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Facial Recognition for Disease Diagnosis Using a Deep Learning Convolutional Neural Network: A systematic Review and Meta-Analysis

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Abstract

Objective: This study aimed to systematically review the literature on facial recognition technology based on deep learning networks in disease diagnosis over the past ten years to identify the objective basis of this application.

Methods: This study followed the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines for literature search and retrieved relevant literature from multiple databases, including PubMed, on November 13, 2023. The search keywords included deep learning convolutional neural networks, facial recognition, and disease recognition. 208 articles on facial recognition technology based on deep learning networks in disease diagnosis over the past ten years were screened, and 22 articles were selected for analysis. The meta-analysis was conducted using Stata 14.0 software.

Results: The study collected 22 articles with a total sample size of 57,539 cases, of which 43,301 were samples with various diseases. The meta-analysis results indicated that the accuracy of deep learning in facial recognition for disease diagnosis was 91.0% [95% CI (87.0%, 95.0%)].

Conclusion: The study results suggested that facial recognition technology based on deep learning networks has high accuracy in disease diagnosis, providing a reference for further development and application of this technology.

Keywords: Disease identification; Face recognition; Review; Convolutional neural network

Abbreviations: FCN: Fully Connected Convolutional Neural Network; PRISMA: Preferred Reporting Items for Systematic Reviews and Meta-Analyses; CNN: Convolutional Neural Networks; AHRQ: Agency for Healthcare Research and Quality; AI: Artificial Intelligence; ML: Machine Learning; DL: Deep Learning; AS: Angelman Syndrome; UPD: Paternal Uniparental Disomy; CdLS: Cornelia de Lange Syndrome; TS: Turner's Syndrome; WBS: Williams-Beuren Syndrome; VGG: network: Visual Geometry Group Network; GSs: Genetic Syndromes; MTCNN: Multitask Cascaded Convolutional Neural Network; KNN: K-Nearest Neighbors; LDM: London Medical Database; NGP: Next Generation Phenotyping; SVM: Support Vector Machines; LM: Language Models; RT: Random Forests; EM: Expectation Maximization; SLR: Systematic Literature Review

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Introduction

Thousands of years ago, the traditional Chinese medicine book 'Huangdi Neijing' recorded that 'Qi and blood in the three hundred and sixty-five veins of the twelve meridians flow into the face and inject into the orifices (facial seven orifices) [1]. In China, many traditional Chinese medicine doctors can understand the patient's condition by observing the patient's face, which is called facial diagnosis. Diseases not only cause abnormalities in human internal structures and physiological functions but also may lead to changes in appearance and deformity, including changes in limbs, trunk, and face [2]. Doctors can make preliminary judgments on specific diseases by identifying the facial features of patients, which play a prompt role in the subsequent diagnosis and treatment of diseases. Hereditary diseases, such as Down syndrome [3], Turner syndrome [4], Nunan syndrome [5] and Williams-Beuren syndrome [6], Angelman syndrome [7], and Corneliade Lange syndrome [8], are the main causes of facial change or deformity. Meanwhile, there are some non-genetic diseases such as acromegaly [9], autism [10], and Alzheimer's disease [11]. Now, more new research focuses on cancer and the new coronavirus. However, the symptoms and facial features of some patients in the early stage of the disease are not prominent, and medical personnel lack experience in identifying the appearance of complex diseases and many other issues [12,13]. Therefore, before the development of convolutional neural-based face recognition technology, the facial features of the disease could not play an important role in the diagnosis of the disease.

In areas where medical resources are scarce, physical examination (especially large-scale equipment inspection) is a luxury, which leads to delays in disease treatment. Even in big cities, due to the difference in personal medical levels and the long queuing time of hospitalization, the treatment of diseases is often delayed [14]. Computer-aided facial diagnosis helps to quickly conduct noninvasive screening of diseases, which provides a reference for doctors to diagnose diseases timely and effectively [15].

Nowadays, face recognition technology has been preliminarily applied to the auxiliary diagnosis of the above diseases [16]. By analyzing the visual features of the face, face recognition technology identifies the identity for human-computer interaction, tracking and monitoring, security monitoring, and identity recognition. In the last ten years, this technology has gradually entered the field of medical diagnosis [17,18].

In this field, face recognition software determines the patient's disease type by extracting the measurement data of specific features of the patient's face, analyzing the patient's face pattern, and comparing it with the disease database. Face recognition technology can identify diseases in time and alleviate the problem of insufficient medical resources. Now, for some diseases, face recognition technology even achieves higher diagnostic accuracy than doctors. Therefore, face recognition can contribute to the early screening of diseases and improve diagnostic accuracy.

This paper aims to provide a comprehensive overview of the current research on the application of face recognition technology to disease diagnosis. Previous surveys were extended by providing a detailed analysis of the history of face recognition and its evolution in the field of medical diagnosis [19]. Also, this study highlighted the potential of face recognition technology in overcoming the challenges of early disease screening and improving diagnostic accuracy. The rest of this paper is organized as follows: Section 2 introduces the meta-analysis design, data collection, and analysis methods. Section 3 provides a summary of the main findings for each study, including sample size, effect size, confidence interval, heterogeneity index, and an overall evaluation of the meta-analysis results for facial recognition in disease diagnosis. The discussion in Section 4 explains the importance of the meta-analysis results and suggests future research directions. Finally, Section 5 emphasizes the importance and contribution of facial recognition in disease diagnosis while acknowledging its limitations.

Material and Methods

In the past ten years, the use of Convolutional Neural Networks (CNNs) for disease recognition has received significant attention from the scientific community. Therefore, this study reviewed publications from the past ten years (2013-2023) because there were fewer studies in this field ten years ago. Ethical approval to conduct the study was obtained from the Ethics Committee of Affiliated Hospital of Shandong University of Traditional Chinese Medicine (2021-074-YK).

Literature Search Strategy

This systematic review was conducted following the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA). A literature search was conducted on December November 13, 2023, and the PubMed library, EBSCO Industries, Elsevier Scopus, Web of Science, and Springlink databases were utilized in this study. These databases were chosen because they provide the most high-impact scientific conference records and journals and cover the field of disease research and machine learning using CNNs. A broad search string was used to avoid missing any potentially exciting research. The search keywords were ("disease" or "disease recognition") and ("convolutional neural network" or "deep learning") and ("face recognition" or "facial diagnosis"). Additionally, other searches were conducted by reviewing the reference lists of all included papers. This comprehensive scope involved a large number of papers, and the most relevant ones were selected using the selection criteria outlined in Table 1. The studies included were cross-sectional and longitudinal, randomized and non-randomized, and had control groups.

Literature Screening and Data Extraction

The authors (Xinru Kong and Qianhui Qiu) read the selected abstracts and searched the full text when there was insufficient information to determine inclusion or exclusion criteria. Then, two authors (Jie Sun and Ziyue Wang) independently reviewed the full publications of the remaining papers and reached a consensus on inclusion criteria. If there was disagreement, a third reviewer (Xianghua Qi and Xiao Ding) would be involved. Finally, the included studies were classified based on their design, sample, method, and results.

Bias Risk Assessment of Included Studies

The Agency for Healthcare Research and Quality (AHRQ) tool for cross-sectional study evaluation was employed in this study to evaluate bias risk in the selected studies. The AHRQ tool consists of 11 items, each with a "yes," "no," or "unclear" response, with "yes" scored as 1 point and "no" or "unclear" scored as 0 points. The total score was out of 11 points, with scores of 8-11 indicating high quality, 4-7 indicating medium quality, and 0-3 indicating low quality.

Table 1: Selection criteria for studies defined in the SLR planning protocol.

ID	Criterion Type		Description
	Inclusion	Exclusion	
C1	√		Studying facial recognition for disease diagnosis through convolutional neural networks.
C2	√		Studying facial recognition for new disease species through convolutional neural networks.
C3	√		Studies describing facial recognition techniques or methods for disease diagnosis using CNN.
C4	√		Studies that present new facial recognition models or architectures for disease diagnosis based on convolutional networks.
C5		√	Studies describing techniques or methods
C6		√	Duplicate publications from multiple sources
C7		√	Studies without qualifying data for extraction
C8		√	Studies that do not use CNN as the main approach

Table 2: Summary of the principal results: disease diagnosis using facial recognition technology.

ID	Disease	Refs	Year	Algorithm	Task of Model	Dataset Characteristics	Disease Image Source	Image source for the control group	View Point of Images	All algorithm Accuracy	Best Accuracy	Predominant Approach
1	Down Syndrome	Bosheng Qin et al.	2020	KNN, SVM and DCNN	Binary Classifier	148 DS patients and 257 healthy images	Face2Gene and the Elife database	CASIA WebFace	Front View	SVM 76.86%, KNN 71.07% and DCNN 95.87%	95.87%	Fine Tune & Transfer Learning
2	Turner syndrome	Zhouxian Pan et al.	2020	DCNN	Binary Classifier	170 TS patients and 1053 healthy images	by the author himself	by the author himself	Front View	DCNN 96.9%	96.9%	Image Segmentation & Transfer Learning
3	Williams-Beuren Syndrome	Hui Liu et al.	2021	VGG16, VGG19, ResNet18, ResNet34 and MobileNetV2	Binary Classifier	104 WBS children,91 cases with other genetic syndromes and 145 healthy children.	by the author himself	by the author himself	Front View	VGG16 90.90%, VGG19 92.70%, ResNet18 87.9%, ResNet34 89.10% and MobileNetV2 85.6%	92.7%	Transfer Learning
4	Noonan Syndrome	Hang Yang et al.	2021	DCNN-Arcface, DCNN-CE, SVM and LR	Binary Classifier	127 NS patients, 163 healthy children, and 130 children with several other dysmorphic syndromes.	by the author himself	by the author himself	Front View	DCNN-Arcface 92.01%, DCNN-CE 85.21%, SVM 82.59% and LR 78.77%	92.01%	New Architecture
5	Angelman syndrome	Diego A. Gomez et al.	2020	Facial recognition system DeepGestalt	Multiclass Classifier	261 AS patients	by the author himself		Front View	DeepGestalt 91%	91%	Transfer Learning
6	Corneliade Lange Syndrome	Ana Latorre Pellicer et al.	2020	Facial recognition system DeepGestalt	Multiclass Classifier	49 CLS patients	by the author himself		Front View	DeepGestalt 97.9%	97.9%	Transfer Learning
7	Genetic Disorders	Yaron Gurovich et al.	2019	DeepGestalt	Multiclass Classifier	17,000 images representing 200 syndromes	London Medical Databases and Face2Gene Library.	London Medical Databases and Face2Gene Library.	Front View	DeepGestalt 91%	91%	New Architecture
8	Genetic syndromes	Dian Hong et al.	2021	VGG16 (Make fine adj)	Binary Classifier	228 Genetic syndromes children and 228 healthy children	by the author himself	by the author himself	Front View	VGG16 88%	88%	Transfer learning
9	Acromegaly	Xiangyi Kong et al.	2017	LM, KNN, SVM, RT, CNNand EM	Binary Classifier	527 Acromegaly patients and 596 healthy	by the author himself	SCUT-FBP dataset	Front View	LM 84%, KNN 93%, SVM 84%, RT 89%, CNN 96% and EM96%	96%	Image Segmentation

10	Autism Spectrum Disorder	Fawaz Wase-lallah Alsaade et al.	2022	Xception, VGG19 and Nasnet Mobile	Binary Classifier	1470 Autistic children 1470 nonautistic children	Kaggle platform,	Kaggle platform	Front View	Xception 91%, VGG19 80% and NASNETMobile78%	91%	Transfer Learning
11	Progeria Syndrome	Dhairya Chandra et al.	2019	LR, CNN, SVM, KNN, RF	Binary Classifier	The 125 images are divided into progeria syndrome and normal faces.	by the author himself	by the author himself	Front View	LR 99.3%, CNN 97.1%, SVM 96%, KNN 97.9% and RF 95.1%	99.3%	Image Segmentation
12	Facial Dermatological Disorders	EvginGoceri et al.	2020	SVM, k-NN, AdaBoost, ScSPM and MD-CNN	Multiclass Classifier	Acne vulgaris, psoriasis, rosacea, seborrheic dermatitis Each disease consists of 101 pictures.	DermNet, Derm-Quest, Derm-Web and Der-moWeb	DermNe, Derm-Quest, Derm-Web and Der-moWeb	Front View and side view	SVM 63.67%k-NN 47.00%, AdaBoost 56.00, ScSPM 70.00%, MD-CNN73.00%	73.00%	Transfer Learning
13	Facial Nerve Paralysis	Anping Song et al.	2018	Inception-v3, Inception-v4, Inception-ResNet-v1, Inception-ResNet-v2, DeepID, ResNet and CNN	Multiclass Classifier	860 FNP patients and 189 controls	FNP dataset	by the author himself	Front View and side view	Inception-ResNet-v1 95.2% Inception-ResNet-v2 95.7% Inception-v3 93.3% DeepID 92.5% Inception-v4 95.3% ResNet 95.0% and CNN97.5%	97.5%	Transfer Learning
14	Cancer	Bin Liang et al.	2022	ResNet (Make fine adj)	Binary Classifier	8124 cancer patients and 8124 controls	by the author himself	Megage date	Front View	ResNet 82%	82%	Transfer learning
15	beta-thalassemia	Bo Jin et al.	2020	AlexNet+ SVM VGG16+ SVM ResNet50+ SVM	Binary Classifier	70 are beta-thalassemia, 70 are healthy	by the author himself	by the author himself	Front View	AlexNet+SVM85.0% VGG16+SVM 95% ResNet50+SVM86.7%	95%	Transfer learning
16	beta-thalassemia, hyperthyroidism, Down syndrome, and leprosy	Bo Jin et al.	2020	AlexNet+ SVM VGG16+ SVM ResNet50+ SVM	Multiclass Classifier	350 images in total, 70 images of each type	by the author himself	by the author himself	Front View	AlexNet+SVM 86.0%, VGG16 + SVM 93.3%, ResNet50+SVM 88.7%	93.3%	Transfer learning
17	Constitution classification	Er-Yang Huan et al.	2020	VGG16 Inception v3, DenseNet121 and ConstitutionNet et al.	Multiclass Classifier	Total 12,730 images	by the author himself	by the author himself	Front View	VGG-16 62.66%, Inception V3 63.79% and DenseNet-121 64.25%.	66.79%	Transfer learning
18	COVID-19 susceptibility	Oleg Kit et al.	2021	ResNet-34(Make fine adj)	Binary Classifier	261 negative and 264 positive	Internet resources	Internet resources	Front View	ResNet-34 84%	84%	Transfer Learning
19	Depression	Xinru Kong et al	2022	CNN, VGG11, VGG19, ResNet and Inception-V3	Binary Classifier	102 Depression patients 1132 controls	by the author himself	by the author himself	Front View	FNC 98.23%, Vgg11 94.40%, Vgg19 97.35%, ResNet50 97.10% and Inception-V3 94.99%	98.23%	Transfer learning
20	Schizophrenia	Xiaofei Zhang et al.	2023	CNN	Binary Classifier	106 schizophrenia patients and 101 healthy controls	by the author himself	by the author himself	Front View	CNN 95.18%	95.18%	Transfer learning
21	Velocardiofacial Syndrome	Rong Min Baek et al.	2023	CNN, ResNet100 and ResNet101	Binary Classifier	98 patients with VCFS and 91 controls	by the author himself	by the author himself	Front View and side view	CNN95.00%, ResNet100 95.00% and ResNet101 85.00%	95.00%	Transfer learning
22	Myasthenia gravis	Annabel M. Ruiter et al	2023	3D-ResNet-50 and 3D-ResNet-34(Make fine adj)	Binary Classifier	70 MG patients and 69 controls	by the author himself	by the author himself	Front View	ResNet-50 80.00% and ResNet-34 87%	87.00%	Transfer learning

Table 3: Healthy Subject Data Set.

Data set	Time of publication	Number of face images and videos	Identity
LFW (Zheng et al., 2017)	2007	13233	5749
YTF(Herrera and Wen, 2020)	2011	3 495 videos	3 495
CASIA-Webface [(Yi et al., 2014)]	2014	494414	10575
VGG2 (Deng et al., 2018)	2015	3310000	9131
IJB-A[(Klare et al., 2015)]	2015	5712 face images and 2085 videos	500
UMDFace [(Bansal et al., 2017)]	2016	367,888	8,277
MegaFace(Kemelmacher-Shlizerman et al., 2016)	2016	4.7M	672000
CelebA (Liu et al., 2018)	2016	202,599	10,177
CFP-FP (Deng et al., 2019)	2016	7000	500
IQIYI(Wang and Lobato, 2019)	2016	500000 videos	5000
MS1M-IBUG (Zhu et al., 2021)	2017	3.8M	85K
Age Database-30 (Moschoglou et al., 2017)	2017	12240	570
CALFW (Zheng et al., 2017)	2017	13233	5749
IJB-B (Whitelam et al., 2017)	2017	11,754 face images and 7,011 videos	1,845
MS1M-ArcFace (Razzhigaev et al., 2020)	2018	5.8M	85K
IMDB-Face (Wang et al., 2018)	2018	1700000	59000
Celeb500k (Cao et al., 2018; Deng et al., 2020)	2018	50000000	500000
CPLFW (Zheng and Deng, 2018)	2018	13233	5749
IJB-C(Maza et al., 2018)	2018	138,000 face images and 11,000 videos	28,936
Glint-Mini (Liu et al., 2022)	2020	5.2M	91K
Asian-Celeb (Le and Kakadiaris, 2020)	2020	2800000	94000
DeepGlint (Deng et al., 2019b)	2020	6.75M	181K

Statistical Methods

Meta-analysis was carried out using Stata 14.0 software. The I^2 value and Q test were used to evaluate heterogeneity among studies. If $P>0.1$ and $I^2<50\%$, a fixed-effects model was selected for meta-analysis. If $P\leq0.1$ and $I^2\geq50\%$, a random-effects model was selected for meta-analysis. An analysis of ACC values was conducted to investigate the sources of heterogeneity. Meanwhile, sensitivity analysis was conducted to evaluate the stability of the results. Additionally, funnel plots and Begg's and Egger's tests were used to evaluate the publication bias. The significance level for meta-analysis was set at $\alpha=0.05$ [20].

Results

Literature Screening Process and Results

During the identification stage, 1227 duplicate articles and book studies were excluded. In the initial screening phase, 98 reviews were excluded because they did not meet the inclusion criteria. During the extraction stage, 60 full-text articles were selected, and 22 articles were summarized after the authors read the full text. The articles were selected following the SLR steps (Figure 1). In the preliminary screening, research papers (n=101) were found among 208 articles, including review papers (n=10), conference papers (n=80), unspecified (n=1), datasets (n=1), editorial materials (n=1), abstracts (n=1), and others (n=13) (Figure 2). The investigation covered research from 2013 to 2023, and 2013 was noted as a milestone for CNN research and its application to disease identification and recognition. At the end of the comprehensive activity, Table 2 was prepared. The table includes the following attributes: ID (study identification), disease type, year, reference (method author), algorithm, task of model, dataset characteristics, disease image source, image source for the control group, view point of images, all algorithm accuracy, best accuracy, and advantageous method. In the "View Point of Images" section, there are three articles with the option "Front View and Side View." These three articles utilize a comprehensive analysis approach to address facial recognition disorders by analyzing front-view and side-view images. The study included 22 articles with a total sample size of 57,539 cases, of which 43,301 were samples with various diseases.

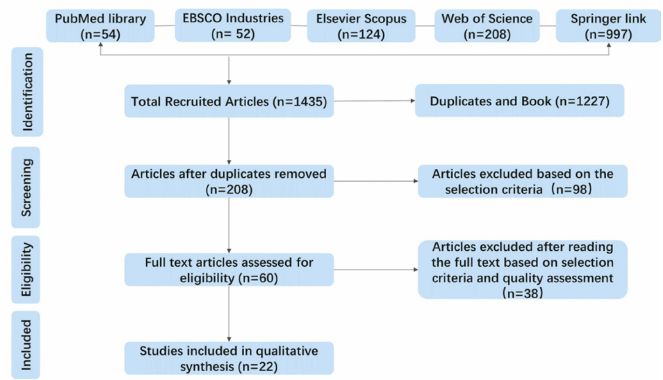

Figure 1: Flowchart of study phases and selections, outlining the process of selecting and analyzing relevant studies for the research. The image was created using the Microsoft Visio software.

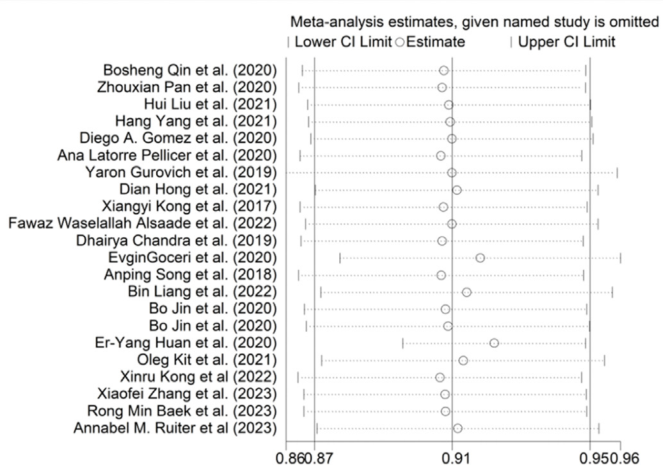

Figure 2: Histogram of all article categories, displaying the distribution of publications across different subject areas. The image was created using the Microsoft Excel software.

Literature Heterogeneity

Through analysis using a random effects model, the heterogeneity result of the 22 articles was I^2=99.6% and P<0.000001. The meta-analysis results indicated that the accuracy of deep learning applied to facial recognition diseases was 91.0% [95% CI (87.0%, 95.0%)], as shown in Figure 3.

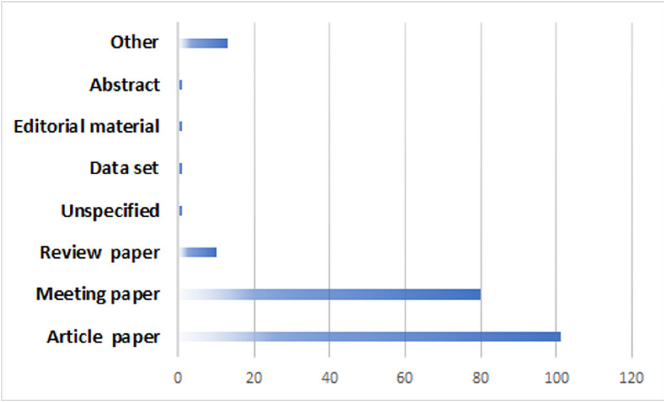

Figure 3: Forest plot of Facial Recognition Diseases Using a deep learning convolutional neural network, showing the effect sizes and confidence intervals of different studies on the topic. The image was created using the Stata 14.0 software tool.

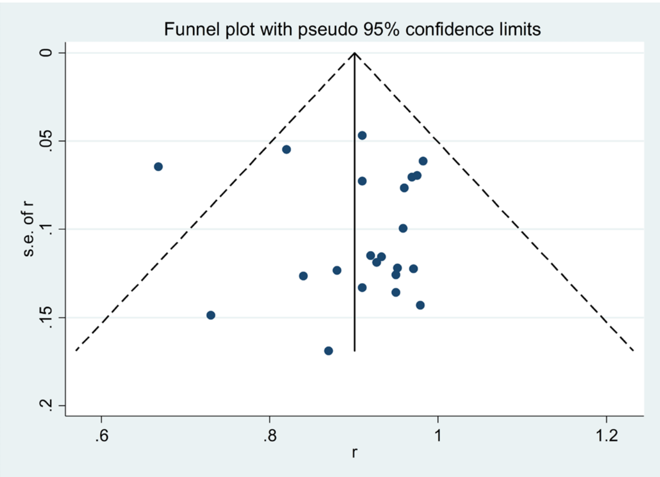

Figure 4: Funnel plot for assessing publication biases, providing a graphical representation of the potential bias in the included studies. The image was created using the Stata 14.0 software tool.

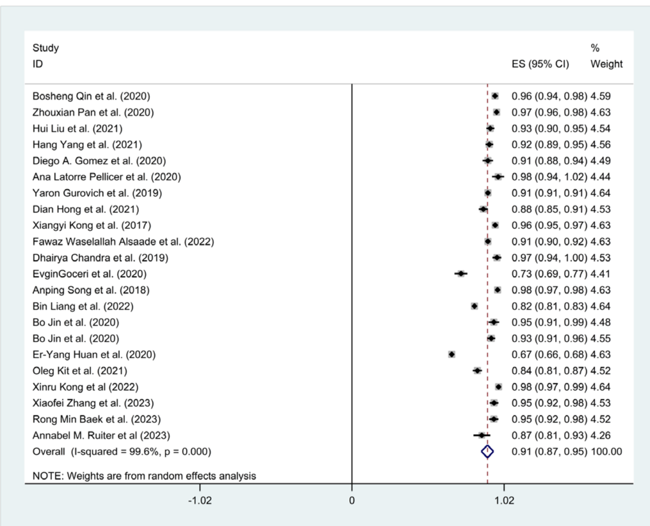

Figure 5: Sensitivity analysis of Facial Recognition Diseases Using a deep learning convolutional neural network, exploring the impact of excluding certain studies or changing analysis methods on the overall results. The image was created using the Stata 14.0 software tool.

Publication Bias Analysis

According to the 22 selected articles, a funnel plot was drawn, and the distribution between studies was asymmetric (Figure 4). Begg's test (P=0.048) indicated a potential publication bias, while Egger's test (P=0.612) indicated no bias, showing stable results.

Sensitivity Analysis

The one-by-one exclusion method was used, and there was no significant change in the results, indicating good stability of the meta-analysis results (Figure 5).

Discussion

The History of AI and Face Recognition

Artificial Intelligence (AI) is the underlying technology for face recognition. AI is a branch of computer science that aims to empower machine logic reasoning [21,22]. The development of AI can be divided into three waves, with the third wave (1993 – present) seeing the maturation of perceptual intelligence through deep learning. Face recognition technology has been studied since the 1950s, and different algorithms and techniques have been proposed for face recognition [23,24]. In the early stages, face recognition was mainly studied as a general pattern recognition problem, with the method based on geometric features being adopted to solve this problem. In recent years, with the development of deep learning algorithms, face recognition technology has achieved unprecedented accuracy, and its applications have extended beyond security monitoring to medical diagnosis [25].

Importance of Adopting AI

Based on the limitations of facial feature extraction, the objective and standardization of facial diagnosis cannot be achieved without interdisciplinary integration with AI. In clinical work, it is difficult for patients to accurately grasp the real crux of the disease due to privacy, forgetting, thinking insignificant, or deliberately concealing. Through AI, deeper and more abstract feature extraction of the depression state of patients can be conducted and qualitative and quantitative analysis of facial features is realized according to feature selection and information filtering. This avoids some inevitable objective effects in the process of facial diagnosis, thus making the diagnosis more standard and accurate. Also, it reduces the workload of doctors and provides accurate auxiliary references for clinical doctors [26].

AI, Mmachine Learning, Deep Llearning, and CNN

AI is a new technical science that studies and develops theories, methods, technologies, and application systems for simulating, extending, and expanding human intelligence [27]. Machine Learning (ML) is a method of artificial intelligence. The algorithm design concept of machine learning is that the computer can automatically 'learn' the algorithm [28]. The core of machine learning is to automatically analyze and obtain rules from data and predict unknown data by using rules. Since learning algorithms usually involve a large number of statistical theories, machine learning and inference statistics are closely related, also known as statistical learning theories. Machine learning is widely used in data mining, biometric identification, search engines, medical diagnosis, and credit card fraud detection. Deep learning is a branch of machine learning, and it imitates human learning methods used to acquire knowledge. The related algorithms are called artificial neural networks because they are inspired by the brain's structure and function. The neural network is a method to realize deep learning. It is a system similar to human brain neurons, and it can complete various computing tasks faster. (4) CNN is a type of feedforward neural network with convolution computation and deep structures, and it has the ability of representation learning. A CNN consists of the input layer, convolution layer, ReLU layer, pooling layer, and full connection layer. Compared with other deep learning structures, CNNs can achieve better results in image and speech recognition. Therefore, the VGG network, residual network, and Inception3 model are used to train and face recognition models [29]. The relationship between AI, ML, DL (Deep Learning), and CNN is illustrated in Figure 6.

Face Recognition

Deep learning was applied to face recognition in 2012, and feature extraction was completed by a neural network. Currently, deep learning has achieved great success in face recognition. Different from traditional recognition methods, this method involves two basic steps: manual feature extraction and classifier judgment, which mainly rely on the characteristics of automatic learning to capture the deep connection of face data [30,31].

CNN is the most common neural network structure for face recognition. It is a deep feedforward model that updates parameters by reverse propagation. To obtain better results, it is generally necessary to design the kernels of the convolution layer and pooling layer and constantly combine the convolution layer, pooling layer, and fully connected layer to obtain better image features [32].

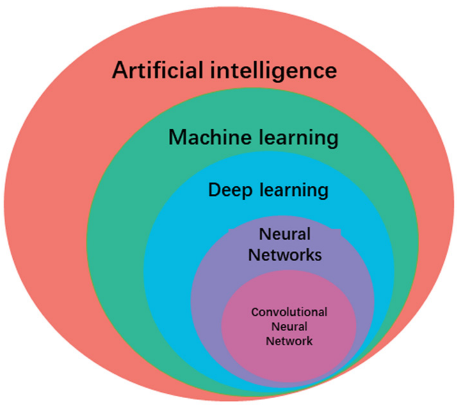

Figure 6: Interrelationship diagrams of AI (Artificial Intelligence), ML (Machine Learning), DL (Deep Learning), and CNN (Convolutional Neural Network), illustrating the connections and distinctions between these related technologies. The image was created using the Microsoft PowerPoint tool.

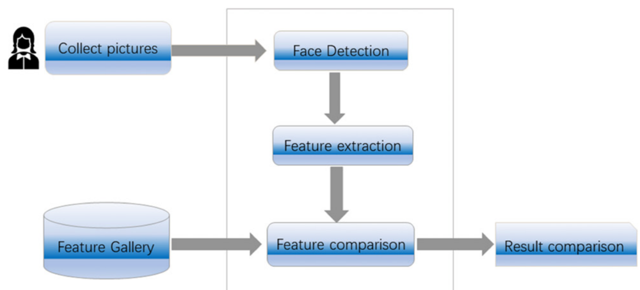

Figure 7: Flowchart of face recognition technology, depicting the steps involved in identifying and verifying individuals based on facial features. The image was created using the Microsoft PowerPoint tool.

Generally, the process of CNN-based face recognition includes image acquisition, face segmentation, feature acquisition, matching, or recognition [33]. Face recognition is to detect and locate the face in static or moving through image acquisition equipment, extract and select facial features through corresponding techniques, and finally match and identify the captured facial features and the face information in the pre-collection and input image library to confirm the identity of the identifier based on geometric features. Image acquisition and face segmentation are the premises of face recognition, and matching and recognition are the keys to face recognition systems (Figure 7).

Application of Deep Learning in Facial Recognition of Genetic Diseases

Since 2017, deep learning has been used in face recognition mainly for diagnosing genetic diseases. Gene mutations can lead to changes in the patient's face, and many genetic diseases are reflected in facial features. In 2019, Yaron Gurovich and Y air Hanani et al. published an article in the Journal of Nature Medicine. They identified the facial phenotype of genetic disorders based on deep learning, which pushed the identification of genetic disorders through face recognition to the climax. In this paper, a facial image analysis framework called DeepGestalt is proposed, which uses computer vision and deep learning algorithms to quantify the similarity of hundreds of syndromes. In the initial experiment, DeepGestalt performed better than clinicians. The model was trained on a dataset consisting of more than 17000 images, which represent more than 200 syndromes and are managed by a community-driven phenotyping platform [34].

Angelman Syndrome (AS), also known as angel syndrome, arises from various genetic mechanisms that disrupt the maternal expression of the UBE3A gene, including deletion, Paternal Uniparental Disomy (UPD), UBE3A pathogenic variants, or imprinting defects. AS is characterized by developmental delays, excessive laughter, speech impairments, distinctive EEG patterns in epilepsy, microcephaly, and various facial features, including a large mouth, prominent tongue, midfacial depression, and protrusion [35]. Researchers analyzed image and molecular data of 261 AS patients aged from 10 months to 32 years using the facial recognition system DeepGestalt, and the analysis results demonstrated significant differences in distinguishing deletion subtypes from UPD or imprinting defects, with less differentiation among UBE3A pathogenic variant subtypes [36].

Cornelia de Lange syndrome (CdLS) is a genetic condition with severe neurodevelopmental issues, including mental retardation and distinctive facial features. It affects around 1/10,000 to 1/30,000 people [37]. Common CdLS facial features include thick eyebrows, long eyelashes, a small nose, and a drooping mouth. Some patients develop normally with a standard head circumference [38]. The application of DeepGestalt and Face2Gene technology is significantly conducive to genetic disease diagnosis and management. In a study of 49 individuals with known CdLS gene variants, Face2Gene showed an accuracy of 97.9% in clinical CdLS diagnosis [39].

Down's syndrome or 21-trisomy syndrome, is a disease caused by additional staining on chromosome 21 [40]. Children who suffer from this disease have obvious special facial signs, such as wide eye distance, low nasal root, small eye fissure, lateral oblique, salivation, etc. Bosheng Qin and Letian Liang developed a recognition method for Down's syndrome using facial images and the CNN network [41]. The training of the network is divided into two main steps: using 10562 subjects' faces to form a general face recognition network, and then using the trained network for transfer learning. The data set of transfer learning is composed of 148 patients with Down's syndrome and 257 healthy subjects. The accuracy of the CNN network in identifying Down syndrome was 95.87 % [42]. Turner's Syndrome (TS) is a disease caused by partial or complete deletion of the X chromosome in female individuals [43]. The most common facial deformities of TS include a high arched palate, low posterior interline, pigmented nevus, small jaw deformity, and posterior rotation ears [44,45]. Zhouxian Pan and Zhen Shen et al. established a facial diagnosis model for TS. The training data included 170 TS patients and 1053 healthy subjects. 2 TS patients and 35 controls were used to test the efficacy in a real clinical environment. Experimental results showed that the average sensitivity and specificity of the study were 96.7 % and 97.0 %, respectively [46].

Williams-Beuren Syndrome (WBS), also known as cocktail syndrome, has a typical 'elves' facial profile. The face of 'Little Elf' is characterized by a wide forehead, periorbital edema, flat nose, etc [47]. Hui Liu and Zi Hua Mo conducted a study involving 104 WBS children, 145 healthy subjects, and 91 patients with other types of genetic syndrome. The classification performance of the face recognition model was evaluated by fivefold cross-validation and compared with human experts. The evaluation results showed that the VGG-19 model achieved the best performance, and the accuracy and precision were 92.7% and 94%, respectively [48]. Hang Yang and Xin-Rong Hud trained automatic facial recognition models on datasets composed of Noonan syndrome patients, healthy children, and subjects with several other malformation syndromes. A new DCNN framework with the arc loss function (DCNN-Arcface) was constructed, and transfer learning and data expansion were adopted in the training process. The DCNN-Arcface model was compared with two traditional machine learning models, the DCNN-CE model, and six doctors. Experimental results indicated that the DCNN-Arcface model achieved high accuracy in distinguishing NS from non-NS [48].

Genetic Syndromes (GSs) are a series of symptoms caused by genetic diseases [49]. Each specific genetic syndrome has specific characteristics according to the development affected by abnormal genes or chromosomes. Many GSs have obvious facial deformities, and facial gestalt can be used as a diagnostic tool for identifying syndromes [50]. Dian Hong and Ying Yi Zheng et al. collected 456 positive photos of 228 GS children and 228 healthy children. Meanwhile, the VGG-16 model was pre-trained by adopting the transfer learning method. Then, the VGGG-16 model was compared with five doctors, and the highest accuracy of the VGGG-16 model in screening GSs was 88% [51]. Rong-Min Baek et al. utilized a deep learning algorithm, the Multitask Cascaded Convolutional Neural Network (MTCNN), to develop a highly accurate facial recognition model for diagnosing Velocardiofacial Syndrome (VCFS). The model exhibited a high accuracy ranging from 94.03% to 99.78%, demonstrating its potential for early detection and treatment of rare genetic diseases in children, thereby offering support to healthcare professionals.

Application of Deep Learning Method in Facial Recognition of Non-Genetic Diseases

In daily social activities, people tend to judge others' health status according to their appearance. Numerous studies have shown that healthy people are often considered more attractive. This can be explained by Darwin's evolutionary theory: this preference evolves because these 'attractive' features provide a signal of the quality of life, especially physical health, and keeping away from people who are considered less attractive (unhealthy) may prevent infectious diseases [52]. Most studies focused on the association between health status and facial features, such as skin color, facial obesity, and symmetry [53,54].

Facial features of acromegaly include tooth spacing widened, protrusion, frontal bone increased, nose increased, zygomatic arch prominent, and skin thickening [55]. Xiangyi Kong and Shun Gong et al. trained several popular machine learning algorithms such as Language Models (LM), K-Nearest Neighbors (KNN), Support Vector Machines (SVM), Random Forests (RT), CNN and EM (Expectation Maximization) with a dataset consisting of 527 patients with acromegaly and 596 normal subjects. Then, the trained model was evaluated by a single dataset, and it was found that the CNN model achieved the highest accuracy of 96 % and specificity of 96 % [56]. As for children with autism, their faces have several characteristics: wider upper face, larger eyes, relatively short middle part of the face (including cheeks and nose), and a wide mouth [57,58]. Fawaz Waselallah Alsaade developed a model to help the community and psychiatrists detect autism through facial feature experiments. The pre-training models of Exception, VGG-19, and NASNET Mobile were used for classification tasks. Then, these models were tested on a dataset consisting of 2940 face images. The Exception model achieved the highest accuracy of 91%.

The most obvious early symptoms of Parkinson's disease include trembling, limb stiffness, motor dysfunction, and gait abnormalities. Many patients with Parkinson's disease often have facial rigidity, no expression, reduced binocular rotation, and reduced blinks [59]. For these patients, it seems that their faces are wearing a stiff mask, so this symptom is also called 'mask face'. Bo Jin and Yue Qu et al. collected facial expression videos from patients with Parkinson's disease and matched controls. Then, relative coordinates and position jitter were adopted to extract facial expression features (facial expression amplitude and jitter of facial small muscles) from the key points of the face. Traditional machine learning algorithms and advanced deep learning algorithms were adopted to diagnose Parkinson's disease. Experimental results indicated that applying the Long- and Short-Term Model (LSTM) neural network to extract key features can achieve an accuracy of 86%. Premature senescence syndrome refers to the phenomenon of physiological aging, physical decline, and psychological weakness in middle-aged people due to various reasons. A general framework for the diagnosis of neonatal premature failure syndrome was proposed in our previous study. Dhairya Chandra proposed an improved VGG-16 model, and they used a variety of machine learning algorithms and feature extraction tools. Results showed that the VGG-16 model obtained the best accuracy of 99.8%.

Facial dermatosis patients such as acne vulgaris, psoriasis, rosacea, and seborrheic dermatitis suffer from serious physiological and social problems. Evgin Goceri studied five common facial skin diseases that can lead to anxiety, depression, and even suicide. The pre-trained DenseNet201 structure and a new loss function were used to classify skin lesions. Experimental results indicated that the method can classify lesions efficiently (the highest accuracy is 95.24%) [60]. Facial neuritis, commonly known as facial paralysis, is a disease characterized by motor dysfunction of facial expression muscles. The general symptom is a skewed mouth and eye, and patients often cannot complete the most basic movements such as raising eyebrows, closing eyes, and drumming mouth [61]. Anping Song and Zuoyu Wu trained a single CNN in an end-to-end manner, with only pixels and disease labels as inputs of the model. The CNN was trained using the dataset of 1049 clinical images. With the help of neurologists, the dataset was divided into seven categories according to the classification criteria. The results of the model matched the level of neurologists, and the classification accuracy was 97.5%.

Cancer has become the second leading cause of death in the world. Most cancers are caused by gene mutation, which affects metabolism and leads to facial changes [62]. Bin Liang and Na Yang et al. collected facial images of cancer patients and established a dataset of cancer facial images. Meanwhile, according to the gender and age distribution of the cancer face image dataset, the face image dataset of non-cancer patients was established by randomly selecting images from the public Megage dataset. A residual neural network was constructed to classify cancer and non-cancer cases. Besides, the guided gradient weighted class activation mapping was used to reveal the related features. The study collected 8124 facial images of cancer patients in total. Experimental results indicated that the average facial obesity of male and female cancer patients is more obvious than that of non-cancer patients. On the test data set, the model accuracy is 82 % [63,64]. The typical features of beta-thalassemia include small foramen, epicanthus, low nose, flat middle face, short nose, smooth middle lip, thin upper lip, and underdeveloped mandible. BO JIN and LEANDRO CRUZ used relatively small datasets for computer-aided facial diagnosis of single and multiple diseases (beta-thalassemia, hyperthyroidism, Down's syndrome, and leprosy). Through deep transfer learning of face recognition, the Top-1 accuracy can reach more than 90%, which is better than the performance of traditional machine learning methods and clinicians [65].

The prevalence of coronavirus 2019 is one of the most severe challenges facing the global health system today. Oleg Kit identified people under 50 years old who were susceptible to 2019 coronavirus disease by a non-invasive method. The pre-trained Dlib face recognition model was employed to analyze 525 facial photos of 2019 patients with different symptoms of the coronavirus disease. Meanwhile, a CNN was used to obtain the face description vector. On the test data set, the accuracy of binary classification for the individual severity of the coronavirus disease is 84%. Constitution classification is the basis and core of constitution research in traditional Chinese medicine. Er-Yang Huan and Gui-Hua Wen proposed a method for constitution classification based on transfer learning. First, the DenseNet-169 model trained with ImageNet was applied [66]. Then, the DenseNet-169 was carefully modified according to the physical characteristics, and the modified model was trained with clinical data to obtain the physical identification network ConstructionNet [67]. To further improve the classification accuracy, the model was combined with VGG-16, Inception v3, and DenseNet-121. Finally, the model was tested according to the idea of ensemble learning, and the composition type of the input face image was determined.

In 2022, researchers including Xinru Kong developed a depression recognition method using facial images and deep convolutional neural networks, which can effectively distinguish depressed patients from healthy individuals through binary classification. The trained neural network models achieved high accuracy, with the accuracy of the FCN model reaching 98.23%, demonstrating their potential for rapid and precise automatic depression identification [68]. Xiaofei Zhang and his colleagues proposed using a deep learning algorithm to recognize Schizophrenia (SCZ) patients based on their facial images. The study is divided into two parts, and the trained CNN achieved an accuracy of 95.18% in classifying "healthy control" or "SCZ patient." The research findings suggest that facial expressions have the potential to serve as indicators of SCZ and could be applied to recognition on mobile devices in clinical and everyday life.

Main Methods

Since 2017, an increasing number of deep learning methods have been applied to identify diseases based on facial features. There are different innovations in the CNN architecture, mainly including parameter optimization, regularization, structural reorganization (convolution layer, pooling layer, and activation function), loss function, and fast processing. Note that different innovative approaches have been explored based on the survey of the latest CNN technologies for disease recognition and classification [69].

Meanwhile, three methods are adopted to design new CNN architectures. One is the structural modification of traditional models for diagnosing Down Syndrome, Noonan Syndrome, and other genetic disorders. Another is transfer learning used to diagnose Williams-Beuren syndrome, Autism spectrum disorder, progeria syndrome, facial dermatological disorders, beta-thalassemia, etc. The other is the construction and validation of deep learning models based on existing studies, such as the models developed for diagnosing genetic disorders, Corneliade Lange syndrome, Langer syndrome, and Angelman syndrome. Besides, other methods such as logistic regression, SVM, random forest, and machine learning are used to diagnose Parkinson's disease [70].

Features of the Ddisease Facial Dataset

As a data-driven method, the face recognition method based on deep learning requires a large amount of training data, and the development of datasets also reflects the progress of face recognition technology. Since 2017, the technology of artificial intelligence has been successively applied to recognize human faces to identify diseases. Meanwhile, many disease datasets have been successively collected, such as the London Medical Database (LDM), the Face2Gene and the elife database, the Kaggle platform, and the FNP dataset. Specifically, LMD contains genetic and clinical photographs and information, which has been collected for decades. In 2016, LMD was specifically integrated into the Face2Gen library to broaden its application scenario. Next Generation Phenotyping (NGP) of FDNA is used by 70% of the world's geneticists in more than 2,000 clinical sites in 130 countries /regions to capture, construct, and analyze complex human physiological data to generate operable genomes. The FDNA database includes unprecedented deep phenotype and genotype information related to more than 10,000 diseases, which are derived from patients in the real world through extensive user networks. Kaggle was released in 2010, and it is an online platform for data mining and forecasting competitions. The dataset of autistic children came from the Kaggle platform, with a total of 2,940 face images, including 1470 face images of autistic children and 1470 face images of healthy children [58]. The FNP dataset comes from clinical images from the Department of Rehabilitation, Shanghai Tenth People's Hospital. Facial nerve paralysis (FNP dataset). It includes images of 377 males and 483 females, where 136 patients are aged under 40, 302 patients are middle-aged, and 422 patients are elderly.

Healthy Subject Data Set

Numerous face datasets have been published for healthy subjects, including LFW, [71] YTF, [72] CASIA-Webface, [73] VGG2, [74] IJB-A, [75] UMDFace, [76] MegaFace, [77] CelebA, [78] CFP-FP, [79] IQIYI, [80] MS1M-IBUG, [81] Age Database-30, [82] CALFW, [71] IJB-B, [83] MS1M-ArcFace, [84] IMDB-Face, [85] Celeb500k, [86,87] CPLFW, [88] IJB-C, [89] Glint-Mini, Asian-Celeb, [91] and DeepGlint. [92] These datasets offer ample sample data for training face recognition algorithms. Table 3 provides a detailed overview of each dataset, including its name, release time, total number of face images, and description.

CNN Algorithms and Frameworks

Early studies tend to use multiple deep convolutional neural networks to learn multiscale fusion features of face images, such as DeepFace and DeepID [93,94]. With the development of deep convolutional neural networks, face recognition methods generally use a single network, and the network structure is mainly ResNet, such as DeepVisage [95], SphereFace [96], CosFace [97], etc. Meanwhile, the research hotspots have shifted from network structure design to loss function design. For example, L-Softmax [98], NormFace [99] ArcFace, [79] and other methods introduce the idea of metric learning into Softmax loss and improve the performance of the face recognition model.

Although CNNs are one of the most commonly used deep learning models in facial recognition tasks, some other methods can also achieve this goal. Therefore, this study explores and compares different deep learning methods to gain a more comprehensive understanding of deep learning applications in facial recognition tasks.

In addition to CNN, other commonly used deep learning methods include Language Models (LM), K-Nearest Neighbors (KNN), Support Vector Machines (SVM), Random Forests (RT), Expectation Maximization (EM), and others [100]. These methods are suitable for different types of data and tasks. LM is usually used for natural language processing tasks, such as text classification and language modeling, while KNN, SVM, RT, and EM are usually used for processing structured or non-image data. CNN is suitable for processing image or video data [101].

Traditional machine learning methods require manual selection or design of features, while CNN can automatically extract features through convolutional and pooling layers, thereby reducing the need for feature engineering and making the model more user-friendly and easier to debug [102]. However, CNN usually has a deep network structure and requires a large amount of training data and computational resources. Meanwhile, traditional machine learning methods usually have relatively simple model structures that are easy to explain and understand, but they may not perform as well as CNN when handling complex image features [103]. For facial recognition tasks, if time series information needs to be considered, such as consecutive frames in a video, LM or RNN models may be more suitable for such tasks. If structured data needs to be processed, KNN, SVM, RT, and EM may be more appropriate. If image processing is required, CNN may be the best choice. Overall, specific task requirements, dataset size, and model performance should be considered in combination when making a selection [104,105].

Prospects, Limitations, and Challenges of Facial Recognition Diseases in Clinical Diagnosis

With the continuous development of facial recognition technology, the application prospects of facial recognition diseases in clinical diagnosis are becoming increasingly broad. Facial recognition diseases can quickly and accurately diagnose various diseases, such as certain genetic diseases, cerebrovascular diseases, and tumors, by analyzing and comparing patients' facial features and other physiological data. Meanwhile, facial recognition diseases can help doctors predict, monitor, and evaluate the prognosis of diseases, providing powerful support for doctors to formulate the best treatment plans. Additionally, facial recognition diseases have advantages such as non-contact, fast speed, and low cost, which can greatly improve the efficiency and accuracy of disease diagnosis, shorten the diagnosis time, and reduce the cost of diagnosis and treatment.

Although facial recognition diseases have great application prospects in clinical diagnosis, there are also some limitations and challenges. Firstly, due to the diversity and complexity of facial features, facial recognition diseases have certain errors and false positive rates, especially in cases where factors such as race, gender, and age are different. Secondly, since facial recognition diseases mainly rely on the quality and quantity of images and data, data acquisition and storage require high costs and technical support, which limits the wide application of facial recognition diseases. Additionally, the clinical validation and standardization of facial recognition diseases need to be further strengthened to ensure their accuracy and safety. Finally, since facial recognition diseases involve sensitive issues such as personal privacy and data security, protecting patient privacy and data security is also a great challenge. Therefore, continuous efforts are needed to strengthen technical research, data management, and legal and regulatory construction, to better promote and apply facial recognition diseases in clinical diagnosis.

Conclusions

The above studies show that face recognition technology has a good clinical application prospect, and it has been applied to the diagnosis of genetic syndrome and even human diseases. As an auxiliary means of disease diagnosis or screening, face recognition technology is efficient, low-cost, and non-invasive.

Currently, facial recognition for disease diagnosis is facing a series of challenges and limitations. Among them, the reliability and accuracy of deep learning algorithms, the completeness and diversity of datasets, and the scalability and generalization ability of algorithms are important research topics. To address these issues, we are exploring various solutions, including improving the performance of existing algorithms, establishing new models, developing better datasets, and proposing new evaluation metrics.

Regarding the current research challenges and directions, our solutions include improving the architecture or parameters of existing algorithms, enhancing the quality and diversity of datasets, and using ensemble learning techniques. The implementation of these methods requires comprehensive consideration of algorithm adaptability and feasibility, as well as in-depth theoretical and experimental analysis of the algorithms to verify their effectiveness and reliability. Therefore, future research will further explore the development direction and optimization methods of deep learning algorithms and strengthen the collection and processing of datasets to improve the generalization ability and application value of algorithms. Through these efforts, the development and application of deep learning technology in facial recognition for disease diagnosis can be further promoted.

Author Statements

Conflict of Interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

Author Contributions

Qianhui Qiu, Xiao Ding and Xinru Kong conceived and designed the project; Xianghua Qi and Ziyue Wang collected the data; Xinru Kong, Jie Sun, and Xiao Ding drafted and revised the paper, supervised the analyses, and suggested revisions of the paper. All the authors have read and approved the final manuscript.

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Data Availability Statement: The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

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