

**Table S1. Primers and DHPLC conditions for alternate *PKHD1* exons**

Exon	Size (bp)	Primer sequences		Genomic DNA nucleotide		DHPLC conditions	
		Forward (5'-3')	Reward (5'-3')	From	To	Temp.	Initial % buffer B
20a (20)	261	GGATGTGGACTCCTCACTACG	AATCCTCCCAGCTGACTGAA	33325	33585	58.6, 62	56, 54
32a (32A)	353	CCACAGGTGCTATGAATTC	GTGACTACTGGTGTGCTG	61321	61673	61, 63.7	57, 56
32a (32B)	365	AAGGCTTCACTTTCATGTATGAA	CCTCACAGTAAGTATGGTCC	61628	61992	59, 60.6	61, 59
32b (32D)	367	CTTCCTTGCAATGTAACTTTT	GATACCAAAGTCCATCTACCTC	62332	62698	57.9, 58.9	59, 59
32b (32E)	441	CTCATCCGGTGCATTGTTC	CCAGAAGTGAAAGGAGCTACCA	62626	63066	58, 62	60, 55
32c (32E)	441	CTCATCCGGTGCATTGTTC	CCAGAAGTGAAAGGAGCTACCA	62626	63066	58, 62	60, 55
<b>38/38a (38.1)</b>	327	TGGGGTTTGGGTAGCTGTAG	CCAAGGCCATGAGACTGTTT	157391	157717	57.6, 59.6	58, 55
<b>38/38a (38.2)</b>	348	GCAGGATGGTGGAGCTTTTA	GCAGACACTTGGCATTTTCA	157589	157936	56.3, 57	60, 59
39a/39b (38)	332	AAATTATCTGGACAACTTTTCTC	AGACCCCAATACAAATGTCCA	175001	175332	59, 60.5	58, 56
<b>39b (39.1)</b>	445	GGTACTGAGTGTGAGCTTGGC	CAGCCACCCAAATTCTCTGTAA	175247	175691	54.9, 55.9	60, 59
39b (39)	278	TTTGAGTGTGCTCAGTTCT	GCAATGCCATCTATCATCAGAC	175612	175889	57, 59.5	57, 55
<b>39b (39.2)</b>	328	CTCACTAATGAGAGGGAGAAGC	TTCTTTAATAGAAATATTTTCCCCAGA	175757	176084	57.3	57
41a (40)	286	CATGCTTTAGTTCTCTGGACTTT	TGCCTTAAACATGGGAGAAA	178101	178386	58.5, 60.5, 61.5	57, 56, 54
<b>41a (41.1)</b>	400	GAGCTATGAGAGGTAAGGGGGT	CACAAAGCTAGTAAGTGGTAGAGAGACA	178319	178718	56.4	60
<b>41a (41.2)</b>	381	GATTCTGGAACAGGCAGTCC	TTCTGTTTTCTGATTCTGATCTTG	178598	178978	57.8, 58.8	59, 58
44a (43)	258	TCAAGGGTGTGAGTTGAGTACA	TTGAGAAAGAACTTTATGCCCTC	183819	184076	56.9, 58.9	56, 54
51a/51b (50)	300	TGATGGGGTTCCTTACTAAATGA	TGGTGATTCTGACTATGTGCTC	239591	239890	58.3, 59.3, 60.3	57, 56, 55
60a (59)	285	GTGGCTGGTGGTTTATATG	GGGTTTGAAGAATTGCCAAG	340670	340954	54.1, 57.4, 58.7	57, 55, 52
<b>62.1</b>	378	TGTCCTGAGACCTTTTTCTTC	TAAAGGGAATGGTGAAGGTG	343981	344358	55.7, 57.2	60, 58
<b>62.2</b>	377	CCTGGGAGTCATCTTTATTTTTTC	CAAACGCCCTGATGTAGTGA	344268	344644	55.5, 58.1	60, 57
<b>62.3</b>	440	CGCCTTTTACTTACTTATCCTGG	CAGCAAACCTTCTGGCTGG	344570	345009	54.7, 57.5	60, 57
<b>63</b>	328	CCACAGTCTGTTGGATGAAAAA	TCTGGGTGAGTAGTTTTGTACG	346942	347269	54.2, 56.5	58, 56
<b>64.1</b>	341	AAACTTGTGTTTAGAGAGAAAATTGTG	TCTCTTGAGACAGTGACCAGAA	405320	405660	56.7, 57.2	58, 57
<b>64.2</b>	323	TTCTTGTCATCTTTGGTTGG	GAGGTGGTGAACACTACAAGCCA	405578	405900	57.5, 58	58.57
<b>64.3</b>	349	TGCGTATAGCTGTGTTCCAA	TGAGTTAAAGAGAACACTGAAGGG	405815	406163	55.8, 56.7, 58.5	58, 57, 55
<b>64.4</b>	339	CATGCTTGATTAAGGCTACTGG	TTCAGTCTGTGGAATGTCA	406098	406436	56, 57.2, 58.8	59, 58, 56
<b>64.5</b>	326	AAGCTGATCTTGGGAAAGGA	GAACCAGTAGGGGACAGAA	406365	406690	56.1, 57.4	58, 57
<b>64.6</b>	346	AACCCCAATTCACAACCAAC	TGGACAGTAATCAACATGAACAA	406630	406975	53.5, 55.5	58, 56
66a (62)	273	AATGGATTGTGGAAAATTGCT	GGCTGAATGCTACATGCTACTT	438315	438587	55.3, 57.3	56, 55
71a (67B)	394	TGCAAGAGACTGGGAAGTGG	CTGGGAACATTCTGCCTTTC	468285	468678	61.1, 62.4	57, 56

GenBank NM\_138694.2, AY129465

Primers of the longest *PKHD1* ORF amplifying (parts of) alternate exons are given in parentheses.

Predicted exons not included in the longest ORF are shown in bold type.