8TH WORLD CONGRESS ON PUBLIC HEALTH AND GLOBAL WELLNESS

November 14-15, 2025 | London, UK

Chanjuan Hao, Ph.D.[1, 2, #, *] Xuyun Hu, Ph.D.[1, 2, #] Ruolan Guo, Ph.D.[1, 2] Zhan Qi, Ph.D.[1] Feng Jin, Ph.D.[3] Xiaofen Zhang, M.S.N.[3] Limin Xie, M.D.[3] Yuanhu Liu, Ph.D., M.D.[3, 4] Xin Ni, Ph.D., M.D.[4,*] **Wei Li, Ph.D., M.D.** [1, 2, *]

[1] Beijing Children's Hospital, Capital Medical University; Laboratory for Genetics of Birth Defects, Beijing Pediatric Research Institute; MOE Key Laboratory of Major Diseases in Children; Center of Genetics and Birth Defects Control, National Center for Children's Health; Beijing 100045, China

[2] Institute of Children's Health, Henan Academy of Medical Sciences, Zhengzhou 451162, Henan, China

[3] Department of Genetics and Reproductive Medicine, Shunyi Women's and Children's Hospital of Beijing Children's Hospital, Beijing 101300, China

[4] Department of Otolaryngology Head and Neck Surgery, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, Beijing 100045, China.

# These authors contributed equally to this work

*Correspondence to: Dr. Chanjuan Hao (hchjhchj@163.com), Wei Li (liwei@bch.com.cn) or Dr. Xin Ni (nixin@bch.com.cn). Beijing Children's Hospital, Capital Medical University, 56 Nan Li Shi Road, Xicheng District, Beijing 100045, China.

Comprehensive Genetic Screening of 465 Disease Genes in 33,894 Newborns in China and An Improved Strategy for Newborn Hearing Screening

Background

Expanded newborn genetic screening has the potential to identify a wide range of inherited conditions early in life. However, the prevalence and distribution of pathogenic variants in large-scale cohorts remain underexplored in the Chinese population. Hearing loss is a prevalent congenital condition. In China, concurrent newborn hearing and limited genetic screening has been implemented during the last decade. However, the role of gene sequencing in this context remains unexplored.

doi.org/10.51219/URForum.2025.Wei-Li

ISBN: 978-1-917892-32-2

Methods

We conducted genetic screening on 33,894 newborns using targeted sequencing of 465 genes associated with monogenic disorders. Variants were classified based on locally optimized American College of Medical Genetics and Genomics (ACMG) guidelines, focusing on pathogenic (P) and likely pathogenic (LP) variants. Key metrics including cumulative carrier rate (CCR), disease-specific prevalence, and regional differences were analyzed. In addition, comprehensive screening for 90 common hearing loss genes was performed on 7501 newborns. Universal newborn hearing screening and limited loci of genetic screening were also compared.

Conclusions

This large-scale study highlights the utility of targeted genetic screening for identifying carrier status and early-onset disease risks in newborns. The findings provide a critical foundation for integrating genetic screening into routine newborn care and for optimizing public health strategies. Combining targeted sequencing with universal newborn hearing screening is technically feasible and clinically useful in identifying newborns with hearing loss, particularly when integrated with genetic counseling and closed-loop management.

Biography

Wei Li, Ph.D., Professor of Beijing Children's Hospital, Capital Medical University. Director of Genetics and Birth Defects Control Center, National Center for Children's Health. Research Interests: Biogenesis of lysosome-related organelles and related diseases; Genetics and pathogenesis of birth defects and rare diseases. He has co-authored 150 SCI-indexed journals with about 10,000 citations. He initiated the first genomic newborn screening program in China in 2015. He has been awarded the Outstanding Achievement Award of China Birth Defects Salvage Foundation (2022).

doi.org/10.51219/URForum.2025.Wei-Li

ISBN: 978-1-917892-32-2