Detection of underspecifications in SNOMED CT concept definitions using language processing

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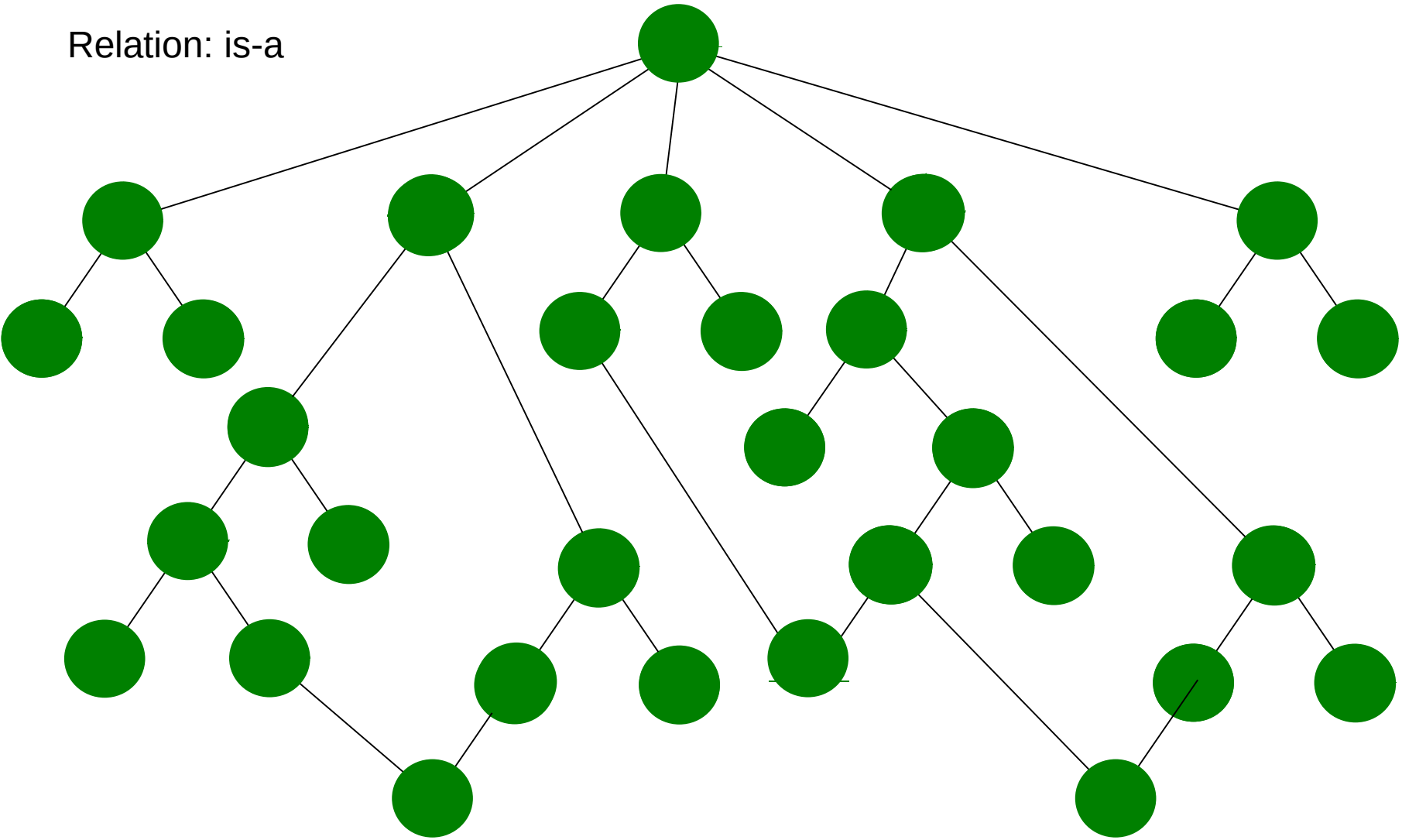
SNOMED CT

- “Standardized Nomenclature of Medicine - Clinical Terms”
- Comprehensive clinical terminology
  (> 300,000 representational units)
- Concepts are arranged in extensive taxonomic (is-a) hierarchies

Taxonomic Structure of SNOMED CT



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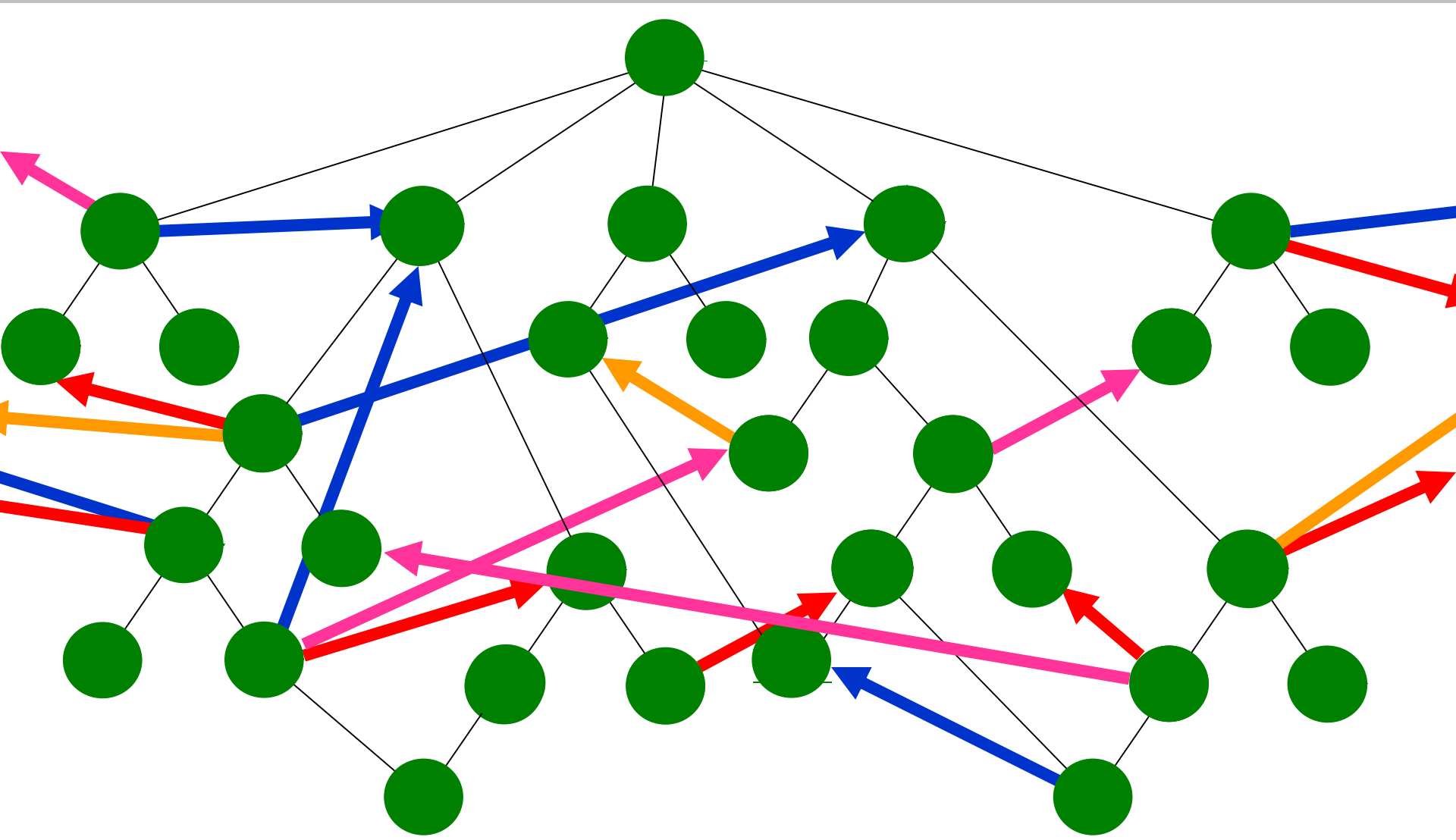
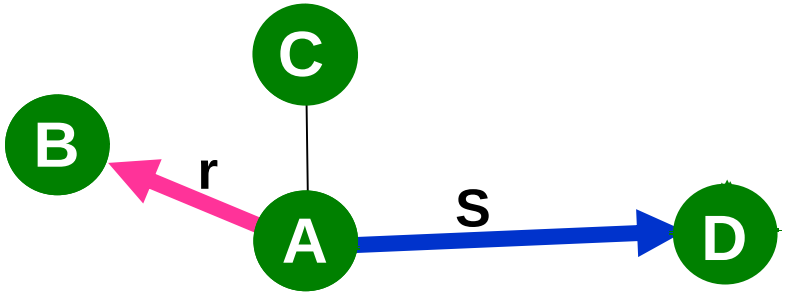
Cross-reference between SNOMED CT concepts



SNOMED CT semantics in a nutshell



A: concept
C: taxonomic parent
B, D: attributes

A – isA – C
A – r – B
A – s – D

A subClassOf
C and
r *some B and*
s *some D*

$\mathcal{EL}+$ description logics

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- Burden of terminology content maintenance and quality assurance
- To be supported by automated approaches

Looking for underspecifications of cross-linkage

- Nearly half (45.2%) of the SNOMED CT concepts (132,125) have no attributes.
- Textual descriptions suggest composed meanings
- Examples:
  - *Cerebral function*
    - only related to its parent *Nervous system function*
    - expected relation with *Brain structure* missing
  - *Hepatitis notification*
    - only related to its parent *Disease notification*
    - expected relation with *Inflammatory disease of liver* missing

Automatic suggestion of attributes

- Source: 01/2009 release of SNOMED CT

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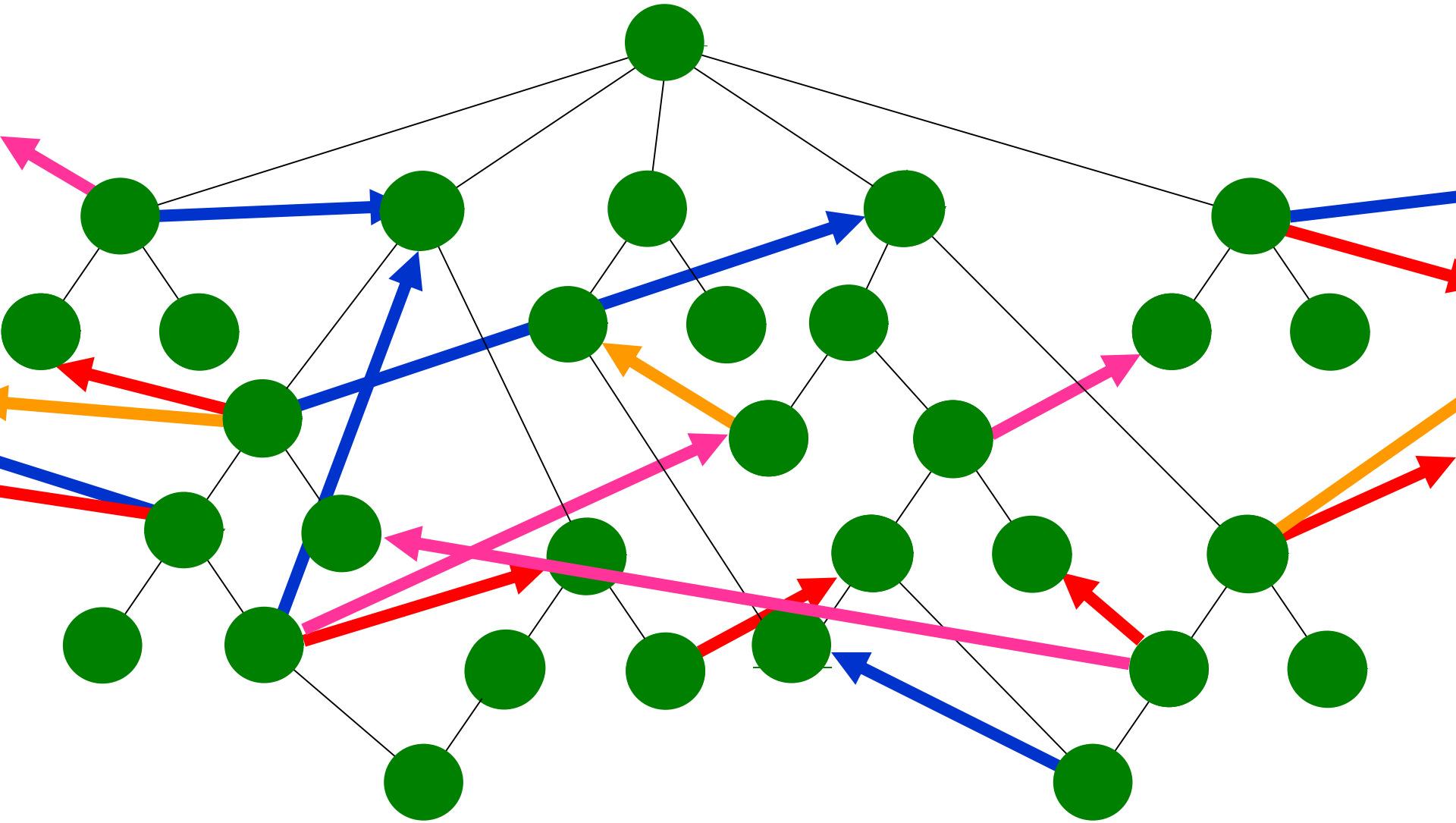
  1. Identify non-attributed concepts

Select non-attributed SNOMED CT concepts



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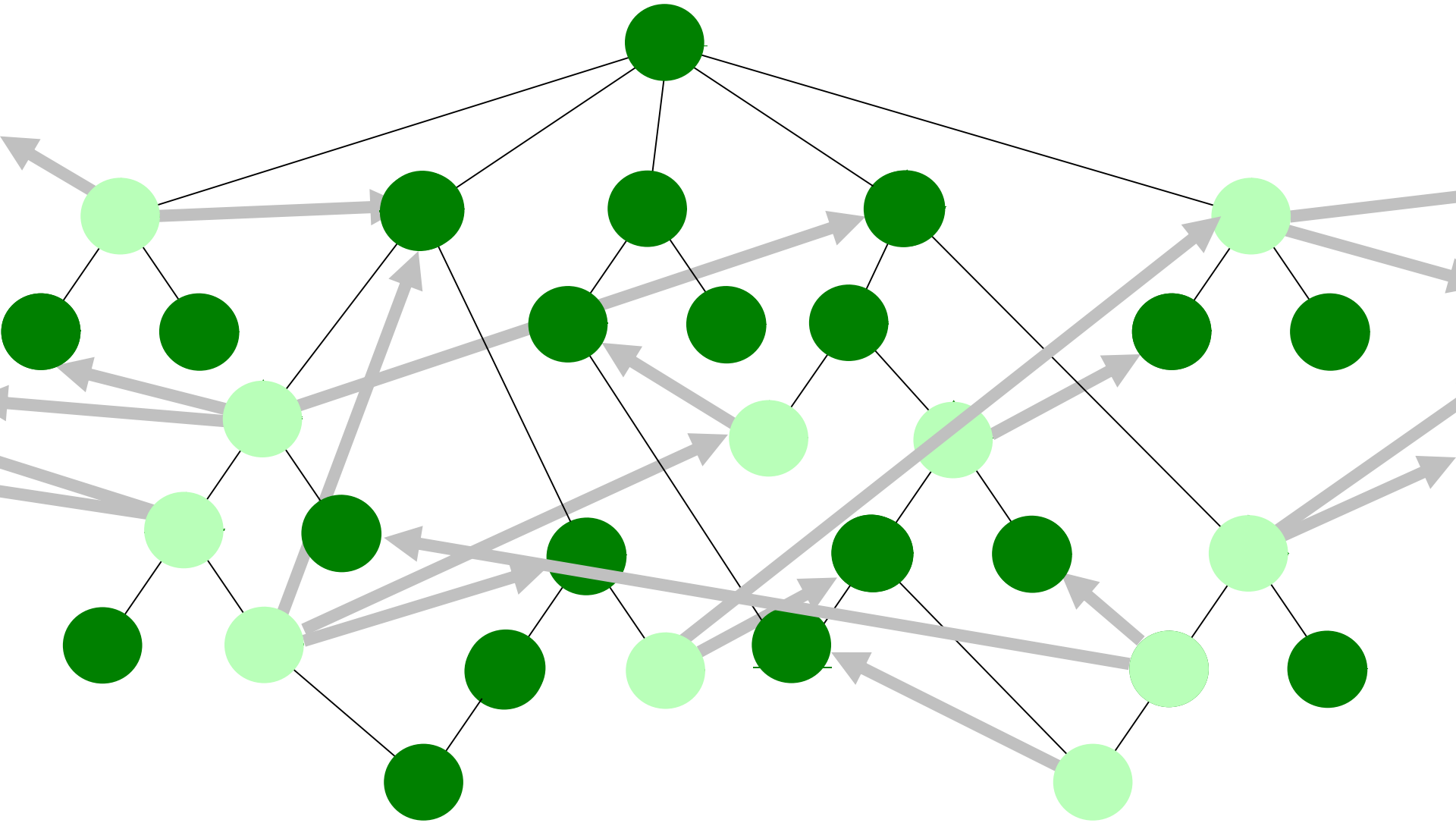

Automatic suggestion of attributes

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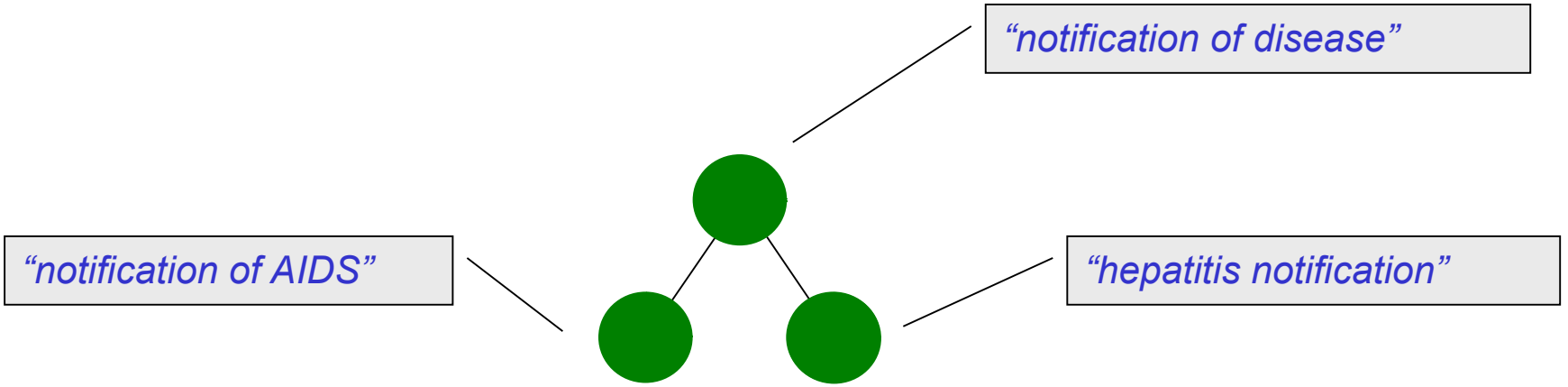
  2. Extract concept names

Extract names of non-attributed concepts



Automatic suggestion of attributes

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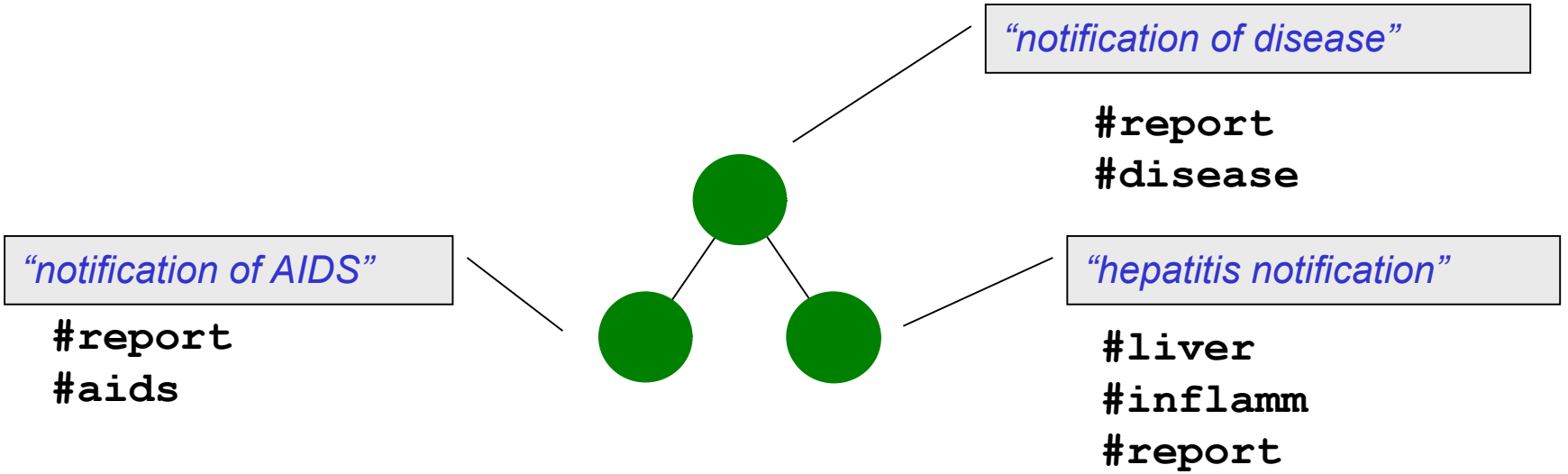
Perform morphosemantic abstraction

Using MorphoSaurus* morphosemantic indexing



*Markó K, Schulz S, Hahn U: MorphoSaurus - Design and Evaluation of an Interlingua-based, Cross-language Document Retrieval Engine for the Medical Domain. Meth Inf Med 4/2005(44): 537-545.

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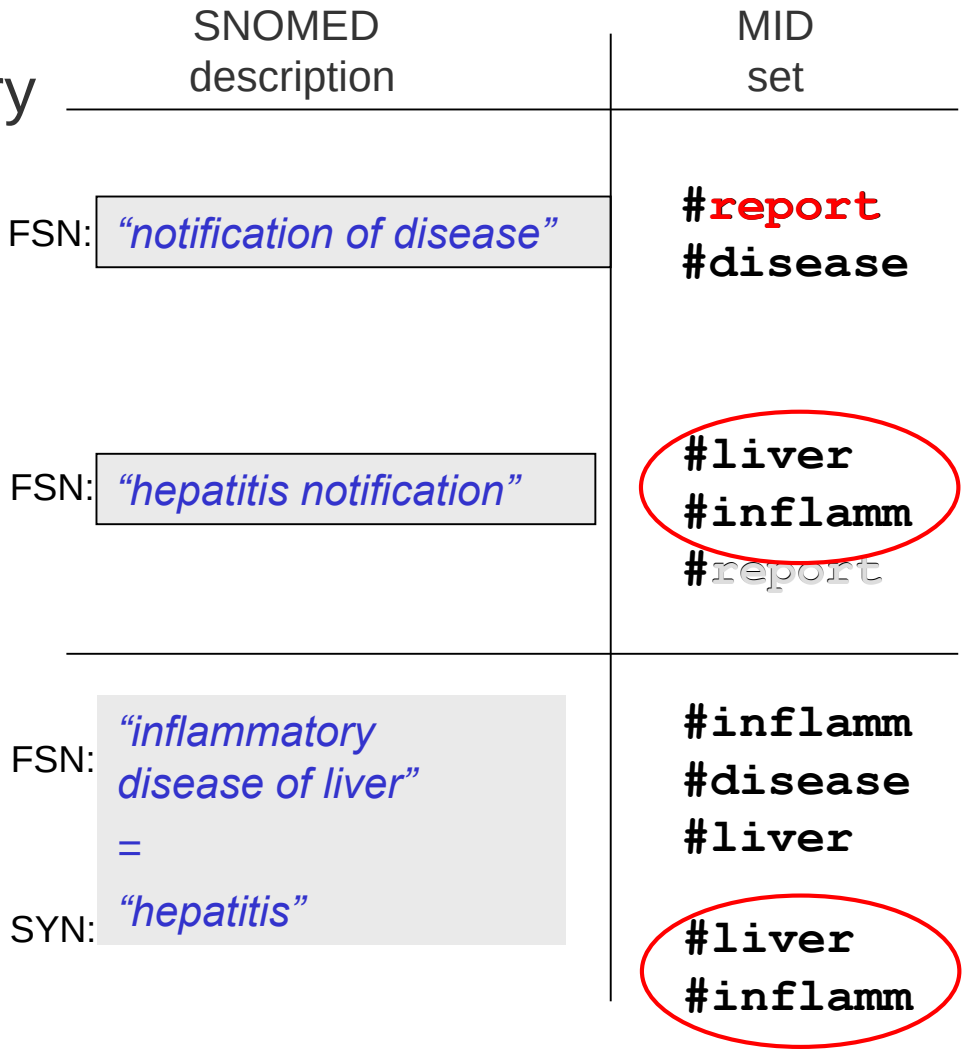
  5. Match reduced child set against MID representations of all SNOMED descriptions

Matching heuristics

- For the FSN MID set of every non-attributed concept:
  - remove MID that occurs in any of this concept's parents
  - check whether the remainder set coincides with the MID representation of some other SNOMED CT concept, considering all descriptions (FSNs, PTs, synonyms)
  - consider this concept a refinement candidate



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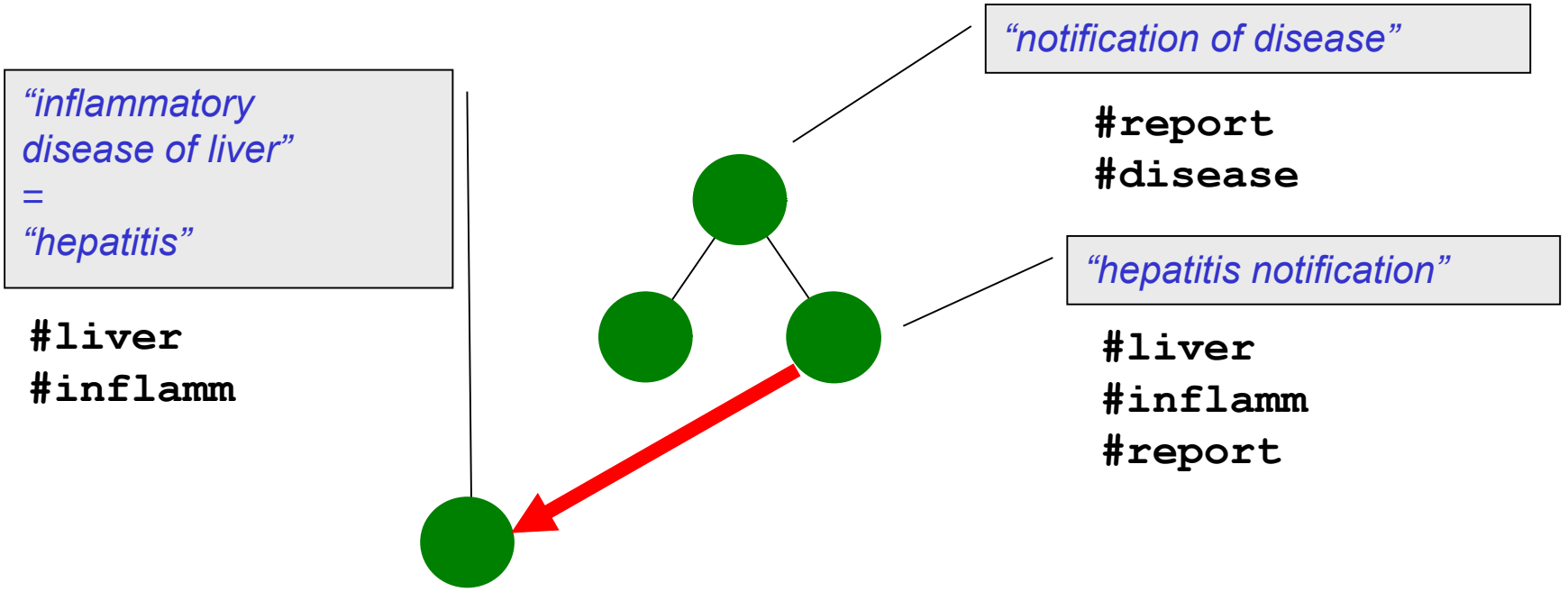
  5. Match reduced child set against MID representations of all SNOMED descriptions
  6. Suggest candidates for refining attributes

Addition of refinement candidate



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Evaluation Methodology

- For each of 14 SNOMED subhierarchies: random sample of 20 underspecified concepts, compared to attribute refinement candidate proposed by the system
- For each of the sample concept verify
  1. whether this concept should be refined
  2. whether one of the suggested refinement candidates can be plausibly used for refinement.
- Performed by two domain experts. Double rating for interrater agreement measurement: 25%

Results of retrieval experiments

Results of retrieval experiments

SNOMED hierarchies	Active Concepts	Non-attributed Concepts n	Non-attributed Concepts %	Refinement candidates n	Refinement candidates %	Analysis of samples(n=20) refine-ment justified	Analysis of samples(n=20) **correct suggest ion**	Sample based estimation refinable concepts	Sample based estimation with correct suggest-ions
Organism	31840	31840	100.0	4973	15.6	0%	**0%**	**0**	**0**
Substance	23554	23554	100.0	8627	36.6	55%	**35%**	**4700**	**3000**
body structure	25637	22386	87.3	15076	58.8	5%	**0%**	**800**	**0**
qualifier value	8823	8823	100.0	3533	40.0	0%	**0%**	**0**	**0**
observable entity	7885	7885	100.0	3647	46.3	70%	**50%**	**2600**	**1800**
Finding	32780	5356	16.3	2253	6.9	90%	**75%**	**2000**	**1700**
physical object	4408	4408	100.0	1339	30.4	85%	**80%**	**1100**	**1100**
morphologic abnormality	4297	4289	99.8	2164	50.4	80%	**60%**	**1700**	**1300**
Occupation	3843	3843	100.0	1330	34.6	75%	**10%**	**1000**	**100**
Product	19310	3541	18.3	686	3.6	100%	**60%**	**700**	**400**
Event	3578	3529	98.6	447	12.5	85%	**45%**	**400**	**200**
Disorder	63874	2812	4.4	1080	1.7	90%	**60%**	**1000**	**600**
Procedure	47764	2256	4.7	1001	2.1	85%	**65%**	**900**	**700**
Others	14511	7603	52.4	2396	16.5	75%	**60%**	**1800**	**1400**
TOTAL	292104	132125	45.2	48552	16.6			**18700**	**12300**

Results

- Interrater agreement (Kohen's kappa):
  - A concept should be refined: 0.55 (low !)
  - There is a proposed refinement candidate: 0.74
- Estimation: approximately 18,000 SNOMED CT concepts can be refined.
- Problematic suggestions:
  - *Macaroni* for *Macaroni maker*
  - *Canada* for *Salmonella canada*
  - *Acyl carnitine* for *Acylcarnitine hydrolase*
  - *First* for *Female first cousin*
    (already fully defined by the intersection of *First cousin* and *Female cousin*)

Conclusions

- Many SNOMED CT concepts are underdefined and can / should be refined
- The proposed methodology was useful to detect underspecifications
- Large difference between SNOMED hierarchies re harvesting and approval of refinement candidates
- “Grey areas”
  - many proposed refinements are debatable
  - only part of refinement candidates not retrieved due to restrictions of the methodology
- Should be considered for future SNOMED CT editing policies

Thank You!
Contact:

Stefan Schulz

http://purl.org/steschu

Acknowledgements:

CNPq, Brazil:
550830/2005-7

BMBF-IB, Germany:
BRA05/022

UTFPR
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