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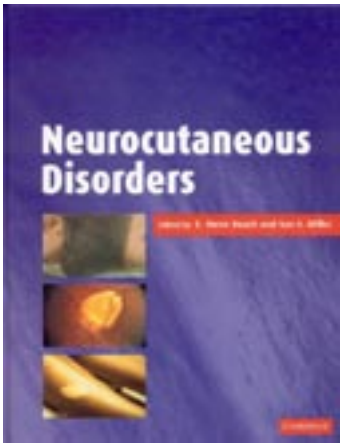

We have a pleasure to thumb through the new edition of the book Neurocutaneous Disorders, a fruit of several-year collaboration of a number of predominantly American as well as Canadian, Italian, Dutch and Brazilian authors. Neurologists, geneticists, ophthalmologists and dermatologists, along with experts from other professions, especially those dealing with disease diagnosis, were engaged in the writing of this book. The book has 42 chapters, each of them accompanied by 30 to 100 references.

In the introduction, the editors stress that congenital or hereditary conditions with lesions of both the skin and the nervous system, both of which are derived from the ectoderm, have been traditionally considered together as neurocutaneous disorders. The earlier term phakomatosis derived from the Greek word phakos meaning “lentil”, “lens” or “mother spot” was devised to describe tuberous sclerosis and Recklinghausen fibromatosis with characteristic cutaneous lesions and the potential for tumor formation. Although the term phakomatosis was widely used for several years, the gradual inclusion of conditions with vascular and other skin lesions and disorders without an increased tumor risk has made the broader concept of neurocutaneous syndrome more appropriate. In the second chapter of the book with data on the genetics of neurocutaneous disorders, the authors stress that there are more than 40 such diseases described in the text, with the majority of them displayng mendelian inheritance. There are ample data on the types of mutations in neurocutaneous disorders, penetration, expressivity, mosaicism, and genetic heterogeneity as well as on X-linkage in this group of diseases. A separate chapter is dedicated to clinical recognition of neurocutaneous disorders and approach to these patients, especially children with genetic diseases in general. There is information on the possibilities of genetic testing, cytogenetics, biochemical and metabolic testing, and gene (DNA) testing. When large numbers of affected individuals are available in a family, as may be in case of NF-1 or NF-2, then a genetic linkage study can highlight particular chromosome regions where the causative gene locus must reside. Recombinant cloning and PCR amplification of genes within this region can proceed, followed by screening for mutations resposible for the disease. Inspection of genes within the chromosome region highlighted by mapping studies may reveal “candidate genes” with tell-tale DNA sequences (motifs) that could be related to the disease.

Special chapters dealing with type I and II neurofibromatosis are enriched with high quality clinical photographs as well as photographs of radiologic and other findings.

The chapter on tuberous sclerosis gives a very comprehensive account of the cutaneous, renal, cardiologic, pulmonary and neurologic symptoms. It has recently been discovered that two gene mutations are involved in tuberous sclerosis, i.e. TSC2 on chromosome 16p13.3 and TSC1 on 9q34. Robert J. Gorlin from Department of Oral Pathology, School of Dentistry, University of Minnesota, has written the chapter on nevoid basal cell carcinoma (Gorlin’s syndrome). Although provided with black-and-white photographs, these are high quality and very illustrative, and accompanied by detailed description of particular symptoms.

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Epidermal nevus syndrome is described in detail. In 30 years since Solomon *et al*. proposed the name “epidermal nevus syndrome”, a second major definitional issue has arisen. Advances in genetics have led to an understanding that the cutaneous manifestations of epidermal nevus syndrome are due to genetic mosaicism. Moreover, several different genetic abnormalities can likely lead to similar phenotypes. The appreciation of genetic heterogeneity in epidermal nevi makes it easier to comprehend the wide variety of both cutaneous and extracutaneous manifestations. It is clear that the condition is not one disease but rather a heterogeneous group defined by a common cutaneous phenotype: the presence of epidermal and adnexal hamartomas that are associated with other organ system involvement.

Separate chapters bring detailed description of multiple endocrine dysplasia type 2, ataxia-telangiectasia, incontinentia pigmenti, pseudoxanthoma elasticum, and hereditary hemorrhagic telangiectasia (Osler-Weber-Rendu syndrome).

The chapter on Ehlers-Danlos syndrome points out that at least ten types of the disease are currently defined on the basis of clinical features, pattern of inheritance, and specific collagen defects. Special attention is paid to vascular manifestations, primarily the potentially life-threatening aneurysms, in Ehlers-Danlos syndrome. A separate chapter is dedicated to hemangiomas as a complex of vascular anomalies. An array of therapeutic options are described, including corticosteroids, interferon alpha-2a or -2b, embolization, and laser therapy (Nd-YAG). Recently, thalidomide has been tried in some forms of hemangioma, because of its anti-angiogenic action. Then, most relevant data on Sturge-Weber syndrome and on a group of neurocutaneous disorders associated with metabolic defects are presented. Lesch-Nyhan syndrome characterized by absence of the hypoxanthine guanine phosphoribosyltransferase enzyme activity is described. Nonepithelializing lip erosions are an important dermatologic symptom.

Some rare neurocutaneous disorders are described in detail, e.g., Menkes disease, fucosidosis, multiple carboxylase deficiency, and homocystinuria due to deficiency of cystathionine β-synthase activity.

Chapter 29 is dedicated to the group of syndromes related with nucleotide excisional repair: xeroderma pigmentosum, Cockayne’s syndrome, and trichothiodystrophy.

The book Neurocutaneous Disorders is an important reading for all those dealing with the issues, including dermatologists, neurologists, pediatricians, geneticists, ophthalmologists, and other specialists. This valuable book should find its place in the library of every clinical department and of all individual professionals engaged in these complex and highly important medical issues. This edition will certainly be followed by new ones because ever newer concepts emerge continuously in this interdisciplinary field of medicine.

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