

Ontological Labels for Automated Diagnosis

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Computational Anatomy Workshop
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Outline

- 1 Introduction
 - Ontologies
 - Biomedical Ontologies
 - Applications
- 2 Methods
 - Computational Cardiac Anatomy
 - Application Ontology Extraction
 - Ontology Atlas Generation
 - Registration
 - Querying
- 3 Results
 - Still Waiting
- 4 Discussion
 - Extensions of This Work
 - Acknowledgements

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What is (an) ontology?

Definition

ontology (n. ä-n-'tä-lə- jē) : A branch of metaphysics concerned with the nature and relations of being.

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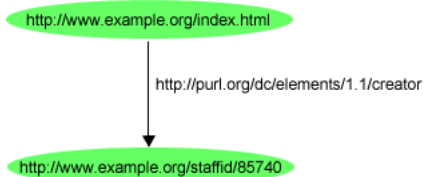
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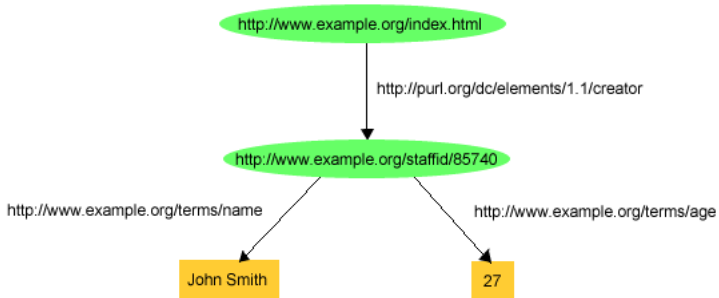
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The Building Blocks: Triples



Description Logics and Directed Graphs



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Examples of Biomedical Ontologies

Example

- SNOMED Clinical Terms
- RadLex
- Cell Behavior Ontology
- **Foundational Model of Anatomy**

National Center for Biomedical Ontology

- <http://www.bioontology.org>
- BioPortal: <http://www.biportal.bioontology.org>

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Existing Use Cases

- Multi-modal (and not just image modalities) annotation
- Unified nomenclature
- Multi-scale querying
 - See Larson and Martone, “Ontologies for neuroscience: what are they and what are they good for?”, *Frontiers in Neuroscience*, 2009.

Automated Reasoning

- Description logics a decidable fragment of first-order logic
- Complex semantic relationships between classes and instances
 - subclass, intersection, union, min/max cardinality, same as, domain, range, ...

Overview: What are we “diagnosing”?

Automatic recognition and location of left-ventricular remodelling, a biomarker for heart failure.

- 1 Computational cardiac anatomy: quantifying left-ventricular remodelling
- 2 Application ontology extraction
- 3 Ontology atlas generation
- 4 Registration
- 5 Querying

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Distinguishing ICM vs. NICM

For details, see Ardenaki et al, “Computational method for identifying and quantifying shape features of human left ventricular remodeling”, *Annals of Biomedical Engineering*, 2009.

- 1 In-vivo MDCT image acquisition
- 2 LV extraction
- 3 Average template (one ES, one ED) generation
- 4 Affine + LDDMM registration
- 5 PCA coefficients distinguish two groups

Tissue Volume Expansion

Imposed on ES template, area of significant expansion in NICM compared to ICM.

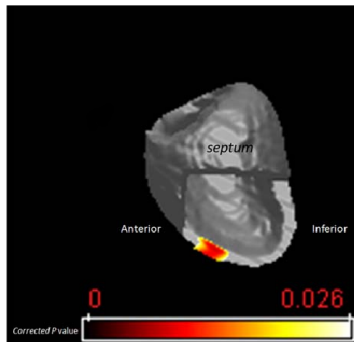


FIGURE 8. Map of the voxel-based significance of tissue expansion for NICM group relative to ICM group. The color scale represents the significance of tissue expansion measured in corrected p values, with yellow representing the least significant area.

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Reference vs. Application Ontologies

- Reference ontology: large, cross-domain body of knowledge
 - Too much information can be a bad thing
- Application ontology: smaller, domain-specific
 - Now, extracted from one or more reference ontologies
 - vSparQL (extends query language with recursive and sub-queries)
 - See Shaw et al “Generating application ontologies from reference ontologies”, *AMIA Symposium*, 2008.

Steps Towards Cardiovascular Ontology

- Myocardium extraction, general term identification



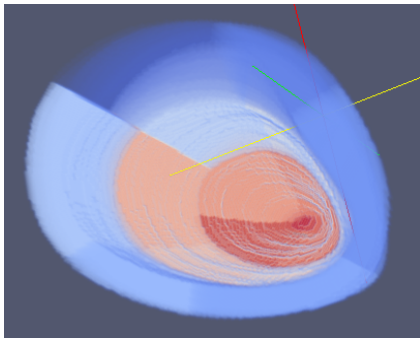
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High-Resolution DTMRI Image

- From Drs. Helm and Winslow.
- LV extracted, hand segmented by Geoffrey Gunter.
- Segmentation recombined, intensities based on region
- Ontological labels via intensity map
 - Need for NIfTI format

Ontology Atlas



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Aligning Atlas With ES Template

- Many attempts, both directions
 - Now focused on Atlas \rightarrow ES
- With and without affine registration
- For details, see <http://wiki.cis.jhu.edu/project/OCV>

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Some Questions We Can Ask

- Programming environment: Java + JENA
 - See <http://github.com/shanest/OntologyCV>
 - Libraries exist for many languages, so can integrate with myriad pipelines
- Sample queries:
 - What is the average T-value per myocardial region?
 - Which regions of myocardium have voxels of significant T-value?
 - Where is Siamak's ROI located?

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Preliminary Failures

- Initial results (from ES \rightarrow Atlas): all basal anterior
- First pass Atlas \rightarrow ES:
 - apical septal
 - mid anteroseptal
 - mid inferoseptal

Next Steps

- Resampled ontology atlas with Siamak's assistance to ensure orientation.
- Landmark-based affine registration.
- Cross fingers

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More to Do

- Finish application ontology extraction
 - Have identified relevant terms in FMA, need to find a non ad-hoc (i.e. biologically motivated) method of extraction
 - Incorporate others: RadLex, Disease Ontology, SNOMED
- Ontology engineering for LDDMM derived metadata
 - And for pipeline description / data location.
- More sophisticated querying.

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Thank You

Questions?