임상유전체 진단의 최신 지견

기 창 석
GC녹십자지놈

Recent update on clinical NGS

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The advancement of clinical genetics and genomics is accompanied by innovations in molecular technologies. Two molecular technologies that have had the greatest impact on the clinical genetics and genomics are the Sanger sequencing technology invented in the mid-1970s and polymerase chain reaction (PCR) technology developed in the mid-1980s. Based on these two technologies, the field of molecular microbiology and molecular diagnostics of genetic conditions has been able to make remarkable progress for about 20 years since 1990s and now clinical genomics field becomes the most rapidly evolving specialty in medicine. Sanger sequencing has been monopolistic position for almost two decades since it was invented by Frederick Sanger in 1977. However, novel sequencing technologies called Next-Generation Sequencing (NGS) finally appeared in early 2000s and they are becoming a power to change the foundation of medical field in conjunction with the 4th Industrial revolution.

Recent advances in NGS technologies have changed clinical genetics and genomics from gene-by-gene approach to syndrome-based gene panel sequencing or whole exome/genome sequencing (WES/WGS). In addition, the novel sequencing technologies have boosted the discovery pace of novel disease genes. Before the WES/WGS is available, a priori information on the causative genes that might underlie a genetic condition is a prerequisite for molecular diagnostics but, theoretically, WES/WGS does not require any information on candidate genes. Although disease-targeted gene panel testing or WES/WGS is still expensive, it may be a cost-effective approach for molecular diagnosis of some genetic disorders with extensive genetic heterogeneity such as inherited neuropathies, muscular disorders, dystonias, hereditary spastic paraplegias, neurodegenerative disorders, etc. In addition, WES/WGS may find unexpected pathogenic variants in genes known to cause different conditions from the initial diagnosis.

In this talk, I'll present recent updates on clinical NGS in terms of novel NGS platforms as well as RNA-seq and sequence interpretation strategies.

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