Smart Genomic Adventures Lead to Precision Medicine

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At the post “next generation sequencing (NGS)” era, a smart hospital must bring genomic medicine to bedside. The power of NGS allows us to dissect an individual genome in disease, cancer and aging conditions to advance our understanding of physiological and pathological mechanisms, and to improve disease prevention, diagnosis, prognosis, and treatment. The journey from DNA/RNA sequences to medical practice is not an easy course. Experts from all areas including molecular biologists, chemists/biochemists, geneticists, software/hardware engineers, bioinformaticians, clinicians, and genetic counselors, need to contribute to the success of the journey.

Clinical applications of NGS in molecular diagnosis are in two major areas; genetic diseases and cancer. Severe genetic diseases usually occur at neonatal period, which requires immediate definitive diagnosis for proper management, including specific panel sequencing, critical trios, critical whole exomes or whole genome sequencing (WES or WGS). While the NGS based diagnosis of cancer may include finding the familial cancer risk genes for prevention, somatic mutations for specific treatment, detection of marker gene/mutations for prognosis and treatment, and liquid biopsy for routine monitoring of cancer progression.

Since the application of NGS to clinical molecular diagnoses a few years ago, many previously unsolved cases have been resolved by the identification of causative genes, which greatly expand the phenotype and genotype of various diseases. Functional investigations in cellular and animal models are required for novel genes to establish their disease mechanisms. In addition, the interpretation of variants of uncertain significance (VUS) is a challenge. As we gradually improve our understanding of disease mechanisms and build up variant databases, we can better correlate laboratory findings to patient’s clinical course. Not only is there continuously growing scientific knowledgebase, but also the ever evolving patients’ clinical phenotypes. Thus, there is a need for constant interactive communications among diagnostic laboratories, clinicians and patients. Development of real time web-based interactive communication tools may suit this purpose.

In conclusion, smart utilization of genomic medicine information in innovative ways can lead to valuable application in precision medicine.