Convergence of genomics and e-health

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Presentation Structure

- Medical informatics
- Bioinformatics
- Synergy between Bioinformatics and Medical Informatics: Biomedical Informatics
- Convergence of Genomics & e-Health: Telegenetics
Introduction

- In this presentation medical informatics/e-health and bioinformatics are compared and contrasted and an opinion is provided for their additionality and the possibility for their collaboration.

- It is pointed out that, while the two scientific areas differ in the histories, in the scientific foundation, and in their methodological approaches in the research in various topics, nevertheless they shared methods and tools, which provides a base for the exchange of experience in their different requirements. The new sector of biomedical informatics gives big promises for the growth of methods of information technology that will be critical in the growth of genomic medicine and molecular medicine.
Medical Informatics

Clinical practice
clinical guidelines

Acquisition
Storage
retrieval
use of information

Medicine

Medical Informatics

Information Science

Computer Science

Resources
devices
methods
Medical Informatics

In medical informatics emphasis is given in
• health information systems,
• hospital information systems,
• smart cards,
• the electronic medical record,
• medical image systems,
• classification and coding systems,
• decision support systems,
• expert systems,
• telemedicine,
• medical education at distance and
• the safety and security of medical data.
Medical Informatics

- Medicine centred discipline
- Has been around since the introduction of computers in hospitals
- It deals with the resources, devices and methods required to optimize the acquisition, storage, retrieval and use of information in health and biomedicine
- Patient centred perspective
- Individualised healthcare
- Used by clinicians in a clinical setting
- Used by patients for self management
- Immediate impact on medicine
Medical Informatics

- The data are clinical (apart from the clinical trials) and the imperfection, the subjectivity, and the uncertainty of retrospective patients data make the results of data mining difficult to be repeated and be transferred. It exists innovative networking (Medline, SUMEX-AIM), documentation standards, glossaries and coding (e.g. UMLS, SNOMED, HL7). Emphasis has been given to medical information systems for patient care in hospitals, to education and telemedicine.
Medical Informatics

- Electronic Medical Records
- Telemedicine
- Virtual healthcare teams
- Evidence Based Medicine
- Health knowledge management (or specialist-oriented information provision)
- Consumer Health Informatics (or citizen-oriented information provision)
- Epidemiological Medical research
Bioinformatics

- Bioinformatics and Computational Biology incorporate the use of techniques including applied mathematics, computer science, artificial intelligence, chemistry, biochemistry, molecular biology and genetics to solve biological problems usually at molecular level.
Bioinformatics

- Biology centred discipline
- Handles large amounts of data generated in the laboratories for biological research
- Biological sequence analysis
- Structural biology /molecular information
- Web based databases
- Completion of the Human Genome Project years ahead of schedule
- Indirect impact on medicine
- Non individualised aspect
Past
Pre-Genomic Era

- Medical informatics and Bioinformatics separate disciplines
- No reason for synergy

Medical informatics
- Classical epidemiology
- Clinical research and practice

Bioinformatics
- Genomic research
Present/Future Post-Genomics

• Medical Informatics and Bioinformatics are no longer enough for advancing genomic medicine
• A new integrative approach is required
• Electronic correlate of phenotypic with genotypic information
• Study interaction between genes and environment (multi-factorial)
• Intelligent integration and combination of all the data generated requires synergy between Medical Informatics and Bioinformatics
Synergy between Medical Informatics and BioInformatics: Biomedical Informatics

Medical Informatics

Bio-Informatics
Levels of biological organization from genes to proteins, cells, tissues, organs and finally the whole organism
<table>
<thead>
<tr>
<th>Health information level</th>
<th>Classical health informatics applications</th>
<th>New genomic data and information</th>
<th>New medical informatics applications</th>
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</table>
| **Population**           | • Public Health & epidemiology databases and surveillance networks  
• Technology assessment, outcomes research | • Genome epidemiology  
• Genetic Screening | • Genome epidemiology databases and networks (CDC-HuGeNet) |
| **Disease**              | • Computerized clinical practice guidelines (CCPGs)  
• Information systems in clinical trials | • New classification of disease based on its molecular causes  
• Clinical trials in pharmacogenetics | • CCPGs including genetic tests and therapy follow-up based on genetic data  
• Decision-making support tools  
• Pharmacogenetics databases |
| **Patient**              | • Computerized patient health record (CPHR) | • Genetic individual profiles (SNPs, mutations) | • Genetic data in the CPHR |
| **Organ Tissue**         | • Pathology lab systems, medical image processing | • Physiological genomics  
• Genetic networks | • Disease models  
• Tumour databanks (Integration of clinical markers and genomic analysis) |
| **Cell**                 | • Imaging in Cytogenetics, histology  
• Microbiology lab information systems | • Gene expression profiling  
• Proteomics | • Molecular classification of disease  
• Information systems in pharmacogenomics (drug R&D)  
• Molecular imaging |
| **Molecule**             | • Biochemistry and genetic tests and laboratory information management systems | • DNA and protein sequences  
• Macromolecular structures | • Facilitating integrated and guided access to relevant databases to health professionals |

Health Information Systems in the age of Post-Genomic Research, Fernando J. Martin-Sanchez,
Ultimate scope
Individualised Healthcare

– Integration of clinical and genetic information
– Molecule to organism modelling
– Disease genetic/environmental component modelling
– Individualised therapies
  • Impact on preventative medicine
– Genetic testing
  • Genetic screening and counselling
  • Patient safety
Knowledge Management and Decision Support Intersection Points in Personalized Medicine

- Patient Encounter (direct care or clinical trial)
- Test ordering and documentation guidance
- Structured Test Result Interpretations
- Integrated Genotypic Phenotypic Database
- Therapeutic ordering and documentation guidance
- Personalized Medicine Decision Support Knowledge Repository

Knowledge Acquisition & Management

- IT Support of Personalized Medicine, John Glaser, 2007
Challenges of IT Support Personalized Medicine

• **Infrastructure and applications**
  – To what degree can we leverage patient care data to support research on the genomic basis of disease?
  – How do you structure, store and “operate” genomic and proteomic data and transactions?
  – What are the methods needed for processing this data?
  – How different will our clinical systems be in ten years?

• **Implementation and care improvement**
  – What is the impact on the safety, quality and efficiency of care?
  – Will we significantly accelerate the discovery process?
  – What steps are needed to manage privacy?
  – How should we educate our medical staff?
  – What are the new issues in our approaches to practice?
  – How should we work with the payers on reimbursement strategies?

• IT Support of Personalized Medicine, John Glaser, 2007
Prioritised Opportunities and Challenges in Biomedical Informatics

- Medical Genetics Databases and Initiatives
- Proteomics Information and Analysis
- Therapeutic Area Focused Initiatives
- Gene Expression Information in Medical Diagnostics & Prognostics
- Modelling & Simulation of Biological Structures, Processes & Diseases
Prioritised Opportunities and Challenges in Biomedical Informatics

• Medical Annotation of Biological Databases
• Functional and Molecular Image Processing
• Integration of Data from Biosensors with Clinical Information Systems
• Integration of patient molecular data in Electronic Health Records
Prioritised Opportunities and Challenges in Biomedical Informatics

- Systems for Clinical Decision Making
- Molecular Information Interfaces for Physicians
- Semantic Interoperability and Ontologies in Biomedicine
- Technologies for Biomedical Information Integration
- Multilevel Modelling and Vertical Information Integration
- Mining Biomedical Literature
- Data Interoperability & Standards
Prioritised Opportunities and Challenges in Biomedical Informatics

- Connecting biobanks to large scale databases to enable data mining.
- Registries linking molecular, familial and clinical data
- Information needs from research in Infectious/Tropical Diseases
- Biodefense information systems and networks
- Addressing information needs from research in infectious and tropical diseases
Prioritised Opportunities and Challenges in Biomedical Informatics

- Patient Risk Profiling and Lifestyle Management
- Identity Confirmation and Personal Genomics
- Informatics to support Pharmacogenetics and Stratified Clinical Trials
- Informatics to enable Medical Device Development and Biosensors
- Applied Pharmaceutical Research
Prioritised Opportunities and Challenges in Biomedical Informatics

- Data Security and Accuracy Considerations
- Post Marketing Surveillance of Drugs and Pharmacovigilance
- Clinical and Ethical Issues related to biomedical data processing
- Health Management Information Systems in the context of genomic medicine (including reimbursement)
Prioritised Opportunities and Challenges in Biomedical Informatics

- Addressing the need for biomedical informatics scientists
- Providing minimal skills to manage health information to researchers and carers
Convergence of Genomics & e-Health: Telegenetics

• Telemedicine services are a reality nowadays, covering many scenarios (e.g. teleconsultation, remote monitoring, training and education, emergency, tele-surgery) and medical specialities (e.g. radiology, cardiology, obstetrics, pathology, psychiatry, genetics).
An integrated concept of Home Tele-assistance

- **TELE-ALARM**
  - Alarm Devices
  - Phone

- **TELE-MONITORING**
  - Monitoring Devices
  - Adapter

- **TELE-PRESENCE**
  - Treatment supervision
  - Remote home care support
  - Physician visit support

- **ACCESS TO INFORMATION**
  - Access Terminal
  - Adapter

- **HOME ENVIRONMENTAL CONTROL**

- **OTHER LEVELS OF HEALTH CARE**
  - TELE-ASSISTENCIA Work Stations
  - TELE-MONITORING Stations / Servers
  - TELE-PRESENCE Routing

- **ACCESS TO INFORMATION PROVIDERS CENTERS**
  - local Information Providers Center

- **Home Environmental Control and Survey**

- **Patients being Rehabilitating**
- **People with disabilities**

- **Home Confined Patients**

- **Chronic Patients**
Tele-alarms

Attention in emergency situations

Fire, smoke, burglary detection..
Health Care Emergency

- ISOLATED FIRST AID SITES
- MOBILE UNITS
- PARAMEDICAL STAFF

TRANSMISSION MEDIA
- GSM
- SATELLITE

HEALTH EMERGENCY COORDINATION CENTRE

EMERGENCY CALLS

SPECIALIZED CARE UNIT

- New applications for these new procedures
- Managerial changes
- Selection of the appropriate HECC

ACADEMY OF ATHENS

BIOMEDICAL RESEARCH FOUNDATION
Convergence of Genomics & e-Health: Telegenetics

• In the domain of genetic medicine there are currently a significant number of services being delivered using telemedicine.

• Services in the domains of cancer genetics, clinical genetics and reproductive genetics can be found in the literature.
Convergence of Genomics & e-Health: Telegenetics

- In fact, many genetic centres that routinely utilize phone consults with physicians and phone interactions with patients to help determine the need for genetic services or to prepare for an appointment, are moving to Internet based services, and incorporating all the needed security and confidentiality requirement.
Convergence of Genomics & e-Health: Telegenetics

• For genetic counsellors and medical geneticists telemedicine is a powerful tool bringing together multiple kinds of distributed information: personal and family history, physical findings, and radiology and pathology results.
“Some of the claims for the medical benefits of genomics have undoubtedly been exaggerated, particularly with respect to the time scales required for them to come to fruition. Because these uncertainties, it is vital that genomics research is not pursued to the detriment of well-established methods of clinical practices, and clinical and epidemiological research. Indeed, for its full exploitation it will need to be integrated into clinical research involving patients and into epidemiological studies in the community.

It is crucially important that a balance is maintained in medical practice and research between genomics and these more conventional and well tried approaches”.
References


• Health Information Systems in the age of Post-Genomic Research, Fernando J. Martin-Sanchez, “Joint Working Group Meeting”, Madrid, June 3, 2002
References

- Telemedicine Glossary 5th edition Glossary of concepts technologies standards and users 5th edn:
References

References

References

References


• Bidgood WD, Jr., et al., Understanding and using DICOM, the data interchange standard for biomedical imaging. J Am Med Inform Assoc. 1997, 4:199-212.


• Godard B, et al., Data storage and DNA banking for biomedical research: Informed consent, Confidentiality, Quality issues, Ownership, Return of benefits. A Professional Perspective. EUROGAPP PROJECT 1999-2000, report; European Society of Human Genetics, 2002
References

References

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