Breakthrough in Medical Genetics

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Next Generation Sequencing (NGS) or massive parallel sequencing (MPS) changes the pace of DNA and RNA sequencing abling to obtain sequencing at the genomics and transcriptomics level. It becomes a cost-effective tool facilitating molecular genetics diagnosis of once really time consuming and expensive. Cancer predisposing gene testing with next-generation multigene panel sequencing is proved to be benefit beyond traditional step testing. The burden is the counseling of the risk and appropriate screening after test revealed a cancer predisposing gene mutation in the families. It also makes research and clinical practice for ‘Precision Medicine’ becomes reality. Requirement of bioinformatician personals and tools to handle the big data and lack of long term clinical information of this somewhat recent technology are the major hurdles but will be soon evolved with global effort leads by ‘Precision Medicine Initiatives’ by the late US president, Mr. Barack.

Gene therapies and genome editing methods comes into the spotlight in the last two years. Many major clinical trials or reports successfully treat severe inherited diseases with gene therapy bring pivotal changes to the gene therapist community after a long stagnation in the field for decades after a tragedy story of a case died from the immune reaction against viral vector used. CRISPR-CAS9 genome editing system makes genetic correction more efficient and precise. It has been used in postnatal gene or immune therapy. The idea of genome editing for human embryo shakes the scientific community after Chinese scientist team revealed their research using human left-over embryos to correct the gene with this technique.

Since Linus Pauling, a double Nobel Laureate published ‘sickle cell anemia, a molecular disease’ in 1949, ‘clinical medicine’ has becomes true ‘molecular medicine’ with the advance in genetic rereading (gene sequencing) and rewriting (gene editing) technologies.