



Application of Deafness Gene Detection in Diagnosis and Genetic Counseling of Hereditary Deafness

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Abstract

Objective To explore the clinical value and genetic counseling value of deafness gene diagnosis in the diagnosis of hereditary deafness. **Methods** the study samples were selected from January 2021 to December 2021. 100 voluntary gene testers were studied in our hospital, and genetic counseling was provided to count the test results. **Results** 23 patients carried gene mutations, 1 GJB2 - 167del homozygous mutation, 8 compound heterozygous mutations and 2 double heterozygous mutations. 12 cases of heterozygous mutation. **Conclusion** the implementation of deafness gene detection in the diagnosis of hereditary deafness can clarify the mutation type and the occurrence probability of deafness in offspring, and provide reference for subsequent marriage and childbirth, which has important clinical value.

Keywords

Hereditary deafness; Gene testing; Prenatal testing; Genetic counseling; Gene mutation

Hereditary deafness is a relatively common congenital disease in clinical practice. In China, the incidence of congenital deafness in newborns is about 0.1%, of which about 50% of the cases are caused by genetic factors [1]. Regardless of the cause, deafness will cause serious cognitive impairment in patients, resulting in a lack of observation and learning ability of the world, which will cause a serious burden on the patient's mental health, life, learning, work and family and society [2]. At present, the treatment of hereditary deafness is difficult and progress has stalled. Most scholars believe that a preventive strategy should be adopted for this disease [3-4]. That is, deafness gene testing should be carried out before pregnancy, before birth and before marriage to clarify the probability of hereditary deafness. At the same time, it can improve the understanding of the types of gene mutations carried by the counselor, patients and their families, under what circumstances they may develop deafness and the probability of occurrence, so as to achieve the purpose of preventing the occurrence of deafness [5]. Based on this, this study used PCR+channel hybridization and direct sequencing to investigate 100 volunteers who underwent gene testing at our hospital between January and December 2021, and provided genetic counseling, in order to confirm the clinical value of implementing gene testing for deafness and the value of genetic counseling in the diagnosis of hereditary deafness. The full text is as follows.

1. Materials and Methods

1.1 General Information

This study included 100 volunteers who underwent genetic testing at our hospital between January and December 2021. The participants ranged in age from 6 to 37 years, with a mean age of (23.33 ± 7.63) years. There were 52 females and 48 males.

1.2 Method

2ml of venous blood should be drawn from the patient the following morning using a disposable sterile negative pressure blood collection tube or a disposable sterile syringe and placed in a sterile container. The blood should be extracted using the Xi'an Tianlong Ex-DNA Whole Blood Genome (3.0) kit. For detailed operating procedures, please refer to the kit instructions.

Use 3 μ L of the extracted DNA sample as a template for PCR amplification; store the remaining DNA sample at -20°C for later use (store at -20°C for no more than 3 months, and freeze-thaw no more than 5 times).

The specific process is as follows:

(1) The PCR products (Group A and Group B) were denatured at 95°C for 5~10 min, then removed and placed in an ice box for later use. The hybridization experiment was carried out at 45°C with the hybridization instrument ready.

(2) Place the accessories as required, lay out the hybridization membrane, add 0.8 mL of hybridization solution preheated to 45°C into the hybridization well, cover with the cover plate and incubate for at least 2 minutes, then turn on the pump to discharge and turn off the water pump;

(3) Add the denatured PCR product DNA (group A and group B) solution prepared in step 1 to 0.8 mL of hybridization solution preheated to 45 °C, mix well, then add it to the membrane, cover with the cover plate and incubate for 20 min, then turn on the pump to perform flow hybridization.

(4) At 45°C, rinse the membrane four times with 0.8 mL of elution buffer 1 (WB1) preheated to 45°C, alternating between the two solutions. After washing with WB1, adjust the temperature of the hybridization solution to 25 °C.

(5) Add 0.5ml of blocking liquid for pre-blocking. When the temperature drops to 30 °C, turn on the pump to drain the liquid, turn off the pump, add 0.5ml of blocking liquid, block for 5 minutes, and then turn on the pump to drain the liquid.

(6) Add 0.5 ml of enzyme-labeled solution, let stand for 5 minutes, then turn on the pump to drain the solution.

(7) Wash 4 times with 0.8 ml of solution A. Set the temperature to 36 °C during the second wash. When the hybridization instrument reaches 36 °C, add 0.5 ml of display solution, close the hybridization instrument lid, and let it develop color for 6 minutes.

(8) Wash three times with 0.8 ml of hybridization solution, and finally wash twice with distilled water.

Note: The temperature of the hybridization solution should be kept at 45°C for all the above operations.

The reagents used in this study were provided by Guangzhou Kaipu Pharmaceutical Technology Co., Ltd., and were used for the *in vitro* qualitative detection of nine mutation sites (mtDNA 1494, mtDNA 1555, SLC26A4 -IVS7(-2), SLC26A4-2168, GJB2-35, GJB2-176, GJB2-235, GJB2-299, GJB3-538) of deafness-related genes (GJB2, GJB3, SLC26A4, mtDNA) in human venous whole blood.

After confirming the patient's genetic information regarding deafness, the examinee's genotype is determined, the mutation site is identified, and the examinee is provided with detailed genetic counseling, as well as various intervention measures and treatment methods.

1.3 Observation indicators

The types of lesions in 100 patients in this study were statistically analyzed.

Standard consultation and answers.

2. Result

2.1 Test Results

Of the 100 individuals who underwent gene chip and direct sequencing testing in this study, 33 underwent premarital testing, 25 underwent preconception testing, and 42 underwent prenatal testing. Among them, 11 patients carried gene mutations: 6 cases of heterozygous SLC26A4 -IVS7 (-2) mutation, 3 cases of heterozygous GJB2-235 mutation, and 2 cases of mtDNA 1555 mutation.

2.2 Consultation and Answers

hearing loss and non-hearing loss: This is a common genetic testing scenario. Taking this consultation as an example, Wang XX and Li XX are a married couple. Li XX has severe hearing impairment, but otherwise has no abnormalities and normal intelligence. Genetic testing revealed that the woman's test was negative, while the man carried the mtDNA 1555 mutation. An investigation of the man's family revealed that his parents have normal hearing and

intelligence, while his mother carries the mtDNA 1555 mutation. The clients were randomly informed about the inheritance patterns of the mtDNA 1555 mutation; if the couple has children, there is a very high probability that their newborn will inherit the mtDNA 1555 mutation, but the probability of deafness is relatively low.

Normal Case: During a premarital checkup, Ms. XX tested positive for GJB2-235 and SLC26A4: IVS7-2A>G mutations, while the man tested negative. They were informed of the inheritance patterns of GJB2 and SLC26A4, and that if they marry and have children, their children will definitely carry GJB2 and SLC26A4. There is also a certain risk of hereditary deafness. If they proceed with the marriage and have children, prenatal testing should be conducted to reduce this risk.

Prenatal checkup: This patient presented with a typical case of hearing impairment in her firstborn child. During genetic counseling for her second pregnancy, the male was found to have a complex heterozygous mutation in mtDNA 1555 and GJB2-235, while the female was negative. Her firstborn child also tested positive for the GJB2-235 heterozygous mutation. Amniocentesis also confirmed the GJB2-235 heterozygous mutation. Given the low probability of GJB2-235, the couple was informed about its inheritance patterns and incidence. They decided to continue the pregnancy. They were also advised that if their newborn marries someone with the same gene mutation in adulthood, their offspring would have a higher probability of deafness, and they should proceed with caution.

3. Discuss

Hereditary deafness is a common special disease in clinical practice. There is currently no effective treatment for this disease. After the onset of the disease, it will have a serious impact on the patient's life and affect their family and society. Therefore, it is necessary to reduce the incidence of hereditary deafness through corresponding intervention measures [6].

Early diagnosis of deafness generally relied on hearing screening, imaging tests, and biochemical examinations. While these tests could determine the severity of deafness and whether it was a structural disorder, they could not identify the cause or differentiate the disease, resulting in very limited effectiveness. They provided almost no reference for clinical intervention guidance, making it difficult to provide relevant guidance based on general screening. Moreover, early diagnostic screenings could not confirm the incidence of deafness in offspring, often failing to provide accurate answers during genetic counseling, which was generally based on experience [7-18]. However, with advancements in medical technology, deafness genes have been gradually identified. Deafness gene screening in East Asian populations has clarified common mutation types, enabling direct clinical application. To this day, genetic screening remains a classic screening method for hereditary diseases. It can identify the type of gene mutation in a person, and based on this, it can analyze the incidence of hereditary deafness in offspring. At the same time, genetic screening can confirm deafness at an early stage, and measures such as cochlear implantation can be taken before language learning, which can effectively improve the language learning ability of people with hereditary deafness, enhance their social adaptability, and reduce the impact of the disease on their families and society.

Proactive premarital and prenatal screening for deafness genes can relatively accurately predict the risk of offspring carrying deafness genes and clarify the probability of deafness. Based on this, reproductive guidance can be provided to relevant populations. This measure should be a universal screening measure in marriages and pregnancies involving individuals with pre-existing deafness or severe hearing impairment. It can assess fetal prognosis, guide eugenic practices, and help reduce the transmission and incidence of hereditary deafness within families. Furthermore, with the advancements in gene chip and gene testing technologies, its application is becoming easier and its diagnostic efficiency higher. In the near future, it may become a basic measure in prenatal checkups.

In conclusion, performing deafness gene testing in the diagnosis of hereditary deafness can identify the mutation type and determine the probability of deafness in offspring, providing a reference for subsequent marriage and reproduction, and has important clinical value.

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