

Table 1. Spectrum of pathogenic variants in *PAH* gene identified in the 157 probands.

Nucleotide change	Protein change	Character	Location	No. of the allele found
c.59_60delinsCC	Q20P	Missense	Exon1	1
c.111_112insG	I38Pfs*19	Frame shift	Exon2	1
163_164insATAT	F55Pfs*3	Frame shift†	Exon2	1
c.168+5G>C	-	Splicing	Exon2	2
c.194T>C	I65T	Missense	Exon3	4
c.194T>G	I65S	Missense	Exon3	1
c.205C>T	P69S	Missense	Exon3	1
c.208_210delTCT	S70del	Deletion	Exon3	4
c.301G>A	D101N	Missense	Exon3	1
c.320A>G	H107R	Missense	Exon3	1
c.331C>T	R111X	Nonsense	Exon3	19
c.440C>T	P147L	Missense	Exon4	1
c.441+3G>C	-	Splicing	Exon4	2
c.442-1G>A	-	Splicing	Exon4	9
c.464G>A	R155H	Missense	Exon5	1
c.466G>C	A156P	Missense	Exon5	2
c.470G>A	R157K	Missense	Exon5	3
c.472C>T	R158W	Missense	Exon5	1
c.482T>C	F161S	Missense	Exon5	3
c.498C>G	Y166X	Nonsense	Exon5	3
c.505C>A	R169S	Missense	Exon5	1
c.510T>A	H170Q	Missense	Exon6	1
c.526C>T	R176X	Nonsense	Exon6	3
c.563G>A	G188D	Missense	Exon6	1
c.611A>G	EX6-96A>G	Splicing	Exon6	27
c.638T>C	L213P	Missense	Exon6	1
c.650G>A	C217Y	Missense	Exon6	1
c.671T>C	I224T	Missense	Exon6	1
c.704A>C	Q235P	Missense	Exon6	1
c.721C>T	R241C	Missense	Exon7	5
c.722delG;c.735G>A	R241Pfs*5	Frameshift	Exon7	2
c.722G>A	R241H	Missense	Exon7	1
c.728G>A	R243Q	Missense	Exon7	54
c.739G>C	G247R	Missense	Exon7	2
c.740G>T	G247V	Missense	Exon7	3
c.755G>A	R252Q	Missense	Exon7	2
c.764T>C	L255S	Missense	Exon7	1
c.770G>T	G257V	Missense	Exon7	2
c.781C>T	R261X	Nonsense	Exon7	1

c.782G>A	R261Q	Missense	Exon7	4
c.838G>A	E280K	Missense	Exon7	1
c.839A>G	E280G	Missense	Exon7	1
c.842+2T>A	-	Splicing	Exon7	10
c.875C>T	P292L	Missense	Exon8	1
c.898G>T	A300S	Missense	Exon8	1
c.904delT	F302Pfs*39	Frame shift	Exon8	1
c.907delT	S303Pfs*38	Frame shift	Exon8	1
c.907T>C	S303P	Missense	Exon8	1
c.913-2A>G	-	Splicing	Exon8	1
c.913-7A>G	-	Splicing	Exon8	1
c.922C>T	L308F	Missense	Exon9	1
c.926C>A	A309D	Missense	Exon9	1
c.940C>A	P314T	Missense	Exon9	1
c.964G>A	A322T	Missense	Exon9	1
c.971T>A	I324N	Missense	Exon10	2
c.975C>G	Y325X	Nonsense	Exon10	2
c.977G>A	W326X	Nonsense	Exon10	1
c.1033G>A	A345T	Missense	Exon10	1
c.1045T>G	S349A	Missense	Exon10	2
c.1066-13T>G	-	Splicing	Exon10	1
c.1066-14C>G	-	Splicing	Exon10	1
c.1066-1G>A	-	Splicing	Exon10	1
c.1068C>A	Y356X	Nonsense	Exon11	17
c.1123C>G	Q375E	Missense	Exon11	1
c.1174T>A	F392I	Missense	Exon11	2
c.1184C>A	A395D	Missense	Exon11	2
c.1197A>T	V399V	Splicing	Exon11	25
c.1199+1G>C	-	Splicing	Exon11	1
c.1199G>A	R400K	Missense	Exon11	1
c.1199G>C	R400T	Missense	Exon11	4
c.1208C>T	A403V	Missense	Exon12	1
c.1222C>T	R408W	Missense	Exon12	4
c.1223G>A	R408Q	Missense	Exon12	6
c.1238G>C	R413P	Missense	Exon12	15
c.1252A>C	T418P	Missense	Exon12	1
c.1256A>G	Q419R	Missense	Exon12	2
c.1301C>A	A434D	Missense	Exon12	5
c.1315+6T>A	-	Splicing	Exon12	1
Exon1 & upstream		Large Deletion		2
Exon4/5		Large Deletion		2

†:Novel variant