
Research and Applications

Evaluation of patient-level retrieval from electronic health record data for a cohort discovery task

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ABSTRACT

Objective: Growing numbers of academic medical centers offer patient cohort discovery tools to their researchers, yet the performance of systems for this use case is not well understood. The objective of this research was to assess patient-level information retrieval methods using electronic health records for different types of cohort definition retrieval.

Materials and Methods: We developed a test collection consisting of about 100 000 patient records and 56 test topics that characterized patient cohort requests for various clinical studies. Automated information retrieval tasks using word-based approaches were performed, varying 4 different parameters for a total of 48 permutations, with performance measured using B-Pref. We subsequently created structured Boolean queries for the 56 topics for performance comparisons. In addition, we performed a more detailed analysis of 10 topics.

Results: The best-performing word-based automated query parameter settings achieved a mean B-Pref of 0.167 across all 56 topics. The way a topic was structured (topic representation) had the largest impact on performance. Performance not only varied widely across topics, but there was also a large variance in sensitivity to parameter settings across the topics. Structured queries generally performed better than automated queries on measures of recall and precision but were still not able to recall all relevant patients found by the automated queries.

Conclusion: While word-based automated methods of cohort retrieval offer an attractive solution to the labor-intensive nature of this task currently used at many medical centers, we generally found suboptimal performance in those approaches, with better performance obtained from structured Boolean queries. Future work will focus on using the test collection to develop and evaluate new approaches to query structure, weighting algorithms, and application of semantic methods.

Key words: information retrieval, patient cohort discovery, electronic health record, structured queries

LAY SUMMARY

- Many academic medical centers use electronic health record data to recruit patients for research studies. This task involves searching the data for patients who meet certain criteria, which can sometimes be quite complex. This task is labor intensive and requires information specialists to design custom queries. The use of automated information retrieval (IR) tools could be an attractive solution to this challenge, but little is known about their effectiveness with patient-level medical data.
- We evaluated the effectiveness of automated IR methods for identification of patients matching 56 different sets of criteria (topics). These topics were derived from actual research requests seen at 2 major medical institutions and were applied to a test set of electronic health record data containing 100 000 patients. For the automated methods, we varied 4 parameters to create 48 different query permutations for each of the 56 topics. We also had an informatics specialist develop custom queries for 10 of the 56 topics for performance comparison to the IR methods.
- We found that the automated IR methods did not perform well at accurately identifying patients who met the 56 sets of criteria, and that the custom queries performed much better at this task. Future work is needed to develop and evaluate new approaches in this area.

INTRODUCTION

Many academic medical centers, including over 90% funded by the National Institutes of Health Clinical & Translational Science Award program, offer patient cohort discovery to their researchers to facilitate clinical research, usually including electronic health record (EHR) data.^{1,2} A number of systems are available to facilitate this task, such as i2b2^{3,4} and TriNetX.⁵ However, the performance of systems and algorithms for this EHR use case is not well studied. It has been shown that typical review of patients for study eligibility is a labor-intensive task, and that automated preprocessing of lists of patients may reduce human time and effort for selection of cohorts.^{6–8}

One challenge for evaluating this use case is the lack of test collections that include data, clinical study descriptions, and relevance judgments for retrieved patients, a problem that has hindered many types of research using EHR data, even in the modern era of ubiquitous EHR adoption.⁹ A major barrier has been the challenge of protecting privacy of the patients from whom the records are from and institutional hesitancy to making such data widely available for informatics research, even in deidentified form.¹⁰ This is especially so for use cases involving processing of textual data within records, including those used on the scale of information retrieval (IR) experiments where corpora of thousands to millions of patient records are typically desired.

There are 2 EHR record collections that have been publicly available, one from the University of Pittsburgh Medical Center (UPMC)¹¹ and the other the Medical Information Mart for Intensive Care-III from the Massachusetts Institute of Technology.¹² Among the uses of the UPMC corpus has been a cohort retrieval for clinical research studies task in a challenge evaluation as part of the annual Text REtrieval Conference (TREC). The TREC Medical Records Track ran in 2011 and 2012, attracting 29 and 24 academic and industry research groups, respectively.^{13,14} Using the University of Pittsburgh collection containing 17 264 encounters containing 93 551 documents (some of which included ICD-9 diagnosis codes, laboratory results, and other structured data), a total of 34 and 47 topics, respectively, by year were developed and relevance judgments performed based on pooled results from participating research groups using the “Cranfield paradigm” common to IR evaluation research.¹⁵ The judgments were performed by physicians enrolled in biomedical informatics educational programs.

A common baseline method for all types of IR experimentation is “word-based” searching, where queries are submitted to the system and output is ranked by a similarity function between query and documents. Word-based methods are in distinction to Boolean searching, which is sometimes called set-based searching, where sets of retrieved documents are combined using Boolean operators. In the TREC Medical Records Track, several domain-specific enhancements on top of word-based queries were found to lead to improved retrieval performance. These included vocabulary normalization specific to the clinical domain, synonym-based query expansion from medical controlled terminology systems such as the Unified Medical Language System Metathesaurus, and recognition of negation.¹⁶ Follow-on research with the test collection found continued improvement in performance from approaches such as query expansion for additional clinical and other corpora¹⁷ as well as use of learning-to-rank methods.¹⁸

One limitation of the TREC Medical Records Track was a limitation of the UPMC corpus, which was retrieval at the encounter (eg, hospital or emergency department visit) and not the patient level. This was due to the deidentification process that broke the links across encounters, a process that also obscured various protected health elements, such as dates, geographic locations, and provider identifiers. Encounter-level retrieval data sets prohibit applying expert judgment and therefore evaluation at the patient level, which is the goal of cohort retrieval. Nonetheless, the TREC Medical Records Track did provide a data set for IR and biomedical informatics researchers to compare different approaches to identifying patient cohorts for recruitment into clinical studies. Unfortunately, the UPMC corpus has been withdrawn from public use (Wendy Chapman, personal communication).

Outside the TREC Medical Records Track, few other evaluations of cohort retrieval have been carried out and published. Some are limited by being document or encounter based, or focus on broadly defined cohorts that may be too general for the clinical research recruitment use case. One analysis using the Medical Information Mart for Intensive Care-III corpus looked at 2 straightforward clinical situations and found accurate retrieval with both structured data extraction and the use of natural language processing (NLP).¹⁹ Another recent approach employed word embeddings and query expansion to define patient cohorts, although used only structured EHR data.²⁰ The 2018 National NLP Clinical

Challenges had a shared task devoted to cohort selection for clinical trials but focused on the complementary task of finding inclusion criteria of clinical trials as opposed to patient-level retrieval.²¹

Another thread of work has focused on making querying easier to carry out, typically through development of natural language or other structured interfaces to the patient data.^{22–25} Other approaches focus on normalizing semantic representation of patient data within the EHR itself²⁶ and applying deep learning to nontopical characteristics of studies and researchers.²⁷ A related area to cohort discovery is patient phenotyping, one of the goals of which is to identify patients for clinical studies.^{28–30} However, the cohort discovery use case has some differences, as some studies have criteria beyond phenotypic attributes, such as age, past treatments, diagnostic criteria, and temporal considerations.

In 2014, Oregon Health & Science University (OHSU) and Mayo Clinic launched a project to use raw (ie, not deidentified) EHR data to perform research in parallel (ie, able to share methods and systems but not data). The OHSU data set has been previously described,³¹ and this paper reports the first results using this data set along with evaluation at the patient level. The Mayo Clinic has reported some of its work, although its retrieval output and relevance judgments were at the encounter level and not the patient level.³²

MATERIALS AND METHODS

The initial overall goal of this work was to assess and compare different approaches to patient-level retrieval by developing a “gold standard” test collection consisting of the 3 usual components of a Cranfield-style IR collection¹⁵: records—in this case patient-level medical records, topics—representations of cohorts to be recruited for clinical studies, and relevance judgments—expert determination of which records were relevant to which topics. Our initial plan was to develop the test collection and apply the methods found to work effectively by research groups in the TREC Medical Records Track. However, upon finding the results for numerous topics applied to this data performed suboptimally, we also developed and evaluated structured Boolean queries, with additional relevance judgments on a subset of topics.

Record collection

As noted in our earlier paper, the patient records originated from OHSU’s Epic (Verona, WI) EHR and were transformed and loaded (without any modification of the underlying structured and textual data) to a research data warehouse.³¹ The study protocol to use the records was approved by the OHSU Institutional Review Board (IRB00011159). To be included in the corpus, patients had to have at least 3 primary care encounters between January 1, 2009 and December 31, 2013, inpatient or outpatient, with at least 5 text note entries. This was done to ensure that records would more likely be comprehensive of their care as opposed to a patient referred to the academic medical for a single consultation.

Both structured and unstructured data were included in the collection. Document types included demographics, vitals, medications (administered, current, and ordered), hospital and ambulatory encounters with associated attributes and diagnoses, clinical notes, problem lists, laboratory and microbiology results, surgery and procedure orders, and result comments. A unique medical record number was used to link the different document types, and each document type could contain multiple data fields. The collection

contained a total of 99 965 unique patients and 6 273 137 associated unique encounters. It originated in a relational database but was extracted into XML format for loading into the open-source IR platform ElasticSearch (v1.7.6) for our experiments.

Topics

The 56 topics used for this research were developed from 5 sources by OHSU and Mayo Clinic as described in our previous paper.³¹ From OHSU, 29 topics were selected from research study data requests submitted by clinical researchers to the Oregon Clinical and Translational Research Institute (OCTRI). From Mayo Clinic, topics were modeled after 2 patient cohorts found in the Mayo Research Data Warehouse, 5 patient cohorts in the Phenotype KnowledgeBase (PheKB), 9 patient cohorts in the Rochester Epidemiology Project (REP), and 12 patient cohorts based on presence of quality measures from the National Quality Forum (NQF).

Each topic was expressed at 3 levels of detail, with the complete list in Supplementary Appendix 1:

Summary statement: 1–3 sentences

Illustrative clinical case

Brief summary plus structured inclusion and exclusion criteria for demographics, diagnoses, medications, and other attributes

Initial runs

As is typically done in Cranfield-style IR experiments, we performed a number of different runs consisting of the text of the topic representation submitted to the ElasticSearch system, which generated ranked output that we limited to 1000 patients per topic. We varied different parameters for different runs by topic representation, text subset, aggregation method, and retrieval model. For the latter, we used a number of common ranking approaches implemented in ElasticSearch and known to be successful both in the TREC Medical Records Track and IR systems generally:

- BM25, also known as Okapi³³
- Divergence from randomness (DFR)³⁴
- Language modeling with Dirichlet smoothing (LMDir)³⁵
- Default Lucene scoring, based on the term frequency-inverse document frequency model³⁶

We performed 48 runs representing all permutations of the following query parameters as described in our previous paper. These representations formed the basis for all queries created for this paper, both manual and automated and include (further referencing in this paper by underlined text):

1. Topic representation: A (summary statement), B (clinical case), or C (detailed criteria, with section headings and numberings removed).
2. Text subset: only clinical notes or all document types (including structured data reporting as text).
3. Aggregation method: patient relevance score calculated by summation (sum) of all documents or by maximum (max) value.
4. Retrieval model: BM25, DFR, LMDir, or Lucene.

Relevance assessment

The relevance assessments were carried out based on the principles discussed in our previous paper.³¹ The initial pools for relevance judging were generated in a similar manner to TREC challenge evaluations, where results from different runs were pooled by selecting from all runs for a given topic the top 15 ranked patients and then

randomly selecting 25% of the next 85 (21 patients) and 1% of the next 900 (9 patients). The process of relevance judging used the locally developed Patient Relevance Assessment Interface.³¹ This system tracked the judgments in a PostgreSQL database and interfaced with the EHR data that were loaded into Elasticsearch. Patient pools for topics were selected for judging and loaded into Patient Relevance Assessment Interface, where all document types could be searched by medical experts to determine patient-level relevance for the topic. Document-level subrelevance could also be assigned in the system. Patients could be assigned one of 3 levels of relevance: definitely relevant, possibly relevant, or not relevant. For retrieval performance metrics, both definitely and possibly relevant patients were considered relevant, since the use case motivating aimed to identify patients who were likely to be candidates for inclusion in clinical studies, and the number of definite plus possibly relevant patients was typically not vastly larger than would be desired for a clinical trial.

Assessment of initial retrieval results

We used the standard `trec_eval` program³⁷ to generate retrieval results for the 48 runs. Because our queries did not exhaustively assess all possible approaches to retrieval, we opted to use the B-Pref measure for results, based on its common usage for IR evaluation when relevance judging is considered to be incomplete.³⁸ B-Pref is a measure that aggregates recall and precision and is calculated based on how many relevant patients are retrieved, in the ranked lists, ahead of the nonrelevant patients in the interval [0,1]. The distribution of B-Pref was evaluated across all 56 topics for each of the 48 runs separately. The intent of this analysis was to assess differences in performance between run parameter settings, and variance within each setting across the 56 topics. We also evaluated the distribution across all of the 48 runs for each of the 56 topics separately to assess differences across topics and variance within each topic across the 48 runs.

As noted in “Results”, the results from these runs were substantially lower than comparable methods applied in the TREC Medical Records Track. This led us to perform additional methods described in the rest of this section that included use of structured Boolean queries on a subset of topics.

Structured queries

Because the word-based query methods that had worked well for the TREC Medical Records Track performed less successfully with this data, we constructed structured Boolean queries for the 56 topics in an iterative manner by one of the authors with clinical experience (SRC). These queries were based on topic representation C, which contained structured data and some free text. These queries were allowed to search all document types that we loaded into Elasticsearch. Since these were structured queries, we did not rank patients returned, and all patients returned were kept in the final query results. Patients retrieved could have been part of the word-based retrieval pools and thus known to be relevant or not, or have not been judged. In addition, some patients not retrieved by the structured queries could have been relevant from retrieval and judgments in the word-based pools. Standard to their definitions, recall for each query was measured as patients retrieved and relevant/patients known to be relevant and precision was measured as patients retrieved and relevant/patients retrieved. We also measured patients retrieved who had not been judged for relevance in the initial pool.

Additional relevance assessment for 10 selected topics

As we discovered that a number of patients retrieved by the structured queries had not been retrieved by the word-based queries and therefore not judged, we selected 10 topics for additional relevance judging of patients returned by the structured queries. These included topics 2, 7, 9, 17, 32, 33, 42, 44, 48, and 52. To build on previous work done in our group, we used 5 topics that had been selected randomly for this previous research,³⁹ while the second 5 topics were selected for diversity in all 5 of our sources for topic definitions (OHSU, Mayo, PheKB, REP, and NQF). The second 5 were also selected based on a higher likelihood to be seen in clinical practice (based on clinician judgment), as compared to other topics in the list of 56.

For these 10 topics our intent was to judge the entire list of patients retrieved by the structured queries. To compare the structured queries to the word-based queries, we used simple precision and recall. B-Pref was not an appropriate measure since the judged structured query patient pools were not ranked. For recall, we combined the relevant patients found in both the structured judged pools and the word-based judged pools. We counted patients judged as definitely or possibly relevant as relevant for all analyses. We also measured relevant patients retrieved in the word-based queries but not in the structured queries.

RESULTS

Word-based query results

Per the Cranfield approach, we performed standard batch runs for the 48 permutations of topic representation, subset, aggregation method, and retrieval model. For relevance judging, the results were pooled by topic. Relevance assessing of patients was done by a physician who took around 30–40 h per topic. Table 1 shows the number, source, summary, and distribution of relevance judgments for a sampling of 10 topics, with the full table of all 56 topics in Supplementary Appendix 2. One topic had no definite or possibly relevant patients and was excluded from further analysis (25). We used the `trec_eval` program to include each topic for each run, along with the relevance judgments, to generate retrieval results for each run.

The highest overall performing run was `b.notes.max.LMDir`, with a mean B-Pref of 0.167. Very close to this run were 2 variations of the Retrieval Model: `b.notes.max.DFR`, and `b.notes.max.Lucene`, although `b.notes.max.BM25` scored lower. At the other end of performance, the `a.notes.sum.BM25` run had a mean B-Pref of 0.106. Figure 1 depicts the median and distribution of B-Pref for all 48 runs across all 56 topics (Figure 1).

There were several performance grouping patterns seen within the 4 parameters. Overall, topic representation B performed better than the other 2 representations. This representation was only composed of text but included a detailed individual case description, along with summary description. There was a tendency for the Retrieval Model BM25 to perform poorer than the other models, primarily with the text subset notes, which was composed of a more limited use of available document types. There was a trend for the aggregation method sum to have lower performance than the method max.

As is commonly seen in IR experiments, the distribution of topics was spread widely (Figure 2). The highest mean B-Pref was for topic 50 (0.895), while 11 topics had essentially a mean B-Pref of 0.0 (ie, most runs retrieved no relevant patients). Two topics consistently had the top 2 highest values for B-Pref for all parameter combina-

Table 1. A sample of the 56 topics with number, source, summary, and pool size, as described in the text

Num	Source	Summary	Pool	Def rel	%	Poss rel	%	Not rel	%
2	OHSU	Adults with IBD who have not had GI surgery	684	63	9.2	4	0.6	617	90.2
7	OHSU	Hereditary hemorrhagic telangiectasia	695	15	2.2	0	0.0	680	97.8
9	OHSU	Children with focal epilepsy with partial seizures	687	31	4.5	13	1.9	643	93.6
17	OHSU	RA on MTX w/o biologic DMARD	704	20	2.8	0	0.0	684	97.2
32	PheKB	ACE inhibitor-induced cough	700	40	5.7	0	0.0	660	94.3
33	PheKB	Children with ADHD on CNS stimulant	732	112	15.3	0	0.0	620	84.7
42	NQF	Elderly patients with dementia on antipsychotic medication	731	24	3.3	0	0.0	707	96.7
44	NQF	COPD with potentially avoidable complication	680	38	5.6	0	0.0	642	94.4
48	REP	Stroke after first MI	698	5	0.7	0	0.0	693	99.3
52	REP	Cataract surgery and prior SSRI use	737	23	3.1	13	1.8	701	95.1

Also shown are number and percentage for definitely relevant, possibly relevant, and not relevant from the initial relevance assessment process. ACE, angiotensin covering enzyme; ADHD, attention deficit hyperactivity disorder; CNS, central nervous system; COPD, chronic obstructive pulmonary disease; DMARD, disease-modifying anti-rheumatic drugs; GI, gastrointestinal; IBD, inflammatory bowel disease; MI, myocardium infarction; MTX, methotrexate; NQF, National Quality Forum; OHSU, Oregon Health & Science University; PheKB, Phenotype KnowledgeBase; RA, rheumatoid arthritis; REP, Rochester Epidemiology Project; SSRI, selective serotonin reuptake inhibitor.

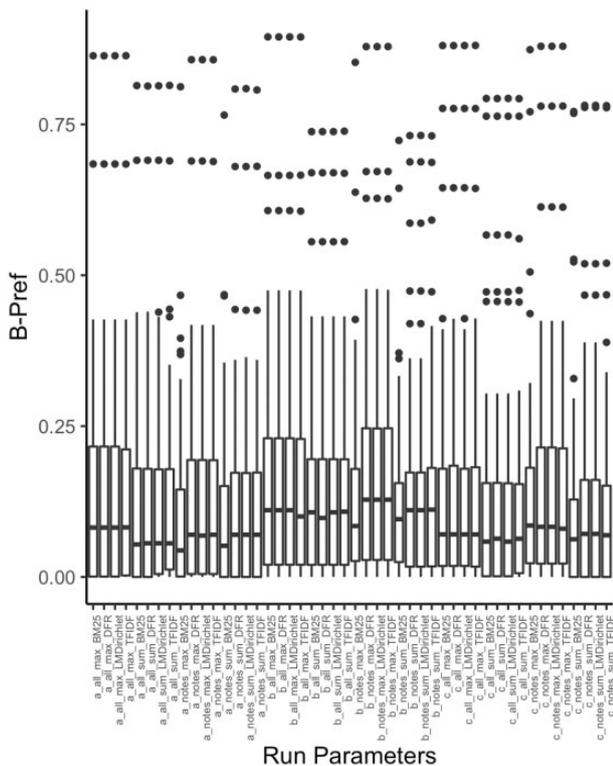


Figure 1. B-Pref distributions for topics within each run. Box ends represent the upper and lower quartile values and whiskers extend 1.5 times the interquartile range. Data points beyond the end of the whiskers are values for individual topics outside the whiskers. The parameter settings are ordered hierarchically first by topic representation (A–C), then text subset (all, notes), then aggregation method (max, sum) and finally the Retrieval Model (BM25, divergence from randomness [DFR], LMDirichlet, and term frequency-inverse document frequency [TFIDF]).

tions within topic representations A and C, topics 50 and 28. For topic representation B, topic 50 was also consistently in the top 2 extreme B-Pref values along with topic 47. These topics did not have complex temporal conditions, medication requirements, or surgery inclusions or exclusions and only required relatively straightforward inclusion/exclusion lists of medical conditions, lab and radiology tests, and demographics.

B-Pref distributions of the 48 parameter combinations (runs) within each topic varied widely in range and shape. Topics 31 and

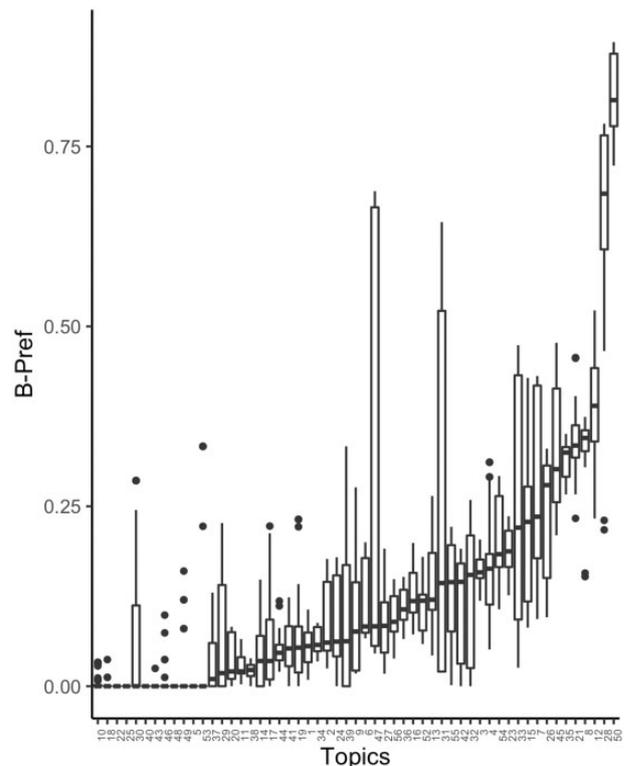


Figure 2. B-Pref distributions for parameter combinations within each topic. Box ends represent the upper and lower quartile values and whiskers extend 1.5 times the interquartile range. Data points beyond the end of the whiskers are values for parameter combinations outside the whiskers. Boxplots are ordered by median B-Pref values.

47 were distinctive, showing much greater variation in performance across parameter settings than the other topics. This variation was entirely due to large differences between topic representations. There was very little performance variation for these 2 topics across the other parameter combinations within each representation.

Structured queries

For each topic, we calculated simple recall and precision on the output of each structured Boolean query (Boolean queries are typically not ranked) using the relevance judgments for the word-based query

pools. As with the word-based queries, a patient was considered relevant if rated definitely or possibly relevant. Table 2 shows an example structured Boolean query for topic 7. Recall for the structured queries varied widely across topics (Figure 3). There was 100% recall of word-based query relevant patients on 8 of the 56

Table 2. Structured Boolean query for topic 7: adults 18–100 years old who have a diagnosis of hereditary hemorrhagic telangiectasia, which is also called Osler-Weber-Rendu syndrome

```
(demographics.BIRTH_DATE: Range[1913-01-01, 1995-12-31])
AND
(
  encounter_diagnoses.DX_ICD = 448.0
  OR
  hospital_encounters.ADMITTING_DX_ICD_CODE = 448.0
  OR
  hospital_encounters.BILL_DISCHARGE_DX_ICD_CODE = 448.0
  OR
  hospital_
    encounters.hospital_encounters.BILL_DX2_ICD_CODE = 448.0
  OR
  hospital_encounters.BILL_DX3_ICD_CODE = 448.0
  OR
  hospital_encounters.BILL_DX4_ICD_CODE = 448.0
  OR
  hospital_encounters.ENCOUNTER_DIAGNOSIS = 448.0
  OR
  problem_list.DX_ICD = 448.0
  OR
  notes.NOTE_TEXT contains "Hereditary hemorrhagic telangiectasia"
  OR
  notes.NOTE_TEXT contains "Osler-Weber-Rendu"
)
```

topics, >50% recall on 35 of the 56 topics, <50% recall on 13 of the 56, one topic (48) with no recall of relevant patients, and 2 topics with no retrieval at all (22, 25).

Precision likewise varied widely across topics for the structured queries (Figure 4). The structured queries outperformed the word-based queries in precision for all topics except 48. Again, topics 22 and 25 did not return any relevant patients. Three topics had 100% precision (29, 34, 46).

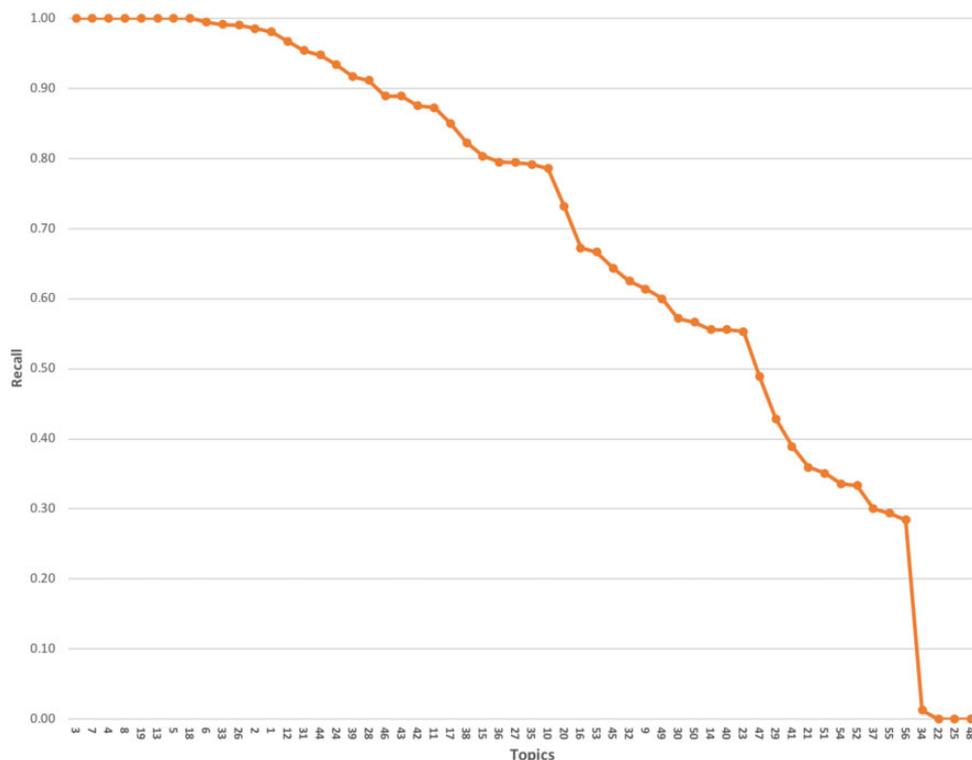
Topics with expanded relevance judgments for the structured queries

Because the structured queries retrieved patients who had not been retrieved by the word-based queries, we did additional relevance judging for 10 selected topics. Due to the large number of patients returned for topic 2 by the structured query (2,578), only a random sample of 750 patients was judged.

Although the structured queries had higher recall than the word-based queries for all 10 topics, these queries did not achieve complete recall of all of the relevant patients for 9 of the 10 topics (Figure 5). The numbers of relevant patients found only in word-based queries was relatively low, compared to the total number of relevant patients (Table 3). This explains the larger number of missed relevant patients for this topic. The structured queries had higher precision for all 10 topics (Figure 6). For topic 52, all patients retrieved by the structured query were judged relevant.

DISCUSSION

We set out to begin this work using word-based query methods that performed well for the TREC Medical Records Track. Our results did not achieve the performance we expected (Figure 1). Overall,



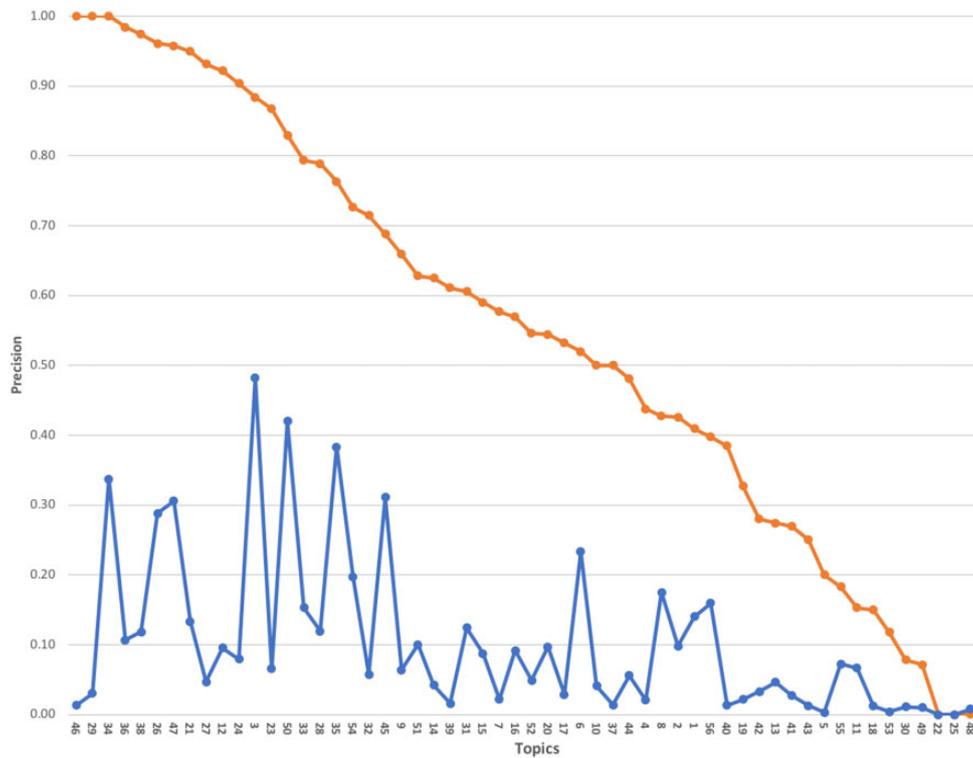


Figure 4. Precision for structured queries (red line) and word-based judged pools (blue line), ordered by structured query precision for each topic.

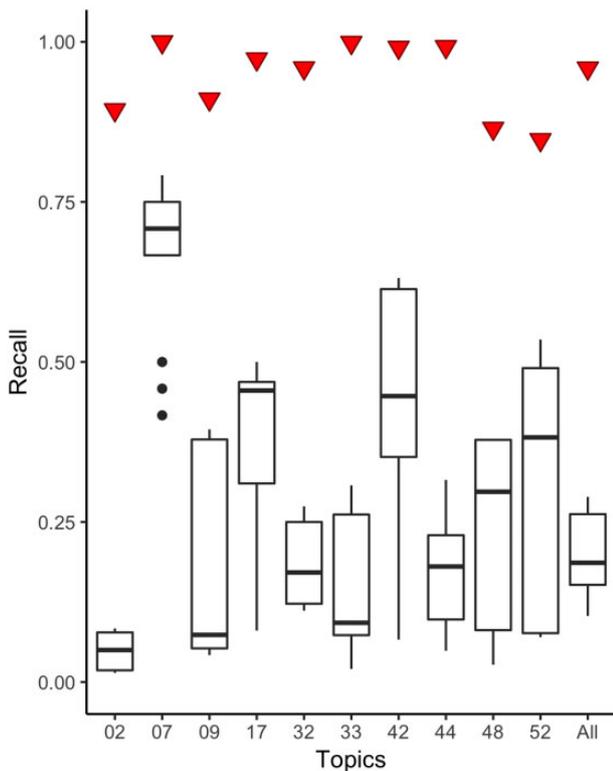


Figure 5. Recall distributions for 10 selected topics based on combined full structured-query relevance judged pools. Red triangles are the values for the structured queries while the box and whisker plots contain the distributions for word-based queries with the original 48 different parameters.

the best results were achieved with the topic representation of the illustrative clinical case formulation (B), with small further improvements for using text subset all and aggregation method max. Within our results, we observed variation common to IR challenge evaluations. Although the overall differences were modest, there was consistently higher values for topic representation B. Likewise, there was small benefit for aggregation method sum versus max. For combinations of parameters, the Retrieval Model BM25 performed worse than the other three. To the extent that these results are generalizable, clinical case formulations are the best query type among word-based methods for the patient cohort discovery task.

Also common to IR challenge evaluation results, reflecting the adage that means and medians can obscure variations, there was a large difference in retrieval performance by topic. As seen in Figure 2, about 10 had very poor performance while 2 had very high performance across all retrieval methods. There was also a substantial range of performance within a number of individual topics.

In the effort to improve our results, we reformulated our queries using structured Boolean approaches developed iteratively. Because pure Boolean queries do not rank their output, we could not directly compare our results with the word-based queries. Instead, we measured standard recall and precision based on the relevance judgments made for patients retrieved by the word-based methods. The results for the structured queries were much better, with a median recall of 0.86 and 8 topics having recall of 1.0 (Figure 3). There were likewise 13 topics with recall under 0.4 and a couple near 0. Precision was not associated with recall for the topics but did vary almost linearly from 1.0 to 0.0 across the topics (Figure 4).

One concern for the structured queries was the use of the relevance judgments only from the word-based query results. As such, we performed additional relevance judgments based on the struc-

Table 3. Ten topics with additional relevance judgments for results from structured Boolean queries

Topic	Structured query patients retrieved	Word-based query relevant	Structured query added relevant	Structured query relevant and retrieved	Recall for structured query	Precision for structured query	Structured query relevant missed
2	750	67	490	438	0.89	0.58	52
7	50	15	24	24	1.00	0.48	0
9	357	44	190	173	0.91	0.48	17
17	110	20	112	109	0.97	0.99	3
32	390	40	368	353	0.96	0.91	15
33	1092	112	983	982	1.00	0.90	1
42	347	24	347	344	0.99	0.99	3
44	378	38	266	264	0.99	0.70	2
48	68	5	37	32	0.86	0.47	5
52	133	36	157	133	0.85	1.00	12

The structured queries retrieved additional patients who were judged for relevance, allowing calculation of recall and precision for these queries as well as determination of numbers found by the word-based queries but missed by the structured queries.

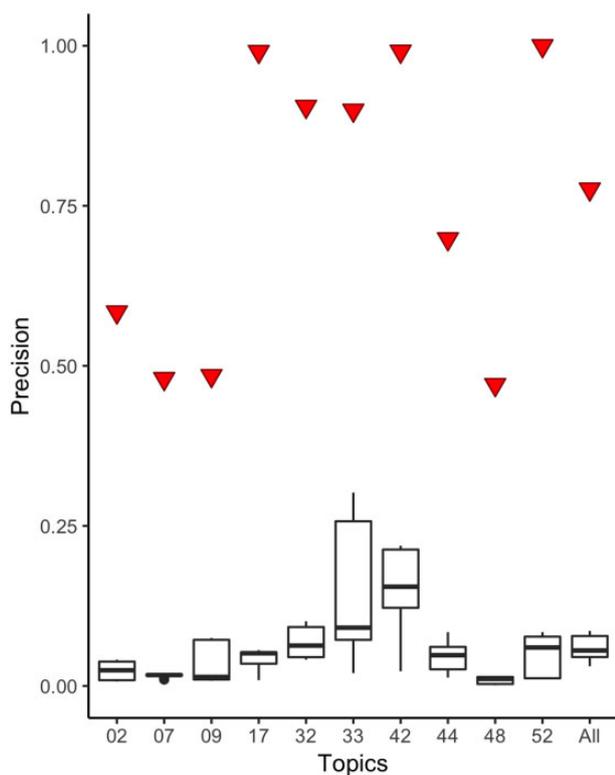


Figure 6. Precision distributions for 10 selected topics based on combined full structured-query relevance judged pools. Red triangles are the values for the structured queries while the box and whisker plots contain the distributions for word-based queries with the original 48 different parameters.

tured query retrieval for 10 topics. This not only would give us a more realistic picture of the performance of these topics but also identify additional patients for relevance judgment for the word-based queries. After the additional judgments, we found that the structured queries had much higher recall than the word-based queries (Figure 5) as well as much higher precision (Figure 6), which was also been found in comparable experiments from Mayo Clinic.³⁹ As precision is sometimes conceptualized as “number needed to read,”⁴⁰ the higher precision for the structured queries means fewer patients would need to be assessed to identify candidates for clinical studies.

There were a number of limitations to this work. Our records were limited to a single academic medical center. There are many additional retrieval methods we could have assessed, but we would not have the resources to carry out the additional relevance judgments required as those additional methods would add new patients to be judged. Finally, there is a global limitation to work with EHR data for these sorts of use cases in that raw, identifiable patient data are not easily sharable such that other researchers could compare their systems and algorithms with ours using our data, although they could apply our methods to their own data.

CONCLUSIONS

Although many medical centers, especially those funded by the Clinical & Translational Science Award program, offer patient cohort discovery tools, this function has not been well studied. This research evaluates patient-level cohort retrieval over a large extract of complete EHR data for an academic medical center, along with 56 diverse information needs. Our results found that structured Boolean queries, searching over unstructured and structured data, outperformed word-based automated methods over the same data. Substantial work remains for defining the best methods for cohort discovery from EHR data, especially in the development of methods that allow automated techniques that do not require users to construct Boolean queries themselves.

Future work will explore additional methods against the baselines established in this paper. One area for possible improvement will be to leverage the semantic approaches using synonym expansion and detection of negation in topics that were found to provide value in the TREC Medical Records Track. Additional opportunities for further work include translating the relatively more-structured topic expression C automatically to Boolean queries. Challenges to developing and evaluating IR methods for this use case include the resources required to perform relevance judgments and the nature of such highly private data that makes their comparison across different research groups difficult. Our future work will continue to develop methods that show promise and evaluate them with real-world topics and relevance judgments. We also plan to classify topic characteristics and assess their role in retrieval performance.

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AUTHOR CONTRIBUTIONS

WRH and SRC conceived of the presented idea. SRC and WRH developed the framework and conducted the analyses. SRC, SDB, AMC, YW, AW, SL, HL, and WRH contributed to the study design, development of the analysis plan, as well as the refinement of the manuscript.

SUPPLEMENTARY MATERIAL

Supplementary material is available at *Journal of the American Medical Informatics Association* online.

CONFLICT OF INTEREST STATEMENT

SRC, AMC, and WRH have research funding from Alnylam Pharmaceuticals that is unrelated to the work described in this paper.

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