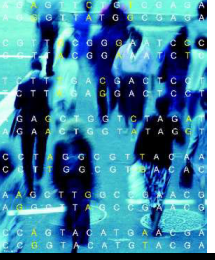

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BioEng 2003 | Fundação Calouste Gulbenkian | 2003.06.26-27

On the requirements of biomedical information tools for health applications: the INFOGENMED case study

Ilídio C. Oliveira[1] (ioliv@ieeta.pt), José Luís Oliveira[1,2], Miguel Santos[3], F. Martin-Sanchez[4], A. Sousa Pereira[1,2]

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Outline

- Motivation
  - Medical and bioinformatics synergy in support of molecular medicine
- INFOGENMED in a nutshell
  - Project goals
- Methods
  - Identification of health application requirements
  - Relevant databases and clinical pathways
- Expected results

A novel life sciences landscape

- New technologies (e.g.: biochips, bioinformatics) and research approaches (e.g.: proteomics, genomics) are revolutionising biomedical research.
- The Human Genome Project is making a unique contribution to the knowledge of the relationships between human genes and physiopathological states.
- A huge wealth of information is being produced
- The integration of genetic information in the clinical environment will give rise to a new clinical practice based on Molecular Medicine and personalised healthcare.

Towards molecular medicine

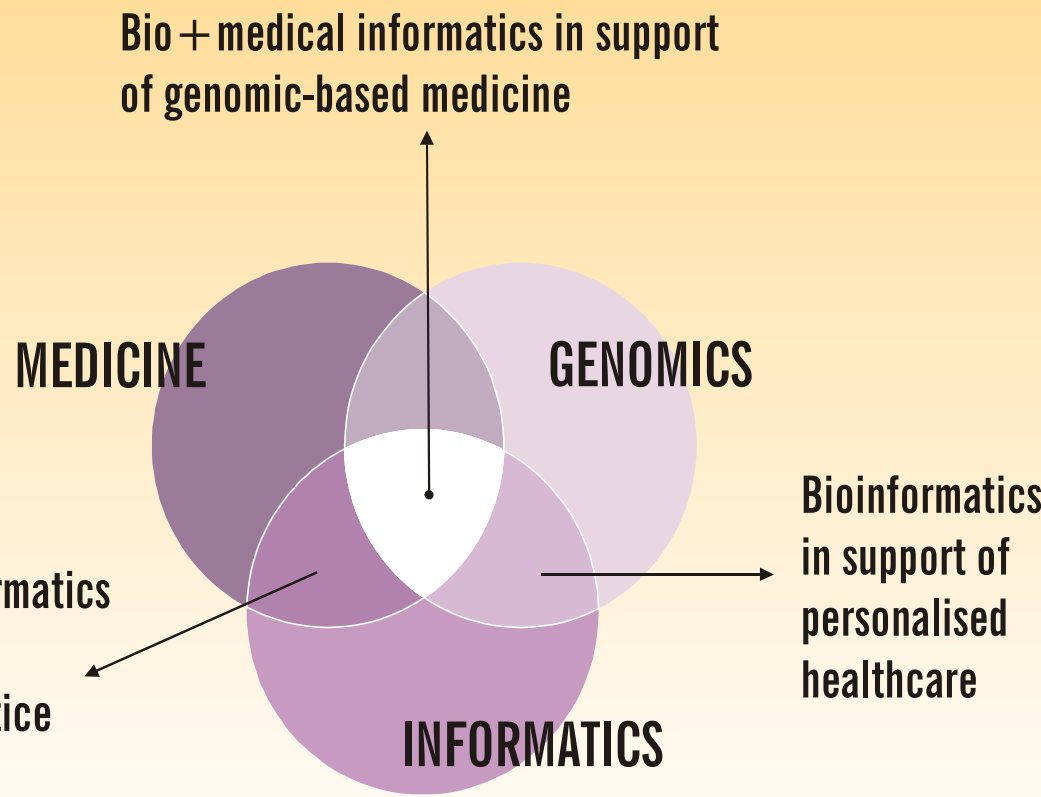
- Genomic medicine is expected to provide:
  - More precise diagnosis, including **genetic tests**;
  - **Personalized drugs** with increased efficiency with minimal inconveniences;
  - Therapy methods acting directly on the genetic code (**gene therapy**).
- The generalization of molecular medicine requires an increased exchange of knowledge between clinical and biological domains

Barriers to knowledge transfer

- Different sources of relevant information are spread (over the Internet)
- Wide range data of formats
- Codification and terminology is not unified, quality is difficult to discern
- Medical coding systems not ready for managing genetic information
- Bioinformatics tools designed for basic research
- Lack of guidance for the physician

The quest for biomedical information tools

- A new bred of information tools, integrated in the clinical workflows, is needed to search, retrieve and relate genetic and clinical data.



INFOGENMED goals

- Unification of heterogeneous biomedical databases under one virtual system:
  - 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 concepts;
  - design of a user-friendly interface and development of “clinical pathways” to help users in resorting to the system for the benefit of their practice.
- Validation of the integrated system in the field of rare genetic diseases.

The consortium

Information Society

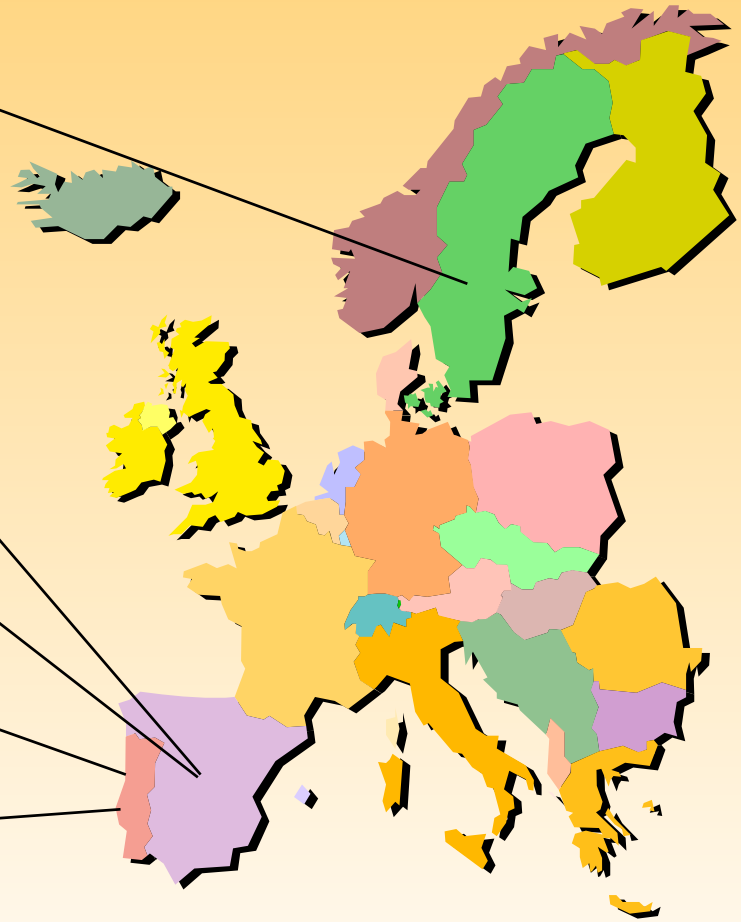
Linkoping Universitet - Sweeden

Universidad Politecnica de Madrid (UPM) - Spain

Instituto de Salud Carlos III (ISCIII) - Spain

IEETA / Univ. Aveiro - Portugal

Genomica / STAB Vida – Portugal



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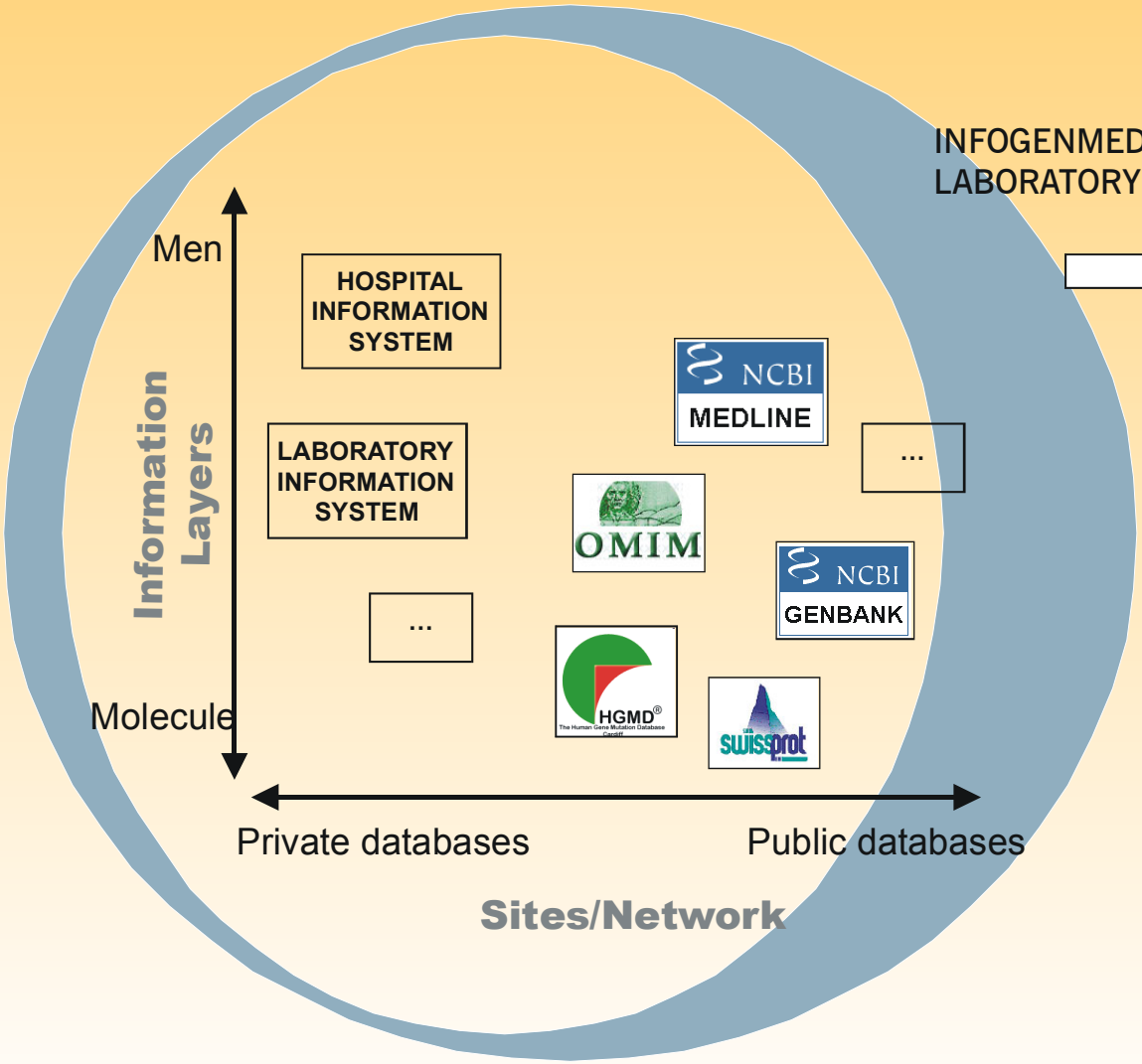
Key competences (roles in the project)

Key competences	Medical Informatics / health telematics	Bioinfor-matics	Distributed data integration	Biomedical market	Clinical app.s (rare genetic deseases)
IEETA / Univ. Aveiro - Portugal	●		●		
Universidad Politecnica de Madrid (UPM) - Spain	●		●		
Instituto de Salud Carlos III (ISCIII) - Spain		●			●
Linkoping Universitet - Sweeden	●	●			
Genomica / STAB Vida – Portugal		●		●	

INFOGENMED approach



INFOGENMED VIRTUAL LABORATORY

Unified querying and information browsing regarding:

- Patient data and conditions
- Genome and sequences
- Protein sequence and structure
- Mutations
- Genetic diseases
- Genetic tests
- Terminology and coding
- Patient counselling
- ...

Health applications' requirements

Requirements analysis:

- Experts Panel interviews
  - 10 experts with complementary backgrounds
- User survey questionnaire
  - 30 replies from professionals in biomedical fields
  - http://www.infogenmed.net/survey

Biologist/Biochemist	10
Medical researcher	9
Medical doctor (practitioner)	3
Bioinformatician	2
Computer scientist	2
Other (Pharmacist, etc.)	4
Total	30

Experts Panel – key inputs

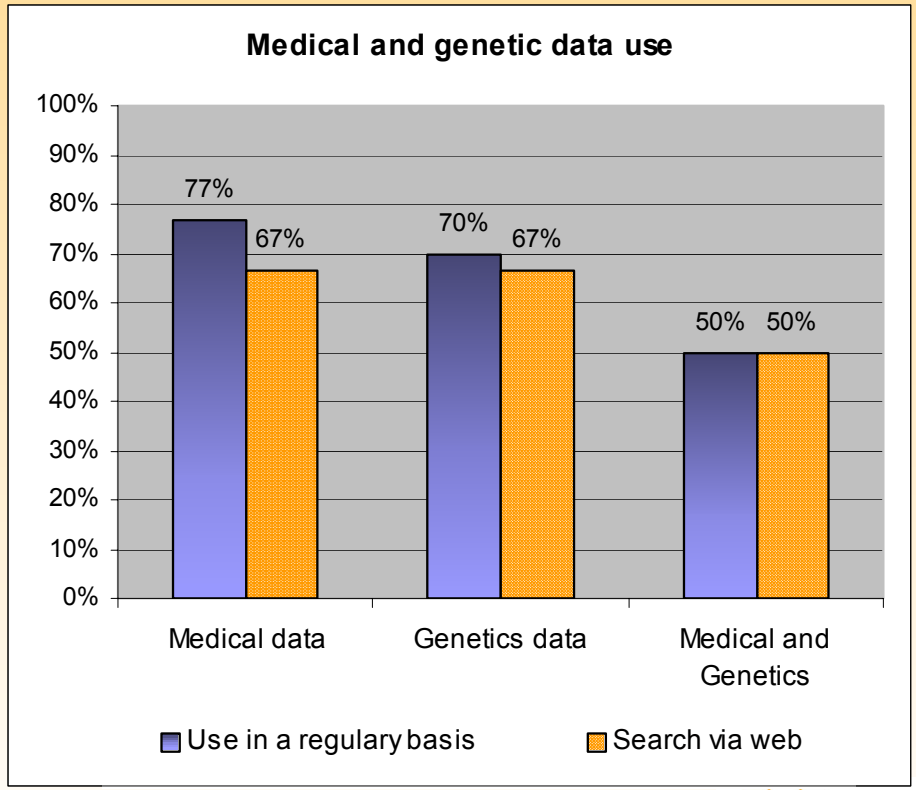
- Too much time is wasted in searches and databases hopping. Information hubs are a definite plus.
- Nomenclature mismatches hinder the seamless use of current biomedical information sources.
  - Across different domains of knowledge (e.g.: clinical and biological)
  - Across databases with semantic related contents (e.g.: no standard nor complete ontology do describe the existing biological data).
- Tools should help users to navigate information at different levels
  - user-friendly interfaces
  - summary and detailed views of data as appropriate

Experts Panel – key inputs 2

- Support for collaboration between experts can be a plus
- Counselling will be important to approach genetics-related issues with patients
- Genetic testing is expected to become a generalized practice in the near future
  - information tools must be prepared

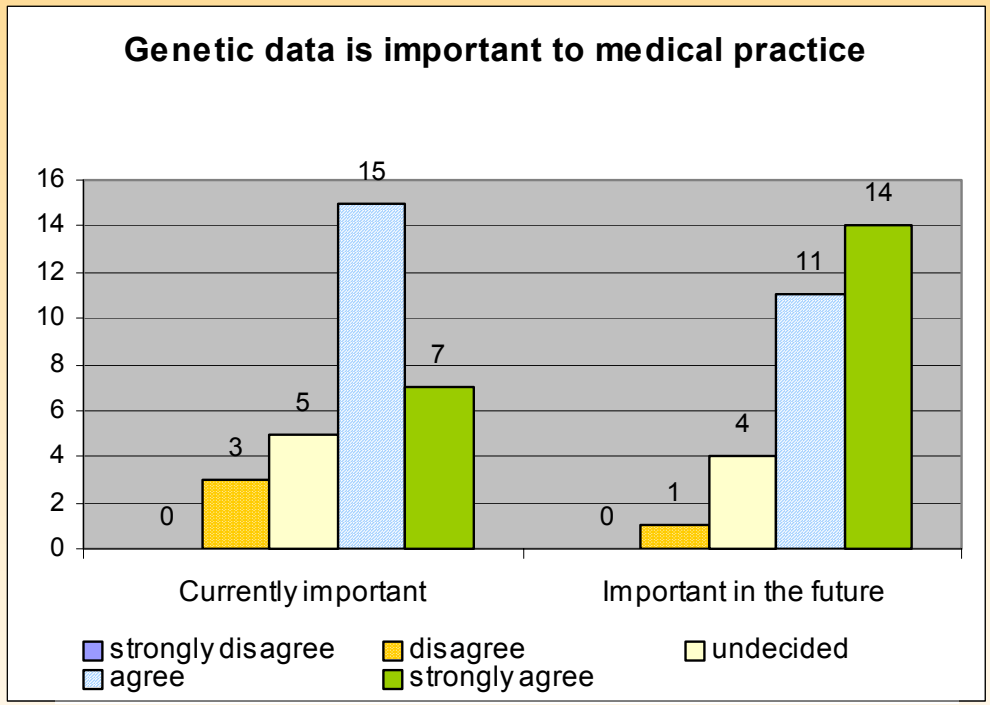
Users survey – key inputs

- 77% search for medical data in a regularly basis
- Genetic data is sought by 70% of respondents
- Note that half of the respondents use both kinds of data regularly
- Almost all respondents that actively seek for biomedical information use the Internet.



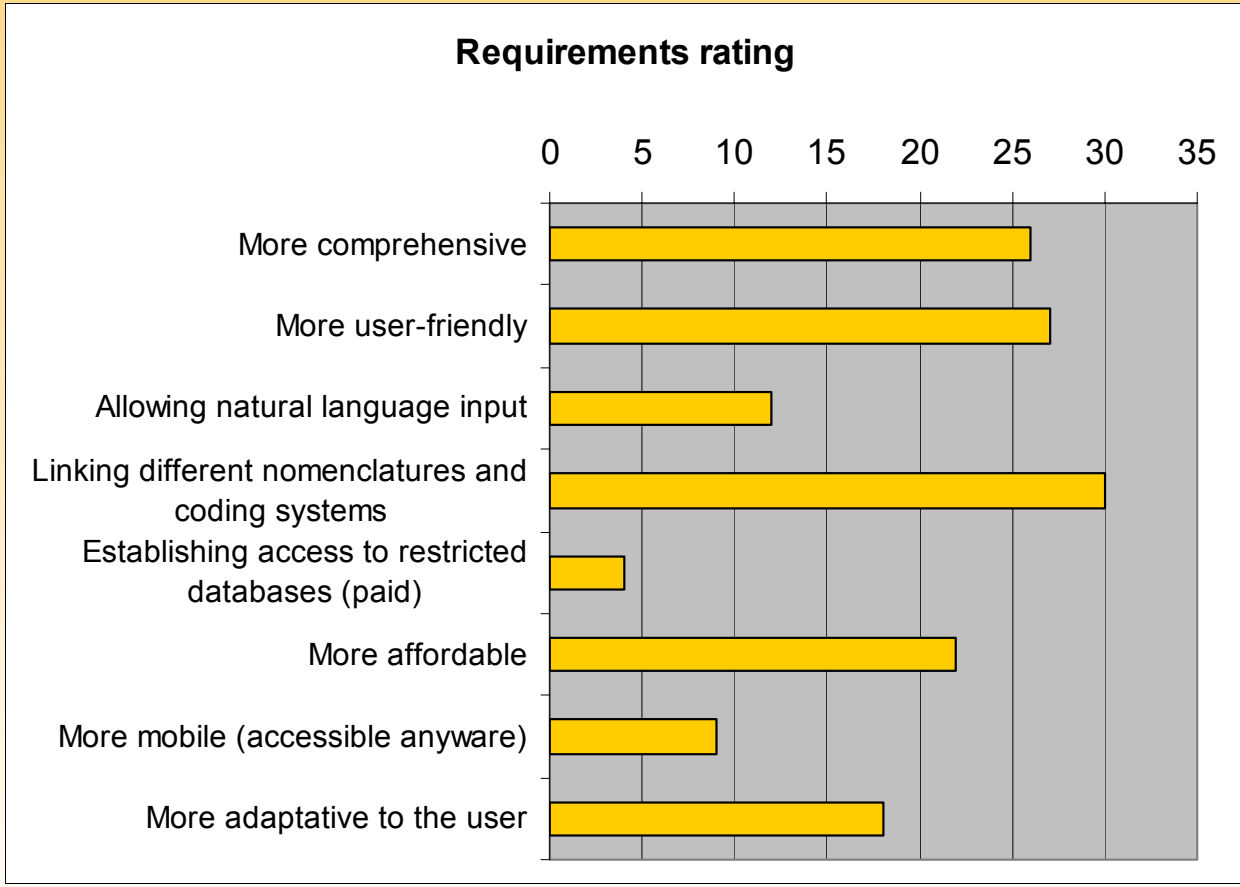
Users survey – key inputs 2

- Currently, there's a real contribution but, still, somewhat moderate
- However, concerning the future role of genetics in medicine, there is a strong conviction that it will play a decisive role.



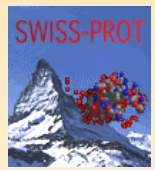
Users survey – key inputs 3

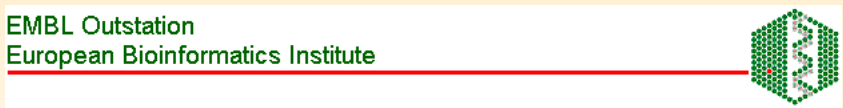
- The ability to link different vocabularies and coding systems rates as the top requirement
- The need for tools that are easy to use was is also stressed.
- Tools available for free is a trend that seams to gain importance among the biomedical community



Sources of Genetic Information

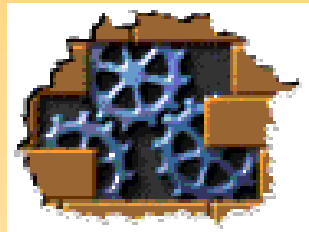
- Genome and sequence databases
  - EMBL
- Protein sequence and structures
  - PDB, SwissProt
- Mutations
  - Central variation databases – HGMD
  - Single Locus Databases
- Genetic diseases
  - OMIM, GeneCards, GeneReviews
  - Genes and Disease
- Genetic tests
  - Geneclinics, EddNal
- SNPs
  - (dbSNP)

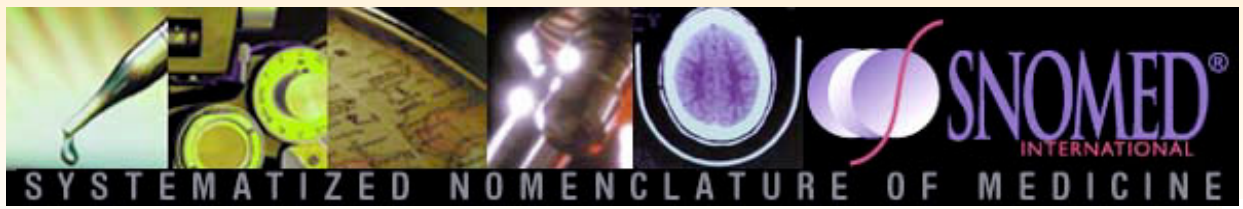




Medical terminology and coding systems

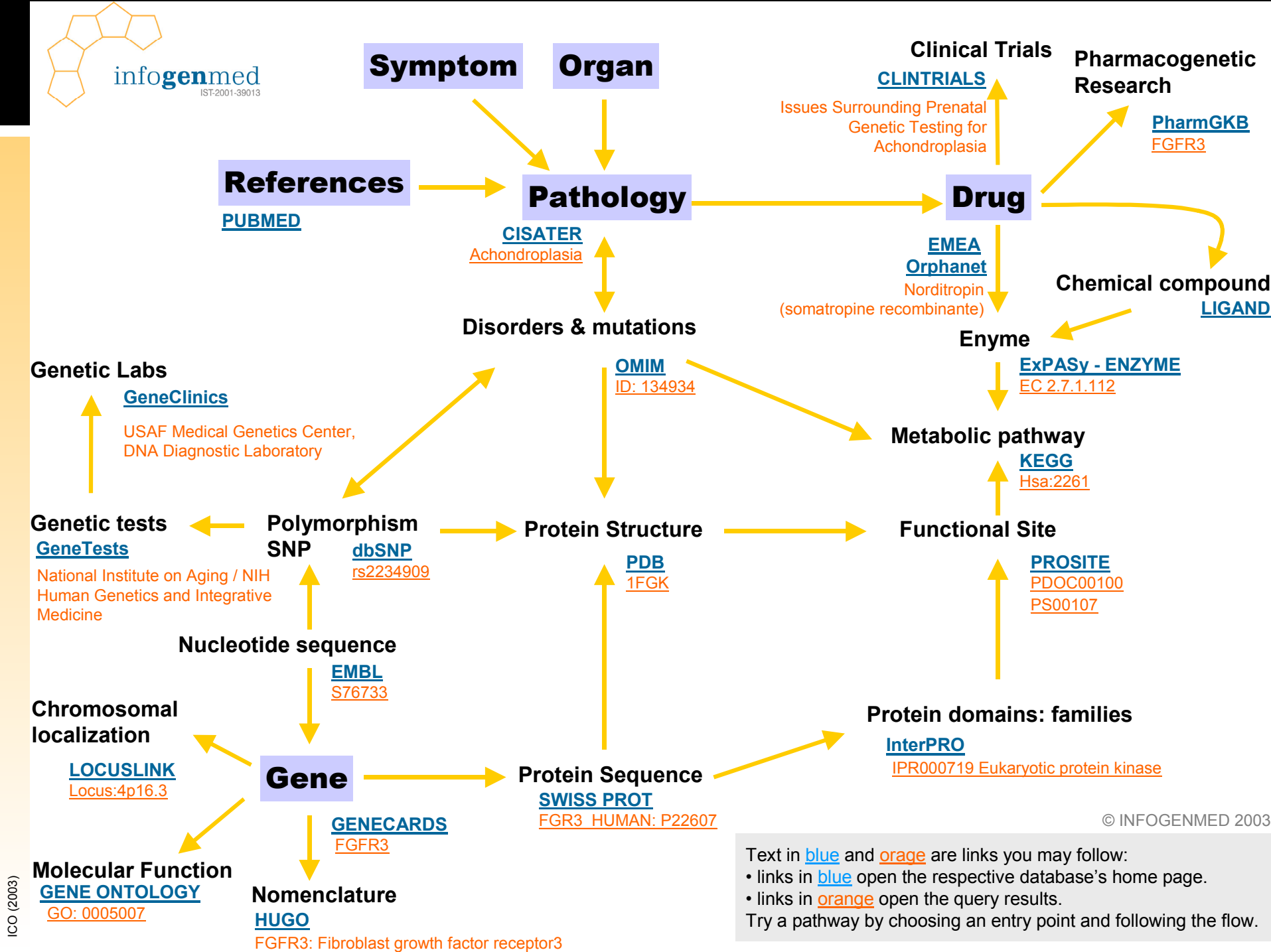
- ICD
- SNOMED
- UMLS
- MeSH
- LOINC
- GALEN





Supporting clinical pathways

- Users start the interaction at a familiar entry point (symptom, organ, drug, pathology), then navigating into deeper levels of detail (disorders & mutations, single nucleotide polymorphisms, etc.), walking through a net of biomedical relevant conceptual links.
- It is the system that autonomously retrieves and relates data for the several concepts, i.e., data sources.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Symptom
Organ
References
PUBMED
Pathology
CISATER
Achondroplasia
Clinical Trials
CLINTRIALS
Issues Surrounding Prenatal Genetic Testing for Achondroplasia
Pharmacogenetic Research
PharmGKB
FGFR3
Drug
EMEA
Orphanet
Norditropin
(somatropine recombinante)
Chemical compound
LIGAND
Disorders & mutations
Enyme
ExPASy - ENZYME
EC 2.7.1.112
Genetic Labs
GeneClinics
USAF Medical Genetics Center, DNA Diagnostic Laboratory
OMIM
ID: 134934
Metabolic pathway
KEGG
Hsa:2261
Genetic tests
GeneTests
National Institute on Aging / NIH Human Genetics and Integrative Medicine
Polymorphism SNP
dbSNP
rs2234909
Protein Structure
PDB
1FGK
Functional Site
PROSITE
PDOC00100
PS00107
Nucleotide sequence
EMBL
S76733
Chromosomal localization
LOCUSLINK
Locus:4p16.3
Gene
Protein Sequence
SWISS PROT
FGR3_HUMAN: P22607
Protein domains: families
InterPRO
IPR000719 Eukaryotic protein kinase
GENECARDS
FGFR3
Molecular Function
GENE ONTOLOGY
GO: 0005007
Nomenclature
HUGO
FGFR3: Fibroblast growth factor receptor3
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Text in blue and orage are links you may follow:
• links in blue open the respective database's home page.
• links in orange open the query results.
Try a pathway by choosing an entry point and following the flow.
ICO (2003)

Expected achievements

- Methods and tools for the integration of clinical and genetic data from heterogeneous remote databases
- A vocabulary server to combine existing terminology systems in Medicine and Genetics
- Novel framework for clinicians to locate, search, access, retrieve and use genomic information in patient care

Summing up

- Medical and bioinformatics need to cooperate to build tools able to access and relate clinical and genetic data
- INFOGENMED aims to facilitate the seamless integration of medical and genetic information, through an easy to use, Web-based environment, tailored to the health professionals.
- A measure of the success of the system will be it ability to support clinical pathways, autonomously retrieving and linking semantic related concepts.

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