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Revelation of Down Syndrome Using Artificial Neural Network

Vincy Devi V. K[1*], Rajesh R[2]

[1]Dept. of Computer Science, Bharathiar University, Coimbatore, India
[2]Dept. of Computer Science, CHRIST, Bangalore, India

Corresponding Author: vincydevi15@gmail.com, Tel.: +917907559811

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Abstract—A disorder in genetic chromosome 21 is popularly known as the Down syndrome (trisomy 21). It results in development and intellectual delays, but if we are able to give the exact care early on, it can make a good difference. Studies show that we can detect the Down syndrome in the early stages of pregnancy by identifying the absence of the fetal nasal bone. The common method followed in this issue is the visual identification of the 'absence' using ultra sonogram image of the nasal bone region. However, the visual identification technique is inefficient and difficult to follow. Thus, image processing based visual extraction technique can play a role in this case. In this paper, we have the raw data, which is employed to train the Back Propagation Neural Network (BPNN). The ultrasonogram images can be analyzed using this feed forward trained neural network and the detection can be made with appreciably low error rates. MATLAB is the platform used in this work for training the Artificial Neural Network (ANN).

Keywords— Down syndrome, Back Propagation Neural Network, Feature extraction, chromosomal abnormalities.

I. Introduction

The American college of obstetricians and gynecologists instructed that every pregnant woman must carry out a screening test regarding fetal aneuploidies [1]. Studies proved that Nasal bone abnormalities in 3 unselected fetuses with Down syndrome can be detected prenatally with the help of ultrasonography [2]. In these studies, nasal bones were undetectable through ultrasonography, however, one had a nasal bone with a measurement that is below the 2.5th percentile of the common distribution for that gestational age.

A report published by Cicero et al. [3] shows that in nearly 73% of Down syndrome cases, the absence of the fetal nasal bone was observed in the first trimester of pregnancy. Only 0.5% of unaffected cases is thus suggested a further improvement in the performance of first-trimester screening. Thus, the data collected by Cicero et al summarizes that detection efficiency become 98% by the inclusion of the nasal bone in first-trimester Down syndrome screening. In order to assess the feasibility of measuring nasal bone length in first-trimester pregnancy Orlandi et al [4] carried out various tests. Their major intention was to confirm if the absence of a fetal nasal bone is a marker for Down syndrome. At the end of their study, they found out that in nearly 94.3% of the cases, the nasal bone assessment was successfully achieved with ultrasound examinations and they further concluded that the absence of the nasal bone can be used as an indicator for Down syndrome in the first trimester of pregnancy.

The sonographic tests at 11 – 24 weeks of gestation founded out that approximately 65% of trisomy 21 fetuses have either absent or short nasal bone [5-8]. Kagan et.al scrutinizes the performance of first- trimester screening for aneuploidies. Their conclusion was, the performance of first-trimester screening for trisomy 21 improves with the assessment of the fetal nasal bone. Section II discusses some major works related to the detection of Down syndrome. Section III explains the methodology employed in this work. Section IV is the results and discussion part of this study and finally, section V concludes the work, along with the suggestion for future work.

II. Related Work

In this work, we employ BPNN for realizing the visual extraction based identification of Down syndrome. The technological progress and the availability of high-capacity microcomputers have accelerated the developments in artificial intelligent diagnostic systems [9, 10]. New computer methodologies are evolved based on the simulation of the human brain (Artificial Neural Networks or ANN) and they overcome the problems arising from the applications of low-level decision making systems [11, 12]. Such low-level systems depend on conventional biostatistics to manage unclear biological models. In such studies, the researchers had developed an artificial intelligent diagnostic system based on neural networks for determining the genetic disorders and mid-pregnancy (16-24th gestational weeks) fetal well-being. This is mainly based upon maternal age, gestational week and 'Triple Test' measurements.

Coppedè et al [13] performed a study using ANNs for identifying the key factors linking folate metabolism to chromosome malsegregation. Here ANNs aims to understand natural processes and uses automated models to recreate those processes. These artificial networks thus follow a method of forecasting with an understanding of the relationship (in particular non- linear relationships) between variables [14-16]. ANNs initially learn a known set of data obtained from a given problem having a known solution (training). Then, the networks, inspired by the analytical processes of the human brain, are able to reconstruct the imprecise rules which may be underlying a complex set of data (testing). Nowadays, ANNs have been successfully using in the medical field. For example, they have been used many different applications, not limited to investigating the predictive values of risk factors related to Alzheimer's disease [17], identifying the placental determinants of fetal growth [18] and to compare between the Alzheimer's disease patients and controls [19]

III. METHODOLOGY

There are many studies conducted by different researchers to see if there any relation between the age of the mother and chance for Down syndrome. The study results are portrayed in Fig 1. It indicates that there is an increase in chance for Down syndrome as the age of mother increases. Its reasons should be biologically analyzed. Thus it is clear that early detection of Down syndrome is very much necessary and needful to society.

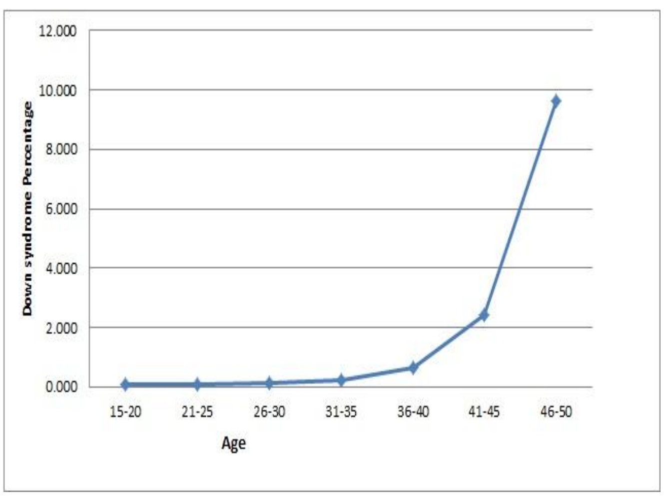

Figure 1. The chance for Down syndrome in different ages of the mother.

ANN is a collection of bonded units motivated by a biological neural system and it consists of vastly interconnected simple processors called neurons. The ANN models are categorized on the basis of,

- Learning rules, which are designated for customizing the weights and biases of the network.
- Training procedure, where a network is actually fine-tuned to do a specified task.

The structure of a neuron model is shown in Fig. 2 and a simple one layer neural network structure is depicted in Fig. 3. In this figure, X_i is the input of network and W_i is a weight of input. The behavior of these neurons is close to that of parallel processing units. Neurons are arranged as different layers and each layer is able to perform different transformation operations on their inputs [20]. In addition, the response of each neuron is characterized by a nonlinear function of the sum of its inputs. These artificial neurons are able to perform simple mathematical operations on its inputs and thus they follow the biological neuron functions and their unique learning process. Each of the neurons can accept a certain number of inputs from distinct neurons and this is proportional to their connection weights. They produce a specific output, which was circulated to multiple other neurons.

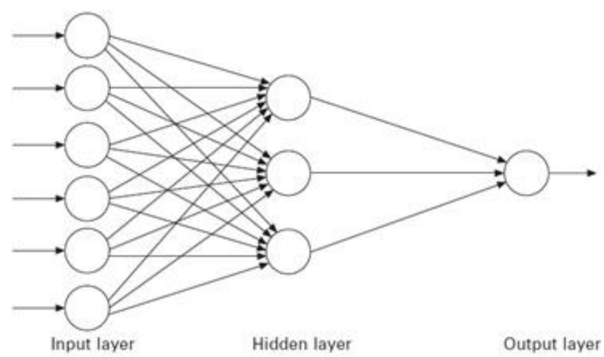

Figure 2. ANN model.

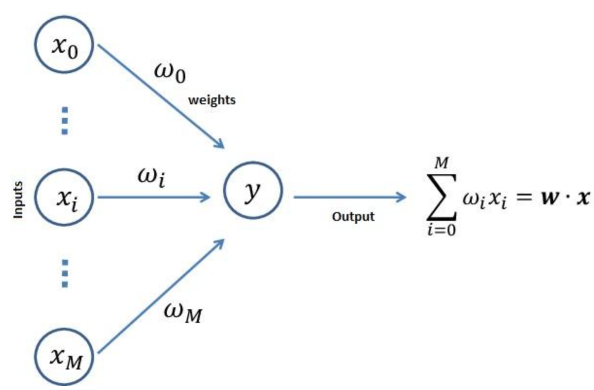

Figure 3. A simple one layer neural network structure.

The neural networks are powerful data modeling tool [21-23] and trained neural networks are able to perform many intelligent assignments similar to what a human brain does. It's important feature is that a neural network gathers knowledge through the process commonly called 'learning'. Information related with neural network is gathered within the inter-neuron network strength. This network strength is known as synaptic weights. One of the powerful merits of NN is the ability to learn and represent Linear and non-linear relationship [24]. Now a day, ANN is a globally accepted rapidly growing field and they are used in a wide range of applications including pattern recognition and classifications, products optimization and circuit design automation [25].

In the current engineering applications, the neural network technique that receiving a wide acceptance is the Back Propagation Neural Network, (BPNN) [26]. A feed-forward connection between the input and hidden layers, which is extended to the output layer, is the basis of the BPNN [27]. If we consider the BP algorithm, its most critical goal is to minimize the mean square error between the anticipated and the desired system responses. In this study, we rely upon a three-layered BPNN structure and it includes input, hidden and output layers. The architecture of BPNN is portrayed in Fig. 4. The input layer of the network receives the information (Figures) from the external sources and handed over them to the neural network for processing. That is the hidden layer, which is present in between the input and output layer processes the whole information received from the input layer [25]. The hidden layer supplies the organized information to the network output layer, and further they are directed to an external receptor. A continues training process establishes a non-linear relation between the given information and its correlative influence parameters based on BPNN.

Neural networks are experimentally trained using a set of algorithms and transfer functions. Through this experiment, it is revealed that for efficiently training the ANN structure, the Levenberg-Marquardt Back-Propagation algorithm with nonlinear sigmoid activation function is effective. A cross-validation technique supervises the learning process of ANN. With this technique, the data sets are divided into three; they are training, validation, and testing. For testing, we use the random sampling method. The weight adjustment of the ANN structure is performed using the training dataset.

In each of the layers, there are multiple ANN nodes to perform intelligent operations. The input function "η" is shown in equation (1) and the nonlinear sigmoid activation function is presented in equation (2).

$$\eta = \sum_{i=1}^{n} W_i X_i + b \quad (1)$$

$$S(\eta) = \frac{1}{1+e^{-\eta}} \quad (2)$$

IV. Results and Discussion

In this work, we investigated various ANN modeling through distinct epochs regarding different network architectures that encompass network algorithms, training, learning and transfer functions, many hidden layers with a number of neurons, various epochs, classifications of data sets and combinations of performances. Figure 4(a)-(d) represent the correlation coefficient for the selected ANN model. Here epoch stands for the measure, how many times the training vectors are used once to amend the weights. Table 1 shows the performance of the ANN model related to MSE for networks within the hidden layers. The gradient, Mu and validation fail parameters for our selected ANN model is shown in Fig.5.

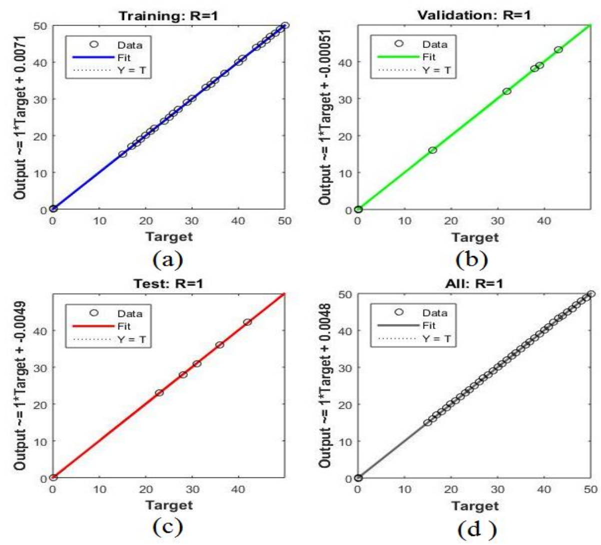

Figure 4. Correlation coefficient for selected ANN model.

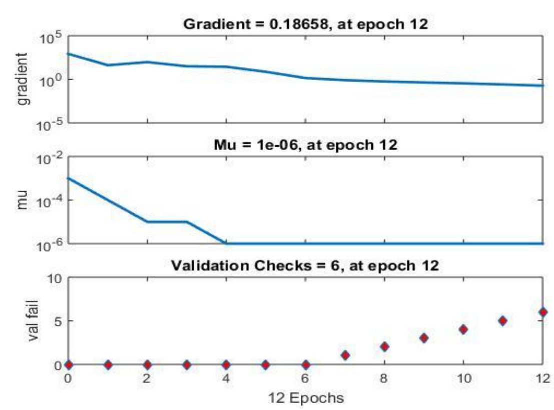

Figure 5. Gradient, Mu, and validation fail parameters for ANN.

Figure 6 portrays the MSE between training, testing and validation data at hidden neurons of 15. The conceptual diagram of our work is shown in Fig. 7.

Table 1. Performance of ANN model with 15 number of hidden neurons.

Corelation Coefficents	MSE	R
Training	9.44691e-4	9.99998e-1
Validation	3.14608e-3	9.99996e-1
Testing	4.55274e-3	9.99995e-1

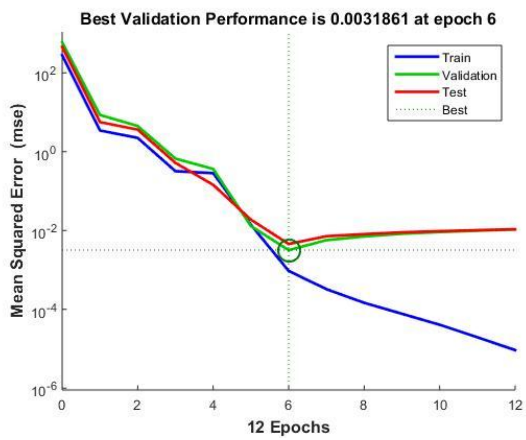

Figure 6. Variation of mean square error with a number of epochs for different hidden neurons.

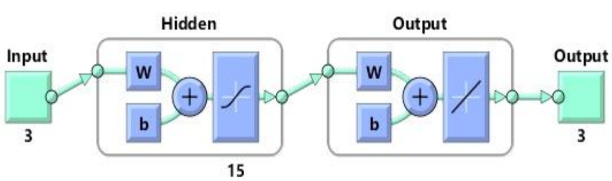

Figure 7. ANN model -conceptual diagram.

V. Conclusion and Future Scope

In this work, we highlighted how the age of mother leads to the possibility for an infant with Down syndrome. Here we signified the optimization of ANN structure by changing the aspects like algorithm, training, learning and transfer functions, hidden layers, dataset separation and performance. For this purpose, we used Levenberg-Marquardt Back-Propagation method for training the ANN model. The major advantage of our intelligent system is that it is possible to train the neurons by various diagnosis centers using their own parameters related to Down syndrome. As a future work, we focus to use more data to train our intelligent system for expanding the ability for prediction.

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Authors Profile

Mrs. Vincy Devi V. K is working as Assistant Professor in the department of Computer Applications , Sree Narayana Gurukulam College of Engineering , Kadayiruppu , Ernakulam . She has 12 years of teaching experience .She is doing her research in Medical Image processing. Her area of interest include Computer Networking, Cloud Computing and Bigdata Analysis.

Dr.Rajesh R is working as Associate Professor of Computer Science in the Department of Computer Science, CHRIST (Deemed to be University) Bangalore, India. Dr.Rajesh research interests are in the areas of Data Structures and Analysis of Algorithms. He has published 33 papers in various Journals and Conferences. Dr. Rajesh is also serving as Managing Editor, Lead Guest Editor, Associate Editor, Editorial board member and Technical Committee member of various National and International Conferences and Journals. He has received Veenus International Foundation's Outstanding Faculty award and Dewang Mehta Education Leadership Award to his credit.

