NEURAL NETWORK SYSTEM FOR DETECTING SIGNS OF HUMAN GENETIC DISEASES BY PHOTO*

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Abstract

This article solves the problem of a neural network system constructing to identify signs of human genetic diseases from a face photography. This study focuses on the recognition of genetic diseases signs associated with the eyes. However, by making insignificant changes and preparing new initial data, it is possible to use the system to process other parts of the face. Such system can be a relatively inexpensive and effective tool for genetic diseases diagnosing with external signs. The paper discusses some aspects of artificial intelligence systems use in medicine. An example of a software solution for diagnosing a disease based on a human face image is given. The task of disease sign recognizing in a photo includes the following main stages: detecting a face in a photo and determining its position, determining key points of the face, highlighting the desired part of the face in order to recognize specific signs of a disease, image transforming an and disease sign recognizing from an image using convolutional neural networks. The main tools used were the following: the Python programming language, libraries for computer vision and machine learning (OpenCV, dlib), the PyTorch machine learning framework. Images from open sources were taken as the initial data. The importance of the initial data quality used for the neural network training is emphasized. The method of processing the initial data is described, the structure of the used convolutional neural network is considered. In the training process, the image recognition accuracy achieved 95%. During testing, the recognition accuracy was 88%. Thus, the developed neural network model can be used for genetic diseases preliminary diagnostics indicated in the work as a doctor's assistant when making a medical diagnosis.

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1. Introduction

Currently, artificial intelligence systems are used in many areas of human activity (Ebrahimi et al., 2022; Holm et al., 2021; Mahmood et al., 2022; Mikhailov et al., 2021; Shabgah et al., 2021): photo processing, voice assistants, information search on the Internet, robotics, medical diagnostics, autonomous driving and many others. Artificial intelligence is most widely used in the medical diagnostic systems construction (Nicholls, 2018; Vatansever et al., 2021; Wissmann et al., 2021). Such systems allow not only making a diagnosis, but also helping to determine the risk of developing certain body pathologies in the early stages.

The features of artificial intelligence and machine learning use in medicine on the example of genetic diseases are described in (Pollard et al., 2021; Rath et al., 2021). A significant part of such diseases have external signs by which they can be identified. These can be some characteristic features on the face, such as a special location or size of the eyes, the size of the forehead, noticeable distortion of the face, etc. In some cases, recognizing the disease at an early stage makes it easier to cure it, or at least slow down its further progression. An intelligent system creation is an inexpensive and effective tool for identifying genetic diseases by external signs.

This paper proposes a neural network system for detecting human genetic diseases signs from a face photography. The use of such systems makes it possible to reduce the material, technical and time costs for the preliminary diagnosis of the patient, and also helps the doctor in making a medical diagnosis (Akhmetvaleev and Katasev, 2018; Daneii et al., 2022; Khorsandi et al., 2021; Ramezani Farani et al., 2022; Ratnawati et al., 2019).

2. Methods

The practical use of artificial intelligence is associated with a number of difficulties, one of which is the task of marking up the initial data used for training (Ghiran et al., 2017; Rezaei et al., 2021). For example, the signs of the disease must be correctly identified on the images, the affected areas must be correctly identified on the X-ray images, etc. In addition, difficulties are associated with the imperfection of legal regulation in the telemedicine technologies field, as well as with the processed information protection.

An important area of artificial intelligence and machine learning application in medicine is genetics (Sookoian and Pirola, 2020; Taghibeikzadehbadr et al., 2020; Tavakol et al., 2019). Although genetic disorders are rare, they account for approximately 80% of all rare diseases. According to statistics, approximately every 17th person has rare diseases (Johnson et al., 2018). Therefore, this area of medicine is not left without attention. In particular, systems to identify genetic diseases in a person's face are currently being developed (Kumov and Samorodov, 2020). Such systems are actively used both on powerful server hardware and on smartphones. The facial recognition technology use can help to reduce the number of undiagnosed genetic diseases.

The Face2Gene system is one of the intelligent systems for recognizing genetic diseases based on a human face image (Javanmard et al., 2022; Latorre-Pellicer et al., 2020; Mishima et al., 2019). It can be presented both as a web application and as a mobile application on Android and iOS operating systems (Fig. 1).

The Face2Gene system for face analysis uses its own DeepGestalt technology (Pantel et al., 2020). The input image is pre-aligned and processed to provide face recognition and landmark detection. After preprocessing, the input image is cut into several areas of the face. Each region is fed to the input of a convolutional neural network to obtain a vector indicating the probabilities of its correspondence to each syndrome. The output vectors of all neural networks are then aggregated.
and sorted to produce the final ranked list of genetic syndromes (Fig. 2). Various studies show that the Face2Gene system works effectively with real images of patients. For example, a study on 49 people with Cornelia de Lange syndrome (CdLS) was conducted (Faridizad et al., 2022; Hosseini and Khamesee, 2022; Latorre-Pellicer et al., 2020). Based on profile images of patients, the CdLS diagnosis was included in the top five predicted syndromes in 97.9% of cases and was even listed as the first prognosis for 83.7% of cases.

Fig. 1. Patient profile in the Face2Gene mobile application

Fig. 2. The image processing order in DeepGestalt
In another study of 17 Japanese people aged 20 to 40, out of 17 eligible patients, Down syndrome was successfully identified by the Face2Gene app as the highest and second most severe disease at 82.2% (14/17) and 100% (17/17) of cases, respectively (Mishima et al., 2019). These results indicate that Face2Gene is doing quite well in this task.

In this work, to solve the problem of human genetic disease signs recognizing from a face photography using a convolutional neural network, the following steps were implemented:

1) finding a face in the photo;
2) determining the key points of the face;
3) highlighting the desired part of the face to recognize specific signs of diseases;
4) transformation of the image for feeding to the neural network input;
5) recognition of a disease sign from the image.

When implementing a convolutional neural network system, the following tools were used:

- Python 3 - a high-level programming language with a large number of libraries for web development, mobile and desktop applications, machine learning, etc.;
- OpenCV - a platform for computer vision and image processing, which has a Python API interface, which allows to use it directly from Python;
- Dlib library - a set of C++ tools that contains machine learning algorithms and allows to use the library in conjunction with the Python programming language;
- PyTorch is a machine learning framework for the Python language.

Special attention is paid to the tools of the Dlib library (King, 2009), since they make it easy to find a face in the image, as well as to highlight key points of the face. In this case, the face model is uniquely specified by 68 points (Fig. 3).

By highlighting key points, it is easy to access images of different parts of the face (Suvorov and Shleymovich, 2020; Petrosyants et al., 2021; Ismagilov et al., 2019). For example, in order to get an image of the left eye, we need to take a rectangle that includes points 37-42. Therefore, in this work the Dlib library was used to carry out image processing in the first three stages. Recognition of disease signs from a photograph (stage 5) was carried out using the PyTorch framework based on a convolutional neural network.
The set of data (images) in the work was obtained using the Google search engine. An expert geneticist identified photographs of people who showed signs of certain diseases. For this, several signs of eye diseases were selected:
- healthy people;
- antimongoloid eye incision;
- blepharophimosis;
- strabismus;
- ptosis.

For each category, on average, 20-40 face images were found: healthy people - 60 images, antimongoloid eye shape - 28, blepharophimosis - 32, strabismus - 31, ptosis - 23. The reason for using a small sample of images is that there is not enough data in the free access.

The found images were placed in directories in accordance with the attribute contained in the image. Since the signs of diseases associated with the eyes were selected for this work, images of the eyes were prepared for the neural network. The set of images in this work was divided into 2 parts in an 80/20 ratio. Thus, the training set contained 80% of the images, and the test set - 20%. It should be noted that the images were not reduced to the same size, since this operation is performed automatically when training the convolutional neural network.

To implement a neural network, an architecture was chosen that included 2 convolutional layers, after each of which there was a subsample layer. Convolutional layers were followed by fully connected layers, the last layer had 5 neurons, since the neural network was designed to classify 5 signs of diseases. Eye images with a dimension of 32 by 64 pixels were submitted to the neural network input. Also, when training the neural network, data augmentation was used (Marastoni et al., 2021; Sabirov et al., 2021; Huang et al., 2021). It led to an artificial increase in samples due to the fact that various distortions were applied to the original images (image rotation, compression, blurring, sharpening, color change). This technique is often used in neural networks training. In this work, only one variant of augmentation was used - horizontal image reflection (Ismagilov et al., 2019; Hosseini, 2022). This allows to double the training sample and reduce the model overfitting effect. Examples of the neural network system operation are presented in Figs. 4 and 5. Figure 5 show the order of processing, highlighting face key points and obtaining images of the eyes.

3. Results and discussion

As a result of training, the neural network for 28 epochs reached 95% accuracy in recognizing the signs of human genetic diseases from a photograph. The average recognition accuracy during testing was 88%. The results of the convolutional neural network operation on images from the testing data set are presented in Table 1.
Table 1. Test images recognition accuracy

<table>
<thead>
<tr>
<th>Signs of eye diseases</th>
<th>Recognition accuracy, %</th>
</tr>
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<tbody>
<tr>
<td>healthy people</td>
<td>82</td>
</tr>
<tr>
<td>antimongoloid eye incision</td>
<td>83</td>
</tr>
<tr>
<td>blepharophimosis</td>
<td>75</td>
</tr>
<tr>
<td>strabismus</td>
<td>100</td>
</tr>
<tr>
<td>ptosis</td>
<td>100</td>
</tr>
</tbody>
</table>

It can be seen that all images with the signs of "Strabismus" and "Ptosis" were recognized correctly, for the sign "Blepharophimosis" the result was 75%, and for healthy people and people with the "Antimongoloid eye cut" sign the percentage of correct recognition reached 82% and 83%, respectively. It should be noted that the recognition result was influenced by the number of examples in the training and testing samples, and it is also possible that several signs of diseases were present in the same photographs. In the future, it is planned to improve the recognition accuracy by increasing the number of images in the samples, as well as increasing the correctness of their labeling.

In this article, a neural network system has been developed to detect signs of human genetic diseases from a face photography. The system uses computer vision and machine learning to recognize signs of eye diseases. However, due to the generality of this approach, its practical application is also possible for recognizing other parts of the face.

The neural network system has demonstrated a sufficiently high accuracy in classifying images corresponding to various signs of human genetic diseases. This indicates the possibility of its practical use to assist a doctor in making a medical diagnosis (Bjorgan and Randeberg, 2015; Marias et al., 2020).

5. Conclusions

Thus, the work has solved the problem of identifying signs of human genetic diseases from a face photography on the basis of constructing a convolutional neural network. The obtained results have shown the effectiveness of the proposed approach to solving the problem.

The neural network model has shown a sufficiently high accuracy in recognizing faces with various signs of human genetic diseases. This indicates its effectiveness and the possibility of practical use in medical diagnostics and imaging systems.

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