

**FAES BIOF509** 

# **Genetic Variants and Cancer**

**Machine Learning Final Project**Mary B. Makarious 16.05.2019

#### **BACKGROUND**

#### WHERE?

The data is from a Kaggle competition that was held ~2 years ago

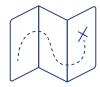
I opted to use this specific dataset because:

- Interesting topic
- Easy to download
- Discussion on different methods used

#### WHAT?

A lot of the ideas that went into this project came from:

- Class materials
- Google
- Kaggle Discussions
- Medium articles
- GitHub Snippets/Tutorials
- What actually worked...



Want to Follow Along?

 $\textbf{Kaggle:} \ \underline{\text{https://bit.ly/2LHDjSd}}$ 

Notebook: <a href="https://bit.lv/2LMLPj9">https://bit.lv/2LMLPj9</a>

### DATA + GOAL



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## Data from Kaggle:

- Information about the genetic variants:
  training\_variants.csv and test\_variants.csv
- Clinical evidence (in text form) that was used to manually classify the variants: training\_text.csv and test\_text.csv

**Goal:** To classify each variant into 1 of 9 mutation classes (unknown to you)

### WORKFLOW

# **Data Input**

## **Preprocessing**

Natural Language Processing

**4-Layer Neural Net** 

Developing the Model

Training the Model

**Predictions on Test Data** 

**Reformatting for Output** 



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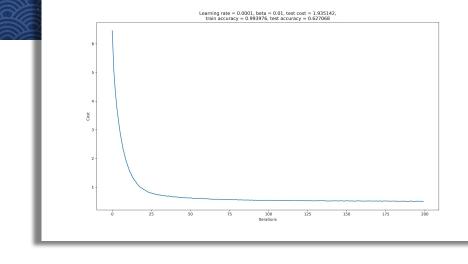
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**Loss per Iteration Plot** 

### MODEL TRAINING + PLOT





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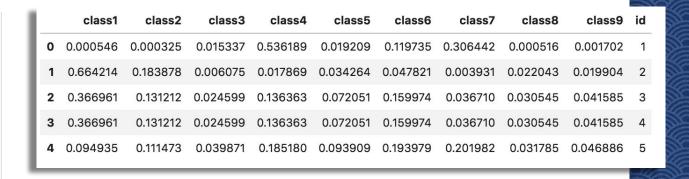
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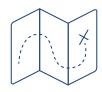
Notebook: https://bit.lv/2LMLPj9

**Personal Interpretation**: Both the NLP and the 4L-NN were successful. The test accuracy is well above chance (~63%).

Would be interesting to see how to improve this (would adding another layer help?)

# **OUTPUT EXAMPLE**





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**Personal Interpretation**: Each variant is reported with the distances between each cluster following NLP+NN (What way would be best to visualize this?)

# **LESSONS LEARNED**

- Machine Learning/Deep Learning can be tough
- There are multiple ways to do something
- This was my first attempt at both NLP and NN
  - There are plenty of resources online explaining ways to further improve your model



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Question for **you**: Ideas on how to best visualize this data?

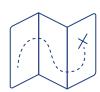
#### **CREDITS**

This project would not have been possible without:

- Martin and Alex
- Kaggle
- Google
- Countless hours of frustration
- Kind people who spend hours cultivating scripts, articles, and tutorials online
- Developers behind all the packages

Any Questions?

GitHub: m-makarious/ML\_GeneticVariants\_in\_Cancer



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