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

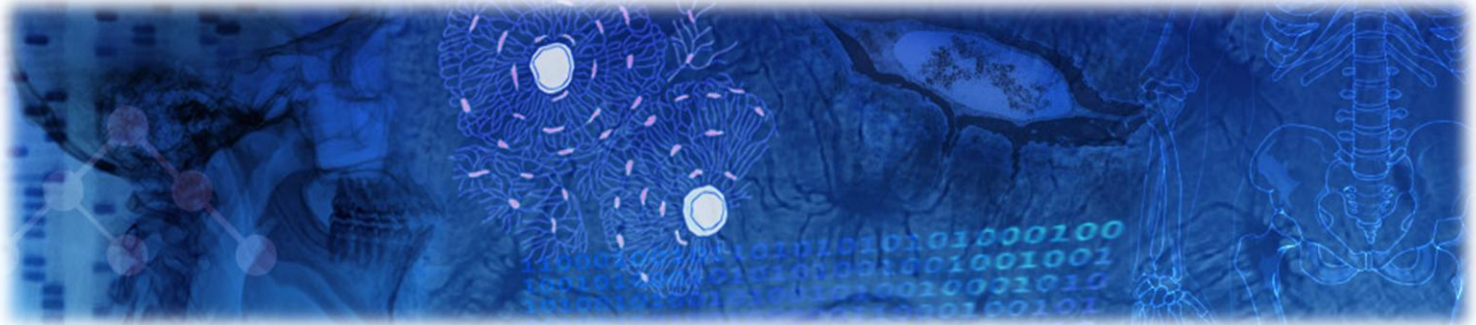
Tudor Groza, Andreas Zankl, Yuan-Fang Li and Jane Hunter

27 Oct. 2011
Bonn, Germany

Skeletal dysplasias ...



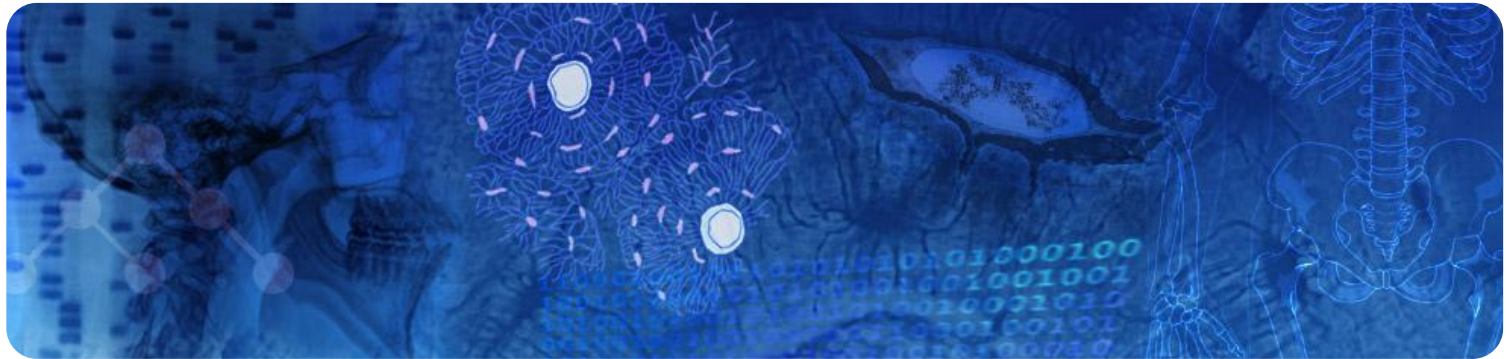
... from the knowledge representation,
curation and discovery perspectives ...



- Challenges & Requirements
- The SKELETOME Platform
- Next steps

Skeletal dysplasias

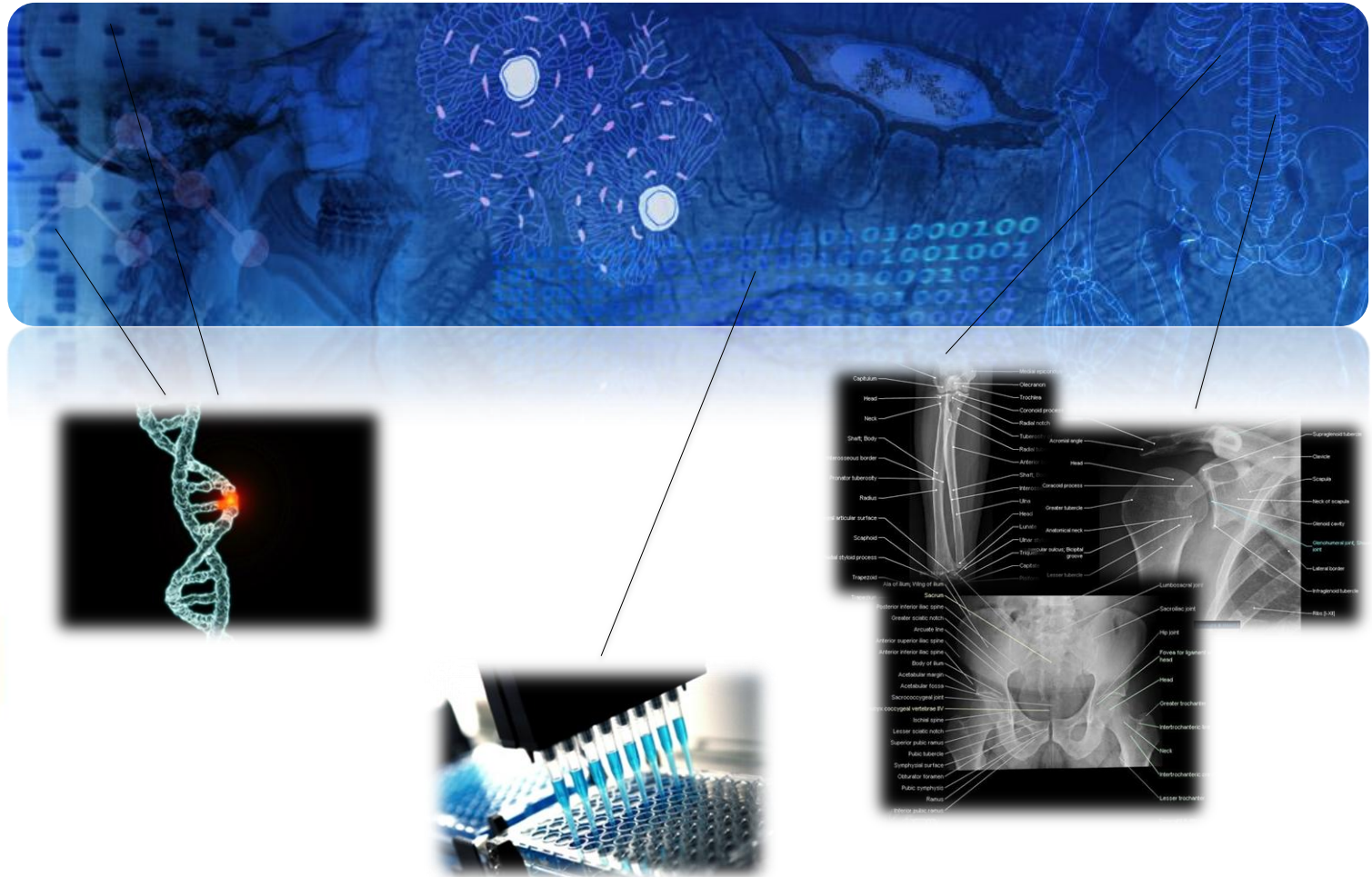
Background



- 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

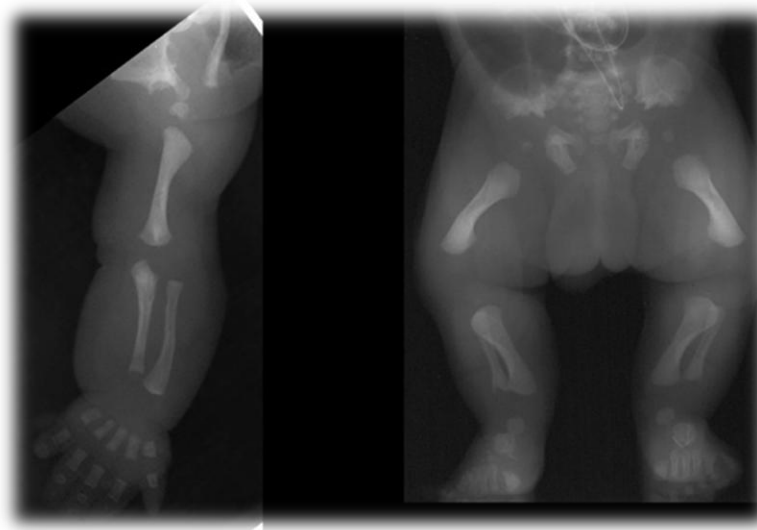
Heterogeneity

Integration



Challenges / Requirements

Common terminology



Gorlin/Hall

Postaxial polydactyly of the hands and feet

Short long bones with **smooth** rounded metaphyses

‘Drumstick’ appearance of the radius and **ulna**

ISDR/Lachman

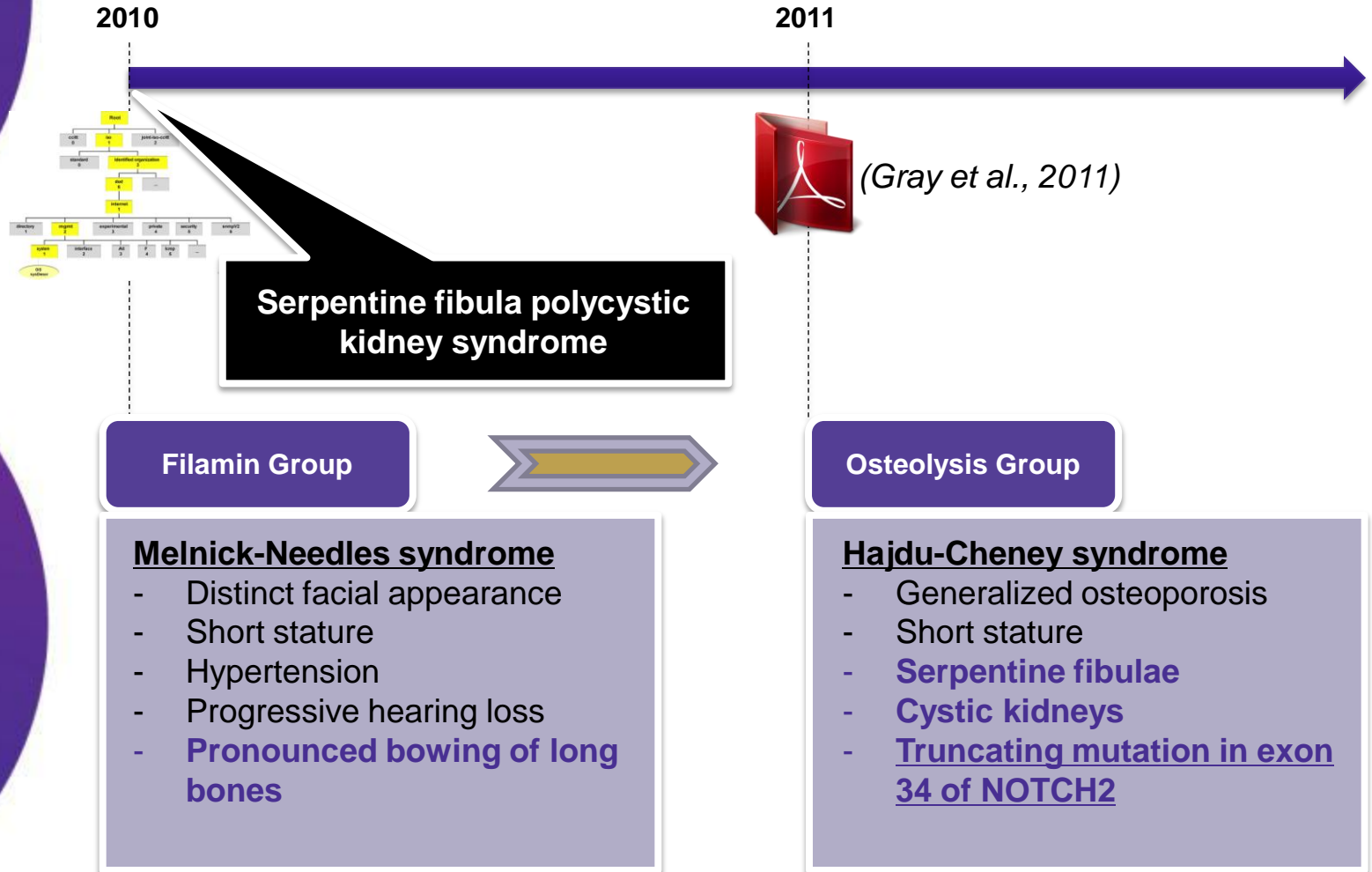
Feet – Polydactyly – postaxial
– **bilateral**

Femur – Metaphysis Proximal – rounded
Femur – Metaphysis distal – rounded

Radius – Metaphysis (Distal) – widened

Challenges / Requirements

Knowledge transfer and evolution





SKELETOME

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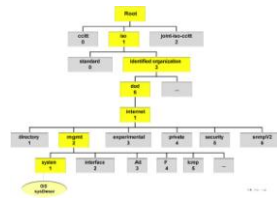
Overview



- **Ontology-based ...**
- **... community-driven knowledge curation platform**
- **Domain knowledge**
- **Patient repository**

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Aims



Knowledge / Data Integration

Common terminology

Decision Support

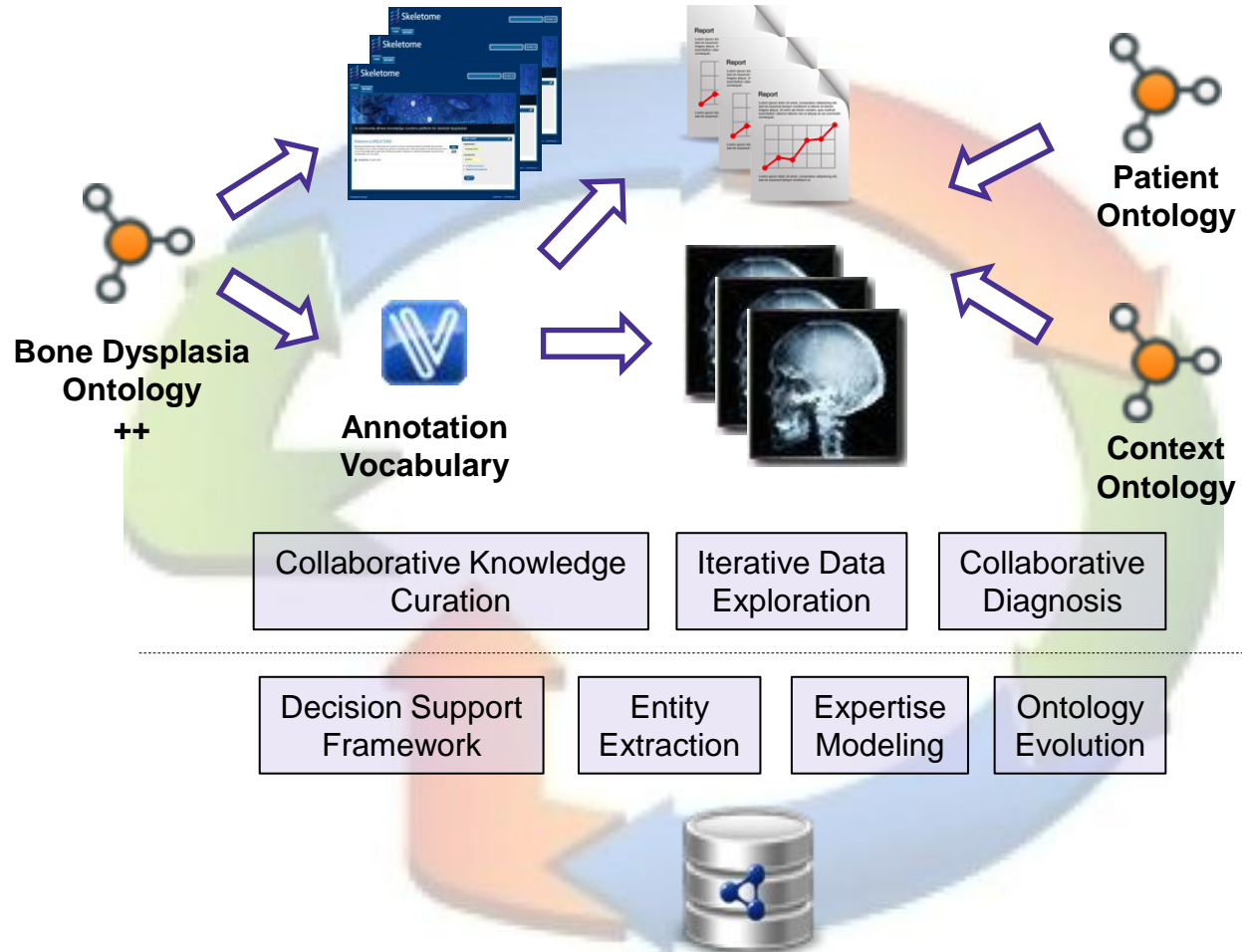
**Knowledge transfer and
sustained knowledge evolution**

Privacy and access control

Provenance and expertise

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Knowledge engineering cycle



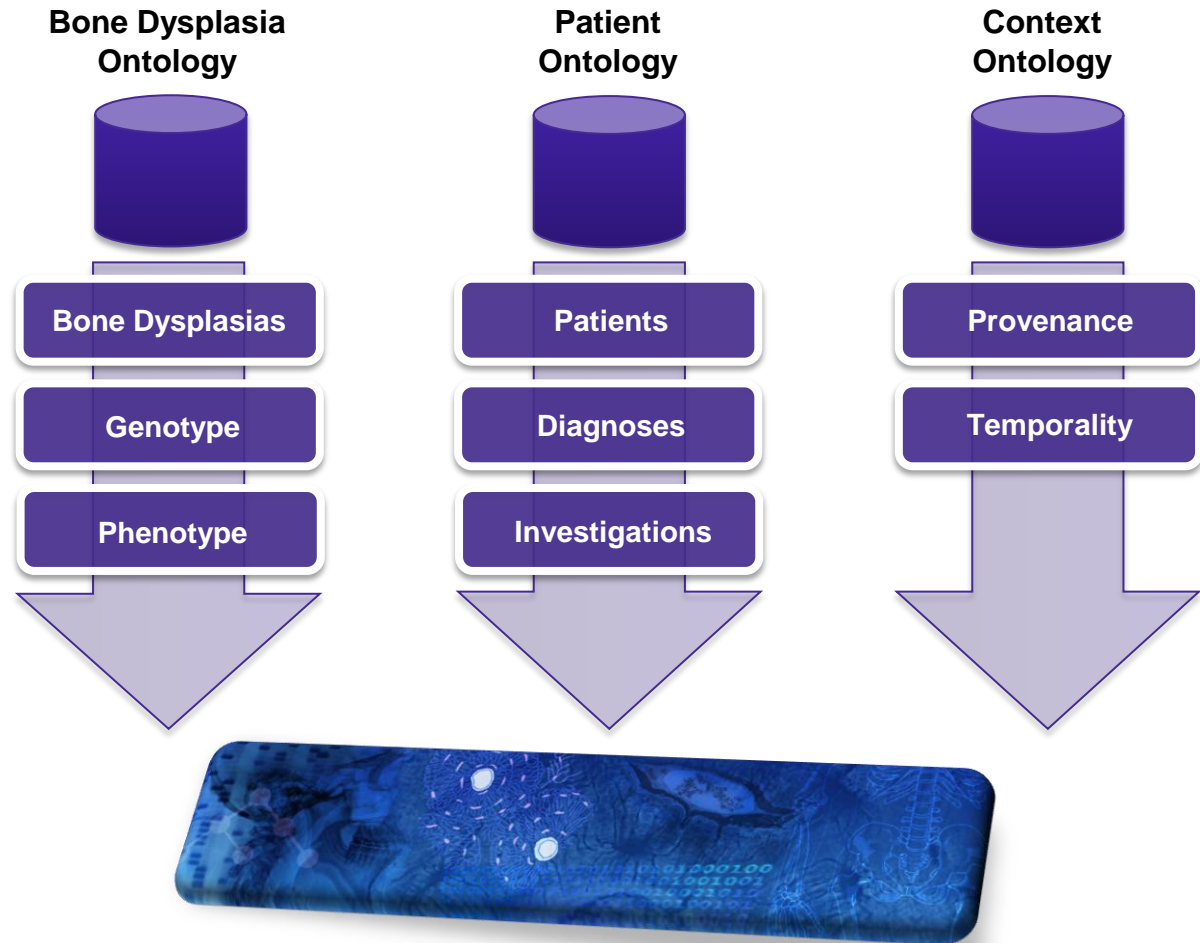
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Ontology set

(Common terminology & Knowledge / Data
integration)

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Ontology set

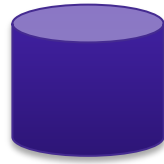


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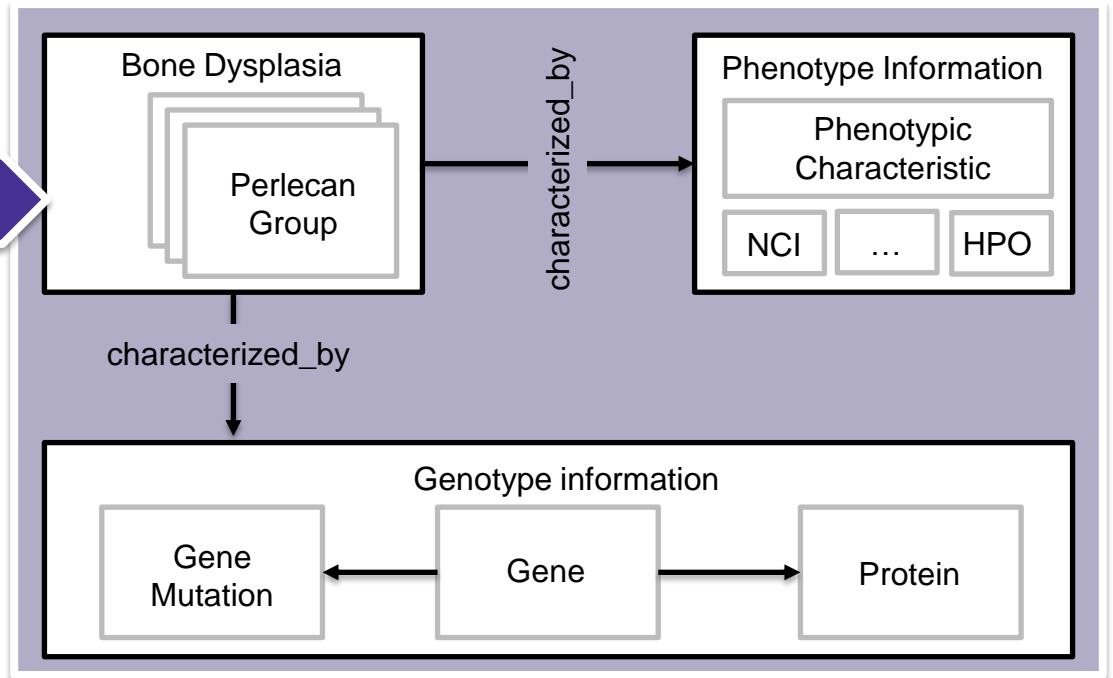
Ontology set



Bone Dysplasia
Ontology



1,200+
concepts

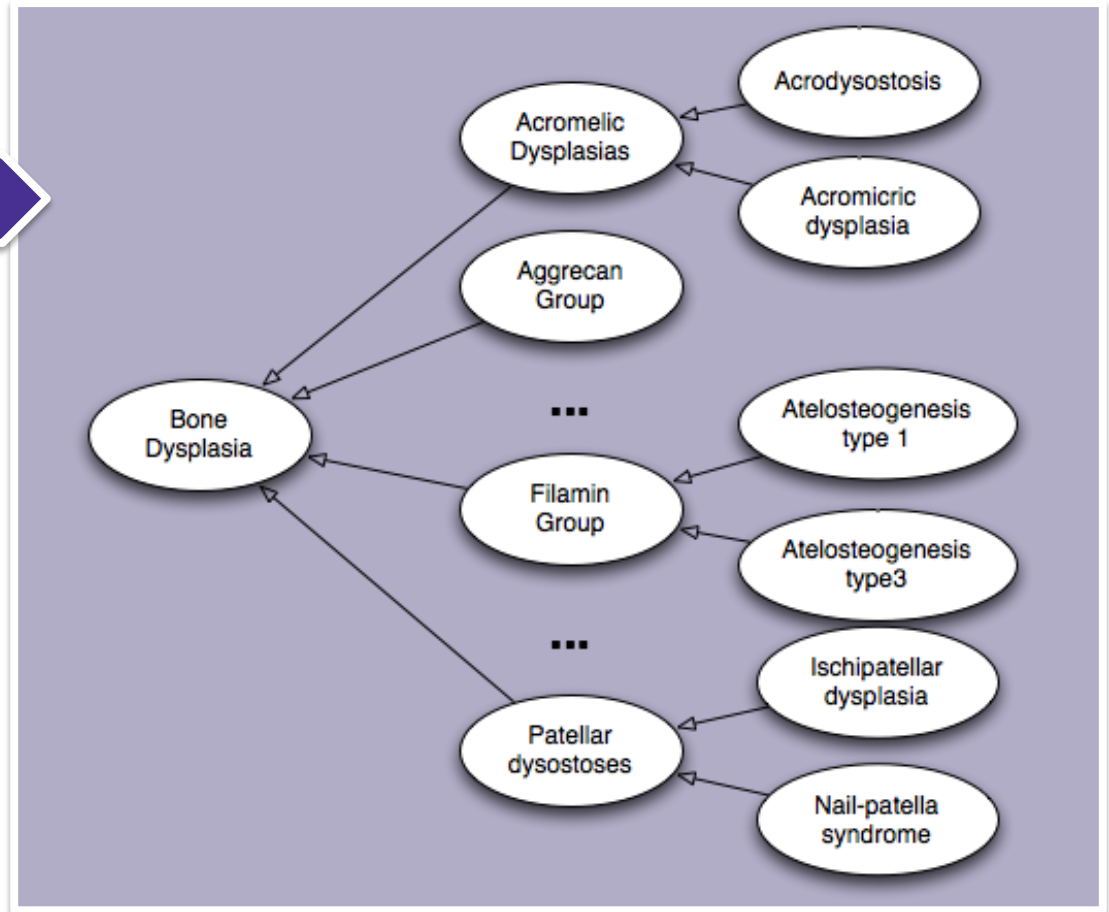
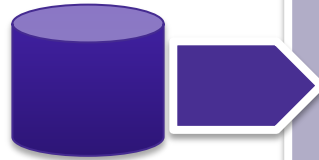


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Ontology set



Bone Dysplasia
Ontology

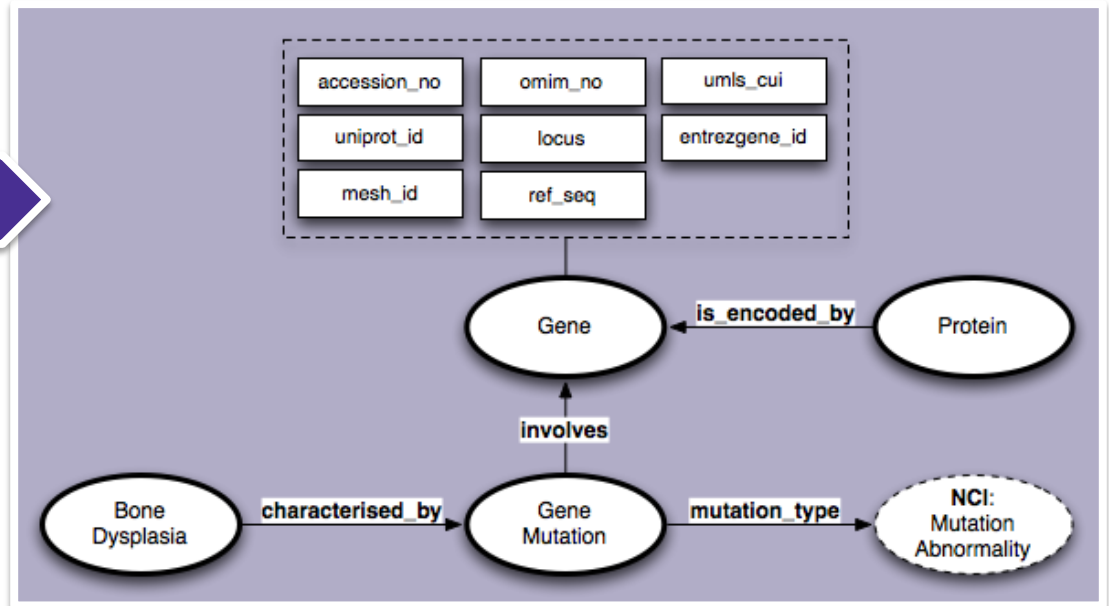
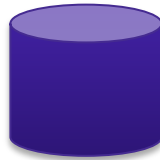


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Ontology set



Bone Dysplasia
Ontology

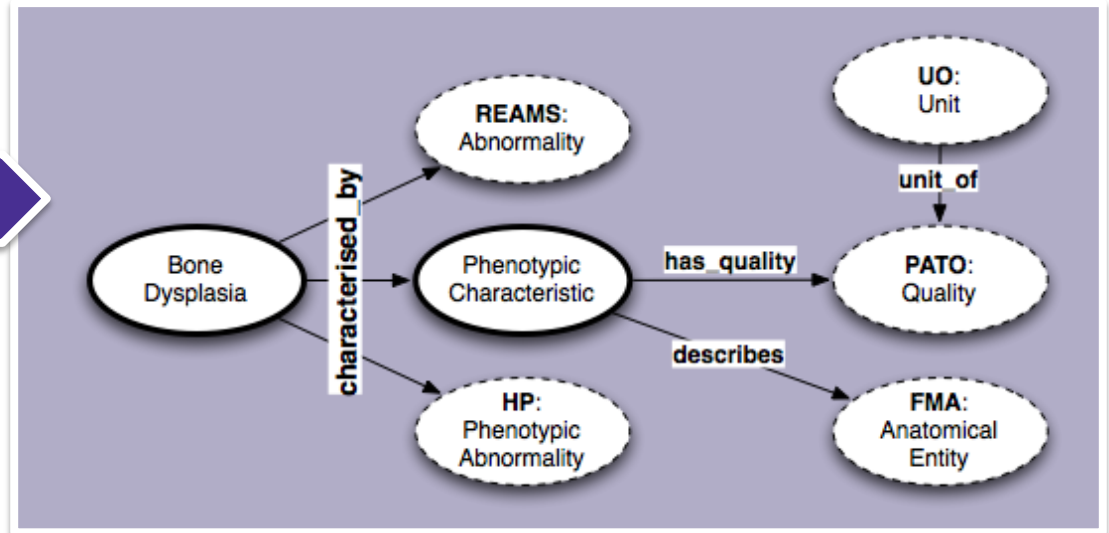
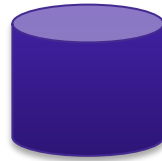


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Ontology set



Bone Dysplasia
Ontology

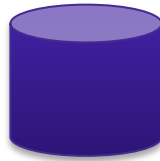


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Ontology set



Bone Dysplasia
Ontology



Class: **Achondroplasia**

SubClassOf:

characterized_by only (**GM000001** or **GM000361** or
HP_0000238 or **HP_0002938**)

SubClassOf:

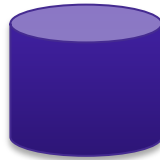
mode_of_inheritance only **HP_0000006** and
mode_of_inheritance some **HP_0000006**

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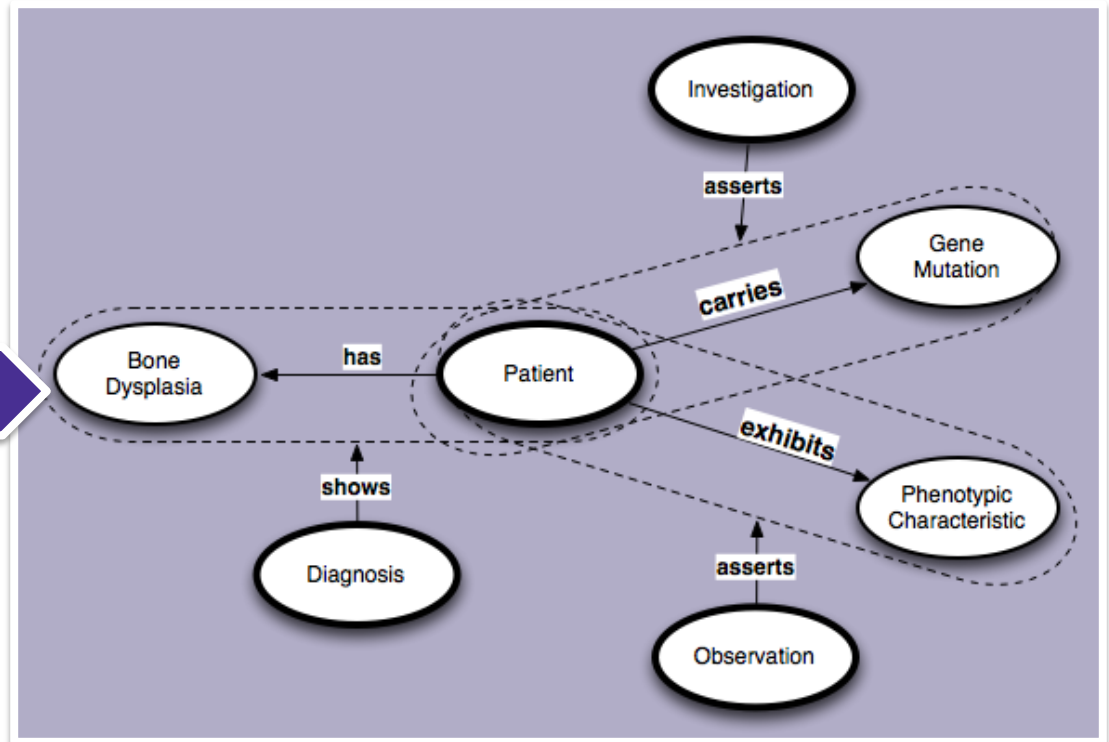
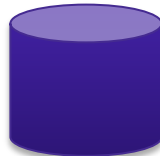
Ontology set



Bone Dysplasia
Ontology



Patient
Ontology

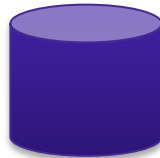


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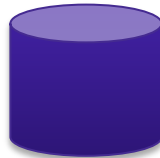
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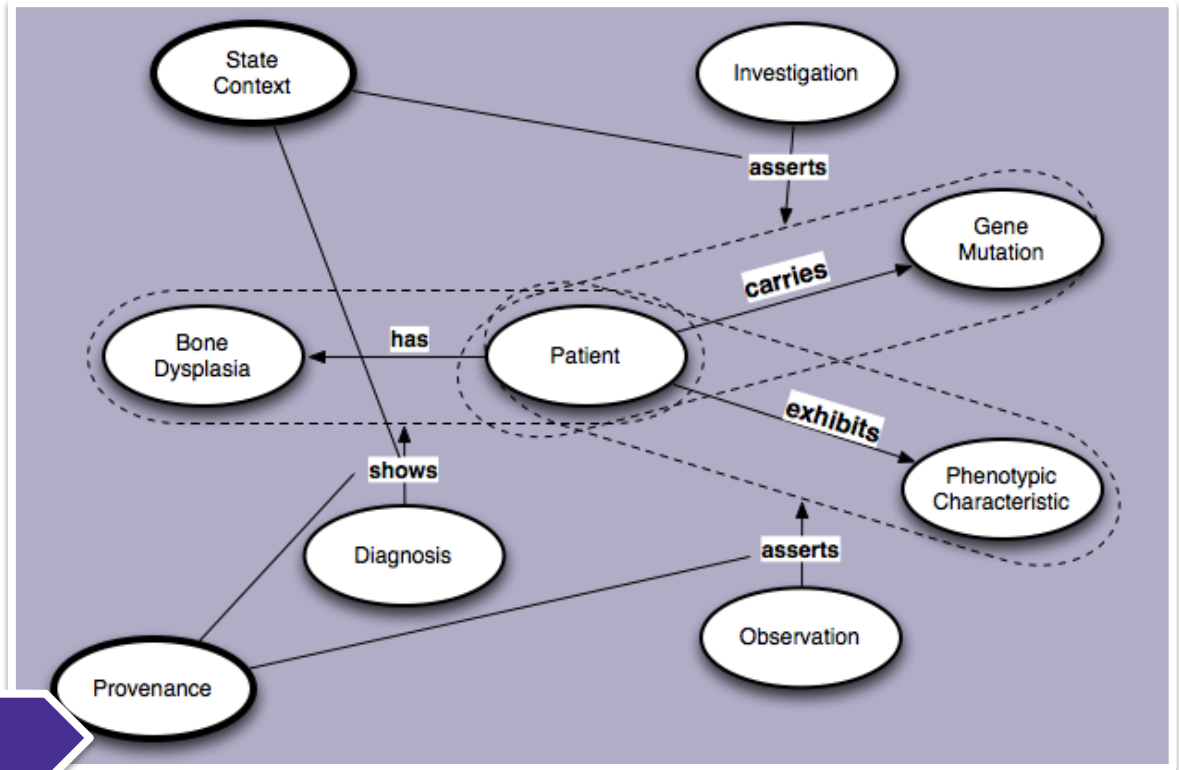
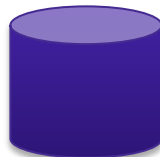
Bone Dysplasia Ontology



Patient Ontology



Context Ontology





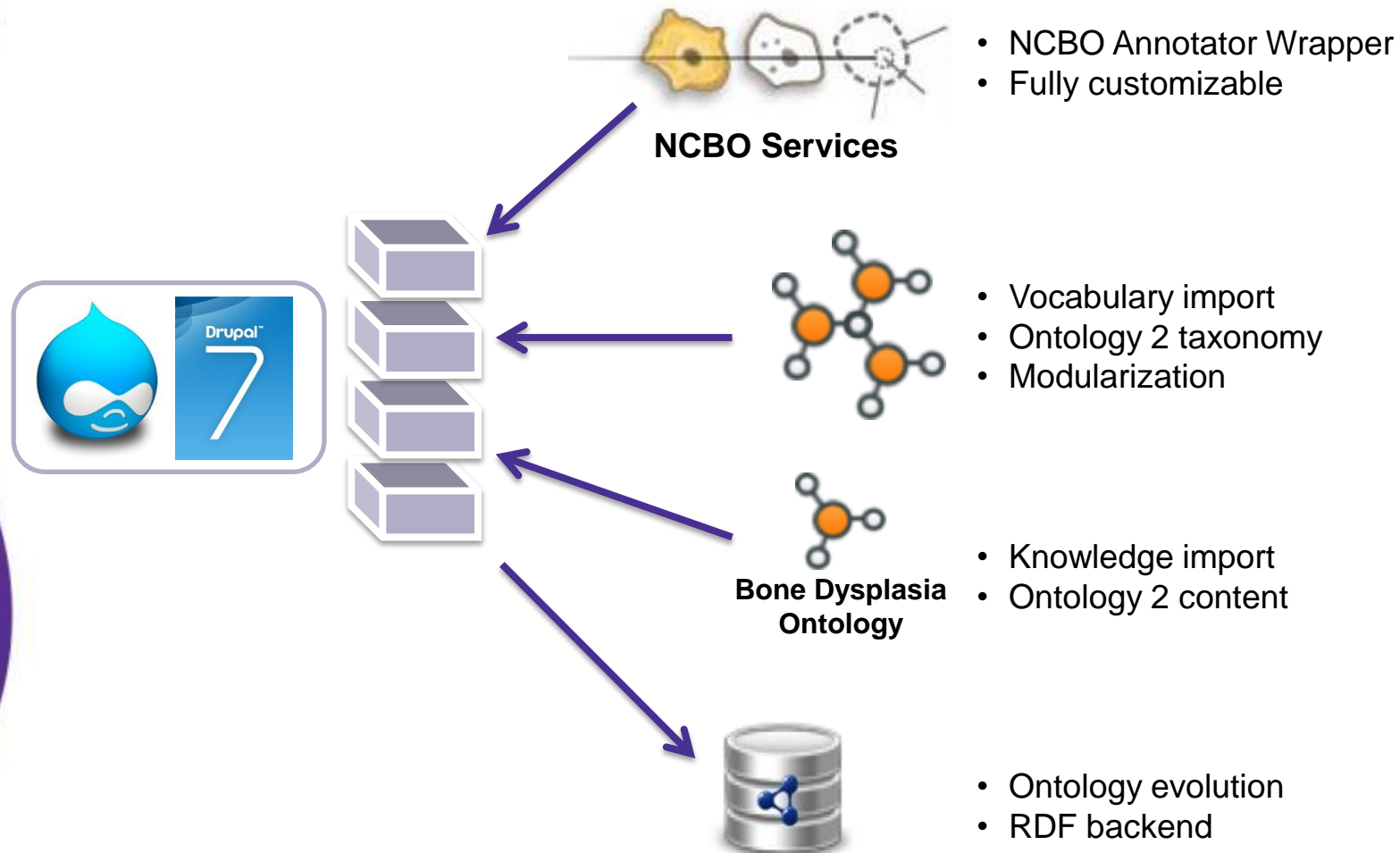
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Community features

(Knowledge transfer and evolution)

A technical perspective

Contributions



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



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Achondroplasia

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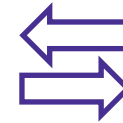
Disease characteristics. Achondroplasia is characterized by abnormal bone growth with disproportionately short arms and legs, a large head, and characteristic facial features including mid-face hypoplasia. In infancy, hypotonia is typical, and acquisition of developmental milestones is often delayed. Intelligence and life span are usually normal, although compromised. In severe cases, upper airway obstruction increases the risk of death in infancy.

Diagnosis/testing. Achondroplasia can be diagnosed by characteristic clinical findings. In individuals who may be too young to diagnose with clinical findings, molecular genetic testing can be used to detect a mutation. Current testing detects mutations in 99% of affected individuals and is available in clinical laboratories.

Management. Recommendations for management of children with achondroplasia include monitoring weight, and head circumference; measures to avoid obesity; MRI or CT for evaluation of spinal cord compression; adenotonsillectomy, continuous positive airway pressure, mask, and tracheostomy to correct obstructive sleep apnea; suboccipital decompression; and lower-limb hyperreflexia or clonus and central hypopnea; surgery to correct scoliosis; and support in socialization and school adjustment.



Bone Dysplasia
Ontology



- Wiki-style editing

Collaborative knowledge curation



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Investigation

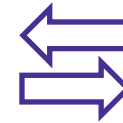
Cervical spine MRI with CSF flow studies is the best investigation to assess symptomatic craniocervical junction compression in children with Achondroplasia.

 Posted By [admin](#)

SEP
02



Bone Dysplasia
Ontology



- Wiki-style editing
- Micro-contributions

Collaborative knowledge curation



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Achondroplasia

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Disease with disproportionate growth and midline often delayed upper airway support

Diagnosis most affected atypical findings detects

Management weight, signs of mask, and lower-limb support

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Administer dysplasias

[Add new Bone Dysplasia](#)
[Add Bone Dysplasia to group](#)
[Remove Bone Dysplasia from group](#)
[Deprecate](#)
[Promote](#)
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BONE DYSPLASIA GROUP

[Abnormal mineralization group](#)

Select the Bone Dysplasia group to contain the

NEW BONE DYSPLASIA NAME

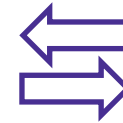
[Investigation](#)

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Bone Dysplasia
Ontology



- Wiki-style editing
- Micro-contributions
- Direct domain knowledge “encoding”

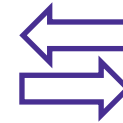
Collaborative knowledge curation



- Alter the ontology structure
- Alter class axioms

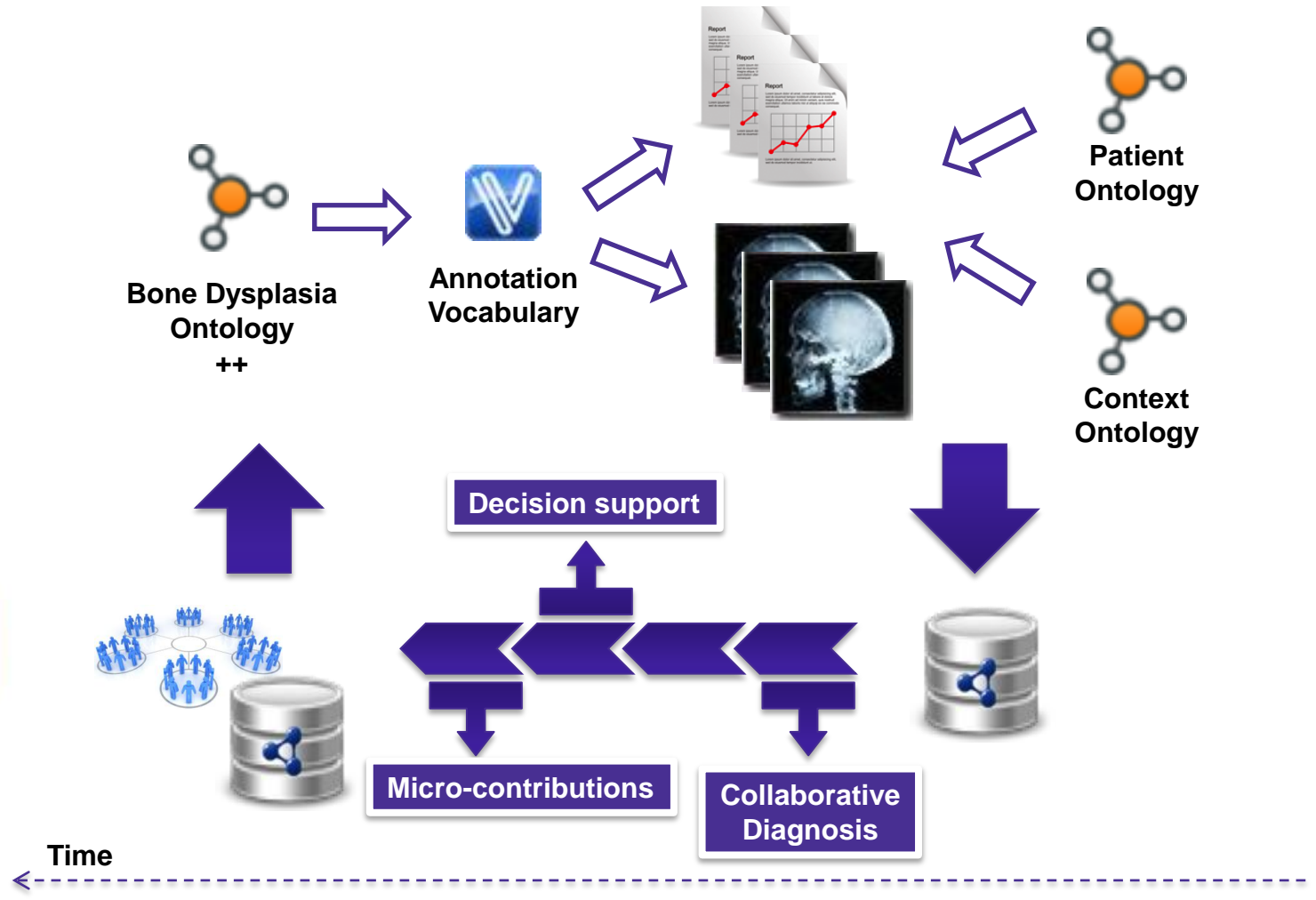


Bone Dysplasia
Ontology



- Wiki-style editing
- Micro-contributions
- Direct domain knowledge “encoding”

Knowledge transfer



Semantic annotation



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[Home](#) » [Add content](#) » Create Clinical summary

Create Clinical summary

TITLE *

Clinical summary

TAGS

, Craniosynostosis, Hypercalciuria, Nephrocalcinosis, Delayed skeletal maturation, Limited elbow movement, Pes planus, Tapered fingers

Suggested Tags

[Craniosynostosis](#) [Hypercalciuria](#) [Nephrocalcinosis](#) [Delayed skeletal maturation](#) [Limited elbow movement](#) [Pes planus](#) [Tapered fingers](#)

OBSERVATIONS ([EDIT SUMMARY](#))

B *I* U ABC [List of icons]

She has a combination of craniosynostosis, hypercalciuria, nephrocalcinosis, osteopaenia, delayed bone age and unusual hands and feet. She has limited elbow mobility, but hyperextensibility of the digits apart from the thumbs and flat feet. I have attached a number of xrays but particularly note the cone epiphyses and the short proximal fifth metacarpals. She has quite long tapering fingers with digitalised thumbs. She is unable to flex the left thumb.

HP:HP_0001363
HP:HP_0002150
HP:HP_0001182

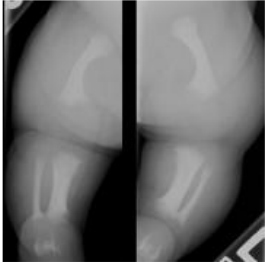
Semantic annotation



[Home](#) » [X-Ray report at 10 weeks](#) » [Edit image Image 24](#)

Edit image Image 24

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TITLE

Image 24

DESCRIPTION

B *I* U **ABC** [List of icons]

Short long bones. Curved femora. Flared metaphyses. Spiky tarsal bones.

Path: p

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TEXT FORMAT Filtered HTML

- Web page addresses and e-mail addresses turn into links automatically.
- Lines and paragraphs break automatically.

TAGS

Short long bones x Flared metaphyses x

[Add](#)

Enter a comma-separated list of words to describe your image.

[SEARCH](#) [ADMIN](#)

ary

osis, Delay

[cinosis](#) [Delayed skeletal maturation](#) [Limited elbow movement](#) [Pes planus](#) [Tapered fingers](#)

[x²](#) [x₂](#) [List of icons]

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HP:HP_0001363
HP:HP_0002150
HP:HP_0001182

HP:HP_0003026

Collaborative diagnosis



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Achondroplasia (diagnosis)

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posted by [Andreas Zankl](#) on [Fri, 10/07/2011 - 13:42](#)

PATIENT: [Case-1](#)

BONE DYSPLASIA: [Achondroplasia](#)

☆☆☆☆☆
You voted 4. Total votes: 1

Comments

[Add new comment](#)

COMMENT *

B *I* U ABC [List Icons] [Link Icon] [Image Icon] [X]

Path:

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- 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

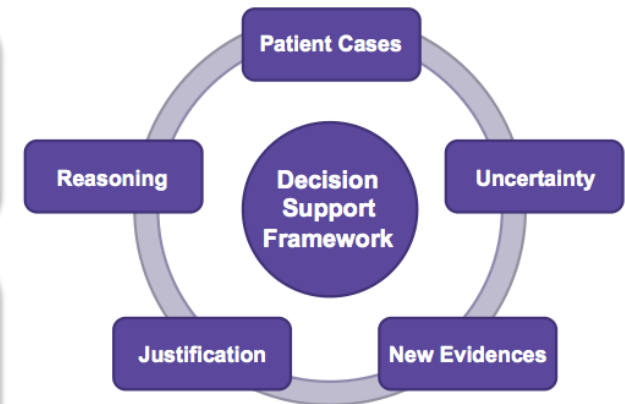


A **ranked list of features** given a particular disorder

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

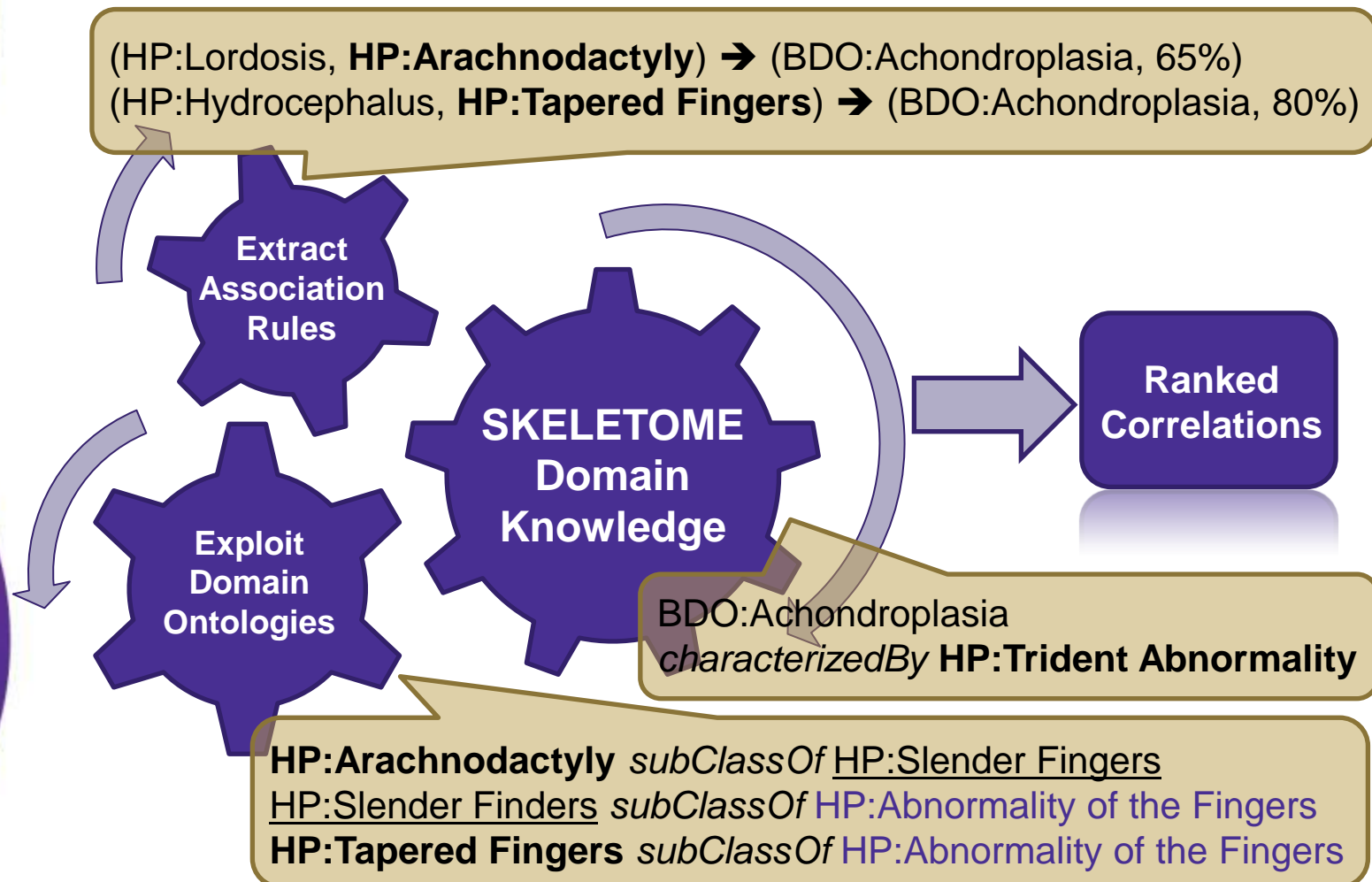
A **ranked list of disorders** given a particular feature

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



Decision support

Overview





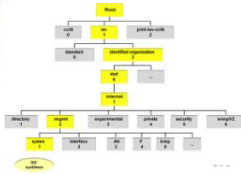
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Future work



Future work

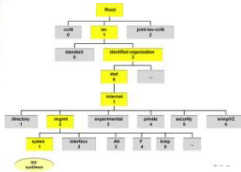
Ontologies



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

Future work

Community



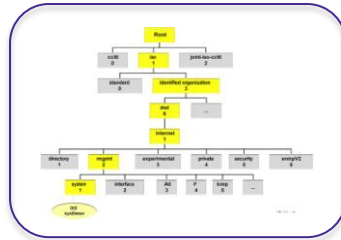
- **Integration with existing resources**
 - International Skeletal Dysplasia Registry
 - European Skeletal Dysplasia Network



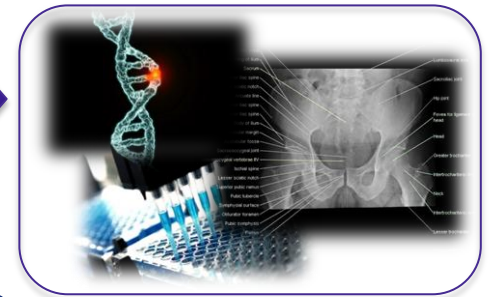
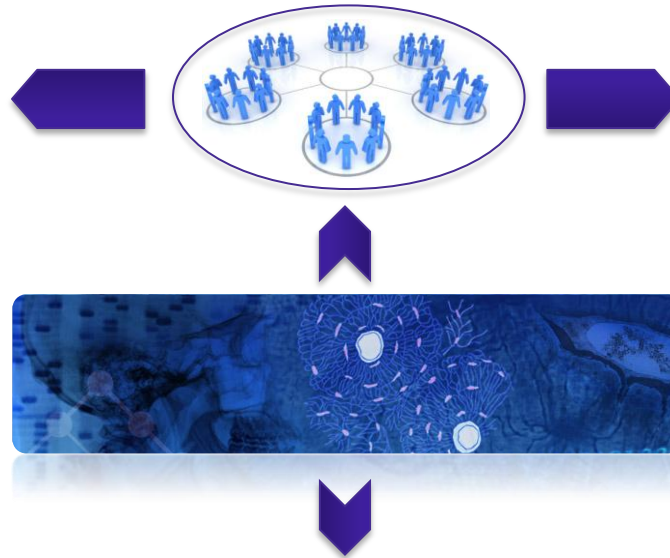
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Summary

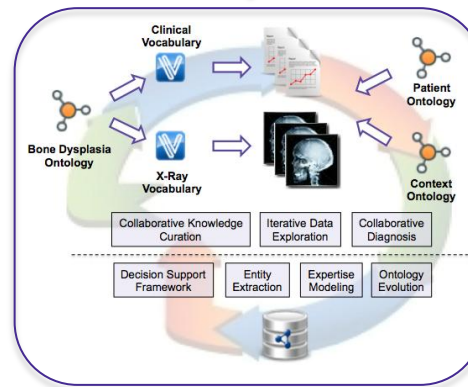
Summary



Existing domain
knowledge



Collaboratively acquired
patient cases
Collaborative diagnosis



Knowledge evolution
Decision support
Iterative data exploration

Thank you!