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Using Semantic Web Technologies to Build a Community-driven Knowledge Curation for the Skeletal Dysplasia Domain

(SKELETOME)

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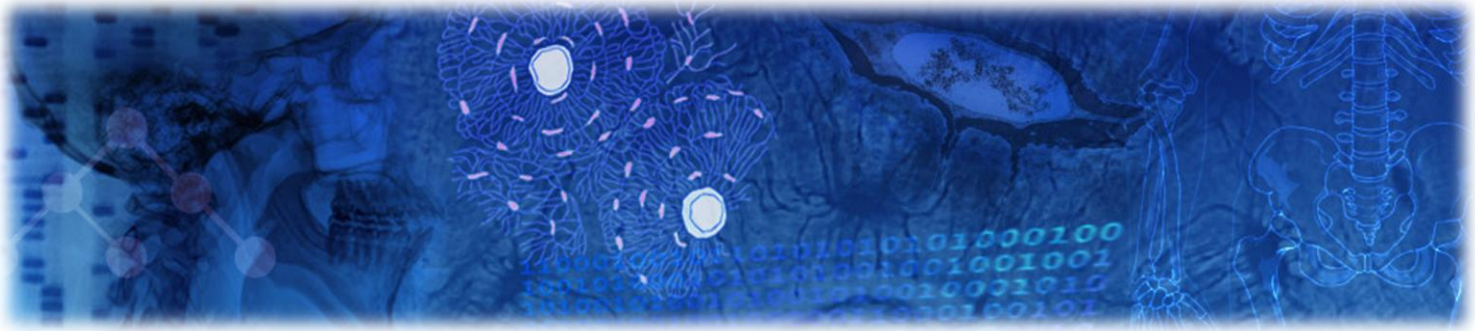
27 Oct. 2011
Bonn, Germany

Skeletal dysplasias …

… from the knowledge representation, curation and discovery perspectives …



- Challenges & Requirements
- The SKELETOME Platform
- Next steps

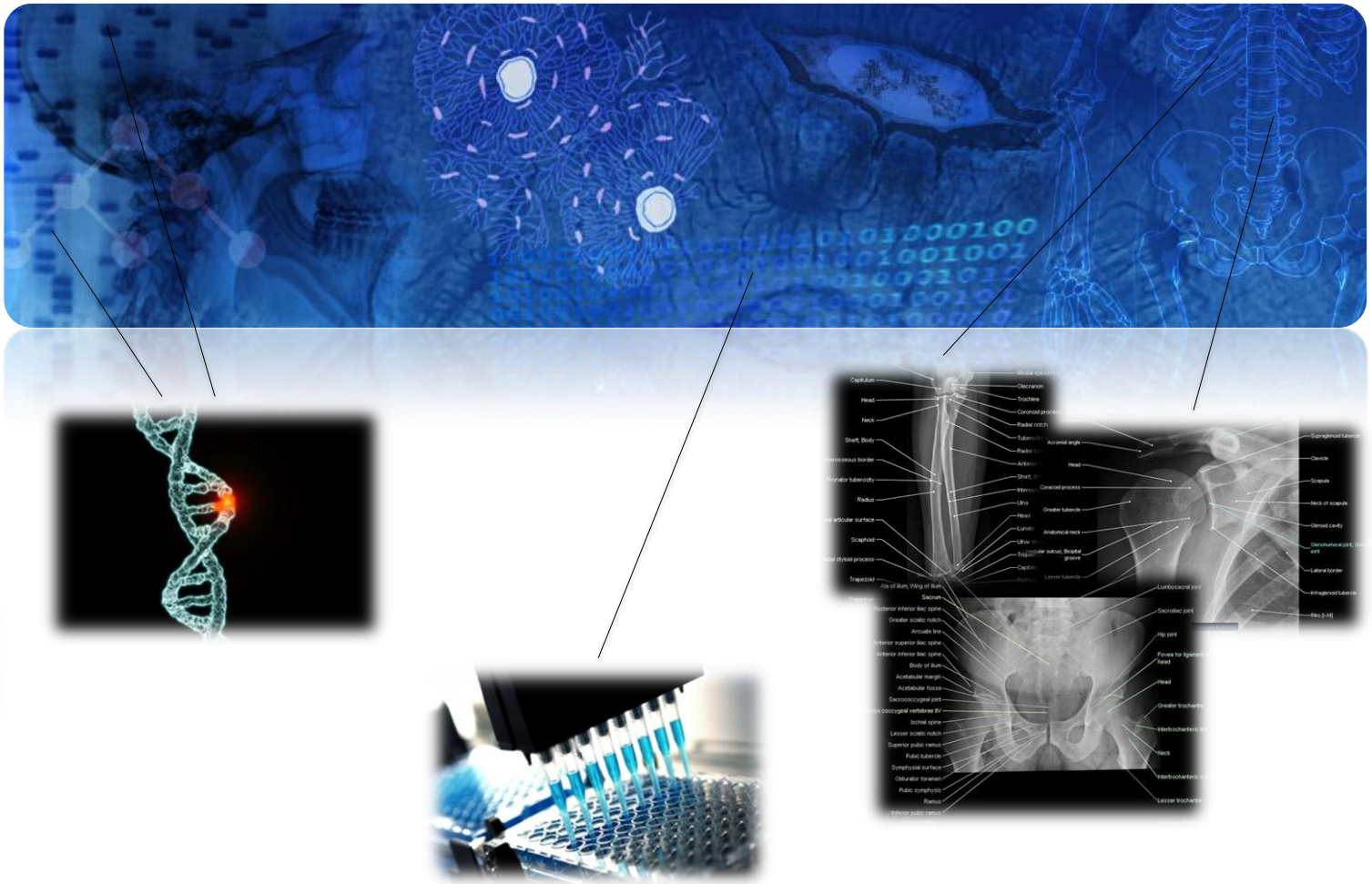
Skeletal dysplasias

Background

- Group of heterogeneous genetic disorders
- 487 types – 40 groups
- Rare (< 1 : 10,000)
- Sparse distribution

Skeletal dysplasias

Background (II)



Skeletal dysplasias

Challenges

Rare Disorders

Few established guidelines

Growing domain knowledge

Decision support

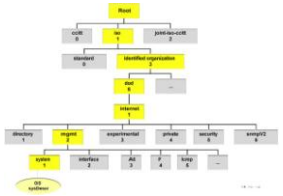
Sparseness

Heterogeneity

Integration

Skeletal dysplasias

Current status



<u>ISDS Nosology</u>
(Warman et al, 2011)

European Skeletal Dysplasia Network

4 years publishing cycle

International Skeletal Dysplasia Registry

- **Groups**
- **Bone Dysplasias**
  - *OMIM*
- **Genes**
  - *Locus*
- **Proteins**

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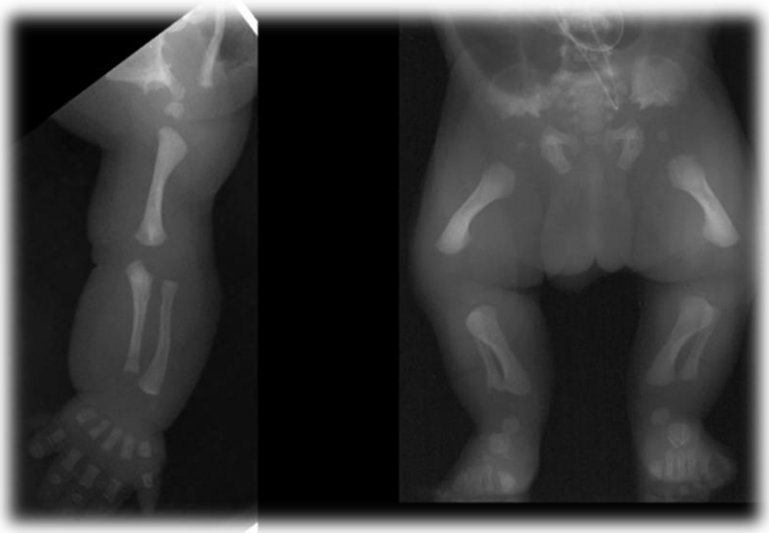
Challenges / Requirements

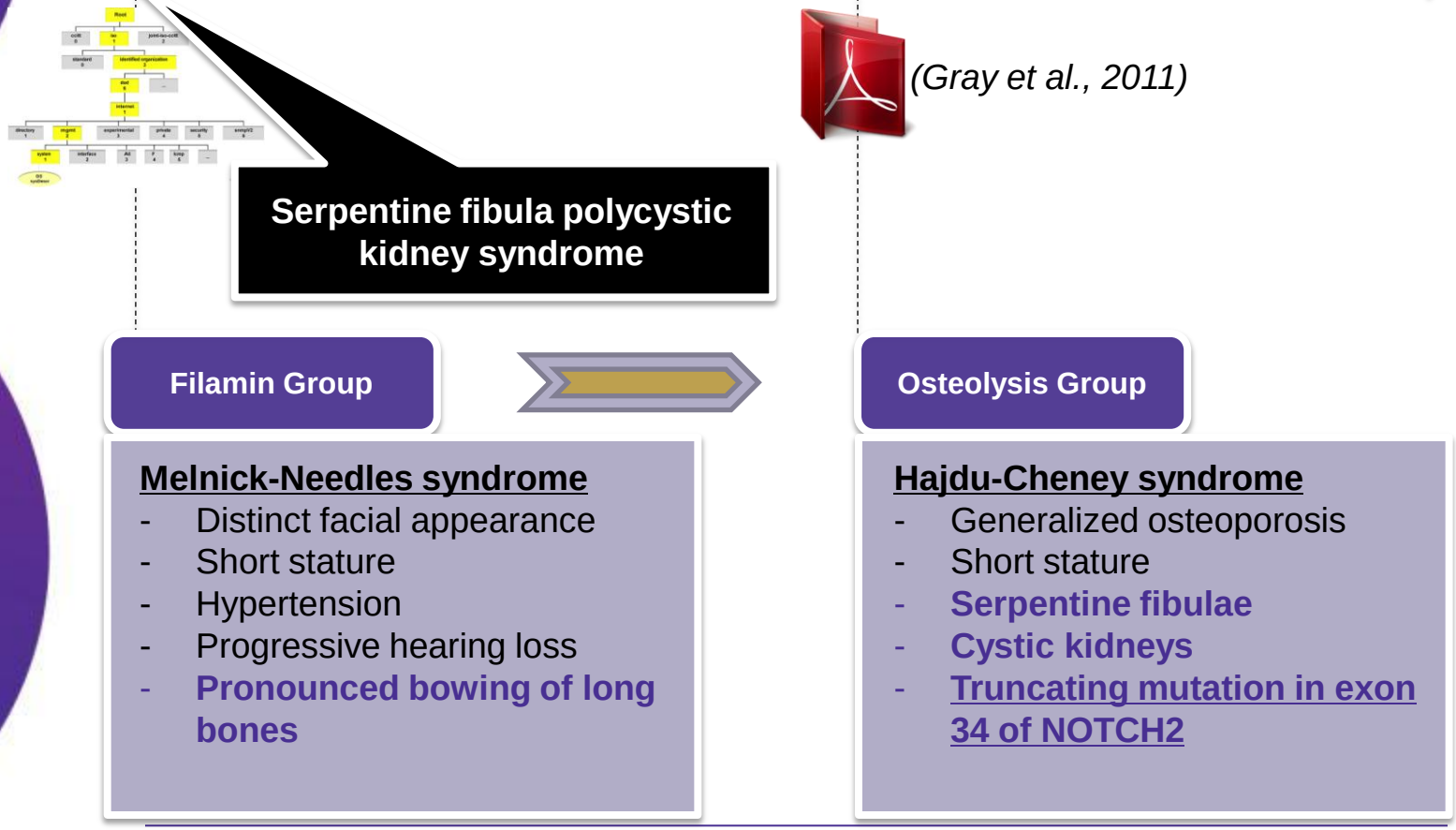
Common terminology



Gorlin/Hall	ISDR/Lachman
Postaxial polydactyly of the hands and feet	Feet – Polydactyly – postaxial – **bilateral**
Short long bones with **smooth** rounded metaphyses	Femur – Metaphysis Proximal – rounded Femur – Metaphysis distal – rounded
'Drumstick' appearance of the radius and **ulna**	Radius – Metaphysis (Distal) – widened

Challenges / Requirements

Knowledge transfer and evolution



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Overview

- Ontology-based …
- … community-driven knowledge curation platform

- Domain knowledge
- Patient repository

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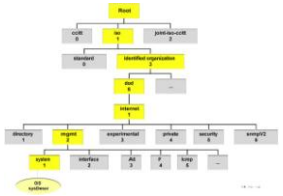
Aims



- Knowledge / Data Integration
- Common terminology
- Decision Support
- Knowledge transfer and sustained knowledge evolution
- Privacy and access control
- Provenance and expertise

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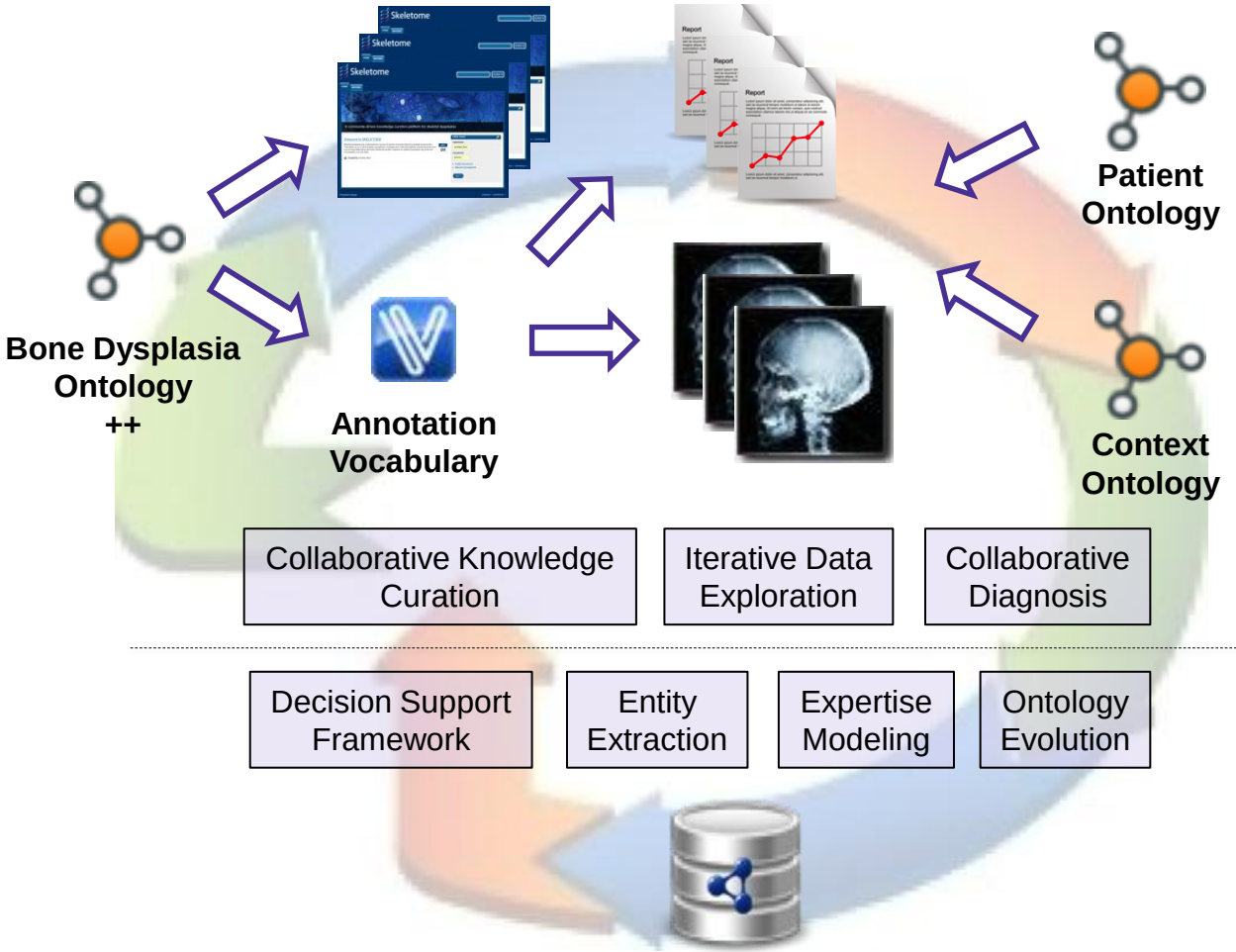
Knowledge engineering cycle



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Ontology set

(Common terminology & Knowledge / Data integration)

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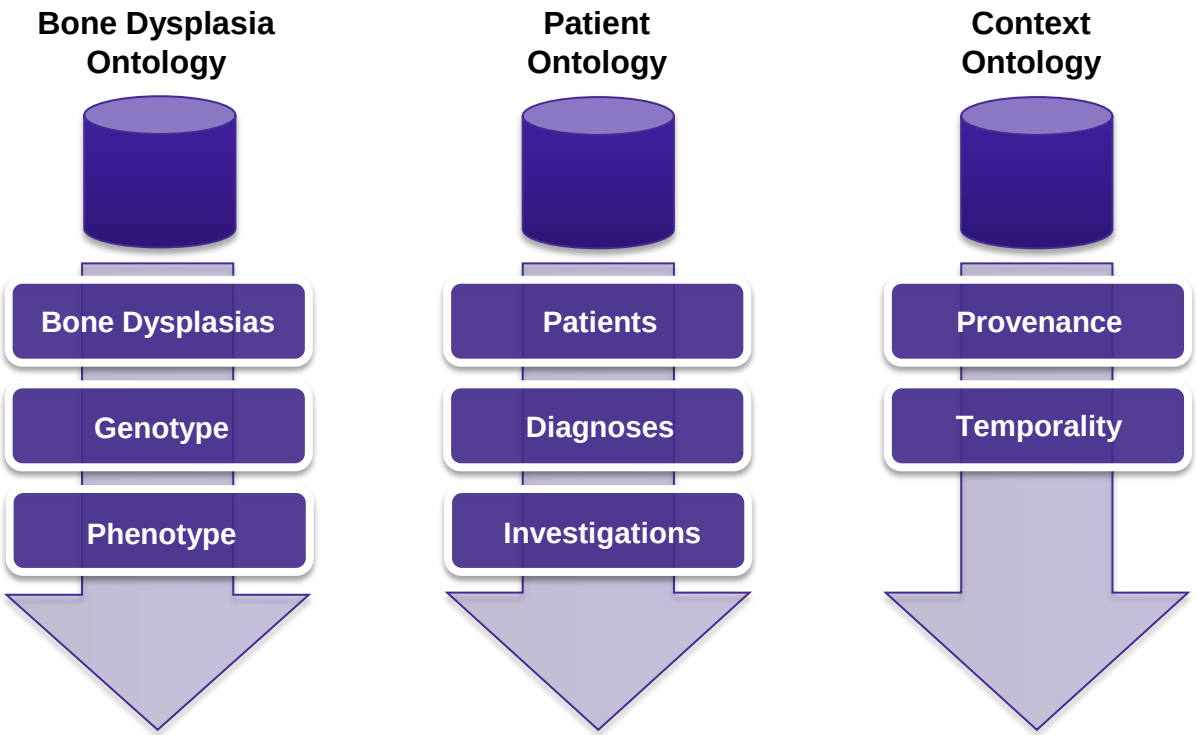
Ontology set



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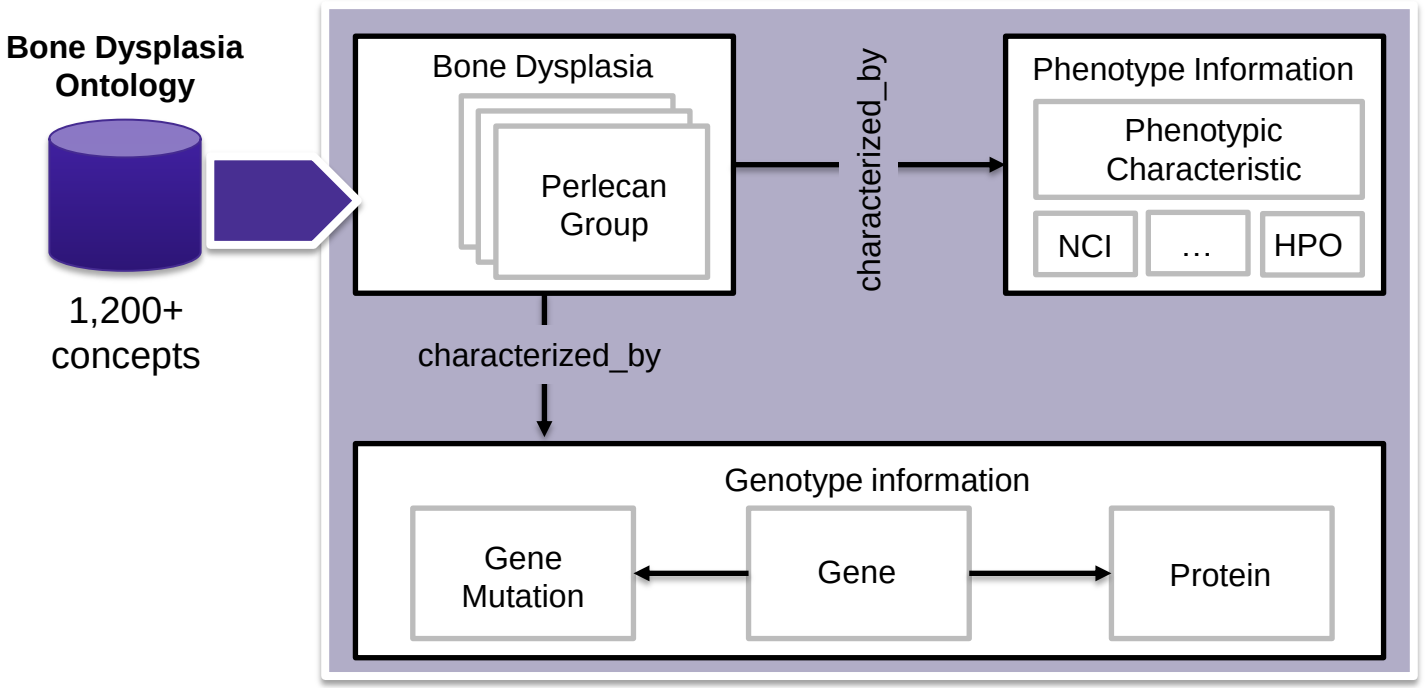
Ontology set



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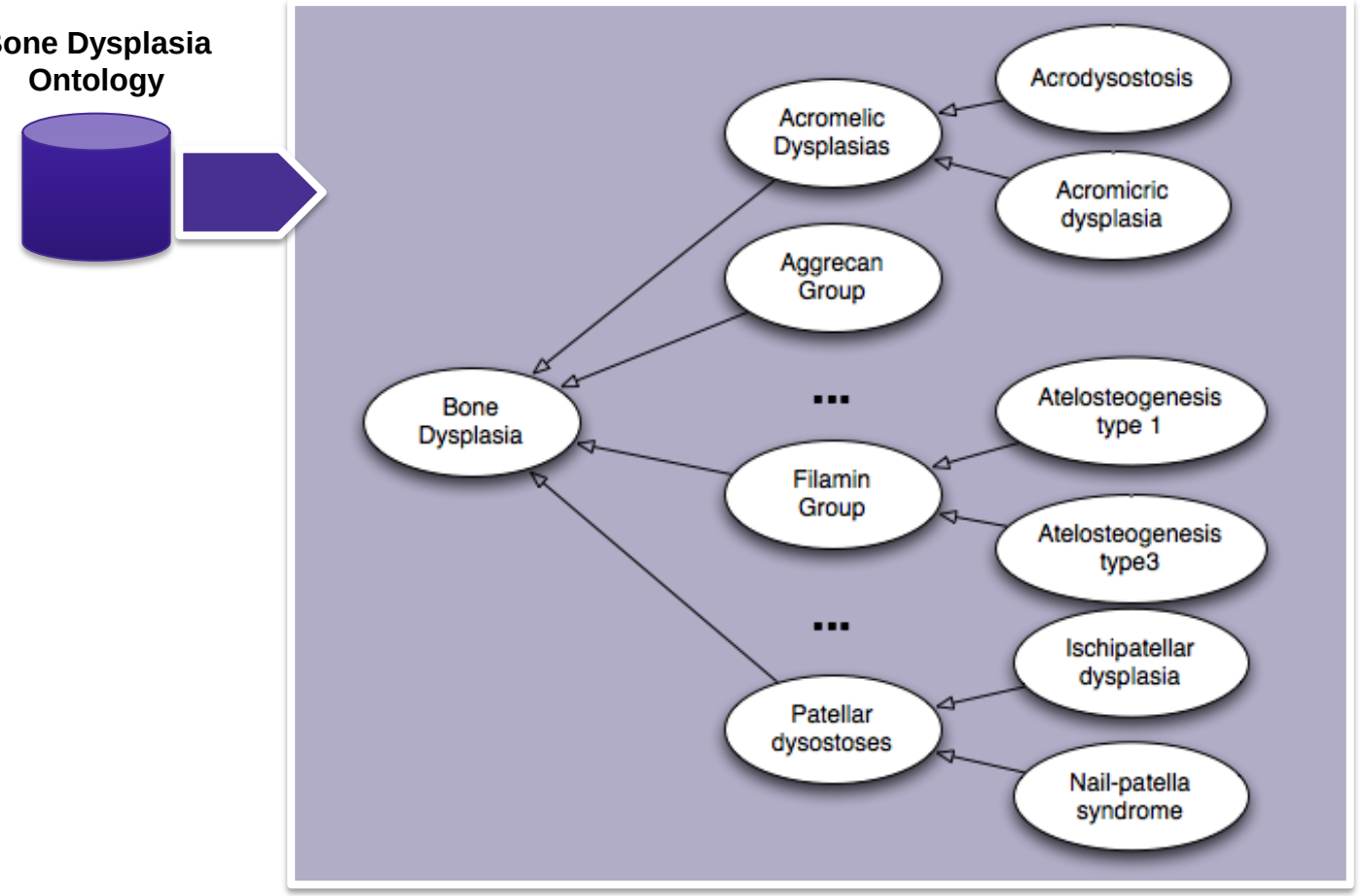
Ontology set

Bone Dysplasia Ontology



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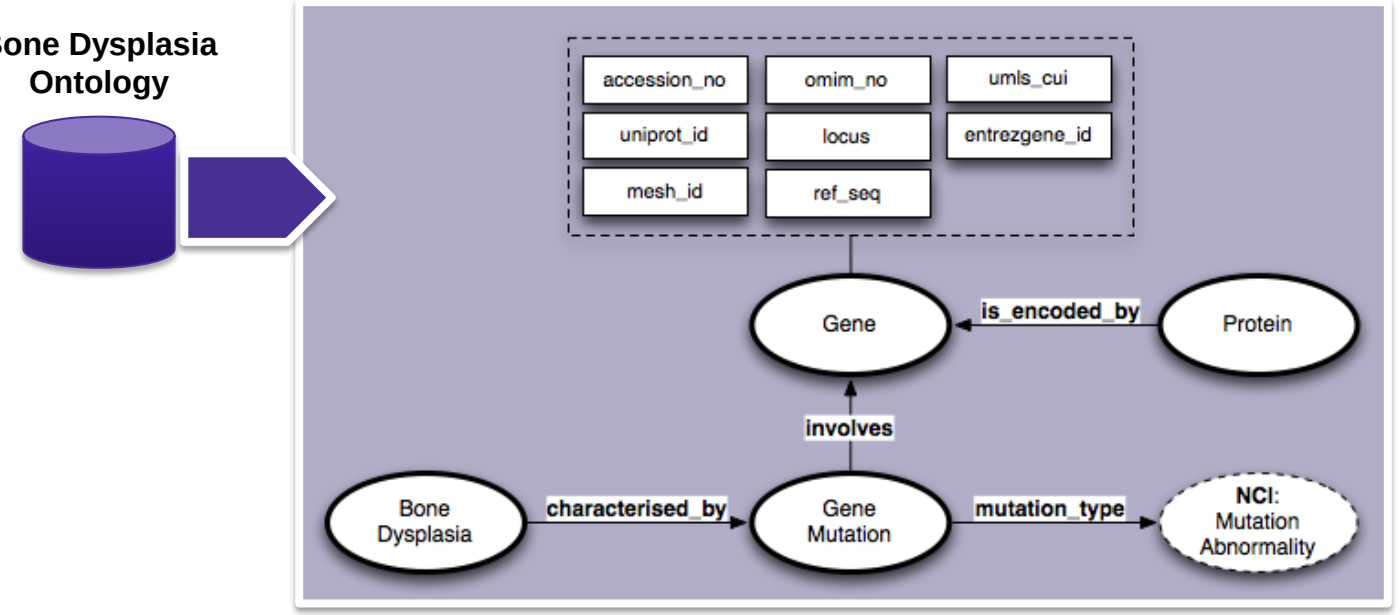
Ontology set

Bone Dysplasia Ontology



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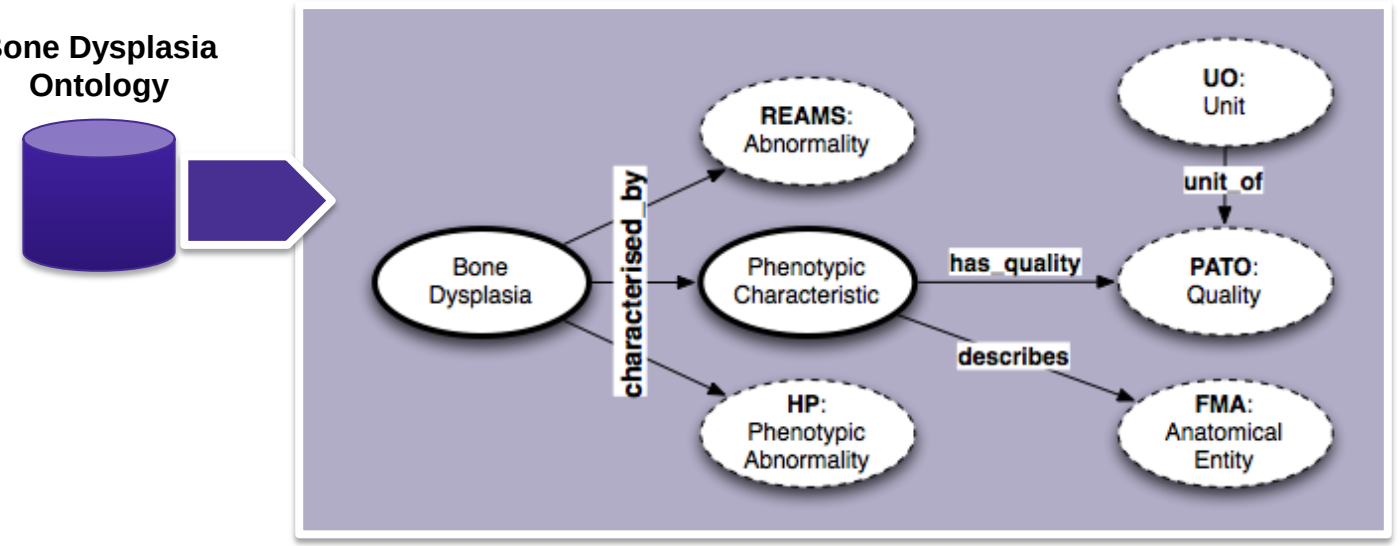
Ontology set

Bone Dysplasia Ontology



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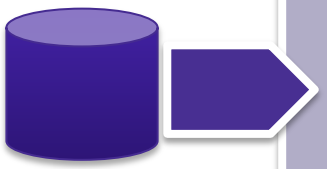
Ontology set

Bone Dysplasia Ontology



Class: **Achondroplasia**
SubClassOf:
characterized_by only (**GM000001** or **GM000361** or **HP_0000238** or **HP_0002938**)
SubClassOf:
mode_of_inheritance only **HP_0000006** and
mode_of_inheritance some **HP_0000006**

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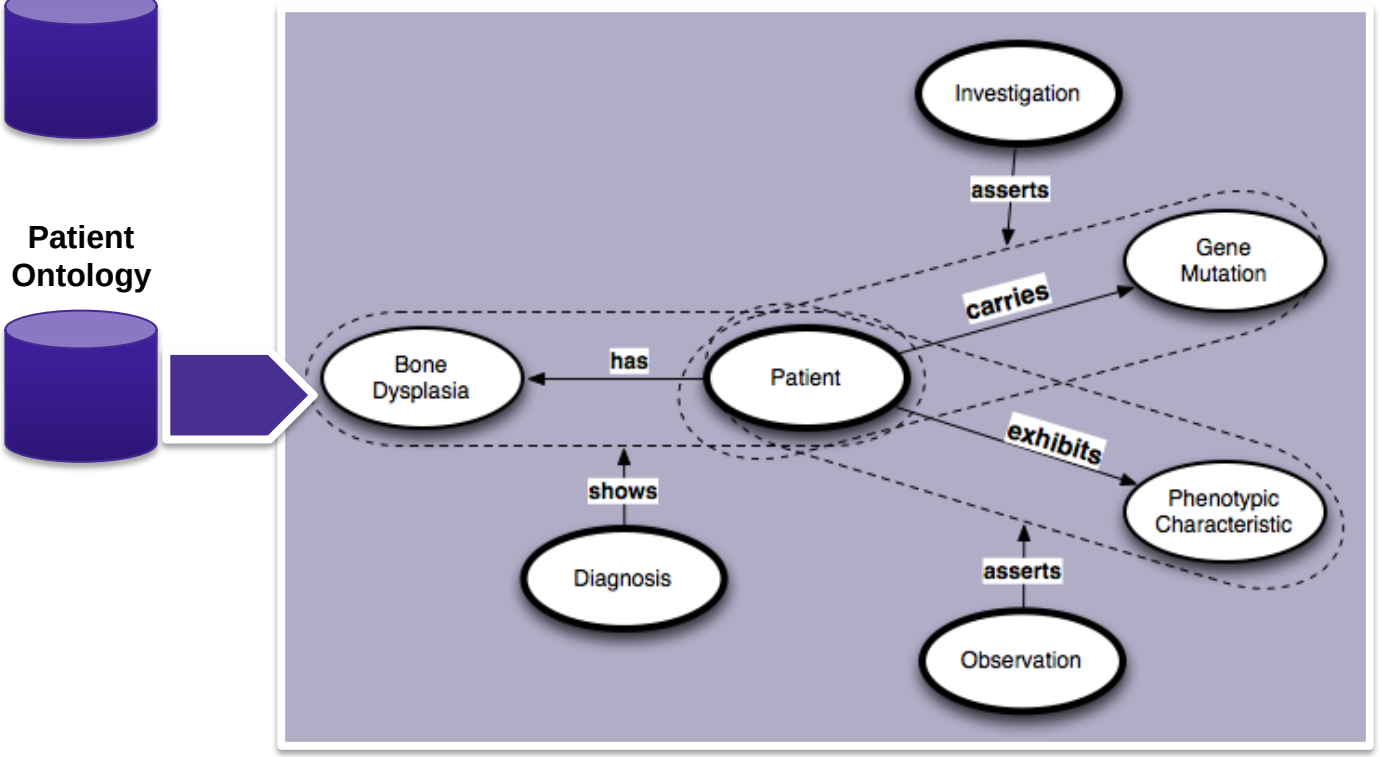
Ontology set

Bone Dysplasia Ontology

Patient Ontology



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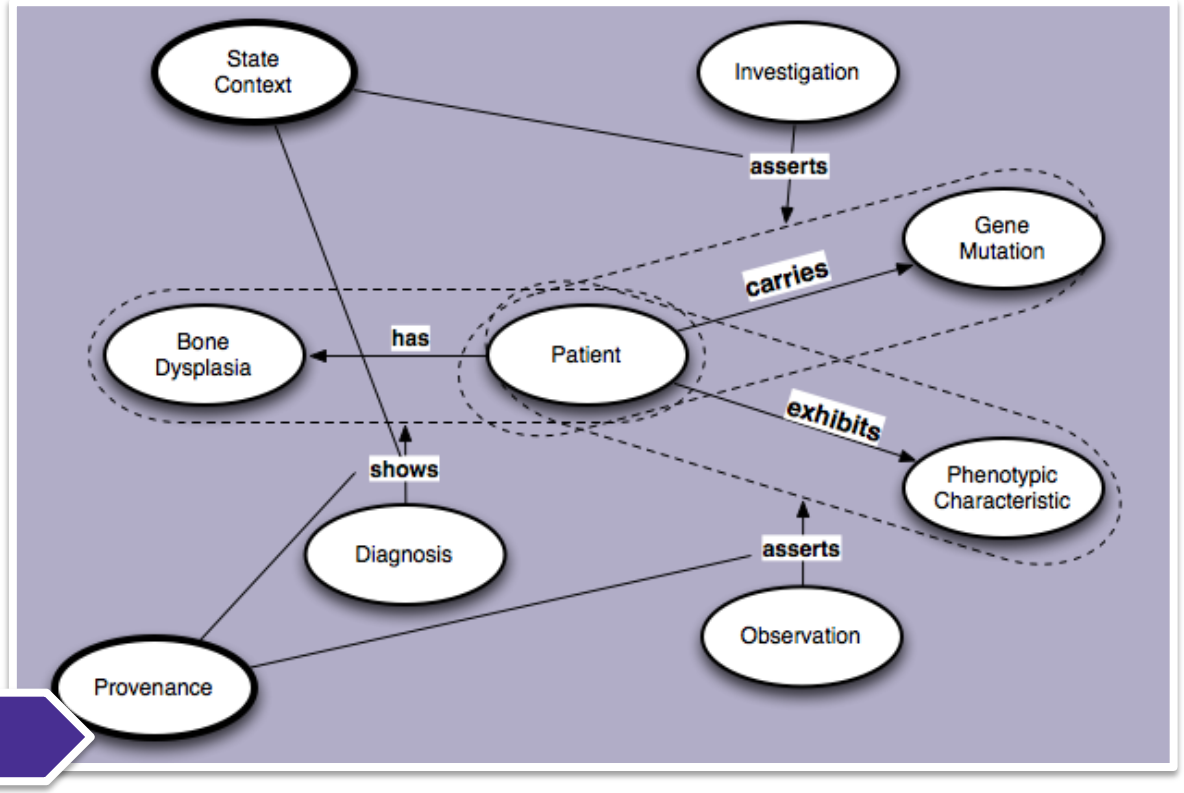
Ontology set

Bone Dysplasia Ontology

Patient Ontology

Context Ontology



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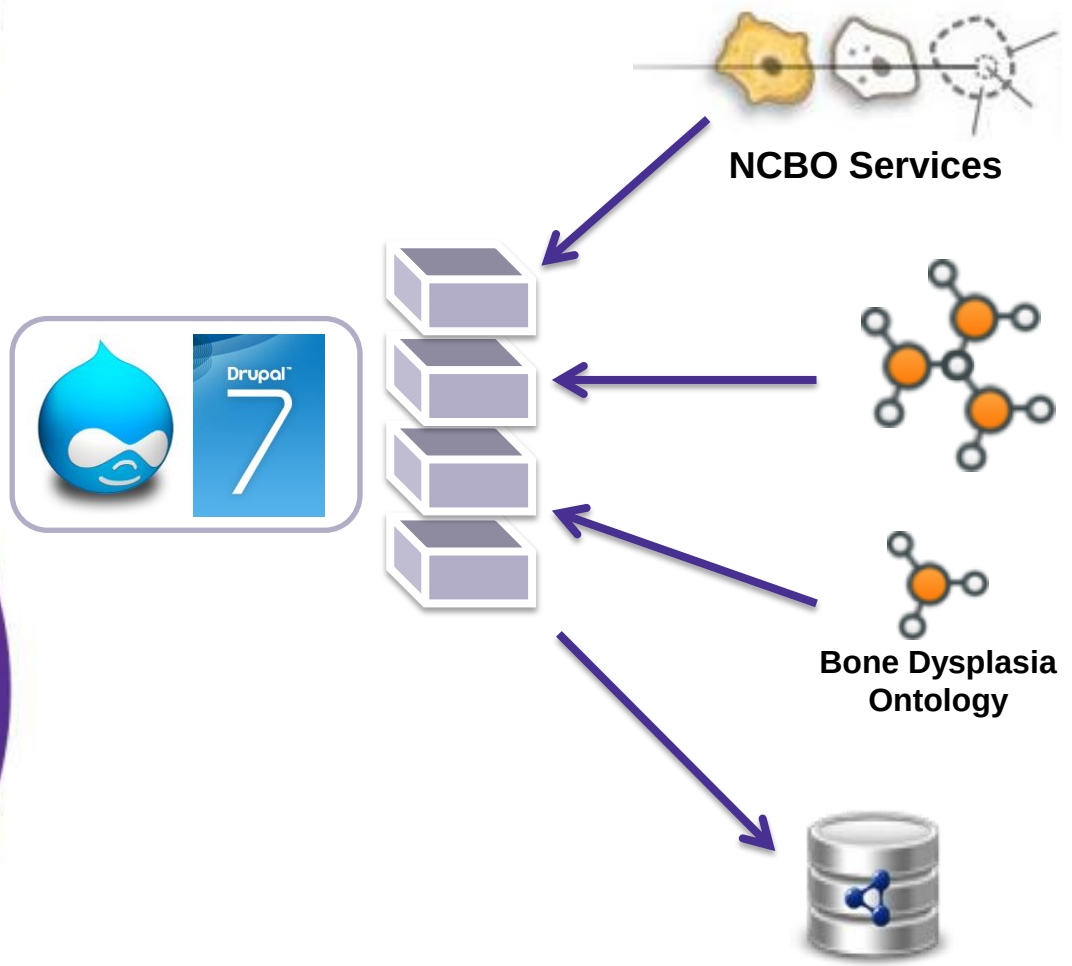
Community features

(Knowledge transfer and evolution)

A technical perspective

Contributions



- NCBO Annotator Wrapper
- Fully customizable

- Vocabulary import
- Ontology 2 taxonomy
- Modularization

- Knowledge import
- Ontology 2 content

- Ontology evolution
- RDF backend

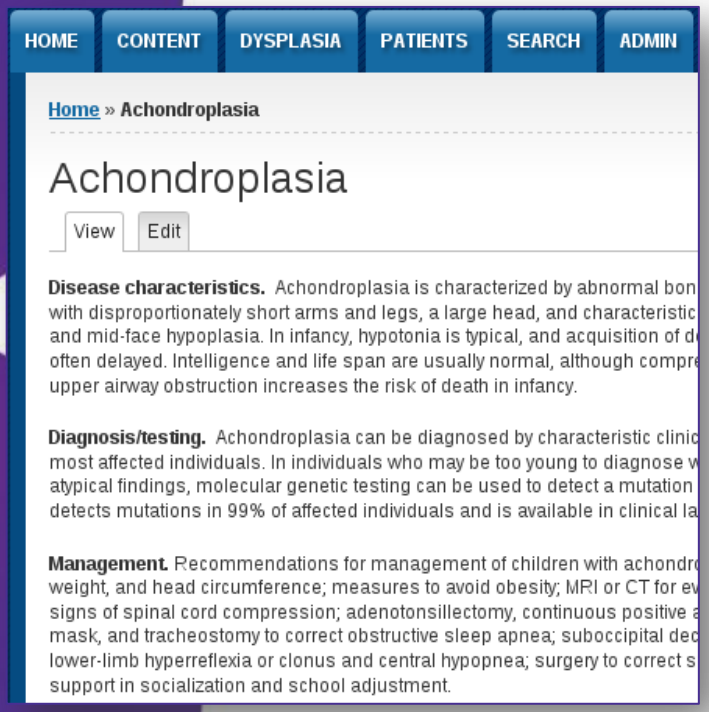
A technical perspective

RDF in Drupal 7 ++

- Core Drupal 7
- Drupal content to RDF via instances
- SPARQL support

- Start from an existing ontology
- Ontology content to Drupal
- Concept-based mapping
- Relations vs. class axioms

Collaborative knowledge curation

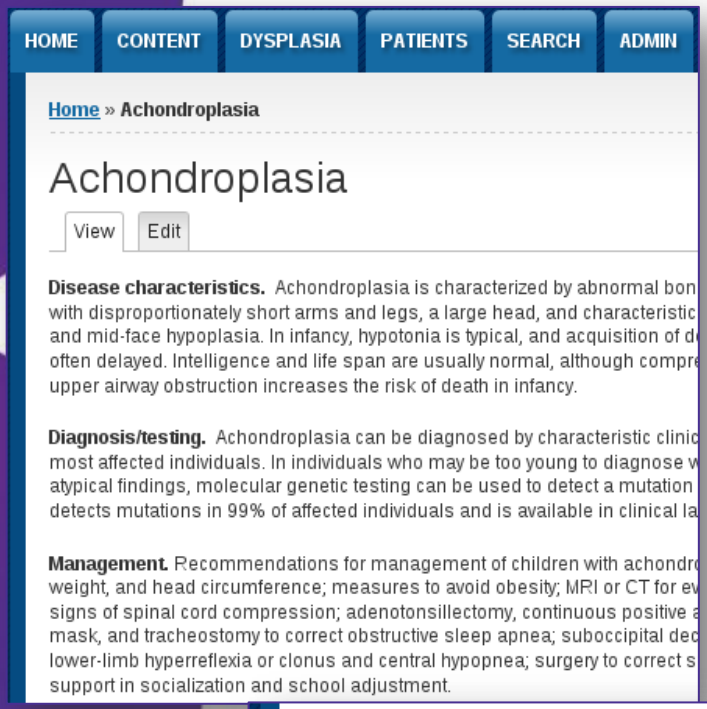


Bone Dysplasia Ontology

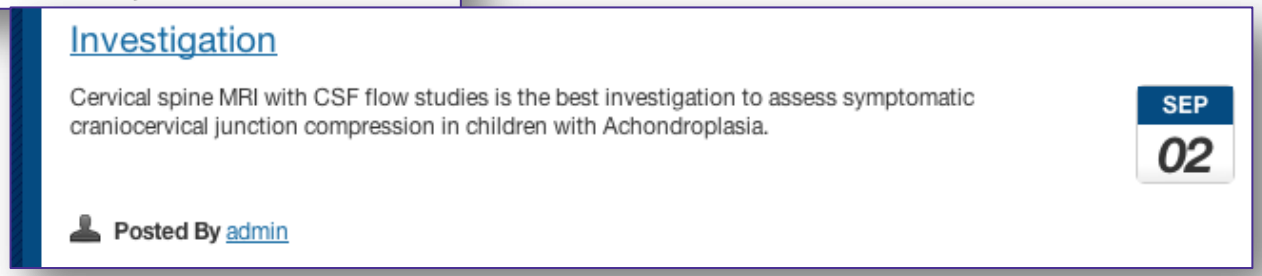
- **Wiki-style editing**

Collaborative knowledge curation

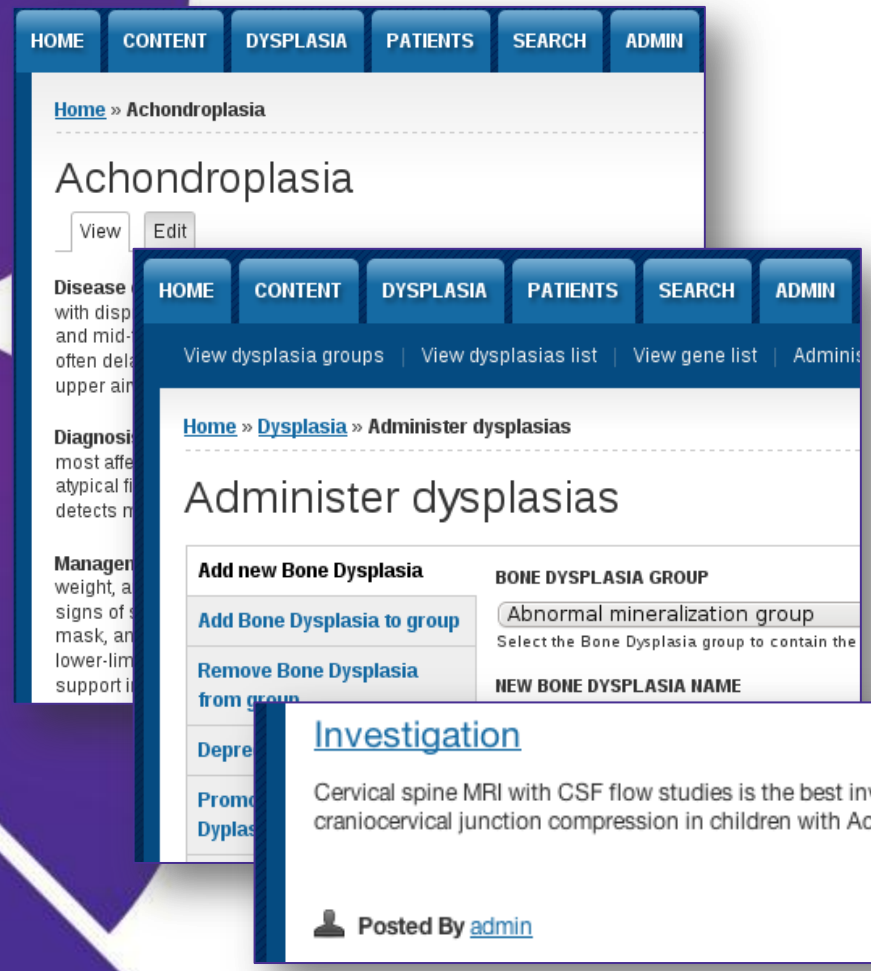
Bone Dysplasia Ontology



- **Wiki-style editing**
- **Micro-contributions**



Collaborative knowledge curation

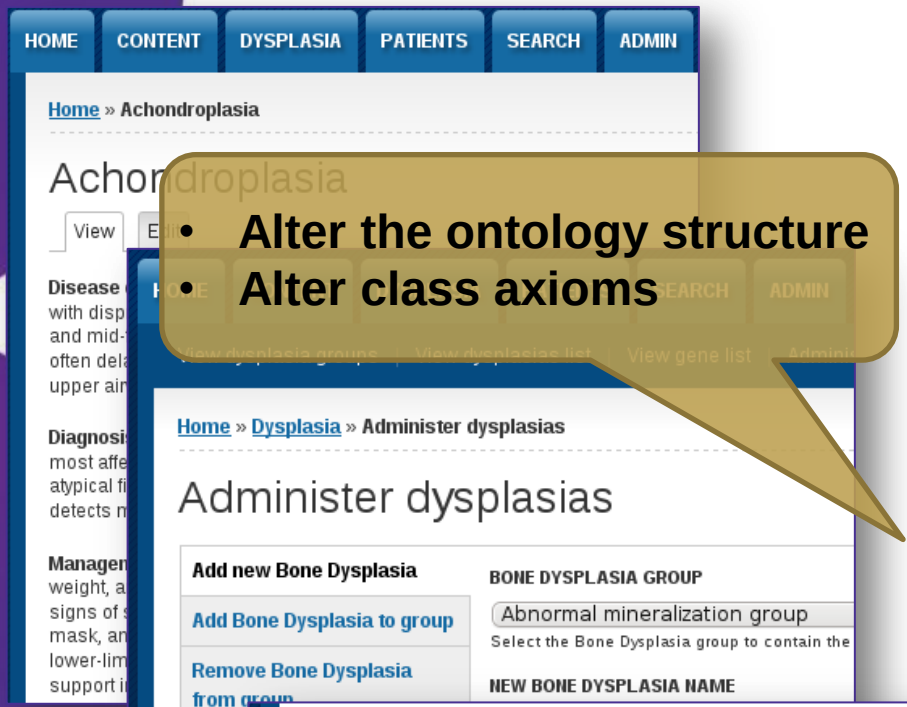


Bone Dysplasia Ontology

- **Wiki-style editing**
- **Micro-contributions**
- **Direct domain knowledge "encoding"**

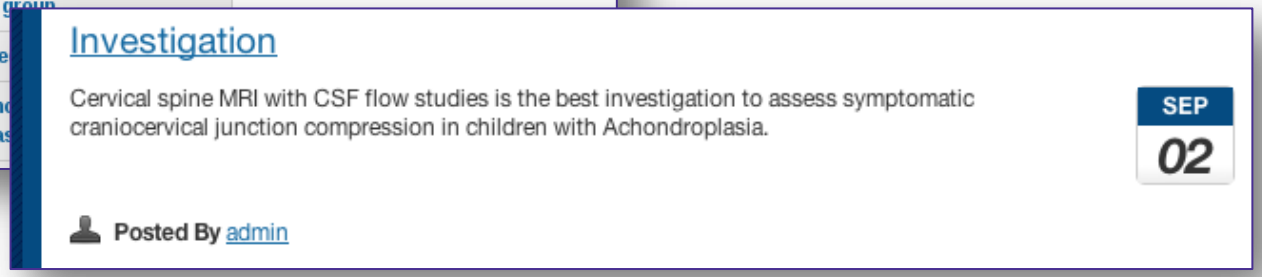
Collaborative knowledge curation

Bone Dysplasia Ontology

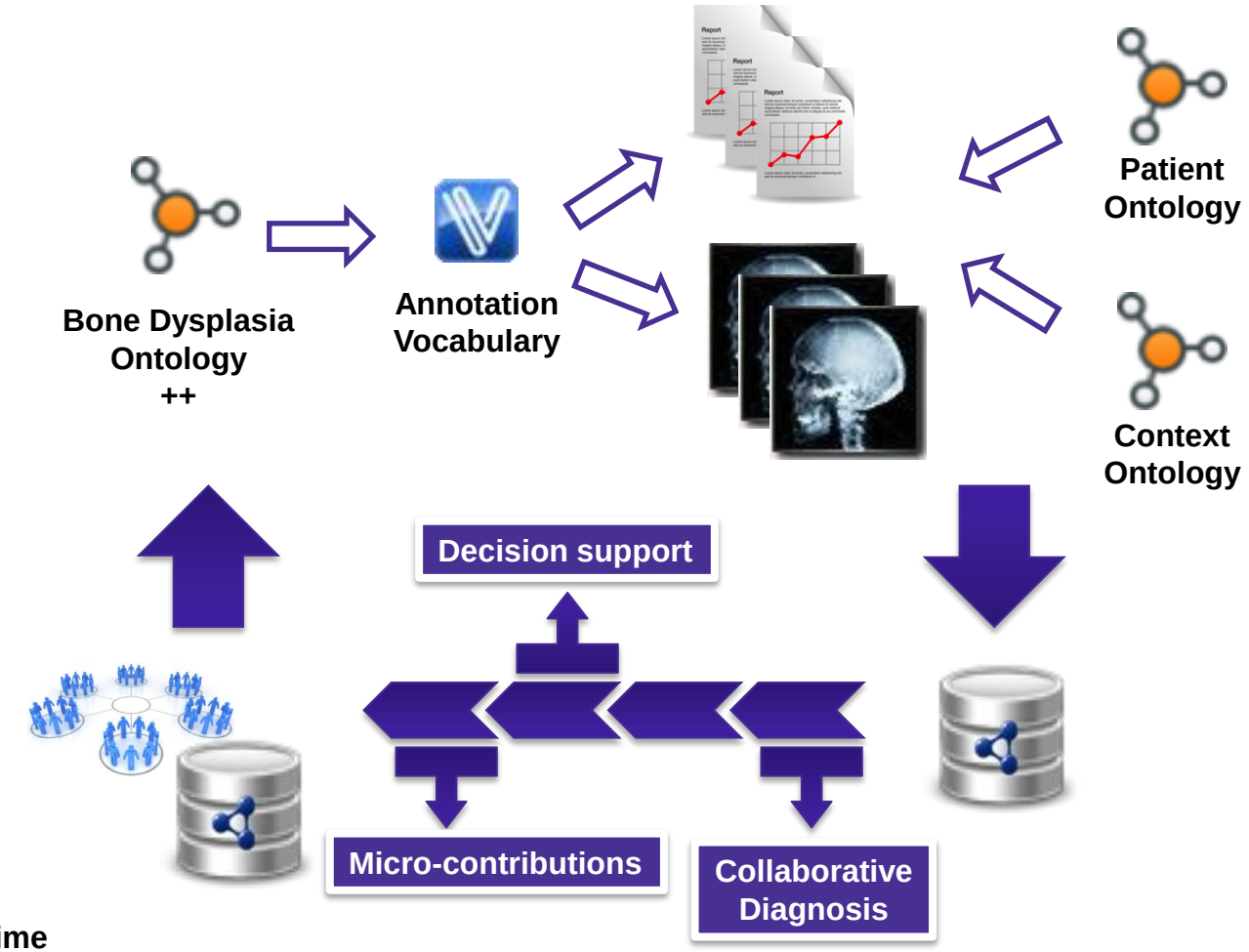


- Alter the ontology structure
- Alter class axioms

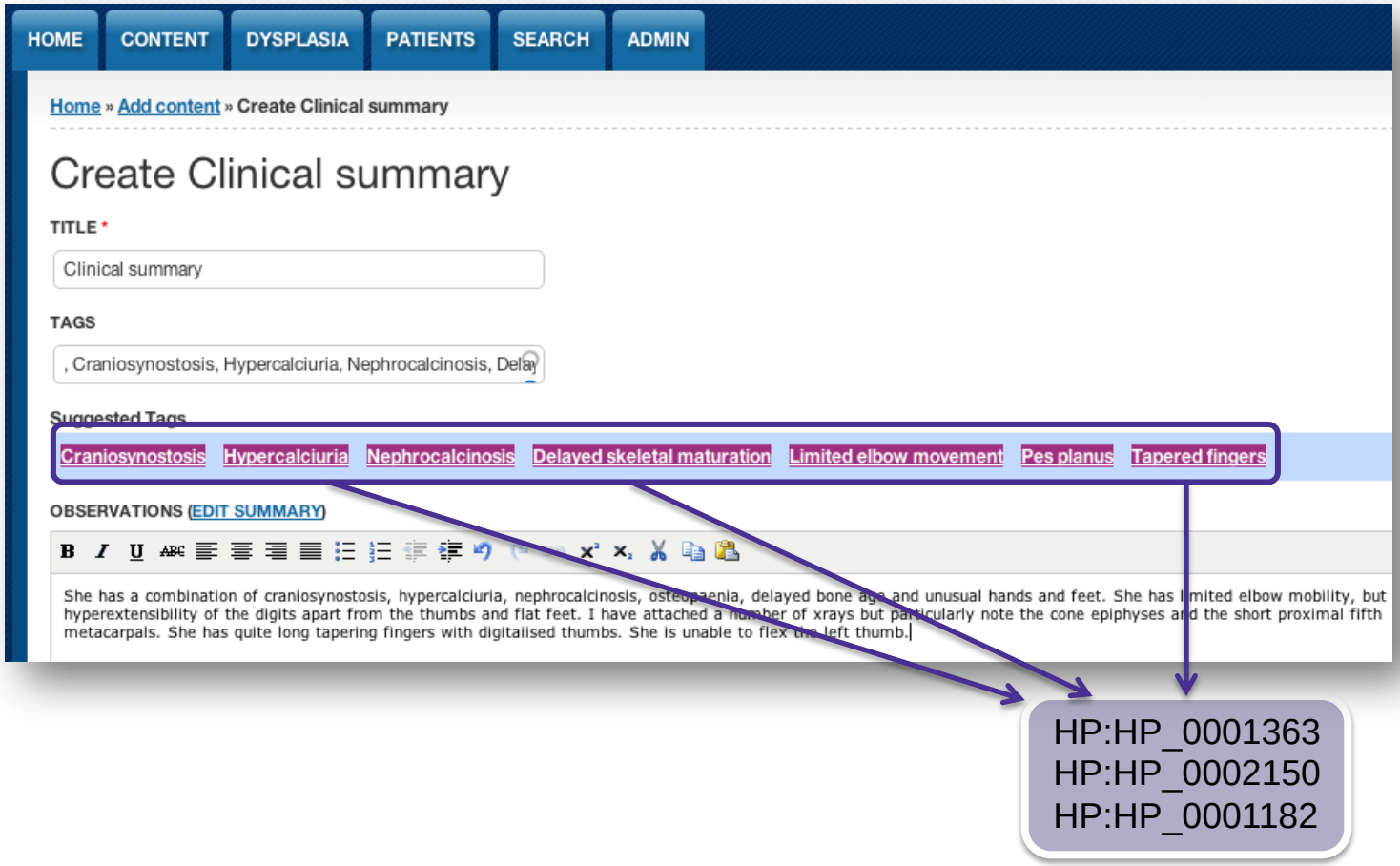
- **Wiki-style editing**
- **Micro-contributions**
- **Direct domain knowledge "encoding"**



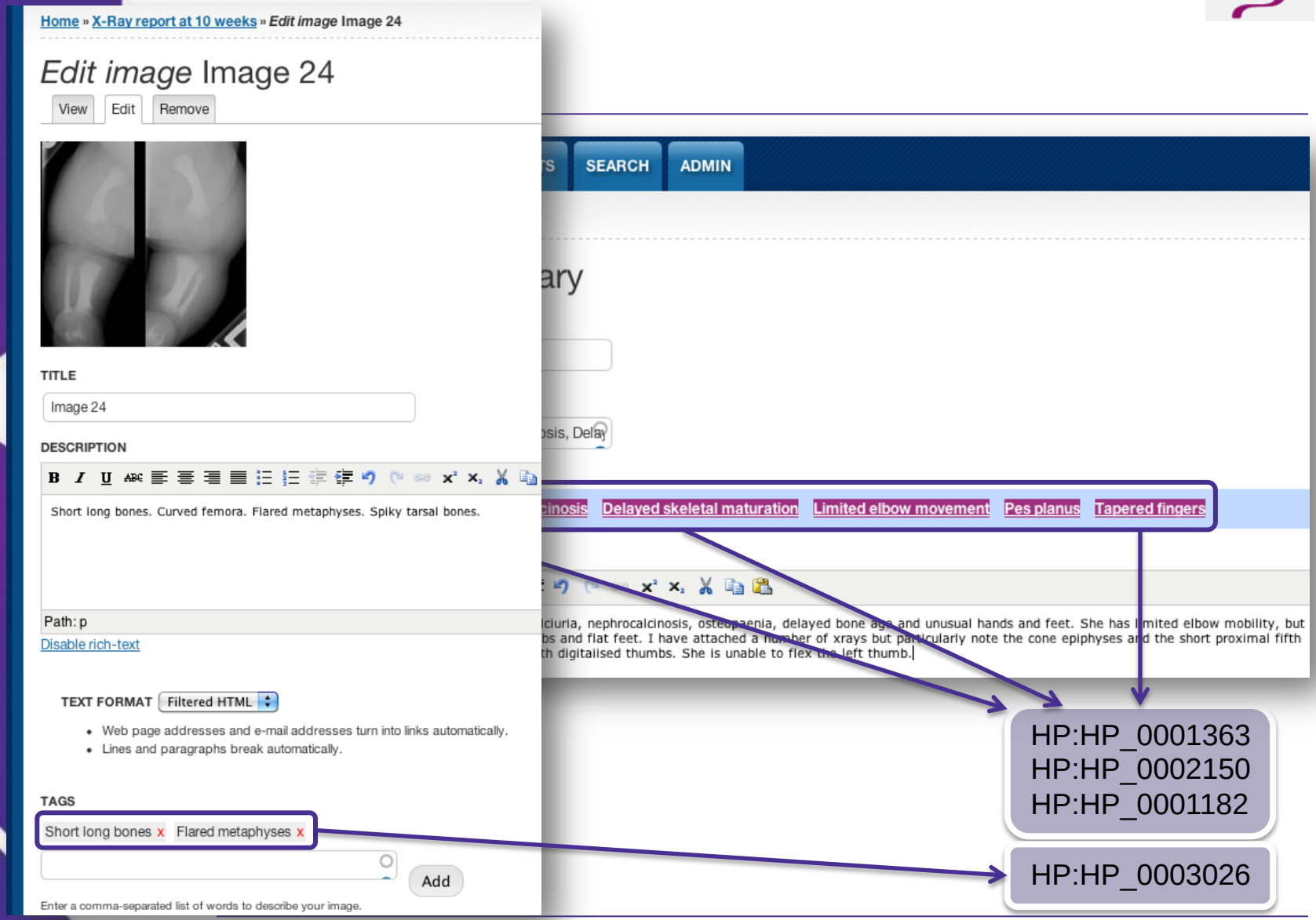
Knowledge transfer



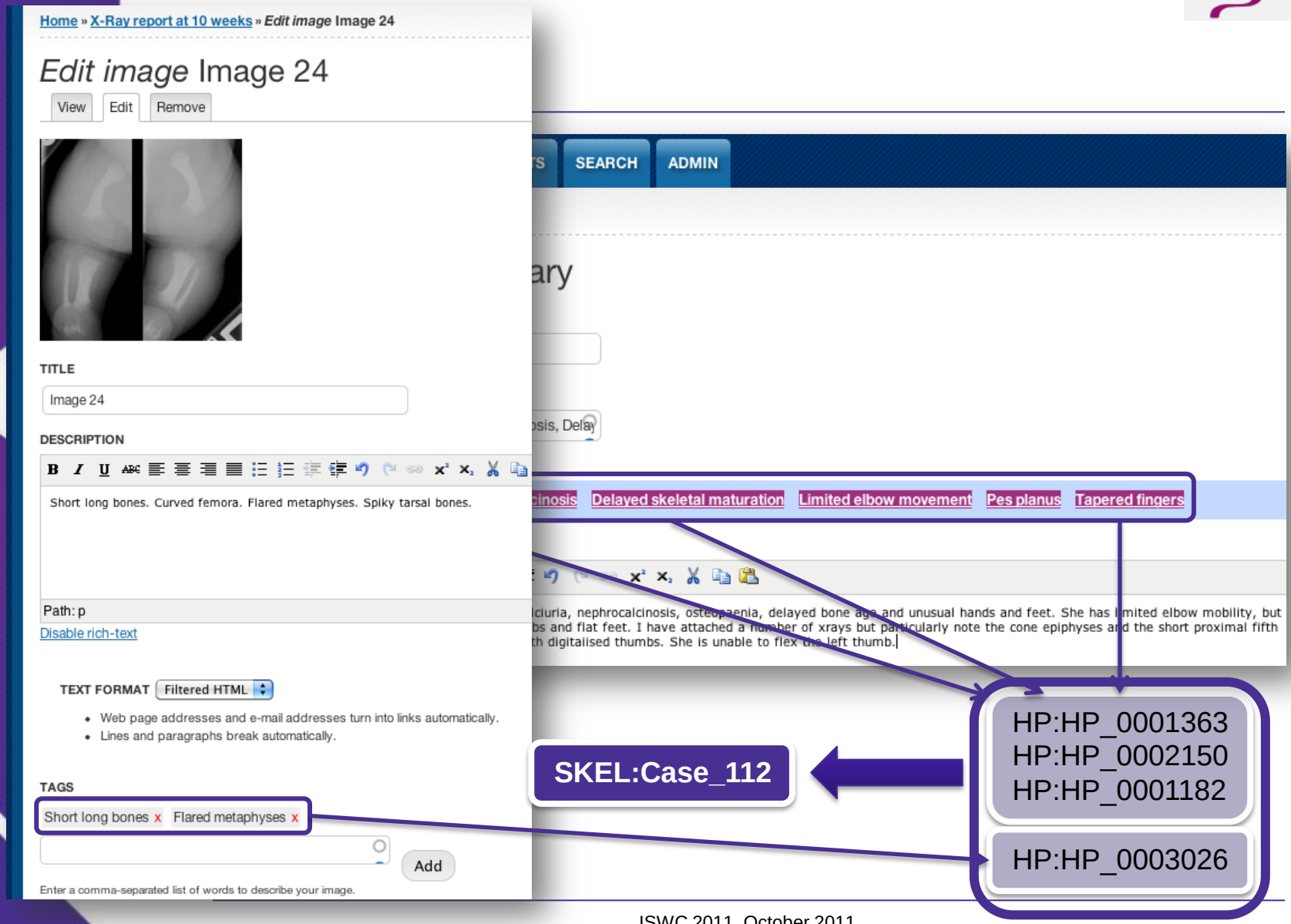
Semantic annotation



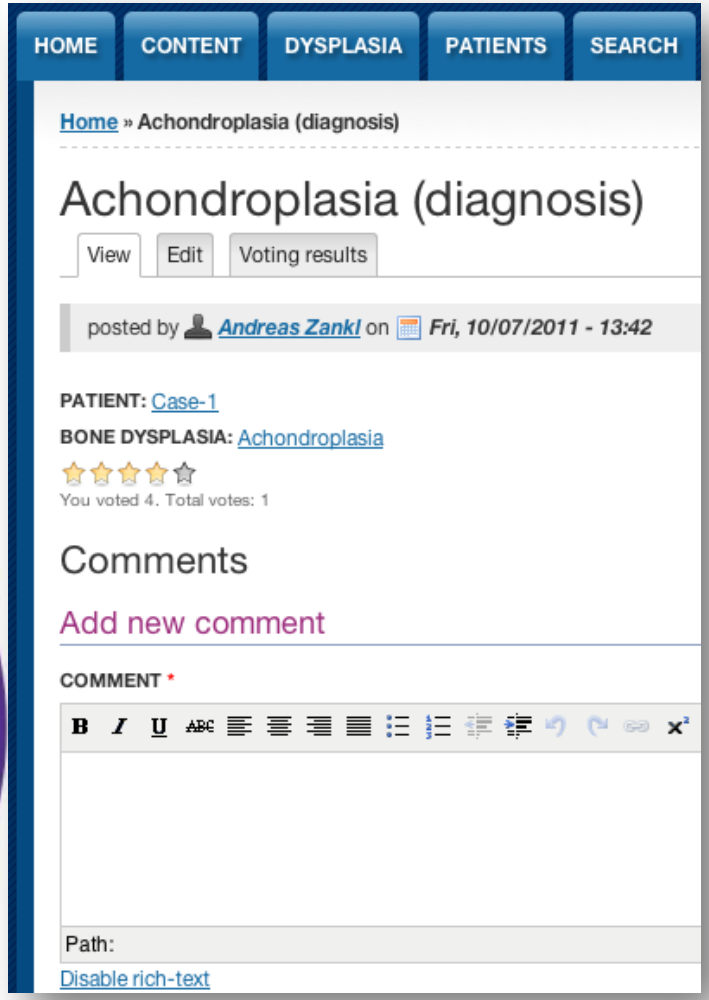
Semantic annotation



Semantic annotation

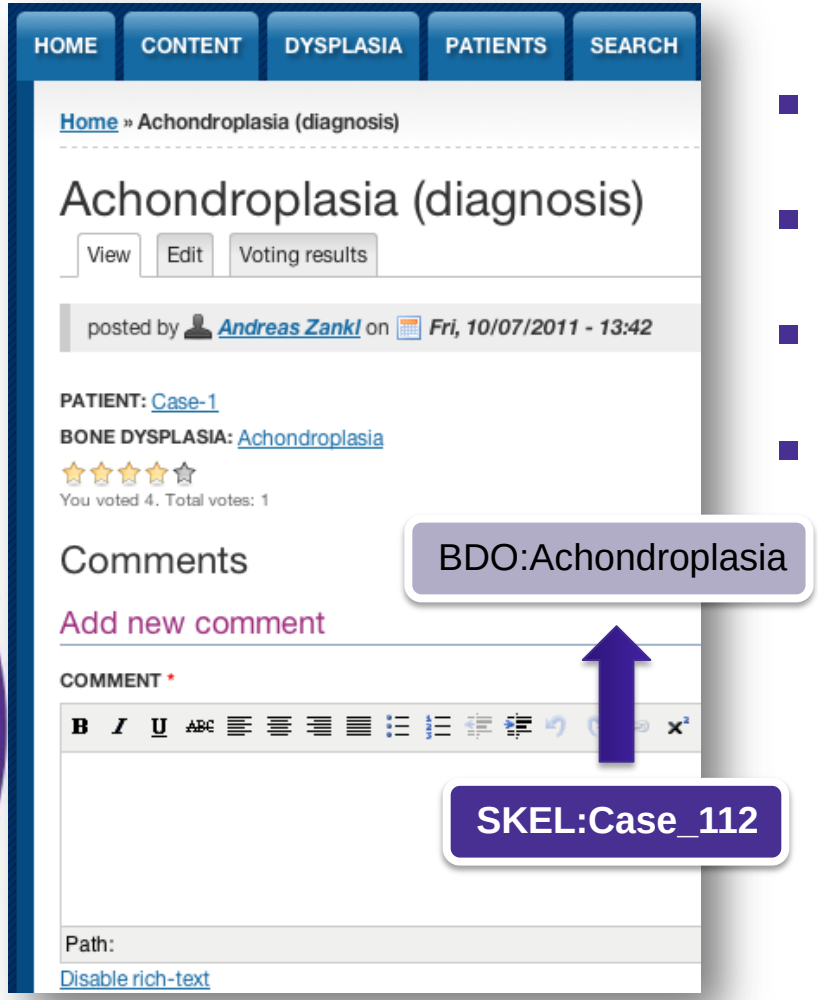


Collaborative diagnosis



- Open discussions
- 5-star rating / voting
- Multiple diagnoses
- Case owner driven

Collaborative diagnosis



- Open discussions
- 5-star rating / voting
- Multiple diagnoses
- Case owner driven

Access control

Authentication

Group-based

- **Intra-group sharing**
- **Inter-group sharing**

Access control

Authentication

Group-based

Individual and role-based

- **Sensitive data**
- **Domain knowledge**

Access control

- Authentication
- Group-based
- Individual and role-based
- Data *filtration*
  - **Patient data ➔ patient descriptions**

Decision support

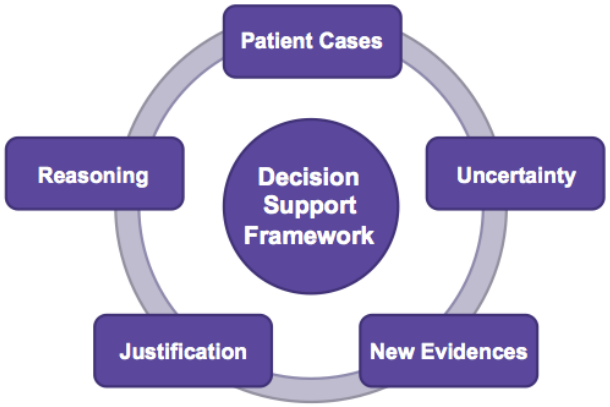
Aims

A ranked list of features given a particular disorder

A probabilistic correlation ranking of a set of features given a disorder

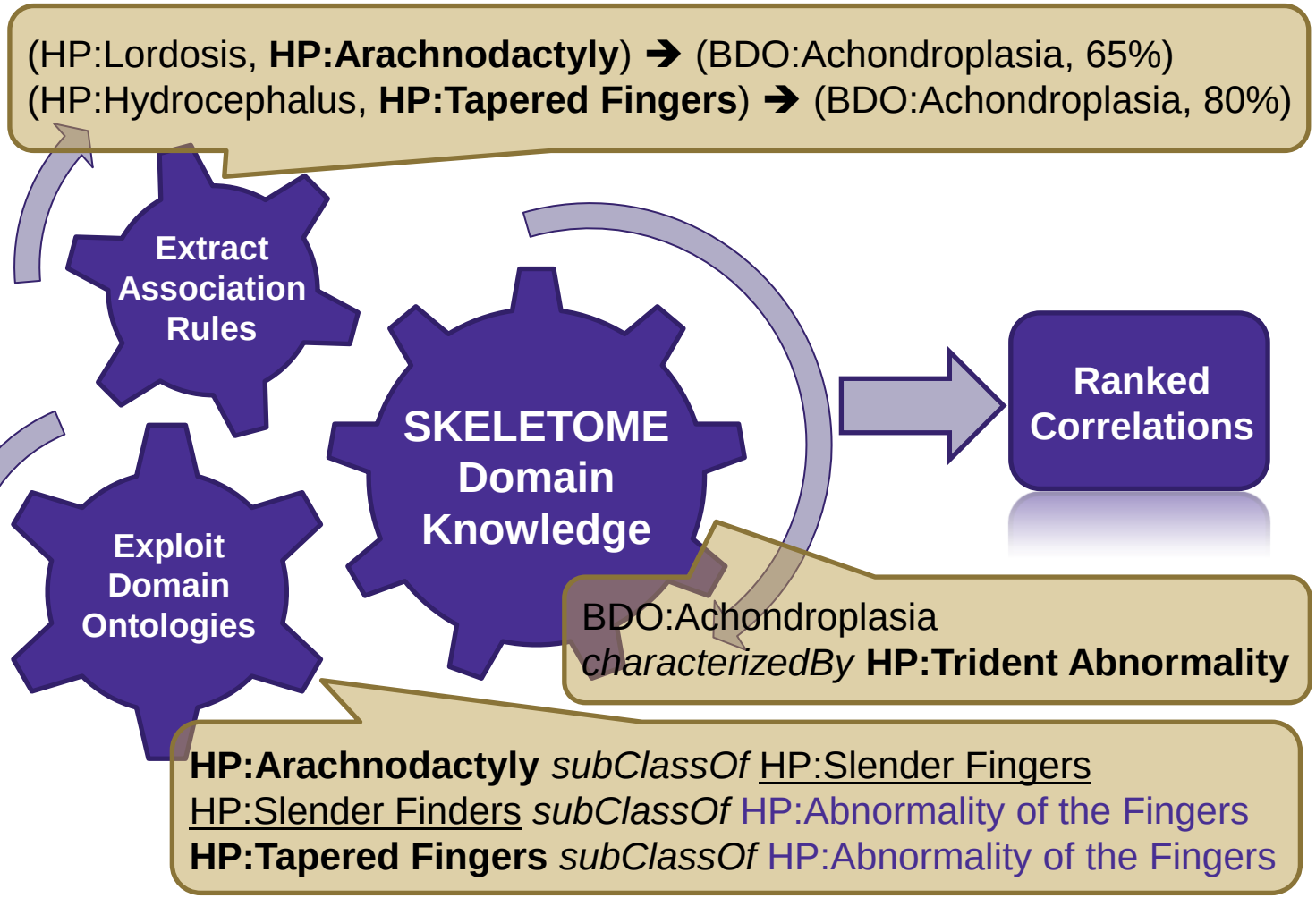
A ranked list of disorders given a particular feature

A probabilistic correlation ranking of a set of disorders given a set of features



Decision support

Overview



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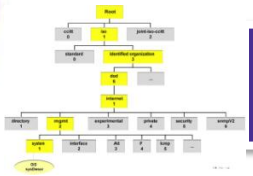
Future work

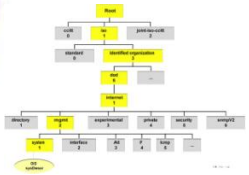
Future work

Ontologies



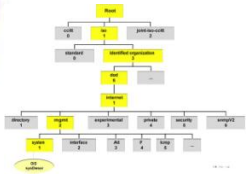
- **Bone Dysplasia Ontology**
  - Protein structures
- **Ontology population**
  - Mining genotype-phenotype associations
  - Mining phenotype-disease associations
- **Linked Data**
  - Proper publishing

Future work
Community



- **Integration with existing resources**
  - International Skeletal Dysplasia Registry
  - European Skeletal Dysplasia Network

Future work
Development



Anatomical entity
part of
Anatomical entity

Quality

Quality

Very short and tapered middle phalanges of the thumb

- **X-Ray tagging & clinical summaries annotation**
  - Mining composite tag structures
- **Advanced ontology evolution support**
  - Protégé 4 as back-end

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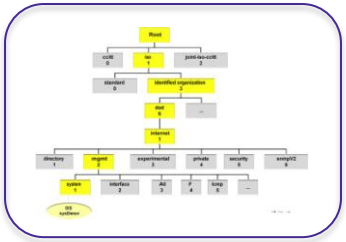
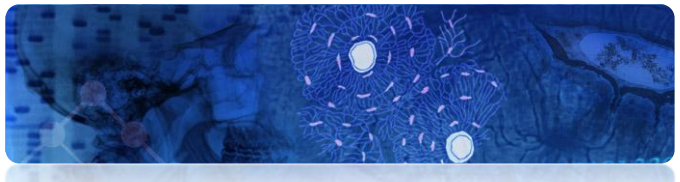
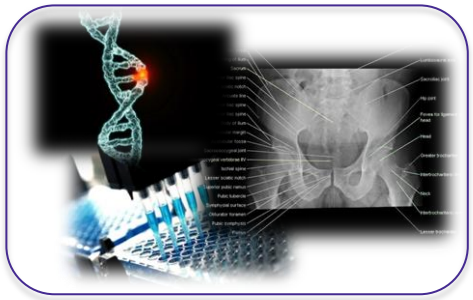
Summary

Summary

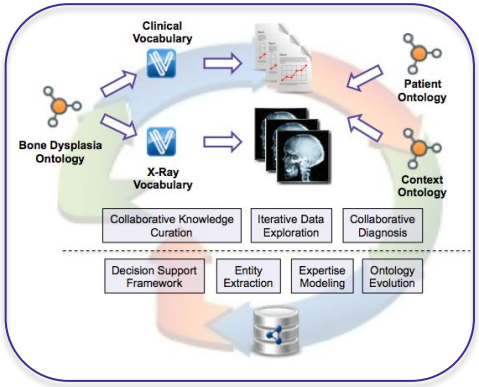


Existing domain knowledge





Collaboratively acquired patient cases
Collaborative diagnosis



Knowledge evolution
Decision support
Iterative data exploration

Thank you!