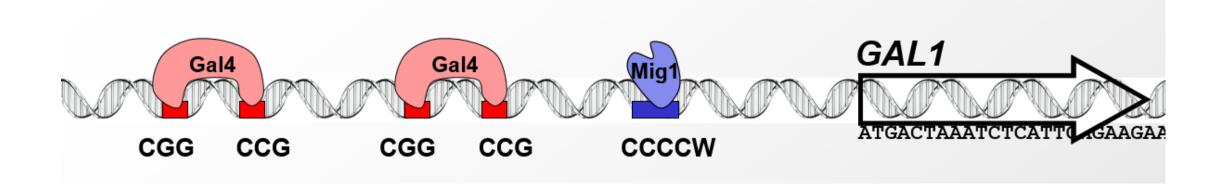
Transcription Factor Bound Regions Prediction: WordToVector Technique with Convolutional Neural Network

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1. Introduction: Motifs & Bound Regions



• Motifs: A pattern, with important biological functions e.g. Promoters

 Transcription Factor Bound Regions (TFBRs): Not only related with motifs

1. Introduction: Relate Bound Regions with Natural Language

- Motif —— 'Word' ,
- Combination of Various Motifs —— 'Sentence'
- Bound Region —— 'Meaning of the Sentence'

Meaning

Sentence

```
SUZ12_N chr21 9437245 9437298 AAAGGCTCT PPARA_3;IKZF2_2;RARG_4
SUZ12_N chr21 9437434 9437516 GTGTTTTTT ZNF384_1;MYC_disc10;YY1_d
MAFK_He chr21 9437491 9437516 TCCAGATGG EGR1_known4;TAL1_known3;
MAFF He chr21 9438106 9438108 AGC
```

Bound Regions Motifs in this Bound Region

2. Methods: Dataset OVERFLOW

Comes from 21st chromosome of human

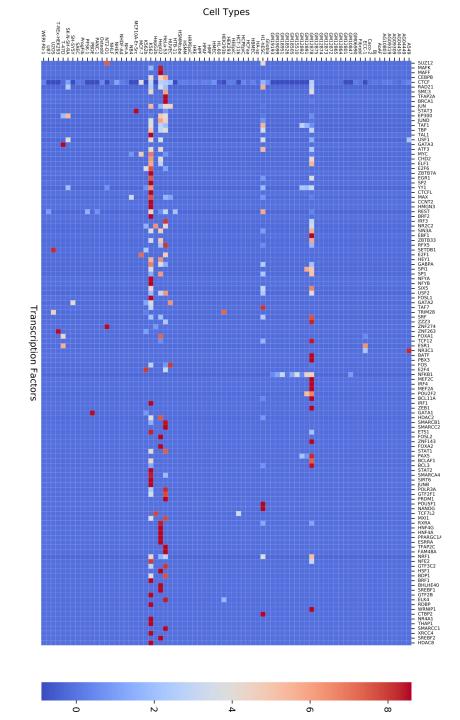
To Process:

• Group motif instances into one motif pattern:

```
CTCF_known1;COMP1_1;CTCF_known2
```

Group TF bound regions with cell types:

```
CTCF_GM12878_encode-Bernstein_seq_hsa
CTCF_HBMEC_encode-Stam_seq_hsa
CTCF_HEEpiC_encode-Stam_seq_hsa
CTCF_HPAF_encode-Stam_seq_hsa
```



Bound Regions 122 Row 2. Methods: Motifs Distribution Column Motifs 602 These Bound Regions are These Bound Regions Very Good Sample Highly Relevant with have many instances For Binary Classification Almost every Motif These Motifs appear Insignificant!

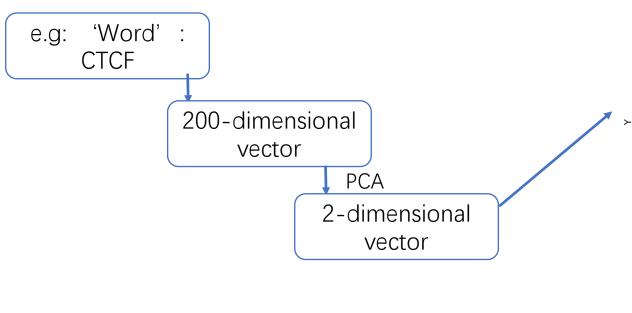
In all Bound Regions

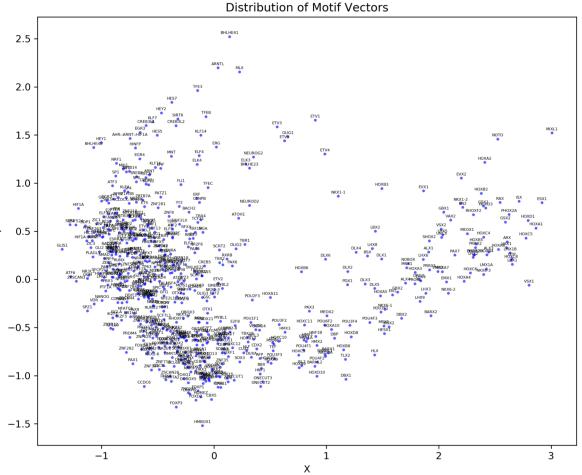
2. Methods: Word2Vec Algorithm

Each motif → A 200-dimensional vector

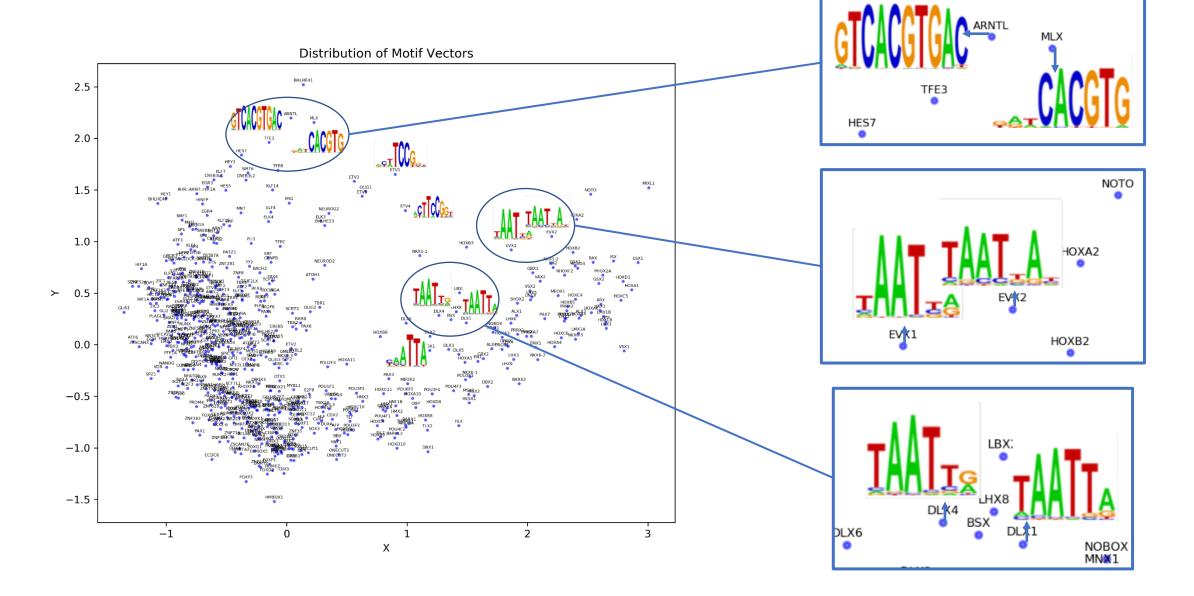
Motifs constantly bound together

 Closer in Distance in the Scatter plot





2. Methods: Distribution of Motifs



2. Methods: Label of the Dataset

• Binary Classification of Bound Regions:

```
CTCF_GM12878_encode-Bernstein_seq_hsa
CTCF_HBMEC_encode-Stam_seq_hsa
CTCF_HEEpiC_encode-Stam_seq_hsa
CTCF_HPAF_encode-Stam_seq_hsa
CTCF_HSMM_encode-Bernstein_seq_hsa
CTCF_NH-A_encode-Bernstein_seq_hsa
CTCF_NHEK_encode-Bernstein_seq_hsa
CTCF_NHEK_encode-Stam_seq_hsa
CTCF_Osteobl_encode-Bernstein_seq_hsa
CTCF_AG09309_encode-Stam_seq_hsa
```

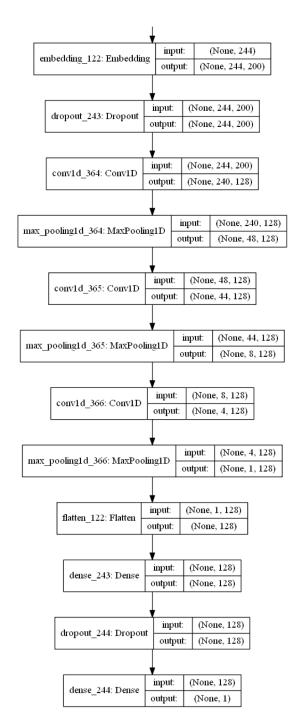
RAD21_GM12878_encode-Myers_seq_hsa_v041610. RAD21_HeLa-S3_encode-Snyder_seq_hsa_lgG-rab RAD21_K562_encode-Snyder_seq_hsa EGR1_K562_encode-Myers_seq_hsa_v041610.1 SP2_K562_encode-Myers_seq_hsa_v041610.2-SC-64

(+): CTCF, 26211, label: 1

(-): Non-CTCF, 38983 ,label: 0

2. Methods: Model Structure

- Sequential
- Embedding: Transfer word index
- CNN: Feature Extraction
- Dropout: Overfitting Prevention
- Sigmoid: Binary Classification



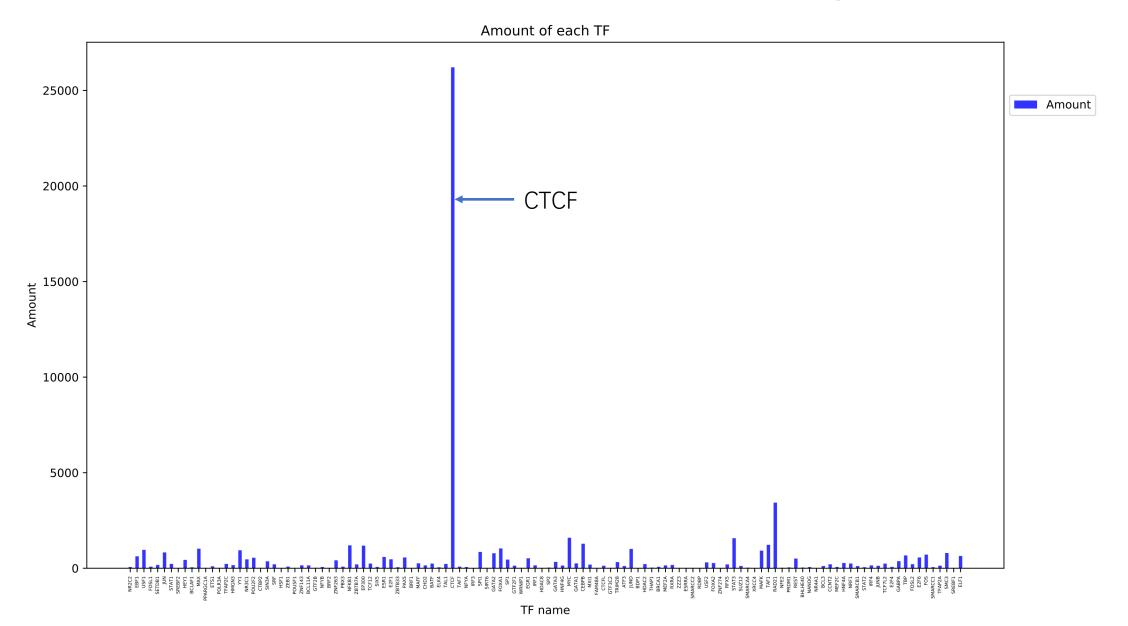
3. Results: Bound Prediction for CTCF Accuracy and Losses

Optimal Accuracy: 87% Model accuracy Model loss 0.50 0.88 0.45 0.86 Overfitting Accuracy 88.0 Overfitting 0.82 0.35 0.80 0.30 0.78 100 20 20 60 80 100 Epoch Epoch

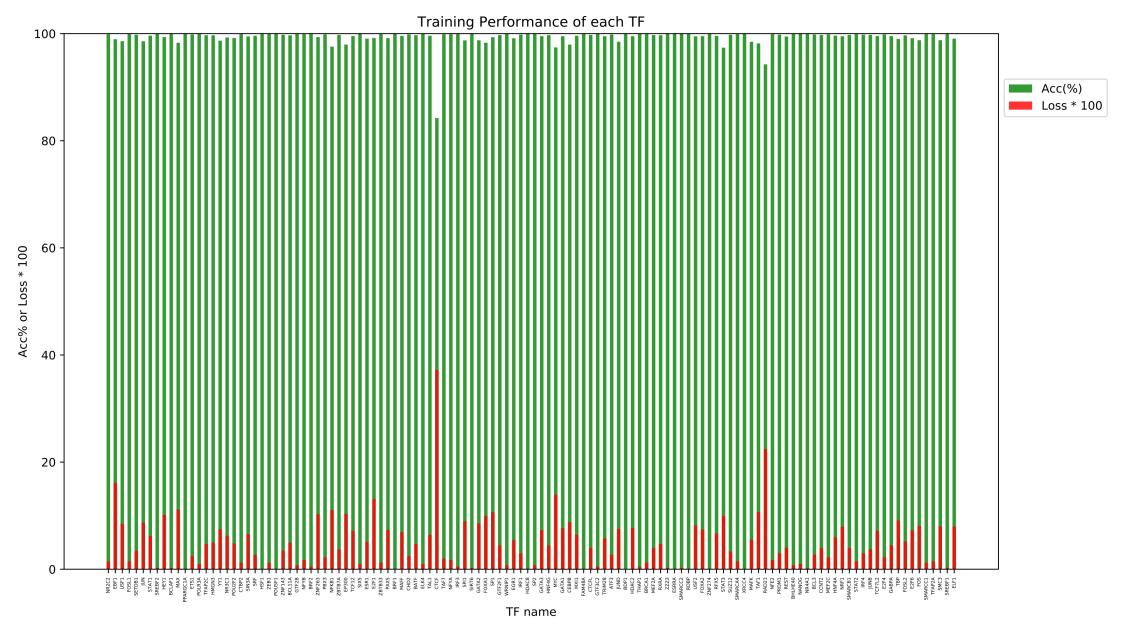
Smoothier After Dropout

But Still Uneven

3. Results: Bound Prediction for All Bound Regions



3. Results: Bound Regions for All Bound Regions



4. Future Goals:

- Data Extension: Expand out datasets to <u>ALL CHROMOSOMES</u>
- Higher Accuracy: Adjust our model till OPTIMAL
- More Features: Comparative Genomics, Epigenomics, Cell Types

5. Outlines:

- 1. Introduction: Motif -> Vector -> Natural Language Processing
- 2. Methods: Word2Vec Algorithm & CNN
- 3. Results: More than 87% Accuracy
- 4. Future Goals: <u>Larger</u> Datasets, <u>Better</u> Model and <u>More</u> Features