CEMARA

a Web dynamic application within a n-tier Architecture for Rare Diseases

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MIE Göteborg May 28th 2008
Rare Diseases

- Prevalence: less than 5/10,000
  - 246,000 patients in the European Union
- Between 5,000 and 8,000 distinct rare diseases
  - 6 to 8% of EU population may be hit
- Approximately 15 millions European citizens suffer or will suffer one day from a rare disease.
- Most of them are genetic diseases.
Rare Diseases Plan

- 132 Reference Centres labeled by the Ministry of Health
- Objectives:
  - monitoring the epidemiology of rare diseases
  - assessing medical activity
  - setting up the national protocols of care
N-tier architecture

Client Tier

Middle Tier

Information System Tier

Web Interface

Servlets JSPs

Component Container

DataBase Handler

Administrator Tools

Data Warehouse

Production Database

Rare Disease Thesaurus - Geographical dictionary

Java

MySQL

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Interoperability – XML scheme
CEMARA Development plan

1. Collecting data
   - Rare diseases professionals
2. Production database
   - Quality control
3. Reorganizing data
   - Data warehouse
4. Interrogating
   - Clinical Research / Decision-maker
5. Reporting
   - Web-GIS

Patients / Families
CEMARA UML Scheme

Core data set

Petals

Thesauri

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CEMARA core data set...

Identification → Double entry detection algorithm

Diagnosis + Medical Information + Activity = Patient File

Family File
...and the “Petals”

Building a cohort for a given disease or for a group of diseases
Thesauri: a collaborative work

- CEMARA
- Orphanet
- Genatlas

patients

a shared vocabulary

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Results

- 30 centres and 100 sites across the French territory
- 256 physicians among 378 users
- More than 19,000 patients recorded within the first year of implementation
- The 3 most prevalent diseases:
  - Neurofibromatosis type 1
  - Osteogenesis imperfecta
  - Mucoviscidosis
Discussion(1)

- Technical aspects:
  - Scalability: structure & loading
  - Confidentiality charter
Discussion(2)

- **Organizational aspects**
  - professionals’ adhesion
  - accompaniment and support

- **Medical aspects:**
  - 1/3 of patients without an identified diagnosis
  - Define and improve protocol of care
  - Better knowledge of not yet identified diseases
  - First CEMARA epidemiological report
Acknowledgments

Members of the steering committee:
Pr A Verloes, Dr M Le Merrer, Pr C Bodemer, Dr G Baujat, Dr E Bourdon-Lanoy, Dr M Gerard-Blanluet, Dr R Salomon

Coordinators, and professionals

Institutional cooperation and fundings: