Ontologies and text mining

Textual components of ontologies

Classes and Relations

Descriptions

$C \sqsubseteq D$

$A \sqsubseteq \exists R.(D \sqcup E)$

$\neg C \sqsubseteq \forall R.A$

Complex Axioms

Textual components of ontologies

Figure from the Human Phenotype Ontology website

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Learning objectives

- Learn applications of the textual component of ontologies
- Learn text mining basics
- Get familiarized with popular text mining methods and their applications

Popular applications

- **Retrieve information from literature in text form**
- **Generate annotations**
- Generate cross references: Manually/Automatically
- Align/expand ontologies

The most common cause of hereditary breast cancer is an inherited mutation in the BRCA1

Popular applications

- Retrieve information from literature in text form
- Generate annotations
- **Generate cross references: Manually/Automatically**
- **Align/expand ontologies**

<div>

Family ontology

Parent
Mother
Father
Brother
Sister

</div>

<div>

Kinship ontology

Parenthood
Brotherhood
Sisterhood

</div>

How to compare/find AND link text?

- Exactly?
  - Brain tumor
  - Brain tumour
- Approximately?
  - How?

Exact match

- Dictionaries

Human Phenotype Ontology
Breast cancer → HP:003002 Breast Carcinoma → HP:003002

Disease Ontology
Breast cancer → DOID:1612 breast tumor → DOID:1612 malignant neoplasm of breast → DOID:1612 malignant tumor of breast→ DOID:1612

Exact match

- Dictionaries
- Example of applications:
  - **Mapping entities from different sources**

Human Phenotype Ontology
Breast cancer → HP:003002 Breast Carcinoma → HP:003002

Disease Ontology
Breast cancer → DOID:1612 breast tumor → DOID:1612 malignant neoplasm of breast → DOID:1612 malignant tumor of breast→ DOID:1612

Exact match

- Dictionaries
- Example of applications:
  - Mapping entities from different sources
  - **Finding mentions in literature and their co-occurings**

Human Phenotype Ontology
Breast cancer → HP:003002 Breast Carcinoma → HP:003002

Breast cancer is more prevalent in females, however, males can also develop breast cancer.

Birth control can increase risk of breast cancer in females.

Any ideas?

Brain tumor was found in a patient.

There are any forms of brain tumour.

Approximate comparison of text

- Exclude unimportant information

Carcinoma of the breast

Carcinoma breast

Approximate comparison of text

- Exclude unimportant information
- Stemming
    - Remove affixes
        - Cancers → cancer
        - Hyperpigmentation → hyperpig
    - Different stems
    - Novel words cannot be stemmed

Approximate comparison of text

- Exclude unimportant information
- Stemming
  - Remove affixes
    - Cancers → cancer
    - Hyperpigmentation → hyperpig
  - Different stems
  - Novel words cannot be stemmed
- Numerical representation
  - Numerical methods
  - How?

Numerical representation and analysis of text

Popular methods:

- Word2Vec
- BERT

Both methods use **embeddings** to represent text

What is an embedding?

- An embedding is a representation of a structure in a different space that **preserves** properties of that structure
- This is done by an embedding function *f*
- *f* preserves some property (structure-preserving)

Why embeddings?

- Perform functions on instances that were not possible in their original form
- Represent instances in a compact dimension
- Visualize instances and their relations
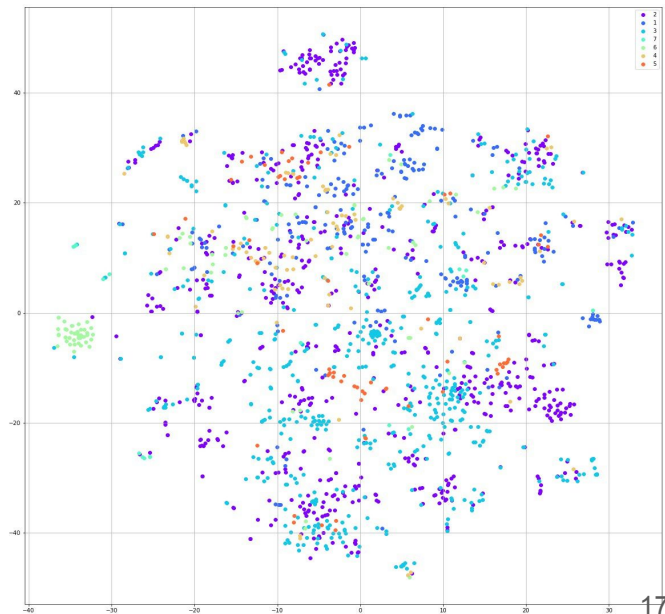
How are embeddings created for text?

- How is text represented numerically?
- Units of text are assigned IDs to create a vocabulary
  - Characters (letters)
  - Words
  - N-grams
  - Sentences
- How can we make this useful for comparison?

How are embeddings created for text?

- How is text represented numerically?
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  - **Characters (letters)**
  - Words
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  - Sentences
- How can we make this useful for comparison?

be bad → 250214

	0
a	1
b	2
c	3
d	4
e	5
f	6
g	7
h	8
i	9

How are embeddings created for text?

- How is text represented numerically?
- Units of text are assigned IDs to create a vocabulary
  - Characters (letters)
  - **Words**
  - N-grams
  - Sentences
- How can we make this useful for comparison?

be bad → 102

	0
be	1
bad	2
good	3
well	4

Word2Vec

- Well-known method
- Generates embeddings that capture co-occurrences based on a corpus
- Embeddings are in the form of *n*-dimensional vectors

Breast cancer is more prevalent in females, however, males can also develop breast cancer.

Birth control can increase risk of breast cancer in females.

Words → embeddings

Breast cancer is more prevalent in females, however, males can also develop breast cancer.
...
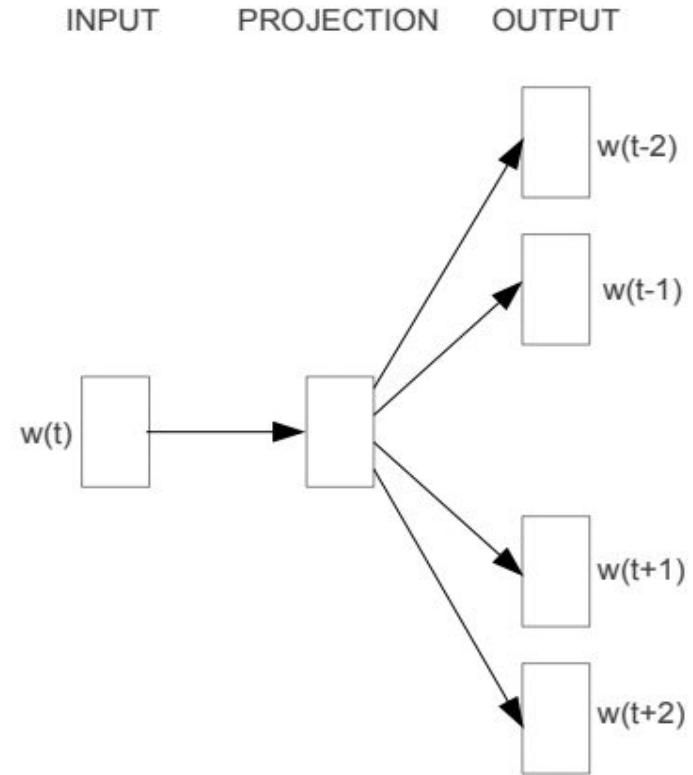Birth control can increase risk of breast cancer in females.

Word2Vec

```
[0.50929456, 0.6771953 , 0.91371871, 0.48265797, 0.18390237]
[0.9146623 , 0.7340195 , 0.78049964, 0.54384624, 0.01162719]
[0.22451245, 0.97085067, 0.79003223, 0.74382914, 0.26143969]
[0.11487895, 0.43190008, 0.86119749, 0.96533036, 0.56099287]
[0.77668599, 0.52129723, 0.71529702, 0.82580858, 0.40596435]
```

Word2Vec

Word2Vec captures co-occurrences

Given a word:

- Capture the words it frequently co-occurred within the given corpus

w(t)

w(t-2)

w(t-1)

w(t+1)

w(t+2)

Skip-gram

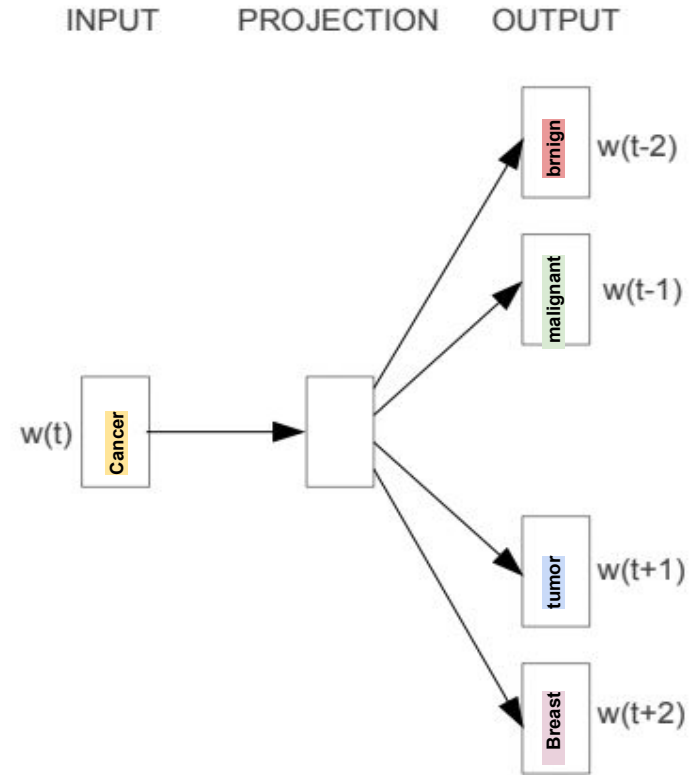Figure from the original paper: Efficient Estimation of Word Representations in Vector Space, Mikolov et al.

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Word2Vec

Word2Vec captures co-occurrences

Given a word:

- Capture the words it frequently co-occurred within the given corpus
- Minimize the cross-entropy loss

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Word2Vec

Many forms of cancer are not malignant but benign.

A breast tumor can be benign.

Breast cancer can be malignant.

	cancer	tumor	benign	malignant
cancer	0	0	1	2

Word2Vec

Many forms of cancer are not malignant but benign.

A breast tumor can be benign.

Breast cancer can be malignant.

	cancer	tumor	benign	malignant
tumor	0	0	1	0

Word2Vec

You can think of this of it as a factorization of a point-wise Mutual Information (PMI) matrix

	cancer	tumor	benign	malignant
cancer	0	0	1	2
tumor	0	0	1	0
benign	1	1	0	1
malignant	2	0	1	0

Word2Vec

You can think of this of it as a factorization of a point-wise Mutual Information (PMI) matrix

$$\text{pmi}(x; y) \equiv \log \frac{p(x, y)}{p(x)p(y)}$$

|	cancer	tumor	benign	malignant
cancer	0	0	1	2
tumor	0	0	1	0
benign	1	1	0	1
malignant	2	0	1	0

Word2Vec

You can think of this of it as a factorization of a Point-wise Mutual Information (PMI) matrix

	Cancer	Benign	Malignant	Tumor	Breast
Cancer	0	2	1	1	4
Benign	2	0	1	1	2
Malignant	1	1	0	2	2
Tumor	1	1	2	0	3
Breast	4	2	2	3	0

$$\text{pmi}(x; y) \equiv \log \frac{p(x, y)}{p(x)p(y)}$$

Word2Vec

You can this of it as a factorization of a
Pointwise Mutual Information (PMI) matrix

Neural Word Embedding as Implicit Matrix Factorization by Levy Omer and Goldberg Yoav
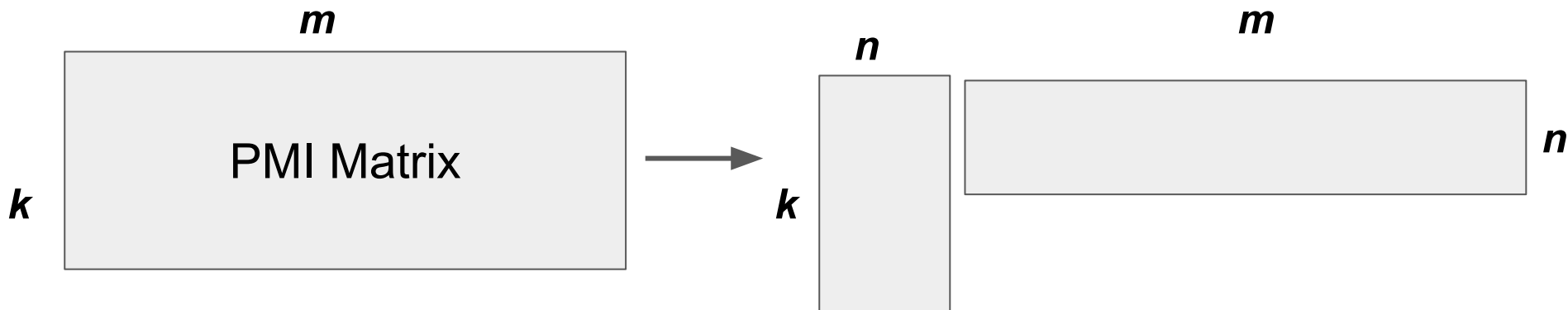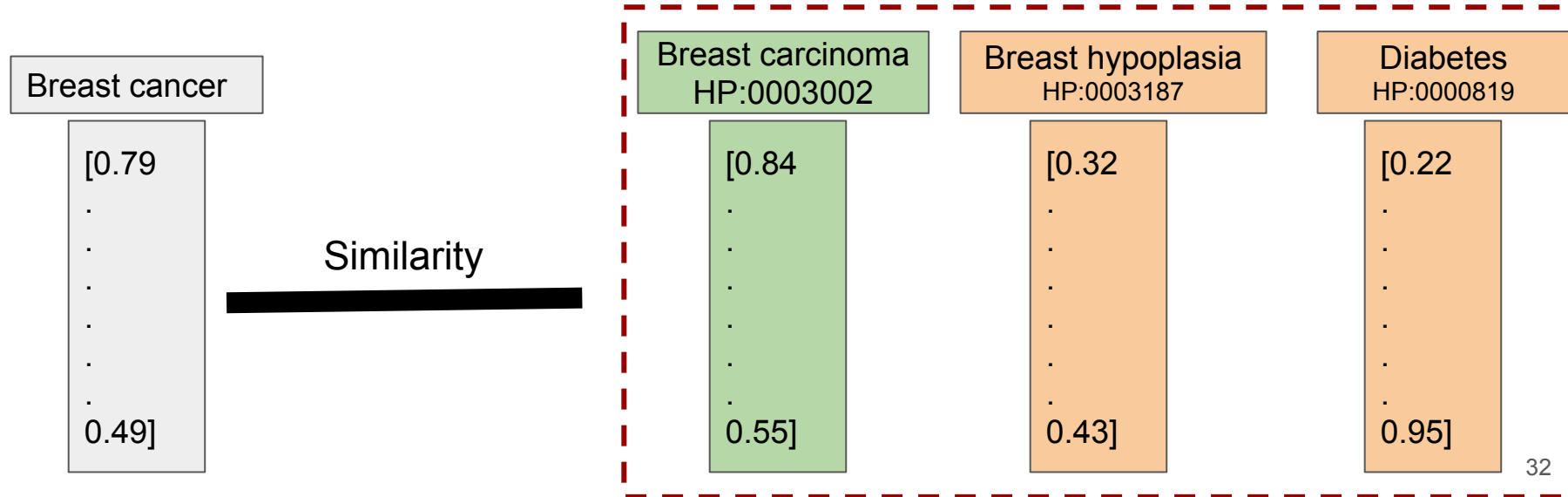
Word2Vec

- Embeddings capture co-occurrences
- Words that appear together frequently have similar vectors
- Language semantics?
- Distance measure can be used:
    - Cosine distance
- Limitations?
    - Fixed representations
    - Context
    - Beyond co-occurrences

Word2Vec applications

- Linking of ontology concept mentions to class IDs
  - Cho, H., Choi, W. & Lee, H. A method for named entity normalization in biomedical articles: application to diseases and plants. *BMC Bioinformatics*

Breast cancer	Similarity	Breast carcinoma HP:0003002	Breast hypoplasia HP:0003187	Diabetes HP:0000819
[0.79 . . . . . . 0.49]		[0.84 . . . . . . 0.55]	[0.32 . . . . . . 0.43]	[0.22 . . . . . . 0.95]

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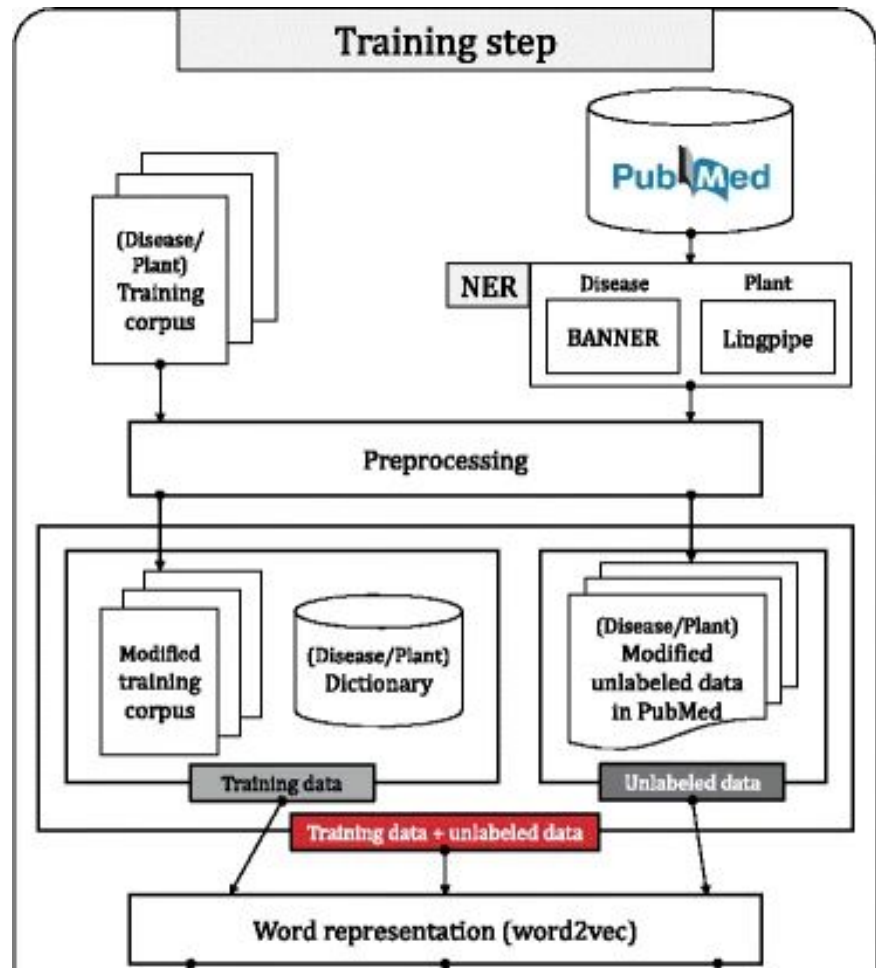

Figure from original paper

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Word2Vec applications

- Linking of ontology concept mentions to class IDs
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- Matching concepts between ontologies
  - **Liao, J., Huang, Y., Wang, H., Li, M. (2021). Matching Ontologies with Word2Vec Model Based on Cosine Similarity. In: , et al. Proceedings of the International Conference on Artificial Intelligence and Computer Vision (AICV2021)**

Word2Vec embeddings are generated
Concepts from two ontologies are aligned based on cosine similarity

Word2Vec applications

- Matching concepts between ontologies
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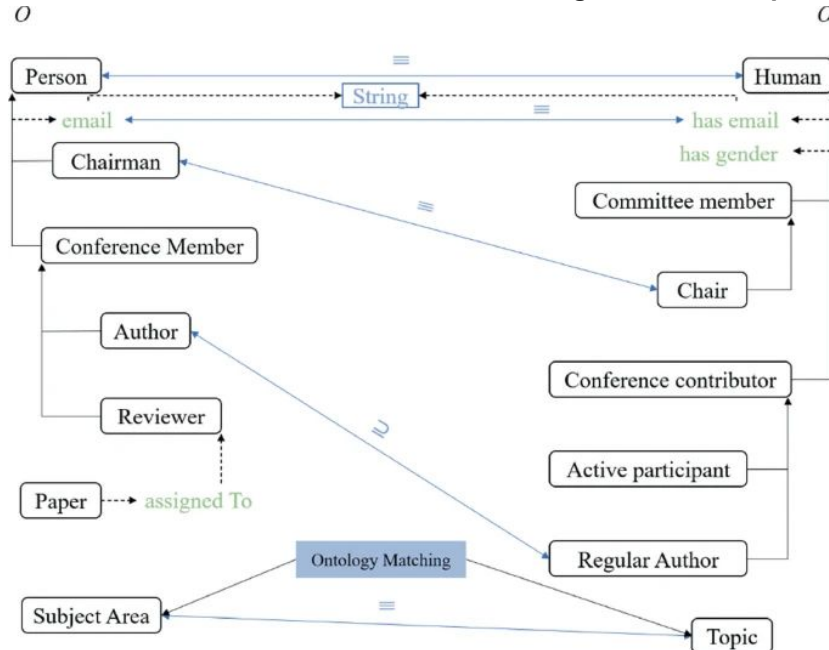

Figure from original paper

Word2Vec applications

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Word2Vec applications

- ## Linking of ontology concept mentions to class IDs
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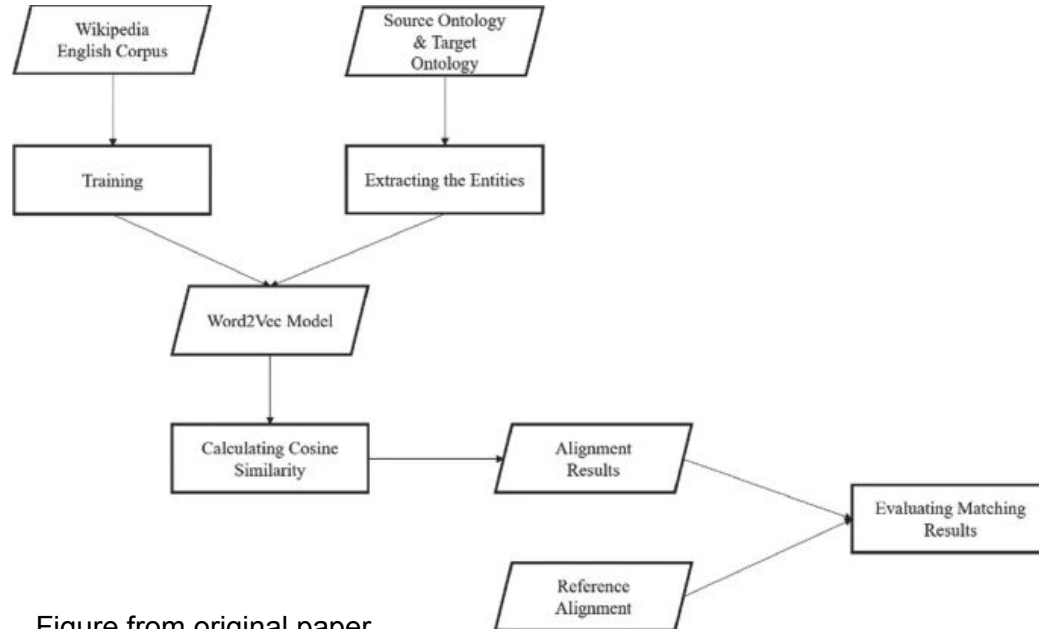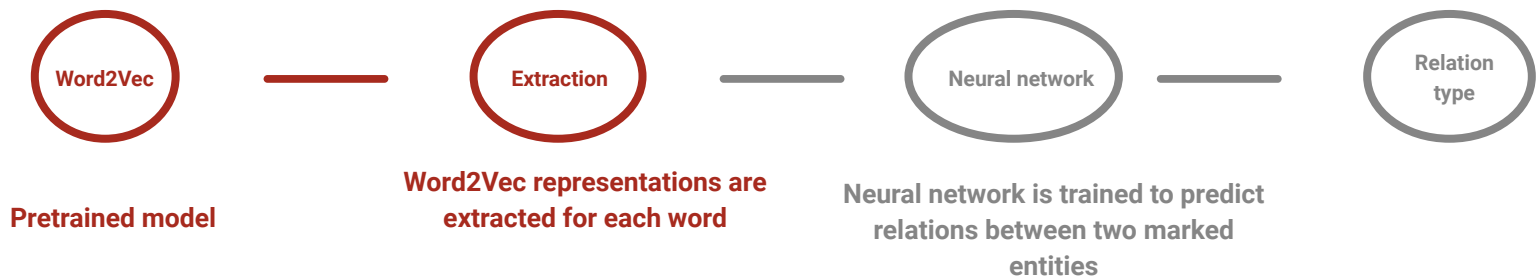- ## Relation extraction between entities from text
  - **Thien Huu Nguyen and Ralph Grishman. 2015. Rela-tion extraction: Perspective from convolutional neu-ral networks. InProceedings of NAACL-HLT.**

Word2Vec embeddings are generated
Neural convolutional models are trained to predict relations

Word2Vec applications

- Relation extraction between entities from text
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Word2Vec

Extraction

Neural network

Relation type

Pretrained model

Word2Vec representations are extracted for each word

Neural network is trained to predict relations between two marked entities

Example

In the morning, the President traveled to Detroit

Word2Vec shortcomings

Static representations

Context agnostic representations

BERT

Bidirectional Encoder Representations from Transformers

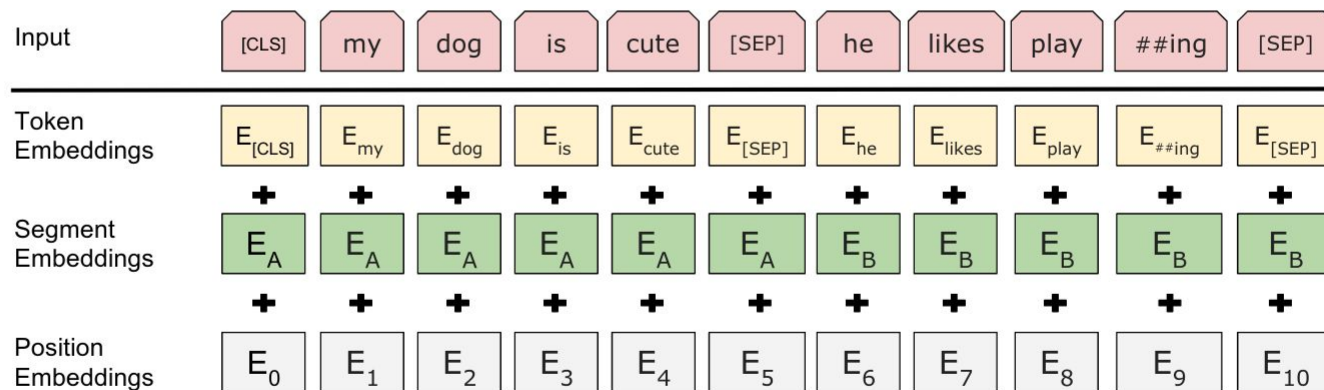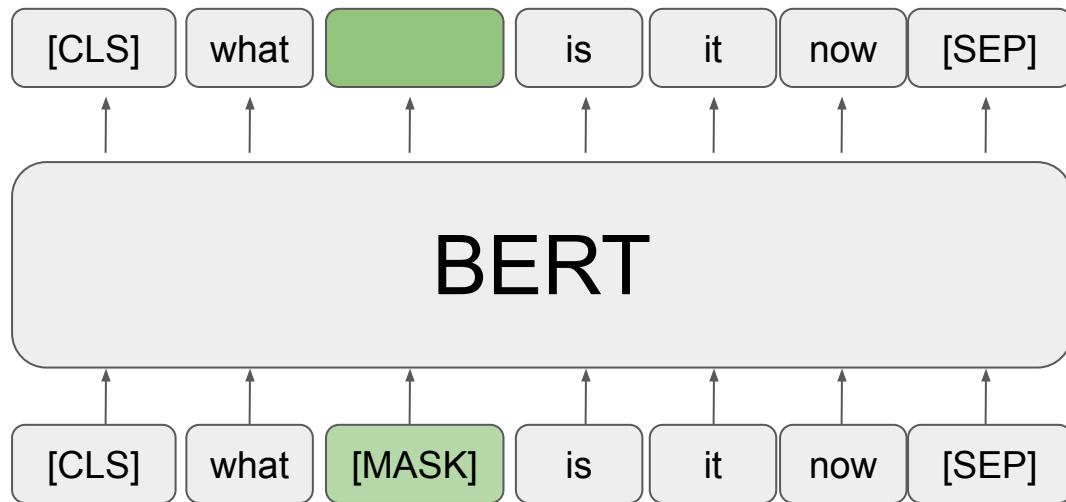| Input	[CLS]	my	dog	is	cute	[SEP]	he	likes	play	##ing	[SEP]
Token Embeddings	$E_{[CLS]}$	E_{my}	E_{dog}	E_{is}	E_{cute}	$E_{[SEP]}$	E_{he}	E_{likes}	E_{play}	$E_{\#\#ing}$	$E_{[SEP]}$
	+	+	+	+	+	+	+	+	+	+	+
Segment Embeddings	E_A	E_A	E_A	E_A	E_A	E_A	E_B	E_B	E_B	E_B	E_B
	+	+	+	+	+	+	+	+	+	+	+
Position Embeddings	E_0	E_1	E_2	E_3	E_4	E_5	E_6	E_7	E_8	E_9	E_{10}

Figure from the original paper: BERT: Pre-training of Deep Bidirectional Transformers for Language Understanding, Devlin et al.
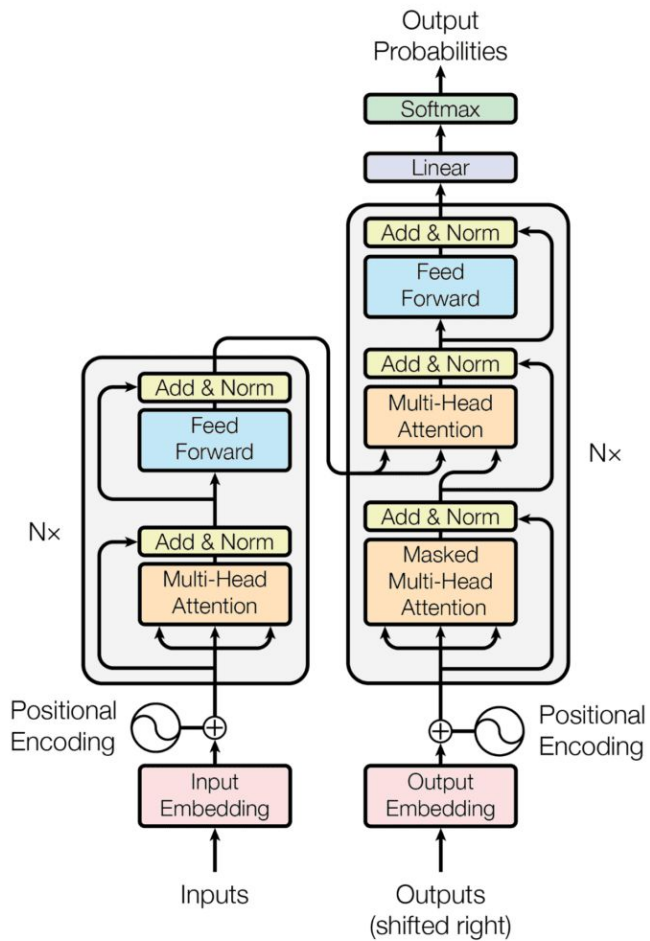
BERT

Masked Language Model (MLM)

BERT

Output
Probabilities

Softmax

Linear

Add & Norm

Feed
Forward

Add & Norm

Multi-Head
Attention

N×

Add & Norm

Feed
Forward

N×

Add & Norm

Multi-Head
Attention

Add & Norm

Masked
Multi-Head
Attention

Positional
Encoding

Input
Embedding

Inputs

Positional
Encoding

Output
Embedding

Outputs
(shifted right)

Figure from the original paper:Attention Is All You Need, Vaswani et al.

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BERT

- Dynamic
- Context-aware
- Word/sentence embeddings

BERT applications

- Finding and linking concept mentions from text to ontology IDs
  - **Ling Luo, Shankai Yan, Po-Ting Lai, Daniel Veltri, Andrew Oler, Sandhya Xirasagar, Rajarshi Ghosh, Morgan Similuk, Peter N Robinson, Zhiyong Lu. PhenoTagger: A Hybrid Method for Phenotype Concept Recognition using Human Phenotype Ontology. Bioinformatics, Volume 37, Issue 13, 1 July 2021, Pages 1884–1890.**

BERT is fine-tuned:
　　Labels and synonyms → positives
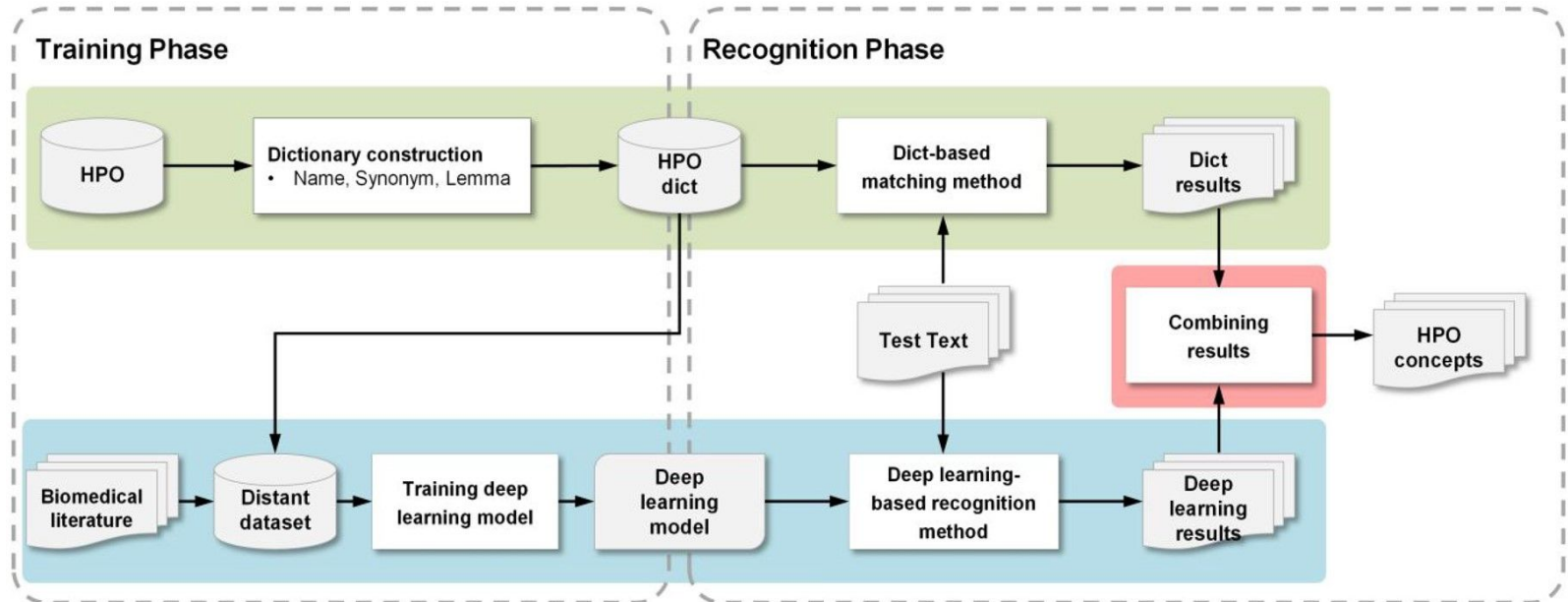　　Negatives are randomly sampled from some corpus

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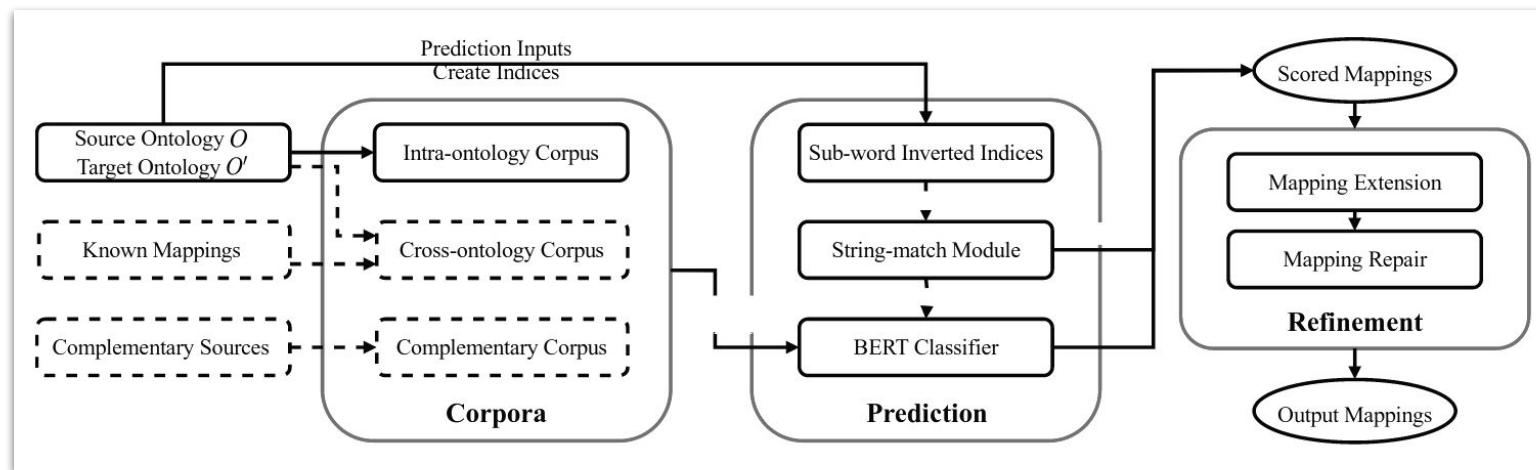
| Dry mouth | → | BERT | → | Xerostomia HP:0000217 |

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BERT is trained on Biomedical corpora
BERT is then fine-tuned using curated tuples

BERT applications

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The most common cause of hereditary breast cancer is an inherited mutation in the BRCA1

The most common cause of @DISEASE$ is an inherited mutation in the @GENE$

BERT

Score

Take home messages

- Textual components of ontologies can help
  - Extract knowledge from literature
  - Transfer knowledge from one source to another
- Methods to represent text
  - Word2Vec
  - BERT
- Important aspects of text:
  - Word meaning
  - Context

Hands-on