

From PAHvdb (December 2012) - www.biopku.org®

Protein variant	Nucleotide aberration	PAH activity (%)*
p.P416Q	c.1247C>A	111
p.V190A	c.569T>C	110
p.K274E	c.820A>G	100
p.R68G	c.202A>G	100
p.P407S	c.1219C>T	94
p.D59Y	c.175G>T	92
p.E76G	c.227A>G	85
p.R53H	c.158G>A	79
p.A313T	c.937G>A	76
p.Y417C	c.1250A>G	76
p.T92I	c.275C>T	76
p.A322G	c.965C>G	75
p.P211T	c.631C>A	72
p.D415N	c.1243G>A	72
p.Q419R	c.1256A>G	71
p.N223Y	c.667A>T	70
p.P69S	c.205C>T	69
p.D143G	c.428A>G	68
p.R68S	c.204A>T	68
p.A403V	c.1208C>T	66
p.G218V	c.653G>T	64
p.V230I	c.688G>A	63
p.E390G	c.1169A>G	62
p.Y414C	c.1241A>G	57
p.A373T	c.1117G>A	56
p.F382L	c.1146C>G	56
p.K398N	c.1194A>C	55
p.P244L	c.731C>T	51
p.V245A	c.734T>C	50
p.L308F	c.922C>T	49
p.F39L	c.117C>G	49
p.R408Q	c.1223G>A	46
p.R155H	c.464G>A	44
p.R261Q	c.782G>A	44
p.H170D	c.508C>G	43
p.R176L	c.527G>T	42
p.T124I	c.371C>T	42
p.R297L	c.890G>T	42
p.A309V	c.926C>T	42
p.E178G	c.533A>G	39
p.I306V	c.916A>G	39
p.L48S	c.143T>C	39
p.L348V	c.1042C>G	35
p.R413P	c.1238G>C	35
p.I65T	c.194T>C	33
p.R413S	c.1237C>A	32
p.A300S	c.898G>T	31
p.V388M	c.1162G>A	28
p.G171A	c.512G>C	27
p.L52S	c.155T>C	27
p.I95del	c.283_285delATC	27

p.A104D	c.311C>A	27
p.A342T	c.1024G>A	26
p.R241C	c.721C>T	25
p.S87R	c.261C>A	25
p.R241H	c.722G>A	23
p.P122Q	c.365C>A	22
p.F39del	c.115_117delTCT	20
p.S70P	c.208T>C	20
p.E178V	c.533A>T	18
p.A395P	c.1183G>C	16
p.G46S	c.136G>A	16
p.E280A	c.839A>C	15
p.P243Q	c.728G>A	14
p.R243Q	c.728G>A	13
p.A47V	c.140C>T	13
p.I174T	c.521T>C	12
p.K42I	c.125A>T	12
p.R158Q	c.473G>A	10
p.I283F	c.847A>T	10
p.L41F	c.121C>T	10
p.F161S	c.482T>C	7
p.V245L	c.733G>C	7
p.V245E	c.734T>A	7
p.L255V	c.763T>G	7
p.L333F	c.997C>T	7
p.A259T	c.775G>A	6
p.R157T	c.470G>C	5
p.G247V	c.740G>T	4
p.Q304P	c.911A>C	4
p.R252G	c.754C>G	3
p.R252Q	c.755G>A	3
p.F299C	c.896T>G	3
p.L255S	c.764T>C	2
p.A259V	c.776C>T	2
p.E280K	c.838G>A	2
p.P281L	c.842C>T	2
IVS7-5T>C	c.843-5T>C	2
p.D282N	c.844G>A	2
p.R408W	c.1222C>T	2
p.D222*	c.663_664delAG	1
p.T278I	c.833C>T	1
p.L311P	c.932T>C	1
p.S349P	c.1045T>C	1
p.R252W	c.754C>T	0
p.Y277D	c.829T>G	0
IVS10-11G>A	c.1066-11G>A	0
*PAH activity is calculated as average of data obtained in eukaryotic cell systems.		