CGT Essential V2.1
Variants List

Gene	Tested Variants
MMACHC NM_015506.2	c.1A>G; c.3G>A; c.81+2T>G; c.82-1G>A; c.90G>A; c.217C>T; c.271dupA; c.273_276delAGAG; c.285dupA; c.292C>T; c.310_313delGACT; c.315C>G; c.328_331delAACC; c.331C>T; c.347T>C; c.352delC; c.388_390delTAC; c.389A>G; c.391C>T; c.394C>T; c.398_399delAA; c.420G>A; c.427C>T; c.445_446delTG; c.440G>A; c.440G>C; c.457C>T; c.464G>A; c.471G>A; c.481C>T; c.482G>A; c.500delC; c.507_519delAGAGGTGCCAGAT; c.547_548delGT; c.565delC; c.565C>A; c.567dupT; c.608G>A; c.609G>A; c.615C>G; c.619dupG; c.616C>T; c.658_660delAAG; c.666C>A; c.688C>T
ACADM NM_001286043.1	c.942A>T; c.199T>C; c.30+2T>C; c.107_113dupGATTTAG; c.118+1G>T; c.157C>T; c.173_174delAG; c.216+2T>G; c.224delT; c.233T>C; c.244dupT; c.250C>T; c.253G>A; c.270_271delCA; c.286+2T>G; c.386-2A>G; c.386-1G>C; c.421_424delATTA; c.419T>C; c.446G>A; c.461C>T; c.486+1G>A; c.486+1G>T; c.525delG; c.530_533delAGTA; c.536delT; c.548_551delCTGA; c.546G>A; c.563T>C; c.568-2A>C; c.568-1G>A; c.676A>G; c.682G>A; c.698+1G>A; c.698+2T>C; c.699-18G>A; c.715C>T; c.716G>T; c.797T>C; c.807+1G>A; c.807+2T>G; c.808-2A>C; c.808-1G>A; c.829T>C; c.833C>T; c.841A>G; c.896A>G; c.898G>A; c.916_928delGCAATGGGAGCTT; c.948+1G>A; c.949-2A>G; c.981_982delAG; c.980G>C; c.1012G>T; c.1025dupT; c.1045-2A>C; c.1048C>T; c.1056_1057delAT; c.1058C>A; c.1076T>C; c.1083delG; c.1084A>G; c.1098_1110dupTAGAATGAGTTAC; c.1111_1112insTAGAATGAGTTAC; c.1111C>T; c.1144delC; c.1144C>T; c.1151C>T; c.1172delA; c.1201_1204delTTAG; c.1213dupG; c.1223T>C; c.1249G>T; c.1288dupT; c.1293+1G>A; c.1294-2A>T; c.1294-1G>C; c.893_902delGTGACGGAGCinsTTTAA; c.708A>C; c.217-1G>A; c.727C>T
DHCR7 NM_001163817.1	c.1426T>C; c.1396G>A; c.1342G>A; c.1337G>A; c.1328G>A; c.1228G>A; c.1222T>C; c.1210C>T; c.1057delG; c.1055G>A; c.1054C>T; c.976G>T; c.964-1G>T; c.964-1G>C; c.963+2T>G; c.963+1G>T; c.939G>A; c.934_935delGT; c.915C>G; c.907G>A; c.906C>G; c.894C>A; c.870G>A; c.866C>T; c.862G>A; c.858G>A; c.841G>A; c.832-1G>C; c.831+2T>A; c.804delT; c.740C>T; c.725G>A; c.724C>T; c.627-1G>A; c.626+1G>T; c.626+1G>C; c.470T>C; c.461C>T; c.461C>G; c.453G>A; c.452G>A; c.440G>A; c.413-1G>A; c.413-2A>G; c.385_412+5delATCCAGGAGGGGGCCGTGACTCCTGCAGGTAGC; c.322-2A>G; c.292C>T; c.278C>T; c.151C>T; c.111G>A; c.99-1G>C; c.89G>C; c.82C>T; c.16C>T; c.3G>A; c.1A>G; c.1084C>T; c.895C>T; c.506C>T; c.1190C>T; c.1112G>A; c.1139G>A; c.1214delA; c.546G>A; c.651C>A; c.696G>A; c.952delT
HBB NM_000518.4	c.440_441dupAC; c.440A>T; c.440A>C; c.439C>G; c.438T>A; c.437A>G; c.436T>C; c.435G>C; c.431A>C; c.428C>A; c.421G>A; c.383A>C; c.371_378delCCCCACCA; c.364G>A; c.364G>T; c.347C>A; c.344T>C; c.343_344delCTinsG; c.341T>A; c.332T>C; c.328delG; c.328G>A; c.323dupG; c.320T>G; c.316-1G>T; c.316-1G>A; c.316-2A>G; c.316-2A>C; c.316-3C>A; c.315+2T>G; c.315+1G>A; c.315+1G>C; c.312C>G; c.306G>C; c.305A>G; c.304G>A; c.302C>T; c.299A>T; c.299A>G; c.299A>C; c.298G>T; c.298G>C; c.298G>A; c.295G>A; c.293A>T; c.287dupA; c.282_283dupTG; c.283G>C; c.277C>T; c.277C>A; c.275T>C; c.271G>T; c.269G>A; c.268A>C; c.257T>C; c.251delG; c.248A>T; c.248A>C; c.247A>G; c.230delC; c.226delC; c.217_221delAGTGAinsT; c.217dupA; c.216dupT; c.208G>A; c.206T>A; c.203_204delTG; c.201delA; c.199A>G; c.194delG; c.190C>T; c.184A>T; c.182T>A; c.179A>C; c.176C>G; c.162delT; c.143_146dupATCT; c.143dupA; c.135delC; c.134C>G; c.130G>T; c.126_129delCTTT; c.128T>C; c.127T>G; c.127T>C; c.114_120delGACCCAG; c.117_118inCC; c.118C>T; c.114G>A; c.113G>A; c.112delT; c.110delC; c.108C>A; c.103G>T; c.93G>T; c.93-17_93-1delTATTTTCCCACCCTTAG; c.93-1G>A; c.93-1G>C; c.93-2A>C; c.93-21G>A; c.92+6T>C; c.92+5G>T; c.92+5G>C; c.92+5G>A; c.92+2T>C; c.92+2T>A; c.92+1G>A; c.92+1G>T; c.92G>C; c.92G>A; c.91A>C; c.90C>T; c.79_80insT; c.80A>G; c.79G>T; c.79G>A; c.75T>A; c.64dupG; c.59A>G; c.52A>T; c.51delC; c.48G>A; c.47G>A; c.46delT; c.45dupG; c.36delT; c.27dupG; c.25_26delAA; c.20delA; c.20A>T; c.19G>A; c.17_18delCT; c.8A>C; c.4delG; c.4G>T; c.3G>A; c.2T>G; c.2T>A; c.2T>C; c.1A>G; c.-50A>C; c.-75G>C; c.-78A>C; c.-78A>G; c.-79A>G; c.-80T>A; c.316-7C>G; c.316-7C>A; c.380T>G; c.-81A>G; c.-29G>A; c.-18C>G; c.*6C>G; c.82G>T; c.-137C>T; c.-137C>A; c.-137C>G; -88C>T; c.316-197C>T; c.316-106C>G (IVS2-745C>G); c.72T>A; c.316-7C>G; c.-151C>T; c.-140C>T; c.-138C>A; c.-138C>T; c.-136C>T; c.-136C>G; c.*110T>C; c.*111A>G ; c.*113A>G
PAH NM_000277.1	c.1355dupA; c.1340C>A; c.1314_1315+4delCAGTAA; c.1315+2T>C; c.1315+1G>A; c.1315+1G>T; c.1301C>A; c.1298dupT; c.1293_1294delGA; c.1282C>T; c.1256A>G; c.1252A>C; c.1249T>C; c.1243G>A; c.1241A>G; c.1240T>C; c.1238G>C; c.1223G>A; c.1222C>T; c.1220C>T; c.1219C>T; c.1209delT; c.1208C>T; c.1200-1G>A; c.1200-8G>A; c.1199+17G>A; c.1199+2T>G; c.1199+1G>C; c.1199+1G>A; c.1198delA; c.1197A>T; c.1196T>C; c.1184C>G; c.1180G>C; c.1180G>T; c.1171_1172delAG; c.1172G>C; c.1171A>G; c.1169A>G; c.1166delC; c.1163T>C; c.1162G>A; c.1162G>C; c.1157A>G; c.1147C>T; c.1139C>T; c.1129delT; c.1114A>T; c.1089delG; c.1076C>G; c.1068C>G; c.1068C>A; c.1066-2A>T; c.1066-3C>T; c.1066-11G>A; c.1065+1G>A; c.1055delG; c.1049C>A; c.1045T>C; c.1042C>G; c.1033G>A; c.1033G>T; c.1028A>G; c.1024delG; c.1025C>A; c.1004A>C; c.997C>T; c.992T>C; c.978G>A; c.977G>A; c.975C>G; c.974A>G; c.970-1G>C; c.970-1G>A; c.967_969delACA; c.955G>T; c.941C>A; c.940C>A; c.934G>T; c.931_932delCT; c.932T>C; c.929C>T; c.926C>T; c.926C>A; c.916delA; c.916A>G; c.913-2A>C; c.913-7A>G; c.912+2T>C; c.912+1G>A; c.910C>T; c.901C>A; c.898G>T; c.896T>G; c.895T>C; c.890G>A; c.887A>G; c.869A>T; c.847A>T; c.844G>A; c.842+5G>A; c.842+4A>G; c.842+3G>C; c.842+2T>A; c.842+1G>A; c.842C>T; c.841C>T; c.841C>G; c.839A>G; c.838G>A; c.837delC; c.836C>T; c.833C>T; c.833C>A; c.830A>G; c.829T>G; c.826A>G; c.824C>T; c.824C>G; c.823C>T; c.818C>T; c.814G>T; c.809G>A; c.806delT; c.806T>A; c.796A>C; c.790delC; c.785T>G; c.782G>A; c.782G>C; c.781C>T; c.781C>G; c.776C>T; c.775G>A; c.770G>T; c.764T>C; c.757G>A; c.755G>A; c.754C>T; c.754C>G; c.745delC; c.745C>T; c.740G>T; c.739G>C; c.737delC; c.737C>A; c.734_735delTGinsCA; c.734T>C; c.734T>A; c.733G>C; c.731C>T; c.728G>A; c.727C>T; c.724C>T; c.722delG; c.722G>A; c.722G>T; c.721C>T; c.694C>T; c.691T>C; c.689T>C; c.688G>A; c.686dupA; c.682G>A; c.673C>A; c.664_665delGA; c.662A>G; c.648C>G; c.638T>C; c.635T>C; c.632delC; c.632C>T; c.631C>A; c.618C>A; c.618C>G; c.612T>G; c.611A>G; c.591G>C; c.580_581delCT; c.581T>C; c.569T>C; c.563G>A; c.561G>A; c.558_559delAT; c.556delA; c.547_548delGAinsTT; c.535T>A; c.533A>G; c.529G>C; c.529G>A; c.527G>T; c.526C>T; c.520A>G; c.511G>A; c.510-2A>G; c.508C>G; c.506G>A; c.505C>T; c.503delA; c.504C>A; c.500A>T; c.498C>G; c.490A>G; c.482T>C; c.473G>A; c.472C>T; c.470_471delGAinsAC; c.464G>A; c.464G>C; c.442-1G>A; c.442-2A>C; c.441+5G>T; c.441+4A>G; c.441+1G>C; c.441+1G>A; c.440C>T; c.434A>T; c.398_401delATCA; c.400C>T; c.385G>T; c.357delC; c.355C>T; c.350delC; c.331C>T; c.329delC; c.320A>G; c.311C>A; c.284_286delTCA; c.283A>T; c.266dupC; c.261C>A; c.250G>T; c.242C>A; c.227A>G; c.226G>T; c.212G>A; c.204A>T; c.196G>T; c.194T>C; c.169G>A; c.169G>T; c.169-2A>G; c.168+5G>C; c.168+1G>A; c.168_168+1delGGinsAA; c.165delT; c.165T>G; c.164T>C; c.143T>C; c.140C>T; c.136G>A; c.127G>T; c.121C>T; c.116_118delTCT; c.117C>G; c.110T>C; c.60+5G>T; c.60+1G>C; c.58C>T; c.47_48delCT; c.3G>C; c.3G>A; c.1A>G; c.442-5C>G; c.1092_1094delTCT; c.912G>A; c.386A>G; c.439C>T; c.1138dupA; c.1139C>A; c.1154T>A; c.2TC; c.360G>A; c.514C>T; c.504C>G; c.693delT; c.898G>A

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Gene	Variants
PMM2 NM_000303.2	c.395T>C; c.1A>G; c.24delC; c.67-2A>T; c.95T>G; c.95_96delTAinsGC; c.104T>A; c.157C>T; c.179-1G>T; c.189delA; c.193G>T; c.205C>T; c.255+1G>A; c.255+2T>C; c.256-2A>G; c.256-1G>C; c.310C>G; c.323C>T; c.324delG; c.338C>T; c.345dupG; c.347+1G>A; c.348-2A>C; c.348-1G>C; c.349G>C; c.357C>A; c.359T>C; c.367C>T; c.385G>A; c.392delC; c.414delA; c.415G>A; c.422G>A; c.430T>C; c.442G>A; c.451_454delGAAA; c.454_455delAA; c.458T>C; c.470T>C; c.484C>T; c.511dupA; c.524-2A>G; c.560G>A; c.563A>G; c.580C>T; c.620T>C; c.623G>C; c.639+1G>A; c.647A>T; c.652C>G; c.653A>T; c.669C>G; c.677C>G; c.691G>A; c.710C>G; c.710C>T; c.722G>C; c.447+5G>A; c.53C>G; c.131T>C; c.355T>C; c.190delT; c.109C>T; c.368G>A; c.26G>A; c.178+1GA; c.179-1G>C; c.192C>G; c.395T>C; c.421C>T; c.435C>G; c.561G>A; c.661T>G; c.72delT; c.97C>T
GAA NM_000152.4	c.1128_1129delGGinsC; c.1129G>C; c.1134C>G; c.1143delC; c.1153delC; c.1156C>T; c.1165delG; c.1192dupC; c.1193delT; c.1194+2T>C; c.1195-1G>A; c.1222A>G; c.1292_1295dupTGCA; c.1309C>T; c.1316T>A; c.1326+1G>A; c.1326+2T>C; c.1411_1414delGAGA; c.1437+1G>A; c.1438-2A>G; c.1438-1G>C; c.1441T>C; c.1447G>A; c.1465G>A; c.1478C>T; c.1496G>A; c.1548G>A; c.1551+1G>C; c.1551+1G>T; c.1552-3C>G; c.1552-2A>G; c.1556T>C; c.1561G>A; c.1564C>A; c.1564C>G; c.1567delT; c.1655T>C; c.1687C>T; c.1754+1G>A; c.1799G>A; c.1802A>C; c.1802C>T; c.1802C>G; c.1824_1828dupATACG; c.1827delC; c.1832G>A; c.1843G>A; c.1856G>A; c.1912G>T; c.1927G>A; c.1933G>C; c.1933G>T; c.1933G>A; c.1935C>A; c.1941C>G; c.1942G>A; c.1961C>A; c.1979G>A; c.2012T>G; c.2014C>T; c.2015G>A; c.2040+1G>T; c.2061delC; c.2104C>T; c.2105G>A; c.2136_2137delGT; c.2140delC; c.2161dupG; c.2173C>T; c.2185delC; c.2188G>T; c.2189+1G>T; c.2213G>A; c.2214G>A; c.2227C>T; c.2242delG; c.2242dupG; c.2237G>A; c.2237G>C; c.2238G>A; c.2238G>C; c.2242G>T; c.2269C>T; c.2281delGinsAT; c.2300delT; c.2331+2T>A; c.2332-1G>C; c.2367dupA; c.2407C>T; c.2495_2496delCA; c.2501_2502delCA; c.2512C>T; c.2544delC; c.2560C>T; c.2608C>T; c.2646+2T>A; c.2662G>T; c.2704_2716dupCAGAAGGTGACTG; c.2704C>T; c.2706delG; c.2815_2816delGT; c.1978C>T; c.1634C>T; c.1822C>T; c.877G>A
AGXT NM_000030.2	c.245G>A; c.847-3C>G; c.698G>A; c.346G>A; c.2T>C; c.2_3delTGinsAT; c.3G>T; c.74T>G; c.77T>C; c.107G>A; c.116_117dupCA; c.122G>A; c.122G>T; c.125G>A; c.130C>T; c.139G>A; c.166-1G>A; c.167T>A; c.187G>C; c.198C>G; c.205C>T; c.209C>A; c.215dupA; c.221_227dupTCACACT; c.242C>A; c.242C>T; c.244G>C; c.248A>G; c.276delT; c.283_285dupGAG; c.283G>A; c.327delG; c.322T>C; c.323G>A; c.324G>T; c.326G>T; c.331C>T; c.332G>A; c.335C>A; c.349G>T; c.353G>A; c.358+1G>T; c.358+2T>G; c.424-2A>G; c.445delG; c.466G>A; c.473C>A; c.473C>T; c.481G>C; c.481G>T; c.481G>A; 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.560C>T; c.570delG; c.568G>A; c.577dupC; c.577delC; c.583A>C; c.584T>G; c.596-2A>G; c.603C>A; c.605T>A; c.612C>A; c.613T>C; c.614C>A; c.614C>T; c.642_645delTCCA; c.646G>A; c.661T>C; c.673_676delAAGG; c.679_680+2delAAGT; c.680+1G>A; c.680+1G>C; c.680+2T>A; c.681-1G>T; c.697C>T; c.698G>T; c.725dupT; c.727G>C; c.731T>C; c.737G>A; c.738G>A; c.744delC; c.752G>A; c.753G>A; c.757T>C; c.776+1G>A; c.776+1G>C; c.776+2T>G; c.777-2A>G; c.777-1G>C; c.798_802delCATCAinsACAATCTCAG; c.806T>C; c.823_824dupAG; c.822G>C; c.834delC; c.844C>T; c.846G>C; c.846+1G>A; c.846+1G>T; c.847-1G>C; c.851T>C; c.853G>T; 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.976delG; c.996G>A; c.997A>T; c.1014C>G; c.1045G>A; c.1071+1G>A; c.1076T>C; c.1079G>A; c.1079G>C; c.1102G>A; c.1125_1126delCG; c.1151T>C; c.1049G>A; c.302T>C
HADHA NM_000182.4	c.337T>C; c.1246C>T; c.1241T>C; c.1235_1236delCT; c.1229C>T; c.1228A>G; c.1225C>G; c.1225C>T; c.1192G>T; c.1188delC; c.1157A>C; c.1124G>C; c.1118G>A; c.1117G>A; c.1095T>A; c.1088G>A; c.1081G>A; c.1081G>C; c.1077delT; c.1072_1074delGAG; c.1072G>A; c.1069C>T; c.1067G>C; c.1066C>T; c.1057_1058delAT; c.1055_1056delCT; c.1037delG; c.1033_1034delTC; c.1025G>A; c.1024C>T; c.1024C>G; c.1023A>C; c.1021G>T; c.1020G>A; c.1018T>C; c.999+2T>C; c.996_999delACAG; c.983G>C; c.982G>C; c.982G>A; c.980A>T; c.980A>G; c.979C>T; c.979C>A; c.974G>A; c.973G>A; c.966C>A; c.959_962delATCA; c.950T>G; c.916C>T; c.902G>A; c.901C>T; c.899T>C; c.896A>G; c.893A>G; c.890C>T
CBS NM_000071.2	c.1552+1G>A; c.1545delG; c.1468-1G>A; c.1359-1G>C; c.1358+2T>C; c.1358+1G>A; c.1330G>A; c.1321A>T; c.1280C>T; c.1224-2A>C; c.1219_1223+8del; c.1223G>A; c.1221delC; c.1218delG; c.1150A>G; c.1136G>A; c.1111G>A; c.1109G>A; c.1087delG; c.1058C>T; c.1051C>G; c.1039G>A; c.1009_1012delATGC; c.1007G>A; c.1006C>T; c.992C>A; c.969G>A; c.959T>C; c.954+2T>G; c.954+1G>A; c.919G>A; c.904G>A; c.903C>G; c.833T>C; c.829-1G>C; c.828+1G>A; c.816T>A; c.797G>A; c.785C>T; c.770C>T; c.738delG; c.737-1G>C; c.736+2T>G; c.707_708delCCinsGGTG; c.700G>A; c.676G>A; c.667-14_667-7delCTCTTTCT; c.572C>T; c.536_553del; c.532-2A>G; c.494G>A; c.467delT; c.451+1G>T; c.442G>A; c.434C>T; c.430G>A; c.415G>A; c.402delG; c.374G>A; c.373C>T; c.362G>A; c.346G>A; c.341C>T; c.325T>C; c.316+1G>A; c.306G>C; c.302T>C; c.253G>A; c.233C>G; c.209+2T>C; c.209+1G>C; c.153_165del; c.162G>A; c.146C>T; c.18_36del; c.28delG; c.19dupC
ARSA NM_000487.5	c.1226C>T; c.869G>A; c.1223_1231delGTGATACCA; c.1229C>T; c.1462C>T; c.1408_1418delGCAGCTGTGAC; c.1344dupC; c.1337delG; c.1283C>T; c.1279C>A; c.1274A>G; c.1264delC; c.1210+1G>A; c.1210+1G>T; c.1114C>T; c.1108-2A>G; c.1107+1delG; c.1036delG; c.1010A>T; c.991G>T; c.986C>T; c.980-2A>C; c.979_979+3delGGTC; c.979+1G>A; c.979G>A; c.960G>A; c.938G>A; c.937C>T; c.929delG; c.905G>T; c.899T>C; c.891delC; c.890C>A; c.883G>A; c.877C>T; c.868C>T; c.862A>C; c.854+1G>T; c.827C>T; c.769C>G; c.763G>A; c.758dupT; c.746T>C; c.739G>A; c.737G>A; c.697C>A; c.674_675dupAT; c.641C>T; c.542T>G; c.526C>T; c.474C>A; c.470C>G; c.467G>A; c.466-2A>G; c.465+2T>A; c.465+1G>A; c.421C>T; c.418delC; c.418dupC; c.410T>C; c.346C>T; c.304delC; c.302delG; c.302G>A; c.293C>T; c.263G>A; c.257G>A; c.256C>T; c.240dupC; c.229G>C; c.227_228insTA; c.225-2A>G; c.224+1G>A; c.211_212delTG; c.206_209delCTCT; c.195delC; c.185_186dupCA; c.109_116delGACCTGGG; c.98T>C; c.1136C>T; c.1174C>T; c.257G>T; c.270C>A; c.774delA
BTD NM_001281723.2	c.449G>A; c.998_1007delACCTTATAAT; c.600_602delCGT; c.44+1G>A; c.44+1G>C; c.44+1G>T; c.104_110delGCGGCTGinsTCC; c.113dupA; c.124_125delCT; c.142G>T; c.166G>T; c.177T>G; c.190G>A; c.190G>T; c.196G>A; c.198G>C; c.208_211dupATCC; c.200A>G; c.241C>T; c.251C>A; c.263T>G; c.268C>T; c.272delA; c.284A>G; c.289C>T; c.304G>A; c.316-1G>T; c.316G>T; c.322C>T; c.329_330dupTA; c.332dupT; c.332T>G; c.340G>A; c.340G>C; c.347G>T; c.378_381dupCATT; c.388T>G; c.399delC; c.412delC; c.426G>A; c.430C>A; c.451T>C; c.460A>C; c.461C>G; c.465G>A; c.472C>T; c.475C>T; c.476G>A; c.496_497delAG; c.534G>T; c.550delA; c.563G>A; c.565C>T; c.590A>G; c.593delC; c.593C>G; c.600delC; c.632G>A; c.637delC; c.635A>G; c.637C>T; c.643delC; c.649C>T; c.652T>A; c.658A>C; c.660C>G; c.670G>C; c.670G>A; c.689A>G; c.699delC; c.707C>T; c.715G>A; c.740G>A; c.761A>G; c.764C>T; c.789C>G; c.800A>T; c.838C>G; c.842T>A; c.842T>G; c.871G>C; c.893T>G; c.901G>C; c.902C>T; c.904A>C; c.935G>A; c.938G>A; c.939delT; c.939T>G; c.1007T>A; c.1055delC; c.1058delC; c.1102_1103dupTC; c.1132C>T; c.1163G>A; c.1164G>A; c.1197_1198delGA; c.1213T>G; c.1233_1247delGGGAAAGGAAGGCTAinsTTCCAATGGCC; c.1245delC; c.1247_1258delATCTCCACGTCT; c.1270dupC; c.1281T>G; c.1290C>A; c.1320T>A; c.1330delG; c.1358_1359delGC; c.1367A>G; c.1374A>C; c.1378dupT; c.1390delA; c.1400dupG; c.1461C>G; c.1464delG; c.1465delT; c.1495C>T; c.1499dupT; c.1514_1518delGGATG; c.1563T>G; c.1601C>T; c.1618C>T; c.1622dupT; c.1274dupA; c.1324delG; c.1447G>T; c.557T>A; c.637delG; c.933delT; c.494dupG; c.442T>C; c.1495dupT; c.1555dupT
SLC26A2 NM_000112.3	c.47C>G; c.55G>T; c.63_64delCA; c.185C>G; c.188delA; c.207delT; c.239_243dupGCAGT; c.255delC; c.325_326delTT; c.331G>T; c.387T>G; c.391delC; c.398C>T; c.403C>A; c.451delT; c.485_486delTG; c.496G>A; c.499delG; c.532C>T; c.541C>T; c.578_581delCCTT; c.611T>G; c.642_643delAA; c.699+2T>C; c.700-2A>G; c.700-1G>C; c.705_711delGATGGGC; c.835C>T; c.906_907delCT; c.1020_1022delTGT; c.1157C>T; c.1242_1245delAAAC; c.1273A>G; c.1361A>C; c.1451G>A; c.1535C>A; c.1724delA; c.1957T>A; c.1976delT; c.1983delA; c.2033G>T; c.2124_2125dupCT; c.117delC; c.1303dupA; c.1639CT; c.1650delG; c.166delAinsCTC; c.1714delC; c.223CT; c.-26+2T>C; c.294G>A; c.2T>C; c.1777G>T; c.1800C>G; c.1878delG; c.854C>A; c.909T>A

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Gene	Variants
GALT NM_000155.3	c.607G>A; c.1A>G; c.18delC; c.25C>T; c.41delCinsTT; c.82G>C; c.82G>T; c.83-2A>G; c.90G>C; c.95T>A; c.98G>A; c.113A>C; c.130G>T; c.130G>A; c.136_140delGCTCA; c.152G>T; c.160C>T; c.163G>T; c.180delG; c.197C>A; c.199C>T; c.203A>C; c.207_214delCCCTCTCA; c.220_221delCT; c.220_221insG; c.221T>C; c.238C>T; c.253-2A>G; c.265T>G; c.265T>C; c.285T>G; c.289_291delAAC; c.289A>G; c.292G>A; c.299C>G; c.307C>T; c.308A>G; c.328+2T>C; c.329-2A>C; c.334dupA; c.337G>A; c.341A>T; c.350T>C; c.354A>C; c.367C>T; c.368G>A; c.374T>C; c.377+1G>T; c.378-2A>T; c.385A>T; c.386T>C; c.392T>G; c.398_399dupCC; c.396C>A; c.400delT; c.404C>G; c.404C>T; c.410dupT; c.413C>T; c.416T>C; c.424A>G; c.425T>A; c.425T>C; c.428C>T; c.442C>T; c.445dupG; c.442C>G; c.443G>A; c.448G>C; c.460T>C; c.460T>G; c.462G>A; c.482T>C; c.490C>T; c.499T>C; c.502G>T; c.502G>A; c.505C>A; c.507+2T>C; c.508-1G>C; c.509T>A; c.509T>C; c.513delT; c.512T>C; c.524G>A; c.528_529insG; c.536G>A; c.539G>T; c.541T>G; c.542C>T; c.550C>G; c.553C>T; c.554C>A; c.556C>T; c.556C>A; c.558C>A; c.562C>T; c.563A>G; c.564+1G>T; c.564+1G>A; c.565_578delGTATGGGCCAGCAG; c.565-2A>G; c.568T>C; c.574A>G; c.575A>G; c.580T>C; c.584T>C; c.594T>C; c.598delC; c.595G>A; c.601C>T; c.602G>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.687G>T; c.687+1G>T; c.687+2T>C; c.688-2A>C; c.691C>T; c.692G>A; c.697G>C; c.719_728delTAGTACTGGT; c.745T>C; c.747G>A; c.748C>A; c.756G>T; c.761dupT; c.770C>T; c.772C>T; c.779_790delATGTGCGGCGGC; c.775C>T; c.776G>A; c.785G>C; c.790_792delCTAinsTAG; c.812A>G; c.814C>G; c.814C>T; c.815G>A; c.820+13A>G; c.821-2A>G; c.824delT; c.833T>A; c.854A>G; c.855G>T; c.865C>T; c.866T>G; c.871G>A; c.872A>T; c.882delT; c.881T>A; c.888C>G; c.899G>A; c.904+1G>T; c.905-2A>G; c.905-1G>A; c.912dupC; c.920C>A; c.922G>A; c.938G>A; c.947G>A; c.948G>A; c.949delC; c.950A>G; c.951G>T; c.952delC; c.957C>A; c.959C>T; c.967T>C; c.967T>G; c.968A>G; c.974C>T; c.976delC; c.979delC; c.980T>C; c.982C>T; c.983G>A; c.985T>C; c.986C>T; c.989C>T; c.997C>G; c.997C>T; c.998G>T; c.998G>A; c.1001A>G; c.1018G>T; c.1018G>A; c.1024C>A; c.1030C>A; c.1034C>A; c.1047delC; c.1048delA; c.1048A>G; c.1052delC; c.1057C>T; c.1059+1G>T; c.1060-1G>A; c.1098C>A; c.1108C>T; c.1138T>C; c.1140A>C; c.1014C>G; c.2T>C
EMD NM_000117.2	c.1A>G; c.3G>A; c.82+1G>T; c.83-2A>G; c.103G>T; c.123C>A; c.130C>T; c.153dupC; c.184dupT; c.187+1G>T; c.239_240insT; c.251_255delTCTAC; c.266-2A>G; c.355C>T; c.419T>A; c.430G>T; c.450-2A>G; c.484C>T; c.512C>A; c.547C>A; c.548C>A; c.600G>A; c.607delC; c.621delG; c.650_654dupTGGGC; c.674_678delTCTGG
GLA NM_000169.2	c.337T>C; c.1246C>T; c.1241T>C; c.1235_1236delCT; c.1229C>T; c.1228A>G; c.1225C>G; c.1225C>T; c.1192G>T; c.1188delC; c.1157A>C; c.1124G>C; c.1118G>A; c.1117G>A; c.1095T>A; c.1088G>A; c.1081G>A; c.1081G>C; c.1077delT; c.1072_1074delGAG; c.1072G>A; c.1069C>T; c.1067G>C; c.1066C>T; c.1057_1058delAT; c.1055_1056delCT; c.1037delG; c.1033_1034delTC; c.1025G>A; c.1024C>T; c.1024C>G; c.1023A>C; c.1021G>T; c.1020G>A; c.1018T>C; c.999+2T>C; c.996_999delACAG; c.983G>C; c.982G>C; c.982G>A; c.980A>T; c.980A>G; c.979C>T; c.979C>A; c.974G>A; c.973G>A; c.966C>A; c.959_962delATCA; c.950T>G; c.916C>T; c.902G>A; c.901C>T; c.899T>C; c.896A>G; c.893A>G; c.890C>T; c.888G>A; c.887T>C; c.886A>G; c.879_880delTTinsAATC; c.865A>T; c.861G>T; c.861G>A; c.847C>T; c.842_844delTAA; c.835C>G; c.830G>A; c.826A>G; c.823delC; c.823C>T; c.815A>G; c.806T>G; c.806T>C; c.802-3_802-2delCA; c.802-2A>G; c.802-2A>T; c.801G>A; c.797A>T; c.791A>T; c.790G>T; c.785G>A; c.784T>C; c.777delA; c.776C>G; c.761T>C; c.748C>T; c.735G>A; c.734G>A; c.730G>C; c.730G>A; c.718_719delAA; c.713G>A; c.707G>A; c.704C>G; c.680G>A; c.680G>C; c.679C>T; c.677G>A; c.670A>G; c.667T>C; c.666C>A; c.662_663delAG; c.661C>T; c.658C>T; c.657C>G; c.647A>G; c.644G>A; c.641C>T; c.640-1G>T; c.640-1G>A; c.638A>G; c.622_623delAT; c.613_621delCCTCTTTAT; c.620A>G; c.620A>C; c.614C>T; c.613C>A; c.610T>C; c.607G>A; c.606T>G; c.605G>A; c.561G>A; c.548G>T; c.548G>A; c.548-1G>A; c.548-2A>G; c.540G>T; c.509A>G; c.485G>A; c.484T>C; c.469C>T; c.466G>A; c.456C>A; c.443G>A; c.439G>A; c.436C>T; c.427G>C; c.427G>A; c.424T>C; c.422C>T; c.394G>A; c.386T>C; c.370-2A>G; c.369+2T>G; c.369+1G>A; c.361_364delGCTA; c.337T>A; c.335G>A; c.335G>T; c.334C>T; c.295dupC; c.290C>T; c.285G>A; c.281G>A; c.274G>T; c.274G>A; c.272T>C; c.272T>A; c.256T>C; c.254G>A; c.244A>T; c.242G>A; c.235G>T; c.195-1G>T; c.195-1G>C
CFTR NM_000492.3	c.-9_14delCGAGAGACCATGCAGAGGTCGCC; c.1A>G; c.2T>C; c.4C>T; c.11C>A; c.14C>T; c.19G>T; c.38C>T; c.53+1G>T; c.56G>A; c.57G>A; c.79G>A; c.79G>T; c.88C>T; c.114C>G; c.115C>T; c.137C>A; c.164+1G>A; c.164+1G>T; c.164+1G>C; c.164+2T>C; c.164+3_164+4insT; c.165-3C>T; c.165-1G>A; c.166G>A; c.169T>G; c.170G>A; c.171G>A; c.174_177delTAGA; c.175dupA; c.178G>A; c.178G>T; c.200C>T; c.223C>T; c.254G>T; c.254G>A; c.262_263delTT; c.263T>A; c.263T>G; c.271G>A; c.273+1G>A; c.273+3A>C; c.274-2A>G; c.274-1G>A; c.274-1G>C; c.274G>A; c.274G>T; c.292C>T; c.293A>G; c.296C>T; c.305T>G; c.313delA; c.325_327delTATinsG; c.326A>G; c.327T>A; c.328G>T; c.328G>C; c.330C>A; c.349C>T; c.350T>C; c.350G>A; c.358G>A; c.377G>A; c.405_406dupAC; c.409delC; c.409_412delCTCC; c.413_415dupTAC; c.416A>G; c.424delA; c.442delA; c.445G>T; c.459_476delAATAGCTATGTTTAGTTT; c.470_483delTTAGTTTGATTTAT; c.476T>C; c.481T>C; c.489+1G>T; c.489+2T>C; c.489+3A>G; c.490-1G>A; c.494T>C; c.531delT; c.532G>A; c.543_546delTAGT; c.571T>G; c.577G>A; c.577G>T; c.579+1G>T; c.579+3A>G; c.579+5G>A; c.580-1G>T; c.595C>T; c.613C>T; c.617T>G; c.647G>A; c.650A>G; c.658C>T; c.680T>G; c.695T>A; c.717delG; c.722_743delGGAGAATGATGATGAAGTACAG; c.723_743+1delGAGAATGATGATGAAGTACAGG; c.739_742dupTACA; c.743+1G>A; c.743+1G>C; c.744-2A>G; c.772A>G; c.803delA; c.805_806delAT; c.828C>A; c.830G>A; c.861_865delCTTAA; c.865A>T; c.869+1G>C; c.870-2A>G; c.870-1G>C; c.888_889dupTC; c.926C>A; c.935_937delTCT; c.933C>G; c.948delT; c.988G>T; c.1000C>T; c.1001G>T; c.1001G>A; c.1006_1007insG; c.1007T>A; c.1013C>T; c.1021_1022dupTC; c.1021T>C; c.1029delC; c.1037T>C; c.1040G>A; c.1040G>C; c.1040G>T; c.1046C>T; c.1055A>G; c.1057C>T; c.1081delT; c.1083delG; c.1093_1094delCT; c.1116+1G>A; c.1117-2A>G; c.1117-1G>A; c.1130dupA; c.1135G>T; c.1141A>T; c.1155_1156dupTA; c.1202G>A; c.1203G>A; c.1209+1G>A; c.1209+1G>T; c.1211delG; c.1219G>T; c.1234_1238delGCAAA; c.1240C>T; c.1301C>G; c.1301_1307delCACTTCT; c.1327_1330dupGATA; c.1340delA; c.1358T>C; c.1364C>A; c.1365_1366delGG; c.1367T>C; c.1373delG; c.1373G>T; c.1393-2A>G; c.1393-1G>A; c.1397C>A; c.1397C>G; c.1400T>C; c.1409_1418delTGATTATGGG; c.1418delG; c.1420G>A; c.1438G>T; c.1466C>A; c.1475C>T; c.1477_1478delCA; c.1477C>T; c.1487G>A; c.1505T>C; c.1519_1521delATC; c.1517T>C; c.1521_1523delCTT; c.1538A>G; c.1545_1546delTA; c.1546A>G; c.1558G>T; c.1558G>A; c.1572C>A; c.1573C>T; c.1579_1584+11del; c.1584+1G>A; c.1585-8G>A; c.1585-2A>G; c.1585-1G>A; c.1624G>T; c.1645A>C; c.1646G>A; c.1646G>T; c.1647T>G; c.1647T>A; c.1648G>T; c.1650delA; c.1651G>A; c.1652delG; c.1652G>A; c.1654C>T; c.1657C>T; c.1670delC; c.1673T>C; c.1675G>A; c.1679G>C; c.1679G>A; c.1679+1G>A; c.1679+1G>C; c.1680-1G>A; c.1680-1G>C; c.1680A>C; c.1682C>A; c.1684G>C; c.1687T>G; c.1687T>A; c.1692delA; c.1703delT; c.1703T>A; c.1705T>G; c.1712T>C; c.1713_1714delAG; c.1721C>A; c.1736A>G; c.1742dup; c.1753G>T; c.1766+1G>A; c.1766+1G>C; c.1766+1G>T; c.1766+2T>C; c.1766+2T>A; c.1766+3A>G; c.1766+5G>T; c.1766G>A; c.1841A>G; c.1911delG; c.1923_1931delCTCAAAACTinsA; c.1943A>T; c.1943delA; c.1973_1985delGAAATTCAATCCTinsAGAAA; c.1986_1989delAACT; c.1990G>T; c.2012delT; c.2017G>T; c.2036G>A; c.2051_2052delAAinsG; c.2052dupA; c.2052delA; c.2053dupC; c.2083dupG; c.2125C>T; c.2128A>T; c.2143C>T; c.2146A>T; c.2158C>T; c.2175dupA; c.2195T>G; c.2215delG; c.2241_2248delGATACTGC; c.2290C>T; c.2291delG; c.2353C>T; c.2374C>T; c.2393dup; c.2395C>T; c.2423_2424dupAT; c.2429delG; c.2443G>T; c.2453delT; c.2463_2464delTG; c.2464G>T; c.2479G>T; c.2483dupA; c.2490+1G>A; c.2490+2T>C; c.2491-2A>C; c.2491G>T; c.2537G>A; c.2538G>A; c.2547C>A; c.2551C>T; c.2552G>T; c.2554dupT; c.2583delT; c.2589_2599delAATTTGGTGCT; c.2601dupA; c.2619+1G>A; c.2620-2A>G; c.2644_2645dupTG; c.2645A>G; c.2657+2_2657+3insA; c.2657+5G>A; c.2658-2A>G; c.2658-1G>C; c.2668C>T; c.2735C>A; c.2737_2738insG; c.2739T>A; c.2763_2764dupAG; c.2777delT;

	c.2780T>C; c.2810dupT; c.2816A>G; c.2825delT; c.2834C>T; c.2845C>T; c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC; c.2875delG; c.2896delA; c.2908G>C; c.2908+1G>A; c.2909-15T>G; c.2909G>A; c.2916_2952delTCTTAATAGATTCTCCAAAGATATAGCAATTTTGGAT; c.2930C>T; c.2932A>T; c.2936A>C; c.2939T>A; c.2982_2988+2delCATCCAGGT; c.2988G>A; c.2988+1G>A; c.2988+1G>C; c.2989-2A>G; c.2989-2A>T; c.2989-1G>A; c.2993delT; c.2997_3000delAATT; c.3000_3014del; c.3002_3003delTG; c.3011_3019delCTATAGCAG; c.3014T>G; c.3017C>A; c.3022delG; c.3039dupC; c.3039delC; c.3038C>A; c.3041A>G; c.3067_3072delATAGTG; c.3095A>G; c.3103C>T; c.3107C>A; c.3124C>T; c.3139_3139+1delGG; c.3139+1delG; c.3140-26A>G; c.3140-1G>A; c.3154T>G; c.3160C>G; c.3161delA; c.3181G>C; c.3184_3188dupCTATG; c.3189G>A; c.3194T>C; c.3196C>T; c.3197G>A; c.3205G>A; c.3209G>A; c.3212A>C; c.3217dupT; c.3230T>C; c.3266G>A; c.3276C>A; c.3276C>G; c.3292T>C; c.3293G>A; c.3294G>C; c.3294G>T; c.3294G>A; c.3299A>C; c.3302T>A; c.3302T>G; c.3304A>T; c.3310G>T; c.3368-2A>G; c.3368-2A>T; c.3435G>A; c.3454G>C; c.3468G>A; c.3468+2dupTc.3468+1G>A; c.3468+5G>A; c.3472C>T; c.3475T>C; c.3476C>T; c.3484C>T; c.3492dupT; c.3495delG; c.3528delC; c.3529A>T; c.3532_3535dupTCAA; c.3536_3539delCCAA; c.3533C>A; c.3587C>G; c.3605delA; c.3611G>A; c.3612G>A; c.3617C>G; c.3659delC; c.3691delT; c.3700A>G; c.3712C>T; c.3717G>A; c.3717+4A>G; c.3717+5G>A; c.3717+12191C>T; c.3718-3T>G; c.3718-1G>A; c.3719T>G; c.3731G>A; c.3744delA; c.3745G>A; c.3747delG; c.3746G>A; c.3752G>A; c.3761T>G; c.3763T>C; c.3764C>A; c.3767dupC; c.3773dupT; c.3806T>A; c.3808delG; c.3808G>A; c.3841C>T; c.3846G>A; c.3848G>T; c.3851_3852dupAA; c.3857T>C; c.3873+1G>A; c.3873+1G>T; c.3873+2T>C; c.3874-8T>A; c.3874-2A>G; c.3883delA; c.3889dupT; c.3900dupT; c.3908delA; c.3907A>C; c.3909C>G; c.3922G>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>G; c.3971T>C; c.3988_3989delCA; c.3988C>T; c.3999delG; c.4004T>C; c.4035_4038dupCCTA; c.4036_4042delCTAAGCC; c.4046G>A; c.4077_4080delTGTTinsAA; c.4086dupT; c.4111G>T; c.4124A>C; c.4127_4131delTGGAT; c.4143C>G; c.4147dupA; c.4144C>T; c.4197_4198delCT; c.4231C>T; c.4234C>T; c.4242+1G>A; c.4242+1G>T; c.4242+2T>C; c.4243-2A>G; c.4251delA; c.4252G>T; c.4300_4301dupAG; c.4364C>G; c.4426C>T
GJB2 NM_004004.5	c.617A>G; c.605G>T; c.551G>A; c.551G>C; c.535G>A; c.439G>A; c.428G>A; c.269T>C; c.235delC; c.224G>A; c.223C>G; c.223C>T; c.172C>G; c.172C>T; c.167delT; c.164C>A; c.136G>A; c.132G>A; c.132G>C; c.131G>A; c.131G>C; c.94C>A; c.94C>T; c.35delG; c.487A>C; c.229T>C; c.71G>A; c.119C>G; c.139G>T; c.169C>T; c.176_191del16; c.19C>T; c.231G>A; c.246C>G; c.250G>C; c.257C>G; c.269dupT; c.283G>A; c.35G>T; c.35dupG; c.370C>T; c.427C>T; c.44A>C; c.493C>T; c.508_511dupAACG; c.516G>A; c.523_533del11; c.564_565delGA; c.632_633delGT; c.-23+1G>A; c.-23G>T
G6PD NM_000402.4	c.1466G>C; c.292G>A; c.1478G>A; c.1466G>T; c.1450C>T; c.1429G>A; c.1408C>T; c.1406G>C; c.1319G>A; c.1270G>C; c.1268G>A; c.1249C>T; c.1246A>G; c.1179C>A; c.1178A>T; c.1172C>T; c.1093G>A; c.1054T>C; c.961G>A; c.934G>C; c.896G>A; c.770G>T; c.727G>T; c.683G>C; c.653C>T; c.632A>T; c.583A>G; c.577G>A; c.473T>C; c.233T>C
FMR1	CGG Triplet Expansion
SMN1	Exon 7 Deletion