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Genomic Technologies for Rare Disease Diagnosis and Personalized Therapeutics

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Genomic medicine has become an essential component of rare disease diagnosis and treatment by enabling the identification of pathogenic variants responsible for inherited disorders. This presentation reviews recent advances in next-generation sequencing, transcriptomics, and multi-omics technologies that support earlier diagnosis and personalized therapeutic interventions. Clinical applications of precision medicine, newborn genomic screening, and biomarker-guided treatment strategies are highlighted through representative case studies. The presentation also discusses the growing role of artificial intelligence in genomic interpretation, international data-sharing initiatives, and collaborative research networks that accelerate therapeutic discovery. Future innovations in genome editing and personalized therapeutics are expected to improve diagnostic accuracy and expand treatment options for patients with rare genetic diseases.

Biography

Jong-Won Kim is an internationally recognized medical geneticist specializing in genomic medicine, molecular diagnostics, and rare inherited disorders. His research integrates advanced genomic technologies into clinical practice to improve diagnosis and personalized treatment for patients with rare diseases. He has authored numerous scientific publications and actively collaborates with international research networks.