Medical Information Retrieval

ESSIR 2023 Lecture

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Outline

Who am I?

Lorraine Goeuriot, lecturer at Laboratoire d'Informatique de Grenoble (LIG), Universit Grenoble Alpes (UGA)
Research interests:

- Domain-based Information Retrieval
- Evaluation of Information Retrieval
- Natural Language Processing
- AI Models for health data

Université Grenoble Alpes

OBJECTIVES

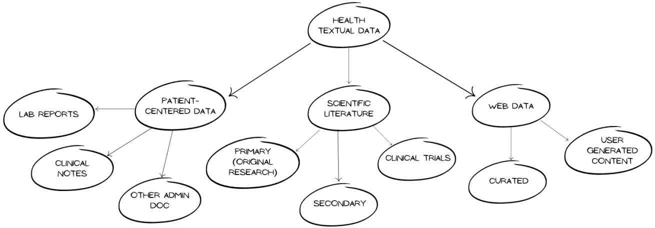
1. Introduce tasks, users and resources in the medical domain
2. Present state-of-the art models and techniques in medical information retrieval and off-the-shelf tools
3. Provide a list of open-source datasets
4. Summarize challenges and research opportunities

Slides, links and references are online:
`https://github.com/lorraine-goeuriot/medical-IR-ESSIR23`

OUTLINE

Medical Information

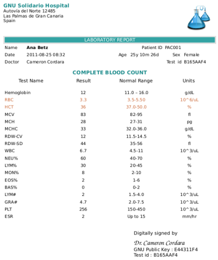
A Classification of Textual Health Information [Hersh, 2010]



- **Patient-specific information**: applies to individual patients. Tells healthcare providers, administrators and researchers about the health and disease of a patient.
  - Structured: laboratory results, vital signs
  - Narrative: history and physical, progress notes, radiology report
- **Knowledge-based information**: has been derived and organized from observational or experimental research. Usually provided in books, journals or *computerized media*.
  - Primary: original research (in journals, books, reports, etc.)
  - Secondary: summaries of research (in review articles, books, practice guidelines, etc.)
- Web data:
  - Curated websites: medical institutions, magazines, specialized websites, Wikipedia
  - User-generated-content: discussion forums, Facebook, Twitter, PatientsLikeMe

Medical Information

Structured Patient-centered Data



GNU Solidario Hospital
Autovía del Norte 12485
Las Palmas de Gran Canaria
Spain

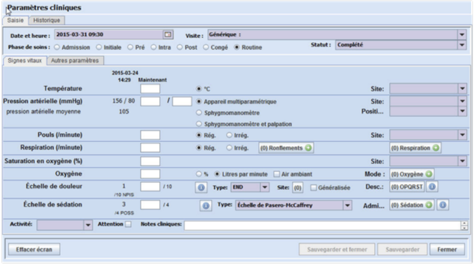
LABORATORY REPORT

Name Ana Betz — Patient ID PAC001
Date 2011-08-25 08:32 — Age 25y 10m 26d — Sex Female
Doctor Cameron Cordara — Test id B165AAF4

COMPLETE BLOOD COUNT

Test Name	Result	Normal Range	Units
Hemoglobin	12	11.0 - 16.0	g/dL
RBC	3.3	3.5-5.50	10^6/uL
HCT	36	37.0-50.0	%
MCV	83	82-95	fl
MCH	28	27-31	pg
MCHC	33	32.0-36.0	g/dL
RDW-CV	12	11.5-14.5	%
RDW-SD	44	35-56	fl
WBC	6.7	4.5-11	10^3/uL
NEU%	60	40-70	%
LYM%	30	20-45	%
MON%	8	2-10	%
EOS%	2	1-6	%
BAS%	0	0-2	%
LYM#	2	1.5-4.0	10^3/uL
GRA#	4.7	2.0-7.5	10^3/uL
PLT	256	150-450	10^3/uL
ESR	2	Up to 15	mm/hr

Digitally signed by
Dr. Cameron Cordara
GNU Public Key : E44311F4
Test id : B165AAF4



HEMATOLOGIE

NUMERATION ET FORMULE LEUCOCYTAIRE
(automate LH Beckman Coulter)

	Résultats	Normes
HEMATIES	5.320.000 /mm3	[illegible]
HEMOGLOBINE	16,5 g/100 ml	13,0 à 17,0
HEMATOCRITE	48,7 %	40,0 à 54,0
LEUCOCYTES	6.000 /mm3	4.000 à 10.000
PLAQUETTES	226.000 /mm3	150.000 à 500.000

VITESSE DE SEDIMENTATION

	Résultats	Normes
1ère heure	8 mm	inf. à 10

CHIMIE DU SANG

	Résultats	Normes
aspect du sérum	normal	
GLYCEMIE	1,08 g/l 5,99 mmol/l	[illegible]
UREE	0,33 g/l 5,48 mmol/l	[illegible]
CREATININEMIE	10 mg/l 88 umol/l	7 à 12 62 à 106
EXPLORATION LIPIDIQUE		
CHOLESTEROL TOTAL	2,86 g/l 7,38 mmol/l	inf. à 2,00 inf. à 5,16
H.D.L.	0,53 g/l 1,37 mmol/l	[illegible]
TRIGLYCERIDES	1,54 g/l 1,76 mmol/l	[illegible]
LDL CHOLESTEROL	2,02 g/l 5,23 mmol/l	[illegible]
PROTEINE C-REACTIVE	inf à 3 mg/l	inf. à 5,0

Medical Information

Narrative Patient-centered Data

```
Admission Date:  [**2015-03-17**]        Discharge Date:  [**2015-03-24**]
Date of Birth:   [**1974-10-03**]        Sex:  F
Service: Neurosurgery
HISTORY OF PRESENT ILLNESS:  The patient is a 40-year-old female with complaints
of headache and dizziness.  In [**2015-01-14**], the patient had headache with
neck stiffness and was unable to walk for 45 minutes.    [...]
PAST MEDICAL HISTORY:   Hypothyroidism.
ALLERGIES:  Penicillin and Bactrim which causes a rash.
MEDICATIONS:  Levoxyl 1.75 mg.
PHYSICAL EXAMINATION:  On physical examination, her blood pressure was 104/73,
pulse 79.  In general, she was a woman in no acute distress.  HEENT:  Nonicteric.
Pupils are equal, round, and reactive to light.  Extraocular movements are full.
[...]

On postoperative day #1, the patient was taken to arteriogram, where she
underwent a cerebral angiogram to evaluate clipping of the aneurysm.  []
DISCHARGE MEDICATIONS:
1. Hydromorphone 2-6 mg po q4h prn.
2. Synthroid 175 mcg po q day.[...]

CONDITION ON DISCHARGE:   Stable.

FOLLOW-UP INSTRUCTIONS:  She will follow up in 10 days for staple removal with
Dr. [**Last Name (STitle) 570**].

(End of Report)
```

Discharge summary extracted from the MIMIC II dataset `https://physionet.org/mimic2/`.

Medical Information

Primary Knowledge-based Documents

Cyberchondria: Studies of the Escalation of Medical Concerns in Web Search

RYEN W. WHITE and ERIC HORVITZ
Microsoft Research

The World Wide Web provides an abundant source of medical information. This information can assist people who are not healthcare professionals to better understand health and illness, and to provide them with feasible explanations for symptoms. However, the Web has the potential to increase the anxieties of people who have little or no medical training, especially when Web search is employed as a diagnostic procedure. We use the term *cyberchondria* to refer to the unfounded *escalation* of concerns about common symptomatology, based on the review of search results and literature on the Web. We performed a large-scale, longitudinal, log-based study of how people search for medical information online, supported by a survey of 515 individuals' health-related search experiences. We focused on the extent to which common, likely innocuous symptoms can escalate into the review of content on serious, rare conditions that are linked to the common symptoms. Our results show that Web search engines have the potential to escalate medical concerns. We show that escalation is associated with the amount and distribution of medical content viewed by users, the presence of escalatory terminology in pages visited, and a user's predisposition to escalate versus to seek more reasonable explanations for ailments. We also demonstrate the persistence of postsession anxiety following escalations and the effect that such anxieties can have on interrupting user's activities across multiple sessions. Our findings underscore the potential costs and challenges of cyberchondria and suggest actionable design implications that hold opportunity for improving the search and navigation experience for people turning to the Web to interpret common symptoms.

Categories and Subject Descriptors: H.3.3 [**Information Storage and Retrieval**]: Information Search and Retrieval—*Search process; query formulation*

General Terms: Human Factors, Experimentation

Additional Key Words and Phrases: Cyberchondria

ACM Reference Format:

White, R. W. and Horvitz, E. 2009. Cyberchondria: Studies of the escalation of medical concerns in Web search. ACM Trans. Inf. Syst. 27, 4, Article 23 (November 2009), 37
DOI = 10.1145/1629096.1629101 http://doi.acm.org/10.1145/1629096.1629101

1. INTRODUCTION

The World Wide Web has the potential to provide valuable medical information to people, where Web sites such as WebMD (http://www.webmd.com) and MSN

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DOI 10.1145/1629096.1629101 http://doi.acm.org/10.1145/1629096.1629101

ACM Transactions on Information Systems, Vol. 27, No. 4, Article 23, Publication date: November 2009.

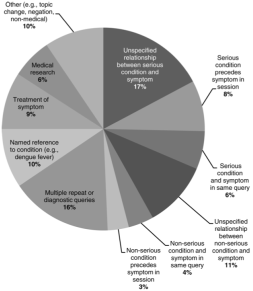

Fig. 1. Distribution of labels assigned to set of hand-labeled no-change sessions.

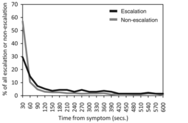

Fig. 2. Temporal distance from initial input of symptom (within session)

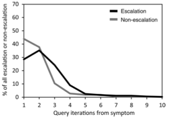

Fig. 3. Query distance from initial input of symptom (within session).

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Berland, G. K., Elliott, M. N., Morales, L. S., Algazy, J. I., Kravitz, R. L., Broder, M. S., Kanouse,

Medical Information

Primary Knowledge-based Documents

Clinical trials

Clinical trials are research studies that test a medical, surgical, or behavioral intervention in people. These trials are the primary way that researchers determine if a new form of treatment or prevention, such as a new drug, diet, or medical device (for example, a pacemaker), is safe and effective in people. Source: `https://www.nia.nih.gov/health/what-are-clinical-trials-and-studies`

Study Overview

Brief Summary:
The purpose of this study is to evaluate whether erythropoietin can help limit the damage to the heart in patients with acute heart attacks.

Detailed Description:
REVEAL is a randomized, double-blinded, placebo-controlled, parallel phase II clinical study that will evaluate the effects of erythropoietin administration on infarct size, left ventricular remodeling and circulating endothelial progenitor cells in patients with large myocardial infarctions (MI). The study will be conducted in two phases: a dose-escalation safety phase and a single dose efficacy phase. Eligible patients who present to the hospital with an acute ST-elevation MI and who agree to participate in this study will be randomly assigned to receive a single infusion of study medication consisting either of erythropoietin or placebo. The size of the infarction and the dimensions of the heart will be assessed by cardiac magnetic resonance imaging (MRI) within 2-6 days of the infusion of the study medication, and again approximately 3 months later.

— Show less

OFFICIAL TITLE

Effects of Erythropoietin on Infarct Size and Left Ventricular Remodeling in Survivors of Large **Myocardial Infarctions**

CONDITIONS

Acute ST Elevation Myocardial Infarction

INTERVENTION / TREATMENT

Drug: Epoetin alfa

STUDY START

2005-09

PRIMARY COMPLETION (ACTUAL)

2009-07

STUDY COMPLETION (ACTUAL)

2011-01

ENROLLMENT (ACTUAL)

223

STUDY TYPE

Interventional

PHASE

Phase 2

OTHER STUDY ID NUMBERS

999905255

05-AG-N255 (Other Identifier) (OTHER: NIH)

Eligibility Criteria

DESCRIPTION

- INCLUSION CRITERIA:

Age greater than 21 years

Acute ST-elevation myocardial infarction

Referral for primary or rescue angioplasty

Revascularization procedure within 8 hours from the onset of ischemic symptoms

TIMI (Thrombolysis in myocardial infarction) flow grade 0 or 1 in the culprit coronary artery at the beginning of coronary angiography

Successful revascularization of infarct-related artery

EXCLUSION CRITERIA:

Clinical indication for erythropoietin

STEMI (ST-elevation myocardial infarction) due to occlusion of a branch vessel

Any history of prior MI, PCI (Percutaneous coronary intervention), CABG (Coronary artery bypass graft), cardiomyopathy, myocarditis, or CHF (congestive heart failure)

Hypersensitivity to human albumin, mammalian cell-derived products, or erythropoietin

Hematocrit greater than 42% in men or greater than 40% in women at the time of study drug administration

Uncontrolled hypertension at the time of study drug administration

AGES ELIGIBLE FOR STUDY

21 Years and older (Adult, Older Adult)

SEXES ELIGIBLE FOR STUDY

All

ACCEPTS HEALTHY VOLUNTEERS

No

Medical Information

Secondary Knowledge-based Documents

- All medical professionals are not researchers: primary resources need to be rephrased, summarized, synthetized
- Summary and reviews of primary resources are published in scientific journals
- Quality issue: the editorial process is not the same for secundary than primary resources
- Other category: clinical practice guidelines (many publications, very little control)

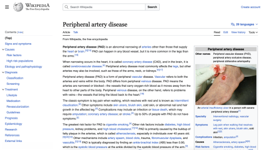
Specific case: Systematic Reviews and Meta-Analysis

- Fragmentation of the scientific literature → difficult to identify all the relevant papers on a topic
- In particular with clinical trials, large amount of publications on a similar condition or treatment
- Systematic reviews tackle a precise question, and describe the complete set of related work and factual approaches
- Meta-analysis compare results at the systematic review scale
- Topics: treatment (63%), causality and security (29%), diagnosis (4,4%), prognosis (2.1%) [Montori et al., 2004]

Medical Information

Curated Websites

1. Health portals
   - E.g. WebMD, Mayoclinic, MedlinePlus
   - Held by public/private institutions or companies
   - Varying quality and trustworthiness
2. Collaborative knowledge bases
   - E.g. Wikipedia, Radiopaedia, wikiDoc...
   - The literature provides strong evidence to position Wikipedia as a prominent health information resource for the public, patients, students, and practitioners seeking health information online [Smith, 2020]
   - 155,000 health articles using 950,000 citations to sources and which collectively received 4.8 billion pageviews in 2013 (all languages included)
3. Newspapers and magazines



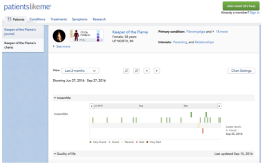
Medical Information

User Generated Content

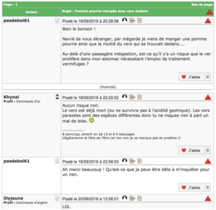
Health topics can be covered on all types of social media:

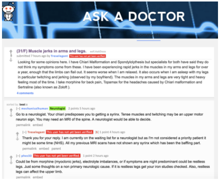
- General social media such as facebook, twitter:

- Medical social media such as PatientsLikeMe:



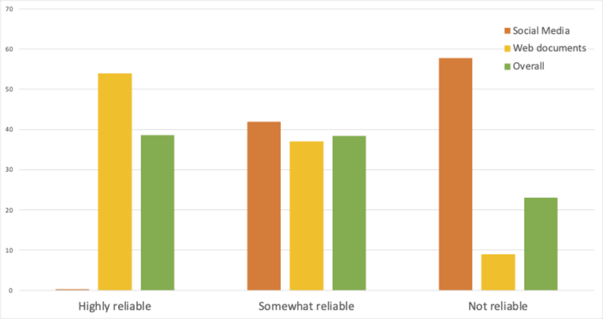
- Discussion forums: where all kinds of users (patients, doctors, students, nurses...) can discuss health topics





Medical Information

Trustworthiness of Health Information



- Within CLEF eHealth, the documents were manually assessed based on their topical relevance, readability and reliability [Goeuriot et al., 2021]
- 55 layuser queries on a broad range of topics
- 250 documents were assessed per query
- Documents in the pool come from CommonCrawl (71%) or social media Twitter and Reddit (29%)
- These results are in line with other studies such as [Scullard et al., 2010]

Medical Information

Certification

How can the quality of health information online be guaranteed?

The organization Health On the Net (HON) certifies the quality and validity of medical websites.
HON manually certifies website according to the following principles:

- Principle 1 : Authority - Give qualifications of authors
- Principle 2 : Complementarity - Information to support, not replace
- Principle 3 : Confidentiality - Respect the privacy of site users
- Principle 4 : Attribution - Cite the sources and dates of medical information
- Principle 5 : Justifiability - Justification of claims / balanced and objective claims
- Principle 6 : Transparency - Accessibility, provide valid contact details
- Principle 7 : Financial disclosure - Provide details of funding
- Principle 8 : Advertising - Clearly distinguish advertising from editorial content

https://www.hon.ch/HONcode/Guidelines/guidelines.html

Medical Information

Medical Textual Data - Summary

Data type	Characteristics	Challenges
	Patient-centered Data	
Structured	Description of patients signs Contains measures in some structured form and sometimes free text	Privacy Numerical data Multiple ambiguous signs Availability
Narrative	Communication between practitioners Record of an event in the patient care Form and content Free text	Privacy Negation and uncertainty Noisy writing Many ambiguities Temporal aspects Availability
	Knowledge-based Data	
Primary	Communication between experts Partially or fully available text Lightly structured - highly standardized High quality	References and links Contains tables and figures Quantity Availability
Secondary	Reliable information Good quality	Broad range of formats and content (and quality)
	Websites	
Curated	Wide range of topics covered All levels of readability Available data	Broad range of formats, content Lower quality
User-generated Content	Anyone can write and publish anything	No quality control Potentially unreliable data Potentially noisy data

Outline

Medical Search

Users and Tasks

Information needs [Hersh, 2010]:

- *Retrospective information needs*:
  - The need for help in solving a certain problem or making a decision
  - The need for background information on a topic
- *Current awareness information needs*:
  - The need to keep up with information in a given subject area

General classification of search queries from [Broder, 2002]:

- Navigational
- Transactional
- Informational

Some of the tasks well explored in IR:

1. Researchers
   - Cohort studies
   - Clinical trials search
   - Systematic reviews
   - Literature search
2. Clinicians
   - Evidence-based medicine
   - Precision medicine
   - Clinical trials search
3. General public
   - Information about a condition
   - Symptom check
   - Treatments
   - Advice and support

Medical Search Queries

Clinical Queries

Clinicians queries:

- Relatively short [Palotti et al., 2016a]
- Not systematically pursued: 51% pursued, and for 78% of them some answers were found [Del Fiol et al., 2014]

Evidence-based Medicine: For a given patient case, clinicians might need evidence in the biomedical literature to support a decision regarding: a diagnosis, a treatment, a test, etc. [Roberts et al., 2015b]:

Topic 1 – Diagnosis
Description: A 44 yo male is brought to the emergency room after multiple bouts of vomiting that has a "coffee ground" appearance. His heart rate is 135 bpm and blood pressure is 70/40 mmHg. Physical exam findings include decreased mental status and cool extremities. He receives a rapid infusion of crystalloid solution followed by packed red blood cell transfusion and is admitted to the ICU for further care.
Summary: A 44-year-old man with coffee-ground emesis, tachycardia, hypoxia, hypotension and cool, clammy extremities.

Precision Medicine: Adapt a clinical decision to a specific patient, upon genetic, environmental, and lifestyle choices (e.g. oncology) [Roberts et al., 2017]

Disease: Liposarcoma **Variant:** CDK4 Amplification **Demographic:** 38-year-old male **Other:** GERD	**Disease:** Colon Cancer **Variant:** KRAS (G13D), BRAF (V600E) **Demographic:** 52-year-old male **Other:** Type II Diabetes, Hypertension

Medical Search Queries

Clinical Queries

Analysis of search queries in an EHR search utility [Natarajan et al., 2010]

- **Navigational queries (14.5%)**: were mostly aiming at retrieving a specific EHR (e.g. using the record number)
- **Transactional queries (0.4%)**: were representing an action (e.g. adding a new note)
- **Information queries (85.1%)**: the most frequent, especially among clinicians and researchers.

Top 5 semantic types of searches

Semantic type	%	Semantic type	%
Laboratory or test result	29.2	Pharmacologic substance	7.5
Disease or syndrome	21.7	Diagnostic procedure	6.2
Body part, organ or organ component	8.1		

Top 10 most frequent queries

Query	%	Query	%
class	9.8	nephrogenic	1.8
nyha	4.5	hysterectomy	1.5
hodgkins	2.9	cva	1.1
iii	2.4	ef	1.0
iv	2.3	hf	0.9

- Very short queries (1.2 term(s) on average in the corpus)
- Many acronyms (*NYHA*) and abbreviations (*tach* for tachycardia)
- Ambiguous (*class*)

Medical Search Queries

PICO Queries

Designed to answer Evidence-based Medicine problems, PICO stands for:

- **P**atient / **P**roblem / **P**opulation
- **I**ntervention
- **C**omparison / **C**ontrol
- **O**utcome

The formulation of a focused clinical question containing well-articulated PICO elements is widely believed to be **the key to efficiently finding high-quality evidence** and also **the key to evidence-based decisions** [Huang et al., 2006].

Example (from [Boudin et al., 2010]):

`"children with pain and fever how does paracetamol compared with ibuprofen affect levels of pain and fever?`

$\longrightarrow$

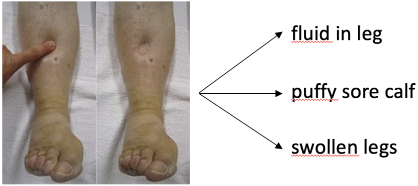
Patient/Problem: `children/pain and fever`
Intervention: `paracetamol`
Comparison: `ibuprofen`
Outcome: `levels of pain and fever`

Medical Search Queries

Layperson queries

Particularities and challenges [Zhang et al., 2012]

- *Conceptual level*: layperson have their own understandings and hypotheses about a particular condition.
- *Terminological level*: layperson's vocabulary doesn't match medical terminologies
- *Lexical level*: queries contain mispelling, partial words, etc.
- Short text (on average less than 3 words), ambiguous



- Circumlocution: "turning around" - when many words are used to describe what could be say with fewer, more precise words [Stanton et al., 2014a]
- Commercial search engine often answer poorly such queries [Zuccon et al., 2015]

[Cartright et al., 2011a]

Topics covered:

- Symptom
- Cause
- Remedy

Types of queries:

- Evidence-directed
- Hypothesis-directed:
  - Diagnosis intent
  - Informational intent

Outline

Semantic Resources

Definitions [Hersh, 2010, Bast et al., 2016]

A concept

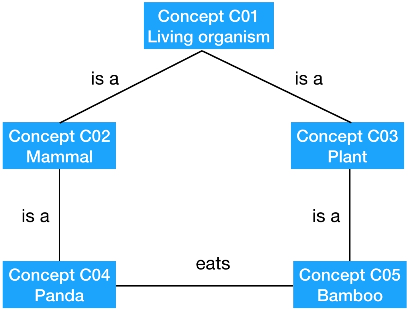
Idea or object that occurs in the world (e.g. *the condition under which human blood pressure is elevated*

A term

String of one or more words that represents a concept (e.g. *hypertension* or *high blood pressure*)

A relationship

Link between 2 concepts (e.g. the *liver* is an *organ*) or terms (e.g. *hypertension* and *high blood pressure* are synonyms)



A Knowledge Base is a collection of records in a database, which typically refers to some kind of knowledge about the world. Records are triples `(subject, predicate, object)`.

- [Bast et al., 2016] calls a *knowledge base* every collection of entities following an ontology.
- A **knowledge-base can be thought as a graph** where entities are the nodes and the relationships are the edges.

Medical Knowledge Resources

Existing Medical thesauri (in English)

1. Medical Subject Headings (MeSH)

- Created by the National Library of Medicine *to index medical documents*
- 28,000 descriptors (concepts) with over 90,000 entry terms
- 3 types of relationships: hierarchical, synonymous, related

Hypertension MeSH Descriptor Data 2018

Details | Qualifiers | MeSH Tree Structures | Concepts

MeSH Heading
Hypertension
Tree Number(s)
C14.907.489
Unique ID
D006973
Annotation
not for intracranial or intraocular pressure; relation to BLOOD PRESSURE: Manual 23.27; Goldblatt kidney is HYPERTENSION, GOLDBLATT see HYPERTENSION, RENOVASCULAR; hypertension with kidney disease is probably HYPERTENSION, RENAL, not HYPERTENSION; venous hypertension: index under VENOUS PRESSURE (IM) & do not coordinate with HYPERTENSION; PREHYPERTENSION is also available
Scope Note
Persistently high systemic arterial BLOOD PRESSURE. Based on multiple readings (BLOOD PRESSURE DETERMINATION), hypertension is currently defined as when SYSTOLIC PRESSURE is consistently greater than 140 mm Hg or when DIASTOLIC PRESSURE is consistently 90 mm Hg or more.
Entry Term(s)
Blood Pressure, High
NLM Classification #
WG 340
See Also
Antihypertensive Agents
Vascular Resistance
Date Established
1966/01/01
Date of Entry
1999/01/01
Revision Date
2010/06/25

The 16 trees in MeSH

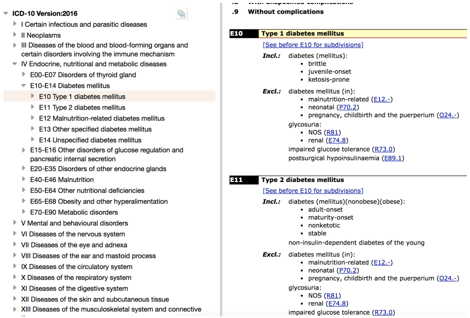
1. Anatomy
2. Organisms
3. Diseases
4. Chemicals and Drugs
5. Analytical, Diagnostic and Therapeutic Techniques and Equipment
6. Psychiatry and Psychology
7. Biological Sciences
8. Natural Sciences
9. Anthropology, Education, Sociology and Social Phenomena
10. Technology, Industry, Agriculture
11. Humanities
12. Information Science
13. Named Groups
14. Health care
15. Publication Characteristics
16. Geographicals

Medical Knowledge Resources

Existing Medical thesauri (in English)

2 International Classification of Medicine (ICD)

- International statistical classification of diseases and health problems
- Coded medical classification including a wide variety of signs, symptoms, trauma, etc.
- Published by the WHO
- Internationaly used to register morbidity and causes and morbidity



ICD-10 Version:2016
- I Certain infectious and parasitic diseases
- II Neoplasms
- III Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
- IV Endocrine, nutritional and metabolic diseases
  - E00-E07 Disorders of thyroid gland
  - E10-E14 Diabetes mellitus
    - E10 Type 1 diabetes mellitus
    - E11 Type 2 diabetes mellitus
    - E12 Malnutrition-related diabetes mellitus
    - E13 Other specified diabetes mellitus
    - E14 Unspecified diabetes mellitus
  - E15-E16 Other disorders of glucose regulation and pancreatic internal secretion
  - E20-E35 Disorders of other endocrine glands
  - E40-E46 Malnutrition
  - E50-E64 Other nutritional deficiencies
  - E65-E68 Obesity and other hyperalimentation
  - E70-E90 Metabolic disorders
- V Mental and behavioural disorders
- VI Diseases of the nervous system
- VII Diseases of the eye and adnexa
- VIII Diseases of the ear and mastoid process
- IX Diseases of the circulatory system
- X Diseases of the respiratory system
- XI Diseases of the digestive system
- XII Diseases of the skin and subcutaneous tissue
- XIII Diseases of the musculoskeletal system and connective

.9 Without complications

E10 Type 1 diabetes mellitus

[See before E10 for subdivisions]

Incl.: diabetes (mellitus):
- brittle
- juvenile-onset
- ketosis-prone

Excl.: diabetes mellitus (in):
- malnutrition-related (E12.-)
- neonatal (P70.2)
- pregnancy, childbirth and the puerperium (O24.-)

glycosuria:
- NOS (R81)
- renal (E74.8)

impaired glucose tolerance (R73.0)
postsurgical hypoinsulinaemia (E89.1)

E11 Type 2 diabetes mellitus

[See before E10 for subdivisions]

Incl.: diabetes (mellitus)(nonobese)(obese):
- adult-onset
- maturity-onset
- nonketotic
- stable

non-insulin-dependent diabetes of the young

Excl.: diabetes mellitus (in):
- malnutrition-related (E12.-)
- neonatal (P70.2)
- pregnancy, childbirth and the puerperium (O24.-)

glycosuria:
- NOS (R81)
- renal (E74.8)

impaired glucose tolerance (R73.0)

ICD Classification

1. Certain infectious and parasitic diseases
2. Neoplasms
3. Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
4. Endocrine, nutritional and metabolic diseases
5. Mental and behavioural disorders
6. Diseases of the nervous system
7. Diseases of the eye and adnexa
8. Diseases of the ear and mastoid process
9. Diseases of the circulatory system
10. Diseases of the respiratory system
11. Diseases of the digestive system
12. Diseases of the skin and subcutaneous tissue
13. Diseases of the musculoskeletal system and connective tissue
14. Diseases of the genitourinary system
15. Pregnancy, childbirth and the puerperium
16. Certain conditions originating in the perinatal period
17. Congenital malformations, deformations and chromosomal abnormalities
18. Symptoms, signs and abnormal clinical and laboratory findings, not elsewhere classified
19. ...

Medical Knowledge Resources

Existing Medical thesauri (in English)

3. Systematized Nomenclature of Medicine (SNOMED): thesaurus designed to process clinical data
4. Cumulative Index to Nursing and Allied Health Literature (CINAHL): classical medical concepts + domain-specific ones
5. EMTREE: European MeSH, used to index EMBASE
6. PsycINFO: psychology and psychiatry thesaurus
7. Gene Ontology: description of biomolecular biology (molecular functions, biological processes, cellular components) - designed to structure the knowledge rather than index content
8. National Cancer Institute (NCI) thesaurus: knowledge model enabling cross-disciplinary communication and collaboration

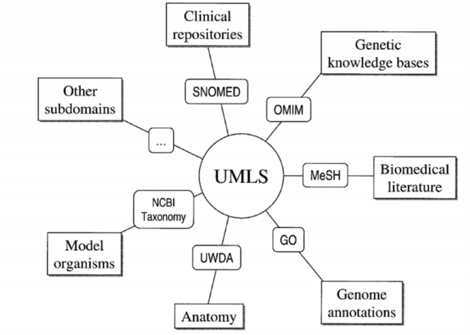
Many thesauri are also available in many well-endowed languages.

Medical Knowledge Resources

Existing Medical thesauri (in English)

The Unified Medical Language System (UMLS)

- Purpose: provide a mecanism to link existing medical thesaurus and controlled vocabularies
- Initiated in 1986 and maintained by the National Library of Medicine
- Contains: a metathesaurus, a semantic network, NLP tools
- Gathers more than 100 thesauri/vocabulary



Bodenreider, O. (2004) The Unified Medical Language System (UMLS): integrating biomedical terminology. *Nucleic Acids Research, 32, D267-D270.*

Medical Knowledge Resources

Existing Medical thesauri (in English)

The Unified Medical Language System (UMLS)

- All the entries corresponding to the same thing are registered as a single concept
- To each concept correspond several terms that represent an expression of the concept
- Each concept is described with: CUI, semantic type, definitions, synonyms, relations
- The information from all the thesauri containing the concept is stored

Concept (CUI)	Term (LUI)	String (SUI)	Atom (AUI)
C0004238 Atrial fibrillation (preferred) Atrial fibrillations Auricular fibrillation Auricular fibrillations	**L0004238** Atrial fibrillation (preferred) Atrial fibrillations	**S0016668** Atrial fibrillation (preferred)	**A0027665** Atrial fibrillation (from MSH)
			A0027667 Atrial fibrillation (from PSY)
		S0016669 Atrial fibrillations (plural variant)	**A0027668** Atrial fibrillations (from MSH)
	L0004327 Auricular fibrillation Auricular fibrillations (synonyms)	**S0016899** Auricular fibrillation (preferred)	**A0027930** Auricular fibrillation (from PSY)
		S0016900 Auricular fibrillations (plural variant)	**A0027932** Auricular fibrillations (from MSH)

Semantic Annotation

Semantic annotation consists in linking documents to knowledge bases by identifying:

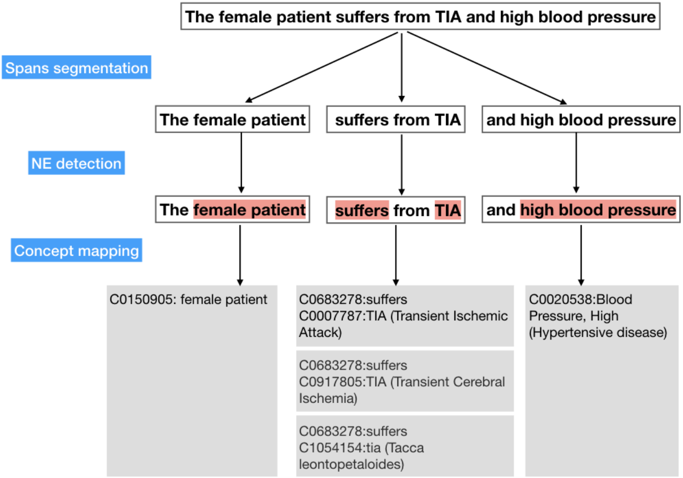
- Entities/concepts (in the document)
- Relationships (implicit or explicit):
  - Between entities: *HK1 involved in glycolytic process*
  - Between an entity and the document:
    - MeSH entities for indexing documents on MEDLINE: *PMID:3207429 is indexed with* `Glucose/metabolism` *and* `Hexokinase/genetics`
    - The ICD code in a medical report

Semantic annotation can be done:

- Automatically
  - Named Entity Recognition and normalization
  - Automatic indexing
- Manually
  - Data curation, manual labelling
  - Manual indexing (majority on MEDLINE)
  - Codes added to patients reports when billing
- Derived from other annotations
  - Using links between concepts and entities

Semantic Annotation

Named Entity Recognition and Linking



Existing Tools

Access to UMLS

- Requires a license (takes up to a few days - free)
- Gives access to the metathesaurus, along with other resources and tools created by the NLM
- `https://www.nlm.nih.gov/research/umls/index.html`

Existing tools for medical named-entity recognition:

Metamap : designed to annotate biomedical literature, interactive, API or batch versions available [Aronson and Lang, 2010]

QuickUMLS : designed to annotate medical text *faster* than other tools [Soldaini and Goharian, 2016]

cTakes : UIMA-based system designed to annotate EHRs [Savova et al., 2010]

SemRep : designed to extract semantic relations from biomedical text (as 3-parts propositions) [Rindflesch and Fiszman, 2003]

MedCAT : Self-supervised NER tool trained on EHR [Kraljevic et al., 2021]

MedSpacy, SciSpacy : python-based medical and biomedical NLP toolkits

SUMMARY

- Many types of data :
  - Patient-specific data
  - Knowledge-based data
  - User-generated data
- Various information needs: search for information, for cohorts, for evidence...
- On the solution side:
  - Some structured knowledge bases: UMLS and all its constituents (and their versions in languages other than English)
  - Several concept annotation tools: MetaMap, cTakes, QuickUMLS...
  - IR models
  - LLMs

Outline

Why is medical IR important?

A largely web-driven activity

- Search engines and social media are popular tools for seeking and sharing information about a range of health conditions
  [De Choudhury et al., 2014, White and Horvitz, 2014]. PewInternet, October 2013:

The % of adult internet users who have looked online in the last 12 months for information about...

55%	Specific disease or medical problem
43	Certain medical treatment or procedure
27	How to lose weight or how to control your weight
25	Health insurance, including private insurance, Medicare or Medicaid
19	Food safety or recalls
16	Drug safety or recalls
16	A drug you saw advertised
15	Medical test results
14	Caring for an aging relative or friend
12	Pregnancy and childbirth
11	How to reduce your health care costs
20	Any other health issue
72	**at least one of the above topics**

- Existence of behavioral patterns suggesting a strong relationship between search behavior and health care [White and Horvitz, 2010, White and Horvitz, 2014]

What makes medical search challenging?
Overview

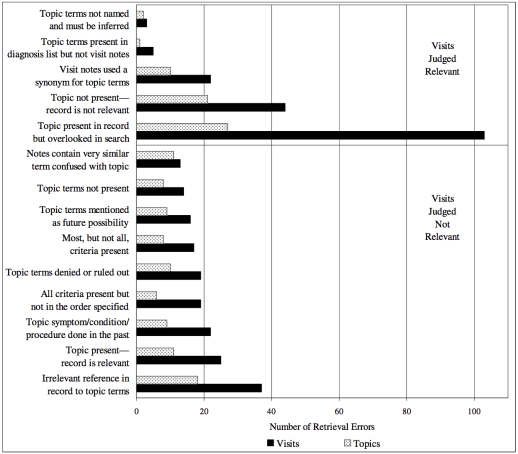
		Lexical representation	Lexical Matching	Result appraisal
Text (Queries/Documents)	High occurrence of lexical variants	■	■	■
	Ambiguity of language	■	■	■
	Significant presence of negation	■	■	■
	Time factors	■	■	■
Search task	Domain-specific task	□	■	■
User	Variability in levels of expertise	■	□	■
	Difficulty of understanding medical language	□	□	■
	Cognitive bias	□	□	■

- Lexical representation [Tamine et al., 2015, Stanton et al., 2014b, Limsopatham et al., 2013, Edinger et al., 2012, Chapman et al., 2001]
- Lexical matching [Edinger et al., 2012, Dinh and Tamine, 2012]
- Result appraisal [Koopman and Zuccon, 2014, Tamine and Chouquet, 2017, Roberts et al., 2015a, W. White and Horvitz, 2009, White and Horvitz, 2013, Cartright et al., 2011b, Palotti et al., 2016b]

What makes medical search challenging?

Lexical representation and matching issues

- Failure analysis from the TREC Medical Records Track [Edinger et al., 2012]
  - Same task across queries: retrieve cohorts of patients fitting criteria similar to those specified for participation in clinical studies
  - Same user's profile: domain-expert (physician)
  - Main results: **both precision errors and recall errors were due to bad lexical representations and lexical mismatches**



What makes medical search challenging?

Domain-specific tasks: need of task-dependent matching?

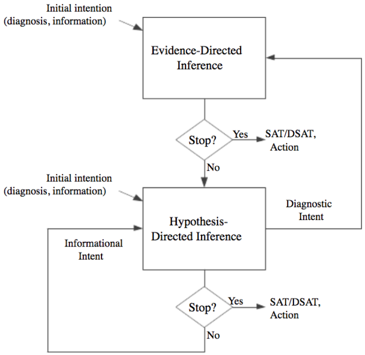
Study of the peculiarities of queries across medical tasks [Tamine et al., 2015]

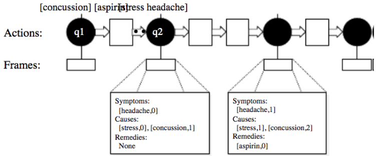
- Study of 4 search tasks:
  - (T1) Retrieve from biomedical scientific literature relevant references that address biology or related protein products (eg., '*Arginin vassopressin*')
  - (T2) Retrieve from medical scientific literature, relevant documents that fit with a patient profile (eg., '*Adult respiratory distress syndroms*')
  - (T3) Identify cohorts in clinical studies for comparative effectiveness research (eg., *Retrieve relevant medical cases including images for differential diagnosis*)
  - (T4) Identify relevant references that deal with typical relations between an entity and a medical process (eg., *What is the role of gene gamma-aminobutyric acid receptors in the process of inhibitory synaptic transmission?*)
- Same user's profile: domain-expert (physician)
- Main results:
  - Queries vary significantly across tasks:
    have multiple topical facets expressed using different levels of specifity w.r.t. medical terminology
  - Query performance significantly vary across tasks:
    however, the shorter and less specific the query is,
    the more difficult it is regardless of the task

What makes medical search challenging?

Domain-specific tasks: need of task-dependent matching?

- Diagnosis, a common medical search setting, is a highly complex task [Cartright et al., 2011b]
- Identifying two medical foci in search sessions corresponding to two iterative and interactive phases:
  - Evidence-directed: findings are merged to build a list of potential explanatory diagnoses ranked by likelihood
  - Hypothesis-directed: list of diagnoses used to guide collection of additional evidence to validate candidate hypotheses



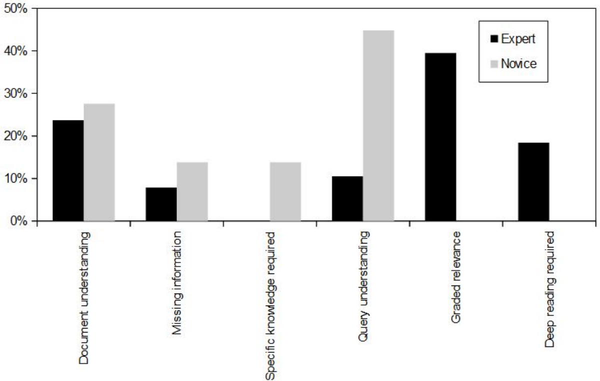


What makes medical search challenging?

Result appraisal: what does make it difficult with respect to user's expertise?

Study the factors of the relevance assessment task difficulty [Tamine and Chouquet, 2017]

- Qualitative analysis of relevance assessments for equivalent information needs
- Different user's profiles: domain-expert (physician), domaine-novice (laypeople)
- Main results:
  - Levels of relevance agreement are low for both experts and novices
  - Better level of relevance agreement among experts than novices
  - More than third of the assessors found the relevance assessment task difficult but the underlying reasons different among experts vs. novices



What makes medical search challenging?

User cognitive bias - Cybercondria

- *Cybercondria: Study the potential of web search to escalate user's medical concerns* [White and Horvitz, 2013, W. White and Horvitz, 2009]
  - Qualitative and quantitative analysis of users' medical search logs and surveys
  - Observe the escalation within session: severity increase in the search terms of evolving medical search sessions
  - Main results:
    - Using Web search to perform diagnosis is a common user activity (more than 24%) while general-purpose ranking functions are based on the presence of lexical query symptoms, not designed for diagnosis inference
    - Common users consider that the system ranks the potential explanatory diagnoses by likelihood

Table I. Probability of Mention of Cause Given Symptom

Symptom	Cause	Web Crawl	Web Search	Domain Search
headache	caffeine withdrawal	.29	.26	.25
	tension	.68	.48	.75
	brain tumor	.03	.26	.00
muscle twitches	benign fasciculation	.53	.12	.34
	muscle strain	.40	.38	.66
	ALS	.07	.50	.00
chest pain	indigestion	.28	.35	.38
	heartburn	.57	.28	.52
	heart attack	.15	.37	.10

Summary of issues

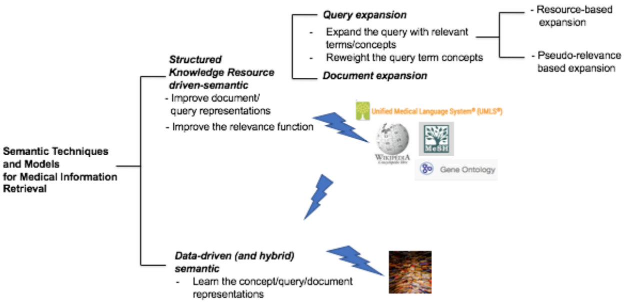
- Semantic gap
  - Vocabulary mismatch between experts and novices
  - Vocabulary mismatch between the query and documents
  - Interpretation of vocabulary in context: negation, lexical variants, time, task peculiarities
- Result appraisal
  - Understand medical language
  - Ability to interpret the results, make accurate inference, assess the credibility
  - Cognitive bias

OUTLINE

Outline

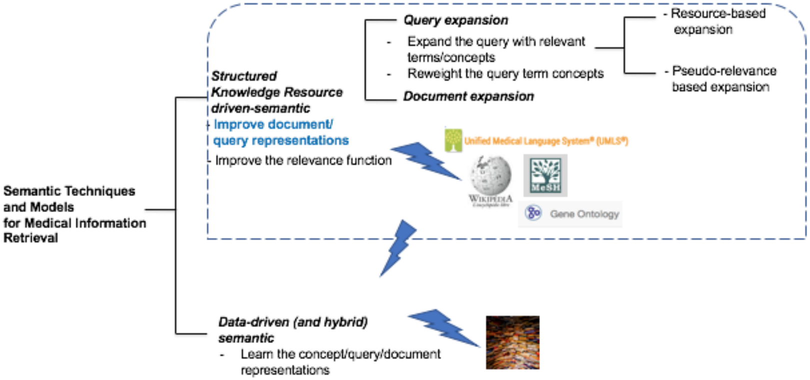
Roadmap

- Traditional IR
  - Q: bag of words
  - D: bag of words
  - RSV(Q,D): Alignment of Q and D
- Semantic (medical) IR
  - Q:
    - Bag of words
    - Bag of words **and concepts/entities**
    - **Embeddings**
  - D:
    - Bag of words
    - Bag of words **and concepts/entities**
    - **Embeddings**
  - RSV(Q,D): **Semantic inference**



Outline

Roadmap


Semantic Techniques and Models for Medical Information Retrieval
Structured Knowledge Resource driven-semantic
Improve document/ query representations
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Query expansion
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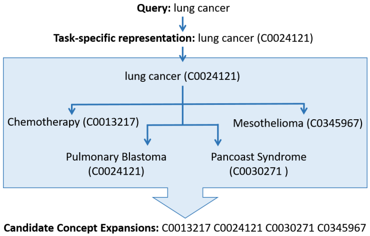
Document expansion
- Resource-based expansion
- Pseudo-relevance based expansion
Unified Medical Language System® (UMLS®)
Wikipedia
MeSH
Gene Ontology
Data-driven (and hybrid) semantic
- Learn the concept/query/document representations

Query/document expansion

- Query/document expansion
  - Enhance the Query/Document using:
    - evidence from related words/terms in semantic resources;
    - relevance feedback signals



QUERY/DOCUMENT EXPANSION

- Query/document expansion
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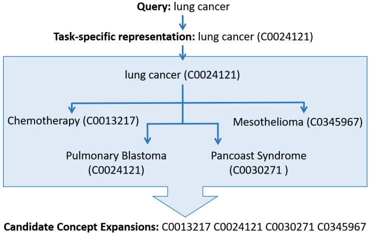
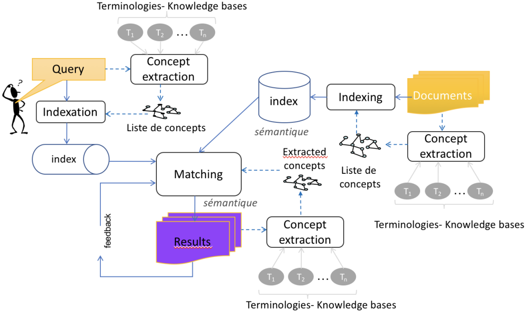


  - The full framework



QUERY/DOCUMENT EXPANSION

- Main impacting factors: [Dinh et al., 2013, Zuccon and Koopman, 2018]
  - Which knowledge-base to use (specialized vs. generic) and how many?
  - Which context to use (global vs. local)?
  - How to select candidate expansion terms and (how to inject them in a retrieval model) ?

Query/document expansion

- Main impacting factors: [Dinh et al., 2013, Zuccon and Koopman, 2018]
  - Which knowledge-base to use (specialized vs. generic) and how many?
  - Which context to use (global vs. local)?
  - How to select candidate expansion terms and (how to inject them in a retrieval model) ?
- Resulting techniques
  - LSMo: Local context, Specialized Mono-Resource [Soldaini et al., 2017, Sondhi et al., 2012]
  - GSMo: Global context, Specialized Mono-Resource [Martinez et al., 2014, Znaidi et al., 2016, Demner-Fushman et al., 2006, Shen et al., 2014, Zhu et al., 2013]
  - GSMu: Global context, Specialized Multiple-Resources [Ando et al., 2005, Huang et al., 2005]
  - LGSMo: Local and Global contexts, Specialized Mono-Resource [Wang and Akella, 2015, Znaidi et al., 2015, Znaidi et al., 2016, Xu et al., 2019]
  - GSGMu: Global context, Specialized General Multiple-Resources [Soldaini et al., 2016]
  - LGSM : Local and Global contexts, Specialized Multiple-Resources [Limsopatham et al., 2013, Dinh and Tamine, 2012, Oh and Jung, 2015, Zhu and Carterette, 2012, Balaneshinkordan and Kotov, 2019, Ai et al., 2006]

		LSMo	GSMo	LGSMo	GSGMu	LGSMu
Context	Local (Pseudo-relevance)	■	□	■	□	■
	Global (Resource)	□	■	■	■	■
Knowledge Base	Specialized	■	■	■	■	■
	General	□	□	□	■	□
	Mono-resource	■	■	■	□	□
	Multiple-resources	□	□	□	■	■

Query/document expansion

Local context, one general resource [Soldaini et al., 2017]

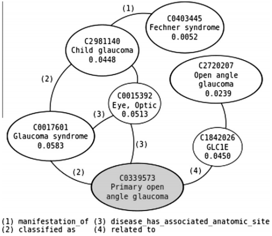
- **Goal:** Query reformulation for clinical decision support
- **Context:** Top N retrieved documents
- **Knowledge-Base**: Wikipedia

- **Key steps: Health Terms Pseudo Relevance Feedback HTPRF**
  - Retrieve the N Top documents w.r.t query Q
  - For each term from the top N documents, compute a score
    $s_j = log_{10}(10 + w_j)\, w_j = \alpha * tf(t_j, Q) + \frac{\beta}{N} \sum_1^N (tf(t_j, D_i) * idf(t_j))$
  - Select the top M terms with the highest score as the candidate expansion terms
  - For each candidate term expansion, compute the likelihood of being health-related. Compute the odds ratio as the proportion of health-related Wikipdia (W_H) documents including term t_i
    $OR(t_i) = \frac{n(t_i, W_H)}{n(t_i, W)}$
  - Consider the top M ranked terms with score $OR(t_j) > \sigma$
  - Expand the query and perform a pseudo-relevance feedback based model
- **Main results/findings**
  - Mapping the wikipedia terms to UMLS semantic types revealed that 75% are present in the UMLS: 32% are symptoms, 20,3% are treatments, 18% are a diagnosis procedure or test,17,1% are diseases
  - The HTPRF parameters do not significantly impact the results
  - Precision oriented with slight improvement (+3,6%) over state-of the best systems in TREC CDS 2014-TREC CDS 2015

Query/document expansion

Global context, one specialized resource [Martinez et al., 2014]

- **Goal**: Searching EHRs
- **Context**: Concepts and relations between concepts
- **Knowledge-Base**: UMLS thesaurus
- **Key steps**
  - Map query words to UMLS semantic types
  - Identify the initial sub-graph based concept including query concepts and related UMLS concepts
  - Assign an uniform probability to the concepts in the sub-graph and then run the Page Rank algorithm
  - Rank the concepts using the Page Rank score
  - Expand the query with the N concepts having the highest PageRank Score
  - Perform a basic retrieval model (eg., TF-IDF, BM25)
- **Main results/findings**
  - Experiments on TREC medical records 2011-2012 show significant improvements (+30% in average)
  - Expansion terms are those related to the query with either taxonomic (eg., synonyms) and not taxonomic (eg., disease has associated anatomic site).
  - Useful expansion in the case of a cohort retrieval task.



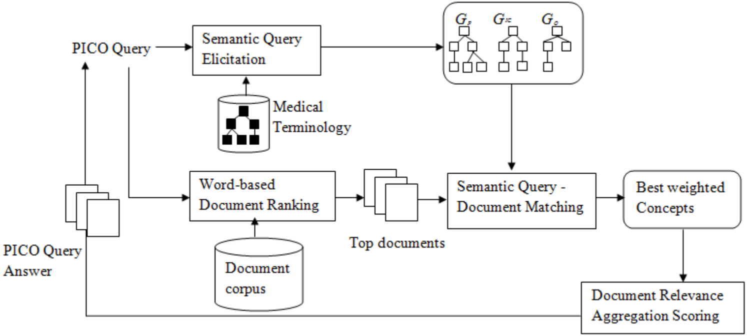
Queries with highest improvement for PageRank, together with the learnt expansion terms and the Bpref increase.

Query	TREC version	Expansion terms	Bpref increase
Hospitalized patients treated for methicillin-resistant Staphylococcus aureus (MRSA) endocarditis	2011	MRSA elsewhere/NOS Personal history of poliomyelitis Personal history of other infectious and parasitic disease	0.931
Patients with Primary Open Angle Glaucoma (POAG)	2012	Eye, Eyeball, Globe, Ocular… Glaucoma syndrome Open cleft glaucoma GLC1E	0.742
Patients with adult respiratory distress syndrome	2012	Immunology Taxonomy Metabolism Historical aspects	0.722

Query/document expansion

Global and local contexts, one specialized resource [Znaidi et al., 2016]

- **Goal:** answer PICO queries for Evidence Based Medicine
- **Context:** Concepts and relations between concepts, Top N retrieved documents
- **Knowledge-Base**: UMLS thesaurus



Query/document expansion

Global and local contexts, one specialized resource [Znaidi et al., 2016]

- **Goal:** answer PICO queries for Evidence Based Medicine
- **Context:** Concepts and relations between concepts, Top N retrieved documents
- **Knowledge-Base**: UMLS thesaurus

- **Key steps: tailored for PICO queries**
  - Map each PICO facet of query Q to UMLS
  - Identify the UMLS query concepts for each facet
  - Build a concept-based tree for each facet
  - Build the candidate query expansion terms using a recursive propagation algorithm from active concept *c* through sub-concepts *csub*
    - Compute $score(c) = +level(csub) * score(s)$
  - Expand the query with the N concepts having the highest score
  - Apply a prioritized-scoring based retrieval

$$RSV_{PICO}(Q, d) = \lambda_P * RSV_P(Q_P, d) + \lambda_{IC} * RSV_{IC}(Q_{IC}, d) + + \lambda_O * RSV_O(Q_O, d)$$

$$\lambda_{IC} = 1, \lambda_P = \lambda_{IC} * RSV(Q^c_{IC}, d), \lambda_O = \lambda_P * RSV(Q^c_P, d)$$

  - Combine the PICO score with a classic score (eg., BM25)

- **Mains findings**
  - Significant improvements (+30%) on a standard PICO collection
  - Slight but non significant improvement in comparison to PICO tailored models
  - The model performs better for relatively short queries (still be long in comparison to classic queries)
  - For long queries, it is more likely that the document matches the query according to different facets but with misleading interpretations of the search intent.

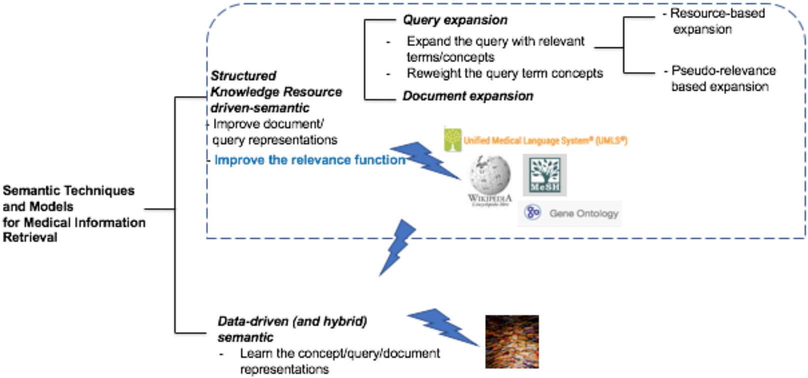
Query/document expansion

Let us discuss

- A large body of state-of-the art work in medical IR rely on query expansion techniques
  - Combine statistical concepts and semantic concepts
  - Exploit generally evidence from multiple resources according to the polyrepresentation view
  - Use generally a local relevance model

- Robustness of query expansion techniques in the medical domain is questionable
  - They do not systematically lead to significant and important improvements over baselines
  - Several impacting factors on performance:
    - The knowledge bases used: general vs. specific
    - The semantic relations exploited: taxonomic vs. non taxonomic
    - The task at hand (collection): searching for cases, scientific literature, etc.

ROADMAP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Relevance ranking

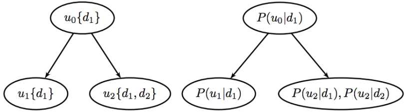
- How to incorporate semantics in the document relevance estimation?
  - Ranking as a semantic inference
    [Goodwin and Harabagiu, 2016, Koopman et al., 2016, Cao et al., 2011]
  - Ranking as learning the discriminant relevant (semantic) features
    [Balaneshin-kordan and Kotov, 2016, Xiong and Callan, 2015, Soldaini and Goharian, 2017]

Relevance ranking

Ranking as a semantic inference: a graph-based approach [Koopman et al., 2016]

- **Goal:** Adhoc medical search
- **Key model components**
  - Graph-based representation of the documents
  - Document ranking as an inference process over related concepts in the graph
  - Knowledge resources with directed relationships between concepts
  - Different types of relationships
- **Key inference rationale:** tune the inference mechanism according to semantic gap issues: lexical mismatch, granularity mismatch, conceptual mismatch
  - Lexical mismatch (eg., *hypertension* vs. *high blood pressure*): association and deductive inference
  - Granularity mismatch (eg., *antipsychotic* and *Diazepman*): introduce uncertainty in the taxonomic (hierarchical eg., IS A) relationships
  - Conceptual mismatch (eg., *treatments* $\rightarrow$ *disease*): deductive inference and logical deduction

- The Graph-based corpus representation



(a) Basic node-document representation. (b) Representation with initial probabilities assigned to node.

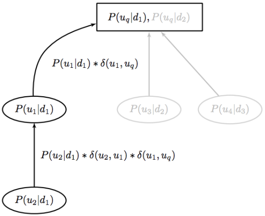
Relevance ranking

Ranking as a semantic inference: a graph-based approach [Koopman et al., 2016]

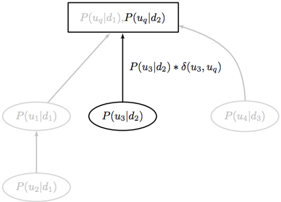
- **The retrieval model**
  - Strength of the association between two information nodes: compute recursively over the graph:

$$\sigma(u,u') = \begin{cases} 1 \text{ if } u = u' \\ \sigma_0(u,u') \text{ if } uRu' \\ argmax_{u_i \in U: uRu_i} \sigma(u,u_i) \times \sigma(u_i,u'), otherwise \end{cases} \quad (1)$$

  - Relevance of document-query $RSV(d,q) = \prod_{u_q \in q} \prod_{u_d \in d} p(u_d \mid d)\sigma(u_d,u_q)$
- **Main results/findings**
  - Effective improvement of queries suffering from the conceptual implication problem
  - Degradation for 'simple' queries do not requiring inference. Inference highlighted general irrelevant concepts



(a) Retrieval process for document d_1.



(b) Retrieval process for document d_2.

Relevance ranking

Ranking as learning the concept-document weighting features [Balaneshin-kordan and Kotov, 2016]

- **Goal:** Retrieve medical literature from clinical cases
- **Key model components**
  - Document ranking as learning the optimal query concept weighting based on the intuition that different concept types have different importance
  - A set of features characterizing query concept computed on the basis of knowledge-bases (Wikipedia, UMLS) as global context and top retrieved documents as local context of the query
- **The learning process**: A multivariate optimization method to train the weights of all features as contributions to fix the concept weight w.r.t. a document
- **The retrieval model**
  - Relevance status value based on the Markov Random Field retrieval framework [Metzler and Croft, 2005]: weighted linear combination of matching scores of concepts types in a query: $sc(Q, D) = \sum_{c \in C_T} \lambda_T(c) * f_T(c, D)$
  - Importance weight of concept c computed as a linear combination of importance features: $\lambda_T(c) \sum_{n=1}^{N} w_{\phi}^{n} \phi^{n}$
  - Consider global features (eg., popularity concept node in the UMLS graph) and local features (eg., Number of top retrieved documents containing concept c)
  - Learn $\lambda_T(c)$ using a multivariate optimization solving problem.
- **Main results/findings**
  - Reasonable performance improvement (5-9%) over the baselines according to search accuracy
  - Exploiting evidence from multiple resources (Wikipedia and UMLS) positively affects performance

Relevance ranking

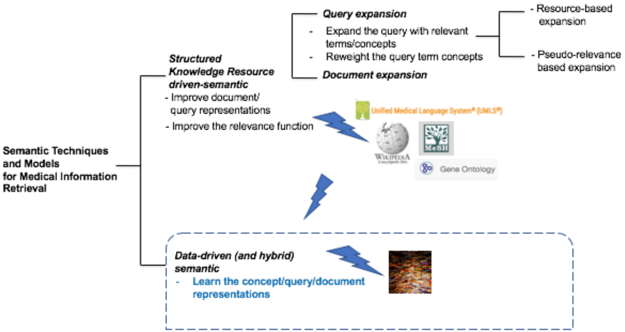
Let us discuss

- **A few work addressed the semantic search at the relevance function level**
  - Identify logical matching between words and concepts
  - Identify relevant semantic features that connect words to concepts, queries to documents

- **Findings: the general trend**
  - High-level inference yields to high computational complexity
  - The good balance between lexical matching and semantic matching is difficult to tune
  - Robustness to concept annotation quality is important

Outline

Learning


Semantic Techniques and Models for Medical Information Retrieval
Structured Knowledge Resource driven-semantic
- Improve document/ query representations
- Improve the relevance function
Query expansion
- Expand the query with relevant terms/concepts
- Reweight the query term concepts
Document expansion
- Resource-based expansion
- Pseudo-relevance based expansion
Unified Medical Language System® (UMLS®)
Wikipedia
MeSH
Gene Ontology
Data-driven (and hybrid) semantic
- Learn the concept/query/document representations

Representation learning for medical search

Overview of early research

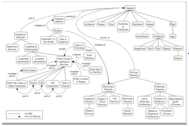
- What do the models learn? (Pre-BERT)
  - *Word, CUI, entity, phrase embeddings*: to learn high-level similarity between information units [De Vine et al., 2014, Limsopatham and Collier, 2016, Liu et al., 2016, Ghosh et al., 2017, Jagannatha and Yu, 2016, Cai et al., 2018, Henry et al., 2018]
  - *Document embeddings*: to improve semantic representations of texts that bridge the gap between data-driven semantic and knowledge resource driven semantic [Minarro-Gimnez et al., 2014, Nguyen et al., 2017, Loza Mencía et al., 2016, Peng et al., 2016, Banerjee et al., 2017, Nguyen et al., 2018a]
  - *Medical objects embeddings*: care events/episodes, disease embeddings [Ghosh et al., 2016, Moen et al., 2015, Choi et al., 2016], patient embeddings [Baytas et al., 2017, Ni et al., 2017, Zhu et al., 2016, Stojanovic et al., 2017, Sushil et al., 2018]
- For which search tasks?
  - Relevance matching RM (eg., document retrieval, care-episode retrieval)
  - Semantic matching (eg., patient similarity)
- Transformer-based IR:
  - Encoding of queries and documents [Jin et al., 2023, Ueda et al., 2021]
  - (Multiple) re-ranking phases [Pradeep et al., 2022, Biester et al., , Tahami et al.,]
  - Query/document classification [Wang et al., 2022]
  - Data augmentation/generation [Pradeep et al., 2022, Karimi, , Bondarenko et al., 2022]
  - Combinations of all the above

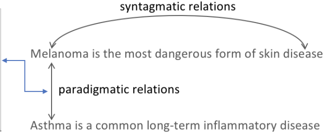
Representation learning for medical search

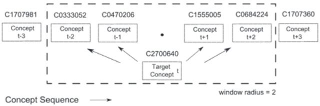
Learning Word, CUI, entity, phrase embeddings

- Different purposes yield to different objective functions
  - Learn readable concept/CUI/entity representations from raw texts: driven by paradigmatic relations provided in knowledge-bases
  - Learn concept and associated poly-senses: learn one vector representation per sense
- Different neural architectures
  - Extension of the CBOW and Skip-Gram models
  - Deep architectures (CNN, RNN, ...)
  - Transformer-based models

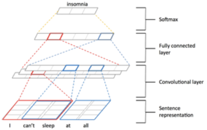
Medical thesaurus



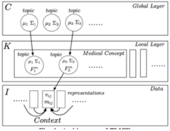




Devine et al. 2014



Limsopatham et al. 2016



Qian et et al. 2017

Representation learning for medical search

Learning Word, CUI, entity, phrase embeddings [De Vine et al., 2014, Liu et al., 2016]

- Learn word meaning representation from language models, in order to perform a concept similarity task - extension of the Skip-Gram model [De Vine et al., 2014]
  - Learn UMLS concept representations from sequences of concepts in annotated texts
  - Maximize the average log probability of the objective function $\frac{1}{2r}\sum_{i=1}^{2r}\sum_{-r\leq j\leq r} log(c_{t+j} \mid c_t)$
  - Valid representations when compared to human-assessments within a concept similarity task
  - Requires huge amount of annotated data.
  - Sensitivity to concept annotation quality?

- Extension of the CBOW model [Liu et al., 2016]
  - Learn concept representations constrained by relations established in a knowledge base
  - Maximize the log probability of the objective function $L = \sum_{i=1}^{T}(log(p(w_t \mid w_{t+k}) + \alpha \sum_{w_s:(w_t,w_s)\in R} wt(w_s \mid w_t)(logp(w_t \mid w_{t\pm k} - logp(w_s \mid w_{s\pm k})))^2)$
    $w_t(w_s \mid w_t) = \frac{f(w_s)}{\sum_{(w_t,w)\in R} f(w)}$
  - Experimental evaluation on IR tasks (query expansion) show: 1) sensitivity to model parameters and collections; 2) ability to select related words in the UMLS thesaurus; 3) slight improvement on a medical document search task

The most similar words to « heart »

CBOW		Online	
Cardiac	0.4891	Cardiac	0.5205
Synergist	0.4494	Hearts	0.5030
Hearts	0.4276	Cor	0.4939
Cardiovascular	0.4096	Synergist	0.4690
Acyanotic	0.3987	Cardiovascular	0.4156
Ouvrier	0.3934	Cerebrovascular	0.4149
Multiorgan	0.3931	Acyanotic	0.3985
Ventricular	0.3837	Ventricular	0.3979
Cardiorespiratory	0.3829	Cardiorespiratory	0.3969
Thrive	0.3766	Biventricular	0.3831

Representation learning for medical search

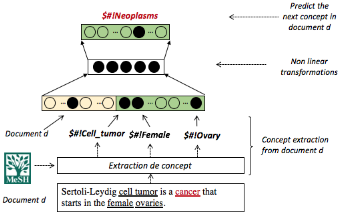
Learning document embeddings [Minarro-Gimnez et al., 2014, Nguyen et al., 2017, Loza Mencía et al., 2016, Peng et al., 2016, Banerjee et al., 2017, Nguyen et al., 2018a]

- Extension of the Doc2Vec model based on an **offline learning** approach [Nguyen et al., 2017]
  - Build the optimal real-valued representation $\hat{d}$ of document d such that the knowledge-based embedding $\hat{d}_i^{(cd2vec)}$ and the corpus-based embedding $\hat{d}^{(PV-DM)}$ are nearby in the latent space. Formally through the minimization problem:

$$\Psi(D) = \sum_{d \in D} \psi(d) = \sum_{d \in D} \left[(1-\beta) \times \|d - \hat{d}^{(cd2vec)}\|^2 + \beta \times \|d - \hat{d}^{(PV-DM)}\|^2 \right]$$

  - Concept-based latent representation of document d is obtained using and extension of the *cd2vec* model. Document vectors $\hat{d}^{(cd2vec)}$ are learned so they allow predicting concepts in their context by maximizing the log-likelihood:

$$\varphi = \sum_{d \in D} logP(d \mid c_1, \ldots, c_m) + \sum_{c_j \in \mathcal{C}_d} logP(c_j \mid c_{j-W} : c_{j+W}, d)$$



Representation learning for medical search

Learning document embeddings [Minarro-Gimnez et al., 2014, Nguyen et al., 2017, Loza Mencía et al., 2016, Peng et al., 2016, Banerjee et al., 2017, Nguyen et al., 2018a]

- Evaluation in a retrieval task [Nguyen et al., 2017]
  - Significant but slight improvement using a query expansion task
  - The model is sensitive to the quality of concept annotation
  - The model allows identifying relevant related concepts

Table: Example of terms/concepts expanded for query 131 in TREC Med

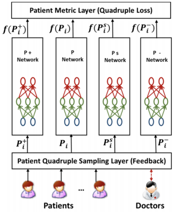
Query text	patients underwent minimally invasive abdominal surgery
Extracted Concepts	Patients; General Surgery;
Added by $Exp_{\hat{d}PV-DM}$	myofascia; ultrasonix; overtube
Added by $Exp_{\hat{d}cd2vec}$	Mesna; Esophageal Sphincter, Upper; Ganglioglioma
Added by Exp_{d}	*umbilical*; *ventral*; *biliary-dilatation*

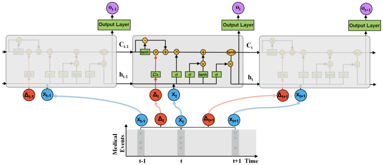
Representation learning for medical search

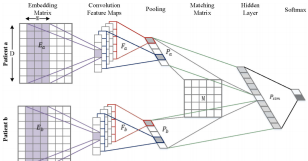
Learning patient profiles, patient similarity

[Baytas et al., 2017, Ni et al., 2017, Zhu et al., 2016, Stojanovic et al., 2017, Sushil et al., 2018]

- Two main objectives
  - Learn patient profile: input (EHR) - output (patient vector) [Baytas et al., 2017, Stojanovic et al., 2017, Sushil et al., 2018]
  - Learn end-to-end patient-patient similarity:
    - Input: EHR patient A, EHR patient B
    - Output: similarity class[Zhu et al., 2016, Ni et al., 2017]
- Input data
  - Heterogeneous patient data: demographic, medication, diagnosis codes etc.
  - Historical data: considering the sequence of medical events with irregular intervals
- Tasks
  - Predict patient mortality, primary diagnosis, length of stay, total incurred charges, ...







Representation learning for medical search

Learning patient profiles, patient similarity

[Baytas et al., 2017, Ni et al., 2017, Zhu et al., 2016, Stojanovic et al., 2017, Sushil et al., 2018]

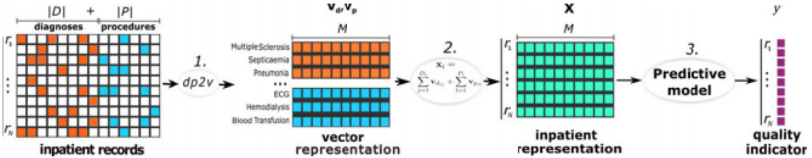
Learning patient vector from patient disease and procedure descriptions [Stojanovic et al., 2017]

- Learn the representations of diseases and procedures (dp2vec): extend the w2vec model by considering patient record as a "sentence" and diseases/procedures as "words"
- Build patient vector: sum the vector representations of in-patient diseases and procedures

Task evaluation

- Predict length of patient stay and total charges as a regression problem (vector representations used as features)
- Predict mortality as a classification problem

Neighbors of *respiratory failure*	Neighbors of *congestive heart failure*
Insertion of endotracheal tube	Insertion of implantable heart assist system
Tracheostomy toilette	Implantation of cardiac resynchronization defibrillator total system (CRT-D)
Other lavage of bronchus and trachea	Implantation of cardiac resynchronization defibrillator pulse generator (CRT-D)
Bronchoscopy through artificial stoma	Insertion of percutaneous external heart assist device
Other oxygen enrichment	Heart transplantation
Other repair and plastic operations on trachea	Excision destruction or exclusion of left atrial appendage (LAA)
Fiber-optic bronchoscopy	Aquapheresis
Infusion of vasopressor agent	Automatic implantable cardioverter-defibrillator (AICD) check
Replacement of tracheostomy tube	Noninvasive programmed electrical stimulation (NIPS)
Replacement of gastrostomy tube	Removal of lead(s) [electrode] without replacement
Complete glossectomy	Endovascular removal of obstruction from head and neck vessel(s)
Other intubation of respiratory tract	Replacement of automatic cardioverter-defibrillator lead(s) only



Representation Learning for Medical Search

Transformer-based models

- Encoding of queries and documents [Jin et al., 2023, Ueda et al., 2021]
- (Multiple) re-ranking phases [Pradeep et al., 2022, Biester et al., , Tahami et al.,]
- Query/document classification [Wang et al., 2022]
- Data augmentation/generation [Pradeep et al., 2022, Karimi, , Bondarenko et al., 2022]
- Combinations of all the above

Off-the-shelf medical models:

- Trained on text: ClinicalBERT, BioBERT, SciBERT, PubMedBERT,
- Including external knowledge: UMLSBERT, Clinical Kb-BERT
- In other languages!
- Check this amazing list: `https://mr-nlp.github.io/posts/2021/05/transformer-based-biomedical-pretrained-language-models-list/`
- (note that it is very complete but not up-to-date)

Representation Learning for Medical Search

Transformer-based models: Clinical Trials Matching [Pradeep et al., 2022]

- **Goal:** Given a patient and the patients electronic health record (EHR) as the query and a collection of actively recruiting clinical trials, return those that the patient is eligible for. (=*Clinical Matching*)
- **Context:** TREC2021 Clinical Trials Track

Their approach is two-fold:

1. Neural query synthesis (NQS): zero-shot document expansion model to generate sentence-long queries from patient descriptions:
   - doc2queryT5 model trained on MS MARCO V2 passage ranking
   - generated queries issued to BM25 + RM3 = first list of candidates
   - **+33% compared to using the EHR as the query**

Patient Description - #23: A 39-year-old man came to the clinic with cough and shortness of breath that was not relieved by his inhaler. · · ·

Query 1: causes for wheezing and shortness of breath
Query 2: what could be wrong when a chef has a cough and is short of breath all of a sudden
Query 3: how often should fluticasone be used for asthma
Query 4: what causes shortness of breath even with inhaler
⋮

Table 2: Examples of synthetic queries for the patient description in Table 1.

2. two-stage neural reranking pipeline trained on clinical trial matching data
   - neural sequence-to-sequence ranking model Med-Mono-T5
   - fine-tuned on CT dataset [Koopman and Zuccon, 2016]
   - model trained and used to find the best segments in each trial from the candidate list
   - final ranking based on the combination of segment's scores
   - **best automatic system at the TREC 2021 Clinical Trials Track**

Representation Learning for Medical Search

Transformer-based models: niomedical literature search [Ueda et al., 2021]

- **Goal:** Leverage biomedical abstract's structure to improve biomedical literature search
- Structure of an abstract: background, methods, results, conclusions
- standard cascading architecture for fine-tuning contextual language models:
  1. for each query q, retrieve initial k abstracts $a \in A$ to be re-ranked: $\mathcal{L} = log(1 + e^{-h_A(q,a^+,a^-)})$
  2. fine-tuning multiple pretrained SciBERT models concurrently: $a = (a|s \in \{B, M, R, C\})$, learn one model h_{A_s} per section
- Comparison of models on TREC PM and TREC COVID: locally fine-tuned features give consistent and significant improvements for the sum aggregator

Table 2: Retrieval effectiveness of different rankings leveraging globally (h_A) and locally ($h_{A_s} \mid s \in S = \{B, M, R, C\}$) fine-tuned SciBERT models as features. The symbols † and ‡ denote significant increases over the global model in the first row for $p < 0.05$ and $p < 0.01$, respectively.

	#	Features	Agg	NDCG	MAP	P@10
PM19	1	$\{h_A(q,a)\}$	–	0.530	0.206	0.456
	2	$\{h_A(q,a)\} \cup \{h_A(q,a_s) \mid s \in S\}$	sum	0.526	0.209	0.462
	3	$\{h_A(q,a)\} \cup \{h_{A_s}(q,a_s) \mid s \in S\}$	sum	**0.569‡**	**0.262‡**	**0.562‡**
	4	$\{h_A(q,a)\} \cup \{h_A(q,a_s) \mid s \in S\}$	max	0.517	0.198	0.428
	5	$\{h_A(q,a)\} \cup \{h_{A_s}(q,a_s) \mid s \in S\}$	max	0.533	0.217	0.465
COVID	1	$\{h_A(q,a)\}$	–	0.467	0.160	0.402
	2	$\{h_A(q,a)\} \cup \{h_A(q,a_s) \mid s \in S\}$	sum	0.486†	0.180‡	0.484‡
	3	$\{h_A(q,a)\} \cup \{h_{A_s}(q,a_s) \mid s \in S\}$	sum	**0.492‡**	**0.192‡**	**0.516‡**
	4	$\{h_A(q,a)\} \cup \{h_A(q,a_s) \mid s \in S\}$	max	0.475	0.169	0.454
	5	$\{h_A(q,a)\} \cup \{h_{A_s}(q,a_s) \mid s \in S\}$	max	0.482†	0.175‡	0.438

Table 3: Impact in retrieval performance on PM19 when removing individual features from our summed feature aggregation. Symbols † and ‡ denote significant decreases from the baseline (first row) for $p < 0.05$ and $p < 0.01$, respectively.

Set of features	NDCG	MAP	P@10
Using all sections	**0.569**	**0.262**	**0.562**
(-) Original Abstract	0.561 (-0.008)‡	0.249 (-0.013)‡	0.532 (-0.030)‡
(-) Background	0.565 (-0.004)	0.254 (-0.008)†	0.547 (-0.015)
(-) Methods	0.564 (-0.005)†	0.252 (-0.010)‡	0.557 (-0.005)
(-) Results	0.563 (-0.006)‡	0.253 (-0.009)‡	0.551 (-0.011)
(-) Conclusions	0.564 (-0.005)‡	0.251 (-0.011)‡	0.540 (-0.022)†

Representation learning for medical search

Let us discuss

- In summary
  - Big trend towards the use of neural models in medical search
  - Learned representations reusable in a wide range of search tasks and prediction tasks
  - Background knowledge (eg., Knowledge-base, expert's assessments) driven learning increases the readability of the representations and the explicability of the learning outcomes

- Pending issues
  - What are the impacting factors? What works vs. fails in the black box?
  - Lack of sufficient amount of labeled data to learn accurate representations (eg., patient similarity, IR tasks)
  - Performance sensitivity to a large size of network parameters, hyper-parameters and models parameters
  - Transfert learning, data augmentation/generation
  - Performance variability across medical tasks

OUTLINE

Outline

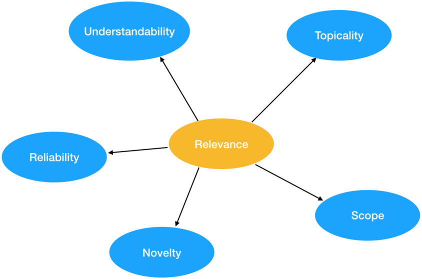
Challenges in Evaluating Medical Information Retrieval

Evaluation at the document level

In classical IR

- From the user point-of-view: a relevant document answer the initial information need
- From the system point-of-view: A relevant document covers the same topic as the query, i.e. contains the query's terms

Relevance has many other dimensions [Zhang et al., 2014]



In the medical domain:

- For patients:
  - Documents must be readable and understandable for a given user
  - The information contained in the documents should be trustworthy
- For medical professionals:
  - Documents must contain up-to-date information
  - Documents must properly cover the topic searched

Challenges in Evaluating Medical Information Retrieval

Each search task has its proper objectives:

- How should the retrieval and the ranking be implemented?
- How should the system be evaluated?

Examples:

- Physician adhoc search: priority given to the rank, P@10, the topicality, scope...
- Patient adhoc search: priority given to the rank, P@10, the topicality, understandability, readability...
- Clinical trials: priority given to the rank, the topicality, the scope, the novelty...
- Systematic reviews: priority given to the recall, the topicality, the scope...

Outline

Summary of the Benchmarking Activities/datasets

Venue	Task	Dataset	Activity
TREC	Genomics adhoc retrieval	Clinical information need Biomedical articles	Terminated
	Genomics passage retrieval	Clinical information need Biomedical articles	Terminated
	Medical records	Patient cohort search	Terminated
	Clinical decision support / Precision medicine	Case reports Biomedical articles	Ongoing
	Health Misinformation	Yes-no questions Web pages	Ongoing
	TREC COVID-19	Round-based biomedical literature Search on COVID-19 topics	Terminated
CLEF	ImageCLEF medical retrieval	Image and medical reports Collection of medical images	Terminated
	CLEF eHealth consumer search	Health information need Large web crawl	Terminated
	CLEF eHealth technological assisted reviews	Boolean queries Biomedical articles	Terminated
*	BioASQ	Annotated biomedical abstracts and QA dataset	Ongoing

IR dataset containing PICO queries: `https://github.com/boudinfl/CLIREC`
TRIP Click: Log files of a biomedical search engine [Rekabsaz et al., 2021]
Clinical trial matching test collection [Koopman and Zuccon, 2016]
The majority of these datasets are still available and can be used for research!

OUTLINE

CONCLUSION

- *A large and growing body of work on Information Retrieval in the medical domain*
  - Focus on task, user profile, information need elicitation in context (time, task, user's expertise, etc.)
  - Model semantic w.r.t. polyrepresentation view: document collections, knowledge bases, users, etc.
  - Shift from lexical matching to semantic matching by considering domain-specific peculiarities
  - Understand relevance assessment facets according to task, user (laypeople vs.expert)
  - Increasing amount of tasks due to a higher collaboration with the medical community
- *Challenges ahead*
  - IR at the service of the medical community:
    - Domain-driven IR models for medical search: Are IR heuristics similar to medical search heuristics?
    - Complex tasks not solved yet
    - Effort in structuring knowledge: dynamic terminologies, languages other than English...
    - Conceptualization of medical search tasks: eg., model hypothetico-deductive approach of medical experts?
  - Neural networks for all
    - Need for big data
    - Black-box: towards explainability
  - Large-scale evaluation under privacy-constraints

Discussion

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