Contents of qPCR validation

Methods

Table S1. PCR Primers for qPCR

Primers	Sequence	Length	Tm
NPHP1-exon5-F	TCAACCGGTGAAGAATACATCG	123	60°C
NPHP1-exon5-R	CTGTTAGGTATGGACATCGACCC		
NPHP1-exon11-F	ACATTTTCATAAGCCGAATTCACAA	192	60°C
NPHP1-exon11-R	GGCGTACATGTCTGCTGAGAA		
AHI1-exon15-F	TGGACGTTTCATGAGAGAATTGTG	161	60°C
AHI1-exon15-R	GCTTTCCTTGACAGCAAACAGC		
AHI1-exon16-F	TCTGCCATATTGGTCCGACA	173	60°C
AHI1-exon16-R	TGCGCAATCATCAGTACATAACC		
β-globin-QF	ACACAACTGTGTTCACTAGC	110	60°C
β-globin-QR	CAACTTCATCCACGTTCACC		

Reaction reagents and condition

The expression level was assessed by real-time Quantitative Fluorescence PCR using SYBR Premis Ex Taq II (Perfect Real Time) (Takara) with ABI 7500 system. Data are presented as mean ± standard deviation of three independent real-time PCR experiments. The PCR cycle was as follows: 10 min 95°C, 1 cycle; 10 s 95°C, 30 s 60°C + fluorescence acquisition, 55 cycles. Values for each gene were normalized to expression level of beta-actin gene (ACTB) via the 2-ΔΔCT method.

Results

As demonstrated in the figures below, qPCR verified the CNV calling by WES.

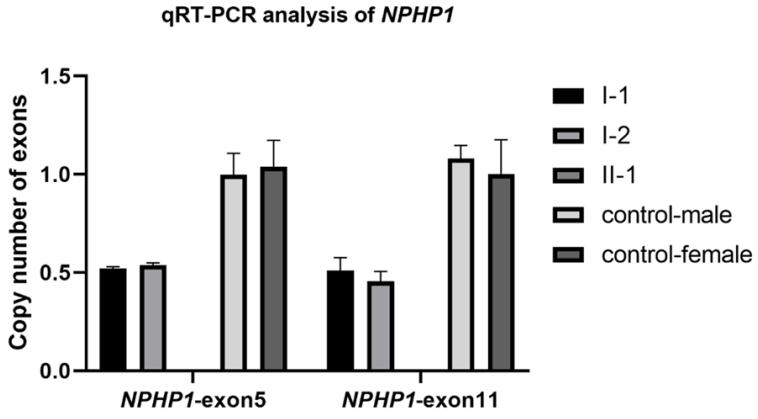

Figure S1. The qPCR results for exon5 and 11 in the NPHP1 gene on the samples from members of Case 2 and controls.

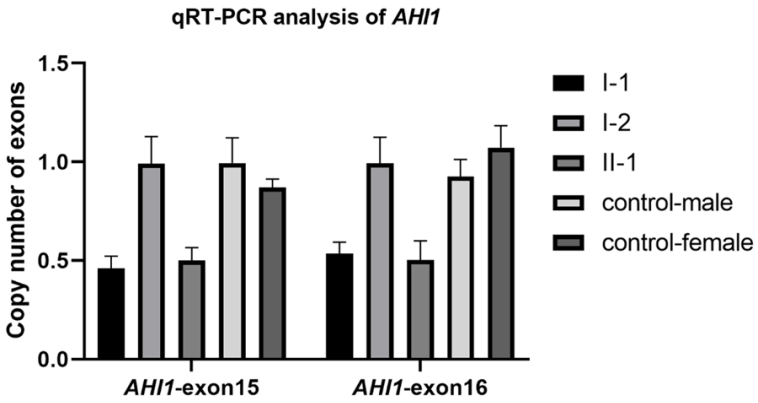

Figure S2. The qPCR results for exon15 and 16 in the *AHI* gene on the samples from members of Case 3 and controls.