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Bio-computational Analysis of Human Genomic Variation in KCNH2 Gene in Chromosome 7

INTRODUCTION

SNP, otherwise known as Single Nucleotide Polymorphism is the single nucleotide substitution of one base for another in the genetic sequence of individuals. Polymorphisms are related to traits specific to a particular ethnicity or population.

Genetic Mutations, are a kind of genetic polymorphism, which is a difference in the genetic DNA sequence amongst individuals. Genetic mutations are more commonly associated with diseases. Hence, studies of SNPs is crucial in the study of health as they are able to predict an individual’s vulnerability to diseases.

KCNH2 is the gene that codes for the voltage-gated potassium channels. Variations along this gene are the cause of numerous cardiac disorders. SNPs in this gene is useful in evaluating risks in other heart-related syndromes.

OBJECTIVE

• Create a coherent computational method that is able to identify common SNPs in chromosome 7 in a population of healthy subjects of Southern Han Chinese origin.
• Single out novel, common SNPs amongst the samples that are not documented in databases.
• Distinguish regions where there are notable changes in amino acid sequences of the translated gene.

METHOD

Galaxy.org, an open source software, is used to process information from the 1000 genome project.

- Extract low coverage data for Chromosome 7 from 39 selected individuals.
- Compute allelic frequencies.
- Filter data samples to obtain SNPs with statistical parameters.
- Compare with human reference genome, hg18.
- Check for changes in amino acid at novel SNP positions.
- Locate positions of novel SNPs.
- Compare with dbSNP 131 to obtain new SNPs present in samples.
- Obtain all SNPs of samples.

RESULTS AND DISCUSSION

A total of 4 novel SNPs were found, of which 1 is found in the exon region. Results also demonstrated that the SNP at position 150286236 observed a change in base from C to G, of which both codons, CGG and CGC, code for Arginine.

CONCLUSION

The results did not yield any significant changes in the amino acid sequence after the gene was being translated. This implies that there is no effective change in the final structure of the proteins that KCNH2 codes for.

Future studies could be directed at establishing the relationship between changes in nucleotide bases and determining how changes at the intron regions could result in changes in the translation process.

Project Title: High Performance Computing for Next Generation Genomic Analysis (Part A)
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