Ontology-supported Clinical Profiling for the Evaluation of Obesity and Related Comorbidities

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Objective

- Development of an ontology-supported knowledge base and related tools
  - To assist physicians in the determination of the health status of a subject
  - To support knowledge transfer between medical researchers and general practitioners
Knowledge base focus

- **Obesity**
  - A disease in which excess body fat has accumulated such that health may be adversely affected
  - Causes nearly **3 million deaths** every year worldwide\(^1\)
  - In Europe, obesity has **reached epidemic proportions**\(^2\)

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Knowledge base design: the ontology

- **Ontology**
  - In Computer Science literature defined as: “an explicit specification of a conceptualization”
  - tied to a domain of interest
  - Built by defining its schema
    - The **concepts** identified in the domain of interest
Collaborative ontology design

- Ontology design has been carried out involving domain experts

- Computer scientists formalized the ontology schema, semantic rules and supporting tools
Ontology design steps

1. Define the domain of interest
2. Identify key concepts
3. Identify relations between concepts
4. Produce the ontology schema
5. Add rules for semantic reasoning
6. Validate and refine the ontology schema
Ontology domain and concepts

The domain of interest
- Human health status in relation to metabolic balance

Concepts identified
- Person
- Body structure
- Physiological profile
- Subject lifestyle
- Nutrition
- Diagnostical tools

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Ontology concepts identified

- Person
  - The main concept around which the whole ontology is based
- Body structure
  - Body Mass Index, Abdomen Circumference, Body Fat, Skeletal Muscle Mass...
- Physiological profile
  - Blood pressure, heart frequency, glycaemia...
- Subject lifestyle
  - Smoker/non smoker, performs physical activity...
- Nutrition
  - Follows some kind of diet
- Diagnostical tools
  - Examinations and techniques adopted to evaluate an individual’s body structure or physiological parameters

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Relations between concepts

- Are defined over individuals, i.e. instances of classes
  1. Specialization relations
     - E.g. “an individual of the class Obese is also in the class Person”
  2. Relations between individuals
     - E.g. “an individual of the class Person is tied to an individual of the class PhysicalActivity through the isEngagedIn relation”
  3. Relations between individuals and values
     - E.g. “an individual of the class Person is tied to a number through the hasBMI relation”
Semantic rules

- Allow to make inferences about properties’ values associated with a class describing a person’s health status
  - “if person $x$ has a body mass index property associated with a value of $y$, and $y$ is between 25 and 29.9, then $x$ is overweight”
Semantic rules

- **38 rules** subdivided in:
  - Individual classification (4)
  - Obesity level detection (3)
  - Central obesity detection (2)
  - Sarcopenia detection (8)
  - Hypertension detection (3)
  - Dyslipidemia detection (5)
  - Diabete detection (2)
  - Insulin resistance detection (1)
  - Metabolic syndrome detection (10)
Ontology implementation - schema

Dashed lines: relations between individuals

Solid lines: specialization relations

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Supporting tools developed

A Web-based application has been developed to simplify the interaction with the ontology
Supporting tools developed
Conclusions

- An ontology-supported medical informatics tool suitable for clinical profiling of patients has been developed.
- The ontology approach allowed to:
  - model the diverse and interconnected aspects of the medical domain related to obesity.
  - Specify semantic rules to infer additional knowledge.
- The knowledge base has been developed in the context of the PICKFIBER EU project\(^1\)
  - The tools have been validated with data provided by the University of Lleida\(^2\) about obesity status and related comorbidities.

**Next step**: interfacing the ontology with already existing ones to better describe aspects related to nutrition and health care.

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\(^1\)PICKFIBER (Platform for International Collaborative Knowledge on Food Improvement, Based on Ecological Resources) project, rif. N° 0120R1/21, subproject in the miniprogramme Innovation4Welfare, co-funded by INTERREG IVC Programme.

\(^2\)University of Lleida (Nutren-Nutrigenomics) – Catalonia (ES) - [http://www.nutren.udl.cat](http://www.nutren.udl.cat)
Classification and data properties are inferred by rules in the form of:

**Antecedent -> Consequent**

- Antecedent and consequent are conjunctions of simple proposition
  - $x$ is a Person

- If the antecedent holds (it is “true”), then the consequent must also hold
  - **IF** $p$ is a Person with a body mass index greater than or equal to 30, **THEN** he/she is obese
## Ontology defined data properties

### Individual's general properties:
- Age
- Sex
- Weight
- Height
- Ethnic Group
- Smokes (Yes, No, Ex)
- Follows some kind of diet (Yes, No)
- Follows a specific therapy (Yes, No)
  - blood pressure control
  - Diabetes
  - Cholesterol control
  - Triglycerides control

### Body composition properties:
- Body Mass Index
- Abdomen Circumference
- Body Fat
- Body Fat Percentage
- Fat Free Mass
- Fat Free Mass Percentage
- Skeletal Muscle Mass
- Skeletal Muscle Index Percentage
- Subcutaneous Adipose Tissue
- Visceral Adipose Tissue

### Physiological properties:
- Systolic Blood Pressure
- Diastolic Blood Pressure
- Heart Frequency
- Basal Calorimetry
- Glycaemia
- Total Cholesterol
- HDL Cholesterol
- HOMA Index
- IL6
- Insulin
- Protein C
- TNAlpha
- Triglycerides
- Heart rate variability
- Leptin
- Total Cholesterol

### Physical activity characterization properties:
- Performs physical activity (Yes, No)
  - Intensity (moderate, strong)
  - Weekly frequency (minutes per week)
- MET
- Code
- Description
Ontology Vs Database

- **Ontology**
  - Focuses on **meaning** and **shared understanding**
  - Oriented towards **human communication** and **Extensibility**

- **Database**
  - Focuses on **data**
  - Oriented towards **storage and querying efficiency**

- **Data has more granularity than concepts**