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Integrating Genomic Medicine into the Diagnosis of Rare Genetic Disorders

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The diagnosis of rare genetic disorders has been significantly enhanced through the integration of genomic medicine into routine clinical practice. Advanced sequencing technologies, combined with improved bioinformatics tools, have increased diagnostic accuracy while reducing the time required to identify disease-causing variants. This presentation discusses the clinical implementation of genomic testing for inherited metabolic and mitochondrial disorders, emphasizing multidisciplinary collaboration between clinicians, geneticists, and laboratory scientists. The presentation also highlights the role of newborn screening, precision diagnostics, and personalized treatment planning in improving patient outcomes. Current challenges, including interpretation of variants of uncertain significance, healthcare accessibility, and ethical considerations surrounding genomic data, are explored. Future directions focus on artificial intelligence-assisted genomic analysis, international collaboration, and precision therapeutics designed to improve the management of rare diseases globally.

Biography

John Christodoulou is an internationally renowned clinical geneticist whose research focuses on inherited metabolic disorders, mitochondrial diseases, and pediatric genomic medicine. He has made significant contributions to integrating genomic technologies into clinical diagnostics and has led numerous multidisciplinary research initiatives in Australia and internationally. His work has advanced precision medicine and improved diagnostic pathways for patients living with rare genetic disorders.