

Allegato 1 - Lista Varianti

Gene, Transcript (n° di varianti)	Varianti testate
<p>MMACHC NM_015506.3 (170)</p>	<p>c.1A>G; c.1A>T; c.2T>C; c.2T>G; c.3G>A; c.14_24del; c.36del; c.48_49del; c.51del; c.51T>A; c.70del; c.72C>A; c.80A>G; c.81G>A; c.81+1G>C; c.81+1G>T; c.81+1G>A; c.81+2T>C; c.81+2T>G; c.82-11_82-8del; c.82-2A>G; c.82del; c.82-1G>A; c.90_97del; c.89G>A; c.90G>A; c.93C>A; c.97G>T; c.126_141del; c.122dup; c.123dup; c.143_146del; c.151_152del; c.158T>C; c.178dupG; c.182G>C; c.189_196dup; c.187delC; c.202C>T; c.217C>T; c.228_231del; c.270dupT; c.271dupA; c.273_276delAGAG; c.273_276del; c.271_272insC; c.276G>A; c.276G>T; c.276+2del; c.276+2T>C; c.277-2A>G; c.277-1G>A; c.281_290del; c.285dupA; c.292C>T; c.310_313delGACT; c.315C>G; c.315C>A; c.321_329delinsACACC; c.328_331delAACC; c.331C>T; c.346C>G; c.346C>A; c.347T>C; c.354_365dup; c.352delC; c.349G>C; c.352C>T; c.364dupC; c.364C>A; c.366T>A; c.366T>G; c.388_390delTAC; c.388_390del; c.384delC; c.388T>C; c.389A>G; c.391C>T; c.394C>T; c.398_399delAA; c.415_416delCCinsTA; c.419G>A; c.420G>A; c.427C>T; c.429+1G>C; c.430-2A>G; c.430-1G>A; c.436_450del; c.433delA; c.435_436delAT; c.434dupT; c.439G>A; c.445_446delTG; c.439G>C; c.440G>A; c.440G>C; c.443_459delinsCTGGGGCTGG; c.449_452dup; c.449T>A; c.451_461del; c.452A>G; c.455_457del; c.457C>T; c.463G>C; c.464G>A; c.466G>A; c.467G>A; c.471G>A; c.471G>C; c.478_481del; c.481C>G; c.481C>T; c.482G>A; c.482G>T; c.487del; c.484G>T; c.497dupT; c.500delC; c.507_519delAGAGGTGCCAGAT; c.506_507del; c.534del; c.541_548delGACTGTGT; c.542_545dup; c.547_548delGT; c.544T>C; c.551_554dupTACC; c.561_572del; c.565delC; c.565C>A; c.565C>T; c.567dupT; c.566G>T; c.566G>A; c.574delC; c.578T>C; c.600del; c.599G>A; c.600G>A; c.603_604delTG; c.604G>T; c.609dup; c.608G>A; c.609G>A; c.614dup; c.616del; c.615C>A; c.615C>G; c.616C>T; c.619dupG; c.619del; c.617G>C; c.617G>A; c.626_627delTG; c.624del; c.626dupT; c.634del; c.640dup; c.643T>C; c.647_649del; c.648dupA; c.649_650del; c.658_660delAAG; c.658_659delinsGC; c.661_664dup; c.666C>A; c.675_679delCTCCA; c.685C>T; c.687del; c.688C>T; c.712C>T.</p>
<p>ACADM NM_000016.6 (362)</p>	<p>c.-6_6delinsACCCCGAGTG; c.-6_6delinsACCCCGAAGG; c.1A>T; c.1A>G; c.2T>G; c.3G>C; c.19C>T; c.24C>A; c.30+1G>C; c.30+2T>G; c.30+2T>C; c.30+4A>G; c.31-2A>G; c.31-1G>C; c.47del; c.65C>G; c.65C>A; c.67C>T; c.74_75del; c.85C>T; c.86G>A; c.86G>T; c.92G>A; c.91C>T; c.107_113dup; c.104T>G; c.117del; c.118G>T; c.118G>C; c.118+1G>T; c.118+10C>A; c.119-1G>C; c.127G>A; c.130C>T; c.136_158dup; c.134A>G; c.135G>C; c.157C>G; c.157C>T; c.158G>A; c.165del; c.166G>C; c.168del; c.173_174del; c.177A>C; c.193dup; c.199T>C; c.201T>A; c.203del; c.203A>G; c.205A>T; c.212del; c.216+1G>A; c.216+1G>T; c.216+2T>C; c.216+2T>G; c.216+5G>T; c.217-2A>C; c.217-2A>G; c.217-1del; c.217-1G>A; c.218A>G; c.221C>T; c.222del; c.224del; c.232dup; c.232A>G; c.233T>C; c.234T>G; c.238A>G; c.241G>A; c.243_250del; c.242_243insT; c.244dup; c.245G>A; c.250C>T; c.253G>C; c.253G>T; c.253G>A; c.254G>A; c.260T>C; c.270_271dup; c.270_271del; c.273dup; c.286+2T>G; c.287-2A>G; c.287-1G>A; c.287-1G>C; c.309del; c.306_307insG; c.308T>A; c.310G>A; c.311A>G; c.322_325del; c.320T>C; c.321_322del; c.321A>C; c.333_345del; c.331G>A; c.331G>T; c.337G>A; c.338C>A; c.341A>G; c.342_343dup; c.343_348del; c.342T>A; c.346T>G; c.347G>A; c.348T>A; c.355dup; c.353G>C; c.362C>T; c.377del; c.383dup; c.387+1del; c.387+1G>A; c.387+1G>T; c.387+2T>C; c.387+2T>A; c.388-14A>G; c.388-5G>A; c.388-3T>G; c.388-1G>A; c.395C>A; c.395C>G; c.403_405del; c.407C>T; c.425del; c.423_425del; c.430_432del; c.426del; c.431_434del; c.430A>T; c.434A>G; c.437del; c.443G>T; c.443G>A; c.449_452del; c.447G>T; c.447G>A; c.454G>T; c.464T>C; c.467del; c.468+1G>A; c.468+2T>C; c.468+2T>G; c.469-2A>G; c.469-2A>C; c.469-1G>A; c.472T>C; c.475del; c.473A>G; c.474T>A; c.484G>T; c.493G>A; c.494C>T; c.503A>G; c.503A>C; c.518del; c.520dup; c.526G>A; c.526G>T; c.530A>G; c.541_542del; c.542A>G; c.554T>C; c.555T>G; c.556_557del; c.558T>A; c.567del; c.572G>A; c.573G>A; c.577A>G; c.580A>G; c.583G>A; c.584G>A; c.589A>G; c.592G>A; c.599G>A; c.599+1G>A; c.599+2T>C; c.599+3A>G; c.599+5G>A; c.600-18G>A; c.600-1G>A; c.600G>A; c.601del; c.608dup; c.603T>A; c.608T>A; c.609A>C; c.613G>C; c.614C>T; c.616C>T; c.617G>A; c.617G>T; c.622G>T; c.652del; c.652G>A; c.653C>G; c.659C>G; c.659C>T; c.661G>A; c.669del; c.668T>C; c.673G>A; c.686del; c.683C>A; c.683C>T; c.698T>C; c.703A>T; c.706A>T; c.708G>C; c.708+1G>A; c.708+2T>G; c.709-13A>G; c.709-2A>G; c.709-2A>C; c.709-1G>A; c.709G>T; c.712_715del; c.717C>G; c.721G>A; c.727C>T; c.728G>A; c.730T>C; c.734C>T; c.739A>G; c.741_742dup; c.742A>G; c.744A>C; c.745G>A; c.755T>G; c.757G>A; c.778_782del; c.794_803delinsTTTAA; c.797A>G; c.799_803del; c.799G>C; c.799G>T; c.799G>A; c.801del; c.817_829del; c.820A>G; c.823_824dup; c.826G>C; c.826_828delinsCC; c.830_831dup; c.835A>T; c.839C>T; c.842G>C; c.843A>T; c.849+1G>A; c.849+2T>C; c.850-2A>G; c.850-1G>C; c.859G>C; c.862del; c.865G>A; c.866T>C; c.882_883del; c.880A>G; c.881G>C; c.890A>G; c.901A>T; c.913G>T; c.926dup; c.927del; c.928G>A; c.945+1G>A; c.946-6T>G; c.946-2A>C; c.949C>G; c.949C>T; c.950A>T; c.957_958del; c.959C>A; c.973dup; c.977T>C; c.982A>G; c.983del; c.983T>C; c.984del; c.985A>C; c.985A>G; c.989_1010del; c.999_1011dup; c.1010dup; c.1010A>C; c.1011C>A; c.1012_1013insTAGAATGAGTTAC; c.1012C>T; c.1018G>A; c.1022_1029del; c.1034A>T; c.1036_1045delinsAAT; c.1040G>T; c.1042del; c.1042C>T; c.1045del; c.1045C>G; c.1045C>T; c.1046G>A; c.1046G>T; c.1049_1055dup; c.1052C>T; c.1054_1057dup; c.1054T>G; c.1055A>G; c.1066A>G; c.1067T>C; c.1068_1071dup; c.1073del; c.1085G>A; c.1097del; c.1102_1105del; c.1103T>G; c.1110dup; c.1114dup; c.1115C>A; c.1117G>A; c.1118T>G; c.1118T>C; c.1120C>T; c.1124T>C; c.1129G>T; c.1133G>A; c.1150G>T; c.1167del; c.1169_1170del; c.1175G>A; c.1177G>A; c.1186dup; c.1184A>G; c.1189_1192del; c.1188C>G; c.1189dup; c.1189T>A; c.1190A>G; c.1190A>C; c.1194+1dup; c.1194+1G>A; c.1194+2T>G; c.1195-2A>C; c.1195-2A>T; c.1195-1G>C; c.1203_1224del; c.1205G>A; c.1207A>G; c.1221_1222del; c.1228_1230del; c.1226_1227del; c.1229T>C; c.1237del; c.1237C>A; c.1237C>T; c.1238G>A; c.1247T>C; c.1250_1256del; c.1250_1253delinsCT; c.1253A>G; c.1257C>A.</p>
<p>DHCR7 NM_001360.3 (277)</p>	<p>c.1426T>A; c.1426T>C; c.1424T>C; c.1409T>A; c.1406G>C; c.1404C>G; c.1399C>T; c.1397T>C; c.1396G>A; c.1385A>G; c.1384T>C; c.1376G>A; c.1365C>A; c.1360A>T; c.1352G>T; c.1351T>C; c.1349_1350delinsTG; c.1349G>T; c.1348del; c.1349G>A; c.1348C>T; c.1342_1344delinsC; c.1342G>A; c.1342G>C; c.1337G>A; c.1337G>C; c.1336C>T; c.1328G>C; c.1328G>T; c.1328G>A; c.1327C>G; c.1327C>T; c.1325A>G; c.1324C>G; c.1293_1307del; c.1295A>G; c.1294T>C; c.1290C>G; c.1277A>C; c.1231_1234del; c.1228G>C; c.1228G>A; c.1224C>G; c.1223A>T; c.1222T>C; c.1221C>A; c.1220A>G; c.1219A>T; c.1214del; c.1211G>T; c.1211G>A; c.1210del; c.1210C>G; c.1210C>A; c.1210C>T; c.1199G>A; c.1190C>T; c.1155_1162del; c.1146C>A; c.1139_1140delinsTA; c.1139G>A; c.1138T>C; c.1113G>A; c.1112G>A; c.1097G>T; c.1090A>G; c.1087C>T; c.1085G>A; c.1084C>G; c.1084C>T; c.1083C>G; c.1083C>A; c.1080_1081del; c.1079T>C; c.1076_1077dup; c.1066del; c.1057del; c.1055G>A; c.1054C>T; c.1033_1042dup; c.1040G>T; c.1039G>A; c.1031del; c.1022T>C; c.1012del; c.1004del; c.998C>T; c.995del; c.991C>T; c.986C>T; c.981C>A; c.977T>G; c.976G>T; c.969_970del; c.964-31_970delinsTGTCACCCCGC; c.970T>C; c.964-1G>T; c.964-1G>C; c.964-2A>C; c.963+2T>G; c.963+1G>A; c.963+1G>T; c.953A>G; c.952del; c.952T>C; c.952T>A; c.949del; c.939G>A; c.938G>A; c.934_935del; c.926G>A; c.925G>A; c.915C>G; c.911G>A; c.908G>A; c.907G>C; c.907G>A; c.907G>T; c.903_906delinsATTT; c.906C>G; c.904T>G; c.902A>G; c.901C>A; c.895C>T; c.894C>A; c.889A>C; c.877A>T; c.874del; c.870G>A; c.866C>T; c.862G>T; c.861C>A; c.858G>A; c.852C>A; c.841G>A; c.839A>G; c.838T>G; c.832-1G>C; c.831+2T>A; c.831+1G>T; c.820_825del; c.822C>A; c.821A>C; c.821A>G; c.818T>G; c.817G>A; c.808A>G;</p>

	<p>c.804del; c.803del; c.782A>G; c.770C>G; c.761_762insT; c.748del; c.744G>A; c.744G>T; c.740C>T; c.739G>T; c.739G>A; c.720_735del; c.730G>A; c.728C>G; c.727C>T; c.725G>T; c.725G>C; c.725G>A; c.724C>T; c.704T>C; c.695_701dup; c.696G>A; c.683G>A; c.682C>T; c.680dup; c.670G>A; c.644_659del; c.655T>C; c.655T>G; c.651C>G; c.651C>A; c.636del; c.634del; c.630del; c.627C>A; c.627-1G>A; c.626+2dup; c.626+2del; c.626+1G>C; c.626+1G>T; c.600C>G; c.592A>G; c.575C>T; c.567T>A; c.546_548del; c.548G>A; c.546G>C; c.546G>T; c.546G>A; c.545G>A; c.533T>A; c.523G>C; c.521T>C; c.506C>T; c.501G>A; c.495_496del; c.474G>A; c.473G>A; c.470T>C; c.461C>T; c.461C>G; c.453G>A; c.452G>A; c.445C>T; c.440G>A; c.439G>A; c.438T>G; c.433A>C; c.429T>G; c.422del; c.413-1G>A; c.413-2A>G; c.385_412+5del; c.412+3A>T; c.412+2T>C; c.401_402del; c.388C>T; c.375C>A; c.375C>G; c.359_374del; c.374A>G; c.373T>C; c.355del; c.356A>T; c.322-1_334del; c.322_330del; c.326T>C; c.322-1G>C; c.322-1G>A; c.322-2A>G; c.321G>C; c.305T>A; c.300T>A; c.296T>C; c.292C>T; c.278C>A; c.278C>T; c.266del; c.248C>A; c.199del; c.197G>A; c.187del; c.187C>T; c.177_178delinsT; c.155_157delinsCCAT; c.152C>T; c.151C>T; c.116C>G; c.111G>A; c.110G>A; c.99G>A; c.99-1G>C; c.98+2_98+6del; c.98+2T>C; c.98+1G>C; c.98+1G>T; c.92G>A; c.91C>T; c.89del; c.89G>A; c.89G>C; c.88G>A; c.82C>T; c.48_78del; c.76C>T; c.16C>T; c.3G>A; c.1A>C; c.1A>G.</p>
<p>HBB NM_000518.5 (409)</p>	<p>c.*132C>T; c.*115_*116del; c.*110_*114del; c.*113A>G; c.*112A>G; c.*110_*111del; c.*111A>G; c.*110_*111insG; c.*110T>C; c.*110T>A; c.*108A>G; c.*93_*105del; c.*32A>C; c.*6C>G; c.440_441dupAC; c.440A>C; c.440A>T; c.439C>G; c.438T>A; c.437A>G; c.436T>A; c.436T>C; c.435G>C; c.429_430delCC; c.431A>G; c.431A>C; c.428C>A; c.422C>T; c.421G>A; c.417dupT; c.397_407del; c.394_404del; c.404T>A; c.389_401del; c.396_397delGA; c.397A>T; c.380_396del; c.395dupA; c.394C>T; c.393T>A; c.389C>T; c.385_388delGCTGinsCCACA; c.388G>C; c.383_385delAGG; c.383A>G; c.383A>G; c.382C>T; c.380T>G; c.380T>A; c.378_379del; c.371_378delCCCCACCA; c.378delA; c.375dupA; c.365A>T; c.364G>A; c.364G>C; c.364G>T; c.363dup; c.354delC; c.349_350insTGAT; c.348_349delinsG; c.345_348dup; c.347C>A; c.339_345dupTGTGCTG; c.346G>C; c.343_344delCTinsG; c.344T>C; c.342dup; c.341T>A; c.340del; c.339T>A; c.337T>C; c.332T>C; c.328delG; c.328G>A; c.328G>C; c.327C>G; c.323dupG; c.323G>A; c.320T>A; c.320T>G; c.319C>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.316-3C>G; c.316-7C>A; c.316-14T>G; c.316-98_316-86dup; c.316-106C>G; c.316-146T>G; c.316-197C>A; c.316-202delT; c.315+4_315+5del; c.315+5G>C; c.315+2delT; c.315+1delG; c.315+2T>C; c.315+2T>G; c.315+1_315+2insACGTTCTC; c.315+1G>A; c.315+1G>C; c.315+1G>T; c.299_315dup; c.312C>G; c.306G>C; c.305A>G; c.304G>A; c.301_302delCCinsTCTGAGAACTT; c.302C>T; c.300T>A; c.299A>C; c.299A>G; c.299A>T; c.298G>A; c.298G>C; c.298G>T; c.292_295dupCAGC; c.272_295dup; c.295G>A; c.294C>G; c.294C>A; c.293A>T; c.287dupA; c.282_283dupTG; c.283G>C; c.282del; c.279_280insGTGG; c.279C>G; c.279C>A; c.277C>T; c.275dupT; c.275T>C; c.273G>C; c.270_271delTG; c.271G>T; c.269del; c.269G>A; c.266_267delTG; c.268A>C; c.263_267dup; c.265_266delCT; c.266dupT; c.266T>C; c.266T>G; c.244_265del; c.263C>T; c.253_260delACCTTTGC; c.258dupT; c.257T>C; c.237_256del; c.255dupC; c.253delA; c.251delG; c.249G>T; c.248A>C; c.248A>T; c.247A>G; c.244delC; c.235delC; c.235dupC; c.233_234del; c.232delC; c.229_230delGC; c.230delC; c.226delC; c.221_224dupATGG; c.224_225insTCCAG; c.217_221delAGTGinsT; c.219_220delTG; c.219dupT; c.217dupA; c.216delT; c.216dupT; c.206_212delTCGGTGC; c.210delT; c.209delG; c.208G>A; c.206T>A; c.206T>C; c.203_204delTG; c.205C>T; c.203T>A; c.201delA; c.202G>A; c.200A>C; c.199A>G; c.199A>T; c.189_195delTCATGGC; c.196A>T; c.194delG; c.191A>G; c.190C>T; c.190C>A; c.184A>T; c.182T>A; c.179delA; c.179A>C; c.165_177delTATGGGCAACCCT; c.178A>T; c.176delC; c.176dupC; c.170_171insGCAACCCTAAGGTG; c.164_168delTTATGinsGGCATCA; c.155_167del; c.166delA; c.166dupA; c.165delT; c.163dupG; c.162delT; c.160G>A; c.158delA; c.155delT; c.153delT; c.150delC; c.147delG; c.143_146dupATCT; c.146delT; c.145_146insATCT; c.143dupA; c.142dupG; c.138delT; c.138dupT; c.137T>C; c.137T>G; c.135delC; c.130dupG; c.126_129delCTTT; c.127_129delTTT; c.129delT; c.129dupT; c.130G>T; c.126_129dup; c.128T>C; c.126delC; c.127T>C; c.127T>G; c.126dup; c.125dupT; c.123delG; c.118_121dup; c.114_120delGACCCAG; c.111_118delTTGGACCC; c.117_118delCC; c.118delC; c.118C>T; c.114del; c.114G>A; c.112delT; c.113G>A; c.110delC; c.110C>A; c.108C>A; c.108C>G; c.107A>G; c.105_106insCTAC; c.102_104del; c.103G>T; c.93-33_101del; c.98T>A; c.98T>C; c.93-21_96del; c.93-30_96delinsGTCCCTGGGCTGTTTTCTACCCCTCAGATTA; c.93-33_96delinsACTGTCCCTGGGCTGTTTTCTACCCCTCAGATTA; c.93-22_95del; c.94delC; c.93-1_94dup; c.93G>T; c.93-17_93-1delTATTTTCCCACCCTTAG; c.93G>C; c.93-1G>A; c.93-1G>C; c.93-2A>C; c.93-2A>G; c.93-3T>G; c.93-15T>G; c.93-21G>A; c.76_92+27del; c.92+6T>C; c.92+5G>A; c.92+5G>C; c.92+5G>T; c.92+2T>A; c.92+2T>C; c.92+2T>G; c.92+1G>A; c.92+1G>C; c.92+1G>T; c.91delA; c.92G>A; c.92G>C; c.92G>T; c.91A>C; c.91A>G; c.89delG; c.90C>T; c.89G>A; c.85delC; c.85dupC; c.86T>A; c.86T>C; c.83C>A; c.82G>T; c.82G>A; c.81G>C; c.79_80insT; c.80A>G; c.78dupT; c.79G>A; c.79G>C; c.68_74delAAGTTGG; c.74delG; c.75T>A; c.71_73delTTG; c.74delGinsCAC; c.70G>T; c.67G>T; c.64dupG; c.61G>A; c.59A>G; c.51delC; c.52A>T; c.50delG; c.50dupG; c.51C>T; c.48G>A; c.46delT; c.47G>A; c.45dupG; c.20_45del; c.44delT; c.44dupT; c.43del; c.36delT; c.33delC; c.33dupC; c.20_32delAGGAGAAGTCTGC; c.30dupT; c.28_29insTA; c.27_28insAGAA; c.27dupG; c.25_26delAA; c.24dupG; c.25A>G; c.-22_23dup; c.22_24del; c.22G>T; c.20_21del; c.20delA; c.18_19delTG; c.19_20delGainsAT; c.20A>T; c.19del; c.17_18delCT; c.19G>A; c.19G>T; c.15_19delinsATCTT; c.17del; c.8_16delinsCTGAGGTGAAGTCTGCTGAGGAGAAGT; c.11dupT; c.9dup; c.8delA; c.4delG; c.3G>A; c.3G>C; c.3G>T; c.2T>A; c.2T>C; c.2T>G; c.1A>C; c.1A>G; c.-18C>G; c.-29G>A; c.-50A>C; c.-77_-76del; c.-75G>T; c.-76A>C; c.-77A>T; c.-78A>C; c.-78A>G; c.-79A>G; c.-79A>C; c.-80T>A; c.-80T>C; c.-80T>G; c.-81A>G; c.-82C>T; c.-121C>T; c.-122T>A; c.-126C>A; c.-136C>T; c.-136C>G; c.-136C>A; c.-137C>A; c.-137C>G; c.-137C>T; c.-138C>A; c.-138C>T; c.-140C>T; c.-140C>G; c.-142C>T; c.-151C>G; c.-151C>A.</p>
<p>PAH NM_000277.3 (882)</p>	<p>c.1357_*2del; c.1355dup; c.1340C>A; c.1339G>A; c.1334del; c.1330C>T; c.1318G>T; c.1316-1G>A; c.1316-1G>C; c.1316-2A>C; c.1316-2A>G; c.1315+6T>A; c.1314_1315+4del; c.1315+5G>C; c.1315+4A>G; c.1315+2T>C; c.1315+1G>A; c.1315+1G>T; c.1306del; c.1304A>T; c.1301C>A; c.1301C>T; c.1298dup; c.1293_1294del; c.1289T>C; c.1285C>A; c.1282del; c.1282C>T; c.1271T>A; c.1262T>G; c.1262T>C; c.1259G>C; c.1259G>T; c.1258A>G; c.1256A>G; c.1253C>A; c.1253C>T; c.1252A>C; c.1250A>G; c.1249T>C; c.1249T>G; c.1247del; c.1247C>T; c.1247C>A; c.1246C>A; c.1244A>T; c.1243G>A; c.1243G>T; c.1242C>A; c.1241A>G; c.1238_1241delinsCCTG; c.1240T>C; c.1238G>A; c.1238G>C; c.1237C>A; c.1232C>G; c.1232C>A; c.1229T>G; c.1228T>A; c.1223G>A; c.1222C>T; c.1215_1219del; c.1220C>T; c.1219C>T; c.1218A>G; c.1217T>C; c.1216A>G; c.1209del; c.1208C>T; c.1204T>G; c.1204T>C; c.1200del; c.1200G>T; c.1200-1G>A; c.1200-1G>C; c.1200-2A>C; c.1200-2A>G; c.1200-3T>G; c.1200-8G>A; c.1199+20G>C; c.1199+17G>A; c.1199+5G>T; c.1199+4A>G; c.1199+2T>G; c.1199+1G>A; c.1199+1G>C; c.1199+1G>T; c.1196_1199del; c.1198del; c.1199G>A; c.1199G>C; c.1198A>C; c.1197A>T; c.1196T>C; c.1194A>G; c.1192A>T; c.1189G>T; c.1187A>G; c.1184C>A; c.1184C>G; c.1183G>C; c.1180del; c.1181A>C; c.1179_1180del; c.1180G>C; c.1180G>T; c.1177_1178insT; c.1176dup; c.1175T>C; c.1174T>A; c.1171_1172del; c.1172G>C; c.1172G>T; c.1171A>G; c.1169A>G; c.1158_1167del; c.1166del; c.1163_1164del; c.1163T>C; c.1162G>A; c.1162G>C; c.1161C>A; c.1161C>G; c.1159T>G; c.1159T>C; c.1157A>G; c.1156T>G; c.1155_1156delinsGC; c.1154T>A; c.1153del; c.1153dup; c.1154T>C; c.1149del; c.1147C>T; c.1146C>G; c.1144T>C; c.1138dup; c.1139C>A; c.1139C>T; c.1129del; c.1130A>G; c.1129T>G; c.1127del; c.1125A>C; c.1123C>G; c.1117_1118del; c.1118C>A; c.1111_1117del; c.1115_1116dup; c.1117G>A;</p>

<p>c.1115C>G; c.1114A>T; c.1111A>T; c.1109A>G; c.1108del; c.1092_1106del; c.1105C>G; c.1102G>A; c.1099del; c.1099dup; c.1098_1099insTCTTATCAGAGAAGCCAAAGCTTCTCCCC; c.1097C>T; c.1097C>A; c.1096C>A; c.1092_1094del; c.1093del; c.1089del; c.1087A>T; c.1085C>G; c.1085C>T; c.1084C>T; c.1084C>A; c.1081A>T; c.1081A>C; c.1078G>T; c.1076C>G; c.1076C>T; c.1074A>T; c.1066-10_1070del; c.1071C>A; c.1069T>G; c.1069T>C; c.1068C>A; c.1068C>G; c.1066-1G>A; c.1066-1G>C; c.1066-1G>T; c.1066-2A>T; c.1066-3C>T; c.1066-7C>A; c.1066-11G>A; c.1066-14C>G; c.1065+3A>G; c.1065+1G>A; c.1065+1G>T; c.1063C>T; c.1057del; c.1056del; c.1055del; c.1054G>T; c.1054G>C; c.1043_1053del; c.1049C>A; c.1049C>G; c.1048T>A; c.1044_1047dup; c.1046C>A; c.1046C>T; c.1045T>C; c.1045T>G; c.1043T>C; c.1042C>G; c.1038del; c.1039C>T; c.1037G>A; c.1033G>A; c.1033G>T; c.1031G>A; c.1030G>A; c.1029T>A; c.1029T>G; c.1028A>G; c.1027T>A; c.1027T>G; c.1024del; c.1025C>A; c.1011_1024dup; c.1024G>C; c.1024G>A; c.1023G>C; c.1022A>C; c.1021A>T; c.1016C>T; c.1013A>C; c.1013A>T; c.1012G>T; c.1007A>G; c.1006C>T; c.1004A>C; c.1002C>A; c.1001G>C; c.998T>C; c.997C>T; c.994G>A; c.992T>C; c.992T>G; c.990G>C; c.982A>G; c.978G>A; c.976del; c.977G>A; c.977G>C; c.973_976del; c.975C>G; c.974A>G; c.971T>A; c.970-1G>A; c.970-1G>C; c.970-1G>T; c.970-2A>G; c.970-2A>C; c.969+5G>A; c.969+2T>G; c.967_969del; c.969+1G>A; c.965C>G; c.964G>A; c.960G>C; c.957_958insTT; c.956A>G; c.955G>T; c.946G>T; c.943G>T; c.941del; c.930_940del; c.941C>A; c.940C>A; c.940C>T; c.938C>A; c.938C>T; c.937G>A; c.935del; c.935G>T; c.935G>A; c.934G>T; c.931_932del; c.932T>C; c.929C>T; c.929C>G; c.926C>A; c.926C>T; c.925G>T; c.925G>A; c.922C>T; c.922C>G; c.920del; c.920G>T; c.920G>A; c.916del; c.916A>G; c.913-2A>C; c.913-2A>G; c.913-7A>G; c.912+2T>C; c.912+1G>A; c.912+1G>C; c.912+1G>T; c.912G>A; c.911A>G; c.910C>T; c.910C>A; c.907del; c.908C>T; c.907T>C; c.907T>G; c.903G>T; c.902A>C; c.901del; c.901C>A; c.901C>T; c.899C>G; c.899C>T; c.898G>A; c.898G>T; c.896T>G; c.895T>C; c.890G>A; c.890G>T; c.889C>T; c.887A>G; c.886G>A; c.884C>G; c.873_880del; c.878del; c.875C>T; c.870T>A; c.870T>G; c.869A>T; c.868C>T; c.865G>A; c.865G>C; c.859C>G; c.859C>A; c.856G>A; c.851G>A; c.850T>C; c.848T>A; c.847A>T; c.847A>G; c.845A>G; c.844G>A; c.843-1G>A; c.843-2A>T; c.843-5T>C; c.842+5G>A; c.842+4A>G; c.842+3G>C; c.842+2T>A; c.842+1G>A; c.842+1G>T; c.842C>T; c.842C>G; c.842C>A; c.841C>G; c.841C>T; c.839_840insTT; c.839A>G; c.839A>C; c.837del; c.838G>A; c.838G>T; c.837dup; c.838G>C; c.836C>T; c.835_836delinsTG; c.835C>G; c.835C>A; c.822_832del; c.833C>A; c.833C>G; c.833C>T; c.832A>G; c.830_831insC; c.830A>G; c.829T>G; c.828G>A; c.827T>A; c.827T>C; c.827T>G; c.826A>G; c.824C>G; c.824C>T; c.823C>T; c.809_820del; c.818C>T; c.810_814del; c.814G>T; c.813T>G; c.812A>G; c.812A>T; c.811C>A; c.810A>T; c.809G>A; c.808A>G; c.806del; c.806T>A; c.805A>C; c.805A>G; c.804C>A; c.803A>G; c.801del; c.801G>C; c.800dup; c.800A>G; c.800A>T; c.799C>G; c.799C>T; c.797C>T; c.796A>C; c.795C>A; c.794G>A; c.792del; c.790del; c.789C>A; c.788T>C; c.785T>G; c.782G>C; c.782G>T; c.781del; c.781C>G; c.781C>T; c.780C>G; c.776C>T; c.775G>A; c.773T>C; c.770G>T; c.764T>C; c.757G>A; c.745_755del; c.755G>A; c.755G>C; c.753_754del; c.754C>G; c.754C>T; c.753del; c.745del; c.746T>A; c.745C>T; c.740del; c.740G>T; c.739G>A; c.739G>C; c.737del; c.737C>A; c.735_737delinsAG; c.737C>T; c.734_735delinsCA; c.733_735delinsCTA; c.722_735delinsCCTCCGACCTGTA; c.734T>A; c.734T>C; c.733G>C; c.733G>A; c.731C>T; c.730C>T; c.728G>A; c.728G>T; c.727C>T; c.724C>T; c.722G>A; c.722G>T; c.721C>T; c.721C>G; c.719T>C; c.718T>G; c.716G>T; c.716G>C; c.716G>A; c.713C>T; c.712A>C; c.712A>G; c.707-12_711del; c.707-1G>A; c.707-2del; c.707-1G>C; c.707-2A>T; c.707-2A>G; c.706+608A>C; c.706+531T>C; c.706+368T>C; c.704A>C; c.703C>T; c.699C>G; c.699C>A; c.697T>A; c.694_696delinsTAA; c.693_694del; c.693del; c.694C>G; c.694C>T; c.692C>T; c.691T>C; c.690_691insG; c.689T>C; c.688G>A; c.677_686del; c.686dup; c.686A>G; c.684A>C; c.682G>A; c.682G>T; c.680T>A; c.679C>G; c.678G>C; c.676del; c.676C>A; c.676C>T; c.667_674dup; c.674C>T; c.673C>A; c.673C>G; c.671T>C; c.670A>G; c.670A>C; c.669del; c.668A>T; c.664_665del; c.666T>G; c.665A>G; c.665A>T; c.662A>G; c.659A>C; c.653G>T; c.650G>T; c.649T>G; c.649T>C; c.648C>G; c.641A>G; c.640G>T; c.638T>C; c.635T>C; c.632del; c.632dup; c.632C>T; c.632C>A; c.631C>A; c.618_625del; c.620A>G; c.618C>A; c.618C>G; c.617A>G; c.615G>C; c.593_614del; c.614A>G; c.592_613del; c.613G>A; c.612T>G; c.610dup; c.611A>G; c.609C>G; c.586_608del; c.607dup; c.608G>A; c.607T>G; c.605C>T; c.604G>A; c.603T>G; c.602A>G; c.601C>T; c.598dup; c.594T>A; c.594T>G; c.591G>C; c.590T>A; c.584dup; c.580_581del; c.581T>C; c.581T>G; c.574_580del; c.578C>T; c.569T>C; c.563del; c.563G>A; c.563G>T; c.562G>A; c.561G>A; c.561G>C; c.558_559del; c.560G>A; c.559T>C; c.556del; c.557C>T; c.556A>G; c.552G>T; c.551del; c.547_548delinsTT; c.548A>G; c.547G>T; c.543_545del; c.545A>G; c.544G>A; c.535_536del; c.535T>A; c.535T>C; c.533A>G; c.533A>T; c.532G>A; c.529G>A; c.529G>C; c.527G>A; c.527G>T; c.527G>C; c.526C>T; c.523C>T; c.521T>C; c.521T>A; c.520A>G; c.516G>C; c.516G>T; c.515A>G; c.514C>T; c.512G>C; c.511G>A; c.511G>T; c.511G>C; c.510T>A; c.510T>G; c.510-1G>C; c.510-1G>A; c.510-2A>G; c.510-5T>A; c.509+1G>A; c.509A>C; c.508C>G; c.506G>A; c.506G>C; c.506G>T; c.505C>A; c.505C>T; c.505C>G; c.504C>G; c.503del; c.504C>A; c.500A>T; c.498C>A; c.498C>G; c.493_496del; c.494C>A; c.493G>A; c.491T>C; c.490A>G; c.482T>C; c.478C>T; c.473G>A; c.472C>T; c.472C>G; c.470_471delinsAC; c.470G>C; c.470G>T; c.467del; c.467C>G; c.467C>T; c.466G>C; c.464G>A; c.464G>C; c.456_463dup; c.463del; c.463C>T; c.462C>A; c.461A>T; c.461A>G; c.460T>C; c.453T>A; c.452A>G; c.450dup; c.451G>C; c.447del; c.443G>A; c.443G>T; c.442G>A; c.442G>C; c.442-1G>A; c.442-1G>C; c.442-2A>C; c.442-2A>G; c.442-5C>G; c.441+6T>A; c.441+5G>T; c.441+5G>C; c.441+5G>A; c.441+4A>G; c.441+3G>C; c.441+2T>G; c.441+2T>A; c.441+1G>A; c.441+1G>C; c.440del; c.440C>T; c.439C>T; c.435C>A; c.434A>T; c.428A>G; c.428A>T; c.398_401del; c.400C>T; c.395C>T; c.386A>G; c.385G>T; c.365del; c.365_366delinsT; c.365C>A; c.364C>T; c.361T>C; c.361T>G; c.360G>A; c.359G>A; c.357del; c.356C>T; c.355C>T; c.353-1G>A; c.353-2A>G; c.353-2A>C; c.353-2A>T; c.353-6T>A; c.352+2dup; c.352+1G>A; c.350_351del; c.350del; c.344_347del; c.346_347del; c.338_341del; c.337A>T; c.331C>T; c.329del; c.329C>T; c.324_328del; c.320A>G; c.320A>C; c.311C>A; c.311C>T; c.308G>A; c.305T>C; c.301G>A; c.293T>C; c.292T>G; c.284_286del; c.280_283del; c.283A>T; c.281T>G; c.277_280del; c.275C>T; c.266_267insG; c.266dup; c.265C>A; c.265C>T; c.261C>A; c.257G>C; c.256del; c.241_256del; c.253A>T; c.250G>T; c.250G>A; c.242C>A; c.241A>C; c.239dup; c.234dup; c.232G>A; c.231T>G; c.227A>G; c.227A>C; c.226G>T; c.224A>G; c.224A>C; c.223G>A; c.223G>C; c.212G>A; c.212G>C; c.208_210del; c.211C>T; c.210del; c.209C>T; c.208T>C; c.206dup; c.205C>T; c.197_204del; c.204A>T; c.202A>G; c.200C>A; c.199T>C; c.199T>G; c.196G>T; c.196G>A; c.190_194del; c.194T>A; c.194T>C; c.194T>G; c.193A>G; c.189_190dup; c.190del; c.187_190delinsCCCA; c.185_188dup; c.184_185insCTGA; c.184del; c.185T>C; c.183C>A; c.183C>G; c.182A>G; c.181A>G; c.171_172del; c.169_171del; c.169G>A; c.169G>T; c.169-1G>A; c.169-2A>G; c.169-13T>G; c.168+5G>A; c.168+5G>C; c.168+2T>C; c.168+2T>A; c.168_168+1delinsAA; c.168+1G>A; c.168+1G>T; c.168G>T; c.165del; c.163_165del; c.165T>G; c.164T>C; c.161T>C; c.157C>A; c.157C>T; c.155del; c.155T>C; c.148A>T; c.143T>C; c.140C>T; c.137del; c.136G>A; c.136G>C; c.131_133del; c.133G>T; c.130G>T; c.127del; c.127G>T; c.124_126del; c.125A>T; c.125A>G; c.122T>C; c.120_121del; c.121C>T; c.116_118del; c.117C>G; c.111_115dup; c.111dup; c.110T>C; c.78del; c.75del; c.72T>A; c.60+5G>C; c.60+5G>T; c.60+5G>A; c.60+4A>T; c.60+2del; c.60+1G>C; c.58C>T; c.47_48del; c.48dup; c.47_48dup; c.47_48insCT; c.43_44insAG; c.36del; c.32T>A; c.30dup; c.23del; c.13del; c.3G>A; c.3G>C; c.2T>C; c.2T>G; c.1A>G; c.1A>T.</p>

<p>PMM2 NM_000303.3 (257)</p>	<p>c.-167G>T; c.1A>G; c.17_28del; c.16_22del; c.24delC; c.26G>A; c.26G>T; c.28del; c.27C>A; c.28C>T; c.34_36delinsTAA; c.34G>C; c.43G>A; c.44G>C; c.52A>G; c.52A>C; c.53C>G; c.55delG; c.59del; c.58C>T; c.61dup; c.61C>G; c.61C>T; c.64C>T; c.66+1delG; c.66_66+1delinsTA; c.66+1G>T; c.66+1G>A; c.67-2A>T; c.67-2A>G; c.67-1G>C; c.72delT; c.73G>T; c.76A>T; c.79G>T; c.91T>C; c.93C>A; c.94C>G; c.95_96delITainsGC; c.95T>G; c.97C>T; c.104T>A; c.109C>T; c.124G>T; c.124G>A; c.127G>A; c.130G>C; c.130G>A; c.131T>C; c.140C>T; c.157C>T; c.160dupG; c.173del; c.178G>T; c.178+1G>A; c.178+2T>G; c.179-25A>G; c.179-2A>G; c.179-1G>C; c.179-1G>T; c.189delA; c.190delT; c.192C>G; c.193G>T; c.193G>A; c.194A>G; c.196dup; c.197_198del; c.201_202del; c.200T>G; c.203T>G; c.205C>T; c.205C>A; c.206C>T; c.208G>T; c.228C>A; c.231del; c.237del; c.243_244del; c.249_250insCTACTTGTGT; c.250A>T; c.253C>T; c.255G>A; c.255+1G>A; c.255+1G>C; c.255+2T>C; c.255+2T>G; c.256-2A>G; c.256-1G>C; c.256-1G>A; c.264_265del; c.284del; c.295_296del; c.296T>A; c.303C>G; c.305A>G; c.309T>A; c.310C>G; c.317A>T; c.317A>G; c.323C>T; c.324delG; c.325A>T; c.328_329insGGGAA; c.337C>A; c.338C>T; c.340A>T; c.345dupG; c.347+1G>A; c.347+2T>C; c.347+5G>A; c.348-2del; c.348-2A>C; c.348-2A>G; c.348-1G>C; c.349G>C; c.349G>T; c.353C>G; c.355T>C; c.356T>C; c.357C>A; c.359T>C; c.360T>G; c.365del; c.367C>T; c.368G>A; c.375dup; c.377_381del; c.385G>A; c.385G>C; c.385G>T; c.386T>C; c.392delC; c.391C>G; c.392C>T; c.394A>T; c.395T>C; c.395T>G; c.414delA; c.415G>A; c.421C>T; c.422G>A; c.422G>T; c.430T>C; c.430T>G; c.431T>G; c.432C>A; c.435C>G; c.436G>T; c.442G>A; c.442G>T; c.447+1G>A; c.447+3dup; c.447+2T>A; c.447+5G>A; c.448-1_448delGA; c.451_454delGAAA; c.448-2A>C; c.448-2A>T; c.448-2A>G; c.448-1G>A; c.454_455delAA; c.451G>T; c.457dup; c.458_462del; c.457A>G; c.458T>C; c.458T>A; c.458T>G; c.459A>G; c.460A>T; c.463C>T; c.467_468delinsT; c.471del; c.470T>C; c.470T>G; c.484C>T; c.488_491delAAGA; c.485G>C; c.511dupA; c.523+1del; c.523G>T; c.523G>C; c.524-2A>G; c.526G>A; c.527G>T; c.527G>A; c.531G>C; c.548T>C; c.549T>G; c.550C>A; c.554A>G; c.556G>A; c.562del; c.560G>A; c.561G>A; c.562G>T; c.563A>G; c.564C>A; c.566_571delinsGTGGATTTC; c.572dup; c.573C>A; c.574T>A; c.574T>C; c.575G>C; c.577C>G; c.580C>T; c.584_585del; c.584A>G; c.599_603del; c.603T>G; c.617del; c.618C>A; c.620T>C; c.623G>C; c.639+1G>A; c.639+2T>C; c.640-15479C>T; c.640-9T>G; c.640G>A; c.640G>C; c.646A>G; c.647A>G; c.647A>T; c.652C>G; c.653A>T; c.658A>G; c.661T>G; c.667G>A; c.672del; c.669C>G; c.669C>A; c.677C>T; c.682G>T; c.682G>A; c.682G>C; c.686A>C; c.687C>G; c.687C>A; c.691G>A; c.691G>C; c.691G>T; c.696del; c.703G>T; c.710C>G; c.710C>T; c.713G>A; c.713G>C; c.715A>T; c.716G>C; c.717G>C; c.717G>T; c.721T>C; c.722G>C; c.723T>G; c.728T>C.</p>
<p>GAA NM_000152.5 (727)</p>	<p>c.-32-17_-32-10delinsTCCCTGCTGAGCCTCTACAGGCCTCCCGC; c.-32-13T>G; c.-32-3C>A; c.-32-2A>G; c.-32-1G>C; c.1A>G; c.1A>T; c.2T>C; c.3G>A; c.3G>T; c.3_4delinsTT; c.4G>T; c.40_47del; c.55del; c.84dup; c.118C>T; c.133del; c.148G>T; c.156_157del; c.169C>T; c.172C>T; c.199del; c.213_217dup; c.236_246del; c.241C>T; c.242del; c.246C>A; c.258del; c.258dup; c.265C>A; c.266G>A; c.266G>C; c.276C>A; c.281_282del; c.282del; c.301G>T; c.304C>T; c.307T>C; c.307T>G; c.308G>A; c.309C>G; c.309C>A; c.316C>G; c.317G>A; c.322T>A; c.323G>A; c.340_341insT; c.342del; c.343C>T; c.352C>T; c.355G>T; c.361C>T; c.365del; c.364A>G; c.371_381del; c.370C>T; c.376del; c.377G>A; c.379_380del; c.378G>A; c.389del; c.393del; c.396dup; c.399C>A; c.424_440del; c.437del; c.444C>G; c.448dup; c.456_457dup; c.461_469del; c.460_465del; c.460C>T; c.461G>C; c.465dup; c.471del; c.482_483del; c.490_524del; c.498_510del; c.511del; c.520_526del; c.525_526del; c.525del; c.528_529del; c.533G>A; c.541_545del; c.546+2_546+5del; c.546G>T; c.546G>C; c.546G>A; c.546+1G>T; c.546+2T>C; c.547-1G>C; c.568C>T; c.569G>A; c.569G>T; c.573C>A; c.584T>A; c.587del; c.596_600delinsGTGT; c.639del; c.643G>T; c.658dup; c.655G>A; c.656G>A; c.670C>T; c.671G>C; c.671G>A; c.671G>T; c.683dup; c.692+1G>C; c.692+1G>A; c.692+1G>T; c.692+2T>C; c.692+5G>T; c.693-2A>C; c.693-1G>A; c.693-1G>C; c.698del; c.700A>G; c.701C>T; c.706del; c.711_712dup; c.715_716del; c.716del; c.722_723del; c.727G>A; c.727_728insCCACCAGGACA; c.729C>A; c.732G>C; c.736del; c.739C>T; c.742del; c.743T>C; c.752C>T; c.755dup; c.759del; c.761C>T; c.763C>T; c.766_785delinsC; c.766_767insC; c.768dup; c.784G>A; c.796C>T; c.797C>T; c.799_803delinsA; c.806_830del; c.827_845del; c.829_851del; c.829_830del; c.836G>A; c.841C>T; c.844G>C; c.853C>T; c.854C>G; c.856del; c.858G>A; c.858+1G>A; c.858+2T>A; c.859-2A>G; c.859-1G>C; c.873_874del; c.871C>T; c.872T>C; c.874T>C; c.875A>G; c.876C>G; c.877G>A; c.877G>C; c.883C>A; c.889_890insG; c.896T>C; c.896T>G; c.904G>T; c.917C>A; c.922C>G; c.922C>T; c.923A>C; c.925G>A; c.925G>T; c.930del; c.934del; c.935del; c.952A>T; c.953T>A; c.953T>C; c.955+1del; c.955+1G>A; c.956-5_957del; c.967C>T; c.971dup; c.971C>T; c.988_998del; c.989G>A; c.990G>C; c.995C>A; c.1000G>A; c.1001G>A; c.1003G>A; c.1003G>T; c.1004G>A; c.1020C>G; c.1030_1031del; c.1051del; c.1054C>T; c.1057del; c.1062C>A; c.1064T>C; c.1075G>T; c.1075G>A; c.1075+1G>A; c.1075+1G>T; c.1075+2T>C; c.1075+2T>A; c.1076-22T>G; c.1076-2A>G; c.1076-1G>A; c.1076-1G>C; c.1082dup; c.1082C>T; c.1082C>G; c.1091del; c.1099del; c.1099T>C; c.1100G>A; c.1105dup; c.1103G>A; c.1106T>A; c.1106T>C; c.1111_1115delinsCCACTGGGG; c.1114C>T; c.1115A>T; c.1117del; c.1123C>A; c.1124G>T; c.1124G>A; c.1130del; c.1128_1129delinsC; c.1129G>C; c.1134C>G; c.1134C>A; c.1143del; c.1153del; c.1156C>T; c.1165del; c.1192del; c.1192dup; c.1190C>T; c.1193del; c.1193T>C; c.1194+2T>C; c.1194+2T>G; c.1194+5G>A; c.1195-2A>G; c.1195-2A>C; c.1195-1G>A; c.1198del; c.1205G>A; c.1210G>A; c.1211A>G; c.1214T>C; c.1216G>A; c.1219T>C; c.1220A>G; c.1221C>A; c.1222A>G; c.1222A>C; c.1225dup; c.1223T>C; c.1226_1227insG; c.1241del; c.1239C>G; c.1243_1244del; c.1274del; c.1280T>C; c.1285C>T; c.1291_1299del; c.1293_1312del; c.1292_1295dup; c.1298_1301delinsTCCTGCACCATGGC; c.1308del; c.1309C>T; c.1309C>A; c.1320_1322del; c.1314C>A; c.1316T>A; c.1326+1G>A; c.1326+2T>C; c.1327-2A>G; c.1336dup; c.1347_1365dup; c.1347_1350del; c.1356_1365del; c.1356del; c.1364A>G; c.1365del; c.1369C>T; c.1370C>A; c.1370C>G; c.1370C>T; c.1374C>A; c.1374_1375delinsTA; c.1376_1377del; c.1375G>C; c.1377dup; c.1376A>T; c.1388_1405del; c.1396del; c.1396dup; c.1396G>T; c.1400T>C; c.1402A>T; c.1407del; c.1408_1410del; c.1411_1414del; c.1417_1420del; c.1420C>T; c.1426_1429delinsGTTGG; c.1431del; c.1432G>A; c.1437G>A; c.1437G>C; c.1437+1G>A; c.1437+2T>C; c.1438-2A>G; c.1438-2A>C; c.1438-1G>T; c.1438-1G>C; c.1438-1G>A; c.1441del; c.1441T>C; c.1441T>A; c.1443G>A; c.1445C>T; c.1445C>G; c.1447G>A; c.1456G>T; c.1456G>C; c.1464dup; c.1463C>T; c.1465G>A; c.1466A>G; c.1477C>G; c.1478C>T; c.1494del; c.1494G>A; c.1496G>A; c.1501_1515del; c.1497G>A; c.1519dup; c.1525C>T; c.1537G>A; c.1538A>G; c.1541del; c.1547G>A; c.1548G>A; c.1551+3_1551+6del; c.1551+1G>T; c.1551+1G>C; c.1551+1G>A; c.1552-3C>G; c.1552-2A>G; c.1552-2A>C; c.1555A>G; c.1556T>C; c.1557_1561dup; c.1557G>A; c.1559del; c.1559A>T; c.1560del; c.1560C>G; c.1561del; c.1561G>A; c.1561G>C; c.1562A>T; c.1563G>T; c.1564C>T; c.1564C>A; c.1564C>G; c.1567del; c.1565C>T; c.1571A>C; c.1575dup; c.1575_1576del; c.1579del; c.1579_1580del; c.1583G>T; c.1583G>C; c.1585T>C; c.1585_1586delinsGT; c.1585T>G; c.1589_1598del; c.1587del; c.1588G>T; c.1589del; c.1594G>A; c.1602_1605delinsAGG; c.1610del; c.1621_1622del; c.1626del; c.1633C>T; c.1634C>T; c.1636+1G>A; c.1637-2A>G; c.1650dup; c.1650del; c.1645G>C; c.1645G>A; c.1646G>A; c.1651_1656del; c.1654del; c.1654C>T; c.1655T>C; c.1657C>T; c.1663G>C; c.1669A>T; c.1670T>G; c.1674_1675del; c.1673G>C; c.1673G>A; c.1687C>T; c.1692del; c.1694_1697del; c.1697C>T; c.1703A>G; c.1703A>T; c.1704dup; c.1705dup; c.1707C>G; c.1709A>G; c.1710C>G; c.1714C>T; c.1716del; c.1716C>G; c.1721T>C; c.1725C>A; c.1727G>A; c.1735G>A; c.1748C>T; c.1754+1_1754+7del; c.1754G>A; c.1754+1G>A; c.1754+2T>A; c.1754+2T>C; c.1755-1G>A; c.1761_1765delinsT; c.1780C>T; c.1781G>A; c.1781G>C; c.1784C>T; c.1798del; c.1796C>A; c.1798C>T; c.1798C>G; c.1799G>A; c.1799G>T; c.1802C>A; c.1802C>G; c.1802C>T; c.1804A>G; c.1809del;</p>

	<p>c.1819_1836del; c.1814del; c.1819G>T; c.1820G>A; c.1824_1828dup; c.1822C>T; c.1826dup; c.1827del; c.1827_1828insA; c.1827C>G; c.1827C>A; c.1829C>T; c.1831G>A; c.1832G>A; c.1836C>G; c.1841C>T; c.1841C>A; c.1846dup; c.1846del; c.1843G>A; c.1844G>A; c.1844G>T; c.1847dup; c.1846G>T; c.1846G>A; c.1848dup; c.1848C>A; c.1853G>A; c.1855A>C; c.1855A>G; c.1856G>A; c.1857C>G; c.1861T>C; c.1874_1882del; c.1880C>T; c.1888+1G>A; c.1895T>G; c.1903A>G; c.1905C>A; c.1905C>G; c.1910_1918del; c.1912G>T; c.1913G>T; c.1913G>A; c.1924G>T; c.1930del; c.1927G>A; c.1927G>T; c.1928G>C; c.1930_1936dup; c.1930G>T; c.1933G>T; c.1933G>C; c.1933G>A; c.1934A>G; c.1935C>A; c.1937del; c.1940G>A; c.1941C>A; c.1941C>G; c.1942G>A; c.1942G>T; c.1943G>A; c.1943G>T; c.1944_1946delinsAA; c.1952del; c.1950_1952delinsT; c.1951_1952delinsT; c.1958C>A; c.1961C>A; c.1962_1964del; c.1961C>G; c.1966del; c.1976_1977del; c.1975_2000dup; c.1978C>T; c.1978C>G; c.1979G>A; c.1987del; c.1987C>T; c.1996dup; c.2003_2013dup; c.2004C>A; c.2012T>G; c.2014C>T; c.2014C>G; c.2015G>A; c.2015G>T; c.2024_2026del; c.2040+1del; c.2040G>A; c.2040+1G>T; c.2040+1G>C; c.2040+2T>G; c.2041-1G>A; c.2045delinsCG; c.2047G>T; c.2051C>T; c.2055C>A; c.2061del; c.2066_2070dup; c.2066_2069del; c.2074C>T; c.2078dup; c.2082del; c.2088del; c.2092_2099del; c.2095dup; c.2096T>C; c.2104C>T; c.2105G>A; c.2105G>T; c.2105G>C; c.2109C>G; c.2109C>A; c.2122dup; c.2122del; c.2130C>A; c.2131A>T; c.2131A>C; c.2131A>G; c.2136_2137del; c.2140del; c.2143C>T; c.2161del; c.2161dup; c.2161G>T; c.2167_2179delinsTGCGACGTGG; c.2167G>A; c.2173del; c.2173C>T; c.2174G>T; c.2185del; c.2188G>T; c.2189+1G>T; c.2189+2T>A; c.2189+3G>C; c.2210C>A; c.2213G>A; c.2214G>A; c.2219_2220del; c.2227C>T; c.2228A>G; c.2229G>C; c.2236T>G; c.2236T>C; c.2242del; c.2242dup; c.2237G>A; c.2237G>C; c.2238G>A; c.2238G>C; c.2238G>T; c.2242G>T; c.2261dup; c.2263del; c.2269C>T; c.2281delinsAT; c.2284G>T; c.2294G>A; c.2296T>C; c.2296T>G; c.2297A>C; c.2297A>G; c.2300del; c.2303C>G; c.2303C>T; c.2313_2322del; c.2314del; c.2314T>C; c.2320dup; c.2319C>A; c.2326C>T; c.2331+2_2331+3del; c.2331+1G>A; c.2331+1G>C; c.2331+2T>A; c.2331+4A>G; c.2332-2A>G; c.2332-1G>C; c.2332-1G>A; c.2345_2346insA; c.2367dup; c.2369_2370delinsGTGA; c.2380del; c.2383G>T; c.2385del; c.2395del; c.2399G>T; c.2402del; c.2408_2426del; c.2407C>T; c.2411G>A; c.2431dup; c.2431del; c.2432del; c.2434G>A; c.2455C>T; c.2456G>C; c.2456G>A; c.2466C>G; c.2470_2471del; c.2476del; c.2477del; c.2479C>T; c.2481+1G>A; c.2481+1G>T; c.2482-2A>G; c.2483del; c.2482-1G>T; c.2495_2496del; c.2501_2502del; c.2502dup; c.2512C>T; c.2544del; c.2545A>T; c.2553_2554delinsAT; c.2554G>T; c.2560C>T; c.2563G>C; c.2577G>A; c.2585del; c.2608C>T; c.2617dup; c.2619C>G; c.2619C>A; c.2623C>T; c.2642_2643delinsC; c.2644_2645insT; c.2646+1G>A; c.2646+2T>A; c.2646+2T>G; c.2647-20T>G; c.2647-7G>A; c.2647-1_2648del; c.2647-2A>G; c.2655_2656del; c.2655_2656delinsT; c.2662G>T; c.2675_2676dup; c.2672del; c.2704_2716dup; c.2701del; c.2702dup; c.2702T>A; c.2704_2709del; c.2704C>T; c.2707_2709del; c.2706del; c.2716G>A; c.2723del; c.2734del; c.2740dup; c.2738C>G; c.2740C>T; c.2741delinsCAG; c.2743C>T; c.2772del; c.2782T>C; c.2799G>A; c.2799+2C>G; c.2799+4A>G; c.2815_2816del; c.2812del; c.2834_2835del; c.2846T>A; c.2853del.</p>
<p>AGXT NM_000030.3 (348)</p>	<p>c.2_3delinsAT; c.2T>C; c.3G>T; c.23_24delinsAT; c.25dup; c.33dup; c.32_33del; c.33del; c.26_27insA; c.28_29delinsA; c.28C>T; c.31_32insT; c.32_33insT; c.32C>G; c.33delinsAAA; c.40dup; c.67C>T; c.74T>G; c.77T>C; c.79G>T; c.83del; c.106C>G; c.106C>T; c.107G>C; c.107G>A; c.116_117dup; c.126dup; c.126del; c.121G>C; c.121G>A; c.122G>T; c.122G>A; c.125G>A; c.130C>T; c.139G>C; c.139G>T; c.139G>A; c.140G>A; c.165+1G>T; c.166-11_166-8delinsTGCATGCAAGAT; c.166-1_172dup; c.166-2A>G; c.166-1G>A; c.167T>A; c.175G>T; c.175G>A; c.187G>C; c.188G>A; c.190A>T; c.191T>A; c.198C>A; c.198C>G; c.205C>T; c.209C>A; c.215dup; c.214A>G; c.214A>C; c.215A>T; c.216C>G; c.221_222del; c.221_227dup; c.224del; c.239_251del; c.238_239insTTGCCAA; c.242C>G; c.242C>T; c.242C>A; c.244G>A; c.244G>C; c.245G>C; c.245_249delinsAGA; c.245G>A; c.248A>G; c.252_253del; c.252T>A; c.254C>A; c.259del; c.268dup; c.276del; c.278_279insCACACTT; c.279del; c.283_285dupGAG; c.283G>T; c.283G>A; c.292G>C; c.299_307dup; c.302T>C; c.305T>A; c.307G>A; c.308G>A; c.322T>C; c.322del; c.323G>A; c.324G>A; c.324G>T; c.326G>A; c.326G>T; c.331C>T; c.332G>A; c.335C>A; c.349dup; c.346G>T; c.346G>A; c.349G>T; c.352C>A; c.352C>T; c.353G>A; c.358G>T; c.358G>A; c.358+1G>T; c.358+2T>G; c.359-1_382del; c.359-2A>G; c.359-1G>A; c.364C>T; c.374_381del; c.371A>C; c.386_395del; c.389del; c.402C>G; c.406_410dup; c.409C>T; c.412G>T; c.416_418del; c.423G>T; c.423+1G>A; c.423+2T>C; c.424-2A>G; c.433del; c.433C>T; c.439A>T; c.445del; c.447_454del; c.449T>C; c.454T>A; c.455T>G; c.457T>G; c.458T>A; c.460del; c.469del; c.466G>T; c.466G>C; c.466G>A; c.467G>C; c.469G>C; c.469G>T; c.473del; c.473C>A; c.473C>T; c.481G>C; c.481G>A; c.481G>T; c.482G>A; c.490C>T; c.497T>C; c.507_508insA; c.508G>C; c.508G>A; c.517T>C; c.518G>A; c.519_520delinsGA; c.519C>A; c.524+1G>A; c.524+2T>A; c.525-2A>T; c.525-2A>G; c.525-1G>A; c.528C>A; c.528C>G; c.534C>G; c.536delinsACTGGAGG; c.547G>A; c.551C>A; c.557_562delinsATCGGT; c.560_561insGGT; c.560C>A; c.560C>T; c.569_570dup; c.570del; c.568G>C; c.568G>A; c.569_570insA; c.569G>A; c.569G>T; c.569G>C; c.577dup; c.577del; c.579_580del; c.583A>C; c.584T>G; c.585G>A; c.593A>C; c.595G>A; c.595+1G>A; c.595+1G>T; c.595+2T>C; c.595+2T>A; c.595+2T>G; c.596-2A>G; c.596-1G>A; c.597_601del; c.598A>G; c.601G>A; c.602A>T; c.603C>A; c.605T>A; c.612C>A; c.613T>C; c.614C>A; c.614C>T; c.622C>T; c.626del; c.627G>T; c.628G>C; c.632T>G; c.638C>A; c.638C>T; c.642del; c.642_645del; c.646G>A; c.653C>A; c.653C>T; c.658A>T; c.662_664del; c.661T>C; c.665dup; c.667A>C; c.673_676del; c.673A>T; c.679_680+2del; c.680+1G>T; c.680+1G>A; c.680+1G>C; c.680+2T>A; c.680+5G>C; c.681-1G>A; c.681-1G>T; c.682_683delinsAA; c.686_688del; c.693C>G; c.697C>T; c.698G>T; c.698G>A; c.706A>T; c.716_718del; c.725dup; c.727G>C; c.731T>A; c.731T>C; c.733_734del; c.737G>A; c.738G>A; c.744del; c.751T>C; c.751_752delinsAA; c.752G>A; c.753G>A; c.757T>C; c.760_761insT; c.766del; c.776+1G>C; c.776+1G>A; c.776+2T>G; c.777-2A>G; c.777-1G>C; c.779A>G; c.781C>G; c.783T>A; c.795del; c.798_802delinsACAATCTCAG; c.807dup; c.806T>C; c.823_824dup; c.823_824del; c.822G>C; c.823A>C; c.824G>T; c.824G>A; c.825C>A; c.826_827delinsGA; c.832del; c.829_830insA; c.830delinsAA; c.834del; c.838del; c.841G>T; c.844C>T; c.846G>C; c.846+1G>A; c.846+1G>T; c.847-3C>G; c.847-2_847-1del; c.848del; c.847-1G>C; c.851T>C; c.853G>T; c.856delinsCC; c.860_861delinsCG; c.863G>A; c.891T>G; c.893T>C; c.907C>T; c.919del; c.922C>T; c.934A>T; c.942+1G>A; c.942+1G>T; c.943-1G>A; c.943-1G>T; c.947T>C; c.949C>T; c.956C>T; c.959_960del; c.960del; c.971_972del; c.973del; c.976del; c.983dup; c.983_988del; c.994_995del; c.996G>A; c.997A>T; c.1007T>A; c.1014C>A; c.1014C>G; c.1045G>A; c.1048G>A; c.1049G>A; c.1071+1G>A; c.1072dup; c.1072_1079del; c.1072-1G>A; c.1076T>C; c.1078C>T; c.1079G>C; c.1079G>A; c.1084G>A; c.1093_1094del; c.1102G>A; c.1110_1111delCG; c.1120G>C; c.1125_1126del; c.1129_1130dup; c.1148C>A; c.1151T>C; c.1153C>T; c.1161C>A.</p>
<p>HADHA NM_000182.5 (228)</p>	<p>c.2229_2233dup; c.2231dup; c.2225_2228dup; c.2227_2228del; c.2222del; c.2220T>A; c.2208T>A; c.2200A>T; c.2198T>C; c.2193_2194dup; c.2183_2190del; c.2167_2168dup; c.2146+2T>C; c.2146+1G>A; c.2134_2138dup; c.2132dup; c.2090_2115del; c.2114T>A; c.2107G>A; c.2063_2087dup; c.2081del; c.2064_2081delinsTGGCCTCTGA; c.2077del; c.2071G>T; c.2059del; c.2039_2040dup; c.2026del; c.2027G>A; c.2026C>T; c.2020dup; c.2020C>T; c.2018del; c.2011del; c.2010del; c.2000+1G>C; c.2000+1G>A; c.1981_1999del; c.1990_1993dup; c.1967del; c.1959dup; c.1944_1945del; c.1937_1940del; c.1921del; c.1916_1919dup; c.1915_1918del; c.1918C>T; c.1917T>A; c.1893del; c.1891A>T; c.1889del; c.1885G>A; c.1844dup; c.1844del; c.1828C>G; c.1814_1815del; c.1811del; c.1793_1794del;</p>

	<p>c.1759_1760del; c.1720_1721del; c.1712T>C; c.1690-17_1690-2del; c.1690-2A>G; c.1689+2T>G; c.1689+2T>C; c.1689+1G>T; c.1678C>T; c.1668_1674dup; c.1653_1668del; c.1654dup; c.1654G>C; c.1644del; c.1645A>G; c.1633_1636delinsGGAAGTCTTGG; c.1621-2A>G; c.1620+2_1620+6del; c.1620+1del; c.1590del; c.1554del; c.1549del; c.1540del; c.1533dup; c.1528G>C; c.1528G>T; c.1523del; c.1519_1520insCTTT; c.1513A>T; c.1491_1495dup; c.1493A>G; c.1480-1G>C; c.1479+1G>T; c.1432del; c.1427_1428del; c.1422dup; c.1418C>A; c.1361_1364dup; c.1351del; c.1344_1345del; c.1336G>T; c.1319dup; c.1261G>T; c.1249del; c.1237A>T; c.1235_1236del; c.1231_1235del; c.1222_1225del; c.1221-1G>C; c.1221-2A>G; c.1220+1G>T; c.1209del; c.1204C>T; c.1202del; c.1195C>T; c.1183dup; c.1167dup; c.1168A>T; c.1165del; c.1152del; c.1132C>T; c.1091_1128del; c.1096_1097dup; c.1086-3_1092del; c.1086-1_1088dup; c.1086-2A>T; c.1070del; c.1059del; c.1052del; c.1036C>T; c.1030del; c.1029C>A; c.1025T>C; c.1002del; c.976-1G>T; c.976-2A>G; c.966_967del; c.960T>A; c.959del; c.944_956del; c.955G>A; c.930_931del; c.919-1G>A; c.919-2A>G; c.918+2T>A; c.914T>A; c.914T>C; c.893del; c.878dup; c.874A>T; c.871C>T; c.862G>T; c.859G>T; c.844_845insA; c.841C>T; c.815C>A; c.800-1_801del; c.800-1G>T; c.799+1G>A; c.761_764del; c.761del; c.721del; c.717C>A; c.703_704dup; c.703C>T; c.690del; c.677-1G>A; c.677-2A>G; c.676+2T>C; c.676+1G>A; c.667G>T; c.658C>T; c.640A>T; c.637_638del; c.612dup; c.574-2A>G; c.573+2T>C; c.573+1G>A; c.566_567delinsA; c.556C>T; c.539C>T; c.531_532insT; c.515del; c.499del; c.488_489del; c.479_482delinsAATA; c.474C>A; c.469C>T; c.453+1G>A; c.451G>T; c.446G>T; c.439G>T; c.407C>G; c.403A>G; c.387_388del; c.370C>T; c.359C>A; c.352_353del; c.340C>T; c.317delinsAGG; c.315-1G>A; c.315-2A>T; c.314+2T>G; c.314+1G>C; c.310dup; c.302G>T; c.294del; c.284_288del; c.274_278del; c.278C>G; c.240G>A; c.238del; c.191dup; c.181-1G>A; c.180_180+5delinsAT; c.180+3A>G; c.180+1G>A; c.160del; c.156_157del; c.157C>T; c.129dup; c.110-1G>A; c.72del; c.70dup; c.68del; c.67+1G>A; c.64C>T; c.28del; c.6_24dup; c.3G>A; c.1A>G.</p>
<p>CBS NM_000071.3 (282)</p>	<p>c.1616T>C; c.1603_1604del; c.1582C>T; c.1576C>T; c.1566del; c.1561del; c.1553-1G>C; c.1553-2A>C; c.1552+1G>A; c.1549del; c.1545del; c.1543C>T; c.1498_1499del; c.1468-1G>A; c.1467+1G>A; c.1456C>T; c.1397C>A; c.1391dup; c.1367T>C; c.1361T>A; c.1359-1G>C; c.1359-2A>C; c.1359-2A>T; c.1358+2T>A; c.1358+2T>C; c.1358+1G>A; c.1357G>A; c.1341del; c.1330G>A; c.1321A>T; c.1318G>T; c.1306G>T; c.1288_1289del; c.1280C>T; c.1259C>A; c.1227_1240del; c.1229G>A; c.1227G>A; c.1226G>A; c.1224-2A>C; c.1219_1223+8del; c.1223+1G>T; c.1223G>A; c.1221del; c.1218del; c.1210G>T; c.1179del; c.1169G>A; c.1152G>C; c.1150A>G; c.1145+1G>A; c.1145+1G>C; c.1136G>A; c.1135C>G; c.1135C>T; c.1126G>A; c.1111G>C; c.1111G>A; c.1109G>A; c.1087del; c.1081G>A; c.1075A>T; c.1064C>T; c.1058C>T; c.1056C>G; c.1051G>C; c.1047T>G; c.1046G>C; c.1045A>G; c.1042G>A; c.1040-2A>G; c.1039+5G>C; c.1039+2T>G; c.1039+2T>C; c.1039+1G>A; c.1039+1G>T; c.1039G>A; c.1036T>C; c.1013T>C; c.1009_1012del; c.1007G>T; c.1007G>C; c.1007G>A; c.1006C>A; c.1005_1006delinsTT; c.1006C>T; c.992C>A; c.992C>T; c.982G>A; c.978del; c.969G>A; c.959T>C; c.955-2A>C; c.955-2A>T; c.955-2A>G; c.954+2T>G; c.954+1G>A; c.949A>G; c.941T>C; c.920G>A; c.920G>C; c.920G>T; c.919G>A; c.913G>A; c.904G>A; c.903C>A; c.903C>G; c.892dup; c.889G>T; c.883C>T; c.869C>A; c.869C>T; c.862G>C; c.862G>A; c.837_841dup; c.836G>T; c.833T>G; c.833T>C; c.829-1G>T; c.829-1G>C; c.808_828+5del; c.828+1G>A; c.825C>A; c.815_816del; c.816T>A; c.808_810del; c.797G>A; c.796A>G; c.785C>G; c.785C>T; c.784A>G; c.776G>A; c.775G>A; c.770C>T; c.740_769del; c.767G>T; c.766G>A; c.754del; c.752T>C; c.738del; c.737-1G>C; c.736_736+8del; c.736+2T>C; c.736+2T>G; c.736+1G>A; c.732_733del; c.727C>T; c.715G>T; c.707_708delinsGGT; c.702del; c.700G>A; c.694C>G; c.689del; c.689T>A; c.685C>A; c.684C>G; c.684C>A; c.683A>G; c.677C>A; c.676G>T; c.676G>A; c.667-1G>C; c.667-14_667-7del; c.650C>T; c.637G>T; c.624G>A; c.606dup; c.580_599del; c.588del; c.572C>A; c.572C>T; c.556dup; c.536_553del; c.532-1G>A; c.532-2A>G; c.531+1G>A; c.531+1G>T; c.529A>T; c.528G>C; c.527A>G; c.526G>T; c.526G>A; c.526G>C; c.503_504del; c.502G>A; c.494G>A; c.493T>G; c.481del; c.478G>A; c.456_477del; c.476_477insCAGGGCCC; c.473C>T; c.472G>A; c.467del; c.463G>A; c.457G>A; c.456C>G; c.452-1G>A; c.452-2A>C; c.451+1G>T; c.451G>A; c.444dup; c.443G>C; c.442G>A; c.437del; c.434C>G; c.434C>T; c.430G>T; c.430G>A; c.430G>C; c.429C>G; c.427dup; c.415G>A; c.407del; c.407T>C; c.403_404delinsG; c.402del; c.393G>C; c.382G>T; c.379del; c.371_374dup; c.374G>A; c.373C>T; c.359_368del; c.369C>A; c.353_362del; c.362G>T; c.362G>A; c.361C>G; c.361C>T; c.359A>G; c.346G>A; c.341C>T; c.340G>T; c.340G>A; c.326G>A; c.325T>C; c.323del; c.317-2A>G; c.316+2_316+3del; c.316+1G>A; c.302T>C; c.301C>G; c.284T>A; c.284T>C; c.263C>G; c.262C>T; c.262C>G; c.258dup; c.256del; c.253G>A; c.233C>G; c.218_230del; c.210-1G>C; c.210-2A>C; c.209+1_209+8del; c.208_209+8del; c.209+2T>C; c.209+1del; c.209+1G>T; c.209dup; c.209+1G>A; c.209+1G>C; c.194A>T; c.194A>G; c.184dup; c.153_165del; c.162G>A; c.156C>A; c.154T>A; c.146C>T; c.140A>C; c.139G>A; c.118del; c.98del; c.95delinsATAAGACAGT; c.78dup; c.18_36del; c.28del; c.19dup; c.19del; c.2T>C.</p>
<p>ARSA NM_000487.6 (360)</p>	<p>c.1492dup; c.1489_1492dup; c.1471T>G; c.1470C>G; c.1468T>C; c.1462C>T; c.1451del; c.1447G>T; c.1428dup; c.1408_1418del; c.1399C>T; c.1388del; c.1378C>T; c.1366C>T; c.1344dup; c.1337del; c.1330dup; c.1323del; c.1314_1315del; c.1293T>A; c.1292A>C; c.1289T>C; c.1288C>T; c.1282_1283dup; c.1283C>T; c.1282C>G; c.1279C>T; c.1279C>A; c.1278del; c.1276G>A; c.1274A>G; c.1269del; c.1264del; c.1264del; c.1238A>G; c.1222_1232del; c.1232C>T; c.1223_1231del; c.1228_1229del; c.1228dup; c.1229C>T; c.1227_1228del; c.1223G>A; c.1223G>C; c.1211-2A>G; c.1210+1G>T; c.1210+1G>A; c.1201_1203del; c.1200C>A; c.1200C>T; c.1199T>C; c.1197C>G; c.1196A>G; c.1195C>T; c.1187A>C; c.1177_1178delinsGG; c.1175G>A; c.1174C>T; c.1174C>G; c.1162G>T; c.1159G>T; c.1156C>T; c.1150G>A; c.1129_1136dup; c.1136del; c.1136C>A; c.1136C>T; c.1130_1132del; c.1128_1129dup; c.1123_1126del; c.1125_1126del; c.1115G>A; c.1114C>T; c.1108-1G>C; c.1108-2A>G; c.1108-3C>G; c.1107+1del; c.1107+1G>C; c.1107+1G>A; c.1107+1G>T; c.1087dup; c.1079T>C; c.1053del; c.1046del; c.1036del; c.1030del; c.1024del; c.1010A>T; c.1007T>C; c.992A>G; c.991G>A; c.991G>T; c.986C>T; c.980-2A>C; c.979_979+3del; c.979+1G>A; c.979G>A; c.979G>T; c.970A>T; c.960G>T; c.960G>A; c.959G>A; c.956_958del; c.958T>G; c.955_956insGCCTTGAT; c.946G>A; c.942G>T; c.937dup; c.938G>A; c.937C>G; c.937C>T; c.931G>A; c.931G>T; c.931G>C; c.929del; c.929G>A; c.929G>T; c.928G>T; c.928G>A; c.926A>G; c.925dup; c.926A>G; c.925G>T; c.925G>A; c.922T>C; c.917C>T; c.916A>G; c.908G>T; c.907G>A; c.905G>A; c.905G>T; c.901C>T; c.901C>G; c.899T>G; c.899T>C; c.891_895del; c.893G>T; c.892G>A; c.892G>T; c.891del; c.890C>A; c.887G>A; c.884G>A; c.883G>A; c.877C>T; c.868_873del; c.869G>C; c.869G>A; c.868C>G; c.868C>T; c.863C>A; c.862A>C; c.855-1G>T; c.855-1G>A; c.854+1dup; c.854+1G>A; c.854+1G>T; c.852T>A; c.851A>G; c.849C>G; c.847G>T; c.842C>T; c.836_837delinsAA; c.827C>T; c.827C>A; c.826A>T; c.809T>C; c.788del; c.774del; c.771T>A; c.771T>G; c.770A>T; c.769G>A; c.769G>C; c.765G>C; c.764A>C; c.763G>A; c.758dup; c.755C>A; c.746T>G; c.746T>C; c.737_744dup; c.739G>A; c.737G>T; c.737G>A; c.736C>T; c.730_731del; c.731C>A; c.718_727del; c.724G>T; c.712C>T; c.701A>G; c.700C>G; c.700C>T; c.697C>A; c.685-1G>A; c.685-2A>G; c.684+1G>A; c.680C>T; c.679T>C; c.677C>T; c.677C>A; c.676G>T; c.674_675dup; c.674A>G; c.662T>C; c.645_660dup; c.641C>T; c.640G>A; c.640G>C; c.632T>C; c.629T>G; c.629T>C; c.622del; c.620C>A; c.617dup; c.609_610del; c.608A>G; c.607T>A; c.607T>C; c.607C>G; c.586del; c.584G>A; c.583del; c.582dup; c.582del; c.581C>G; c.581C>A; c.580C>T; c.578C>G; c.577C>A; c.574C>T; c.572_573insTGTCTCTTATA; c.545del; c.545C>G; c.544C>A; c.542dup; c.542T>G; c.536del; c.526C>T; c.514G>T; c.495_501del; c.494dup; c.488G>C; c.466-4_482del;</p>

	<p>c.475C>T; c.474C>A; c.470C>T; c.470C>G; c.467delG; c.467G>A; c.466-2A>G; c.465+2T>A; c.465+1G>A; c.465G>A; c.460G>C; c.460G>A; c.460G>T; c.451dup; c.449C>G; c.449C>T; c.448C>T; c.442G>A; c.436T>C; c.434G>A; c.433C>T; c.433C>G; c.427T>C; c.393_425del; c.422del; c.421C>G; c.418del; c.418dup; c.418C>G; c.413C>T; c.413C>G; c.413C>A; c.412C>G; c.412C>A; c.412C>T; c.410T>C; c.397_406del; c.392G>A; c.386G>A; c.385G>A; c.378G>A; c.369_374delinsAACCTTGGG; c.374A>C; c.371G>A; c.370G>A; c.370G>T; c.365T>C; c.349_350dup; c.346C>T; c.338T>C; c.335T>A; c.311del; c.304del; c.302_303delinsTT; c.302dup; c.302del; c.302G>T; c.302G>A; c.301G>T; c.295dup; c.292_293delinsCT; c.293C>T; c.272del; c.270C>A; c.263del; c.263G>A; c.257G>T; c.257G>A; c.256C>G; c.256C>T; c.251C>T; c.251C>G; c.248T>C; c.245G>T; c.245G>A; c.244del; c.244C>T; c.240dup; c.229G>C; c.227_228insTA; c.225-1G>C; c.225-2A>G; c.224+2T>A; c.224+1G>T; c.224+1G>A; c.211_212del; c.208_209del; c.206_209del; c.209T>C; c.200del; c.195del; c.195C>G; c.185_186dup; c.185_186del; c.179G>C; c.178C>T; c.157_164del; c.160C>G; c.157C>T; c.136T>C; c.135C>G; c.129_133del; c.133A>G; c.122A>G; c.116dup; c.109_116del; c.116G>A; c.110_111insACTGGGCTGCTATGGGCA; c.109G>C; c.98T>C; c.93C>G; c.61del; c.34del; c.13del; c.2T>C.</p>
<p>BTD NM_001370658.1 (306)</p>	<p>c.-148C>T; c.-24A>T; c.-17_-17+3del; c.-18del; c.-18A>T; c.-17+1del; c.-17+1G>A; c.-17+1G>C; c.-17+1G>T; c.-17+2T>C; c.-6del; c.38_44del; c.38_44delinsTCC; c.40_41del; c.41_44del; c.40G>A; c.44_45del; c.47dup; c.48C>G; c.52del; c.56_59dup; c.58_59del; c.59T>C; c.70_71del; c.76G>T; c.91_92del; c.100G>T; c.106_107del; c.111T>G; c.124G>A; c.124G>T; c.125T>C; c.130G>A; c.132G>C; c.142_145dup; c.143_146dup; c.167del; c.175C>T; c.177_178del; c.176G>A; c.185C>A; c.185C>T; c.197T>G; c.202C>T; c.203del; c.206del; c.218A>G; c.220G>T; c.223C>T; c.229G>A; c.238G>A; c.239C>T; c.248del; c.249+1G>A; c.249+1G>T; c.249+2C>G; c.250-15del; c.250-1G>T; c.253_254dup; c.253G>A; c.256C>T; c.263_264dup; c.262A>G; c.266dup; c.265G>C; c.266T>G; c.270del; c.274G>C; c.281G>T; c.282del; c.304A>G; c.312_315dup; c.320C>T; c.322T>G; c.333del; c.346del; c.346C>T; c.347dup; c.359G>A; c.360G>A; c.364C>T; c.364C>A; c.368G>T; c.382C>T; c.383G>A; c.385T>C; c.394A>C; c.395C>G; c.399G>A; c.399+2T>G; c.400-2A>G; c.406C>T; c.409C>T; c.410G>A; c.416G>A; c.419G>C; c.419G>A; c.419G>T; c.424G>C; c.425C>T; c.430_431del; c.430del; c.451G>A; c.4458T>G; c.466A>T; c.468G>T; c.477_478delinsAA; c.484del; c.496T>C; c.496T>G; c.497G>A; c.500del; c.499C>T; c.500C>T; c.516C>A; c.522C>G; c.523A>G; c.524A>G; c.527del; c.527C>G; c.528_542del; c.534_536del; c.534del; c.535G>A; c.545A>T; c.565C>T; c.566G>A; c.569A>G; c.571del; c.571C>T; c.572G>A; c.577del; c.581A>G; c.583C>T; c.586T>A; c.588C>G; c.589_591delinsAGTA; c.592G>C; c.594G>C; c.604G>A; c.604G>C; c.622G>T; c.623A>G; c.633del; c.635T>C; c.640A>G; c.641C>T; c.649G>A; c.671C>T; c.674G>A; c.675C>A; c.683dup; c.682A>C; c.683T>C; c.683A>G; c.686del; c.695A>G; c.697C>T; c.698C>T; c.704T>C; c.705C>G; c.710T>A; c.723C>G; c.734A>T; c.743dup; c.751G>T; c.754T>G; c.755G>A; c.772C>G; c.776T>A; c.776T>G; c.798del; c.799del; c.805G>C; c.806C>T; c.827T>G; c.835G>C; c.836C>T; c.838A>C; c.869G>A; c.873del; c.875G>T; c.875G>A; c.881A>T; c.887del; c.896C>T; c.901dup; c.906C>G; c.908A>G; c.920dup; c.932_941del; c.941_942del; c.946C>T; c.950del; c.989del; c.992del; c.1000del; c.1021T>G; c.1025T>A; c.1030dup; c.1036_1037dup; c.1066C>T; c.1069G>T; c.1082_1097del; c.1092dup; c.1097G>A; c.1098G>A; c.1103dup; c.1108_1109insTGGG; c.1110_1111dup; c.1130_1155del; c.1131_1132del; c.1137G>A; c.1147T>G; c.1154T>C; c.1167_1181delinsTCCAATGGCC; c.1181_1192del; c.1178G>C; c.1179del; c.1189G>T; c.1192T>C; c.1193G>C; c.1204dup; c.1207del; c.1207T>C; c.1208G>C; c.1211G>A; c.1211G>C; c.1215T>G; c.1224C>A; c.1247_1248del; c.1246G>A; c.1252T>C; c.1253A>G; c.1254T>A; c.1256C>A; c.1264del; c.1259T>C; c.1268T>C; c.1270G>T; c.1270G>C; c.1271A>G; c.1273G>A; c.1274G>A; c.1274G>T; c.1279C>T; c.1280A>G; c.1292_1293del; c.1292G>A; c.1298dup; c.1300T>G; c.1301A>G; c.1308A>C; c.1309G>A; c.1312dup; c.1324del; c.1334dup; c.1334del; c.1350dup; c.1352G>T; c.1372G>C; c.1372G>A; c.1386dup; c.1387G>T; c.1396_1415del; c.1395C>G; c.1398del; c.1399del; c.1399T>C; c.1400G>A; c.1401G>A; c.1403G>A; c.1406A>G; c.1406A>C; c.1415C>T; c.1419del; c.1429C>T; c.1433dup; c.1448_1452del; c.1451T>A; c.1465C>T; c.1466C>G; c.1495dup; c.1497T>G; c.1535C>T; c.1541C>T; c.1543C>G; c.1543C>T; c.1547_1548del; c.1550G>A; c.1552C>A; c.1552C>T; c.1553G>A; c.1553G>T; c.1556dup; c.1559A>G; c.1567G>C; c.1568A>G; c.1568A>T; c.1569C>A; c.*159G>A; c.3G>A; c.1167G>A; c.1333G>T; c.1503del.</p>
<p>SLC26A2 NM_000112.4 (229)</p>	<p>c.-26+1G>A; c.-26+2T>C; c.2T>C; c.3G>T; c.15_19delTAAAG; c.47C>G; c.55G>T; c.58_62dup; c.63_64delCA; c.78_88dup; c.69del; c.100del; c.104C>G; c.110del; c.111del; c.117delC; c.118A>T; c.121C>T; c.134del; c.136_137insTT; c.138dupT; c.139C>T; c.145delA; c.165_166insCT; c.166delAinsCTC; c.185C>G; c.188delA; c.207delT; c.213_219del; c.218del; c.214A>T; c.223del; c.223C>T; c.239_243dupGCAGT; c.235C>T; c.241del; c.255delC; c.272_276delinsATTT; c.277del; c.293G>A; c.294G>A; c.299delC; c.306C>G; c.309_327del; c.314del; c.325_326delTT; c.331G>T; c.347del; c.373C>T; c.374A>T; c.387T>G; c.391delC; c.399_402dup; c.395T>C; c.398C>T; c.426C>A; c.438delT; c.438dupT; c.437_438del; c.451delT; c.469T>C; c.485_486delTG; c.484G>T; c.496G>A; c.499delG; c.532C>T; c.541C>T; c.567dup; c.578_581delCCTT; c.611T>G; c.613_617del; c.638del; c.642_643delAA; c.695dup; c.697C>T; c.699+2T>C; c.700-2A>G; c.700-1G>C; c.705_711delGATGGGC; c.736_739delGTCT; c.746C>G; c.746C>A; c.749A>T; c.758T>A; c.764G>A; c.767T>C; c.776G>T; c.796dup; c.807dup; c.819delT; c.822del; c.835C>T; c.840dup; c.854C>A; c.870delG; c.874_875dup; c.892del; c.906_907delCT; c.909T>A; c.916_929dup; c.918delC; c.922delA; c.925delC; c.931T>C; c.961del; c.1020_1022delTGT; c.1034T>A; c.1060G>T; c.1064_1065insAAAAA; c.1114A>T; c.1142dup; c.1147_1150del; c.1155del; c.1157C>T; c.1157C>G; c.1217_1223del; c.1240_1244del; c.1242_1245delAAAC; c.1246C>T; c.1272dup; c.1273A>G; c.1277T>C; c.1277T>A; c.1283del; c.1303dupA; c.1306del; c.1311dupT; c.1338delA; c.1336A>T; c.1343C>G; c.1343C>A; c.1361A>C; c.1375dup; c.1375_1377dup; c.1382C>T; c.1394delT; c.1393_1394del; c.1397dup; c.1421delT; c.1428T>G; c.1432delC; c.1434del; c.1441delA; c.1441dup; c.1448T>C; c.1451G>A; c.1483dup; c.1487_1488insGGCG; c.1509_1514del; c.1513T>C; c.1514G>A; c.1535_1536del; c.1535C>A; c.1537_1541dupGTTAT; c.1544G>A; c.1565C>T; c.1592del; c.1596delT; c.1607_1610delGTTT; c.1621del; c.1625_1650delinsAACACCA; c.1630del; c.1639C>T; c.1649delA; c.1650delG; c.1655C>A; c.1691_1698dup; c.1707C>G; c.1714delC; c.1720del; c.1724delA; c.1734del; c.1764C>A; c.1767C>A; c.1773_1776del; c.1772del; c.1777G>T; c.1796T>A; c.1805_1808del; c.1800C>G; c.1800C>A; c.1806_1809delAACT; c.1810_1811del; c.1817delC; c.1831delG; c.1847del; c.1859delA; c.1878delG; c.1926delT; c.1931del; c.1950delG; c.1955_1958delACTG; c.1956_1957dupCT; c.1957T>A; c.1957T>G; c.1958G>A; c.1968del; c.1976delT; c.1982delC; c.1983delA; c.1987G>A; c.1998_1999delAC; c.1994dup; c.1994A>C; c.2004_2007del; c.2017_2018delGA; c.2018A>T; c.2032_2033delinsCT; c.2033G>T; c.2050del; c.2053C>T; c.2056del; c.2057_2061delinsAGAA; c.2065_2066del; c.2065_2068delinsTC; c.2069_2070delTG; c.2085del; c.2087dup; c.2088_2089delinsGT; c.2089G>T; c.2095del; c.2097T>A; c.2105dup; c.2109del; c.2120_2121delTTT; c.2124_2125dupCT; c.2125_2126del; c.2143G>A; c.2144C>T.</p>

CFTR
NM_000492.4
(1539)

c.-9_14delCGAGAGACCATGCGAGGTCGCC; c.1A>G; c.2T>A; c.2T>C; c.2T>G; c.3G>A; c.4delC; c.4C>T; c.9del; c.11C>A; c.13C>T; c.14C>G; c.14C>T; c.15del; c.19G>T; c.25dupG; c.38C>T; c.40_44del; c.42del; c.40A>T; c.43del; c.49_50dupTT; c.50delT; c.50dupT; c.49_50delTT; c.44T>C; c.44T>A; c.51delC; c.53+1G>C; c.53+1G>T; c.53+2T>C; c.52+2T>G; c.54-2A>G; c.54-1G>A; c.54-1G>T; c.56G>A; c.57G>A; c.57G>T; c.71del; c.71T>A; c.79G>A; c.79G>T; c.87dup; c.88C>T; c.89dup; c.97G>T; c.100_117del; c.112_113del; c.114C>G; c.115C>T; c.131A>G; c.137C>A; c.142_145del; c.147_150delATCT; c.148T>C; c.149C>A; c.156dup; c.156_163dup; c.156del; c.154A>T; c.164+1G>A; c.164+1G>C; c.164+1G>T; c.164+2dup; c.164+2T>C; c.164+2T>G; c.164+2T>A; c.164+4dup; c.164+3_164+4insT; c.165-2delA; c.165-2A>C; c.165-2A>G; c.165-1G>A; c.166G>A; c.166G>T; c.168delA; c.169T>G; c.169T>C; c.170G>A; c.171G>A; c.174_177delTAGA; c.175dupA; c.179_180delAG; c.177dup; c.178G>T; c.178G>A; c.185dup; c.198del; c.200C>T; c.202A>T; c.206T>G; c.217delC; c.220C>T; c.221G>C; c.223C>T; c.233delT; c.233dupT; c.234dupC; c.234del; c.236G>A; c.243delT; c.253G>T; c.253G>C; c.254G>A; c.254G>T; c.262_263delTT; c.264_268del; c.263T>A; c.263T>C; c.263T>G; c.269T>A; c.270del; c.271G>A; c.273G>C; c.273+1G>A; c.273+1G>C; c.273+2T>G; c.273+2T>C; c.273+3A>C; c.274-5_274-2delTGTA; c.274-2A>C; c.274-2A>G; c.274del; c.274-1G>A; c.274-1G>C; c.274-1G>T; c.274G>A; c.274G>T; c.276A>T; c.285dup; c.283A>T; c.292C>T; c.293A>G; c.300_301del; c.296C>T; c.296C>G; c.303dupA; c.302T>G; c.305T>G; c.305T>C; c.308del; c.307G>T; c.310delA; c.313delA; c.314T>A; c.319_326delGCTTCCTA; c.320C>G; c.323C>T; c.326_327del; c.325_327delTATinsG; c.325T>C; c.325T>A; c.326A>G; c.327T>A; c.327T>G; c.328G>C; c.328G>T; c.329A>T; c.330C>A; c.330C>G; c.331C>G; c.332del; c.332C>T; c.340A>T; c.346G>A; c.349C>G; c.349C>T; c.350G>A; c.350G>C; c.350G>T; c.354dup; c.357del; c.358G>A; c.366T>A; c.367dup; c.370G>A; c.376G>A; c.377G>A; c.380_382dupTAT; c.387del; c.393del; c.394_398del; c.396del; c.405_406dupAC; c.409_412delCTCC; c.409delC; c.410_416delTCTACA; c.410T>C; c.413_415dupTAC; c.416A>G; c.416A>T; c.422C>A; c.424delA; c.429delT; c.433delC; c.433C>G; c.434T>A; c.441_442del; c.442delA; c.443T>A; c.445G>A; c.445G>T; c.450dup; c.449T>A; c.451C>T; c.454A>G; c.459_476delAATAGCTATGTTTAGTTT; c.458G>A; c.461dup; c.470_483delTTAGTTTGATTTAT; c.476dup; c.473G>A; c.476T>C; c.476T>A; c.480_483del; c.481T>G; c.482del; c.482A>G; c.483_484insCC; c.484A>G; c.488delA; c.487A>G; c.488A>T; c.489+1G>A; c.489+1G>T; c.489+2T>C; c.489+2T>G; c.489+3A>G; c.490-3T>G; c.490-2A>C; c.490-2A>G; c.490-1G>A; c.490-1G>T; c.490-1G>C; c.494del; c.494T>C; c.498del; c.502_503del; c.503C>G; c.506dupG; c.509G>A; c.522_526del; c.520_530del; c.523A>G; c.531delT; c.531dupT; c.532G>A; c.532G>C; c.534_538del; c.533G>A; c.535C>T; c.543_546delTAGT; c.543_556del; c.544dupA; c.548del; c.551_555del; c.550del; c.553dup; c.558del; c.560delA; c.558C>G; c.567C>A; c.571T>G; c.574G>T; c.578_579+5del; c.575A>G; c.577G>A; c.577G>T; c.579_579+3del; c.579+1G>A; c.579+1G>T; c.579+1_579+2insACAT; c.579+3A>G; c.579+5G>A; c.580-2A>C; c.580-2A>G; c.580-1G>T; c.580-1G>C; c.580G>A; c.580G>T; c.581_582del; c.581G>T; c.586dupG; c.592G>A; c.592G>C; c.595C>T; c.596A>G; c.598T>A; c.601delG; c.604_605del; c.606G>A; c.614dup; c.613C>T; c.613C>A; c.617del; c.614C>T; c.614C>G; c.617T>G; c.618dup; c.617T>A; c.619C>T; c.647G>A; c.653T>A; c.657dupA; c.658C>T; c.662del; c.671delT; c.675T>A; c.680T>G; c.695T>A; c.708delT; c.708dup; c.709C>G; c.717delG; c.723_743+1delGAGAATGATGAGTACAGG; c.719T>G; c.739_742dupTACA; c.741C>A; c.742A>G; c.742A>T; c.743G>C; c.743+1G>A; c.743+1G>C; c.743+2T>C; c.744-14_744-3del; c.744-9_744-2delinsTACG; c.744-2A>G; c.748C>T; c.751A>T; c.755delCinsGCTGGGAAGAT; c.761del; c.764del; c.769G>T; c.772del; c.772A>G; c.773delG; c.777del; c.794T>G; c.803delA; c.805_806delAT; c.808C>T; c.810del; c.811delT; c.817A>T; c.825C>G; c.828C>A; c.830G>A; c.831G>A; c.835G>T; c.850dupA; c.850del; c.844G>T; c.848_860delAAATGATGAAAAinsTG; c.860dupA; c.861_865del; c.859A>T; c.863T>G; c.865_869delAGACA; c.866_870del; c.865A>T; c.868C>T; c.869+1_869+3del; c.869+1G>C; c.869+1G>T; c.869+3A>T; c.869+5G>A; c.870-1113_870-1110del; c.870-12T>A; c.870-2A>G; c.870-1G>C; c.874_875delGA; c.874G>T; c.888_889dupTC; c.892A>T; c.902dup; c.905_906insA; c.908_911del; c.912C>G; c.915_916del; c.922_926del; c.927del; c.926C>A; c.935_937delTCT; c.935_937del; c.926C>G; c.930delC; c.931T>C; c.933C>G; c.933C>A; c.941G>A; c.941G>T; c.948delT; c.948dup; c.950T>A; c.959delT; c.959T>A; c.960dup; c.962del; c.972del; c.980del; c.982del; c.987delA; c.989dup; c.988G>T; c.1000C>T; c.1001G>A; c.1001G>T; c.1008_1019dupATTCACCACCAT; c.1006_1007insG; c.1007T>A; c.1012_1013del; c.1013C>T; c.1021_1022dupTC; c.1021T>C; c.1025_1026insTA; c.1029delC; c.1037T>C; c.1039C>T; c.1040G>A; c.1040G>C; c.1040G>T; c.1045G>C; c.1053_1054del; c.1052C>T; c.1054C>T; c.1055G>A; c.1057C>T; c.1066_1071del; c.1067G>A; c.1069del; c.1068G>A; c.1075C>A; c.1075_1079delinsAAAAA; c.1078del; c.1076A>G; c.1079C>A; c.1080del; c.1081delT; c.1081T>C; c.1083delG; c.1082G>A; c.1084_1088dupTATGA; c.1083G>A; c.1086T>A; c.1086T>G; c.1093_1094delCT; c.1090T>C; c.1097del; c.1115delA; c.1114C>T; c.1116G>T; c.1116_1116+1insATCAA; c.1116+1G>A; c.1116+1G>T; c.1116+1G>C; c.1116+2T>C; c.1116+2T>A; c.1117-5A>G; c.1117-2A>G; c.1117-1G>A; c.1117-1G>T; c.1117G>A; c.1118dup; c.1125A>C; c.1126C>T; c.1130dupA; c.1130del; c.1132C>T; c.1135G>T; c.1140T>A; c.1141A>T; c.1150G>T; c.1155_1156dupTA; c.1157dup; c.1155T>A; c.1159_1160del; c.1162_1168delACGACTA; c.1162_1163delinsTA; c.1171G>T; c.1175T>G; c.1177del; c.1189del; c.1190dup; c.1197delC; c.1202G>A; c.1203G>A; c.1209G>C; c.1209G>T; c.1209+1G>A; c.1209+1G>T; c.1209+1G>C; c.1209+2T>G; c.1209+2T>A; c.1210-2_1210-1delAG; c.1210-3C>G; c.1210-2A>C; c.1211delG; c.1210-1G>C; c.1210-1G>T; c.1219G>T; c.1221del; c.1224_1227del; c.1227_1228del; c.1234_1238delGCAAAA; c.1234del; c.1235delC; c.1243_1247del; c.1239dupA; c.1240C>T; c.1244del; c.1248dupT; c.1261dup; c.1287del; c.1290del; c.1301_1307delCACTTCT; c.1301C>A; c.1301C>G; c.1302del; c.1313del; c.1319_1323del; c.1315C>T; c.1329_1350del; c.1322T>C; c.1324A>T; c.1327_1330dupGATA; c.1327G>T; c.1330_1331delAT; c.1333_1336del; c.1331T>C; c.1334del; c.1340delA; c.1343_1350del; c.1347_1350delAAGA; c.1344_1345delinsTT; c.1351G>T; c.1352G>T; c.1358T>C; c.1360_1387del; c.1364C>A; c.1365_1366delGG; c.1366del; c.1364C>T; c.1366G>T; c.1368del; c.1367T>C; c.1375_1383delTCCACTGGA; c.1369G>C; c.1373delG; c.1373G>T; c.1373G>A; c.1375_1385del; c.1390A>G; c.1391A>C; c.1392G>T; c.1392+1del; c.1392+1G>A; c.1393-2A>G; c.1393-1G>A; c.1393C>A; c.1397C>G; c.1397C>T; c.1398A>T; c.1399C>T; c.1400T>C; c.1405A>G; c.1409_1418delTGATTATGGG; c.1409T>G; c.1411A>T; c.1411A>G; c.1415dupT; c.1418delG; c.1420G>A; c.1420G>T; c.1423del; c.1424del; c.1428G>T; c.1435G>C; c.1435G>T; c.1439del; c.1438G>T; c.1438G>A; c.1439G>A; c.1446dup; c.1447A>G; c.1447A>T; c.1465_1469del; c.1464_1476del; c.1466C>A; c.1469delT; c.1468_1471del; c.1470_1471del; c.1472_1482del; c.1471T>C; c.1475C>T; c.1477_1478delCA; c.1477C>T; c.1478A>G; c.1478A>T; c.1483del; c.1486delT; c.1487G>A; c.1488G>A; c.1493_1507del; c.1490T>A; c.1499G>A; c.1502C>T; c.1505T>A; c.1505T>C; c.1505T>G; c.1508A>G; c.1510G>T; c.1519_1521delATC; c.1519_1521del; c.1516A>C; c.1517T>A; c.1517T>G; c.1517T>C; c.1518C>G; c.1519A>T; c.1521_1523delCTT; c.1523T>G; c.1525G>C; c.1526G>T; c.1530_1531delTT; c.1529T>G; c.1534T>C; c.1538A>G; c.1543_1555delTATAGATACAGAA; c.1540G>T; c.1545_1546delTA; c.1547_1550del; c.1547_1548del; c.1546A>G; c.1546_1548delAGAinsT; c.1550A>G; c.1551C>A; c.1551C>G; c.1558G>A; c.1558G>T; c.1559T>A; c.1559T>C; c.1562T>C; c.1570T>C; c.1573del; c.1571G>A; c.1572C>A; c.1579_1584+11del; c.1573C>T; c.1574A>C; c.1579G>T; c.1580A>G; c.1582G>T; c.1584+1G>A; c.1584+1G>T; c.1584+2T>C; c.1585-9412A>G; c.1585-8G>A; c.1585-2A>G; c.1585-2A>T; c.1585-1G>A; c.1585-1G>T; c.1589T>A; c.1592_1593delinsG; c.1609G>A; c.1616_1617dupTA; c.1615del; c.1616_1617insTT; c.1622T>C; c.1625del; c.1624G>T; c.1642_1643delCT; c.1645A>C; c.1646G>A; c.1646G>T; c.1647T>A; c.1647T>G; c.1648G>T; c.1650delA; c.1652delG; c.1651G>A; c.1652G>A;

<p>c.1652G>T; c.1654C>A; c.1654C>T; c.1656delA; c.1657C>T; c.1663A>T; c.1670delC; c.1672_1673del; c.1673T>C; c.1674delA; c.1673T>A; c.1675G>A; c.1679G>A; c.1679G>C; c.1679+1G>A; c.1679+1G>C; c.1679+2T>C; c.1680-886A>G; c.1680-877T>G; c.1680-2A>C; c.1680-1G>A; c.1680-1G>C; c.1680A>C; c.1680A>T; c.1682dup; c.1682C>A; c.1682C>T; c.1684G>C; c.1687T>A; c.1687T>C; c.1687T>G; c.1688A>G; c.1692delA; c.1689C>A; c.1694A>G; c.1697C>A; c.1700A>C; c.1703delT; c.1703T>A; c.1705T>C; c.1705T>G; c.1706A>G; c.1709del; c.1707T>A; c.1709T>A; c.1713_1714delAG; c.1712T>C; c.1714G>A; c.1720C>T; c.1721C>A; c.1724T>A; c.1727G>T; c.1731C>A; c.1733_1734delTA; c.1733T>C; c.1735_1736del; c.1735G>C; c.1736A>G; c.1738del; c.1742dup; c.1752dup; c.1753G>T; c.1760T>C; c.1762G>T; c.1763A>T; c.1766G>A; c.1766G>T; c.1766+1G>A; c.1766+1G>C; c.1766+1G>T; c.1766+2del; c.1766+2T>A; c.1766+2T>C; c.1766+2T>G; c.1766+3A>G; c.1766+3A>T; c.1766+5G>T; c.1766+5G>C; c.1767-2A>G; c.1767-1G>A; c.1769G>C; c.1778_1779del; c.1781T>C; c.1786_1787del; c.1792_1798delAAAACTA; c.1792A>T; c.1800del; c.1801delA; c.1801A>T; c.1802T>C; c.1807del; c.1810_1826del; c.1811del; c.1811C>T; c.1822del; c.1825C>T; c.1826A>G; c.1826A>T; c.1837G>A; c.1841A>G; c.1846del; c.1846dup; c.1853T>C; c.1860T>G; c.1861G>T; c.1865G>A; c.1867dup; c.1867_1869delinsG; c.1871_1878del; c.1874_1875del; c.1884del; c.1882G>A; c.1882G>C; c.1883G>C; c.1891dup; c.1893_1897del; c.1904delA; c.1900C>T; c.1909C>T; c.1911delG; c.1920_1921dupTA; c.1923_1931delCTCAAACTinsA; c.1924_1931del; c.1937del; c.1936G>T; c.1941T>A; c.1943delA; c.1943A>T; c.1949del; c.1954C>T; c.1966G>T; c.1969A>T; c.1970del; c.1972dup; c.1973del; c.1973_1985delGAAATTCATCTinsAGAAA; c.1976del; c.1977_1986del; c.1979C>G; c.1981delA; c.1986_1989delAACT; c.1987_2000del; c.1990G>T; c.1993A>T; c.2000_2001del; c.1997T>A; c.2012delT; c.2012T>A; c.2017G>T; c.2019_2022dup; c.2036G>A; c.2044delA; c.2044dupA; c.2045delC; c.2045dupC; c.2051_2052delAA; c.2051_2052dupAA; c.2052delA; c.2052dupA; c.2049_2052del; c.2051_2052delAAinsG; c.2053dupC; c.2053C>T; c.2057_2058delinsA; c.2061_2062insTTTTTTTTTTTTTTTTTTTTTTT; c.2058_2061dup; c.2061del; c.2062A>T; c.2062_2066delinsTTTTTGTGTTG; c.2065C>T; c.2074G>T; c.2083dupG; c.2083del; c.2089delA; c.2089dupA; c.2086_2089del; c.2091del; c.2125C>T; c.2126G>A; c.2128A>T; c.2143C>T; c.2145_2146delAAinsGT; c.2146A>T; c.2148del; c.2156T>A; c.2158C>T; c.2161del; c.2175dupA; c.2184_2185insA; c.2188G>T; c.2195T>G; c.2203delA; c.2210C>T; c.2213T>G; c.2215delG; c.2233G>T; c.2241_2248delGATCATCG; c.2241del; c.2249_2256delCTCGCATC; c.2249C>T; c.2261del; c.2277del; c.2278dup; c.2283dup; c.2290C>T; c.2291delG; c.2299C>T; c.2301del; c.2303dup; c.2324_2325delAC; c.2327C>G; c.2335del; c.2335C>T; c.2337del; c.2341C>T; c.2349dup; c.2353C>T; c.2373delA; c.2374C>T; c.2379del; c.2380del; c.2382_2383del; c.2393dup; c.2395C>T; c.2410G>T; c.2423_2424dupAT; c.2417A>G; c.2425del; c.2426C>A; c.2428A>T; c.2429delG; c.2435dup; c.2438del; c.2440C>T; c.2443delG; c.2443G>T; c.2450delG; c.2453delT; c.2454_2455insT; c.2455G>T; c.2463_2464delITG; c.2464G>T; c.2464G>A; c.2467G>T; c.2472del; c.2475_2478dupCGAA; c.2476G>T; c.2479G>T; c.2483dupA; c.2489dup; c.2486T>A; c.2488A>T; c.2490+1G>A; c.2490+1G>T; c.2490+1G>C; c.2490+2T>C; c.2491-2A>C; c.2491-2A>G; c.2491G>T; c.2496C>A; c.2502delT; c.2502dupT; c.2508delT; c.2523_2548del; c.2537G>A; c.2538G>A; c.2547_2555del; c.2547C>A; c.2551C>T; c.2552G>T; c.2554dupT; c.2555del; c.2556T>G; c.2560dup; c.2562del; c.2573delG; c.2576_2588del; c.2583delT; c.2589_2599delAATTTGGTCTG; c.2592_2593del; c.2600T>A; c.2600T>G; c.2601dupA; c.2605dup; c.2605del; c.2611dup; c.2615del; c.2617G>T; c.2619+1G>A; c.2619+2T>C; c.2619+2T>A; c.2620-2A>G; c.2620-1G>T; c.2637_2644del; c.2639_2640dupTG; c.2644_2645dupTG; c.2645G>A; c.2646G>A; c.2657+1_2657+2insA; c.2657+5G>A; c.2658-2A>G; c.2658-1G>C; c.2658-1G>T; c.2668C>T; c.2672del; c.2679G>T; c.2700T>A; c.2705del; c.2732_2733insA; c.2733_2734insA; c.2735C>A; c.2737_2738insG; c.2738A>G; c.2739T>A; c.2739T>G; c.2742T>G; c.2743_2745delinsA; c.2755del; c.2757C>G; c.2757C>A; c.2758G>A; c.2758G>T; c.2763_2764dupAG; c.2768C>A; c.2777delT; c.2776_2777del; c.2780T>C; c.2797A>G; c.2803_2813del; c.2805_2810delACCACTinsTCAGA; c.2810dupT; c.2812dupG; c.2813T>G; c.2816del; c.2816A>G; c.2819_2823del; c.2816_2817delinsC; c.2818_2831del; c.2818dup; c.2818_2819del; c.2819_2820insA; c.2822delT; c.2824del; c.2825delT; c.2828del; c.2834C>T; c.2836A>T; c.2845C>T; c.2848del; c.2846A>T; c.2854delA; c.2855T>C; c.2856G>C; c.2856G>T; c.2856G>A; c.2859_2890del; c.2869_2870insG; c.2872C>T; c.2875delG; c.2879_2882delCTAT; c.2883_2886dup; c.2885C>A; c.2885C>G; c.2891_2892delinsA; c.2896delA; c.2897del; c.2899_2900delinsA; c.2900T>C; c.2907A>C; c.2908G>C; c.2908G>A; c.2908+1G>A; c.2909-15T>G; c.2909_2924dup; c.2909delG; c.2909-2A>T; c.2909-1G>C; c.2909-1G>A; c.2909G>A; c.2916_2952delTCTTAATAGATTCTCAAAGATATAGCAATTTGGAT; c.2917C>T; c.2924_2925delGA; c.2929T>C; c.2930C>T; c.2932A>T; c.2936A>C; c.2936A>T; c.2939T>A; c.2948del; c.2956del; c.2958del; c.2959_2960dup; c.2964dup; c.2968dup; c.2977G>T; c.2982_2988+2delCATCCAGGT; c.2986dupC; c.2986C>A; c.2988+1G>A; c.2988+1G>C; c.2988+1G>T; c.2988+2del; c.2988+2T>C; c.2988+2T>G; c.2989-3C>G; c.2989-2A>G; c.2989-2A>T; c.2989-2A>C; c.2989-1G>A; c.2989-1G>T; c.2993delT; c.2997_3000delAATT; c.2994del; c.3000_3014del; c.2998del; c.3002_3003delTG; c.3008del; c.3007G>T; c.3011_3019delCTATAGCAG; c.3011delC; c.3014dupT; c.3014T>G; c.3017C>A; c.3022delG; c.3032T>G; c.3033dup; c.3039delC; c.3039dupC; c.3038C>A; c.3038C>T; c.3047T>C; c.3061C>T; c.3061C>A; c.3067_3072delATAGTG; c.3068_3072delTAGTG; c.3068T>G; c.3078del; c.3081T>G; c.3083T>G; c.3095A>G; c.3103C>T; c.3106del; c.3106A>G; c.3107C>A; c.3107C>T; c.3110C>A; c.3124C>T; c.3131A>G; c.3134C>A; c.3136G>T; c.3139_3139+1delGG; c.3139+1delG; c.3139+1G>A; c.3139+1G>C; c.3139+2T>C; c.3140-26A>G; c.3140-16T>A; c.3140-11A>G; c.3140-1G>A; c.3154T>G; c.3158C>T; c.3160C>G; c.3161delA; c.3168del; c.3176T>G; c.3180del; c.3185_3191del; c.3181G>C; c.3181G>A; c.3184_3188dupCTATG; c.3188G>A; c.3189G>A; c.3191_3192insTTTTAAGCTTAAAAGG; c.3194T>C; c.3194T>G; c.3196C>T; c.3197G>A; c.3197G>T; c.3200_3205del; c.3200C>T; c.3205G>A; c.3208C>T; c.3209G>A; c.3209G>C; c.3211C>T; c.3212A>C; c.3217dupT; c.3218A>G; c.3220T>C; c.3222T>A; c.3222T>G; c.3229_3230delCT; c.3227_3231del; c.3231_3232delGT; c.3230T>C; c.3254A>G; c.3254A>C; c.3257C>T; c.3259G>C; c.3263dupA; c.3264delC; c.3266G>A; c.3267G>A; c.3275_3290del; c.3274T>C; c.3277del; c.3276C>A; c.3276C>G; c.3281C>G; c.3287del; c.3290_3305del; c.3292T>C; c.3293G>A; c.3294del; c.3293G>C; c.3293G>T; c.3294G>A; c.3294G>C; c.3294G>T; c.3297C>A; c.3301del; c.3299A>C; c.3302T>A; c.3302T>G; c.3303_3304delinsAG; c.3304A>T; c.3310G>T; c.3311A>T; c.3315del; c.3319T>C; c.3322_3323del; c.3322del; c.3324delC; c.3330del; c.3343dupA; c.3351_3352del; c.3353C>T; c.3353C>G; c.3357_3359delinsA; c.3365del; c.3367+1G>A; c.3367+1G>T; c.3367+2T>A; c.3367+2T>C; c.3368-2A>G; c.3368-2A>T; c.3368-1G>A; c.3382A>T; c.3384_3388del; c.3389_3402del; c.3392_3396dup; c.3397del; c.3398_3399del; c.3400_3401delACinsGTA; c.3404T>A; c.3407_3422del; c.3406G>A; c.3411_3414del; c.3409A>G; c.3410T>G; c.3415A>G; c.3419_3421delTGA; c.3419T>A; c.3421_3424dupAGTA; c.3428T>A; c.3430C>T; c.3434G>C; c.3434G>A; c.3435G>A; c.3444del; c.3454G>C; c.3458T>A; c.3461A>G; c.3468G>A; c.3468_3468+6delinsATAAAAAGCTTATAAAAAG; c.3468+1G>A; c.3468+1G>T; c.3468+2dupT; c.3468+2T>C; c.3468+5G>A; c.3469-1304C>G; c.3469-3_3469-2del; c.3469-2A>G; c.3469-2A>C; c.3469-1G>A; c.3469-1G>C; c.3472C>T; c.3475T>C; c.3476C>T; c.3484C>T; c.3486_3487delAG; c.3485G>T; c.3492dupT; c.3492del; c.3493A>T; c.3493_3494delinsT; c.3495delG; c.3497del; c.3525_3537del; c.3528delC; c.3530delA; c.3529A>T; c.3532_3535dupTCAA; c.3536_3539delCCAA; c.3533C>A; c.3533C>G; c.3540delA; c.3539_3544del; c.3546delC; c.3546C>G; c.3546C>A; c.3547A>T; c.3556C>T; c.3567dup; c.3569_3570delIT; c.3571dup; c.3587C>G; c.3587C>A; c.3592del; c.3598_3599delAAinsG; c.3600del; c.3599_3600delAAinsG; c.3602del; c.3605delA; c.3611G>A; c.3612G>A; c.3615del; c.3617C>A; c.3617C>G; c.3618_3619delAG; c.3623delG; c.3623dup; c.3622_3623delinsT; c.3623G>A;</p>

	<p>c.3639dup; c.3658dup; c.3659delC; c.3661_3662del; c.3672dup; c.3682G>T; c.3691delT; c.3697_3698del; c.3700A>G; c.3708del; c.3712C>T; c.3717G>A; c.3717+1G>A; c.3717+4A>G; c.3717+5G>A; c.3717+40A>G; c.3718-2477C>T; c.3718-5_3725del; c.3718-3T>G; c.3718-1G>A; c.3718-1G>C; c.3719T>G; c.3724del; c.3725del; c.3728T>A; c.3730G>A; c.3731G>A; c.3731G>T; c.3736del; c.3737C>T; c.3739G>A; c.3739G>T; c.3741_3745dup; c.3741_3742insATCAG; c.3744delA; c.3743C>A; c.3743C>G; c.3747delG; c.3745G>A; c.3745G>C; c.3745_3750delinsC; c.3746G>A; c.3752G>A; c.3754dup; c.3758dup; c.3761T>G; c.3763T>C; c.3764C>A; c.3767dupC; c.3773dupT; c.3773del; c.3782del; c.3782_3783del; c.3788del; c.3792del; c.3793G>T; c.3793G>A; c.3795del; c.3794G>A; c.3796G>T; c.3806T>A; c.3808delG; c.3809A>T; c.3816_3817del; c.3822G>A; c.3827del; c.3829del; c.3836T>A; c.3838C>T; c.3841C>T; c.3844T>G; c.3844T>C; c.3845G>A; c.3846G>A; c.3846G>C; c.3848G>T; c.3851_3852dupAA; c.3849G>C; c.3857T>C; c.3859G>T; c.3868C>T; c.3871C>T; c.3872A>G; c.3873G>C; c.3873+1G>A; c.3873+1G>T; c.3873+2T>C; c.3874-4522A>G; c.3874-2A>C; c.3874-2A>G; c.3874-1G>A; c.3876del; c.3883_3886delATTT; c.3883delA; c.3883_3884insG; c.3889dupT; c.3889del; c.3891dupT; c.3896C>T; c.3900dupT; c.3908delA; c.3908dup; c.3904A>T; c.3907A>C; c.3907A>T; c.3908A>T; c.3909C>G; c.3917_3918dup; c.3921T>A; c.3922G>T; c.3925C>T; c.3929G>A; c.3930G>A; c.3932_3933delinsAATATG; c.3937C>T; c.3944_3951delTATGGAAA; c.3947G>A; c.3949A>T; c.3957del; c.3957_3958insAGGG; c.3963+1G>A; c.3963+1G>C; c.3963+1G>T; c.3963+2T>A; c.3963+2T>G; c.3964-3C>G; c.3965_3969dup; c.3964-1_3964delinsTT; c.3971T>C; c.3972delC; c.3976del; c.3988_3989delCA; c.3988C>T; c.3999delG; c.3997G>T; c.4004T>C; c.4007_4010del; c.4023del; c.4028delG; c.4028dup; c.4033_4034del; c.4032T>A; c.4035_4038dupCCTA; c.4036_4042delCTAAGCC; c.4036dup; c.4046del; c.4046G>A; c.4052del; c.4051A>T; c.4054C>T; c.4056G>T; c.4058delT; c.4056G>C; c.4065_4066del; c.4071delT; c.4071_4073delinsAA; c.4078_4079del; c.4077_4080delTGTTinsAA; c.4078delG; c.4086dupT; c.4090del; c.4097T>C; c.4097T>A; c.4102delinsTGCTTGA; c.4111G>T; c.4124A>C; c.4127_4131delTGGAT; c.4129G>C; c.4136+1G>A; c.4139del; c.4143C>G; c.4144C>T; c.4147dupA; c.4168C>T; c.4170del; c.4197_4198delCT; c.4193T>G; c.4200_4201delTG; c.4199delG; c.4201G>T; c.4225G>T; c.4230C>A; 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.4252delG; c.4252G>T; c.4262_4263del; c.4262T>A; c.4272C>A; c.4272C>G; c.4272C>G; c.4276T>C; c.4280T>C; c.4300_4301dupAG; c.4297G>T; c.4303dupA; c.4307_4320delTCTTCCGGCAAGCC; c.4312del; c.4315C>T; c.4325del; c.4329delC; c.4339delG; c.4340del; c.4364C>A; c.4364C>G; c.4374dup; c.4387_4388del; c.4389delG; c.4392del; c.4400delT; c.4408G>T; c.4417G>T; c.4426C>T; c.4433C>G; c.4443G>C; c.366+10297C>T.</p>
<p>GALT NM_000155.4 (419)</p>	<p>c.1A>G; c.1A>C; c.2T>C; c.2T>G; c.14_23dupGAACCCATCC; c.18delC; c.20del; c.25C>T; c.28C>T; c.29_48delinsGACC; c.37C>T; c.40_41insT; c.41delCinsTT; c.50del; c.71_72insA; c.82G>A; c.82G>C; c.82G>T; c.82+2delT; c.82+2T>G; c.83-2A>G; c.83-1G>C; c.86A>G; c.90G>C; c.91C>A; c.95T>A; c.98G>A; c.100T>A; c.101A>G; c.107C>T; c.113A>C; c.118G>A; c.118G>T; c.127_130del; c.130G>A; c.130G>T; c.132delG; c.136_140delGCTCA; c.133T>C; c.134C>A; c.134C>T; c.142C>A; c.142C>T; c.151C>T; c.152G>A; c.152G>T; c.155C>G; c.157T>A; c.158G>A; c.160C>T; c.164delG; c.163G>T; c.165_174del; c.166C>T; c.173A>T; c.180delG; c.189dupG; c.197C>A; c.199C>T; c.200del; c.200G>A; c.203A>C; c.207_214delCCCTCTCA; c.209del; c.213dup; c.220_221delCT; c.220_221insG; c.220C>G; c.221T>C; c.231_233delinsT; c.238C>T; c.241G>C; c.241G>A; c.252+1G>A; c.252+2T>C; c.252+2T>G; c.253-2A>G; c.253-1G>A; c.262dup; c.260C>G; c.265T>C; c.265T>G; c.267C>G; c.285T>G; c.289_291delAAC; c.289A>G; c.290A>G; c.290A>T; c.291C>G; c.292G>A; c.292G>C; c.299C>G; c.307C>T; c.327del; c.328+1G>C; c.328+2T>C; c.328+2T>G; 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.367C>G; c.368G>A; c.368G>C; c.370G>T; c.374T>C; c.377+1G>T; c.377+1G>A; c.378-2A>T; c.378-2A>G; c.379A>G; c.384_388del; c.382G>A; c.385A>T; c.386T>C; c.392T>G; c.394C>T; c.398_399dupCC; c.396C>A; c.400delT; c.403T>G; c.404C>G; c.404C>T; c.406G>C; c.410dupT; c.412del; c.413C>T; c.413C>G; c.416T>C; c.424A>G; c.424A>C; c.425T>A; c.425T>C; c.428C>T; c.428C>G; c.434_435del; c.439A>G; c.442C>G; c.442C>T; c.445dupG; c.443G>A; c.448G>C; c.450del; c.452T>C; c.460T>C; c.460T>G; c.462G>A; c.465_474dup; c.467C>A; c.468_469delinsT; c.482T>C; c.490C>T; c.499T>C; c.502_504delGTG; c.501G>A; c.501G>C; c.502G>A; c.502G>T; c.505C>A; c.507G>C; c.507+1G>T; c.507+2T>C; c.508-29del; c.508-2_509del; c.508-1G>C; c.508-1G>A; c.509T>A; c.509T>C; c.513delT; c.513dupT; c.512T>C; c.514G>T; c.524G>A; c.528_529insG; c.530T>C; c.533T>G; c.535G>C; c.536G>A; c.539G>T; c.541T>G; c.542C>T; c.550dup; c.550del; c.547C>A; c.550C>G; c.556del; c.552C>A; c.553C>T; c.554C>A; c.554C>T; c.556C>A; c.556C>T; c.557A>G; c.558C>A; c.561_564+9delCCAGGTAAGGGTG; c.558C>G; c.561C>A; c.562C>T; c.563A>G; c.564+1G>A; c.564+1G>T; c.565_578delGTATGGCCAGCAG; c.565-2A>G; c.568T>C; c.576_589del; c.572C>T; c.574A>G; c.575G>A; c.580T>C; c.584T>C; c.586C>G; c.587C>T; c.590A>G; c.593T>C; c.594T>G; c.595G>A; c.598delC; c.598C>T; c.601C>T; c.602G>A; c.604G>A; c.607G>A; c.608A>G; c.610C>T; c.611G>C; c.615_616del; c.616C>T; c.617A>G; c.619C>T; c.625T>G; c.626A>C; c.626A>G; c.627T>A; c.634C>T; c.635A>C; c.645_646insGAGT; c.649_653dup; c.650T>C; c.652delC; c.652_653del; c.658dupG; c.658G>A; c.665G>A; c.667C>A; c.667C>T; c.670delC; c.668G>A; c.669_670del; c.670C>T; c.677T>C; c.680_681del; c.680T>C; c.684del; c.687G>T; c.687+1G>T; c.687+2T>C; c.688-2A>C; c.688-1G>C; c.691C>G; c.691C>T; c.692G>A; c.697G>C; c.708_709del; c.707G>T; c.719_728delTAGTACTGGT; c.717G>T; c.719T>G; c.737G>A; c.745T>C; c.747G>A; c.748C>A; c.752_753delACinsCT; c.754del; c.752A>C; c.756G>T; c.761dupT; c.768_770delGCC; c.772del; c.770C>T; c.772C>T; c.779_790delATGTGCGCGCGG; c.775C>T; c.776G>A; c.785G>C; c.790delC; c.790_792delCTAinsTAG; c.792_793insG; c.794del; c.796G>T; c.812A>G; c.813G>C; c.814del; c.814C>G; c.814C>T; c.815G>A; c.815G>C; c.815G>T; c.820+2T>C; c.820+13A>G; c.821-7A>G; c.821-2A>G; c.821-1del; c.821-1G>A; c.824delT; c.823C>G; c.833T>A; c.835A>G; c.836T>G; c.844C>G; c.849_850del; c.853A>T; c.854A>G; c.854A>C; c.855G>T; c.856_864delTATGACAAC; c.857A>G; c.860A>G; c.865C>T; c.866T>G; c.870del; c.871G>A; c.872A>T; c.882delT; c.881T>A; c.883C>T; c.883C>A; c.888C>G; c.888C>A; c.892A>G; c.893T>C; c.894G>A; c.895G>A; c.896G>A; c.899G>A; c.900G>A; c.900G>T; c.904+1G>A; c.904+1G>T; c.904+2T>G; c.905-2A>G; c.905-1G>A; c.912dupC; c.910C>G; c.910C>T; c.912_913insGA; c.920C>A; c.921dup; c.938G>A; c.939G>A; c.947G>A; c.948G>A; c.949delC; c.949C>T; c.950A>G; c.951G>T; c.952delC; c.957C>A; c.957C>G; c.958G>A; c.959C>T; c.961C>T; c.967T>C; c.967T>G; c.968A>G; c.971C>T; c.979_986delCTGCGCTC; 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>A; c.998G>T; c.1001A>G; c.1006A>T; c.1006A>G; c.1014C>G; c.1014C>T; c.1018G>A; c.1018G>T; c.1024C>A; c.1030C>A; c.1030C>T; c.1034C>A; c.1047delC; c.1046T>G; c.1048delA; c.1048A>G; c.1052delC; c.1049C>G; c.1049C>T; c.1054G>C; c.1055A>G; c.1057C>T; c.1059+1G>T; c.1060-2A>G; c.1060-1G>A; c.1072delC; c.1087G>T; c.1087G>A; c.1098C>A; c.1098C>G; c.1108C>T; c.1117_1118del; c.1138T>C; c.1139G>C; c.1140A>C.</p>

<p>GJB2 NM_004004.6 (272)</p>	<p>c.668_671delAAAA; c.647_650delGATA; c.645delT; c.641T>G; c.630_634del; c.632_633delGT; c.632G>A; c.626A>C; c.624del; c.617A>G; c.614T>C; c.610del; c.560_605dup; c.605G>T; c.604T>C; c.592_600delGTCTGGAGinsCAGTGTTCATGACATTC; c.592_600delinsCAGTGTTCATGACATTC; c.598G>A; c.598G>T; c.596G>T; c.595T>C; c.595T>G; c.592G>A; c.585G>T; c.585G>A; c.584T>C; c.583A>G; c.583A>T; c.575_576delCA; c.576delA; c.568_571del; c.564_565delGA; c.563A>G; c.560A>G; c.551G>A; c.551G>C; c.550C>T; c.548C>T; c.535G>A; c.523_533delCCCAACACTGT; c.533T>C; c.532G>T; c.527A>G; c.526A>G; c.524C>G; c.518C>G; c.517C>T; c.516G>A; c.516G>C; c.514delT; c.514T>A; c.508_511dupAACG; c.506G>A; c.505T>C; c.498_499dup; c.500T>C; c.486_493dup; c.490C>T; c.487A>G; c.487A>C; c.458_475dup; c.474C>G; c.468_472del; c.465T>A; c.464A>G; c.453_460delinsAGAGAAGAC; c.456C>A; c.456C>G; c.449del; c.440A>G; c.439G>A; c.428G>A; c.428G>C; c.427C>T; c.426C>A; c.420C>G; c.416G>A; c.409A>C; c.408C>A; c.407dup; c.402delG; c.400T>C; c.399G>A; c.389G>C; c.388G>C; c.385G>T; c.382A>G; c.380G>T; c.379C>T; c.370C>T; c.365A>T; c.358_360delGAG; c.358_360del; c.353_356dup; c.355G>A; c.355G>T; c.345dup; c.338_341dupGTGA; c.334_335delAA; c.314_329del; c.328del; c.327_328delGGinsA; c.313_326delAAGTTCATCAAGGG; c.326G>T; c.310_323delAGGAAGTTCATCAA; c.299_300delAT; c.298delC; c.299A>T; c.298C>T; c.290dupA; c.280_284dupCACGT; c.284T>C; c.283G>A; c.279G>A; c.277A>G; c.277A>C; c.269dupT; c.269T>C; c.268del; c.266T>C; c.265C>T; c.263C>T; c.257delC; c.257C>G; c.256A>G; c.253T>C; c.251T>C; c.250G>A; c.250G>C; c.250G>T; c.247_249del; c.246C>G; c.244A>G; c.241C>G; c.239dupA; c.239A>C; c.238C>T; c.238C>A; c.235delC; c.236T>A; c.232dupG; c.233C>A; c.232G>A; c.232G>T; c.231G>A; c.230G>A; c.229T>C; c.227T>C; c.224G>A; c.223C>G; c.223C>T; c.218A>G; c.206_212dup; c.208C>G; c.207C>G; c.206T>C; c.205T>C; c.200dup; c.196G>C; c.196G>T; c.196G>A; c.195C>G; c.195C>A; c.194A>G; c.193T>C; c.176_191del16; c.188T>C; c.187G>C; c.187G>T; c.187G>A; c.178T>G; c.176G>A; c.176G>C; c.176G>T; c.175G>A; c.175G>C; c.172C>G; c.172C>T; c.169C>T; c.167delT; c.164C>A; c.162C>A; c.161del; c.161A>G; c.155_158del; c.158G>A; c.158G>T; c.157T>A; c.148G>A; c.148G>T; c.145del; c.137_141delATGAG; c.139G>T; c.139G>A; c.136G>A; c.136G>C; c.134delG; c.134G>A; c.134G>T; c.133G>A; c.130_131delTG; c.132G>A; c.132G>C; c.131G>A; c.131G>C; c.125_127delAGG; c.125_127del; c.128T>C; c.119C>A; c.119C>G; c.118G>A; c.110T>C; c.109G>A; c.109G>T; c.108del; c.104T>G; c.101T>C; c.101T>G; c.101T>A; c.98T>C; c.95G>A; c.95G>T; c.94C>A; c.94C>T; c.93del; c.89T>C; c.85_87delTTC; c.88A>T; c.82C>G; c.79_82delinsAGA; c.76del; c.72G>A; c.72G>C; c.71G>A; c.31_68del; c.66G>T; c.53_63del; c.51_62delCACCAGCATTGGinsA; c.60T>G; c.59T>C; c.56G>C; c.56G>T; c.49_50delTTC; c.50C>T; c.32_45delGGGGTGTGAACAAA; c.44A>C; c.41dup; c.41A>G; c.40A>G; c.37G>A; c.35delG; c.35dupG; c.35G>T; c.35G>A; c.34G>C; c.34G>T; c.28del; c.23C>T; c.19C>T; c.11delG; c.9G>A; c.8G>A; c.3G>A; c.2T>C; c.2T>G; c.1A>G; c.1A>T; c.-22-2A>C; c.-22-2A>G; c.-23+2T>G; c.-23+1G>A; c.-23G>T.</p>
<p>GJB6 NM_001110219.3 (16)</p>	<p>c.487delC; c.485dupA; c.443delC; c.383_384delTA; c.346A>T; c.322C>T; c.263C>T; c.261dupA; c.223C>T; c.176G>A; c.169C>T; c.148G>C; c.110T>A; c.63del; c.31G>A; c.14C>T.</p>
<p>HEXA* NM_000520.6 (280)</p>	<p>c.1559G>A; c.1549dup; c.1543C>T; c.1540del; c.1537C>T; c.1528C>T; c.1525_1526+27del; c.1526+2T>C; c.1526+1G>A; c.1526+1G>T; c.1511G>T; c.1511G>A; c.1510del; c.1510C>T; c.1502C>G; c.1499del; c.1496G>A; c.1495C>T; c.1491T>G; c.1455G>A; c.1454G>A; c.1429_1445del; c.1444G>T; c.1444G>A; c.1435del; c.1432G>A; c.1422G>C; c.1422-1G>T; c.1422-2A>G; c.1421+1G>T; c.1421+1G>C; c.1421G>A; c.1419_1420del; c.1395C>G; c.1393G>A; c.1389T>A; c.1385A>T; c.1382G>T; c.1378T>C; c.1364del; c.1363G>A; c.1361G>A; c.1360G>C; c.1360G>A; c.1349del; c.1342C>T; c.1338del; c.1333del; c.1330+2T>C; c.1330+1G>A; c.1307_1308del; c.1305C>T; c.1302C>G; c.1294A>T; c.1293G>A; c.1278_1281dup; c.1275_1278dup; c.1274_1277dup; c.1260G>A; c.1260G>C; c.1259G>A; c.1258del; c.1257dup; c.1250_1251del; c.1220del; c.1217_1220delinsA; c.1214_1215delinsG; c.1194_1197del; c.1178G>C; c.1177C>T; c.1176G>A; c.1171G>A; c.1169_1171delinsT; c.1168del; c.1168C>T; c.1160_1161del; c.1150C>T; c.1147-1G>T; c.1142_1146del; c.1140del; c.1123del; c.1123G>T; c.1121A>C; c.1120C>T; c.1119G>A; c.1118G>A; c.1114del; c.1100_1111del; c.1098T>G; c.1098T>A; c.1092_1093del; c.1088dup; c.1074-7_1074-3del; c.1073+1G>C; c.1073+1G>T; c.1073+1G>A; c.1069C>T; c.1039_1056del; c.1053_1054delinsTCCAGCTTGA; c.1048C>T; c.1043_1046del; c.1031T>C; c.1030T>A; c.1024A>T; c.1014del; c.989_1010dup; c.1002del; c.996C>A; c.995A>T; c.987G>A; c.987-1G>C; c.986+3A>G; c.986+2T>C; c.986+1G>C; c.986+1G>A; c.986G>A; c.977del; c.972T>A; c.967G>A; c.965A>G; c.965A>T; c.962_964del; c.964G>T; c.964G>A; c.962G>A; c.958G>T; c.947dup; c.938del; c.929_930del; c.915_917del; c.917T>G; c.898_905del; c.902T>G; c.862G>T; c.855_856insTC; c.844G>T; c.834C>G; c.822_823del; c.814G>A; c.814G>T; c.806G>A; c.806-1G>A; c.806-1G>C; c.806-7G>A; c.805+1G>C; c.805+1G>A; c.805G>C; c.805G>A; c.797G>A; c.788C>T; c.786C>G; c.778C>T; c.772G>C; c.766G>T; c.755G>A; c.754C>T; c.749del; c.749G>A; c.748G>A; c.743del; c.739C>T; c.736G>A; c.735C>A; c.718_719insT; c.718A>T; c.709C>T; c.677C>T; c.672+2T>C; c.672+1del; c.672+1G>T; c.672+1G>A; c.672G>T; c.638A>G; c.637T>C; c.631_634del; c.632T>C; c.629C>G; c.629C>T; c.617T>A; c.616del; c.616G>C; c.611A>G; c.609G>A; c.608G>A; c.607T>G; c.605A>G; c.604del; c.604C>T; c.598G>A; c.590A>C; c.574G>C; c.571-1G>T; c.570+1G>A; c.568_569dup; c.570G>A; c.568_569del; c.570G>T; c.551_552del; c.546dup; c.540C>G; c.539del; c.538T>C; c.536A>G; c.533G>T; c.533G>A; c.532C>T; c.530C>G; c.524A>C; c.521dup; c.514_515del; c.509G>A; c.508C>T; c.496del; c.492del; c.477_478del; c.465del; c.460-1G>T; c.460-1G>A; c.460-2A>G; c.459+24G>C; c.459+5G>A; c.459+4A>C; c.459+2dup; c.459+1G>A; c.450_456del; c.451G>T; c.441G>A; c.436del; c.430C>T; c.426del; c.425_426del; c.422del; c.421_422insAGGAGGCC; c.413-1G>A; c.413-2A>G; c.412+1G>T; c.409C>T; c.400_401del; c.395_398del; c.398G>A; c.397del; c.380T>G; c.379C>T; c.370C>T; c.347A>G; c.346+1G>A; c.346+1G>T; c.346+1G>C; c.340G>A; c.316C>T; c.314del; c.310_313dup; c.293_294del; c.289_290del; c.288del; c.284T>A; c.276dup; c.271del; c.254-1G>C; c.254-2A>G; c.254-34G>C; c.237_253+7del; c.253-1G>A; c.226_242del; c.233G>A; c.228del; c.207_208del; c.196del; c.196C>T; c.183_193del; c.187G>T; c.173G>A; c.170dup; c.163C>T; c.155C>A; c.144C>A; c.144C>G; c.139_140delinsT; c.139C>T.</p>
<p>EMD NM_000117.3 (102)</p>	<p>c.1A>G; c.2T>G; c.3G>A; c.12C>A; c.12C>G; c.16delG; c.22del; c.60delC; c.77dupT; c.79del; c.82G>A; c.82+1G>A; c.82+1G>T; c.83-2A>C; c.83-2A>G; c.101dupA; c.102C>G; c.103G>T; c.110_112del; c.116_143del; c.121_155del; c.123C>A; c.125del; c.130C>T; c.135dupG; c.139_140dup; c.143dup; c.153dupC; c.153del; c.177_178delTA; c.178del; c.184dupT; c.187+1G>T; c.187+1G>A; c.187+2T>G; c.217_218dupAT; c.217dup; c.236_239del; c.239_240insT; c.251_255delTCTAC; c.256C>T; c.262A>T; c.265+2T>A; c.266-3A>G; c.266-2A>G; c.284_298delATGAAGAGAGCTACT; c.282C>G; c.282C>A; c.313del; c.359_362del; c.355C>T; c.359C>A; c.391_397dup; c.397C>T; c.399+1G>T; c.399+2T>C; c.406del; c.419T>A; c.428_430del; c.430G>T; c.439_449+3del; c.441C>A; c.449+1G>A; c.450-2A>G; c.450-1G>A; c.468_471delCCGG; c.475_476delAG; c.483C>G; c.484C>T; c.506_507del; c.512C>A; c.512C>G; c.545_547del; c.546T>A; c.547C>A; c.548C>A; c.548C>T; c.561_567dup; c.570dup; c.570del; c.581_582delCA; c.581C>G; c.600G>A; c.607delC; c.615del; c.619delC; c.621delG; c.631del; c.643_653dup; c.640_644dupGGGGC; c.643del; c.650_654dupTGGGGC; c.650_654del; c.651_655dup; c.662_663insT; c.674_678delTCTGG; c.674del; c.676_677del; c.682C>T; c.703_704insA; c.740_743del; c.46_82+6del.</p>

<p>GLA NM_000169.3 (1159)</p>	<p>c.1287_1288dup; c.1284_1287delACTT; c.1288T>C; c.1285delC; c.1277_1278del; c.1278del; c.1250T>G; c.1250T>C; c.1246C>T; c.1244T>C; c.1241del; c.1240_1241del; c.1241T>C; c.1235_1236delCT; c.1235C>T; c.1235C>A; c.1234A>C; c.1232G>A; c.1231G>C; c.1229C>T; c.1229C>A; c.1228A>G; c.1228A>C; c.1226C>T; c.1225C>G; c.1225C>T; c.1225C>A; c.1223del; c.1220T>G; c.1220T>A; c.1213_1217del; c.1212_1214delAAG; c.1213del; c.1210dup; c.1208del; c.1208T>C; c.1197_1202dup; c.1201dupT; c.1202C>G; c.1202C>A; c.1199_1200del; c.1197G>A; c.1193_1196del; c.1196G>A; c.1194del; c.1192G>T; c.1191T>G; c.1189dup; c.1188delC; c.1187del; c.1185dup; c.1184G>A; c.1181_1183dup; c.1182del; c.1176_1179del; c.1177_1178insG; c.1176del; c.1176G>T; c.1171A>G; c.1169dup; c.1168G>A; c.1167dup; c.1166delC; c.1166C>T; c.1163_1165del; c.1165C>G; c.1163T>A; c.1160T>C; c.1156_1157del; c.1157A>C; c.1156C>T; c.1153A>C; c.1151_1152delinsAT; c.1151T>A; c.1147_1149delITTC; c.1145_1149del; c.1139_1149del; c.1146C>G; c.1134_1145dup; c.1146C>A; c.1145G>C; c.1145G>A; c.1125_1140del; c.1139del; c.1134T>A; c.1133G>A; c.1132dup; c.1133G>T; c.1132T>C; c.1130C>A; c.1124_1129del; c.1125_1128dupAGTG; c.1121_1123del; c.1077_1120del; c.1119dup; c.1118del; c.1118G>A; c.1118G>T; c.1080_1117del; c.1117G>A; c.1117G>C; c.1116_1117delinsAT; c.1115T>G; c.1115T>A; c.1090_1103del; c.1093_1101dup; c.1102delinsTTATAC; c.1100dup; c.1081_1100del; c.1098del; c.1086_1098del; c.1085_1098del; c.1095del; c.1095T>A; c.1095T>G; c.1094dup; c.1091_1092del; c.1088_1091dup; c.1088del; c.1088G>A; c.1088G>C; c.1087C>T; c.1077_1086del; c.1085C>T; c.1084C>A; c.1080_1082del; c.1082del; c.1082G>A; c.1082G>C; c.1067_1082delinsTACTCTTAT; c.1081G>A; c.1081G>C; c.1081G>T; c.1069_1079del; c.1079G>A; c.1078_1079insT; c.1077delT; c.1078G>C; c.1078G>A; c.1078G>T; c.1062_1076delAAACCGGCAGGAGAT; c.1076T>C; c.1074_1075del; c.1072_1074delGAG; c.1073A>G; c.1073A>C; c.1072G>A; c.1072G>T; c.1069C>T; c.1068dup; c.1067G>A; c.1067G>C; c.1066C>T; c.1066C>G; c.1064dup; c.1065C>A; c.1061T>A; c.1057_1058delAT; c.1032_1058del; c.1055_1057dup; c.1055_1056delCT; c.1055C>G; c.1055C>A; c.1054G>C; c.1051_1052del; c.1049del; c.1048G>A; c.1048G>C; c.1047G>A; c.1046G>A; c.1045_1046insA; c.1045T>C; c.1045T>A; c.1043_1044insG; c.1042dup; c.1042G>C; c.1041dup; c.1040dupT; c.1037delG; c.1031_1034dupTCTC; c.1033_1034delTCT; c.1034C>G; c.1033T>C; c.1031dup; c.1031T>C; c.1028del; c.1017_1027delGTGGGAACGAC; c.1028C>T; c.1016_1026del; c.1025del; c.1025G>A; c.1025G>T; c.1023delA; c.1024C>G; c.1024C>T; c.1023A>C; c.1022A>G; c.1021dup; c.1021_1022insT; c.1021del; c.1021G>A; c.1021G>T; c.1019_1020insA; c.1020G>A; c.1019G>A; c.1019G>C; c.1018T>C; c.1016T>G; c.1012G>A; c.1012G>T; c.1010T>C; c.1000-1G>A; c.1000-1G>C; c.1000-2A>T; c.1000-2A>G; c.1000-10G>A; c.999+2T>C; c.999+1del; c.996_999delACAG; c.999+1G>T; c.999+1G>A; c.997dup; c.997C>T; c.994dupA; c.993_994del; c.994del; c.988delC; c.988C>T; c.987C>A; c.986del; c.984del; c.983G>C; c.983G>A; c.982G>T; c.982G>C; c.982G>T; c.981A>C; c.980A>G; c.980A>T; c.979C>A; c.979C>T; c.974dup; c.974G>A; c.973G>A; c.973G>C; c.971T>A; c.969del; c.946_966del; c.966C>A; c.966C>G; c.964del; c.964G>C; c.963_964delinsCA; c.959_962delATCA; c.963G>C; c.962A>G; c.962A>T; c.961C>G; c.961C>T; c.960T>G; c.959A>T; c.958A>T; c.956T>C; c.950_954dupTTGCC; c.955A>T; c.952del; c.950T>C; c.950T>G; c.950T>A; c.949del; c.947dup; c.946_947insACGACACATCAGCCCTCAAGCCAAAGCTCTCTTCAGGATAAGGACG; c.947T>G; c.947T>A; c.946del; c.936G>C; c.936G>T; c.935A>G; c.934C>T; c.932T>G; c.932T>C; c.931dup; c.931del; c.931C>T; c.931C>G; c.929T>G; c.928C>G; c.928C>T; c.927del; c.925G>C; c.924A>C; c.924A>T; c.909_918del; c.916C>T; c.914del; c.895_908del; c.908T>A; c.908T>C; c.908T>G; c.907A>T; c.903_904insG; c.902dupG; c.902G>A; c.902G>C; c.901del; c.902G>T; c.901C>G; c.901C>T; c.899T>C; c.899T>A; c.898C>T; c.897C>A; c.896A>G; c.896A>T; c.894T>A; c.894T>G; c.893_894insG; c.893A>G; c.892_893insT; c.892A>C; c.892A>G; c.890C>T; c.890C>A; c.889T>C; c.888G>A; c.888G>T; c.886_887del; c.887T>C; c.886A>G; c.886A>T; c.885del; c.884del; c.884T>G; c.881T>G; c.879_880delTTinsAATC; c.878C>T; c.878C>A; c.877C>T; c.877C>A; c.877C>G; c.875C>T; c.874G>C; c.871G>A; c.870G>A; c.870G>C; c.869T>C; c.868A>C; c.868A>T; c.866T>G; c.865A>T; c.864dup; c.863delC; c.862G>C; c.861G>A; c.861G>T; c.861G>C; c.860G>C; c.860G>A; c.859T>C; c.859T>G; c.854C>A; c.853G>C; c.851T>C; c.850A>G; c.848dupA; c.848A>C; c.846_847del; c.847C>T; c.842_844delTAA; c.845C>T; c.844A>C; c.844A>G; c.842dup; c.837_839del; c.838C>A; c.837G>T; c.837G>C; c.836A>G; c.835C>G; c.835C>T; c.835C>A; c.834del; c.833dup; c.833del; c.831G>A; c.830G>A; c.828C>A; c.827G>C; c.827G>A; c.826A>G; c.824_825del; c.825del; c.823delC; c.824T>A; c.823C>T; c.821G>T; c.821G>A; c.820G>C; c.820G>A; c.819T>G; c.819T>A; c.818T>C; c.817T>C; c.816C>A; c.815A>G; c.815A>T; c.812G>T; c.812G>A; c.812G>C; c.811_812delinsCTCA; c.811G>A; c.811G>T; c.810T>G; c.809T>C; c.807del; c.803_806delTAGT; c.803_806del; c.806T>C; c.806T>G; c.805G>A; c.805G>C; c.802-3_804delCAGTTAinsGGCACTTT; c.804A>C; c.803T>C; c.802-3_802-2delCA; c.802-1G>C; c.802-2A>G; c.802-2A>T; c.801+48T>G; c.801+4A>G; c.801+3A>G; c.801+2dup; c.801+2T>C; c.801+1G>C; c.801+1G>T; c.801+1G>A; c.800delT; c.801G>A; c.800T>G; c.800T>C; c.798T>A; c.797A>T; c.797A>C; c.797A>G; c.796G>A; c.796G>T; c.794C>G; c.793C>T; c.791A>T; c.791A>C; c.790G>T; c.788A>G; c.786dupG; c.786del; c.786G>C; c.785G>A; c.785G>T; c.784T>C; c.782del; c.782dup; c.782G>A; c.782G>T; c.781G>T; c.779G>A; c.779G>T; c.779G>C; c.777_778del; c.777delA; c.778G>A; c.778G>C; c.774_775delAC; c.776C>G; c.776C>T; c.738_775del; c.773G>T; c.772G>C; c.772G>A; c.772G>T; c.770C>T; c.769dup; c.770G>A; c.769G>C; c.763del; c.761_762del; c.761T>C; c.760dup; c.759del; c.758T>C; c.758T>G; c.757del; c.751G>T; c.749A>C; c.748C>A; c.748C>T; c.746_747del; c.747C>A; c.744_745del; c.734_742dup; c.740C>G; c.739T>C; c.737C>T; c.736del; c.735G>A; c.734G>A; c.733_734insC; c.733del; c.733T>C; c.730G>A; c.730G>C; c.728T>G; c.728T>C; c.728T>A; c.725T>C; c.725T>A; c.723dupT; c.722del; c.718_719delAA; c.719del; c.716_718del; c.718A>T; c.717A>G; c.716T>C; c.713G>A; c.712A>T; c.702_709del; c.708_709delinsAT; c.708G>T; c.708G>A; c.708G>C; c.707G>A; c.707G>T; c.706T>A; c.706T>C; c.704C>G; c.704C>A; c.704C>T; c.700_702del; c.702T>G; c.700G>T; c.695T>C; c.692A>C; c.692A>T; c.692A>G; c.691G>A; c.688G>A; c.687del; c.685T>G; c.683dup; c.680G>A; c.680G>C; c.679C>T; c.678del; c.678G>T; c.678G>A; c.677G>A; c.676T>A; c.676T>C; c.676T>G; c.675C>A; c.674A>G; c.672T>G; c.671del; c.671A>C; c.671A>G; c.670A>G; c.670A>C; c.668G>A; c.668G>C; c.668G>T; c.667T>C; c.667T>G; c.666del; c.666C>A; c.666C>G; c.662_663delAG; c.664T>G; c.661C>T; c.659G>C; c.658C>T; c.657C>G; c.656T>C; c.656T>A; c.655del; c.648T>A; c.647A>G; c.646dup; c.646T>G; c.645T>A; c.644A>G; c.641C>T; c.640-1delG; c.640-1G>A; c.640-1G>T; c.640-1G>C; c.640-2A>G; c.640-2A>T; c.640-2A>C; c.640-3C>G; c.640-11T>A; c.640-394C>T; c.640-801G>A; c.640-814T>C; c.640-859C>T; c.639+5G>A; c.639+5G>T; c.639+4A>T; c.639+2T>C; c.639+1G>C; c.639+1G>A; c.639G>C; c.638del; c.638A>G; c.638A>T; c.634C>T; c.630delC; c.620_630delATATGTGGCCinsCACT; c.627G>A; c.618_627delinsA; c.622_623delAT; c.623T>G; c.623T>A; c.613_621delCCTCTTTAT; c.621dup; c.621T>A; c.618_621delinsAAA; c.620A>C; c.620A>G; c.618_619del; c.619T>C; c.617T>C; c.614del; c.614C>T; c.614C>G; c.613C>A; c.613C>G; c.613C>T; c.612G>T; c.612G>C; c.612G>A; c.611G>A; c.610T>C; c.606_607del; c.607G>A; c.607G>T; c.606del; c.606T>G; c.605G>A; c.605G>C; c.604T>C; c.602C>T; c.602C>A; c.596del; c.596T>C; c.595dup; c.593T>C; c.584_585insAATAG; c.584dup; c.584G>T; c.581C>T; c.579del; c.573del; c.572T>A; c.572T>C; c.568del; c.564del; c.559_564dup; c.562del; c.561G>A; c.561G>T; c.559_560del; c.560T>C; c.560T>G; c.560T>A; c.559A>G; c.554del; c.548-3_553del; c.553A>T; c.551A>G; c.550T>C; c.550T>G; c.550T>A; c.548delG; c.548G>A; c.548G>T; c.548G>C; c.548-1G>A; c.548-1G>T; c.548-1G>C; c.548-2A>G; c.539_547+9del; c.547+4A>G; c.547+1G>A; c.547+1G>T; c.547+1G>C; c.547G>A; c.547G>T; c.539_547delinsC; c.540G>T; c.539del; c.530T>A; c.525_526del; c.522dup; c.522T>A; c.521G>A; c.520del; c.520T>C; c.520T>G; c.519C>A; c.516T>G; c.515G>A; c.515G>C; c.515G>T; c.514T>G; c.514T>C; c.512G>A;</p>
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	<p>c.512G>T; c.511G>A; c.511G>C; c.511G>T; c.509A>G; c.509A>T; c.508G>A; c.508G>C; c.506T>C; c.504A>C; c.503A>G; c.502A>C; c.502A>T; c.500T>C; c.500T>A; c.486_498del; c.496_497delCTinsGG; c.496C>G; c.494A>T; c.494A>G; c.493G>T; c.493G>C; c.491T>G; c.490G>T; c.488del; c.488G>T; c.487G>C; c.487G>T; c.486G>A; c.486G>T; c.486G>C; c.484_485del; c.482_485del; c.484delT; c.485G>A; c.485G>C; c.484T>C; c.484T>G; c.479C>A; c.477del; c.476T>C; c.476T>G; c.469_470del; c.469C>T; c.467C>T; c.467C>A; c.466del; c.466G>A; c.465T>A; c.463G>C; c.461_462insCA; c.460del; c.457_459del; c.454_456del; c.456C>A; c.455A>C; c.455A>G; c.454T>G; c.454T>C; c.453C>G; c.453C>A; c.452del; c.442_450del; c.449del; c.447del; c.444T>G; c.443G>A; c.440G>T; c.439G>A; c.437del; c.436C>T; c.431del; c.431G>T; c.428dup; c.427G>A; c.427G>C; c.426dup; c.426C>G; c.426C>T; c.425G>A; c.425G>T; c.424T>C; c.422C>T; c.422C>A; c.421A>C; c.413dup; c.413del; c.398_413delTTTATGCAGATGTTGGinsCTGC; c.413G>A; c.413G>T; c.412G>A; c.409del; c.408T>G; c.408T>A; c.406G>T; c.406G>C; c.404C>T; c.402T>G; c.402T>A; c.401A>C; c.401A>G; c.400del; c.400T>C; c.400T>G; c.398T>A; c.396del; c.395G>A; c.395G>C; c.394G>A; c.394G>C; c.392T>G; c.392T>C; c.389_390insT; c.386_389dup; c.388A>G; c.386T>C; c.383G>A; c.379A>T; c.370_377del; c.376A>T; c.375del; c.374A>T; c.374A>C; c.371T>G; c.371T>A; c.370del; c.370-1G>A; c.370-1G>T; c.370-2A>G; c.370-2A>C; c.369+4_369+5del; c.369+2T>G; c.369+1G>A; c.369+1G>T; c.369+1G>C; c.361_364delGCTA; c.359_364del; c.363del; c.361G>C; c.359_361delinsCAA; c.361G>A; c.358C>G; c.355C>T; c.337_354del; c.350_355delinsA; c.352C>T; c.351T>G; c.350T>G; c.339del; c.338T>C; c.337T>A; c.337T>C; c.335G>A; c.335G>T; c.334C>T; c.334C>G; c.334C>A; c.330_331del; c.331C>T; c.329del; c.317_327del; c.326A>G; c.323_324insCAGA; c.318_319del; c.319C>T; c.317T>G; c.314_315del; c.313A>T; c.307G>T; c.304_305del; c.305del; c.304del; c.305C>A; c.305C>G; c.297_300del; c.298del; c.299G>A; c.299G>C; c.297_298del; c.298A>T; c.295delC; c.295dupC; c.295C>T; c.290del; c.290C>T; c.289G>C; c.285G>A; c.285G>T; c.284G>C; c.284G>T; c.284G>A; c.283T>C; c.283T>G; c.281G>A; c.281G>C; c.281G>T; c.280T>A; c.280T>C; c.279C>G; c.261_278del; c.278A>G; c.278A>T; c.273_276dupTGAT; c.277G>A; c.277G>T; c.273_276del; c.275A>T; c.275A>G; c.274G>A; c.274G>T; c.274G>C; c.272T>A; c.272T>C; c.270dup; c.270del; c.270C>G; c.270C>A; c.267_268dup; c.269G>A; c.265delC; c.266T>G; c.266T>C; c.266T>A; c.265C>T; c.264C>G; c.263A>G; c.262T>G; c.260del; c.258T>G; c.257A>T; c.257A>G; c.256del; c.256T>G; c.256T>C; c.254G>A; c.254G>C; c.254G>T; c.253G>A; c.244A>T; c.243_244delinsAT; c.243G>T; c.243G>C; c.243G>A; c.242G>A; c.242G>C; c.241T>C; c.239G>A; c.237del; c.235G>T; c.233C>G; c.224_228delinsCA; c.227T>C; c.227T>G; c.218C>T; c.218C>A; c.215del; c.215T>G; c.214del; c.214A>G; c.207del; c.207C>A; c.206T>C; c.205T>C; c.203T>C; c.202C>T; c.197A>G; c.196G>A; c.195-1G>C; c.195-1G>T; c.195-1G>A; c.195-2A>G; c.194+2T>C; c.194+1G>A; c.194+1G>T; c.194G>C; c.194G>T; c.190A>T; c.188G>A; c.188G>C; c.187T>A; c.187T>G; c.184dup; c.184del; c.181_182dup; c.178C>T; c.176del; c.176A>T; c.175G>A; c.172delG; c.167_172del; c.164_171delACTGCCAGinsTCTGCCTA; c.165_170del; c.170A>C; c.169C>T; c.168C>A; c.167G>A; c.167G>T; c.166T>G; c.162del; c.161T>C; c.157_160del; c.160C>T; c.158del; c.157_158delinsC; c.156C>G; c.156C>A; c.154dup; c.155G>A; c.155G>C; c.155G>T; c.154del; c.154T>C; c.154T>G; c.154T>A; c.153G>A; c.153G>T; c.153G>C; c.152T>A; c.151_152delinsG; c.149T>G; c.147_148insCCC; c.146G>C; c.146G>T; c.145C>G; c.145C>A; c.145C>T; c.144G>C; c.144G>T; c.135_142del; c.143A>G; c.142G>A; c.141G>C; c.141G>T; c.141G>A; c.140G>T; c.139dup; c.140G>A; c.139T>C; c.139T>G; c.125_137delTGGGCTGGCTGCA; c.134_138delinsGCTCG; c.137A>G; c.137A>T; c.137A>C; c.136C>T; c.134T>C; c.128_132del; c.132G>A; c.132G>T; c.131G>A; c.130T>G; c.128delG; c.128G>T; c.128G>A; c.127G>A; c.127G>C; c.123_126dup; c.126G>C; c.126G>A; c.124_125del; c.125T>C; c.125T>G; c.125T>A; c.124A>C; c.124A>G; c.123del; c.122C>T; c.116_119del; c.119C>G; c.119C>T; c.119C>A; c.118C>T; c.116C>G; c.114del; c.109del; c.110C>T; c.109G>A; c.109G>C; c.109G>T; c.108G>C; c.108G>T; c.107T>G; c.107T>C; c.101_105del; c.104G>A; c.104G>T; c.103G>A; c.103G>C; c.102T>A; c.102T>G; c.101A>G; c.100A>C; c.98A>G; c.97G>T; c.97G>C; c.95T>C; c.95T>A; c.92C>T; c.85dupG; c.85del; c.59_84del; c.80delC; c.77dup; c.73del; c.59_73del; c.59_72dupCCCTCGTTCTCTGG; c.59_72del; c.72G>A; c.71G>A; c.62T>C; c.62T>G; c.51_49delGCTGGCGCTTGG; c.44C>A; c.44C>G; c.43G>C; c.34_42del; c.41T>C; c.36C>G; c.32del; c.26delA; c.25del; c.9_19del; c.19G>T; c.18del; c.4C>T; c.3G>A; c.1_2del; c.2T>C; c.2T>A; c.1A>G; c.1A>C.</p>
<p>G6PD NM_001360016.2 (252)</p>	<p>c.1490_1492del; c.1478_1479insAGGAGGCAGA; c.1466C>T; c.1465C>T; c.1463G>T; c.1457+7_1458-43del; c.1442C>G; c.1441C>G; c.1438A>T; c.1400C>G; c.1388G>T; c.1388G>A; c.1387C>T; c.1387C>A; c.1381G>A; c.1380G>C; c.1378G>T; c.1376del; c.1376G>A; c.1376G>T; c.1376G>C; c.1375C>G; c.1375C>T; c.1372C>T; c.1367A>T; c.1366G>C; c.1366G>A; c.1361G>T; c.1361G>C; c.1361G>A; c.1360C>T; c.1358T>A; c.1354_1358del; c.1351C>T; c.1347G>C; c.1346A>G; c.1342A>G; c.1340G>T; c.1339G>A; c.1330G>A; c.1318C>T; c.1315C>T; c.1292T>G; c.1291G>A; c.1288-2A>T; c.1288-2A>G; c.1285A>G; c.1284C>A; c.1283dup; c.1246G>A; c.1229G>C; c.1229G>A; c.1228G>T; c.1226C>G; c.1226C>A; c.1225C>T; c.1220A>C; c.1215G>A; c.1205C>A; c.1193A>G; c.1192G>A; c.1187C>G; c.1187C>T; c.1186C>G; c.1183C>T; c.1180G>C; c.1178G>A; c.1177C>G; c.1177C>T; c.1175T>C; c.1166A>G; c.1160G>A; c.1159C>T; c.1156A>G; c.1155C>G; c.1154G>T; c.1153T>G; c.1153T>C; c.1143C>G; c.1141T>C; c.1139T>C; c.1138A>G; c.1132G>A; c.1118T>C; c.1102G>A; c.1084_1101del; c.1094G>A; c.1089C>G; c.1089C>A; c.1088A>T; c.1082C>T; c.1081G>A; c.1067T>A; c.1063A>G; c.1057C>T; c.1054dup; c.1052G>T; c.1048G>C; c.1030G>A; c.1028_1029del; c.1024C>T; c.1006A>G; c.1004C>A; c.1003G>A; c.1000_1002del; c.995C>T; c.982G>A; c.957_980del; c.973G>A; c.968T>C; c.964T>C; c.962G>T; c.962G>A; c.949G>A; c.946G>A; c.921G>C; c.920A>C; c.916G>A; c.910G>T; c.882del; c.871G>A; c.864+44del; c.864+43G>A; c.854G>A; c.853C>T; c.851T>C; c.848A>G; c.848A>T; c.844G>T; c.844G>C; c.835A>G; c.835A>T; c.833C>T; c.832T>C; c.827C>T; c.826C>T; c.825G>C; c.821A>T; c.820G>A; c.811G>C; c.806G>A; c.770G>T; c.770G>A; c.769C>G; c.743G>A; c.735_741del; c.739G>A; c.737G>T; c.725_730del; c.713A>G; c.703C>T; c.697G>C; c.695G>A; c.691G>A; c.691G>C; c.686_688del; c.680G>T; c.680G>A; c.679C>G; c.679C>T; c.660C>G; c.655A>G; c.648T>G; c.637G>C; c.634A>G; c.611G>A; c.598G>A; c.595A>G; c.593G>A; c.593G>C; c.592C>A; c.592C>T; c.586A>T; c.585G>C; c.573C>G; c.565_567del; c.563C>T; c.551C>G; c.551C>T; c.542A>T; c.536G>A; c.527A>G; c.520_522del; c.519C>G; c.517T>C; c.514C>T; c.497G>A; c.496C>T; c.493A>G; c.488G>A; c.487G>A; c.486-17del; c.486-17C>T; c.473G>A; c.464A>C; c.463C>G; c.448G>A; c.442G>A; c.430C>G; c.412T>G; c.409C>T; c.407G>A; c.406C>T; c.404A>G; c.404A>C; c.403A>C; c.392G>T; c.383T>G; c.383T>C; c.371A>G; c.356A>C; c.354C>G; c.353A>C; c.352T>C; c.323T>A; c.312C>T; c.305T>C; c.290A>T; c.283_285del; c.274C>T; c.242G>A; c.241C>T; c.235G>C; c.227C>A; c.224T>C; c.219C>T; c.209A>G; c.208T>C; c.202G>A; c.197T>G; c.196T>A; c.193A>G; c.187G>A; c.185C>T; c.185C>A; c.179T>C; c.170G>A; c.152C>T; c.148C>T; c.143T>C; c.131C>G; c.130G>A; c.108_110del; c.95A>G; c.94C>G; c.94C>T; c.49C>T; c.34G>T; c.31C>T; c.7G>A; c.7C>T; c.-9+2T>G.</p>

<p>OTC NM_000531.6 (476)</p>	<p>c.-157T>G; c.-142G>A; c.-139A>G; c.-106C>A; c.1A>G; c.1A>T; c.2T>C; c.3G>A; c.29_32delACAA; c.42delT; c.50_53del; c.53delA; c.67C>T; c.72_77+18del; c.77G>A; c.77G>C; c.77G>T; c.77+1G>A; c.77+1G>T; c.77+2dup; c.77+3_77+6del; c.77+4A>C; c.77+5G>C; c.78-3C>G; c.78-2A>G; c.78-1G>C; c.78-1G>T; c.85C>T; c.94C>T; c.106C>T; c.115G>T; c.116G>A; c.116G>C; c.116G>T; c.118C>T; c.119G>A; c.119G>T; c.122A>G; c.125T>C; c.127C>T; c.128T>C; c.131C>T; c.133C>G; c.134T>C; c.140delA; c.134T>G; c.140_141insG; c.140A>T; 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.156A>T; c.158T>C; c.158T>G; c.163T>G; c.167T>C; c.170T>A; c.174G>A; c.177dup; c.177dupA; c.179C>T; c.179C>G; c.179C>A; c.188T>C; c.190A>T; c.200T>G; c.205C>T; c.208A>T; c.214G>A; c.216+1delG; c.216+1G>A; c.216+1G>T; c.217-2dupA; c.217-2A>G; c.217-1G>A; c.227T>C; c.228A>C; c.231G>T; c.232C>T; c.236G>A; c.238A>G; c.240G>T; c.241T>C; c.243_245delCTT; c.245T>G; c.247G>C; c.248G>A; c.258dup; c.254T>C; c.261_262del; c.259G>A; c.262A>T; 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.278C>G; c.281G>C; c.284T>C; c.286T>C; c.298+1_298+5delGTAAG; c.298+1delG; c.298G>T; c.298+1G>A; c.298+1G>T; c.298+2T>C; c.298+2T>G; c.299-2A>G; c.299-1G>A; c.299G>A; c.303del; c.305C>A; c.314G>T; c.316G>A; c.317G>A; c.317G>T; c.330delT; c.327T>A; c.332T>C; c.350A>G; c.350A>T; c.350A>C; c.359_360delTTG; c.364_365insTT; c.365A>G; c.376delG; c.376G>A; c.377A>G; c.378C>A; c.386G>A; c.386G>C; c.386G>T; c.386+1G>A; c.386+1G>T; c.386+2T>C; c.386+5G>A; c.386+7663T>G; c.387-2A>C; c.387-2A>G; c.387-2A>T; c.387-1G>A; c.390_392dupATT; c.391_397dupTTGTCTA; c.392T>C; c.394T>C; c.395C>T; c.395C>A; c.403delG; c.403G>C; c.404C>A; c.407A>T; c.409G>A; c.416T>C; c.418G>C; c.419C>T; c.421C>G; c.421C>T; c.422G>A; c.422G>C; c.425T>A; c.429T>A; c.430A>T; c.437C>G; c.437C>A; 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.477T>G; c.478A>T; c.479T>A; c.479T>C; c.479T>G; c.480C>G; c.481A>G; c.482A>G; c.484G>A; c.484G>C; c.485G>C; c.485G>A; c.485G>T; c.488T>A; c.488T>G; c.490T>C; c.491C>G; c.491C>T; c.493G>C; c.493G>T; c.494A>G; c.501C>A; c.501C>G; c.502C>T; c.503A>C; c.503A>G; c.504T>A; c.505C>T; c.505C>G; c.506C>T; c.514A>T; c.516C>G; c.517C>G; c.520G>C; c.523G>C; c.523G>T; c.524A>G; c.524A>T; c.526T>C; c.527A>G; c.532_537delACGCTC; c.533C>T; c.535C>G; c.535C>T; c.536T>C; c.539A>C; c.540G>C; c.540+1G>C; c.540+1G>T; c.540+1G>A; c.540+2T>A; c.540+2T>C; c.540+265G>A; c.541-2A>G; c.542A>G; c.545A>T; c.547T>G; c.548A>G; c.562_563delGG; c.562G>C; c.562G>A; c.563G>T; c.563G>C; c.568dupA; c.571C>T; c.576C>G; c.577T>C; c.577T>G; c.578G>A; c.579G>T; c.586delG; c.583G>A; c.584G>C; c.586G>A; c.586G>T; c.587A>T; c.588T>A; c.589G>A; c.589G>T; c.590G>A; c.593A>T; c.593A>G; c.594C>A; c.595A>G; c.596A>G; c.596A>T; c.602T>C; c.604C>T; c.604C>G; c.605A>C; c.607T>C; c.609del; c.608C>T; c.608C>G; c.613A>G; c.614T>C; c.614T>A; c.615G>C; c.617T>A; c.617T>G; c.618G>T; c.618G>A; c.618G>C; c.620G>A; c.621C>A; c.622G>T; c.622G>A; c.626C>A; c.626C>T; c.628A>C; c.628A>G; c.629A>T; c.629A>C; c.634G>T; c.635G>T; c.640C>T; c.643C>T; c.645dupT; c.646C>G; c.650C>A; c.653_663del; c.652G>C; c.658C>G; c.659C>T; c.663G>A; c.663G>C; c.663G>T; c.663+1G>A; c.663+1G>T; c.663+2T>C; c.665delG; c.664-1G>A; c.673C>A; c.673C>T; c.674C>A; c.674C>G; c.674C>T; c.698C>T; c.700G>T; c.709del; c.709_712delinsCCT; c.709_712delGCCAinsCCT; c.714dup; c.716A>G; c.716A>T; c.717G>A; c.717G>C; c.717G>T; c.717+1G>A; c.717+1G>T; c.717+2T>C; c.717+3A>G; c.717+8_717+23del; c.718-2_731del; c.718-2A>G; c.725C>T; c.731_739delTGTTGCTGA; c.731T>A; c.731T>C; c.740C>A; c.740C>G; c.749dup; c.749dupC; c.757G>A; c.757G>C; c.759delA; c.758C>A; c.764A>C; c.766G>T; c.779T>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.803T>A; c.804G>C; c.806G>A; c.808del; c.808C>T; c.809A>C; c.813_815del; c.817_819delGAG; c.814G>T; c.817_819del; c.818delA; c.822del; c.826A>T; c.829C>T; c.830G>A; c.830G>T; c.833_864delinsCC; c.835C>T; c.844del; c.848del; c.853C>T; c.860_861dupCA; c.865_866del; c.867G>T; c.867+1G>A; c.867+1G>T; c.867+1126A>G; c.868-2A>T; c.876delA; c.882delT; c.890_893delACTG; c.892_893delTTG; c.893G>C; c.903A>T; c.904C>T; c.904C>G; c.905A>G; c.905A>T; c.906delC; c.906C>G; c.907T>C; c.907T>G; c.908G>A; c.912G>T; c.914C>A; c.914C>G; c.919A>G; c.929_931del; c.928G>T; c.929A>G; c.929A>T; c.931G>A; c.941_943delAAG; c.943G>T; c.943G>A; c.944T>A; c.944T>C; c.944T>G; c.946T>G; c.947T>C; c.953C>T; c.958C>T; c.959G>C; c.959G>T; c.961T>C; c.962C>A; c.962C>T; c.970_979del; c.976G>A; c.982G>T; c.988A>G; c.991A>T; c.994T>A; c.995G>A; c.996G>A; c.1005G>A; c.1005+1G>T; c.1005+2T>C; c.1006-3C>G; c.1006-2A>G; c.1006-1G>A; c.1006G>T; c.1009G>C; c.1009G>T; c.1015G>A; c.1015G>C; c.1018T>C; c.1020_1028del; c.1019C>T; c.1022T>C; c.1027A>G; c.1028C>T; c.1028C>A; c.1028C>G; c.1033T>C; c.1033T>G; c.1034A>G; c.1034A>T; c.1039C>A; c.1039C>T; c.1042C>T; c.1052delA; c.1061T>G; c.1063T>G.</p>
<p>FMR1</p>	<p>Espansione triplette CGG</p>
<p>SMN1</p>	<p>Delezione dell'esone 7</p>

*La regione chr15:72668198-72668313 nel gene *HEXA* non può essere rilevata.