Genetic Variant Classifications

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Genetic variants come from the changes of DNA sequences

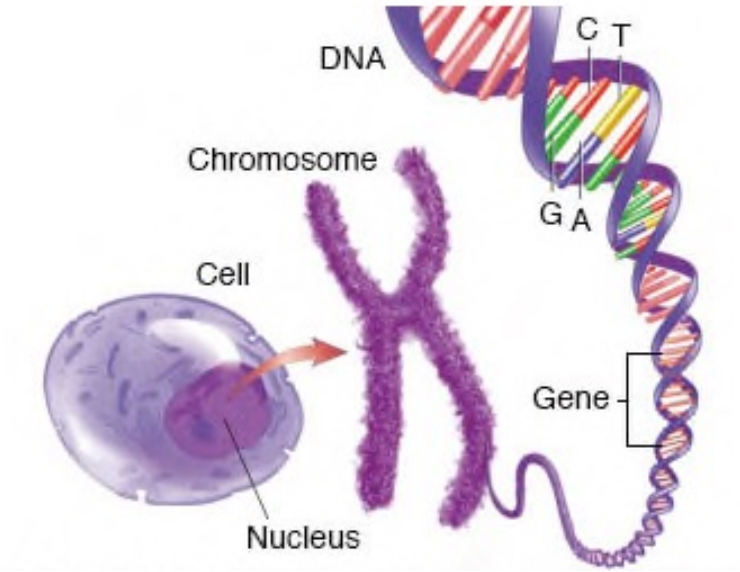
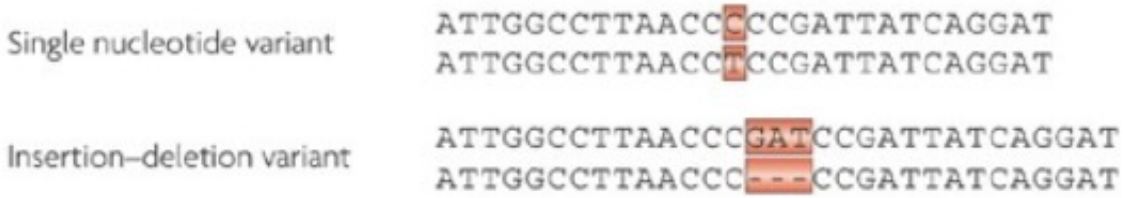
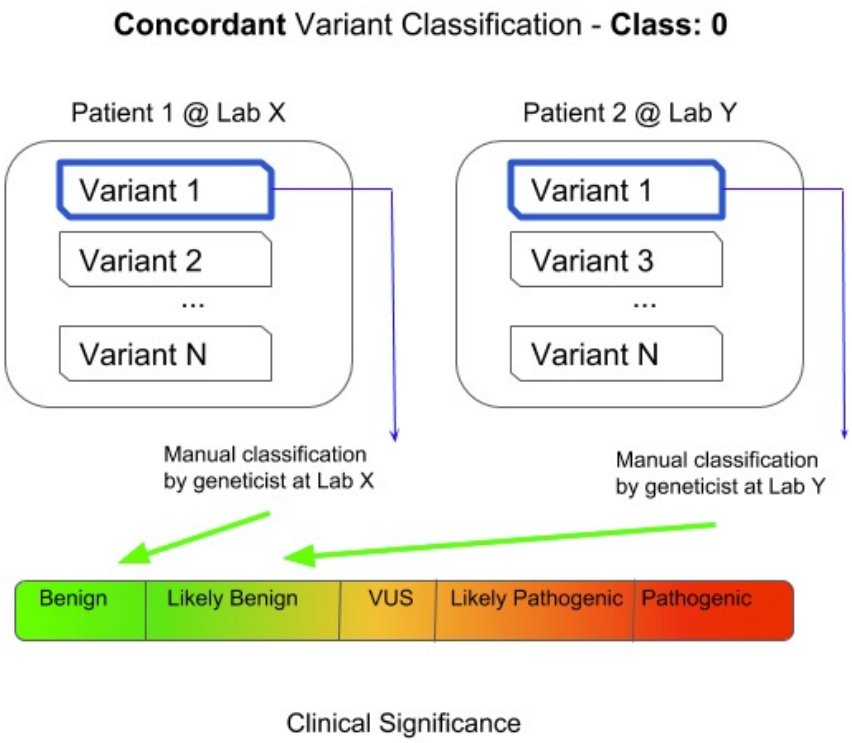
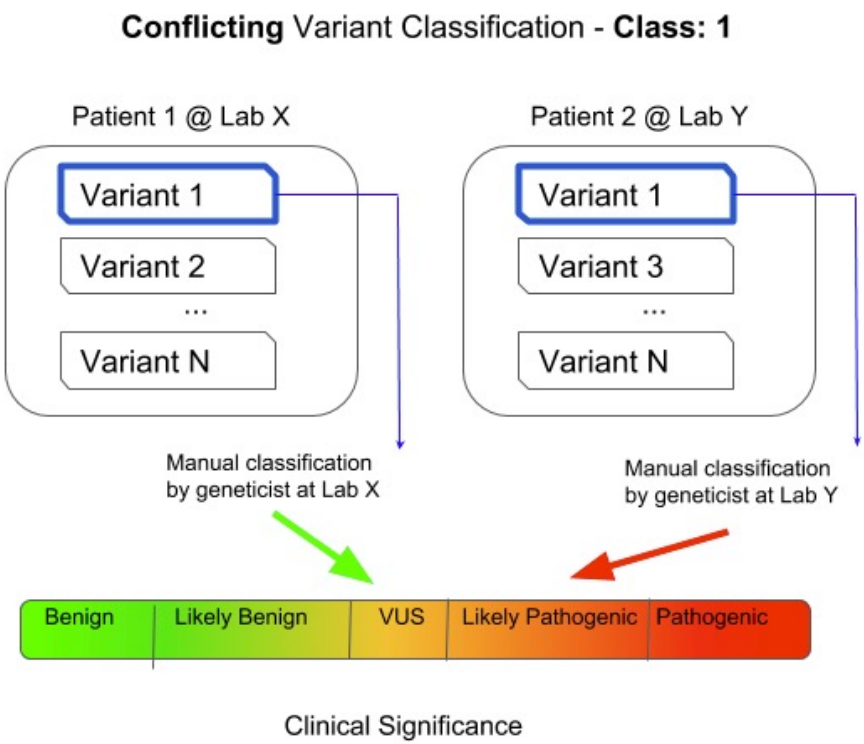


Figure 1: Classes of human genetic variants.



Genetic variants are classified manually which resulting in conflicting classification





Data exploration analysis

clinvar_conflicting.var (Kaggle)
(46 features)

Remove features

1. Redundant
2. Not correlated

```
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RangeIndex: 65188 entries, 0 to 65187
Data columns (total 9 columns):
 #   Column      Non-Null Count  Dtype
---  ------      --------------  -----
 0   CHROM       65188 non-null  object
 1   CLNVC       65188 non-null  object
 2   MC          64342 non-null  object
 3   IMPACT      65188 non-null  object
 4   SYMBOL      65172 non-null  object
 5   AF_ESP      65188 non-null  float64
 6   LoFtool     60975 non-null  float64
 7   CADD_PHRED  64096 non-null  float64
 8   CLASS       65188 non-null  int64
dtypes: float64(3), int64(1), object(5)
memory usage: 4.5+ MB
```

CLNVC - Variant Type
MC - Molecular consequence
IMPACT - the impact of the variants
SYMBOL - Gene Name

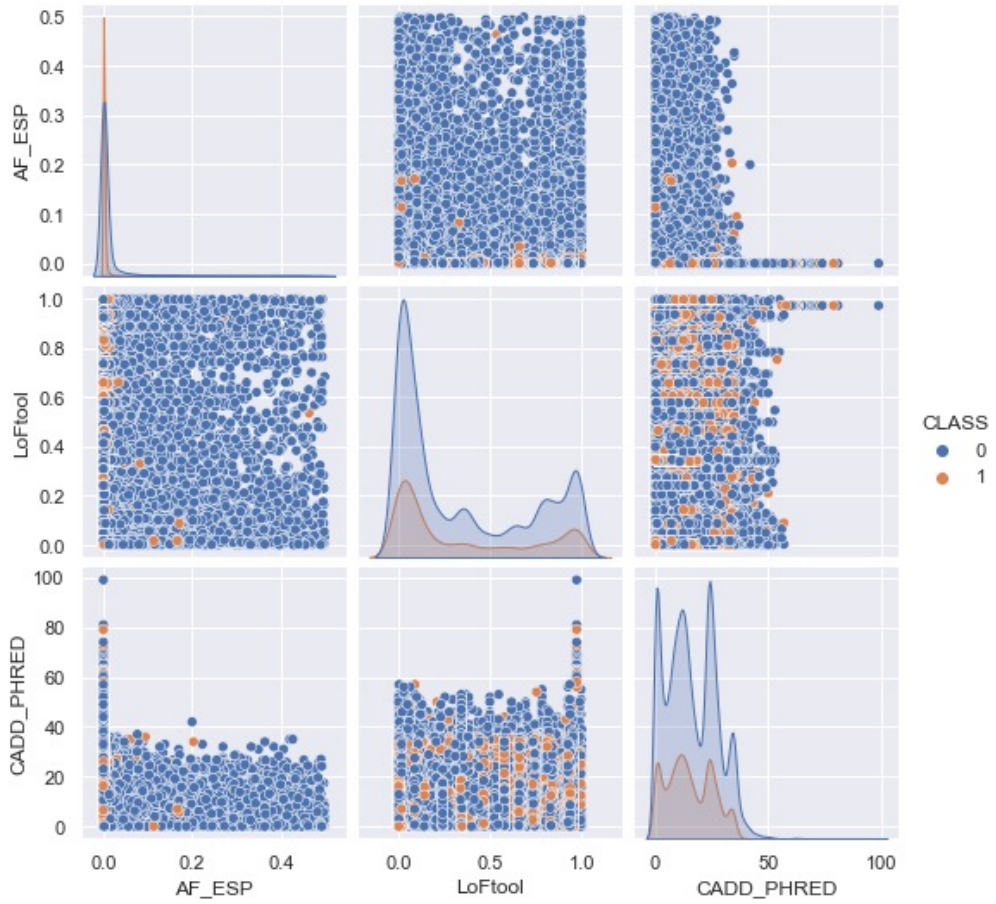
AF_ESP - Allele frequencies
LoFtool - Loss of Function tolerance score
CADD_PHRED - Scoring the deleteriousness of the variants

AF_ESP and SYMBOL are more distinguishable among 2 classes

Chi2 test for categorical features (p-value)

	CLASS
CHROM	1.407244e-05
CLASS	NaN
IMPACT	1.856664e-191
SYMBOL	6.362397e-309

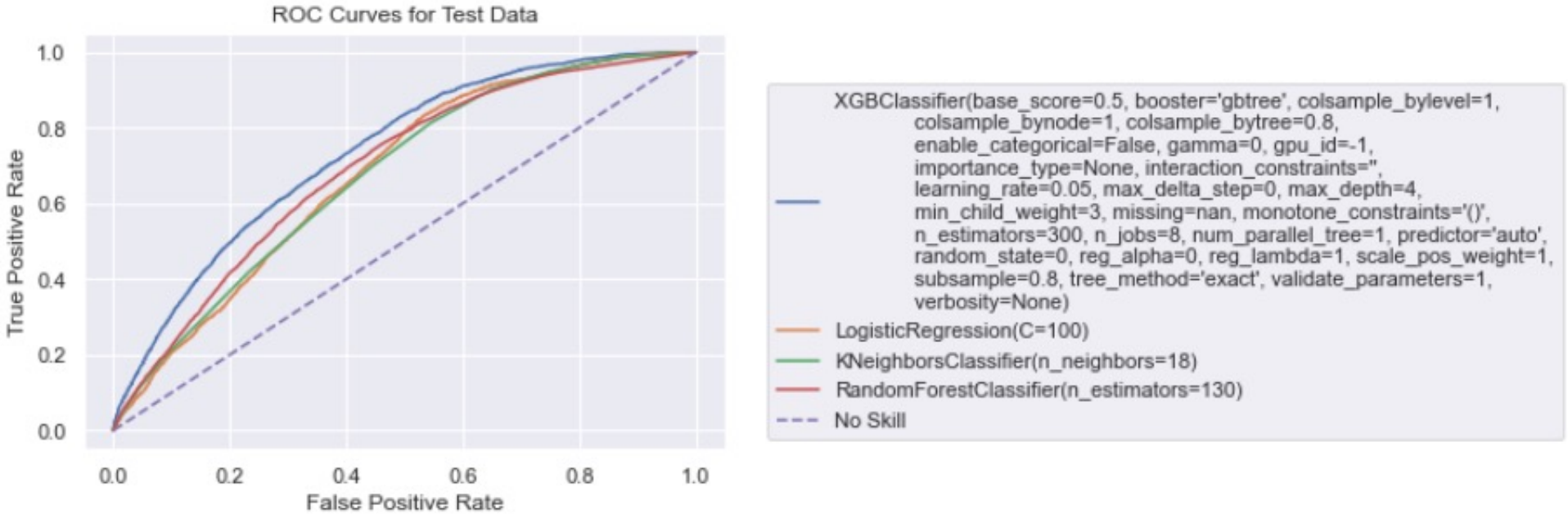
Pairplot for numerical features

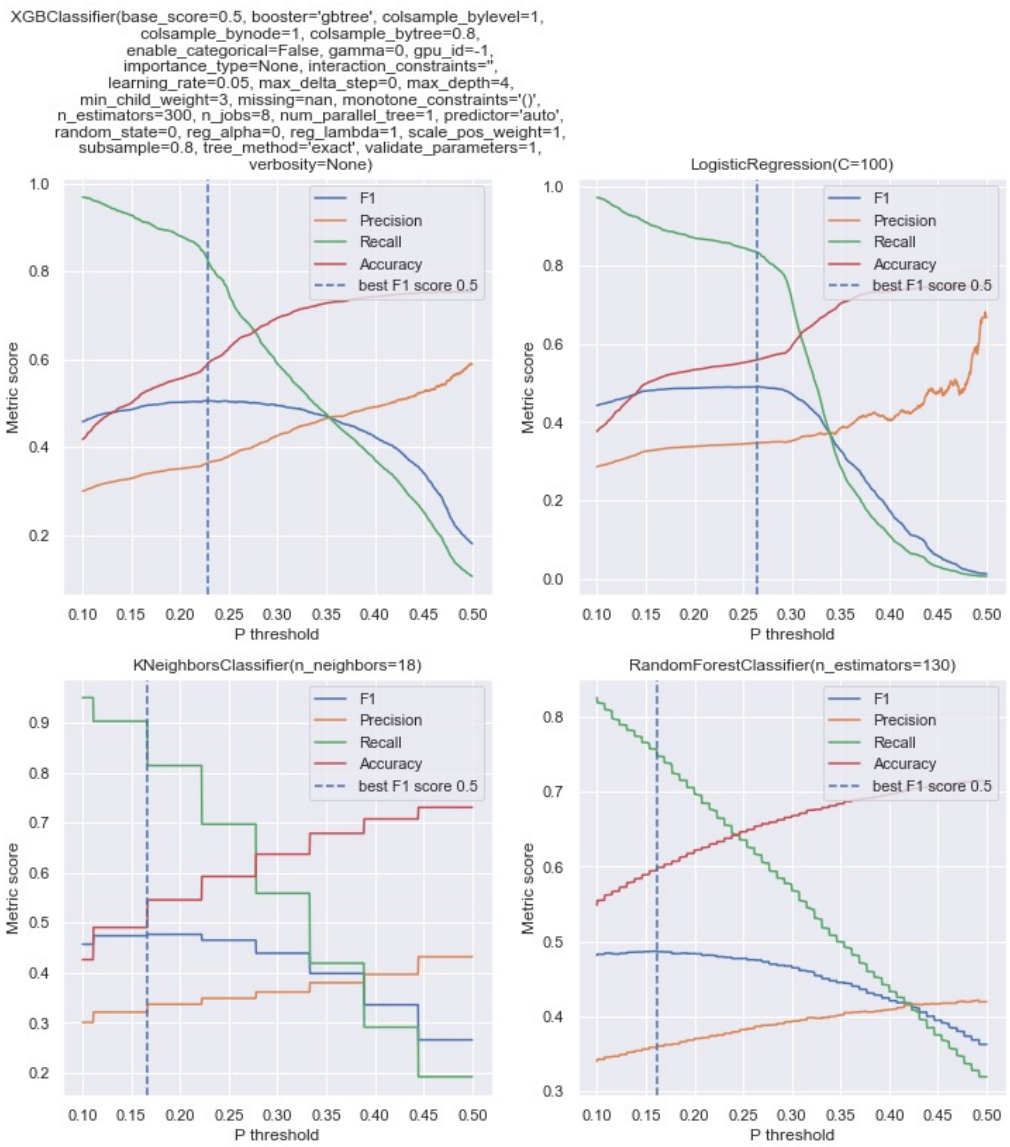


Data preprocessing

- Convert categorical feature into dummies variables
- Scale the numerical data
- Class rebalance
- Model fitting

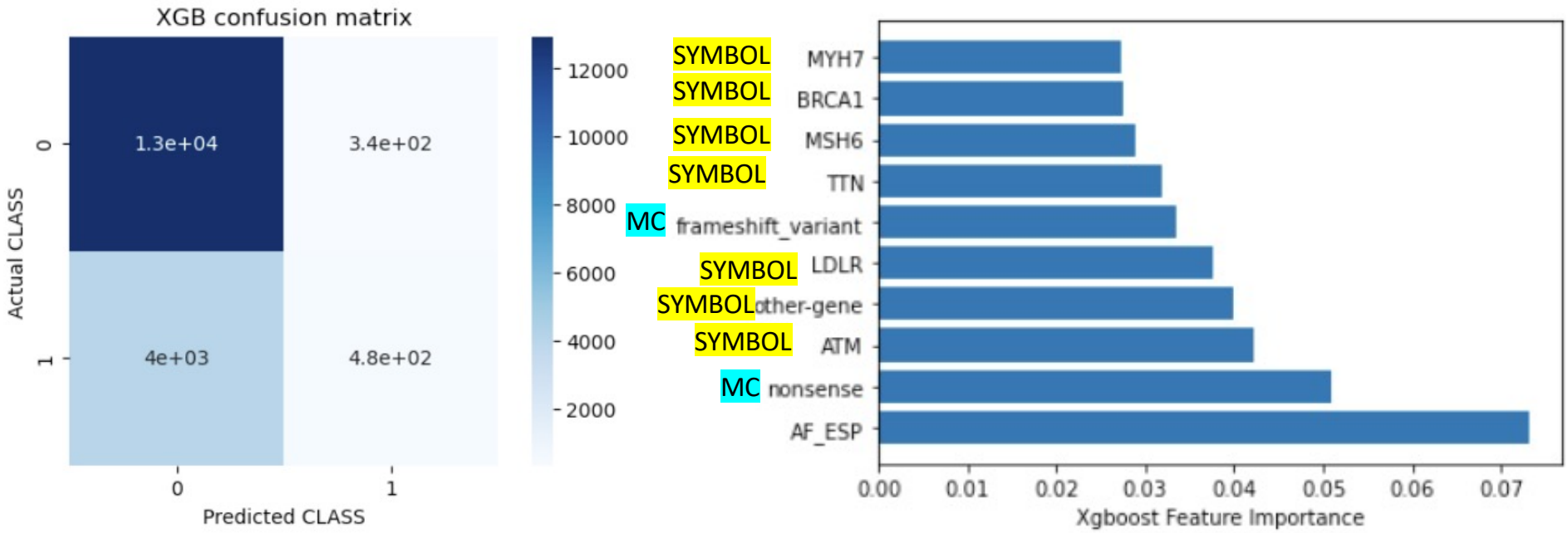
XGBoost tree has better performance for the prediction





All 4 models have similar F1 scores

AF (variant frequency), symbol (gene names) and MC(molecular consequences) are the important features



Questions