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Multiple facial image features-based recognition for the automatic diagnosis of turner syndrome

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ABSTRACT

Because of the diversity of individual clinical symptoms and the lack of reliable diagnostic criteria based on the clinical features of appearance, the initial diagnosis of Turner syndrome (TS) mainly depends on the clinical characteristic of height, resulting in many patients with Turner syndrome being diagnosed with other diseases, such as dwarfism. To improve objectivity, reduce the burden on well-experienced endocrinologists, allow screening of suspected patients in under-developed areas and provide TS patients with early detection and early treatment, a facial image analysis-based computer-aided system for automatic face classification is proposed. The system is composed of facial image pre-processing, image feature extraction, and automatic classification. First, several unique appearance features are identified in different facial regions based on clinical observations by endocrinologists, including ocular distance, epicanthus, and the numbers and sizes of melanocytic nevi. Based on the characteristics, we trained a 68 feature-points face model. Then, distance between points, Gabor wavelet filtering and spot detection are applied to extract global features and local features, respectively, and Gabor features are reduced by principal component analysis (PCA). Finally, Support Vector Machine (SVM) and the Adaboost algorithm are used for classification. Although all subjects involved in this trial are Chinese, the method achieves an average accuracy of 84.6% on the training set and 83.4% on the testing set based on K-fold cross- validation. The sustainable acquisition and accessibility of face images used for research is one of our advantages. We believe that this work can serve as an important reference for other assistant diagnosis systems related to facial images.

1. Introduction

In recent years, with the development of big data and intelligent medical technology, the use of big data to assist in diagnoses is receiving more and more attention. In particular, computer-aided healthcare systems may provide useful information from a large amount of data, especially in under-developed areas with few medical resources. An example is the automatic face classification of Turner syndrome based on facial image analysis. Turner syndrome (TS) is one of the most common sex chromosome abnormalities in females [1,2]. In addition to 45,X, the karyotype of TS may have a variety of chimaeras, such as 45,X/46,XX. TS is caused by a partly or completely missing X chromosome [3]. It occurs in approximately 1/2500 to 1.11/5000 live female births [3]. According to statistics, there are more than 400 clinical features in patients with Turner syndrome. Because of the diversity of individual clinical symptoms and the lack of reliable diagnostic criteria based on the clinical features of appearance, currently, the primary clinical clue for diagnosis in childhood is short stature, resulting in many patients with Turner syndrome being diagnosed with other diseases, such as dwarfism, in the early stages of diagnosis. Although some patients were diagnosed in infancy and early childhood, most of them were not diagnosed until the age of 10, and approximately

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22% of patients were reported to have had diagnosis delayed until after the age of 12. The earlier the diagnosis is, the better it will be for treatment. A large-scale screening method that enables objective judgement to be made for face images is urgently needed. In this paper, we demonstrated a possible solution: a computer-aided healthcare system for mass screening based on facial image analysis.

Turner syndrome is the most common chromosomal-aberrant disease. TS patients may have different clinical manifestations because of different karyotypes. Endocrinologist H.H.Turner first described the syndrome in 1938. Turner found that those patients had the same physical characteristics, such as webbed neck, torticollis, facial asymmetry, etc. [5]. To commemorate the contribution of Turner, the descendants named the disease Turner syndrome. In 1964, TS was recognized as occurring because of a chromosomal abnormality. In the more than 400 clinical features of patients with TS, there are some facial features different from the ordinary. Based on these special facial features, we can realize automatic face classification using image processing technology and pattern recognition algorithms.

TS patients' diagnoses mainly rely on peripheral blood lymphocyte chromosome karyotype analysis, and typical clinical manifestations, among them karyotype analysis, take a long time and are expensive. The study found that approximately 90% of TS patients have the main clinical features of short stature and gonadal hypoplasia [6]. For short stature, the best treatment time is before puberty. The majority of patients had slow height growth in childhood. In preschool they significantly lagged behind same sex children of the same age, and after adolescence height did not significantly increase. The average height of the untreated TS patients was 140 cm, which was approximately 20 cm shorter than the average height of normal women. For patients with TS, a reliable method of promoting growth rate is rhGH (recombinant human growth hormone); children can improve their height and even achieve normal height. However, the therapeutic effect of rhGH may be related to the age at onset of treatment, karyotype, duration of treatment, body mass index (BMI), and the dose of rhGH. The patients in their early years have been of significantly short stature; therefore to make patients obtain the biggest benefit, we evaluate in detail the patient's height, bone age and development; once diagnosed as TS, therapy with rhGH can be provided. The younger the age at diagnosis, the better the treatment.

For gonadal dysgenesis, oestrogen therapy can not only promote the development of secondary sexual characteristics, thereby reducing the psychological burden on TS patients but can also promote the absorption and deposition of calcium to effectively increase bone density and reduce the incidence of fractures. Currently, the start time and dose of oestrogen replacement therapy is still controversial; a reasonable time to begin the use of oestrogen will benefit the patient's physical and mental health. Therefore, for patients, early diagnosis is very helpful in providing for a reasonable arrangement of oestrogen treatment time. Because of the abnormal chromosome structure or number in TS patients, there is no cure at present, only symptomatic treatment. The results of this study can help patients with TS achieve early detection, early treatment, regular monitoring and prevention of complications, thereby alleviating the tremendous psychological pressure and long-term financial burden on TS patients.

In this paper, the realization of automatic face classification, mainly using face recognition-related technology, is described. In recent years, with the rapid development of science and technology and the rise of computer artificial intelligence, facial recognition, as an important branch of biometrics and a hot topic of artificial intelligence, is widely used in business, information security, public safety, human-computer interaction and other fields. It is still a bold attempt to apply facial recognition to medical care. The sustainable acquisition and accessibility of face images is one of our advantages. We have established a database of patients and can continually obtain a significant number of facial images for scientific research. In the study of TS patients, the researchers focus more attention on the patients' chromosome karyotype, growth and development, and some other clinical features. Meanwhile, there are very few TS studies regarding patients' facial features that would qualify as guidance for evidence-based recommendations. A few studies refer to some TS patients' facial features as different from ordinary, such as facial asymmetry [5], epicanthus [6–9], melanocytic nevus [9], and ocular distance [10]. Although these features were mentioned in the studies, there was no quantitative comparison between TS patients and healthy subjects regarding these facial features. At the same time, through the analysis and comparison of a large amount of data, we found that the geometric characteristics of the forehead and the nasal bridge of the patient were different from those of normal people.

However, because there is little work reported on TS facial features, we engaged in extensive reading of references and found some studies of facial images for other endocrine diseases. Kosilek et al. [11–15] found that Cushing Syndrome and Acromegaly patients had some facial features different from healthy subjects and used FIDA (Facial Image Diagnostic Aid) to formulate automatic facial image classification methods. They performed a semi-automatic analysis of the images by comparing texture and geometry features within a grid of nodes placed on the facial images and finally achieved a satisfactory initial classification accuracy. Chen et al. [16] found that the visual difference between the faces of chronic fatigue syndrome (CFS) patients and healthy people can be used to diagnose CFS. In improving recognition accuracy, they apply a fusion of hybrid facial features methods. Some of the methods used in this paper are mainly referenced from their research. Lina et al. [18] used automated FDNA (Facial Dysmorphology Novel Analysis) technology to recognize facial images with Cornelia de Lange Syndrome (CdLS) and achieved a satisfactory initial classification accuracy. With reference to the above methods and specific facial features of TS patients, we designed an automatic facial classification system for TS.

Fig. 1 shows the facial images of typical TS patients. In spite of the fact that all participants or guardians have signed a consent form, because the patients are younger, to protect the patient's privacy we have made an eye mask for the image. Because of the different karyotypes and individual differences, different TS patients show different clinical symptoms. In images (a), (b), and (c), all patients show the epicanthus in their eye region. Epicanthus is the name for a skin fold of the upper eyelid, covering the inner corner (medial canthus) of the eye. One of the primary facial features that is often closely associated with the epicanthus is elevation of the nasal bridge. Often, a lower-rooted nasal bridge is more likely to cause epicanthus, and a higher-rooted nasal bridge is less likely to do so, but many TS patients with epicanthus have a higher-rooted nasal bridge, such as in image (b). Furthermore, TS patients also have many abnormal and various sizes of melanocytic nevi. A melanocytic nevus is a type of lesion that contains nevus cells (a type of melanocyte). There are also several facial features concerning distance, such as the distance between the eyes and the width of the forehead. By selecting appropriate features in facial images, it is possible to find a method to recognize TS patients automatically.

The contribution of this paper can be summarized as follows: (1) We propose a computer-aided system for automatic face classification based on facial image analysis, because it improves the objectivity of diagnosis, reduces the burden on well-experienced endocrinologists, enables large-scale screening in under-developed areas and make possible early detection and early treatment of TS patients. We firmly believe that it would be helpful and worthwhile for other medical diagnosis problems to use similar approaches. (2) A set of suitable experimental analysis methods and flows are summed up for facial image automatic classification, where two feature extraction methods, based respectively on the global features and the local features, are investigated. Fusion of multiple facial features to improve the accuracy of recognition alleviates degraded recognition accuracy because of noise, illumination, and non-universality [16,19,20]. Therefore, in this paper we propose a Multiple Facial (MF) features extraction and