Inherited Disorders of Biogenic Amines: A Practical Guide

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in collaboration with
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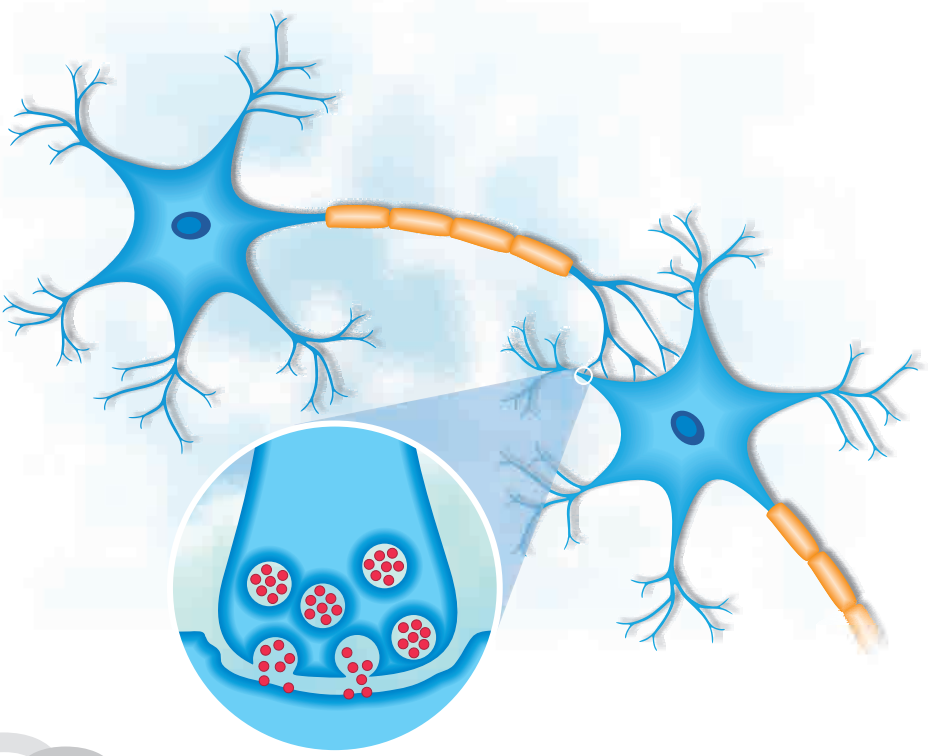

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UNI-MED Verlag AG, one of the leading medical publishing companies in Germany, presents its highly successful series of scientific textbooks, covering all medical subjects. The authors are specialists in their fields and present the topics precisely, comprehensively, and with the facility of quick reference in mind. The books will be most useful for all doctors who wish to keep up to date with the latest developments in medicine.

Preface and acknowledgements

The first cases of children with a form of phenylketonuria that was accompanied by a complex severe progressive neurologic illness unresponsive to dietary treatment were reported in the 1970s. It was already hypothesized that these children lacked tetrahydrobiopterin, the cofactor required not only for the enzyme, phenylalanine hydroxylase, but also for tyrosine hydroxylase and tryptophan hydroxylase, the rate-limiting enzymes in the biosynthetic pathways for biogenic amines, catecholamine neurotransmitters, and serotonin. Thereafter, a plethora of primary defects in the metabolism of biogenic amines, leading to neurotransmitter imbalances, were discovered.

Increased understanding of these concepts led to the development of specific pharmacologic treatments for some of these diseases, although this was very limited at first. In general, the disease would take its course and genetic counseling was virtually all that could be offered. In recent years, new genetic and metabolic techniques have underpinned amazing progress in diagnostic approaches and, crucially, a much better understanding of the underlying pathophysiology. New and successful therapeutic approaches have followed, including intracerebral gene therapy.

The "Working Group on Neurotransmitter-related Disorders", together with lay support groups, moved this field forward so quickly that we saw a need for a concise, specific, but comprehensive and up-to date reference resource on inherited disorders of biogenic amines. In this book, we have sought to combine brief, but solid, information on the biochemical and molecular background of these disorders, together with a description of their clinical presentation, treatment, monitoring and disease course. We hope you find our book interesting, informative and useful.

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