



Gene Discovery in Complex Disease

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DESCRIPTION

Complex or multifactorial diseases are ultimately defined as diseases that are determined by a variety of genetic and environmental factors. There are many techniques and strategies that can be used to detect genetic factors that affect complex diseases, but these techniques and strategies have their own limitations. The name "complex illness" alone shows that it is not easy to decipher the results of related studies. Ultimately, it is a difficult endeavor to demonstrate and accurately characterize the contribution of a factor to a complex disease, as the effects of one factor can be confused by another. However, if the two very fundamental differences are observed, genetic dissection of complex diseases will be much easier. The first difference is the complexity at the individual level and the complexity at the group level. The second difference is the difference between the two components of the gene discovery gene identification and characterization of gene effects. Genetic epidemiology as a research discipline addresses both elements of gene discovery for complex diseases, but is particularly well suited for characterizing genetic effects at the population level.

Non-coding regulators such as transcription enhancers are important for accurate regulation of gene expression. Transcriptional regulation is a very complex process, often mediated by sequences of enhancer elements that can be separated from regulatory genes above the mega base. *Drosophila* studies have shown that important developmental genes are often regulated by multiple "shadow" enhancers. It has an excessive pattern of activity and is therefore protected from genetic disorders. Recent studies suggest that similar tissue structures may be present in the mammalian genome. Mammalian developmental genes have been reported to be

located closer to more enhancer elements than the average gene. Finally, differences in enhancer element activity are often not reflected in changes in gene expression.

Consistent with the important role played by non-coding enhancer elements in gene regulation, many groups have shown that enhancers are rich in common disease-related gene variants in contrast. Rare disease, related mutants are primarily localized within proteins the coding regions of the identified genes, and studies using Whole Exome Sequencing (WES) have led to genes and disease. Mutations are effectively associated with conditions such as epilepsy, idiopathic pulmonary fibrosis, It is based on the hypothesis that the uncoding transcriptional regulatory landscape of a gene reflects the characteristics of the gene itself. Therefore, we attempted to construct a human gene regulation and evaluate its ability to prioritize genes important in human disease. Determining which regulatory element is associated with target gene, remains a significant issue in the field of genomics and gene regulation. Many approaches have been developed but most methods are currently limited to a number, depending on the enhancer element or organization type. Genes associated with human disease are enriched with a set of genes that are not evolutionarily conserved (eg, genes with high number of enhancers) or genes with high total number of enhancer for nucleotides, the number of preserved enhancer nucleotides is counted in the same way.

The functioning human body is a very complex biological phenomenon. A set of biochemical networks, physiological systems, and organs need to work together in a complex and coordinated way for the human body to thrive. There are many properties of the human body that require to control of various biochemical and physiological mechanisms.

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