Special Issue

Functional Non-Coding SNPs in Health and Diseases

Message from the Guest Editor

Genome-wide association studies have revealed many associations between single-nucleotide polymorphisms in human genome and predisposition to complex diseases, such as autoimmunity, cardiovascular disease, and cancer. These data have strong potential to improve our knowledge of the molecular mechanisms of these diseases and to provide drug developers with new therapy targets. However, a great amount of additional research and conceptualization are needed to transform the statistical association data into valuable fundamental and practical output. Which particular SNPs in LD groups are functional and truly causative? Are they found among common variants or do rare polymorphisms mediate the disease risk? Can an isolated variant significantly influence the disease molecular pathways or do SNPs in strong LD function as haplogroups? Expression of which genes and in which cells and cell states do noncoding functional variants affect? And is the transcriptional effect of noncoding variants mediated only by altering transcription factors binding, or are other mechanisms also involved? Dr. Marina A. Afanasyeva

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