

Transcriptomic Profiling with Molecular Stratification in Autoimmune Diseases

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DESCRIPTION

Transcriptomics, which provides a link between the genome, the proteome, and the cellular phenotype, is the analysis of the RNA transcripts produced by the genotype at a specific time. It is a combination of processes that has developed significantly together with genomics, proteomics, and metabolomics. Transcriptomics is the study of each RNA molecule within a cell. The information required to produce proteins and perform other important activities in the cell is contained RNA, which is copied from fragments of DNA. Transcriptomics is used to learn more about the way genes are activated in various types of cells and this also may influence the development of diseases like cancer [1,2].

Transcriptomics profiling is the study of the transcriptome, which is the entire group of RNA. It is also known as expression profiling as it defines the levels of mRNA expression in a particular cell population. Hybridization- or sequencing-based techniques are commonly used for transcriptome profiling. In hybridization-based techniques, complementary probe sequences are linked to fluorescently labelled fragments either in solution or on a solid surface, such as a microarray. These approaches have limitations such as low resolution, low specificity, and low sensitivity. Later, Sanger sequencing-based approaches such as Serial Analysis of Gene Expression (SAGE), Cap Analysis of Gene Expression (CAGE) and Massively Parallel Signature Sequencing (MPSS) were developed but although, these approaches have serious limitations, including the inability to distinguish between isoforms and consideration of partial transcript structures for gene expression [3].

RNA Sequencing (RNASeq) has become a potent technique for analyzing the transcriptome with the development of Next Generation Sequencing (NGS), a technology that provides the sequencing of millions of nucleotide fragments simultaneously. Despite the high throughput and easy operation of microarrays, RNASeq offers numerous advantages over microarrays. The computational workflow for a study of reference-based transcriptome profiling typically begins with the use of a suitable read aligner to align the quality-checked reads to the reference

genome or transcriptome. The adjusted peruses are then used to quantitate the genomic highlights (qualities/isoforms). Before comparing different experimental conditions, the quantity of features needs to be normalized.

Systemic Autoimmune Diseases (SADs) are a group of chronic inflammatory conditions with autoimmune etiology and numerous common clinical features, making diagnosis and treatment difficult. Due to the diversity of molecular mechanisms within the same disease class, it is particularly challenging to develop new treatments or improve existing ones. Reclassifying these conditions at the molecular level, which may lead to a more uniform stratification in terms of pathological molecular pathways, is the first step toward developing a precision medicine strategy for Systemic Autoimmune Diseases (SADs) [4].

The transaction of DNA methylation designs and environmental factors and between these, is determinant in the regulation of the immune system. This, the low concordance for autoimmunity in monozygotic twins, and the fact that the genetic contribution to disease is dependent on regulatory variants with very small effects suggest that epigenetic regulation may play a significant role in the onset of these diseases. As a result, data on DNA methylation may be a useful marker for reclassifying autoimmune disorders on a molecular level.

CONCLUSION

Patients with systemic autoimmune diseases can be divided into three stable disease clusters based on distinct molecular patterns that distinguish between various molecular disease mechanisms. A paradigm shift in our understanding of systemic autoimmune diseases is marked by these findings, which have significant implications for upcoming clinical trials and the investigation of nonresponse to therapy. Early diagnosis and effective treatment are inhibited by clinical heterogeneity, which is characteristic of systemic autoimmune diseases. These issues may be addressed if patients could be divided into groups based on molecular pattern. The goal of this research was to find molecular clusters that could be used to reclassify systemic autoimmune diseases without regard to the clinical diagnosis.

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