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·论　　著·

# 957例新生儿听力和聋病易感基因联合筛查结果分析

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【摘要】 **目的**　探讨新生儿听力和聋病易感基因联合筛查的临床意义。**方法**　选择2014年1~l2月出生后42 d进行听力复查的957例新生儿，听力复筛采用畸变产物耳声发射(DPOAE)结合自动判别听性脑干反应(AABR)。新生儿在出生后3 d内均已采集足跟血检测9个常见耳聋基因突变位点，包括GJB2基因(35 del G、176 del 16、235 del C、299 delAT)、GJB3基因(538 C>T)、SLC26A4基因(IVS7-2A>G、2 168 A>G)、线粒体DNA 12S rRNA基因(1 555 A>G、1 494 C>T)。**结果**　听力复筛通过904例，未通过53例，复筛通过率为94.46%。突变携带者50例，携带率为5.22%。听力复筛通过人群中检测出突变携带者45例，携带率为4.98%；听力复筛未通过人群中检测出突变携带者5例，携带率为9.43%。**结论**　新生儿听力和聋病易感基因联合筛查，可发现部分听力筛查不能发现的高危耳聋新生儿和迟发性耳聋新生儿，并可进行婚育及用药指导。

【关键词】 新生儿；耳聋；听力筛查；基因
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**Study of universal newborn hearing screening and deafness predisposing gene screening in 957 newborns.** *YUAN Jing-jing, ZHANG Fan, ZHANG Miao, BAO Shi-ping. Department of Otorhinolaryngology-Head and Neck Surgery, Beijing Youan Hospital, Capital Medical University, Beijing 100069, CHINA*

【**Abstract**】 **Objective**　To investigate the clinic significance of universal newborn screening with deafness predisposing genes in newborns. **Methods**　A total of 957 newborns underwent hearing screening at 42 d after birth in our hospital from January 2014 to December 2014. Distortion product otoacoustic emissions (DPOAE) combined with auto-auditory brainstem response (AABR) was used in hearing screening. These newborns had been taken blood sample at heel at 3 d after birth for detecting 9 common deafness genes mutation, including GJB2 (35 del G, 176 del 16, 235 del C, 299 delAT), GJB3 (538 C>T), SLC26A4 (IVS7-2A>G, 2 168 A>G) and mitochondria DNA 12S rRNA (1 555 A>G, 1 494 C>T). **Results**　In 957 newborns of hearing screening, 904 newborns showed "pass", while 53 showed "refer". The rate of showing "pass" was 94.46%. Mutation of deafness predisposing genes were detected in 50 cases, and the positive detection rate was 5.22%. Forty-five cases of mutations were detected in the newborns who passed hearing screening, with the positive detection rate of 4.98%. Five cases of mutations were detected in the newborns who did not pass hearing screening, with the positive detection rate of 9.43%. **Conclusion**　Combined screening of neonatal hearing and deafness predisposing genes can detect newborns with high risk of hearing loss and delayed deafness, which has an important guiding effect in marriage, childbearing and medication.

【**Key words**】 Newborn; Deafness; Hearing screening; Gene

听力是日常生活中至关重要的能力，听觉障碍严重影响生活质量。据2006年全国第二次残疾人抽样调查显示，我国听力及言语残疾人口约为2 780万，其中7岁以下的聋儿约有80万[1]。耳聋有遗传和环境因素等多种病因，据报道儿童期感音神经性聋的发病率为1‰，重度先天性聋中有一半以上是遗传病因[2-3]。我国开展普遍的新生儿听力筛查已经超过10年，取得了重大成效。但在新生儿听力筛查中存在一些局限，并非所

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