

Table 1. Clinical characteristics and variants of PKU patients

No.	Phenotype	Known mutation*	c.1199+502A>T	c.1065+241C>A	c.706+368T>C
P01	mPKU	c.1199G>A	heterozygous	wild type	wild type
P02	cPKU	c.728G>A	heterozygous	wild type	wild type
P03	mPKU	c.208_210del	wild type	heterozygous	wild type
P04	cPKU	c.1197A>T	wild type	wild type	heterozygous
P05	cPKU	c.728G>A	heterozygous	wild type	wild type
P06	mPKU	-	wild type	wild type	wild type
P07	mPKU	c.694C>T	heterozygous	wild type	wild type
P08	cPKU	c.728G>A	heterozygous	wild type	wild type
P09	mPKU	c.782G>A	wild type	wild type	wild type
P10	mPKU	IVS7+2T>A	heterozygous	wild type	wild type

Note: cPKU= classic PKU, mPKU=mild PKU, *Identified in previous reported[Yan et al.,2019], - unknown mutation.