

# X连锁隐性遗传聋哑 (deaf-mute) 家系的遗传学特征分析 Genetic Analysis in a Chinese Deaf-Mute Family with X linked Recessive Inheritance

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## 摘要

在进行中国人群的遗传性耳聋发病情况的调查中, 发现了一个5代隔代遗传的聋哑家系 (L021家系)。研究中调查家系成员64人, 对其中的31人进行了系统的听力学检查, 发现聋哑男性8位, 听力表型为全聋及极重度聋, 获得家系成员的血样31人份。家系图谱分析显示该家系为X连锁隐性遗传性耳聋家系, 为先天性聋哑疾病分子病理机制的研究提供了模板。

Abstract: In studying genetic factors in hearing loss among Chinese hearing-impaired population, a Chinese family with deaf-mute that had been reversion inherited through five generations was found (named pedigree L021). X linked recessive inheritance was hypothesized to be the transmission in this family. A total of 64 members in this family were investigated. Of these, audiometric evaluation was performed on 31 members, including 8 males with deaf-mute. Most affected individuals showed deafness or profound sensorineural hearing loss. Blood samples were obtained from 31 consented individuals in this family. Pedigree analysis indicates a X-linked recessive inheritance pattern in pedigree L021. The pedigree described herein provides an excellent model for further study on the molecular mechanism of congenital deaf-mute.

关键词 [聋哑](#) [遗传性耳聋](#) [X连锁](#) [基因定位](#) [Key words](#) [Deaf-mute](#) [Hereditary Hearing Loss](#) [X linked](#) [Gene Mapping](#)

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## Key words

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