IT5007: Software Engineering on Application Architecture

Course Project Interim Evaluation

Machine Learning Genomics App

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1. **Problem Statement:**

Our “ML-based genomics analysis app” aims to utilize state-of-the-art machine learning technology in other research areas.

In terms of research side novelty we are working on developing machine learning algorithm based ancestry breakdowns . This is also informed by domain knowledge of genomics, history and ethnography.

Many researchers and data analysts may have difficulty in running off-the-shelf machine learning models, among other reasons because they need to install the development tools, packages and have to become familiar with the programming languages. There are online tools such as “Visualgo" which implement all kinds of algorithms to help students easily understand algorithms and use the tool. However, there are not such open and free tools in the Genomics research area. Our project is to help them out by implementing all the things together. To help research and analysis with relative ease without having to download any development tools and installing related packages.

We propose an app with 3 major functionalities

1. Genetic Distance Tool:

It is to calculate the genetic distance of the input sample to that with other samples. Will return the nearest neighbors upon pressing a button. The whole distance matrix upon entering n samples

2. Genetic PCA Tool:

This functionality will make the genetic PCA plot
Since PCA is a linear dimensionality reduction method the relative distances are comparable
We might look into some novel things such as making Umap and t-SNE plots for further fine-grained visualization if time permits.

3. Ancestry Breakdown:

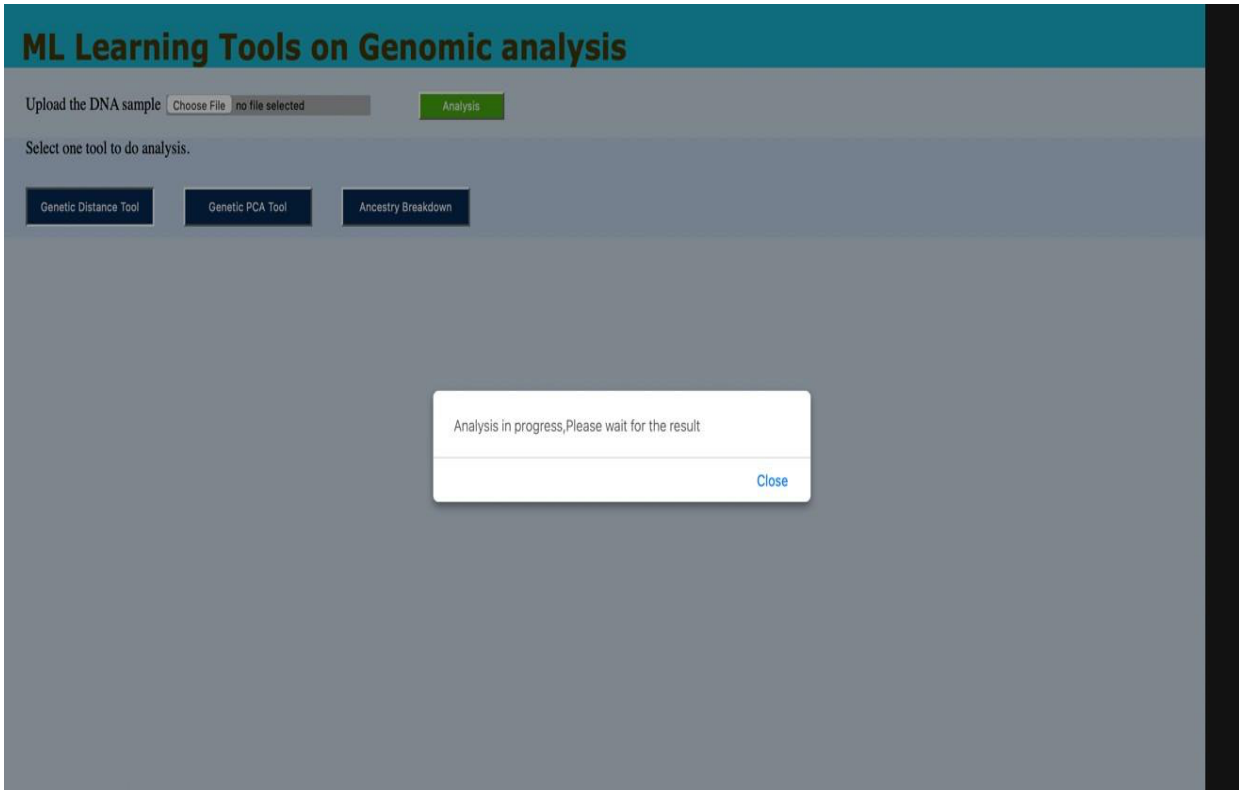
Fuzzification is performed after the machine learning clustering is done. This is after the models have been made and the clusters have been generated. This allows us to come up with the ancestry percentages. Various models and model specifications will be attempted. Domain knowledge comes into play in terms of the results making sense.. The ancestral components are selected such that they correspond to realistic source populations.

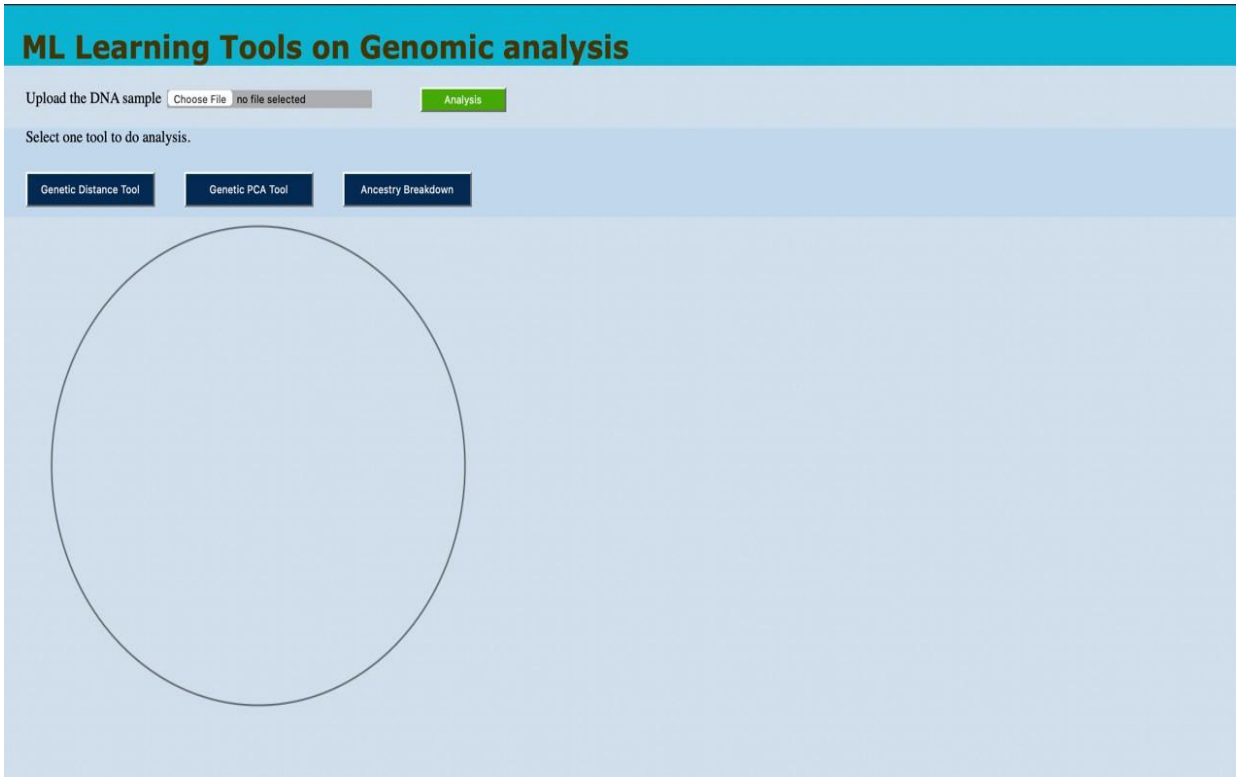
Will look into using various machine learning algorithms to develop the ancestry tool such as:

Fuzzy c means	GaussianMixture
SpectralClustering	AffinityPropagation
AgglomerativeClustering	Birch
DBSCAN	K-means
MiniBatchKMeans	MeanShift
OPTICS	SpectralClustering

2. Solution Architecture

The frontend has been designed to be simple and easy to use. Users need only to upload their DNA sample followed by selecting a machine learning tool(“Genetic Distance Tool”, “Genetic PCA Tool”, “Ancestry Breakdown”) and then click on the “Analysis” button, our machine learning backend will output the results



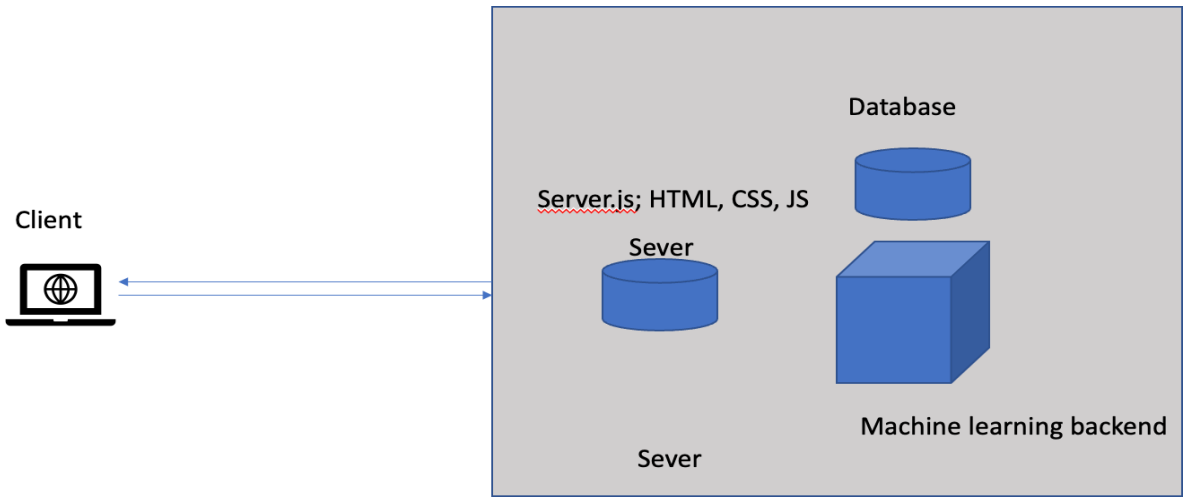


We plan to implement the machine learning model as the backend and deploy it

The generation of ancestries involves fuzzification of results from unsupervised learning algorithms

This step is done ex-post the clustering for ease of computation

The architecture will be as given below:



3. **Legal/Other Aspects**:

Open source libraries to be used:

Numpy

Pandas

Matplotlib

Scikit-Learn

Umap

etc.

Various data processing, genomics and machine learning packages will be used.

4. Competition Analysis

Vahaduo has a separate custom PCA tool and a Admixture tool which has the genetic distance functionality

The research originality is terms of the ancestry breakdown.
There is no competition for the machine learning based ancestry breakdowns to the best of our knowledge.
(https://vahaduo.github.io/vahaduo/)

5. Feature List

Project Schedule			
Documentation	**PIC**	**Due date**	**Status**
Project discussion report	HUANG DAN and SUPRATIK BHATTACHARYA	23-Feb	Done
Interim Evaluation report	HUANG DAN and SUPRATIK BHATTACHARYA	01-Mar	Done
Final Evaluation report	HUANG DAN and SUPRATIK BHATTACHARYA	01-Apr	Onging
FrontEnd			
Mainpage	HUANG DAN	31-Mar	Onging
Genetic Distance Tool	HUANG DAN	31-Mar	Onging
Genetic PCA Tool	HUANG DAN	31-Mar	Onging
Ancestry Breakdown	HUANG DAN	31-Mar	Onging
BackEnd			
Backend related work	HUANG DAN and SUPRATIK BHATTACHARYA	23-Apr	Onging
Genetic Distance Tool	SUPRATIK BHATTACHARYA	23-Apr	Onging
Genetic PCA Tool	SUPRATIK BHATTACHARYA	23-Apr	Onging
Ancestry Breakdown	SUPRATIK BHATTACHARYA	23-Apr	Onging

6. Git Repository :

The Repository on Github is given below:
https://github.com/dandelion793/IT5007CourseProject

Class Participation (2 points): Participation in the class discussions on MS Teams will be evaluated. Cases where the student writes an explanatory post for the peers will be considered:
We have attended the discussion meeting with the professor at 3:15pm on 23 Feb.

Appendix A:

Data sources-Ancient DNA samples:

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- Narasimhan, V. M., Patterson, N., Moorjani, P., Rohland, N., Bernardos, R., Mallick, S., ... & Reich, D. (2019). The formation of human populations in South and Central Asia. *Science*, *365*(6457), eaat7487.
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