

# From Targeted Testing To Targeted Analysis: The Era of Comprehensive Clinical Genomics Analysis Is Coming

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Received date: Jan 24, 2018; Accepted date: Jan 27, 2018; Published date: Jan 31, 2018

Citation: Hongxing L, Yu Z (2018) From Targeted Testing To Targeted Analysis: The Era of Comprehensive Clinical Genomics Analysis Is Coming. Genet Mol Biol Res Vol No: 1 Iss No: 1: PDF No: 3

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## Editorial

With the widely application of next generation sequencing (NGS) technology in clinic and the progress of molecular mechanisms research in tumour, more and more molecular markers have been identified and used in clinical diagnosis and treatment decisions [1-4].

Detection of dozens or hundreds of genes by targeted sequencing has been widely used in the diagnosis and treatment of various oncological and genetic diseases, and reports of genomics analysis based on whole exome sequencing (WES), whole genome sequencing (WGS), and transcriptome sequencing (RNA-seq) is also significantly increased.

### Advantages and challenges of genomics analysis

With the significant decrease of cost and the improvement of analytical capabilities, the application of WES, WGS and RNA-seq increased gradually which provides more comprehensive genetic information.

Compared with traditional medical testings, genomics analysis brings new concepts and challenges. Traditional medical testings (biochemistry, immunology and targeted sequencing, etc.) basically belong to targeted testing that require the designation of a limited number of targets and corresponding tests which are expected to be relevant to the disease or phenotype. Genetic testing is the first to proceed with a methodologically comprehensive omics-based analysis that covers the real whole of genome, exome and transcriptome rather than specific targets. However, the huge amount of data generated by the omics sequencing (about 12G for 100 × WES data and about 90G for 30 × WGS data) which brings a great challenge to the analysis and interpretation of information.

### Interpretation of the connotation of genomics analysis

In previous targeted testings, each test provides a limited amount of information, and there is generally no information redundancy. However, genomics testing is different; you can first

obtain more comprehensive information in one respect, and then extract abundant information to guide the diagnosis and treatment of diseases from different angles according to the needs. Genomics has brought revolutionary advances from "targeted testing" to "targeted analysis" in terms of testing and analysis modes.

We can use the genomics testing to analyze genetic disease and genetic predisposing factors of tumor, somatic mutation, genetic pharmacogenomics, minor histocompatibility antigens (mHAg), tumor neoantigen prediction as well as telomere length and chimeric rate [5-11].

### The era of comprehensive clinical genomics analysis is coming

Now the cost of WES is close to the cost of targeted sequencing of dozens or hundreds of genes, but the amount of information is greatly increased. The advantages of omics testing and analysis are that it can provide a variety of information of clinical diagnosis and treatment for patients by analysis of multiple aspects in one testing and analysis of several times in one testing, which can achieve the most economical cost and the information-price-performance ratio. At the same time, genomics data can also be used as a treasure trove of information, accumulating valuable resources for further medical research.

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