Chapter 3.3
Genomic Databanks for Biomedical Informatics

Andrea Maffezzoli
Politecnico di Milano, Italy

Marco Masseroli
Politecnico di Milano, Italy

ABSTRACT

In the area of medical informatics, the recent ICT (information and communication technology) tools and systems supporting knowledge on sciences involved in the study of genes, chromosomes, and protein's expression level in various organisms, that is genomics and proteomics, are becoming necessary to develop new prospects for the comprehension of mechanisms lying at the base of biological processes which cause a disease. This can allow more effective diagnostic and treatment methods and also personalized pharmacological therapies. At this purpose, the mutual intervention of different sciences, such as biology, medicine, engineering, informatics and mathematics, becomes an indispensable step: The development of a science embracing all these fields is identified in bioinformatics, which was conceived for the analysis, storage and processing of huge amount of biological data. The achievement of all the aforementioned operations involves the creation of the so-called genomic or proteomic databanks, which represent a major source of information on nucleotide sequences, as well as biological, clinical, physiological and bibliographical annotations related to singular sequences. There are different types of databanks based on their peculiar characteristics and features (such as primary and derivative or specialized databanks), and several ways to access data stored in these databanks; there are also specific bioinformatics databank-based tools developed to perform searching operations and to extract significant information, in order to summarize and compare gene annotations related to the causes of a disease and finally to identify a list of the most significant genes as cause of disease.

MEDICAL INFORMATICS AND E-HEALTH IN THE POST-GENOMIC ERA

The subject of medical informatics (MI) plays a very important role among the several branches of bioengineering because it deals with the practical and theoretical issues of the implementation of ICT in a wide range of solutions in the medical field, as well as in the more general health sector.

The constant evolution and improving of the technologies involved make it possible to guess that medical informatics will become increasingly important in the years to come. Consequently, in the last years, the complexity of medical-informatics solutions causes them to always be referred to as belonging to the so-called e-health field. This term hints at a wider role and groups all the numerous activities arising from MI applications.

E-health responds to growing demands for quality health services, patient mobility, data recording and processing, and finally for the more rational management of the economic resources and human efforts destined to these services.

E-health tools and applications can provide fast and easy access to electronic health records at the point of need: They can support diagnosis by noninvasive imaging-based systems, they support surgeons in planning clinical interventions using patient-specific digital data, and they provide access to specialized resources for education and training. Digital data transfer enables more effective networking among clinical institutions across the world and the creation of a global network of centers of reference. Electronic health records also enable the extraction of information for research, management, public-health, or other related statistics of benefit to health professionals.

More generally, e-health aims to improve the overall quality, productivity, and efficiency of the sector.

For these purposes, new knowledge and skilled personnel are needed, as well as essential, in order to implement new technologies and gain adequate financial support, from both the private and public sectors. This will carry very important consequences for the industries operating in the field, the so-called e-health industries, which represent today the third industrial force in the European health sector, and which will bring new life to ICT industries.

Examples of the operational capabilities of MI are as follows:

- Communication networks in hospitals
- Medical databases
- Telemedicine services
- MI Web-based portals
- ICT tools and systems dealing with the patient
    - Diagnosis
    - Monitoring
    - Treatment
    - Prevention

In particular, if we refer to the ICT tools and systems (Lacroix, 2002), the related applications can generate new and undiscovered developments, especially if viewed from the perspective of the latter discoveries carried out after human-genome sequencing and in the post-genomic era, with the subsequent developments of genomics, the systematic study of genes, chromosomes, and nucleic acids in an organism, and proteomics, which analyzes the proteins' expression levels in the biological processes of an organism.

In the postgenomic era, the complete sequencing of the human genome has helped to develop new prospects for the comprehension of the many mechanisms existing at the base of biological processes that cause disease.

A new vision of pharmacology has also started, supporting so-called pharmacogenetics and pharmacogenomics, which study how an individual's genetic inheritance affects the body's response to drugs. Pharmacogenomics focuses on a treatment that must be personalized according to the genetic profile of the patient: This could potentially reveal