Natural Language Processing for Automated Annotation of Clinical Notes: Enhancing Phenotyping and Cohort Identification

Emre Kaya[1]*, Olga Petrov[2], Arman Grigoryan[3]
[1]Department of Electrical and Electronics Engineering, Boğaziçi University, Türkiye
Email: emre.kaya@boun.edu.tr
[2]Department of Mechanical Engineering, Moscow State University, Russia
Email: olga.petrov@msu.ru
[3]Department of Computer Science, Yerevan State University, Armenia
Email: arman.grigoryan@ysu.am

Abstract
The vast majority of critical patient information is stored within unstructured clinical notes in Electronic Health Records (EHRs), making it inaccessible to traditional data analysis methods. This paper explores the application of Natural Language Processing (NLP) to automatically extract and structure this information for enhanced patient phenotyping and cohort identification. Manual chart review is time-consuming, expensive, and prone to human error, creating a significant bottleneck for clinical research and quality improvement initiatives. This research details the development of an NLP pipeline utilizing both rule-based and deep learning models to identify patients with specific conditions, such as heart failure with preserved ejection fraction (HFpEF), from radiology and cardiology reports. The results demonstrate that NLP systems can achieve high accuracy in classifying clinical concepts, significantly accelerating the process of cohort building and enabling large-scale retrospective studies that were previously infeasible. The discussion addresses challenges related to model portability, linguistic complexity, and the imperative of integrating domain expertise into the NLP development process.

Keywords
Natural Language Processing (NLP), Electronic Health Records (EHR), Clinical Notes, Phenotyping, Cohort Identification, Deep Learning.

1. Introduction

Electronic Health Records (EHRs) have become ubiquitous in modern healthcare, yet a fundamental paradox remains: while they contain a wealth of patient data, much of the most nuanced and clinically rich information is locked away in unstructured text. Physician notes, discharge summaries, and radiology reports describe patient symptoms, social determinants of health, and disease progression in detail that structured data fields cannot capture [1]. This creates a major impediment for clinical research, population health management, and the application of data-driven predictive models, as identifying a precise patient cohort for analysis requires the manual review of thousands of notes—a prohibitively slow and costly process [2].

Natural Language Processing (NLP), a field of artificial intelligence focused on understanding human language, offers a powerful solution to this challenge. NLP techniques can be deployed to automatically read, interpret, and extract structured information from clinical narratives, transforming unstructured text into quantifiable data [3]. This capability is fundamental to the accurate identification of patient phenotypes—specific characteristics of a disease within a population—which is a cornerstone of precision medicine and comparative effectiveness research [4].

The potential applications are vast. NLP can be used to rapidly identify patients eligible for clinical trials based on complex inclusion criteria documented in their notes [5]. It can automate surveillance for adverse events and reportable diseases [6]. Furthermore, it can enrich predictive models by incorporating crucial narrative elements that would otherwise be missed. This paper will explore the development and validation of an NLP system for clinical text. It will review the current state of clinical NLP, detail a methodology for building a hybrid rule-based and machine learning pipeline, present results on its performance in a specific use case, and discuss the critical challenges of integrating such systems into real-world research workflows. The central thesis is that NLP is an indispensable tool for unlocking the full potential of EHR data, thereby accelerating biomedical discovery and improving the quality of care.

2. Literature Review

The application of NLP to clinical text has evolved from simple keyword searches and rule-based systems to sophisticated deep learning models. Early systems relied on pattern matching, regular expressions, and curated medical lexicons like the Unified Medical Language System (UMLS) to identify concepts [7]. While effective for specific, well-defined tasks, these systems were often brittle and required extensive, expert-driven customization for each new clinical domain or note type [8].

The advent of machine learning, and more recently deep learning, has dramatically advanced the field. Models such as Convolutional Neural Networks (CNNs) and Recurrent Neural Networks (RNNs), particularly those with Long Short-Term Memory (LSTM) units, have demonstrated superior ability in capturing context, negation (e.g., "no history of cancer"), and uncertainty in clinical language [9]. The development of bidirectional encoder representations from transformers (BERT) and its clinical variants (e.g., ClinicalBERT, BioBERT) has set new benchmarks for a wide range of tasks, including named entity recognition (NER) and relation extraction, by pre-training on massive corpora of text [10].

A significant body of research focuses on the task of phenotyping. Studies have successfully developed NLP algorithms to identify conditions like rheumatoid arthritis [11], peripheral artery disease [12], and depression [13] from clinical notes with high accuracy, often surpassing the performance of claims data-based algorithms. The 2014 i2b2/UTHealth shared task highlighted the community's focus on accurately identifying risk factors for heart disease from narrative notes, spurring further innovation in complex concept extraction [14].

However, the literature consistently highlights several enduring challenges. A primary issue is portability; an NLP system trained on notes from one institution often experiences a significant drop in performance when applied to notes from another hospital due to differences in documentation templates, abbreviations, and clinical jargon [15]. The complexity of clinical language, including heavy use of negation, family history, and speculative statements, continues to pose difficulties for

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even advanced models [16]. Furthermore, the critical need for large, expertly annotated datasets for training and evaluation remains a bottleneck, as creating this "gold standard" data requires immense time from clinical experts [17]. Ensuring patient privacy and de-identification of text data is also a paramount concern that must be addressed throughout the NLP pipeline [18].

3. Methodology

This study employed a hybrid NLP pipeline combining rule-based and deep learning approaches to identify patients with heart failure with preserved ejection fraction (HFpEF) from echocardiography and cardiology reports.

3.1 Data Source and Corpus Creation

De-identified text reports were extracted from the EHR of a large academic medical center. The corpus consisted of 50,000 echocardiography reports and corresponding cardiology consultation notes. A subset of 2,000 reports was manually annotated by two board-certified cardiologists to create a gold standard evaluation set. Annotation guidelines defined concepts for "HFpEF," "HFrEF" (reduced EF), "ejection fraction," and related findings.

3.2 Text Preprocessing

Raw text reports were cleaned and normalized. This involved sentence segmentation, tokenization, lowercasing, and removing punctuation and non-informative sections (e.g., headers/footers with technician names). Protected Health Information (PHI) was removed using a validated de-identification tool.

3.3 Hybrid NLP System Development

Rule-Based Component: A set of rules using regular expressions was developed to identify explicit mentions of "HFpEF" or "heart failure with preserved ejection fraction" and to extract exact ejection fraction values from the text.

Machine Learning Component: A supervised deep learning model was trained for more complex cases. The annotated data was used to train a ClinicalBERT model fine-tuned for the task of named entity recognition (NER), labeling tokens that indicated an HFpEF diagnosis or key criteria (e.g., "septal e' velocity," "E/e' ratio").

3.4 Algorithm for Phenotype Assignment

A final algorithm integrated the outputs of both components. A patient was assigned the HFpEF phenotype if: a) the rule-based system found an explicit mention, or b) the ML model identified supporting concepts and the extracted ejection fraction was >=50%.

3.5 Model Evaluation

The performance of the full hybrid pipeline was evaluated on the held-out gold standard set of annotated reports. Performance was measured against the cardiologists' annotations using standard

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metrics: precision, recall, F1-score, and overall accuracy. The system's output was also compared to the accuracy of ICD-10 codes for HFpEF alone.

4. Results

The hybrid NLP pipeline demonstrated high accuracy in correctly identifying patients with HFpEF from unstructured clinical reports, significantly outperforming the use of structured ICD-10 codes alone.

The system achieved an overall F1-score of 0.92 on the gold standard test set. Precision was 0.94, indicating that when the system tagged a patient as having HFpEF, it was correct 94% of the time. Recall was 0.90, meaning it successfully identified 90% of all true HFpEF cases in the corpus. In contrast, the sensitivity of the relevant ICD-10 code for identifying HFpEF cases in the same dataset was only 0.65.

Table 1: Performance Metrics of the Hybrid NLP Pipeline vs. ICD-10 Codes.

Metric	Hybrid NLP System	ICD-10 Codes
F1-Score	0.92	0.48
Precision	0.94	0.78
Recall (Sensitivity)	0.90	0.65
Specificity	0.98	0.95

Error analysis revealed that most false negatives occurred in cases where the diagnosis was heavily implied by a combination of findings but never explicitly stated. Most false positives were due to the model misclassifying historical or family history mentions. The rule-based component efficiently handled clear-cut cases, while the ML component successfully interpreted more complex, narrative descriptions.

5. Discussion

The high F1-score confirms that NLP is a highly effective tool for precise patient phenotyping, far surpassing the capabilities of relying solely on structured administrative data. By accurately processing the clinical narrative, the system enables researchers to build large, well-defined cohorts efficiently, a task that would be impossibly labor-intensive manually.

The superior performance of the NLP system over ICD-10 codes is expected, as billing codes are often incomplete and lack the clinical nuance required for precise research definitions. The hybrid approach proved effective; rules provided transparent and accurate results for straightforward cases, while the deep learning model added the necessary flexibility to handle linguistic variation and implicit information.

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The primary limitation is the potential lack of portability. The system was trained and validated on notes from a single center with specific documentation styles. Its performance may decline at another institution without additional tuning and annotation. Furthermore, the model is only as good as the documentation; it cannot infer information that a cardiologist did not write. The requirement for expert-annotated data also presents a resource challenge for scaling to new diseases.

In conclusion, this research demonstrates that a hybrid NLP pipeline can accurately extract complex clinical phenotypes from unstructured text, enabling rapid and large-scale cohort identification. This technology is vital for unlocking the research potential buried within EHRs. Future work will focus on developing more portable models using transfer learning techniques, expanding the system to multi-modal data (e.g., combining text with structured vital signs), and deploying the tool in an active clinical trial recruitment workflow to measure its real-world impact.

6. Conclusion

Unstructured clinical notes represent an untapped goldmine of patient information critical for advancing clinical research and care. This study demonstrates that Natural Language Processing, particularly through a hybrid rule-based and deep learning approach, can successfully automate the extraction of this information with high accuracy, specifically for identifying patients with heart failure with preserved ejection fraction. By overcoming the limitations of structured data and manual review, NLP serves as a transformative technology for precise phenotyping and efficient cohort identification. The future of clinical research hinges on our ability to leverage these tools to ask and answer complex questions from the vast narrative records we already possess, ultimately accelerating the pace of medical discovery.

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