Deafness genetic testing and prenatal diagnosis analysis of fourteen families
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Objective
The aim of this study is to discuss the role of the deafness genetic testing in disease screening and prenatal diagnosis, and provide genetic counseling to patients and their families.

Methods
Peripheral blood of 14 family members, the fetal amniotic fluid and fine hair were collected. Hereditary deafness gene chip and gene sequencing were used to detect the most common deafness genes in Chinese people. Genetic counseling was provided for the fetus prognosis according to the tests.

Results
There were 14 probands among 14 family members. We found out 9 cases of 235delC gene homozygous mutations, 4 cases of IVS7-2A>G gene homozygous mutations and 1 case of 235 delC and 299 delAT compound heterozygous mutation. There was 235 delC heterozygous mutation on both sides in 9 couples, IVS7-2A>G heterozygous mutation in 4 couples, 235delC and 299 delAT mutation in 1 couple. In 14 cases of prenatal diagnosis, 235 delC heterozygous mutation in 6 cases, 235 delC homozygous mutation in 2 cases, IVS7-2A>G heterozygous in 2 cases, IVS7-2A>G homozygous mutation in 1 cases, 299 delAT heterozygous mutation in 1 cases and 2 cases of wild type. The overall detection rate of abnormal 96.43% (54/56), prenatal diagnosis of abnormal rate was 85.71% (12/14), homozygous mutation rate of 21.43% (3/14).

Conclusion
When deafness genetic testing is applied in disease screening, the disease and the definite etiology can be found and the disease progression can be slowed down through early intervention. When it is applied in prenatal diagnosis, fetal prognosis can be evaluated and accurate genetic counseling can be provided.