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SNP discovery with fuzzy inference through computational model for decision making

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Abstract

SNPs discovery requires bioinformatics tools to be applied to different cases, with the ability to analyze "reads" from different sources and to establish reliable measures. These tools work with different methodologies concerning distinct attributes. However, similar results are expected, even when dealing with a same data set, but it's not unusual to yield different results, which leads to uncertainty in decision making when the results are discordant. This text describes a fuzzy inference decision model applied to assist decision making in cases when information is conflicting and also in the confirmation of coincident information.

Differences between specific base pairs of different aligned sequences are the most common type of generic variability. Such differences, know as single nucleotide polymorphisms (SNPs), are important in the study of variability of species, because they may cause functional or phenotypic changes, which, by their turn, can result in evolutionary or biochemical effects on the individuals of the species. The use of computational algorithms to SNPs investigation is a widespread practice and the Polyphred and Polybayes programs stand out, because they are widely used. Thus, it is expected these programs show similar results when they are using the same data set, despite used different methods, but it isn't unusual show conflicting results. The PhD thesis "Computational models to the identification of genomic information associate to the resistance to cattle tick", propose a fuzzy inference model to aid in the decision process, using the fuzzyMorphic.pl, a computational tool, write in Perl language, which allow the modeling and implementation of fuzzy inference system and this text shows the computational model, inference fuzzy model specifically, proposed and developed in context of quoted thesis, to aid decision support from previous or primary results to SNPs discovery. The model proposed explores these results to aid decision support when the results are divergent or confirm them when they are similar. The computational model is based in a two steps methodological procedure, like several protocols for data mining: the data pre-processor section and the fuzzy inference section. The data pre-processor section provides extraction, integration, selection, completion and deletion procedures, which are the common procedures to preprocessing for data mining and, depending on the characteristics of research, requirements for the processing of data. The second section brings the fuzzy system itself, which includes the fuzzification, inference and defuzzification procedures. The fuzzification processes can be described by membership functions composed by fuzzy sets in a standard format, to the inference process can be used the Mamdani's or Larsen's inference models and the defuzzification process can be represented by an output function with fuzzy sets in standard format and, furthermore, using the "center of maxima" as the defuzzification method, because it takes "multiply shots" on the output

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function. The Polyphred's method search for positions in sequences where were detected more than one nucleotide and the Polybayes' method look for polymorphic sites by evaluating the different nucleotides within cross-sections of a multiple alignment. However, both methods do not consider the base quality in the sequence consensus resulting of the alignment. The described model in this text combines this base quality with the previous results obtained from the Polybayes and Polyphred, setting new attributes to SNPs identification.

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The second International Society for Computational Biology Latin American regional meeting (ISCB-Latin America) took place in March 17th-21st 2012 in Santiago, Chile. More than 250 people attended, primarily from countries in Latin America. As a scientific conference with an international audience, all oral and poster presentations, as well as all printed materials, were in English.

The major aim of ISCB-Latin America 2012 was to deeply motivate and inspire young Latin American students and post-docs to conduct the best research possible in the areas of Bioinformatics and Computational Biology.

The first two days of the meeting (March 17-18) were dedicated to hands-on practical tutorials and workshops covering different topics of interest.

The main conference took place March 19th-21st and featured the following six topic sessions:

Session I. Comparative Genomics and Evolution

Session II. Genomics, Proteomics, Metagenomics and Metabolomics

Session III. Macromolecule Structure/Function Prediction

Session IV. Computer Aided Drug Design and Docking Simulations

Session V. Biomedicine and Immunoinformatics

Session VI. Functional Genomics and Systems Biology

Each session had two keynote speakers and six oral presentations. There were also two poster sessions.

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