Newborn Screening for Congenital Hearing Impairment: A 17-Year Experience for Improving Quality of Healthcare

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Abstract

Background: To improve healthcare quality, a preventive intervention system has been developed globally with routine newborn screening, diagnosis, intervention, and rehabilitation for congenital hearing impairment. An intervention program with Newborn Hearing Screening (NHS) was implemented in Guangdong Province, China, in 1999, along with molecular screening for common inherited deafness mutations since 2011. Methods: We evaluated the clinical screening data for congenital hearing impairment and the results of genetic testing for deafness genes that were collected by the Guangdong provincial administrative agencies and stored in the provincial healthcare intervention system. Results: NHS rates varied from <30% in eastern Guangdong to >95% in the provincial capitol. This difference in rates correlated primarily with the regional per capita Gross Domestic Product (GDP; R²=0.69, p<.001) and the average maternal hospitalization days (R²=0.11, p=.01). The province-wide screening rate increased from 30.0% in 2008 to 80.58% in 2016. Combined screening for hearing and deafness genes was conducted in several hospitals in developed regions. The total mutation rate of hearing and deafness genes ranged from 3.08% to 4.5%, with GJB2 235 del C being the most frequently encountered mutation. Three hospitals conducted genetic screening of pregnant women for hearing and deafness genes, and the rate of mutation carrier ranged from 3.16% to 4.17%. Conclusion: To further improve healthcare quality, hospitals are recommended to implement hearing screening and diagnostic procedures, networking platforms, genetic diagnostic testing, and genetic counseling services. Furthermore, there is an urgent need for more referral molecular testing laboratories to perform molecular diagnosis of hearing loss.


Introduction

Hearing impairment is a common congenital anomaly that can impede the interpersonal interactions and social development of children, occurring in approximately one to three infants per 1,000 live births. Prevention of and intervention for congenitally impaired hearing has become a global public health concern [1-3]. The coverage rate of neonatal hearing screening was over 95% in 2012 in the U.S. [4], whereas it ranged from below 30% to more than 95% in 2011 in different regions of Italy [4]. In 1999, China’s Ministry of Health specified that “Newborn Hearing Screening (NHS) should be included as a routine test in maternal and childcare to ensure early detection and early intervention of hearing impairment” [5]. In 2007, the National Program for Hearing Impairment Prevention and Rehabilitation (2007-2015) policy declaration released jointly by China’s Ministry of Health, Ministry of Education, and Ministry of Civil Affairs stated that the NHS rate should increase by 30% over the 2005 level by 2015 in geographic areas where NHS is already available [6]. Since 2000, the Chinese central government has developed various regulations, national plans, guidelines, and technical criteria for a universal NHS program. However, implementation of the program was at the discretion of provincial or local governments, which vary significantly in their economic development and available resources to implement these programs [7]. Guangdong is the most populous province in China, with a population of 107,240,000 residents [8]. The province is a popular destination for migrant workers because it has been a leader in policy reform in China and offers desirable employment opportunities owing, in part, to its special economic zone status. The large population and high population mobility make implementation of prevention and intervention programs particularly challenging in the province. Hence, with the aim of providing guidance for future development, we reviewed the progress and performance of Guangdong’s prevention and intervention system for neonatal deafness over 17 years, from 1999 to 2016. We identified weaknesses, made forward-looking recommendations, and devised a model that should improve the program’s quality in the future.

Materials and Methods

Ethical standards

The authors assert that all procedures were conducted in compliance with the ethical standards of the relevant national and institutional guidelines on human experimentation and with the Helsinki Declaration. The authors obtained written informed consent from the guardians of the newborns in the study. This study was approved by the Hospital Ethical Committee of the Third Affiliated Hospital of Sun Yat-sen University.

Data collection

Data presented in this study were extracted from the provincial NHS database, which was developed by the provincial government agency. This database contains information about NHS programs in four administrative districts of Guangdong (the east, the west, the north, and the Pearl Delta districts). For the NHS program, the database collected information on the numbers of livebirths and the numbers and percentages of newborns who received primary NHS each year. The Pearl Delta district includes the cities of Guangzhou, Foshan, Dongguan, Shenzhen, Zhongshan, Zhuhai, Huizhou, Zhaoqing, and Jiangmen, which comprise the leading economic region and contain the largest number of university-affiliated academic hospitals and tertiary hospitals in this province. To assess the NHS status and influential factors of each administrative region, we analyzed days of maternal hospitalization, Gross Domestic Product (GDP), number of trained staff with NHS knowledge, and NHS rates per calendar year. We also evaluated the local medical records regarding implementation of NHS and hearing diagnosis in children.

Deafness genetic screening

Information regarding molecular screening for common genetic loci/mutations for deafness was collected from questionnaires from 22 of 32 university-affiliated hospitals that were invited to complete questionnaires (68.8% response rate). The remaining 10 facilities were followed by telephone interview, resulting in a 100% questionnaire completion rate. The collected information consisted of (1) when and with whom the genetic screening was conducted, (2) which genetic loci were screened, (3) how the screening results were shared, and (4) How genetic counseling was performed.

Literature Review

The SinoMed, PubMed, China National Knowledge Infrastructure, and Wanfang Data databases and the VIP Chinese Science and Technology Journal Database were searched with the key words “newborn, neonatal, infant,” “hearing screening,” “otoacoustic emission,” “auto-auditory brainstem response,” and “deafness genetic screening” for “Guangdong Province.” The publications in the search results were screened to remove articles with duplicate samples, review articles, and monographs. Only publications that used data from health care providers located in Guangdong were used for subsequent analyses. Outcome measures included the Primary Screening Rate (PSR), secondary screening rate, deafness detection rate, and mutation carrier rate.
Statistical Analysis

Statistical analysis was performed with SPSS 19.0 software (SPSS, Chicago, IL, USA). To determine the factors influencing the PSR, multiple stepwise regression analysis was conducted with the independent variables, including average maternal hospitalization days, regional GDP per capita, and regional number of trained staff with knowledge of NHS.

Results

Implementation of NHS in Guangdong province (2009-2016)

Between 1999 and 2001, NHS was conducted in six metropolitan cities in Guangdong province, all of which were located within the Pearl Delta district. By the end of 2004, NHS was being conducted throughout the entire Pearl Delta area. Implementation of NHS in the west district began in 2005, financial coverage for which was not achieved until 2007. Subsequently, NHS was implemented in the east district, starting with the three coastal cities surrounding Shantou University, between 2007 and 2009. Finally, NHS was implemented in the inland northern area between 2009 and 2011, thereby marking achievement of province-wide NHS in 2011 (Figure 1). More than 75% (98/130) of the tertiary hospitals in Guangdong province are concentrated in the Pearl Delta district, and nearly 35% (45/130) are in Guangzhou, the capital city of the province. Furthermore, the distribution of tertiary hospitals is closely related to regional GDP (Figures 2A and 2B).

Figure 1: Course of NHS implementation in Guangdong province. The thick line defines the Pearl Delta area, including Guangzhou, Foshan, Dongguan, Shenzhen, Zhongshan, Zhuhai, Huizhou, Zhaoqing, and Jiangmen.

Figure 2: Hospitals and GDP of Guangdong Province. (A) Distribution of all 130 tertiary hospitals in Guangdong Province in 2014. Among the 130 tertiary hospitals in Guangdong Province, 98 (75.4%) were concentrated in the Pearl Delta Area, and 45 were in Guangzhou, the province capital. (B) Regional GDP structure of Guangdong Province in 2014. The Pearl Delta accounted for 78.9% of the provincial GDP.

Comparison of screening rates among the provincial capital and administrative districts

Between 2008 and 2016, the number of screened newborns in the entire province increased from 300,000 to 999,530 (Figure 3). The provincial PSR increased gradually but remained below 80% in 2015. Meanwhile, the PSR in the capital city was more than 90% and reached 100% after 2012 (Figure 4). The PSRs from the four administrative districts varied, ranging from <30% in the Chaozhou-Shantou area (eastern district) to > 80% in the Pearl Delta region (Figure 4). Multiple stepwise regression analysis indicated that the average number of maternal hospitalization days ($R^2=0.11$, $p=.01$) and the regional GDP per capita ($R^2=0.69$, $p <.001$) were associated with the PSR, whereas the regional number of trained staff with knowledge of NHS was not a significantly related factor. The rate of secondary screening, rate of detection, and diagnostic techniques employed in Pearl Delta area hospitals are shown in Table 1. The primary screening pass rate was stable across the hospitals in the area (81.3-87.4%). The number of diagnostic techniques employed varied from two to four, and detection rates varied widely from 0.128% to 6.86%.
**Figure 3:** Live births and screened cases in Guangdong, 2008-2016. Newborn screenings increased rapidly from 480,061 cases to 858,420 cases between 2011 and 2014.

**Figure 4:** Comparisons of newborn hearing Primary Screening Rates (PSRs) among Guangdong’s provincial capital and four administrative regions, 2008-2016. The PSR in the province (green line) increased rapidly from 39.7% to 67.1% between 2011 and 2014. The PSRs of the administrative districts varied widely, ranging from <30% in eastern Guangdong to >80% in the Pearl Delta region. PSRs in the capital were consistently >90%.
### Table 1: NHS and diagnosis rates in testing facilities in the Pearl Delta area.

<table>
<thead>
<tr>
<th>Screening facility (year range, no. cases)</th>
<th>Primary screening Pass rate (%)</th>
<th>Secondary screening rate (%)</th>
<th>Detection rate (%)</th>
<th>Diagnostic techniques</th>
</tr>
</thead>
<tbody>
<tr>
<td>The Third Affiliated Hospital of Sun Yat-sen University (2006-2014, 39,200)</td>
<td>86.6</td>
<td>85</td>
<td>0.22</td>
<td>ABR, OAE, acoustic immittance, 40-Hz AERP</td>
</tr>
<tr>
<td>Maternal and Child Hospital of Guangdong Province (2012-2013, 18,049)</td>
<td>81.3</td>
<td>84</td>
<td>4.9*</td>
<td>OAE, ASSR</td>
</tr>
<tr>
<td>Guangzhou Women and Children Medical Center (2009, 163,768)</td>
<td>86.54</td>
<td>64.1%</td>
<td>0.22</td>
<td>ABR, OAE, acoustic immittance, 40-Hz AERP</td>
</tr>
<tr>
<td>Zhuhai Maternal and Child Health Hospital (2002-2004, 4,666)</td>
<td>85.5</td>
<td>88</td>
<td>6.86*</td>
<td>ABR, OAE, acoustic immittance, ASSR</td>
</tr>
<tr>
<td>Jiangmen Central Hospital (2009-2011, 4,676)</td>
<td>85.07</td>
<td>47.75</td>
<td>0.128</td>
<td>ABR, OAE, acoustic immittance, ASSR</td>
</tr>
<tr>
<td>Four hospitals in Pearl Delta (2001-2003, 33,706)</td>
<td>85.45</td>
<td>78</td>
<td>1.32</td>
<td>OAE, ASSR</td>
</tr>
<tr>
<td>Hospital of Traditional Chinese Medicine of Zhongshan (2010-2011, 29,657)</td>
<td>87.4</td>
<td>88.9</td>
<td>0.13</td>
<td>ABR, OAE, acoustic immittance, ASSR</td>
</tr>
<tr>
<td>Total (2001-2014, 293,722)</td>
<td>85.41</td>
<td>76.53</td>
<td>1.97</td>
<td>*</td>
</tr>
</tbody>
</table>

*Data were collected from governmental statistics and published articles. All searched articles were related to the Pearl Delta area. The diagnosis rate was calculated on the basis of the number of cases. Hearing loss may occur in one or both ears. Confirmed cases included referred cases from other hospitals that failed NHS. Combinations of five objective diagnostics were used to cross-validate the nature and degree of hearing loss. ABR: Auditory Brainstem Response; OAE: Otoacoustic Emission; ASSR: Auditory Steady-State Response; AERP: Auditory Event-Related Potential.

### Government guidance and promotion

In 2007, the Grassroots Health and Maternal and Child Health Division of the Guangdong Provincial Public Health Commission convened an expert panel, and an expert team was assembled by the provincial governmental administrative agency to develop hearing-diagnostic centers for children from birth to 3 years of age. To address multiple problems, such as deficits in secondary screening, follow-up diagnosis, and intervention, the centers would be expected to accept referred newborns who did not pass hearing screening tests in less-developed areas. In addition, the panel was also deemed responsible for providing workshops to train center staff across the province in hearing screening procedures and to improve their diagnostic knowledge. In early 2008, selected hospitals that had performed NHS in the Pearl Delta area were reviewed according to the requirements of the “Technical Specifications for Newborn Screening” released by China’s Ministry of Health in 2004. A total of 16 hospitals in the province were accredited to conduct hearing tests for children ages 0 to 3 years by the end of 2012, which promoted hearing screening tests in less-developed areas. Hospitals with members on the panel have held 20 screening/diagnosis training workshops since 2007, benefiting up to 3,200 trainees. Although participation in the workshops has created opportunities for trainees to establish contact with screening facilities and diagnostic centers, no permanent referral or guidance relationships have been established. As a result, several children who did not pass NHS did not obtain timely follow-up services in diagnosis, intervention, and consulting. In June 2015, the Guangdong provincial government spent ¥1.6 billion on free birth defect screening, including NHS, in 84 poor counties. In November of the same year, the Guangdong provincial government contributed 39 NHS devices to poverty-stricken counties. Local governments also provide continued support for the NHS project. Since 2014, a number of economically developed regions (e.g., the cities Huizhou, Zhongshan, Foshan, Zhuhai, and Guangzhou) implemented free NHS for babies born within their jurisdictions. After more than 10 years of expansion of the NHS program, Guangdong has established a stratified system of hospital management according to screening agency levels (Figure 5).
Figure 5: Management of NHS in Guangdong province. After more than 10 years of expanding the NHS program, Guangdong has established a stratified hospital management model.

Development and promotion of NHS information management software

A research team at Zhuhai Maternal and Child Health Hospital began developing a hearing screening information management system in 2007 [9]. In 2010, this system was patented in China and has been adopted in several hospitals. With this system, diagnostic centers in Zhongshan city and Zhuhai city now accept referred newborns who do not pass NHS in their local facilities and accept follow-up referrals for newborns of migrant workers in their jurisdiction. This network system is expanding coverage gradually, but has not yet been adopted across the entire province.

Coverage and effectiveness of genetic screening for deafness

The Maternal and Child Health Hospital of Guangdong Province began conducting screening tests for neonatal deafness-susceptibility genes in 2011. Subsequently, Guangzhou Women and Children Medical Center and Foshan Maternal and Child Health Hospital, the Third Affiliated Hospital of Sun Yat-sen University, and Zhuhai Maternal and Child Health Hospital followed suit in 2012, 2013, and 2014, respectively. Neonatal screening for deafness genes is now offered as a routine genetic screening test in these five hospitals. The Third Affiliated Hospital of Sun Yat-sen University (Unit 1), the Maternal and Child Health Hospital of Guangdong Province (Unit 2), and the Foshan Maternal and Child Health Hospital (Unit 3) used nine loci of four genes to screen for deafness genes, and their screening results are shown in Table 2. Six additional hospitals provide genetic screening for children with a confirmed diagnosis of hearing impairment; however, their results have not been reported formally. The results from three hospitals have indicated that the total mutation rate ranges from 3.08% to 4.5%, with GJB2 235 del C having the highest mutation rates (1.65-2.1%), which were higher than that in the general Chinese population (0-1%). The identified mutations are summarized in Table 2. Three hospitals (the Maternal and Child Health Hospital of Guangdong Province, Southern affiliated hospital, Southern Medical University, and Foshan Nanhai Maternity and Childcare) conduct deafness gene screening for pregnant women, with the aim of implementing secondary prevention of hereditary deafness, as shown in Figure 6. The Maternity and Childcare Hospital of Guangdong Province reported a 4.17% carrier rate among 7,263 women of childbearing age. Further genetic tests were performed on the husbands of 95 of these carriers. Eight couples who were found to harbor deafness-causing mutations in the same gene received prenatal diagnosis [10]. Foshan Nanhai Maternity and Childcare Center screened 3,950 expectant mothers and obtained a carrier rate of 3.16%. Further genetic testing was performed in two of these carriers’ husbands. No formal genetic testing reports have been released from Nanfang Hospital yet. Follow-up genetic testing occurred at the Third Affiliated Hospital of Sun Yat-sen University. To further enrich and clarify the combination of different GJB2 or SLC26A4 mutations and their auditory phenotypes, PCR sequencing was conducted for newborns with congenital deafness with GJB2 or SLC26A4 single heterozygous mutations, and the deafness gene targeting technique will be used.
to sequence the known deafness genes in congenitally deaf newborns with negative deafness genetic screens. We identified 15 cases with mtDNA12S rRNA mutations. The infants’ families were provided with medical guidance brochures advising them to avoid the use of potentially ototoxic drugs. GJB2 heterozygous mutations were observed in 145 newborns, and their guardians were instructed to observe and monitor their auditory behavior. Three children with late-onset hearing loss, i.e., hearing loss that follows a passed newborn hearing screening and occurs before until 6 years of age, were identified in follow-up examinations, which enabled the implementation of early intervention methods. In addition, SLC26A4 heterozygous mutations were detected in 49 newborns, and their guardians were given advice regarding how they could avoid daily triggers (such as exposure in ototoxic drugs, injury or foreign object in the ear, or exposure to loud noise) and how to observe their auditory behavior. Late-onset hearing loss was identified in one patient with an SLC26A4 heterozygous mutation who was diagnosed with bilateral enlarged vestibular aqueducts on the basis of temporal computed tomography findings.

<table>
<thead>
<tr>
<th>Nucleotide change</th>
<th>Unit 1 N = 8,200</th>
<th>Unit 2 N = 17,659</th>
<th>Unit 3 N = 10,238</th>
<th>General population (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>GJB2 35 del G</td>
<td>0.01</td>
<td>0.01</td>
<td>-</td>
<td>0-0.2% [17,18]</td>
</tr>
<tr>
<td>176 del 16</td>
<td>0.08</td>
<td>0.1</td>
<td>0.06</td>
<td>-</td>
</tr>
<tr>
<td>235 del C</td>
<td>2.10</td>
<td>1.77</td>
<td>1.65</td>
<td>0.1% [17,18]</td>
</tr>
<tr>
<td>299 del AT</td>
<td>0.34</td>
<td>0.39</td>
<td>0.20</td>
<td>0-0.5% [17,18]</td>
</tr>
<tr>
<td>GJB3 538 C &gt; T</td>
<td>0.11</td>
<td>0.12</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>SLC26A4 2168 A &gt; G</td>
<td>0.16</td>
<td>0.26</td>
<td>0.12</td>
<td>-</td>
</tr>
<tr>
<td>IVS7-2 A &gt; G</td>
<td>0.95</td>
<td>1.16</td>
<td>0.11</td>
<td>-</td>
</tr>
<tr>
<td>mtDNA12S 1494 C &gt; T</td>
<td>0.03</td>
<td>0.04</td>
<td>0.01</td>
<td>-</td>
</tr>
<tr>
<td>rRNA 1555 A &gt; G</td>
<td>0.21</td>
<td>0.03</td>
<td>0.22</td>
<td>-</td>
</tr>
<tr>
<td>Total</td>
<td>4.20</td>
<td>4.50</td>
<td>3.08</td>
<td></td>
</tr>
</tbody>
</table>

Table 2: Deafness gene screening.

Figure 6: Work-flow chart of the deafness genetic screening protocol for pregnant women. Three hospitals conduct deafness gene screening for pregnant women, with the aim of implementing secondary prevention of hereditary deafness.

Genetic counseling services after combined screening for hearing and deafness genes

Genetic counseling in the five hospitals where genetic screening for deafness is offered routinely includes reporting of prenatal diagnostic laboratory screening results to parents or guardians and a preliminary consultation followed by deafness-related consulting and intervention guidance for children with hearing impairment at the otology clinic.

Interventions for, and health care of, confirmed neonatal hearing impairment

Children who are confirmed to have permanent sensorineural deafness are managed with hearing aids or cochlear implants, depending on the degree of their present hearing loss. These interventions are paid for by government funding in cases in which the children’s families are impoverished. Otherwise, they are paid for by the family because the devices are not yet covered by medical insurance. The goal of NHS is to ensure that hearing-impaired children will achieve optimal language development. Thus, although NHS begins as a medical intervention, patient outcomes are dependent upon language rehabilitation and educational support. Outcomes are limited by the fact that there is a shortage of hearing rehabilitation institutions and personnel. By the end of 2014, only 68 hearing rehabilitation facilities were providing hearing rehabilitation services for deaf children in Guangdong province, with a combined annual charge capacity of only 1,222 cases [8], which is insufficient to meet the demand. Consequently, a Department of Hearing was established in June of 2014 at Sun Yat-sen University’s Xinhua Institute with the purpose of training hearing rehabilitation professionals.
Discussion

Progress and factors affecting NHS in Guangdong

NHS began in the central Pearl Delta area in 1999 and gradually spread throughout the entire province over the 12-year period from 1999 through 2011. Implementation was initially focused in the Pearl Delta area and west Guangdong, and then spread to the east and north Guangdong regions, as shown in Figure 1. Implementation of NHS was propelled by university-affiliated hospitals first, and the course of implementation progress was closely related to the distribution of university-affiliated hospitals and other tertiary hospitals, which played a leading and important role in the adoption of NHS (Figure 2A). The distribution of tertiary hospitals was closely related to regional GDP, as shown in Fig. 2B. Screening rates increased annually, and by 2012, more than 820,000 newborns were receiving hearing screening annually in the province, as shown in Fig. 3. With the observed overall hearing impairment detection rate of 1.97% (Table 1), 5,787 hearing-impaired children benefited from NHS and received appropriate medical intervention, thereby reducing or avoiding deafness-related developmental and communicative sequelae. However, NHS implementation differs considerably across regions, with rates in some remote mountainous areas being less than 33% of the rate observed for the capitol city (Figure 4). There are several possible causes for this disparity. First, newborn families often fail to recognize the importance of screening and may be reluctant to have their children screened. Guardian education level is highly related to the decision to undergo NHS. Second, the costs of NHS are primarily covered by families and only partially covered by medical insurance [11]. Therefore, families with financial difficulties may choose against NHS due to prohibitive costs. Typically, hospitalization periods for delivering mothers are less than 3 days for low-income families. In this study, we found that regional GDP per capita and average hospitalization days were both significant factors related to screening rate. Third, whether the staff receives professional training also affects NHS rates. After 2011, governmental and administrative guidance and promotion led to a marked increase in screening rates, with the provincial PSR reaching approximately 70% by the end of 2014 (Figure 4).

Newborn hearing secondary screening, diagnosis, and related factors

NHS enables early detection and early intervention for hearing loss; however, secondary screening and diagnosis are also crucial. At the end of the study period, several NHS weaknesses remained, including insufficient secondary screening, follow-up diagnosis, and intervention after primary NHS, particularly in less-developed regions. After three rounds of discussions, the expert panel convened by the provincial government in 2007 (introduced above) proposed the following three solutions: create hearing diagnosis centers for infants and toddlers (ages 0–3 years) in conditional areas; provide expert guidance at regular intervals to personnel involved with NHS in less-developed areas; and provide annual NHS and early childhood hearing diagnosis training courses. By the end of 2012, a total of 16 hospitals in the province were accredited to conduct hearing tests for children 0 to 3 years of age. Furthermore, six experts provided direction guidance hospital staff in three less-developed administrative areas, and steering group members have held 16 training workshops on hearing screening and diagnosis in multiple hospitals, benefiting over 2,500 trainees. These steps resolved, in part, the referral problem for children from less-developed areas who failed the NHS tests. However, 16 diagnostic centers do not have stable cooperative relationships with screening facilities across the province; consequently, the total secondary screening and referral rates were still below 80% at the end of the study period (Table 1). The development and promotion of information management software may further improve referral rates. A new screening network platform was piloted in selected hospitals in the Pearl Delta area, a convenient and efficient location for secondary screening and referrals; however, the network platform has not been adopted across the entire province. With the aim of implementing a network platform across the entire province, a trial-basis network management operation mechanism was established in 2014 for information-sharing and collaboration between government agencies, expert group and screening facilities, and diagnostic centers, which is available to centers across the entire province (Figure 5). The present data show significant variation in the diagnostic techniques employed and the detection rates across the selected hospitals, as shown in Table 1. No uniform standards regarding the combination of techniques have been implemented for hearing diagnosis. For example, several hospitals depend on only otoacoustic emission and auditory steady-state response results for diagnosis (Table 1).

Achievements and shortcomings of combined hearing and genetic screening for deafness in newborns

Five hospitals in Guangdong began routinely conducting combined hearing and genetic screening for deafness in 2011, with another six hospitals providing genetic testing and diagnosis for deafness in children with confirmed hearing impairment. According to the screening results of three hospitals (Units 1, 2, and 3) that use the same screening method, the mutation rate of deafness susceptibility genes in newborns ranged from 3.08% to 4.5%, with the GJB2 235 del C mutation being observed the most often (1.46-2.1%), as shown in Table 2. The Third Affiliated Hospital of Sun Yat-sen University conducted follow-up examinations of infants with positive genetic screening results and observed that late-onset
Suggested measures to improve the quality of deafness prevention and control in Guangdong

A multi-pronged approach should be utilized to improve deafness prevention. First, the network management platform for deafness screening must be improved. Furthermore, information sharing and interaction among government agencies, expert groups, and screening facilities must be promoted. Second, better training must be provided to staff who are involved with hearing screening, genetic screening, diagnosis, and genetic counseling. Finally, experts should be asked to develop education materials. Furthermore, mass media resources, such as the internet, television, radio, and WeChat, should be utilized to promote deafness-related knowledge; free leaflets should be distributed in less-developed areas to raise public awareness of deafness screening.

Hearing screening, diagnosis and intervention measures

We suggest the following methods to improve hearing screening, diagnosis, and intervention measures. First, a standardized operating procedure and a uniform combination of diagnostic techniques and criteria should be developed. Furthermore, individual expert panel members should oversee the implementation of this system in particular areas of the province. Each expert could oversee the implementation and provide guidance and technical consulting regarding the implementation of the screening process in two to three prefecture-level cities. Finally, the appropriate government agencies should examine the causes of low screening rates, allocate funds to poor areas, and replace outdated equipment.

Improve genetic screening and diagnosis

We recommend universal screening for nine loci within four genes in hospitals. Children with a confirmed hearing impairment or a family history of deafness should be targeted for screening. Furthermore, four to five molecular diagnostics laboratories are needed to provide comprehensive molecular diagnostics throughout the province, including coverage of the eastern, western, southern, and northern regions of Guangdong as well as the Pearl Delta area. Standard molecular diagnostic procedures and guidelines should be developed for all such laboratories to follow. The goal is to establish within 3-5 years of effort a government-led, coordinated prevention and intervention system for newborn deafness with uniform screening and diagnostic procedures and unified diagnostic criteria. This healthcare model should be supported by a well-functioning network management platform, and related training should be provided by experts.

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Conflict of Interest

All authors claimed no conflict of interest.

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2. Primary ear and hearing care summary.


