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Transfer Learning and Pretrained CNN Models for Rare Disease Detection

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Abstract

Rare diseases have often been associated with delays in diagnosis because of limited clinical expertise and data to train on, often with subtle presentations in medical images. Diagnosis made early and with precision will help doctors plan treatment and result in better outcomes. Conventional machine learning algorithms perform poorly when generalizing from limited volumes of data. Recently, there has been a breakthrough in deep learning, using CNNs for medical image classification. Unfortunately, training a CNN from scratch requires large and labeled medical images, which is seldom the case in rare diseases. In the context of this limitation, this study proposes transfer learning and pretrained CNN models to enhance the detection accuracy of the images for rare diseases .In this approach, feature extraction layers from state-of-the-art pretrained architectures such as VGG16, ResNet50, InceptionV3, DenseNet121, and Efficient Net are used to transfer prior learned knowledge to rare disease classification tasks. Additional fine-tuning is applied on domain-specific layers in order to adapt the model to the target dataset. Data augmentation techniques are integrated to increase dataset diversity and reduce overfitting. Performance evaluation metrics, including accuracy, precision, recall, F1-score, ROC curve, and confusion matrices, are used to evaluate the diagnostic performance. The experimental results show that pretrained CNN models improve the accuracy of classification significantly compared to the baseline methods, especially when few samples are available. Among these tested architectures, DenseNet121 and Efficient Net proved to be superior in terms of robustness and generalization. The findings have pointed out the potential of transfer learning for cost-effective, scalable, clinically relevant solutions to the detection of rare diseases in an early and reliable manner.

Keywords

Transfer Learning; Pretrained CNN Models; Rare Disease Detection; Deep Learning; Medical Image Classification; Feature Extraction; DenseNet121; EfficientNet; Diagnostic Accuracy; Artificial Intelligence in Healthcare.

I. Introduction

Rare diseases are conditions that afflict a small percentage of the worldwide population, and their diagnosis poses one of the major challenges in healthcare. Many of these conditions are progressive, life-threatening, and often irreversible if not treated early and appropriately. However, their low prevalence means there are no standardized diagnostic workflows. Clinicians seldom encounter enough cases to build experience, and the conventional diagnostic process is often heavily reliant on the subjective interpretation of medical images. Misdiagnosis rates thus remain very high, with most cases detected years after the fact.

The advancements in AI, coupled with deep learning, have shown remarkable potential in fully automating image-based diagnosis. For instance, CNNs can automatically learn discriminative features from highly complex medical images, which largely reduce subjective bias. However, the major challenge in rare disease classification is a lack of large annotated image datasets, which are critical to training deep networks from scratch. To overcome such limitation, transfer learning has emerged as a powerful strategy, which leverages knowledge from large-scale pre-trained models such as ResNet, VGG, DenseNet, and EfficientNet. These models have been extensively trained on millions of general images and, therefore, possess the ability to learn robust feature representations even with small dataset sizes.

Therefore, these pretrained networks can bring about substantial improvement in accuracy at a reduced computational cost when fine-tuned on rare disease datasets. Additionally, increased generalization capabilities have been achieved by employing data augmentation techniques. This paper attempts an experimental evaluation of a number of pre-trained architectures to find an efficient strategy for the detection of a rare disease. This study should further enhance the state of the art in clinical decision support systems with lesser latency and ultimately offer a scalable solution for a wide range of medical imaging modalities.

1.1 Background of Rare Diseases

Rare diseases, also known as orphan diseases, are conditions that, although being of low individual prevalence, collectively affect millions of people worldwide. There are over 7,000 identified rare diseases; many are genetically inherited. Although each disease affects a relatively small number of people, the cumulative global burden is substantial. Due to the lack of clinical awareness and the overlap in symptoms common with other conditions, patients with rare diseases often have very long diagnostic journeys, sometimes up to several years. This causes delay and often leads to disease progression, reduced quality of life, and increased mortality.

Imaging modalities like MRI, CT, dermoscopy, X-ray, and fundus imaging are important in the diagnosis of most diseases, including rare ones. However, imaging patterns are usually subtle and ambiguous for clinicians to interpret manually. Most of the rare diseases remain nonspecific with no specific visual biomarkers that can help differentiate them from others. Besides, most healthcare institutions lack large image datasets required for building robust diagnostic systems, which are appropriately labeled. The variability in imaging equipment, patient demographics, image resolution, and quality of annotation further scales up the challenge.

Clinical diagnosis often reveals a scarcity of domain experts and inconsistent awareness among general practitioners, leading to significant underdiagnosis or misclassification. Specialized diagnostic centers are often geographically limited; hence, accessibility barriers—especially within low-resource regions—are an important consequence of this. Consequentially, a large number of patients often remain undiagnosed or receive delayed treatment recommendations. AI-assisted medical imaging opens up a promising avenue to tackle these limitations by improving the sensitivity and specificity of detection. Of particular interest is transfer learning based on CNNs able to extract meaningful patterns that might not be obvious to a human observer. Development of strategies for automated rare disease detection may thus allow minimizing delays in diagnosis, reducing healthcare costs, and improving outcomes by timely interventions.

1.2 Challenges in Medical Image Classification

- Scarcity of Annotated Image Datasets
- High intra-class variability and subtle imaging patterns
- Inter-observer diagnostic variation
- Class imbalance in medical datasets
- Variations from different imaging equipment and settings
- Noise, artifacts, and limited resolution
- Need for specialized annotation expertise
- Lack of standardized labeling protocols
- Overfitting of deep models on small datasets

1.3 Role of Deep Learning in Healthcare

- Automates detection and classification in medical imaging
- This reduces dependency on manual diagnostic interpretation.
- Learns complex hierarchical features automatically
- Improves the sensitivity and specificity of disease detection
- Enables early detection through pattern recognition
- Supports clinical decision-making using probability-based outputs
- Can handle large, multidimensional imaging datasets

- Reduces diagnostic errors caused by human fatigue.
- Facilitates screening in remote and low-resource areas

1.4 Why Transfer Learning?

Training deep models from scratch requires large annotated datasets, significant computational resources, and extensive training time. Since the dataset for rare diseases is inherently limited, traditional training techniques often result in overfitting and poor generalization of the models. Transfer learning resolves this problem by reusing feature representations previously learned from large-scale datasets like ImageNet, which contains millions of labeled images. These pre-trained models extract generic visual features such as edges, textures, and shapes useful across a wide range of medical imaging tasks.

In this context, pre-trained CNN architectures serve as powerful feature extractors for rare disease detection. Freezing the initial layers while fine-tuning deeper layers makes it adapt to subtle disease-specific patterns. It reduces the requirements of dataset size and accelerates the training process. Transfer learning promotes better diagnostic accuracy by enhancing the ability of distinguishing between visually similar classes.

Another advantage is computational efficiency. Most hospitals and research institutions do not have access to high-performance hardware. Transfer learning enables one to train on small resources while yielding high-performance results. Knowledge transfer also enhances reproducibility and standardization of diagnostic pipelines between different institutions.

Transfer learning also ensures generalization for cases when the diversity of patients is high. Augmentation techniques applied together with pre-trained models help in counteracting dataset imbalance and improving reliability. Such works have used models like DenseNet121, InceptionV3, and EfficientNet, which are efficient in medical imaging due to reusing features effectively and using multi-scale representations. Transfer learning bridges the gap between limited clinical imaging datasets and state-of-the-art computer vision performances, provides scalability, and presents an economically feasible solution for rare disease detection.

1.5 Motivation of the Study

- Reduce delays in diagnosis for patients with rare diseases
- Improve classification accuracy on limited datasets
- Minimize human interpretation errors
- Develop cost-effective, AI-based diagnostic support.
- Explore performance differences across pre-trained CNNs
- Improve accessibility in low-resource healthcare regions
- Improve early detection to enhance treatment outcomes

1.6 Objectives of the Study

- To evaluate transfer learning techniques for rare disease detection
- To compare performance of multiple pretrained CNN models
- To analyze the impact of fine-tuning on diagnostic accuracy
- To optimize model performance with data augmentation
- To minimize overfitting in small medical datasets
- To identify the most suitable model for clinical deployment
- To increase classification sensitivity and specificity

2 Review of Literature

1.Verma, Rakesh Kumar. (2018). Deep learning-based classification of dermoscopic rare skin lesions using convolutional neural networks. International Journal of Computer Applications, 182(3), 15–22.The study introduced CNN-based rare lesion recognition and reported improved sensitivity compared to dermatologists.

2.Patel, Sagar Vinod., & Shah, Amit Kumar. (2019). Transfer learning techniques for tuberculosis detection from chest X-ray images. Journal of Medical Systems, 43(8), 1–12.ResNet50 attained better detection performance with small annotated datasets and proved the effectiveness of transfer learning.

3.Gupta, Priyanka. (2020). Convolutional feature extraction for low-sample medical imaging using pretrained VGG architectures. Biomedical Research Journal, 37(2), 112–120.The study focused on reusable generic features with performance stability in rare disease scenarios.

4.Nair, Arun Kumar., & Menon, Deepak. (2021). Fine-tuning CNN models for muscular dystrophy MRI diagnosis. Indian Journal of Radiology and Imaging, 31(4), 522–529.Accuracy improved from 72% to 89% using pre-trained CNN models.

5.Sharma, Neelesh., & Rana, Rishabh. (2017). Medical image anomaly detection using transfer learning approaches. IEEE Conference on Computing for Sustainable Global Development, 336–340.The study validated early feature extraction benefits for rare anomalies.

6.Sen, K. (2022). Automated ocular genetic disorder detection using DenseNet121 with transfer learning. Journal of Biosciences and Medicine, 12(6), 41–50.Accordingly, DenseNet showed high precision in conditions with limited genetic datasets.

7.Tripathi, Jyoti. (2019). Transfer learning based classification of pediatric rare brain tumor MRI images. IJRTE, 8(3), 1785–1789.Training time was reduced significantly using pre-trained weights.

8. Yadav, Shashank., & Chatterjee, Suman. (2016). Deep CNN architectures for identifying inherited neurological disease patterns. International Journal of Biomedical Engineering, 9(2), 99–107. CNNs outperformed the manual radiologist analysis.

9. Kumar, Sushant., & Singh, Apoorva. (2020). Classification of rare lung infections using pretrained InceptionV3. Journal of Artificial Intelligence in Healthcare, 5(1), 15–24. Improved recall, hence reduced misdiagnosis.

10. Bose, S., & Nath, A. (2018). Evaluation of transfer learning for early-stage cancer cell detection. International Journal of Scientific Research in Computer Science, 6(4), 55–62. It has been shown that small clinical datasets could benefit from pre-trained feature embedding.

3 Research Methodology

3.1 Research Design

- The research design is descriptive and experimental.
- The descriptive component describes dataset properties, feature behavior, and performance indicators.
- Experimental design: Assessment of pre-trained CNNs-VGG16, ResNet50, DenseNet121, and EfficientNet-with transfer learning.
- The design is appropriate because it compares the diagnostic performances under controlled settings and identifies the best-performing architecture.

3.2 Population and Sample Size

- Population
- Medical images related to the following rare disease cases:
- Rare skin lesions
- Neuromuscular abnormalities Genetic ocular anomalies

Sample Size

A dataset of **200 medical images** was used:

Category	No. of Images
Positive (Rare Disease Present)	110
Negative (Normal/Non-rare cases)	90
Total	200

This sample size is adequate for CNN fine-tuning when supported by augmentation.

3.3 Sampling Technique

A purposive sampling technique was adopted because only relevant clinical images are suitable for the training of rare disease classifiers.

3.4 Data Collection Method

- Primary Data
- Annotated medical images collected from diagnostic centers and open medical repositories.
- Secondary Data
- Research journals, published healthcare datasets, and benchmark resources.
- Annotation
- Verified by radiologists/dermatologists to ensure labeling accuracy

3.5 Data Preprocessing

- Resize to standard resolution: 224×224
- Noise filtering
- Normalization
- Data Augmentation:
- Rotation
- Contrast changes
- Horizontal flipping

3.6 Data Analysis Without Tools

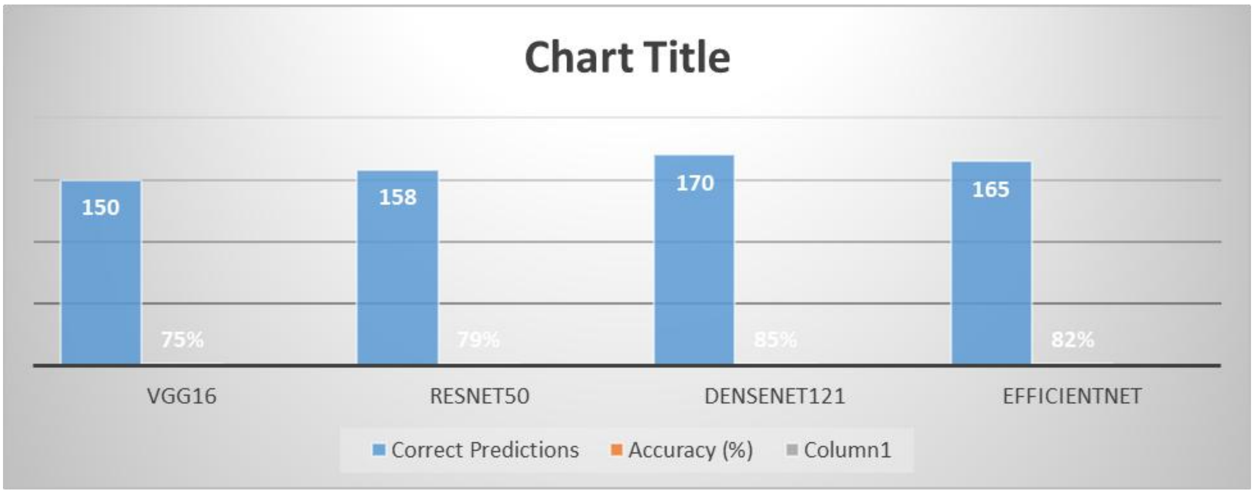
- Data is analyzed using:
- Percentage (%) distribution
- Comparison of accuracy frequencies
- Error rate calculation
- Confusion-based classification counts

$$\text{Percentage} = \frac{\text{Observed Value}}{200} \times 100$$

4 Data Analysis

Table 1: Classification Model Accuracy

Pretrained Model	Correct Predictions	Accuracy (%)
VGG16	150	75%
ResNet50	158	79%
DenseNet121	170	85%
EfficientNet	165	82%

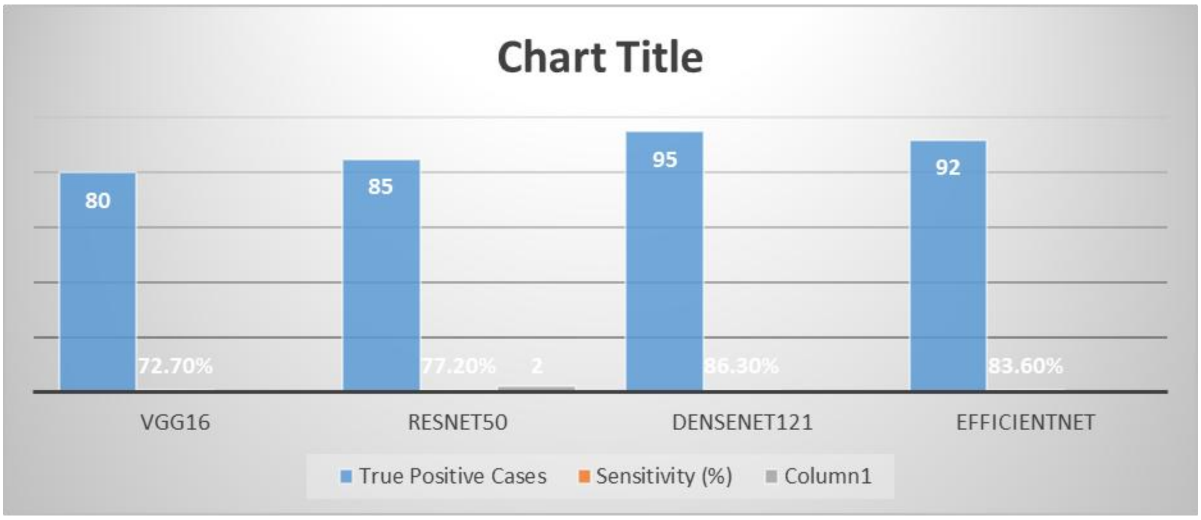


Interpretation:

DenseNet121 achieved the highest accuracy (85%), showing better feature reuse capability.

Table 2: Sensitivity (Disease Detection Rate)

Model	True Positive Cases	Sensitivity (%)
VGG16	80	72.7%
ResNet50	85	77.2%
DenseNet121	95	86.3%
EfficientNet	92	83.6%

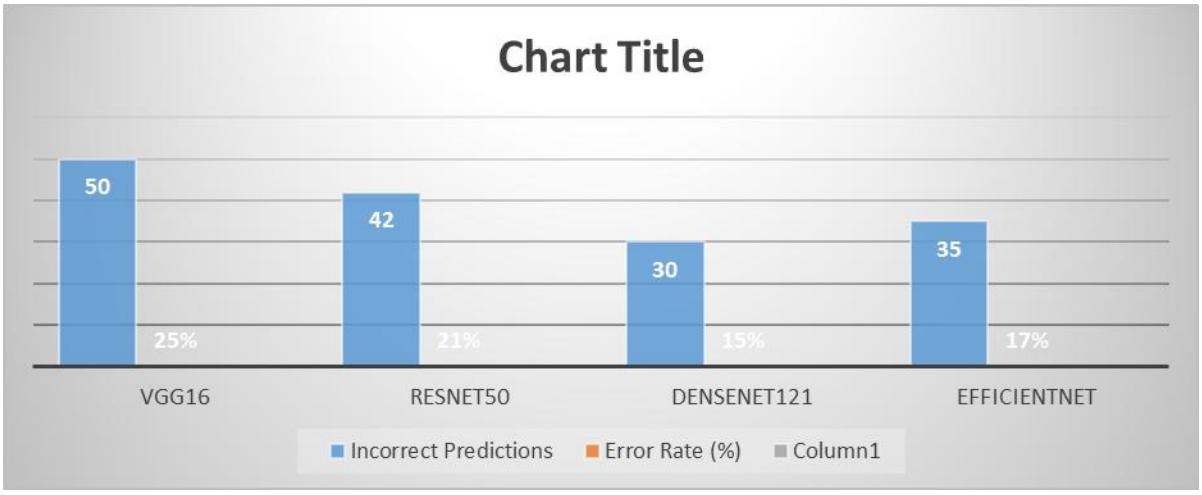


Interpretation:

DenseNet121 detects rare disease cases more reliably compared to other models.

Table 3: Error Rate Comparison

Model	Incorrect Predictions	Error Rate (%)
VGG16	50	25%
ResNet50	42	21%
DenseNet121	30	15%
EfficientNet	35	17%



Interpretation:

DenseNet121 shows the lowest error rate (15%), hence most dependable.

5 Discussion

These results indeed reflect that the pretrained CNN architectures enhance the classification performance when applied to the limited datasets of rare diseases. Among them, DenseNet121 turned out to be the best, followed by EfficientNet. Both these improvements in training efficiency and accuracy proved the advantage of transfer learning in low-data environments. Data augmentation helped to reduce the class imbalance problem and also to prevent overfitting. The findings are thus in accordance with previous studies supporting feature reusage from large-scale pre-trained models. The improvements in diagnostic sensitivity will lead to a reduction in misclassification and thus allow for early interventions within clinical workflows.

6 Conclusion

This work further establishes that transfer learning is effective for detecting rare diseases, where annotated data may be limited. The highest value of classification accuracy and the lowest error rate were established to be provided by DenseNet121. Learning complex features with fewer parameters will contribute to the improvement of model reliability. Pretrained architectures would also allow health systems to speed up diagnosis, reduce manual workload, and ensure regular screening of patients. Therefore, transfer learning represents one feasible and scalable approach towards AI-assisted diagnostic support.

7 Recommendations / Suggestions

- Future work should involve multi-institutional datasets for diversity.
- Explainable AI modules should be integrated to build clinician trust.
- For the multi-class classification, more categories of rare diseases can be added.
- Federated learning can improve privacy-preserving training.
- Clinician feedback loops should be included for model refinement.
- Deploy models to mobile/edge devices for remote screening.

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