**ISSN No. 2456-0472**
**The Engineering Journal of Application & Scopes, Volume 3, Issue 1, May 2018**

# DETECTION OF DOWN SYNDROME FROM FACIAL IMAGES USING MACHINE LEARNING

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**Abstract— Down syndrome is a genetic disorder disease and it is also known as 'Trisomy 21', caused by the presence of all or parts of a third copy of chromosome 21. It is typically associated with physical growth delays, characteristic facial features and mild to moderate intellectual disability". Down syndrome can be identified during pregnancy by prenatal screening followed by diagnostic testing or after birth by direct observation and genetic testing. Down syndrome has a private facial view, thus it can be recognized by using facial features. This project is based on Image Processing and Machine Learning. In this we have used some sample images and applied some image processing techniques to detect the presence of Down syndrome through MATLAB. We have calculated the accuracy of all the techniques we have used and chosen the technique having maximum accuracy which is GIST descriptor. This is a very challenging problem as the similarity between the faces of people with Down syndrome and not Down syndrome people is not very considerable. Therefore, we used the GIST approach for feature extraction which is a very effective feature descriptor. In this way, we have developed an efficient system to recognize Down syndrome.**

**Keywords— Down Syndrome Detection; Histogram Of Oriented Gradients; Local Binary Pattern; GIST; Feature Extraction; Classification.**

## I. INTRODUCTION

Down syndrome is a genetic and rare disease and its symptoms and effects are not common and well known among people. So, our main target is to make a system that can automatically detect Down syndrome in a person. We have made a software that detects the presence of Down syndrome disease in people from their facial images. Individual with Down syndrome tends to be below average skill and time spent in activities and motivational factors likely play a role. People of Down syndrome tend to lead sedentary lifestyles [1] and have a high incidence of obesity and being over-weight. Despite the common characteristics, it is important to remember that people with Down's syndrome more closely resemble their parents and siblings in appearance, than other people with Down's syndrome [2]. This means, they are also very similar to people who have not Down syndrome, which makes problem complex.

In the study we tried to recognize Down syndrome from images of faces. We searched the literature but there are only a few studies about this subject. Computer-based recognition of dysmorphic faces was implemented by Hartman, Dagmar, Rolf, Christoph, and Bernhard (2003) that used bunch graph-matching algorithm for describing specific facial pattern. We studied face samples and have tried three different techniques combined with three different classifiers and selected GIST descriptor, which is an effective descriptor for extracting facial features along with linear Support Vector Machine classifier.

a)  b) 

**Fig. 1 a) Down, b) Not down syndrome face samples**

## II. LITERATURE REVIEW

A study of the various developmental disorders that are considered for the experiment along with a description of the previous computer vision approaches for detecting these Disabilities have been presented in this section

Developmental disorders
A brief description of the developmental disorders that were considered for the experiments, along with their causes and symptoms has been enlisted below.
Cerebral Palsy (CP)
Cerebral Palsy can be defined as a developmental disorder affecting the movement and posture of a human being the major causes may include head injuries before or after birth, head trauma caused due to infection during pregnancy, oxygen deprivation or complications due to premature delivery [3]**.** The common symptoms include difficulty in common movements such as swallowing, speech disorders. Cerebral palsy leads to stiff and weak muscles resulting in lower coordination during muscle movement[4].
Autism Spectrum Disorder (ASD)
Autism Spectrum Disorder is a neurological condition that includes a wide variety of disorders. Major causes of ASD include genetic predisposition and environmental triggers such as premature exposure to alcohol or a certain medication in the womb[5]. The main

characteristics include impairment of social and communication skills, repetitive behavior and restricted interests, lack of empathy, poor coordination and in severe cases may also lead to a speech disorder [6]. In many cases subjects with ASD have an aberrancy in the brain size and have poor boundaries between the temporal and frontal lobes.

Down syndrome (DS)

Down syndrome affects a specific region in the brain called the hippocampus which is essential for memory and learning. The syndrome is caused due to an abnormal cell di vision resulting into extra genetic material from the chromosome21.Common symptoms includes speech loss, immune deficiency, thyroid disease, flaccid muscles, hearing loss[7]. It does cause a different facial appearance encompassing lazy eye or spots, abnormally large tongue. Also it is characterized by intellectual and developmental delays which includes learning disability and speech impairment in the child. Annually about 6,000 children are born with Down syndrome in the United States[8].

Fetal Alcohol Spectrum Syndrome (FASD)

The main characteristics of FASD include hyper activity, lack of focus, heart problems, kidney defects, behavioral problems along with low IQ and poor coordination. These are a group of conditions that are caused due to the exposure of the child to alcohol in the mother's womb[9]. The subjects have shown abnormal facial representations in terms of facial width, small eye openings and thin upper lips [10].

Intellectual disabilities (ID)

This disability is distinctively identified by restrained behavior in intellectual and adaptive functioning. The major causes include abnormal genetic conditions, problems faced during pregnancy, complication occurring while birth and exposure to toxicity. The major symptoms include facing difficulty while thinking and understanding. Also certain life skills such as some conceptual, social and practical adeptness may bear impact[11].Behavioral symptoms includes restlessness, hyperactivity or impulsivity. The developments in these patients are slower than normal patients and these subjects may face difficulties while communicating, grasping and applying mathematical and verbal skills.

Progeria (PG)

This is an extremely rare progressive genetic disorder that leads to rapid aging in children. It is caused by the genetic mutation of the LMNA gene slow growth, hair loss are some of the symptoms that can be seen during the early years of a child. People may suffer from skin rashes, dwarfism, enlarged head, pains in the chest, and deformity of bones. This disorder can't be cured however medication can delay progression.

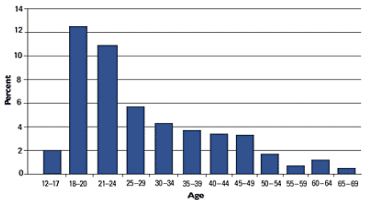


**Fig. 2.Histogram of percentage of Down syndrome affected people VS their age**

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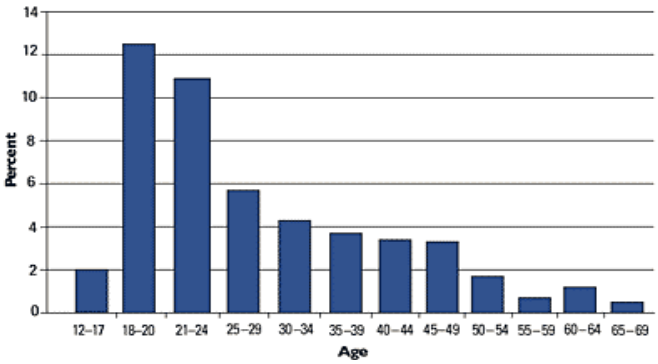


**Fig. 3. Histogram of percentage of Down syndrome affected people VS their age**

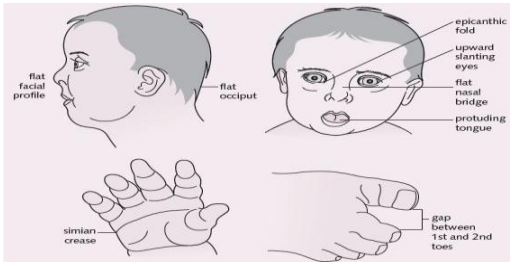


**Fig. 4. Symptoms of Down syndrome**

## III. PROPOSED METHODOLGY

Histogram of Oriented Gradients

The histogram of oriented gradients (HOG) is a feature descriptor used in computer Vision and image processing for the purpose of object detection. The technique counts occurrences of gradient orientation in localized portions of an image. This method is similar to that of edge orientation histograms, scale-invariant feature transform descriptors, and shape contexts, but differs in that it is computed on a dense grid of uniformly spaced cells and uses overlapping local contrast normalization for improved accuracy.

The essential thought behind the histogram of oriented gradients descriptor is that local object appearance and shape within an image can be described by the distribution of intensity gradients or edge directions. The image is divided into small connected regions called cells, and for the pixels within each cell, a histogram of gradient directions is compiled. The descriptor is the concatenation of these histograms. For improved accuracy, the local histograms can be contrast-normalized by calculating a measure of the intensity across a larger region of the image, called a block, and then using this value to normalize all cells within the block. This normalization results in better invariance to changes in illumination and shadowing.

SVM classifier

The final step in object recognition using histogram of oriented gradient descriptors is to feed the descriptors into some recognition system based on supervised learning. The support vector machine (SVM) classifier is a binary classifier which looks for an optimal hyper plane as a decision function. Once trained on images containing some particular object, the SVM classifier can make decisions regarding the presence of an object, such as a human, in additional test images.

Local Binary Pattern

Local binary patterns (LBP) are a type of visual descriptor used for classification in computer vision. The local binary pattern (LBP) operator is defined as a gray-scale invariant texture measure, derived from a general definition of texture in a local neighborhood. Through its recent extensions, the LBP operator has been made into a really powerful measure of image texture, showing excellent results in many empirical studies. The LBP operator can be seen as a unifying approach to the traditionally divergent statistical and structural models of texture analysis. Perhaps the most important property of the LBP operator in real world applications is its tolerance against illumination changes. Another equally important property is its computational simplicity, which makes it possible to analyze images in challenging real-time settings. The LBP method has already been used in a large number of applications all over the world, including visual inspection, image retrieval, remote sensing, biomedical image analysis, face image analysis, motion analysis, environment modeling, and outdoor scene analysis [12].

The original LBP operator was introduced by Ojala, Pietikäinen and Harwood (1996). LBP operator is defined as a gray-scale measure, derived from a general definition of facial expression in a local neighborhood. For each pixel in an image, a binary code is produced by thresholding its value with the value of the center pixel. Different LBP operators can be defined according to the neighbors.

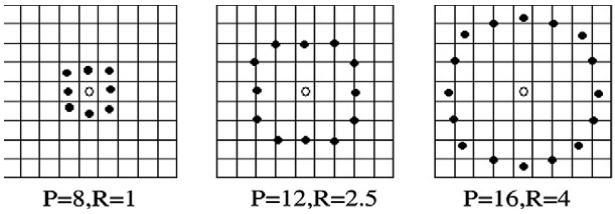


**Fig.5. Different LBP Operators**

An LBP pattern is a uniform pattern if it contains at most two bitwise transitions from 0 to 1 or 1 to 0 at its binary representation when the binary string is considered circular. For example, 11100001 (with 2 transitions) is a uniform pattern, whereas 11110101 (with 4 transitions) is a non-uniform pattern. There are 58 uniform LBP code patterns for 8-bits LBP code, and 256 - 58 = 198 non-uniform LBP patterns [13].

GIST Descriptor

"The GIST Descriptor is a low dimensional representation of the scene, which does not require any form of segmentation." [14]

Given an input image, a GIST descriptor is computed by convolving the image with 32 Gabor filters at 4 scales, 8 orientations, producing 32 feature maps of the same size of the input image. Each feature map is divided into 16 regions (by a 4x4 grid), and then average the feature values within each region. Finally the 16 averaged values of all 32 feature maps are concatenated, resulting in a 16x32=512 GIST descriptor.

k-Nearest Neighbors Classifier
In pattern recognition, the k-nearest neighbor's algorithm (k-NN) is a non-parametric method used for classification and regression. In both cases, the input consists of the k closest training examples in the feature space. The output depends on whether k-NN is used for classification or regression.

In k-NN classification, the output is a class membership. An object is classified by a majority vote of its neighbors, with the object being assigned to the class most common among its k nearest neighbors (k is a positive integer, typically small). If k = 1, then the object is simply assigned to the class of that single nearest neighbor.

We have kept the value of k=1 for our project.

We have implemented the k-NN classifier along with the three object identifying

techniques viz. HOG, LBP and GIST descriptor and recorded different accuracies.

Random Forest Classifier
Random forests or random decision forests are an ensemble learning method for classification, regression and other tasks, that operate by constructing a multitude of decision trees at training time and outputting the class that is the mode of the classes (classification) or mean prediction (regression) of the individual trees. Random decision forests correct for decision trees' habit of over fitting to their training set. We have implemented the Random Forest Classifier along with the three object identifying techniques viz. HOG, LBP and GIST descriptor. The function we have used for computing the random forest classifier in MATLAB is fitensemble. We have supplied as input the training set, the training label, 'bag' as the method, 100 as the no. of learning cycles, 'tree' as the learner template and 'classification' as the type of unsupervised learning method.

The complete project has been divided into a series of sections.

## IV. EXPERIMENTAL RESULT

The results obtained from the different techniques are summarized in Table 1.

**Table 1: Table showing the accuracies obtained from the different techniques**

| S. No. | Technique Applied | Accuracy obtained (in %) |
|---|---|---|
| 1 | HOG with SVM Classifier | 92.200 |
| 2 | LBP with SVM Classifier | 92.200 |
| 3 | GIST Descriptor with SVM Classifier | 99.600 |
| 4 | HOG with k-NN Classifier | 82.030 |
| 5 | LBP with k-NN Classifier | 96.800 |
| 6 | GIST Descriptor with k-NN Classifier | 96.800 |
| 7 | HOG with Random Forest Classifier | 88.800 |
| 8 | LBP with Random Forest Classifier | 99.200 |
| 9 | GIST Descriptor with Random Forest Classifier | 98.200 |

## V. CONCLUSION

We have developed a highly efficient system and obtained successful results for the proposed database by comparing nine different techniques. The highest accuracy was obtained by applying GIST descriptor with SVM classifier while the lowest accuracy was obtained by applying HOG with KNN classifier. Different facial expressions, different races, different light conditions, facial wears such as glasses, hair etc. make the problem more difficult but the system gives true outputs in these conditions. Hence, the system will be able to recognize Down syndrome correctly in most of the images irrespective of their differences. In future work, the system database can be widened. Furthermore, the system can be extended for different visual medical problems.

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