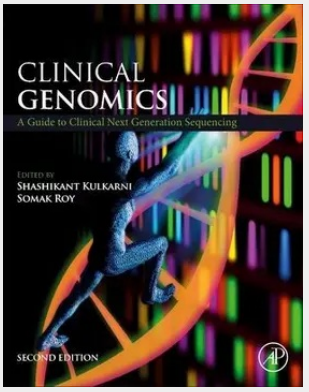
Kulkarni / Roy

Clinical Genomics

A Guide to Clinical Next Generation Sequencing



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Clinical Genomics, Second Edition is a comprehensive, practical reference [AN1] designed specifically for laboratory directors, clinical medical directors, and genomic medicine professionals. Edited by renowned genomic pathology leaders Kulkarni and Roy, this updated edition offers clear guidance on the current technical, bioinformatic, clinical, regulatory, and reimbursement considerations involved in implementing next-generation sequencing technologies within clinical molecular diagnostic laboratories. Structured for ease of reference, this book covers targeted gene sequencing methods, whole exome/genome approaches, emerging technologies such as long-read sequencing, and critical considerations for assay validation. Detailed sections address bioinformatics workflows, variant detection and interpretation methodologies, integration with clinical informatics systems, and essential IT infrastructure, including cloud-based solutions. This book also thoroughly addresses practical topics essential to laboratory management, such as regulatory compliance, ethical frameworks, billing strategies, and reimbursement paradigms, helping directors navigate the evolving landscape of genomic diagnostics.

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