Single Nucleotide Polymorphism (SNP) and Its Preview

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Commentary

The announcement of human genome in June of 2000 pleasantly sent shock waves throughout the scholarly community and the global world population. It announced the completion of a draft of human genome, along with the arrival of post-genomic era. In fact, in the post-genomic era, we are focusing on DNA sequences, as these sequences in the human body determine the specific trait of an individual. To fully understand the significance of these sequences in the human body, we must first truly understand the ‘single nucleotide polymorphisms’ (Single Nucleotide Polymorphism, SNP).

What are the so-called single nucleotide polymorphisms? They are actually the positions of some genes in DNA sequences with mutation(s). When mutations occur in certain positions, they often give rise to some kind of disease or even diseases. The human genome is normally composed of about three billion base pairs of nucleotide bases: A, T, G, and C. There is a single nucleotide polymorphism in about every 500 bp ~ 1000 bp (bp: base pair), therefore, the chance of the appearance of single nucleotide polymorphisms in the human genome is quite high [1]. SNPs are very important in the studies of human health. They can also be used to track the inheritance of disease genes within families. In the post-genomic era, it is worthwhile to focus the research to identify SNPs associated with complex diseases such as heart disease, diabetes, and cancer. A single nucleotide polymorphism in the post-genomic era really requires a major focus of research.

Future Prospects

1. The Analysis, diagnosis and prevention against diseases

Once the human data of single nucleotide polymorphism become more complete, we can accomplish the distribution pattern study for single nucleotide polymorphism in association through the data study of bioinformatics with the comparison between normal controls and patients. With such an intensive study, whereby, one can thus identify the cause of many genetic diseases, a prior diagnosis of some latent ethnic and vulnerable factors can hopefully be gotten under control.

2. As to the development of new drugs by pharmaceutical companies, it reveals such new drugs can also hopefully be helpful in removing toxic aldehydes including acetaldehyde, an intermediate of ethanol metabolism. Nearly 40% of East Asians inherit an inactive ALDH2 variant, which has a lysine-for-glutamate substitution at position 487 (E487K), and show a characteristic alcohol flush reaction after drinking and a higher risk for gastrointestinal cancers. Here there has been a report on the characterization of knocking mice in which the ALDH2/E487K mutation is inserted into the endogenous murine Aldh2 locus. These mutants recapitulate essentially all human phenotypes including impaired clearance of acetaldehyde, increased sensitivity to acute or chronic alcohol-induced toxicity, and reduced ALDH2 expression due to a dominant-negative effect of the mutation. When treated with a chemical carcinogen, these mutants exhibit increased DNA damage response in hepatocytes, pronounced liver injury, and accelerated development of hepatocellular carcinoma (HCC). Importantly, ALDH2 protein levels are also significantly lower.

3. Molecular genetic map markers: In 2002, from 15th to 18th of October, the U S National Geography Club conducted a global human gene (DNA) genetic program; this was led by internationally renowned geneticist, Dr. Spencer Wells, who had been to Taiwan and Hong Kong where the work of an Asian DNA sampling was carried out. This DNA research can be traced back to human ancestors about 60,000 years ago, and the ancestors’ DNA evolution process has been explored.

4. The establishment of such a so called ‘household’ record would need to transfer maps or so called ‘Atlases’ after applying as a marker. When an SNP is adjacent to the coding sequence of a gene, the SNP would be very likely linked together with the hereditary gene; the researchers then can compare the atlas with that of its counterpart from the control group. Hence, it will be essential to have further study of any individual pattern change, or to track any difference that may be related to genetic factors. To draw atlases of the effective SNP requires an estimated number between 100,000 to 1,000,000 SNP, with the ideal number ranging between 600 thousands and 100 million [2].

5. Diagramming of SNPs on biological networks with the intention to assimilate information about SNPs and protein arrangement mutations in biological networks, there are some developed node characteristic files for Cytoscape that permit the conjuring up of such informations in the background of networks. The utilization such node attribute files encompassing protein annotations permits the ID of the nodes in the network that have mutations and/or natural disparities. Such information on all the annotations existing for each SNP in the attribute files; these annotations can be castoff to visualize, screen and search the network. As aforementioned, in the pathway representation all dissimilar states of a protein appear as different nodes. Hence, the information about the protein mutation and ordinary variation of a given protein, on top of all the consistent nodes in the pathway. The UniProt identifier was used for this mapping and hence any pathway, protein-protein interaction network or network model containing UniProt identifiers can be prolonged with the attribute files, according to Masaru Katoh (2007) Dysregulation of stem cell signaling network due to germline mutation, SNP, helicobacter pylori infection, epigenetic change, and genetic alteration in gastric cancer, Cancer Biology & Therapy.

6. Mitochondrial aldehyde dehydrogenase 2 (ALDH2) in the liver removes toxic aldehydes including acetaldehyde, an intermediate of ethanol metabolism. Nearly 40% of East Asians inherit an inactive ALDH2’2 variant, which has a lysine-for-glutamate substitution at position 487 (E487K), and show a characteristic alcohol flush reaction after drinking and a higher risk for gastrointestinal cancers. Here there has been a report on the characterization of knocking mice in which the ALDH2/E487K mutation is inserted into the endogenous murine Aldh2 locus. These mutants recapitulate essentially all human phenotypes including impaired clearance of acetaldehyde, increased sensitivity to acute or chronic alcohol-induced toxicity, and reduced ALDH2 expression due to a dominant-negative effect of the mutation. When treated with a chemical carcinogen, these mutants exhibit increased DNA damage response in hepatocytes, pronounced liver injury, and accelerated development of hepatocellular carcinoma (HCC). Importantly, ALDH2 protein levels are also significantly lower.
in patient HCC than in peritumor or normal liver tissues [3]. Our results reveal that ALDH2 functions as a tumour suppressor by maintaining genomic stability in the liver, and the common human ALDH2 variant would present a significant risk factor for hepatocarcinogenesis. The aforementioned study suggests that the ALDH2*2 allele-alcohol interaction may be an even greater human public health hazard than previously appreciated, as per the ALDH2*2 allele-alcohol interaction may be an even greater human public health hazard than previously appreciated [5].

7. In the post-genomic sequencing era, one of the exciting directions to fully understand the meaning that underlying the genomic sequences is deciphering the epigenetic codes, including 5 meC/5 hmeC DNA methylation, protein/histone post-translational modifications and expression/splicing of non-coding transcripts. For instance, single nucleotide polymorphisms (SNPs) play a critical role in the study of imprinted genes. Imprinted genes refer to the mono-allelic expression of some genes according to their parent-of-origin, and the differentially existed SNPs on either maternal or paternal allele facilitate the investigation of the transcriptional origin at the imprinted loci. In mammals, as normal expression of imprinted genes is directly regulated by the DNA methylation that presents at the imprinted control regions (ICRs), 3 it is interesting to investigate the relationship between the establishment of DNA methylation and the variability of SNPs [4]. Strikingly, recent observation revealed that apart from DNA methyl transferases (DNMTs), some histone modifiers are also involved in the maintenance of imprinted DNA methylation, 4 which suggested that the differentially existed SNPs might also influence the deposit of histone modifications. Taken together, investigation of the link between SNPs and epigenetic markers might create a novel direction towards the fully understanding of human genetic codes.

**Conclusion**

With the advance in biotechnology, a variety of life experiences gradually has at least partially unraveled the mystery of creatures, but SNP is still the key to unlock such mystery. If this mystery can be totally unlocked, it will bring more understanding in the prevention of disease in human body, the treatment of various diseases, and ultimately the development of a variety of drugs that can expect to extend life span of human being. The development of biotechnology vastly improves the longevity of human life. The improvements of daily diet and living environment also have significant benefits. We are looking forward to seeing further advancement in the field of biotechnology and also have a high degree of anticipation that the development of a safe and effective cancer drug therapy will not remain elusive in the near future. As well, the ALDH2*2 allele-alcohol interaction may be an even greater human public health hazard than previously appreciated.

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