T; c.1543delC; c.1544G>A; c.1559+1G>T; c.1559+2T>C; c.155C>A; c.1561C>T; c.1570C>T; c.1582dupC; c.173G>A; c.187G>T; c.196C>T; c.233G>A; c.253+1G>A; c.254-1G>C; c.254-2A>G; c.309dupG; c.343_346dupTGTA; c.349C>T; c.373G>A; c.379+1G>A; c.379+1G>C; c.379+1G>T; c.380A>G; c.413T>G; c.430delT; c.442C>T; c.446-2A>G; c.455delC; c.458_459delTT; c.459delT; c.463C>T; c.469delG; c.492+5G>A; c.493-1G>A; c.493-1G>T; c.498delT; c.541C>T; c.542G>A; c.557A>C; c.563C>G; c.565C>T; c.566G>A; c.566G>T; c.571T>C; c.573C>G; c.579dupA; c.584_585delCT; c.603+1G>A; c.604-1G>T; c.649delG; c.649G>C; c.662C>T; c.664_667delTTCC; c.665T>C; c.670T>C; c.705+1G>A; c.742C>T; c.751_752insT; c.772C>T; c.781G>A; c.782G>A; c.787C>T; c.805G>C; c.821C>T; c.838+1G>A; c.838+1G>C; c.838G>A; c.838G>C; c.839-1G>C; c.839G>A; c.931_938delTTCATGAG; c.935T>G; c.948_950delCTT; c.962_963delCT; c.980dupA; c.995_997delGAG; c.997G>A; c.997G>T.
EMD NM_000117.2	c.101dupA; c.103G>T; c.123C>A; c.12C>A; c.12C>G; c.130C>T; c.135dupG; c.153dupC; c.16delG; c.177_178delTA; c.184dupT; c.187+1G>T; c.1A>G; c.217_218dupAT; c.239_240insT; c.251_255delTCTAC; c.265+2T>A; c.266-2A>G; c.284_298delATGAAGAGAGCTACT; c.355C>T; c.3G>A; c.419T>A; c.430G>T; c.450-2A>G; c.468_471delCCGG; c.475_476delAG; c.484C>T; c.512C>A; c.547C>A; c.548C>A; c.581_582delCA; c.600G>A; c.607delC; c.60delC; c.619delC; c.621delG; c.640_644dupGGGGC; c.650_654dupTGGGC; c.674_678delTCTGG; c.703_704insA; c.77dupT; c.82+1G>A; c.82+1G>T; c.82G>A; c.83-2A>C; c.83-2A>G.
GLA NM_000169.2	c.1017_1027delGTGGGAACGAC; c.1018T>C; c.1019_1020insA; c.101A>G; c.1020G>A; c.1021G>A; c.1021G>T; c.1023A>C; c.1023delA; c.1024C>G; c.1024C>T; c.1025G>A; c.1031_1034dupTCTC; c.1033_1034delTC; c.1037delG; c.1040dupT; c.1042G>C; c.1045T>C; c.1048G>A; c.104G>A; c.1055_1056delCT; c.1057_1058delAT; c.1062_1076delAAACCGGCAGGAGAT; c.1066C>T; c.1067G>A; c.1067G>C; c.1069C>T; c.1072_1074delGAG; c.1072G>A; c.1072G>T; c.1077delT; c.107T>G; c.1081G>A; c.1081G>C; c.1085C>T; c.1087C>T; c.1088G>A; c.1095T>A; c.109G>C; c.1117G>A; c.1118G>A; c.1124G>C; c.1125_1128dupAGTG; c.1147_1149delTTC; c.1157A>C; c.1166delC; c.1188delC; c.118C>T; c.1192G>T; c.119C>G; c.119C>T; c.1201dupT; c.1212_1214delAAG; c.1225C>G; c.1225C>T; c.1228A>G; c.1229C>T; c.1235_1236delCT; c.1241T>C; c.1244T>C; c.1246C>T; c.124A>C; c.125_137delTGGGCTGGCTGCA; c.125T>C; c.126G>C; c.127G>A; c.1284_1287delACTT; c.1285delC; c.128delG; c.128G>T; c.131G>A; c.132G>A; c.137A>G; c.137A>T; c.141G>C; c.146G>C; c.153G>A; c.153G>T; c.154T>C; c.154T>G; c.164_171delACTGCCAGinsTCTGCCTA; c.166T>G; c.169C>T; c.172delG; c.188G>A; c.190A>T; c.194G>C; c.195-1G>C; c.195-1G>T; c.196G>C; c.19G>T; c.235G>T; c.242G>A; c.244A>T; c.254G>A; c.256T>C; c.265delC; c.266T>G; c.26delA; c.272T>A; c.272T>C; c.273_276dupTGAT; c.274G>A; c.274G>T; c.277G>A; c.281G>A; c.285G>A; c.290C>T; c.295C>T; c.295delC; c.295dupC; c.298A>T; c.2T>C; c.307G>T; c.31_43dupGGCTGCGCGCTTG; c.334C>T; c.335G>A; c.335G>T; c.337T>A; c.337T>C; c.350T>G; c.361_364delGCTA; c.369+1G>A; c.369+2T>G; c.370-2A>G; c.386T>C; c.394G>A; c.398_413delTTTATGCAGATGTTGGinsCTGC; c.402T>G; c.404C>T; c.406G>T; c.413G>A; c.41T>C; c.422C>T; c.424T>C; c.427G>A; c.427G>C; c.431G>T; c.436C>T; c.439G>A; c.443G>A; c.444T>G; c.456C>A; c.466G>A; c.469C>T; c.476T>C; c.476T>G; c.484delT; c.484T>C; c.485A>G; c.490G>T; c.496_497delCTinsGG; c.50_54delGCTTC; c.509A>G; c.520T>C; c.540G>T; c.547G>A; c.548-1G>A; c.548-2A>G; c.548G>A; c.548G>T; c.560T>C; c.561G>A; c.58G>C; c.59_72dupCCCTCGTTTCCTGG; c.593T>C; c.59C>A; c.604T>C; c.605G>A; c.606T>G; c.607G>A; c.610T>C; c.613_621delCCTCTTTAT; c.613C>A; c.614C>T; c.619T>C; c.620_630delATATGTGGCCinsCACT; c.620A>C; c.620A>G; c.622_623delAT; c.62T>C; c.630delC; c.638A>G; c.640-1delG; c.640-1G>A; c.640-1G>T; c.641C>T; c.644A>G; c.646T>G; c.647A>G; c.657C>G; c.658C>T; c.661C>T; c.662_663delAG; c.666C>A; c.667T>C; c.668G>A; c.670A>G; c.677G>A; c.679C>T; c.680G>A; c.680G>C; c.688G>A; c.695T>C; c.704C>G; c.706T>A; c.706T>C; c.707G>A; c.708G>T; c.713G>A; c.717A>G; c.718_719delAA; c.723dupT; c.730G>A; c.730G>C; c.734G>A; c.735G>A; c.748C>A; c.748C>T; c.749A>C; c.751G>T; c.758T>C; c.761T>C; c.770C>T; c.774_775delAC; c.776C>G; c.777delA; c.782G>A; c.782G>T; c.784T>C; c.785G>A; c.786dupG; c.790G>T; c.791A>T; c.797A>T; c.800delT; c.801+1G>C; c.801+3A>G; c.801G>A; c.802-2A>G; c.802-2A>T; c.802-3_802-2delCA; c.802-3_804delCAGTTAinsGGCAACTTT; c.803_806delTAGT; c.805G>A; c.806T>C; c.806T>G; c.80delC; c.811G>A; c.815A>G; c.818T>C; c.823C>T; c.823delC; c.826A>G; c.830G>A; c.835C>G; c.842_844delTAA; c.847C>T; c.848dupA; c.851T>C; c.85dupG; c.861G>A; c.861G>T; c.863delC; c.865A>T; c.869T>C; c.870G>A; c.874G>C; c.879_880delTTinsAATC; c.886A>G; c.887T>C; c.888G>A; c.889T>C; c.890C>T; c.893A>G; c.894T>A; c.896A>G; c.899T>C; c.901C>G; c.901C>T; c.902dupG; c.902G>A; c.916C>T; c.950_954dupTTGCC; c.950T>C; c.950T>G; c.959_962delATCA; c.95T>C; c.961C>G; c.962A>G; c.966C>A; c.966C>G; c.973G>A; c.974G>A; c.979C>T; c.979C>T; c.980A>G; c.980A>T; c.982G>A; c.982G>C; c.983G>C; c.988delC; c.98A>G; c.994dupA; c.996_999delACAG; c.999+1G>T; c.999+2T>C.
G6PD NM_000402.4	c.292G>A; c.1054T>C; c.1093G>A; c.1172C>T; c.1178A>T; c.1179C>A; c.1192G>A; c.1243T>C; c.1246A>G; c.1249C>T; c.1268G>A; c.1270G>C; c.1282G>A; c.1319G>A; c.1406G>C; c.1408C>T; c.1429G>A; c.1437G>C; c.1450C>T; c.1466G>C; c.1466G>T; c.1468G>T; c.1478A>G; c.1555C>T; c.188T>C; c.221C>G; c.233T>C; c.299A>G; c.473T>C; c.494A>C; c.538G>A; c.577G>A; c.583A>G; c.607T>C; c.632A>T; c.653C>T; c.683G>A; c.683G>C; c.727G>T; c.770G>T; c.896G>A; c.934G>C; c.938A>T; c.961G>A
OTC NM_000531.6	c.77+2dup; c.77+3_77+6del; c.77+4A>C; c.77+5G>C; c.78-3C>G; c.125T>C; c.177dup; c.298+1delG; c.403G>C; c.429T>A; c.488T>A; c.505C>T; c.535C>G; c.674C>A; c.709del; c.709_712delinsCCT; c.717+3A>G; c.749dup; c.844del; c.860_861dupCA; c.1006-3C>G; c.1005+1G>T; c.1005+2T>C; c.1005G>A; c.1006-1G>A; c.1006G>T; c.1009G>C; c.1009G>T; c.1015G>A; c.1015G>C; c.1018T>C; c.1022T>C; c.1028C>A; c.1033T>C; c.1033T>G; c.1034A>G; c.1039C>A; c.1042C>T; c.1052delA; c.1061T>G; c.106C>T; c.115G>T; c.116G>C; c.118C>T; c.119G>A; c.122A>G; c.127C>T; c.128T>C; c.131C>T; c.133C>G; c.134T>C; c.140_141insG; c.140A>T; c.140delA; c.143_144delTT; c.143T>C; c.145A>C; c.148G>A; c.148G>T; c.154G>A; c.154G>T; c.155A>G; c.158T>C; c.158T>G; c.163T>G; c.167T>C; c.170T>A; c.174G>A; c.177dupA; c.179C>T; c.188T>C; c.1A>G; c.1A>T; c.200T>G; c.205C>T; c.214G>A; c.216+1delG; c.216+1G>A; c.216+1G>T; c.217-1G>A; c.217-2A>G; c.217-2dupA; c.227T>C; c.228A>C; c.231G>T; c.232C>T; c.236G>A; c.238A>G; c.240G>T; c.243_245delCTT; c.245T>G; c.247G>C; c.248G>A; c.254T>C; c.259G>A; c.264A>T; c.268A>G; c.269G>A; c.270T>G; c.271delA; c.274C>G; c.274C>T; c.275G>A; c.275G>C; c.275G>T; c.277A>G; c.281G>C; c.284T>C; c.29_32delACAA; c.298+1_298+5delGTAAG; c.298+1G>A; c.298+1G>T; c.298+2T>G; c.298G>T;

	c.299G>A; c.2T>C; c.305C>A; c.314G>T; c.316G>A; c.317G>A; c.317G>T; c.327T>A; c.330delT; c.332T>C; c.350A>G; c.350A>T; c.359_360delTG; c.364_365insTT; c.365A>G; c.376delG; c.377A>G; c.386+1G>A; c.386+1G>T; c.386+2T>C; c.386G>A; c.386G>C; c.386G>T; c.387-1G>A; c.387-2A>C; c.387-2A>G; c.387-2A>T; c.390_392dupATT; c.391_397dupTTGTCTA; c.392T>C; c.394T>C; c.395C>T; c.3G>A; c.403delG; c.404C>A; c.407A>T; c.409G>A; c.416T>C; c.418G>C; c.421C>G; c.421C>T; c.422G>A; c.422G>C; c.425T>A; c.42delT; c.430A>T; c.437C>G; c.443T>C; c.443T>G; c.444G>C; c.444G>T; c.451delC; c.452T>G; c.455C>T; c.460G>T; c.463G>C; c.463G>T; c.464C>A; c.473C>T; c.476T>C; c.479T>A; c.479T>C; c.479T>G; c.481A>G; c.482A>G; c.484G>A; c.484G>C; c.485G>A; c.488T>G; c.490T>C; c.491C>G; c.491C>T; c.493G>C; c.493G>T; c.494A>G; c.502C>T; c.501C>A; c.501C>G; c.503A>C; c.503A>G; c.504T>A; c.505C>G; c.506C>T; c.514A>T; c.516C>G; c.517C>G; c.520G>C; c.524A>G; c.524A>T; c.526T>C; c.527A>G; c.532_537delACGCTC; c.533C>T; c.535C>T; c.536T>C; c.53delA; c.540+1G>C; c.540+1G>T; c.540+2T>A; c.540+2T>C; c.540G>C; c.541-2A>G; c.542A>G; c.545A>T; c.547T>G; c.548A>G; c.562_563delGG; c.562G>C; c.563G>T; c.568dupA; c.571C>T; c.576C>G; c.577T>C; c.577T>G; c.578G>A; c.583G>A; c.584G>C; c.586delG; c.586G>A; c.586G>T; c.587A>T; c.589G>A; c.589G>T; c.590G>A; c.593A>T; c.594C>A; c.595A>G; c.596A>G; c.602T>C; c.604C>T; c.605A>C; c.608C>G; c.613A>G; c.615G>C; c.617T>A; c.617T>G; c.618G>C; c.620G>A; c.621C>A; c.622G>A; c.626C>A; c.626C>T; c.628A>C; c.634G>T; c.635G>T; c.640C>T; c.643C>T; c.645dupT; c.646C>G; c.650C>A; c.658C>G; c.659C>T; c.663+1G>A; c.663+1G>T; c.663+2T>C; c.663G>A; c.663G>C; c.664-1G>A; c.665delG; c.673C>A; c.673C>T; c.674C>G; c.674C>T; c.67C>T; c.698C>T; c.700G>T; c.709_712delGCCAinsCCT; c.716A>G; c.716A>T; c.717+1G>A; c.717+1G>T; c.717+2T>C; c.717G>A; c.717G>C; c.718-2A>G; c.725C>T; c.731_739delTGTTGCTGA; c.731T>A; c.740C>A; c.740C>G; c.749dupC; c.757G>A; c.757G>C; c.759delA; c.764A>C; c.766G>T; c.77+1G>A; c.77+1G>T; c.779T>C; c.77G>A; c.77G>C; c.78-1G>C; c.780_781delAAinsG; c.785C>A; c.785C>T; c.787G>A; c.788A>G; c.790A>G; c.791C>A; c.791C>T; c.793T>C; c.794G>T; c.795G>A; c.799A>C; c.803T>C; c.806G>A; c.808C>T; c.809A>C; c.817_819delGAG; c.818delA; c.829C>T; c.830G>A; c.830G>T; c.835C>T; c.867+1G>A; c.867+1G>T; c.867G>T; c.868-2A>T; c.876delA; c.882delT; c.890_893delACTG; c.892_893delTG; c.893G>C; c.903A>T; c.904C>T; c.905A>G; c.905A>T; c.906C>G; c.906delC; c.907T>C; c.907T>G; c.908G>A; c.912G>T; c.914C>A; c.914C>G; c.919A>G; c.928G>T; c.929A>G; c.931G>A; c.941_943delAAG; c.943G>T; c.944T>A; c.944T>C; c.944T>G; c.946T>G; c.947T>C; c.94C>T; c.953C>T; c.958C>T; c.959G>C; c.959G>T; c.961T>C; c.962C>A; c.976G>A; c.982G>T; c.988A>G; c.991A>T; c.994T>A; c.995G>A; c.996G>A.
FMR1	CGG Triplet Expansion
SMN1	Exon 7 Deletion

* The region chr15:72668198-72668313 in *HEXA* gene cannot be detected