Assertion modeling and its role in clinical phenotype identification

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Abstract
This paper describes an approach to assertion classification and an empirical study on the impact this task has on phenotype identification, a real world application in the clinical domain. The task of assertion classification is to assign to each medical concept mentioned in a clinical report (e.g., pneumonia; chest pain) a specific assertion category (e.g., present, absent, and possible). To improve the classification of medical assertions, we propose several new features that capture the semantic properties of special cue words highly indicative of a specific assertion category. The results obtained outperform the current state-of-the-art results for this task. Furthermore, we confirm the intuition that assertion classification contributes in significantly improving the results of phenotype identification from free-text clinical records.

1. Introduction
There is a great amount of information captured in physicians’ comments made during health care. Increasingly, researchers are finding valuable uses by mining and aggregating this data in clinical and translational studies which lead to improved patient care, but progress is slow since study cohorts first need to be constructed, and currently, this can only be done through manual annotation of the patient records. As a result, there is a great deal of interest in identifying critical illness phenotypes, and other medical concepts, automatically. The task cannot be handled by simple key word matching, since the phenotype may be mentioned even when the physician is ruling it out or specifying the conditions under which the phenotype is observed.

An important contribution to address this issue is the Informatics for Integrating Biology and the Bedside (i2b2)/Veteran’s Affairs (VA) challenge. Each year, this challenge focuses on a specific problem to enable clinical information extraction. The 2010 i2b2/VA challenge introduced assertion classification as the shared task, formulated such that each medical concept mentioned in a clinical report (e.g., asthma) is associated with a specific assertion category [1]; 21 teams competed in this task, confirming that assertion classification is perceived to be an important task. The assertion categories that were included in the task are: present, absent, conditional, hypothetical, possible, and not associated with the patient (not patient, for short). As observed, because it requires the identification of both negated and uncertain medical concepts, the task of classifying medical assertions is closely related to both tasks of negation detection and hedge (or speculation) identification that were recently studied in [2,3].

While the systems report good results, error analysis suggests that the performance can still be improved, particularly for the categories that express hedging (i.e., conditional, hypothetical, and possible). In this paper, we introduce several novel features that explore the syntactic information encoded in dependency trees in relation to special cue words for these categories. We find that each of the feature classes added provides significant improvement.

With our new assertion classifier, we return to the task at hand, to automatically identify critical illness phenotypes, specifically pneumonia; while pneumonia identification is of great value to clinical researchers, there are many other phenotypes (e.g., sepsis) that can be identified using the same methodology. In this paper, we describe how we map from the six assertion categories to distinguishing positive from negative cases of pneumonia. We also experiment with adding assertion classification to various feature representations for this task, and find that assertion classification provides significant improvement in all experiments, underscoring the importance of semantic phenomena on the interpretation of clinical text.

2. Related work
The task of identifying an assertion category for a given medical concept has been extensively studied in the past several years due
to its importance and direct application in the clinical domain. One of the earlier tools developed in this direction is NegExpander [4]. This tool employs rules based on part-of-speech tags with the purpose of distinguishing positive from negative expressions in clinical text. Later on, Mutalik et al. [5] proposed NegFinder – a tool relying on methodologies for parsing formal computer languages which encodes various rules used for extracting a large set of patterns associated with negated medical concepts.

A more simple but powerful regular expression-based tool for deciding on a negated concept is NegEx [6]. Since at the core of the NegEx algorithm lies a list of rules matching negative expressions which can be easily modified, this tool drew favorable attention to be enhanced with additional functionalities. In this regard, Golding and Chapman [7] incorporated machine learning techniques into the NegEx algorithm, Chapman et al. [8] proposed Context, a generalized version of NegEx, and Uzuner et al. [9] implemented the Extended NegEx (ENegEx) tool in order to handle alter-association assertions. Uzuner et al. [9] also compared ENegEx with a machine learning approach for assertion classification.

More recently, Elkin et al. [10] extended the NegFinder and NegEx tools by employing a negation ontology. In their study, Elkin et al. also performed an error analysis in order to determine the most common cases where negation is not properly identified. Also, Huang and Lowe [11] proposed an approach using regular expressions matching structured information that is encoded into syntactic parse trees. This way, they were able to identify not only the negated medical concepts situated into a close proximity from a negation signal (such as no or not), but also the ones located distantly from these signals.

The performances of the tools described above are, however, difficult to compare because most of these tools are not publicly available. Also, the access to the datasets on which these tools were evaluated is restricted due to the policies imposed by clinical data warehouses. On the other hand, the initiative of the i2b2/VA organizers to provide a standard, publicly available dataset of deidentified medical records gave to the researchers from the biomedical informatics field the opportunity to create a new generation of assertion classification tools which can be ranked on a common evaluation platform. The most successful approaches participating in the 2010 i2b2/VA challenge for the assertion classification task lie in performing significant feature engineering. In concordance with this, the medical concepts were specified, and the documents were already segmented into sentences and tokenized.

Example 1. Example of an artificially created clinical report emphasizing assertion annotations of medical concepts.

| s1 | History of present illness: |
| s2 | For the past several years, the patient has continued to have significant exercise intolerance which has limited his ability to work. |
| s3 | He does become slightly short of breath when lifting weights. (conditional) |
| s4 | Family history: |
| s5 | One of his brothers has had a myocardial infarction. (not patient) |
| s6 | Physical examination: |
| s7 | On physical examination, the patient has no fever. (absent) |
| s8 | Lungs revealed occasional wheezing bilaterally at both bases. (present) |
| s9 | Hospital course: |
| s10 | He was also started on antibiotics for possible bronchitis or community-acquired pneumonia. (possible) |
| s11 | Discharge Medications: |
| s12 | Lasix 40 mg Tablet Sig: One (1) Tablet PO once a day as needed for shortness of breath or wheezing. (hypothetical) |

3. Classifying medical assertions

To enable the study of assertion classification, the i2b2/VA organizers provided a set of 826 documents annotated by medical experts, which was split into 349 and 477 training and test documents respectively, and 827 non-annotated documents. Overall, the training and test datasets contain 11,968 and 18,550 annotated concepts, respectively. The documents consist of deidentified discharge summaries and progress notes, collected from three different institutions. The set of annotated documents have the following distribution of assertion categories: approximately 69% of concepts are present, ~20% absent, ~4.5% hypothetical and possible, and <1% conditional and not patient. For the assertion classification task, the medical concepts were specified, and the documents were already segmented into sentences and tokenized.

In Example 1, we show an artificially created clinical report with sentences extracted from the i2b2/VA dataset. These sentences represent examples for each assertion category, in which the medical problems are emphasized in boldface. For instance, the medical problem from the sentence s3 is annotated as conditional because it is experienced by the patient only under specific conditions (in this case, only when the patient performs intense physical activities). As is also observed in Example 1, a clinical report can be structured into sections. Here s1, s4, s5, s8, and s10 represent examples of section headers which mark the beginning of a new section in the report. These types of section headers are usually seen in discharge summaries.

3.1. Features

As noted by many participating teams in the assertion classification task, an important factor to obtaining top results for this task lies in performing significant feature engineering. In concordance with this observation, we implemented a wide diversity of features that explore the contextual information of a medical concept at sentence and report level. To extract these features, we first pre-processed the documents using SPLAT [15], a state-of-the-art NLP platform which include lemmatization, Porter stemming,
and constituent and dependency parsing. Then, based on an initial set of features, we trained an SVM multi-class classifier with default parameter settings [16] and employed an optimization method to decide which features from the initial set provide the best performing results. This optimization method is based on a greedy algorithm that iteratively selects the most salient features from the initial set. For determining the final set of features, we ran this feature selection method over the training set using a 10-fold cross validation evaluation scheme. The features from this final set are listed in Table 1.

In the remaining part of this section, we describe in detail the features from Table 1. For a better representation, we grouped the features based on their common characteristics and on the impact they have on improving the classifier’s performance.

3.1.1. Basic features (f1–9)

This set of features encodes the surrounding contextual information of the medical concept at the sentence level. For instance, the feature f2 associated with the concept from S3 in Example 1 is “‘lift weight’, which represents expressions occurring right after a concept such as when, after, or while lifting weights.” The set of binary features f1 tries to determine if the concept is part of a list, or if it is adjacent to a question mark, which, in medical language, often indicates uncertainty. The list of negative prefixes used for extracting the binary features f1 is: ab, de, di, il, in, ir, re, un, no, mel, mal, and mis. This list is identical with the list used by Kim et al. [14]. One important feature is f6, which is based on the output generated by the ConText algorithm [8]. This algorithm uses regular expressions to identify three clinical properties of a clinical condition: negation, temporality, and experiencer.

3.1.2. Section features (f10–12)

Since clinical records are often structured into sections, extracting features that identify the section headers proved to be useful for specific assertion categories. For instance, most of the medical concepts present in allergies sections are annotated as conditional; similarly, many concepts in the family history sections are not associated with the patient.

In this dataset, identifying section headers in clinical notes is relatively straightforward. Most of the section headers were selected from the sentences that end with a colon; however, the sentences that represent a special set of expressions (e.g., instructions, family history, medications, discharge instructions, etc.) or the ones that start with these expressions followed by a colon are also marked as section headers.

3.1.3. Category specific features (f13)

Due to the fact that the distribution of the assertion categories is highly non-uniform, the multi-class classifier has the tendency to favor the features covering the majority categories and is not able to assign sufficient weight to some of the features designed for instances corresponding to the minority categories. To address this issue, we built a simple analysis tool that extracts all possible adjacent n-grams to the left and right of the medical concepts from the training set and counts how many times each n-gram g is associated with every assertion category c. Based on these frequency counts, denoted as f\(g, c\), we computed a score that measures how relevant an n-gram is for a specific category.2 For instance, the left uni-gram no has a score of .99 (~1000/1009) for absent because it occurred 1000 times for this category, 3 times each for present and not patient, and 1 time each for conditional, possible, and hypothetical.

After computing the scores, we built a binary feature for each of the six categories. One such feature is true for a given concept if the concept has an adjacent n-gram relevant to the category associated with the feature. For an n-gram to be relevant to one of the six categories, it needs to be counted at least three times and the score for the category to be >.95.

In addition, we ran greedy optimization techniques for the most relevant features on the training set and used them as heuristics for predicting the instances associated with the minority categories. Examples of highly relevant features associated with the conditional category and used for this purpose are the bi-grams occurring to the left of the medical concept on exertion and with exertion.

3.1.4. Assertion focus features (f14–19)

This novel set of features tries to capture whether a medical concept is within the semantic influence of a special word expression that is able to signal a specific assertion category. The word expressions we identified as being able to signal an assertion category are the negation cues (e.g., not, without, and absence of) from the BioScope corpus [17], the speculative (or hedge) cues (e.g., suggest, possible, and might) from the same corpus, the temporal sig-

2 The score of an n-gram g over a category c is given by f(g, c)/\(\sum_{c'} f(g, c')\). This is a simplification to the method that computes correlation scores based on statistical tests.
nals (e.g., after, while, on, and at) from TimeBank [18], and a list of kinship terms (e.g., father, mother, sister, and aunt) from the Longman English Dictionary. We refer to all these word expressions as assertion cues.

Based on this list of special words, the features f_{14-19} extract various forms of knowledge that encode the connection between an assertion cue and a medical concept in order to help the classification algorithm decide whether or not the concept is within the focus of the assertion cue. To our knowledge, this is the first system to make use of syntactic features in the assertion classification task. The idea of assertion focus is inspired from the semantic theory of negation. According to Quirk et al. [19], the focus of negation is the most contrastive part from the negation scope whereas the negation scope is the part of the language that encodes the meaning of a negative item. A recent study on identifying the negation focus is described in [20].

Although most of these features explore the syntactic information encoded in dependency trees, we also extracted path features from constituent trees. However, those features did not end up being selected in the final feature set.

4. Assertion classification results

Table 2 shows the results achieved by our system on both training and test sets. The results for each category are reported in terms of precision (P) and recall (R) whereas the results measuring the performance over all assertion categories (listed as ‘overall’ in the table) are reported in terms of macro- and micro-averaged F-measure (macroF and microF in the table). The micro-averaged F-measure is computed by averaging over all the medical assertions and therefore it assigns an equal weight to each data instance. In consequence, this measure is dominated by the results associated with the majority categories. On the other hand, the macro-averaged F-measure gives an equal weight for each assertion category by computing locally an F-measure for each such category and then averaging these measures to obtain the final score. Since the micro-averaged F-measure is considered as the primary measure for assertion classification, the feature selection optimization is performed based on this measure.

The first four rows in Table 2 show the cross validation results over the training instances. To measure the impact of each feature set, we split the experiments by cumulatively adding each set to over the training instances. To measure the impact of each feature is performed based on this measure.

When adding the second row from the training set, where the former was annotated as the category 63.74% (109) were classified as present. In fact, these misclassifications accounted for 91.6% of the conditional false negatives. Our analysis of the system predictions over the training set revealed that most of the errors for the conditional category correspond to instances where the condition is expressed in text after the medical problem and is introduced by a preposition or subordinating conjunction followed by a construction expressing a physical activity (with climbing, after exercise, when taking, while doing exercise, when getting up, etc.). We tried to capture these cases by first looking for expressions that match a prepositional phrase or subordinating clause and having a (NP IN NP) or (SBAR WHADVP S) syntactic structure. Then, we tested whether the head of the noun phrase or of the clause s is a hyponym of the synset representing a human action or activity in WordNet [22]. However, this feature was not able to make a clear separation between conditional and present instances due to the fact that many present instances matched the above criteria even after we tried to narrow down the list of activity hyponyms.

Inconsistencies in annotations for the conditional category was another factor that negatively influenced the classifier’s performance. One such annotation inconsistency is shown by Example 1 and Example 2 from the training set, where the former was annotated as conditional and the latter as present.

(1) The patient is allergic to Ciprofloxacin, morphine sulfate and Droperidol.

(2) The patient is allergic to prednisone and penicillin.

A similar example of inconsistency for allergic was also presented in [23].

Table 2

<table>
<thead>
<tr>
<th>System configuration</th>
<th>Absent</th>
<th>Not patient</th>
<th>Conditional</th>
<th>Hypothetical</th>
<th>Possible</th>
<th>Present</th>
<th>Overall</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>P</td>
<td>R</td>
<td>P</td>
<td>R</td>
<td>P</td>
<td>R</td>
<td>P</td>
</tr>
<tr>
<td>Training set</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Basic</td>
<td>95.77</td>
<td>95.66</td>
<td>85.48</td>
<td>57.61</td>
<td>76.19</td>
<td>31.07</td>
<td>94.26</td>
</tr>
<tr>
<td>+Section</td>
<td>96.20</td>
<td>95.78</td>
<td>92.68</td>
<td>82.61</td>
<td>73.33</td>
<td>32.04</td>
<td>95.36</td>
</tr>
<tr>
<td>+Category specific</td>
<td>96.50</td>
<td>95.78</td>
<td>92.94</td>
<td>85.87</td>
<td>80.39</td>
<td>39.81</td>
<td>95.51</td>
</tr>
<tr>
<td>+Assertion focus</td>
<td>96.87</td>
<td>96.37</td>
<td>95.18</td>
<td>85.87</td>
<td>82.35</td>
<td>40.78</td>
<td>95.54</td>
</tr>
<tr>
<td>Test set</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Kim et al.</td>
<td>96.31</td>
<td>94.71</td>
<td>97.52</td>
<td>81.38</td>
<td>81.25</td>
<td>30.41</td>
<td>92.07</td>
</tr>
<tr>
<td>Our results</td>
<td>95.71</td>
<td>93.88</td>
<td>91.79</td>
<td>84.83</td>
<td>80.00</td>
<td>30.41</td>
<td>92.42</td>
</tr>
</tbody>
</table>

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5. Positive and negative assertion categories

In practical clinical applications it is often required to identify a patient as being either positive or negative for a specific phenotype by analyzing the clinical information encoded into the patient reports. Therefore, in order to use our assertion classifier for such types of applications, we first need to determine what is the best way of mapping the six assertion categories into the positive and negative classes. From all \( \sum_{k=1}^{5} \binom{6}{k} = 2^6 - 2 - 62 \) ways of performing these mappings, we identified only two of them as more plausible. Since it is obvious that present belongs to positive and absent and not patient to negative, the two mappings encode whether the assertion categories that express hedging (i.e., conditional, hypothetical, and possible) are attached to the positive or negative class. We denote as hedge\( ^{+} \) and hedge\( ^{-} \) the mappings where the hedge categories are attached to positive and negative, respectively. Another aspect that we considered is whether to perform these two mappings before training the assertion classifier (i.e., building a binary predictor) or after learning the assertion model (i.e., mapping the assertion predictors).

Table 3 shows the results of the assertion classifier based on the mapping configurations described above on the i2b2/VA dataset. As can be noticed, the results when performing the two mappings after learning the assertion model, slightly outperform the results corresponding to the before configuration. Also, for both after and before configurations, the hedge\( ^{+} \) mapping is better predicted by the classifier than the hedge\( ^{-} \) mapping. Although mapping the hedge classes to the positive class gives slightly better performance, it remains of course to be seen which mapping results in the best performance in the context of the clinical application introduced in the following section.

6. A clinical case study

In this section, we extend our previous work described in [24] and investigate the role of assertion classification in identifying complex illness phenotypes such as pneumonia. Although our current study focuses on pneumonia identification, the methodology we propose for this task can be easily adapted for detecting other phenotypes as well.

Using all the reports associated with a patient, the task of pneumonia identification is to classify the patient as positive or negative for pneumonia. Because this type of application is resource intensive and, at the same time, requires real-time assessments, automatic methods for detecting different types of pneumonia are currently needed in clinical research and by hospitals for pneumonia surveillance.

6.1. Framework for pneumonia identification

The simplest method for deciding whether a patient is positive for pneumonia is to extract the pneumonia expressions from the patient clinical notes, and their assertion values, as described in Section 3. However, in some cases, pneumonia expressions are not even mentioned in the reports. When present, it is in fact often noted that clinical notes will use hedging terms even when there is a fairly high certainty for a patient to be positive for pneumonia, for reasons of liability or because conclusive test results are yet to arrive.

6.1.1. Statistical feature selection

To capture the relevant clinical information for pneumonia identification, we developed a supervised learning framework in which the features associated with a patient correspond to uni-grams and bi-grams of words and Unified Medical Language System (UMLS) concepts from the patient notes. For processing clinical data and classification we used the same NLP platform and SVM classifier as for assertion classification, and for extracting the UMLS concepts we used the 2011 version of MetaMap [25]. In this process, we configured MetaMap such that only the UMLS concepts having the highest mapping score for each match are considered.

Unlike a conventional learning framework, however, we also implemented a methodology to select only the most informative features for pneumonia identification. Specifically, this methodology uses statistical hypothesis testing to measure the association strength between each feature from the training set and the two categories of this task. As a result, the features will be ranked based on those values such that the ones with a strong association to the two categories will be on top. Finally, only the most relevant features that are within a specific threshold will be selected for training. This methodology, called statistical feature selection, has been successfully applied in text categorization [26,27].

To rank the set of features associated with a feature type (e.g., word bi-grams), we constructed a contingency table for each feature from the set and used the \( t \)-statistic\(^3 \) to determine whether there is an association between the feature and the two categories. For instance, Table 4 lists the top 5 uni-grams, bi-grams, and UMLS concepts ranked by this statistical test. As can be observed, many of these features are closely linked to the known causes, clinical signs, and symptoms of pneumonia.

6.1.2. Assertion of pneumonia expressions

In order to determine whether the task of assertion classification plays a key role in pneumonia identification, we implemented a binary feature, called the assert feature, which assigns to each patient

\[^{3}\text{ We also experimented with } \chi^2 \text{ and Fisher exact test, but these measures did not perform as well as the } t \text{-test.}\]

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### Table 3

<table>
<thead>
<tr>
<th>Mapping configuration</th>
<th>Positive</th>
<th></th>
<th></th>
<th>Negative</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>P</td>
<td>R</td>
<td>F</td>
<td>P</td>
<td>R</td>
<td>F</td>
</tr>
<tr>
<td>Before</td>
<td>94.65</td>
<td>97.59</td>
<td>96.10</td>
<td>93.87</td>
<td>87.00</td>
<td>90.31</td>
</tr>
<tr>
<td>Hedge( ^{+} )</td>
<td>92.21</td>
<td>99.01</td>
<td>98.61</td>
<td>95.95</td>
<td>92.89</td>
<td>94.40</td>
</tr>
<tr>
<td>Hedge( ^{-} )</td>
<td>94.51</td>
<td>98.28</td>
<td>96.36</td>
<td>95.53</td>
<td>86.55</td>
<td>90.82</td>
</tr>
<tr>
<td>After</td>
<td>94.40</td>
<td>98.93</td>
<td>98.67</td>
<td>95.70</td>
<td>93.66</td>
<td>94.67</td>
</tr>
</tbody>
</table>

### Table 4

<table>
<thead>
<tr>
<th>Top 5 most informative uni-grams, bi-grams and UMLS concepts according to ( t )-statistic.</th>
<th>Uni-gram</th>
<th>Bi-gram</th>
<th>UMLS concept</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sputum</td>
<td>Sputum cx</td>
<td>Microbial sputum culture</td>
<td></td>
</tr>
<tr>
<td>Suctioning</td>
<td>Sputum culture</td>
<td>Sputum</td>
<td></td>
</tr>
<tr>
<td>H1N1</td>
<td>H1N1</td>
<td>Influenza</td>
<td></td>
</tr>
<tr>
<td>Ventilatory</td>
<td>H1N1 Influenza</td>
<td>Influenza</td>
<td></td>
</tr>
<tr>
<td>Consolidation</td>
<td>Acquired pneumonia</td>
<td>Influenza preparation</td>
<td></td>
</tr>
</tbody>
</table>
tient a label corresponding to positive or negative pneumonia. For this purpose, we selected from the clinical reports only the medical expressions identified by MetaMap that have the same identifier as the pneumonia concept (CUI:C0032285) in the UMLS Metathesaurus [28]. For a more complete set, we also ran simple regular expressions to identify the word pneumonia, an abbreviation often used by physicians for pneumonia but which is not tagged as a pneumonia concept in UMLS Metathesaurus yet. After we ran the assertion classifier for all the pneumonia concepts of a patient, we counted how many times each of the six assertion values were identified, and then we mapped the most frequent value to one of the two categories of pneumonia identification as described in Section 5. For the binary features corresponding to patients with no pneumonia concepts identified in their reports, we assigned a default value of negative pneumonia.

### 6.2. Pneumonia identification experiments

Although the F-measure is the primary measure for this task, we also report the negative predictive value (NPV) and specificity (Spec.), since accurately identifying patients for negative pneumonia is equally important in the clinical domain. Furthermore, we measured the statistical significance using the same randomization test as for assertion classification.

The dataset used in our study consists of 3442 reports (e.g., admission notes, discharge summaries, and daily progress notes) corresponding to a cohort of 236 patients. The annotation was performed at patient level by an experienced medical expert based on the information encoded in the patient reports. From the 236 patients, 44 were identified as positive and the remaining 192 as negative for pneumonia.

#### 6.2.1. Baselines

A simple baseline we considered is a rule-based approach which uses as criteria for pneumonia identification the value of the assert feature. Since the dataset of 236 patients was also used by Yetisgen-Yildiz et al. [29] for the same task, we considered their approach as another baseline for our system. In their supervised approach, Yetisgen-Yildiz et al. [29] extracted word n-grams, UMLS concepts, and their corresponding semantic types, and represented the feature vector associated with a patient as a ‘bag of words’ from various combinations of the above mentioned feature types. Using a 5-fold cross validation scheme, their system reached the best performing results when only the word n-grams were considered. For an accurate comparison, we used the same evaluation scheme and dataset split as Yetisgen-Yildiz et al. [29] in all our experiments.

### Table 5

Baseline results for pneumonia identification.

<table>
<thead>
<tr>
<th>System configuration</th>
<th>P</th>
<th>R</th>
<th>NPV</th>
<th>Spec.</th>
<th>F</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yetisgen-Yildiz et al.</td>
<td>73.90</td>
<td>38.60</td>
<td>87.32</td>
<td>96.90</td>
<td>50.70</td>
</tr>
<tr>
<td>Assert rule (hedge)</td>
<td>44.22</td>
<td>86.36</td>
<td>95.89</td>
<td>72.92</td>
<td>56.72</td>
</tr>
<tr>
<td>Assert rule (hedge')</td>
<td>34.11</td>
<td>100.00</td>
<td>100.00</td>
<td>55.73</td>
<td>50.87</td>
</tr>
</tbody>
</table>

Table 5 shows the results of the two baselines described above. As observed, the results of the rule-based approach using both mapping configurations outperform the results of Yetisgen-Yildiz et al. [29]. Furthermore, these results indicate that mapping the hedge assertions to the negative category constitute a better choice for pneumonia identification. Based on this observation, we used the hedge mapping for all the experiments in the next section involving the assert feature. Because the size of the dataset used in this study is relatively small, none of the baseline results are statistically significant from the other.

#### 6.2.2. Experimenting with the assert feature

We performed a set of experiments to assess the benefit of using the assert feature in combination with other feature types. In this regard, we studied how the performance of our system evolves for these experiments when using various threshold values on the ranked word lists. Fig. 1 shows this experimental study. For each experiment, we considered 27 different values that capture the variation of the threshold for selecting from a range of 10 to 40,000 significant word n-grams. For instance, if the feature extractor selects the first 30 features from the ranked lists of word n-grams, our system will achieve 70.71 F-measure (left plot, words + assert experiment, threshold = 30). In all the experiments shown in this figure, we used a threshold value of 50 for the selection of the concept n-grams.

Although there exist noisy features in the ranked lists of words and concept n-grams, which cause some fluctuation in the performance across the range of threshold values, the experiments using the assert feature show substantial improvements over the other experiments. In Fig. 1, it is also worth observing that the results for the last threshold values are close to the results achieved by Yetisgen-Yildiz et al. [29] since, similar to the configuration of their system, our feature extractor selects almost the entire initial set of features.

Table 6 lists the best performing results as well as the aggregate results over all the threshold values considered. For the concepts and concepts + assert experiments, we used a limited set of threshold values (up to 10,000), since the total number of concepts extracted is relatively smaller than the number of word n-grams. The thresholds for the word and concept lists are denoted in this table as $w_{th}$ and $c_{th}$, respectively. To compute the aggregate results, we used one contingency table with the results generated when considering all threshold values. For example, a total of 236 x 27 system predictions were considered for all the experiments involving word n-grams. For this configuration, the results when adding the assert feature are statistically significant at

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Fig. 1. A study on the impact of the performance results of feature type combinations when using various threshold values on the ranked list of word n-grams.

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The best performing results and the aggregate results for pneumonia identification. The differences in performance for the aggregate results when adding the assert feature are statistically significant at p < .001.

<table>
<thead>
<tr>
<th>Feature set</th>
<th>Best results</th>
<th>Aggregate results</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>wth, wthb</td>
<td>p, r, npv, spec, f</td>
</tr>
<tr>
<td>Concepts</td>
<td>– 5000</td>
<td>60.71 77.72 94.44 88.54 68.00</td>
</tr>
<tr>
<td>Concepts + assert</td>
<td>– 5000</td>
<td>67.31 79.55 95.11 91.15 72.92</td>
</tr>
<tr>
<td>Words</td>
<td>10,000</td>
<td>85.00 77.27 94.90 96.88 80.95</td>
</tr>
<tr>
<td>Words + assert</td>
<td>5000</td>
<td>– 87.80 81.82 95.90 97.40 84.71</td>
</tr>
<tr>
<td>Words + concepts</td>
<td>10,000 50</td>
<td>82.22 84.09 96.34 95.83 83.15</td>
</tr>
<tr>
<td>Words + concepts + assert</td>
<td>10,000 50</td>
<td>90.24 84.09 96.41 97.92 87.06</td>
</tr>
</tbody>
</table>

p < .001. Also, all the best performing results are statistically significant when compared with the baseline results.

7. Discussion

Our study of assertion classification is fundamentally a study of semantics. Although a medical concept may not at first be conceived of as a predication, it may be viewed as a state that is true to some degree, or not, for a specific patient. Thus, our problem is related to determining the truth-conditionality of predication, which is a fundamental process in understanding text. What we have found in our experiments is that ideas, the truth-conditions of the medical concept matter when trying to determine whether or not the patient exhibits that phenotype. In fact, as we observe in Fig. 1, the assertion feature helps for all possible representations for modeling the phenotype classification. Currently, the assertion feature is present only for medical concepts denoting pneumonia, but as there are other concepts that are also predictive of pneumonia, we expect that expanding our assertion feature to all concepts will show yet further improvements.

Previous studies have used simple word-based features for assertion classification, usually where the cue words are found within some window of words before and after the medical concept. Table 2 captures our experiments that show that each of the sets of features we propose in addition to the baseline features provides a statistically significant performance improvement. To our knowledge, this is the first study to include syntactic features for modeling assertion classification, which had a positive impact on predicting all assertion categories. As we mention in the text, modeling the conditional category is particularly challenging, so it is encouraging to see improvement using syntactic features.

Lastly, we find that it is the application in which the semantic feature is used, that ultimately determines the best mapping from the theoretically motivated six assertion classes to the practically-oriented two classes, positive and negative. In the context of our application for predicting pneumonia, it is better to model the phenotype by mapping the hedge classes to a negative instance of pneumonia.

References


Although our findings clearly indicate that assertion classification plays a key role in pneumonia identification, previous studies did not find this task helpful in improving various other clinical applications [30].