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Nota di contenuto	Molecular Diagnosis of Deafness: Impact of Gene Identification -- Genetic Epidemiology of Deafness Genes and Clinical Applications -- Molecular auditory function -- Clinical Benefit of Molecular Diagnosis: Precision Medicine in Deafness -- Gap junction and channel transporter-related genes -- KCNQ4, GJB2, GJB6, KCNE1, KCNQ1, SLC26A4 -- Stereocilia-related genes CDH23, MYO7A, USH1C, PCDH15, USH1G, USH2A, ADGRV1, WHRN, CLRN1, MYO15A, MYO6, TMC1, STRC, ACTG1, DIAPH1, LOXHD1, PTPRQ, ESPN, MYO3A -- Neurotransmission-related genes OTOF, SLC17A8, PJVK -- Transcription factor and related genes EYA1, SIX1, PAX3, SOX10, MITF, SNAI2 CHD7, POU3F4, POU4F3, EYA4 -- Cell adhesion molecule and extracellular matrix-related genes COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, TECTA, OTOA, OTOG, OTOGL, COL4A3, COL4A4, COL4A5 -- Uncategorized important deafness genes WFS1, COCH, TMPRSS3, CRYM, NOG, mitochondrial -- Cochlear Implantation from the perspective of the responsible gene -- Gene therapy and mechanism-based drugs: Toward more precise treatment based on

molecular diagnosis.

Sommario/riassunto

This book sheds new light on the molecular diagnosis of deafness, emphasizing the impact of gene identification and its clinical application. Through this comprehensive volume, readers will gain insights into the genetic underpinnings of hearing loss and the latest advancements in genetic testing and counseling. The chapters cover topics such as the genetic epidemiology of deafness genes, non-syndromic and syndromic hearing loss, and the role of genetic testing in cochlear implantation. The author delves into specific genes like GJB2, SLC26A4, and MYO7A, providing a detailed analysis of their clinical implications. This book also explores future directions in genetic screening, making it an indispensable resource for understanding the complexities of genetic hearing loss. Molecular Diagnosis of Deafness is essential for ENT clinicians, geneticists, and genetic counselors seeking to enhance their knowledge of deafness genes and their clinical applications. It offers a practical guide to navigating the challenges of genetic heterogeneity in hearing loss and provides valuable insights for improving patient care through advanced genetic testing and counseling.