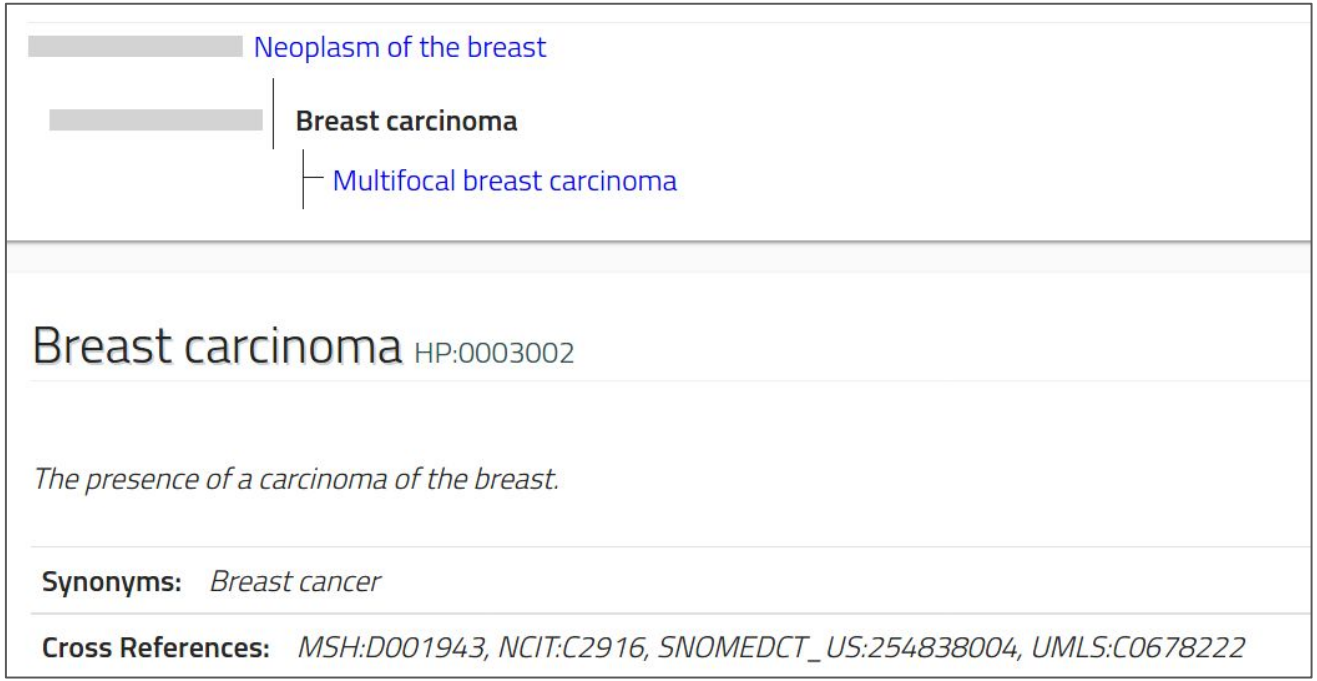
Ontologies and text mining

Textual components of ontologies



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

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

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
- 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
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  - Different stems
  - Novel words cannot be stemmed
- Numerical representation
  - Numerical methods
  - How?

Numerical representation and analysis of text

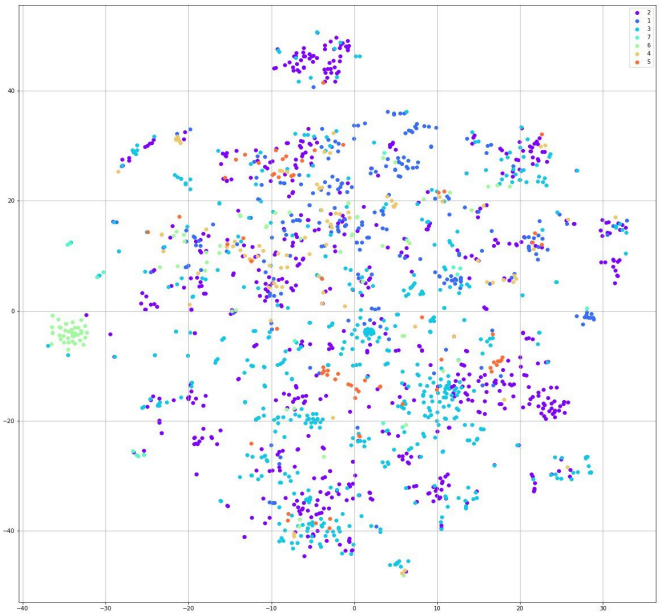
Popular methods:

- Word2Vec
- BERT

Both methods use embeddings to represent text

What is an embedding?

- An embedding ***Y*** is a representation of an instance ***X*** in a different space that preserves ***X***'s 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)
  - N-grams (words)
  - Sentences
- How can we make this useful for comparison?

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  - Words
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- 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?

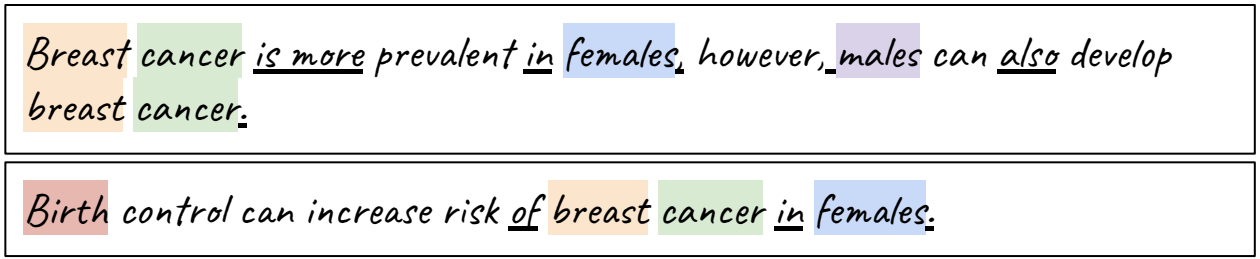
- How is text represented numerically?
- Units of text are assigned IDs to create a vocabulary
  - Characters (letters)
  - **Words**
  - N-grams
  - Sentences
- How can we make this useful for comparison?

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



Word2Vec

Word2Vec captures co-occurrences

Given a word:

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

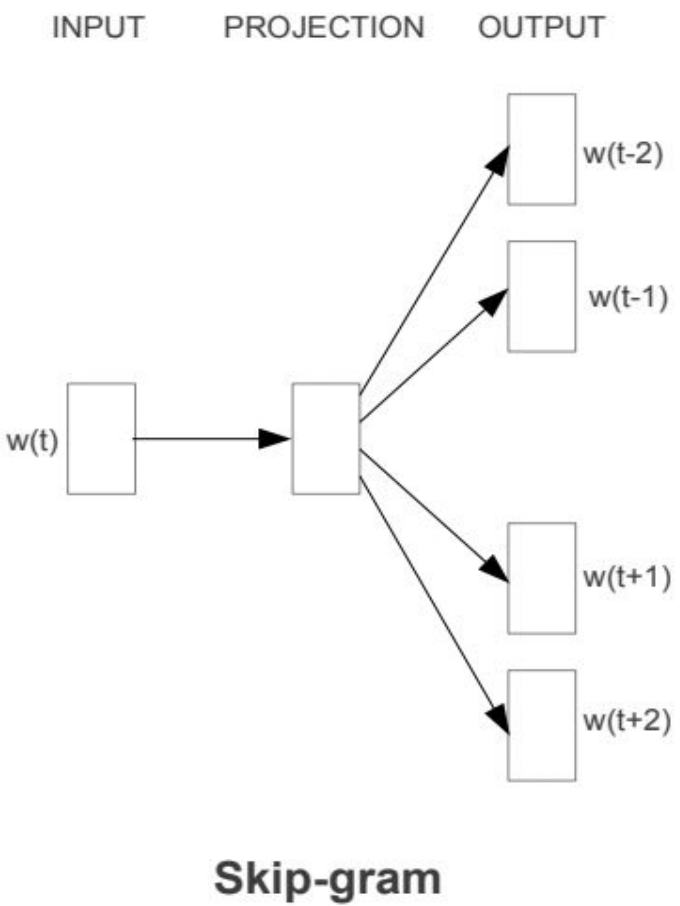


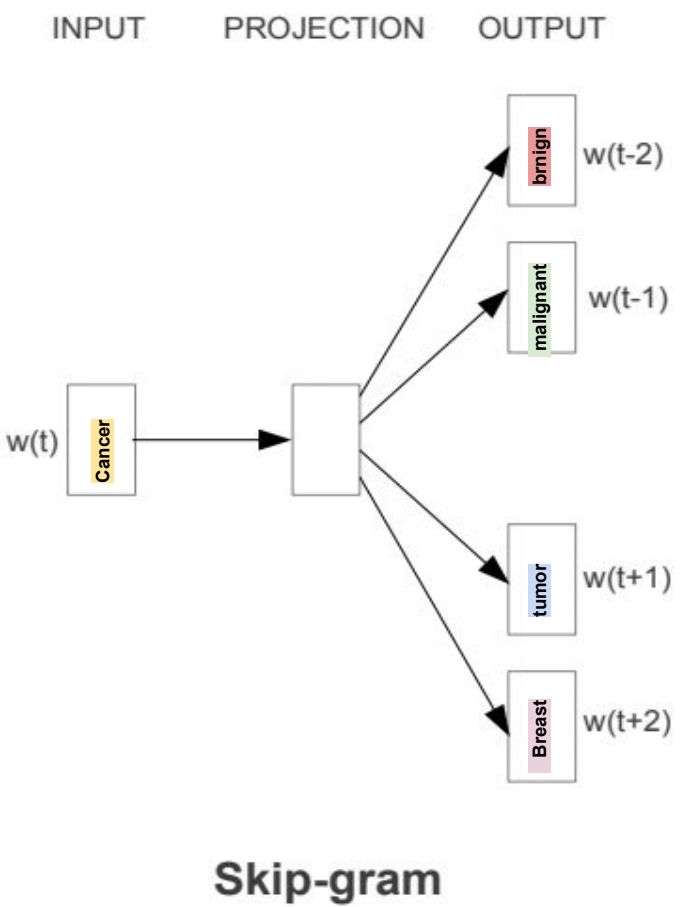
Figure from the original paper: Efficient Estimation of Word Representations in Vector Space, Mikolov et al.

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

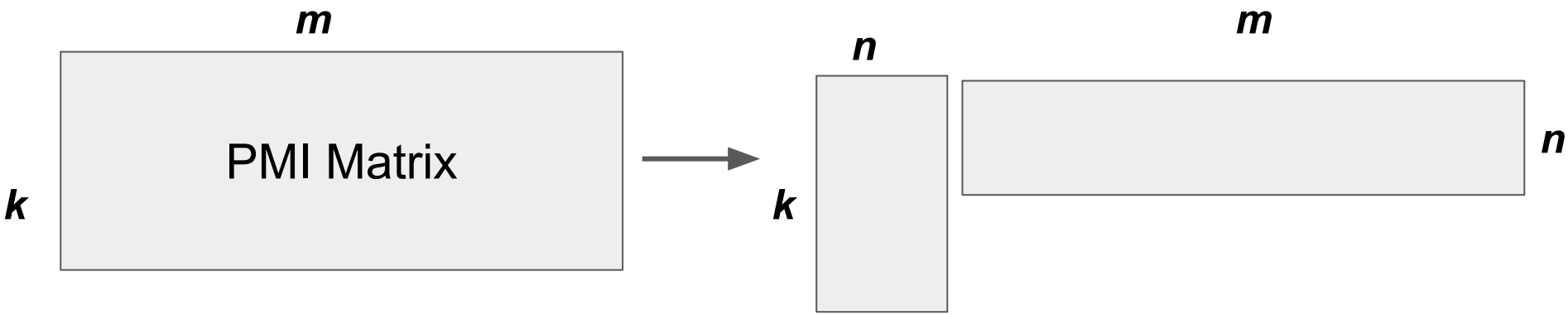
You can this of it as a factorization of a Pointwise Mutual Information (PMI) matrix

	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

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

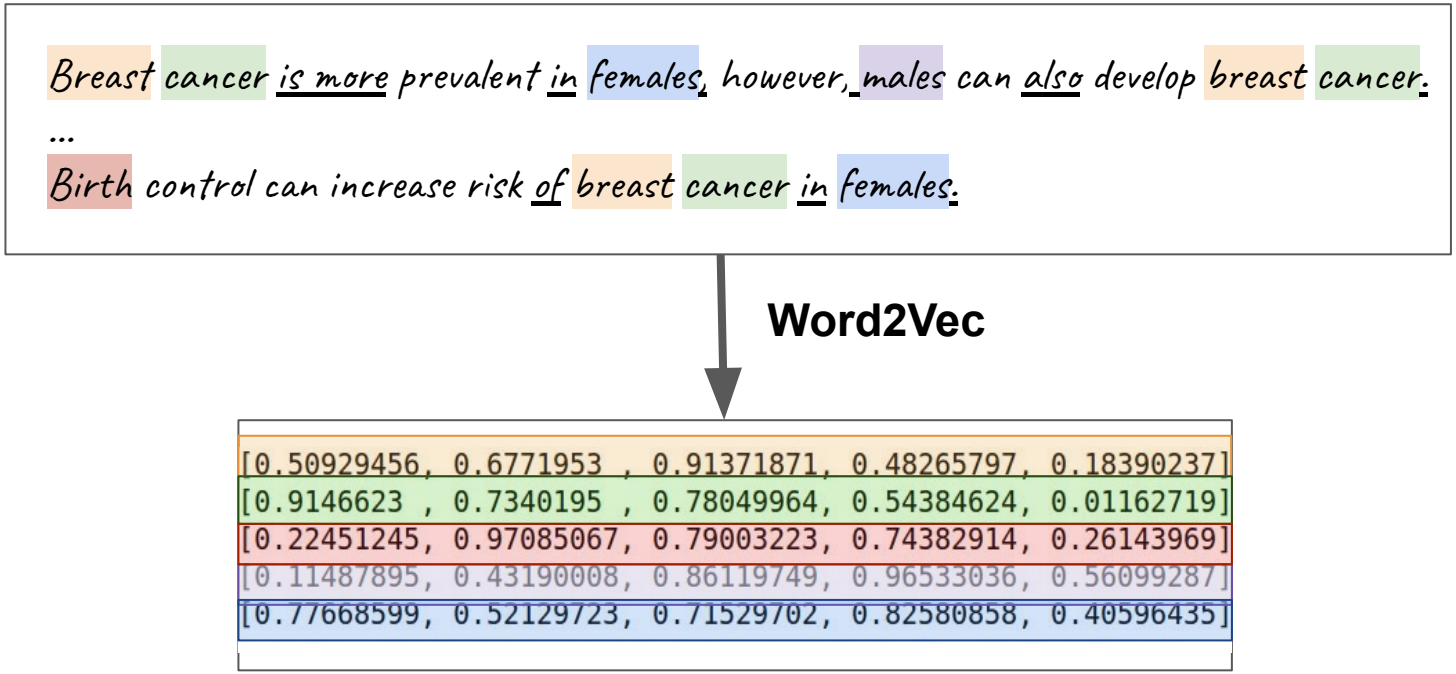
Word2Vec

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

Words → embeddings



Word2Vec

- Embeddings capture co-occurrences
- Words that appear together frequently have similar vectors
- Language semantics?
- Distance measure can be used:
  - Edit 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***

Word2Vec embeddings are generated
Cosine similarity is used to rank candidate class IDs

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

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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 shortcomings

Static representations

Context agnostic representations

BERT

Bidirectional Encoder Representations from Transformers

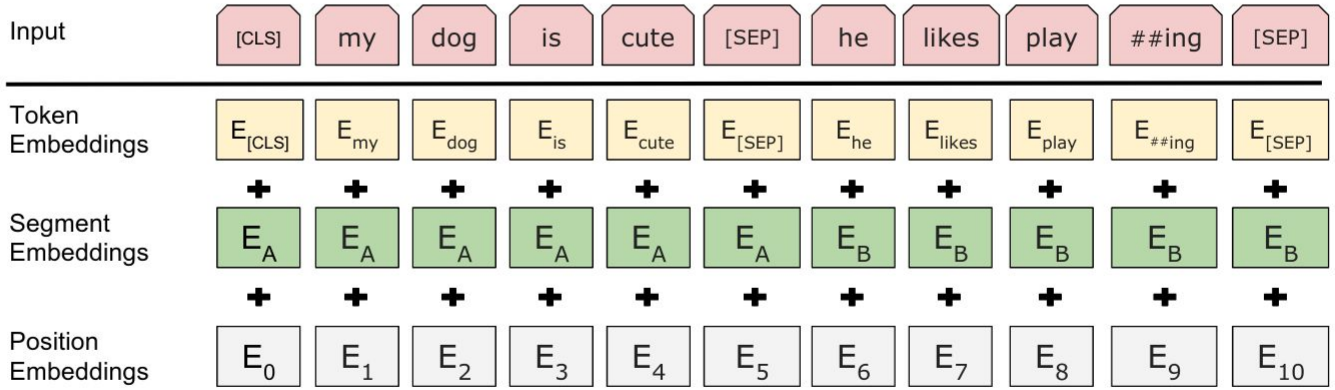
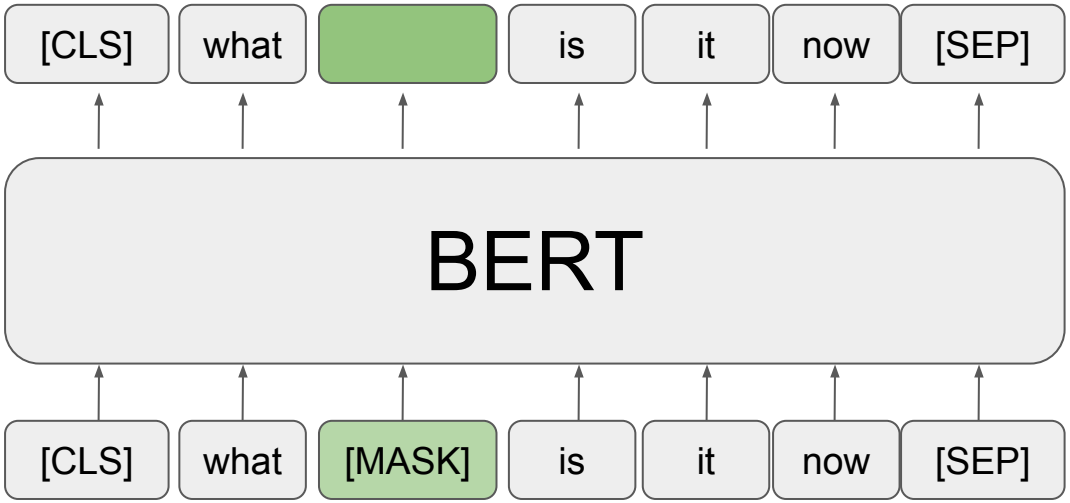


Figure from the original paper: BERT: Pre-training of Deep Bidirectional Transformers for Language Understanding, Devlin et al.

BERT

Masked Language Model (MLM)



BERT

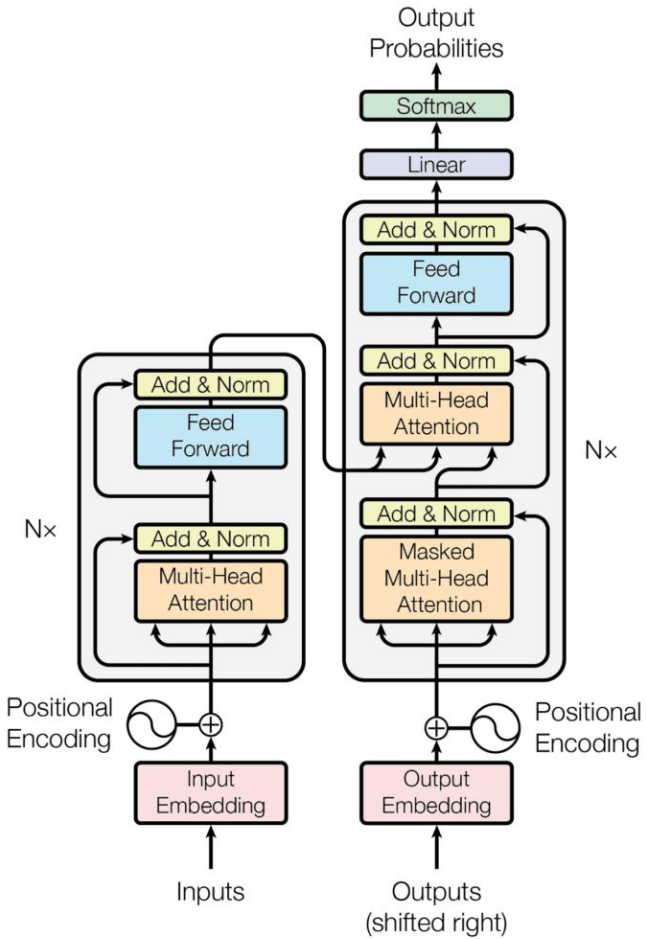


Figure from the original paper:Attention Is All You Need, Vaswani et al.

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

BERT applications

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- Relation extraction between entities from text
  - **Jinhyuk Lee, Wonjin Yoon, Sungdong Kim, Donghyeon Kim, Sunkyu Kim, Chan Ho So, Jaewoo Kang, BioBERT: a pre-trained biomedical language representation model for biomedical text mining, Bioinformatics, Volume 36, Issue 4, 15 February 2020.**

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

BERT applications

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- Matching concepts between ontologies
  - **He, Y., Chen, J., Antonyrajah, D., & Horrocks, I. (2022). BERTMap: A BERT-based ontology alignment system. Proceedings of the . AAAI Conference on Artificial Intelligence**

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