

Down Syndrome: A Multidisciplinary Perspective

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Abstract

Trisomy 21, a genetic disorder resulting from a chromosomal abnormality, is one of the most common forms of mental disability in the United States. Individuals with Down syndrome frequently present with a constellation of medical problems including conductive hearing loss and, to a lesser degree, sensorineural hearing loss. As part of a health care team, audiologists must be sensitive to and aware of medical conditions prior to establishing intervention strategies. Medical conditions, by necessity, precede audiologic interventions and, therefore, a close working relationship among team members is critical. Yet, audiologic and communication interventions should be established at the earliest possible time for maximizing an individual's development potential. This article stresses the importance of a multidisciplinary team in the provision of services so that prevention of further disabilities, improved outcomes of medical interventions, and appropriate rehabilitative and educational planning may ensue.

Key Words: Amplification, audiologic intervention, Down syndrome, hearing impairment

Trisomy 21, a genetic disorder resulting from a chromosomal abnormality, is one of the common forms of mental disability in the United States, with an incidence of approximately 1/700 live births. In 1866, Langdon Down prepared the first adequate description of this syndrome — hence, the name, Down syndrome. The correct diagnosis of Down syndrome can often be made at birth, fostering early identification and immediate rehabilitative planning.

Otolaryngologic and audiologic problems are common in Down syndrome and constitute potential additional disability. These topics are the focus of this paper and emphasize the importance of early and regular re-evaluation of the audiologic status of every child with Down syn-

drome. Skilled pediatric audiologic and otolaryngologic management in association with appropriate communication intervention can greatly enhance the social and cognitive function of children with Down syndrome.

GENETIC CONSIDERATIONS

The genetic imbalance leading to Down syndrome can be represented by either Full 21 trisomy (nondisjunction), Partial 21 trisomy (translocation), or mosaicism (postfertilization nondisjunction). When nondisjunction occurs during the first meiotic division of reproductive cells, both #21 chromosomes end up in one cell. Instead of both cells having 23 chromosomes, one cell has 24 chromosomes, and the other has 22. The loss of a #21 chromosome makes it highly unlikely for an egg containing 22 chromosomes to survive. However, the egg with 24 chromosomes can survive. Union with a cell containing 23 chromosomes results in a child with 47 chromosomes who has Down syndrome or trisomy 21.

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