CEMARA

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

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for the CEMARA steering committee

eHealth beyond the horizon - get IT there

MIE Göteborg May 28th 2008

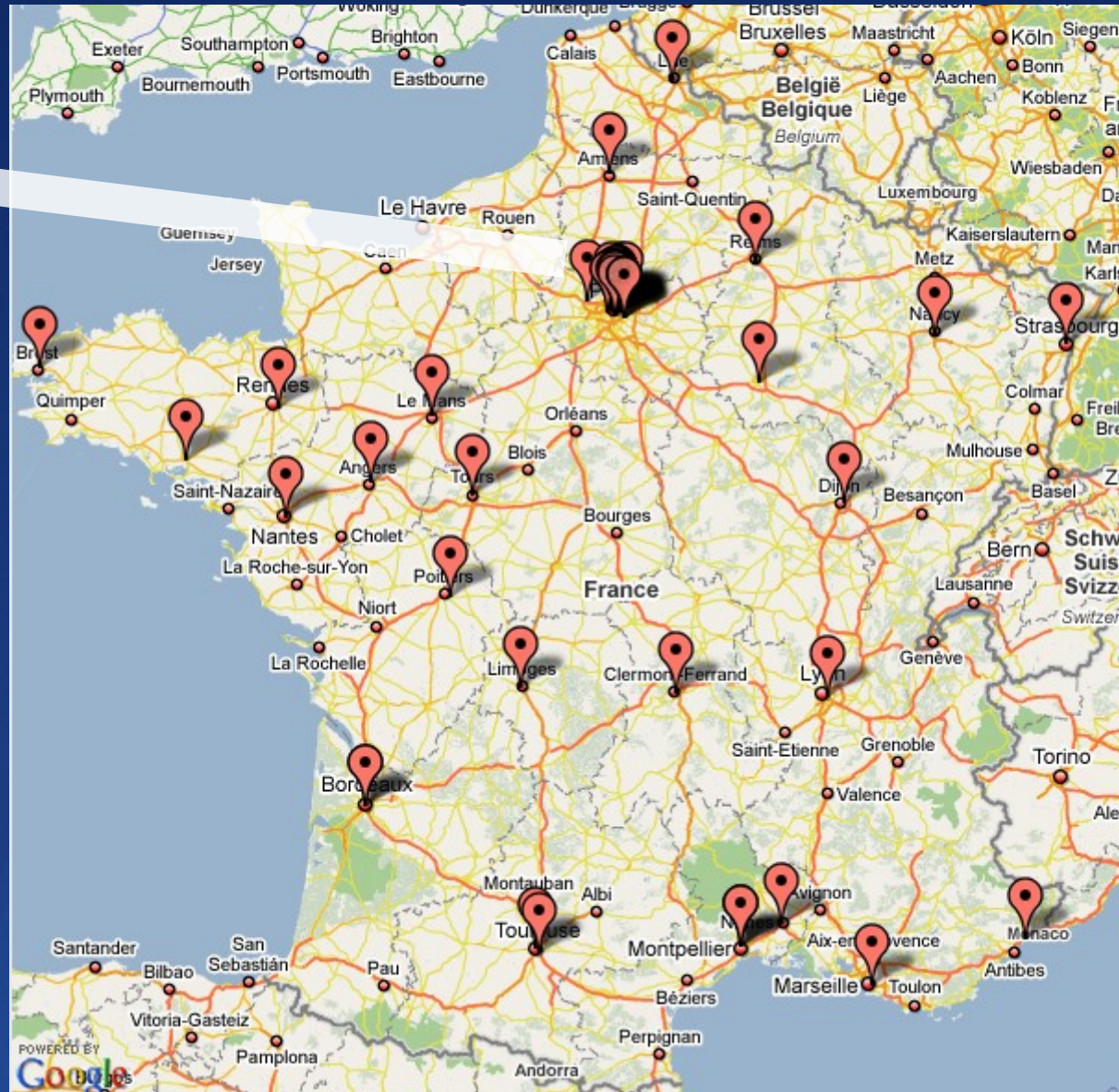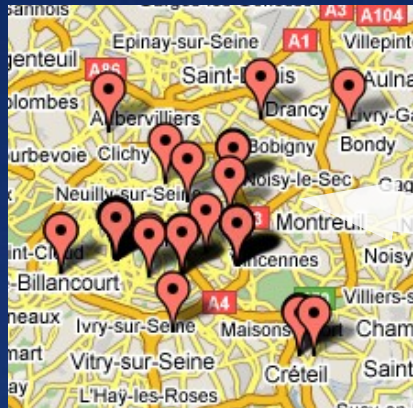CEMARA
orphanet GENATLAS

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.
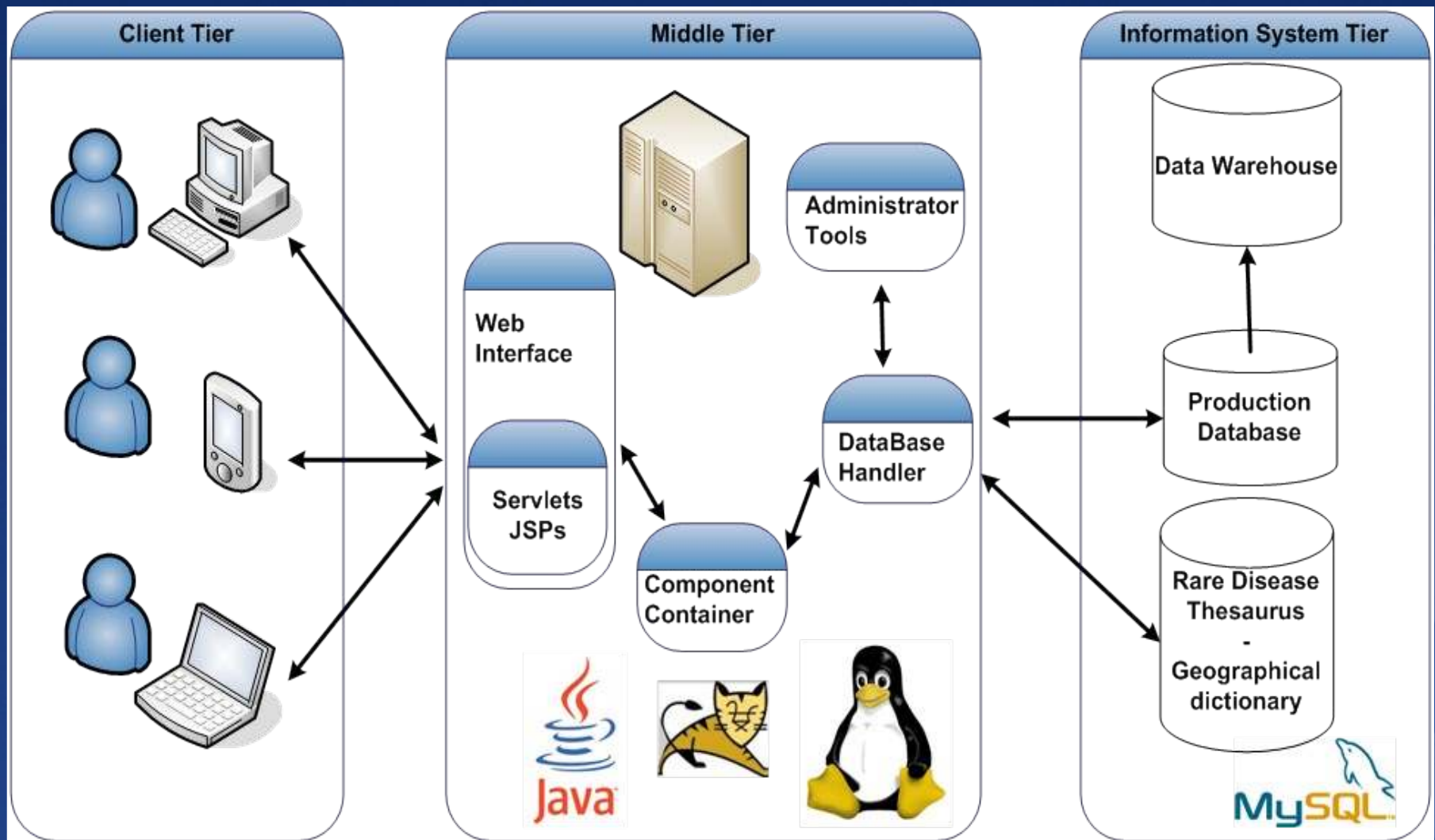
- Most of them are genetic diseases

Rare Diseases Plan

- 132 Reference Centres labeled by the Ministry of Health

- Objectives:
  - monitoring the epidemiology of rare diseases
  - assessing medical activity
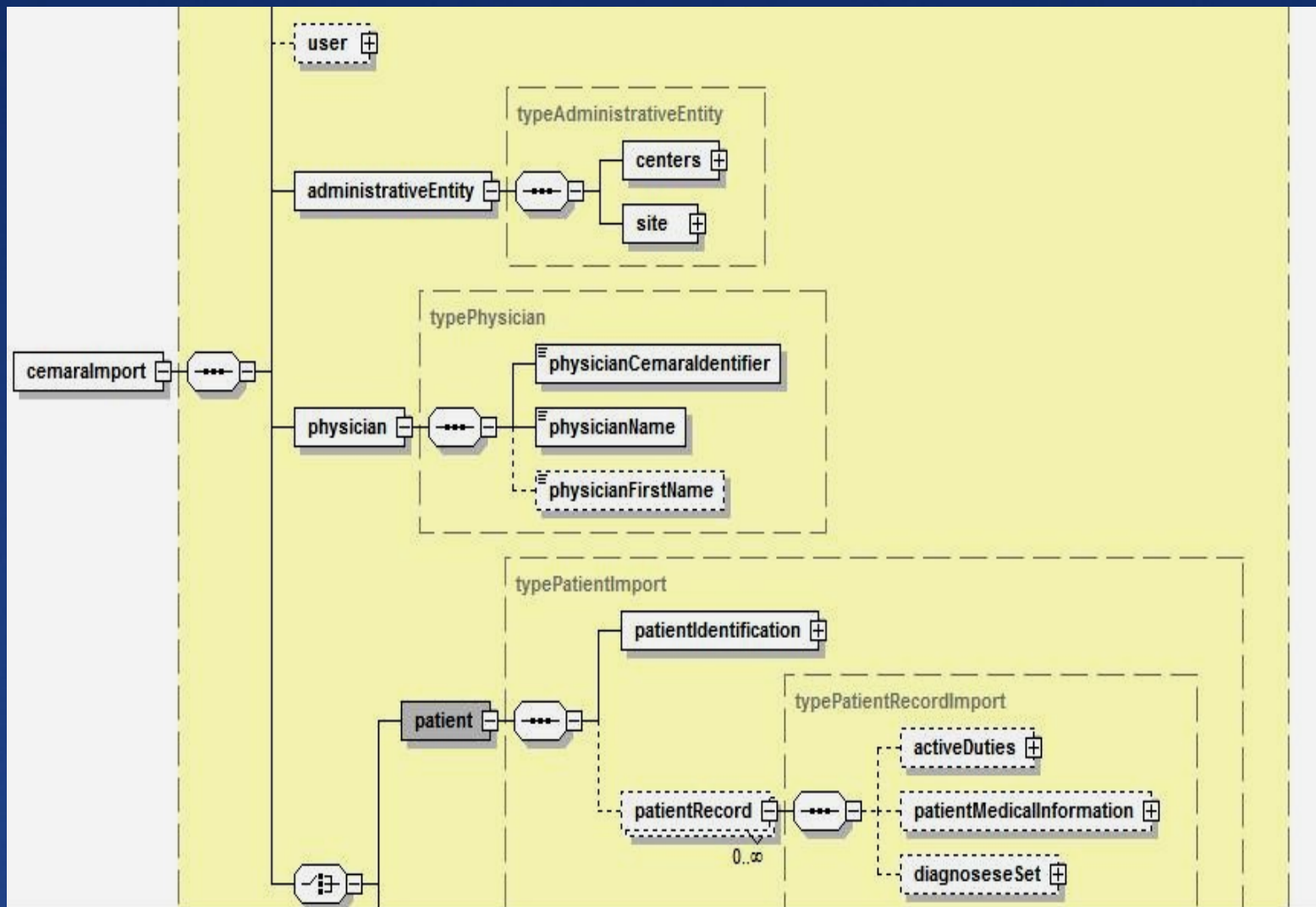 - setting up the national protocols of care

CEMARA Professional Network
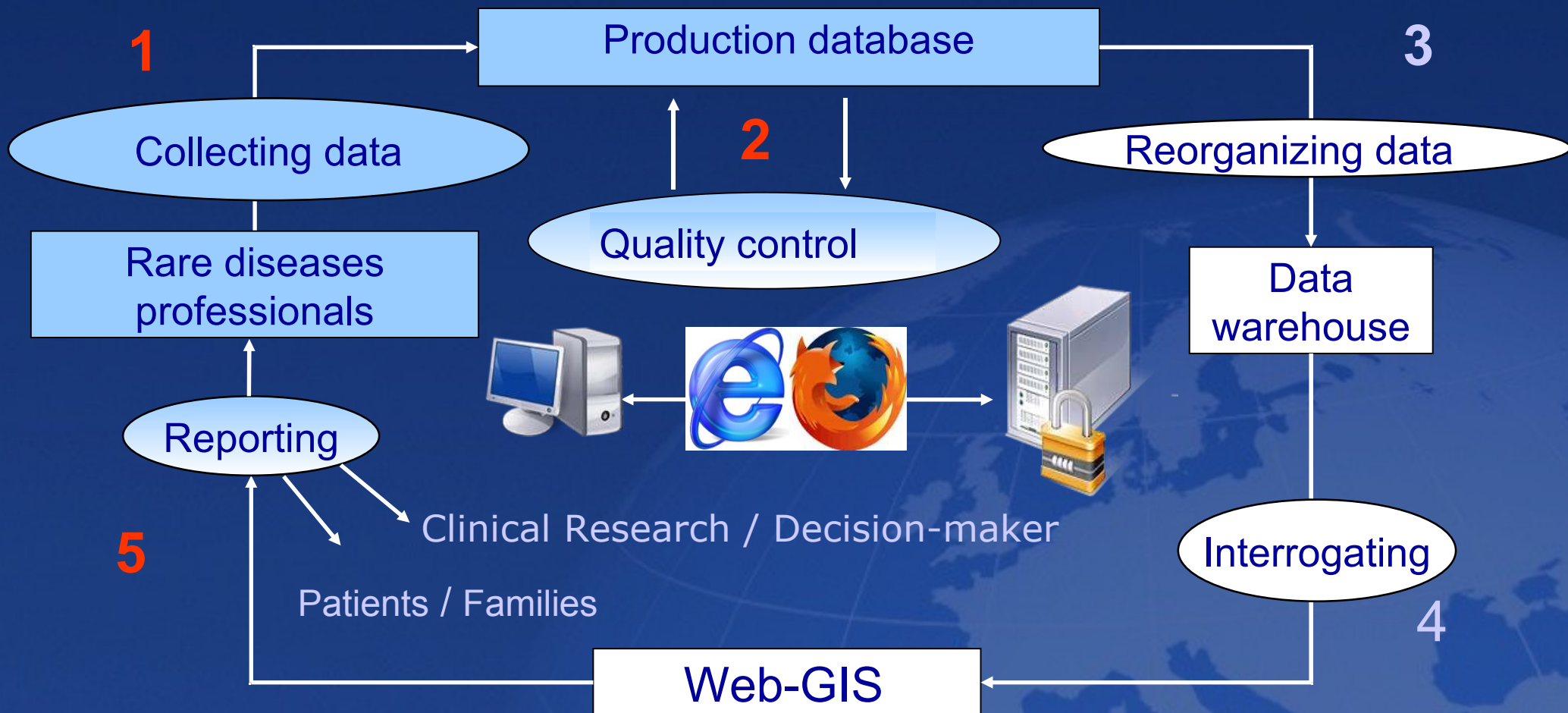

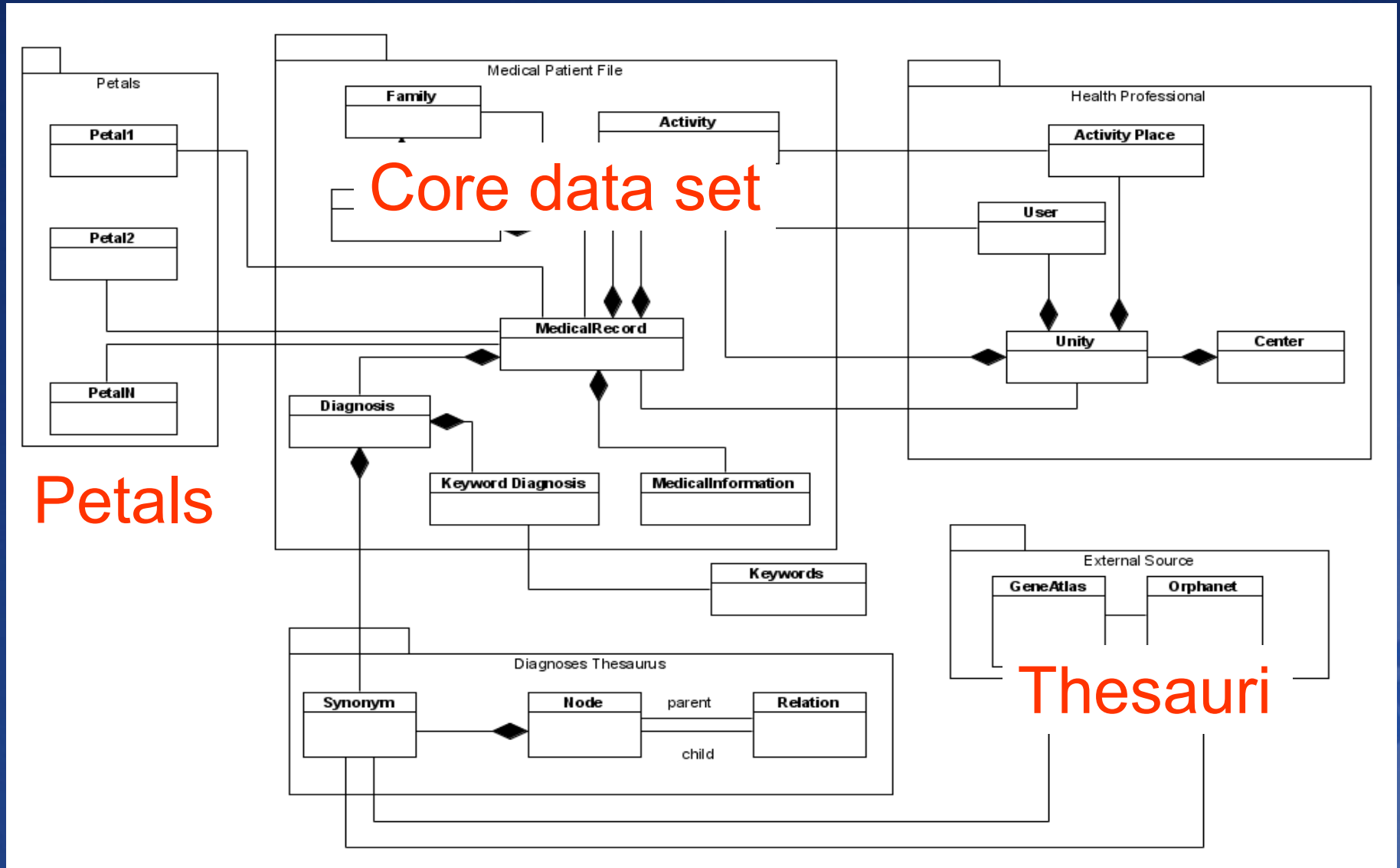C Messiaen - Necker-Enfants Malades Paris

N-tier architecture

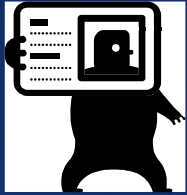# Interoperability – XML scheme
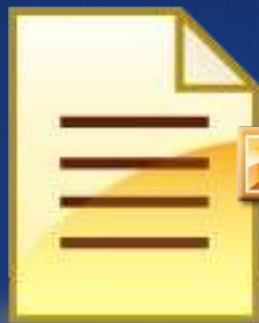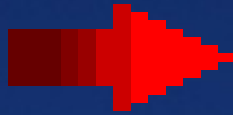
CEMARA Development plan

CEMARA UML Scheme

CEMARA core data set...

Identification

Double entry detection algorithm

Diagnosis + Medical Information + Activity = Patient File

Family File

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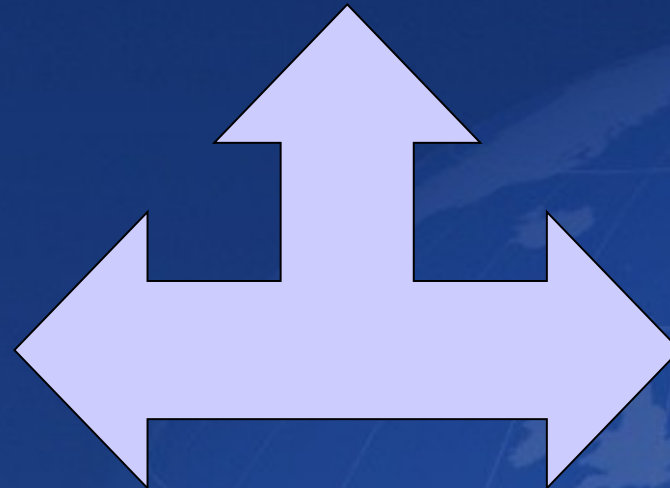
…and the "Petals"

Building a cohort for a given disease or for a group of diseases

Thesauri : a collaborative work

CEMARA patients

orphanet diseases

GENATLAS Université René Descartes - Paris genes

a shared vocabulary

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

CNiL

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

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