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Inherited Disorders of Biogenic Amines: A Practical Guide

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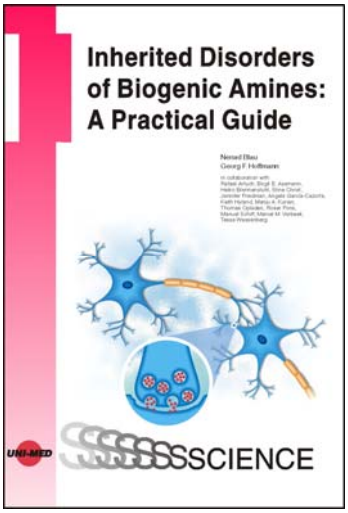

Disorders of biogenic amines have been recognized as important causes of severe, progressive encephalopathies mostly of early onset. The past decade has seen immense progress, unravelling of new diseases, as well as major advantages in diagnostics and patient care, with gene therapy just begun.
The diagnosis of these disorders is almost exclusively based on clinical signs and symptoms leading to measurement of metabolites in CSF. Whilst the majority of the identified disorders are due to inherited enzyme deficiencies, defects in transport of active compounds have also been reported. There is still widespread uncertainty about when to perform specialized CSF investigations and what to investigate. Next generation sequencing is now starting to complement CSF analysis and will take a greater role in the primary diagnostics in the future.
The main focus of this book is the clinical and biochemical approach to these disorders. We wanted to provide key information and recommendations on therapy, monitoring and follow-up and hope for quicker and improved therapy and by that outcome of affected individuals.

Klinische Lehrbuchreihe
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