Chapter 11
Towards an Intelligent Integrated Approach for Clinical Decision Support

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ABSTRACT

Nowadays, making use of big data is becoming mainstream in different enterprises and industry sectors. The medical sector is no exception. Specifically, medical services, which generate and process enormous volumes of medical information and medical device data, have been quickening big data utilization. In this chapter, we present a concept of an intelligent integrated system for direct support of decision making of physicians. This is a work in progress and the focus is on decision support for pharmacogenomics, which is the study of the relationship between a specific person’s genetic makeup and his or her response to drug treatment. Further, we discuss a research direction considering the current shortcomings of clinical decision support systems.

INTRODUCTION

Everyone’s talking about the value of big data in medical sector. However, as the data piles up – most of it is isolated in different silos, and health systems are struggling to turn big data from a concept into a reality. By definition, big data in medical sector refers to electronic health data sets so large and complex that they are difficult to manage with traditional software and/or hardware; nor can they be easily managed with traditional or common data management tools and methods (Akerkar, 2013a). Gradually, health-related data will be generated and accumulated, resulting in an enormous volume of data. The current medical data includes personal medical records, radiology images, clinical trial data FDA submissions, human genetics and population data genomic sequences, etc. Moreover, new types of big data, such as 3D imaging, genomics and biometric sensor readings, are also stimulating this exponential growth.

Predictably, it takes over 10 years and a billion dollars to develop a new medical treatment for a specific disease. This is because most medications that look promising turn out not to work for many
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Figure 1. Every person is unique, and drugs that are beneficial for some turn out to be ineffective or unsafe for others.

The diagram illustrates the concept of patient clusters based on drug response. Different clusters are represented with different outcomes: noxious but useful, noxious and not useful, not noxious and not useful, and not noxious and useful.

of the patients, and, even worse, some of the patients are harmed by certain drugs (see Figure 1). 50% of medication is prescribed sub-optimally, because it has no effect in particular clusters of people. A sizable fraction of this variability in medication efficacy and safety can be accounted for by individual differences in the ‘code’ that drives the human organism: the genome. This is addressed by an R&D domain of rapidly increasing importance and popularity: pharmacogenomics (Shin et al., 2009). The promise of pharmacogenomics is to know personal drug response in advance, optimizing the efficiency of medical treatments and avoiding harm. The term pharmacogenomics is often used interchangeably with pharmacogenetics. Although both terms relate to drug response based on genetic influences, pharmacogenetics focuses on single drug-gene interactions, while pharmacogenomics encompasses a more genome-wide association approach, incorporating genomics and epigenetics while dealing with the effects of multiple genes on drug response (Shin et al., 2009).

However, understanding a human genome is not trivial. The genetic code of each of us is made up of three billion letters. How can we help medical doctors and drug developers understand what the three billion letters in each patient’s genetic code mean and what the implications of these characters are for finding the best possible medical treatments?

It is becoming increasingly easier and cheaper to obtain the individual genetic code of individual patients. Advances in genetic sequencing in the last decade have made it possible to extract this code faster and cheaper than ever before. Figure 2 shows the cost for sequencing a full human genome since 2001 on a logarithmic scale. It is clear that the cost of sequencing a full human genome is, since 2007, decreased rapidly than exponentially.

Unfortunately, the interpretation of the large quantities of data generated by genetic testing is still associated with many difficulties and costs. This led commentators to speculate that while the $1,000 personal genome has been reached, the current methods for interpreting the data could cost $100,000 for each patient, rendering it infeasible for clinical practice (Mardis, 2010). Furthermore, it is becoming
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