infogenmed

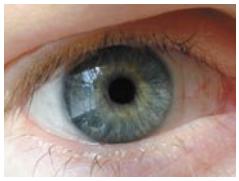
bringing together

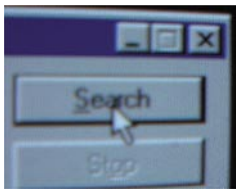
genetic and clinical information

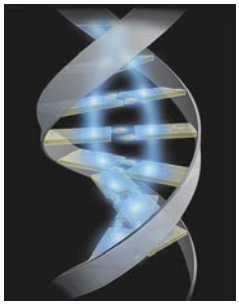
for medical applications

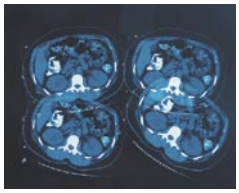
infogenmed
IST 2001 39013

Project brochure November 2002







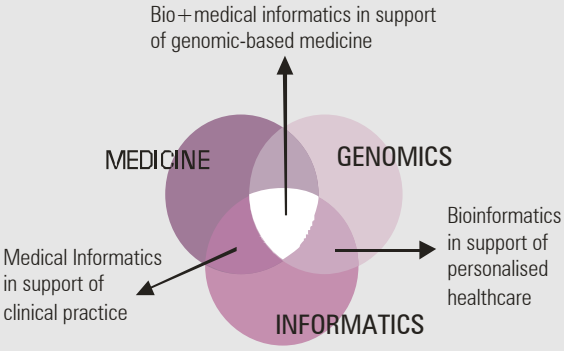


Infogenmed in a nutshell

The increasing availability of genetic and clinical information, both in public and in-house databases, demands for practical information technology tools, able to give clinicians integrated and comprehensive access to previously scattered and unconnected data.
INFOGENMED is building a software environment to access and integrate genetic and medical information for health applications. Medical information regarding diseases will be easily accessible and related genetic information, a type of valuable data the clinicians are mostly unfamiliar with, will also be located, retrieved and presented in an unified, user-friendly way.
This will empower medical practice, research, knowledge development and the collaboration between bioinformatics and medical informatics, paving the way for individualized medicine: as the understanding of the genetics base of drug action increases and genetic information is added to patient records, both patient-tailoring of medical action and population studies of genetic epidemiology will be improved.
This innovative approach will be tested in the field of rare genetic diseases. It will bridge the gap between scientific fields, retrieving and placing relevant medical and genetics information where it is of most use: in the clinicians' hands, for the patients' benefit.

Problem and motivation

Medical informaticians have long been providing physicians with computing aids for patient care and management, while bioinformatics experts have more recently been building and managing large databases of genetic information, as the Human Genome Project exemplifies.
There has been little collaborative effort between those professionals. Infogenmed is one of the first such attempts, building methods and integrated tools that enable the access to and the use of dispersed, heterogeneous databases, to improve clinical and research practice.



Infogenmed concept. Medicine, genomics and informatics are merging together to create new sinergies for patient care: knowledge empowered genomic-based medicine, supported by practical IT tools.

Technical challenges

For Infogenmed to enable the envisioned access to medical and genetic information from scattered and heterogeneous sources by professionals with different backgrounds, a number of complex problems have to be overcome:

- Relevant information is distributed (along local, remote and Web-based databases)
  Adequate tools and methods will locate, access, retrieve and integrate information
- Relevant information is presented in a wide heterogeneity of formats, codification and terminologies
  A vocabulary server will be set up and format conversion rules established

• Dissemination will be determined both by effectiveness and ease of use

A virtual repository and a single user-friendly interface will abstract the user from the underlying complexity of the system

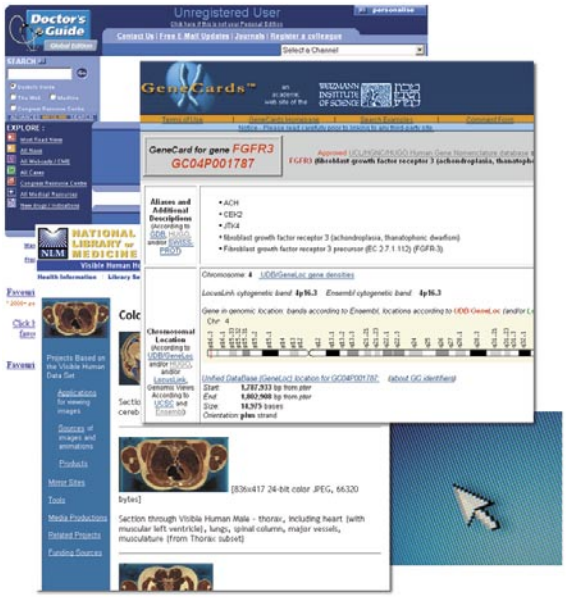
• Most targeted users are unfamiliar with (the particulars of) genetic information

An internal assistant will provide basic guidance

Products and achievements

Having started in September 2002, Infogenmed will reach conclusion in September 2004, pioneering:

- the design of a system to support the search and linkage of the contents of scattered databases, including the
  - — development of methods and tools to locate, access and integrate distributed (medical and genetic) data;
  - — construction of a vocabulary server for the association of (medical and genetic) terms (ontologies);
  - — design of a user-friendly interface and
  - — development of an "assistant" to help users in resorting to such methods and tools for the benefit of their practice (e.g.: flowchart representation for pathway visualization);
- the field-validation of the entire system in the field of rare genetic diseases.



A net of biomedical knowledge. Currently, amazing quantities of information in genetics and health are available in the Internet. Infogenmed is developing the tools to bring that knowledge to every day clinical practice.

Infogenmed consortium

The Infogenmed consortium joins institutions with complementary backgrounds, expertise and research skills, including:

- three universities;
- a national health institution;
- and an industrial partner.

This multinational team is working to support health professionals and enhance their ability to prevent, diagnose, care and rehabilitate patients: in the end, objectives of the European Information Society Technologies (IST) programme, Key Action 1 Systems & Services for the Citizen/Health.

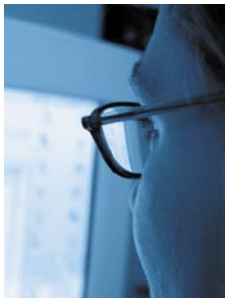
IEETA is a leading research organization of University of Aveiro, Portugal. It is particularly renowned for its work in the fields of signal processing, telematics and biomedical engineering. It is the project coordinator and participates in several technical tasks.
http://www.ieeta.pt

Universidad Politecnica de Madrid (UPM) is a leading Spanish university in the fields of engineering and applied informatics. UPM, through the Department of Artificial Intelligence (DIA), is the main responsible partner for designing and implementing the tools being developed in the project.
http://www.dia.fi.upm.es

Instituto de Salud Carlos III (ISC) is the Spanish national institute of health. It provides strong expertise in Bioinformatics and clinical applications of genomics. The role of ISCIII in the project is coordinating the database creation and the clinical testing in hospitals.
http://www.isciii.es/biotic

Linkoping Universitet brings the Swedish experience and expertise in biomedical engineering, bioinformatics and telematics in Healthcare. Linkoping is particularly active in analyzing the confidentiality and security issues of the project.
http://www.imt.liu.se

Genomica is a private Portuguese company exploiting and transferring to wide range of users the latest developments in genomics and genetics. Accordingly, it is mostly concerned with the dissemination and exploitation of project results.



A vision for the future

Mr. *S*. enters your consulting room. He has a family history of an inherited condition, *C*, a rare genetic disease, and is afraid he might be afflicted too.

This is a very specific condition and you wish to look into the most recent developments on the subject. On the medical side, you want to double check if no known symptoms or early warning signs are going unnoticed. On the genetics side, you would like information about the possibility of a predictive genetic test and related pharmacogenomics.

You turn to your computer, access the Infogenmed environment browser and quickly key in the standard name of the condition, in your own language.

According to your instruction, the informatics application autonomously ponders on the best way to provide help. You need not be familiar with genetics databases or other: the application connects to several free-access Internet databases on your behalf and requests and retrieves relevant information on the condition. And since all the information is presented in an integrated web page, you almost forget it comes from such different sources.

The search confirms that the symptoms and the condition are associated with gene *G*. You know that researchers sometimes dub that gene with other names but you need not worry about it: the vocabulary server provides the necessary correspondence and interconnections.

There is a novel genetic test of this condition: if Mr. *S*. so wishes, he can be screened (in one of the application-identified laboratories) and you will be able to suggest adequate lifestyle changes at this early stage so as to lessen the severity of the genetic disease, if necessary. Also, you might advise adequate psychological help to guide Mr. *S*. through the puzzling consequences of genetic testing: the results might recommend decisive (and intrusive) preventive action over some condition not yet apparent in his body.

Meanwhile, the application flashes the possible subsequent routes for you to proceed along the research protocol. You opt for "Molecule association". You are now shown the molecules associated with the gene disorder, so you decide to find out what related pharmaceutical options are currently available and discover that there is a new medicine just out.

At this stage, you allow the application to access a basic genetic profile of Mr. *S*., already present in your local server, to check for the compatibility of the new medicine to his individual characteristics, as well as to assess the best dosage for him.

You do so in confidence, since all personal information of the patient file is encrypted and his privacy is preserved as the query takes place. A few seconds later you find out that the new medicine should have significant therapeutic effects in Mr. *S*.' case.

You have reasons to smile as you turn and tell Mr. *S*. that there is increased hope for him in this new approach, even if he is found to carry the defective gene.

Get in touch

António Sousa Pereira - coordinator
IEETA - Instituto de Eng. Electrónica e Telemática de Aveiro
Universidade de Aveiro
Campus Universitário de Santiago
P3810-193 Aveiro - Portugal
Phone: +351 234 370 500
Fax: +351 234 370 545
E-mail: asp@ieeta.pt

More information

http://www.infogenmed.net

Questions, suggestions, collaboration? Pay us a visit, at our web site. You will find further information about the project and useful links for related resources.

Aknowledgement

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European Commission

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