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EMILIA - ROMAGNA
Istituto Ortopedico Rizzoli di Bologna
Istituto di Ricovero e Cura a Carattere Scientifico

NETTAB

GePhCARD & BioMIMS:

a combined platform that support research on hereditary diseases

October 14th – NETTAB 2011

Marina Mordenti – Rizzoli Orthopaedic Institute

Difficulty & delay in diagnosis

No data exchange

- Partial data gathering
- No data integration
- Reduced data merging
- Few information

No statistical analyses

Inadequate treatments

Increase knowledge on Hereditary Diseases

- collect clinical and genealogical data of each patient /family

- increase molecular screening on blood/tissue samples

Our focus is to define a correlation between clinical data (Phenotype) and genetic screening (Genotype)

Short overview on Hereditary Rare Diseases

- less than one in 2000
- 25 million people are affected by them
- 7000 diseases are rare
- Most involve skeleton
- Mostly are not curable, chronic, life-threatening

Multiple Osteochondromas - MO

- cartilaginous caps on long bones
- huge inter/intra-familiar clinical variability (3 class each divided in 2 sub-class)
- in less than 5% of the patients a progression into a SPC
- Mutations on EXT1/EXT2 genes
- Mutated proteins for bone growth

Osteogenesis Imperfecta - OI

- heterogeneous disorder
- susceptibility to fracture, bone fragility
- 4 clinical types, expanded into 7
- caused by mutations in COL1A1 or COL1A2 genes
- mutated chains of type I collagen, structural protein of bone

IT FOR SUPPORTING REASERCH IN HRD

- Store genomic data
- Store clinical data
- Create a data model to integrate clinical and genomic data in a standard way to allow heterogeneous application interoperability
- Correlate genomic data to clinical data in a patient centric view

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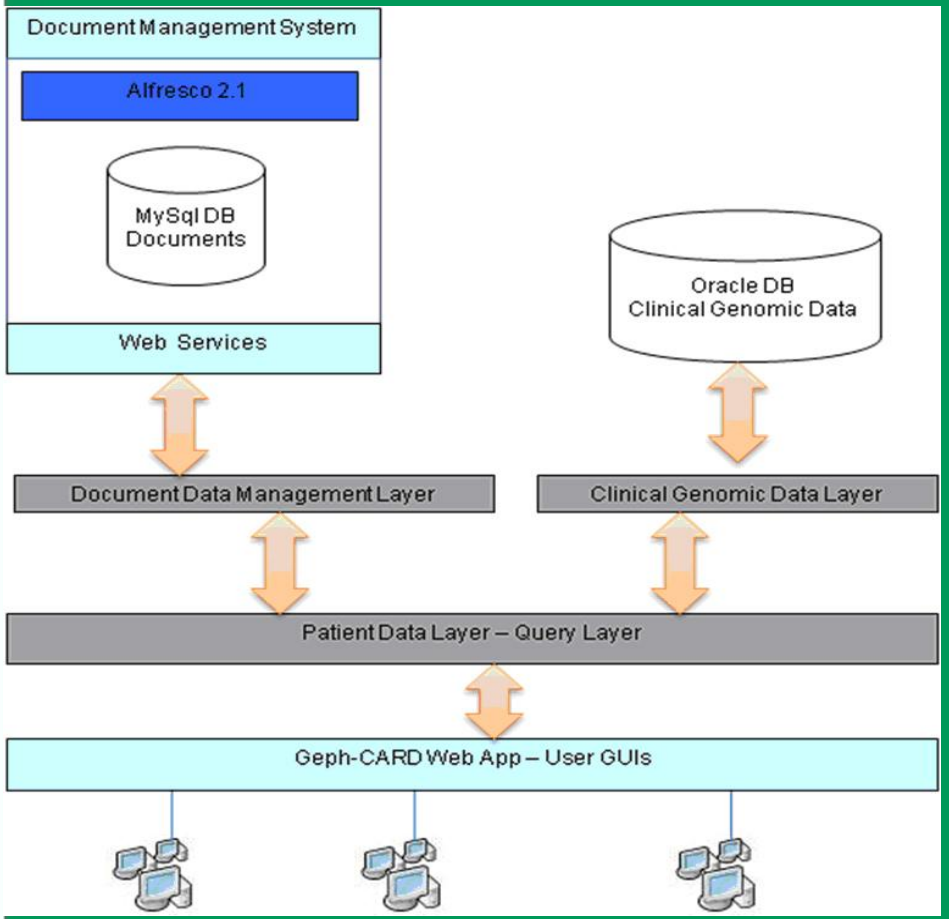
GePhCARD: IT PLATFORM FOR COLLECTION

designed as services (Web Services) and developed according to SOA principles



a relational database to store clinical, genomic and genealogic data of patients

a relational database to store and index digital documents

a document management system based on Alfresco 2.1 framework

a web application

GePhCARD: IT PLATFORM FOR COLLECTION

- **GENEALOGICAL DATA DOMAIN**

To store general information on each family and to guarantee the possibility to compare clinical and genomic data inside the same family

- **PERSONAL DATA DOMAIN & PATIENT PANEL**

To store a complete set of private data for each patient or relative. Some fields are mandatory to identify each patient univocally

GePhCARD: IT PLATFORM FOR COLLECTION

- **CLINICAL CHART**

2 sections: a left navigation panel structured as a tree with data distributed in sub-sections and a right section created to visualize the sub-section's details

- **DOCUMENTAL DATA DOMAIN**

an existing professional open source CMS Alfresco for storing document and a full index based searching system to perform both full text and metadata searches way

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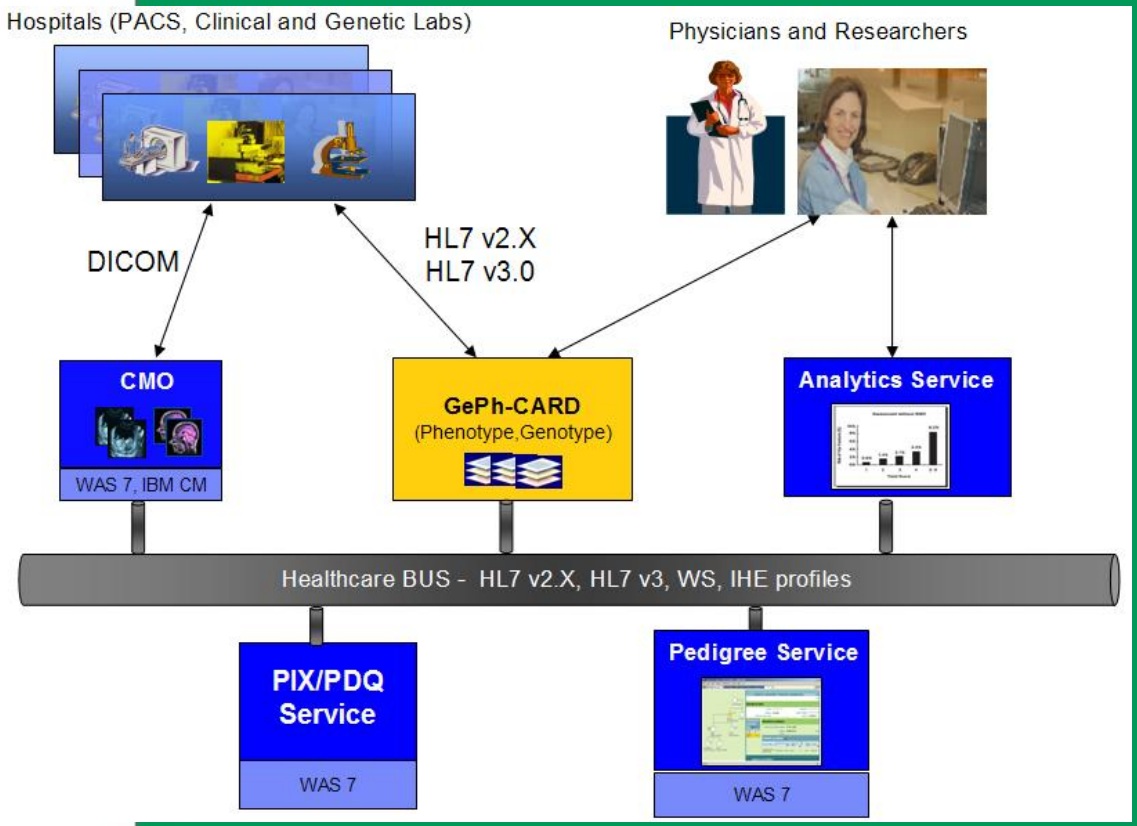
BIOMIMS: IT PLATFORM FOR COLLECTION



relational DB for archiving clinical and genetic data

a Light MPI Server (Master Patient Index) for interoperability

a Content Manager for storage of clinical and genetic raw data

an innovative tool for pedigree analysis and clustering

a Web based UI interface
a Medical Imaging Repository (CMO) (secure DICOM based communication

BIOMIMS: IT PLATFORM FOR COLLECTION

DICOM COMUNICATION

To collect and integrate medical images (upload and retrieve from the appropriate system service in DICOM format)

MASTER PATIENT INDEX

To ensure the correct identification of patients and their data in a standard manner

BIOMIMS: IT PLATFORM FOR COLLECTION

PATIENT IDENTIFIER

IHE patient identifier Cross-Reference (PIX) and Patient Demographic Query (PDQ) transactions. To enable interoperability and cross-institutional information sharing (preserving security and privacy)

PEDIGREE ANALYTICS

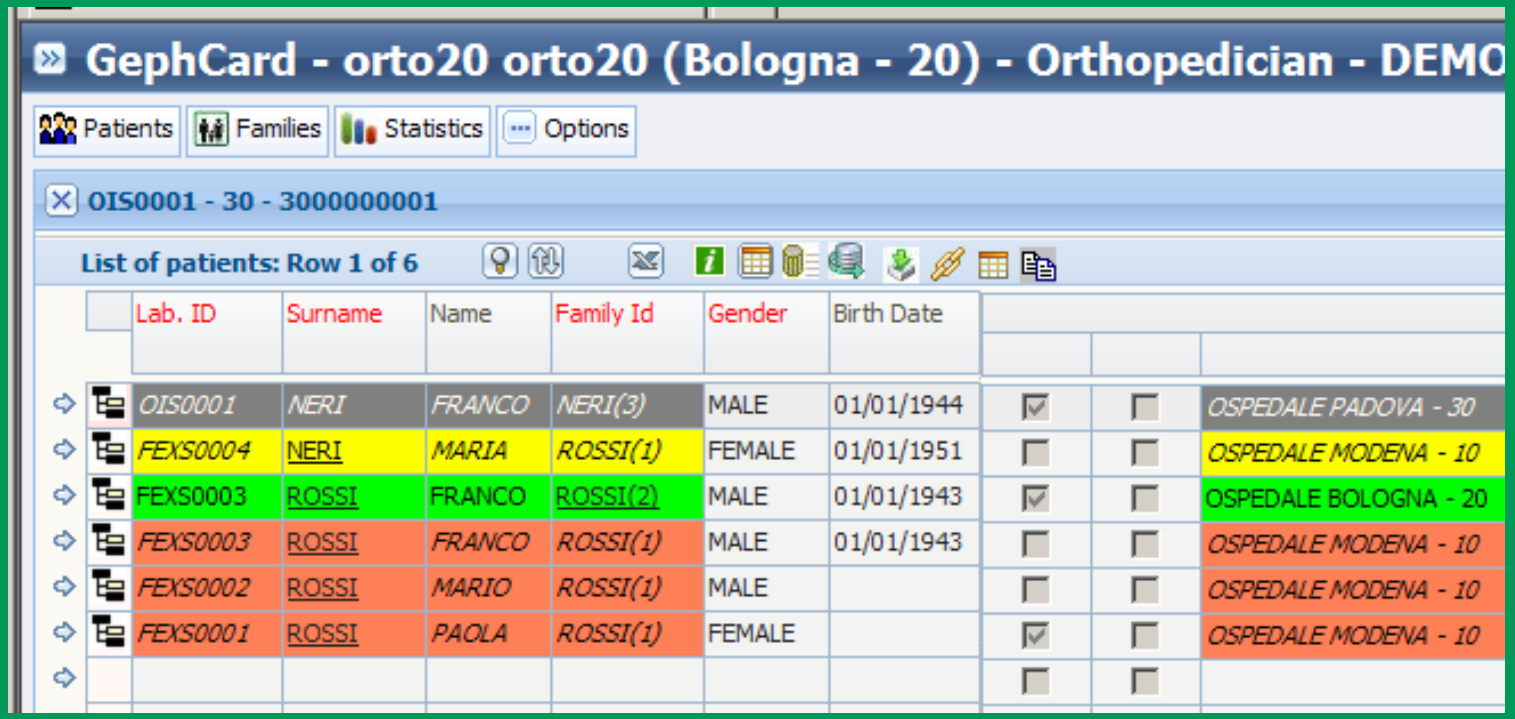
to manage genealogic trees for an healthcare related pedigree creation, management and analysis

GePhCARD & BioMIMS

They work in concert to:

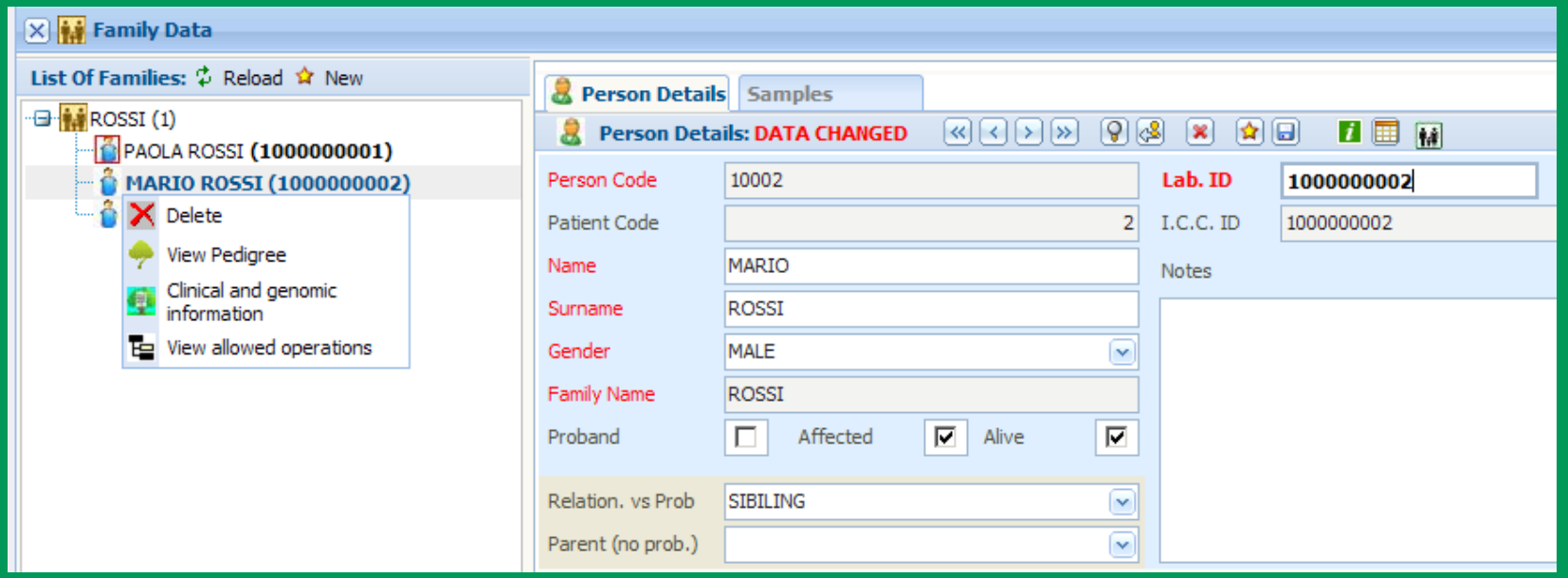
- collect data
- support a set of sophisticated and federated queries (include a combination of different types of information)
- store interesting queries
- extrapolate data
- analyse data

PATIENT DATA ACCESSIBILITY



Lab. ID	Surname	Name	Family Id	Gender	Birth Date			
OIS0001	NERI	FRANCO	NERI(3)	MALE	01/01/1944	☑	☐	OSPEDALE PADOVA - 30
FEXS0004	NERI	MARIA	ROSSI(1)	FEMALE	01/01/1951	☐	☐	OSPEDALE MODENA - 10
FEXS0003	ROSSI	FRANCO	ROSSI(2)	MALE	01/01/1943	☑	☐	OSPEDALE BOLOGNA - 20
FEXS0003	ROSSI	FRANCO	ROSSI(1)	MALE	01/01/1943	☐	☐	OSPEDALE MODENA - 10
FEXS0002	ROSSI	MARIO	ROSSI(1)	MALE		☐	☐	OSPEDALE MODENA - 10
FEXS0001	ROSSI	PAOLA	ROSSI(1)	FEMALE		☑	☐	OSPEDALE MODENA - 10
						☐	☐	

PATIENT'S FAMILY INTERFACE



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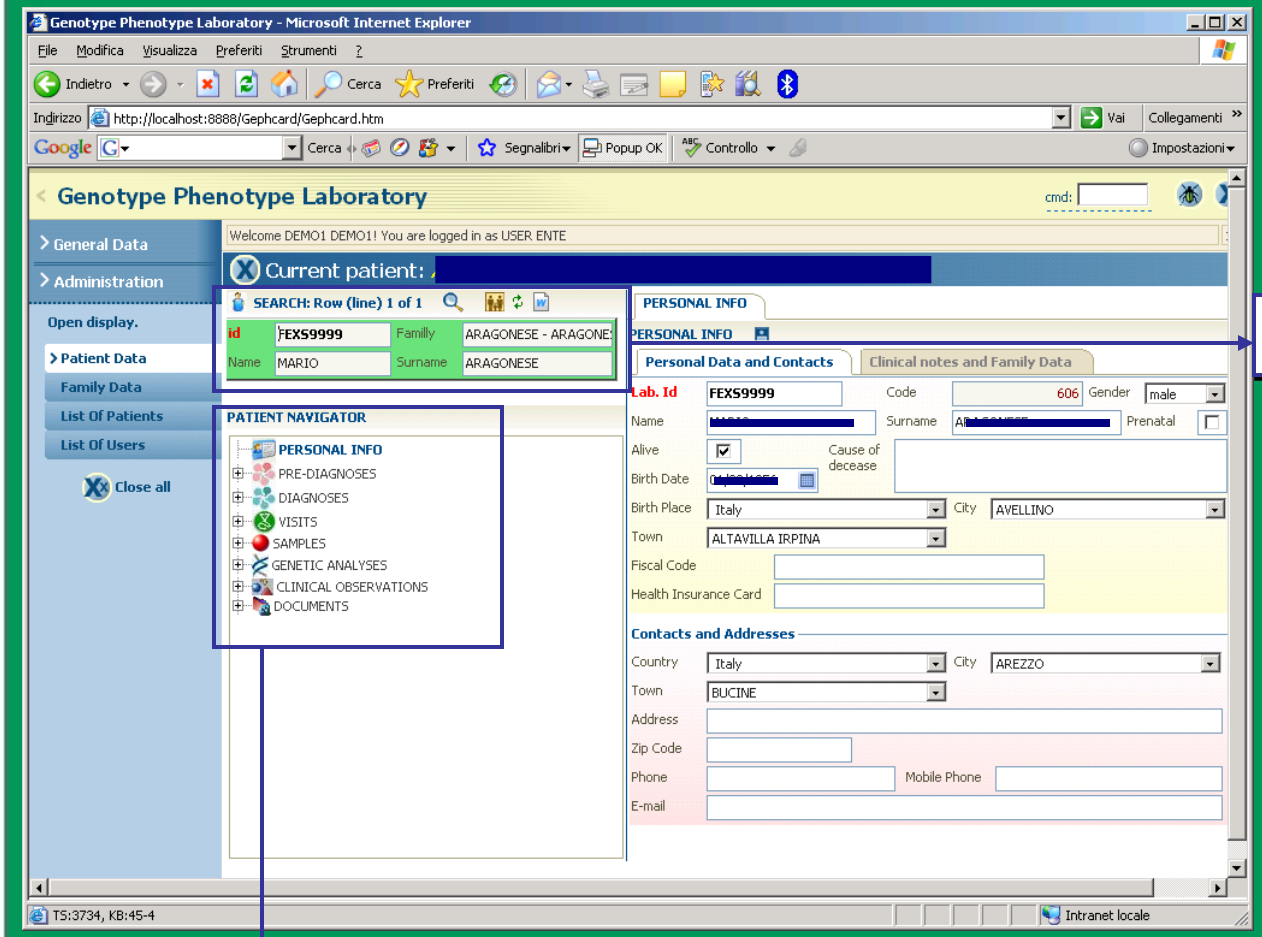
PATIENT PANEL



patient search panel

patient navigator tree

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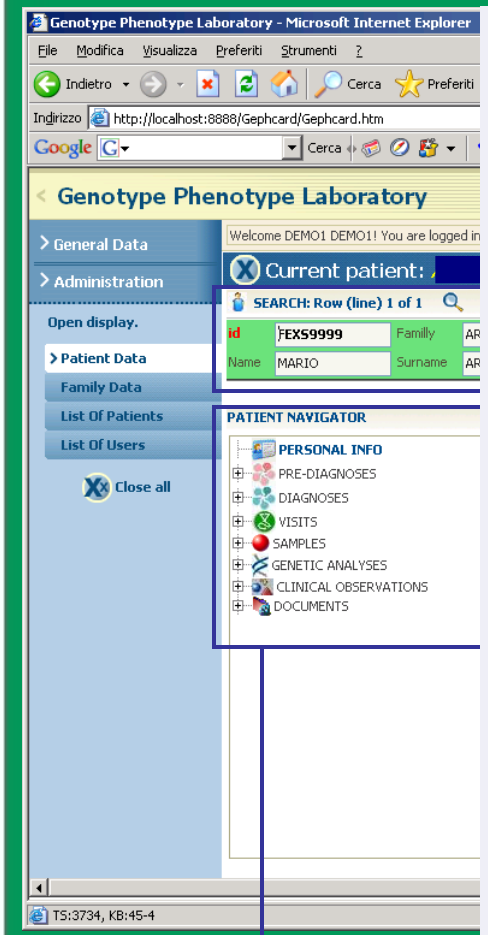
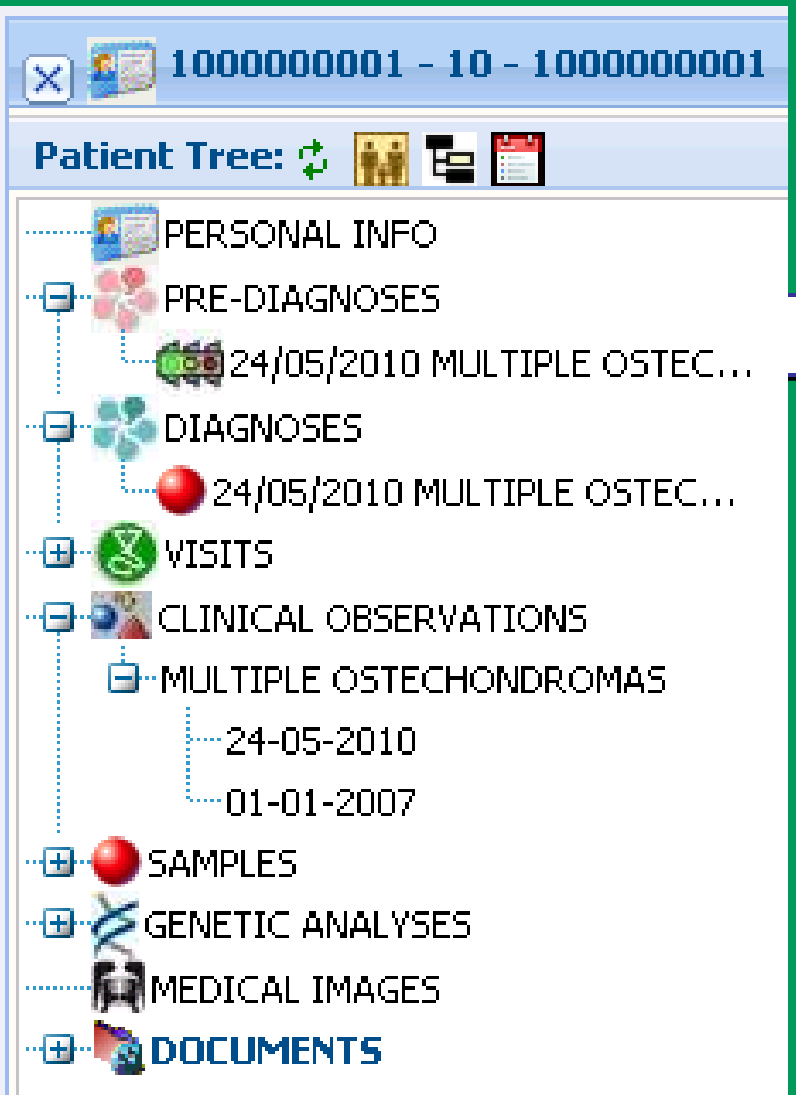
PATIENT PANEL





patient search panel

patient
t

PERSONAL DATA DOMAIN

Genotype Phenotype Laboratory - Patient Pop Up -- Finestra di dialogo pagina Web

Patient Pop Up

Patient: The changes of the data have been registered on the database.

Patient: Revised data.

Identification Info

Laboratory Id	FEXS9999	Name	Mario	Surname	[redacted]
Patient Code		Prenatal ☐	Alive ☑	Cause of Decease	
Gender	male	Birth Date	[redacted]		
Birth Place	Italy	City	AVELLINO	Town	ALTAVILLA IRPINA
Fiscal Code		Health Insurance Card			

Contacts and Addresses

Country	Italy	City Of Residence	AREZZO	Town	BUCINE
Address				Zip Code	
Phone		Mobile Phone		E-mail	

Some clinical Notes

Institutional Clinical Chart Id	ZQ465546729
Other Diseases	
Notes	

Family Data

Proband	☑	Family Name	[redacted]

Personal data

Family data

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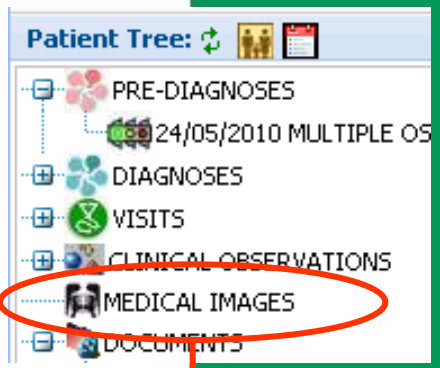
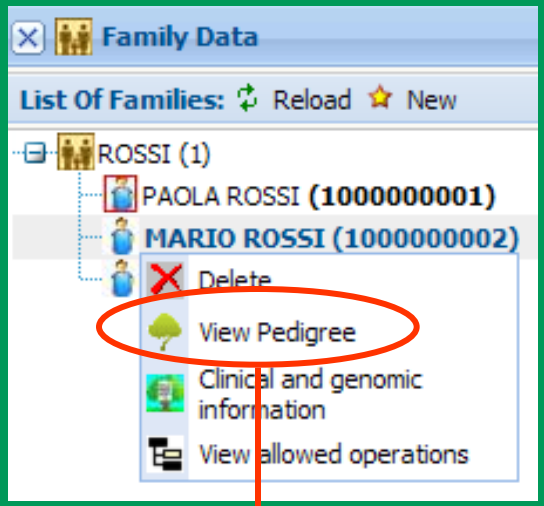
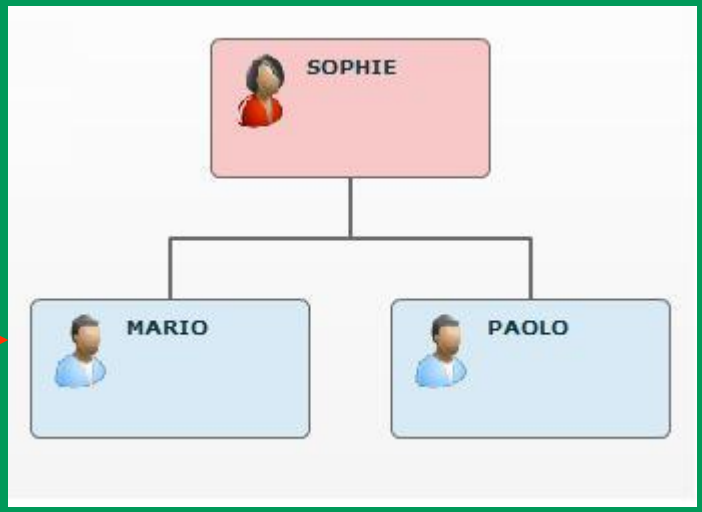
DICOM IMAGES



PEDIGREE TOOL





OI CLINICL DATA

ALL | DETAILS (as OI)

DETAILED OBSERVATIONS: Row 1 of 1

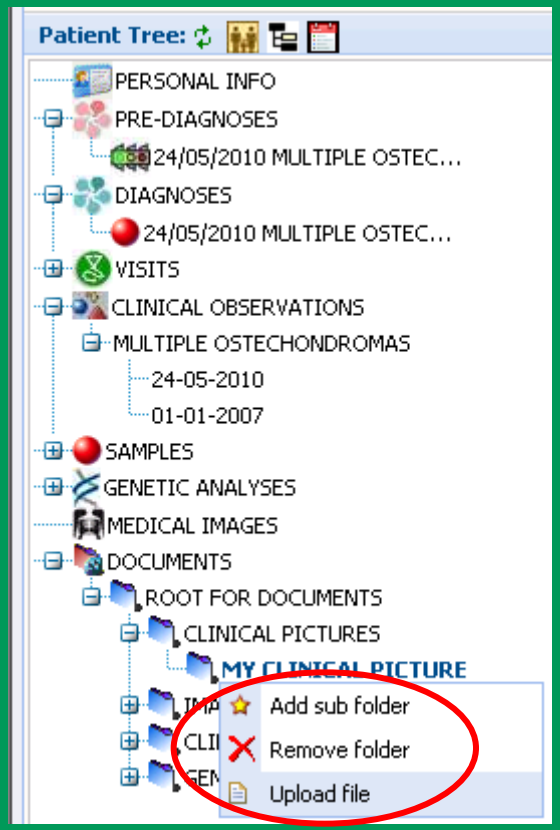
Date	23/11/2009	Disease	OSTEOGENESIS IMPERFECTA	N° of fractures	
Deafness	YES	Details		Sclera	
Dentin. Imper.		Details			
Cutis Laxa		Skin		R. C. Def.	
Card. Lesions		Details		Rhizomely	
Worm. Bones		Sp. Def.	KYPHOSIS	Frn. Boss.	
Triang. Facies		Max. Def.		Jn. Hyperl.	
BD		Z Score		T Score	BMD
Notes	Dall'89 ha avuto fratture multiple in seguito a traumi diretti (anche se di lieve entità)				
Height		Weight		Height Percentile	

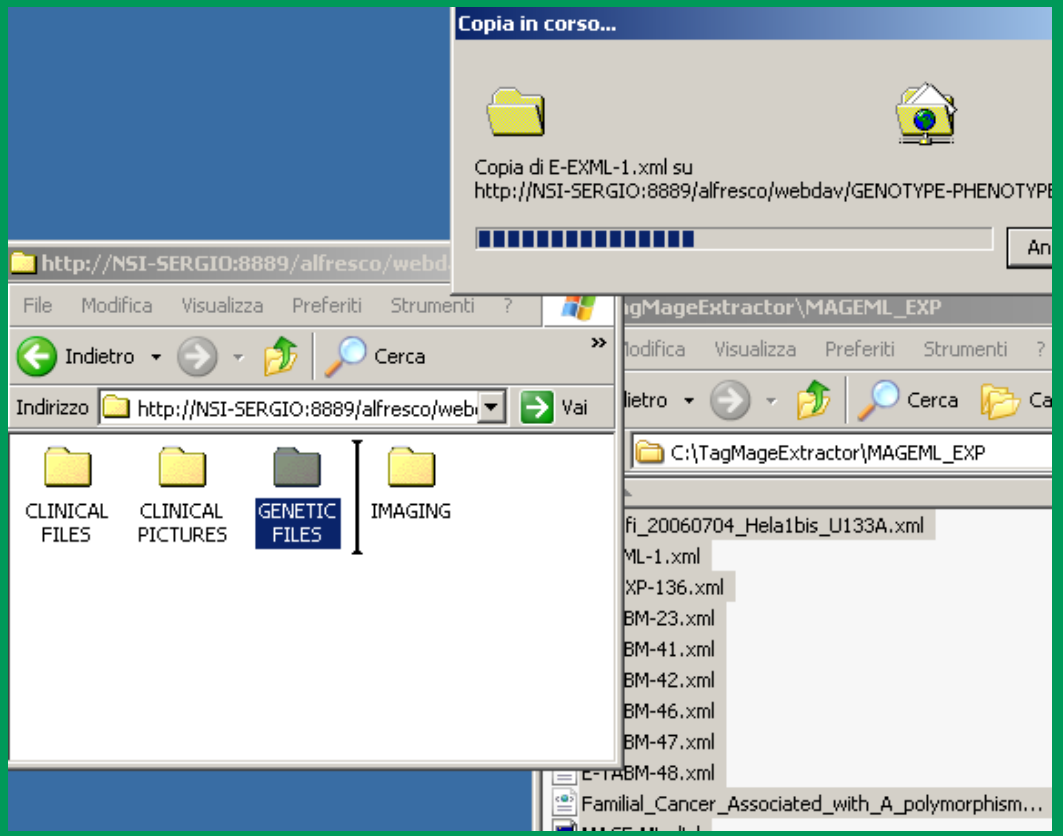
Sites - Locations - Sides | Limitations

Sites - Locations - Sides: Row 1 of 3

Site	N°	Age	Location	Side	N° fractures	Level of pain
RIBS			UNKNOWN	unknown		
HUMERUS			UNKNOWN	unknown		

ALFRESCO


Patient Tree:
PERSONAL INFO
PRE-DIAGNOSES
24/05/2010 MULTIPLE OSTEC...
DIAGNOSES
24/05/2010 MULTIPLE OSTEC...
VISITS
CLINICAL OBSERVATIONS
MULTIPLE OSTECHONDROMAS
24-05-2010
01-01-2007
SAMPLES
GENETIC ANALYSES
MEDICAL IMAGES
DOCUMENTS
ROOT FOR DOCUMENTS
CLINICAL PICTURES
Add sub folder
Remove folder
Upload file


Copia in corso...
Copia di E-EXML-1.xml su
http://NSI-SERGIO:8889/alfresco/webdav/GENOTYPE-PHENOTYPE
File Modifica Visualizza Preferiti Strumenti
Indietro
Cerca
Indirizzo
http://NSI-SERGIO:8889/alfresco/webd
Vai
CLINICAL FILES
CLINICAL PICTURES
GENETIC FILES
IMAGING
C:\TagMageExtractor\MAGEML_EXP
fi_20060704_Hela1bis_U133A.xml
ML-1.xml
XP-136.xml
BM-23.xml
BM-41.xml
BM-42.xml
BM-46.xml
BM-47.xml
BM-48.xml
Familial_Cancer_Associated_with_A_polymorphism...

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RARE HEREDITARY DISEASES

Lack of data for meaningful research

Collaboration among centres

IT PLATFORM FOR COLLABORATION

The data accessibility

➔Role Based Access Control (RBAC) system

➔ enables users from different organizations with customized access rights to patients' information according the user profile or role

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GePh-CARD

Genotype-Phenotype Correlation, Analyses, Research Database

IT PLATFORM

to organize and screen genetic, genealogical, and clinical data

genotype-phenotype patient characterization to a personalized healthcare vision

BioMIMS

BioMarker Imaging Management System

IT PLATFORM

to merge information from dispersed hospitals (pedigree, imaging, etc)

clinical and genealogical characterization to a personalized healthcare vision

MO RESULTS

1. Male patients have more severe manifestations than female, from an inter- and an intra-familial point of view

2. EXT1 mutations are associated with a more severe form and correlate to specific clinical manifestations

3. Class III patients usually have low height

4. Negative Familiarity refers to Class III

OIS RESULTS

1. Quantitative genetic defects (Frameshift, Duplication, Initiating methionine, Nonsense, SpliceSite, SpliceVariant) are usual for Class I patients

2. Qualitative genetic defects (In-frame insertion, In-frame deletion, In-frame insertion-deletion, Missense) are usual for Class II

OUTCOMES 1

- More accurate and precise data → A statistical analyses dataset → Better **disease overview** and help in **differential diagnosis**
- Increased patient and family dataset → **Genotype-Phenotype Correlation** & Study on Hereditary
- Patient-Centric & Family-Centric Approach → **Patient's quality of life**

OUTCOMES 2

- Logging tool thorough an authentication system → Multilevel access profile system (different roles - different domains - different datasets) → **Data Legal Protection**
- Web-accessibility (user-friendly interface) → **Input from different locations**
- Service Oriented Architecture (SOA) → Possibility of **future implementations** and incorporations of configurable modules → Pairing of new tecniques & new modules

OUTCOMES 3

- To purpose innovative research directions → To decide the future health-related strategies
- Multi-language engine and multi-organization structure → Increased gathering of data and data merge
- Advanced algorithms → Correlation patterns → Pedigree analytics
- Articulated queries system → Possibility of store queries → Reload interesting results

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THANKS!!!!

Department of
Medical Genetics
&
Skeletal Rare
Diseases

> n s i

IBM