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The Application of Integration Approaches through Biomedical Informatics in Health

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Abstract

INFOBIOMED is an European Network of Excellence (NoE) funded by the European Commission within the sixth Framework Programme. The main objective of this collaborative project is to create an stable and lasting European structure in Biomedical Informatics (BMI). This new discipline is based on the synergy between Bioinformatics (BI) and Medical informatics (MI).

Using all the relevant information, from the clinical records level down to the genomic, proteomic and molecular information levels, BMI is providing the technical and scientific infrastructure, as well as the necessary knowledge to develop new useful instruments and methods to improve healthcare advancing towards individualized medicine.

The main challenge of INFOBIOMED is to infer, based on the direct application in four pilots, the line of action and collaboration for BMI to fully export the synergies between BI and MI. In this paper, we describe the facts that support the consolidation of BMI as a relevant discipline for future European healthcare.

Keywords:

Biomedical Informatics, Genomic Medicine, INFOBIOMED

Introduction

The technology and resources, the increased knowledge extracted from large amounts of genetic and proteomic and phenotypic data are transforming the practice of medicine mostly characterized up to now by treating symptoms but also looking into the most fundamental causes and understanding the mechanisms of diseases [1]

In this context the EU funded the Network of Excellence (NoE) INFOBIOMED in order to consolidate the challenge of "Structuring European Biomedical Informatics to Support Individualised Healthcare" setting a durable structure for a collaborative approach at the European level and mobilising the critical mass of resources necessary for enabling the collaborative approach. This NoE consists of a consortium of 17 groups related with BI, MI and BMI research areas, that supports the consolidation of BMI as a relevant scientific discipline to be taken into account for future healthcare [2].

This is, BMI provides the technical and scientific infrastructure, as well as the necessary knowledge to produce new useful instruments and methods to improve healthcare in order to advance towards individualized medicine.

Further information about INFOBIOMED project is available through its web page http://www.infobiomed.org/

Materials and methods

The network has been structured by activities that cover all the significant aspects that are relevant to both MI and BI and that have the potential to provide a space for synergy between them. These aspects are envisaged for promoting integration of the research carried out in the fields of reference and they can be included in blocks of activities or workpackages (WP): NoE management (WP1); dissemination and communication (WP2); training and mobility (WP3); Data interoperability (WP4); methods, technologies and tools (WP5) and pilots applications (WP6).

Work Packages 4 and 5 have a horizontal design, the former dealing with data interoperability and the latter concerned with methods, technologies and tools. WP6 of the NoE introduces a novel vertical approach to biomedical research and healthcare.

The work in WP6 can be summarized as the application of data interoperability and management and the development of methods, technologies and tools in four pilots as 'proof of concept' of the pursued integrative vision of BMI that aim to cover the whole range of information levels from molecule to population.

Considering the different levels of heterogeneity due to BI and MI perspectives, the INFOBIOMED consortium aims in WP4 to integrate genomic, proteomic, clinical and population databases. Responding to the needs, different approaches have been made according to information linkage, data and query translation. Several of these databases containing all information levels related with the research of the diseases of interest were developed.

Due to the importance and success of ontologies in scientific research, data integration is evolving towards

ontology based methods that can solve the problems associated with syntactic and semantics gaps across data sources. This task is also relevant in INFOBIOMED and it has been developed an specific tool called Ontofusion.

INFOBIOMED efforts related with key technologies, methods and tools (WP5) were directed towards three main goals:

1. Data analysis and information retrieval: to identify, define and index the computational tools required for database integration, using and developing techniques of data and text mining, machine learning, knowledge extraction and other systems.

Diseasecard is an example of an information retrieval tool for accessing and integrating genetic and medical information for health applications (http://www.diseasecard.org) along multiple databases. Also, INFOBIOMED partners have developed the PIANA (Protein Interactions And Network Analysis) software framework that facilitates the work with protein interaction networks.

2. Image visualisation and analysis in order to enhance and develop image analysis approaches related with medical and genetic imaging techniques, like the DIA application (Dental Images Analyser) that extracts information from digitised radiographs that have been converted to the DICOM standard. Radiographic data in DICOM can be accessed by any computer system, which may be an important advantage in the studies of complex diseases, where multiple databases exist.

3. Information systems and decision support tools to extract knowledge from clinical and genomic available data sources. New models and systems to computerize clinical practice guidelines that will be a step towards genomic based enhancement of clinical decision making.

There are complementary activities for promoting integration of BMI research like dissemination and communication activities and training and mobility programmes in order to spread the knowledge generated in BMI. In this area some tools have been adapted to the needs of INFOBIOMED such as the LINK3D application (www.link3d.net). It can be used as a research tool to allow researchers to hold virtual meetings in a secure way. The INFOBIOMED website houses the European Biomedical Informatics Gateway for the BMI community and other stakeholders. It provides a central repository of reference materials and a notice board for announcing BMI events worldwide.

Results

There are four pilots in INFOBIOMED WP6 that represent the demonstration of the BMI approach to biomedical research and medical practice. The four INFOBIOMED pilots were based on several clinically relevant areas like pharmainformatics, genomics and microbiology, genomics and chronic inflammation and genomics and colon cancer.

Pharmainformatics (WP 6.1) carries out research on the impact of BMI in the drug discovery process, focussing in two case studies (Complex Regional Pain Syndrome and Nuclear Hormone Receptors) that establish an information continuum from pathology to pathway to target to ligand/ approved drug.

The approximation given by this pilot is mainly an example of translational research since its discoveries begin at “the bench” with basic research—in which scientists study those diseases at a molecular level—then progress to the individual level.

Genomics and Microbiology (WP 6.2) focuses its BMI activity on the study of comparative and functional genomic approaches combined with proteomic strategies in order to improve clinical evaluation strategies through a comprehensive pathway-related with host/pathogen interaction. The pathogens selected for this pilot are Cytomegalovirus and HIV.

In this pilot application, INFOBIOMED concentrates its efforts on the interferon pathway, combining host and virus genotype data with clinical data in order to find new markers of host immunity and viral therapy resistance. For this reason this study tries to improve clinical management obtaining information from molecular to individual level.

Genomics and chronic inflammation (WP 6.3) investigates the susceptibility to adult chronic periodontitis. A data warehouse was built combining many different causes that contribute to this complex inflammatory disease: genetics, infection, environment, intermediate phenotypes and disease phenotype. In order to gain more insight in periodontal disease, to design new treatment strategies and to devise preventive methods, this pilot focussed on the clinical management and disease prevention approach from the population to the molecular level information.

Genomics and colon cancer (WP 6.4) studies Hereditary Non Polyposis Colon Cancer. It uses the accumulated knowledge to build an infrastructure based on standards (e.g transmission of pedigrees including geno and phenotypes) that have been developed in XLM. To facilitate the use of this tool in different countries it is based on the international standard HL7. This pilot is mostly oriented towards public health, in particular towards prevention and exchane of information from the population to the molecular level.

Conclusions

The INFOBIOMED Network of Excellence is contributing to create a stable and lasting European BMI structure to

support individualised healthcare. Overcoming the obstacles and deploying the promised benefits of genomic revolution to society by combining the experience in both BI and MI developments, the methods and technologies used within the BMI integrative approach allow a direct application in the development of genomic medicine.

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