Unsupervised Numerical Reasoning to Extract Phenotypes from Clinical Text by Leveraging External Knowledge

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Abstract Extracting phenotypes from clinical text has been shown to be useful for a variety of clinical use cases such as identifying patients with rare diseases. However, reasoning with numerical values remains challenging for phenotyping in clinical text, for example, temperature 102°F representing Fever. Current state-of-the-art phenotyping models are able to detect general phenotypes, but perform poorly when they detect phenotypes requiring numerical reasoning. We present a novel unsupervised methodology leveraging external knowledge and contextualized word embeddings from ClinicalBERT for numerical reasoning in a variety of phenotypic contexts. Comparing against unsupervised benchmarks, it shows a substantial performance improvement with absolute gains on generalized Recall and F1 scores up to 79% and 71%, respectively. In the supervised setting, it also surpasses the performance of alternative approaches with absolute gains on generalized Recall and F1 scores up to 70% and 44%, respectively.

Key words: Numerical Reasoning, Phenotyping, Contextualized Word Embeddings, Unsupervised Learning, Natural Language Processing, Deep Learning
1 Introduction

Extracting phenotypes from clinical text has been shown crucial for many clinical use cases [37] such as ICU in-hospital mortality prediction, remaining length of stay prediction, decompensation prediction and identifying patients with rare diseases. There are several challenges in extracting phenotypes such as handling a wide variety of phenotypic contexts, ambiguities, long term dependencies between phenotypes, and so on. Numerical reasoning is one of the key challenges as many of the phenotypes rely on bedside measurements such as temperature, blood pressure, heart rate, breathing rate, serum creatinine, hematocrit, glucose levels. We call these terms numeric entities. As these phenotypes require deep reasoning with the numbers, they are often missed or incorrectly predicted by the existing phenotype extraction methods [3][32][15][6][2][17][35].

Existing phenotype extraction methods such as Neural Concept Recognizer (NCR) [2] which are mostly based on state-of-the-art (SOTA) machine learning (ML) and natural language processing (NLP) technologies exploit non-contextualized word embeddings. These methods cannot detect contextual synonyms of the phenotypes which can be mentioned in various ways by clinicians. For example, previous SOTA phenotyping models like NCR and NCBO [15] can capture the phenotype Fever from the sentence “patient is detected with fever” but fail to capture the same from the sentence “patient is reported to have high temperature”. Similarly, in the sentence, “patient is reported to have high temperature in the room with low temperature”, only the former instance of temperature is a phenotype, while the latter is not. The recent study [38] demonstrates the capability of contextualized embeddings (BERT-based [7]) to differentiate the two instances by context. However, none of these methods above are specifically designed to reason with numbers in clinical text, for example, “temperature 102F” representing Fever. While the contextualized embeddings are useful for reasoning in different contexts, they are not sufficient to address numerical reasoning.

In practice, the numerical reasoning for clinical context has specific challenges. First, clinical text may have accumulation of multiple numeric examples in a condensed context such as “Physical examination: temperature 97.5, blood pressure 124/55, pulse 79, respirations 18, O2 saturation 99% on room air.”. In addition, the numeric examples can be mentioned in a variety of different contexts such as “temperature of 102F”, “temperature is 102F”, “temperature is recorded as 102F”, “temperature is found to be 102F”, which make it more challenging to identify the (numeric entity, number) pair namely (temperature, 102F) in this case. Moreover, numbers in clinical text may not be necessarily connected with phenotypes. For example, the number in “patient required 4 days of hospitalization” is not relevant to any phenotype.

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1 In the medical text, the word “phenotype” refers to deviations from normal morphology, physiology, or behaviour, such as skin rash, hypoxemia, neoplasm, etc. [30]. Please note the difference of the phenotypic information to the diagnosis information expressed in ICD-10 codes [25] as the former contributes to the latter.
To the best of our knowledge, previous studies have not addressed these challenges and we propose the first deep learning based (BERT-based) unsupervised methodology in this paper to accurately extract phenotypes by numerical reasoning from various contexts using external knowledge for clinical natural language processing. In summary, our main contributions are as follows:

1. We propose a new approach to accurately detect phenotypes requiring numerical reasoning using natural language processing and deep learning techniques.
2. The approach is unsupervised and does not require manual data labelling.
3. Our approach can detect phenotypes from a variety of different contexts as it uses contextualized word embeddings.

2 Related Work

Phenotyping: Extraction of phenotypes from text has been addressed using several strategies. Shallow matching using linguistic patterns was used extensively by cTAKES [32], MetaMap [3], and Clinphen [6]. Then, the shallow matching was extended to semantic analysis by leveraging non-contextualized word embeddings by the works of [2, 17, 35]. For example, Neural Concept Recognizer (NCR) [2] uses a convolutional neural network (CNN) to build non-contextualized embeddings by leveraging hierarchical medical concepts from biomedical ontologies like Human Phenotype Ontology (HPO) [16]. Finally, [38] showed that using contextualized embeddings from ClinicalBERT [11] helps to detect contextual synonyms of the phenotypes from the text. Similarly, some other works [22, 36, 10, 20] also exploited ClinicalBERT or BioBERT [19] for phenotype detection, but all of these works focus on a limited set or group of phenotypes. None of these methods addresses phenotyping requiring numerical reasoning, so we extend the work and leverage external knowledge with ClinicalBERT to extract phenotypes requiring deep numerical reasoning in different textual contexts.

Numerical Reasoning: Recent works publish new datasets for numerical reasoning [39] and utilise deep learning based models to develop the numerical reasoning skills [34, 9, 31, 13] in respective domains other than the clinical domain. For example, [11] shows gains by using artificially created data on various tasks involving numeracy such as math word problems and reading comprehension. Other works [8, 12, 28] designed special modules for numerical reasoning in text which were then integrated with neural networks. Overall, these models have shown advancements in the respective domains for specialized problems but they did not incorporate clinical knowledge with specific extensive reasoning for clinical applications [33].
3 Methodology

This section presents our unsupervised method for numerical reasoning (NR) in clinical textual notes to extract phenotypes. Figure 1 shows the architecture of the proposed method which includes four steps (1) one-time creation of external knowledge connecting numeric entities and phenotypes, (2) extraction of numbers and lexical candidates for numeric entities from text, (3) creation of contextualized embeddings for numeric entities and lexical candidates and (4) linking candidates to numeric entities by embedding similarity and then determining corresponding phenotypes. This section elaborates all of the above steps.

3.1 External Knowledge

Phenotypes can often be inferred from ranges of numerical values together with numeric entities which are mentioned in clinical text. For example, the clinicians may mention the numeric entity temperature and the value “102 Fahrenheit” in clinical notes to suggest a patient is suffering from the phenotype Fever (HP:0001945). Therefore, we create an external knowledge base to formalise such connections between phenotypes, numeric entities and numerical values.
Table 1: Examples of numeric entities that are used in the study with normal reference range and units. For example, the normal range of body temperature is 36.4 to 37.3 in Celsius or 97.5 to 99.1 in Fahrenheit. The ID column corresponds to that in Table 2.

Table 2: Examples of numeric entities that are used in the study with phenotype labels (including HPO ID and HPO name). Each numeric entity is connected with three phenotype concepts. For example, a patient has Hypothermia (Fever) if their body temperature is lower (higher) than the lower (upper) normal limit. If the body temperature of the patient is inside the normal range, it is negation of the general phenotype Abnormality of temperature regulation. The ID column corresponds to Table 1.

We first manually collect a list of 33 most frequent numeric entities such as temperature, heart rate, breathing rate, serum anion gap and platelet and their corresponding normal reference ranges (values and units) from the website of National Health Service of UK and MIMIC-III database. Table 1 shows examples of numeric entities and their corresponding lower/upper bounds with units. For example, the normal body temperature has a lower bound 97.5 Fahrenheit (36.4 Celsius) and an upper bound 99.1 Fahrenheit (37.3 Celsius).

Those numeric entities are then manually mapped with phenotypes which are defined and standardised by Human Phenotype Ontology (HPO). In most cases, a numeric entity corresponds to three phenotypes depending on whether the actual measurement is lower than, higher than or within the normal reference range. If the actual measurement is lower (higher) than the lower (upper) bound, it means the relevant phenotype is affirmed. For example, the phenotype Hypothermia (HP:0002045) or Fever (HP:0001945) is affirmed if the body temperature is lower than 97.5 Fahrenheit (36.4 Celsius) or higher than 99.1 Fahrenheit (37.3 Celsius).

2 Accessed in November 2021: https://www.nhs.uk
or higher than the normal range, respectively. Otherwise, if the body temperature is inside the normal range, the general phenotype Abnormality of Temperature Regulation (HP:0004370), which is the parent phenotype of Hypothermia (HP:0002045) or Fever (HP:0001945), is negated. Table 2 demonstrates examples of the connections between numeric entities and phenotypes.

Both Table 1 and Table 2 are validated by three expert clinicians with consensus for authenticity and consistency. The external knowledge is created at one time and prior to other steps, which makes the external knowledge reusable.

3.2 Number and Lexical Candidates Extraction

We then extract numbers and their corresponding lexical candidates which are likely to be numeric entities from clinical text. For example, in the input sentence “her pyrexia increased to 102F and she was begun on levofloxacin”, the number “102” is connected with “pyrexia” which is contextually similar to the numeric entity “temperature”. The regular expression patterns are created to extract the numbers which typically appear in alpha-numeric format such as “pyrexia increased to 102F” and “heart rate in 90s”. The numbers which are dates or part of specific clinical concepts such as “vitamin B12”, “O2 saturation” are excluded by using a predefined dictionary of alpha-numeric words [24] as the numbers are not relevant to phenotypes.

After the extraction of numbers, the lexical candidates connected to these numbers are extracted using syntactic analysis. As shown in Figure 2, we focus on (proper) nouns, adjectives and verbs that are connected with syntactic connections to the extracted numbers (heads or children in the syntactic tree). As a special case, we allow one additional hop from the extracted number via the dependency relation ‘obl’ which stands for oblique nominal. For example, in Figure 2 the words “pyrexia”, “increased”, and “begun” are extracted as lexical candidates because they are connected to the extracted number “102F” and therefore are likely to represent numeric entities.

The list of extracted lexical candidates is passed to the following steps to decide the corresponding numeric entities based on context. As the extraction method of lexical candidates is designed to encourage more extraction to increase recall so
that no important word is missed, not all of the lexical candidates will eventually correspond to a numeric entity.

3.3 Contextualized Embeddings for Numeric Entities and Lexical Candidates

We use contextualized embeddings (ClinicalBERT \cite{1}) of numeric entities and lexical candidates to measure their similarity and decide which numeric entity should be assigned to the extracted lexical candidates from the input sentence.

The objective of the model is to learn a semantic space where all possible expressions (including names and synonyms) of one numeric entity are clustered while the expressions of different numeric entities are differentiated. To achieve this, we use ClinicalBERT finetuned with Semantic Textual Similarity (STS) objective defined as follows:

\[
L(e_i, s_j) = \frac{1}{|E| |S|} \sum_{i=1}^{|E|} \sum_{j=1}^{|S|} \left( \cos(h_{e_i}, h_{s_j}) - y_{e_i, s_j} \right)^2,
\]

where \( y_{e_i, s_j} = \begin{cases} 1, & \text{if } s_j \text{ is a synonym of } e_i \\ 0, & \text{otherwise} \end{cases} \) (1)

where \( h_{e_i} \) represents contextualized embedding for the \( i \)th numeric entity \( e_i \) in \( E \). Similarly, \( h_{s_j} \) represents contextualized embedding for the \( j \)th synonym \( s_j \) in \( S \). The ground truth label \( y_{e_i, s_j} \) is 1 if the synonym \( s_j \) is one of the synonyms of the numeric entity \( e_i \) and 0 if otherwise. The loss function aims to maximise the cosine similarity between numeric entities and their corresponding synonyms and minimise the similarity between numeric entities and irrelevant synonyms. The collection of \( \{h_{e_i} \mid e_i \in E\} \) is used as the reference contextualized embeddings of numeric entities created once.

As the training data, we collect all synonyms \( S = \{s_1, s_2, \ldots, s_j \} \) of all the numeric entities \( E = \{e_1, e_2, \ldots, e_i \} \) (listed in Table 2) by connecting the HPO IDs with Unified Medical Language System (UMLS) \cite{5}.

During inference, lexical candidates extracted from input sentences are fed into the finetuned ClinicalBERT based model to produce their contextualized embeddings.

3.4 Embedding Similarity and Deterministic HPO Assignment

Embeddings pairs are formed by Cartesian product of the contextualized embeddings of lexical candidates and reference contextualized embeddings of numeric entities. Then cosine similarity is computed between all the pairs. The pair with the maximum
cosine score above a pre-set threshold gives the selected lexical candidate which in turn gives the corresponding numeric entity.

A sentence may have multiple numbers connected with their corresponding numeric entities. We simply consider the lexical candidates (corresponding to each number) as an independent case for the above Cartesian product which helps extracting multiple candidate numeric entities from a single sentence.

After measuring similarity of embeddings and determining the numeric entities, we deterministically assign the phenotype depending if the corresponding number is lower than the lower bound, inside the normal range, or higher than the upper bound. For example, in Figure 1, the lexical candidate “pyrexia” is extracted and the numeric entity “temperature” is assigned based on contextualized embedding. As the number “102°F” is higher than the upper bound “99.1”, the phenotype Fever (HP:0001945) is eventually assigned.

We also enhance the HPO assignment process by handling different units of numbers (e.g. Fahrenheit and Celsius) because sometimes the units are not explicitly mentioned in text. Therefore, we decide the unit by comparing the ratio of the number to the extreme ends of the normal reference ranges in different units. For example, normal range for temperature is (36.4, 37.3) in Celsius and (97.5, 99.1) in Fahrenheit. If a given number is 92, then we take the ratios as the following. The unit giving the smaller ratio (Fahrenheit in this case) is then used to determine HPO assignment.

\[
\frac{\text{number}}{\text{upper_bound_celsius}} = \frac{92}{37.3} = 2.5 \quad \text{and} \quad \frac{97.5}{\text{number}} = \frac{97.5}{92} = 1.1
\]

| Primary Phenotype | Unit   | Granular Range | Granular Phenotype          |
|-------------------|--------|----------------|----------------------------|
| HPO ID            | HPO Name | Lower | Upper | HPO ID | HPO Name                      |
| HP:0012664        | Reduced ejection fraction | %     | 0     | 29.9   | HP:0012666 | Severely reduced ejection fraction |
| HP:0012664        | Reduced ejection fraction | %     | 30    | 39.9   | HP:0012665 | Moderately reduced ejection fraction |
| HP:0012664        | Reduced ejection fraction | %     | 40    | 49.9   | HP:0012663 | Mildly reduced ejection fraction  |
| HP:0001945        | Fever   | celsius  | 37.4  | 38     | HP:0011134 | Low-grade fever                  |
| HP:0001945        | Fever   | fahrenheit | 99.2  | 100.4  | HP:0011134 | Low-grade fever                  |

Table 3: A list of granular phenotypes under primary phenotypes. For example, the reduced ejection fraction can be further divided into three sub-phenotypes by severity based on the actual percentage mentioned in clinical text.

Moreover, we consider granular phenotypes based on granular sub-ranges as shown in Table 3.
4 Experiment Design

4.1 Datasets

| Test Set (Unsupervised Setting) | Test Set (Supervised Setting) |
|---------------------------------|------------------------------|
| EHRs   | All phenotypes | NR-specific phenotypes | EHRs   | All phenotypes | NR-specific phenotypes |
| 705   | 20,926         | 1,121                    | 170   | 5,047          | 322                      |

Table 4: Statistics (counts) of the test sets in the unsupervised and supervised setting, respectively. The test set in the unsupervised setting includes all manually annotated EHRs. The test set in the supervised setting is a subset of that in the unsupervised setting because some annotated EHRs are used to finetune the baseline models. Please note only Numerical Reasoning (NR) specific phenotypes are used for evaluation as the other phenotypes are not related with numbers in clinical narratives.

We use clinical textual notes from the publicly available MIMIC-III database [14]. In the unsupervised setting, we collected 705 EHR textual notes with 20,926 gold phenotype annotations as shown in Table 4. The gold phenotype annotations were created by three expert clinicians with consensus and the clinicians were specifically asked to identify contextual synonyms of phenotypes such as “drop in blood pressure” and “BP of 79/48” for Hypotension (HP:0002615). Out of these phenotype annotations, we select a subset with 1,121 phenotype annotations (i.e., NR specific phenotypes) which require numerical reasoning based on two criteria: (1) the annotated phenotypes are among one of the HPO IDs that require numerical reasoning as mentioned in Table 2 and Table 3 and (2) the corresponding textual spans of phenotypes contain numbers. The test set in the unsupervised setting is used to compare the proposed NR model with previous unsupervised baseline methods.

In the supervised setting, as 535 out of 705 manually annotated EHRs are used to finetune the baseline methods (like ClinicalBERT), the remaining 170 EHRs are used for testing. In other words, the test set in the supervised setting is the subset of that in the unsupervised setting. Though, the proposed NR model is strictly unsupervised, we compare it with supervised baselines to rigorously assess its performance.

4.2 Implementation Details

We use the Stanford Stanza [27][40] library to extract the lexical candidates for numeric entities using syntactic analysis. In syntactic analysis, we only focus on nouns, adjectives and verbs that are “NOUN”, “PROPN”, “ADJ”, and “VERB” as marked by the Part of speech (POS) tagger and we also optimise the process by adding words with the dependency relation ‘compound’ to capture multi-word phrases like “heart rate” and “blood pressure”. Then, we use Semantic Textual Similarity (STS) model.
from Sentence Transformers [29] library to finetune the ClinicalBERT embeddings with cosine similarity up to 4 epochs using their default hyperparameter[1] along with a train and validation batch size of 16 and 1000 evaluation steps. Mean pooling is used to get embeddings of multi-word UMLS synonyms. The threshold for embedding similarity is set as 0.9 empirically. The implementation of the proposed method also uses some other third-party libraries including PyTorch [26] and spaCy.

4.3 Baselines and Evaluation Methods

We compare the proposed NR model with previous state-of-the-art phenotyping models. In the unsupervised setting, the proposed NR model is compared with unsupervised baselines including NCBO [15], NCR [2] and the unsupervised model by [38]. In the supervised setting, the proposed NR model is compared with the finetuned ClinicalBERT [1] (which is finetuned for phenotyping) and the supervised model by [38]. The NCBO, NCR and finetuned ClinicalBERT are selected as they show better performance than other baseline phenotyping methods (including cTAKES [32], MetaMap [3], Clinphen [6], MedCAT [17], BERT [7], BioBERT [19], SciBERT [4]) in corresponding settings as demonstrated by [38]. Please note the work by [38] publishes one unsupervised and one supervised model hence we compare the proposed NR model with both. We decide not to compare with recent numerical reasoning models (such as [34, 9, 31, 13]) as none of them incorporates clinical knowledge and we find it costly to adapt them to the clinical domain.

We first evaluate the proposed NR model against the baselines by using micro-averaged Precision, Recall and F1-score at the document level. To ensure comparison with previous studies, we follow the practice by [21] and compute the metrics by the following two strategies. (1) Exact Matches: Only the exact same HPO annotations against the gold standard annotations are counted as correct; (2) Generalized Matches: the gold standard annotations as well as predicted HPO annotations are extended to include all ancestor HPO concepts until the root concept Phenotypic Abnormality (HP:0000118) (exclusive) in the HPO hierarchy. All the extended HPO annotations are then de-duplicated and added to the list of gold standard and predicted HPO annotations respectively for evaluation. By the generalized matches, the prediction of HPO concepts which are children, neighbours or ancestors of the target HPO concepts also receives credits.
5 Results and Discussion

5.1 Quantitative Analysis

We report our quantitative results in Table 5 where we evaluate the NR model in the unsupervised setting. We also compare the NR model with the baselines – NCBO, NCR, and unsupervised model by [38] but they perform poorly on the unsupervised test set with straight 0 on all the metrics. This is expected as they are not designed to handle numbers. The NR model performs significantly better than all of them achieving 69% recall and 59% F1 using exact metrics, while 79% recall and 71% F1 using generalized metrics. Precision is relatively lower as we focus on recall to extract more phenotypes, which is motivated by the preference that a model is sensitive to capture more phenotypic features of patients rather than missing ones for better accuracy in downstream clinical use cases [37]. Overall, the NR model shows huge gains which is useful in the absence of costly annotated data.

We also compare the unsupervised NR model with the previous state-of-the-art supervised baseline methods. First, we compare the NR model with the supervised model by [38] which is finetuned with annotated data. This comparison is shown in Table 6 on the supervised test set. Though the supervised model by [38] outperforms its unsupervised version, the proposed unsupervised NR model performs better than the supervised baseline with gains of 12.5% and 5.7% on exact and generalized recall, respectively. However, there is a drop in precision which results in the comparable F1 scores. Moreover, using a combination of both the models achieves the best performance improving score by 21.5% and 14.3% on exact and generalized recall, respectively, and 4.3% and 0.7% gains on exact and generalized F1 scores, respectively. Then, the NR model is compared against the finetuned ClinicalBERT [1] which is finetuned to detect phenotypes. The combination of NR model and supervised model by [38] surpasses the performance of the baseline with gains of 66.4% and 69.7% on exact and generalized recall, respectively and 40% and 44.2% gains on exact and generalized F1 scores, respectively, as shown in Table 6. These results highlight the impact of the NR model which shows better performance than the supervised models eliminating the need of costly human annotations of phenotypes.

| Model                  | Exact         | Generalized   |
|------------------------|---------------|---------------|
|                         | Precision     | Recall        | F1    | Precision | Recall   | F1    |
| NCBO                   | 0             | 0             | 0     | 0         | 0        | 0     |
| NCR                    | 0             | 0             | 0     | 0         | 0        | 0     |
| [38] (unsupervised)    | 0             | 0             | 0     | 0         | 0        | 0     |
| Numerical Reasoning (NR)| 0.5176        | 0.6879        | 0.5907| 0.6479    | 0.7907   | 0.7122|

Table 5: In the unsupervised setting, the comparison of baselines NCBO, NCR, and [38] (unsupervised) with proposed Numerical Reasoning (NR) model shows the superior performance of NR model. Interestingly but not surprisingly, the baseline methods produce zero accuracy as they are not designed to reason by numbers.
| Model                          | Exact | Generalized |
|-------------------------------|-------|-------------|
|                              | Precision | Recall | F1   | Precision | Recall | F1   |
| Finetuned ClinicalBERT        | 0.8235 | 0.181 | 0.2968 | 1.000     | 0.2229 | 0.3646 |
| [38] (supervised)             | 0.6791 | 0.6293 | 0.6532 | 0.8245    | 0.7762 | 0.7996 |
| Numerical Reasoning (NR)      | 0.5952 | 0.7543 | 0.6664 | 0.7290    | 0.8339 | 0.7780 |
| [38] (supervised) + NR        | 0.5921 | 0.8448 | 0.6963 | 0.7175    | 0.9201 | 0.8062 |

Table 6: The comparison of supervised baselines with the proposed Numerical Reasoning (NR) model in the supervised setting shows that the NR model increases recall significantly by finding more phenotypes even without supervision. Please note supervised setting refers to a subset of unsupervised setting test set which is created to compare unsupervised NR with the supervised baselines.

5.2 Qualitative Analysis

We investigate the numerical reasoning capabilities of the proposed NR model and other baseline methods by eye-balling example sentences having different contexts. In the sentence “patient has a temperature of 102F.”, NCR, NCBO, and [38] (unsupervised) do not detect any phenotype. But after adding the word high, i.e., “patient has a high temperature of 102F.”, [38] (unsupervised) correctly detects the phenotype Fever (HP:0001945). However, the predicted textual span is “high temperature” only ignoring the number 102F. It indicates that the [38] (unsupervised) relies on context without considering numbers, while NCR and NCBO still do not detect any phenotype. When the word “temperature” is changed to “fever” and the whole sentence becomes “patient has a high fever of 102F.”, all three unsupervised baseline methods can correctly detect the phenotype Fever (HP:0001945) though the the number is still ignored in the predicted textual span. Overall, we observe all the unsupervised baseline methods solely rely on the textual content by ignoring the numbers, though [38] (unsupervised) can find contextual synonyms of phenotypes.

In contrast, the proposed NR model correctly detects the phenotype from all the three variants of the original sentence with the correct textual spans which include numbers. More precisely, the target textual spans are “temperature of 102F.”, “temperature of 102F.”, and “fever of 102F” with the phenotype Fever (HP:0001945) for the three sentences above, respectively. We observe the similar behavior given the sentence “patient has a breathing rate of 27.” with the phenotype Tachypnea (HP:0002789) as well as “patient has a serum creatinine of 1.7.” with the phenotype Elevated serum creatinine (HP:0003259). The model [38] (unsupervised) detects the phenotype (still ignoring the numbers) when an indicative word like “high” is added, while NCBO and NCR miss the annotations with the exception for the latter sentence where NCR detects the phenotype after “high” is added to the sentence. In short, the results suggest that the proposed NR model reasons with the numbers effectively in different contexts without supervision.

The supervised model by [38] overall performs much better with reasonable accuracies than the unsupervised baselines which give straight 0 scores. However, it still lacks the capabilities to reason with numbers. For instance, though the [38] (super-
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(unsupervised) correctly predicts the phenotype Fever (HP:0001945) from the sentence “patient has a temperature of 102F.”, if the number in the sentence is changed from 102F to 92F and the target phenotype is therefore changed to Hypothermia (HP:0002045), the (supervised) still predicts fever mistakenly. Similar incorrect predictions are observed when the target phenotype is changed from Tachypnea (HP:0002789) to Bradypnea (HP:0046507) and from Elevated serum creatinine (HP:0003259) to Decreased serum creatinine (HP:0012101). We hypothesize Fever is far more common than Hypothermia in the training data, so the model is finetuned with bias towards the highly frequent phenotypes. This may result in the inflation of the scores in Table 4 for (supervised) which overestimates its numerical reasoning capabilities. Based on the observation, we conclude the supervision without additional tailored learning objectives is not sufficient to obtain the numerical reasoning capabilities.

However, there are some cases where the NR model fails to produce accurate predictions. For example, in the text - “Pt still with scant bibasilar crackles. Sat @ 97% on 2L NG. Continuing with oral HTN meds and Dig.”, the model predicts Abnormal blood oxygen level (HP:0500165) to negate the phenotype “Sat @ 97%” as 97% is within normal reference range for blood oxygen, i.e., 95%-100%. However, the correct phenotype is Hypoxemia (HP:0012418) as the patient achieved this normal range using some external oxygen which implies from the phrase “2L NG”.

5.3 Ablation Studies

We conduct two ablation studies to probe the benefit of contextualized embeddings and the learning objective for finetuning in Equation 1.

| NR Model with                                | Exact            | Generalized        |
|----------------------------------------------|------------------|--------------------|
|                                              | Precision        | Recall             | F1    | Precision | Recall | F1    |
| Keyword based shallow matching               | 0.6854           | 0.2641             | 0.3813| 0.7745    | 0.3449 | 0.4773|
| Pretrained contextualized embeddings         | 0.5065           | 0.3758             | 0.4314| 0.6006    | 0.465  | 0.5241|
| Finetuned contextualized embeddings (used by the final NR model) | 0.5176 | 0.6879 | 0.5907 | 0.6479 | 0.7907 | 0.7122|

Table 7: Ablation studies on the unsupervised test set. Comparison of Numerical Reasoning (NR) model variants using keyword based shallow matching of lexical candidates with numeric entities, pretrained contextualized embeddings and finetuned contextualized embeddings. The finetuned contextualized embeddings substantially outperform other two methods and is incorporated into the final NR model.

To evaluate the usage of contextualized embeddings with cosine similarity to connect lexical candidates with numeric entities as described in Section 3.4, we ablate the contextualized embeddings and instead we use keyword based shallow matching to connect lexical candidates with numeric entities. Table 7 shows that the ablated method results in significant performance drop, more precisely, in terms of
Fig. 3: UMAP visualization of pretrained and finetuned contextualized embeddings of numeric entities and their UMLS synonyms by pretrained and finetuned Clinical-BERT, respectively. Finetuning leads to better differentiation of numeric entities in the semantic space which helps the NR model to identify them with higher accuracy.

In Table 7, we also compare the difference between pretrained and finetuned contextualized embeddings. The pretrained embeddings are generated by the pretrained ClinicalBERT model without finetuning and the finetuned embeddings are generated after finetuning ClinicalBERT using Semantic Textual Similarity (STS) Equation 1 as mentioned in Section 3.3. As shown in Table 7, the pretrained contextualized embeddings perform poorly with a drop on exact Recall from 68.8% to 37.6% and F1 from 59.1% to 43.1% on unsupervised test set. For better interpretation, we visualize the pretrained and finetuned contextualized embeddings of numeric entities and their corresponding UMLS synonyms in Figure 3 by using Uniform Manifold Approximation andProjection (UMAP) dimensionality reduction [23]. We find that, by the pretrained contextualized embeddings most of the numeric entities are spread out unevenly in the space. For example, the data points for (general) cholesterol, low-density lipoprotein cholesterol, and high-density lipoprotein cholesterol are intermixed. On the other hand, the finetuned contextualized embeddings form well segregated clusters which means it is easier to predict a corresponding numeric entity of lexical candidates (connected with a number) using cosine similarity without collisions. Overall, it confirms that pretrained contextualized embeddings are not sufficient to connect lexical candidates with numeric entities effectively without the proposed learning objective for finetuning in Equation 1.
6 Conclusions and Future Works

Numerical reasoning is critical to capture critical phenotypes such as bedside measurement from clinical text. Current state-of-the-art phenotyping models are not designed to reason with numbers, and thus all of them perform poorly in detecting the phenotypes that require numerical reasoning. The proposed unsupervised model shows substantial gains over these models due to its explicit design to reason with numbers by leveraging external knowledge. The proposed model can be potentially generalized to other biomedical NLP tasks that require numerical reasoning from text. The model can be further extended to consider document level context and dynamic external knowledge base.

Acknowledgements

We would like to thank Dr. Garima Gupta, Dr. Deepa (M.R.S.H) and Dr. Ashok (M.S.) for helping us create gold-standard phenotype annotation data and validate the external knowledge for numerical reasoning.

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