International Research Journal of Engineering and Technology (IRJET) e-ISSN: 2395-0056
Volume: 09 Issue: 01 | Jan 2022 www.irjet.net p-ISSN: 2395-0072

A Comprehensive Survey on Down Syndrome Identification using Facial Features

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***Abstract*—**Down's syndrome is a genetically inherited disorder that causes a unique facial appearance, intellectual disability, and delays in development. Early and precise diagnosis of this genetic disorder is important to the patients and their families to bring about extra care and catered health management. The structure of the face is a major indicator in the detection of Down syndrome defects paving the door for computer-aided diagnosis which relies on facial image analysis. This, in turn, significantly simplifies the identification process. Through this paper, we investigate different methods for the identification of Down syndrome by using facial photographs with the help of various algorithms and techniques. This study briefs us about the important techniques that have been proposed so far and the problems that were faced during their implementation.

***Index Terms*—Image processing, Machine learning, Computer vision**

I.INTRODUCTION

Down syndrome is a disorder caused by chromosomal defects in which a child is born with an extra chromosome 21. This extra copy alters the way the infant's brain and body develops, posing physical and mental obstacles for the kid. A somewhat flattened face, eyes shaped like an almond that slant up, a short neck, tiny ears, the arch of the nose tends to be flat, the tongue stays out of the mouth most of the time, little white flecks on the iris of the eye, small feet and hands, and others are the common physical characteristics of Down syndrome.

In the clinic, doctors must distinguish between patients with Down Syndrome and those who are healthy. Prenatal diagnostics can detect Down syndrome during pregnancy while chromosomal analysis can diagnose it after birth. To verify this diagnosis, a chromosome test called Karyotype test will be conducted. However, the identification of non classical representations of Down syndrome is tongue-tied by clinical experts' prior experience. Furthermore, chromosome testing is expensive, takes a lot of time, and is difficult, and many rural or small health institutions lack access to these tools. Hence, the need for utilization of computer-aided systems with health professionals becomes increasingly necessary.

Machine Learning models based on anything as simple as a photograph of the patient can aid in the accurate detection of Down Syndrome. Early discovery leads to a big decrease in Down syndrome-related deaths. Furthermore, it adds an improved quality of life, thanks to appropriate treatment. This research investigates different approaches to create a best automated Down syndrome detection approach based on 2- Dimensional facial images.

II. LITERATURE SURVEY

Qian Zhao et al. [1] proposed a technique that defined 17 anatomical landmarks on the face. The photos were then analysed for geometric properties. Local texture characteristics around each landmark were extracted with the help of Con- tourlet transformation. A local binary pattern was applied to maintain the spatial information for each anatomic landmark. After smoothing the images, a multi-layer Contourlet trans- form was used. More information was obtained by combining local textural and geometrical properties. An SVM classifier was used to differentiate between the two classes. This project works on a small-scale data set and to provide a more thorough validation of this method, it should be applied and tested on a larger scale of data.

Vlad Dima et al. [2] adopted some common face recognition methods like Eigenfaces and Local Binary Patterns to dis- criminate between the two classes. kNN and SVM classifiers along with Radial Basis Function (RBF) and a third degree polynomial kernel have been used for classification. The paper studies various approaches to pull out features from the images which include traditional Local Binary Pattern (LBP), a blend of Local Binary Pattern(LBP) and Discrete Wavelet Transform (DWT), an Eigenface technique based on projection method that uses ATT, and a projection method based on ATT's Local Binary Pattern features. All the above methods perform well in differentiating the two classes. However, in the presence of noisy data, projection methods perform better compared to the combination of LBP and DWT. As a further improvement, large volumes of data should be used to boost the project's potential and use case.

Sayed Mohamad Tabatabaei et al. [3] implemented a quick system for detecting Down syndrome. For experimental purposes, this paper uses a custom data set of 200 photographs for each class. Higher-order directional derivative local binary patterns (DLBPs) is the proposed texture feature extraction approach. The study conducted two experiments. The primary method applied the simplest first-order directional derivative local binary patterns. The second method hired the second- order directional derivative local binary patterns in addition to the first-order directional derivative local binary patterns. The outcomes of the first and second tests were compared. After feature extraction in both situations, an SVM classification was performed to distinguish between the two classes, DS and non-DS. When the outcomes of these two strategies were examined, it was clear that the second approach had a considerable impact on the accuracy.

Qian Zhao et al. [4] proposed a method for detecting Down syndrome using non-standard front-viewed facial images and some common ML algorithms. The approach consists of four parts which are landmark detection, extraction of features, categorization, and assessment. The approach is based on a supervised learning scheme. First, a local model is used to pinpoint the landmarks on the face. Then, using size-variant windows, textural and geometrical features of the face were obtained from these landmarks. Following feature extraction, Down syndrome-specific traits were identified. Finally, the results of the various classifiers like linear SVM, SVM based on radial basis function (RBF) kernel, random forest (RF), k-nearest neighbor (k-NN), were compared and evaluated. Throughout the data set, leave-one-out validation is used. Accuracy, recall, precision, and other parameters were used to assess the performance. Local and blended texture features were shown to have the best performance when employing the SVM classifier with RBF.

Qian Zhao et al. [5] proposed a method based on landmark analysis and various algorithms, this work proposes a straightforward and conventional evaluation method. To classify DS from non-DS using facial pictures, an ICA-based Hierarchical Constrained Local Model (HCLM) was proposed and deployed. Three models were presented for landmark detection which are Principal Component Analysis based CLM, Independent Component Analysis based HCLM, and Independent Component Analysis based CLM. Using Independent Component Analysis vs. Principal Component Analysis with CLM, in addition to HCLM vs. CLM with Independent Component Analysis, resulted in considerable improvements. Specific geometric characteristics and texture features based on local binary patterns were retrieved and selected after landmarks were located using Hierarchical Constrained Local Model with ICA. Throughout the data set, leave-one-subject validation was used. Various classifiers like kNN, SVM, linear discriminant analysis, and random decision forests were used to evaluate the performance.

Jadisha Yarif Ramírez Cornejo et al. [6] proposed a four- stage methodology that includes facial detection, extraction of features, reduction of these features, and finally classification. The methodology is based on a method that employs a cascade function that has been trained on both the classes of images. Relevant characteristics are chosen using a variation of the Adaboost learning method, in which a cascade classifier made up of multiple stages decides whether or not a window contains a face. The discovered face landmarks are used to create a geometric descriptor. The geometric depiction uses thirteen two-dimensional fiducial points on the face, for example, 2 points for eyebrows, 1 point for the tip of the nose, 4 points for the corners of the eyes, and so on. The experiment employs two techniques for feature reduction which are linear discriminant analysis (LDA) and principal component analysis (PCA). The paper studies and compares the results of two classifiers, kNN and SVM. After dimensionality reduction, the classifiers were applied to the feature matrix to generate a comparison between the observed recognition rates. The results obtained by the combination of geometrical and textural features is better compared to the results obtained using solely geometric features. Textural features appear to have taken precedence over geometric features.

Ling Li et al. [7] proposed a technique based on "cascaded framework of voting isolation forests and logistic regression (CVIFLR)". To achieve good accuracy, 3 stages of CVIFLR were implemented. The pre-judgement stage was used for identifying the anomalies, with isolation forests. The identified anomalies here, consisted of few mistaken ones too. Next, model ensemble was done where each isolation forest was trained with a split of negative data in the training set. Hyperparameter tuning and evaluation verified if all samples categorized as "must be negative" were real negative ones. However, the "may be positive" samples were still under suspicion and were taken to the next stage for final evaluation. In final judgement phase, a LR model recognises the true and false positive instances from the samples under suspicion.

Bosheng Qin et al. [8] proposed a method that makes use of images of the face and deep CNN to quantify the problem of binarily distinguishing subjects with Down syndrome from healthy subjects through ungoverned two-dimensional images. The training algorithm proposed consisted of 3 steps: image pre-processing, the general training of network for recognition of face, and the training of network for identification of DS anomaly. A Deep CNN was used to draw out facial features for prediction process. When a raw image was pre-processed and input to the facial recognition network. It calculated the similarity of input images to DS affected and healthy unaffected faces in training dataset. After a general recognition network for the face was trained using DCNN, the final layer of DCNN was improvised to suit the requirements of DS anomaly identification.

Bing Feng et al. [9] proposed and designed a CNN with 9 layers and 2 merging branch CNN models. These 2 models took 2 chromosome single-nucleotide polymorphism maps as inputs. A singular chromosome SNP map was taken as the in- putted image by each branch's model. Later on, the 2 branches of CNN model were converged together to a single model which became another layer. Conventional ML Algorithms like random forest, SVM, and decision tree algorithms were also proposed for the same purpose, with the same genotyping data. Though these models achieved decent accuracy scores, their false negative rates were too high. Furthermore, the CNN model was able to fetch out visual patterns regionally and local motifs distinctively from small regions on the above- mentioned maps.

Nattariya Vorravanpreecha et al. [10] proposed a method based on the tool Face2Gene which compares features of a patient's face to a reserved database for possible matches with this genetic syndrome. It used DCNN that was able to compare 300 different syndromic models to 2D photographs. In this model, the analysis of the 2D image started with the detection of the face and getting rid of the background. Then facial regions were identified by cropping the inputted image and fed to the DCNN. The vectors that were the output of all regional DCNNs were then accumulated to receive a list of rankings of around 30 possible syndrome matches, in an order going from most to least likely. When DS appeared inside the top 10 syndrome-matches, it was distinguished as cut-off point for results to be taken as positive in the study. However, the application used in this study failed to recognize a majority of profile photographs and was concluded to be successful only for frontal facial photos.

Kurt Burçin et al. [11] have addressed the implementation of the Local Binary Pattern approach in this paper. The local Binary pattern approach is a simple yet effective texture opera- tor which can recognize minute differences. LBP runs in a 3*3 block size, in which the center pixel is used as a threshold for neighboring pixels. Binary code is generated for each pixel. One (1) is set for values higher or equal to the threshold, and for values lower than the threshold, zero (0) is set. The resultant binary value is converted into a decimal value, the new image represents the characteristics of the original image more effectively. The improved system is developed by using two classification techniques i.e., Euclidean Distance and Changed Manhattan distance methods which effectively classifies the DS and non-DS categories. As a part of future enhancement, the framework can be enhanced for the detection of different sets of medical problems.

Subhiksha Ramanathan et al. [12] adopted a method that focused on pulling out suggestions from the first trimester (prototypic) test reports of patients. Features are extracted with the help of Optical Character Recognition which converts the pdf files to machine-readable format. The reports are later put under different clustering techniques like Hierarchical cluster- ing, K-medoids, DBSCAN (Density-based spatial clustering of applications with noise) and K-means. It is inferred that K means gives better results with an accuracy of 86 percent. While the follow-ups with supervised techniques applied for the un-normalized data to obtain posterior probability indicates that, Naive Bayes gives more accuracy compared to others. As a part of future enhancement, the real data can comprise a data set of more patients fake reports can be generated by ADASYN process and pre-processing model can be made more adaptable to accept the input reports in any given structure according to the study of this paper.

Pushkar Shukla et al. [13] focused on developing a model for recognizing development disorders-which refers to chronic disabilities i.e., impairment of certain parts of the brain or parts of the body or both. The framework deploys Deep Convolutional Neural Network for extraction of features and support vector method as a classification technique. This study demonstrated a high accuracy score after five cross-fold validation. The model can be further improved by including audio-video visuals as data sets.

Olalekan Agbolade et al. [14] displayed a study of facial recognition within the field of mongolism, which consisted of reliable feature extraction, facial point detection, and clas- sification. From their literature review, most of the works used data on a small scale to train images, which is not advisable in deep learning. It was observed that the geometric representation achieved commendable performance for DS recognition within the feature extraction process, also the SVM or SVM based classifiers depicted a better recognition accuracy compared to classifiers that used neural networks. The approach that involved neural networks offered many advantages such as a combined approach for classification and feature extraction with flexible approaches for finding moderate solutions which are not linear.

Qian Zhao et al. [15] proposed a model in which features of Geometric and textural nature are extracted using anatomical landmarks to detect the structure of the face. To locate the landmarks independent component analysis (ICA) is used which is a hierarchical constrained local model. To select the dominant ICA, a data-driven ordering is done. The Accuracy increases due to the hierarchical structure by fitting different models into separate groups. Various classifiers are tested to differentiate between Down Syndrome and a Normal person. This model was also evaluated against different genetic diseases and yielded high performance.

Cerrolaza Juan et al. [16] proposed a general idea about detecting genetic Syndromes is set up using facial images by the combination of both textures and facial features. They have used the 2 Dimensional extension of Linear Discriminant Analysis(LDA) to extract landmark-specific local Binary patterns. It further uses Image filters and soft Neighborhood Weighting matrices that further increase the accuracy of the predictions. It achieved an accuracy of 0.95 on a data set of 145 cases.

Safak Saraydemir et al. [17] used the famous Gabor Wavelet Transform(GTW) for feature extraction. Later the dimension- ality of the features is decreased using Principal component analysis(PCA). The LDA or Linear Discriminant Analysis is used to derive a new dimension that has extremely important information. SVM and kNN are used to implement classifica- tion. What sets this paper apart from the rest is that feature selection is implemented before Principal component Analysis due to the correlation between components of the feature vectors. It can be enhanced by using the linear kernel for SVM and Euclidean distance metric for kNN.

Jin Bo et al. [18] applied Deep learning models to 2D facial images to identify various diseases. They have made use of Deep Transfer learning(DTL) models which gave them an accuracy of 90 percent by using Convolutional Neural network(CNN) as a feature extractor on a data set of 350 facial images thereby making it better than its predecessors using conventional machine learning algorithms.

III. CONCLUSION

The many methodologies and procedures utilised in the detection of Down syndrome using face photographs have been discussed through this research. To obtain increased accuracy, several algorithms and augmentation strategies have been evaluated against various amounts of data. The majority of surveys yielded good results, which, with further refinement, could be utilised in the medical industry to differentiate Down Syndrome from non-Down Syndrome merely based on facial photos. It also gives us an overview of the key strategies and issues encountered during the implementation of the algorithms.

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