ISRCTN89197487 https://doi.org/10.1186/ISRCTN89197487

Multidimensional analysis of screening results of deafness susceptibility genes in 3066 newborns of different altitudes and nationalities in Xining, Qinghai

Submission date
31/10/2025

Recruitment status
No longer recruiting

Registration date
14/11/2025

Overall study status
Completed

Last Edited
25/02/2026

Condition category
Ear, Nose and Throat

[_] Prospectively registered
[_] Protocol
[_] Statistical analysis plan
[X] Results
[_] Individual participant data

Plain English summary of protocol

Background and study aims
Xining City, Qinghai Province is located in a high-altitude hypoxic (low oxygen) area. Deafness is a common sensory disorder in newborns, and genetic factors are one of the main causes of congenital deafness. At present, there is a lack of large-scale and systematic research on the distribution of deafness-related genes in newborns in this specific high-altitude region. This study aims to fill this gap by using high-throughput sequencing technology to carry out large-scale deafness-related gene screening for newborns in Xining. The specific goals include: first, finding out the rate of newborns carrying deafness-related genes and the types of gene variations in the local area; second, providing targeted health education and clinical guidance for parents of children who may be at risk of deafness based on different gene characteristics, so as to achieve early diagnosis, early prevention and early intervention of deafness; third, providing scientific basic data for the research on prevention and treatment of deafness in Qinghai Province.

Who can participate?
The study recruits newborns of multiple ethnic groups living in different altitude areas of Qinghai Province. The specific inclusion criteria are:
1. Newborns born during the study period (March 2023 to March 2025)
2. Newborns whose families live in different altitude areas of Qinghai (including low-altitude, medium-altitude and high-altitude areas)
3. Newborns of various ethnic groups in Qinghai (such as Tibetan, Hui, Han, Salar, Tu, Mongolian, etc)
4. The guardians of the newborns have fully understood the purpose and process of the study and signed the informed consent form
5. The heel blood samples collected meet the quality requirements of the study (can be used for subsequent gene detection).

What does the study involve?
After the guardians of the newborns sign the informed consent form, the research team of the Affiliated Hospital of Qinghai University will collect a small amount of heel blood from the newborns. The collected heel blood will be made into dried blood spots for subsequent gene detection. Then, the research team will use high-throughput sequencing technology to detect common deafness-causing genes in these dried blood spot samples. After the detection is completed, the research team will analyze the detection data from multiple perspectives such as altitude, ethnicity and genotype. For newborns found to carry deafness susceptibility genes, the research team will provide targeted health education and clinical guidance to their parents, such as explaining the risk of deafness, suggesting follow-up observation and prevention measures.

What are the possible benefits and risks of participating?
Possible benefits:
1. For the participating newborns, if they carry deafness susceptibility genes, they can be found early through the study. Parents can obtain professional health education and clinical guidance in a timely manner, which helps to take preventive measures as early as possible and reduce the risk of deafness onset
2. The research results can provide a basis for the prevention and treatment of deafness in Qinghai Province and ultimately benefit more local families and newborns.
Possible risks:
1. The process of collecting heel blood may cause slight pain or temporary redness at the collection site for the newborn, but this discomfort will disappear quickly
2. There is a risk of personal information leakage in the research process, but the research team will strictly anonymize and encrypt all sample information and detection data to ensure the privacy of participants and will not disclose any personal information to third parties.

Where is the study run from?
Affiliated Hospital of Qinghai University (China)

When is the study starting and how long is it expected to run for?
March 2022 to December 2025

Who is funding the study?
Specific funding information of the study has not been provided yet and will be supplemented after confirmation. The study is funded by the Qinghai Provincial Department of Science and Technology (Project No.: 2023-SF-129) (China)

Who is the main contact?
Yi Wang, wegreatgroup@163.com

Contact information

Type(s)
Public, Scientific, Principal investigator

Contact name
Ms Yi Wang

Contact details
No. 29 Tongren Road
Chengxi District

Xining City
China
810001
+86 (0)13639716896
wegreatgroup@163.com

Additional identifiers

Study information

Scientific Title
Multidimensional analysis of screening results of deafness susceptibility genes in 3066 newborns of different altitudes and nationalities in Xining, Qinghai

Study objectives
1. Conduct hearing screening and deafness-susceptible gene screening for newborns born in Qinghai University Affiliated Hospital, identify newborns with deafness or at high risk of deafness, and provide further follow-up, guidance and intervention.
2. Obtain epidemiological data of newborns with deafness or deafness-susceptible gene carriers born in Qinghai University Affiliated Hospital.
3. Establish a multidisciplinary team (MDT) diagnosis and treatment model for newborn deafness in Qinghai.
4. Provide genetic counseling for deafness-susceptible gene carriers and their parents.

Ethics approval required
Ethics approval required

Ethics approval(s)
Approved 05/03/2022, Research Ethics Committee of Qinghai University Affiliated Hospital (REC) (No. 29 Tongren Road, Chengxi District, Xining, 810001, China; +86 (0)971 6162033; qdfydb001@163.com), ref: P-SL-202282

Primary study design
Observational

Study design
Observational epidemiological study

Study type(s)
Screening

Health condition(s) or problem(s) studied
Deafness

Interventions
From March 2023 to March 2025, the research team of the Affiliated Hospital of Qinghai University strictly abided by ethical norms. With the full informed consent of the participants, the team systematically and continuously collected heel blood samples from multi-ethnic newborns living in areas at different altitudes, and successfully obtained 3,615 complete blood samples required for the study. Subsequently, high-throughput sequencing technology was

applied to detect these samples, and accurate data on deafness susceptibility-related genes were obtained. During the analysis phase, the research team took altitude (low altitude, medium altitude, high altitude), ethnicity (Tibetan, Hui, Han, Salar, Tu, etc), and genotype (15 loci of 4 common deafness-causing genes: GJB2, SLC26A4, mitochondrial 12SrRNA, and GJB3) as the core dimensions. By comprehensively using statistical analysis and bioinformatics methods, a systematic multi-dimensional analysis was conducted to deeply explore the association characteristics between different factors and the carriage of deafness susceptibility genes.

Intervention Type
Genetic

Primary outcome(s)
Neonatal deafness susceptibility genes detected using high-throughput sequencing of neonatal heel blood samples collected at a single timepoint. The results are categorized as carrying deafness susceptibility genes (positive) or not carrying deafness susceptibility genes (negative).

Key secondary outcome(s)
There are no secondary outcome measures

Completion date
31/12/2025

Eligibility

Key inclusion criteria
Obtained written informed consent from the children's parents

Participant type(s)
Population

Healthy volunteers allowed
No

Age group
Neonate

Sex
All

Total final enrolment
3615

Key exclusion criteria
Among the 549 collected newborn heel blood dried blood spot specimens, some were unqualified and cannot be used for subsequent testing

Date of first enrolment
01/03/2025

Date of final enrolment

01/03/2025

Locations

Countries of recruitment
China

Study participating centre
National Genetic Testing Technology Application and Demonstration Center of the South Campus, Qinghai University Affiliated Hospital
No 29 Tongren Road
Chengxi District
Xining City
China
810012

Sponsor information

Organisation
Qinghai Provincial Department of Science and Technology

Funder(s)

Funder type
Government

Funder Name
Qinghai Provincial Department of Science and Technology

Results and Publications

Individual participant data (IPD) sharing plan
Relevant datasets generated and/or analyzed during the current study will be available upon reasonable request from Yi Wang (wegreatgroup@163.com).

IPD sharing plan summary
Available on request

Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?

Results article	24/02/2026	25/02/2026	Yes	No