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Nidāna - A System For Detection Of Genetic Disorders With Prominent Facial Features Using AI

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Abstract

Artificial Intelligence along with facial analysis techniques have lately been at par with the capabilities of medical experts in the identification of various genetic syndromes. So far, these techniques could identify some of the diseases by extracting the facial features of an individual, restricting their role in the medical field where a lot of diagnoses should be considered. We've developed a portal that would help in early detection of genetic disorders or at least reach a strong hypothesis if not an exact diagnosis using AI, computer vision and CNN, that gauges similarities of genetic syndromes on the basis of unconstrained 2D facial images. The results predicted by the portal can be used in cohesion with other medical diagnoses, behavioural and growth analysis for precision in the diagnosis of genetic syndromes.
Keywords: *Artificial intelligence, CNN, Genetic Syndromes, Transfer Learning*

1. Introduction

Diseases induced by concealed genetic disorders influence the majority of individuals throughout their life. Below, we explicitly focus on genetic syndromes with prominent facial features. These disorders affect nearly 8% of the population. From the 60 crore people affected by rare genetic diseases, India has around 10% of them. All these alarming figures and we still don't see proper care given to them. Many affected people exhibit signs that impair their well-being and standard of their life. Primal diagnosis is vital to avoid sudden emergence of possible health disorders, such as serious congenital heart defects, respiratory problems, slow and stunted development, amongst various other issues. Though most of them cannot be cured completely, we can surely help to improve their lives with our little gestures. Early diagnosis is beneficial for the patients because peculiar avoidance and monitoring programs help avoid the incidence of potential health issues, like respiratory problems, and developmental delays, among others.

Most of these syndromes seem to have distinct facial phenotypes [1], which provides comprehensive information to genetic specialists, which helps them in diagnosing a particular genetic disease [2]. Genetic researchers have either sought a solid diagnosis for more specific or unusual syndromes, or at most a clear hypothesis, based on the facial features of the individual. In most cases, though, patients visit a genetic expert years later, after initial symptoms emerged. Oftentimes, due to the subtle nature of these syndromes and a wide range of potential disorders, attaining the appropriate diagnosis includes prolonged and valuable handiwork and analysis which may take several ages or even a few decades (the diagnostic odyssey).

Identification of unclassical presentations of typical syndromes, or occasional disorders, may be limited by prior experience of the particular genetic specialist. Computer systems are used as an aid or reference for medical specialists thereby increasing extensively. Nidāna guarantees to provide expert knowledge and awareness to healthcare professionals.

Facial appearance is a crucial aspect of diagnosing the syndrome. Using computer vision for facial recognition has tremendous potential to this point. Over the past few years, computer vision research has dealt with the issue of facial recognition.

The identification of a particular genetic syndrome from facial phenotypes is analogous to typical facial identification. However, in reality, the creation of the Syndrome Identification System is difficult due to many reasons, emphasizing limited data, the subtlety of facial features and ethnic diversity.

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A key element for the successful implementation of such systems has been proven to be the size of the datasets. This allows for the training of stable, accurate models with CNN algorithms. In the case of genetic syndromes, due to the prevalence of those genetic disorders, it's not possible to compile such extensive datasets. Potential populations of size and stigma are much smaller, as is the wide variation in the general population of patients per condition. Another difficulty in some of the syndromes is the subtlety of facial features, combined with the fact that some of the syndromes do not have clinically defined, distinctive facial phenotype.

2. Problem Definition

New computer-aided identification analyses of the condition do not resolve real-world issues by classifying hundreds of syndromes from unconstrained images. We address issues such as classifying between average and disabled people or diagnosing only one illness, usually using restricted pictures taken.

There are three main issues we wish to address here.

Problem 1: Other population vs. Single syndrome subjects - a binary classification problem of classifying the given subject with a peculiar syndrome from unaffected subjects (subjects with unaltered genetic structure) or individuals with other syndromes [4][6].

Problem 2: Normal subjects vs. Syndromic Subjects - a binary classification problem of classifying the given subject with any syndrome from normal (subjects with unaltered genetic structure) individuals [4].

Problem 3: Classifying multiple syndromes - a multi-class classification issue of identifying the appropriate syndrome from various considered potential syndromes.

Our application is mostly focused on addressing issues in 3rd problem definition but also shows potential on the first problem [5].

3. Proposed System

The proposed system is designed to develop a portal where the medical experts and normal individuals can be registered as users, although the views and the services provided to both of them would be different. As a medical expert, the user can use our system for the initial diagnosis of an individual to identify whether the individual is affected by a genetic disorder or to reach a certain hypothesis on which rare genetic disorder the individual has. The medical expert would have a record of each affected individual in the form of cases that will be stored in a centralized database. As a normal individual or a medical expert, the user would have access to upload a facial image which would be then analyzed by our system along with that, the user would have to answer certain questions which would be used for analyzing their behaviour and growth patterns, all these combined inputs would then be processed by the system and a tentative result would be shown to the user in the form of a graph or a confidence value of which genetic syndrome the individual might have. If the user is a normal individual, depending on the summarized data they will have an option to make an appointment to consult a doctor/medical expert for which the user has to give consent that he/she allows his details to be accessed by the medical expert

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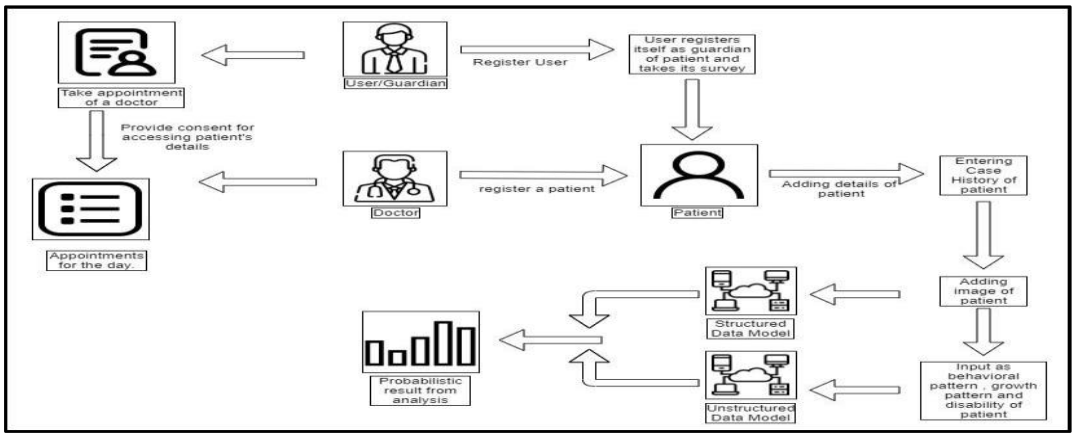

Figure 1: Block Diagram for Nidāna

3.1 Methodology

There are two core modules of our system which are used to analyze the data given by the user and generate the summarized results based on that. The first module is basically a CNN model based on transfer learning [7] which takes frontal facial images of an individual as an input. Now the image given by the user as input cannot be directly passed to the model as it would be a real-world unconstrained two-dimensional image, hence the image is passed to a preprocessing phase where the initial stage is to detect the face of the patient from the input image and then crop the region of interest which only covers the frontal facial region of the individual and the final step in the preprocessing phase is to scale each facial cropped region to a fixed size of 224 × 224 to use it as an input to our model.

We have now used the ResNet-50 architecture for the CNN platform, which is based on a residual learning network and is easier to refine and thus allows for the training of deeper networks resulting in an overall improvement in network capacity and performance.

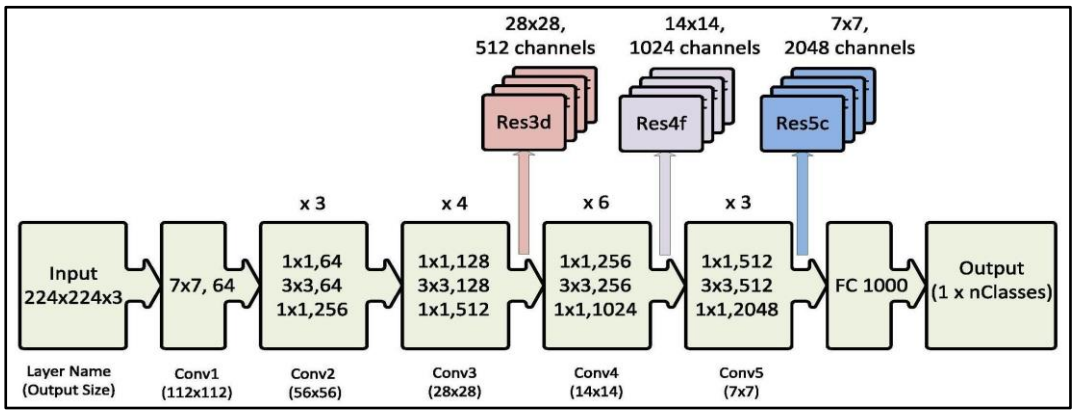

Figure 2: ResNet50 Architecture

The model is trained using Keras [8] VGGface [10] with TensorFlow [9] as the backend. The base model uses 4 Fully Connected layers on top of VGGFace which is a predefined classification of the algorithm by Keras. The Transfer Learning approach has the ability to reuse already learned features again. SGD optimizer with a small learning rate of 0.0001, the momentum of 0.9 and a softmax activation function was used to optimize the training process of the model. The callback was defined in Keras for reducing the learning rate by 0.1 whenever the validation loss stopped improving. The batch size was set at 32 in each case. We try to minimize cross-entropy loss as a basis of good classification. The classifier used was the built-in Keras (v2) library for deep learning. A total of 200 epochs was run and the accuracy was established around 100 epochs. We calculated prediction confidence values for each image belonging to the 8 classes. The 8 classes of genetic syndromes that we will be classifying are **Angelman, Apert, CDL, Down, FragileX, Progeria, Treacher Collins, Williams.**

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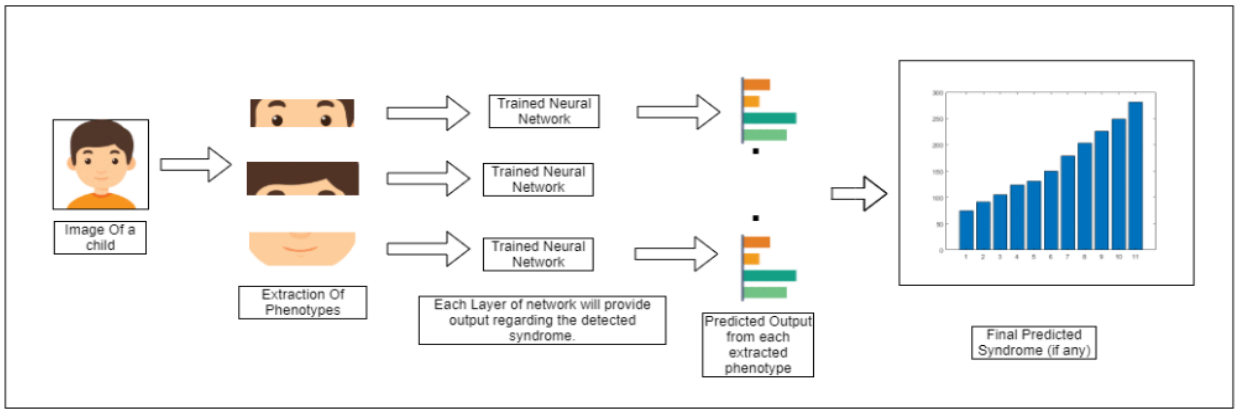

Figure 3: Methodolog

The Second module is based on the unstructured data obtained from users in the form of answers to some particular questions based on the difficulties faced by the individual, their growth and behavioural patterns. Based on this data provided by the user our module would generate probability scores on the basis of similarity with significant characteristics of a particular genetic syndrome.

The combined results of both the modules are then represented in a summarized form of confidence values and graphs to the users. These results would assist in the process of early diagnosis of a genetic syndrome or providing support in achieving a firm hypothesis.

3.2 Implementation

The proposed model uses the SITW_eLIFE dataset [3] (excluding the Gorlin Collection) provided by Cristoffer Nellaker from Oxford University. We used 80% of the data for training and 20% for testing. The below images are some samples from the dataset that we've used for training our system.

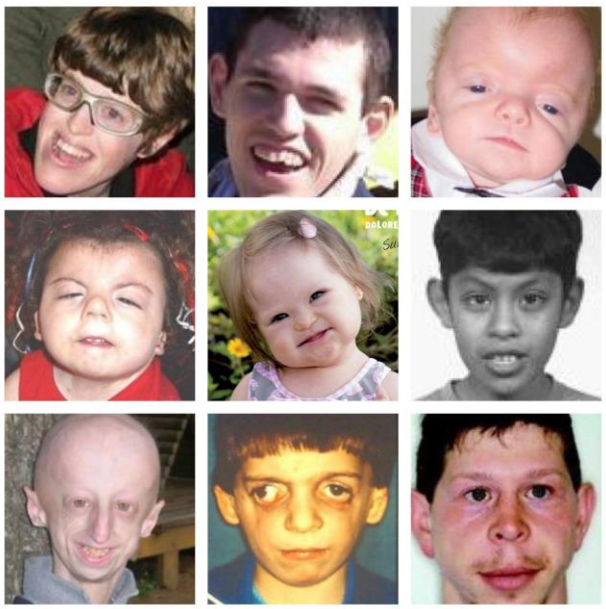

Figure 4. Sample Images from the SITW_eLIFE dataset

Apart from the image dataset, we've also used some unstructured data from the users in the form of answers to a simple questionnaire in which the users are asked to select the symptoms the think the individual displays. These symptoms are classified into three categories such as Physical disabilities, Behavioral patterns, Growth patterns. Based on this data provided by the users one of the modules in

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our system generates confidence values based on the similarity of symptoms with significant characteristics of a particular syndrome.

4. Results and Discussion

As there is no adequate comparative criterion it is unable to compare the above procedures with respect to the accuracy, we strive to develop and deliver the finest system which ensures maximum accuracy and performance. Although performance can be assessed through cross-validation and the generalizability of the models is still unclear. Therefore, the amount of syndromes assisted in a program is important for medical use.

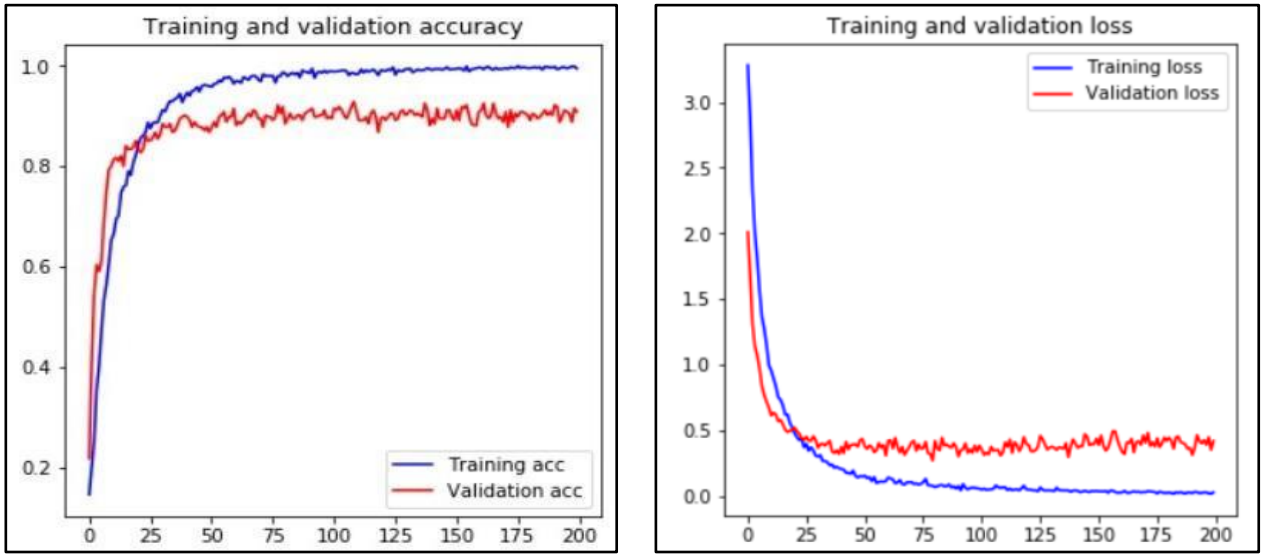

Fig 5: Training and validation accuracy and loss graphs

Now the classification of the images by the system in validation phase can be represented in the form of a confusion matrix in which true labels are represented along the rows and the predicted labels are represented along with the columns.

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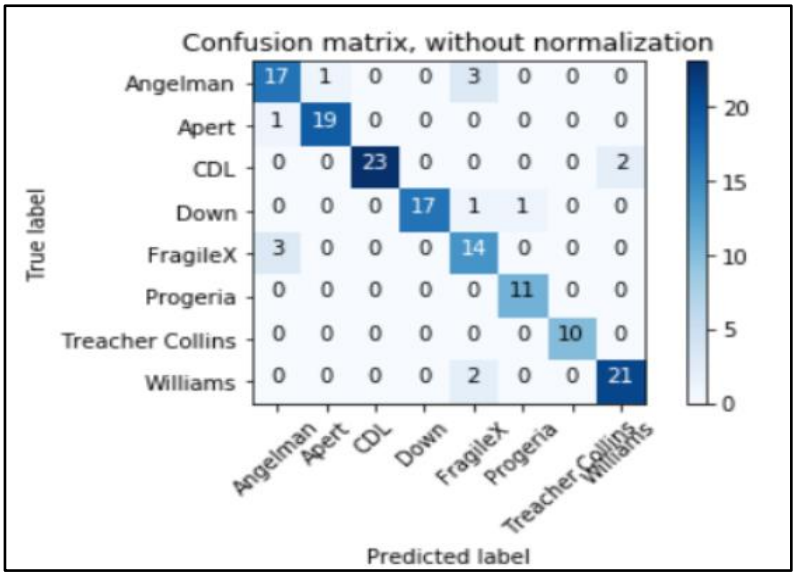

Fig 6: Confusion matrix of results predicted by the system

5. Conclusion and Future Scope

We believe this system will, therefore have a common use case for medical use, taking the prevalence and diagnostic challenges of various syndromes into account. Patients with such Artificial Intelligence (AI) technologies will improve the manner in which rare genetic syndromes and other genetically-caused diseases are researched and explored. The possibility to explain phénotype in a standard way opens the way to the emerging field of precision medicine.

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