Predicting the Origin of individuals from Genetic data

Team 17; https://github.com/Annilo/POrigGen

Ami Sild

1st year Masters student,
Data Science

Danat Yermakovich

3rd year PhD student,
Centre for Genomics,
Evolution and Medicine,
Institute of Genomics

Agnes Annilo

1st year Masters student,
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Grayson Felt

1st year Masters student,
Actuarial and Financial
Engineering

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Genealogical geographical origin

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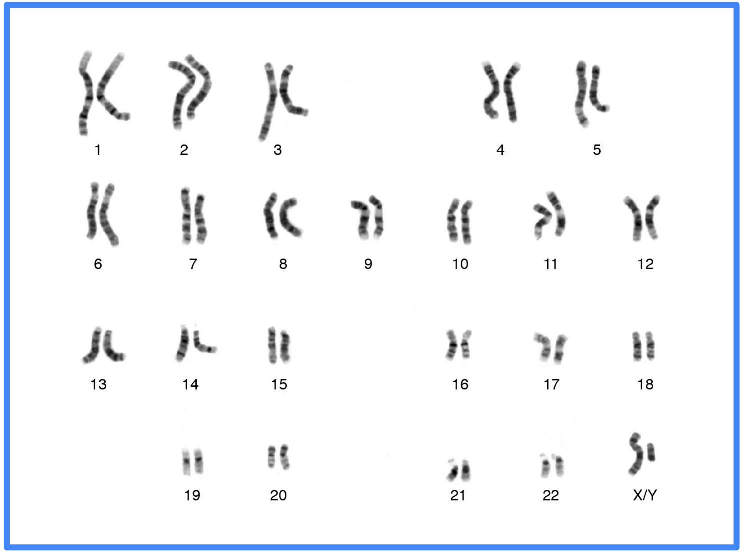
due to:
gradient changing
of
genetic population
structure
across world



Human genome

https://www.genome.gov/genetics-glossary/Karyotype

Approach

Predicting sample's population label from genetic data

80%
train

3200 samples (observations)
26 Pops across 5 SuperPop
50-150 samples per Pop

~10 millions genetic
variations i.e. features

2561 / 641

<table>
<tr><th>chr_pos_ref_alt</th><th>1:58771:T:C</th><th>1:183401:C:G</th><th>1:186291:G:A</th><th>1:281912:C:G</th></tr>
<tr><th>SampleID</th><th></th><th></th><th></th><th></th></tr>
<tr><td>HG00097</td><td>1|1</td><td>0|0</td><td>0|0</td><td>0|0</td></tr>
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Approach

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3200 samples (observations)
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80% train

Genetic feature preprocessing (MAF < 0.05, LD pruning)

70 000 features

PCA

Train and evaluate different models

Outcome

predicting a sample's population label (1000G)

10 KK => 70K

2561 / 641

chr_pos_ref_alt	1:58771:T:C	1:183401:C:G	1:186291:G:A	1:281912:C:G
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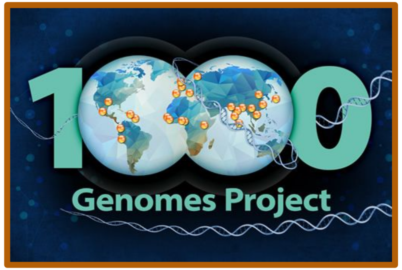
HG00102	1|1	0|0	0|0	1|0
HG00103	0|0	0|0	0|0	0|0
HG00105	0|1	0|0	0|0	1|0
HG00106	0|1	0|1	0|0	0|0
HG00107	1|0	0|0	0|0	1|0

Approach

Predicting sample's population label from genetic data



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PCA

Visualisation

Train and evaluate different models

CV-tuning of hyperparameters and N of PCs

Outcome

predicting a sample's population label (1000G)

2561 / 641

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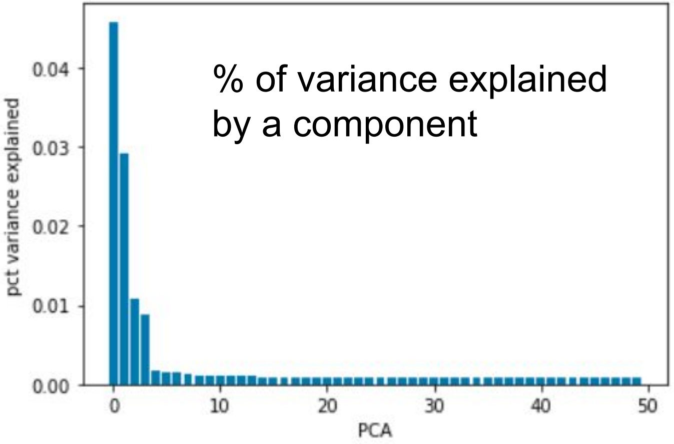
HG00107	1|0	0|0	0|0	1|0

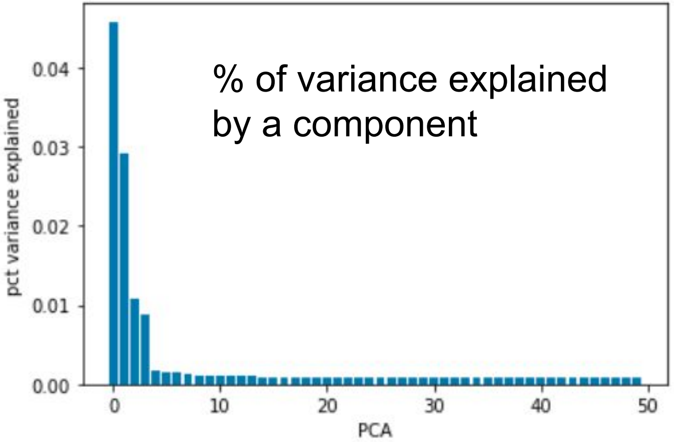
Results: Data PCs

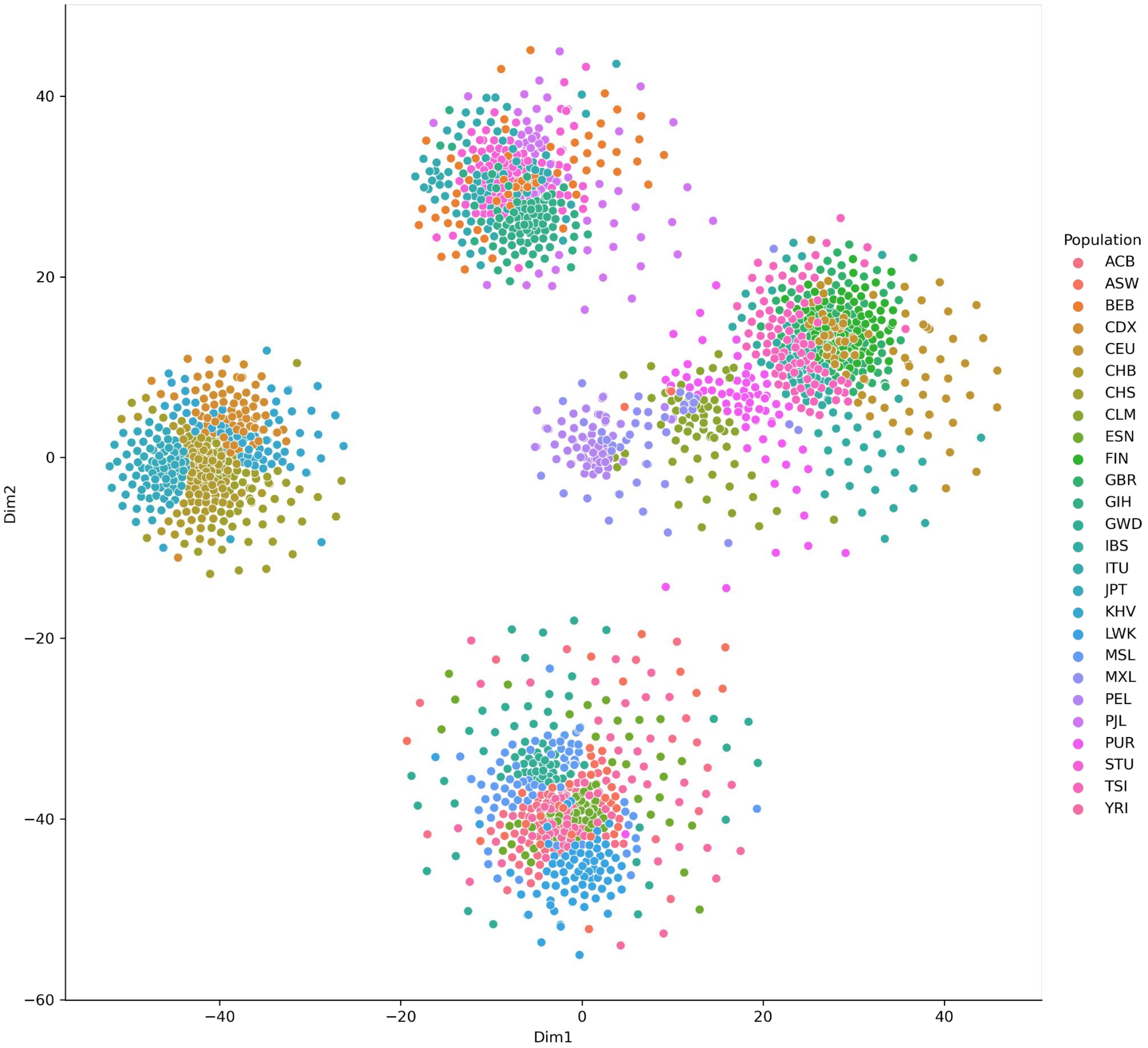
2561 train samples from 1000G:
all PCs



Results: Data tSNE

2561 train samples from 1000G: **all PCs**





Results: Data tSNE

2561 train samples from 1000G:
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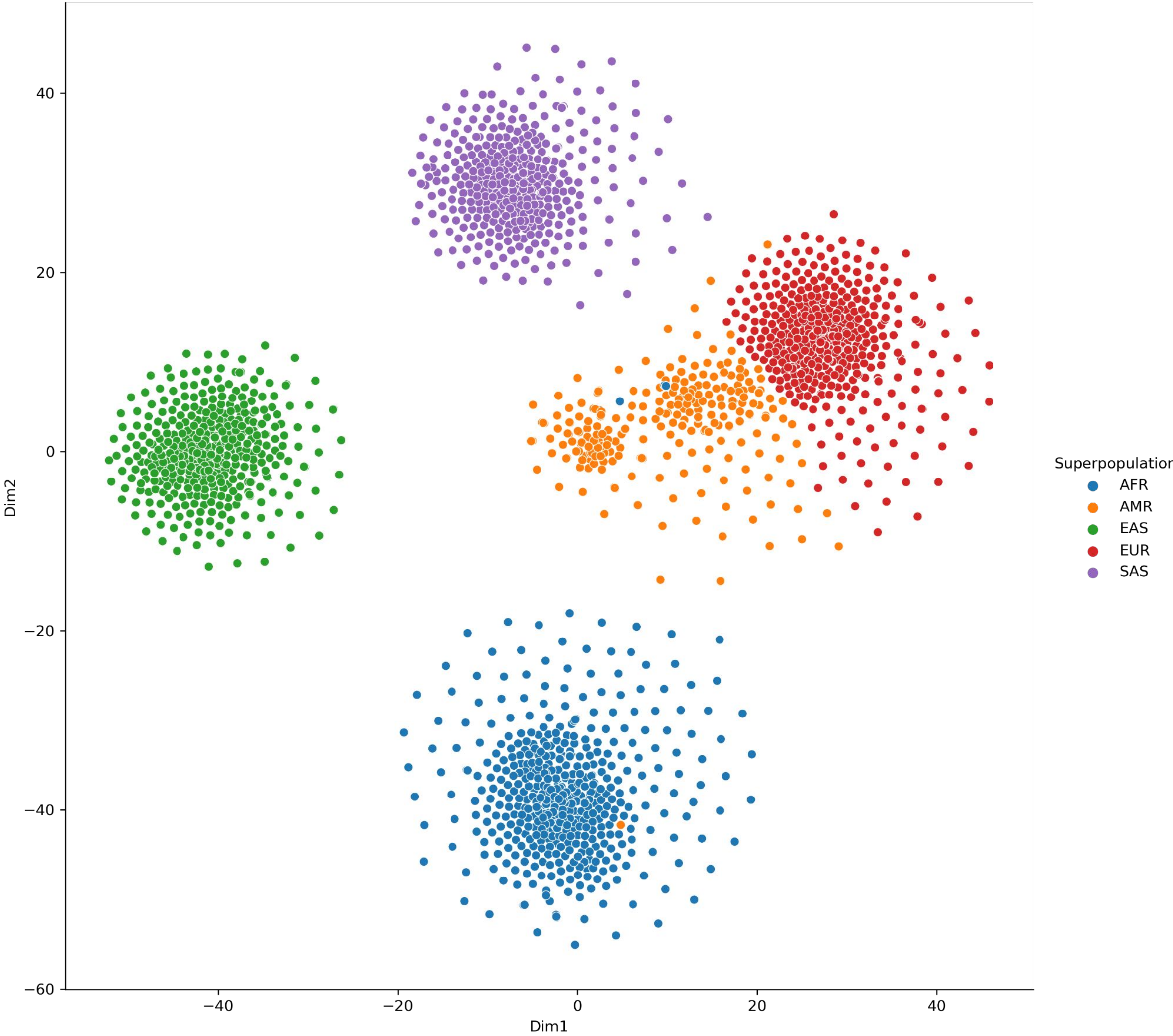
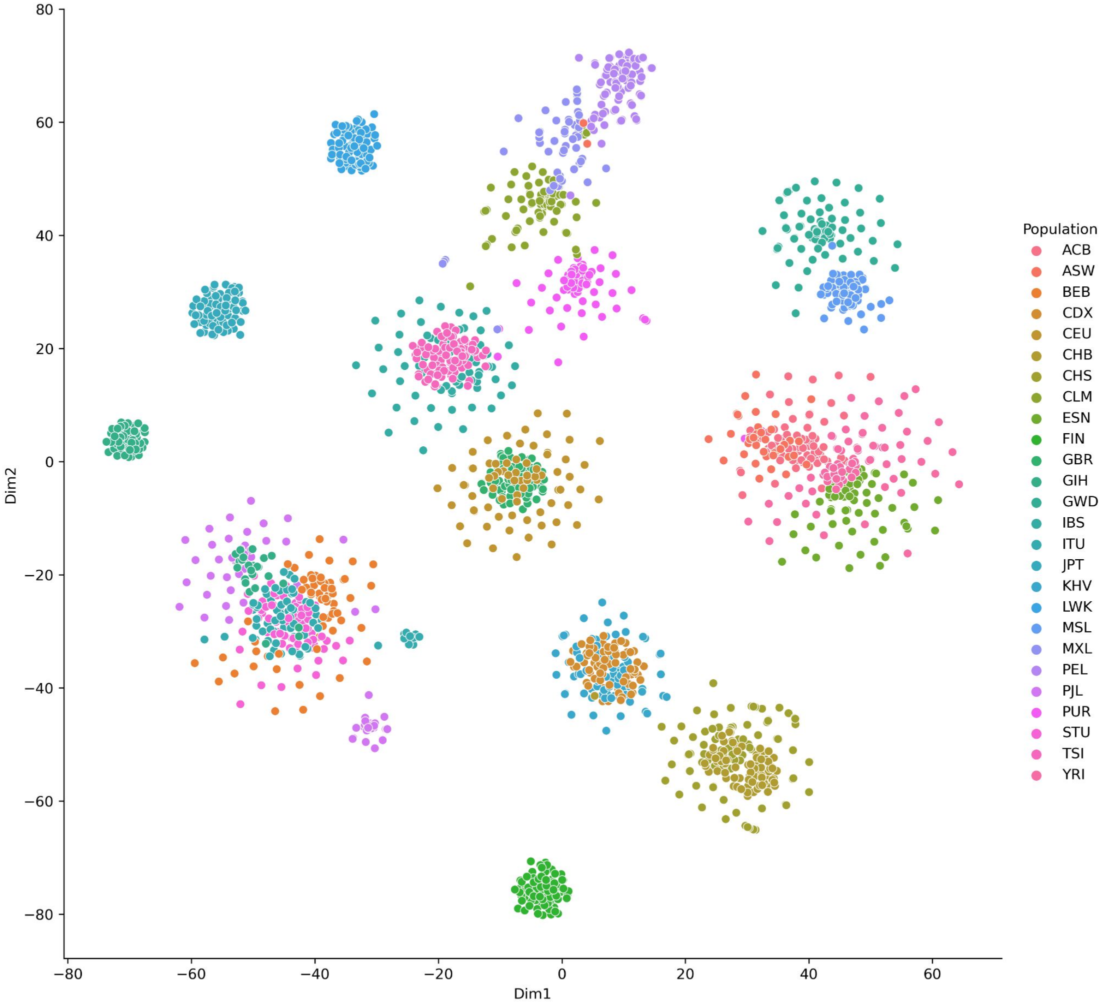


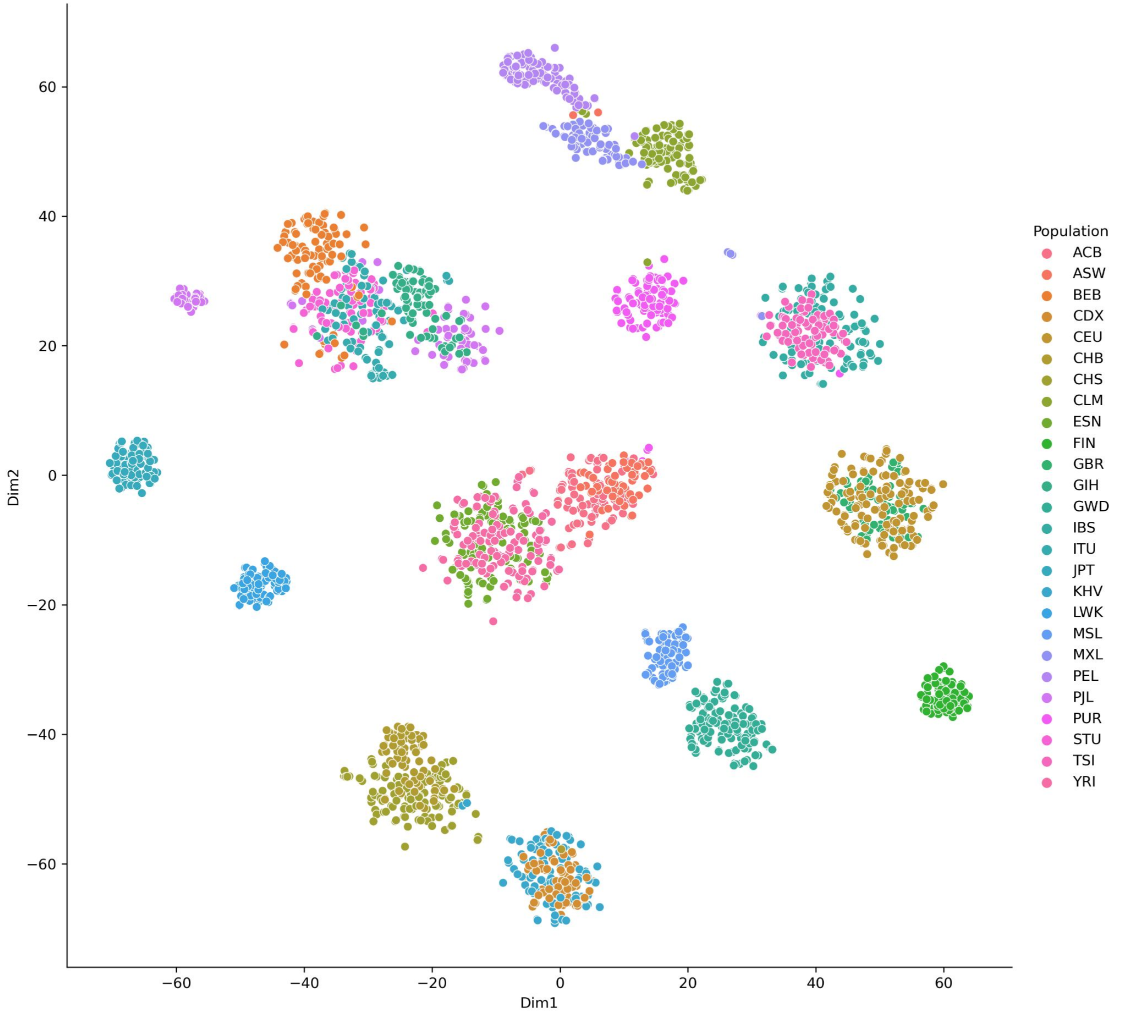
Results: Data tSNE

2561 train samples from 1000G:
50 PCs



Results: Data tSNE

2561 train samples from 1000G:
20 PCs

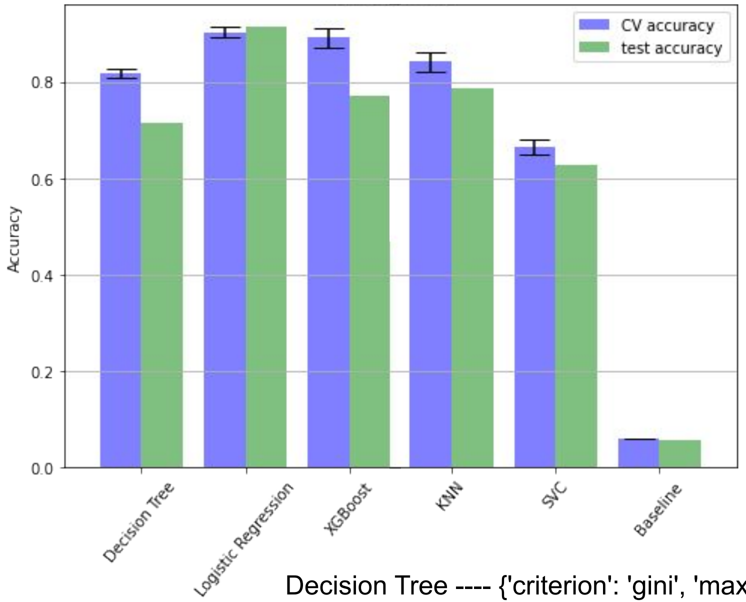


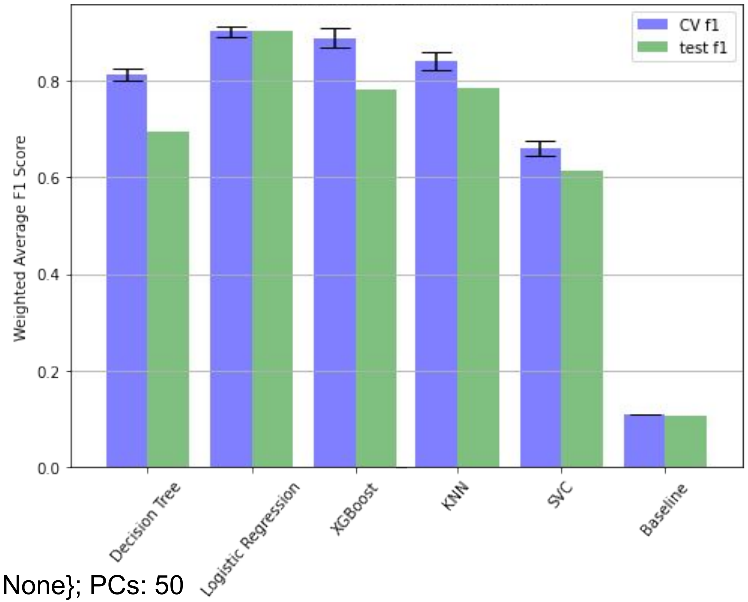
Results: Models

Accuracy

GridSearchCV, 4 folds

Weighted Average F1

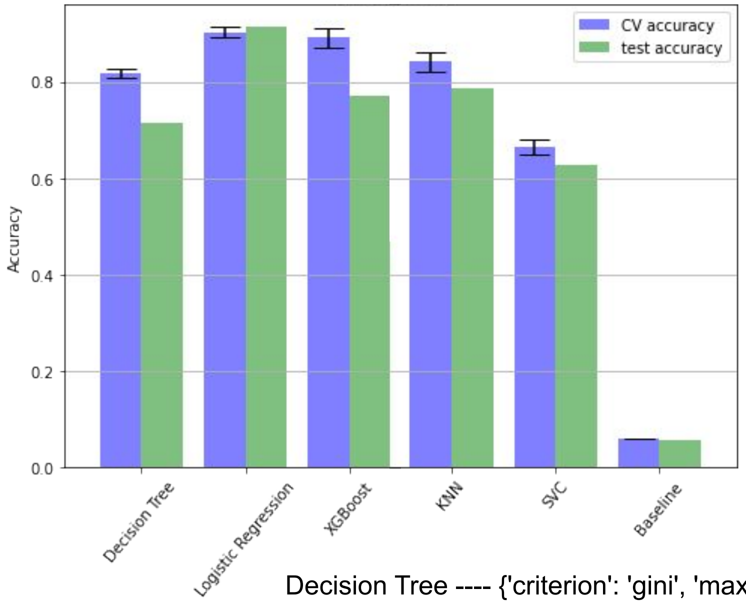


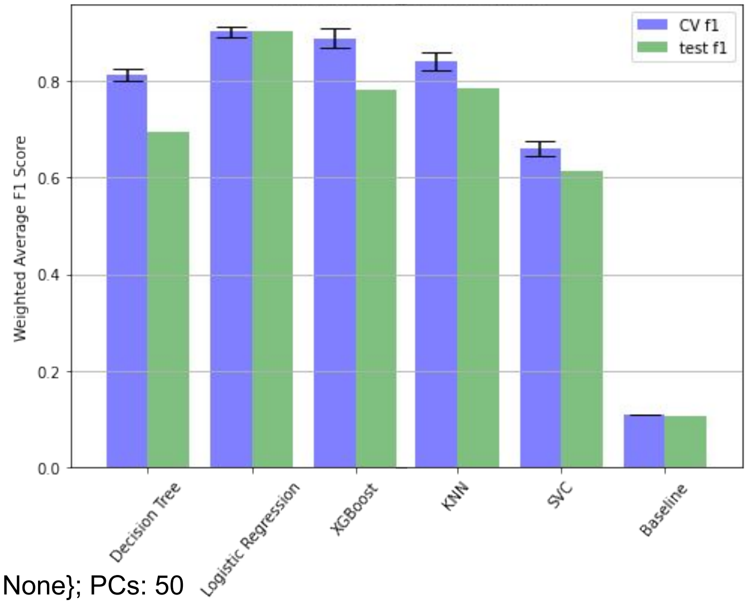


Decision Tree ---- {'criterion': 'gini', 'max_depth': None}; PCs: 50
Logistic Regression ---- {'penalty': 'l2', 'solver': 'saga'}; PCs: 1000
XGBoost ---- {'gamma': 0.5, 'max_depth': '100'}; PCs: 500
KNN ---- {'n_neighbors': 2, 'weights': 'distance'}; PCs: 50
SVC ---- {'kernel': 'poly'}; PCs: 5
Baseline ---- always predict largest class

Results: Models

Accuracy *GridSearchCV, 4 folds* Weighted Average F1





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SVC ---- {'kernel': 'poly'}; PCs: 5

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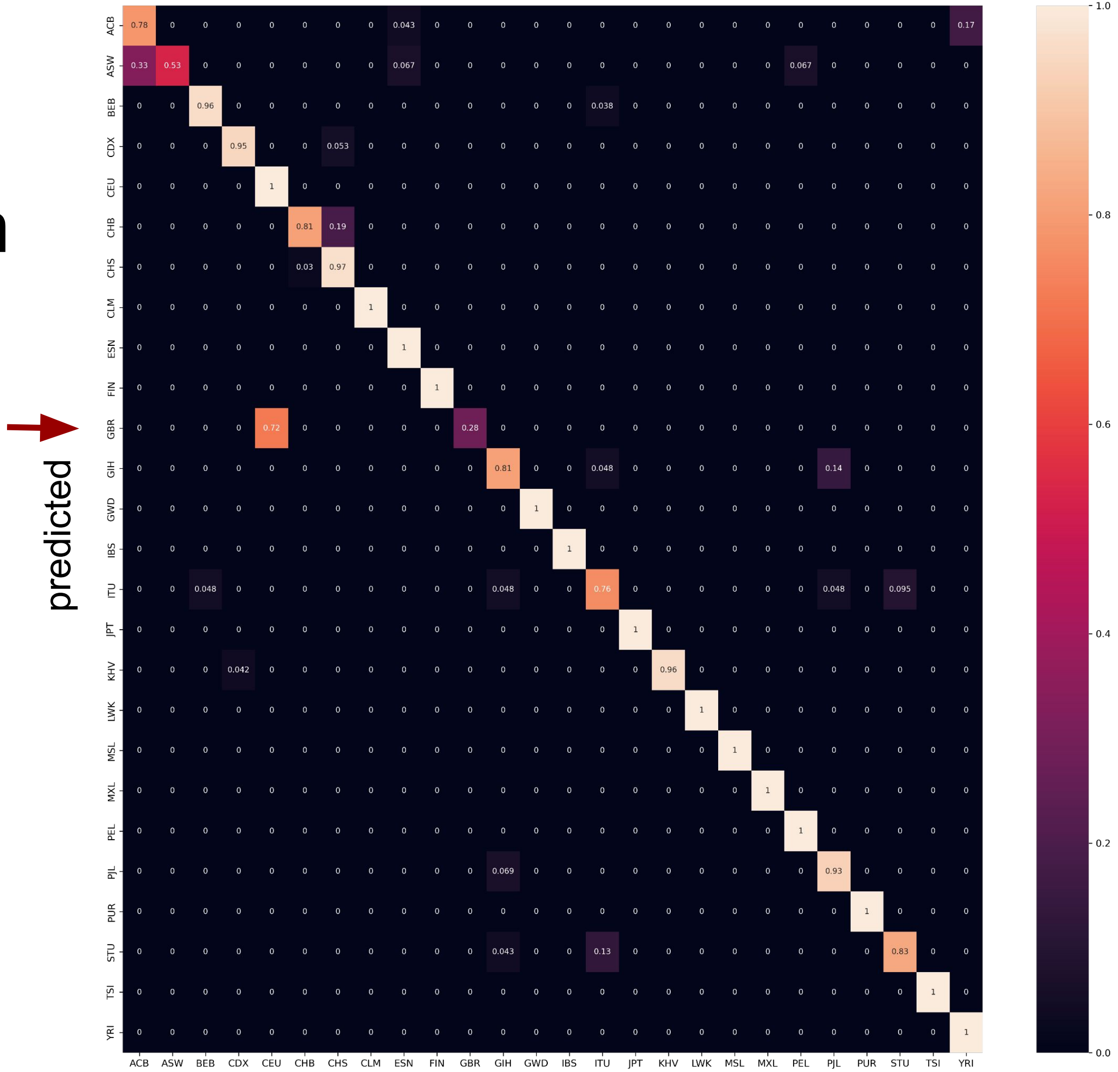
Results:
Best LogRegression Confusion Matrix



Main Lessons

- Different stages of problems complexity have their own best types of models
- In multiclassification, primary efforts can be devoted to distinguishing the most similar classes
- Ensembles have potential in multiclassifaction
- Large datasets require a large RAM amount

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- Different stages of problems complexity have their own best types of models
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Thank you for your attention!

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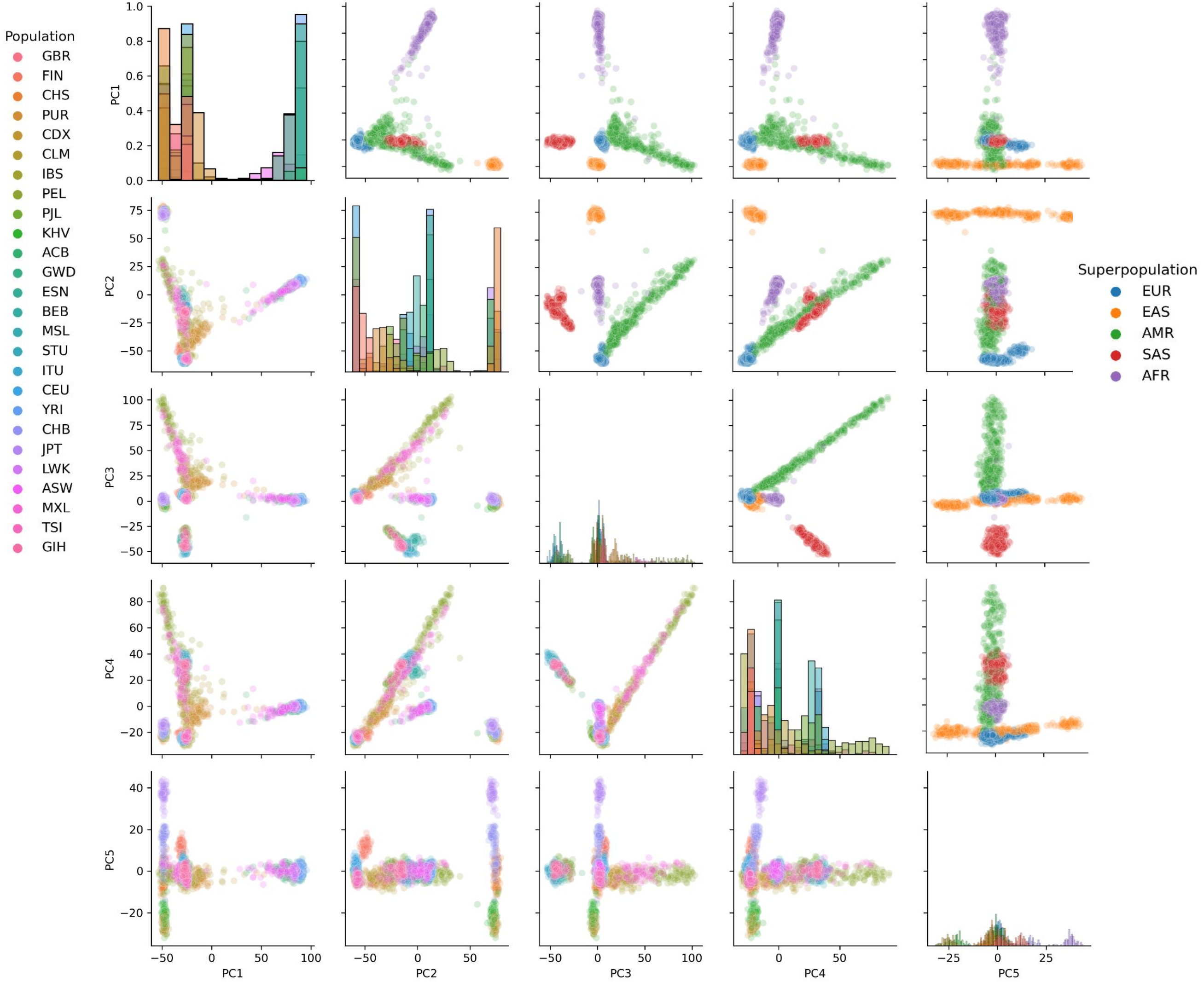
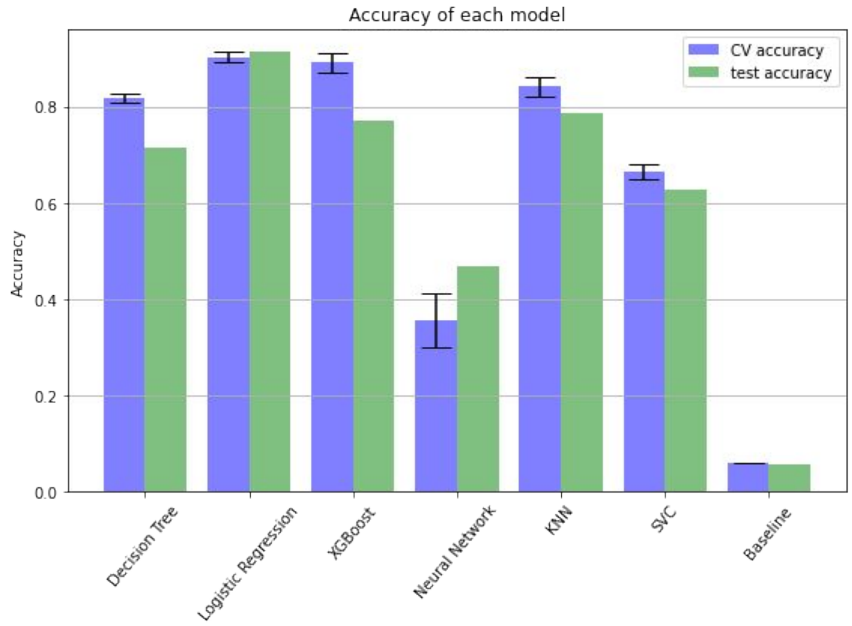
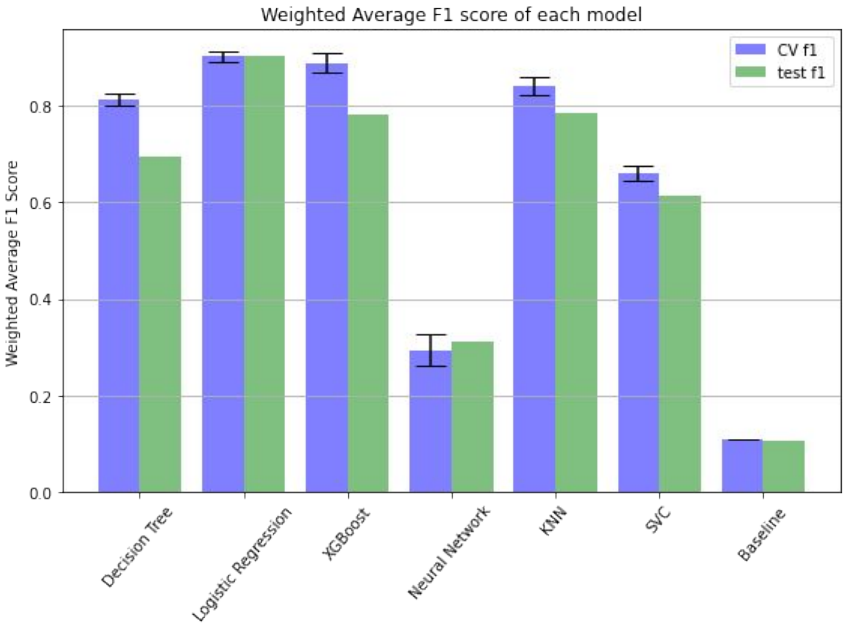
Results: Data PCs

2561 train samples from 1000G:
26 populations
5 Superpopulations



Results: Models





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XGBoost ---- {'gamma': 0.5, 'max_depth': '100'}; PCs: 500
#Neural Network ---- {'activation': 'relu', 'solver': 'adam'}; PCs: 1000
KNN ---- {'n_neighbors': 2, 'weights': 'distance'}; PCs: 50
SVC ---- {'kernel': 'poly'}; PCs: 5
Baseline ---- always predict largest class

Results: Ensemble from RF, KNN, LogR