Condition: Hereditary deafness

Inheritance:

Genetically heterogeneous; most common single gene nonsyndromic cause due to mutation of *GJB2* encoding connexin26.

Genetic etiology:

Genetically heterogeneous.

Frequency:

Prelingual deafness affects approximately 1/1,000 children.

Clinical features:

Deafness is divided into sensorineural and conductive types. Approximately 50% of prelingual deafness is due to genetic causes, and approximately 70% of this consists of isolated deafness, referred to as “nonsyndromic.” Approximately half of this is DFNB1 due to *GJB2* mutation. Other forms are associated with additional features, such as thyroid disease (Pendred syndrome) or blindness (Usher syndrome).

Management:

Supportive care; cochlear implants are successful in some children with prelingual deafness.

Genetic counseling:

Based on specific genetic cause; molecular genetic testing available for many of the causative genes.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.