Health Search

From Consumers to Clinicians

Slides available at https://ielab.io/russir2018-health-searchtutorial/

Guido Zuccon Queensland University of Technology

y @guidozuc



Outline

- Dealing with the semantic gap: exploiting the semantics of medical language
 - concept based search & inference, query expansion, learning to rank
- Dealing with the nuances of **medical language**
 - negation, family history, understandability
- Understanding and aiding query formulation
 - query variations, query reformulation, query clarification, query suggestion, query intent, query difficulty, task-based solutions

Dealing with the semantic gap

Exploiting semantics of medical language

- What are medical concepts, where are they defined
- Why use concepts
- Why concepts and terms

Medical concepts

- Medical concepts are defined in domain knowledge resource
- Capture the key aspects of the domain or some specific sub-domain
- Relationships between concepts capture associations

Implicit VS Explicit Semantics

- Explicit semantics: structured human representation of knowledge and its concepts
 - e.g., medical terminologies
- Implicit Semantics: draw representation of words/concepts from data
 - e.g., distributional/latent semantic models

Key Medical Terminologies

Medical Subject Headings (MeSH)

Controlled vocabulary for indexing journal articles

Mainly used by researchers and clinicians searching the literature.



SNOMED CT

Formal medical ontology: ~500,000 concepts ~3,000,000 relationships



ICD

International Statistical Classification of Diseases and Related Health Problems (ICD)

Diagnosis classification from World Health Organisation

Used extensively in billing

Chanter	Placks	Title
Chapter	Blocks	The
I	A00-	Certain infectious and parasitic diseases
	B99	
II	C00-	Neoplasms
	D48	
ш	DEO	Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
	D50-	
	D89	
IV	E00-	Endocrine, nutritional and metabolic
	E90	diseases
v	F00-	Mental and behavioural disorders
	F99	
VI	G00-	Diseases of the nervous system
	G99	
VII	H00-	Diseases of the eye and adnexa
	H59	
VIII	H60-	Diseases of the ear and mastoid process
	H95	
IX	100-199	Diseases of the circulatory system
x	J00-	Diseases of the respiratory system
	J99	
XI	K00-	Diseases of the digestive system
	K93	
XII	L00-	Diseases of the skin and subcutaneous
	L99	tissue

Unified Medical Language System (UMLS)

• UMLS is a compendium of many controlled vocabularies in the biomedical sciences



- Combined many terminologies under one umbrella
- UMLS concept grouped into higher level semantic types
 - Concept: Myocardial Infarction [C0027051] of type Disease or Syndrome [T047]
 - <u>https://uts.nlm.nih.gov//metathesaurus.html</u>

An important note

- These resources contain information that can help characterise medical language
 - Synonyms of a term
 - Relationship between terms/concepts
- Rarely do these resources contain information that directly answers questions like
 - What is the drug of choice for condition x?
 - What is the cause of symptom x?
 - What test is indicated in situation x?
 - How should I treat condition x (not limited to drug treatment)?

- How should I manage condition x (not specifying diagnostic or therapeutic)?
- What is the cause of physical finding x?
- What is the cause of test finding x?
- Can drug x cause (adverse) finding y?
- Could this patient have condition x?
- That is, they **do not directly resolve the clinical questions** presented in [Ely et al., 2000] taxonomy
- They capture truisms/**universal facts**, not subjective knowledge/things that could change over time 12

Convert Terms to Concepts (aka Concept Mapping)



Concept extraction/mapping tools

- Metamap National Library of Medicine [Aronson&Lang, 2010]
 - Extensive configuration option; <u>but</u>: default options tuned for biomedical literature, not necessarily websites or clinical text
 - Can be slow and unstable
- QuickUMLS [Soldaini&Goharian, 2016]
 - Modern computationally efficient mapper
 - Shown in the hands-on session
- **SemRep** to extract relations between concepts [Rindflesch&Fiszman, 2003]
 - <subject, object, relation> from 27.9M PubMed articles stored into SemMedDB: <u>https://skr3.nlm.nih.gov/SemMedDB/</u>
- Others exist: cTakes [Savova et al., 2010], Ontoserver [McBride et al., 2012], etc.

Concept Mapping as an IR problem

"...the patient had headaches and was home ..."



[Mirhosseini et al., 2014]

to generate at least one mapping)

Practical - part 1

- In this hands-on session, we will:
 - 1. Take a collection of clinical trials, annotate them with medical concepts, producing documents with both term and concept representation.
- In part 2, we will use these results to:
 - 2. Index these documents in Elasticsearch with multi term/concepts fields.
 - 3. Search Elaticsearch with either term or concept, demonstrating semantic search capabilities.
 - 4. Play a bit more (maybe)
- Instructions: <u>https://ielab.io/russir2018-health-search-tutorial/hands-on/</u>

Implicit Medical Concept Representations: Word Embeddings

- [Pyysalo et al., 2013]: word2vec and random indexing on very large corpus of biomedical scientific literature. <u>http://bio.nlplab.org</u>
- [De Vine et al., 2014]: word2vec on medical journal abstracts (embedding for UMLS)
 - Learns embedding of a concept, from co-occurrence with concepts
- [Zuccon et al., 2015, b]: word2vec on TREC Medical Records Track. <u>http://zuccon.net/ntlm.html</u>
- [Choi et al., 2016]: word2vec on medical claims (embedding for ICD), clinical narratives (embedding for UMLS) <u>https://github.com/clinicalml/embeddings</u>
- [Beam et al., 2018]: cui2vec (variation of word2vec) on 60M insurance claims + 20M health records + 1.7M full text biomedical articles. https://figshare.com/s/00d69861786cd0156d81
- Nuances of medical word embeddings:
 - [Chiu et al., 2016]: bigger corpora do not necessarily produce better biomedical word embeddings

Concept-based IR

Two types for Concept-based Retrieval

- Concept Augmented Term-based Retrieval e.g. [Ravindran&Gauch, 2004]
 - Maintain the original term representation of documents.
 - Use a concept-based approach to improve the query representation.
- Pure Concept-based Retrieval
 - Map the terms in documents to higher-level concepts
 - Retrieval is then done in 'concept space' rather than 'term space'
 - SAPHIRE system [Hersh&Hickam, 1995]
 - Language modelling concepts [Meij et al., 2010]

Combining Text and Concept Representations

[Limsopatham et al., 2013c]: learning framework that combines bag-of-words and bag-of-concepts representations on per-query basis

- 1. Linear combination model for merging scores from the two representations
- 2. Features: QPPs for both representations
- Regression to infer model parameters (Gradient Boosted Regression Trees)

Exploiting concept hierarchies



[Zuccon et al., 2012]

Semantic Inference for IR

Concept-based retrieval that exploits ontology relationships

- Inferring conceptual relationships [Limsopatham et al., 2013]
- Information Retrieval as Semantic Inference [Koopman et al., 2016]
 - both: expand queries by inferring additional conceptual relationships from KB, but in different ways
 - [Limsopatham et al., 2013] also infers relationships
 - from collection of medical free-text, and
 - via PRF

"This is a 62-year-old gentleman who has Type I DM and is on hemodialysis. He is currently taking Avapro''

- Hemodialysis \checkmark
- DM? Diabetes mellitus?
- Avapro? Hypertension!

Inferring conceptual relationships [Limsopatham et al., 2013]

- For KB: use semantic relationships of concepts to represent the relationships between concepts.
- For free-text: MetaMap to identify concepts from the free-text, then infer relationships by co-occurence/association rules





Practical - part 2

- Let's resume from where we left in part 1, and let's do:
 - 1. Index these documents in Elasticsearch with multi term/concepts fields.
 - 2. Search Elaticsearch with either term or concept, demonstrating semantic search capabilities.
 - 3. Play a bit more (maybe)

 Instructions: <u>https://ielab.io/russir2018-health-search-</u> <u>tutorial/hands-on/</u>

Choices in KB Query Expansion

- Many other approaches to do inference over KB data
- [Jimmy et al., 2018] consider the Entity Query Feature Expansion model [Dalton et al., 2014] and the influence settings choices have



Choices in KB Query Expansion Findings for CHS

- For CHS, EQFE based on **UMLS** is more effective than based on Wikipedia.
 - Choice 1: Index all UMLS concepts
 - Choice 2: Use all uni-, bi-, and tri-grams of the original queries
 - Choice 3: Map mentions to UMLS aliases
 - Choice 4: Source expansion from the UMLS title
 - Choice 5: Add **relevance feedback** terms

Knowledge based vs data-driven Query Expansion



Combine multiple-evidences in the collection that refer to the same case

Zhu&Carterette, 2012]



- Ranking generated for each document, individually
- Ranking generated for an aggregated case

Fused into new ranking

• Online possible in situations where multiple documents are available for one case (e.g. with health records, where case=patient)

Adaptively Combine (or not) Records of a Case [Limsopatham et al., 2013b]

- Choose between:
 - 1. Combine records for a patient, then rank patient
 - 2. Rank records, then identify patients based on relevance of records ranking
- Classifier to learn to select which ranking approach to use, depending on query
- Features: query difficulty measures (QPPs), number of medical concepts in query

Different, diverse corpora used for query expansion

[Zhu et al., 2014]

- Mixture of relevance models to **combine evidence from** ulletdifferent collections to derive query expansions
 - Collections: Mayo Clinic health records (39M), TREC Genomics • (166K), ClueWeb09B (44M), TREC Medical Records (100K)
- Findings:
 - Access to large clinical corpus significantly improves query expansion
 - The more difficult the query, the more it benefit expansion benefits from auxiliary collections
 - "use all available data" is sub-optimal: value in collection \bullet curation

Measure the usefulness of different collections [Limsopatham et al., 2015]

- Automatically decide which collection to use for query expansion evidence
 - 14 different document collections, from domain-specific (e.g. MEDLINE abstracts) to generic (e.g. blogs and webpages)
 - But they are not all useful, and not to the same extent to generate query expansion terms
- Techniques based on resource selection and learning to rank

Co-occurences, Latent Methods & Word2vec

- (Co-occurrence of) concepts as a graph -> application of link analysis methods [Koopman et al., 2012; Martinez et al., 2014]
- Explicit and latent concepts [Balaneshinkordan&Kotov, 2016]
- Word **embeddings** and concept embeddings [Zuccon et al., 2015, b; Nguyen et al., 2017]

Co-occurence Graphs, Semantic Graphs and Page Rank

- [Koopman et al., 2012]:
 - Build concept graph from document concepts as they co-occur in document
 - 2. Run Pagerank
 - 3. Use Pagerank scores as additional weights for retrieval
- [Martinez et al., 2014]:
 - 1. Build concept graph from query concepts and related concepts in UMLS
 - 2. Run Pagerank
 - 3. Rank concepts using page rank scores; select top K concepts as query expansion
- Analysis shows expansion terms selected by Pagerank: taxonomic (eg., synonyms) and not taxonomic (eg., disease has associated anatomic site).

Explicit and Latent Concepts

- [Balaneshin-kordan&Kotov, 2016]: different concept types/ sources (KBs, PRF) should have different weights
- Builds upon Markov Random Field retrieval [Metzler&Croft, 2005]
- Different features for different semantic types + topical features of KB graphs, and statistics of concepts in collection
- Learns optimal query concept weight using multivariate optimisation
- Base approach (without optimisation) best system at TREC CDS 2015
Word Embeddings and Concept Embeddings: Neural Translation LM [Zuccon et al., 2015, b]

$$p_t(w|d) = \sum_{u \in d} p_t(w|u) p(u|d)$$

use Word Embeddings for computing this



Constraining word embeddings by prior knowledge

- [Liu et al., 2016]: learn concept embeddings constrained by relations in KB (UMLS)
- Results in a modified CBOW
- Use word embeddings to re-rank results: interpolate original relevance score with similarity based on embeddings
- Experiments only limited to synonym relations & singleword concepts

Concept-Driven Medical Document Embeddings

[Nguyen et al., 2017]: optimises document representation for medical content

- Uses neural-based approach (akin to *doc2vec*) to create embedding that captures latent relations from concepts and terms in text.
- Uses embedding to identify top documents
- Extract top words and concepts from top documents to produce expansions



Learning to Rank

[Soldaini&Goharian, 2017]: compares 5 LTR in CHS context:

- LTR: logistic regression, random forests, LambdaMART, AdaRank, ListNet
- Features: statistical (36 features), statistical health (9), UMLS (26), latent semantic analysis (2), word embeddings (4).
- LambdaMART performed best; all features required

Dealing with the nuances of medical language



- Negated content best handled by:
 - Not removing negated content (as is commonly done)
 - Indexing positive, negated & family history content separately [Limsopatham et al., 2012]
 - Weighting content separately [Koopman & Zuccon, 2014]

PICO

- PICO: framework for formulating clinical questions
 P: Patient/Problem (P) (e.g., males aged 20-50)
 I: Intervention (e.g., weight loss drug)
 C: Comparison (e.g., controlled exercise regime)
 O: Outcome (e.g., weight loss)
- Exploiting PICO elements in IR:
 - Language modelling based **content weighting** [Boudin et al., 2010]
 - Tagging PICO elements for IR "I" & "P" elements most beneficial for retrieval
 - Field retrieval based on PICO [Scells et al., 2017b]
 - promising, but needs method to predict which keywords require
 PICO annotations
 RobotReviewer [Marshall et al., 20]



RobotReviewer [Marshall et al., 2015]: Algorithm for extracting PICO elements from free-text

Readability & Understandability

- Laypeople do not necessarily understand medical documents that clinicians would understand
- Need to retrieve documents that are both understandable and relevant
- [Palotti et al., 2016 b]: LTR with two sets of features:
 - Estimate relevance: standard IR features
 - Estimate understandability: features based on readability measures and medical lexical aspects

Understanding and aiding query formulation

What would search for?



Enter your search terms at http://chs.ielab.webfactional.com/

"Circumlocutory" queries

Symptom Group	Crowdsourced Circumlocutory Queries						
alopecia	baldness in multiple spots, circular bald spots, loss of hair on scalp in an inch width round						
angular cheilitis edema	broken lips, dry cracked lips, lip sores, sores around mouth fluid in leg, puffy sore calf, swollen legs						
exophthalmos	bulging eye, eye balls coming out, swollen eye, swollen eye balls						
hematoma	hand turned dark blue, neck hematoma, large purple bruise on arm						
jaundice	yellow eyes, eye illness, white part of the eye turned green						
psoriasis	red dry skin, dry irritated skin on scalp, silvery-white scalp $+$ inner ear						
urticaria	hives all over body, skin rash on chest, extreme red rash on arm						

[Stanton et al., 2014]

How effective are Google & Bing at Health Search?

System	ndcg@1		ndc	g@5	ndcg	j@10	P@	<u>گ</u> 5	P@	010	
	Rel	Hrel	Rel	Hrel	Rel	Hrel	Rel	Hre	Rel	Hrel	
Bing	.3846	.2308	.3812	.2654	.3802	.2764	.4385	.27 9	.4308	.2769	
Google	.3846	.3077	.4242	.3142	.4252	.3138	.5000	.315	.4923	.3115	

Effectiveness of two widely used commercial search engines when prompted with (circumlocutory) medical queries are discounted at self-diagnosis purposes. Results are averaged over 26 queries. P@k stands for precision at rank k; ndcg@k stands for normalised discounted cumulative gain at rank k.

[Zuccon et al., 2015]

Performance per query



Only highly relevant



[Zuccon et al., 2015]

Query Recommendation

[Zeng et al, 2006]: recommend queries based on UMLS and query log (CHS task)

• Leads to higher user satisfaction and query success rate



Query Reformulation

[Soldaini et al., 2015]: compares the effectiveness of 7 query reformulation techniques (CDS task)

- 1. **UMLS Concepts Selection** (MMselect): remove all terms with no mapping to any UMLS concepts
- 2. Health-related terms selection (HT): compute ratio of associated Wikipedia page P being health-related over being not-health-related. Retain only query terms with ratio ≥ 2 .
- 3. Query Quality Predictors (QQP): use QPPs as features of SVMrank to select query terms.
- 4. **Faster QQP**: rank sub-queries using MI and retains the top 50. In addition to QQP features, add features: UMLS concepts found, UMLS sem-types found, HT ratio, and MeSH found.

Query Reformulation

[Soldaini et al., 2015]: compares the effectiveness of 7 query reformulation techniques (CDS task)

- 5. **UMLS Concepts Extraction** (MMexpand): append the preferred terms UMLS query concepts to expand original query
- 6. **Pseudo Relevance Feedback** (PRF): weight terms in top 10 initial results, rank and add top 20 terms not in original query.
- 7. Health Terms PRF (HT-PRF): as PRF, but candidate expansion terms filtered health term ratio
 - This is empirically identified as the best technique
 - The HT component in general seems effective

Query Reformulation with deep learning [Soldaini et al., 2017]: considers short clinical notes as queries (CDS task)

- 1. Generate candidate terms using PRF
- 2. Train supervised neural network to predict Weight Relevance Ratio (WRR) of candidate terms: importance of term in relevant documents
- 3. For representations it uses word embeddings, statistical features over multiple collections, syntactical and semantical features
 - The neural network approach and HT-PRF perform similarly

Query Clarification

[Soldaini et al., 2016]: add the most appropriate expert expression to queries submitted by users

- Acquire expert expressions from 3 KBs: behavioral (logs), MedSyn, and DBpedia
- Select expression with the highest probabilities of appearing in health-related Wikipedia pages, using logistic regression classifier
- Finding through user study evaluation (CHS task):
 - Expressions from all 3 KBs improve rate of correct answers (behavioural KB best)
 - number of correct answers significantly increases when users clicked HON-certified websites

Query Reduction

- [Koopman et al., 2017 c]: reduce verbose clinical queries (health records, CDS task) using generic & domain-specific methods
- Reduce to only UMLS Medical Concepts & Tasked UMLS
- Combined model UMLS + IDF-r (proportion of top-ranked IDF terms retained)
- Comparison vs human-generated queries: human generated queries significantly more effective
 - per-query parameter learning promising
 - automated reduction handicapped in that they only use terms from narrative

Query Reduction

[Soldaini et al., 2017 b]: use convolutional neural networks (CNN) to reduce queries (CDS task)

- Queries are short clinical notes
- CNN is used to estimate the importance of each query term
- Given a query, a relevant document and a non-relevant document:
 - 1. Use CNN to determine weights terms in query
 - 2. Use term weights to score relevant and non-relevant documents
 - 3. Back-propagate a loss if non-relevant document is scored higher than relevant document

Query Rewriting

[Scells&Zuccon, 2018]: through a chain of transformation, generates better (Boolean) queries (for systematic reviews compilation)

- Defines set of transformations: mostly syntactic transformations
- Selects transformations based on: heuristics, classifier, learning to rank
- Large gains possible by transforming queries



Query Difficulty

 [Boudin et al., 2012]: predictor that exploits MeSH structure to ascertain how difficult queries are — estimates query variability and specificity

$$MeSH-QD(Q, \mathcal{T}) = \sum_{t \in Q} \underbrace{\frac{df(t)}{\sum_{t' \in V(t)} df(t')} \cdot ln\left(1 + \frac{N}{df(t)}\right)}_{t' \in V(t)} \cdot \underbrace{ln\left(1 + \frac{N}{df(t)}\right)}_{\text{length}(t)} \cdot \underbrace{\frac{depth(t)}{depth(t)}}_{\text{length}(t)}$$

- V(t): set of alternative expressions of the concept t; depth/length in MeSH
- coverage of thesaurus & concept mapping influence quality
- [Scells et al., 2018]: **standard predictors** for QPP and QVPP (V=variation) in systematic reviews compilation
 - Predictors **not suited** to the domain-specific nature of the task
 - Identifying best performing variations hard task

Task based retrieval

- Research on how clinicians' query shows a set of standard query types [Ely et al., 2000]
- Can be simplified to three clinical tasks:

i.searching for diagnoses given a list of symptoms;

ii.searching for relevant tests given a patient's situation

iii.searching for effective **treatments** given a particular condition.

• These can be exploited in a retrieval scenario...

Tasked-based retrieval

 Concept-based approach but "focusing only on medical concepts essential for the information need of a medical search task" [Limsopatham et al., 2013]

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Aspects of the Medical Decision Criteria Malaria reemergence in the Peruvi MetaMap's Semantic Type Symptom Diagnostic test Diagnosis Treatment 2640690 01/01/1999 Emerging Infectious Diseases http://ww Body Location or Region ~ V V Body Part, Organ, or Organ Component 1 1 Clinical Drug 1 **Diagnostic Procedure** ✓ Abstract Disease or Syndrome Finding ~ Epidemic malaria has rapidly emerged in Loreto Departme 1 1 Health Care Activity of malaria cases in South America (after Brazil), most from Injury or Poisoning 1 Intellectual Product 1 fourfold in Peru. Plasmodium falciparum infection, which Medical Device 1 became the dominant Plasmodium infection in the highes Mental or Behavioral Dysfunction ✓ has also increased during this epidemic in Loreto. Moreov Neoplastic Process 1 strains have emerged, which require development of effice Pathologic Function Pharmacologic Substance 1 Sign or Symptom ✓ Therapeutic or Preventive Procedure User Interface Retrieval Task-oriented retrieval Field-based inverted file index Significant concept estimation Clinician searcher



How does a good health query look like?

- [Tamine&Chouquete, 2017] found that in health search, query quality is influenced by medical expertise
- [Koopman et al., 2017] studied the querying behaviour of 4 clinicians
 - most effective clinicians those who entered short queries (but retrieval models optimised for short queries)
 - most effective clinicians those who inferred novel keywords most likely to appear in relevant documents
 - most effective clinicians posed queries around treatments rather than diagnoses (but influenced by task: searching for clinical trials)