Chapter 1

Bioinformatics as Applied to Medicine: Challenges Faced Moving from Big Data to Smart Data to Wise Data

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ABSTRACT

The emerging advances of Bioinformatics have already contributed toward the establishment of better next generation medicine and medical systems by putting emphasis on improvement of prognosis, diagnosis and therapy of diseases including better management of medical systems. The purpose of this chapter is to explore ways by which the use of Bioinformatics and Smart Data Analysis will provide an overview and solutions to challenges in the fields of genomics, medicine and Health Informatics. The focus of this chapter would be on Smart Data Analysis and ways needed to filter out the noise. The chapter addresses challenges researchers and data analysts are facing in terms of the developed computational methods used to extract insights from NGS and high-throughput screening data. In this chapter the concept “Wise Data” is proposed reflecting the distinction between individual health and wellness on the one hand, and social improvement, cohesion and sustainability on the other, leading to more effective medical systems, healthier individuals and more socially cohesive societies.

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INTRODUCTION

Nowadays, there is an increased need to improve healthcare and the way clinical decisions are made for the welfare of patients by improving diagnosis, therapies and prognosis of diseases so as to advance personalized medicine and targeted drug/gene therapy. The demands of Big Data analytics incorporating the latest computational intelligence and statistical methodologies together with data mining and machine learning methodologies are high and quite challenging. Especially due to the rapid developments of biomedical research, there seems to be an ever-increasing demand of various computational and mathematical approaches to analyze and integrate the resulting large-scale data with the molecular and bioinformatics basis of clinical science. In fact, the latest developments in Bioinformatics require that we move from a data management organizational culture to a learning organization culture. This perspective requires better and more effective collaboration among various stakeholders with the researchers who should be able to solve problems in an effective and flexible manner by efficiently collecting and analyzing data to meaningfully optimize and automate solutions. Big Data as a concept needs to be communicated to a wider community of stakeholders. Big Data is usually defined around four aspects: data volume, data velocity, data veracity and data value. Veracity and value aspects of Big Data deal with the quality and the usefulness of Big Data. Those two aspects make management a major challenge for most enterprises in terms of quality. The decisive criterion here isn’t necessarily the amount of (big) data, but its valuable content (smart) see Figure 1. Turning this data from big to smart is the challenge that needs to be addressed today in order to lead to real time assistance hoping to improve medicine and provide better more personalized treatment.

The main focus of this chapter will be on Smart Data Analysis (focusing on veracity and value) and the various ways needed to filter out the noise and hold the valuable data. That way, one can expect that problems would be solved more effectively ranging from business problems to the improvement of healthcare operations. Smart Data Analysis will open new avenues to optimize computing capacities, explore molecular biology, genomics and proteomics and Health Informatics applications and implications. Smart Data Analysis combined with declines in the cost of generating genomic and proteomic data have made the approaches to DNA sequencing, RNA-sequencing, and high-throughput screening and protein analysis more efficient and effective in terms of analyzing data but also have created new challenges in data analysis. Formal approaches to modeling and simulating regulatory processes in biological systems are supported by the emerging field of Systems Biology which has accelerated the convergence of discovery science with clinical medicine and their connection to improvement of medical systems. The chapter addresses these challenges researchers and data analysts are facing in terms of the developed computational methods they use to extract insights from next-generation sequencing (NGS) and high-throughput screening data. However, a great challenge remains: how to apply Smart Data in ways that lead to wise decisions concerning individual wellness and social welfare, and not focused on market imperatives. This leads to a critical perspective on Smart Data. As a consequence, in this chapter a new concept is proposed (i.e. “Wise Data”) see Figure 1, which aside of the focus of Smart Data on veracity and value also reflects the distinction between individual health and wellness on the one hand, and social improvement, cohesion and longevity on the other, leading to more efficient medical systems, healthier individuals and more socially cohesive societies. Therefore, the overall objective of the chapter is to help communicate and disseminate the importance of establishing the biological relevance of genomic and proteomic discoveries and related specific gene expression to realizing the clinical potential to make possible targeted therapy, personalized medicine and enhancement of human wellness.