

表 3 非综合征耳聋 GJB2 致病性错义突变

Table 3 The pathogenic missense mutations of GJB2 in nonsyndromic hearing loss

隐性遗传突变			显性遗传突变	功能未明的突变
Recessive mutation			Dominant mutation	Unknown mutation
T8M	Y65X	K122I	W44S	N54I
G12V	W77R	Q124X	W44C	V84A
K15T	W77X	R127H	R75Q	T123N
S19T	L79P	W133X	R143Q	E129K
I20T	Q80X	Y136X	M163L	Y155X
W24X	Q80P	S139N	D179N	M163V
V27I+E114G	Q80R	R143W	R184Q	A171T
R32C	I82M	E147K	C202F	F191L
R32L	V84L	E147X		A197S
R32H	S85P	R165W		
V37I	A88S	W172X		
A40E	L90V	C174R		
A40G	L90P	P175T		
W44X	M93I	V178A		
G45E	V95M	R184W		
E47X	Y97X	R184P		
E47K	H100Y	S199F		
Q57X	H100L	I203K		
C64X	E101G	N206S		
Y65X	S113R	L214P		