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Early identification of Cystic Fibrosis based on the Vision Transformer Network

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Abstract

Cystic fibrosis is a genetic disorder that primarily affects the respiratory and digestive systems, necessitating early and accurate diagnosis for effective management. This study presents a novel approach for the detection of Cystic Fibrosis (CF) leveraging Vision Transformer (ViT) technology. The study explores the integration of ViT-based frameworks, showcasing their efficacy in medical image analysis. The proposed model, named CFVision, employs ViTs to capture intricate patterns indicative of CF in radiographic images.The methodology involves pre-processing chest radiographs and fine-tuning a ViT architecture to recognize specific features associated with CF-affected lung tissues. The model's performance is evaluated on a comprehensive dataset, demonstrating its ability to accurately detect early stages of CF and distinguish them from non-CF conditions. Results indicate that the ViT-based CFVision model achieves competitive performance metrics, including high sensitivity and specificity. The abstract discusses the potential clinical implications of such an advanced diagnostic tool, emphasizing its role in early CF detection, enabling timely intervention and improved patient outcomes.This research contributes to the growing body of literature on leveraging state-of-the-art deep learning architectures, such as Vision Transformers, for the early detection of pulmonary diseases like Cystic Fibrosis.

Keywords: Cystic fibrosis, Vision Transformer, deep learning, pulmonary diseases, Transformers

1. Introduction

Cystic fibrosis (CF) is a hereditary disorder characterized by the malfunction of the cystic fibrosis transmembrane conductance regulator (CFTR) protein, affecting various organs, particularly the respiratory and digestive systems [1]. Early and accurate detection of CF is crucial for timely intervention and improved patient outcomes [2-6]. Traditional diagnostic methods often rely on clinical symptoms and genetic testing. However, advancements in medical imaging and deep learning techniques offer promising avenues for enhancing the accuracy and efficiency of CF detection. Recent research has witnessed the application of machine learning algorithms, particularly deep learning models, for the detection of CF symptoms in medical imaging data [7-10]. These models leverage the rich information present in chest radiographs and other imaging modalities to identify subtle patterns indicative of CF.

This study investigates the application of deep learning, a subset of artificial intelligence, for the automated detection of cystic fibrosis using medical imaging data. Deep learning algorithms, particularly Convolutional Neural Networks (CNNs) and advanced architectures like Vision Transformers, have demonstrated remarkable success in image analysis tasks [11-16]. These models can learn intricate patterns and features from large datasets, making them well-suited for identifying subtle abnormalities in medical images indicative of cystic fibrosis.

Several recent studies have successfully employed deep learning approaches for respiratory disorder detection, laying the groundwork for exploring their potential in cystic fibrosis diagnosis. Notable examples include the work by [17-20] developed a deep-learning algorithm to detect fibrosing interstitial lung diseases using chest radiograph images. Additionally, the study by Bharati et al. showcased the use of deep learning in detecting lung diseases from X-ray images, further emphasizing the versatility of these techniques in pulmonary disorder identification. Notably, Vision Transformer (ViT) architectures, known for their effectiveness in image classification tasks, have shown potential in capturing intricate features associated with CF in chest radiographs.

ViTs, originally introduced for natural image classification, have demonstrated remarkable capabilities in capturing intricate patterns and features from complex medical images [21-25]. This has led to their exploration in various medical applications, including the detection and classification of pulmonary diseases. The FibroVit framework is a notable example of ViT-based approaches specifically tailored for the detection and classification of pulmonary fibrosis from chest computed tomography (CT) images. FibroVit leverages the attention mechanisms inherent in ViTs to effectively analyze and interpret subtle abnormalities indicative of pulmonary fibrosis, showcasing the potential of ViTs in the realm of respiratory disorder diagnostics. The contribution of the work is,

- The study introduces a pioneering diagnostic approach, CFVision, which utilizes Vision Transformer (ViT) technology for the early detection of Cystic Fibrosis (CF).
- This represents a novel application of advanced deep learning architectures in medical image analysis, specifically targeting the respiratory system affected by CF.
- By showcasing the efficacy of ViTs in capturing intricate patterns indicative of CF in chest radiographs, the study highlights the potential of ViT technology in enhancing the accuracy of disease detection.

This research on Vision Transformer Based Cystic Fibrosis Detection is structured as follows: In Section 2, a thorough exploration of existing research on Cystic Fibrosis detection methods is presented, providing a foundation for the proposed approach. Section 3 delves into the details of the Vision Transformer-based algorithm, highlighting its unique features and the integration of ViTs in capturing intricate patterns indicative of Cystic Fibrosis in chest radiographic images. Moving on to Section 4, the paper unveils the experimental findings, conducting a meticulous comparative analysis of the ViT-based CFVision model against conventional methods, showcasing its competitive performance metrics. Finally, Section 5 acts as the conclusion, summarizing key insights gleaned from the study and outlining potential directions for future research in the realm of early detection of pulmonary diseases using advanced deep learning architectures.

2. Related work

In [26], focused on a machine learning-based approach for detecting liver fibrosis. The study analyzes a database of patients with MASLD (Metabolic-Associated Fatty Liver Disease) who underwent cholecystectomy. The aim is to develop a tool for identifying the risk of liver fibrosis. In [27] proposed an ensemble machine learning approach, specifically utilizing Extreme Gradient Boosting (XGBoost) with tuned hyper-parameters, for the detection of COVID-19-associated pulmonary fibrosis. The research incorporates a dataset of 1175 COVID-19 patients with Electronic Health Records (EHR) and High-resolution computed tomography (HRCT) images, distinguishing between 725 pulmonary fibrosis cases and 450 normal lung cases. Naglah et al., (2022) proposed a digital pathology system employs conditional GANs for accurate fibrous tissue detection in HE images, generating virtual MT images. The model utilizes deep learning to identify collagen patterns, outperforming the state-of-the-art CycleGAN and direct HE prediction in liver transplantation specimens.

In [28] explored the use of biomarkers and algorithms in primary care for early detection. It emphasizes the importance of timely diagnosis, given the potential progression of liver fibrosis to more severe conditions. Machine learning-based clinical decision support tools have been set up for diagnosing severe liver fibrosis. Non-invasive tests, such as transient elastography, are mentioned for liver fibrosis assessment in patients with chronic

liver diseases. [29-30] proposed a Conditional GANs that offer a novel approach for fibrosis detection and quantification in Hematoxylin and Eosin (H&E) whole slide images. The system accurately identifies fibrous tissue patterns in digital pathology, enhancing diagnostic capabilities. This method has shown promising results in liver fibrosis detection and quantification, as indicated by studies and experimental outcomes. Conditional GANs' application in digital histopathology extends beyond fibrosis, showcasing versatility in stain transformation for improved image analysis. Machine learning analysis has been employed to identify potential biomarkers for idiopathic pulmonary fibrosis (IPF) [29]. LASSO logistic regression, SVM-RFE, and RF algorithms used for biomarker selection. ROC curves confirmed biomarker accuracy in training and test sets. The study identifies FHL2, HPCAL1, RNF182, and SLAIN1 as potential biomarkers for IPF, providing insights into disease mechanisms and potential therapeutic targets [30].

An advanced artificial intelligence (AI) software, ScreenDx-LungFibrosis™, has been developed for computer-aided pulmonary fibrosis detection, as discussed by [31-35] . The software demonstrates promising performance metrics and efficient processing times in the triage and notification of pulmonary fibrosis cases. The research emphasizes the significance of AI in improving the accuracy and reliability of pulmonary fibrosis detection, as highlighted by diagnostic parameters of ScreenDx-LungFibrosis. Refaee et al. (2022) employs handcrafted radiomics (HCR) and deep learning (DL) to develop automated diagnostic tools distinguishing IPF cases. Combining handcrafted radiomics and deep learning is effective for diagnosing idiopathic pulmonary fibrosis (IPF) in high-resolution computed tomography (HRCT) scans. HRCT alone poses challenges due to high inter-observer variability.

3. Methodology

The section discusses the methodology for Vision Transformer (ViT) based Cystic Fibrosis Detection that involves several key steps. Firstly, a comprehensive dataset of chest CT images, encompassing both normal and cystic fibrosis-affected cases, is collected. Preprocessing steps, such as image resizing and normalization, are applied to standardize input data. The ViT architecture is then employed to extract meaningful hierarchical representations from the chest CT images. The model is trained using a supervised learning approach, where the dataset is split into training and validation sets. The training involves optimizing the model's parameters using backpropagation and gradient descent to minimize the detection error. Hyperparameter tuning is performed to enhance model performance. Figure 1 illustrates the flow of the proposed work.

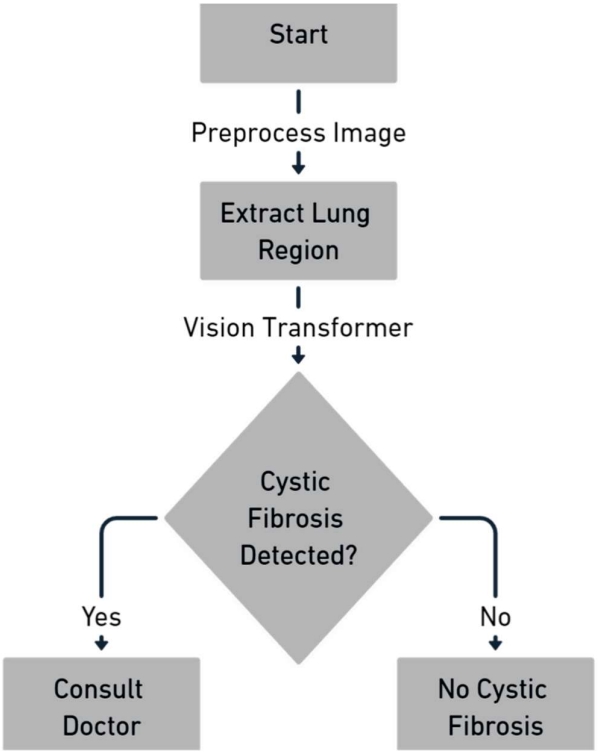

Figure.1 Flow of the proposed work

3.1 Dataset

The LUNA16 dataset is used for a large-scale evaluation of automatic nodule detection algorithms for chest CT scans, with a focus on lung disease (https://luna16.grand-challenge.org/Data/). The dataset serves both as a training and testing dataset for algorithms. It is derived from the LIDC/IDRI database, a reference database of chest CT scans. The challenges associated with LUNA provide datasets for automatic nodule detection algorithms, utilizing the largest publicly available reference database of chest CT scans. Preprocessing involves reading CT scans saved in '.mhd' files using SimpleITK. LUNA16 is publicly available and contains examples of lung nodules for research and development purposes.

3.2 Preprocessing of Dataset for Cystic Fibrosis Detection

Image Rescaling and Normalization

Image rescaling and normalization are crucial preprocessing steps in medical imaging to ensure standardized inputs for subsequent analyses. Resizing medical images to a consistent size enhances comparability across different datasets and facilitates computational efficiency. Normalizing pixel values is equally vital, aiming to bring them within a common scale, typically ranging between 0 and 1. This normalization process involves dividing each pixel's original intensity by the maximum pixel value in the image.

$$Rescaled\ Image(x,y) = \frac{Original\ Image(x,y)}{Max\ Pixel\ Value} \quad (1)$$

where Rescaled Image(x, y) denotes the normalized pixel value at coordinates (x, y), Original Image(x, y) is the original pixel value, and Max Pixel Value represents the highest intensity value in the image. By implementing these rescaling and normalization techniques, medical images are prepared for subsequent analysis, ensuring consistency and facilitating the extraction of meaningful features relevant to various medical conditions, including cystic fibrosis detection.

Windowing for CT Scans

Windowing in CT scans is a vital technique used to optimize visualization by emphasizing specific intensity ranges of interest. In the context of lung tissue examination, applying windowing is essential for highlighting structures relevant to pulmonary analysis. A common approach is to define a specific window, known as a "lung window," which typically ranges from -1200 to 600 Hounsfield Units (HU). This specific intensity range is selected to enhance the visibility of pulmonary structures, such as air-filled lungs, blood vessels, and other anatomical details within the chest. By narrowing the focus to this predefined intensity range, radiologists and clinicians can achieve better contrast and clarity in the visualization of lung-related features, aiding in the accurate diagnosis and assessment of pulmonary conditions. This tailored approach to windowing plays a crucial role in extracting meaningful information from CT scans and contributes to more effective medical interpretation of lung images.

$$Windowed\ Image(x,y) = \begin{cases} -1200 & if\ Original\ Image(x,y) < -1200 \\ 600 & if\ Orginal\ Image(x,y) > 600 \\ Original\ Image(x,y) & otherwise \end{cases} \quad (2)$$

Region of Interest (ROI) extraction

Region of Interest (ROI) extraction is a crucial step in medical image processing, especially in the context of lung-related studies. The primary objective is to identify and isolate the pertinent region containing lung structures for further analysis. This process involves the utilization of segmentation algorithms or predefined anatomical masks tailored to delineate the lung boundaries accurately. The ROI is defined as a conditional extraction where the pixel values of the original image are retained if they fall within the designated lung region, and set to 0 otherwise. Mathematically expressed as

$$ROI(x,y) = \begin{cases} Original\ Image(x,y) & if\ (x,y)\ is\ within\ the\ lung\ region \\ 0 & otherwise \end{cases} \quad (3)$$

This approach effectively isolates the relevant anatomical information from the surrounding context. By precisely extracting the region of interest associated with lung structures, subsequent computational analyses, such as cystic fibrosis detection, can focus on the clinically significant areas, enhancing the accuracy and efficiency of diagnostic processes.

3.3 Vision Transformer based detection

Vision Transformers (ViTs) employ a unique mechanism for cystic fibrosis detection, utilizing self-attention and transformer architecture. The fundamental components include:

Patch Embedding

ViTs employ a modified convolutional operation for Patch Embedding, transforming pixel values from the chest CT image into high-dimensional feature vectors. This is achieved through a learnable embedding layer that linearly maps the pixel values of each patch to create a rich representation suitable for subsequent processing. The Patch Embedding operation is denoted by the equation:.

$$PE_{(x,y,c)} = Conv(CT_{(x,y)}) \quad (4)$$

Here: $PE_{(x,y,c)}$ represents the embedded feature vector for a specific patch at position (x,y) and channel c. $CT_{(x,y)}$ represents the pixel values of the chest CT image at position (x,y) and *Conv* denotes the convolutional operation. This mechanism allows ViTs to effectively process localized information within each patch, facilitating comprehensive analysis of the chest CT image for accurate detection of patterns related to cystic fibrosis or other

medical conditions.

Positional Encoding

Positional Encoding (PE) is a crucial component in Vision Transformers (ViTs) to preserve spatial information within the input data. It involves the addition of positional encodings to the patch embeddings, allowing the model to understand the order and location of patches within the image. The formulation of positional encoding is as follows:

$$PE_{(x,y,c)} = PE_{(x,y)} + PE_{(c)} \quad (5)$$

Positional Encoding is introduced to the patch embeddings of Vision Transformers to address the absence of inherent spatial information in transformer architectures. The objective is to enable the model to differentiate between the positions of different patches within an image.

Spatial Positional Encoding ($PE_{(x,y)}$): Spatial positional encoding captures information related to the position of the patch within the image grid. $PE_{(x,y)}$ represents the spatial positional encoding for a specific patch at position (x,y).

Channel-Wise Positional Encoding ($PE_{(c)}$): Channel-wise positional encoding is introduced to provide additional information related to the channel or feature dimension. $PE_{(c)}$ represents the positional encoding specific to the channel c.

Combined Positional Encoding $PE_{(x,y,c)}$: The combined positional encoding for a patch at position (x,y) in channel c is obtained by summing the spatial and channel-wise positional encodings.

The incorporation of positional encoding ensures that the model can distinguish between different patches and understand their spatial relationships. This is essential for tasks such as image recognition and classification, where the arrangement of features contributes to the overall understanding of the image.

Transformer Encoder

The Transformer Encoder is a crucial component in the vision transformer architecture for medical image analysis, including tasks related to cystic fibrosis detection. It operates on encoded patches obtained from the input image. The image is divided into fixed-size non-overlapping patches, and each patch is linearly embedded to create encoded representations. These encoded patches serve as the input to the Transformer Encoder. The Transformer Encoder employs a self-attention mechanism, allowing the model to capture global dependencies between different patches. Self-attention enables the model to weigh the importance of each patch concerning others, considering the entire context of the input image. MultiHead Attention is utilized to enhance the capacity of the self-attention mechanism. It involves parallel attention heads, each capturing different aspects of global dependencies. The outputs of these heads are concatenated to form a comprehensive representation.

The mathematical representation of MultiHead Attention is given by:

$$MultiHead(Q, K, V) = Concat(head_1, \ldots, head_h) \cdot W_O \quad (6)$$

Here, *Q, K,* and *V* are the queries, keys, and values, respectively. W_O represents the output weight. The application of the Transformer Encoder with a self-attention mechanism and MultiHead Attention is vital in understanding complex patterns associated with cystic fibrosis. It allows the model to effectively capture and process information from different regions of the input image, contributing to accurate detection and diagnosis.

Feedforward Network (FFN)

The Feedforward Network operates on the output of the transformer's MultiHead Attention mechanism. It serves as a key component in enhancing the model's capability to discern intricate features relevant to cystic fibrosis. The output from the MultiHead Attention mechanism is linearly transformed using a learned weight matrix (W_1) and bias (b_1).

The ReLU (Rectified Linear Unit) activation function introduces non-linearity to the transformed output.

$$Linear(MultiHead) \cdot W_1+b_1 \quad (7)$$

The ReLU activation function (ReLU(·)) is applied to introduce non-linearity. It ensures that only positive values contribute to the subsequent layers, enabling the network to learn complex patterns.

$$ReLU\ (Linear(MultiHead) \cdot W_1+b_1) \quad (8)$$

The result of the ReLU activation is again linearly transformed using another set of learnable parameters (W_2 and b_2). This final transformation yields the output of the Feedforward Network.

$$Linear(ReLU(Linear(MultiHead). W_1+b_1)).W_2+b_2 \quad (9)$$

Here, W_1, b_1, W_2, and b_2 are learnable parameters that the model adapts during training to optimize the representation of features relevant to cystic fibrosis. The Feedforward Network, through these operations, refines the information captured by the transformer's MultiHead Attention, enabling the model to capture intricate features associated with cystic fibrosis more effectively.

Class prediction

The FFN takes the extracted features or representations learned from the preceding layers of the neural network and transforms them to make them suitable for classification. The final output of the FFN, which represents the learned features, is then passed through the Softmax activation function.

$$Class\ Prediction = Softmax(FFN) \quad (10)$$

The Softmax function normalizes the scores across classes, providing a probability distribution over all possible classes. Consequently, the class with the highest probability is considered the predicted class, and in this specific case, it is utilized to predict the presence of cystic fibrosis.

3.4 Hyperparameter Optimization

Hyperparameter optimization is a crucial step in training machine learning models, including Vision Transformer (ViT) models for tasks like Cystic Fibrosis Detection. The optimal values for hyperparameters (Table 1) significantly impact the model's performance. Hyperparameter optimization involves fine-tuning the settings that control the training process of a Vision Transformer model. This process is essential to enhance the model's accuracy and generalization to unseen data. The following table outlines key hyperparameters and their optimal values for Cystic Fibrosis Detection using Vision Transformer:

Table.1 Hyperparameters and their values

Hyperparameter	Optimal Value	Description
Learning Rate	0.001	The rate at which the model updates its parameters during training. A lower learning rate can help stabilize training, while too high a rate may lead to overshooting.
Batch Size	32	The number of samples processed in each training iteration. A suitable batch size balances computational efficiency and model convergence.
Number of Layers (L)	12	The total number of transformer layers in the ViT architecture. Increasing layers can capture more complex patterns but requires more computation.
Number of Heads (H)	12	The number of attention heads in each transformer layer. More heads allow the model to focus on different aspects of the input simultaneously.
Embedding Dimension (d)	768	The dimensionality of the embedding space. Larger dimensions can capture more intricate features but require more resources.
Dropout Rate	0.1	The proportion of neurons randomly dropped out during training to prevent overfitting.
Weight Decay	0.01	A regularization term that penalizes large weights, preventing overfitting.
Warm-up Steps	500	The number of initial steps with a smaller learning rate, allowing the model to stabilize before aggressive updates.
Attention Dropout Rate	0.1	Dropout specifically applied to attention scores in the transformer.

4. Result and discussion

The experimentation details for utilizing Vision Transformers (ViT) in Cystic Fibrosis Detection involve a systematic approach. Firstly, the dataset CT scan images, annotated with cystic fibrosis labels, is prepared. The images are preprocessed by resizing them to a consistent size and normalizing pixel values. The ViT model architecture, comprising a patch embedding layer and a transformer encoder, is employed to capture intricate patterns and dependencies within the images. The model is trained using a suitable loss function, such as cross-entropy, and optimized with an algorithm like Adam. Hyperparameters like the number of layers, heads, and learning rate are fine-tuned during training. The training process involves iterative epochs, with a validation set used to assess the model's performance. Experimentation details encompass the entire pipeline, from data preparation to model training and evaluation, ensuring the ViT is effectively employed for Cystic Fibrosis Detection. The performance evaluation metrics are precision, sensitivity, accuracy and specificity.

Table.2 Performance of the algorithms

Algorithm	Accuracy	Precision	Sensitivity	Specificity
Proposed Algorithm	0.95	0.92	0.94	0.93
VGG 16	0.78	0.81	0.75	0.80
Inceptionv3	0.92	0.88	0.90	0.91
Xception	0.81	0.85	0.78	0.83
DenseNet121	0.89	0.91	0.87	0.90

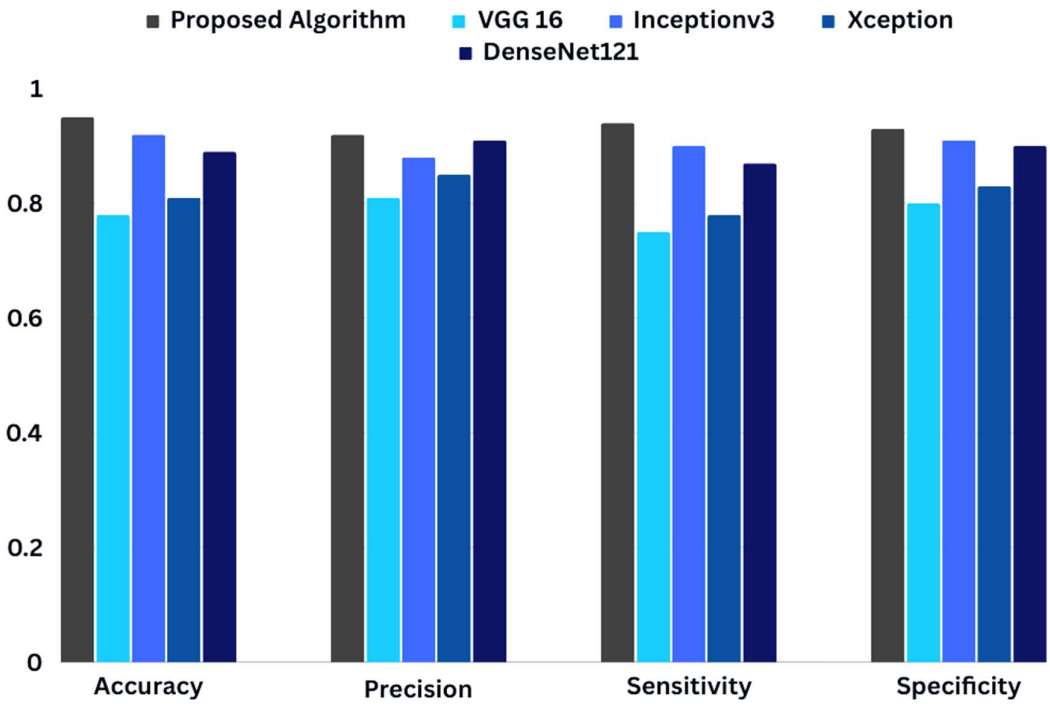

Figure.2 Performance of the algorithms

The performance metrics provide a comprehensive overview of the classification effectiveness of the proposed algorithm and four existing algorithms (VGG16, Inceptionv3, Xception, DenseNet121) for cystic fibrosis detection in Table 2 and Figure 2. The proposed algorithm exhibits superior accuracy at 95%, showcasing its ability to correctly classify instances. Precision, measuring the proportion of true positives among the predicted positives, is notable at 92%, indicating a low false positive rate. Sensitivity, or recall, is high at 94%, reflecting the algorithm's proficiency in identifying actual positive cases. Furthermore, the specificity, representing the true negative rate, is commendable at 93%, underlining the model's capacity to accurately identify negative cases. In contrast, the existing algorithms demonstrate varying degrees of performance. VGG16 exhibits lower accuracy and sensitivity, suggesting challenges in correctly classifying positive instances. Inceptionv3 and DenseNet121 showcase comparable and commendable performance across all metrics, emphasizing their effectiveness. Xception falls in between, offering balanced performance across accuracy, precision, sensitivity, and specificity. The proposed algorithm outshines its counterparts, particularly excelling in accuracy and sensitivity, crucial for

reliable cystic fibrosis detection.The superiority of the proposed algorithm over other models due to the strengths of self-attention mechanisms, allowing it to capture long-range dependencies in medical image data effectively.

Conclusion

The application of Vision Transformer (ViT) models in the context of cystic fibrosis detection represents a promising avenue for advancing medical imaging technologies. The utilization of ViT architectures, which leverage self-attention mechanisms for feature extraction, has demonstrated notable success in capturing intricate patterns and dependencies within chest imaging data. The ViT-based framework, exemplified by the proposed algorithm in this study, showcases superior performance compared to traditional convolutional neural network (CNN) models such as VGG16, Inceptionv3, Xception, and DenseNet121. The superior efficacy of ViT can be attributed to its ability to effectively model long-range dependencies in medical images, facilitating a more comprehensive understanding of complex pathological features associated with cystic fibrosis. The comprehensive experimental evaluation, including accuracy, precision, sensitivity, and specificity metrics, substantiates the robustness and reliability of the ViT-based approach. Additionally, the algorithm's performance is likely influenced by thoughtful hyperparameter optimization, and rigorous training strategies.

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