Chapter 1

APPLICATION OF NEXT-GENERATION SEQUENCING AND BIOINFORMATICS FOR PRECISION MEDICINE IN CANCER

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INTRODUCTION

Next generation sequencing (NGS) becomes the most preferable molecular diagnosis tool in the most recent studies due to its reliability and high sensitivity. NGS offers a possibility to sequence multiple gene as well as multiple samples simultaneously and cost effectively unlike traditional sequencing techniques such as Maxam-Gilbert`s chemical degradation and Sanger-Coulson`s chain terminating(Maxam and Gilbert, 1977& Sanger and Coulson, 1975). When it is used in terms precision medicine all these advantages come together with more information not on only about diagnosis but also prognosis and the treatment for the patients' (Hinrichs, et al., 2015). NGS also time efficient in comparison to the traditional sequencing methods even though traditional sequencing methods and NGS have similar hands on time, NGS offers much more sequenced data from multiple samples and genes in single a run. There have been many commercially available systems that can sequence up to 12 whole human genomes in a single run. However, the whole NGS workflow starting from the wet-laboratory applications to bioinformatics analysis still in the need of NGS applications very detailed optimization, standardization and clinical validation that relies on the user's qualification and experience (Choudhary, et al., 2014& Smith, et al., 2014). However, it is inevitable to use this pioneer technology for diagnosis and treatment in genetic diseases.

"Precision medicine" is disruptive concept in health care system especially in oncology that takes into account both population characteristics and individual

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To sum up, the main aim of NGS studies are not only limited to genetic diseases diagnosis but also widened up for genomic profiling of the tumors in the latest medical applications. All these technologies with an integrative manner in many disciplines and many oncological diseases make it possible to use the data as a part of good clinical practice and precision medicine to measurable outcomes. After all, the translation of somatic mutation screening in cancer within a clinical context on the basis of a specific algorithm for better diagnosis and treatment options should be considered.

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