KSN SEOUL, KOREA 2023
April 27(Thu) - 30(Sun), 2023
Coex, Seoul, Korea

Submission No.: GD01-9112
Session Title: Genetic Disease
Date & Time, Place: April 27 (Thu), 13:00 - 15:00, Room 5

Diagnostic Approach for Genetic Disease by Translating Whole Genome Big Data

SOYOUNG LEE
Hallym University Sacred Heart Hospital, Korea, Republic of

Molecular diagnoses, or genetic tests, are necessary to confirm genetic diseases, and provide appropriate management and genetic counselling in clinical filed. Historically, clinical genetics has relied on single gene molecular tests and cytogenetic tests. Next-generation sequencing (NGS) is a massively parallel sequencing technology, which enables determine the order of nucleotides in entire genomes of DNA or RNA. Therefore, clinicians prefer to perform simultaneous sequencing of multiple genes or whole exomes and genomes. Although panel sequencing or whole exome sequencing (WES) based on NGS are still used for genetic diagnosis, demands for whole genome sequencing (WGS) is increasing, especially in patients with rare or undiagnosed diseases.
WGS is known a comprehensive method for analyzing entire genomes, and diagnostic yield is higher than other genetic tests. In principle, WGS allows the detection of disease relevant genomic variant. First, WGS is able to determine single nucletide variants and small indel in non-coding regions including regulatory regions or repeated sequences as well as coding regions. Second, WGS is more useful to detect structural variants, and could show relatively accurate breakpoints in deep intronic or intergenic regions. Third, complex genomic rearrangements and transposable elements are detected, which were difficult to be found by previous genetic studies. Finally, WGS enables large size copy number variations without other cytogenic tests such as chromosomal study or microarray. However, genetic diagnosis using WGS is still challenging. WGS is still expensive although getting cheaper. In addition, our understandings for human genomics are immatue even though rapid evolution. Diagnostic approach using WGS generate large amounts of data, but standardized NGS technology and tools for analyzing and interpreting WGS big data are neither fully developed nor established yet. Moreover, it requires accurate clinical information and close communication between clinicians and bioinformaticians or geneticists to interpret selected variants including secondary findings.
WGS is a powerful method to discover genetic causes of diseases, hypothesizes disease mechanisms, suggests appropriate management, provides genetic counseling, and develop new drugs or treatment. To this end, it is thought that 4C (Clinical information, Curation, Cumulation, Communication) is important to analyze WGS big data in clinical field.