Use of machine learning methods and virtual reality to analyze genetic characteristics[*]

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Abstract

Recent advancements in computer vision and virtual reality (VR) have introduced new possibilities for diagnosing genetic disorders based on facial feature analysis—phenotypic characteristics. This study provides an overview of practical implementations of VR in medicine, as well as facial image processing methods, including preprocessing, key point detection, and classification using machine learning algorithms such as Support Vector Machines (SVM) and Convolutional Neural Networks (CNN). The potential integration of VR into clinical practice is examined, including the development of interactive training scenarios for physicians and the application of 3D modeling for analyzing rare genetic syndromes. The study discusses the prospects of implementing VR simulations for testing facial anomaly recognition algorithms and remote patient diagnosis. Additionally, key challenges related to algorithm accuracy, the accessibility of VR solutions, and the need for inclusive datasets are highlighted. The integration of VR and machine learning into the diagnostic process enhances the accuracy of medical decision-making and expands the potential of personalized medicine.

Keywords

machine learning, genetic disorder diagnosis, facial feature analysis, virtual reality, VR simulations, 3D modeling, CNN, medical diagnostics

1. Introduction

Virtual reality technologies open up wide opportunities for the rehabilitation of patients with various impairments, providing significant advantages over traditional methods. VR provides full immersion in a virtual environment, simulating conditions close to real ones, which improves the perception of exercises, stimulates participation and simplifies the implementation of complex tasks.

One of the key advantages is the individualization of therapy. Unlike traditional methods, it creates realistic scenarios tailored to individual patient needs, taking into account their cognitive and physical capabilities. In addition, VR creates a safe space, minimizing the risk of injury, which is especially important for patients with impaired motor coordination, such as after a stroke or with Parkinson's disease. For example, a stroke survivor relearning hand coordination can engage in VR-based exercises that simulate everyday tasks, such as picking up objects or preparing a meal, in a risk-free environment. Similarly, patients with Parkinson’s disease can practice movement control through virtual balance and gait training exercises, helping them regain confidence in their mobility.

VR technologies allow for an objective assessment of rehabilitation progress, recording parameters such as speed and accuracy of movements, which simplifies the adjustment of the treatment program. Additionally, One of the most compelling aspects of VR-based rehabilitation is its ability to boost patient motivation through gamification. VR overcomes this challenge by integrating interactive, game-like elements into therapy sessions, making the process more engaging - especially for children with cognitive impairments.

Another advantage of VR is its potential for remote rehabilitation. Patients who live in rural areas, have mobility limitations, or struggle with transportation can access therapy sessions from

[*] *AIT 2025: 1st International Workshop on Application of Immersive Technology, March 5, 2025, Almaty Kazakhstan*

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home using VR headsets. This increased accessibility reduces barriers to consistent treatment, leading to better long-term recovery outcomes. VR technologies show high potential for the rehabilitation of patients after stroke, with Parkinson's disease and children with cognitive disorders, combining safety, effectiveness and an innovative approach.

This article is devoted to the development a novel VR-based respiratory rehabilitation system that integrates Strelnikova breathing exercises into an interactive virtual environment using Meta Quest 3.

In recent years, there has been a rapid increase in interest in the application of virtual reality (VR) and computer vision technologies in medicine. The number of publications on the topic of "VR technology in medicine" in the PubMed database increased from 58 in 2017 to 145 in the first half of 2021 [1, 2].

Modern methods for diagnosing genetic diseases require significant resources, time, and the involvement of highly qualified specialists. Moreover, traditional approaches based on genetic testing and clinical analysis involve high financial costs, limiting accessibility for patients in certain regions and social groups. For example, according to a study by Smith et al. (2021), the cost of genetic testing in some countries ranges from $500 to $2,000 per patient, creating a substantial financial burden on healthcare systems [3]. In Almaty, in major private medical laboratories such as Invivo and Invitro, the cost of testing for major hereditary diseases is 328,730 tenge [4]. Whole Genome Sequencing with a geneticist's report costs 538,040 tenge and 690,000 ₸, respectively [5]

Traditionally, genetic research faces challenges related to processing vast amounts of information and the complexity of determining relationships between genes, proteins, and other biomolecules. Analytical procedures often lack clarity in identifying hidden patterns and dynamic processes. Furthermore, such procedures can take considerable time, sometimes, with results requiring several days to weeks, significantly delaying treatment initiation and reducing its effectiveness.

The integration of VR and computer vision offers the possibility of creating interactive and immersive environments in which researchers can:

1. Interactively visualize multidimensional genetic data in a three-dimensional space, facilitating a deeper understanding of their structure and relationships.
2. Detect anomalies and patterns using computer vision algorithms adapted for the specificity of biological data.

The use of VR and machine learning methods not only automates the analysis process but also enables the creation of interactive tools for physicians and researchers. A study by Ivanov et al. (2023) demonstrated that the implementation of VR technologies in the genetic disease diagnostic process can reduce analysis time by up to 40% compared to traditional methods [6].

Thus, the integration of VR and computer vision into medical diagnostics represents a promising direction that can reduce diagnostic costs through process automation, shorten the waiting time for analysis results, accelerate treatment initiation, and improve diagnostic accuracy through machine learning algorithms and immersive technologies.

The motivation for this research is high, as the application of VR and computer vision in genetic studies can significantly accelerate the discovery of new genetic patterns and relationships, which is of great value for both fundamental science and practical medicine. A better understanding of the genetic characteristics of individual patients contributes to the development of personalized treatment approaches, which is a key focus of modern medicine.

The aim of this review is to explore the potential of virtual reality and computer vision for the automated analysis of facial phenotypic features associated with genetic diseases. The following sections analyze existing VR and machine learning technologies in medical diagnostics, identify key computer vision methods applicable to facial anomaly analysis, and examine successful case studies of implementing these technologies in clinical practice.

2. Overview of Technologies and Methods

Virtual reality (VR) is actively being implemented in medical practice, offering new opportunities for diagnostics, surgical planning, and professional training. VR enables the creation of interactive 3D models of a patient's anatomical structures, allowing physicians to thoroughly examine specific cases before performing surgical interventions. For instance, the Surgical Theater platform is used for neurosurgical planning, enabling the development of interactive 3D and VR models [7]. At the Stanford Simulation and Virtual Reality Center for Neurosurgery, this technology is employed to create 360-degree virtual models of patients' brains, enhancing surgical planning and improving treatment efficiency.

Figure 1: Precision VR™, Surgical Theater's VR medical visualization platform in use

Also, numerical modeling plays a significant role in simulating biological systems. For example, [8] conducted a study using ANSYS Fluent to simulate blood flow in coronary arteries, demonstrating the importance of computational fluid dynamics (CFD) in cardiovascular analysis and supporting the broader integration of modeling technologies into medical research.

Additionally, VR is used in the rehabilitation of patients after strokes and injuries. Specialized programs allow patients to control their movements in a virtual environment, which contributes to motor function recovery. According to a study by Dolganov and Karpova, the use of VR in conjunction with standard rehabilitation programs in the acute phase of a stroke improves upper limb function and reduces limitations in daily activities [9]. In another case, these technologies have demonstrated effectiveness in rehabilitation and biofeedback therapy for patients with impaired fine motor skills after an ischemic stroke [10].

Machine learning methods, combined with computer vision, play a crucial role in facial data analysis for diagnosing various diseases, including genetic syndromes. The key algorithms include neural networks, Support Vector Machines (SVM), and the k-Nearest Neighbors (kNN) algorithm. Additionally, convolutional neural networks (CNN) enable automatic recognition and classification of facial features due to their ability to extract complex patterns from data and process spatial dependencies [11]. First of all, the architecture consists of a Convolutional Layer that highlights spatial features such as edges, textures, and shapes by applying filters to the input data (Figure 2). Then comes the Pooling Layer, which reduces the dimension of the data, preserving the most significant features, which reduces computational complexity. The third Fully Connected Layer converts selected features into classes or results, for example, for the diagnosis of syndromes.

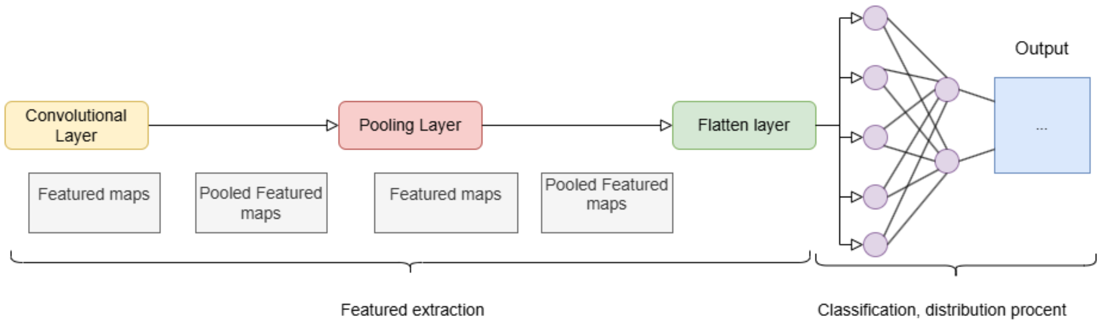

Figure 2: The typical architecture of CNN

SVM is widely used for high-accuracy data classification and regression tasks, particularly when dealing with limited datasets. However, it has limitations—it is computationally intensive for large datasets and sensitive to the choice of kernel and its parameters. The core principle of SVM is finding a hyperplane that best separates data points into different classes [12]. Key components of an SVM model include:

1. Hyperplane – a decision boundary that maximally separates different classes in multidimensional space.
2. Support vectors – data points closest to the hyperplane that determine its position.
3. Regularization parameter (C) – balances the margin maximization between classes and classification error minimization.
4. Kernel functions – transform data into higher-dimensional space where classes become linearly separable. Examples include linear, polynomial, and Radial Basis Function (RBF) kernels.

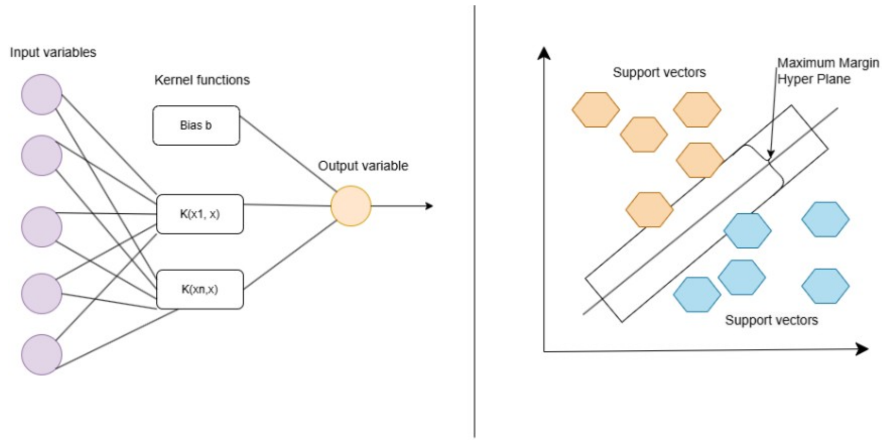

Figure 3: The typical architecture of SVM algorithm

Advanced CNN models such as ResNet and Inception offer improved accuracy in medical image analysis, exceeding 90% in certain diagnostic tasks [13]. ResNet is a deep network with residual connections that solve the problem of gradient attenuation. Inception uses multiscale convolutions, which allows to identify features of different levels of complexity at the same time. This makes the model universal for analyzing various types of data. EfficientNet optimizes accuracy and computational costs by using compound scaling, i.e. changing the depth, width, and resolution of the network at the same time.

Various methods are employed for 3D facial scanning, each with its advantages and limitations. Light Detection and Ranging (LiDAR) utilizes laser pulses to measure distances to objects, creating highly accurate three-dimensional models [14]. This method provides superior precision and detail, making it particularly useful for tasks that require precise facial geometry reconstruction. However, LiDAR technology is expensive and often requires specialized equipment and controlled scanning environments. Additionally, LiDAR devices can be sensitive to bright sunlight or atmospheric interference, affecting their performance in outdoor conditions. The second method -photogrammetry, is based on analyzing multiple images of an object taken from different angles. These images are then processed using specialized software to create a 3D model [15]. Compared to LiDAR, photogrammetry is more accessible, as it requires only standard photographic equipment. However, its accuracy and level of detail may be lower than that of LiDAR, especially if the image quality is poor or the number of captured photos is insufficient. Additionally, the processing time for generating a model can be computationally intensive and time-consuming.

Third method - 3D Reconstruction Deep learning techniques enable the reconstruction of 3D facial models from 2D images. Algorithms such as CNNs are trained on large datasets to predict three-dimensional facial structures based on two-dimensional photographs [16]. These approaches are rapidly evolving and have shown promising results, allowing the generation of 3D models without the need for specialized scanning equipment. However, the accuracy of such models depends on the quality and diversity of training data, as well as the complexity of the neural network architecture.

3. Challenges and Risks

Despite the rapid development of computer vision and VR technologies, their application in medical diagnostics faces several challenges. First and foremost, the accuracy and reliability of algorithms capable of identifying facial features characteristic of genetic disorders are crucial. Achieving this requires large datasets and thorough model validation. The limited availability of datasets for rare syndromes poses difficulties in model generalization. Studies indicate that machine learning models can exhibit bias based on ethnicity, gender, and age, primarily due to imbalanced training datasets [17].

One key recommendation is the implementation of bias mitigation techniques and the development of more inclusive datasets. This is particularly important in regions with diverse ethnic compositions, such as Kazakhstan and Central Asia, where ethnic characteristics may influence the expression of facial features. Research suggests that data augmentation can improve algorithm accuracy by up to 15% [18].

Secondly, high-precision VR devices and computer vision systems can be expensive, limiting their use in low-resource regions. The cost of specialized hardware and software may hinder widespread adoption, making affordability a key challenge for integrating these technologies into clinical settings. Thirdly, medical professionals must understand how AI-driven algorithms make decisions to build trust in automated diagnostic methods. The interpretability and transparency of these algorithms are essential for their acceptance in medical practice.

Additionally, the use of facial images in medical diagnostics raises ethical and legal concerns. Strict data privacy regulations, such as the General Data Protection Regulation (GDPR), must be adhered to when collecting and processing biometric data. Violations of privacy in facial image data handling can have significant consequences for patients and their families. Addressing these ethical concerns is critical for ensuring the responsible deployment of AI and VR technologies in medical diagnostics.

4. Phenotypic Features in Diagnosis

Phenotypic manifestations refer to a set of external traits that arise from the interaction of genes influencing growth, development, and the function of tissues and organs. Human facial structures are shaped by complex genetic interactions, which determine not only basic proportions but also unique facial characteristics. For instance, mutations in genes involved in morphogenesis, such as TCF4 or FGFR2, can lead to craniofacial deformities.

One well-known example is Down syndrome, which results from trisomy of 21st chromosome. It is characterized by a flat facial profile, almond-shaped eyes, a short nose, and distinctive hand features. These physical traits help physicians diagnose the condition at an early stage.

Another case is Marfan syndrome, caused by a mutation in the FBN1 gene. This disorder is associated with tall stature, long fingers (arachnodactyly), and distinct facial features such as a narrow lower jaw. Marfan syndrome is also linked to systemic complications, including cardiovascular abnormalities.

According to the American Journal of Medical Genetics, approximately 60% of genetic syndromes exhibit distinct external phenotypic features [19]. These findings emphasize the crucial role of facial analysis in diagnosis, particularly for rare diseases where genetic testing may not always be available. Thus, machine learning (ML) and computer vision provide new opportunities for automated diagnosis, enabling the identification of patterns associated with specific genetic conditions.

5. Successful Cases in Disease Diagnosis and Specialist Training

One of the most successful cases in computer vision-based genetic diagnostics is DeepGestalt, a system that utilizes convolutional neural networks (CNNs) for facial image analysis. It has been tested on over 10,000 images, achieving 91% accuracy [20]. Another notable example is Face2Gene, which is widely used in clinical practice. This system enables the diagnosis of genetic disorders based on facial images by leveraging large annotated databases. These databases contain phenotypic markers, including facial characteristics and other manifestations of genetic syndromes. According to clinical trials, Face2Gene has demonstrated an accuracy of 89% for Down syndrome and 82% for Noonan syndrome [11]. Both systems rely on large biometric datasets, allowing them to learn from diverse phenotypic patterns and improve diagnostic precision.

In the field of medical training, the VR-NRP platform was developed for neonatal resuscitation training [21]. It provides a realistic and interactive VR environment, where medical professionals can practice life-saving techniques on newborns, increasing their confidence and efficiency in real-world scenarios. Another example is a mixed reality system for medical procedures, such as central venous catheter insertion [22]. This technology allows remote experts to guide local practitioners through medical procedures, enhancing training quality and reducing errors. Additionally, the SONIA system provides interactive VR-based neuroanatomy training [23]. It enables students and educators to explore complex brain structures, improving comprehension and knowledge retention.

6. Computer Vision Image Analysis Methods and Virtual Reality Integration

Various types of data are used for genetic disease analysis, each contributing to different aspects of facial feature recognition and classification:

1. 2D Photographs constitute the primary data type for most machine learning-based facial analysis systems. Modern research in machine learning for facial image processing relies on publicly available datasets such as Labeled Faces in the Wild (LFW) and CelebA [24]. These datasets provide high-quality images that are widely used for training and testing deep learning models.
2. 3D Images allow for a more precise analysis by creating volumetric models of facial structures. For example, FaceBase is a database containing 3D facial scans of individuals with craniofacial anomalies. Additionally, the 3D Facial Alignment in the Wild (3DFAW) dataset includes 3D face scans captured in various expressions and lighting conditions, making it valuable for developing robust models resistant to external factors.
3. Biometric Data – key facial landmarks, such as the distance between critical facial points, are used to build models for facial structure analysis and disease detection.

Stages of Data Processing:

- Preprocessing – This step involves noise removal, image normalization, and facial alignment, ensuring consistency across the dataset.
- Augmentation – Since high-quality medical datasets are often limited, augmentation is used to expand training data. This includes image transformations such as rotation, scaling, and noise addition. Tools like Albumentations and TensorFlow ImageDataGenerator are widely applied to enhance model robustness, particularly in scenarios with small datasets.
- Data Cleaning – The removal of duplicate images and mislabeling corrections ensures that only high-quality and accurately labeled data are used for training.

Facial landmark detection involves identifying specific facial points, such as the eyes, nose, and mouth, to determine the structure of the face. This process consists of preliminary face localization using models like MTCNN (Multi-task Cascaded Convolutional Neural Network) or Haar-Cascade. A multi-task cascaded deep convolutional neural network (Figure 4) is a method consisting of three convolutional networks that work in stages: first, coarse detection, then refinement, and finally localization of key points. MTCNN, implemented in PyTorch or TensorFlow, detects faces and simultaneously identifies the eyes, nose, and mouth. It is particularly effective for handling variations in pose, lighting, and occlusion. Haar-Cascade, on the other hand, analyzes pixel groupings to detect potential face regions by applying pretrained feature classifiers. Although Haar-Cascade is computationally efficient, it is generally less accurate compared to deep learning-based models like MTCNN.

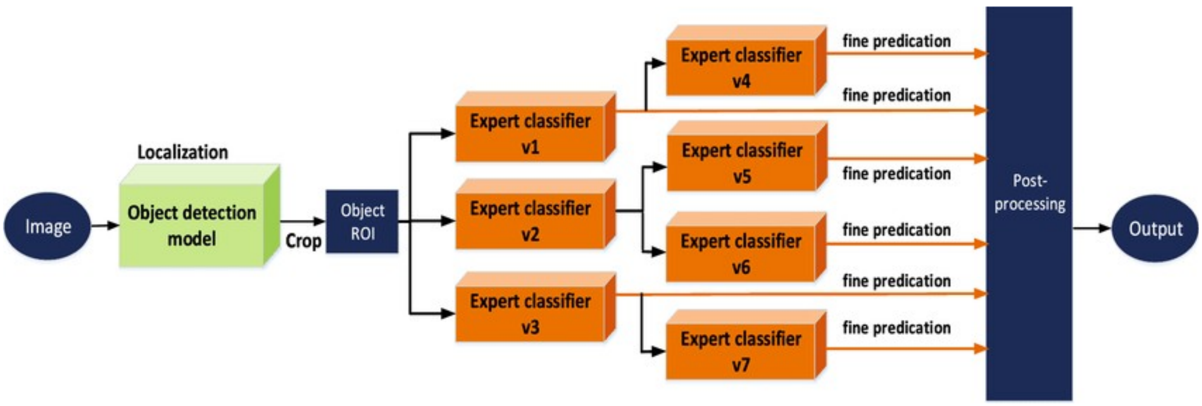

Figure 4: Multi-task cascade deep convolutional neural (MTCD-CNN) architecture [25]

The second stage involves detecting key points, such as the corners of the eyes, the tip of the nose, and the mouth. Convolutional neural networks trained on annotated datasets such as 300-W, where facial landmarks were manually labeled, are used for this task [26]. For more diverse conditions, including images taken from different angles, the Annotated Facial Landmarks in the Wild (AFLW) dataset is applied. Feature extraction refers to identifying significant image characteristics, including: geometric features – distances between key facial points; gradients and textures – methods such as Histogram of Oriented Gradients (HOG) and Local Binary Patterns (LBP). On one hand, HOG extracts gradient directions in small image regions to derive texture-based characteristics. On the other hand, LBP compares pixel values within a small window, generating binary patterns [27]. Tools such as OpenCV, a library used for preprocessing, face localization, and feature analysis, and Dlib, which predicts 68 facial landmarks for structural analysis, are widely used for implementing these methods.

Various metrics are used to assess the efficiency of developed models, including accuracy, recall, and the F-measure. Validation is performed on test datasets that were not used during the training process to ensure objective evaluation. Additionally, cross-validation methods are applied to enhance the reliability of the results.

The integration of computer vision and VR into medical practice enhances diagnostic accuracy and training efficiency, providing new tools for analyzing and visualizing complex medical data.

Thus, the combination of VR, computer vision, and advanced 3D scanning methods opens new perspectives for medical diagnostics and treatment, contributing to greater precision and efficiency in medical procedures.

7. Prospects for future research

Despite significant advancements in computer vision and VR, several unresolved challenges require further investigation:

1. Enhancing the accuracy of facial feature recognition algorithms – Developing more balanced datasets that consider ethnic and age diversity among patients.
2. Creating more accessible VR systems that can be implemented in clinical settings.
3. Developing VR interfaces that enable interaction with patient biometric data for more precise diagnostics.
4. Analyzing the impact of VR simulations on clinical decision-making and diagnostic accuracy.

Further research in VR and computer vision could greatly improve the precision and accessibility of genetic disease diagnostics while also enhancing medical education. The integration of these technologies into clinical practice will contribute to the development of personalized medicine and improve the overall quality of healthcare services.

8. Conclusion

This study provides a comprehensive review of modern approaches to the analysis of facial phenotypic features associated with genetic diseases using machine learning and computer vision. Key image processing techniques, including preprocessing, key point detection, and classification using deep learning models, have been examined. Additionally, the role of VR in medical diagnostics, patient rehabilitation, and physician training has been analyzed.

The application of VR and computer vision in facial data analysis enhances diagnostic accuracy, automates the recognition of genetic syndromes, and enables the development of interactive educational tools for medical professionals. The use of advanced machine learning algorithms, such as CNN, ResNet, and Inception, demonstrates high effectiveness in facial recognition and anomaly classification tasks.

The integration of VR into the diagnosis and treatment of genetic diseases can be implemented in the following ways:

1. VR Simulations for Facial Anomaly Recognition Testing. Utilization of 3D modeling of facial structures for analyzing phenotypic features of rare genetic disorders.
2. VR in Clinical Practice. Development of VR applications that allow physicians to interact with 3D facial models of patients in real-time. Creation of VR platforms for telemedicine, enabling remote consultations and diagnostics.
3. VR-Based Training and Simulations. Immersive VR training systems for teaching physicians methods for diagnosing genetic diseases. Development of virtual case studies with real patient data to enhance the qualification of medical specialists.

Declaration on Generative AI

The author(s) have not employed any Generative AI tools.

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