Teaching medicine with a terminology/ontology portal

http://www.hetop.eu

Grosjean J, Merabti T, Griffon N, Dahamna B, Darmoni SJ
Rouen University Hospital & TIBS, LITIS EA 4108, Normandy University

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Introduction

- Terminologies and ontologies, important source of knowledge.

- Indexing and reasoning

- Terminology portals: Bioportal (english), HMTP (French/english)

- Teaching?
  - Anatomy (FMA)
  - Rare diseases (Orphanet, HPO)
Methods: modelling

Meta-modelling (upper level modelling)

For each Terminology or Ontology (T/O): study the original model and create a specific parser program

Database (metamodel implementation)
Methods: web service

Terminologies / Ontologies

32 T/O:
1.3 million concepts,
4 millions relations,
23 languages

DB

WebService

Model multi-
terminologies
-discipline
-lingual

• Curators
• Physicians
• Translators
• T/O editors
• Students?
Methods: evaluation

- Taught to medical student
- “How do you grade portal interest in teaching ?”
- “How do you grade the portal ergonomics ?”
- Analogical scale (0-100)
Results: HETOP

- HETOP is a free portal providing access to 32 Health terminologies/ontologies including: MeSH, ICD-10, FMA, Orphanet, HPO, SNOMED int… (free access for academic use only)

- HETOP provides the usual data for each concept: preferred terms, original code, synonyms, definitions and other attributes, relations and hierarchies.
Results : HETOP

- Cross-lingual functions: querying in any of the 23 available languages including English, French, German, Spanish, Portuguese, Italian, Swedish, Arabic, Russian…

- Access to other resources on the Internet (PubMed, CISMeF, etc.) with infobutton system (InfoRoute)

- Bilingual version used daily by over 500 unique users => about 7,000 queries per day
Results: Teaching anatomy

**Quadriiceps femoris** (FMA Entity)

- **Description**
- **Hierarchies**
- **Relations**
- **Resources**

**French term:**
- Muscle quadriceps fémoral

**English term:**
- Quadriiceps femoris

**Original code:** 22428

**Synonyms:**
- Suggest a new synonym

**FMA Synonym**
- Quadriiceps

**CUI UMLS**
- French
- C0224440

**UMLS Synonym**
- French
- Muscle crural
- Muscle vaste externe
- Muscle vaste médial de la cuisse

**Contextual access to bioportal**

**Synonyms**
- Muscle droit antérieur de la cuisse
- Muscle vaste intermédiaire de la cuisse
- Muscle quadriceps
- Muscle vaste interne
- Muscle vaste latéral de la cuisse
- Muscle quadriceps crural
- Muscle vaste latéral
- Muscle vaste médial
## Results: Teaching anatomy

### Quadriiceps femoris (FMA Entity)

<table>
<thead>
<tr>
<th>Description</th>
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</table>

**Relations (full):** Intra-terminology, Inter-terminology

**Secondary segmental supply(ies) (1):**
- L2 (FMA Entity)

**Systemic part(s) (1):**
- Musculature of anterior compartment of thigh (FMA Entity)

**Member of (1):**
- Musculature of thigh (FMA Entity)

**Regional part (4):**
- Rectus femoris (FMA Entity)
- Vastus intermedius (FMA Entity)
- Vastus lateralis (FMA Entity)
- Vastus medialis (FMA Entity)

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### Results: Teaching anatomy

#### L2 (FMA Entity)

<table>
<thead>
<tr>
<th>Description</th>
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<th>Relations</th>
<th>Resources</th>
</tr>
</thead>
<tbody>
<tr>
<td>Relations (full):</td>
<td>Intra-terminology</td>
<td>Inter-terminology</td>
<td></td>
</tr>
</tbody>
</table>

#### Secondary segmental supply of (1)

- Quadriceps femoris  
  - FMA Entity

#### Segmental supply of (17)

- Adductor brevis  
  - FMA Entity
- Iliacus  
  - FMA Entity
- Pectineus  
  - FMA Entity
- Second lateral lumbar intertransversarius  
  - FMA Entity
- Adductor longus  
  - FMA Entity
- Iliocostalis lumborum  
  - FMA Entity
- Psoas major  
  - FMA Entity
- Second medial lumbar intertransversarius  
  - FMA Entity
- Adductor magnus  
  - FMA Entity
- L2 dermatome  
  - FMA Entity
- Quadratus lumborum  
  - FMA Entity

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Results: Teaching rare diseases

**Cystic fibrosis** (Orphanet Disease)

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<th>Description</th>
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<th>Resources</th>
</tr>
</thead>
</table>

**French term:**
Mucoviscidose

**English term:**
Cystic fibrosis

**Original code:**
586 (PROD)

**Synonyms:**
[Suggest a new synonym]

**ORPHA Synonym**
French
Fibrose kystique du pancréas

**Specific attributes:**

**Transmission**
- French
- Autosomique récessif
- Autosomal recessive
- Référence MIM
- French

**Prévalence**
- French
- 1-5 / 10 000

**Apparition**
- French
- Néonatal/petite enfance

Contextual access

Orphanet Knowledge
Results: Teaching rare diseases

**Cystic fibrosis** (Orphanet Disease)

- **Description**
- **Hierarchies**
- **Relations**
- **Resources**

- Full tree

  - Orphanet Diseases top tree
  - Channelopathy
    - Non-pore-loop channelopathy
    - Non-pore-loop channelopathy due to epithelial Cl- channel CFTR anomaly
      - Cystic fibrosis
  - Rare cardiac disease
    - Cardiomyopathy
      - Secondary cardiomyopathy
        - Endocrine disease with cardiomyopathy
          - Diabetes mellitus
          - Exocrine pancreatic disease with diabetes
            - Cystic fibrosis
  - Rare gastroenterologic disease
    - Rare pancreatic disease
      - Cystic fibrosis
  - Rare genetic disease
Results: Teaching rare diseases

Cystic fibrosis (Orphanet Disease)

Related gene(s) (1)
- CFTR

Related sign(s) (9)
- Agammaglobulinemia/hypogammaglobulinemia
- B-cell deficiency
- Repeate respiratory infections
- Structural anomalies of the liver and the biliary tract
- Structural anomalies of the pancreas
- Hepatomegaly/liver enlargement (excluding storage disease)
- Immunodeficiency/increased susceptibility to infections/recurrent infections
- Lung fibrosis
- Malabsorption/chronic diarrhea/seatorrhea

ICD-10 link(s) (1)

A pour Phenotype(s) HPO (16)
- Asthma
- Chronic lung disease
- Male infertility
- Autosomal recessive inheritance
- Cor pulmonale
- Meconium ileus
- Biliary cirrhosis
- Dehydration
- Pancreatic insufficiency
- Bronchiectasis
- Distal intestinal obstruction syndrome
- Rectal prolapse
- Bronchopulmonary infection
- Failure to thrive
- Hypercalciuria
- Recurrent pneumonia

Orphanet signs (9)

HPO signs (16)
Results: Teaching rare diseases

**CFTR (Orphanet Gene)**

- **Description**
- **Hierarchies**
- **Relations**
- **Resources**

**Relations (full):**
- Intra-terminology
- Inter-terminology

**Related disease(s) (3):**
- Congenital bilateral absence of vas deferens
- Cystic fibrosis
- Hereditary chronic pancreatitis

Orphanet diseases (3)
Results: evaluation

2010 and 2011 evaluation of HETOP (Mean)

<table>
<thead>
<tr>
<th></th>
<th>2010</th>
<th>2011</th>
<th>Overall</th>
</tr>
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<tbody>
<tr>
<td>Interest in teaching</td>
<td>79.9</td>
<td>87.5</td>
<td>83.7</td>
</tr>
<tr>
<td>Ergonomics</td>
<td>57.2</td>
<td>55.5</td>
<td>56.35</td>
</tr>
<tr>
<td>n</td>
<td>25</td>
<td>11</td>
<td>36</td>
</tr>
</tbody>
</table>

“How do you grade portal interest in teaching?”
“How do you grade the portal ergonomics?”
Strenghts and limits

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- Orphanet available in English, French, Italian, Spanish and Portuguese → Teaching in many countries
- Extension to other fields with other terminologies/ontologies (NCIT, SNOMED CT)
- Cross-lingual navigation

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- An improvable design
Conclusion

- Part of the curriculum at Rouen Medical School
- Many other uses:
  - Indexing
  - Translating
  - Coding
  - Demonstration tool: show a new terminology, an automatic translation...
Thank you!

Any questions?

nicolas.griffon@chu-rouen.fr

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