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Research Article

An Automated Identification of Muscular Atrophy and Muscular Dystrophy Disease in Fetus using Deep Learning Approach

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ABSTRACT

Muscular Atrophy (MA) and Muscular dystrophy (MD) diseases are genetic diseases. These diseases are commonly diagnosed with the help of techniques such as chorionic villus sampling (CVS) and amniocentesis, vitro fertilization (IVF), Intrauterine insemination (IUI) in fetus level. Among these techniques, chorionic villus sampling (CVS) and amniocentesis recommends the diagnosis of muscular dystrophy and muscular atrophy through identification of the patterns that exist in fetus. However, while there is a dearth of information about disease-specific patterns, there are overlaps among the patterns of different diseases. Therefore, Deep learning techniques can be used in the diagnosis of muscular dystrophies, muscular atrophy which enables us to analyze, learn, and predict for the future. In this scenario, the current research paper an automated muscular dystrophy detection and muscular antropy model using Convolutional neural networks (CNN)method and Restricted Boltzmann machines (RBM). These models have been proposed to act as an automated deep learning (DL) model that examines the chorionic villus sampling (CVS) and amniocentesis data.

Keywords: Muscular Atrophy, Muscular dystrophy, chorionic villus sampling, amniocentesis, CNN.

INTRODUCTION

Babies and young toddlers are most commonly affected by spinal muscular atrophy, a condition that impairs muscle movement. In families, parents pass down SMA to their offspring. (Omid Omrani *et al* ,2009) determined the frequency of SMN and neuronal apoptosis inhibitory protein (NAIP) gene deletions in Iranian SMA patients. In 68 out of 75 cases, homozygous deletion of SMN1 exons 7 and/or 8 was found (90%) of the time. In 40/54 type I, 2/8 type II, and 1/13 type III individuals, deletion of exon 5 of the NAIP gene was detected. Mutations in a gene known as the survival motor neurone gene 1 (SMN1) are the cause of it. We inherit one gene from each parent; genes are paired. They determine things like your body's functioning and the colour of your eyes and hair. A protein produced by the SMN1 gene aids in the proper function of the neurones that regulate our muscles. Movement issues and muscle weakness may result from a gene mutation. Two damaged copies of this gene, one from the mother and one from the father, are present in a kid with SMA. A human operator must manually outline each muscle in order to measure muscle mass in CT, particularly for the psoas muscle. (Nicholas C, et al.,2020 Wang)designd a deep neural network which describes On clinical CT images of a diseased population, it may enable the automation of muscle measures. These automated psoas size measures were significant predictors of death in cirrhosis patients, demonstrating the potential for broader therapeutic application of this technology. More automation is required to enhance the precision and repeatability of muscle measures as well as to make adoption and integration into clinical care easier. By using CNN techniques, he can predict the muscle mass in Liver. (Voulodimos A *et all,2018*) describes an outline of some of the most important deep learning techniques for computer vision applications, including stacked denoising autoencoders, deep belief networks, deep Boltzmann machines, and convolutional neural networks. (Mahmoud Shekari Khaniani et al ,2016) evaluated a methodology for families with homozygous deletion of the SMN1 gene, the use of SMN1 deletion detection using a straightforward PCR assay may be recommended for prenatal prediction.(

Rashna S Dastu *et al.*,2006) defines the initial line of inquiry for confirming a clinical diagnosis of SMA is now SMN gene deletion investigations. The diagnosis of SMA is confirmed by the presence of homozygous deletions of exons 7 and/or 8 of the SMN1 gene, even in patients with atypical clinical characteristics. NAIP gene deletions are helpful in prognostic prediction since they were primarily observed in individuals with severe illness. (Jiang W, *et al* ,2013) defines In terms of prenatal diagnosis, the quantitative PCR approach's results were just as trustworthy as those obtained with the PCR-RFLP method. The quantitative PCR approach can provide additional details on SMA carrier status that align with linkage analysis findings. (Saul Ullman M.D, *et al,1985*), his team describes a new technique called Chorioniv villus sampling (CVS) which is used to identify the genetic disorders. (Juliann M Savatt et al ,2021) describes genetic testing for neuro developmental disorders by using Chromosomal micro array methods. Diana (W. Bianchi *et al,2005*) evaluates Meternal serum screening test which is used to give the prediction for gentic disorders. (G. Coratti, *et al.*,2023) evaluates the prediction to classify the types of SMA using different machine learning algorithms. (Bram De Wel,et al,2023) different Deep learning approaches like (convolutional randomforest classifier,U-net model segmentation)are used to classify thetypes Muscular dystrophys.(YiRang Shin *et al.*,2021) uses Ultrasongrahpy methods ,AI,ML,and DL techniques like deep convolutional neural networks (DCNN),KNN algorithms to classigy the genetic diseases. (Nwawka *et al,2016*) proposed Musculoskeletal Ultrasound methods to identify the geneticdisorders. (M. Zaidman *et al.*2012) proposed Reliable Measurements of Calibrated Muscle Backscatter from Different Ultrasound Systems to classify the genetic disorders. (S Pillen *et al.*,2007) proposed Diagnostic value in childhood neuromuscular disease*Neuromuscul Disorder using different machine learning techniques*. (PanelK et al.,2020) proposed Qualitative and quantitative muscle ultrasound in patients with Duchenne muscular dystrophy using Deep learning techniques. (Wen-Chin Weng *et al,2019*) proposed Instantaneous frequency as a new approach for evaluating the clinical severity of Duchenne muscular dystrophy through ultrasound imaging and CNN. (Fukuyama Y Et al,1981) proposed Percutaneous needle muscle biopsy in the diagnosis of neuromuscular disorders in children. (A Krizhevsky *et al,2012*) proposed ImageNet Classification with Deep Convolutional Neural Networks. (Rahul Chauhan *et al,2018*) Convolutional Neural Network (CNN) for Image Detection and Recognition. (Anu Sayal, *et al,2023*) prposed Neural Network and Deep Learning.

The majority of the previous studies focused on identifying the genetic disorders like spinal muscular antrophy and muscular dystrophy using different biological methods through AI, ML and DL methods. In contrast this paper introduces advanced ML and DL Algorithms tailored for realtime Chronionic villus sampling and amniocentesis testing methods at fetus level to identify genetic disorders like muscular antrophy and muscular dystrophy. The paper is structured as follows: Section 2 details the proposed method, section 3 presents results and discussion, and section 4 offers conclusions.

METHODS

The proposed method is shown Figure 1. Chroionic villus sampling and Amniocentesis are used to detect Muscular antrophy and muscular dystrophy diseases as shown in fig (1).

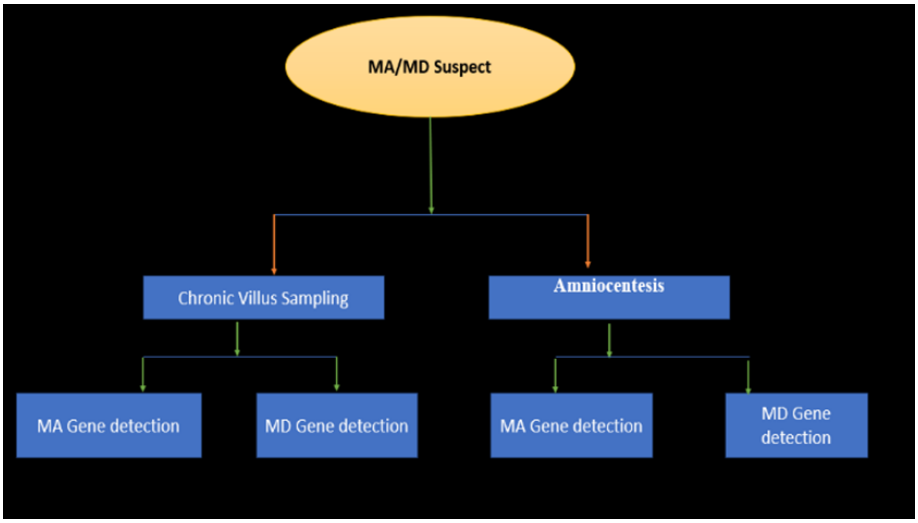

Fig 1: Proposed Methodology for Diagnosis of Muscular Atrophy and Muscular dystrophy

Chorionic Villus Sampling (CVS)

Typically, between weeks 10 and 14 of pregnancy, a little tissue sample is taken from the placenta through the cervix or abdomen that is as shown in fig (2). Between weeks 10 and 14 of pregnancy, we can do this. To assist in removing a little portion of placental tissue, doctor will use ultrasonography. Foetal DNA is found in tiny structures called chorionic villi found in the tissue. SMA testing will be performed on this DNA.

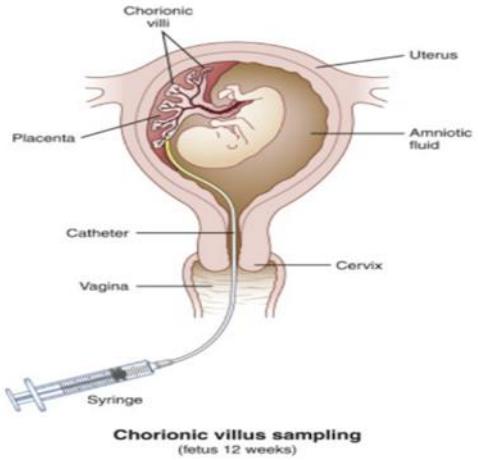

Fig 2. Chorionic villus sampling

Amniocentesis

Typically, between weeks 16 and 20, a tiny amount of amniotic fluid is extracted from the abdomen using a fine needle. This test can be performed 16–20 weeks during pregnancy. Doctor will place a tiny needle into your abdomen under the supervision of an ultrasound. A small amount of amniotic fluid containing foetal DNA is extracted by the needle. After that, the fluid is examined for SMA. SMA genetic mutation diagram as shown in below of fig (3).

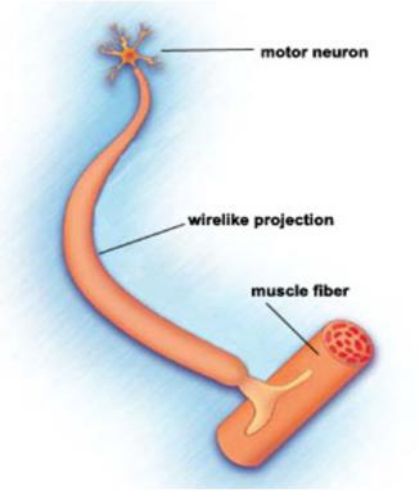

Fig -3: SMA (Spinal Muscle Atrophy)

Deep Learning Algorithms

After performing chroionic villu sampling we applied ML classifiers SVM for SMA classification. Later we applied CNN and RNN also for Muscular antrophy and muscular dystrophy classification. All the applied models are evaluation for accuracy.

ML Classifiers

Regression analysis and data classification both benefit greatly from the usage of SVMs. One significant benefit of SVMs is that, during the learning phase, they acquire a subset of support vectors, which is frequently a very tiny

portion of the original data set. A tiny data collection is used to create this set of support vectors, which reflect a specific classification problem. Generalised Support vector machine as shown in fig(4).

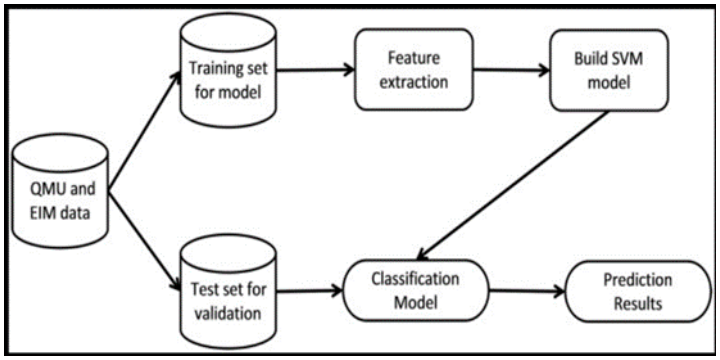

Fig -4: Generalised Support vector machine

CNN and RNN

CNN and RNN are most popular methods in Deep learning Methodology.Chroinic villus samling image dataset is applied to CNN network as shown in fig 5.CNN is the extended version of ANN.Convoloutonal layer apply the filter to the input image (CVS) to extract the features. The final prediction is made by the fully connected layer after the Convolutional layer applies filters to the input image to extract features and the Pooling layer downsamples the image to minimise computation. The network uses gradient descent and backpropagation to find the best filters.

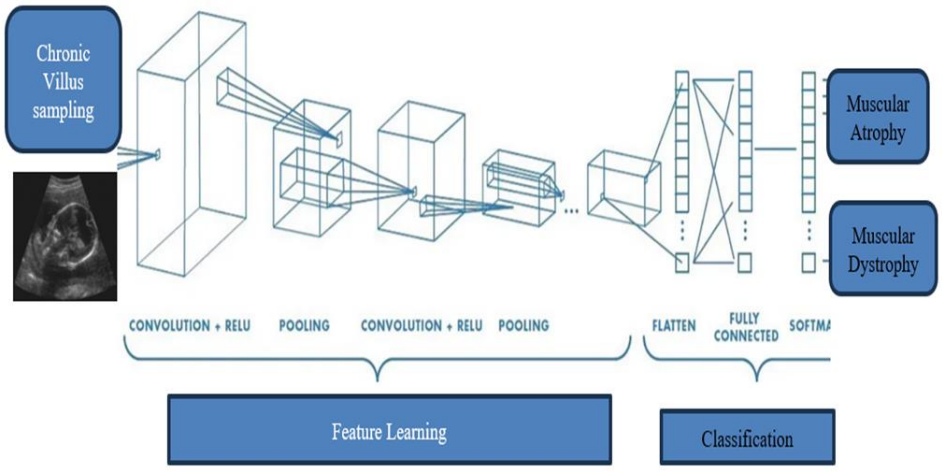

Fig -5: Classification using CNN

RESULTS AND DISCUSSION

Creation of dataset

A single institute used multiplex ligation-dependent probe amplification based copy number analysis of the 79 DMD exons to assess a population of 12,362 women. When a single exon loss was suspected, the primer region was sequenced consecutively Fig-6. A single institute investigated a sample of 12,362 women employing copy number analysis of the 79 DMD exons based on multiplex ligation-dependent probe amplification. When a single exon loss was suspected, the primer region was sequenced consecutively.

Healthy women of childbearing age who underwent testing at the Genetic Institute of Meir Medical Centre in Israel between July 2020 and August 2021 were included in the study cohort.The Israeli Ministry of Health (MOH) provided funding for the screening.Every lady had to disclose her ethnic background and any genetic disease history in her family. Our genetic institute's referred population is ethnically varied and accurately reflects the majority of Israel's ethnic groups. Different regions of Israel have varying proportions of various ethnic groups. In our cohort,

the Arab population makes up 33.2% of the total, whereas the Israeli population makes up 21% .Electromyography (EMG), muscle biopsy, genetic testing, and creatin kinase (CK) levels are used to diagnose Duchenne muscular dystrophy 75 patients with a range of neurological and muscular conditions (myopathies in 7, lower motor neurone diseases in 22, upper motor neurone illnesses in 6, and unlocalized or miscellaneous disorders in 13) underwent percutaneous needle biopsies, and the procedure's value as a diagnostic technique was assessed.

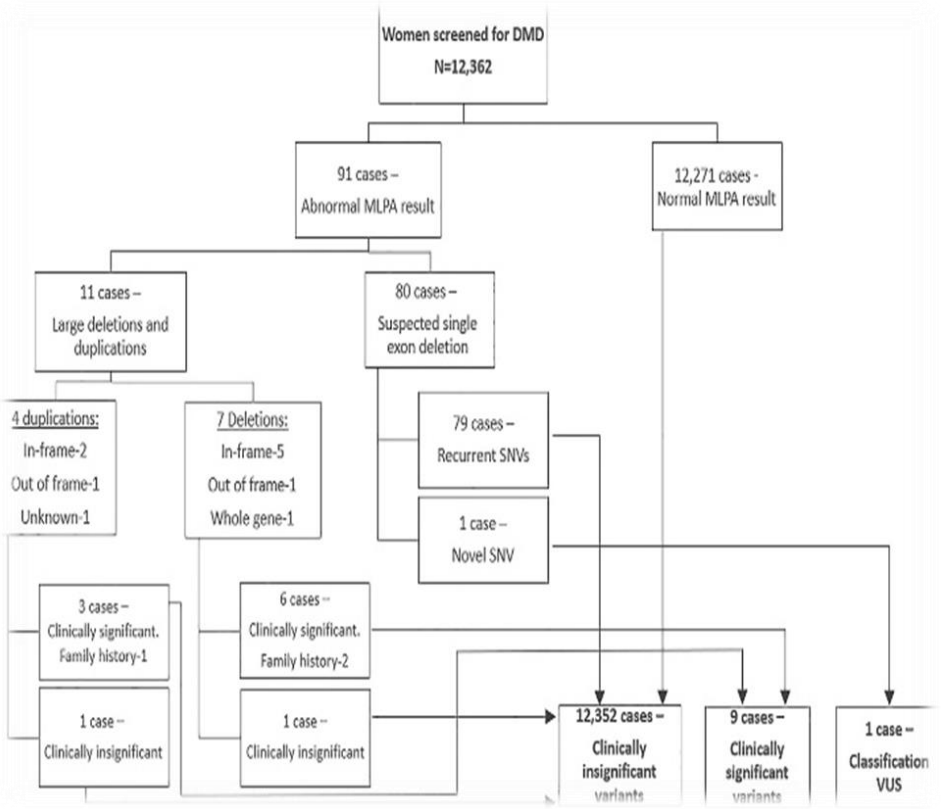

Fig-6: DMD cases

Chronical villus sampling Procedure

A little tissue sample is taken from the placenta through the cervix or abdomen during the chronic villus sampling procedure, which typically takes place between weeks 10 and 14 of pregnancy. Between weeks 10 and 14 of your pregnancy, you can do this. Feed this sample into the convolutional neural network. CNN's primary goal is to categorise diseases by selecting features, convolution, pooling, and building a fully connected model before doing classification. some samples of (CVS) as shown in fig-7.

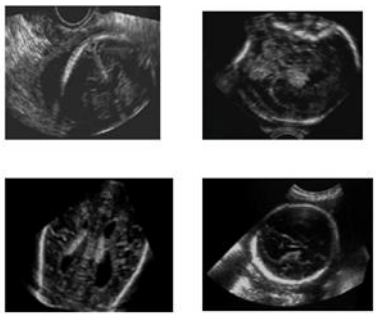

Fig-7: Different images of Chronic villus sampling

Applying CVS samples to CNN

Now apply the CVS samples to our CNN model. As illustrated in the fig-8 first step is import the libraries like pandas, numpy, matplotlib, sklearn, OpenCV.After that importing the dataset which is related CVS sampling which contains two classes like Muscular Atrophy and Muscular Dystrophy effected images.After that data visualization, In order to

construct the classifier for each class, we will attempt to comprehend and visualise a few of the images that have been supplied as shown in fig-9.

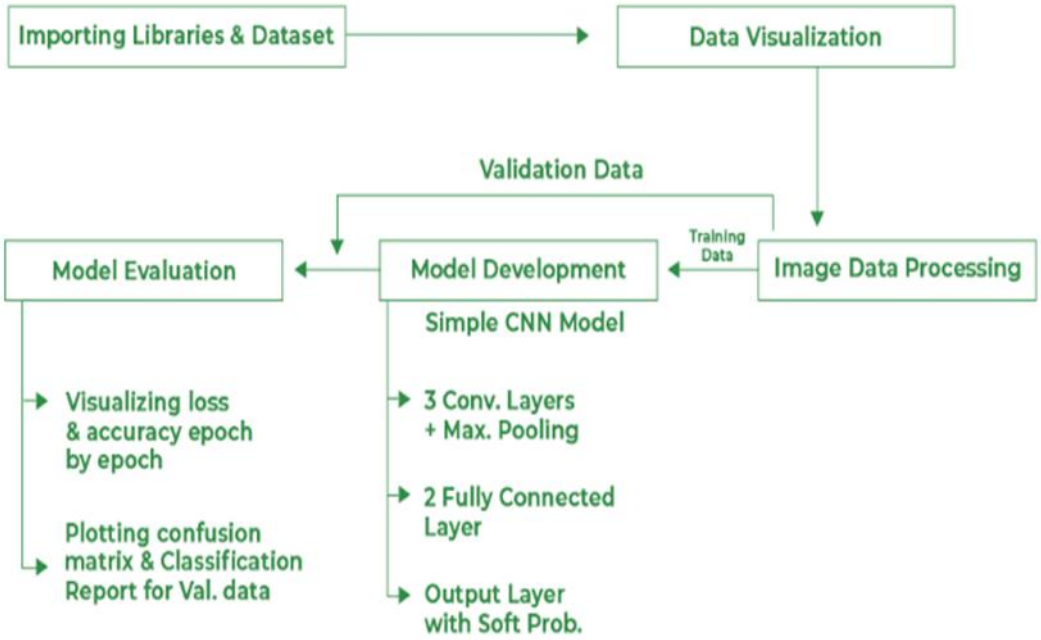

Fig-8: Flowchart of applying CVS samples to CNN model

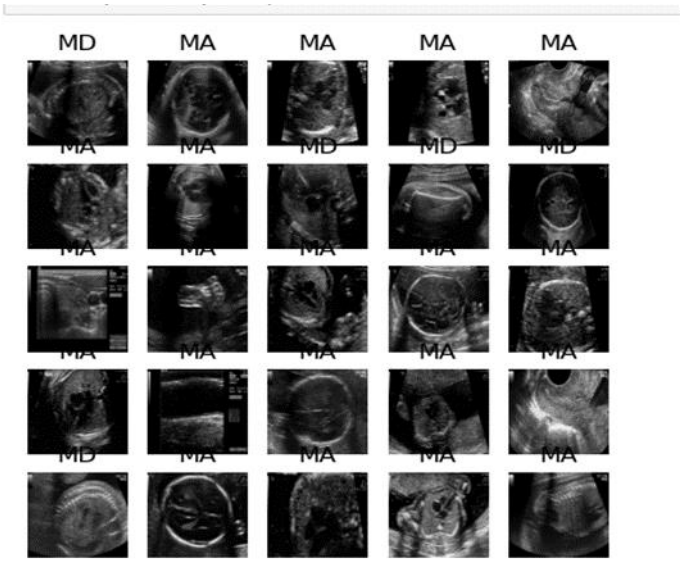

Fig-9: Data visualization of MA and MD

Feature Extraction

In Deep learning, feature extraction is a technique that lowers processing resource requirements without sacrificing significant or pertinent data. In order to analyse data efficiently, feature extraction aids in reducing the dimensionality of the data. Stated differently, feature extraction is the process of developing new features that more effectively extract the key information from the original data. Finding the linear feature combinations that best divide two or more classes of objects or events is the goal of linear discriminant analysis, or LDA. One essential part of convolutional neural networks (CNNs) that lowers the spatial dimensions of feature maps is the pooling layer. Usually, the convolutional layer comes before it.

Model Development

To build CNN Model we use Tensorflow Library and Keras framework activities. Here we implement Sequential model which contains 3 convolutional layers, flatten layer, two fully connected layers, dropout layer and one output layer. summary of our model is as shown in the fig-10.

```
model.summary()
Model: "sequential"
_________________________________________________________________
 Layer (type)                Output Shape              Param #
=================================================================
 rescaling_1 (Rescaling)     (None, 180, 180, 3)       0

 conv2d (Conv2D)             (None, 180, 180, 16)      448

 max_pooling2d (MaxPooling2D  (None, 90, 90, 16)       0
 )

 conv2d_1 (Conv2D)           (None, 90, 90, 32)        4640

 max_pooling2d_1 (MaxPooling  (None, 45, 45, 32)       0
 2D)

 conv2d_2 (Conv2D)           (None, 45, 45, 64)        18496

 max_pooling2d_2 (MaxPooling  (None, 22, 22, 64)       0
 2D)

 flatten (Flatten)           (None, 30976)             0

 dropout (Dropout)           (None, 30976)             0

 dense (Dense)               (None, 128)               3965056

 dense_1 (Dense)             (None, 2)                 258

=================================================================
Total params: 3,988,898
Trainable params: 3,988,898
Non-trainable params: 0
_________________________________________________________________
```

Fig-10: Sequential Model

Visualizing the loss and Accuracy epoch by Epoch

Optimizer, Loss and metrics are used to calculate the accuracy epoch by epoch in CNN model as shown in below fig-11.

```
Epoch 1/5
209/209 [==============================] - 261s 1s/step - loss: 0.7278 - accuracy: 0.6318 - val_loss: 0.8551 - val_accuracy: 0.
3906
Epoch 2/5
209/209 [==============================] - 230s 1s/step - loss: 0.6289 - accuracy: 0.6862 - val_loss: 1.0177 - val_accuracy: 0.
3906
Epoch 3/5
209/209 [==============================] - 234s 1s/step - loss: 0.6252 - accuracy: 0.6862 - val_loss: 0.7977 - val_accuracy: 0.
3906
Epoch 4/5
209/209 [==============================] - 227s 1s/step - loss: 0.6240 - accuracy: 0.6862 - val_loss: 0.8548 - val_accuracy: 0.
3906
Epoch 5/5
209/209 [==============================] - 226s 1s/step - loss: 0.5803 - accuracy: 0.6983 - val_loss: 0.8464 - val_accuracy: 0.
4103
```

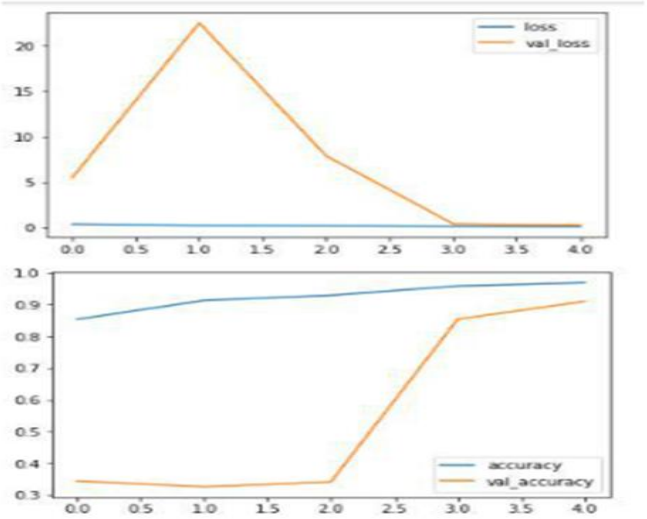

Fig-11 Acuuracy Model

CONCLUSION

Using our Convolutional Neural network of Deep Learning, we can quickly determine the disorders of muscular atrophy and muscular dystrophy that affect that baby based on the results of testing of chronic villus sample on the woman's foetus. However, it might provide 70–80% classification accuracy. Amniocentesis, the second method of fetal genetic testing, will be the subject of my upcoming research study.

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