Evaluation using the BeFree corpus

GAD dataset

To obtain a large benchmark of Gene Disease Associations along with associated sentences from literature, we used the corpus generated by BeFree system based on Genetic Association Database (GAD)

----

imports

```
import tensorflow as tf
import keras
from keras.models import load_model
from keras.callbacks import ModelCheckpoint, EarlyStopping
from keras_tqdm import TQDMNotebookCallback
import numpy as np
np.random.seed(1337)
from keras_tqdm import TQDMNotebookCallback
import nltk
import xml.etree.ElementTree as ET
import pandas as pd
import os
import string
from nltk.tokenize import TreebankWordTokenizer
from numpy.random import random_sample
import re
import pickle
from sklearn.metrics import accuracy_score
from sklearn.metrics import classification_report

from keras.layers import Embedding, Flatten,LSTM, GRU
from keras.layers.convolutional import Conv2D, MaxPooling2D
from tensorflow.keras.utils import to_categorical
from tensorflow.keras.models import Sequential, Model
from keras.layers import Dense, Dropout, Activation,  Input, merge,Conv1D,MaxPooling1D,GlobalMaxPooling1D,Convolution1D
from keras import regularizers
from sklearn.metrics import precision_recall_fscore_support
from sklearn.model_selection import StratifiedKFold
import matplotlib.pyplot as plt
from keras.layers import Concatenate, concatenate
from keras import backend as K
from keras.layers import multiply
from keras.layers import merge
from keras.layers.core import *
from keras.layers.recurrent import LSTM
from keras.models import *
random_seed=1337
```

Define Callback functions to generate Measures

```
from keras import backend as K

def f1(y_true, y_pred):
    def recall(y_true, y_pred):
        true_positives = K.sum(K.round(K.clip(y_true * y_pred, 0, 1)))
        possible_positives = K.sum(K.round(K.clip(y_true, 0, 1)))
        recall = true_positives / (possible_positives + K.epsilon())
        return recall

    def precision(y_true, y_pred):
        true_positives = K.sum(K.round(K.clip(y_true * y_pred, 0, 1)))
        predicted_positives = K.sum(K.round(K.clip(y_pred, 0, 1)))
        precision = true_positives / (predicted_positives + K.epsilon())
        return precision
```

```
    precision = precision(y_true, y_pred)
    recall = recall(y_true, y_pred)
    return 2*((precision*recall)/(precision+recall+K.epsilon()))
```

Experiments to reproduce the results of Table 7

Evaluation results for multi-class classification

Load Prerocssed Data

```
with open('../data/pickles/befree_3class_crawl-300d-2M.pickle', 'rb') as handle:
    gene_id_list = pickle.load(handle)
    gene_symbol_list = pickle.load(handle)
    disease_id_list = pickle.load(handle)
    X_train = pickle.load(handle)
    distance1_vectors = pickle.load(handle)
    distance2_vectors = pickle.load(handle)
    Y_train = pickle.load(handle)
    word_list = pickle.load(handle)
    word_vectors = pickle.load(handle)
    word_dict = pickle.load(handle)
    distance1_dict = pickle.load(handle)
    distance2_dict = pickle.load(handle)
    label_dict = pickle.load(handle)
    MAX_SEQUENCE_LENGTH = pickle.load(handle)
print ("word_vectors",len(word_vectors))
```

```
word_vectors 6766
```

Create Position Embedding Vectors

```
import keras
from keras_pos_embd import TrigPosEmbedding

model = keras.models.Sequential()
model.add(TrigPosEmbedding(
    input_shape=(None,),
    output_dim=20,                      # The dimension of embeddings.
    mode=TrigPosEmbedding.MODE_EXPAND,  # Use `expand` mode
    name='Pos-Embd',
))
model.compile('adam', keras.losses.mae, {})
```

```
d1_train_embedded=model.predict(distance1_vectors)
```

```
d1_train_embedded.shape
```

```
d2_train_embedded=model.predict(distance2_vectors)
```

```
d2_train_embedded.shape
```

```
(5330, 81, 20)
```

Prepare Word Embedding Layer

```
EMBEDDING_DIM=word_vectors.shape[1]
print("EMBEDDING_DIM=",EMBEDDING_DIM)
embedding_matrix=word_vectors

def create_embedding_layer(l2_reg=0.01,use_pretrained=True,is_trainable=False):

    if use_pretrained:
        return Embedding(len(word_dict) ,EMBEDDING_DIM,weights=[embedding_matrix],input_length=MAX_SEQUENCE_LENGTH,trainable=is_trainable,em
```

```
    else:
        return Embedding(len(word_dict) ,EMBEDDING_DIM,input_length=MAX_SEQUENCE_LENGTH)
```

```
EMBEDDING_DIM= 300
```

Prepare Attention Mechanism

```
INPUT_DIM = 2
TIME_STEPS = MAX_SEQUENCE_LENGTH
def attentionNew(inputs):
    inputs = Lambda(lambda x: tf.keras.backend.tanh(x))(inputs)
    input_dim = int(inputs.shape[2])
    a = Permute((2, 1))(inputs)
    a = Dense(TIME_STEPS, activation='softmax')(a)
    a_probs = Permute((2, 1))(a)
    output_attention_mul = multiply([inputs, a_probs])
    output_attention_mul = Lambda(lambda x: tf.keras.backend.tanh(x))(output_attention_mul)
    return output_attention_mul
```

Create the Model

```
# set parameter for metric calculation, 'macro' for multiclass classification
param='macro'
from keras.optimizers import Adam
def build_model():

    sequence_input = Input(shape=(MAX_SEQUENCE_LENGTH,), dtype='int32')
    embedding_layer=create_embedding_layer(use_pretrained=True,is_trainable=False)
    embedded_sequences = embedding_layer(sequence_input)


    pos_embedd_1=Input(shape=(MAX_SEQUENCE_LENGTH,20), dtype='float32')
    pos_embedd_2=Input(shape=(MAX_SEQUENCE_LENGTH,20), dtype='float32')

    embedded_sequences = concatenate([embedded_sequences,pos_embedd_1,pos_embedd_2])


    x = Conv1D(64, 5, activation='relu')(embedded_sequences)
    x = MaxPooling1D(3)(x)
    x = Dropout(0.4)(x)
    conv_sequence_w5=GlobalMaxPooling1D()(x)    #x = Flatten()(x)



    x = Conv1D(128, 3, activation='relu')(embedded_sequences)
    x = MaxPooling1D(3)(x)
    x = Dropout(0.4)(x)
    conv_sequence_w4=GlobalMaxPooling1D()(x)    #x = Flatten()(x)



    x = Conv1D(256, 3, activation='relu')(embedded_sequences)
    x = MaxPooling1D(3)(x)
    x = Dropout(0.4)(x)
    conv_sequence_w3=GlobalMaxPooling1D()(x)    #x = Flatten()(x)

    forward = GRU(100, recurrent_dropout=0.1, return_sequences=True)(embedded_sequences)
    backward = GRU(100, go_backwards=True, recurrent_dropout=0.1, return_sequences=True)(embedded_sequences)
    lstm_gru_sequence = concatenate([forward, backward], axis=-1)
    # Apply attention mechanism
    attention_output = attentionNew(lstm_gru_sequence)  # Shape: (None, MAX_SEQUENCE_LENGTH, 200)
    attention_pooled = GlobalMaxPooling1D()(attention_output)  # Shape: (None, 200)

    merge = concatenate([conv_sequence_w5,conv_sequence_w4,conv_sequence_w3,attention_pooled])
    x = Dense(128, activation='relu', kernel_regularizer=regularizers.l2(0.05))(merge)
    x = Dropout(0.4)(x)
    preds = Dense(3, activation='softmax')(x)
    model = Model(inputs=[sequence_input, pos_embedd_1,pos_embedd_2],outputs=preds)
    opt=tf.keras.optimizers.Adam(learning_rate=0.001)
    model.compile(loss='categorical_crossentropy',optimizer=opt,metrics=['acc',f1])
```

```
    return model
```

```
model = build_model()
model.summary()
```

```
Model: "model_1"
__________________________________________________________________________________________________
Layer (type)                    Output Shape         Param #     Connected to
==================================================================================================
input_1 (InputLayer)            (None, 81)           0
__________________________________________________________________________________________________
embedding_1 (Embedding)         (None, 81, 300)      2029800     input_1[0][0]
__________________________________________________________________________________________________
input_2 (InputLayer)            (None, 81, 20)       0
__________________________________________________________________________________________________
input_3 (InputLayer)            (None, 81, 20)       0
__________________________________________________________________________________________________
concatenate_1 (Concatenate)     (None, 81, 340)      0           embedding_1[0][0]
                                                                 input_2[0][0]
                                                                 input_3[0][0]
__________________________________________________________________________________________________
gru_1 (GRU)                     (None, 81, 100)      132300      concatenate_1[0][0]
__________________________________________________________________________________________________
gru_2 (GRU)                     (None, 81, 100)      132300      concatenate_1[0][0]
__________________________________________________________________________________________________
concatenate_2 (Concatenate)     (None, 81, 200)      0           gru_1[0][0]
                                                                 gru_2[0][0]
__________________________________________________________________________________________________
lambda_1 (Lambda)               (None, 81, 200)      0           concatenate_2[0][0]
__________________________________________________________________________________________________
permute_1 (Permute)             (None, 200, 81)      0           lambda_1[0][0]
__________________________________________________________________________________________________
dense_1 (Dense)                 (None, 200, 81)      6642        permute_1[0][0]
__________________________________________________________________________________________________
conv1d_1 (Conv1D)               (None, 77, 64)       108864      concatenate_1[0][0]
__________________________________________________________________________________________________
conv1d_2 (Conv1D)               (None, 79, 128)      130688      concatenate_1[0][0]
__________________________________________________________________________________________________
conv1d_3 (Conv1D)               (None, 79, 256)      261376      concatenate_1[0][0]
__________________________________________________________________________________________________
permute_2 (Permute)             (None, 81, 200)      0           dense_1[0][0]
__________________________________________________________________________________________________
max_pooling1d_1 (MaxPooling1D)  (None, 25, 64)       0           conv1d_1[0][0]
__________________________________________________________________________________________________
max_pooling1d_2 (MaxPooling1D)  (None, 26, 128)      0           conv1d_2[0][0]
__________________________________________________________________________________________________
max_pooling1d_3 (MaxPooling1D)  (None, 26, 256)      0           conv1d_3[0][0]
__________________________________________________________________________________________________
multiply_1 (Multiply)           (None, 81, 200)      0           lambda_1[0][0]
                                                                 permute_2[0][0]
__________________________________________________________________________________________________
dropout_1 (Dropout)             (None, 25, 64)       0           max_pooling1d_1[0][0]
__________________________________________________________________________________________________
dropout_2 (Dropout)             (None, 26, 128)      0           max_pooling1d_2[0][0]
__________________________________________________________________________________________________
dropout_3 (Dropout)             (None, 26, 256)      0           max_pooling1d_3[0][0]
__________________________________________________________________________________________________
lambda_2 (Lambda)               (None, 81, 200)      0           multiply_1[0][0]
__________________________________________________________________________________________________
global_max_pooling1d_1 (GlobalM (None, 64)           0           dropout_1[0][0]
__________________________________________________________________________________________________
global_max_pooling1d_2 (GlobalM (None, 128)          0           dropout_2[0][0]
__________________________________________________________________________________________________
```

```
validation_split_rate = 0.1
skf = StratifiedKFold(n_splits=5,shuffle=True, random_state=42)
Y = [np.argmax(y, axis=None, out=None) for y in Y_train]
for tr_index, te_index in skf.split(X_train,Y):
    test_index = te_index
    train_index = tr_index

trainRate = (len(train_index)/len(Y))*100
testRate = (len(test_index)/len(Y))*100
print ("TrainRate:{:.2f}% testRate:{:.2f}% validation:{:.2f}%  ".format(trainRate,testRate, trainRate*validation_split_rate))
X_train, X_test = X_train[train_index], X_train[test_index]
pos_train1, pos_test1 = d1_train_embedded[train_index], d1_train_embedded[test_index]
pos_train2, pos_test2 = d2_train_embedded[train_index], d2_train_embedded[test_index]
y_train, y_test = Y_train[train_index], Y_train[test_index]
```

```
# # Saving the training data split as a pickle file
# training_data = {
#     'X_train': X_train,
#     'pos_train1': pos_train1,
#     'pos_train2': pos_train2,
#     'y_train': y_train
# }

# with open('training_data12.pkl', 'wb') as f:
#     pickle.dump(training_data, f)

# # Saving the testing data split as a pickle file
# testing_data = {
#     'X_test': X_test,
#     'pos_test1': pos_test1,
#     'pos_test2': pos_test2,
#     'y_test': y_test
# }

# with open('testing_data12.pkl', 'wb') as f:
#     pickle.dump(testing_data, f)
```

```
TrainRate:80.00% testRate:20.00% validation:8.00%
```

```
# Load the training data from the pickle file
with open('training_data.pkl', 'rb') as f:
    train_data = pickle.load(f)

# Load the testing data from the pickle file
with open('testing_data.pkl', 'rb') as f:
    test_data = pickle.load(f)

# Extract data from the loaded dictionaries
X_train = train_data['X_train']
pos_train1 = train_data['pos_train1']
pos_train2 = train_data['pos_train2']
y_train = train_data['y_train']


X_test = test_data['X_test']
pos_test1 = test_data['pos_test1']
pos_test2 = test_data['pos_test2']
y_test = test_data['y_test']
print(X_train.shape)
print(X_test.shape)
```

```
(4264, 81)
(1066, 81)
```

Run the Evaluation using 10 fold Cross Validation

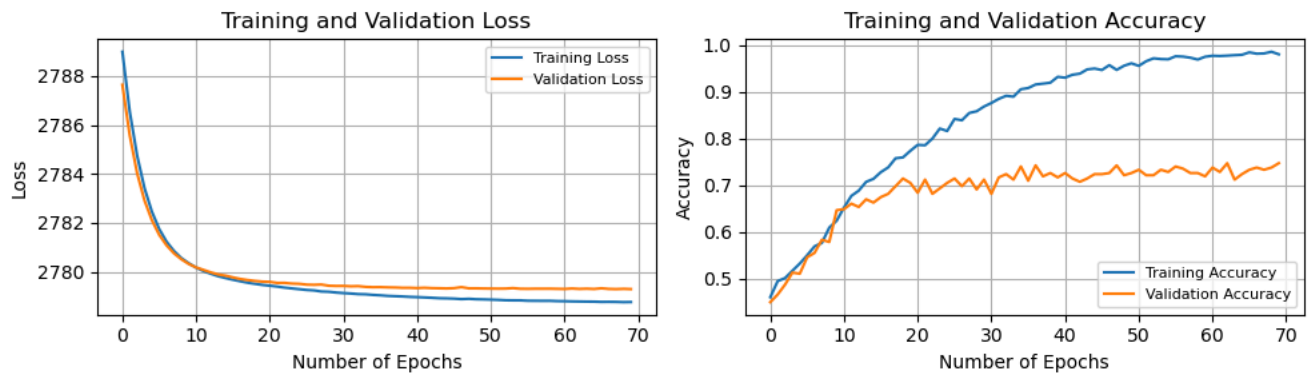
```
MaxEpochs =70
batchsize =32
validation_split_rate = 0.1
history=model.fit([X_train,pos_train1,pos_train2], y_train,validation_split=validation_split_rate ,epochs=MaxEpochs, batch_size=batchsize,ve
```

```
Train on 3837 samples, validate on 427 samples
Epoch 1/70
3837/3837 [==============================] - 13s 3ms/step - loss: 2788.9912 - acc: 0.4603 - f1: 0.3225 - val_loss: 2787.6417 - val_ac
Epoch 2/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2786.5415 - acc: 0.4947 - f1: 0.3315 - val_loss: 2785.5688 - val_ac
Epoch 3/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2784.7480 - acc: 0.5012 - f1: 0.3639 - val_loss: 2784.0269 - val_ac
Epoch 4/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2783.4213 - acc: 0.5168 - f1: 0.3868 - val_loss: 2782.9149 - val_ac
Epoch 5/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2782.4602 - acc: 0.5322 - f1: 0.4078 - val_loss: 2782.1099 - val_ac
```

```
Epoch 6/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2781.7462 - acc: 0.5499 - f1: 0.4356 - val_loss: 2781.5087 - val_ac
Epoch 7/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2781.2296 - acc: 0.5689 - f1: 0.4716 - val_loss: 2781.0789 - val_ac
Epoch 8/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2780.8509 - acc: 0.5765 - f1: 0.4766 - val_loss: 2780.7536 - val_ac
Epoch 9/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2780.5605 - acc: 0.6091 - f1: 0.5289 - val_loss: 2780.5136 - val_ac
Epoch 10/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2780.3518 - acc: 0.6239 - f1: 0.5461 - val_loss: 2780.3217 - val_ac
Epoch 11/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2780.1723 - acc: 0.6521 - f1: 0.5922 - val_loss: 2780.1805 - val_ac
Epoch 12/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2780.0332 - acc: 0.6771 - f1: 0.6199 - val_loss: 2780.0783 - val_ac
Epoch 13/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.9204 - acc: 0.6883 - f1: 0.6448 - val_loss: 2779.9770 - val_ac
Epoch 14/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.8196 - acc: 0.7071 - f1: 0.6743 - val_loss: 2779.8919 - val_ac
Epoch 15/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.7449 - acc: 0.7136 - f1: 0.6865 - val_loss: 2779.8436 - val_ac
Epoch 16/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.6775 - acc: 0.7279 - f1: 0.7070 - val_loss: 2779.7722 - val_ac
Epoch 17/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.6145 - acc: 0.7381 - f1: 0.7181 - val_loss: 2779.7089 - val_ac
Epoch 18/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.5547 - acc: 0.7576 - f1: 0.7416 - val_loss: 2779.6645 - val_ac
Epoch 19/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.5092 - acc: 0.7594 - f1: 0.7453 - val_loss: 2779.6280 - val_ac
Epoch 20/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.4653 - acc: 0.7735 - f1: 0.7636 - val_loss: 2779.6003 - val_ac
Epoch 21/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.4331 - acc: 0.7863 - f1: 0.7682 - val_loss: 2779.5905 - val_ac
Epoch 22/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.3985 - acc: 0.7855 - f1: 0.7738 - val_loss: 2779.5470 - val_ac
Epoch 23/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.3554 - acc: 0.7998 - f1: 0.7852 - val_loss: 2779.5485 - val_ac
Epoch 24/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.3221 - acc: 0.8212 - f1: 0.8119 - val_loss: 2779.5212 - val_ac
Epoch 25/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.2907 - acc: 0.8160 - f1: 0.8082 - val_loss: 2779.5106 - val_ac
Epoch 26/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.2590 - acc: 0.8418 - f1: 0.8311 - val_loss: 2779.4767 - val_ac
Epoch 27/70
3837/3837 [==============================] - 12s 3ms/step - loss: 2779.2405 - acc: 0.8389 - f1: 0.8290 - val_loss: 2779.4700 - val_ac
Epoch 28/70
```

```python
import matplotlib.pyplot as plt

# Training & Validation accuracy
train_loss = history.history['loss']
val_loss = history.history['val_loss']
train_acc = history.history['acc']
val_acc = history.history['val_acc']
epochs = len(train_loss)

xc = range(epochs)

plt.figure(figsize=(10, 3))

# Loss subplot
plt.subplot(1, 2, 1)
plt.plot(xc, train_loss, label='Training Loss')
plt.plot(xc, val_loss, label='Validation Loss')
plt.xlabel('Number of Epochs', fontsize=10)
plt.ylabel('Loss', fontsize=10)
plt.title('Training and Validation Loss', fontsize=12)
plt.legend(fontsize=8)
plt.grid(True)

# Accuracy subplot
plt.subplot(1, 2, 2)
plt.plot(xc, train_acc, label='Training Accuracy')
plt.plot(xc, val_acc, label='Validation Accuracy')
plt.xlabel('Number of Epochs', fontsize=10)
plt.ylabel('Accuracy', fontsize=10)
plt.title('Training and Validation Accuracy', fontsize=12)
plt.legend(fontsize=8, loc='lower right')  # Change position to lower right
plt.grid(True)
```

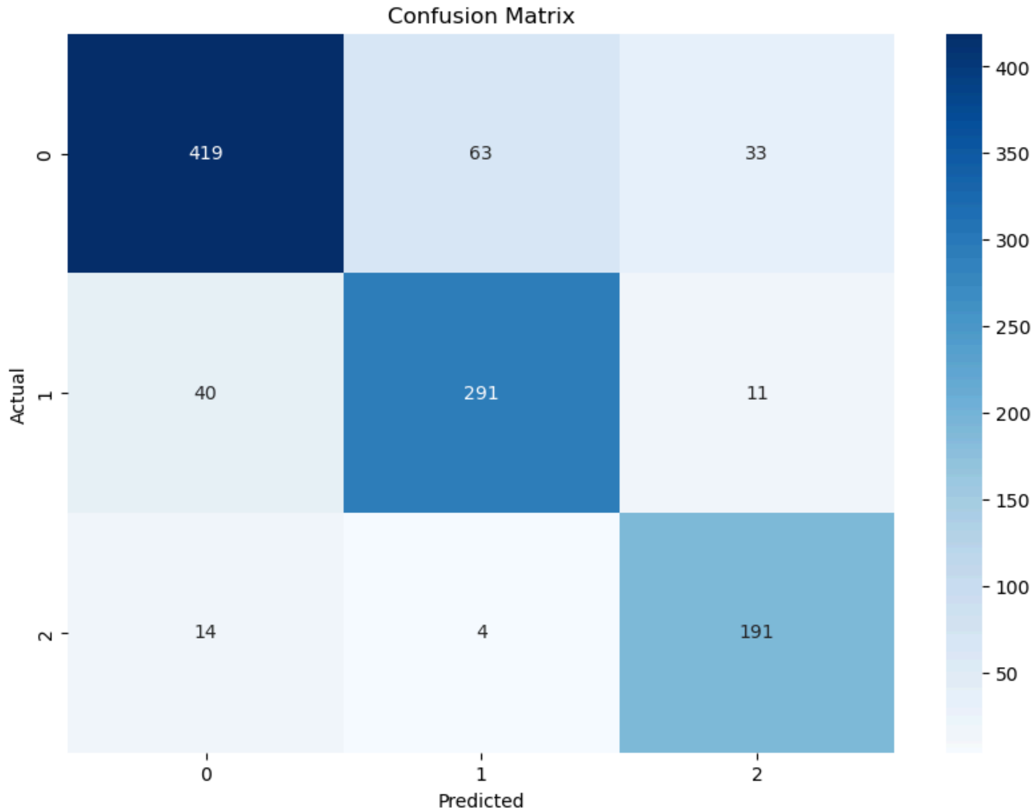
```
plt.tight_layout()
plt.show()
```




```
predicted = np.argmax(model.predict([X_test,pos_test1,pos_test2]), axis=1)
y_test_to_label = np.argmax(y_test, axis=1)
prec, reca, fscore, sup = precision_recall_fscore_support(y_test_to_label, predicted, average=param)
# Generate the classification report as a dictionary
report_dict = classification_report(y_test_to_label, predicted, output_dict=True)

# Create a new dictionary to hold the formatted values
formatted_report_dict = {}

# Iterate over the items in the report dictionary
for key, value in report_dict.items():
    if isinstance(value, dict):
        # Format the nested dictionary values
        formatted_report_dict[key] = {sub_key: f"{sub_value:.4f}" for sub_key, sub_value in value.items()}
    else:
        # Format the top-level dictionary values
        formatted_report_dict[key] = f"{value:.4f}"

# Create a string representation of the formatted dictionary
formatted_report_str = classification_report(y_test_to_label, predicted, digits=4)

# Print the formatted classification report
print(formatted_report_str)
print(" Precision:{:.2f}% Recall:{:.2f}% Fscore:{:.2f}% ".format(prec*100, reca*100, fscore*100))
```

```
              precision    recall  f1-score   support

           0     0.8858    0.8136    0.8482       515
           1     0.8128    0.8509    0.8314       342
           2     0.8128    0.9139    0.8604       209

    accuracy                         0.8452      1066
   macro avg     0.8372    0.8594    0.8467      1066
weighted avg     0.8481    0.8452    0.8452      1066

 Precision:83.72% Recall:85.94% Fscore:84.67%
```

```
import numpy as np
import matplotlib.pyplot as plt
import seaborn as sns
from sklearn.metrics import classification_report, confusion_matrix, precision_recall_fscore_support
from sklearn.model_selection import StratifiedKFold
# Calculate and visualize the confusion matrix
cm = confusion_matrix(y_test_to_label, predicted)
plt.figure(figsize=(10, 7))
sns.heatmap(cm, annot=True, fmt='d', cmap='Blues', xticklabels=['0', '1','2'], yticklabels=['0', '1','2'])
plt.xlabel('Predicted')
plt.ylabel('Actual')
plt.title('Confusion Matrix')
plt.show()

# Print precision, recall, and f-score
prec, reca, fscore, sup = precision_recall_fscore_support(y_test_to_label, predicted, average=param)
print(" Precision:{:.2f}% Recall:{:.2f}% Fscore:{:.2f}% ".format(prec*100, reca*100, fscore*100))
```




```
Precision:83.72% Recall:85.94% Fscore:84.67%
```

Start coding or generate with AI.