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AI in Medicine

Future of Healthcare by AI



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Speech Title	Whole Genome Sequencing for Rare Disease Diagnosis	
Abstract(200 words) :		
<p>Rare diseases are collectively common, affecting approximately 3% of the global population, yet their clinical heterogeneity and overlapping phenotypes often complicate timely and accurate diagnosis. Conventional genetic testing methods, such as targeted gene panels and whole-exome sequencing (WES), have significantly improved diagnostic yield but remain limited. These approaches primarily focus on coding regions and may miss pathogenic variants in regulatory regions, structural variants, repeat expansions, and mitochondrial genome alterations.</p> <p>Whole-genome sequencing (WGS) offers a comprehensive, unbiased assessment of the entire genome and has demonstrated superior performance in diagnosing genetically unresolved rare diseases. WGS enhances variant detection across diverse classes, including noncoding, structural, and complex rearrangements, thereby increasing diagnostic yield, particularly in patients with atypical presentations or negative WES results. Integration of WGS with advanced bioinformatics, transcriptomic and epigenomic data, and phenotype-driven interpretation further refines diagnostic accuracy and facilitates discovery of novel disease genes.</p> <p>Future trends include the implementation of WGS as a first-tier diagnostic test, incorporation of long-read sequencing for resolving repetitive or structurally complex regions, and the application of artificial intelligence to improve variant prioritization. As sequencing costs decline and data-sharing frameworks evolve, WGS is expected to become an essential tool for precision medicine, enabling earlier diagnoses, optimized management strategies, and novel therapeutic opportunities for patients with rare diseases.</p>		