

SUPPLEMENTAL TABLE

Table S1 TaqMan™ primer and probe (FAM labeled) sequences employed for real-time quantitative PCR to determine allele copy numbers for *HPS6* and surrounding genomic areas in DNA from patients HPS51 and HPS107 (see Figure 1f for genomic location of the assays)

Target area	Primers and Probes	Sequence (5'-3')	HPS51 allele number	HPS107 allele number
<i>ORF67</i> ex1	ORF67ex1-F	ATGATGATGGGCCATTGGATGT	2	2
	ORF67ex1-R	TGTCTCATAGGCTTCACACCAAAA		
	ORF67ex1-probe	CAGTGCCGGTGTCTACT		
<i>HPS6</i> 5UTRa	<i>HPS6</i> 5UTRa-F	GTCCAACCTCTCCTTTTCAGTGTGTA	2	2
	<i>HPS6</i> 5UTRa-R	GCAACTTCTAGGAAAGATACAGAGTGAT		
	<i>HPS6</i> 5UTRa-probe	CCTCTGACAGTTAACCTAC		
<i>HPS6</i> 5UTRb	<i>HPS6</i> 5UTRb-F	CGGCATACGCAGCTTCAG	2	2
	<i>HPS6</i> 5UTRb-R	CCCCGAGGCGGTTCTG		
	<i>HPS6</i> 5UTRb-probe	CTCCCTCCCTCTCCCC		
<i>HPS6</i> 5UTRc	<i>HPS6</i> 5UTRc-F	CCAAGGCCAATCCCTCTTTATGAG	2	1
	<i>HPS6</i> 5UTRc-R	CAGTGAAACAGAAATGGATGCAAGT		
	<i>HPS6</i> 5UTRc-probe	TCCCCGGCCCCCTACCT		
<i>HPS6</i> ex1A*	<i>HPS6</i> ex1A-F	Unknown (Hs00544576_s1)*	2	1
	<i>HPS6</i> ex1A-R	Unknown (Hs00544576_s1)*		
	<i>HPS6</i> ex1A-probe	Targeting <i>HPS6</i> nt.763 in NM_024747*		
<i>HPS6</i> ex1B*	<i>HPS6</i> ex1B-F	Unknown (Hs00949672_s1)*	2	1
	<i>HPS6</i> ex1B-R	Unknown (Hs00949672_s1)*		
	<i>HPS6</i> ex1B-probe	Targeting <i>HPS6</i> nt.827 in NM_024747*		
<i>HPS6</i> 3UTRa	<i>HPS6</i> 3UTRa-F	AGAACAGTAGCTTGACTCCAGGTA	2	1
	<i>HPS6</i> 3UTRa-R	AGAACTTGGATGGCCCTTGTC		
	<i>HPS6</i> 3UTRa-probe	CCTCAAGCCAGGCTGC		
<i>HPS6</i> 3UTRb	<i>HPS6</i> 3UTRb-F	GGCCGAGCTCCCTTACTTG	2	1
	<i>HPS6</i> 3UTRb-R	GGACCAGCTGGACCAAGAG		
	<i>HPS6</i> 3UTRb-probe	TTGCTGCCCTGTGTCTG		
<i>HPS6</i> 3UTRc	<i>HPS6</i> 3UTRc-F	AGGACATATGAGCAGTTACCCAGAT	2	2
	<i>HPS6</i> 3UTRc-R	GGCACATCAGCACATCAGGTA		
	<i>HPS6</i> 3UTRc-probe	AAGGCAACATTCCCC		
<i>LDB1</i> ex9	LDB1ex9-F	CAGATCCAGAATACATACCCTTGCA	2	2
	LDB1ex9-R	TGTCTCATAGGCTTCACACCAAAA		
	LDB1ex9-probe	CAGTGCCGGTGTCTACT		

*Assays-on-demand (pre-manufactured) from Applied Biosystems