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Nota di contenuto	Preface -- 1. Epidemiology of Ectodermal Dysplasias -- 2. What is and isn’t an ectodermal dysplasia? -- 3. X-linked hypohidrotic ectodermal dysplasia: the pathogenic role of the EDA gene -- 4. X Linked-Hypohidrotic Ectodermal Dysplasia - Clinical Features -- 5. Incontinentia pigmenti -- 6. Genetic basis and molecular diagnosis of p63-associated ectodermal dysplasia -- 7. Focal Dermal Hypoplasia (FDH) and WNT10A-related ED -- 8. Nectinopathies: ectodermal dysplasia syndromes caused by mutations in PVRL1/4 genes encoding cell adhesion molecules nectins-1/4 -- 9. Other syndromes with prominent ectodermal dysplasia signs -- 10. Ectodermal Dysplasias: Orofacial Manifestations and Management -- 11. Oral and dental

phenotype in ectodermal dysplasias -- 12. Ocular manifestations in hypohidrotic ectodermal dysplasia -- 13. Othorynolaryngologic manifestations in hypohidrotic ectodermal dysplasia -- 14. Broad phenotypic spectrum of Ectodermal Dysplasias in childhood and adolescence -- 15. ED Practical Management -- 16. Molecular therapies -- 17. Patient Advocacy Organizations: One Story of Success.

Sommario/riassunto

Ectodermal Dysplasias (ED) are a diverse group of genetic disorders characterized by congenital defects of two or more ectodermal structures (e.g., sweat gland, tooth, nail, hair), that count about 100 different manifestations. Therefore, the multidisciplinary aspect is a novel but crucial approach to correctly diagnose and treat this kind of diseases and eventually direct patients to specialized centers. A new classification has been proposed as well as new therapeutic options, such as the first prenatal protein replacement therapy for a genetic disease, X-linked hypohidrotic ectodermal dysplasia. The chapters of this book address all relevant topics, starting with epidemiology and embryology, disease classification, molecular biology, EDA1-associated ED, WNT10A-related ED, and p63-related ED. A chapter on ED caused by defects of structural proteins is included and one specifically devoted to differential diagnoses. Specific chapters describe diagnostic assessments and treatment: odontostomatological signs and therapy, ophthalmological or otorhinolaringoiatric signs, other organ involvement, neurological and neuropsychological issues. The final part is dedicated to the most recent developments in molecular therapy and the extremely important role of parents' and patients' associations. Written by internationally renowned experts, this handy resource will be of valuable help for a variety of specialists who deal with ectodermal dysplasias in their daily clinical work, e.g., pediatricians, dermatologists, ENT-specialists, dentists.