

Table SI. Oligonucleotide primers used for *PAH* gene sequencing.

Primers	Forward primer (5'-3')	Reverse primer 5'-3')	Product size (bp)
Ex 1	ACCCTTTGTAACCTCTTCTCCT	ACTGAGCTTATGAAACCAGGAAG	283
Ex 2	AAAGAGTTCATGCTTGCTTTGT	TCCAGATCCTGTGTTCTTTTCA	297
Ex 3	TCCTGTTCTGGTTCTGCATCT	ATGAAGACAGTGTGGAGTACTT	311
Ex 4	CAGCCCACTTGCCATCAC	TTCCAGCCCTCGTGTAAT	346
Ex 5	TAACCAAGGGAAGGAGACATGC	AACTGGATGAGGGCAAGGGA	264
Ex 6	CTCCCTCTGCTAACCTAACCTG	TCATACTTGCCCTCCACATACTTG	372
Ex 7	ATCTGAAGCCAAGTCTGCCT	AGCCAGCAATGAACCCAAAC	364
Ex 8	GCCCATTCCTCATGTAGAAAGAC	TAGCTTCCCAGAACCACACA	312
Ex 9	GGCCACCCATCACCTTTT	TCCAGGGGAGTAGGAAAGTT	234
Ex 10	CCCTCCAGAAACACCCTCAT	GAATGAGTTCACAGGTTGCA	362
Ex 11	GTGTAGAAGGAATCGGGGTGAG	GGAGAACATGGGAGAGAACTG	390
Ex 12	GAGGTGTCCGTGTTCCCTAAA	GTTCTTCTCCATCAATGAACCCA	380
Ex 13	GCTCCTTGAAAGTTGGGGTG	AAGGTTTGCTTTTCGGACTTTT	270
M13	TGTAAAACGACGGCCAGT	CAGGAAACAGCTATGACC	

Table SII. Summary of demographic and clinical characteristics of patients with PKU.

Family	Consanguinity ^a	Patients (n=77)	Age ^b	Sex	Allele 1	Allele 2	APVs	Observed phenotypes	Predicted phenotypes
PKU-1	First cousins	PKU-1-1 (Index)	10 Mo	M	c.143T>C	c.143T>C	2	Classic PKU	Classic-Mild PKU
		PKU-1-2 (Index)	11 Mo	M	c.143T>C	c.143T>C	2	Classic PKU	Classic-Mild PKU
PKU-2	First cousins	PKU-2-1 (Index)	2 Mo	M	c.143T>C	c.143T>C	2	Classic PKU	Classic-Mild PKU
PKU-3	Second cousins	PKU-3-1 (Index)	6 Y	M	c.116_118del	c.116_118del	0.7	Classic PKU	Classic PKU
PKU-4	Second cousins	PKU-4-1 (Index)	6 Y	M	c.155del	c.155del	0	Classic PKU	Classic PKU
PKU-5	First cousins	PKU-5-1 (Index)	2 Mo	F	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
PKU-6	First cousins	PKU-6-1 (Index)	1 Y	M	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
PKU-7	First cousins	PKU-7-1 (Index)	26 Y	M	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
		PKU-7-2 (Index)	30 Y	F	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
PKU-8	First cousins	PKU-8-1 (Index)	1 Y	F	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
PKU-9	Not relatives	PKU-9-1 (Index)	14 Y	M	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
		PKU-9-2 (Index)	10 Y	M	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
PKU-10	First cousins	PKU-10-1 (Index)	20 Y	F	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
PKU-11	First cousins	PKU-11-1 (Index)	17 Y	F	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
PKU-12	First cousins	PKU-12-1 (Index)	8 Y	M	c.168_168+1delinsAA	c.168_168+1delinsAA	0	Classic PKU	Classic PKU
PKU-13	First cousins	PKU-13-1 (Index)	20 Y	M	c.169_171del	c.169_171del	0	Classic PKU	Classic PKU
		PKU-13-2 (Index)	8 Y	M	c.169_171del	c.169_171del	0	Classic PKU	Classic PKU
PKU-14	First cousins	PKU-14-1 (Index)	5 Y	M	c.169_171del	c.169_171del	0	Classic PKU	Classic PKU
PKU-15	First cousins	PKU-15-1 (Index)	1 Y	F	c.169_171del	c.169_171del	0	Classic PKU	Classic PKU
PKU-16	First cousins	PKU-16-1 (Index)	6 Y	F	c.169_171del	c.169_171del	0	Classic PKU	Classic PKU

		PKU-16-2 (Index)	1 Mo	M	c.169_171del	c.169_171del	0	Classic PKU	Classic PKU
PKU-17	First cousins	PKU-17-1 (Index)	8 Y	M	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
		PKU-17-2 (Index)	14 Y	F	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
PKU-18	Second cousins	PKU-18-1 (Index)	3 Y	F	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
PKU-19	First cousins	PKU-19-1 (Index)	17 Y	F	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
		PKU-19-2 (Index)	4 Y	M	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
		PKU-19-3 (Index)	19 Y	F	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
		PKU-19-4 (Index)	21 Y	M	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
PKU-20	Second cousins	PKU-20-1 (Index)	9 Y	F	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
		PKU-20-2 (Index)	9 Y	M	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
PKU-21	First cousins	PKU-21-1 (Index)	11 Y	F	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
		PKU-21-2 (Index)	2 Mo	M	c.441+5G>T	c.441+5G>T	4.2	Classic PKU	Classic PKU
PKU-22	First cousins	PKU-22-1 (Index)	1 Y	F	c.509+1G>A	c.509+1G>A	0	Classic PKU	Classic PKU
		PKU-22-2 (Index)	3 Y	F	c.509+1G>A	c.509+1G>A	0	Classic PKU	Classic PKU
PKU-23	Second cousins	PKU-23-1 (Index)	9 Y	M	c.526C>T	c.526C>T	0	Classic PKU	Classic PKU
PKU-24	Second cousins	PKU-24-1 (Index)	16 Y	M	c.526C>T	c.526C>T	0	Classic PKU	Classic PKU
		PKU-24-2 (Index)	6 Y	F	c.526C>T	c.526C>T	0	Classic PKU	Classic PKU
PKU-25	Second cousins	PKU-25-1 (Index)	3 Y	M	c.526C>T	c.526C>T	0	Classic PKU	Classic PKU

PKU-26	Second cousins	PKU-26-1 (Index)	6 Y	M	c.526C>T	c.526C>T	0	Classic PKU	Classic PKU
PKU-27	Second cousins	PKU-27-1 (Index)	17 Y	M	c.593_614del	c.593_614del	0	Classic PKU	Classic PKU
		PKU-27-2 (Index)	11 Y	M	c.593_614del	c.593_614del	0	Classic PKU	Classic PKU
		PKU-27-3 (Index)	11 Y	M	c.593_614del	c.593_614del	0	Classic PKU	Classic PKU
		PKU-27-4 (Index)	11 Y	M	c.593_614del	c.593_614del	0	Classic PKU	Classic PKU
PKU-28	First cousins	PKU-28-1 (Index)	2 Y	F	c.781del	c.781del	NA	NA	NA
		PKU-28-2 (Index)	1 Y	M	c.781del	c.781del	NA	NA	NA
PKU-29	First cousins	PKU-29-1 (Index)	8 Y	F	c.782G>A	c.782G>A	1.7	Classic PKU	Classic PKU
PKU-30	First cousins	PKU-30-1 (Index)	17 Y	F	c.810A>T	c.810A>T	0	Classic PKU	Classic PKU
PKU-31	First cousins	PKU-31-1 (Index)	13 Y	M	c.810A>T	c.810A>T	0	Classic PKU	Classic PKU
		PKU-31-2 (Index)	5 Y	M	c.810A>T	c.810A>T	0	Classic PKU	Classic PKU
PKU-32	First cousins	PKU-32-1 (Index)	8 Y	M	c.810A>T	c.810A>T	0	Classic PKU	Classic PKU
PKU-33	First cousins	PKU-33-1 (Index)	3 Y	M	c.842C>T	c.842C>T	0	Classic PKU	Classic PKU
PKU-34	First cousins	PKU-34-1 (Index)	2 Y	M	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
PKU-35	Second cousins	PKU-35-1 (Index)	4 Y	M	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
PKU-36	Second cousins	PKU-36-1 (Index)	11 Y	M	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
		PKU-36-2 (Index)	7 Y	F	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
PKU-37	Not relatives	PKU-37-1 (Index)	23 Y	F	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU

PKU-38	Second cousins	PKU-38-1 (Index)	8 Y	M	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
		PKU-38-2 (Index)	5 Y	F	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
		PKU-38-3 (Index)	3 Y	F	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
PKU-39	First cousins	PKU-39-1 (Index)	5 Y	M	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
		PKU-39-2 (Index)	3 Y	M	c.1066-11G>A	c.1066-11G>A	0	Classic PKU	Classic PKU
PKU-40	Second cousins	PKU-40-1 (Index)	18 Y	M	c.1089del	c.1089del	0	Classic PKU	Classic PKU
		PKU-40-2 (Index)	16 Y	M	c.1089del	c.1089del	0	Classic PKU	Classic PKU
PKU-41	First cousins	PKU-41-1 (Index)	4 Y	M	c.1162G>A	c.1162G>A	2	Classic PKU	Classic PKU
		PKU-41-2 (Index)	15 Y	M	c.1162G>A	c.1162G>A	2	Classic PKU	Classic PKU
		PKU-41-3 (Index)	17 Y	F	c.1162G>A	c.1162G>A	2	Classic PKU	Classic PKU
PKU-42	First cousins	PKU-42-1 (Index)	11 Y	F	c.1162G>A	c.1162G>A	2	Classic PKU	Classic PKU
PKU-43	First cousins	PKU-43-1 (Index)	6 Y	M	c.1199+1G>C	c.1199+1G>C	0	Classic PKU	Classic PKU
PKU-44	Second cousins	PKU-44-1 (Index)	8 Y	M	c.168_168+1delinsAA	c.169_171del	NA	NA	NA
PKU-45	Not relatives	PKU-45-1 (Index)	13 Y	M	c.526C>T	c.441+5G>T	NA	NA	NA
PKU-46	Not relatives	PKU-46-1 (Index)	11 Y	M	c.782G>A	c.838G>A	NA	NA	NA
PKU-47	Second cousins	PKU-47-1 (Index)	3 Y	F	c.727C>T	c.441+5G>T	NA	NA	NA
		PKU-47-1 (Index)	2 Y	M	c.727C>T	c.441+5G>T	NA	NA	NA
		PKU-47-1 (Index)	1 Y	F	c.727C>T	c.441+5G>T	NA	NA	NA
PKU-48	Not relatives	PKU-48-1 (Index)	1 Mo	M	c.898G>T	c.1066-11G>A	NA	NA	NA

PKU-49	Not relatives	PKU-49-1 (Index)	2 Mo	F	c.969+5G>A	c.1208C>T	NA	NA	NA
PKU-50	Not relatives	PKU-50-1 (Index)	3 Mo	F	c.1066-11G>A	c.1222C>T	NA	NA	NA

^a, Parental consanguinity. ^b, age at investigations. PKU, phenylketonuria; Mo, months; Y, year, M, male; F, female; NA, not available.