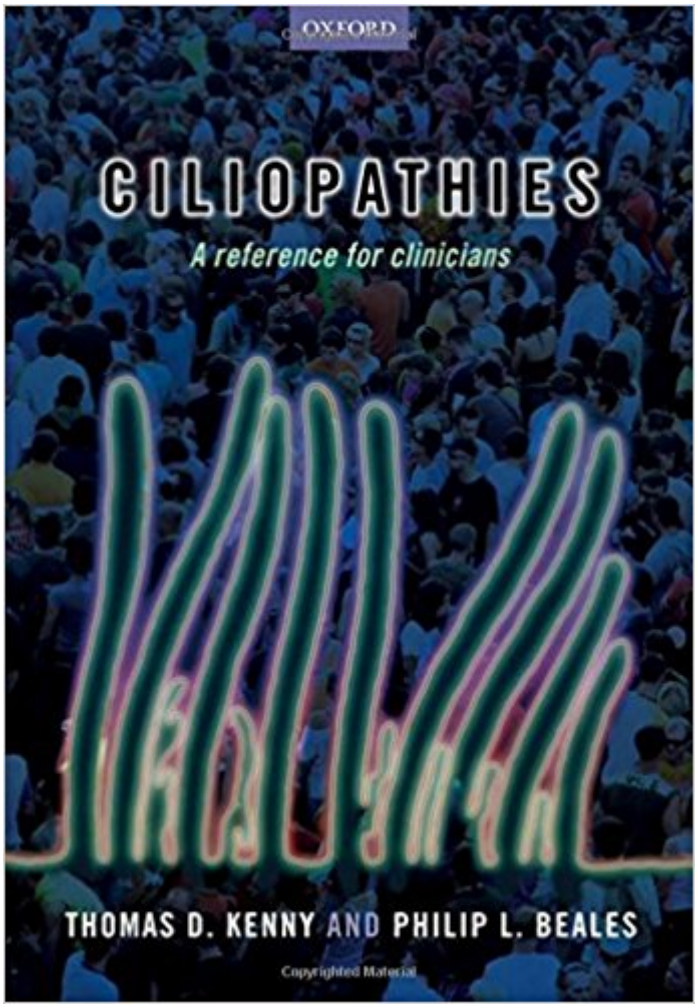
Ciliopathies: A Reference For Clinicians



Synopsis

The ciliopathies are a group of rare diseases that often affect multiple systems within the body, and are caused by defects in the function or structure of cilia. When cilia go wrong, there are profound consequences; these are discussed in detail for the first time in Ciliopathies: a reference for clinicians.The book provides a clinical overview and reference to this newly emergent group of disorders ranging from Alstrom syndrome to putative ciliopathic disorders. Each chapter provides an in-depth discussion on a specific disorder, including the latest scientific research together with a description of its features, and practical guidelines on diagnosis. The authors also examine the evidence for dysfunction of cilia in cancer and more common disorders.Ciliopathies: a reference for clinicians will appeal to those involved in the care of patients with ciliopathies, including specialists in the fields of nephrology, diabetes, cardiology, and ophthalmology, and non-clinical researchers interested in cilia biology.

Book Information

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Customer Reviews

"This is a valuable book on a lesser-known group of disorders in the medical and research communities known as ciliopathies. The editors Kenny and Beale have led a pioneering effort with this book in a field that still requires much "digging" to understand the causes and effects. We also commend the contributors to this book named below for their initiatives in their particular specialties." --Biz India

Thomas D. Kenny, Medical Adviser, NHS Specialised Services, UK,Philip L. Beales, Professor of Medical and Molecular Genetics, UCL Institute of Child Health, London, UKPhilip Beales is Professor of Medical and Molecular Genetics at UCL and Wellcome Trust Senior Research Fellow in Clinical Science. He is Director of the Centre for Translational Genomics (GOSgene) and Head of the Cilia Disorders Laboratory at the UCL Institute of Child Health. He is best known for his clinical and genetic research into rare diseases especially, the ciliopathies, culminating in novel gene discoveries for Bardet-Biedl syndrome, Jeune Asphyxiating Thoracic Dystrophy, Cranioectodermal dysplasia, Acrocallosal Syndrome and several other disorders. He, with colleagues, was the first to attribute the Bardet-Biedl syndrome phenotype to dysfunctional primary cilia. Philip is a consultant in clinical genetics at Great Ormond Street Hospital for Children and Guys Hospital; National lead for the Department of Health specialist commissioned Bardet-Biedl syndrome clinical and diagnostic service; chairman of the UCL Rare Diseases Steering Committee and co-editor in Chief of CILIA.