# Implementing image processing for detecting people with down syndrome

# Abstract

Down syndrome is a genetic disorder that affects 1 in every 1000 babies born worldwide [1]. The cases of down syndrome have been on the rise steadily for the last couple of years. This paper proposes a way to identify people suffering from down syndrome using their images by implementing basic techniques of Image Processing. For this purpose, we have used convolution neural network to identify if an individual is suffering from down syndrome.

# Introduction

Down syndrome (DS) is a genetic disorder caused by an extra copy of chromosome present in the cells of the concerned person. The problem being addressed here is to identify people affected with down syndrome, so that they can be provided special care and help.

The cases of Down syndrome have been steadily on the rise for some years. According to data available one in every 691 babies in USA is born with Down syndrome, which is approximately 6000 infants per year [2]. Currently there are about 40,000 people with down syndrome living in USA [2]. Therefore, there must be some efficient way to recognize people affected with Down syndrome, so that they may be given special attention.

With such large number of people affected with Down syndrome, it is very hard for the concerned authorities and organizations to keep track of all those people suffering from this condition. The earlier approaches fail because they were not designed to keep track of such people that work in different parts of the world at different times due to increasing globalization.

Earlier the life expectancy of people with Down syndrome was around 20 years [1]. Therefore, these people were not so involved in mainstream life. As a result, there was not much requirement to address this problem. But with the increasing medical facilities and new discoveries in the field of medicines, the life expectancy of people with Down syndrome has dramatically increased to 60 years in the last decade [1]. This increase in life expectancy has led to increase in job opportunities for such people. With the increasing globalization, these people are working in different parts of the world. Therefore, it becomes necessary to employ some measures to identify such people and provide them with the best facilities available.

The approach involves using basic techniques of Image Processing to identify people with Down syndrome using their images, so that they can be attended to. The benefit of such models is

- It can aid in the recognition of people with Down Syndrome in common places like airports, tourist attractions, restaurants, etc.
- It can be used for the initial diagnosis of people suffering from Down syndrome in the areas where specialists may not be available.

This model is based on the Convolution Neural Network (CNN) [10] based analysis of the image of people to identify people suffering with down syndrome.

Most of the experiments conducted earlier consisted of a relatively small data set. This model contributes to the development based on the detection of Down syndrome using deep learning algorithm [10]. For this purpose, this makes use of the Convolution Neural Network as its base algorithm. Data set consisting of the images of normal people as well as images of people suffering from Down syndrome has been used for training as well as testing purposes for more accurate results.

The section 2.1 describes about the Down syndrome, mentioning its characteristics and effects. Section 2.2 mentions the related work earlier in this field. Section 3 describes the system architecture on which the experiment was conducted. Section 4 describes about the experimental setup including its data set and the steps for the experiment. Section 5 describes the results achieved from the experiment.

Facial dysmorphism is a common feature in subjects with Down syndrome. In a study conducted by [], it was observed that subjects with Down syndrome have significant distortion near the eye and mouth region. Taking the proposed analogy into consideration, the proposed model builds a model to extract those regions. Deep representations are then extracted from these regions and are used for building the model.

The existing work on Down syndrome have conducted this experiment on a relatively smaller data-set. The proposed model takes into consideration a data-set of 853 subjects with down syndrome.

The remaining part of the paper has been organized as follows

Section two describes the various causes, symptoms and attributes of humans with down syndrome. A discussion of the existing work on recognizing humans with Down syndrome has been carried out in Section 3. The proposed architecture has been described in Section 4. The various experiments that were conducted have been presented and discussed in Section 5.

# 2. Literature Survey

A description and detailed study of Down syndrome is given below:

# 2.1. Down Syndrome

Down Syndrome is a chromosomal disorder. It is caused due to the presence of extra chromosome 21 in the cells of the affected person. The extra copy can be partial or full. The person with Down syndrome has forty-seven chromosomes instead of the normal forty-six chromosomes in a healthy individual. When a baby develops it receives twenty-three pairs of chromosomes (half from father and half from mother). When the cells divide these pairs of chromosomes are equally partitioned. But in some people chromosome 21 does not get partitioned properly. This gives rise to abnormal chromosome numbers in the individual resulting in Down Syndrome. Worldwide 1 in every 1000 babies is born with Down syndrome [2]. People with Down syndrome develop various physical and mental characteristics which include:

#### **Physical Characteristics**

- Flat facial features
- Small head and ears
- Short neck
- Abnormally large tongue
- Eyes that slant upward
- Poor muscle tone
- Short height

#### **Mental Characteristics**

- Short attention span
- Impulsive behavior
- Slow learning
- Delayed language and speech development

Down syndrome is peculiarized by cognitive disabilities i.e. intellectual and development delays which includes learning disabilities and speech impairment. This is because Down syndrome affects hippocampus region of the brain that is responsible for memory and learning processes.

People suffering from Down syndrome are also at a greater risk of suffering from following health conditions

- Leukemia
- Obesity
- Chronic constipation
- Sleep apnea (Interrupted breathing during sleep)
- Poor vision
- Cataracts
- Strabismus
- Anemia
- Congenital heart defects
- Hearing loss
- Thyroid diseases

There are three different types of chromosomal patterns resulting into Down syndrome.

#### Trisomy 21

Trisomy 21 is a condition where a person has forty-seven chromosomes instead of normal forty-six i.e. (more than twenty-three sets of chromosomes), which is caused by a faulty cell division. Chromosome 21 is one amongst the given twenty-three pairs of chromosomes and is also the smallest human autosome (not a sex chromosome). A normal person has two copies of this chromosome but a person suffering from down syndrome has three copies of chromosome 21, because at the time of fertilization the 21<sup>st</sup> chromosomes of the parent are unable to separate, thus forming a group containing three chromosomes. The extra chromosome is simulated within each cell present in the body through cell division. Trisomy 21 is the most common amongst people suffering from Down syndrome. It accounts for 95% of all cases.

#### Mosaicism

Mosaicism is the condition in which the individual with down syndrome has two different types of cells. The first with 46 chromosomes (twenty-three sets) whereas the second ones are with forty-seven chromosomes which contains an extra copy of 21<sup>st</sup> chromosome. This is the rarest of all three as It constitutes 1% of all Down syndrome cases.

#### Translocation

It constitutes 3-4% of the total cases of down syndrome. This condition arises when the part of chromosome 21<sup>st</sup> pair merges with the chromosome of 14<sup>th</sup> pair. While the number of chromosomes in the cell remains 46, the presence of extra part of chromosome 21 causes Down syndrome.

#### 2.2 Related Work

A petite amount of work has been done to detect down syndrome by executing the basic concepts of image processing to diagnose the population suffering from the disorder. The technique of Gabor Wavelets for representation extraction was given by Safak et al. which was followed by the Principle component analysis and LDA and then obtaining results by applying SVM (support vector machine) and K-nearest neighbour on 15 healthy individuals and 15 individuals with down syndrome. The results were quite promising with a total of 97.3% and 96% respectively. A model made with the help LBP and template matching for down syndrome detection from cropped images was then proposed by Brucin and Vasif. The concept of cropped images helped us perceive the important features of human face with the help of local binary pattern. Model given by Shukla et al. that used CNN, GIST and LBP with SVM

(Support Vector Machine) classifier to obtain results simultaneously. This system was then used to test the results for six different disorders and had some auspicious outcomes.

# **3.System Architecture**

The complete research was conducted on a machine having the following architecture:

i5-72000U

clock speed-2.50GHz

8GB DDR4 RAM

4GB Nvidia 940MX DDR5 GPU

For this research both the global as well as the local facial features have been considered to account for the variety of features in people suffering from Down syndrome. This model is based on the [3] that conducted experiments to study the eye and ear patterns of people suffering from down syndrome and to find the differences between normal people and people suffering from Down syndrome. In the proposed model, all the images were cropped to represent three different regions of the faces of test subjects. All three cropped regions along with the full image were then provided as input for the CNN for extracting essential features, which were then combined to form single feature vector. The entire framework can be divide into three parts: preprocessing, extraction of facial features and classification.

#### 3.1 Preprocessing

For detecting cropping faces from the original image, an advanced face detector based on mixture of trees was used. Input was a single image and the output of the detector was the location of 68 different facial key points. These key points were then used for cropping the image.

## 3.2 Extraction of facial features using Convolution Neural Network

The recent researches [4,5] have proved the superiority of CNN over other methods like HOG [6] and LBP [7] for facial representation. However, CNN requires a lot of data for its implementation. Therefore, for our purpose we used pre-trained models for efficient training of algorithm as our data set was relatively small. It is based on the pre-trained Alex-Net model [8]. All the cropped images were resized to 227x227 as it is the requirement of Alex-Net model.

The network is calibrated on the LFW database [9]. This database consists of 5749 people providing 13,233 images out of which 1680 people had more than one image. The calibration does not cause any structural change in Alex-Net model.

The initial learning rate for the final layer was equal to 0.001 and it was 0.0001 for the remaining layers. The original network was trained for 10000 iterations with a dropout of 0.5 and the momentum of 0.7. In this model, we extracted different features using different convolution neural networks and finally all the features were concatenated together into a single feature vector that was used to train and test the model.

## 3.3 Support Vector Machines for classification

Support Vector Machines are an ideal choice for classification. Support Vector Machines work by identifying an optimal hyperplane that divides the data into different classes. Since we had only two classes we used SVM as a binary classifier. The feature vector obtained from CNN were then classified using SVM classifier.

# 4. Experimental Setup

A description of various experimental scenarios is described below.

## 4.1 Data-Set description

The data was collected with the consent of various organizations working for the development of people with special needs in Uttarakhand region of India. Apart from the various organizations the images were also connected from various sources on internet. The data is annotated based on gender as well for detailed study.

## 4.2 Experimental Scenarios

To obtain more accurate result, the experiment was conducted in two different scenarios. The experiment was conducted on a machine with i5 7<sup>th</sup> generation processor having NVIDIA 940 MX GPU.

**1)Scenario 1:** In this scenario, the model was trained and tested on 800 photos of different people suffering from Down syndrome as well as on 800 photos of normal people. The purpose of this experiment was to differentiate people suffering with down syndrome from normal healthy people.

**2)Scenario 2:** In this scenario, the entire data was bifurcated based on gender and then the model was trained and tested to differentiate normal people from those suffering with Down syndrome. The results of both the gender were recorded separately.

## **5.Experimental Results**

The results of the experiments conducted under the two scenarios are described below.

## 5.1 Experimental Scenario 1

A dataset comprising of 800 pictures of both normal people as well as of those suffering from Down syndrome was used for training the model and an equal number of image of both the categories were used for testing the model. The training and testing data were divided in the ratio of 1:1. The model gave

an accuracy of 98.47 in correctly identifying the images of people with Down syndrome. Table 3 gives the comparison of the proposed method with other methods available.

HOG with SVM classifier	60.42
LBP with SVM classifier	91.28
GIST with SVM classifier	92.86
GIST with KNN classifier	87.19
GIST with Random forest classifier	94.02
CNN with SVM classifier	98.47
CNN with KNN classifier	70.62
CNN with Random forest classifier	95.02

## 5.2 Experimental Scenario 2

A dataset comprising of \*\*\*\* pictures of females consisting of normal as well as of those suffering from Down syndrome were used for training as well as testing the model. Similarly, \*\*\* pictures of males consisting of both normal as well as ones suffering from Down syndrome were used for testing and training the model. The results obtained from both the cases are tabulated below.

Table for male dataset:

HOG with SVM classifier	78.14
LBP with SVM classifier	92.03
GIST with SVM classifier	92.23

GIST with KNN classifier	84.73
GIST with Random forest classifier	94.06
CNN with SVM Classifier	96.95
CNN with KNN classifier	84.75
CNN with Random forest classifier	95.39

Table for female dataset:

HOG with SVM classifier	66.47
BP with SVM classifier	91.37
GIST with SVM classifier	90.78
GIST with KNN classifier	86.86
GIST with Random forest classifier	95.10
CNN with SVM Classifier	96.15
CNN with KNN classifier	85.32
CNN with Random forest classifier	96.33

# 6. Analysis

An analysis of results in different scenarios is presented below.

## 6.1 Scenario 1

The proposed model achieved an accuracy of 98.47, better than all other available models for calculation of image features and their classification.

## 6.2 Scenario 2

The model obtained an accuracy of \*\*\*\* for the dataset consisting of images of only males, while it obtained an accuracy of \*\*\*\* for the dataset consisting of images of only females.

# 7. Conclusion

The proposed framework uses the convolution neural network for both local and global feature extraction. Comparison was made with other proposed models of image feature extraction, but this model outperformed all the other models. The experiment was conducted in two parts : one was with all the images ,second part was conducted after dividing the dataset on the basis of gender

The results clearly signify the importance and future of deep learning frameworks in the field of detecting Down syndrome. Such models can be used quite cost effectively in our daily lives to identify people with suffering with down syndrome and improving their lives.

## References

[1]- www.who.int/genomics/public/geneticdiseases/en/index1.html

#### [2]- <u>www.ndss.org</u>

[3]- M. M. Cohen and R. A. Winer. Dental and facial characteristics in downs syndrome (mongolism). J Dent Res, 44(1):197–208, 1965.

 [4] A. Sharif Razavian, H. Azizpour, J. Sullivan, and S. Carlsson.
Cnn features off-the-shelf: an astounding baseline for recognition. In Proceedings of the IEEE Conference on Computer Vision and Pattern RecognitionWorkshops, pages 806– 813, 2014.

[5] W. Ouyang and X. Wang. Joint deep learning for pedestrian detection. In Proceedings of the IEEE International Conference on Computer Vision, pages 2056–2063, 2013.

[6] N. Dalal and B. Triggs. Histograms of oriented gradients for human detection. In 2005 IEEE Computer Society Conference on Computer Vision and Pattern Recognition (CVPR'05), volume 1, pages 886–893. IEEE, 2005.

[7] T. Ojala, M. Pietikainen, and T. Maenpaa. Multiresolution gray-scale and rotation invariant texture classification with local binary patterns. IEEE Transactions on pattern analysis and machine intelligence, 24(7):971–987, 2002.

[8] A. Krizhevsky, I. Sutskever, and G. E. Hinton. Imagenet classification with deep convolutional neural networks. In Advances in neural information processing systems, pages 1097–1105, 2012.

[9] G. B. Huang, M. Ramesh, T. Berg, and E. Learned-Miller. Labeled faces in the wild: A database for studying face recognition in unconstrained environments. Technical report,

Technical Report 07-49, University of Massachusetts, Amherst, 2007.

[10] A. Vedaldi and K. Lenc. Matconvnet: Convolutional neural networks for matlab. In Proceedings of the 23rd ACM international conference on Multimedia, pages 689–692. ACM, 2015.