

Machine Learning and Deep Learning Approaches for Anomaly Detection in Medical Image Analysis: Advancing Diagnostic Healthcare

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Abstract: -Anomaly is an event or a behaviour that deviates from the normal, in this situation the problem arises that need to be solved. however, the underlying causes remains elusive. And it is very important to detect these anomalies. The definition of anomaly varies between domains. This paper addresses the medical anomaly which is a challenge in medical imaging modalities that need to be resolved for accurate disease diagnosis, Medical anomaly can be described as a structural or functional abnormality that is ambiguous. This paper describes various ML and DL approaches for anomaly detection for various diseases using imaging modalities and help researchers and medical practitioners to choose an appropriate technique for diagnosing the disease early with high accuracy.

Keywords: Medical anomalies, Anomaly detection, Medical imaging modalities, machine learning, deep learning

1. Introduction

Medical anomalies are referred to deviations that are considered abnormal or typical in the human body. These anomalies can be present at birth known as congenital anomalies or can be developed later. They are often sign of a disease, abnormalities or malfunctions of organs and tissues. Further medical anomalies can also be evident of various issues such as structural deformities, strange pattern in medical images, abnormal lab results or improper physiological functions. Anomaly detection in healthcare is crucial, it can be either fraud detection in health care [1] or detecting an error in a medical report. In medical diagnosis there are various diagnostic procedures that include non-invasive lab tests, imaging tests, and invasive endoscopy, biopsy. Using Machine learning model with medical imaging modalities like X-Ray, CT scan, MRI has a great potential to revolutionize the health care system [2]. Disease with high incident rates such as Diabetes, Heart disease, Alzheimer's, Neurological degeneration, brain tumours as well as very common cancers like Lung, breast cancer and so on has piqued the inquisitiveness of ML researchers worldwide. Machine learning uses prediction techniques in healthcare industry to enhance the prediction of disease [3][4]. In our study we discuss Medical imaging, the use of Machine Learning and Deep learning models and frameworks in disease diagnosis, and these models combined with image modalities have a great potential and are gaining worldwide recognition in biomedical research and clinical practices thus capable of even screening, prognosis and prediction of disease in healthcare system.

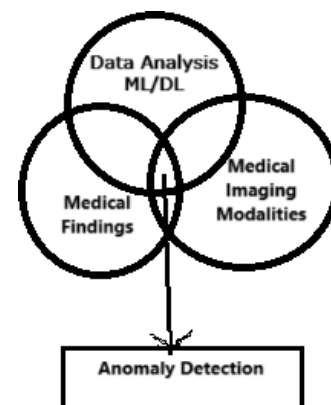


Fig 1. Anomaly Detection in Health care system

In fig 1. Using imaging modalities in conjunction with clinical results and data analysis methods such as machine learning, anomaly detection becomes transparent, effective for early disease detection and prediction.

2. Medical Imaging in Diagnostic Radiology

Medical imaging is a technique or practice of imaging internal organs or tissues of the body for clinical analysis and medical intervention. Radiology is a branch of medicine that combines medical imaging to diagnose and treat disease. There are different types of advanced imaging techniques like X-ray, CT (Computer Tomography), PET (Poisson Emission Tomography), MRI (Magnetic Resonance Imaging), Ultrasound etc.,

2.1. X-ray :- X-rays make use of a type of radiation called electromagnetic waves as these wave lengths are shorter the visible light can pass through most tissue types in human body. Because the calcium in bones is denser, x-rays are scattered by it. The negative image of the film, which is positioned across from the x-ray

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source, makes areas that are exposed to more light look darker. The lung and muscle seem darker and the portions of bone appear brighter as more x-rays pass through them. These X-rays are used for detecting bone fractures, tumours, pneumonia and dental issues.

2.2. Computer Tomography (CT) Scan: - It is also known as X-Ray CT. Hounsfield invented the CT scan prototype in 1969[5]. It produces cross-sectional images using narrow beam of X-rays. The obtained cross-sectional image slices are stacked together to produce 3D representation, they are more informative compared to that of conventional X-ray. CT scan is one of the best methods in detection and monitoring various types of cancers like kidney, bladder, neck, head and brain. They tend to be accurate in managing abdominal illness such as cancer of stomach, oesophagus and rectum when compared to other modalities.

2.3. Magnetic Resonance Imaging (MRI): - By using magnetic resonance electromagnetic signals are obtained. These signals produced from human organs help to rebuild the high resolution with detailed human organ structure. They are specially used for diagnosis,

staging of tumours like rectal, prostate, [6].

2.4. Positron Emission Tomography (PET): A radioactive tracer is injected, swallowed, or inhaled, it becomes lodged in the tissues of the body. Gamma rays are created when positrons from the unstable nuclei of the tracer mix with electrons. A 3D image of the tracer is produced by a computer when a ring of detectors in the PET scanner picks up these gamma rays. They are used in evaluation of Thyroid, Colon, Oesophageal, Melanoma, cervical cancers and to detect heart, brain disorders.

2.5. Medical Ultrasound: Sound waves are produced by the transducer using electrical energy. When an array of piezoelectric crystals receives an electric signal, high frequency sound waves are produced, which causes the transducer to vibrate. As these waves move through the body, various tissues with various properties at various depths reflect them back. These crystals pick up reflected sound waves and produce an electric signal that a computer may interpret as an image. These images can be used to detect abnormalities in the pelvic, heart, rectal, blood vessels, breasts, abdominal organs etc.,

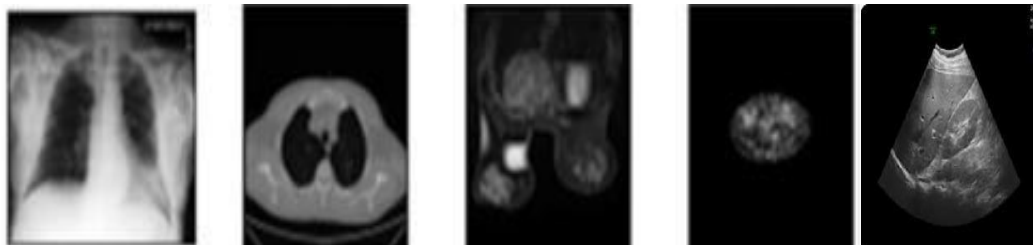


Fig 1. Medical Imaging Modalities. (Chest X-Ray, CT, MRI, PET scan, Ultrasound)

3. Machine Learning Techniques in Medical Image Analysis

Machine learning algorithms allow computer to learn pattern from data without being explicitly programmed. In order to provide precise diagnosis and classification, a supervised algorithm learns from the annotated data in images to predict output variables based on input features. In order to find underlying patterns and clusters in medical images, unsupervised algorithms examine the

unlabelled data to reveal hidden structures and relationships. Reinforcement learning is an agent-environment interaction dynamic that uses iterative decision-making to maximize cumulative rewards. To detect patterns in medical image analysis, traditional machine learning techniques mostly rely on supervised learning, in which a model is taught to carry out ML tasks like classification and segmentation.

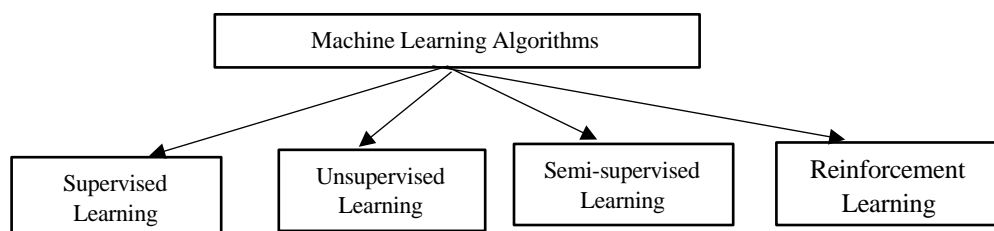


Fig 2: - ML algorithms in Medical image analysis

3.1 Supervised Learning

Supervised learning uses labelled data to train algorithms and to predict outcomes and recognize patterns. In medical image analysis it is frequently used for disease classification, image segmentation. They enable correct delineation of anatomical, pathological regions in medical images for segmentation task to precise treatment and planning.

3.2 Unsupervised Learning

Unsupervised learning uncovers the patterns and structures within unlabelled data without the knowledge of output labels. It is used for tumour classification and detection, mass detection in breast cancer. This helps to understand complex anatomical and pathological structures.

3.3 Semi-Supervised Learning

Semi-supervised learning approach is a combination of both supervised and unsupervised learning. It makes use of labelled and unlabelled data to train the models. It is used in classification, artificial image generation tasks, classification. It requires careful consideration for designing models and to balance labelled and unlabelled data.

3.4 Reinforcement Learning

Reinforcement learning is used when a decision needs to be made in uncertainty and the models must be able to learn from its action and experiences. It is used to design patient regime treatment, breast lesion detection and extraction task here the learning agents learn from actions, brain tumour detection.

Feature engineering is necessary in ML models to manually extract features from images (shape, intensity, texture). Commonly used machine learning algorithms for medical image analysis are mentioned in Table 1.

ML method	Description	Task/Process	Application in healthcare
Support Vector Machine (SVM)	Finds optimal hyperplane that separates different classes.	Classification	Brain Tumour Classification in MRI [7], CT scans [8]
Naïve Bayes	Probabilistic model based on Bayes theorem	Classification	Pneumonia detection and classification using X-rays [9]
Random Forest (RF)	Ensemble learning method to enhance prediction accuracy	Classification Segmentation	Tissue Segmentation in CT Scans [10]
K-Nearest Neighbor(KNN)	Uses proximity to classification	Classification	Classify Breast Cancer using Histopathological images [11]
K-Means Clustering	It groups unlabelled dataset to clusters	Segmentation	Bone Segmentation in CT liver images [12]

Table 1: Popular ML algorithms used in Medical Image Analysis

4. Deep Learning Approaches in Medical Image Analysis

Deep Learning algorithms have revolutionized medical image analysis by enabling automated feature extraction and achieving state-of-art performance on a range of medical image analysis applications. Deep learning techniques, such as CNNs and RNNs, outperform traditional machine learning algorithms and are capable of autonomously learning hierarchical characteristics from unprocessed medical images. CNNs are commonly used for image classification, segmentation, and registration tasks in medical image analysis. RNNs are used in medical image analysis and other tasks involving sequence data, including time-series data and medical reports.

4.1. Convolution Neural Networks (CNN)

CNNs have many layers of filters to acquire the local feature from the given input image and merge them to build more abstract representations. The initial CNN layers learn basic features like edges, corners while the higher layers learn high level features like object shapes and structures. They are used for image enhancement, segmentation, 3D image analysis etc.,

4.2. Generative Adversarial Network (GAN)

GAN is an unsupervised learning to create new data that mimic the provided data. It consists of two components namely a) generator – Network that is responsible to produce new samples b) Discriminator-Network distinguishes between real and generated data. They are used for Data Augmentation, anomaly detection, image synthesis, image reconstruction.

4.3. Recurrent Neural Network (RNN)

RNN are used to analyse time series or sequential data, Long Short-Term Memory (LSTM) is the standard RNN used for time series prediction in financial data [13], they address vanishing gradient problem using gates to control the flow of information and are well suited for task requiring long term dependencies. RNN are characterized by their ability to maintain hidden state to capture previous inputs to consider temporal dependencies. They are used in dynamic image analysis, video image analysis, improve diagnostic accuracy, understand disease progression.

4.4 Autoencoders (AE)

AEs are type of RNN used for mainly for unsupervised learning and are designed to learn efficient input data representations for the purpose of feature learning or dimension reduction. It has two components i) Encoder- to learn features from input data ii) Decoder- produce output closely to its original input. They are used for, feature extraction, anomaly detection, image reconstruction, image denoising.

4.5 Transfer learning

Transfer learning includes models pre-trained on large, general and annotated dataset like ImageNet that contains millions of images. Even when ImageNet contains natural images more than medical images, the primary layers of these pre-trained models learn general features of image such as shapes, features, textures which can be useful for medical imaging tasks like fine tuning. They are used for classification, segmentation, localisation and disease detection.

DL Architecture	Description	Task/process	Application in Healthcare
Convolutional Neural Network (CNN)	Extracts spatial features using layers of CNN	Anomaly Detection, Classification, Segmentation	Detection of brain tumours, retinal disease, lung nodule[14]
RNN	It's a type of ANN to process sequential data	Sequential Analysis	Disease flow detection Using fMRI [15]
Autoencoder	Used to compress and reconstruct data	Image Denoising Anomaly detection	Identify anomalies in ultrasound [16]
U-Net	For segmentation encoder-decoder architecture is used	Image Segmentation	Segments tumours, lesions using MRI and CT scans [17]
Generative Adversarial Network (GAN)	Uses generator network and discriminator network to identify real data	Image Generation Image Enhancement	Data augmentation using chest x-rays [18]

Table 2: Popular DL architecture used in Medical image analysis

Deep learning has become indispensable for playing crucial role in medical image analysis. Table 2. Shows **5 Overview of ML and DL tools and Frameworks in Medical Image Analysis**

Traditional based machine learning approaches often use libraries scikit-learn, OpenCV, XGBoost for classification, segmentation, registration purposes and deep learning architectures use several frameworks like

various DL architecture and DL tasks and its applications in healthcare
keras, Caffee, Pylearn, TensorFlow, pytorch for models like CNN, U-Net to automate feature extraction and segmentation. Python is the most common language used for DL. Table 3 shows the analysis of various ML and DL tools, frameworks and the use cases used in medical image analysis.

Tool/Library	Framework	Category	Description	Medical Image analysis use case
DICOM Toolkit	Python	Image Processing	Collection of libraries for processing DICOM images	Handling DICOM images
Open CV	Python, C++	Machine Learning	Library for processing real time images	Feature extraction, Pre-processing images
Scikit- learn	Python	Machine Learning	ML library for classification	Feature based classification using Random Forest (RF) and Support vector machine (SVM)
Matplotlib	Python	Data Visualization	Plotting library for image data visualization and model performance	Visualization of feature maps, Segmentation maps
TensorFlow	TensorFlow Keras	Deep Learning	Open source Library to build deep neural networks	Segmentation using U- Net, CNN's for disease detection
Pytorch	pytorch	Deep Learning	Deep learning framework with computation graph	Medical image analysis, image segmentation
Keras	Tensorflow, Theano	Deep learning	API to build deep learning models	CNN medical image classification
AutoML	Tensorflow, scikit-learn	Machine learning	Automated ML framework for model selection	Automated model selection in medical images
SimpleTK	C++, Python	Machine Learning	image registration and segmentation	Image registration and segmentation in radiological images
XGBoost	Python, R	Machine Learning	Implement gradient boosting trees	Medical image classification. anomaly detection
Caffe	Caffe	Deep Learning	Framework optimised for training deep networks	X-ray, CT, MRI image classification

Table 3: ML and DL tools used in Medical Image Analysis

6 ML and DL techniques for anomaly detection in Medical imaging modalities in disease detection

Anomaly detection is an observation of irregular event that deviates from normal behaviour, it is important to find these anomalies [19]. In recent times deep learning has dominant approach over traditional ML algorithms, though both are used for detection along with

classification of disease. The benefits result from deep learning's capacity to manage sizable and varied datasets, automatically extract hierarchical features from complex data, and deliver exceptional accuracy and scalability. A detailed description of ML and DL techniques, medical anomalies, medical imaging modalities and disease diagnosis is given in Table 4.

Author	Imaging Modality	Medical Anomaly	ML/DL Techniques	Disease Diagnostic Tasks	Performance metrics
[20]	Ultrasound	Breast tumour	<ul style="list-style-type: none"> • ANN • SVM • RF • 10-fold cross validation 	Classify benign and Malignant lesion	Accuracy SVM 77.7% RF:78.5%
[21]	CT Scan, X-Rays	Covid-19	<ul style="list-style-type: none"> • RNN • CNN • Hybrid DL model 	Classification for Covid-19 Detection	Accuracy with CT image: 94% Accuracy with X-rays: 98%
[22]	PET scan	Parkinson Disease	<ul style="list-style-type: none"> • VGG-16 based CNN 	PSD Detection	Specificity:97.5% Accuracy:84.6% Sensitivity:71.6% Precision:96.7%
[23]	MRI	Alzheimer's Disease	<ul style="list-style-type: none"> • CNN • ResNet101 • DenseNet121 • VGG-16 	AD detection	CNN Accuracy:97.60% Recall :97% AUC: 99.26%
[24]	Chest X- Rays	Tuberculosis (TB)	<ul style="list-style-type: none"> • ViT_Base_EfficientNet_B1_2 24 	Detection of TB	Accuracy: 97.72%
[25]	Chest X-rays	Lung Disease	<ul style="list-style-type: none"> • VDSNET 	Lung disease detection	Accuracy: 73%
[26]	X-ray	Covid-19	<ul style="list-style-type: none"> • COVIDX-Net 	Detection of Covid19	MobileNetV2-60% DenseNet-201-90% ResNetV2-70% InceptionV3- 50% Xception-80% VGG-19 –90% InceptionResNetV2-80%
[27]	CT	Interstitial Lung Disease	<ul style="list-style-type: none"> • 3D CNN with Grad-CAM images 	Classification of ILD	3D-CNN-97.17%
[28]	CT Scan & Chest X-Ray	Covid-19 & Pneumonia	<ul style="list-style-type: none"> • TL with DCNN • EfficientNet 	Classification of pneumonia & Covid-19	EfficientNet- 99%

Table 4: ML and DL technique for disease diagnosis in Medical Image Analysis

6. ML and DL, Medical Imaging Modalities and Cancer Types

Cancer is a disease in which abnormal cells grow uncontrollably spreading to other parts of the body leading to persons death. Cancer detection is a specific application within disease detection where the actual goal is to identify abnormal tumors using diagnostic method. There are more than 100 different types of cancer in which lung, breast, brain cancer is the most common types of cancer accounting for highest mortality. Advanced diagnostics for lung cancer and other cancers often involve integration of multiple

imaging modalities like CT, X-ray, MRI. ML and DL are making significant progress in improving early detection, increasing diagnostic accuracy and predicting patient outcomes in Cancer Diagnosis. Lung cancer can be diagnosed using CT, MRI, PET. ML Lung cancer is detected using ML, for segmentation marker-controlled watershed is used and binary classifier is used for classification. Dataset is trained with various algorithms like SVM, KNN, Decision Tree, Logistic Regression, Naïve Bayes and Random Forest. A performance accuracy of 88.5% is achieved with Random Forest [29]. Detection using DL, CNN is used to detect lung nodule to

be cancerous and non-cancerous, Ensemble 2DCNN consists of three different layers, kernels and pooling techniques gave a combined accuracy of 95% [10]. Classification and prediction of lung cancer, geometric filter is used for image processing, k-means clustering for segmentation and for Classification ANN, KNN, RF were used, ANN model produced accurate results [30]. Deep learning models ResNet-50, EfficientNet-B3, ResNet-101 along with transfer learning is used for lung cancer prediction, fusion model for classification along with ResNet-101 achieve 90% precision [31]. DenseNet and adaptive boosting algorithm is used for lung nodule classification and detection with 90.5% accuracy [32]. Breast cancer is detected using X-Ray(mammography), ultrasound, MRI. Detection of breast cancer using MRI dataset, six categorization models like RF, DT, SVM, KNN, LR, SVC. The best accuracy result was achieved by RF with 96.49% accuracy [33]. A new deep learning-based model was proposed to classify breast cancer with two types of datasets, the proposed model achieved 93% accuracy with ultrasound images and 95% accuracy on histopathological images. [34]. Brain tumor can be detected using MRI and CT. MRI brain tumor images were used. Image was pre-processed using median filter and segmented using thresholding and k-means clustering algorithm, features were extracted using GLCM and SVM to classify lung cancer using SVM and the model achieved an accuracy rate of 80% [35]. Using MRI image dataset YOLOv7 with transfer learning model is proposed to detect presence of brain tumor with 99.5% accuracy. [36]. A hybrid model was used for Liver cancer detection, CT images taken from biopsy and CNN with pre-trained model was used, the proposed model performed well with an accuracy of 99.5%, precision value 86.4%, recall value of 97.9%[37]. From detecting, classifying and segmenting tumors to predicting survival rates and treatment planning ML and DL models are indispensable tools in modern healthcare.

7. Conclusion: -

This paper offers insightful information about current research on medical anomaly detection in imaging modalities, which is being revolutionized in the healthcare industry by machine learning and deep learning approaches. ML uses learning techniques and DL acquires the details on how machines react. The significance of Anomaly detection in medical diagnosis is to enable the clinicians or physicians to use the ML and DL techniques for accurate detection, classification and prediction of any kind of medical anomalies in Medical Imaging. In ML, the features have to be extracted and from raw images that may be prone to error or lead to a noisy data but in DL automatically features are extracted from raw images leading to better disease detection even if dataset are large and noisy. Even though DL is a subset of ML, Deep learning algorithms are extensively used than conventional machine learning algorithms like convolutional network architectures with considerable data augmentation, can outperform human experts in terms of performance. By leveraging deep learning approaches, medical image analysis can achieve a higher accuracy, better generalization and significantly benefiting the clinicians and radiologists. However, as

these technologies advance and aid in the disease diagnosis, issues with data accessibility, model interpretability, and clinical integration continue to be crucial areas of attention.

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