# Analysis of selected articles

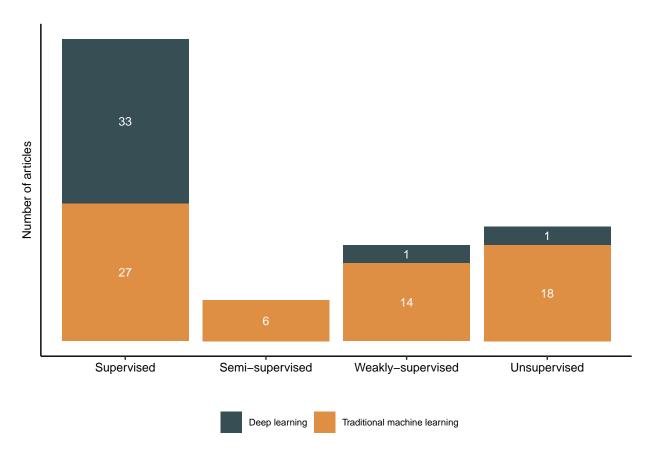
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## 1 Machine learning (ML) methods

#### 1.1 ML paradigms



#### 1.2 Traditional ML methods

Table 1: Common traditional machine learning methods (Count > 1)

| ML                | Traditional ML method         | Count |
|-------------------|-------------------------------|-------|
| Supervised        | Random forest                 | 14    |
| Supervised        | Logistic regression           | 11    |
| Supervised        | SVM                           | 11    |
| Supervised        | L1 logistic regression        | 8     |
| Supervised        | Decision trees                | 4     |
| Supervised        | XGBoost                       | 4     |
| Supervised        | Naive Bayes                   | 3     |
| Weakly-supervised | PheNorm                       | 3     |
| Weakly-supervised | MAP                           | 2     |
| Weakly-supervised | Random forest                 | 2     |
| Unsupervised      | LDA                           | 5     |
| Unsupervised      | K-means                       | 4     |
| Unsupervised      | UPGMA Hierarchical clustering | 2     |

## [1] "There are 18 papers using multiple traditional machine learning methods"

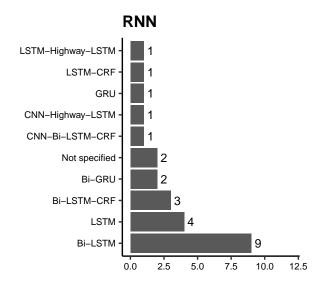
### $1.3\quad \text{Deep learning (DL) methods}$

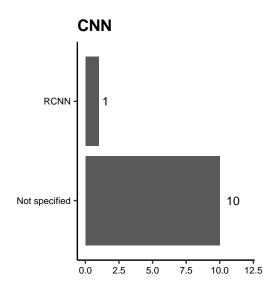
Table 2: Common deep learning methods (Count > 1)

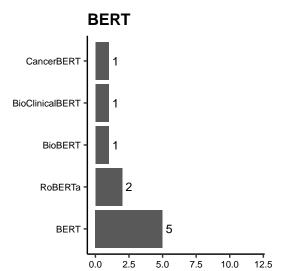
| DL method | ML         | Count |
|-----------|------------|-------|
| BERT      | Supervised | 7     |
| CNN       | Supervised | 11    |
| FFNN      | Supervised | 3     |
| RNN       | Supervised | 19    |

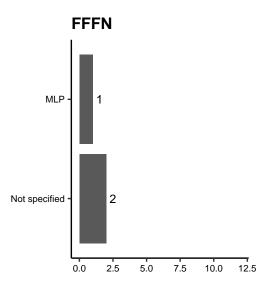
## [1] "There are 5 papers using multiple deep learning methods"

#### 1.3.1 Neural network variants



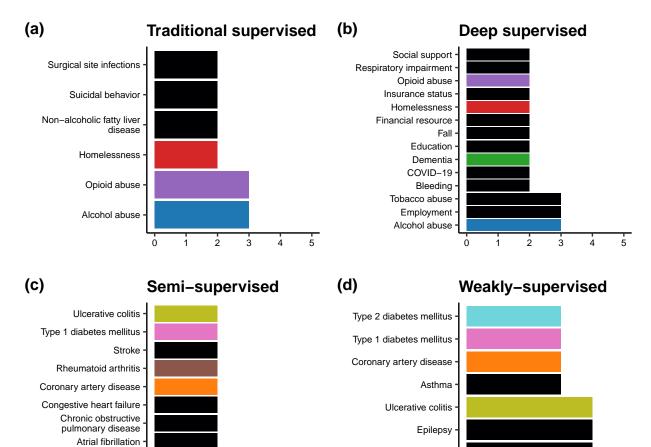






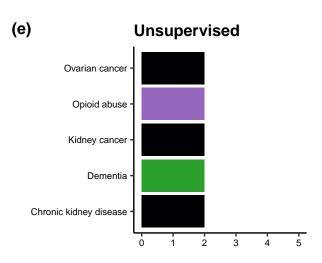
#### 2 Phenotypes

#### 2.1 Phenotypes considered across ML paradigms



Crohn's disease

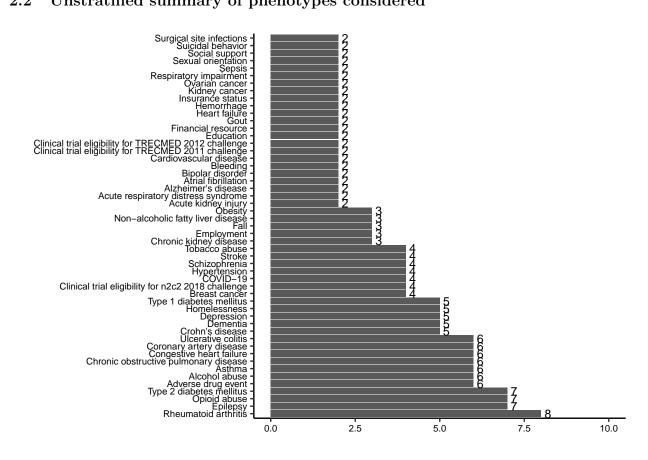
Rheumatoid arthritis



Alzheimer's disease

Type 2 diabetes mellitus

#### 2.2 Unstratified summary of phenotypes considered



#### 3 Data sources

#### 3.1 Use of structured and unstructured data

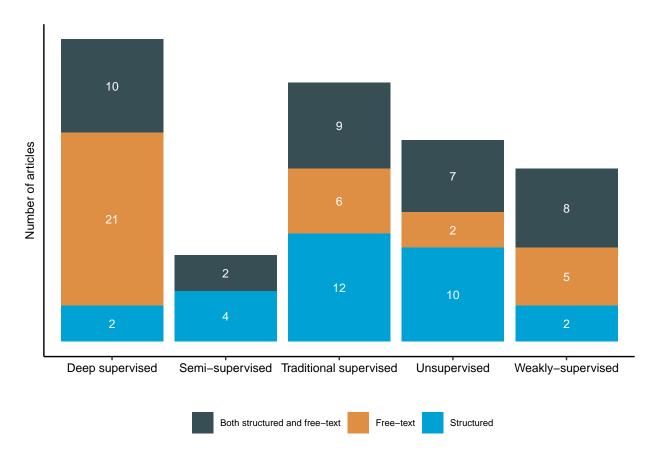
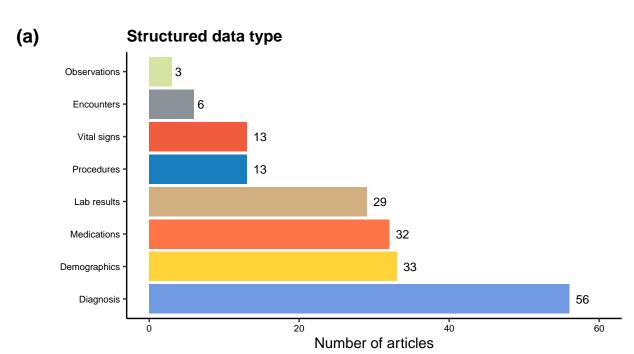


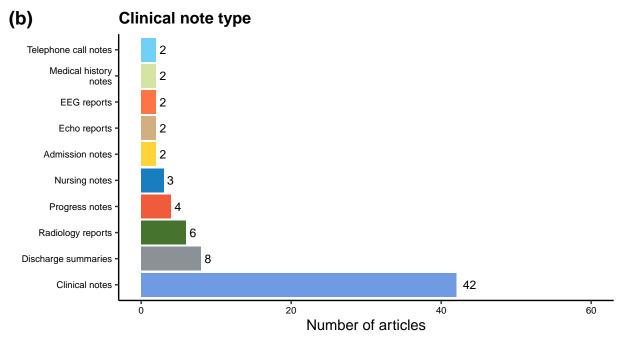
Table 3: Use of structured and unstructured data

| Data                          | Count |
|-------------------------------|-------|
| Both structured and free-text | 36    |
| Free-text                     | 34    |
| Structured                    | 30    |

#### 3.2 Structured and unstructured data types

- ## [1] "There are 50 papers using multiple structured data types"
- ## [1] "There are 13 papers using multiple unstructured data types"





| Terminology unnested                  | Supervised<br>Traditional<br>machine<br>learning | Unsupervised<br>Traditional<br>machine<br>learning | Supervised<br>Deep<br>learning | Weakly-<br>supervised<br>Traditional<br>machine<br>learning | Semi-<br>supervised<br>Traditional<br>machine<br>learning | Count |
|---------------------------------------|--|--|--------------------------------|---|---|-------|
| UMLS                                  | 11   | 3  | 8                              | 8   | 1   | 31    |
| ICD-9                                 | 6  | 5  | 4                              | 4   | 2   | 21    |
| ICD-9/10                              | 11   | 1  | 3                              | 0   | 2   | 17    |
| SNOMED-<br>CT                         | 2  | 3  | 4                              | 3   | 0   | 12    |
| RxNorm                                | 3  | 1  | 2                              | 2   | 1   | 9     |
| CPT                                   | 2  | 0  | 3                              | 2   | 0   | 7     |
| Phecode                               | 0  | 2  | 0                              | 3   | 2   | 7     |
| ICD                                   | 0  | 1  | 0                              | 4   | 0   | 5     |
| ICD-9-CM                              | 1  | 2  | 0                              | 1   | 0   | 4     |
| LOINC                                 | 3  | 0  | 0                              | 1   | 0   | 4     |
| ICD-10                                | 0  | 0  | 1                              | 1   | 1   | 3     |
| ATC (Anatomical therapeutic chemical) | 2  | 0  | 0                              | 0   | 0   | 2     |
| NDC<br>(National<br>drug<br>codes)    | 2  | 0  | 0                              | 0   | 0   | 2     |

### 3.3 Terminologies

## [1] "There are 37 papers using multiple terminologies"

| NLP<br>software                | Supervised<br>Deep<br>learning | Weakly-<br>supervised<br>Traditional<br>machine<br>learning | Supervised<br>Traditional<br>machine<br>learning | Semisupervised Traditional machine learning | Unsupervised Traditional machine learning | Count                  |
|--------------------------------|--------------------------------|---|--|---|---|------------------------|
| cTAKES NegEx NILE NLTK MetaMap | 8<br>0<br>0<br>4<br>1          | 0<br>2<br>5<br>0  | 8<br>3<br>1<br>0<br>3                            | 1<br>0<br>0<br>0<br>0                       | 2<br>1<br>0<br>1<br>0                     | 19<br>6<br>6<br>5<br>4 |
| Stanford<br>CoreNLP            | 2                              | 0   | 0  | 0   | 0   | 2                      |

#### 3.4 Natural language processing (NLP) software

## [1] "There are 7 papers using multiple NLP software"

#### 3.5 Embeddings

Embeddings were only used in deep supervised articles.

| Embedding training data       | Count |
|-------------------------------|-------|
| Unstructured EHR              | 11    |
| Biomedical literature         | 10    |
| MIMIC-III database (internal) | 7     |
| MIMIC-III database (external) | 6     |
| Wikipedia                     | 6     |
| Structured EHR                | 2     |

## [1] "There are 7 papers using multiple embedding training data"

| Embedding               | Count         |
|-------------------------|---------------|
| Word2vec                | 19            |
| GloVe<br>BERT           | 6<br>5        |
| RoBERTa                 | 3             |
| BioBERT BioClinicalBERT | $\frac{2}{2}$ |
| FastText                | $\frac{2}{2}$ |
| Not specified           | 2             |

## [1] "There are 11 papers using multiple embedding training methods"

### $3.6\quad {\rm Openly\text{-}available\ data}$

#### 3.6.1 Competition data

## [1] "There are 2 papers using multiple competition data"

| Competition data name          | Supervised<br>Traditional<br>machine<br>learning | Supervised<br>Deep<br>learning | Count |
|--------------------------------|--|--------------------------------|-------|
| 2018 n2c2<br>track 2           | 0  | 6                              | 6     |
| 2018 n2c2<br>track 1           | 1  | 3                              | 4     |
| TRECMED 2011                   | 1  | 1                              | 2     |
| TRECMED 2012                   | 1  | 1                              | 2     |
| 2008 i2b2                      | 1  | 0                              | 1     |
| 2012<br>physionet<br>Challenge | 0  | 1                              | 1     |

| Data<br>source        | Supervised<br>Deep<br>learning | Supervised<br>Traditional<br>machine<br>learning | Weakly-<br>supervised<br>Deep<br>learning | Weakly-<br>supervised<br>Traditional<br>machine<br>learning | Unsupervised Traditional machine learning | Count |
|-----------------------|--------------------------------|--|---|---|---|-------|
| MIMIC-III<br>database | 9                              | 1  | 1   | 1   | 3   | 15    |
| MTSamples<br>database | 1                              | 0  | 0   | 0   | 0   | 1     |

#### 3.6.2 Other publicly available data sources

#### 3.7 Private data sources and demographics reporting

## [1] "71 articles did not use openly available data"

## [1] "Among these 71 articles, 38 reported demographics"

#### 3.8 Institutions

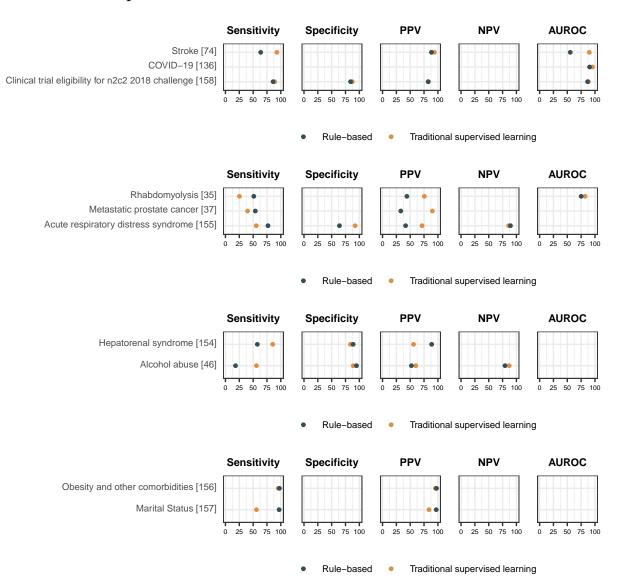
| Country    | Count |
|------------|-------|
| US         | 94    |
| France     | 2     |
| Canada     | 1     |
| China      | 1     |
| Germany    | 1     |
| Israel     | 1     |
| Italy      | 1     |
| Korean     | 1     |
| Netherland | 1     |
| Singapore  | 1     |
| Spain      | 1     |

#### 3.9 Data sources summary across different ML paradigms

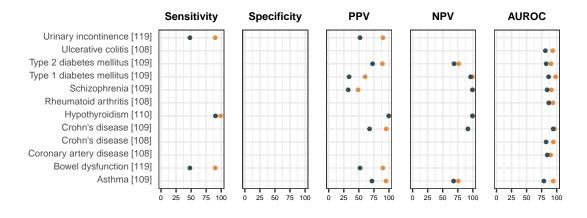
|       | Total<br>number<br>of papers | Used<br>free-text | Used<br>NLP<br>software | Used<br>competi-<br>tion data | Used<br>multisite<br>data | Used<br>open<br>data | Used<br>private<br>single-<br>site data | Compared<br>to rule-<br>based<br>algo-<br>rithms | Comapred<br>to tradi-<br>tional<br>ML | Reported patient demographic | Released<br>open<br>code |
|-------|------------------------------|-------------------|-------------------------|-------------------------------|---------------------------|----------------------|---|--|---------------------------------------|------------------------------|--------------------------|
| TSL   | 27                           | 15                | 14                      | 3                             | 1                         | 1                    | 22                                      | 10   | 0                                     | 13                           | 4                        |
| DSL   | 33                           | 31                | 18                      | 11                            | 1                         | 9                    | 12                                      | 2  | 20                                    | 5                            | 9                        |
| SSL   | 6                            | 2                 | 1                       | 0                             | 0                         | 0                    | 6                                       | 1  | 0                                     | 3                            | 0                        |
| WSL   | 15                           | 13                | 10                      | 0                             | 3                         | 2                    | 10                                      | 8  | 1                                     | 4                            | 3                        |
| USL   | 19                           | 9                 | 4                       | 0                             | 3                         | 3                    | 13                                      | 0  | 0                                     | 13                           | 4                        |
| Total | 100                          | 70                | 47                      | 14                            | 8                         | 15                   | 63                                      | 21   | 21                                    | 38                           | 20                       |

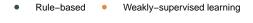
### 4 Reporting and evaluation

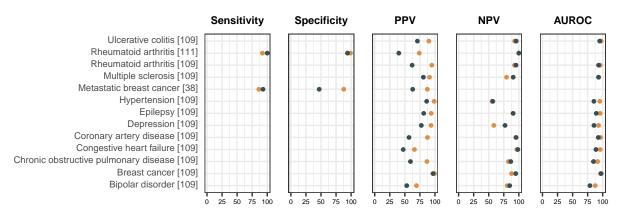
#### 4.1 Traditonal supervised ML vs. rule-based



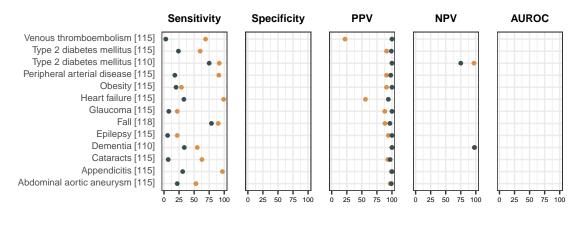
#### 4.2 Weakly-supervised ML vs. rule-based





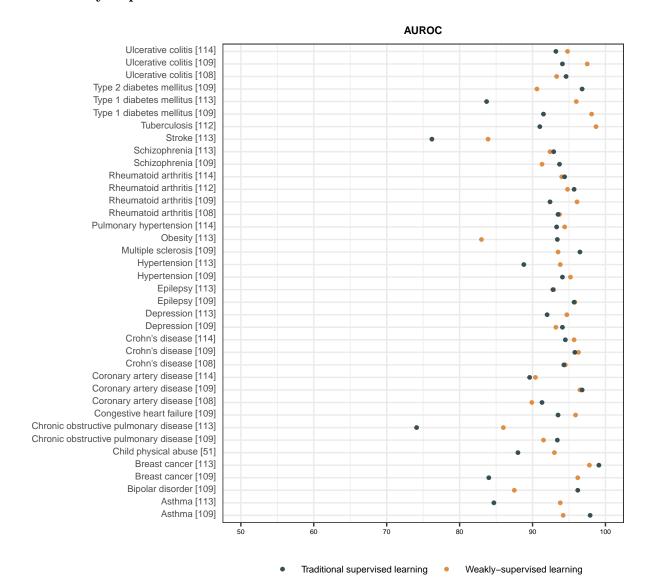




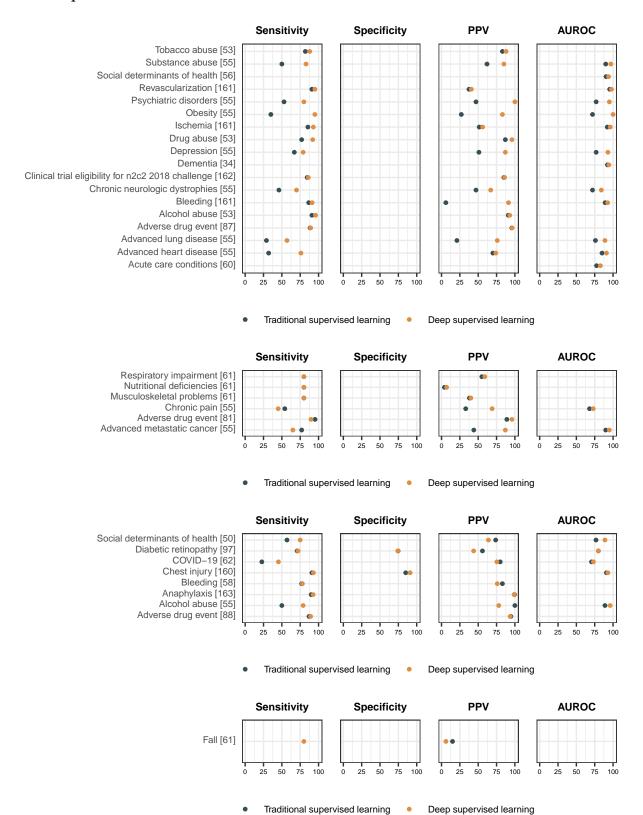


Rule-based
 Weakly-supervised learning

#### 4.3 Weakly-supervised ML vs. traditional



#### 4.4 Deep ML vs. traditional



| Model       | Supervised | Supervised | Weakly-    | Weakly-    | Semi-      | Count |
|-------------|------------|------------|------------|------------|------------|-------|
| perfor-     | Deep       | Tradi-     | supervised | supervised | supervised |       |
| mance       | learning   | tional     | Deep       | Tradi-     | Tradi-     |       |
| metrics     |            | machine    | learning   | tional     | tional     |       |
|             |            | learning   |            | machine    | machine    |       |
|             |            |            |            | learning   | learning   |       |
| Precision   | 26         | 23         | 0          | 8          | 4          | 61    |
| Recall      | 25         | 23         | 1          | 7          | 2          | 58    |
| AUROC       | 11         | 15         | 1          | 10         | 5          | 42    |
| F-score     | 26         | 9          | 0          | 7          | 0          | 42    |
| Specificity | 6          | 11         | 1          | 1          | 0          | 19    |
| Accuracy    | 4          | 8          | 1          | 4          | 0          | 17    |
| NPV         | 1          | 7          | 0          | 5          | 2          | 15    |
| AUPRC       | 4          | 2          | 0          | 2          | 0          | 8     |
| Calibration | 2          | 3          | 0          | 0          | 0          | 5     |
| plots       |            |            |            |            |            |       |
| Log loss    | 1          | 1          | 0          | 0          | 1          | 3     |
| Brier       | 1          | 1          | 0          | 0          | 0          | 2     |
| score       |            |            |            |            |            |       |
| Hamming     | 2          | 0          | 0          | 0          | 0          | 2     |
| loss        |            |            |            |            |            |       |
| Matthews    | 1          | 1          | 0          | 0          | 0          | 2     |
| Correla-    |            |            |            |            |            |       |
| tion        |            |            |            |            |            |       |
| Coeffi-     |            |            |            |            |            |       |
| cient       |            |            |            |            |            |       |
| Normalized  | 1          | 1          | 0          | 0          | 0          | 2     |
| dis-        |            |            |            |            |            |       |
| counted     |            |            |            |            |            |       |
| cumula-     |            |            |            |            |            |       |
| tive gain   |            |            |            |            |            |       |

# 5 Model performance metric reporting