Convolutional Neural Network approach for Cancer subtype Prediction

Naresh Santosh Shet,Rakesh U,Akhileshkumar Patil,K.Sai Sudhir

Information Science and Engineering ,RVITM Bangaluru, India

Abstract - Cancer subtype prediction is a crucial aspect of cancer diagnosis and treatment, as it can inform personalized treatment plans and improve patient outcomes. It can be categorized in many forms. If we detect the subtype of cancer in the early stage it helps in many ways. We will be majorly focusing on 30 types of cancer some of them are Adrenocortical carcinoma, Bladder Urothelial Carcinoma, Breast invasive carcinoma, Ovarian serous cystadenocarcinoma, Pancreatic adenocarcinoma, Pheochromocytoma and Paraganglioma, Prostate adenocarcinoma, Sarcoma, Skin Cutaneous Melanoma.

In this, we will be creating a deep learning model where the TCGA RNA-Seq dataset is used to train the model. Initially, the taken data will go through pre-processing. Where Missing values have been added and null values have been dropped followed by Feature selection and Normalization. Using the data processed Heat map will be created. The gene value will be stored in the NumPy array and then using matplotlip show () is used to create 2-Dimentional arrays. Using those data model will be trained and accuracy been calculated. Our findings suggest that our predictive model can be a valuable tool in guiding personalized cancer treatment and improving patient outcomes.

Index Terms – Genetic Algorithm (GA), Convolution Neural Network (CNN), The Cancer Genome Atlas Program (TCGA) , Area Under the Curve (AUC), Receiver Operating Characteristic (ROC)

I. INTRODUCTION

Cancer subtype prediction is a critical task in cancer research that involves using advanced computational and analytical techniques to classify tumors based on their genetic and molecular characteristics. Cancer is a complex disease, and the different subtypes of cancer can have unique clinical and biological features, which can impact the diagnosis, prognosis, and treatment of patients. Identifying the correct subtype of cancer is essential for personalised and effective treatment.

The subtype classification process involves analyzing the genomic, transcriptomic, and proteomic profiles of cancer cells to identify specific molecular features and biomarkers that are characteristic of different cancer subtypes. Machine learning algorithms and other computational methods are then applied to the data to develop predictive models that can accurately classify tumors into specific subtypes. These models can be used to guide clinical decision-making and develop targeted therapies that are tailored to the specific molecular features of each patient's tumor.

Cancer subtype prediction is a rapidly evolving field that has the potential to transform cancer diagnosis and treatment. Advances in genomics, bioinformatics, and machine learning are enabling researchers to identify new biomarkers and develop more accurate predictive models, which can help improve patient outcomes and reduce the burden of cancer worldwide.

II. RELATED WORKS

For classifying the subtypes of cancer the authors of research papers used the GA/KNN approach.The main algorithms used as a base for subtype classification is the genetic algorithm(GA) and the KNN algorithm. These algorithms were capable of identifying multiple groups of genes which could accurately categorise over majority of data from multiple tumours in a dataset just by using RNA-Seq of genes.

To help identify and categorize subtypes of cancer the authors of paper made use of unsupervised learning with the help of data from TCGA.The main advantage of this approach is the ability to automatically create features from data from many types of cancer to help in identifying a particular type.

The authors of research papers have also made use of TCGA dataset to identify 30+ different types of cancer. The results show that the overall accuracy is 95.8%.

The authors of research papers also made use of TCGA data from about 31 various types of cancer patients, as well as healthy tissue RNA Seq data from GTEx.The input for this training model is the expression data of selected genes. This data is then converted into RGB colours by converting gene expression levels into a binary format. This algorithm has an accuracy of 97%.

III. METHODS

Here the dataset is prepared which will be used for training the model so that the subtypes of cancer could be predicted as accurately as possible. The main aim of our project is proper identification of subtypes of cancer with good accuracy. Data was obtained from UCSE Xena platform which includes RNA-Seq data from resources like TCGA.

The data undergoes pre-processing to ensure the accuracy of the model during prediction. The various pre-processing techniques involved are removal of null values present in the dataset, selection of ideal genes that meet the requirements from the available genes. The final pre-processing step that the dataset undergoes is normalizing the selected genes.The dataset undergoes pre-processing to make sure that there is no redundant data present in the dataset and thus ensuring efficient prediction of subtypes of data.

The given input that is obtained after the pre-processing is converted into heat maps. To create heat maps the data present in the CSV file is first transposed. The patient ids are represented in rows and various types of genes are represented in columns. The gene values are now sent to a array which is converted into images using matplotlib function. The main reason for conversion of gene values into images is that it makes things easier for the model to accurately predict the subtypes of cancer.

The usage of images enables us to have a clear understanding about the dataset through just one look. Before starting the training of the model it is ensured that the heat map images are of specific order i.e. 244*244 pixels. The CNN models takes ideal samples from different tumour labels. The samples are split in a specific ration so that it could be used for both testing and training.

The main objective of our project is to ensure that our CNN model can properly identify the subtypes of cancer and to improve its accuracy as much as possible when compared with the existing model that is used for prediction of subtypes of cancer.

Layer (type)	Output Shape	Param #
conv2d (Conv2D)	(None, 224, 224, 16)	448
max_pooling2d (MaxPooling2D)	(None, 112, 112, 16)	0
batch_normalization (BatchNormalization)	(None, 112, 112, 16)	64
conv2d_1 (Conv2D)	(None, 112, 112, 32)	4640
max_pooling2d_1 (MaxPooling2D)	(None, 56, 56, 32)	0
batch_normalization_1 (BatchNormalization)	(None, 56, 56, 32)	128
conv2d_2 (Conv2D)	(None, 56, 56, 64)	18496
max_pooling2d_2 (MaxPooling2D)	(None, 28, 28, 64)	0
batch_normalization_2 (BatchNormalization)	(None, 28, 28, 64)	256
conv2d_3 (Conv2D)	(None, 28, 28, 64)	36928
max_pooling2d_3 (MaxPooling2D)	(None, 14, 14, 64)	0
batch_normalization_3 (BatchNormalization)	(None, 14, 14, 64)	256
conv2d_4 (Conv2D)	(None, 14, 14, 128)	73856
max_pooling2d_4 (MaxPooling2D)	(None, 7, 7, 128)	0
batch_normalization_4 (BatchNormalization)	(None, 7, 7, 128)	512
conv2d_5 (Conv2D)	(None, 7, 7, 128)	147584
max_pooling2d_5 (MaxPooling2D)	(None, 3, 3, 128)	0
batch_normalization_5 (BatchNormalization)	(None, 3, 3, 128)	512
conv2d_6 (Conv2D)	(None, 3, 3, 256)	295168
max_pooling2d_6 (MaxPooling2D)	(None, 1, 1, 256)	0
batch_normalization_6 (BatchNormalization)	(None, 1, 1, 256)	1024
conv2d_7 (Conv2D)	(None, 1, 1, 256)	590080
max_pooling2d_7 (MaxPooling2D)	(None, 1, 1, 256)	0
batch_normalization_7 (BatchNormalization)	(None, 1, 1, 256)	1024
flatten (Flatten)	(None, 256)	0
dense (Dense)	(None, 33)	8481
dropout (Dropout)	(None, 33)	0
dense_1 (Dense)	(None, 33)	1122

Total params: 1,180,579
Trainable params: 1,178,691
Non-trainable params: 1,888

Table 1 Architecture of CNN Model

3.1 CNN TRAINING

Table and Figure represent the results of training a deep learning architecture on a dataset of 17,906 samples that are evenly distributed between normal and tumor labels. The dataset is split into training and testing sets using an 80:20 ratio. The deep learning architecture is optimized to classify the cancer subtypes based on gene expression data.

The deep learning architecture used in this study is not specified in the information provided, but it likely involves a combination of convolutional layers, pooling layers, and fully connected layers. These layers work together to extract features from the gene expression data and classify the samples into normal and tumor subtypes.

After 40 epochs of training, the accuracy of the model on the testing set reached 97.7%, which is a high level of accuracy and suggests that the model is able to accurately classify the cancer subtypes based on the gene expression data. The accuracy and loss plots shown in Figure indicate that the model is performing well and is not overfitting to the training data.

Overall, these results are promising and suggest that the deep learning architecture is effective for cancer subtype prediction based on gene expression data. However, it is important to note that further evaluation and validation is necessary before this approach can be used in clinical practice.

3.2 PERFORMANCE MEASUREMENT

Figure displays the Receiver Operating Characteristic (ROC) curve for our model, and the Area Under the Curve (AUC) value was calculated to be 0.97. We also computed additional performance measures from the confusion matrix generated by test sample predictions. Our model achieved a precision and recall of 98% for tumor prediction, as shown in Table .

In the literature, various approaches have been proposed for classifying tumor and normal samples based on gene expression data, ranging from simpler machine learning methods to more complex deep learning networks. Typically, these approaches involve preprocessing the gene expression data through an irreversible manipulation such as normalization and mapping data points to a different domain using techniques like Principal Component Analysis (PCA) or t-Distributed Stochastic Neighbor Embedding (t-SNE). In contrast, our method involves a minimal and reversible change to the gene expression data. The RGB mapping is reversible and does not require normalization or any dimensionality reduction techniques. Table presents a comparison of our approach with several other approaches, both in terms of preprocessing and classification steps. Although the study by Elbashir et al (Normalization + CNN) achieved the highest accuracy, their sample dataset was problematic, and our approach produced better overall results.

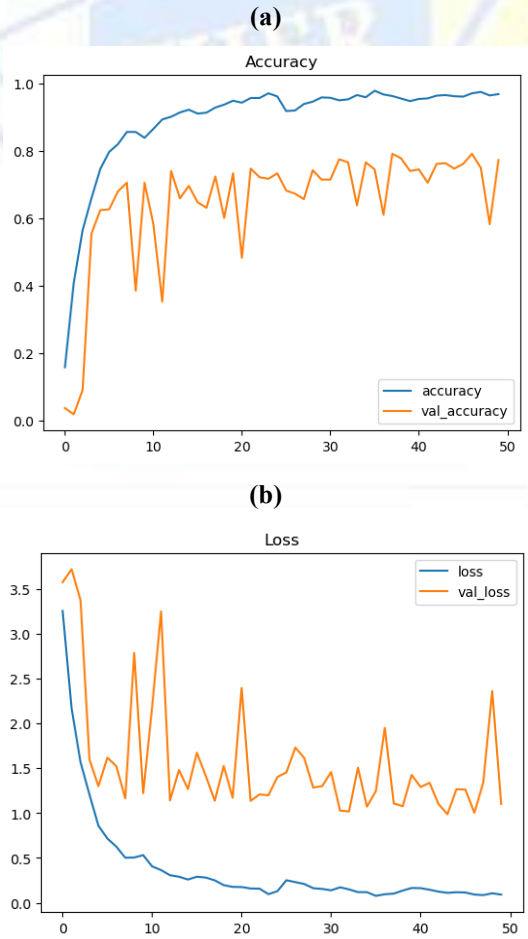


Figure 3. Model accuracy (a) and loss (b) plots.

Accuracy:	0.75918
Precision:	0.78247
Recall:	0.75918
F1 Score:	0.75419
Cohen Kappa Score:	0.7512

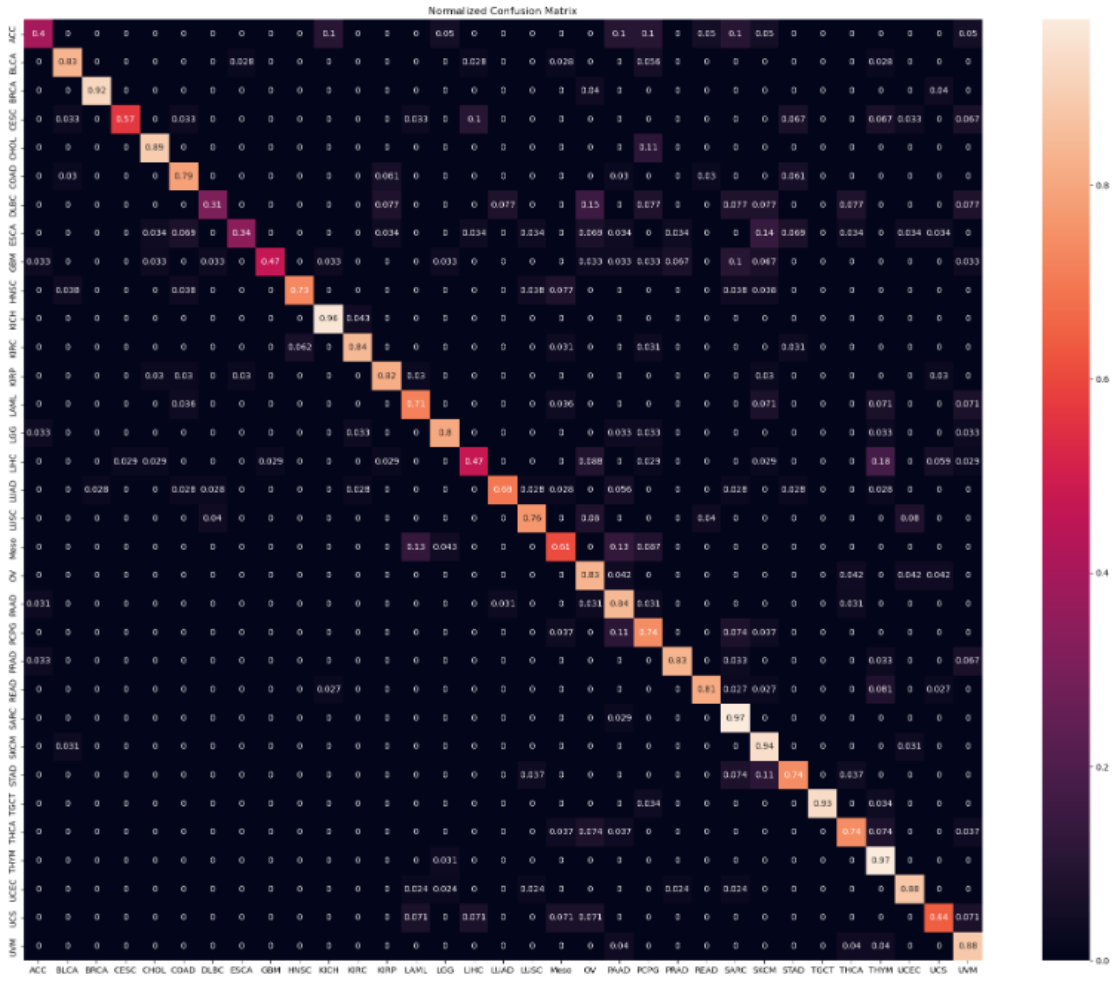
Table 2. Accuracy, Precision, Recall, F1 score, Cohen kappa Score

	precision	recall	f1-score	support
ACC	0.67	0.40	0.50	20
BLCA	0.88	0.83	0.86	36
BRCA	0.96	0.92	0.94	25
CESC	0.94	0.57	0.71	30
CHOL	0.67	0.89	0.76	9
COAD	0.79	0.79	0.79	33
DLBC	0.57	0.31	0.40	13
ESCA	0.83	0.34	0.49	29
GBM	0.93	0.47	0.62	30
HNSC	0.90	0.73	0.81	26
KICH	0.85	0.96	0.90	23
KIRC	0.90	0.84	0.87	32
KIRP	0.84	0.82	0.83	33
LAML	0.74	0.71	0.73	28
LGG	0.83	0.80	0.81	30
LIHC	0.73	0.47	0.57	34
LUAD	0.93	0.69	0.79	36
LUSC	0.79	0.76	0.78	25
Meso	0.61	0.61	0.61	23
OV	0.57	0.83	0.68	24
PAAD	0.60	0.84	0.70	32
PCPG	0.59	0.74	0.66	27
PRAD	0.86	0.83	0.85	30
READ	0.91	0.81	0.86	37
SARC	0.69	0.97	0.80	34
SKCM	0.62	0.94	0.75	32
STAD	0.71	0.74	0.73	27
TGCT	1.00	0.93	0.96	29
THCA	0.77	0.74	0.75	27
THYM	0.60	0.97	0.74	32
UCEC	0.86	0.88	0.87	41
UCS	0.56	0.64	0.60	14
UVM	0.63	0.88	0.73	25

Precision, Recall and F1-Score for each of the 33 Cancer classes

accuracy			0.76	926
macro avg	0.77	0.75	0.74	926
weighted avg	0.78	0.76	0.75	926

Overall Accuracy of Model



Confusion matrix

IV. RESULT

Accuracy of CNN Model is 74.83%.

V. CONCLUSION

Cancer is a complex disease that can manifest in various forms, with different subtypes exhibiting unique characteristics and behaviors. It is crucial to identify these subtypes accurately and efficiently to develop personalized treatment plans for cancer patients. Traditional methods of identifying cancer subtypes involve time-consuming and labor-intensive procedures that may not yield accurate results. Therefore, there is a need for novel approaches that can expedite the identification process and provide accurate results.

In recent years, deep learning-based models have gained popularity in the field of cancer research due to their ability to process large datasets and extract meaningful insights. In this paper, the researchers implemented a convolutional neural network (CNN) model based on deep learning and tested it on the TCGA RNA-Seq dataset to identify cancer subtypes. The TCGA RNA-Seq dataset is a multiclass dataset that contains gene expression profiles of several cancer types.

The proposed CNN model uses a deep learning approach to extract features from the gene expression data and classify the cancer subtypes accurately. The model achieved a test accuracy of 74.83%, which indicates its potential to identify cancer subtypes quickly and efficiently. The results of this study suggest that deep learning-based models can provide accurate and efficient identification of cancer subtypes, which can aid in the development of personalized treatment plans for cancer patients.

Overall, the proposed CNN model provides a promising approach to identify cancer subtypes quickly and efficiently. The use of deep learning-based models in cancer research is an exciting area of study that has the potential to revolutionize cancer treatment and improve patient outcomes.

V. REFERENCES

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