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Content

- Background
- Objectives
- Methodology
- Outputs

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Background

- New technologies (Biochips, bioinformatics) and research approaches (Proteomics, Genomics,) are revolutionising biomedical research.

- The Human Genome Project is making a main contribution to the knowledge of the relationships between human genes and physiopathological states.

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Background

- The integration of these massive amounts of genetic information in the clinical environment will give rise to a new clinical practice based on Molecular Medicine and personalised healthcare.

- Diagnosis will be more precise and include genetic testing that could be done using biochip technology at the point of care and therapy methods will include personalized drugs.

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Barriers

- Different sources of relevant information are spread over the Internet
- Wide range of formats difficulting data interchange
- Codification and terminology is not unified, quality is difficult to discern

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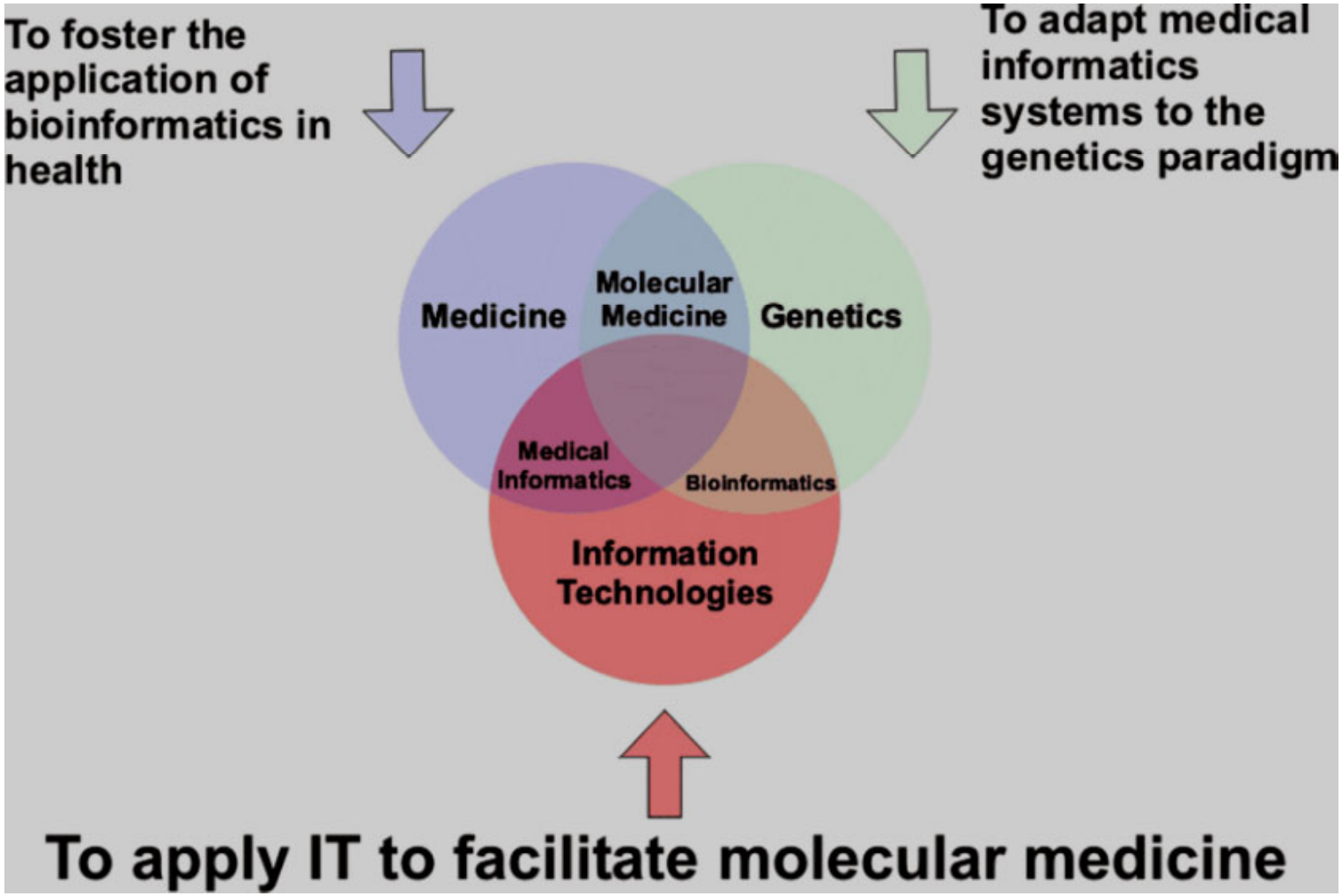
Barriers

- Medical coding systems not ready for managing genetic information
- Bioinformatic tools designed for researchers
- Lack of guidance for the physician


To foster the application of bioinformatics in health
To adapt medical informatics systems to the genetics paradigm
Medicine
Molecular Medicine
Genetics
Medical Informatics
Bioinformatics
Information Technologies
To apply IT to facilitate molecular medicine

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Objectives

1. Determination of the needs of genetic and medical information in various pathologies, considered as “rare genetic diseases”, in health environments.

2. Design of the methods and development of tools for the integration of heterogeneous databases over Internet.

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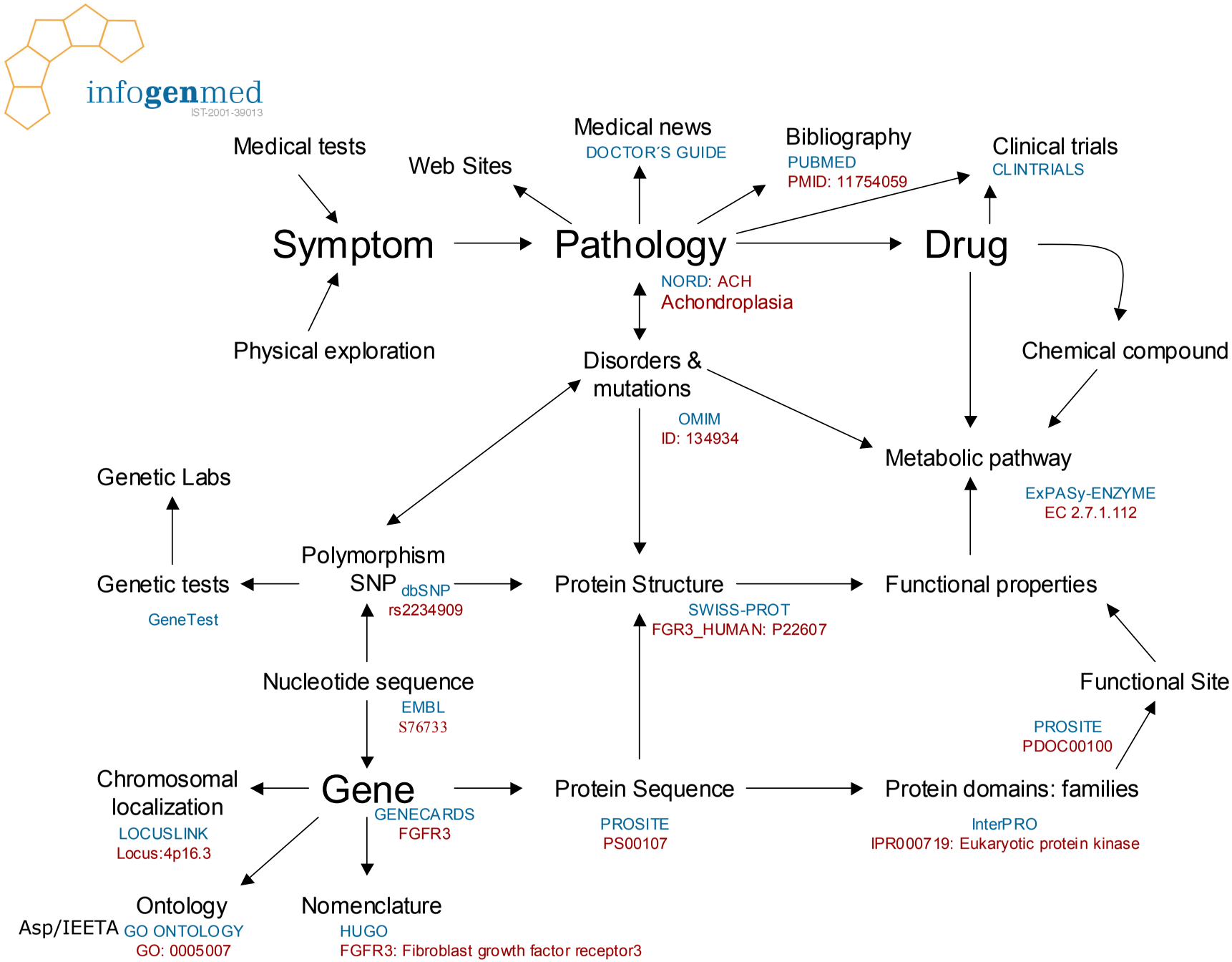
Objectives

3. Design and implementation of an interface to aid users to search, find and retrieve the contents of remote databases, based on a vocabulary server for the integration of medical and genetic terms and concepts.

4. Development of an assistant to help health practitioners to use the designed methods and tools.

5. Integration of the complete system and validation in the area of rare genetic diseases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Medical tests
Web Sites
Medical news
DOCTOR´S GUIDE
Bibliography
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PMID: 11754059
Clinical trials
CLINTRIALS
Symptom
Pathology
Drug
NORD: ACH
Achondroplasia
Physical exploration
Disorders & mutations
Chemical compound
OMIM
ID: 134934
Metabolic pathway
ExPASy-ENZYME
EC 2.7.1.112
Genetic Labs
Polymorphism
SNP
dbSNP
rs2234909
Genetic tests
GeneTest
Protein Structure
SWISS-PROT
FGR3_HUMAN: P22607
Functional properties
Nucleotide sequence
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S76733
Functional Site
PROSITE
PDOC00100
Chromosomal localization
LOCUSLINK
Locus:4p16.3
Gene
GENECARDS
FGFR3
Protein Sequence
PROSITE
PS00107
Protein domains: families
InterPRO
IPR000719: Eukaryotic protein kinase
Ontology
Asp/IEETA
GO ONTOLOGY
GO: 0005007
Nomenclature
HUGO
FGFR3: Fibroblast growth factor receptor3

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Methodology

- Databases
  - Clinical (patient information) PHRs
  - Medical (disease information) (Rare diseases)
  - Genetics (dbSNP)
  - Hybrids (OMIM)
- Panel of users

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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)

GeneCards™

HGMD™ The Human Gene Mutation Database Cardiff

GENETests

PDB™ PROTEIN DATA BANK

SWISS-PROT

EMBL Outstation
European Bioinformatics Institute

OMIM
Online Mendelian Inheritance in Man
Johns Hopkins University

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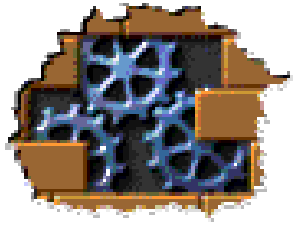
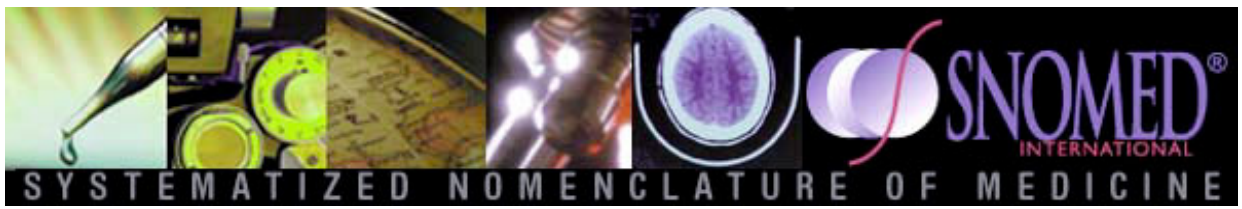
Medical terminology and coding systems



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

UMLS Unified Medical Language System

MEDICAL SUBJECT HEADINGS MeSH



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Technical issues

- WEB-based user interface
- Internet/intranet, TCP/IP protocol
- JAVA compliant and CORBA middleware
- Open and distributed system based on reusable components
- Object-oriented design and implementation
- Adaptable to PC, MAC and UNIX user workstations
- State-of-the-art security guidelines

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Expected achievements

- Integration of clinical and genetic info from heterogeneous remote databases
- Improving existing methods:
  - Medical/Genetics virtual databases
  - Data warehouse
  - Distributed DBs
- A vocabulary server that aims 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

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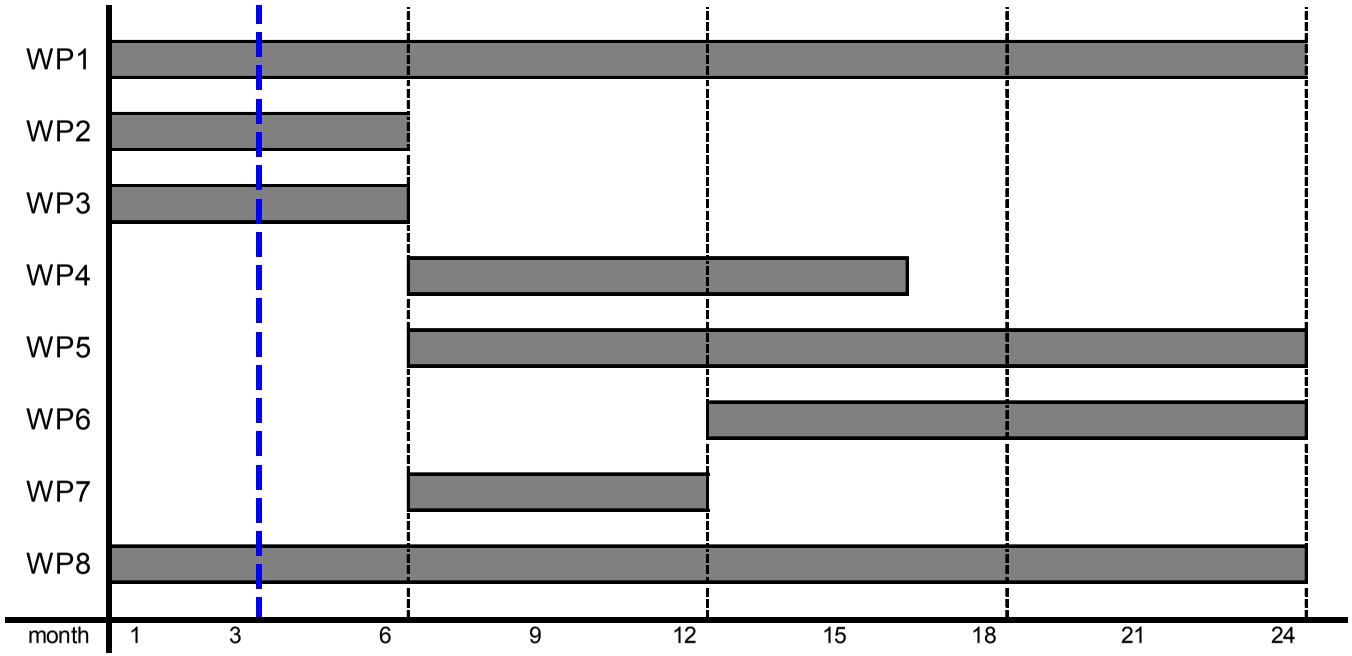
Current status

Project Timetable



WP1 - Project Management and Coordination
WP2 - Analysis of the state of the art
WP3 - Functional analysis and user requirements
WP4 - Database creation and validation
WP5 - Design and implementation of the informatic tools
WP6 - Clinical testing and evaluation
WP7 - Analysis of confidentiality and security issues
WP8 - Dissemination and exploitation plan

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WP3 Functional analysis and user requirements

- Elaboration of structured questionaire to help user requirement discussion with clinicians
- The questionaire incorporates a Vision document and draft clinical protocol
- Survey on clinicians expectations based on questionaire
- If you are interested in colaborating please visit project's website: **www.infogenmed.net**

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Project information

- The project started in September 2002 and will last 24 monthes

- Funded by the EU under the Framework Programme 5 - *Information Society Technologies (IST) Programme*

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Consortium

- Universidade de Aveiro – IEETA (Coordinator)
- UPM - Universidad Politecnica de Madrid
- ISCIII – Instituto de Salud Carlos III
- Linköping University
- STAB Vida – GENOMICA

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