ABSTRACT

1 in 500 newborns are born deaf or with a hearing loss great enough to affect speech and language development. Most of this hearing loss is genetic and non-syndromic. High throughput genetic testing has revolutionized the diagnosis of congenital sensorineural hearing loss. There are over 80 identified genes and 1000 mutations associated with nonsyndromic genetic hearing loss, making directed genetic testing an expensive, protracted and low yield endeavor. Comprehensive genetic testing (CGT) has streamlined the workup of patients with both bilateral sensorineural hearing loss (SNHL) as well as auditory neuropathy spectrum disorder (ANSD). CGT spares patients unnecessary imaging studies and referrals and is now the single best test after an audiogram to order in the evaluation of hearing loss. Continued improvements in the process have brought costs to the level of directed genetic tests. A specific genetic diagnosis provides patients and families with answers as to causes and provides them with tools to plan for the future. As experience with CGT has evolved, specific genetic diagnoses also inform treatment decisions. For example, certain mutations are known to predict success of cochlear implantation. In the future, genetic testing will be the foundation for gene therapies to prevent or reverse hearing loss.