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Study of Human Genetics and Relations with Complex Diseases

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Opinion

Study of human genetics has developed significantly rapid during the past decades. In one perspective, Biotechnology and the branch of Genetic engineering has been pushed to another new era due to the fact that more and more new and mature methods came into being in process [1]. Prospective methodology made the study of human genetics more reliable and the mechanism behind complex diseases is being more and more well investigated and discovered [2]. In another aspect, bioscience has reached a relatively high level in the trend of big data times [3]. From perspective of bioinformatics and computational genetics, theoretical frameworks and pipelines are becoming much more applicable and preferable for both biostatisticians and biologists to exploit since previous results are more than promising [4].

A while ago, transcriptome research has been heavily focused to the discovery of human disease mechanisms [2]. A good amount of investigations is remarkable in this area and gene-level studies really revealed a lot of genetic mechanisms of human diseases such as tumor, obesity, diabetes, heart defects, mutation, transferring the defect gene from one generation to next one, schizophrenia, etc. Gene expression technologies such as microarray and RNA-sequencing etc. are widely used. Related methodologies are well developed and summarized [5]. Combing with efficient high-throughput data oriented analysis methods such as single-cell gene expression analysis and so on, transcriptomic analysis has become extremely popular in this field. With developments of genome wide association studies (GWASs), thousands of associations between genetic variants and complex human traits are known.

A literature review for statistical methods of rare variant association studies could be found in [6]. However, translating the associations into epistemology of human disease mechanisms has fundamental challenges due to the fact that information and knowledge of noncoding regions of the human genome in both technical experiments and bioinformatics theories are still relatively exile [7-8]. Thus, studies in the area are nowadays especially active and trying to formation of interdisciplinary innovative techniques which will stand for the upcoming challenges that would be disease detection, disease diagnosis and gene expression profile.

Epigenomic study is another area that geneticists pay much attention to. Expression quantitative trait loci (eQTLs) are important since they influence the expression level of genes [9]. Many tools that employ statistics methods show up to investigate sequence variants in eQTLs. Genotype-phenotype study meets the demand of discovering the associations between genotype and human diseases as well [10].

Also, people have already found some relations between genotypic information and gene expressions, which make it possible to impute transcriptome through genotypes [11]. While till date, integrating epigenomic information into this process is still not well covered. More generally standard and efficient pipelines to link common traits and disease variants as well as causal genes are in high demand presently [12-14] as people need to break through the current bottleneck and promote the study in the field to a platform of higher level.

This special issue of our journal intends to make contribution for community of human genetic study and complex human diseases. The special issue is also associated to detection, diagnosis, development of new techniques, development of new drug relating drug screening to that diseases, the applicable treatment to cure disease.

Hope the techniques and methods that rose among our authors and the professional experiences shared by them therein could give impetus to the study and provide reference in this area.

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