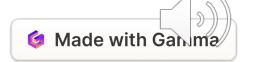


# Deciphering Genetic Codes Across Species: A Machine Learning Approach with k-Means Clustering of DNA Sequences

Genomic sequencing is a burgeoning field, amassing nearly inconceivable amounts of data. These massive sets of information require intensive sorting to facilitate understanding. Machine learning serves as an excellent tool for this purpose. Machine learning methods are increasingly valuable in classifying DNA sequences, so we will attempt to do this across these distinct but related species (humans, chimpanzees, and dogs).

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## **Background/History**

1

#### **Genomic Sequencing Technology**

Genomic sequencing technology has rapidly improved in recent years, leading to a notable expansion of DNA data availability. With so much information available, processing it quickly and effectively is tantamount in the field.

2

#### **Machine Learning in Genomics**

Machine learning has the potential to be a powerful method in refining understanding about genes, their interactions, their effect on bodily mechanisms, their evolutionary implications, and more.

3

#### **Novice Approach**

Due to the author's novice in genomics, this project will attempt a straightforward k-means clustering approach.



## **Data Explanation**

#### **Genetic Sequences**

We are looking at the DNA sequences, made up of nucleotides (ACGT), that are essentially a language for directing the actions of the body.

#### **NLP Techniques**

NLP techniques can be applied to identify patterns within k-mers, such as frequency counts, which can be analogous to word frequencies in text analysis.

#### **Sample Sequences**

This project elects to use sample sequences (as opposed to full genomes) to test various models' efficacy in classification.





## Methodology

1 Data Preprocessing

Import datasets with Pandas and review distribution of classes among datasets.

**2** Data Transformation

A Bag of Words model is created using CountVectorizer, which is a method to convert a collection of text documents to a matrix of token counts.

3 Modeling

Perform k-means clustering utilizing Kmeans and MiniBatchKmeans and determine the best k value through the elbow method.



### **Analysis**

# Human Genetic Clustering

The human genetic data shows a dominant cluster, which we've labeled as Cluster 1, encompassing a significant majority of the sequences with 4298 instances.

# **Chimpanzee Genetic Clustering**

The chimpanzee data also exhibits a primary cluster, with Cluster 1 containing 1646 sequences.

#### **Dog Genetic Clustering**

The canine sequences
demonstrate a very distinct
pattern, with a primary cluster
(Cluster 1 with 811 sequences)
and several minor clusters or
single instances.



### **Conclusion & Limitations**

#### **1** Genetic Evolution

The clustering analysis has the potential to identify regions of genetic similarity and divergence, which can further our understanding of genetic evolution and the underlying biological functions of these sequences.

#### **2** Intersection of Fields

This project's approach, at the intersection of genomics and computational linguistics, underscores the innovative potential of NLP in biological sciences.

#### 3 Limitations

Despite its limitations, it has the potential to serve as valuable experience for the author and could benefit the greater understanding of fellow scholars interested in this exciting intersection of fields.





### **Challenges & Ethical Assessment**

1 Data Processing

The data used in this project is limited and, due to its nature, needs to be encoded in a specific manner, then converted again.

2 Preprocessing Impact

The initial analysis suggests that the preprocessing step could be influencing the clustering outcomes significantly.

**3** Future Investigations

Further investigations will include revising the preprocessing pipeline and experimenting with other clustering techniques to find the most representative clustering solution for our data.

**4** Ethical Considerations

It is crucial to maintain anonymity of DNA sequence sources to protect privacy. Transparency and detailed documentation will ensure our work contributes positively to genomic research and adheres to regulatory standards.



# **Future Uses/Additional Applications**

**1** Health and Wellness

If this method is proven to be accurate and reproducible, it could be applied to other animals, plants, bacteria, and fungi, guiding environmental conservation, sustainability, and technology.

2 Breakthroughs

These applications could guide breakthroughs in health, wellness, biotechnology, and possibly even immortality.

3 Refinement

As it is refined, it could be useful to compare clustering of genotypical sequences against abnormal sequences to see where they vary.

### Recommendations

1 Reproduction with Larger Datasets

Ideally, this method would be reproduced with larger datasets (full genomes) and data sourced from more subjects. 2 Refinement of Methodology

For each attempt, feature selection, conversion, and vectorization could be refined. Being able to visualize a fuller assessment of the clustering is also recommended.

3 Subject Matter Expertise

Having a subject matter expert present during these explorations and experiments would also be ideal.

## **Implementation Plan**

1 Research Expansion

To expand on this research, one would need to consider more sophisticated bioinformatics techniques, access to full genomes, and the computational resources to process them.

**2** Collaboration

Furthermore, collaboration with geneticists would be invaluable in interpreting the biological significance of the clusters identified by the machine learning models.

**3** Ethical Considerations

Detailed documentation is essential for reproducibility as well as peer review. The goal of this project is to contribute positively to genomic research, so the author and future collaborators must uphold their responsibility to ethically use the information and tools at our fingertips.

### Questions

- 1. Will you explain more about Natural Language Processing and its mechanisms?
- 2. Could you elaborate on the implications of the skewed data observed in your analysis?
- 3. What approaches could improve the efficacy of this methodology?
- 4. Why are the assumptions listed critical for the methodology of this project?
- 5. How might the findings of successful k-means clustering contribute to the existing body of knowledge in genomic research and machine learning?
- 6. Has this methodology been implemented on a larger scale in other studies?
- 7. What other types of clustering methods would be applicable to this data?
- 8. What additional machine learning methods could be attempted with this data?
- 9. What are the potential real-world applications of this research on human health and environmental sustainability?
- 10. What are the broader ethical implications of using machine learning in genomic sequencing, particularly concerning data privacy?



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