Supplementary Materials and methods

gap-PCR

A gap-PCR was carried out to identify the large scale (Approximately 11.7 kb) exon 4–10 del in *IKBKG* (NM_001099857.5). The specific PCR using the F1R1 primers will produce a 3101-bp amplicon for exon 4-10 del allele. The same primers did not work for the intact allele because of too long distance of target genomic position, which was calculated as 14785bp.The specific PCR using the F1R2 primers will produce a 3308-bp amplicon for wild-type (WT) allele (Supplementary Fig.1). Meanwhile, a 524bp amplicon was amplified with G6PD-F1R1 primer for internal control. all primers were designed using Primer5 software. the gap-PCR primer sequences used（5’-3’）:

F1：GAGGACCAATACCGAGCATCTGA；

R1：CCCACGCTCTCCAGTGTGAAAG；

R2：AGAGCCTGGCATTCCTTAGTGG；

G6PD-F1：GACGGGGACACTGACTTCTGA；

G6PD-R1：AAAGGCGGTGTTTCGTGGA.

In brief, The gap-PCR was performed using the Premix Taq PCR Kit (RR003Q，Takara，dalian, China). In all the PCRs, 50 ng genomic DNA was used in a total reaction volume of 25 μL. The reaction system of gap-PCR was as follows: 95 °C for 1 min，1 cycle； 95 °C for 10 seconds，60 °C for 10 seconds, 72 °C for 3 min 30 seconds ,35 cycle；72 °C for 3 min 30 seconds , 4 °C for 5 min , 1 cycle. After all PCRs was completed，the reaction products were separated by 1% agarose gel electrophoresis.The specificity of the amplified fragment was confirmed using Sanger sequencing.

Long and accurate polymerase chain reaction(LA-PCR)

The LA-PCR was performed using the Platinum™ SuperFi™ II PCR Master Mix (12368010，Thermo Fisher Scientific，Waltham, MA, USA).The reaction system of LA-PCR was as follows: 95 °C for 1 min，1 cycle； 95 °C for 10 seconds，60 °C for 10 seconds, 72 °C for 9 min 30 seconds ,35 cycle；72 °C for 9 min 30 seconds , 4 °C for 5 min , 1 cycle.After all PCRs was completed，the reaction products were separated by 1% agarose gel electrophoresis. Cut the target band with a blade and recycle it. Then ,the specificity of the amplified fragment was confirmed using Sanger sequencing. the LA-PCR primer sequences used （5’-3’）： F1：GAGGACCAATACCGAGCATCTGA； R3：TCCAGGTGGCATCCCAGTTG；seq_exon10-R：CACTAATGCCCCTCACTTTCT.

Supplementary Figure 1 Genomic structures of *IKBKG* and *IKBKGP1*

Compared with *IKBKG* (NM:001099857.5), *IKBKGP1* (ENST000000612193.1) did not contain the exon 1 and exon 2. Hence，the forward primer F1 was located in intron 2 of the *IKBKG* gene and outside of *IKBKGP1* ’s homology regions.The reverse primer R1 was located at 5063bp downstream of exon10.The expected intact WT allele was 14785bp for F1R1 primers，but it did not work for the intact allele because of too long distance of target genomic position. The specific PCR using the F1R1 primers will produce a 3101-bp Amplicon for exon 4-10 deletion allele.A reverse primer R2 was located in exon4，the specific PCR using the F1R2 primers will produce a 3308-bp amplicon for wild-type allele. A reverse primer R3 was located in exon10, the specific PCR using the F1R3 primers will produce a 9254-bp amplicon. And there were two identical MER67B repeat sequences in exon3 and downstream of exon10.