A summary of EHR-based phenotyping article annotation

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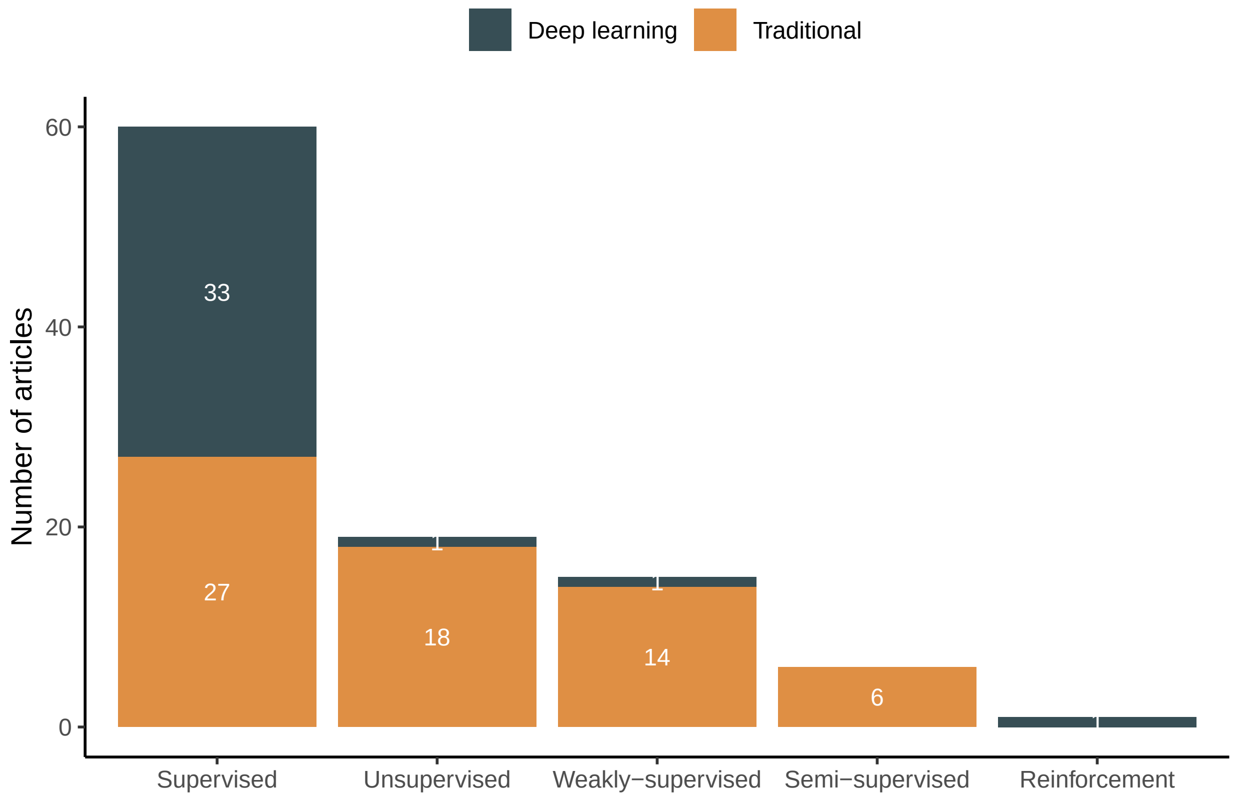
Contents

1 Overview



1.1 Traditional ML method

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
Unsupervised	LDA	5
Unsupervised	Hierarchical clustering	4
Unsupervised	K-means	4
Weakly-supervised	PheNorm	3
Weakly-supervised	MAP	2
Weakly-supervised	Random forest	2

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

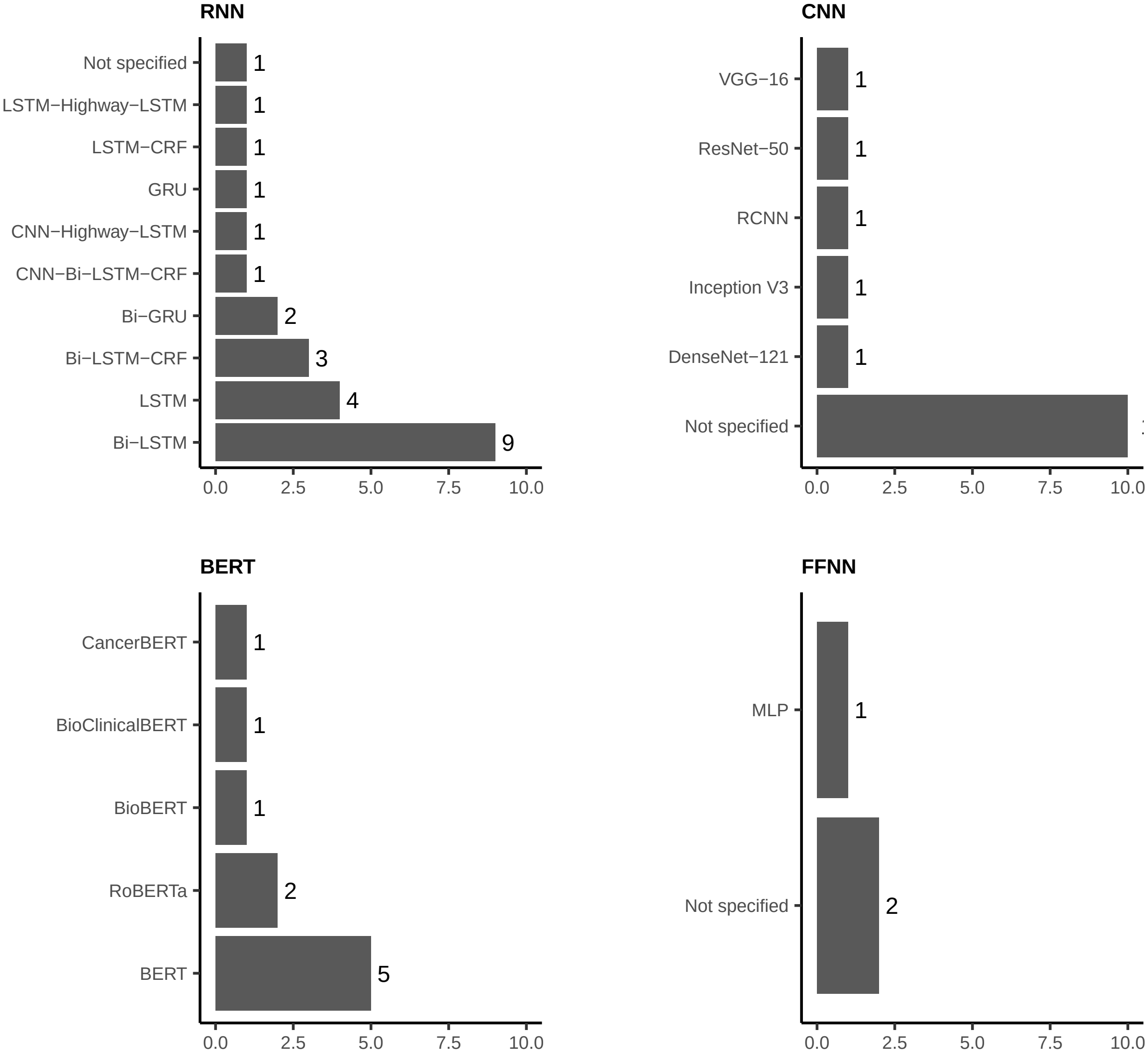
1.2 DL method

Table 2: Deep learning methods

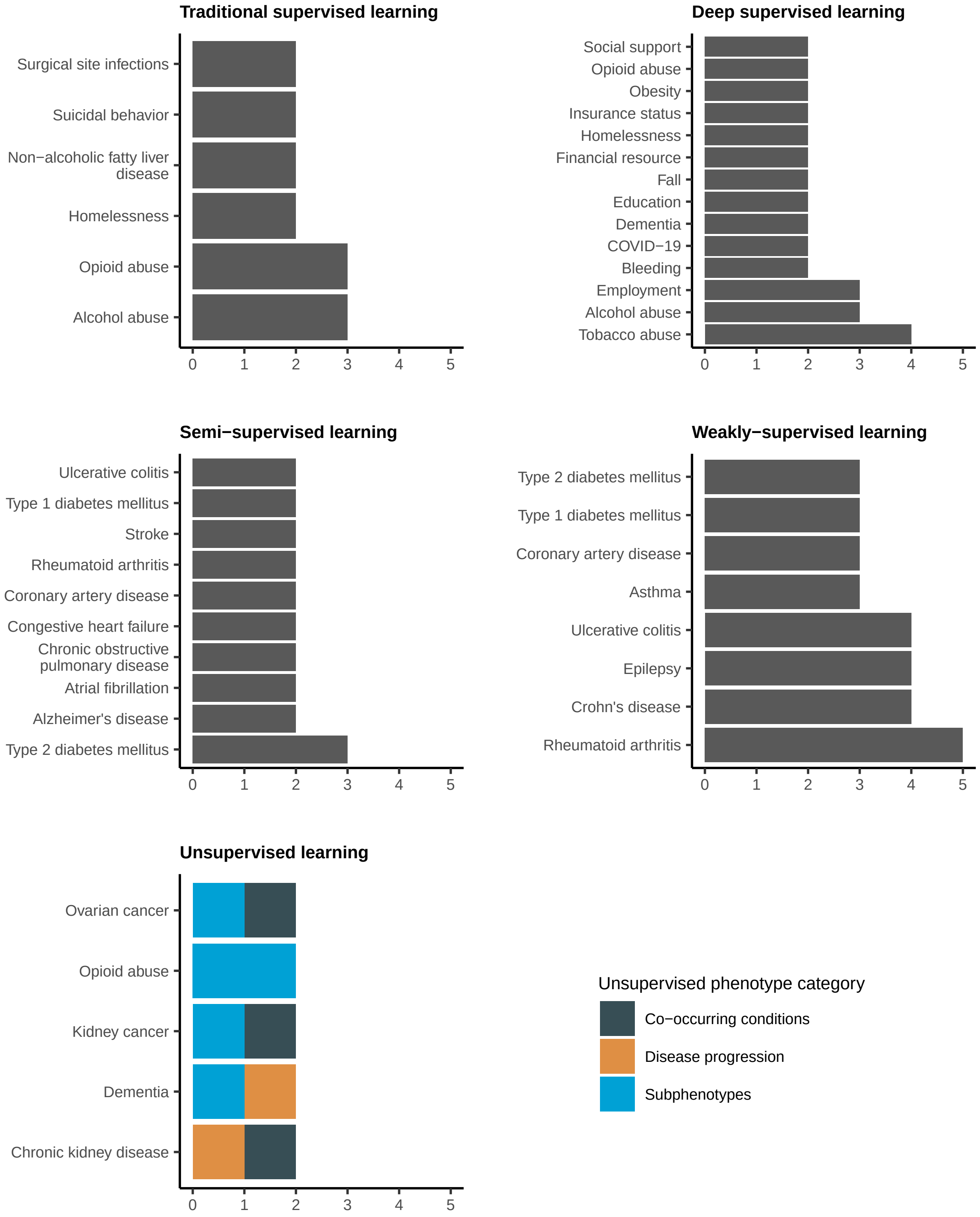
DL method	ML	Count
BERT	Supervised	7
CNN	Supervised	12
FFNN	Supervised	3
RNN	Supervised	18

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

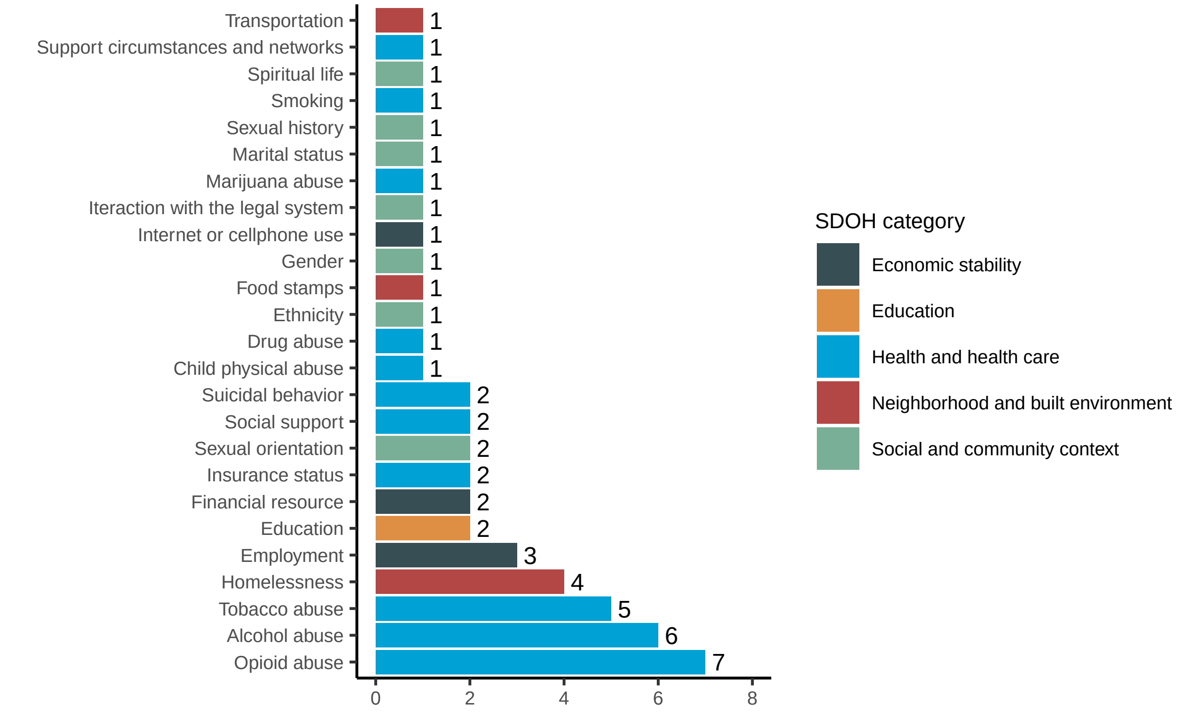
1.2.1 Deep neural network variants

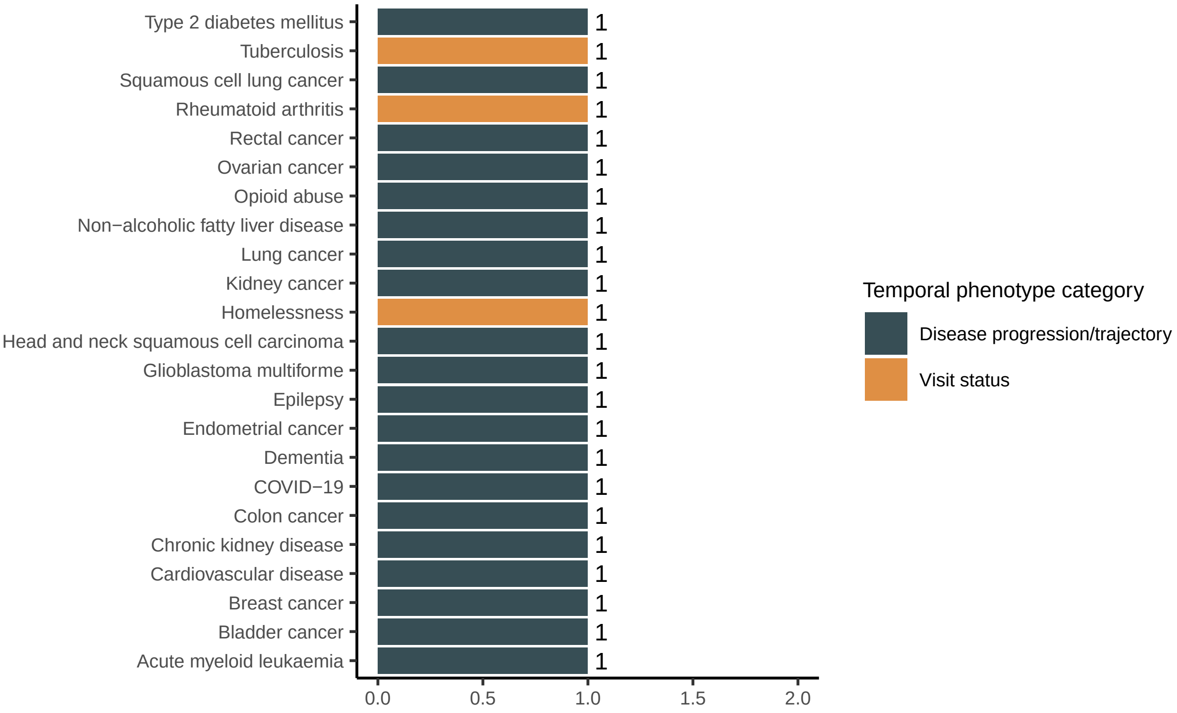


2 Phenotype


Traditional supervised learning
Surgical site infections
Suicidal behavior
Non−alcoholic fatty liver disease
Homelessness
Opioid abuse
Alcohol abuse
0
1
2
3
4
5
Deep supervised learning
Social support
Opioid abuse
Obesity
Insurance status
Homelessness
Financial resource
Fall
Education
Dementia
COVID−19
Bleeding
Employment
Alcohol abuse
Tobacco abuse
0
1
2
3
4
5
Semi−supervised learning
Ulcerative colitis
Type 1 diabetes mellitus
Stroke
Rheumatoid arthritis
Coronary artery disease
Congestive heart failure
Chronic obstructive pulmonary disease
Atrial fibrillation
Alzheimer's disease
Type 2 diabetes mellitus
0
1
2
3
4
5
Weakly−supervised learning
Type 2 diabetes mellitus
Type 1 diabetes mellitus
Coronary artery disease
Asthma
Ulcerative colitis
Epilepsy
Crohn's disease
Rheumatoid arthritis
0
1
2
3
4
5
Unsupervised learning
Ovarian cancer
Opioid abuse
Kidney cancer
Dementia
Chronic kidney disease
0
1
2
3
4
5
Unsupervised phenotype category
Co−occurring conditions
Disease progression
Subphenotypes

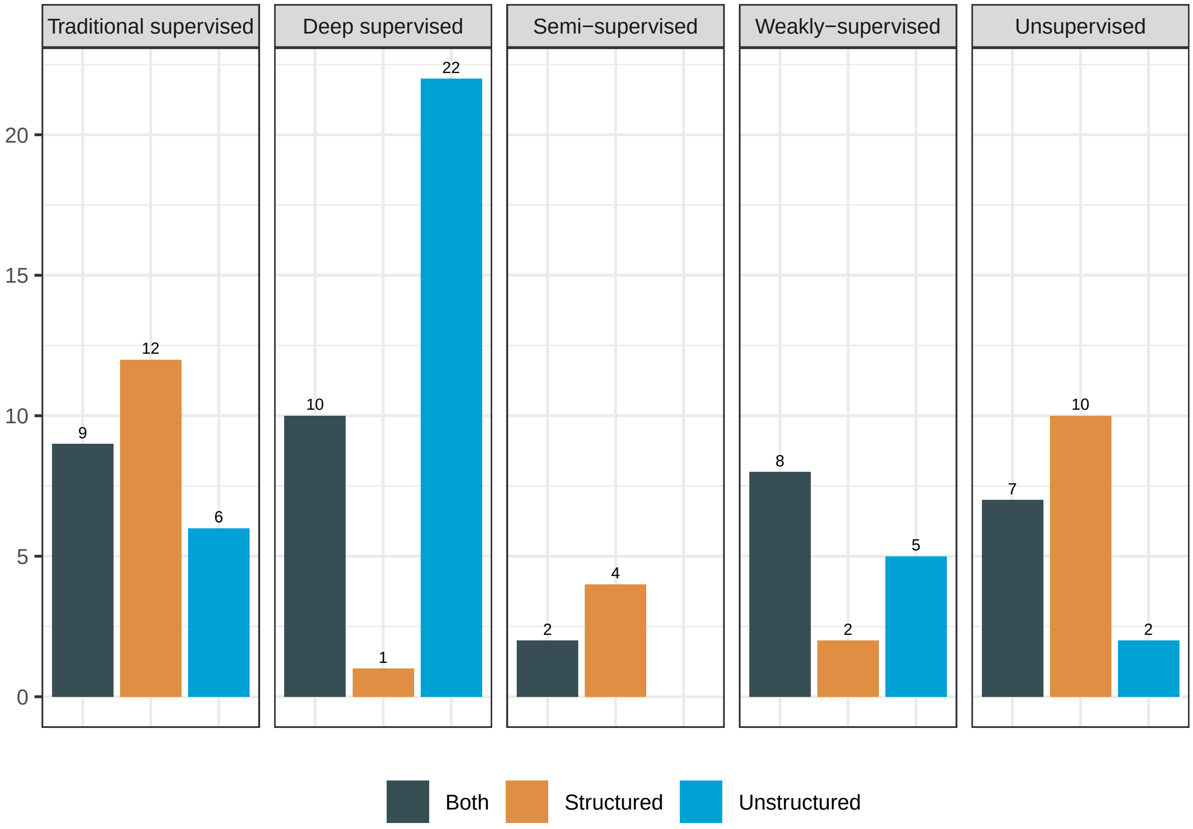
2.1 More nuanced phenotype





3 Data source

3.1 Summary

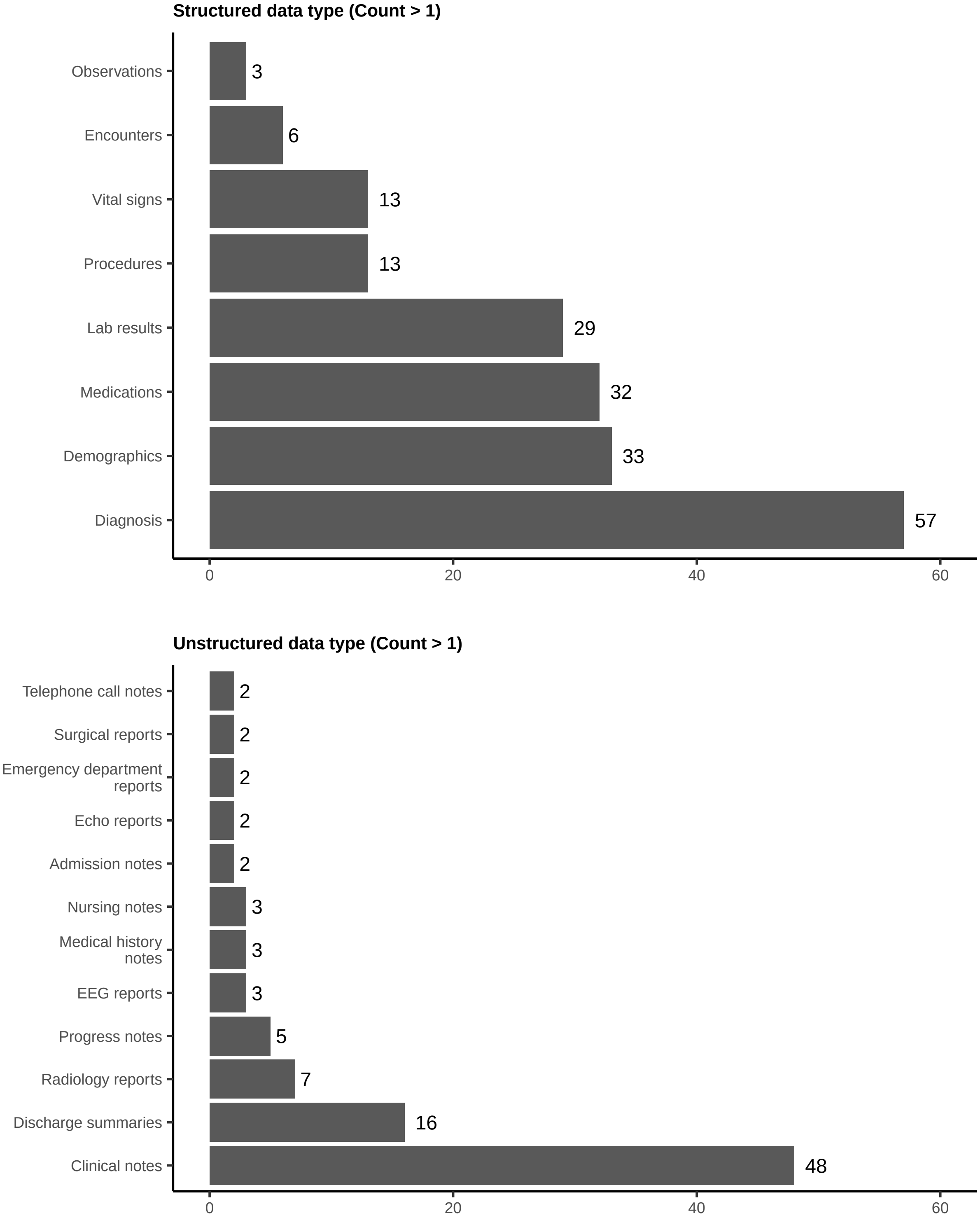


```
## [1] "There are 101 papers using machine learning models"
## [1] "There are 71 papers using machine learning models with unstructured data"
## [1] "There are 47 papers using machine learning models with NLP software"
## [1] "There are 14 papers using machine learning models with competition data"
## [1] "There are 18 papers using machine learning models with data from multiple sites"
## [1] "There are 29 papers using machine learning models with openly available data"
## [1] "There are 64 papers using machine learning models with data from private single site"
## [1] "-----------------------------"
## [1] "There are 20 papers machine learning models compared with rule-based algorithms"
## [1] "There are 21 papers machine learning models compared with traditional ML algorithms"
## [1] "-----------------------------"
## [1] "There are 45 papers reported machine learning models demographics"
## [1] "There are 20 papers released machine learning models source code"
```

3.2 Structured and unstructured data type

```
## [1] "There are 50 papers using multiple structured data type"
```

```
## [1] "There are 15 papers using multiple unstructured data type"
```


Structured data type (Count > 1)
Observations
3
Encounters
6
Vital signs
13
Procedures
13
Lab results
29
Medications
32
Demographics
33
Diagnosis
57
0
20
40
60
Unstructured data type (Count > 1)
Telephone call notes
2
Surgical reports
2
Emergency department reports
2
Echo reports
2
Admission notes
2
Nursing notes
3
Medical history notes
3
EEG reports
3
Progress notes
5
Radiology reports
7
Discharge summaries
16
Clinical notes
48
0
20
40
60

3.2.1 Traditional supervised learning

```
## [1] "There are 27 papers using traditional supervised learning"
## [1] "There are 15 papers using traditional supervised learning with unstructured data"
## [1] "There are 14 papers using traditional supervised learning with NLP software"
## [1] "There are 3 papers using traditional supervised learning with competition data"
## [1] "There are 2 papers using traditional supervised learning with data from multiple sites"
## [1] "There are 4 papers using traditional supervised learning with openly available data"
## [1] "There are 22 papers using traditional supervised learning with data from private single site"
## [1] "-----------------------------"
## [1] "There are 10 papers traditional supervised learning compared with rule-based algorithms"
## [1] "There are 0 papers traditional supervised learning compared with traditional ML algorithms"
## [1] "-----------------------------"
## [1] "There are 13 papers reported traditional supervised learning demographics"
## [1] "There are 4 papers released traditional supervised learning source code"
```

3.2.2 Deep supervised learning

```
## [1] "There are 33 papers using deep supervised learning"
## [1] "There are 32 papers using deep supervised learning with unstructured data"
## [1] "There are 18 papers using deep supervised learning with NLP software"
## [1] "There are 11 papers using deep supervised learning with competition data"
## [1] "There are 9 papers using deep supervised learning with data from multiple sites"
## [1] "There are 19 papers using deep supervised learning with openly available data"
## [1] "There are 13 papers using deep supervised learning with data from private single site"
## [1] "-----------------------------"
## [1] "There are 2 papers deep supervised learning compared with rule-based algorithms"
## [1] "There are 19 papers deep supervised learning compared with traditional ML algorithms"
## [1] "-----------------------------"
## [1] "There are 9 papers reported deep supervised learning demographics"
## [1] "There are 8 papers released deep supervised learning source code"
```

3.2.3 Semi-supervised learning

```
## [1] "There are 6 papers using semi-supervised learning"
## [1] "There are 2 papers using semi-supervised learning with unstructured data"
## [1] "There are 1 papers using semi-supervised learning with NLP software"
## [1] "There are 0 papers using semi-supervised learning with competition data"
## [1] "There are 0 papers using semi-supervised learning with data from multiple sites"
## [1] "There are 0 papers using semi-supervised learning with openly available data"
## [1] "There are 6 papers using semi-supervised learning with data from private single site"
## [1] "-----------------------------"
## [1] "There are 1 papers semi-supervised learning compared with rule-based algorithms"
## [1] "There are 0 papers semi-supervised learning compared with traditional ML algorithms"
## [1] "-----------------------------"
## [1] "There are 3 papers reported semi-supervised learning demographics"
## [1] "There are 0 papers released semi-supervised learning source code"
```

3.2.4 Weakly-supervised learning

```
## [1] "There are 15 papers using weakly-supervised learning"
## [1] "There are 13 papers using weakly-supervised learning with unstructured data"
## [1] "There are 10 papers using weakly-supervised learning with NLP software"
## [1] "There are 0 papers using weakly-supervised learning with competition data"
## [1] "There are 4 papers using weakly-supervised learning with data from multiple sites"
## [1] "There are 2 papers using weakly-supervised learning with openly available data"
## [1] "There are 10 papers using weakly-supervised learning with data from private single site"
## [1] "----------------------------"
## [1] "There are 7 papers weakly-supervised learning compared with rule-based algorithms"
## [1] "There are 1 papers weakly-supervised learning compared with traditional ML algorithms"
## [1] "----------------------------"
## [1] "There are 4 papers reported weakly-supervised learning demographics"
## [1] "There are 3 papers released weakly-supervised learning source code"
```

3.2.5 Unsupervised learning

```
## [1] "There are 19 papers using unsupervised learning"
## [1] "There are 9 papers using unsupervised learning with unstructured data"
## [1] "There are 4 papers using unsupervised learning with NLP software"
## [1] "There are 0 papers using unsupervised learning with competition data"
## [1] "There are 3 papers using unsupervised learning with data from multiple sites"
## [1] "There are 3 papers using unsupervised learning with openly available data"
## [1] "There are 13 papers using unsupervised learning with data from private single site"
## [1] "----------------------------"
## [1] "There are 0 papers unsupervised learning compared with rule-based algorithms"
## [1] "There are 0 papers unsupervised learning compared with traditional ML algorithms"
## [1] "----------------------------"
## [1] "There are 15 papers reported unsupervised learning demographics"
## [1] "There are 4 papers released unsupervised learning source code"
```

3.3 Openly-available data

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

Competition data name	Supervised Traditional	Supervised Deep learning	Count
2018 n2c2 track 1	1	3	4
2018 n2c2 track 2	0	6	6
TRECMED 2011	1	1	2
TRECMED 2012	1	1	2

Data source	Count
MIMIC-III database	21
MTSamples database	1

```
## [1] "There are 1 papers using multiple Openly data"
```

Data source	Reinforcement Deep learning	Supervised Deep learning	Supervised Traditional	Unsupervised Traditional	Weakly-supervised Deep learning	Weakly-supervised Traditional	Count
MIMIC-III database	1	14	1	3	1	1	21

4 NLP software

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

NLP software	Supervised Deep learning	Weakly-supervised Traditional	Semi-supervised Traditional	Supervised Traditional	Unsupervised Traditional	Count
cTAKES	8	0	1	8	2	19
MetaMap	1	0	0	3	0	4
NegEx	0	2	0	3	1	6
NILE	0	5	0	1	0	6
NLTK	4	0	0	0	1	5
Stanford CoreNLP	2	0	0	0	0	2

5 Emebddings

Embeddings were only used in deep supervised articles.

Embedding training data	Count
Unstructured EHR	13
MIMIC-III database	12
Biomedical literature	10
Wikipedia	6
Structured EHR	2

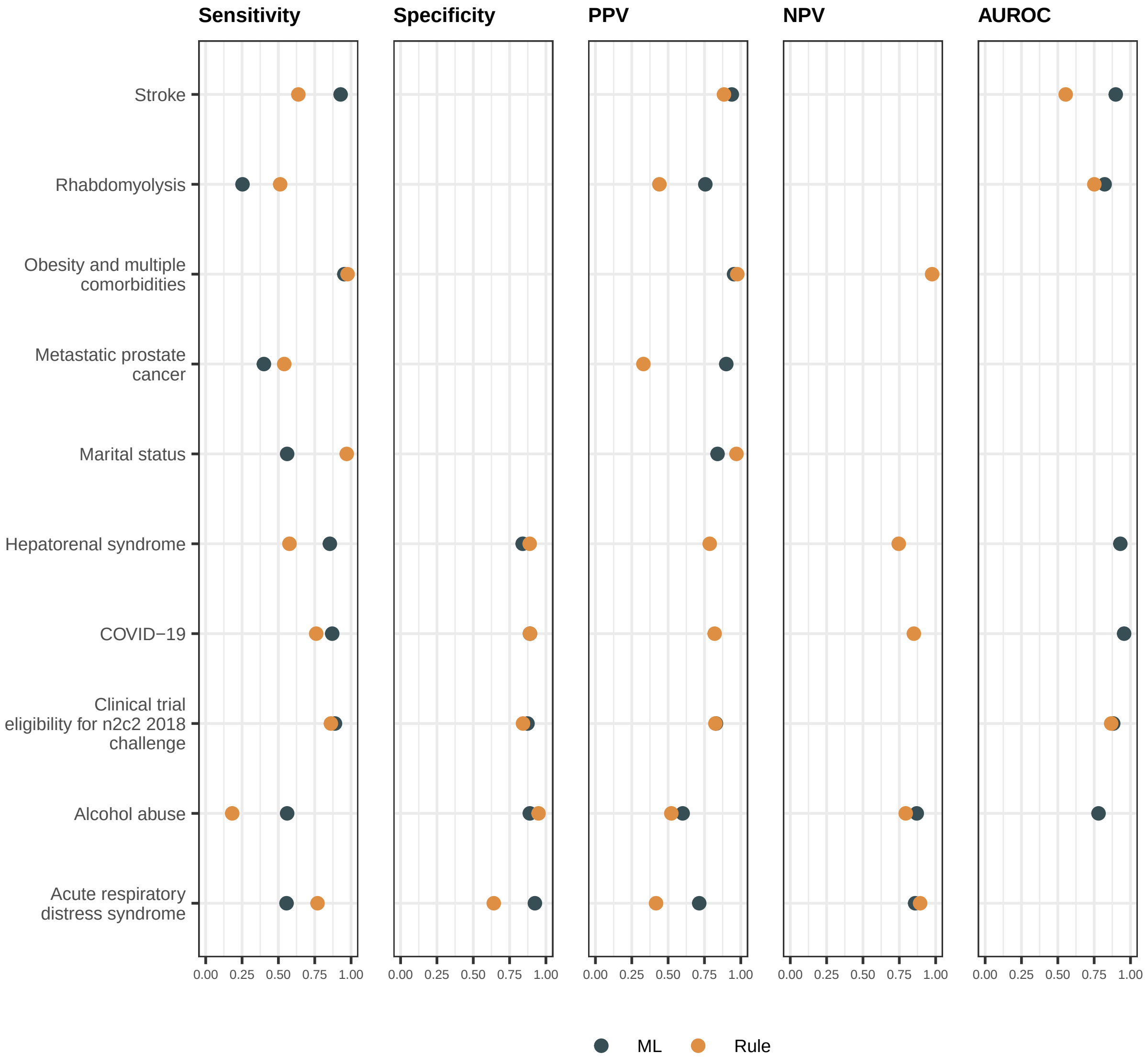
```
## [1] "There are 7 papers using multiple embedding training data"
```

Embedding	Count
Word2vec	19
GloVe	6
BERT	5
RoBERTa	3
BioBERT	2
BioClinicalBERT	2
FastText	2
Not specified	2

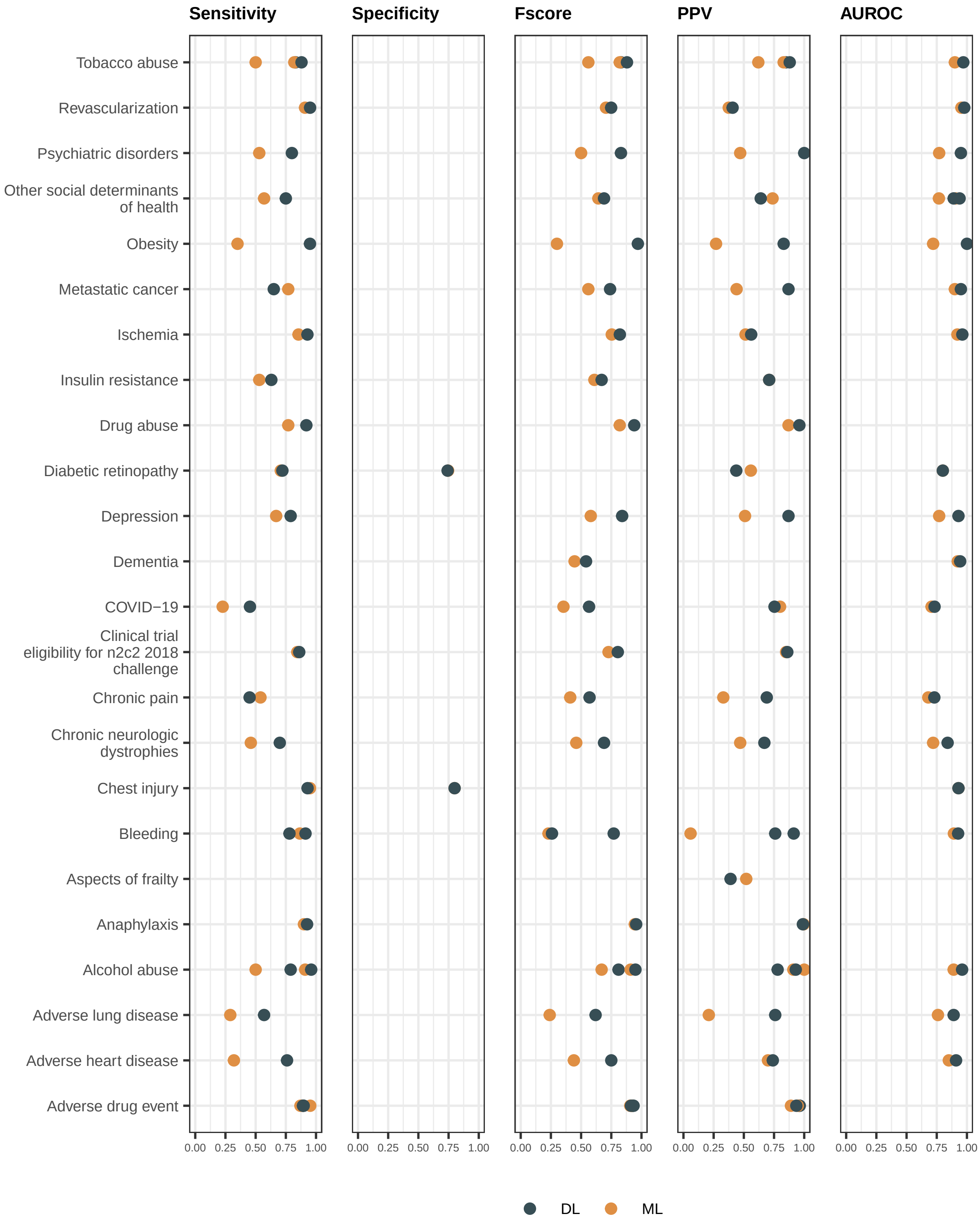
```
## [1] "There are 11 papers using multiple embedding training methods"
```

6 Validation and comparison

6.1 Traditonal supervised ML vs. rule-based



6.2 Deep supervised ML vs. supervised ML



Model performance metrics	Supervised Deep learning	Supervised Traditional	Weakly-supervised Deep learning	Weakly-supervised Traditional	Reinforcement Deep learning	Unsupervised Traditional	Semi-supervised Traditional	Count
Accuracy	5	8	1	4	0	0	0	18
AUPRC	3	2	0	2	1	1	0	9
AUROC	10	15	1	10	1	0	5	42
F-score	26	9	0	7	0	0	0	42
NPV	1	7	0	5	0	0	2	15
Precision	26	23	0	8	0	0	4	61
Recall	26	23	1	7	0	0	2	59
Specificity	7	11	1	1	0	0	0	20

7 Reporting

```
## There are 45 papers reported demographcis, 0.4455
```

```
## There are 20 papers reported demographcis, 0.198
```