GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness Explained

GJB2-related DFNB1 nonsyndromic hearing loss and deafness is an inherited condition that affects a part of the inner ear called the cochlea, preventing auditory information from being transmitted to the brain. This disorder causes mild to severe sensorineural hearing loss that is present from birth but not progressive. Management may consist of hearing aids or cochlear implantation for individuals with profound deafness.

How the Genetics Work

GJB2-related DFNB1 nonsyndromic hearing loss and deafness is an autosomal recessive disorder caused by pathogenic variants in the GJB2 gene. In general, individuals have two copies of the GJB2 gene. Carriers of GJB2-related DFNB1 nonsyndromic hearing loss and deafness have a single variant in one copy of the GJB2 gene while individuals with GJB2-related DFNB1 nonsyndromic hearing loss and deafness have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.