GePh-CARD
Genotype Phenotype Correlation, Analysis and Research Database

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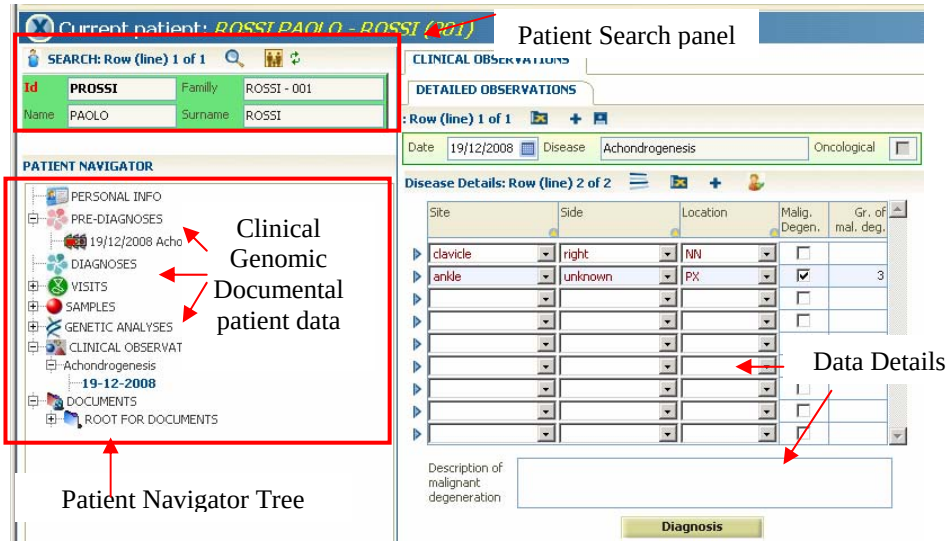
Analysis of inherited diseases and their associated phenotypes is of great importance to gain knowledge of underlying genetic interactions. GePh-CARD is a web application (https://gephcard.nsi-online.it/) based on a patient centric approach that gives the user some powerful tools based on a friendly interface to organize, focalize and screen genetic, genealogical, and clinical data for patients suffering from rare skeletal diseases.
The application has been tested on Hereditary Multiple Exostosis disease, a skeletal disorder characterized by formation of various cartilaginous caps, usually located at the meta-epiphysis of the long bones but can be easily configured to manage other diseases belonging to other clinical domains.



1. Description of the product

GePh-CARD is a multi organization web application software developed to support orthopaedics, clinicians and lab scientists and it is articulated in five main domains:

i. **personal data domain**: to store personal data for each patient or relative.
ii. **clinical data domain**: to store the patient's clinical data depending on the type of disease the patient is suffering from. Pluggable custom modules make it possible to extend the application to any kind of clinical data set for any kind of disease;
iii. **genetic data domain**: to store the patient's genetic data depending on the type of the genetic method used. Pluggable custom modules make it possible to extend the application to any kind of genetic data set for any kind of genetic analysis method;
iv. **genealogical data domain**: to create a pedigree for the patient's family, leading the users to easier understand the relationships between the relatives;
v. **documental data domain**: to manage different kinds of clinical and genetic documents.

Due to simple **clinical**, **genetic** and **documental domain extension mechanisms** the software is highly configurable for different diseases belonging to different clinical domains and for different genetic analysis methods that could become available in the future.

2. Innovative aspects of the product

Due to its highly configurable modules, GePh-CARD makes it possible to easily extend both the clinical domain and the genetic one due to two kinds of extension mechanism:

- metadata extension for existing diseases and existing genetic analysis methods;
- domain extension for both diseases and genetic methods' analysis.

The **multi-language engine** and the **multi-organization structure** make the software suitable for being used by different clinical and research organizations that manage their own patients keeping them separated by patients belonging to different organizations.
A Role Based Access Control system is used to provide different users of different organizations with customized access rights to the different modules of the application.

3. Main advantages of the offer

With its dynamic mechanisms, the application is suitable for being used by different clinical genomic organizations that are involved into studying different kinds of diseases and using different kinds of genetic analysis methods. The integration with an existing professional open source CMS (Content Management System) like Alfresco makes the product a complete solution for storing any kind of document. A full index based searching system based on Alfresco will be available to the final user to perform both full text and metadata searches. As a result GePh-CARD could be considered a complete solution with low costs for further customizations.
In addition, data can be easily wrapped into HL7 v3 XML documents performing clinical and genomic data exchange adopting one of the most important international standard.

4. Technology keywords

GePh-CARD, clinical domain, genomic domain, genotype phenotype correlation

5. Current stage of development

GePh-CARD is a mature product for storing and analysing patients clinical and genomic data to study the existence of a genotype-phenotype correlation.

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