

SUPPLEMENTARY MATERIALS

Supplementary Tables S1 and S2

Supplementary Figure S1

SUPPLEMENTARY TABLE S1. Information of the 11 mutations detected by ASPUA

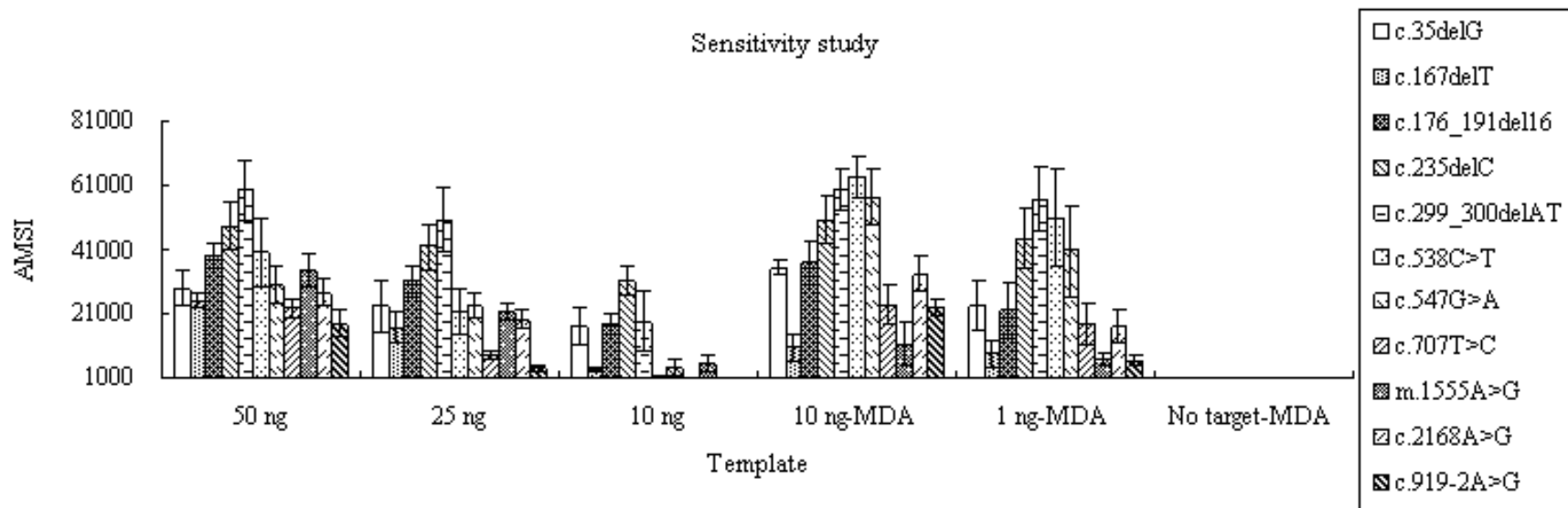
Gene and GenBank Accession	Nucleotide change	Amino acid change
GJB2 NM_004004.4	c.35delG	Frameshift
	c.167delT	Frameshift
	c.176_191del16	Frameshift
	c.235delC	Frameshift
	c.299_300delAT	Frameshift
GJB3 NM_024009.2	c.538C>T	p.Arg180X
	c.547G>A	p.Glu183Lys
SLC26A4 NM_000441.1	c.707T>C	p.Leu236 Pro
	c.2168A>G	p.His723Arg
	c.919-2A>G	Splice acceptor
MTRNR1 NC_001807.4	m.1555A>G	—

Nucleotide +1 in the cDNA reference sequence is the A of the ATG translation initiation codon.

SUPPLEMENTARY TABLE S2. The clinical information of the patient samples

Gene	Mutation^a	Clinical information	Number of subjects
GJB2	c.35delG hom	deaf	14
GJB2	c.35delG het	carriers	4
GJB2	c.35delG het	deaf	9
GJB2	c.35delG/c.299_300delAT	deaf	1
GJB2	c.35delG/c.167delT	deaf	2
GJB2	c.167delT hom	deaf	5
GJB2	c.167delT het	carriers	5
GJB2	c.167delT het	deaf	1
GJB2	c.176_191del16 hom	deaf	2
GJB2	c.176_191del16 het	deaf	3
GJB2	c.235delC/c.176_191del16	deaf	6
GJB2	c.235delC hom	deaf	11
GJB2	c.235delC het	deaf	5
GJB2	c.235delC/c.299_300delAT	deaf	3
GJB2	c.299_300delAT hom	deaf	3
GJB2	c.299_300delAT het	deaf	4
GJB3	c.538C>T (p.Arg180X) het	deaf	6
GJB3	c.547G>A (p.Glu183Lys) het	deaf	9
SLC26A4	c.707T>C (p.Leu236Pro) het	deaf	1
SLC26A4	c.919-2A>G hom	deaf	8
SLC26A4	c.919-2A>G het	deaf	4
SLC26A4	c.919-2A>G /c.2168A>G (p.His723Arg)	deaf	8
SLC26A4	c.2168A>G (p.His723Arg) het	deaf	6
MTRNR1	m.1555A>G hom	deaf	11
MTRNR1	m.1555A>G hom	nomal	1
	No mutations detected	nomal	9
Total			141

^a het stands for heterozygous; hom represents homozygous (hom represents homoplasmic for the m.1555A>G mutation).



SUPPLEMENTARY FIGURE S1. Detection sensitivity of ASPUA in genomic DNA. 50 ng, 25 ng and 10 ng of a wild-type gDNA were assayed, respectively. 10 ng and 1 ng of the gDNA and a target-free control were amplified by multiple displacement amplification (MDA), and then the MDA products were detected by ASPUA.