Phenotyping of Clinical Notes with Improved Document Classification Models Using Contextualized Neural Language Models

Andriy Mulyar\textsuperscript{1} * Elliot Schumacher\textsuperscript{2} Masoud Rouhizadeh\textsuperscript{3} Mark Dredze\textsuperscript{2}  \\
\textsuperscript{1}Department of Computer Science, Virginia Commonwealth University  \\
\textsuperscript{2}Department of Computer Science, Johns Hopkins University  \\
\textsuperscript{3}Institute for Clinical and Translational Research, Johns Hopkins University

Abstract

Clinical notes contain an extensive record of a patient’s health status, such as smoking status or the presence of heart conditions. However, this detail is not replicated within the structured data of electronic health systems. Phenotyping, the extraction of patient conditions from free clinical text, is a critical task which supports a variety of downstream applications such as decision support and secondary use of medical records. Previous work has resulted in systems which are high performing but require hand engineering, often of rules\cite{Uzuner2008, Uzuner2009}. Recent work in pretrained contextualized language models\cite{Devlin2019} have enabled advances in representing text for a variety of tasks. We therefore explore several architectures for modeling phenotyping that rely solely on BERT representations of the clinical note, removing the need for manual engineering. We find these architectures are competitive with or outperform existing state of the art methods on two phenotyping tasks.

1 Introduction

Electronic Health Record (EHR) systems contain a wealth of health information about patients, including structured data (e.g. demographics, medical codes, lab results) and unstructured text in the form of clinical notes. While structured data includes information that characterizes medical conditions of the patient, it often does not include numerous characteristics of interest that are typically contained in the clinical notes. For example, while a clinician may not code the smoking status of a patient, it can appear in the notes if it was discussed during the visit. Since the mention of smoking status appears in unstructured text, the phrasing can vary: “The patient smokes regularly” or “He reports a history of smoking.” Including these characteristics as structured data can support both improved patient care and secondary use of medical records.

The task of automatic identification of specific phenomic traits within patients is called phenotyping. Phenotyping is critical to cohort selection, in which a study selects a population from an EHR system for further study, e.g. men over 50 who smoke. There have been multiple shared tasks, including identifying smoking status\cite{Uzuner2008} and obesity related co-morbidities\cite{Uzuner2009}, which have produced high-performing systems for obesity\cite{Ware2009, Solt2009} and smoking status\cite{Clark2008, Yao2018}. While these successes are promising, the uses of phenotyping are vast, for which an almost unlimited number of phenomic traits can be useful for patient care or cohort selection. Therefore, systems that require extensive preprocessing (e.g abbreviation and negation detection) or feature engineering (e.g. detection of temporal phrases) targeted at specific tasks may have limited utility in supporting a diverse range of phenomic traits.

*Work performed as a visiting student at Johns Hopkins University.
Figure 1: Our model architecture for a document classifier for phenomic traits based on repeated application of BERT to spans in a clinical note. Working bottom-up, a document is tokenized and chunked into spans according to the maximum BERT sequence length. An encoding function $f((\hat{h}_p))$ condenses the unrolled language model hidden state sequence $(\hat{h}_p)$. Not shown, this encoded representation is then fed into a layer of perceptrons with tanh activation for classification.

Recent work on contextualized neural language models [Devlin et al. [2019], Peters et al. [2018], Dai et al. [2019]] has led to systems which construct representations of text data that can support many tasks, with task specific training data only needed to train a final prediction layer. These models have been applied in the clinical space [Alsentzer et al. [2019]]. However, these models – which require building representations across the entire input – have only been used for sentences or short segments of text. Little work has applied these models to entire documents or entire clinical notes.

We develop a phenotyping system based on neural contextualized representations of language. We utilize a clinically fine-tuned [Alsentzer et al. [2019]] version of BERT (Devlin et al. [2019]). BERT can only be applied directly to relatively short spans of text. Since the phenomic trait can be contained anywhere in the clinical note, we explore ways of combining BERT representations from multiple segments into a single document-level representation. Recent BERT based document classification architectures [Adhikari et al. [2019]] are not suited for clinical notes as they consider only the first few sentences of the text thus cannot capture information relayed further into the document. We evaluate our approach on two domains of phenomic traits (obesity co-morbidities and smoking) and find that our best approach to document level modeling outperforms previous state of the art systems.

2 Phenotyping of Clinical Notes with BERT

We frame phenotyping as a classification problem, where for each phenomic trait for a clinical note our system produces a label, either binary or multi-class. Our classifier uses BERT to generate a sequence of representations of the text document. This representation sequence is condensed into a single document representation and then fed to our classifier. We first describe how BERT is applied to the document, and then describe several methods to construct a single document representation from the BERT output.

BERT considers a fixed length of text, e.g. $\ell = 512$ WordPieces/subwords, where tokens are subdivided using the WordPiece algorithm [Wu et al. [2016]]. BERT passes these subwords through its multiple transformer layers and produces both a per subword representation, as well as a representation meant to summarize the entire input: CLS. We divide the clinical document into chunks, where each chunk is of length $\ell$ (with the exception of possibly the last) and is passed into BERT. The result of this process is that we have a representation for each subword of input, as well as a CLS representation for each text chunk. This process is shown in Figure 1.

The next step requires the combination of the output from each BERT segment into a single document embedding. Previous approaches consider only the first two sentences as the basis for the representation [Wu and Dredze [2019]]. However, since the phenomic trait can be contained anywhere in the document, we need to combine all CLS embeddings into a single representation. Additionally, since clinical notes can be of arbitrary length, we need a general method to collapse this sequence of segment representations into a single representation. This combination function is represented in Figure 1 as $f((\hat{h}_p))$. 
We consider four options for \( f(\langle \vec{h}_p \rangle) \). The input to each function is a sequence of the CLS embeddings. Each element of the sequence itself summarizes the document segment that element encodes.

\[ f_{\text{max}} : \text{A dimension-wise max over all CLS embeddings.} \]
\[ f_{\text{id}} : \text{The identity function (a concatenation of all CLS embedding).} \]
\[ f_{\text{Transformer}} : \text{A dimension-wise max of the output sequence in the encoder layer of a Transformer [Vaswani et al. 2017].} \]
\[ f_{\text{LSTM}} : \text{The last hidden state of an LSTM run left-to-right over the CLS embeddings.} \]

Each of these four architectures are identical up to the choice of encoding function \( f \). In our proposed architectures (Figure 1), we use a frozen (no parameter fine-tuning) instance of ClinicalBERT [Alsentzer et al. 2019] - a BERT language model fine-tuned over biomedical and clinical domain text. We use BERT’s maximum word piece input length of 512, using 510 tokens with a padding on each side. The output of each CLS combination function \( f \) is linearly projected (via a layer of perceptrons) into the label space followed by a hyperbolic tangent \( (\tanh) \) activation. This entire architecture is trained on the available training set, which updates the parameters of the linear projection and if applicable \( f \), e.g. the encoder layer of the transformer or the LSTM model parameters. During training, we randomly dropout [Srivastava et al. 2014] weights with probability \( \frac{1}{10} \) in the projection and use binary cross entropy loss to estimate the target label distribution.

2.1 Implementation details

During training we utilize the BERT-base hidden size (768 dimensions) throughout all relevant internal hidden states in the \( f_{\text{LSTM}} \) and \( f_{\text{Transformer}} \) architectures and a dropout probability of .1 in the projection layer. During prediction we apply a given label if the hyperbolic tangent of the projections component corresponding to the label exceeds a threshold of 0; otherwise, the document does not receive the corresponding label. We trained all architectures on an NVIDIA Tesla M40 GPU. All architectures took approximately 24 hours of wall time (with negligible CPU computation) to converge to the performance reported in Table 4 on the N2C2 2006 training set and approximately 36 hours of wall time to converge on the N2C2 2008 training set.

3 Evaluation

We consider two clinical note phenotyping datasets released as part of shared tasks by N2C2 (formally named I2B2): 2006 Smoker Identification [Uzuner et al. 2008] and 2008 Obesity Risk Factors [Uzuner 2009]. The datasets are publicly available[3]. Smoking consists of a single prediction task: select smoking status from four fine-grained options: past smoker, current smoker, non-smoker, and status unclear. Obesity contains a label for obesity and 14 co-occurring morbidities (e.g. congestive heart failure), so a clinical note can have 0 or more applicable labels. For the obesity dataset we train and evaluate only using the “intuitive judgments” labels as these provide the largest annotation coverage over the data.

Each dataset contains a pre-defined training set identical to the data available to participants during the shared task competition period. We train each architecture over each respective training set, and report micro-averaged \( F_1 \) for each architecture on the evaluation set after 1000 training epochs (a suitable number selected during development). We do not perform any additional hyper-parameter optimization since there is no development set. We include for comparison both the best system at the shared tasks and subsequent work with CNNs for these tasks.

Cognizant of the tendency to overfit a shared task dataset after years of research, we report an additional result using a different training scheme for our best architecture. We take the training set and divide it into five partitions. We train the model for 1000 epochs from the same random initialization five times, each time withholding a different partition. We report the mean and variance of the micro-averaged \( F_1 \) when evaluated over the challenge evaluation set across these models. This gives us a sense of the variance expected due to the specific data instances supplied during training, contextualizing the single number obtained from a single training run. Additionally, a single data partition from this validation scheme was precisely the partition utilized for model development.

[3] https://portal.dbmi.hms.harvard.edu/data-sets/
4 Results and Discussion

Table 1: Phenotyping results (micro-averaged $F_1$) of our architectures trained on the respective shared task training sets and evaluated on the evaluation sets.

|                     | I2B2 2006: Smoking | I2B2 2008: Obesity |
|---------------------|--------------------|--------------------|
| $f_{\text{max}}$   | 50.3               | 74.7               |
| $f_{\text{I}}$     | 82.9               | 76.8               |
| $f_{\text{Transformer}}$ | 75.9            | **97.7**           |
| $f_{\text{LSTM}}$  | **98.1** (97.1 ± .48) | **99.7** (93.9 ± .59) |
|                     | Shared Task 1\textsuperscript{st} Place | 90.0               | 95.0               |
|                     | Majority Label Baseline | 81.0               | 74.4               |
|                     | DocBert [Adhikari et al. 2019] | 80.2               | 67.6               |
|                     | CNN [Wang et al. 2019] | 77.0               | –                  |
|                     | CNN + Rules [Yao et al. 2018] | –                  | 96.2               |

We showcase the competitiveness of our architectures with respect to previous methods in Table 1. In both shared tasks, top submissions consisted mainly of hand created regular expression and rules for each label. The top performing system in I2B2 2006 [Clark et al. 2008] utilized handcrafted regular expressions, rules and feature sets to train per-label binary support vector machines. The top performing system in I2B2 2008 [Ware et al. 2009] consisted of purely hand engineered rules for each label. As a simple baseline, we report the performance of predicting the majority occurring label across all training instances at evaluation. DocBert does not outperform either baseline. This is because it utilizes only the first 510 document tokens hence cannot capture any label indicating signal further into the document. For the I2B2 2006 dataset, a recent system [Wang et al. 2019] explored a CNN architecture with word2vec representations but did not outperform the majority label or shared task baseline. Similarly, [Yao et al. 2018] utilized a CNN with word2vec alongside handcrafted features to achieve state of the art performance on I2B2 2008.

Our approach achieves state-of-the-art performance on smoking and obesity. Both naive architectures ($f_{\text{max}}$ and $f_{\text{I}}$) failed to outperform most shared task system baselines across both tasks. In both tasks, $f_{\text{LSTM}}$ performed best. To the best of our knowledge, $f_{\text{LSTM}}$ beats the state of the art on I2B2 2006 by 7% and on I2B2 2008 by 4%.

The multi-head attention based Transformer encoder architecture $f_{\text{Transformer}}$ is competitive but does not outperform top ranking systems on the I2B2 2006 task. We hypothesize this lower than expected performance is associated to lost signal when the dimension-wise max combines the multi-head attention weighted encoder output. Additionally, the utilization of multi-head attention results in the loss of temporal information amongst document sub-chunks (ex. certain document sections always precede others) which may contribute to the observed performance reduction.

The near perfect generalization performance of $f_{\text{LSTM}}$ on both tasks suggests that these datasets are no longer suitable benchmarks for evaluating the effectiveness of novel phenotyping systems based on predictive performance. In light of this, we suggest that these datasets be retired as measures of progress in phenotyping systems. However, it would still be of practical interest to engineer less computationally intensive systems that could maintain similar predictive performance. For example, work on distilling BERT into a smaller and faster model [Sanh et al. 2019].

5 Conclusion

We explore and contribute several document classification architectures that combine representations from state of the art language models. We find that treating document encoding as a sequence modeling task over sequential document chunks is an effective framework for document representation agnostic of the sequence model. All of our proposed architectures perform competitively with previous task baselines. Notably, we beat state of the art by a large margin on a well known smoking status phenotyping task and achieve near perfect generalization performance on obesity phenotyping without any task specific engineering. We make our Pytorch implementation publicly available.

[https://github.com/AndriyMulyar/bert_document_classification](https://github.com/AndriyMulyar/bert_document_classification)
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