A summary of EHR-based phenotyping article annotation

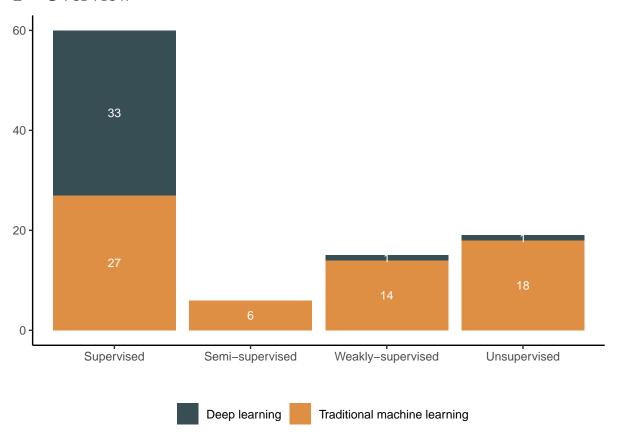
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1 Overview



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
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"

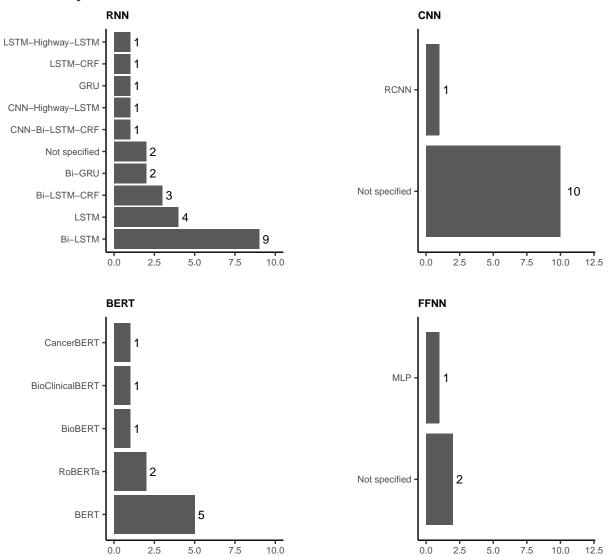
1.2 DL method

Table 2: Deep supervised learning methods

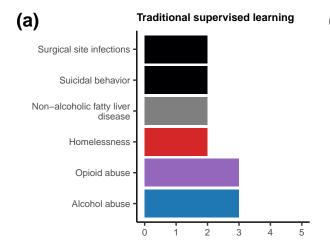
DL method	ML	Count
BERT	Supervised	7
CNN	Supervised	11
FFNN	Supervised	3
RNN	Supervised	19

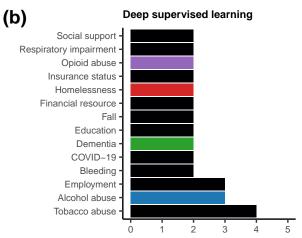
[1] "There are 5 papers using multiple deep learning methods"

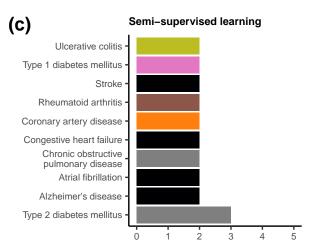
1.2.1 Deep neural network variants

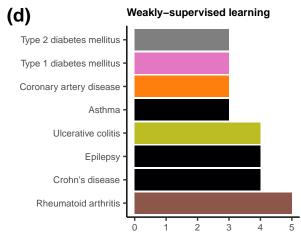


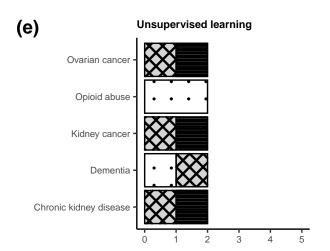
2 Phenotype











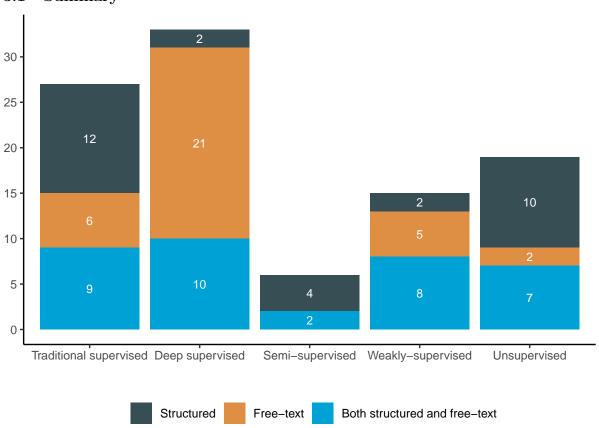
Unsupervised phenotype category Co–occurring conditions Disease progression Subphenotypes

2.1 More nuanced phenotype

	Total number of papers	Used free-text	Used NLP software	Used competi- tion data	Used multisite data	Used open data	Used private single- site data	Compared to rule- based algo- rithms	Comapred to tradi- tional ML	Reported patient demographic	Released open code
TSL	27	15	14	3	2	4	22	10	0	13	4
DSL	33	31	18	11	9	20	12	2	20	9	9
SSL	6	2	1	0	0	0	6	1	0	3	0
WSL	15	13	10	0	4	2	10	8	1	4	3
USL	19	9	4	0	3	3	13	0	0	15	4
Total	100	70	47	14	18	29	63	21	21	44	20

3 Data source

3.1 Summary

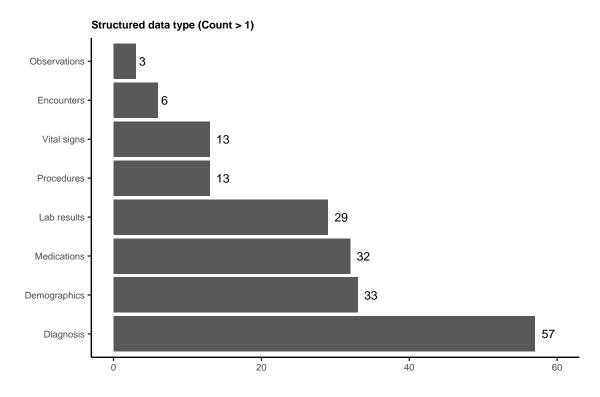


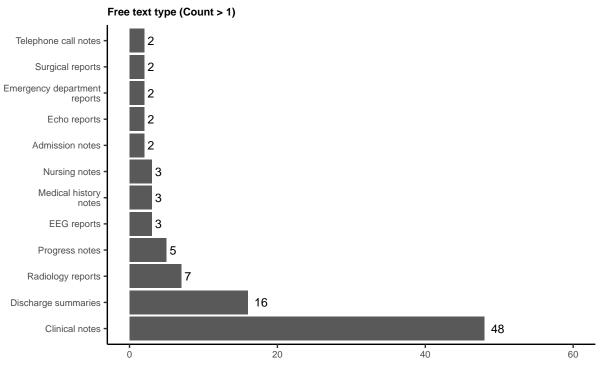
TSL = Traditional supervised learning. DSL = Deep supervised learning. DRL = Reinforcement deep learning. SSL = Semi-supervised learning. WSL = Weakly-supervised learning. US = Unsupervised learning.

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"





Data source	Supervised Deep learning	Supervised Traditional machine	Weakly- supervised Deep	Weakly- supervised Traditional	Unsupervised Traditional machine	Count
	icarining	learning	learning	machine	learning	
		learning	learning	learning	learning	
MIMIC-III database	15	1	1	1	3	21
MTSamples database	1	0	0	0	0	1

3.3 Openly-available data

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

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

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

4 NLP software

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

5 Emebddings

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

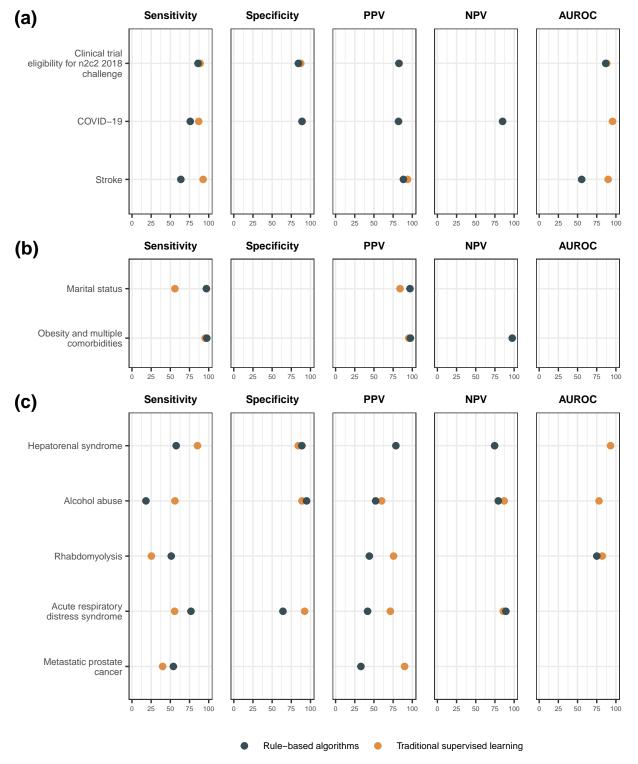
[1] "There are 7 papers using multiple embedding training data"

Embedding	Count
Word2vec	19
GloVe	6
BERT	5
RoBERTa	3
BioBERT	2
${\bf BioClinical BERT}$	2
FastText	2
Not specified	2

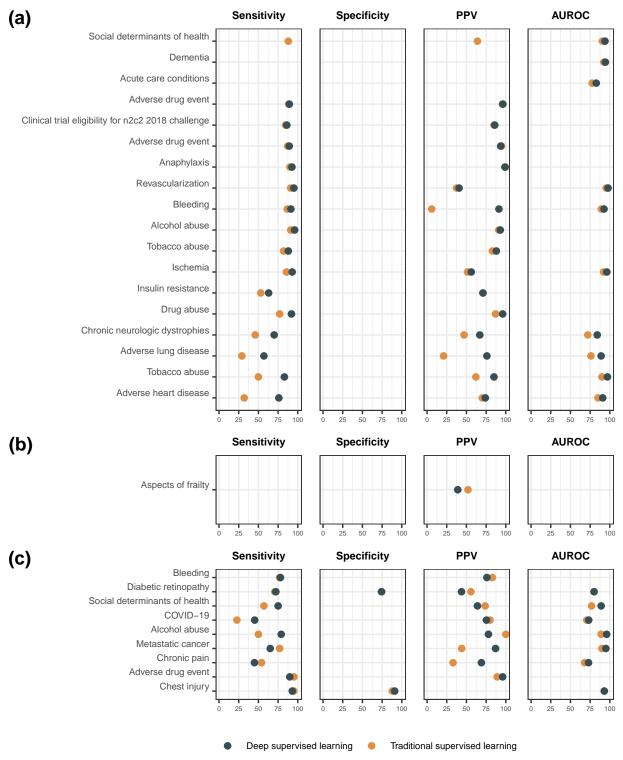
[1] "There are 11 papers using multiple embedding training methods"

6 Validation and comparison

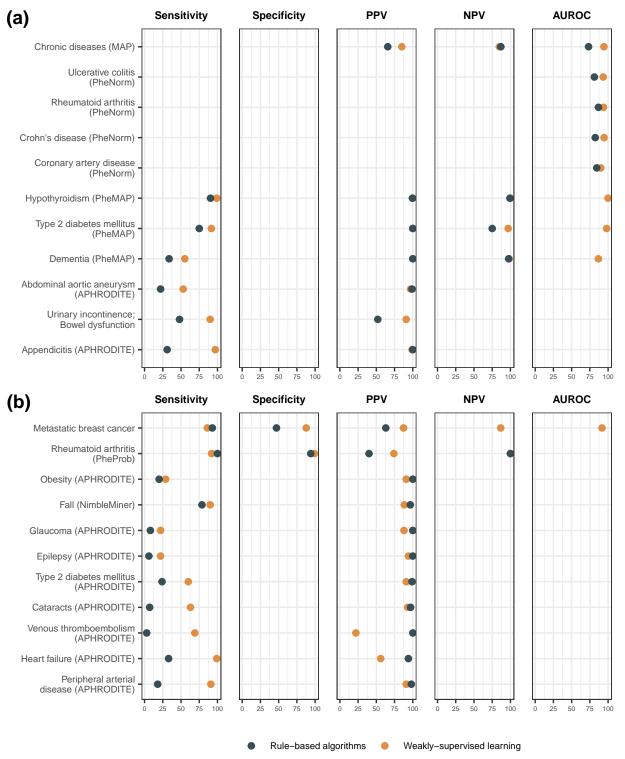
6.1 Traditonal supervised ML vs. rule-based



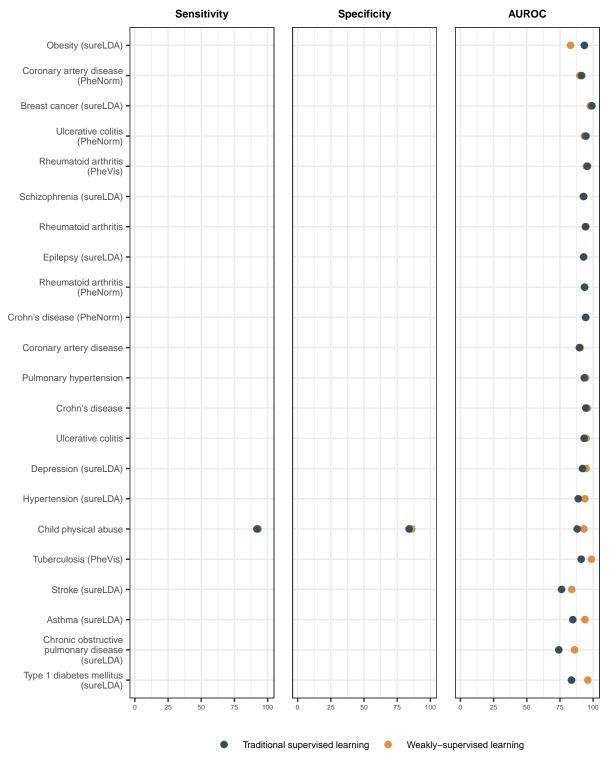
6.2 Deep supervised ML vs. traditional supervised ML



6.3 Weakly-supervised ML vs. rule-based algorithms



6.4 Weakly-supervised ML vs. traditional supervised ML



7 Model performance metric reporting

Model per-	Supervised	Supervised	Weakly-	Weakly-	Semi-	Count
formance	Deep	Tradi-	supervised	supervised	supervised	
metrics	learning	tional	Deep	Tradi-	Tradi-	
		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 score	1	1	0	0	0	2
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						