

A PERSONAL GENOMIC INFORMATION ANALYSIS AND MANAGEMENT SYSTEM FOR HEALTHCARE PURPOSES

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Abstract: Currently, a large amount of personal genomic data can be generated at an affordable price in a short period of time due to the improvement in the DNA sequencing technologies. Abundant research results on genetic diseases have been published in recent years. Therefore, it is eventually possible to integrate multiple types of information together and apply them into genomic-based personalized healthcare. However, this is still a very challenging task for healthcare professionals because the desired information is hidden in highly complex and heterogeneous genomic data sets and spread in various databases, which were typically created for researchers. In this research project, a personal genomic information management and analysis system is created for healthcare professionals, especially physicians. To properly design such a system, an exploratory survey was conducted to identify the current status of physicians in using genomics in their clinical practice and to collect their expectations about the features of a patient genomic information system. The results of this study indicated that physicians have sufficient knowledge in genomics and they are interested in incorporating genomics into their clinical practice. The results also indicated that a well-designed patient genomic information system with desired features can help physicians to incorporate genomics into their clinical practice. Based on the survey findings, a personal genomic information system was created for the purpose of managing and analyzing patient genomic data. In this system, we first created an integrated database, and then developed data analysis algorithms to extract clinical information from patient genetic variation data, including disease-associated genetic variations and pharmacogenomic associations. Physicians can conveniently identify the genetic reasons for diseases and determine personalized treatment options based on the information provided by the system. A usability study was