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NLP and ML in Medicine for the Classification of Genetic Mutations in Cancerous Tumors

Much has been said in recent years about how precision medicine, specifically genetic testing, will disrupt the treatment of diseases such as cancer. However, this is still only partially happening due to the enormous amount of manual work still required. In this project, we aim to take personalized medicine to its full potential.

Once sequenced, a cancerous tumor can have thousands of genetic mutations. The challenge is to distinguish the mutations that contribute to tumor growth. Currently, the interpretation of genetic mutations is being done manually. This is a time-consuming task where a clinical pathologist has to manually review and classify each genetic mutation based on evidence from text-based clinical literature.

The Memorial Sloan Kettering Cancer Center has provided an expert-annotated knowledge base where world-class researchers and oncologists have manually annotated thousands of mutations. In this project, we will develop a Machine Learning algorithm that, using this knowledge base as a reference, will automatically classify genetic variations.

The complete dataset can be found at:

[https://www.kaggle.com/c/msk-redefining-cancer-treatment/data]

Data Description

There are 9 different classes in which a genetic mutation can be classified.

The training and test data consist of 2 different files. The first one (train/test_variants) provides information about the genetic mutations, while the second one (train/test_text) provides the clinical evidence that experts use to classify the genetic mutations.

The two files are linked by the ID column. For example, the genetic mutation located on the row with ID=15 in the variants file was classified using the clinical evidence from the row with ID=15 in the text file.

To make things more challenging, some of the test data samples were computer-generated to prevent manual labeling. We should ignore the samples that were generated by the computer.

File Details

- **training_variants**: The descriptions of genetic mutations used for training are separated by commas. The fields are **ID** (used to link with the evidence dataset), **Gene** (the gene where

the genetic mutation is located), **Variation** (the amino acid change of this mutation), **Class** (the class from 1-9 to which the genetic change has been classified).

- **training_text**: The content of the clinical evidence used to classify the genetic mutations is delimited by double bars (||). The fields are **ID** (used to link with the variants dataset) and **Text** (the clinical evidence used to classify the genetic mutations).
- **test_variants**: The descriptions of genetic mutations used for testing are separated by commas. The fields are **ID** (used to link with the evidence dataset), **Gene** (the gene where the genetic mutation is located), **Variation** (the amino acid change of this mutation).
- **test_text:** The content of the clinical evidence used to classify the genetic mutations is delimited by double bars (||). The fields are **ID** (used to link with the variants dataset) and **Text** (the clinical evidence used to classify the genetic mutations).

Loading the Packages

```
from platform import python_version
print('A Versão da Linguagem Python utilizada neste projeto é ', python_version())
```

```
A Versão da Linguagem Python utilizada neste projeto é  3.9.16
```

```
# Manipuluação e Vizualização de Dados
import pandas as pd
import numpy as np
import matplotlib.pyplot as plt
import matplotlib.gridspec as gridspec
import string
import plotly.express as px
import plotly.graph_objects as go
from plotly.subplots import make_subplots
```

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```
# Pré - Processamento de Texto
import re
import nltk
import sklearn
import wordcloud
from wordcloud import WordCloud, STOPWORDS
from nltk.tokenize import word_tokenize
from nltk.corpus import stopwords
from sklearn.feature_extraction.text import TfidfVectorizer
from sklearn.preprocessing import Normalizer
from sklearn.preprocessing import LabelEncoder
from imblearn.over_sampling import ADASYN
from sklearn.model_selection import train_test_split

# Modelos de Machine Learning
from sklearn.naive_bayes import MultinomialNB
from sklearn.linear_model import SGDClassifier
from sklearn.neighbors import KNeighborsClassifier
from sklearn.ensemble import RandomForestClassifier

# Métricas dos Modelos
from sklearn.metrics import ConfusionMatrixDisplay, confusion_matrix
from sklearn.metrics import roc_auc_score
from sklearn.metrics import log_loss

# Retirando Avisos
import warnings
warnings.filterwarnings('ignore')
```

```
# Registrando as Versões dos Pacotes
%reload_ext watermark
%watermark -a "Thiago Bulgarelli" --iversions
```

```
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matplotlib: 3.6.2
pandas    : 1.5.2
wordcloud : 1.8.2.2
nltk      : 3.7
re        : 2.2.1
numpy     : 1.23.5
plotly    : 5.9.0
sklearn   : 1.0.2
```

Loading the Data

```
# Carregando os Datasets
train_Ev = pd.read_csv('dados/training_text', sep="\|\|", engine='python')
train_Var = pd.read_csv('dados/training_variants', sep=',')
test_Ev = pd.read_csv('dados/test_text', sep='\|\|',  engine='python')
test_Var = pd.read_csv('dados/test_variants', sep=',')
```

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```
# Visualizando as Evidências de Treino
print(train_Ev.info())
train_Ev.head()
```

```
<class 'pandas.core.frame.DataFrame'>
RangeIndex: 3321 entries, 0 to 3320
Data columns (total 2 columns):
 #   Column  Non-Null Count  Dtype 
---  ------  --------------  ----- 
 0   ID      3321 non-null   int64 
 1   Text    3316 non-null   object
dtypes: int64(1), object(1)
memory usage: 52.0+ KB
None
```

	ID	Text
0	0	Cyclin-dependent kinases (CDKs) regulate a var...
1	1	Abstract Background Non-small cell lung canc...
2	2	Abstract Background Non-small cell lung canc...
3	3	Recent evidence has demonstrated that acquired...
4	4	Oncogenic mutations in the monomeric Casitas B...

```
# Visualizando as Variantes de Treino
print(train_Var.info())
train_Var.head()
```

```
<class 'pandas.core.frame.DataFrame'>
RangeIndex: 3321 entries, 0 to 3320
Data columns (total 4 columns):
 #   Column     Non-Null Count  Dtype 
---  ------     --------------  ----- 
 0   ID         3321 non-null   int64 
 1   Gene       3321 non-null   object
 2   Variation  3321 non-null   object
 3   Class      3321 non-null   int64 
dtypes: int64(2), object(2)
memory usage: 103.9+ KB
None
```

	ID	Gene	Variation	Class
0	0	FAM58A	Truncating Mutations	1
1	1	CBL	W802*	2
2	2	CBL	Q249E	2
3	3	CBL	N454D	3
4	4	CBL	L399V	4

```
# Visualizando as Evidências de Teste
print(test_Ev.info())
test_Ev.head()
```

```
<class 'pandas.core.frame.DataFrame'>
RangeIndex: 5668 entries, 0 to 5667
Data columns (total 2 columns):
 #   Column  Non-Null Count  Dtype 
---  ------  --------------  ----- 
 0   ID      5668 non-null   int64 
 1   Text    5667 non-null   object
dtypes: int64(1), object(1)
memory usage: 88.7+ KB
None
```

	ID	Text
0	0	2. This mutation resulted in a myeloproliferat...
1	1	Abstract The Large Tumor Suppressor 1 (LATS1)...
2	2	Vascular endothelial growth factor receptor (V...
3	3	Inflammatory myofibroblastic tumor (IMT) is a ...
4	4	Abstract Retinoblastoma is a pediatric retina...

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```
# Visualizando as Variantes de Teste
print(test_Var.info())
test_Var.head()
```

```
<class 'pandas.core.frame.DataFrame'>
RangeIndex: 5668 entries, 0 to 5667
Data columns (total 3 columns):
 #   Column     Non-Null Count  Dtype 
---  ------     --------------  ----- 
 0   ID         5668 non-null   int64 
 1   Gene       5668 non-null   object
 2   Variation  5668 non-null   object
dtypes: int64(1), object(2)
memory usage: 133.0+ KB
None
```

	ID	Gene	Variation
0	0	ACSL4	R570S
1	1	NAGLU	P521L
2	2	PAH	L333F
3	3	ING1	A148D
4	4	TMEM216	G77A

```
# Unindo o Dataset de Treino para Limpeza e Transformação
df = train_Ev.merge(train_Var, how='right', on='ID')

# Visualizando Dataset de Treino Completo
print(df.info())
df.head()
```

```
<class 'pandas.core.frame.DataFrame'>
Int64Index: 3321 entries, 0 to 3320
Data columns (total 5 columns):
 #   Column     Non-Null Count  Dtype 
---  ------     --------------  ----- 
 0   ID         3321 non-null   int64 
 1   Text       3316 non-null   object
 2   Gene       3321 non-null   object
 3   Variation  3321 non-null   object
 4   Class      3321 non-null   int64 
dtypes: int64(2), object(3)
memory usage: 155.7+ KB
None
```

	ID	Text	Gene	Variation	Class
0	0	Cyclin-dependent kinases (CDKs) regulate a var...	FAM58A	Truncating Mutations	1
1	1	Abstract Background Non-small cell lung canc...	CBL	W802*	2
2	2	Abstract Background Non-small cell lung canc...	CBL	Q249E	2
3	3	Recent evidence has demonstrated that acquired...	CBL	N454D	3
4	4	Oncogenic mutations in the monomeric Casitas B...	CBL	L399V	4

```
# Unindo o Dataset de Teste para deixar aos moldes do Dataset de Treino
dft = test_Ev.merge(test_Var, how='right', on='ID')

# Visualizando Dataset de Teste Completo
print(dft.info())
dft.head()
```

```
<class 'pandas.core.frame.DataFrame'>
Int64Index: 5668 entries, 0 to 5667
Data columns (total 4 columns):
 #   Column     Non-Null Count  Dtype 
---  ------     --------------  ----- 
 0   ID         5668 non-null   int64 
 1   Text       5667 non-null   object
 2   Gene       5668 non-null   object
 3   Variation  5668 non-null   object
dtypes: int64(1), object(3)
memory usage: 221.4+ KB
None
```

	ID	Text	Gene	Variation
0	0	2. This mutation resulted in a myeloproliferat...	ACSL4	R570S
1	1	Abstract The Large Tumor Suppressor 1 (LATS1)...	NAGLU	P521L
2	2	Vascular endothelial growth factor receptor (V...	PAH	L333F
3	3	Inflammatory myofibroblastic tumor (IMT) is a ...	ING1	A148D
4	4	Abstract Retinoblastoma is a pediatric retina...	TMEM216	G77A

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Data Cleaning and Transformation

Let's apply Text Processing techniques to keep only the most important words from each observation, removing special characters or unnecessary punctuation that does not contribute to the understanding of the context.

We will also check for missing data and determine if they need to be replaced or eliminated from our analysis.

```
# Analisando as Informações Básicas do Dataset
df.info()
```

```
<class 'pandas.core.frame.DataFrame'>
Int64Index: 3321 entries, 0 to 3320
Data columns (total 5 columns):
 #   Column     Non-Null Count  Dtype 
---  ------     --------------  ----- 
 0   ID         3321 non-null   int64 
 1   Text       3316 non-null   object
 2   Gene       3321 non-null   object
 3   Variation  3321 non-null   object
 4   Class      3321 non-null   int64 
dtypes: int64(2), object(3)
memory usage: 155.7+ KB
```

```
# Verificando dados Ausentes
df.isna().sum()
```

```
ID           0
Text         5
Gene         0
Variation    0
Class        0
dtype: int64
```

```
# Excluindo as Observações sem Informação
df.dropna(inplace=True)
print(df.isna().sum())
```

```
ID           0
Text         0
Gene         0
Variation    0
Class        0
dtype: int64
```

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```
# Carregando as StopWords em Ingles
nltk.download('stopwords')
SW = set(stopwords.words('english'))
```

```
[nltk_data] Downloading package stopwords to
[nltk_data]     C:\Users\Bugath\AppData\Roaming\nltk_data...
[nltk_data]   Package stopwords is already up-to-date!
```

```
# Função de Processar o texto
def text_process(text):
    text = str(text)

    # Transforma o Texto em Letras Minúsculas
    text = text.lower()

    # Remove as Pontuações
    text = re.sub('[%s]' % re.escape(string.punctuation), ' ',
                  text)

    # Remove as Tags HTML
    text = re.sub('<.*?>+', ' ', text)

    # Remove Caracteres Especiais
    text = re.sub('[^a-zA-Z0-9\n]', ' ', text)

    # Remove Multiplos Espaços
    text = re.sub(r'\s+', ' ', text)

    # Tokenização do Texto
    text_tokens = word_tokenize(text)

    # Separa as Sentenças em Palavras
    text = text.split()

    #Remove as StopWords
    tw = [word for word in text_tokens if word is not SW]
    text = (' ').join(tw)

    return text
```

```
# Aplicando a Função text_process no Dataset de Treino
df['Text'] = df['Text'].apply(text_process)
print(df.info())
print(df.isnull().sum())
df.head()
```

```
<class 'pandas.core.frame.DataFrame'>
Int64Index: 3316 entries, 0 to 3320
Data columns (total 5 columns):
 #   Column     Non-Null Count  Dtype
---  ------     --------------  -----
 0   ID         3316 non-null   int64
 1   Text       3316 non-null   object
 2   Gene       3316 non-null   object
 3   Variation  3316 non-null   object
 4   Class      3316 non-null   int64
dtypes: int64(2), object(3)
memory usage: 155.4+ KB
None
ID           0
Text         0
Gene         0
Variation    0
Class        0
dtype: int64
```

	ID	Text	Gene	Variation	Class
0	0	cyclin dependent kinases cdks regulate a varie...	FAM58A	Truncating Mutations	1
1	1	abstract background non small cell lung cancer...	CBL	W802*	2
2	2	abstract background non small cell lung cancer...	CBL	Q249E	2
3	3	recent evidence has demonstrated that acquired...	CBL	N454D	3
4	4	oncogenic mutations in the monomeric casitas b...	CBL	L399V	4

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Clean and organized data! We are ready to gain a better understanding of the data and proceed with our analysis.

Exploratory Data Analysis - EDA

Our objective here is to explore the data and uncover hidden insights in the relationships between variables.

```
# Criando uma Cópia do Dataset Original
dfEDA = df.copy()
```

Variable Gene

```
# Analisando os dados Unicos
dfEDA['Gene'].value_counts()
```

```
BRCA1     264
TP53      163
EGFR      141
PTEN      126
BRCA2     125
          ...
RICTOR      1
PIK3R3      1
PPM1D       1
WHSC1       1
FAM58A      1
Name: Gene, Length: 262, dtype: int64
```

```
# Distribuição dos tipos de Genes
fig = px.bar(dfEDA['Gene'].value_counts(), title='Frequência dos Tipos de Genes')
fig.update_xaxes(title_text = 'Genes')
fig.update_yaxes(title_text = 'Contagem de Observações')
```

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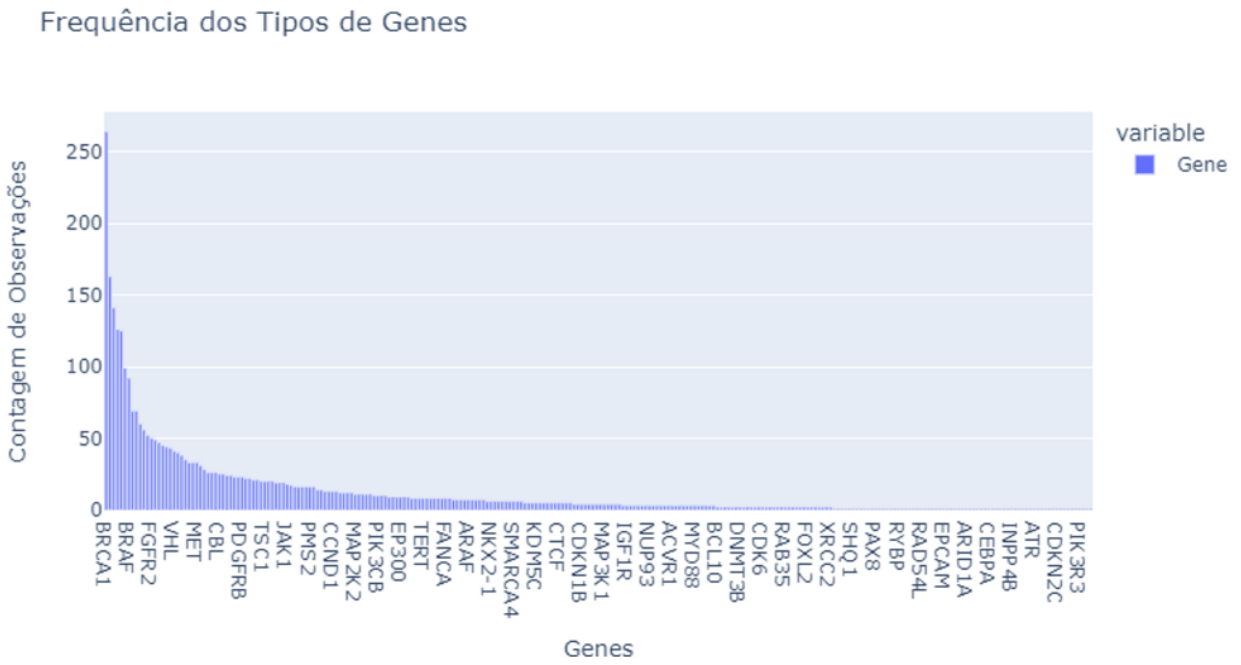
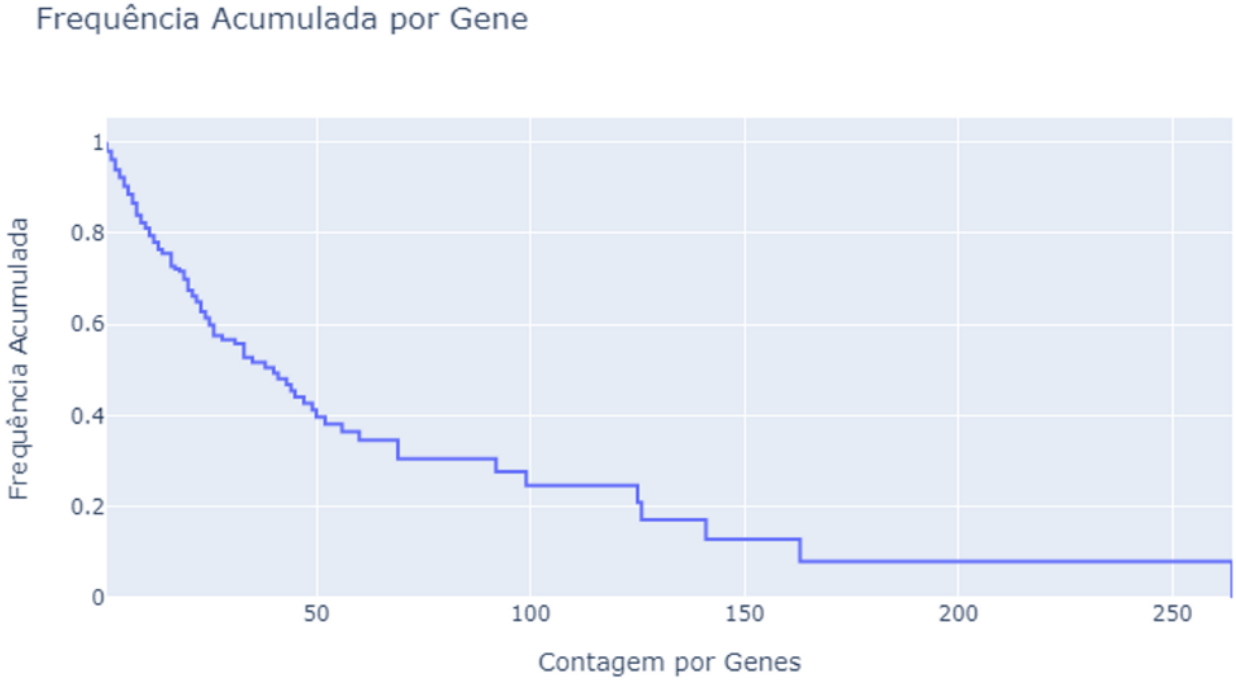



```
# Frequência Acumulada por Tipo de Gene
y = dfEDA['Gene'].value_counts(normalize=True)
fig = px.ecdf(x = dfEDA['Gene'].value_counts(),
              y = y,
              ecdfnorm=None,
              ecdfmode='complementary',
              title='Frequência Acumulada por Gene')
fig.update_xaxes(title_text = 'Contagem por Genes')
fig.update_yaxes(title_text = 'Frequência Acumulada')
```



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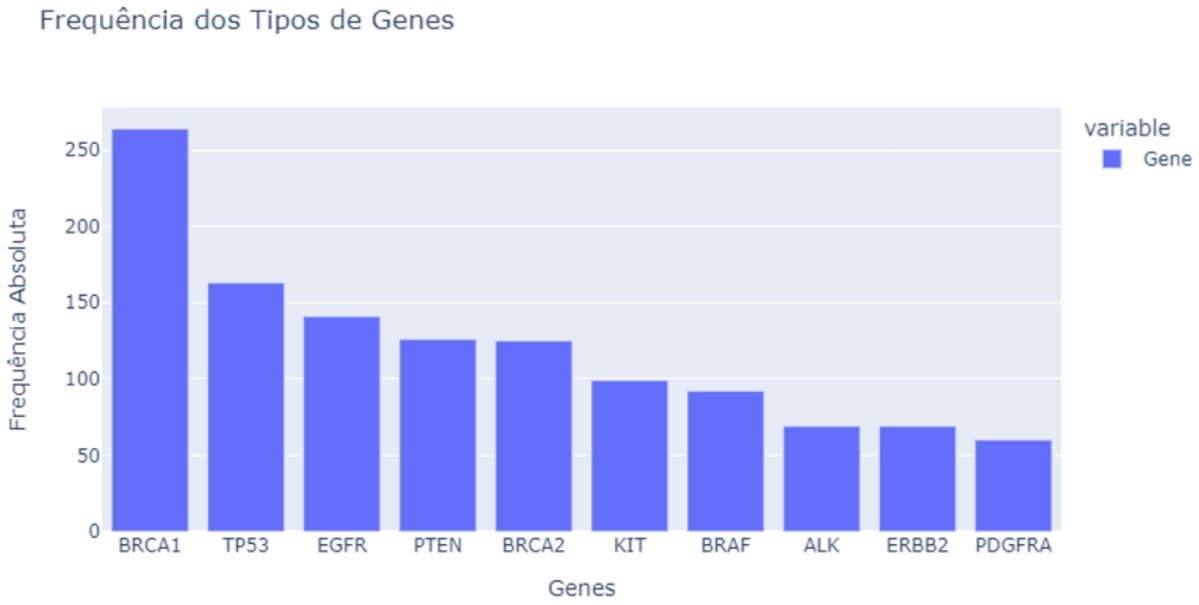
Insight → Based on the provided statement, we can say that the genes with more than 16 occurrences in our dataset account for 75% of the classified cases.

```
# Frequência Absoluta dos tipos de Genes 10+
dfEDA1 = dfEDA['Gene'].value_counts().copy()
dfEDA1 = dfEDA1.head(10)
fig = px.bar(dfEDA1, title='Frequência dos Tipos de Genes')
fig.update_xaxes(title_text = 'Genes')
fig.update_yaxes(title_text = 'Frequência Absoluta')
```



Insight → The most common genes, which are BRCA1, TP53, EGFR, PTEN, BRCA2, KIT, BRAF, ALK, ERBB2, and PDGFRA, account for 34% of our observations.

Variable Variation

```
# Analisando os dados Unicos
dfEDA2 = dfEDA.copy()
dfEDA2['Variation'].value_counts()
```

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```
Truncating Mutations    92
Deletion                74
Amplification           70
Fusions                 34
Overexpression           6
                        ..
H1094R                   1
M1250T                   1
PTPRZ1-MET Fusion        1
H1106D                   1
K83E                     1
Name: Variation, Length: 2993, dtype: int64
```

```
# Frequência Acumulada por Tipo de Variação
y = dfEDA2['Variation'].value_counts(normalize=True)
x = dfEDA2['Variation'].value_counts()
fig = px.ecdf(x= x,
              y=y,
              ecdfnorm=None,
              ecdfmode='complementary',
              title='Frequência Acumulada por Variação')
fig.update_xaxes(title_text = 'Contagem por Variação')
fig.update_yaxes(title_text = 'Frequência Acumulada')
```

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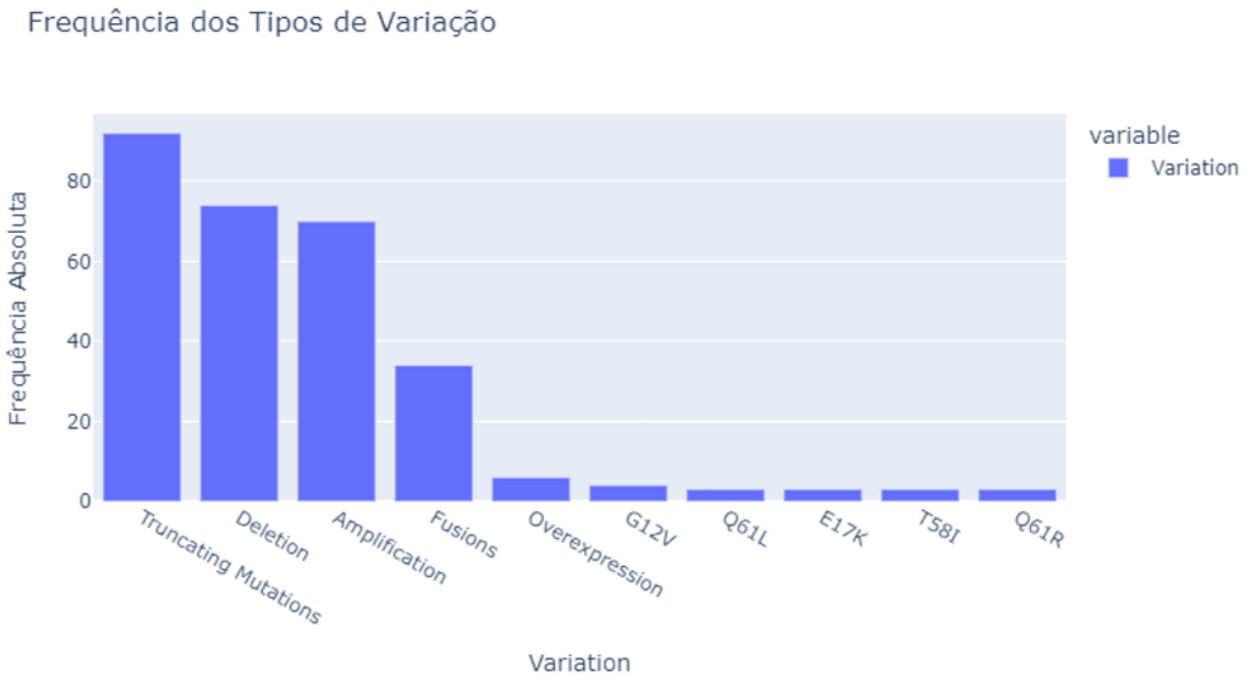
```
# Frequência Absoluta dos tipos de Variação 10+
x = dfEDA2['Variation'].value_counts().head(10)
fig = px.bar(x, title='Frequência dos Tipos de Variação')
fig.update_xaxes(title_text = 'Variation')
fig.update_yaxes(title_text = 'Frequência Absoluta')
```



Insight → The most common variations, which are Truncating Mutations, Deletion, Amplification, and Fusions, account for 8% of our observations.

Insight → It is clear that we have almost 1 unique variation for each observation, as we have 2993 different variations for 3316 observations.

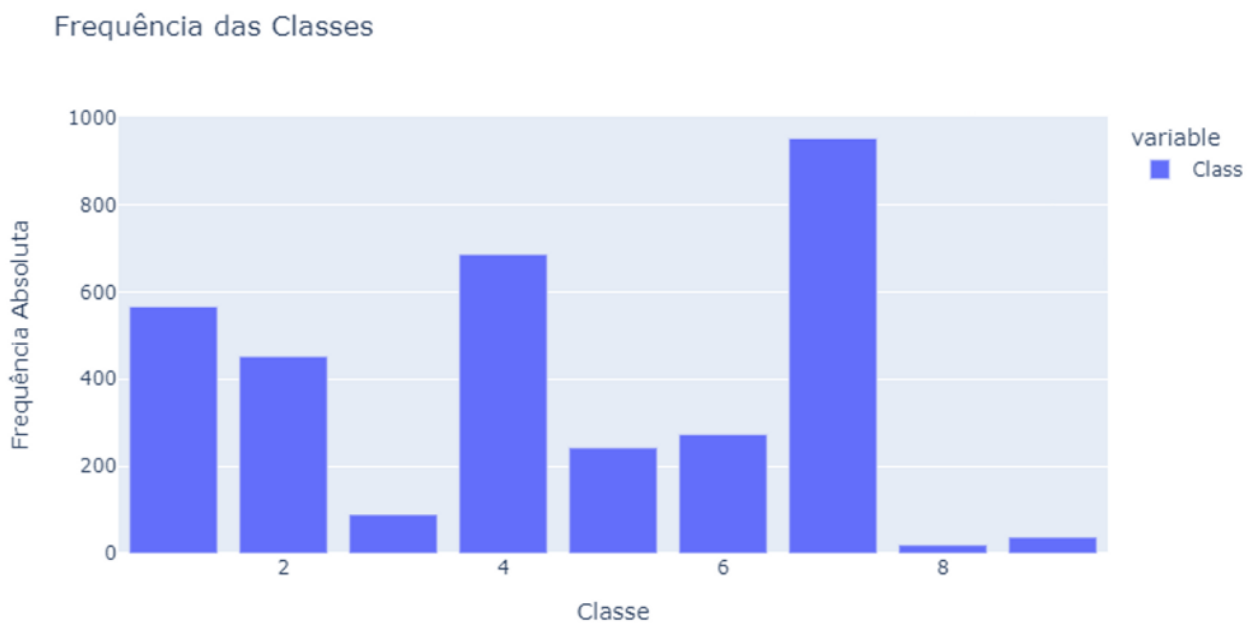
Variable Class - Target

To understand the distribution of our response variable, let's examine its distribution and identify if there is a need to balance the classes.

```
# Analisando as Quantidades de Cada Classe
dfEDA3 = dfEDA.copy()
dfEDA3['Class'].value_counts()
```

```
7    952
4    686
1    566
2    452
6    273
5    242
3     89
9     37
8     19
Name: Class, dtype: int64
```

```
# Distribuição dos tipos de Classe
fig = px.bar(dfEDA3['Class'].value_counts(), title='Frequência das Classes')
fig.update_xaxes(title_text = 'Classe')
fig.update_yaxes(title_text = 'Frequência Absoluta')
```



Insight → We can observe that the response variable exhibits a clear class imbalance. Therefore, it is necessary to consider appropriate strategies during the preprocessing stage to address this issue.

Let's further examine the top 5+ types of genes per class to see if there are any distinct patterns or behaviors.

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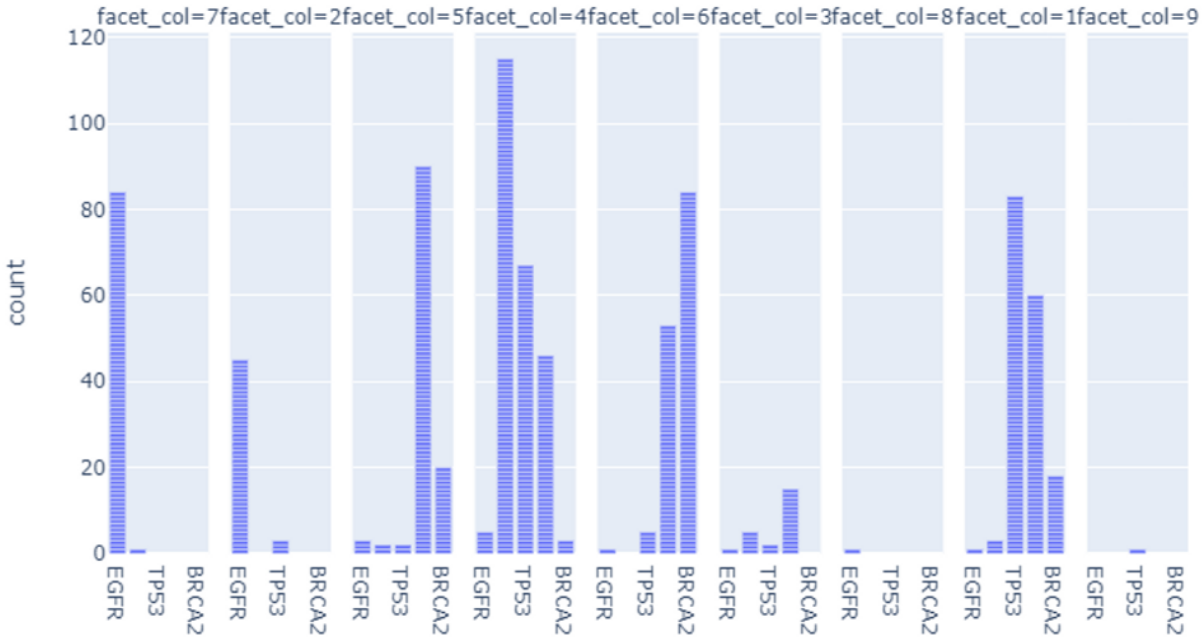
```
# Preparando um Dataframe para os 5 Mais Frequentes Genes
dfEDA4 = dfEDA.copy()
filtro = ['BRCA1', 'TP53', 'EGFR', 'PTEN', 'BRCA2']
dfEDA4 = dfEDA4[dfEDA4['Gene'].isin(filtro)]
dfEDA4
```

	ID	Text	Gene	Variation	Class
138	138	non small cell lung cancer is the leading caus...	EGFR	L747_T751delinsP	7
139	139	in contrast to other primary epidermal growth ...	EGFR	S752_I759del	2
140	140	the accurate determination of perfluoroalkyl s...	EGFR	I491M	5
141	141	in contrast to other primary epidermal growth ...	EGFR	D770_P772dup	7
142	142	purpose clinical features of epidermal growth ...	EGFR	G719A	7
...	...	...	...	...	...

```
# Analisando os 5 Genes de Maior Frequência, em relação as Classes
fig = px.bar(x=dfEDA4['Gene'], facet_col=dfEDA4['Class'])
fig.show()
```



We have a dataset that is quite diluted across the classes, which does not provide us with much information about the data patterns.

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Variable Text

Let's focus on observing the "Text" variable for the most frequent genes since the volume of data is too large for our computational capacity. The key is to gain an understanding of the most commonly used terms within the dataset and identify any interesting insights.

```
# Criando uma cópia do Dataset com os Genes Mais Frequentes com a Variável Text
dfEDA5 = dfEDA4['Text'].copy()

# Visualizando as 5 primeiras observações
dfEDA5.head()
```

```
138    non small cell lung cancer is the leading caus...
139    in contrast to other primary epidermal growth ...
140    the accurate determination of perfluoroalkyl s...
141    in contrast to other primary epidermal growth ...
142    purpose clinical features of epidermal growth ...
Name: Text, dtype: object
```

```
# Criando nosso Volume de Palavras
DataCloud = " ".join(review for review in dfEDA5)

# Verificando o Comprimento da Lista de Palavras
len(DataCloud)
```

```
48095828
```

We have 48 million words in our sample.

```
# Criando uma WordCloud
stopwordswd=set(STOPWORDS)
WD = WordCloud(stopwords=stopwordswd, background_color='white').generate(DataCloud)
plt.figure(figsize=(12,12))
plt.imshow(WD, interpolation='bilinear')
plt.axis('off')
plt.show()
```

We can identify some highly frequent words such as "mutation," and genes that we have identified based on their frequency, such as "p53," "brca1," "brct," and "egfr." Additionally, there are impactful words such as "patient," "wild-type," "family," and "history."

Given that this is a highly technical and complex text, we acknowledge that our tokenization and stopwords may have some limitations. However, we believe it is sufficient to proceed with our preprocessing and achieve good predictive results. Let's move forward!

Preprocessing and Data Preparation

Let's prepare the data for our predictive modeling.

As we observed during EDA, we have an imbalanced variable "Class" with 9 possible classes. However, since we are working with non-relational data and text variables, it is not feasible to generate synthetic data. Undersampling would also be impractical due to the significant class imbalance. Therefore, we will proceed with the imbalanced response variable and aim to achieve the best possible performance in our machine learning models.

Since the goal of the project is to use literature texts to classify genes and their mutations, we will perform TF-IDF vectorization only on the "Text" variable. After that, we will split the data into training and testing sets. Once these steps are completed, we will be ready to start developing our machine learning models.

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```
# Aplicando Vetorização nos dados com TF-IDF na Variável Text
vetorizador = TfidfVectorizer().fit(df['Text'])
X = vetorizador.transform(df['Text'])
y = df.Class
```

```
# Verificando o Shape da Variável de Entrada
X.shape
```

```
(3316, 153173)
```

```
# Separando os dados em Treino e Teste
X_train, X_test, y_train, y_test = train_test_split(X, y, test_size=0.3, random_state=0)
```

Great! We are all set to start developing our machine learning models. Let's proceed with the model development process!

Machine Learning

Sure! Let's work with the algorithms K-Nearest Neighbors, Random Forest Classifier, Multinomial Naive Bayes, and Stochastic Gradient Descent.

For each model, we will calculate the following metrics:

- Log Loss: The logarithmic loss between the actual and predicted values.

- ROC AUC: The accuracy of the model based on the ROC probability curve.

We will compile the results into a data frame to decide which model to deploy with new data. Let's proceed with model training and evaluation.

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Model 00 - K-Nearest Neighbors

```
# Testando o Melhor Valor de K
klist = [3, 10, 20, 30, 50, 80, 100, 200, 300, 500, 800, 1000]

# Lista de Log Loss de K
LogLoss = []
rAUC = []

# Loop de Teste
for k in klist:

  # Treinando o modelo KNN com cada valor de k
  modelo = KNeighborsClassifier(n_neighbors = k)
  modelo.fit(X_train, y_train)

  # Previsões com o modelo
  pred = modelo.predict_proba(X_test)

  # Avaliando o modelo com a lista de Métricas
  score1 = log_loss(y_test, pred, eps=1e-15)
  score2 = roc_auc_score(y_test, pred, multi_class='ovr')
  print(f'As métricas para k={k} são:\nLog Loss {score1} \nrAUC = {score2}\n------------------------\n')
  LogLoss.append(score1)
  rAUC.append(score2)
```

```
As métricas para k=3 são:
Log Loss 7.2469291862376375
rAUC = 0.8317891744856349
------------------------

As métricas para k=10 são:
Log Loss 3.461789199254402
rAUC = 0.8543974087100542
------------------------

As métricas para k=20 são:
Log Loss 2.273187752857395
rAUC = 0.854930572989245
------------------------

As métricas para k=30 são:
Log Loss 2.0759173896354843
rAUC = 0.8412752632396319
------------------------

As métricas para k=50 são:
Log Loss 1.761707324536837
rAUC = 0.8305992023758364
------------------------

As métricas para k=80 são:
Log Loss 1.7398057398708717
rAUC = 0.8181349965632221
------------------------

As métricas para k=100 são:
Log Loss 1.7840949125406764
rAUC = 0.8121780263484553
------------------------

As métricas para k=200 são:
Log Loss 1.810393434413759
rAUC = 0.7986984307944747
------------------------

As métricas para k=300 são:
Log Loss 1.8018078038309575
rAUC = 0.8002878299844858
------------------------

As métricas para k=500 são:
Log Loss 1.7519217969140624
rAUC = 0.8132767997118974
------------------------

As métricas para k=800 são:
Log Loss 1.7759019671578624
rAUC = 0.7675602297593834
------------------------

As métricas para k=1000 são:
Log Loss 1.7893641506349194
rAUC = 0.789524106770118
------------------------
```

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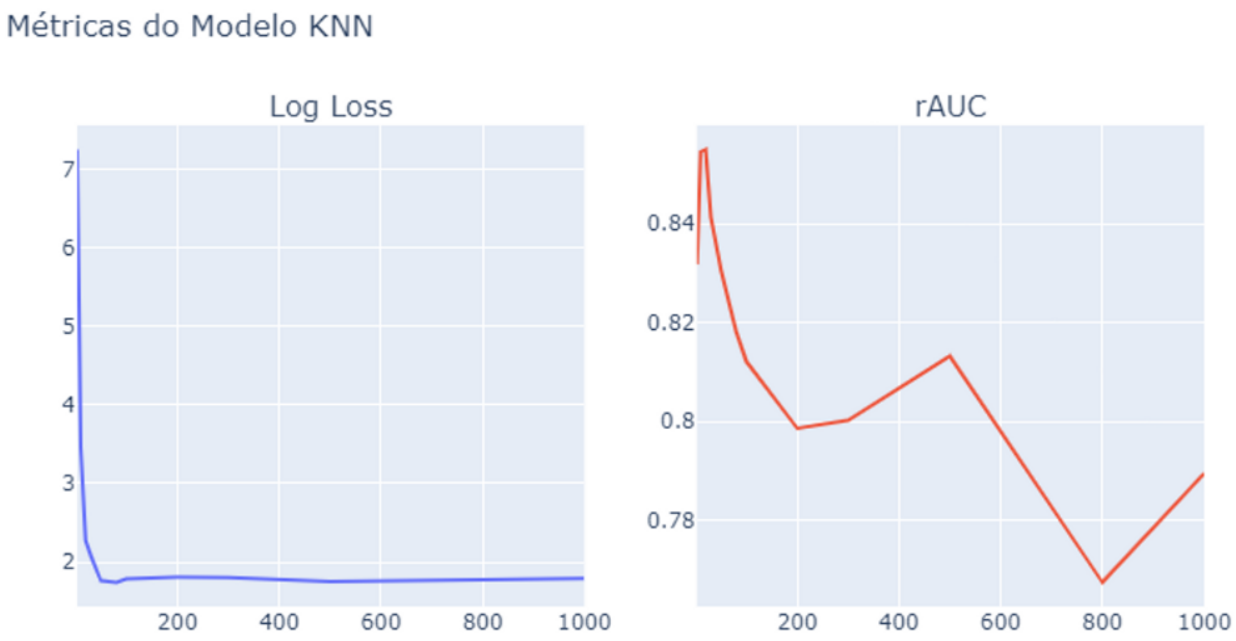
```
# Visão Gráfica das Métricas
dic = {'K': klist, 'Log Loss': LogLoss, 'rAUC': rAUC}
data = pd.DataFrame(dic)
fig = make_subplots(rows=1, cols=2, subplot_titles=['Log Loss', 'rAUC'])
fig.add_trace(go.Scatter(x = data['K'], y = data['Log Loss'], mode='lines', showlegend=False), row=1, col=1)
fig.update_yaxes(showticklabels = True)
fig.add_trace(go.Scatter(x = data['K'], y = data['rAUC'], mode='lines', showlegend=False), row=1, col=2)
fig.update_layout(title_text = 'Métricas do Modelo KNN')
fig.show()
```



The logarithmic loss (Log Loss) measures how closely the predicted probability aligns with the actual value. In other words, the greater the divergence between the predicted probability and the actual value, the higher the log loss. Therefore, we aim for a lower log loss.

The ROC AUC curve helps determine the model's accuracy level, with a higher value indicating better accuracy.

That being said, you have identified an ideal value of K = 80 for the K-Nearest Neighbors algorithm. We will proceed with training and evaluating the model using this value.

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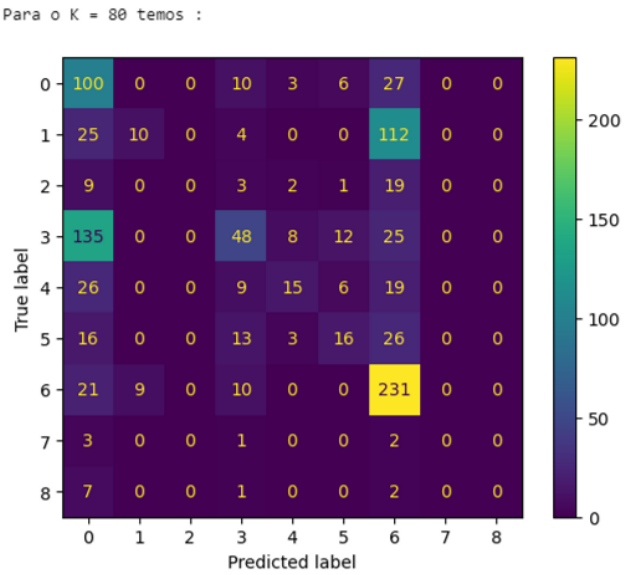
```
# Criando o Modelo KNN com K - Otimizado
K=80
print(f'Para o K = {K} temos :')

# Treinamento do Modelo KNN Otimizado
modeloKNN = KNeighborsClassifier(n_neighbors = K)
modeloKNN.fit(X_train,y_train)

# Previsão do Modelo KNN Otimizado
predKNN = modeloKNN.predict(X_test)
predKNN_proba = modeloKNN.predict_proba(X_test)

# Métricas do Modelo KNN Otimizado
score1KNN = log_loss(y_test, predKNN_proba, eps=1e-15)
score2KNN = roc_auc_score(y_test, predKNN_proba, multi_class='ovr')

# Matriz de Confusão
disp = ConfusionMatrixDisplay(confusion_matrix(y_test,predKNN))
disp.plot()
plt.show()
```

```
Para o K = 80 temos :
```



We can observe from the Confusion Matrix that our model is making more accurate predictions for classes 7 and 1. This is due to the imbalanced nature of the output variable, where the model learns more about the classes with higher frequency compared to the classes with lower frequency.

The class imbalance can affect the model's ability to accurately predict minority classes. It is important to address this issue through techniques such as oversampling, undersampling, or using class weights to give more importance to the minority classes during training.

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By addressing the class imbalance, we can potentially improve the model's performance in predicting the classes with lower frequency.

```
# Salvando os Resultados em um Dataset de Compilação
resultado = {'Modelo KNN': [score1KNN, score2KNN]}
resultado = pd.DataFrame(resultado, index=['Log Loss', 'rAUC'])
resultado
```

	Modelo KNN
Log Loss	1.739806
rAUC	0.818135

Model 01 - Random Forest Classifier

```
# Testando o Melhor Valor dos Estimadores
Elist = [3, 10, 20, 30, 50, 80, 100, 200, 300, 500, 800, 1000]

# Lista de Log Loss de E
LogLoss = []
rAUC = []

# Loop de Teste
for E in Elist:

  # Treinando o modelo RF com cada valor de E
  modelo = RandomForestClassifier(n_estimators= E, max_depth=9)
  modelo.fit(X_train, y_train)
  # Previsões com o modelo
  pred = modelo.predict_proba(X_test)
  # Avaliando o modelo com a lista de Métricas
  score1 = log_loss(y_test, pred, eps=1e-15)
  score2 = roc_auc_score(y_test, pred, multi_class='ovr')
  print(f'As métricas para E={E} são:\nLog Loss {score1} \nrAUC = {score2}\n------------------------\n')
  LogLoss.append(score1)
  rAUC.append(score2)
```

```
As métricas para E=3 são:
Log Loss 1.588718501755847
rAUC = 0.7680705398708116
------------------------

As métricas para E=10 são:
Log Loss 1.3618397728860574
rAUC = 0.8628919630568463
------------------------

As métricas para E=20 são:
Log Loss 1.3538509871367217
rAUC = 0.8729000781936662
------------------------

As métricas para E=30 são:
Log Loss 1.3155455928765336
rAUC = 0.874587127306645
------------------------

As métricas para E=50 são:
Log Loss 1.2923752135684154
rAUC = 0.8952951072526827
------------------------

As métricas para E=80 são:
Log Loss 1.3129939160878585
rAUC = 0.8860501431291453
------------------------

As métricas para E=100 são:
Log Loss 1.3013142923385632
rAUC = 0.8892347701411101
------------------------

As métricas para E=200 são:
Log Loss 1.3065416928682023
rAUC = 0.8936404731094836
------------------------

As métricas para E=300 são:
Log Loss 1.30033888946557
rAUC = 0.8958949986257753
------------------------

As métricas para E=500 são:
Log Loss 1.3010230268713627
rAUC = 0.9019048545870412
------------------------

As métricas para E=800 são:
Log Loss 1.3007150430575682
rAUC = 0.9023410984245687
------------------------

As métricas para E=1000 são:
Log Loss 1.3004709209953975
rAUC = 0.9033806573122884
------------------------
```

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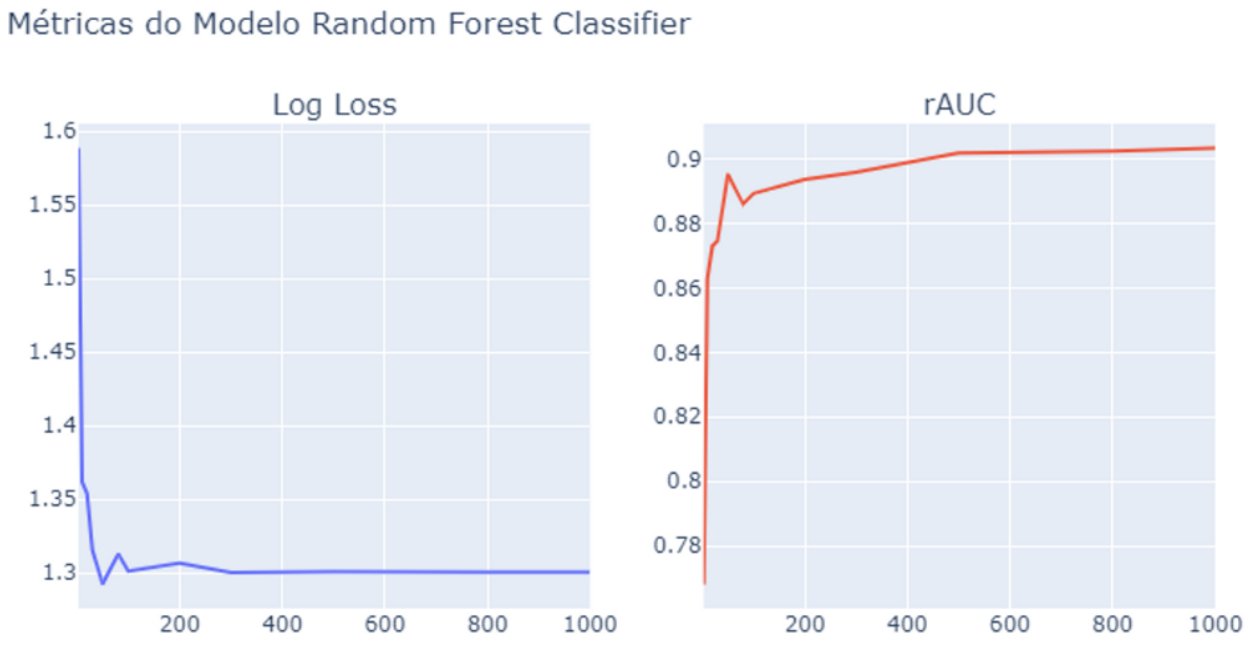
```
# Visão Gráfica das Métricas
dic = {'E': Elist, 'Log Loss': LogLoss, 'rAUC': rAUC}
data = pd.DataFrame(dic)
fig = make_subplots(rows=1, cols=2, subplot_titles=['Log Loss', 'rAUC'])
fig.add_trace(go.Scatter(x = data['E'], y = data['Log Loss'], mode='lines', showlegend=False), row=1, col=1)
fig.update_yaxes(showticklabels = True)
fig.add_trace(go.Scatter(x = data['E'], y = data['rAUC'], mode='lines', showlegend=False), row=1, col=2)
fig.update_layout(title_text = 'Métricas do Modelo Random Forest Classifier')
fig.show()
```



We can conclude that the best value for the number of estimators (E) is 50, as it results in the lowest log loss and a good accuracy of 89.5% with ROC AUC.

By evaluating the model's performance with different values of E, we have determined that E = 50 provides the best balance between log loss and accuracy. This suggests that using 50 estimators in the Random Forest Classifier yields favorable results for our problem.

It's important to note that the choice of the optimal value for the number of estimators may vary depending on the specific dataset and problem at hand. It's always recommended to experiment with different values and evaluate their performance to determine the most suitable value for the given task.

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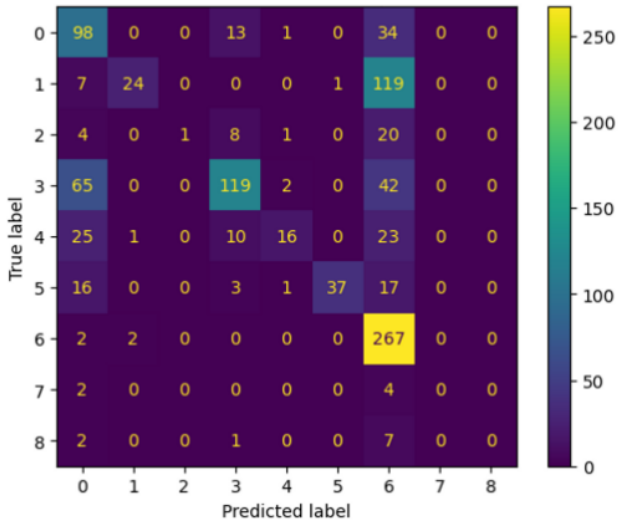
```
# Criando o Modelo RF com E otimizado
E=50
print(f'Para o Número de Estimadores E = {E} temos :')

# Treinamento do Modelo RF
modeloRF = RandomForestClassifier(n_estimators= E, max_depth= 9)
modeloRF.fit(X_train,y_train)

# Previsão do Modelo RF
predRF = modeloRF.predict(X_test)
predRF_proba = modeloRF.predict_proba(X_test)

# Métricas do Modelo KNN Otimizado
score1RF = log_loss(y_test, predRF_proba, eps=1e-15)
score2RF = roc_auc_score(y_test, predRF_proba, multi_class='ovr')

# Matriz de Confusão
disp = ConfusionMatrixDisplay(confusion_matrix(y_test,predRF))
disp.plot()
plt.show()
```

```
Para o Número de Estimadores E = 50 temos :
```



We can observe from the Confusion Matrix that our model is making more accurate predictions for classes 1, 4, 6, and 7 compared to the K-Nearest Neighbors (KNN) model. This indicates an improvement in performance with the Random Forest Classifier.

The Random Forest model is able to capture more complex patterns in the data and make more accurate predictions for multiple classes. This is reflected in the improved performance for certain classes in the Confusion Matrix.

```
# Salvando os Resultados no Dataset de Compilação
resultado['Modelo RF'] = [score1RF, score2RF]
resultado
```

	Modelo KNN	Modelo RF
Log Loss	1.739806	1.315174
rAUC	0.818135	0.877773

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Model 02 - Multinomial Naive Bayes

```
# Testando o Melhor Valor de Alpha
alpha = np.arange(0.001, 0.01, 0.001)

# Lista de Log Loss de K
LogLoss = []
rAUC = []

# Loop de Teste
for a in alpha:

  # Treinando o modelo MNB com cada valor de k
  modelo = MultinomialNB(alpha= a)
  modelo.fit(X_train, y_train)

  # Previsões com o modelo MNB
  pred = modelo.predict_proba(X_test)

  # Avaliando o modelo com a lista de Métricas
  score1 = log_loss(y_test, pred, eps=1e-15)
  score2 = roc_auc_score(y_test, pred, multi_class='ovr')
  print(f'As métricas para aplha = {a} são:\nLog Loss {score1} \nrAUC = {score2}\n-----------------------\n')
  LogLoss.append(score1)
  rAUC.append(score2)
```

```
As métricas para aplha = 0.001 são:
Log Loss 1.986331007604535
rAUC = 0.8901304350215504
-----------------------

As métricas para aplha = 0.002 são:
Log Loss 1.8695063115405373
rAUC = 0.8921189515750536
-----------------------

As métricas para aplha = 0.003 são:
Log Loss 1.826223437315264
rAUC = 0.8937109694172661
-----------------------

As métricas para aplha = 0.004 são:
Log Loss 1.8107515103646306
rAUC = 0.8945787406669246
-----------------------

As métricas para aplha = 0.005 são:
Log Loss 1.8097576895227006
rAUC = 0.8948926441100631
-----------------------

As métricas para aplha = 0.006 são:
Log Loss 1.817951905715058
rAUC = 0.8946954990481765
-----------------------

As métricas para aplha = 0.007 são:
Log Loss 1.8321227527237165
rAUC = 0.8940473881777038
-----------------------

As métricas para aplha = 0.008 são:
Log Loss 1.8503222674362616
rAUC = 0.8926344686971448
-----------------------

As métricas para aplha = 0.009000000000000001 são:
Log Loss 1.870994858693849
rAUC = 0.8911577865613949
-----------------------
```

```
# Visão Gráfica das Métricas
dic = {'alpha': alpha, 'Log Loss': LogLoss, 'rAUC': rAUC}
data = pd.DataFrame(dic)
fig = make_subplots(rows=1, cols=2, subplot_titles=['Log Loss', 'rAUC'])
fig.add_trace(go.Scatter(x = data['alpha'], y = data['Log Loss'], mode='lines', showlegend=False), row=1, col=1)
fig.update_yaxes(showticklabels = True)
fig.add_trace(go.Scatter(x = data['alpha'], y = data['rAUC'], mode='lines', showlegend=False), row=1, col=2)
fig.update_layout(title_text = 'Métricas do Modelo Multinomial Naive Bayes')
fig.show()
```

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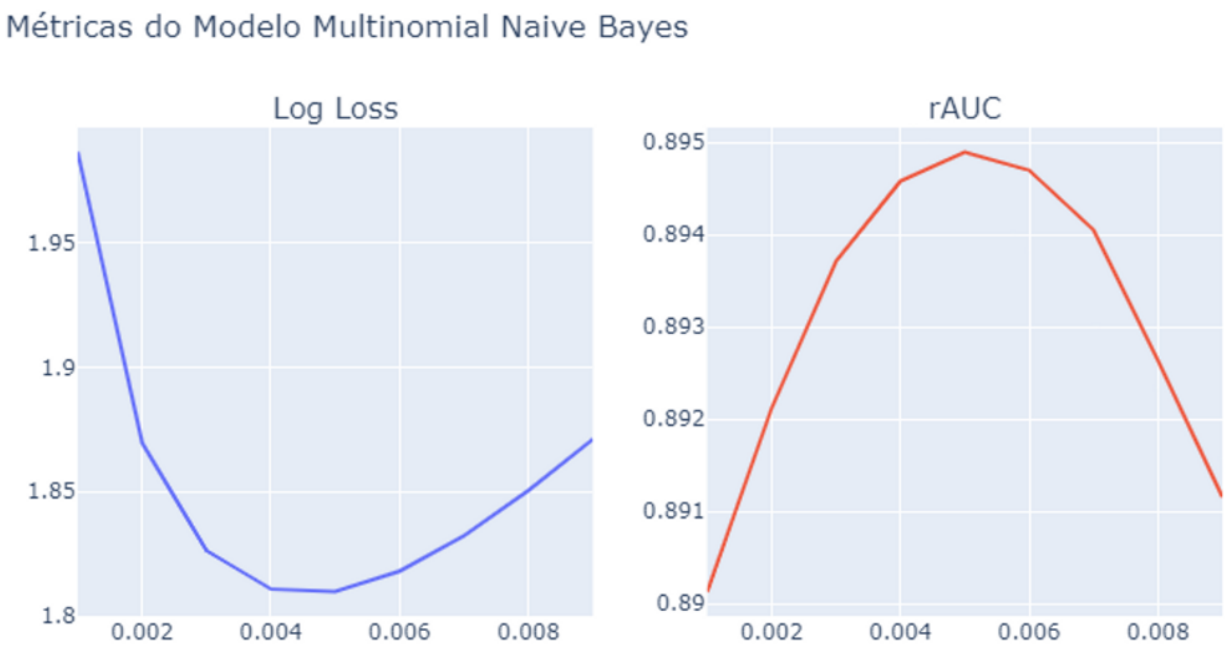


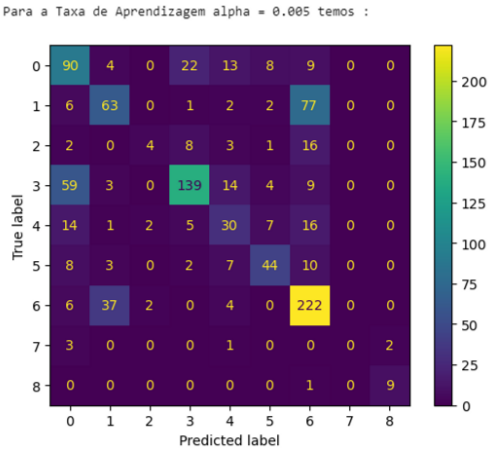
We can conclude that a value of alpha equal to 0.005 is suitable as it results in a log loss of 1.81 and an rAUC of 89.5%.

```
# Criando o Modelo MNB com alpha otimizado
A = 0.005
print(f'Para a Taxa de Aprendizagem alpha = {A} temos :')

# Treinamento do Modelo MNB
modeloMNB = MultinomialNB(alpha = A)
modeloMNB.fit(X_train,y_train)

# Previsão do Modelo MNB
predMNB = modeloMNB.predict(X_test)
predMNB_proba = modeloMNB.predict_proba(X_test)

# Métricas do Modelo MNB Otimizado
score1MNB = log_loss(y_test, predMNB_proba, eps=1e-15)
score2MNB = roc_auc_score(y_test, predMNB_proba, multi_class='ovr')
# Matriz de Confusão
disp = ConfusionMatrixDisplay(confusion_matrix(y_test,predMNB))
disp.plot()
plt.show()
```



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We have made progress in our predictions, achieving more correct predictions and fewer errors for all classes! Although we obtained a higher Log Loss compared to the previous models, we have achieved the highest accuracy so far.

The trade-off between Log Loss and accuracy is a common consideration in machine learning. While a lower Log Loss indicates better probability estimation, a higher accuracy demonstrates the model's ability to make correct predictions overall.

In this case, even though the Log Loss is slightly higher, the improved accuracy suggests that the Stochastic Gradient Descent (SGD) model performs better in terms of overall prediction accuracy.

```
# Salvando os Resultados no Dataset de Compilação
resultado['Modelo MNB'] = [score1MNB, score2MNB]
resultado
```

	Modelo KNN	Modelo RF	Modelo MNB
Log Loss	1.739806	1.315174	1.809758
rAUC	0.818135	0.877773	0.894893

Model 03 - Stochastic Gradient Descent Classifier

```
# Testando o Melhor Valor de Alpha
alpha = np.arange(1e-6, 1e-4, 1e-5)

# Lista de Log Loss de K
LogLoss = []
rAUC = []

# Loop de Teste
for a in alpha:

  # Treinando o modelo MNB com cada valor de k
  modelo = SGDClassifier(loss = 'log' ,alpha = a ,penalty = 'l2' ,shuffle = True , class_weight = 'balanced', random_state = 0)
  modelo.fit(X_train, y_train)

  # Previsões com o modelo MNB
  pred = modelo.predict_proba(X_test)

  # Avaliando o modelo com a lista de Métricas
  score1 = log_loss(y_test, pred, eps=1e-15)
  score2 = roc_auc_score(y_test, pred, multi_class='ovr')
  print(f'As métricas para aplha = {a} são:\nLog Loss {score1} \nrAUC = {score2}\n------------------------\n')
  LogLoss.append(score1)
  rAUC.append(score2)
```

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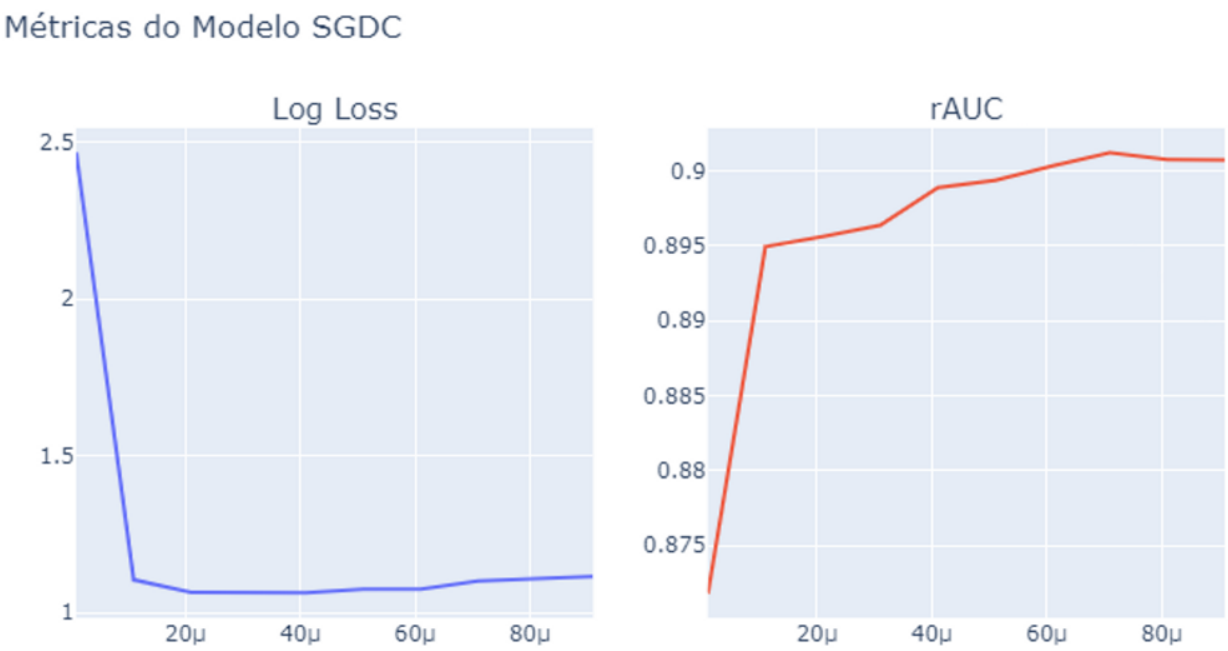
```
As métricas para aplha = 1e-06 são:
Log Loss 2.465576114086081
rAUC = 0.8718152520486088
------------------------

As métricas para aplha = 1.1000000000000001e-05 são:
Log Loss 1.103728365277631
rAUC = 0.894958588837818
------------------------

As métricas para aplha = 2.1000000000000002e-05 são:
Log Loss 1.063389495247917
rAUC = 0.8956276703115074
------------------------

As métricas para aplha = 3.1e-05 são:
Log Loss 1.0609805418305134
rAUC = 0.8963684957964951
------------------------

As métricas para aplha = 4.1e-05 são:
Log Loss 1.0621328273267892
rAUC = 0.8988846138414817
------------------------

As métricas para aplha = 5.1e-05 são:
Log Loss 1.0736573068759645
rAUC = 0.8993695541944818
------------------------

As métricas para aplha = 6.1000000000000005e-05 são:
Log Loss 1.0746356388066716
rAUC = 0.9003517855495805
------------------------

As métricas para aplha = 7.1e-05 são:
Log Loss 1.099807015677233
rAUC = 0.9012197126788707
------------------------

As métricas para aplha = 8.1e-05 são:
Log Loss 1.1091348310813096
rAUC = 0.9007558419403193
------------------------

As métricas para aplha = 9.1e-05 são:
Log Loss 1.114259172155043
rAUC = 0.900738893445205
------------------------
```

```
# Visão Gráfica das Métricas
dic = {'alpha': alpha, 'Log Loss': LogLoss, 'rAUC': rAUC}
data = pd.DataFrame(dic)
fig = make_subplots(rows=1, cols=2, subplot_titles=['Log Loss', 'rAUC'])
fig.add_trace(go.Scatter(x = data['alpha'], y = data['Log Loss'], mode='lines', showlegend=False), row=1, col=1)
fig.update_yaxes(showticklabels = True)
fig.add_trace(go.Scatter(x = data['alpha'], y = data['rAUC'], mode='lines', showlegend=False), row=1, col=2)
fig.update_layout(title_text = 'Métricas do Modelo SGDC')
fig.show()
```



We can determine that the best learning rate alpha is 2.1e-5, which gives us a Log Loss close to 1.06 and an rAUC of 89.5%.

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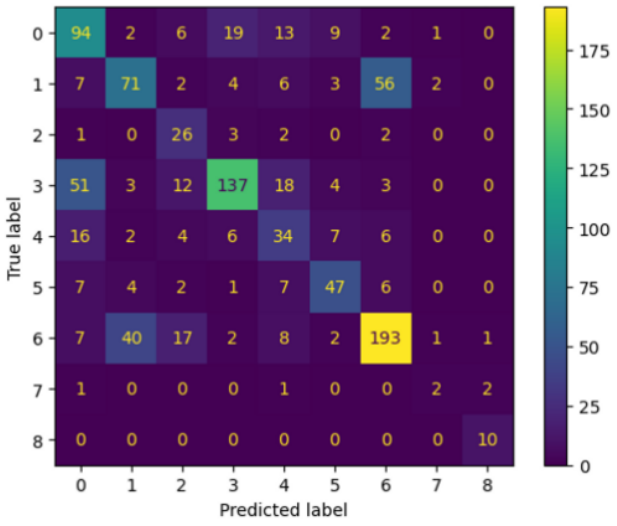
```
# Criando o Modelo SGDC com alpha otimizado
A = 2.1e-5
print(f'Para a Taxa de Aprendizagem alpha {A} temos :')

# Treinamento do Modelo SGDC
modeloSGDC = SGDClassifier(alpha = A, loss = 'log' ,penalty = 'l2' ,shuffle = True , class_weight = 'balanced', random_state = 0)
modeloSGDC.fit(X_train,y_train)

# Previsão do Modelo SGDC
predSGDC = modeloSGDC.predict(X_test)
predSGDC_proba = modeloSGDC.predict_proba(X_test)

# Métricas do Modelo SGDC Otimizado
score1SGDC = log_loss(y_test, predSGDC_proba, eps=1e-15)
score2SGDC = roc_auc_score(y_test, predSGDC_proba, multi_class='ovr')
# Matriz de Confusão
disp = ConfusionMatrixDisplay(confusion_matrix(y_test,predSGDC))
disp.plot()
plt.show()
```

```
Para a Taxa de Aprendizagem alpha 2.1e-05 temos :
```



From the Confusion Matrix, we can see that our model made many more correct predictions than incorrect ones in all classes. We also obtained the lowest Log Loss and the highest Accuracy among all the trained models.

```
# Salvando os Resultados no Dataset de Compilação
resultado['Modelo SGDC'] = [score1SGDC, score2SGDC]
resultado
```

	Modelo KNN	Modelo RF	Modelo MNB	Modelo SGDC
Log Loss	1.739806	1.315174	1.809758	1.063389
rAUC	0.818135	0.877773	0.894893	0.895628

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Great! With the SGDC model selected as the best model, we can proceed with the classification of new data and deliver the results to our end client. Using the trained SGDC model, we can now make predictions on unseen data by inputting the relevant features (genes, variations) into the model.

Once the predictions are generated, they can be presented to the end client along with any additional information or visualizations that might be useful for interpretation. It's important to communicate the limitations and assumptions of the model to the client to ensure they have a clear understanding of the predicted results.

Model Deploy

```
# Verificando se Temos dados Ausentes
df = dft.copy()
df.isna().sum()
```

```
ID           0
Text         1
Gene         0
Variation    0
dtype: int64
```

```
# Eliminando dados Ausentes
df = df.dropna()
df.isna().sum()
```

```
ID           0
Text         0
Gene         0
Variation    0
dtype: int64
```

```
# Aplicando a Função text_process no Dataset
df['Text'] = df['Text'].apply(text_process)
print(df.info())
print(df.isnull().sum())
df.head()
```

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```
<class 'pandas.core.frame.DataFrame'>
Int64Index: 5667 entries, 0 to 5667
Data columns (total 4 columns):
 #   Column     Non-Null Count  Dtype 
---  ------     --------------  ----- 
 0   ID         5667 non-null   int64 
 1   Text       5667 non-null   object
 2   Gene       5667 non-null   object
 3   Variation  5667 non-null   object
dtypes: int64(1), object(3)
memory usage: 221.4+ KB
None
ID           0
Text         0
Gene         0
Variation    0
dtype: int64
```

	ID	Text	Gene	Variation
0	0	2 this mutation resulted in a myeloproliferati...	ACSL4	R570S
1	1	abstract the large tumor suppressor 1 lats1 is...	NAGLU	P521L
2	2	vascular endothelial growth factor receptor ve...	PAH	L333F
3	3	inflammatory myofibroblastic tumor imt is a ne...	ING1	A148D
4	4	abstract retinoblastoma is a pediatric retinal...	TMEM216	G77A

```
# Aplicando Vetorização nos dados com TF-IDF na Variável Text
X = vetorizador.transform(df.Text)
X
```

```
<5667x153173 sparse matrix of type '<class 'numpy.float64'>'
	with 10485176 stored elements in Compressed Sparse Row format>
```

```
# Classificando os dados com o Modelo SGDC
Classe = modeloSGDC.predict(X)
Classe = pd.Series(Classe)
```

```
# Organizando a Planilha de Entrega para o Cliente
DadosClassificados = dft.drop(columns=['Text'])
DadosClassificados['Classe do Cancer'] = Classe
DadosClassificados.head(10)
```

	ID	Gene	Variation	Classe do Cancer
0	0	ACSL4	R570S	7.0
1	1	NAGLU	P521L	4.0
2	2	PAH	L333F	2.0
3	3	ING1	A148D	7.0
4	4	TMEM216	G77A	4.0
5	5	CD40LG	A123E	4.0
6	6	KLF11	T220M	7.0
7	7	SGCB	T151R	4.0
8	8	CLCF1	R197L	1.0
9	9	SDHAF1	R55P	7.0

Project Delivered, Happy Client! Let's move on to the next one!!