

**Table S4. Prediction of three splice site variants of *PKHD1*.**

Exon	Nucleotide Change	The Human Splicing Finder	SpliceAI	MaxEntScan
22	c.2141-3T>C	NO significant impact on splicing signals	Get new WT Acceptor site	cut-off < 3.0
47	c.7351-2A>T	broken WT Acceptor site decreasing the score from 86.45 to 58.58	broken WT Acceptor site	cut-off $\geq$ 3.0
61	c.11174+5G>A	broken WT Donor site decreasing the score from 92.39 to 82.83	broken WT Donor site	cut-off $\geq$ 3.0