# CNN based Face Recognition System for Patients with Down and William Syndrome

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## ABSTRACT

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Keywords: Convolutional Neural Network Down syndrome Face recognition William syndrome Down syndrome, also known as trisomy genetic condition, is a genetic disorder that affects many people. Williams syndrome is a hereditary disorder that can affect anyone at birth. It marks medical and cognitive issues, such as cardiovascular illness, developmental delays, and learning impairments. This is accompanied by exceptional verbal abilities, a gregarious attitude, and a passion for music. Down syndrome and William Syndrome are both genetic illnesses. However, it can be distinguished from the arrangement of chromosome 21. Down syndrome and William syndrome can also be identified by recognizing faces, or facial characteristics, such as observing particular facial features. Therefore, this research develops Convolutional Neural Network (CNN) architectures to recognize Down syndrome and William syndrome using a facial recognition approach. A total of 480 facial photos were used in the study, with 390 images used for training data and 90 images used for testing data. The identification class is divided into three categories, Down syndrome, William syndrome, and normal. There are 160 photos in each patient class. This research presents two CNN architectures using a grayscale image of 256×256 pixels. The first CNN architecture comprises 12 layers, while the second comprises 15 layers. The average accuracy results with 12 layers were 91% by attempting to train and test six times. With 15 layers, the average accuracy value is 89%. In comparison, the first architecture has the highest accuracy value.

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## I. Introduction

Down syndrome, also known as trisomy genetic condition, is a genetic disorder that affects many people. An additional chromosome 21 causes trisomy genetic disease [1]. The extra chromosome increases the number of particular proteins, interfering with the body's natural growth. It may also result in predetermined changes in brain development. These abnormalities can also lead to developmental delays, learning impairments, heart problems, and blood cancer. Race, country, religion, or socioeconomic level have no bearing on this illness [2][3].

Williams syndrome is a hereditary disorder that can affect anyone at birth. It characterizes by medical and cognitive issues, such as cardiovascular illness, developmental delays, and learning impairments [4]. This is accompanied by exceptional verbal abilities, a gregarious attitude, and a passion for music. Williams syndrome is a neurogenetic condition that has been extensively researched. Williams syndrome, also known as Williams-Beuren syndrome or Infantile Hypercalcemia, is a multisystem neurodevelopmental condition that causes intellectual impairment

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[5][6][7]. Williams syndrome is a rare genetic illness that manifests in various symptoms and learning difficulties [8]. The heart, blood vessels, kidneys, and other body organs can all be affected in children with Williams syndrome. In addition, people with Williams syndrome have distinct traits on their faces, such as a different nose, mouth, and facial features [9][10][11][12]. Moreover, the children with Williams syndrome have a flat nasal bridge, short nose with a large tip, large mouth with full lips, small chin, small and spaced teeth, missing or crooked teeth, uneven eye, creases covering the corners of the eyes, and a white starburst pattern surrounds the iris or colored area of the eye [13][14]. Moreover, Down syndrome has physical characteristics such as a smaller head shape than normal people with a flat area at the nape of the neck, a bigger fontanelle that closes slower (average age two years), a slanted eye shape with a corner of the eye, and the centerfolds into the shape of a small mouth with a long tongue, giving it a projecting appearance [15][16]. Physical and mental characteristics of people with Down Syndrome include: face characteristics that are flat, head and ears are small, short neckline, a huge tongue, upward-slanting eyes, muscle tone is poor, and the person is short [17]. Furthermore, the mental characteristics of Down syndrome include short attention span, impulsive behavior, sluggish learning, and delayed language and speech development [18].

Cognitive problems, such as intellectual and developmental delays, learning disabilities, and speech disorders, are unique to Down syndrome. Down syndrome impairs the hippocampus, which is essential for memory and learning [19]. People with Down Syndrome are more likely to have the following health problems: Thyroid disease, Leukemia, Obesity, Chronic Constipation, Sleep Apnea, Poor vision, Cataracts, Strabismus, Anemia, Congenital heart defects, and Hearing loss [20][21]. Down syndrome and Williams syndrome can be diagnosed by detecting the face, or facial characteristics, such as picking up particular facial traits [22][23].

Downs syndrome and William syndrome are both genetic illnesses. However, it can be precisely detected with the arrangement of chromosome 21. There is an additional chromosome 21 in Down syndrome. Williams syndrome can manifest in many body parts, including the face, heart, and other organs. It can also have an impact on a child's learning abilities [24]. Facial landmarks can be used to identify people with Down Syndrome automatically using a non-standard snapshot of a patient's face. Facial landmarks are used for a local model, and then geometric feature extraction is performed based on anatomy landmarks and texture features from binary patterns. After feature selection, multiple classifiers are utilized to distinguish between Down syndrome and normal cases. The accuracy of the SVM classification with the RBF kernel employing texture characteristics was 94.6% [20].

Other studies have found that facial characteristics can identify people with Down syndrome. Down syndrome affects one out of every 1000 newborns born across the world. In the recent decade, the number of people diagnosed with Down syndrome has increased. It has been noted that people with Down syndrome exhibit various face traits. A face feature-based approach for detecting patients with Down's Syndrome. The Deep Convolutional Neural Network extracts and merges deep representations of different face areas. The Random Forest-based pipeline was then used to classify the merged representations. This model was evaluated on a dataset of over 800 people with Down syndrome and recognized 98.47% of them [19].

Previous studies have shown that face characteristics and landmark features can distinguish down syndrome and normal. Therefore, this research will design a system that can recognize Down syndrome and William Syndrome using a facial recognition approach and a Deep Learning algorithm Convolutional Neural Network. Previous research has already performed similar cases. However, this study distinguishes the syndrome into three categories: Down syndrome, William syndrome, and normal. In addition, the CNN network design used in this study differs from earlier studies.

#### **II. Method**

In this study, three categories of data were used: Down Syndrome, William Syndrome, and Normal. Input data or datasets, as well as test data, are retrieved via the website. The Down Syndrome, William Syndrome, and Normal classifications were created due to this research. The training and testing data storage for Convolutional Neural Network (CNN) technique is illustrated in Figure 1.

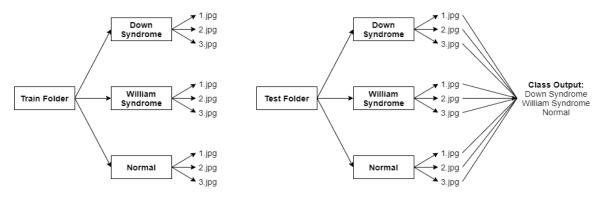


Fig. 1. Image data storage for CNN algorithm

This system will detect Down Syndrome, William Syndrome, and Normal using the CNN technique. In Figure 2, the stages of CNN's identification of these three classifications are as follows:

- 1. The RGB picture dataset is processed, transformed to grayscale, and size equalized, with a 256×256 input size.
- 2. Following processing, each class is organized into a folder, subsequently collected into a Train folder, as shown in Figure 1.
- 3. CNN training is responsible for all photos in each folder and the Train folder. Several distinct architectures will be used during the training phase. Figure 2 shows the CNN architectural model.
- 4. The fourth step is testing the data.

The architectures of the CNN network design, as indicated in Figure 2, will be as follows: Convolutional layer, Activation layer, Max Pooling layer, and Fully Connected layer. In terms of functionality, a convolutional neural network is similar to a neural network (artificial neural network) [25]. Weights, biases, and activation functions are examples of neurons in CNN. A CNN has a convolution layer, a pooling layer, an activation layer, and a fully linked layer. Figures 2 and Figure 3 present the CNN network architecture. The following process is the training and testing process. This research employs two alternative architectures. Figure 3 shows the first architecture, while Figure 4 shows the second architecture.

The system to be constructed is a CNN algorithm-based identification system for classified Down's Syndrome, William's Syndrome, and Normal. The total number of images used in this investigation was 480, with a grayscale resolution of 256×256. Table 1 shows the complete data distribution and is separated into two categories: training and testing.

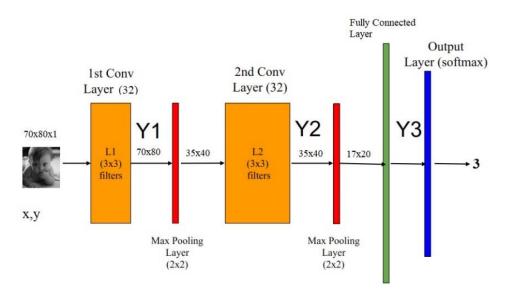


Fig. 2. CNN model for syndrome identification

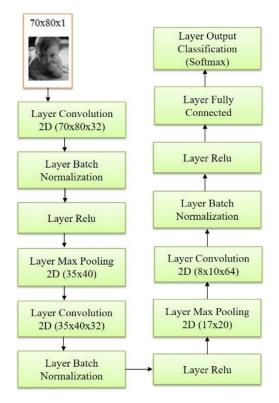


Fig. 3. CNN first architecture

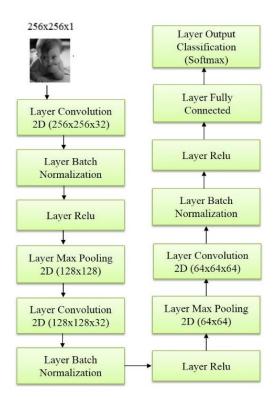


Fig. 4. CNN second architecture

Table 2 shows the image dataset that comprises three sorts of photographs: Down syndrome, William syndrome, and normal facial images. A grayscale image is used as the training and testing input. All photos are equal in size to  $256 \times 256$  pixels. The images are initially processed by being converted to grayscale and equalizing their sizes.

No	Classification	Training	Testing
1	Down Syndrome	130	30
2	William Syndrome	130	30
3	Normal	130	30
	Total	390	90

Table 1. Dataset distribution

#### Table 2. Example of image dataset

No	Down Syndrome	William Syndrome	Normal
1			
2			
3		- Contraction of the second se	E

# **III. Results and Discussions**

An RGB image is used, transformed to grayscale, and equalized to  $256 \times 256$  pixels. Two alternative architectures are used in the CNN training and testing procedure. Figure 3 illustrates the first architecture, and Figure 4 shows the second architecture. The first architecture uses 20 epochs (Figure 5) for training and 50 epochs for testing (Figure 6). The same epochs are also applied in the second architecture.

The first layer in Figure 4 is the input layer, and the input image is [70 80 1] and grayscale image with a resolution of  $256 \times 256$  pixels. Each convolution layer has various nodes or filters. For example, the convolution layer in Figure 4 has 32 nodes with a  $3 \times 3$  filter size. Figure 4 further indicates that the size of the pooling layer, for example, differs from other pooling layers in terms of nodes, yet both have a  $2 \times 2$  filter size. The pooling layer's results will change the image's size from  $256 \times 256$  to  $35 \times 40$ .

CNN was tested six times for training and testing and for identifying test data. Table 3 shows the test results, which show the average accuracy of the two CNN architectural models. In general, it shows that the 12 layer's accuracy outperforms the networks with 15 layers.

12 Layer (%)	15 layer (%)
0.94	0.88
0.92	0.91
0.86	0.89
0.91	0.89
0.94	0.88
0.92	0.91

Table 3. CNN accuracy

Epoch	1	Iteration	1 T	Time Elapsed (hh:mm:ss)	I I	Mini-batch Accuracy	1	Mini-batch Loss	I I	Base Learning Rate
1	1	1	I.	00:01:09	I	26.56%	1	1.4486	T	1.0000e-04
20	1	40	÷.	00:40:39	1	99.22%	÷.	0.0835	T	1.0000e-04

accuracy =

0.9394

Fig. 5. 20 Epoch for training the first architecture

Epoch	1	Iteration	1 T	Time Elapsed (hh:mm:ss)	T T	Mini-batch Accuracy	1	Mini-batch Loss	T T	Base Learning Rate
1	1	1	1	00:01:09	I	26.56%	i.	1.4486	1	1.0000e-04
20	1	40	1	00:40:39	1	99.22%	T.	0.0835	Т	1.0000e-04



# **IV. Conclusion**

Two CNN architectures are proposed in this study. With an average accuracy of 91%, the first CNN architecture was trained and evaluated six times. While, the accuracy of the second architecture, which includes 15 layers, is 89% percent on average. The first CNN architecture has the highest accuracy to provide effective identification. The future research will identify Down Syndrome, William Syndrome, and Normal using CNN transfer learning.

# **Declarations**

## Author contribution

All authors contributed equally as the main contributor of this paper. All authors read and approved the final paper.

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#### Conflict of interest

The authors declare no known conflict of financial interest or personal relationships that could have appeared to influence the work reported in this paper.

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## References

- P. N. Alexandrov, M. E. Percy, and W. J. Lukiw, "Chromosome 21-Encoded microRNAs (mRNAs): Impact on Down's Syndrome and Trisomy-21 Linked Disease," *Cell. Mol. Neurobiol.*, vol. 38, no. 3, pp. 769–774, Jul. 2017.
- [2] V. Dima, A. Ignat, and C. Rusu, "Identifying Down Syndrome Cases by Combined Use of Face Recognition Methods," in Advances in Intelligent Systems and Computing, 2018, pp. 472–482.
- [3] J. Y. R. Cornejo, H. Pedrini, A. Machado-Lima, and F. de L. dos S. Nunes, "Down syndrome detection based on facial features using a geometric descriptor," *J. Med. Imaging*, vol. 4, no. 04, p. 1, Dec. 2017.
- [4] R. P. Thom, B. R. Pober, and C. J. McDougle, "Psychopharmacology of Williams syndrome: safety, tolerability, and effectiveness," *Expert Opin. Drug Saf.*, vol. 20, no. 3, pp. 293–306, Mar. 2021.

- [5] M. Lugo et al., "Social, neurodevelopmental, endocrine, and head size differences associated with atypical deletions in Williams–Beuren syndrome," Am. J. Med. Genet. Part A, vol. 182, no. 5, pp. 1008–1020, May 2020.
- [6] S. Kaya, K. Orhan, and F. Tulga öz, "Williams-Beuren Syndrome: A Case Report," Cumhur. Dent. J., pp. 481–485, Dec. 2019.
- [7] C. G. Del Cole, S. C. Caetano, W. Ribeiro, A. M. E. e. Kümmer, and A. P. Jackowski, "Adolescent adaptive behavior profiles in Williams–Beuren syndrome, Down syndrome, and autism spectrum disorder," *Child Adolesc. Psychiatry Ment. Health*, vol. 11, no. 1, p. 40, Dec. 2017.
- [8] H. Liu *et al.*, "Automatic Facial Recognition of Williams-Beuren Syndrome Based on Deep Convolutional Neural Networks," *Front. Pediatr.*, vol. 9, no. May, pp. 1–7, 2021.
- [9] D. Dimitriou, H. C. Leonard, A. Karmiloff-Smith, M. H. Johnson, and M. S. C. Thomas, "Atypical development of configural face recognition in children with autism, Down syndrome and Williams syndrome," *J. Intellect. Disabil. Res.*, vol. 59, no. 5, pp. 422–438, May 2015.
- [10] A. Santos, C. Silva, D. Rosset, and C. Deruelle, "Just another face in the crowd: Evidence for decreased detection of angry faces in children with Williams syndrome," *Neuropsychologia*, vol. 48, no. 4, pp. 1071–1078, Mar. 2010.
- [11] N. Shalev, A. Steele, A. C. Nobre, A. Karmiloff-Smith, K. Cornish, and G. Scerif, "Dynamic sustained attention markers differentiate atypical development: The case of Williams syndrome and Down's syndrome," *Neuropsychologia*, vol. 132, p. 107148, Sep. 2019.
- [12] C. Ji, D. Yao, M. Li, W. Chen, S. Lin, and Z. Zhao, "A study on facial features of children with Williams syndrome in China based on three-dimensional anthropometric measurement technology," *Am. J. Med. Genet. Part A*, vol. 182, no. 9, pp. 2102–2109, Sep. 2020.
- [13] Vincy Devi V.K and Rajesh R, "A study on Down syndrome detection based on Artificial Neural Network in Ultra sonogram images," in 2016 International Conference on Data Mining and Advanced Computing (SAPIENCE), Mar. 2016, pp. 204–209.
- [14] P. Shukla, T. Gupta, A. Saini, P. Singh, and R. Balasubramanian, "A Deep Learning Frame-Work for Recognizing Developmental Disorders," in 2017 IEEE Winter Conference on Applications of Computer Vision (WACV), Mar. 2017, pp. 705–714.
- [15] Ş. Saraydemir, N. Taşpınar, O. Eroğul, H. Kayserili, and N. Dinçkan, "Down Syndrome Diagnosis Based on Gabor Wavelet Transform," J. Med. Syst., vol. 36, no. 5, pp. 3205–3213, Oct. 2012.
- [16] R. Al-Shawaf and W. Al-Faleh, "Craniofacial characteristics in Saudi Down's syndrome," King Saud Univ. J. Dent. Sci., vol. 2, no. 1–2, pp. 17–22, Jul. 2011.
- [17] S. O. Wajuihian, "Down syndrome: An overview," African Vis. Eye Heal., vol. 75, no. 1, Mar. 2016.
- [18] J. D. Santoro *et al.*, "Neurologic complications of Down syndrome: a systematic review," J. Neurol., vol. 268, no. 12, pp. 4495–4509, Dec. 2021.
- [19] A. Mittal, H. Gaur, and M. Mishra, "Detection of Down Syndrome Using Deep Facial Recognition," in Advances in Intelligent Systems and Computing, 2020, pp. 119–130.
- [20] Q. Zhao, K. Rosenbaum, R. Sze, D. Zand, M. Summar, and M. G. Linguraru, "Down syndrome detection from facial photographs using machine learning techniques," in *Medical Imaging 2013: Computer-Aided Diagnosis*, Feb. 2013, p. 867003.
- [21] Q. Zhao et al., "Automated down syndrome detection using facial photographs," in 2013 35<sup>th</sup> Annual International Conference of the IEEE Engineering in Medicine and Biology Society (EMBC), Jul. 2013, pp. 3670–3673.
- [22] S. M. Tabatabaei and A. Chalechale, "Using DLBP texture descriptors and SVM for Down syndrome recognition," in 2014 4<sup>th</sup> International Conference on Computer and Knowledge Engineering (ICCKE), Oct. 2014, pp. 554–558.
- [23] W. Song *et al.*, "Multiple facial image features-based recognition for the automatic diagnosis of turner syndrome," *Comput. Ind.*, vol. 100, pp. 85–95, Sep. 2018.
- [24] J. Grieco, M. Pulsifer, K. Seligsohn, B. Skotko, and A. Schwartz, "Down syndrome: Cognitive and behavioral functioning across the lifespan," Am. J. Med. Genet. Part C Semin. Med. Genet., vol. 169, no. 2, pp. 135–149, Jun. 2015.
- [25] A. Qayyum, S. M. Anwar, M. Awais, and M. Majid, "Medical image retrieval using deep convolutional neural network," *Neurocomputing*, vol. 266, pp. 8–20, Nov. 2017.