NEUROACANTHO CYTOSIS SYNDROMES
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Why have a book on the neuroacanthocytosis syndromes? Most physicians likely think that they would never see a patient with any of these rare disorders. But this is, of course, the first reason for having and reading such a book. If a physician does not know about a disorder, then even if a patient with it does appear, the diagnosis will be missed. And, why should the book be called Neuroacanthocytosis Syndromes? Isn’t there only one disorder of neuroacanthocytosis? As the reader of this book will find out, if they don’t know already, there are a number of neurologic disorders with acanthocytes, and neuroacanthocytosis proper has at least two distinct forms with separate defined genetic basis (chorea-acanthocytosis and the McLeod syndrome).

Neuroacanthocytosis was first described in the United States. Estes, Morley, Levine and Emerson reported a family from New England in 1967, and more details were given in an article by Levine, Estes and Looney in 1968. Critchley, Clark and Wikler presented a family from Kentucky at the American Neurological Association in 1967 and published their full paper in 1968. Now the disorder has been found all over the world.

Neuroacanthocytosis (proper) clinically can be characterized by chorea particularly in the orofacial region, tics including vocalizations and lip and tongue biting, dysarthria, dysphagia, seizures, peripheral neuropathy, myopathy with raised plasma CK and behavioral and cognitive dysfunction. If the whole syndrome is present, it should be relatively easy to diagnose, but when only partial it can masquerade as many other conditions.

In this book, the neurology of the neuroacanthocytosis syndromes is expanded in detail with a good overview of the differential diagnosis. Readers of the book will be able to find out about the pathophysiology of the clinical manifestations, cell biology of acanthocytes, the genes that cause the disorder and their biology, possible animal models, relationship to Huntington’s disease, and approaches to therapy including surgery and gene therapy.

Many thanks to Dr. A. Danek for putting this all together and the team of experts that he has gotten to contribute to this useful work.
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