Predicting Genetic Disorders Using Decision Tree Model with Bayesian Network

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Outline

- Introduction
- Background
- Research Question
- Data Collection
- Model Building
- Model Evaluation
- Results and Discussion
- Conclusion

Genetic Disorder

Definition

- Caused by a mutation in the deoxyribonuclease acid or change in number or structure of chromosomes.
- Mutation can occur as a result of:
- environmental factors
- genetic inheritance
- chromosomal damage

Impact on Health

- Early prediction may help in early intervention and treatment
- Provide valuable information for patients and family to make informed decision about their health and lifestyle
- Opportunity for genetic counselling

Decision Tree Model

Decision trees are non-parametric supervised learning model for classification and regression.

Aim: Create a model to predict the value of a target variable based values of the predictor variables

- Pros:

- simple to understand and interpret
- trees can be visualized
- require little preparation of data
- Performs well even if assumptions are

somehow violated

- Cons:

- can cause overfitting
- can be unstable due to small variation in data
- can create biased trees if certain classes dominate

Goal

To use Decision Tree Model to predict genetic disorders from the given dataset

Data Features

- The Data consisted of a train dataset and test dataset
- It contained medical information of children with genetic disorders
- The patient's age ranges from 0 – 14 years old
- Raw Train data : 45 variables; 22083 observations
- Raw Test data: 43 variables; 9463 observations

Data was collected from 27 different

hospitals across the US

- 3 different genetic disorders

Cleaned Data: 43 variables; 10410 observations

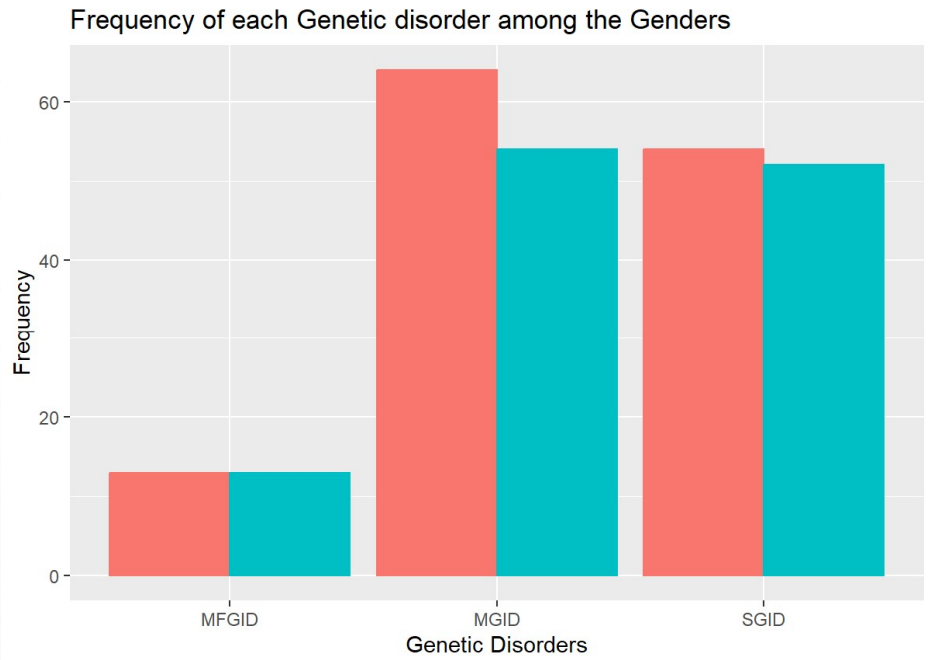
- Converted characters & integer variables into factors
- Replaced missing data with NA

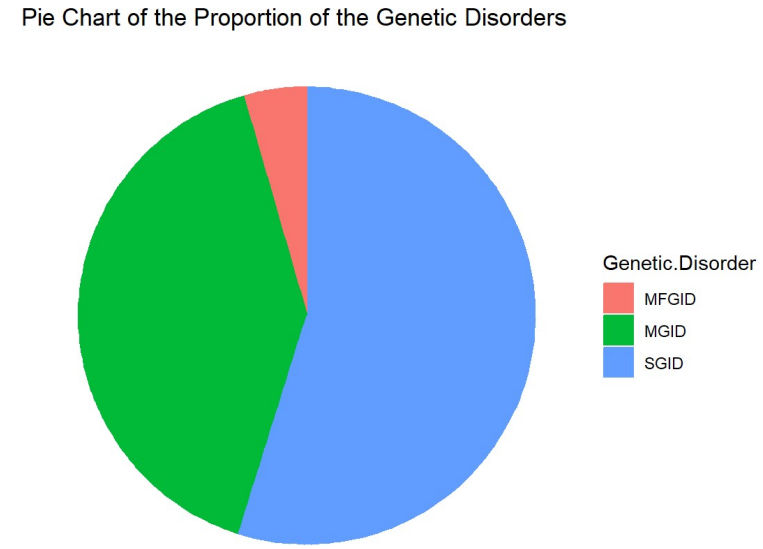
Feature Selection

- Bayesian Network – Conditional Probability Table (CPT)
- Using CPT & Bayesian network, 11 variables were selected

- Updated cleaned data: 11 variables; 10410 observations

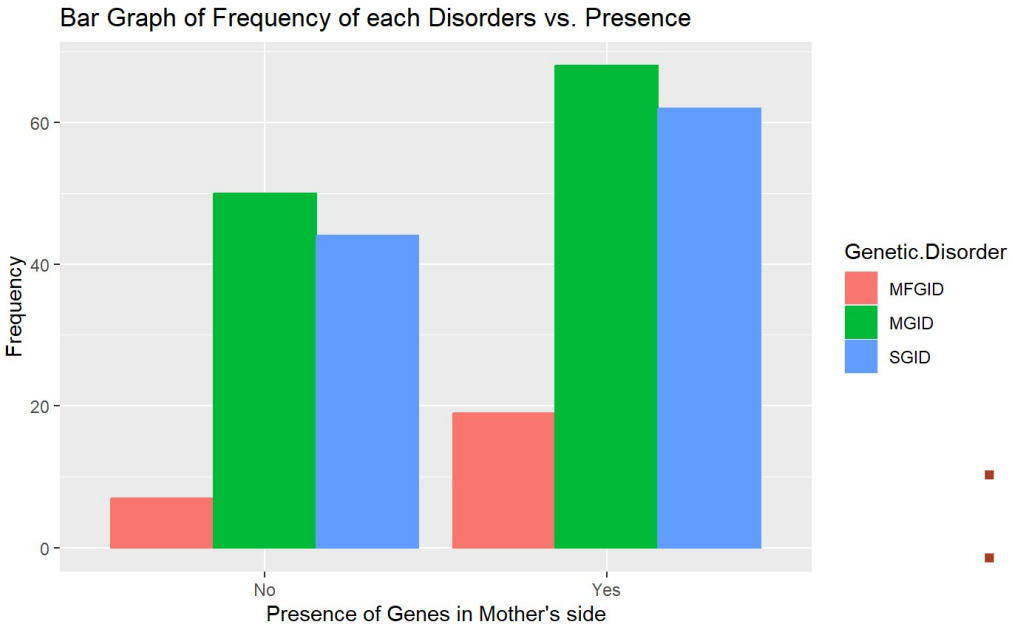
Data Visualization

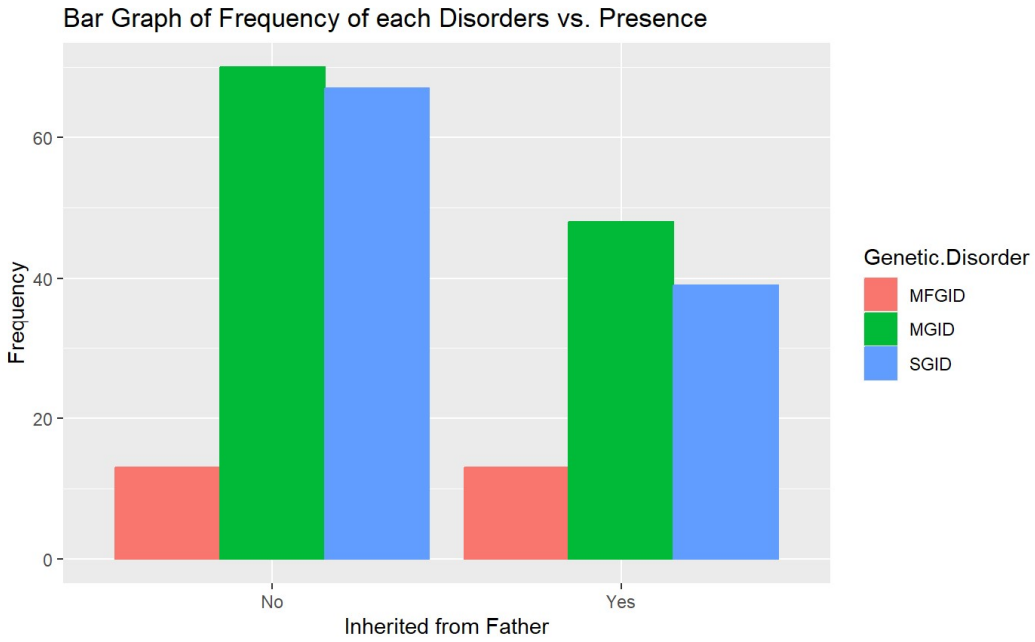




- MFGID - Multifactorial genetic inheritance disorders
- SGID - Single-gene inheritance diseases
- MGID - Mitochondrial genetic inheritance disorders

Data Visualization





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- To develop the model:
- 80% of dataset to train the model
- Train Data: 11 variable; 8376 observations.
- Subset of relevant variables for predicting the target variable
- Genes.in.mother.s.side; Inherited.from.father; Maternal.gene ; Paternal.gene; Symptom.1; Symptom.2; Symptom.3; Symptom.4; Symptom.5
- Tuned hyperparameters to avoid overfitting;

Model Building:

Decision Tree

- Learns simple decision rules based on the data features
- Each node on the tree is a data feature that plays a role in the outcome
- The leaf nodes are the outcomes -> targeted variable

- To test the model,

- Test data: 9 variables; 5318 observations

- Test Data: 11 variable; 2034 observations (20%)

- Confusion Matrix (Error Matrix) comprises of:

- Accuracy of the model
- Confidence Interval of the accuracy
- No Information Rate: Baseline accuracy
- P-value [Acc > NIR]: evaluates significance of model compared to NIR
- Kappa: agreement between predictions and actual values. 1 = perfect; 0 = most likely a chance
- MT P-value: Biased or not; small p-value indicates strong evidence of model bias

Result:

Confusion Matrix (Error Matrix)

Provides thorough analysis of true positive, true negative, false negative and false positive predictions.

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Confusion Matrix and Statistics

          Reference
Prediction MFGID MGID SGID
     MFGID   272  158  172
     MGID    258 2768 1651
     SGID    225  557  898

Overall Statistics

               Accuracy : 0.5659
                 95% CI : (0.5541, 0.5776)
    No Information Rate : 0.5005
    P-Value [Acc > NIR] : < 2.2e-16

                  Kappa : 0.2246

 Mcnemar's Test P-Value : < 2.2e-16

Statistics by Class:

                     Class: MFGID Class: MGID Class: SGID
Sensitivity               0.36026      0.7947      0.3300
Specificity               0.94681      0.4508      0.8155
Pos Pred Value            0.45183      0.5918      0.5345
Neg Pred Value            0.92402      0.6867      0.6547
Prevalence                0.10849      0.5005      0.3910
Detection Rate            0.03909      0.3978      0.1290
Detection Prevalence      0.08651      0.6721      0.2414
Balanced Accuracy         0.65354      0.6228      0.5728
```

Confusion Matrix

Sensitivity: proportion of actual (+) that are correctly identified

Specificity: : proportion of actual (-) that are correctly identified

Pos Pred Value: proportion of (+) predictions that are correct

Neg Pred Value: proportion of (-) predictions that are correct

Prevalence: proportion that belongs to (+ class

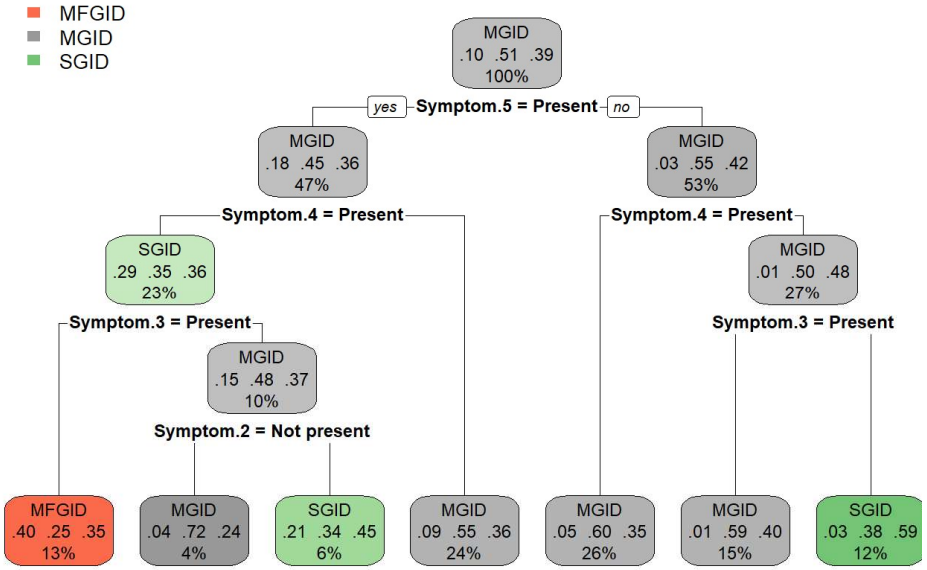
Detection Rate: # of true (+) predictions by model/ total # of instances

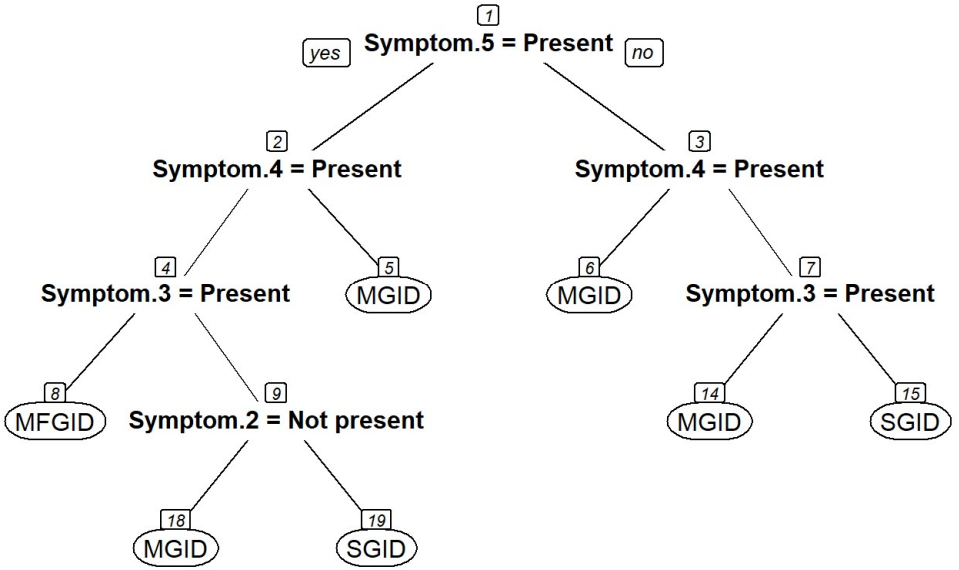
Detection Prevalence: # of (+) predictions by model/ total # of instances

Balanced Accuracy: average of sensitivity and specificity

Decision Tree of Test Dataset

*** Optimized decision tree**





Conclusion

- Accuracy can be improved by adding more highly correlated variables to create the decision tree.

1

- Accuracy can be improved by transforming existing features to increase the performance of the model

2

- Accuracy can be improved by handling imbalanced classes.

3

Citations

- Kumar, A. (2021). Predict the genetic disorders dataset-of genomes. www.kaggle.com. https://www.kaggle.com/datasets/aibuzz/predict-the-genetic-disorders-datasetof-genomes?rvi=1
- Decision Tree Models in Python — Build, Visualize, Evaluate | by Mustafa Adel Amer | Towards Data Science