



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 X linked Recessive Inheritance

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），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 had been reversion inherited through five generations was found (named pedigree L021). X linked recessive inheritance was observed in the transmission in this family. A total of 64 members in this family were investigated. Of these, audiometric examination of 31 members, including 8 males with deaf-mute. Most affected individuals showed deafness or profound sensorineural hearing loss. Samples were obtained from 31 consented individuals in this family. Pedigree analysis indicates a X-linked recessive inheritance. The pedigree described herein provides an excellent model for further study on the molecular mechanism.

[存档文本](#)