CLINICAL INVESTIGATION

Health and genomics: Functional genomics

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Editorial

The use of functional genomics in clinical illness diagnosis and prognosis enables advancement in all aspects of biology in health and disease. This Special Issue of IJMS on "Functional Genomics in Health and Disease" is important since it attempts to cover the entire biology of coding and non-coding genes.

Maestri describe a Long-Read Sequencing Approach for Direct Haplotype Phasing in Clinical Settings in this issue. This has significant translational implications because several samples can be analysed. In clinical contexts, these predictive indicators can be utilised to select patients for therapy groups.

Vyse and colleagues discuss how trans-ancestral fine-mapping and epigenetic annotation can be utilised to identify functionally relevant risk alleles in Systemic Lupus Erythematosus.

Using an integrative approach, Dudea-Simon and colleagues reveal the diagnostic and prognostic potential of important core genes for cervical cancer. The potential interaction of miRNAs with important signalling pathways in infected monocyte-derived macrophages and peripheral mononuclear blood, allowing the creation of novel biomarkers that can distinguish HIV-1 and HIV-2 infection based on an altered miRNA pattern.

Zayed demonstrates a new bioinformatics method for determining the molecular genetic susceptibility pattern in asthma. This study led to the discovery of new genetic risk factors for the development of moderate-to-severe asthma, providing a foundation for better asthma treatment.

Bioinformatics study that focuses on the important genes and biological processes linked to dementia. The phosphodiesterase 4D-Interacting Protein was found as a crucial factor for the frontal brain dementia switch gene in this investigation.

The roles of extracellular vesicle biology are discussed by Chitoiu and colleagues, which is a particularly interesting topic in light of the recent success of multi-omics data.

It provides an overview of genome-wide association studies, focusing on triple-negative breast cancer, and provides substantial and fresh information for both research and clinical trials.

As a result, the goal of this Special Issue is to provide answers to many of our readers' questions about this topic, as well as to open up new research routes, resulting in key findings in this continuing revolution.

It is critical to first identify function in order to comprehend functional genomics. Graur, et al. define function in two different ways in their paper. "Selected effect" and "causal role" are two terms that come to mind. The function for which a trait (DNA, RNA, protein, and so on) is selected is referred to as the "selected effect" function. The function that a trait is sufficient and essential for is referred to as the "causal role" function. The most common definition of function in functional genomics is "causal role."

Function-related features of the genome, such as mutation and polymorphism (such as single nucleotide polymorphism (SNP) analysis), as well as the measurement of molecular activities, are all part of functional genomics. Transcriptomics (gene expression), proteomics (protein production), and metabolomics are some of the "-omics" included in this category. Multiplex methods are commonly used in functional genomics to determine the amount of many or all gene products such as mRNAs or proteins in a biological sample. A more targeted functional genomics method might use sequencing as a readout of activity to examine the function of all variants of one gene and quantify the impact of mutations. These measuring modalities work together to quantify numerous biological processes and better understand gene and protein activities and interactions.