

Table 2 In silico analysis of the 269 G>A mutation in AKRUC

Mutation	Amino acid change	Polyphen-2 <sup>a</sup>	SIFT <sup>b</sup>	Mutation taster <sup>c</sup>	ExAC	1000G	gnomAD
c.G269A	p.R90Q	possible damaging (0.651)	affect protein function (0.00)	disease causing (0.9999)	0	0	0

a Polyphen-2 (<http://genetics.bwh.harvard.edu/pph2/>).

b SIFT (<http://gsift.jcvi.org>).

c Mutation taster (<http://www.mutationtaster.org/>)