

protein_variant_in_BIOPKU	cDNA_Aberration	apv	Phenotype
p.M1V	c.1A>G	0	cPKU
p.M1L	c.1A>T	0	cPKU
p.M1R	c.2T>G	0	cPKU
p.M1T	c.2T>C	0	cPKU
p.M1I	c.3G>A	0	cPKU
p.V5Sfs*3	c.13del	0	cPKU
p.N8Ifs*30	c.23_24delinsT	0	cPKU
p.N8Tfs*30	c.23del	0	cPKU
p.R13Qfs*5	c.30dup	0	cPKU
p.L11*	c.32T>A	0	cPKU
p.R13Gfs*25	c.36del	0	cPKU
p.L15Qfs*24	c.43_44insAG	0	cPKU
p.P15Pfs*24	c.43_44insCT	0	cPKU
p.S16Lfs*22	c.46del	0	cPKU
p.S16*	c.47_48del	0	cPKU
p.D17Lfs*22	c.47_48dup	0	cPKU
p.D17*	c.48dup	0	cPKU
p.D17Lfs*22	c.47_48insCT	0	cPKU
p.Q20Kfs*17	c.57_60+1del	0	cPKU
p.Q20*	c.58C>T	0	cPKU
p.Q20Pfs*7	c.59delinsCC	0	cPKU
IVS1+1G>C	c.60+1G>C	0	cPKU
IVS1+4A>T	c.60+4A>T	0	cPKU
IVS1+5G>A	c.60+5G>A	0	cPKU
IVS1+5G>T	c.60+5G>T	0	cPKU
IVS1+5G>C	c.60+5G>C	0	cPKU
IVS1+5_+6del	c.60+5_60+6del	0	cPKU
IVS1-13del9	c.61-13del	0	cPKU
IVS1-1G>A	c.61-1G>A	0	cPKU
p.E26Lfs*13	c.73_74dup	0	cPKU
p.I25Mfs*13	c.75del	0	cPKU
p.I25Mfs*13	c.74del	0	cPKU
p.C29*	c.87C>A	0	cPKU
p.S36*	c.107C>A	0	cPKU
p.I38Dfs*19	c.111dup	0	cPKU
p.S40L	c.119C>T	0	cPKU
p.S40*	c.119C>A	0	cPKU

p.L41Qfs*15	c.120_121del	0	cPKU
p.L41F	c.121C>T	0	cPKU
p.K42del	c.124_126del	0	cPKU
p.E43*	c.127G>T	0	cPKU
p.E43Kfs*18	c.127del	0	cPKU
p.G46Vfs*15	c.137del	0	cPKU
p.K50*	c.148A>T	0	cPKU
p.L52Cfs*9	c.155del	0	cPKU
p.E54Tfs*9	c.160_163del	0	cPKU
p.F55Rfs*5	c.162_165del	0	cPKU
p.F55del	c.163_165del	0	cPKU
p.F55Yfs*3	c.163_164insATAT	0	cPKU
p.F55Lfs*6	c.165del	0	cPKU
p.E56Rfs*5	c.166del	0	cPKU
p.E57Rfs*3	c.168_168+1delinsAA	0	cPKU
IVS2+1G>A	c.168+1G>A	0	cPKU
IVS2+1G>T	c.168+1G>T	0	cPKU
IVS2+1G>C	c.168+1G>C	0	cPKU
IVS2+2T>A	c.168+2T>A	0	cPKU
IVS2+5G>A	c.168+5G>A	0	cPKU
IVS2+5G>C	c.168+5G>C	0	cPKU
IVS2+5G>T	c.168+5G>T	0	cPKU
IVS2+6T>G	c.168+6T>G	0	cPKU
IVS2-13T>G	c.169-13T>G	0	cPKU
IVS2-2A>G	c.169-2A>G	0	cPKU
IVS2-2A>T	c.169-2A>T	0	cPKU
IVS2-1G>A	c.169-1G>A	0	cPKU
p.E57*	c.169G>T	0	cPKU
p.E57Gfs*5	c.169_170insGA	0	cPKU
p.E57Pfs*5	c.169_182del	0	cPKU
p.E57Mfs*3	c.169_172del	0	cPKU
p.E57Cfs*77	c.169_352del	0	cPKU
p.E57Cfs*77	c.170_352+1del	0	cPKU
p.N58*	c.171_172del	0	cPKU
p.D58_L62del	c.173_187del	0	cPKU
p.L62*	c.184del	0	cPKU
p.L62Pfs*6	c.184_185insCTGA	0	cPKU
p.L62Pfs*3	c.185_189del	0	cPKU

p.H64Dfs*4	c.185_188dup	0	cPKU
p.T63_H64delinsPN	c.187_190delinsCCCA	0	cPKU
p.H64Pfs*10	c.189_190dup	0	cPKU
p.H64Tfs*9	c.190del	0	cPKU
p.H64*	c.190_194del	0	cPKU
p.I65Kfs*9	c.194_201del	0	cPKU
p.E66*	c.196G>T	0	cPKU
p.E66Afs*8	c.197_204del	0	cPKU
p.E66Dfs*8	c.198_205del	0	cPKU
p.E66Dfs*8	c.209_211del	0	cPKU
p.P69_S70dup	c.206_211dup	0	cPKU
p.S70Ffs*7	c.206dup	0	cPKU
p.S70P	c.208T>C	0	cPKU
p.R71Vfs*2	c.210del	0	cPKU
p.K73Qfs*4	c.216_217insC	0	cPKU
p.E76*	c.226G>T	0	cPKU
p.Y77*	c.231T>G	0	cPKU
p.E78Ffs*13	c.232_235del	0	cPKU
p.E78Vfs*13	c.233_236del	0	cPKU
p.F79Ifs*7	c.234dup	0	cPKU
p.T81Hfs*5	c.239dup	0	cPKU
p.T81Vfs*6	c.241_256del	0	cPKU
p.L83Wfs*9	c.248del	0	cPKU
p.D84Vfs*8	c.251del	0	cPKU
p.K85*	c.253A>T	0	cPKU
p.R86Lfs*6	c.257del	0	cPKU
p.A90Cfs*12	c.266dup	0	cPKU
p.A90Cfs*12	c.266_267insG	0	cPKU
p.Q93Sfs*5	c.277_280del	0	cPKU
p.N93Kfs*5	c.279_282del	0	cPKU
p.I94Sfs*4	c.280_283del	0	cPKU
p.A104_V106del	c.310_318del	0	cPKU
p.A104V	c.311C>T	0	cPKU
p.E108*	c.322G>T	0	cPKU
p.E108Dfs*4	c.324_328del	0	cPKU
p.S110Yfs*85	c.329del	0	cPKU
p.R111*	c.331C>T	0	cPKU
p.D112Efs*2	c.335dup	0	cPKU

p.K113*	c.337A>T	0	cPKU
p.K115Rfs*81	c.338_341del	0	cPKU
p.K115Tfs*79	c.344_347del	0	cPKU
p.D116Hfs*29	c.346_347del	0	cPKU
p.T117Kfs*78	c.350del	0	cPKU
p.T117S*28	c.350_351del	0	cPKU
IVS3+1G>A	c.352+1G>A	0	cPKU
IVS3-2A>G	c.353-2A>G	0	cPKU
IVS3-1G>A	c.353-1G>A	0	cPKU
IVS3-1G>C	c.353-1G>C	0	cPKU
IVS3-1G>T	c.353-1G>T	0	cPKU
p.W120Gfs*75	c.354del	0	cPKU
p.W120Gfs*75	c.357del	0	cPKU
p.W120Gfs*75	c.358del	0	cPKU
p.W120*	c.359G>A	0	cPKU
p.P122Q	c.365C>A	0	cPKU
p.P122Qfs*73	c.365del	0	cPKU
p.P122Lfs*73	c.365_366delinsT	0	cPKU
p.R123Efs*72	c.367del	0	cPKU
p.Q126*	c.376C>T	0	cPKU
p.N133Rfs*61	c.398_401del	0	cPKU
p.Q134*	c.400C>T	0	cPKU
p.E141*	c.421G>T	0	cPKU
p.D145Tfs*50	c.433del	0	cPKU
p.P147S	c.439C>T	0	cPKU
p.P147Lfs*48	c.440del	0	cPKU
IVS4+1delGT	c.441+1del	0	cPKU
IVS4+1G>A	c.441+1G>A	0	cPKU
IVS4+1G>C	c.441+1G>C	0	cPKU
IVS4+1G>T	c.441+1G>T	0	cPKU
IVS4+2T>G	c.441+2T>G	0	cPKU
IVS4+2T>A	c.441+2T>A	0	cPKU
IVS4+2T>C	c.441+2T>C	0	cPKU
IVS4+3G>C	c.441+3G>C	0	cPKU
IVS4+4A>G	c.441+4A>G	0	cPKU
IVS4+5G>T	c.441+5G>T	0	cPKU
IVS4+6T>C	c.441+6T>C	0	cPKU
IVS4+6T>A	c.441+6T>A	0	cPKU

IVS4-3T>G	c.442-3T>G	0	cPKU
IVS4-2A>C	c.442-2A>C	0	cPKU
IVS4-2A>G	c.442-2A>G	0	cPKU
IVS4-1G>A	c.442-1G>A	0	cPKU
IVS4-1G>C	c.442-1G>C	0	cPKU
IVS4_Ex7del	c.442+?_842-?del	0	cPKU
p.G148Lfs*105	c.442-?_706+?del	0	cPKU
p.G148V	c.443G>T	0	cPKU
p.G148Wfs*29	c.442-2914_509+1173del4155ins268	0	cPKU
p.D151RfsTer13	c.450dup	0	cPKU
p.D151Ifs*44	c.451del	0	cPKU
p.P152Lfs*43	c.453del	0	cPKU
p.R155Lfs*43	c.456_463dup	0	cPKU
p.Y154*	c.462C>A	0	cPKU
p.R155Vfs*40	c.463del	0	cPKU
p.R155P	c.464G>C	0	cPKU
p.A156Efs*39	c.467del	0	cPKU
p.R157K	c.470G>A	0	cPKU
p.R157S	c.471A>C	0	cPKU
p.R158W	c.472C>T	0	cPKU
p.R158Q	c.473G>A	0	cPKU
p.K159*	c.475A>T	0	cPKU
p.Q160*	c.478C>T	0	cPKU
p.A165T	c.493G>A	0	cPKU
p.A165Vfs*35	c.493_494insT	0	cPKU
p.A165Tfs*29	c.493_496del	0	cPKU
p.Y166*	c.498C>A	0	cPKU
p.Y166*	c.498C>G	0	cPKU
p.Y168Sfs*27	c.503del	0	cPKU
p.Y168*	c.504C>A	0	cPKU
p.R169Pfs*26	c.506del	0	cPKU
IVS5+1delG	c.509+1del	0	cPKU
IVS5+1G>A	c.509+1G>A	0	cPKU
IVS5+1G>C	c.509+1G>C	0	cPKU
IVS5+5del	c.509+5del	0	cPKU
p.G148Wfs*29	c.442-1377_509+226del	0	cPKU
p.G148Wfs*29	c.442-1463_509+257del	0	cPKU
IVS5+781del	c.442-102_509+781del	0	cPKU

p.G148Wfs*29	c.442-4403_509+883del	0	cPKU
p.H170Qfs*37	c.510-735_912+434del	0	cPKU
IVS5-6T>G	c.510-6T>G	0	cPKU
IVS5-6T>A	c.510-6T>A	0	cPKU
IVS5-2A>G	c.510-2A>G	0	cPKU
IVS5-1G>A	c.510-1G>A	0	cPKU
p.H170Q	c.510T>A	0	cPKU
Ex6dup	c.510_706dup	0	cPKU
p.H170Q	c.510T>G	0	cPKU
p.G171A	c.512G>C	0	cPKU
p.Q172*	c.514C>T	0	cPKU
p.R176*	c.526C>T	0	cPKU
p.Y179H	c.535T>C	0	cPKU
p.Y179Hfs*20	c.535_536del	0	cPKU
p.M180Ifs*18	c.540_544del	0	cPKU
p.E181Kfs*13	c.541_544del	0	cPKU
p.E183del	c.543_545del	0	cPKU
p.E183*	c.547G>T	0	cPKU
p.K184Rfs*11	c.551del	0	cPKU
p.T186Hfs*9	c.556del	0	cPKU
p.W187Gfs*8	c.558del	0	cPKU
p.W187Gfs*12	c.558_559del	0	cPKU
p.W187R	c.559T>C	0	cPKU
p.W187R	c.559T>A	0	cPKU
p.W187*	c.560G>A	0	cPKU
p.W187*	c.561G>A	0	cPKU
p.G188Afs*7	c.563del	0	cPKU
p.V190G	c.569T>G	0	cPKU
p.F191Lfs*4	c.573del	0	cPKU
p.K192*	c.574_580del	0	cPKU
p.L194Efs*5	c.580_581del	0	cPKU
p.L194Dfs*6	c.580delinsGA	0	cPKU
p.S196Vfs*4	c.584dup	0	cPKU
p.S196Lfs*2	c.586_608del	0	cPKU
p.L197*	c.590T>A	0	cPKU
p.L197*	c.590_612del	0	cPKU
p.Y198Vfs*9	c.591_613del	0	cPKU
p.Y198Sfs*136	c.592_613del	0	cPKU

p.Y198Cfs*136	c.593_614del	0	cPKU
p.Y198*	c.594T>G	0	cPKU
p.T200Nfs*6	c.598dup	0	cPKU
p.H201Qfs*5	c.602dup	0	cPKU
p.H201Q	c.603T>A	0	cPKU
p.C203Lfs*3	c.607dup	0	cPKU
p.C203G	c.607T>G	0	cPKU
p.C203*	c.609C>A	0	cPKU
p.Y204Lfs*2	c.610dup	0	cPKU
p.Y204_T236delinsS	c.611A>G	0	cPKU
p.Y204*	c.612T>G	0	cPKU
p.E205K	c.613G>A	0	cPKU
p.Y206*	c.618C>A	0	cPKU
p.Y206*	c.618C>G	0	cPKU
p.I209Tfs*6	c.625_626insC	0	cPKU
p.P211Hfs*130	c.632del	0	cPKU
p.L212Tfs*3	c.632dup	0	cPKU
p.L212P	c.635T>C	0	cPKU
p.E214Kfs*127	c.636del	0	cPKU
p.E214*	c.640G>T	0	cPKU
p.Y216*	c.648C>G	0	cPKU
p.G218Afs*123	c.653del	0	cPKU
p.D222*	c.664_665del	0	cPKU
p.Q226Tfs*118	c.667_674dup	0	cPKU
p.N223Kfs*118	c.669del	0	cPKU
p.I224T	c.671T>C	0	cPKU
p.P225T	c.673C>A	0	cPKU
p.Q226*	c.676C>T	0	cPKU
p.Q226K	c.676C>A	0	cPKU
p.Q226Sfs*115	c.676del	0	cPKU
p.Q226Pfs*112	c.677_686del	0	cPKU
p.E228*	c.682G>T	0	cPKU
p.D229Efs*54	c.686dup	0	cPKU
p.S231Vfs*52	c.690_691insG	0	cPKU
p.S231F	c.692C>T	0	cPKU
p.Q232Ifs*50	c.693_694del	0	cPKU
p.Q232Nfs*109	c.693del	0	cPKU
p.Q232*	c.694C>T	0	cPKU

p.Q232del	c.694_696del	0	cPKU
p.Q235*	c.703C>T	0	cPKU
IVS6+1G>T	c.706+1G>T	0	cPKU
IVS6+2T>A	c.706+2T>A	0	cPKU
IVS6+368T>C	c.706+368T>C	0	cPKU
IVS6+629A>C	c.706+629A>C	0	cPKU
IVS6-2A>G	c.707-2A>G	0	cPKU
IVS6-2del	c.707-2del	0	cPKU
IVS6-2A>T	c.707-2A>T	0	cPKU
IVS6-1G>A	c.707-1G>A	0	cPKU
IVS6-1G>C	c.707-1G>C	0	cPKU
p.T238P	c.712A>C	0	cPKU
p.G239A	c.716G>C	0	cPKU
p.G239D	c.716G>A	0	cPKU
p.R241Pfs*100	c.722del	0	cPKU
p.R241Pfs*5	c.722_735delinsCCTCCGACCTGTA	0	cPKU
p.L242F	c.724C>T	0	cPKU
p.L242Sfs*99	c.724del	0	cPKU
p.R243*	c.727C>T	0	cPKU
p.R243L	c.728G>T	0	cPKU
p.P244S	c.730C>T	0	cPKU
p.V245E	c.734T>A	0	cPKU
p.V245Afs*34	c.734_744del	0	cPKU
p.A246Vfs*95	c.737del	0	cPKU
p.G247D	c.740G>A	0	cPKU
p.G247V	c.740G>T	0	cPKU
p.G247Afs*94	c.740del	0	cPKU
p.L248Rfs*93	c.743del	0	cPKU
p.L249Ffs*92	c.745del	0	cPKU
p.L249P	c.746T>C	0	cPKU
p.R252Gfs*30	c.753_754del	0	cPKU
p.R252Gfs*89	c.753del	0	cPKU
p.R252G	c.754C>G	0	cPKU
p.R252W	c.754C>T	0	cPKU
p.R252P	c.755G>C	0	cPKU
p.R252Q	c.755G>A	0	cPKU
p.L255S	c.764T>C	0	cPKU
p.G257V	c.770G>T	0	cPKU

p.G257D	c.770G>A	0	cPKU
p.A259T	c.775G>A	0	cPKU
p.A259V	c.776C>T	0	cPKU
p.R261*	c.781C>T	0	cPKU
p.R261G	c.781C>G	0	cPKU
p.R261Efs*80	c.781del	0	cPKU
p.F263L	c.787T>C	0	cPKU
p.F263L	c.789C>G	0	cPKU
p.F263L	c.789C>A	0	cPKU
p.H264Tfs*77	c.790del	0	cPKU
p.C265Y	c.794G>A	0	cPKU
p.C265*	c.795C>A	0	cPKU
p.T266E	c.796_797delinsGA	0	cPKU
p.Q267*	c.799C>T	0	cPKU
p.Q267R	c.800A>G	0	cPKU
p.Q267Hfs*74	c.801del	0	cPKU
p.Y268C	c.803A>G	0	cPKU
p.Y268*	c.804C>A	0	cPKU
p.I269Tfs*72	c.806del	0	cPKU
p.R270K	c.809G>A	0	cPKU
p.H271fs*10	c.810_814del	0	cPKU
p.R270S	c.810A>T	0	cPKU
p.H271L	c.812A>T	0	cPKU
p.G272*	c.814G>T	0	cPKU
p.K274Nfs*5	c.822_832del	0	cPKU
p.M276Pfs*66	c.823_825delinsCCCCC	0	cPKU
p.Y277C	c.830A>G	0	cPKU
p.T278Yfs*5	c.830_831insC	0	cPKU
p.T278I	c.833C>T	0	cPKU
p.T278N	c.833C>A	0	cPKU
p.E280-IVS7+3fsdel8	c.838del8bp(842+4del)???	0	cPKU
p.E280Nfs*61	c.837del	0	cPKU
p.E280Rfs*3	c.837dup	0	cPKU
p.E280Nfs*61	c.833del	0	cPKU
p.E280_IVS7+3fs	c.838_842+3del	0	cPKU
p.E280K	c.838G>A	0	cPKU
p.E280Dfs*3	c.839_840insT	0	cPKU
p.E280G	c.839A>G	0	cPKU

p.E280A	c.839A>C	0	cPKU
p.P281S	c.841C>T	0	cPKU
p.P281A	c.841C>G	0	cPKU
p.P281fs	c.841_842+1del	0	cPKU
p.P281L	c.842C>T	0	cPKU
IVS7+1G>A	c.842+1G>A	0	cPKU
IVS7+1G>T	c.842+1G>T	0	cPKU
IVS7+2T>A	c.842+2T>A	0	cPKU
IVS7+2T>C	c.842+2T>C	0	cPKU
IVS7+3G>C	c.842+3G>C	0	cPKU
IVS7+4A>G	c.842+4A>G	0	cPKU
IVS7+6T>A	c.842+6T>A	0	cPKU
IVS7-13_843-10del	c.843-13_843-10del	0	cPKU
IVS7-4del	c.843-4del	0	cPKU
IVS7-2A>T	c.843-2A>T	0	cPKU
IVS7-2A>G	c.843-2A>G	0	cPKU
IVS7-1G>A	c.843-1G>A	0	cPKU
p.D282*	c.843dup	0	cPKU
p.D282N	c.844G>A	0	cPKU
p.I283F	c.847A>T	0	cPKU
p.I283N	c.848T>A	0	cPKU
p.C284R	c.850T>C	0	cPKU
p.C284*	c.852C>A	0	cPKU
p.E286K	c.856G>A	0	cPKU
p.H290L	c.869A>T	0	cPKU
p.P292Ffs*21	c.873_880del	0	cPKU
p.L293Cfs*48	c.878del	0	cPKU
p.S295*	c.884C>G	0	cPKU
p.D296G	c.887A>G	0	cPKU
p.F299del	c.895_897del	0	cPKU
p.F299C	c.896T>G	0	cPKU
p.Q301*	c.901C>T	0	cPKU
p.Q301P	c.902A>C	0	cPKU
p.S303Pfs*38	c.907del	0	cPKU
p.F302fs*39	c.904del	0	cPKU
p.Q304*	c.910C>T	0	cPKU
p.Q304P	c.911A>C	0	cPKU
IVS8+1G>A	c.912+1G>A	0	cPKU

IVS8+1G>T	c.912+1G>T	0	cPKU
IVS8+1G>C	c.912+1G>C	0	cPKU
IVS8+2T>C	c.912+2T>C	0	cPKU
IVS8+2T>A	c.912+2T>A	0	cPKU
IVS8+3A>C	c.912+3A>C	0	cPKU
IVS8-8A>G	c.913-8A>G	0	cPKU
IVS8-7A>G	c.913-7A>G	0	cPKU
IVS8-5T>G	c.913-5T>G	0	cPKU
IVS8-2A>G	c.913-2A>G	0	cPKU
IVS8-2A>C	c.913-2A>C	0	cPKU
IVS8-2del	c.913-2del	0	cPKU
p.E305*	c.913G>T	0	cPKU
p.Ex9dup	c.914_969+1dup	0	cPKU
p.I306Lfs*35	c.916del	0	cPKU
p.G307Afs*34	c.920del	0	cPKU
p.A309Pfs*32	c.925del	0	cPKU
p.S310_L311del	c.927_932del	0	cPKU
p.L311*	c.930_940del	0	cPKU
p.L311Gfs*4	c.931_932del	0	cPKU
p.L311P	c.932T>C	0	cPKU
p.L311R	c.932T>G	0	cPKU
p.G312Vfs*29	c.935del	0	cPKU
p.G312V	c.935G>T	0	cPKU
p.P314Lfs*27	c.941del	0	cPKU
p.E316*	c.946G>T	0	cPKU
p.E319*	c.955G>T	0	cPKU
p.T323del	c.967_969del	0	cPKU
IVS9+1G>A	c.969+1G>A	0	cPKU
IVS9+2dup	c.969+2dup	0	cPKU
IVS9+2T>G	c.969+2T>G	0	cPKU
IVS9+5G>A	c.969+5G>A	0	cPKU
IVS9+6T>A	c.969+6T>A	0	cPKU
IVS9-7A>G	c.970-7A>G	0	cPKU
IVS9-6T>G	c.970-6T>G	0	cPKU
IVS9-5T>A	c.970-5T>A	0	cPKU
IVS9-2A>G	c.970-2A>G	0	cPKU
IVS9-2A>C	c.970-2A>C	0	cPKU
IVS9-1G>T	c.970-1G>T	0	cPKU

IVS9-1G>A	c.970-1G>A	0	cPKU
IVS9-1G>C	c.970-1G>C	0	cPKU
p.I324N	c.971T>A	0	cPKU
p.Y325C	c.974A>G	0	cPKU
p.Y325*	c.975C>G	0	cPKU
p.Y325*	c.975C>A	0	cPKU
p.W326Gfs*15	c.976del	0	cPKU
p.W326*	c.977G>A	0	cPKU
p.W326*	c.978G>A	0	cPKU
p.V329Gfs*24	c.986_987del	0	cPKU
p.G332V	c.995G>T	0	cPKU
p.C334*	c.1002C>A	0	cPKU
p.Q336*	c.1006C>T	0	cPKU
p.K341*	c.1021A>T	0	cPKU
p.K341Rfs*59	c.1022del	0	cPKU
p.K341Gfs*12	c.1021_1022del	0	cPKU
p.A342P	c.1024G>C	0	cPKU
p.A342Hfs*58	c.1024del	0	cPKU
p.A342Ffs*58	c.1023del	0	cPKU
p.Y343*	c.1029T>A	0	cPKU
p.Y343*	c.1029T>G	0	cPKU
p.G344S	c.1030G>A	0	cPKU
p.G346Vfs*5	c.1037_1044del	0	cPKU
p.L347Sfs*53	c.1038del	0	cPKU
p.L348Cfs*52	c.1042del	0	cPKU
p.L348Rfs*2	c.1043_1053del	0	cPKU
p.S350Vfs*5	c.1044_1047dup	0	cPKU
p.S349P	c.1045T>C	0	cPKU
p.S349*	c.1046C>A	0	cPKU
p.S349L	c.1046C>T	0	cPKU
p.S350T	c.1048T>A	0	cPKU
p.S350Y	c.1049C>A	0	cPKU
p.G352Vfs*48	c.1055del	0	cPKU
p.E353Nfs*47	c.1056del	0	cPKU
p.E353Nfs*47	c.1057del	0	cPKU
p.E353*	c.1057G>T	0	cPKU
p.E353Vfs*42	c.1057_1058insTCAG	0	cPKU
p.L354Ffs*40	c.1061dup	0	cPKU

p.Q355*	c.1063C>T	0	cPKU
IVS10+1G>A	c.1065+1G>A	0	cPKU
IVS10+1G>T	c.1065+1G>T	0	cPKU
IVS10+3A>G	c.1065+3A>G	0	cPKU
IVS10-14C>G	c.1066-14C>G	0	cPKU
IVS10-11G>A	c.1066-11G>A	0	cPKU
IVS10-10G>A	c.1066-10G>A	0	cPKU
IVS10-7C>A	c.1066-7C>A	0	cPKU
IVS10-3C>G	c.1066-3C>G	0	cPKU
IVS10-2A>T	c.1066-2A>T	0	cPKU
IVS10-2A>G	c.1066-2A>G	0	cPKU
IVS10-1G>A	c.1066-1G>A	0	cPKU
IVS10-1G>C	c.1066-1G>C	0	cPKU
IVS10-1G>T	c.1066-1G>T	0	cPKU
p.Y356H	c.1066T>C	0	cPKU
p.Y356*	c.1068C>A	0	cPKU
p.Y356*	c.1068C>G	0	cPKU
p.C357R	c.1069T>C	0	cPKU
p.C357*	c.1071C>A	0	cPKU
p.L358*	c.1073T>G	0	cPKU
p.S359*	c.1076C>G	0	cPKU
p.E360*	c.1078G>T	0	cPKU
p.K361*	c.1081A>T	0	cPKU
p.P362T	c.1084C>A	0	cPKU
p.K363Afs*30	c.1087_1088del	0	cPKU
p.K363*	c.1087A>T	0	cPKU
p.K363Nfs*37	c.1089del	0	cPKU
p.L365_L369del	c.1092_1106del	0	cPKU
p.L365del	c.1092_1094del	0	cPKU
p.L364del	c.1090_1092del	0	cPKU
p.L367Sfs*43	AGCTTCTCCCC	0	cPKU
p.L367Wfs*33	c.1099del	0	cPKU
p.L367Pfs*27	c.1099dup	0	cPKU
p.E370Afs*25	c.1103_1106dup	0	cPKU
p.K371Pfs*27	c.1111_1117del	0	cPKU
p.K371*	c.1111A>T	0	cPKU
p.K371R	c.1112A>G	0	cPKU
p.A373Hfs*20	c.1117_1118del	0	cPKU

p.I374Yfs*20	c.1119_1120insT	0	cPKU
p.N376lfs*24	c.1127del	0	cPKU
p.Y377Tfs*23	c.1129del	0	cPKU
p.T378Cfs*15	c.1132_1133del	0	cPKU
p.T380Gfs*13	c.1138_1139del	0	cPKU
p.T380Rfs*20	c.1138del	0	cPKU
p.Q383*	c.1147C>T	0	cPKU
p.Q383Hfs*17	c.1149del	0	cPKU
p.L385Cfs*15	c.1153del	0	cPKU
p.L385P	c.1154T>C	0	cPKU
p.Y386C	c.1157A>G	0	cPKU
p.Y386Ffs*14	c.1157del	0	cPKU
p.Y387H	c.1159T>C	0	cPKU
p.Y387*	c.1161C>A	0	cPKU
p.Y387*	c.1161C>G	0	cPKU
p.V388Gfs*5	c.1163_1164del	0	cPKU
p.A389Efs*11	c.1166del	0	cPKU
p.E390Dfs*4	c.1169_1170insC	0	cPKU
p.S391Ffs*2	c.1171_1172del	0	cPKU
p.N393*	c.1176dup	0	cPKU
p.N393lfs*2	c.1177_1178insT	0	cPKU
p.D394Mfs*6	c.1180del	0	cPKU
p.K398*	c.1192A>T	0	cPKU
p.V399A	c.1196T>C	0	cPKU
p.V399Gfs*52	c.1196_1199del	0	cPKU
p.R400Gfs*52	c.1198del	0	cPKU
IVS11+1G>T	c.1199+1G>T	0	cPKU
IVS11+1G>C	c.1199+1G>C	0	cPKU
IVS11+1G>A	c.1199+1G>A	0	cPKU
IVS11+2T>C	c.1199+2T>C	0	cPKU
IVS11+2T>G	c.1199+2T>G	0	cPKU
IVS11+4A>G	c.1199+4A>G	0	cPKU
IVS11+5G>A	c.1199+5G>A	0	cPKU
IVS11+5G>T	c.1199+5G>T	0	cPKU
IVS11-8G>A	c.1200-8G>A	0	cPKU
IVS11-2A>G	c.1200-2A>G	0	cPKU
IVS11-1G>A	c.1200-1G>A	0	cPKU
IVS11-1G>T	c.1200-1G>T	0	cPKU

p.N401Tfs*51	c.1200del	0	cPKU
p.N401_S439del	c.1200-?_1315+?del	0	cPKU
p.A404Pfs*48	c.1209del	0	cPKU
p.I406Sfs*15	c.1215_1219del	0	cPKU
p.I406Sfs*15	c.1216_1220del	0	cPKU
p.I406T	c.1217T>C	0	cPKU
p.P407Lfs*45	c.1220del	0	cPKU
p.R408W	c.1222C>T	0	cPKU
p.R408Gfs*44	c.1222del	0	cPKU
p.R408L	c.1223G>T	0	cPKU
p.S411*	c.1232C>G	0	cPKU
p.S411*	c.1232C>A	0	cPKU
p.R413P	c.1238G>C	0	cPKU
p.Y414*	c.1242C>A	0	cPKU
p.Y414*	c.1242C>G	0	cPKU
p.P416Hfs*36	c.1247del	0	cPKU
p.P416Hfs*36	c.1245del	0	cPKU
p.Y417*	c.1251C>A	0	cPKU
p.Q419*	c.1255C>T	0	cPKU
p.L424Wfs*28	c.1269del	0	cPKU
p.L424*	c.1271T>A	0	cPKU
p.Q428Sfs*24	c.1282del	0	cPKU
p.Q428*	c.1282C>T	0	cPKU
p.K431Nfs*4	c.1293_1294del	0	cPKU
p.L433Ffs*3	c.1298dup	0	cPKU
p.S436Pfs*16	c.1306del	0	cPKU
IVS12+1G>A	c.1315+1G>A	0	cPKU
IVS12+1G>T	c.1315+1G>T	0	cPKU
IVS12+2T>C	c.1315+2T>C	0	cPKU
IVS12+2T>A	c.1315+2T>A	0	cPKU
IVS12+4A>G	c.1315+4A>G	0	cPKU
IVS12+5G<A	c.1315+5G>A	0	cPKU
IVS12-5T>C	c.1316-5T>C	0	cPKU
IVS12-2A>C	c.1316-2A>C	0	cPKU
IVS12-2A>G	c.1316-2A>G	0	cPKU
IVS12-1G>A	c.1316-1G>A	0	cPKU
IVS12-1G>C	c.1316-1G>C	0	cPKU
IVS12-1G>T	c.1316-1G>T	0	cPKU

p.E440*	c.1318G>T	0	cPKU
p.G442Wfs*47	c.1323dup	0	cPKU
p.L448Rfs*3	c.1342_1345del	0	cPKU
p.*453Vext*35	c.1355dup	0	cPKU
p.K452Nfs*33	c.1356del	0	cPKU
p.*453Pext*33	c.1357_1361del	0	cPKU
Ex1_3del	c.(1-1393_1-405)_(352+33_370)del	0	cPKU
Ex?del	c.-95-3279_-95-83del	0	cPKU
Ex1del	c.1-?_60+?del	0	cPKU
Ex1_2del	c.1-?_168+?del	0	cPKU
Ex3del	c.169-4949_352+1467delinsGGCACCTG	0	cPKU
Ex3del	c.169_4951delins8	0	cPKU
Ex3del	c.169-2_352+?del	0	cPKU
Ex3del4765	c.169-6125_169-1361del	0	cPKU
Ex4del	c.215_223delTAAAGAAAG	0	cPKU
Ex4_5del	c.(441+1_442-1)_(509+1_510-1)del	0	cPKU
Ex4-8del	c.353-?_912+?del	0	cPKU
Ex5del	c.442-?_509 + ?del	0	cPKU
Ex5del	c.442_1556del1881	0	cPKU
Ex5del4232ins268	c.442_509del168	0	cPKU
Ex5_Ex6del	g.11653bpdel	0	cPKU
Ex5_Ex6del	c.456_706+138del11653	0	cPKU
Ex?del	c.443_509+1del	0	cPKU
Ex6del7831	c.509+4140_706+510del7831	0	cPKU
EX6del7793insGG	c.510-6708_706+888del7793insGG	0	cPKU
Ex6del	c.510-?_706+?del	0	cPKU
Ex6_7_8del	c.?	0	cPKU
Ex9_11del	g.~6.7kdel	0	cPKU
Ex10_11_12_13del	c.970-?_1359+?	0	cPKU
Ex12del	c.1314_1315+4del	0	cPKU
Ex12_13del	c.1200-?_1359+?del	0	cPKU
PAHdel	c.-27778delPAH	0	cPKU
p.F39L	c.117C>G	0.2	cPKU
p.L213P	c.638T>C	0.2	cPKU
IVS7-5T>C	c.843-5T>C	0.2	cPKU
p.F39L	c.115T>C	0.3	cPKU
p.V412F	c.1234G>T	0.3	cPKU
p.R243Q	c.728G>A	0.4	cPKU

p.Y277D	c.829T>G	0.4	cPKU
IVS10-3C>T	c.1066-3C>T	0.4	cPKU
p.A395P	c.1183G>C	0.4	cPKU
p.S231P	c.691T>C	0.5	cPKU
p.L194P	c.581T>C	0.6	cPKU
p.P281R	c.842C>G	0.6	cPKU
p.F39del	c.116_118del	0.8	cPKU
p.F39del	c.115_117del	0.8	cPKU
p.V399=	c.1197A>T	0.9	cPKU
p.Y154H	c.460T>C	1.0	cPKU
IVS3-2A>T	c.353-2A>T	1.2	cPKU
p.F161S	c.482T>C	1.2	cPKU
p.P211L	c.632C>T	1.2	cPKU
p.H271R	c.812A>G	1.2	cPKU
p.P292L	c.875C>T	1.2	cPKU
p.E56D	c.168G>T	1.3	cPKU
p.Q304K	c.910C>A	1.4	cPKU
p.L348V	c.1042C>G	1.4	cPKU
p.T372R	c.1115C>G	1.4	cPKU
p.I65T	c.194T>C	1.5	cPKU
p.A165D	c.494C>A	1.5	cPKU
p.R261Q	c.782G>A	1.5	cPKU
IVS11+502A>T	c.1199+502A>T	1.5	cPKU
p.G46S	c.136G>A	1.6	cPKU
IVS2+2T>C	c.168+2T>C	1.7	cPKU
IVS2+19T>C	c.168+19T>C	1.7	cPKU
p.T81P	c.241A>C	1.7	cPKU
p.R169G	c.505C>G	1.7	cPKU
p.G171R	c.511G>C	1.7	cPKU
p.Y206C	c.617A>G	1.7	cPKU
p.H220P	c.659A>C	1.7	cPKU
p.L287V	c.859C>G	1.7	cPKU
p.G346R	c.1036G>C	1.7	cPKU
p.I406V	c.1216A>G	1.7	cPKU
p.I95del	c.284_286del	1.8	cPKU
p.A156P	c.466G>C	1.8	cPKU
IVS7+5G>A	c.842+5G>A	1.8	cPKU
p.L48S	c.143T>C	1.9	cPKU

p.G218V	c.653G>T	1.9	cPKU
p.M276R	c.827T>G	2.0	cPKU
p.Y387D	c.1159T>G	2.0	cPKU
p.K398=	c.1194A>G	2.0	cPKU
IVS12-2A>C	c.1200-2A>C	2.0	cPKU
p.Q419P	c.1256A>C	2.0	cPKU
p.*453Pext*33	c.1357_*2del	2.0	cPKU
p.*453Pext*33	c.1357_*2del	2.0	cPKU
p.V388M	c.1162G>A	2.1	cPKU
IVS11-1G>C	c.1200-1G>C	2.1	cPKU
p.A345T	c.1033G>A	2.2	cPKU
p.G247R	c.739G>C	2.3	cPKU
p.A342T	c.1024G>A	2.3	cPKU
p.S70del	c.206_208del	2.4	cPKU
p.A434D	c.1301C>A	2.4	cPKU
p.Q20H	c.60G>C	2.5	cPKU
p.P147L	c.440C>T	2.5	cPKU
p.H170R	c.509A>G	2.5	cPKU
p.L197F	c.591G>C	2.5	cPKU
p.F233L	c.697T>C	2.5	cPKU
p.F233L	c.699C>G	2.5	cPKU
p.L249F	c.745C>T	2.5	cPKU
p.R270I	c.809G>T	2.5	cPKU
p.G307D	c.920G>A	2.5	cPKU
p.S310F	c.929C>T	2.5	cPKU
p.P314A	c.940C>G	2.5	cPKU
p.G344D	c.1031G>A	2.5	cPKU
p.R400T	c.1199G>C	2.5	cPKU
p.P416Q	c.1247C>A	2.5	cPKU
p.R261P	c.782G>C	2.6	cPKU/mPKU
p.R400K	c.1199G>A	2.6	cPKU/mPKU
p.A104D	c.311C>A	2.7	cPKU/mPKU
p.Q304=	c.912G>A	3.0	mPKU
p.S349A	c.1045T>G	3.1	mPKU
p.I65S	c.194T>G	3.3	mPKU
p.E76D	c.228G>C	3.3	mPKU
p.L98V	c.292T>G	3.3	mPKU
p.H146Y	c.436C>T	3.3	mPKU

p.G188D	c.563G>A	3.3	mPKU
p.Q226H	c.678G>C	3.3	mPKU
p.V230A	c.689T>C	3.3	mPKU
p.A309D	c.926C>A	3.3	mPKU
p.A309V	c.926C>T	3.3	mPKU
p.A322D	c.965C>A	3.3	mPKU
p.S67P	c.199T>C	3.7	mPKU
p.V291M	c.871G>A	3.8	mPKU
p.S391I	c.1172G>T	3.8	mPKU
p.P244L	c.731C>T	3.9	mPKU
IVS7-14_-11del	c.843-14_-11del	4.0	mPKU
p.S303P	c.907T>C	4.0	mPKU
p.D101N	c.301G>A	4.1	mPKU
p.Y343C	c.1028A>G	4.2	mPKU
p.I174N	c.521T>A	4.3	mPKU
p.T418P	c.1252A>C	4.4	mPKU
p.K320N	c.960G>C	4.5	mPKU
p.F331S	c.992T>C	4.5	mPKU
IVS4+5G>A	c.441+5G>A	4.6	mPKU
p.P314T	c.940C>A	4.7	mPKU
p.R408Q	c.1223G>A	4.7	mPKU
p.T63P	c.187A>C	5.0	mPKU
p.R68G	c.202A>G	5.0	mPKU
p.S70del	c.208_210del	5.0	mPKU
p.D84Y	c.250G>T	5.0	mPKU
p.I95F	c.283A>T	5.0	mPKU
p.G103D	c.308G>A	5.0	mPKU
p.D129Y	c.385G>T	5.0	mPKU
p.D129G	c.386A>G	5.0	mPKU
p.D143G	c.428A>G	5.0	mPKU
p.I164V	c.490A>G	5.0	mPKU
IVS6+531T>C	c.706+531T>C	5.0	mPKU
p.T238A	c.712A>G	5.0	mPKU
p.R241H	c.722G>A	5.0	mPKU
p.P275S	c.823C>T	5.0	mPKU
p.P275R	c.824C>G	5.0	mPKU
p.M276V	c.826A>G	5.0	mPKU
p.H285Y	c.853C>T	5.0	mPKU

p.G312C	c.934G>T	5.0	mPKU
p.A313T	c.937G>A	5.0	mPKU
p.D338Y	c.1012G>T	5.0	mPKU
p.A345S	c.1033G>T	5.0	mPKU
p.G352R	c.1054G>C	5.0	mPKU
p.G352C	c.1054G>T	5.0	mPKU
IVS10+241C>A	c.1065+241C>A	5.0	mPKU
p.C357Y	c.1070G>A	5.0	mPKU
p.L358F	c.1074A>T	5.0	mPKU
p.Q375H	c.1125A>C	5.0	mPKU
p.D394Y	c.1180G>T	5.0	mPKU
p.A395G	c.1184C>G	5.0	mPKU
p.Y417H	c.1249T>C	5.0	mPKU
p.L444F	c.1330C>T	5.0	mPKU
p.R241C	c.721C>T	5.1	mPKU
p.Y414C	c.1241A>G	5.1	mPKU
p.P407S	c.1219C>T	5.3	mPKU
IVS12+6T>A	c.1315+6T>A	5.3	mPKU
p.R68S	c.204A>T	5.4	mPKU
p.M1I	c.3G>C	5.5	mPKU
p.M1I	c.3G>T	5.5	mPKU
p.M276K	c.827T>A	5.6	mPKU
p.P119S	c.355C>T	5.7	mPKU
p.L430P	c.1289T>C	5.7	mPKU
p.R169C	c.505C>T	5.8	mPKU
p.Q375E	c.1123C>G	5.9	mPKU
p.V245M	c.733G>A	6.0	mPKU
IVS4-5C>G	c.442-5C>G	6.1	mPKU
p.Q419R	c.1256A>G	6.1	mPKU
p.N167I	c.500A>T	6.2	mPKU
p.I421T	c.1262T>C	6.2	mPKU
p.D222G	c.665A>G	6.4	mPKU
p.L308F	c.922C>T	6.4	mPKU
p.Y168H	c.502T>C	6.7	mPKU/HPA
p.I174V	c.520A>G	6.7	mPKU/HPA
p.S310C	c.929C>G	6.7	mPKU/HPA
p.A373T	c.1117G>A	6.7	mPKU/HPA
p.K396R	c.1187A>G	6.7	mPKU/HPA

p.R413S	c.1237C>A	6.7	mPKU/HPA
p.E390G	c.1169A>G	6.8	mPKU/HPA
p.E76G	c.227A>G	7.1	HPA
p.Q172H	c.516G>C	7.1	HPA
p.V190A	c.569T>C	7.1	HPA
p.K363N	c.1089G>C	7.1	HPA
p.V177L	c.529G>C	7.2	HPA
p.H107R	c.320A>G	7.5	HPA
p.E178G	c.533A>G	7.5	HPA
p.T372S	c.1114A>T	8.1	HPA
p.F55L	c.165T>G	8.2	HPA
p.T92I	c.275C>T	8.3	HPA
p.I94S	c.281T>G	8.3	HPA
p.E182K	c.544G>A	8.3	HPA
p.V388L	c.1162G>C	8.3	HPA
IVS11+17G>A	c.1199+17G>A	8.3	HPA
p.I65V	c.193A>G	8.8	HPA
p.R169S	c.505C>A	9.0	HPA
p.R176Q	c.527G>A	9.0	HPA
p.T186I	c.557C>T	9.0	HPA
p.F240V	c.718T>G	9.0	HPA
p.P366H	c.1097C>A	9.0	HPA
p.R413H	c.1238G>A	9.0	HPA
p.N61K	c.183C>A	9.2	HPA
IVS10+258C>A	c.1065+258C>A	9.3	HPA
p.I406M	c.1218A>G	9.3	HPA
p.R53H	c.158G>A	9.4	HPA
p.A322T	c.964G>A	9.6	HPA
p.F392I	c.1174T>A	9.6	HPA
p.S87R	c.261C>A	9.7	HPA
p.A300S	c.898G>T	9.7	HPA
p.A403V	c.1208C>T	9.7	HPA
p.R176L	c.527G>T	9.8	HPA
p.V230I	c.688G>A	9.8	HPA
p.I306V	c.916A>G	9.8	HPA
p.D415N	c.1243G>A	9.8	HPA
p.P211T	c.631C>A	9.9	HPA
p.V245A	c.734T>C	9.9	HPA

p.T380M	c.1139C>T	9.9	HPA
p.V45A	c.134T>C	10	HPA
p.A47V	c.140C>T	10	HPA
p.E66K	c.196G>A	10	HPA
p.R71H	c.212G>A	10	HPA
p.S87R	c.259A>C	10	HPA
p.L98S	c.293T>C	10	HPA
p.S110L	c.329C>T	10	HPA
p.A132V	c.395C>T	10	HPA
p.D145V	c.434A>T	10	HPA
p.R155H	c.464G>A	10	HPA
p.R169H	c.506G>A	10	HPA
p.R169L	c.506G>T	10	HPA
p.H170D	c.508C>G	10	HPA
p.V177M	c.529G>A	10	HPA
p.T193I	c.578C>T	10	HPA
p.H201Y	c.601C>T	10	HPA
p.C203Y	c.608G>A	10	HPA
p.C217G	c.649T>G	10	HPA
p.E221G	c.662A>G	10	HPA
p.V245A	c.734_735inv	10	HPA
p.L258P	c.773T>C	10	HPA
p.I269L	c.805A>C	10	HPA
p.M276T	c.827T>C	10	HPA
p.G289R	c.865G>C	10	HPA
p.G289R	c.865G>A	10	HPA
p.R297C	c.889C>T	10	HPA
p.R297H	c.890G>A	10	HPA
p.A313V	c.938C>T	10	HPA
p.P314S	c.940C>T	10	HPA
p.A322G	c.965C>G	10	HPA
p.V379A	c.1136T>C	10	HPA
p.D394H	c.1180G>C	10	HPA
p.A395S	c.1183G>T	10	HPA