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Multi Regressive Splines and Quantum Correlated Prenatal Diagnosis in the Identification of Chromosomal Abnormalities

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Abstract: Prenatal diagnosis is the identification of the chromosomal abnormalities has undergone a number of changes in the last few years. Irrespective of the healthcare system, in prenatal medicine, the advantages and disadvantages of chromosomal abnormalities must be comprehended. This is because chromosomes are found in the nucleus of cells that performs as a carrier of genetic information. Several machine learning and deep learning techniques has been explored in the recent past to establish prediction models and the challenges to assess chromosomal abnormalities. Despite the advantages that machine learning and deep learning offers in screening for measuring disorders, it must be recognized that it does not adequately address many other chromosomal disorders and any of the structural fetal anomalies in an accurate and precise manner. To fill this gap in this work a method called, Multi-regressive Splines and Quantum Correlated (MS-QC) prenatal diagnosis in the identification of chromosomal abnormalities is proposed. The MS-QC method is split into two sections, namely, preprocessing the data and pertinent chromosomal feature extraction for accurate and precise chromosomal abnormalities. Initially, with the input obtained from the prenatal cytogenetic Data set a machine learning based Multivariate Logistic Regression Spline function is applied that can capture complicated and multifaceted nature. The Multivariate Logistic Regression Spline function by including multiple independent variables can account for more characteristics that influence the dependent variable and hence minimize the overall error and bias involved in analyzing the risk factors for Prenatal Diagnosis in the Identification of the Chromosomal Abnormalities. With the preprocessed data, correlation based feature selection to evaluate distinct subsets on the basis of Quantum Distribution function according to features-class correlations is measured for extracting the most pertinent chromosomal features from fetal cells. The Quantum Distribution function here assists in extracting the most probable chromosomal features from fetal cells therefore obtaining more precise information of fetal disorders indicators. Simulations will be performed to validate the proposed method in Python language in terms of precision by 18%, recall by 26% and diagnosis accuracy by 15% respectively.

Keywords*: Chromosome, Machine Learning, Deep Learning, Penatal Cytogenetic, Multivariate Logistic Regression Spline, Quantum Distribution*

1. Introduction

Karyotyping is a significant mechanism in cytogenetic practice for early diagnosis of genetic diseases. Chromosomal karyotype is paramount in ascertaining whether a newborn has a genetic disorder or not. A deep convolutional neural networks (DCNN) method was proposed in [1] with the purpose of classifying chromosomes in an automatic fashion, therefore improving the overall accuracy. Despite improvement in accuracy training time was not focused.

In [2], feature fusion classifier with dynamic weights (FFCDW) for measuring the abnormality in chromosome was presented. Also for detecting chromosomal abnormality, post augmentation of data using three deep learning techniques, ResNet, SENet, and VGG19, the three trained models were fused using arbitrary weighting model. Using this technique resulted in the improvement of overall precision. Despite improvements observed in terms of precision, the accuracy aspects were not focused. Yet another machine learning algorithm employing quantum processer was presented in [3] to focus on the time factor.

A growing number of predictive mechanisms have been designed with data sourced acquired from electronic health records, digital devices and diagnostic imaging. In this review, the latest materials and methods were explored using machine learning, also the algorithms to demonstrate prediction models and the issues to challenges to appraise fetal well-being, predicting and diagnosis of obstetric diseases such as preterm birth and fetal growth restriction was presented in [4]. Yet another method to focus on the area under receiver operating

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characteristic curve employing neural network in prenatal diagnosis was designed in [5].

In spite of the dominances that cell free DNA analysis offered in screening for common trisomies, it must be ascertained that it does not provide mechanism for several other chromosomal disorders and structural fetal anomalies.

To focus on this aspect antenatal screening for observing abnormalities in chromosome was presented in [6]. Prenatal microarray analysis as second-tier diagnostic test was applied in [7] to focus on the chromosomal microarray analysis with higher detection rate.A comprehensive assessment of genetic characteristics from genetic disorder diagnosis to prognosis prediction employing different machine learning techniques was investigated in [8]. Yet another method using copy number variation sequencing was proposed in [9] to focus on the precision and accuracy aspects. Timely and precise genetic disorders detection can result in remedial measures and circumvent irreversible occurrences. Hence, precise and low-risk diagnostic mechanisms for detecting these diseases in the fetal period are indispensable.

In [10], an integrated artificial neural network (ANN) and genetic algorithm (GA) were employed for predicting down-syndrome via first-trimester screening test. With this integrated mechanisms not only resulted in the improvement of sensitivity but also minimized the error factor in a significant manner. Noninvasive prenatal diagnosis (NIPD) works towards in detecting fetal-associated genetic disorders prior birth. This is performed by detecting markers in pregnant woman peripheral blood, captivating the holding the possible in minimizing the fetal birth defect risks. A review of Artificial Intelligence (AI) techniques for genomic medicine was investigated in [11].
Owing to the constraints of sensitivity in traditional methods, separation mechanisms on the basis of micro have been designed as novel mechanisms for isolating and enhancing chromosomal disorders was presented in [12].

Yet another method for observing chromosomal abnormalities employing deep learning techniques was investigated in [13]. Prenatal diagnosis employing ultrasound imaging is essential for abnormal identification of fetal. However, due to constrained coverage, this mechanism faces issues. To address on this issue, based on region of interest a mechanism for prenatal genetic disorder detection was presented in [14]. A method to improve clinic detection rate was proposed in [15] using deep learning. With this deep learning technique ensured mechanism for early risk assessment for identifying high risk fetuses affected by genetic diseases.

Despite the prenatal diagnosis in identifying chromosomal abnormalities maintained higher level of accuracy, the precision and recall involved gets compromised with the increase in the training process. Hence, to effective handle this issue, both precision and recall while ensuring prenatal diagnosis in the identification of chromosomal abnormalities, a learning technique has to be designed. To solve this issue, in this work, Multi-regressive Splines and Quantum Correlated (MS-QC) for Prenatal Diagnosis in the identification of chromosomal abnormalities is proposed.

1.1 Contributions of the work

Based on the above aspects, to summarize, the major contributions of our research work are as listed below.

- This work utilizes Multi-regressive Splines and Quantum Correlated (MS-QC) for prenatal diagnosis in the identification of chromosomal abnormalities. An efficient solution has been proposed for four different diagnostic planes.
- The proposed Multi-regressive Splines and Quantum Correlated (MS-QC) combines Multivariate Logistic Regression Spline-based preprocessing and pertinent chromosomal feature extraction employing Quantum Distribution. Feature extraction has been carried out by combining Quantum Distribution and Pearson Correlation therefore reducing the dimensionality and extracting probable chromosome feature extraction.
- The detection module is run over the Python high-level general-purpose programming language and validated the MS-QC method through a simulated environment.
- The prenatal diagnosis in the identification of chromosome abnormalities results compared with other chromosome abnormalities methods which show that the proposed chromosome abnormality method performs efficient and significant detection than others in terms of precision, recall, accuracy and training time.

1.2 Organization of the work

The remainder of this paper is organized as follows. Section 2 contextualizes machine, deep and ensemble learning techniques for prenatal diagnosis in identifying chromosomal abnormalities and reviews the related

works. Section 3 introduces our novel FPUS23 dataset and describes our proposed method Multi-regressive Splines and Quantum Correlated (MS-QC) for prenatal diagnosis in identifying chromosomal abnormalities. Section 4 provides with the experimental setup and implementation details provided in Section 5. A detail comparative analysis using table values and graphical representations is provided in Section 6. Finally, Section 7 presents the final remarks to conclude this paper.

2. Related Works

As far as the human genetic material is concerned, chromosomes are considered as the conveyors. Some of the chromosomal abnormalities include numerical and structural abnormalities, on one hand numerical abnormalities are said to occur with an increase or decrease in the chromosome number whereas on the other hand structural abnormalities are said to occur owing to the breakage and reunion of chromosome fragments.

Basic recognition performance was done employing support vector machine and followed by which deep convolution neural network was applied in [16] for abnormality detection with minimal error. Yet another method focusing on the detection error was proposed in [17] employing transfer learning. A comprehensive review of prenatal diagnosis and fetal defects was investigated in [18]. However the false negative rate involved in obtaining chromosomal abnormalities was not focused. To concentrate on this issue, computational intelligence approach for prenatal diagnosis was presented in [19]. Two novel techniques like, segmentation employing semantic mechanism and landmark localization were utilized in [20] for designing an AI model with the purpose of measuring the selected markers and estimate the diagnostic value for fetal abnormalities. Also receiver operating characteristic curves were used for screening of fetal abnormality.

The employment of Artificial Neural Networks (ANNs) [21] for classifying and instituting associations between genome and phenotype has been explored more predominantly over the past few decades. The reason for this is that ANNs are proven to be good approximates of complicated functions and as a result classification are said to be done without the requirement for explicitly defined input-output model.

Yet another method to assess the chromosomal detection effectiveness and application of clinical value of non-invasive prenatal testing (NIPT) for foetal was proposed in [22] to focus on the accuracy aspects. A hybrid optimization model employing DCNN and hill climbing strategy was proposed in [23] to focus on the accuracy aspects. A holistic review on prenatal diagnosis in China during the period of 2014 – 2020 was presented in [24]. Novel classification of abnormalities found in congenital, mastering the features of sonographic of each subtype and prenatal screening techniques in addition to providing assistance to appropriate counseling and management was presented in [25].

Two distinct cell morphology-based machine learning techniques to focus both on the accuracy and receiver operating characteristic curve was proposed in [26]. An innovative Bayesian mechanism was applied in [27] to perform prenatal diagnosis of genetic diseases, therefore resulting in achieve clinically acceptable accuracy. Despite improvement observed in terms of detection accuracy, however, training time was not focused. A seven year retrospective study on prenatal diagnosis using chromosomal abnormalities was presented in [28]. An extension of previous binary approaches for detection of chromosome was designed in [29]. Yet another method to comprehend technical basis and clinical indications employing cumulative diagnostic mechanism was proposed in [30] to yield an accurate chromosomal abnormality detection model. However, the error involved in diagnosis was not focused.

Motivated by the above said issues and to address on the aspects like precision, recall, training time and accuracy, in this work a method called, Multi-regressive Splines and Quantum Correlated (MS-QC) prenatal diagnosis in the identification of chromosomal abnormalities is designed. The elaborate description is provided in the following sections.

3. Methodology

One of the prominent facilitations in medical genetics was the evolution of prenatal diagnostic mechanisms that utilize several invasive and non-invasive mechanisms. The objective of prenatal diagnosis is to impart information of the genetic abnormalities of the fetus even at the early stage for the termination of pregnancy to be possible and to circumvent the children birth with genetic disorders. Motivated by this for accurate and precise identification of chromosomal abnormalities, in this work a method called, Multi-regressive Splines and Quantum Correlated (MS-QC) prenatal diagnosis is proposed. Figure 1 shows the structure of Multi-regressive Splines and Quantum Correlated (MS-QC) prenatal diagnosis method.

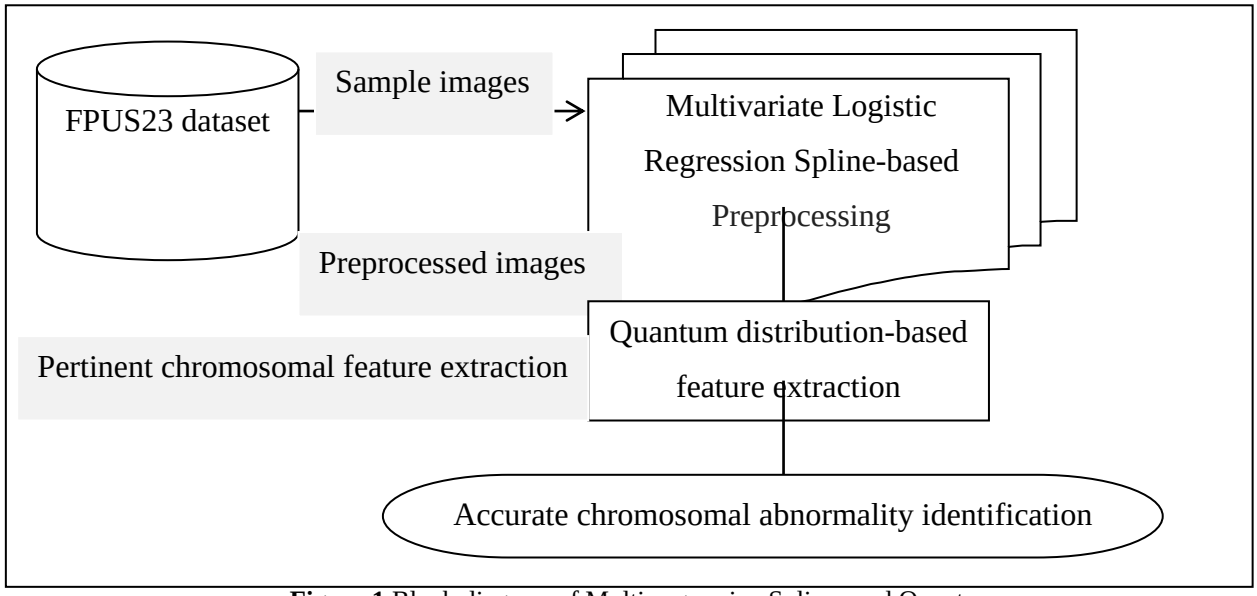

Figure 1 Block diagram of Multi-regressive Splines and Quantum Correlated (MS-QC) method

As shown in the above figure the proposed MS-QC method for identifying chromosomal abnormalities with respect to prenatal diagnosis, the entire process is split into two sections, namely, preprocessing and feature extraction. With the raw sample images obtained from FPUS23 dataset the images are subjected to preprocessing and feature extraction to prenatal diagnosis for chromosomal disorders. First, the raw images are subjected to Multivariate Logistic Regression Spline-based Preprocessing model by splitting the domain of image (i.e., brain's Biparietal Diameter (BPD), estimate the Abdominal Circumference (AC), estimates the fetus' Femur Length (FL)) into adjoining regions and fitting a polynomial to each region (i.e., diagnostic plane) separately with the purpose of identifying the true underlying structure by removing the noisy background elements.

Second with the preprocessed data as input, Quantum Distribution-based Feature extraction algorithm is applied with the intent for extracting the most pertinent chromosomal features from fetal cells. The elaborate description of the proposed MS-QC method is provided in the following sections.

3.1 Dataset description extracting the most pertinent chromosomal features

The FPUS23 dataset employed in our work has been utilized in training sonographers to assess the fetus development and condition. In this work, a 23-week and 10 to 12 week old phantom as a mid-pregnancy scan is imparted to check for fetus anomalies. The fetus model includes full skeletal structure to assess the fetus' anatomy (like head, arms legs, and abdomen). The probe is positioned on the phantom abdomen surface and navigated to the diagnostic planes that can be used for the measurement of brain's Biparietal Diameter (BPD), estimate the Abdominal Circumference (AC), estimates the fetus' Femur Length (FL).

Also the sequences are labeled as a correct diagnostic plane for one of three biometric parameters (BPD, AC, FL) or as a non-diagnostic plane. This is due to the reason that the number of data samples acquired for each of the diagnostic planes is smaller than the non-diagnostic plane output class. As a result the samples were augmented to ensure equal representation of data across all classes. In addition after the acquisition of several frames at diagnostic plane more information are obtained to obtain the head, abdomen, arms, and legs, for capturing the fetal anatomies.

Moreover, the phantom abdomen was also rotated and positioned in four probable orientations (i.e., head up view front 'huvf', head up view back 'huvb', head down view front 'hdvf'and head down view back 'hdvb') respectively. Also, the images are tagged with the anatomies, such as heads, arms, legs, and abdomen and finally, the images are in due course comprehensive annotated with boxes representing to ascertain their bounds and clearly evaluate biometric parameters later, such as BPD, AC, FL, at the correct diagnostic plane. On the other hand, images that do not keep in check any relevant fetus information are not annotated. Table 1

given below lists the FPUS23 dataset overview and the number of input samples present in each class i.e., Diagnostic Plane, Fetus Orientation, Fetus Anatomy and Anatomy Bounds employing box annotation respectively.

Table 1 FPUS23 dataset overview

Total (5265)				
Diagnostic plane	AC Plane (1386)	BPD Plane (1280)	FL Plane (1281)	No Plane (1318)
Total (15728)				
Fetus orientation	Hdvb (3757)	Hdvf (3235)	Huvb (3980)	Huvf (4756)
Total (9317)				
Fetus anatomy	Head (3003)	Arms (1629)	Legs (2159)	Abdomen (2526)
Total (9455)				
Anatomy bounds	Head (4370)	Arms (4853)	Legs (4572)	Abdomen (6435)

As provided in the above table, the dataset employed in our work, is split in the ratio of '$8:1:1$' with respect to training, validation and testing for three cases (i.e., diagnostic plane, fetus orientation and fetus anatomy). Finally in case of the anatomy bounds we use 80% for training and 20% for validating.

3.2 Multivariate Logistic Regression Spline-based preprocessing model

Prenatal diagnosis makes use of different types of materials and methods to ascertain the health and condition of an unborn fetus. In the absence of proficiency acquired prenatal diagnosis, there could be an unexpected result for the mother or the fetus or both. Chromosomal abnormalities or malformations are disarrangements in the normal chromosomal content and are the most prevalent source of genetic diseases in humans. Despite certain chromosomal abnormalities do not cause disease in carriers although they may result in exorbitant proportions of chromosomal disorder in child.

In this work a Multivariate Logistic Regression Spline function model is proposed to focus on the error and bias involved in analyzing the risk factors by taking into consideration the intensity gradient and obtaining distinct fetus orientations by means of image gradients is designed. Initially, with the input obtained from the prenatal cytogenetic Data set a machine learning based Multivariate Logistic Regression Spline function is applied that can capture complicated and multifaceted nature

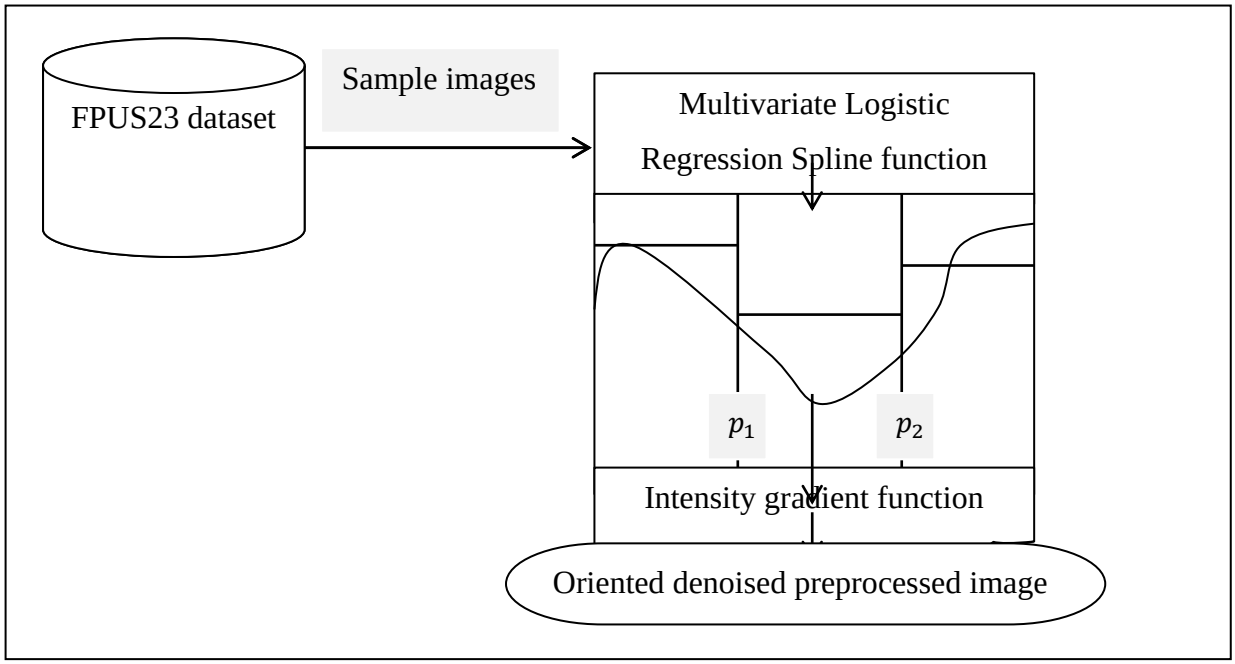

Figure 2 Block diagram of Multivariate Logistic Regression Spline-based preprocessing

The Multivariate Logistic Regression Spline function by including multiple independent variables can account for more characteristics that influence the dependent variable and hence minimize the overall error and bias involved in analyzing the risk factors for Prenatal Diagnosis in the Identification of the Chromosomal Abnormalities. Figure 2 shows the block diagram of Multivariate Logistic Regression Spline-based preprocessing model.

As illustrated in the above figure, the raw sample images obtained from the FPUS23 dataset are initially subjected to Multivariate Logistic Regression Spline function by including multiple independent variables accounting for more characteristics and hence influencing the dependent variable by introducing two different points 'p_1' and 'p_2'. This in turn would result in the true underlying structure. Next, the obtained underlying structured is provided as input to the intensity gradient function with the purpose of acquiring four different fetus orientations for further processing.

Let us consider sample images obtained from FPUS23 dataset from the corresponding diagnostic plane, involving four different types of images, namely, AC Plane, FL Plane, BPD plane and NO plane respectively. With the sample images provided as input the objective here remains in minimizing the overall training time and boost the accuracy. With this objective the sample images are initially subjected to Multivariate Logistic Regression Spline function that with the aid of piecewise constant fit eliminates the noise by including multiple independent variables for more characteristics that influence the dependent variable. Followed by which, to produce different orientations of the fetus the noise eliminated images are subjected to image gradient to produce the output resultant images to focus on the accuracy aspects. To start with the aid of Multivariate Logistic Regression we modeled our function in the form as given below.

$$f(SI) = \sum_{m=1,i=1}^{M,n} \alpha_m SI_i \qquad (1)$$

From the above equation (1) '$f(SI)$' the multivariate (i.e., here represents the sample images in the form of either NO_PLANE or FL_PLANE or AC_PLANE or BPD_PLANE) function is initially evolved by means of a weight 'α_m' with each weight here representing the distinct diagnostic plane.

Here '$\alpha_m = 1\ [NO\_PLANE], 2\ [FL\_PLANE], 3[AC\_PLANE], 4[BPD\_PLANE]$' respectively.

Followed by which dimensionality reduction is performed by retaining the most significant features and eliminating the unnecessary features removes noise in the image data. This dimensionality reduction for the

corresponding diagnostic plane is mathematically formulated as given below.

$$f(SI)[DR] = \sum_{m=1}^{M} \alpha_m HD_m(SI) \quad (2)$$

$$HD_m(SI): R^P \rightarrow R \quad (3)$$

From the above equations (2) and (3), the resultant function with dimensionality reduction '$f(SI)[DR]$' is formulated by mapping from higher dimensional space 'R^P' to lower dimensional space 'R' respectively via mapping function ' $HD_m(SI)$ '. In addition to dimensionality reduction in our work focus is also made on concentrating fetus orientation employing intensity gradient. To obtain different orientation of fetus (i.e., huvb, huvf, hdvb, hdvf) intensity gradient is applied to the dimensionality reduced resultant features by observing a sharp different in the values of intensity to adjacent pixels. This difference in intensity values denotes the transition between one image features to another image feature, to be more specific performing the transition from the foreground of resultant image feature to the background of resultant feature therefore generating four different orientations. The sample images are initially split into three regions separated at points 'p_1' and 'p_2'. The points 'p_1' and 'p_2' are selected in such a manner that it is equidistant to each other with respect to the overall sample images. Then the model fit employing the four basic functions for each of the orientation of fetus (i.e., huvb, huvf, hdvb, hdvf) is mathematically formulated as given below.

$$HD_1(SI) = I(SI < p_1) \quad (4)$$

$$HD_2 = I(p_1 \leq SI < p_2) \quad (5)$$

$$HD_3 = I(p_2 \leq SI)) \quad (6)$$

$$HD_4(SI) = HD_1(SI)SI \quad (7)$$

From the above equations (4), (5), (6) and (7) the true underlying structure using piecewise constant function is obtained for each sample image 'SI' into adjoining regions. Followed by which the gradient of high dimensionality mapped resultant feature images for each true underlying structure is obtained as given below.

$$\nabla f[HD_1(SI)] = \begin{bmatrix} g_p[HD_1(SI)] \\ g_q[HD_1(SI)] \end{bmatrix} = \begin{bmatrix} \frac{\partial f}{\partial p}[HD_1(SI)] \\ \frac{\partial f}{\partial q}[HD_1(SI)] \end{bmatrix} \quad (8)$$

From the above equation (8), the gradient resultant image feature vector for high dimensionality mapped results at '$HD_1(SI)$' is obtained. In a similar manner the gradient resultant image feature vector for high dimensionality mapped results at '$HD_2(SI)$', '$HD_3(SI)$' and ' $HD_4(SI)$ ' are arrived at. The pseudo code representation of Multivariate Logistic Regression Spline-based preprocessing is given below.

Input: Dataset ' DS ', Sample Images ' $SI = \{SI_1, SI_2, \dots, SI_n\}$ ', NO_PLANE, FL_PLANE, AC_PLANE, BPD_PLANE $\in SI$
Output: Computationally-efficient preprocessed images
Step 1: **Initialize** 'n' Step 2: **Begin** Step 3: **For**each Dataset 'DS' with Sample Images 'SI' **//Dimensionality reduction** Step 4: Formulate Multivariate Logistic Regression for sample images involved in simulation as given in equation (1) Step 5: Perform dimensionality reduction by mapping as given in equations (2) and (3) **//Fetus orientation using intensity gradient** Step 6: Evaluate true underlying structure using piecewise constant function as given in equations (4), (5), (6) and (7) Step 7: Evaluate gradient of high dimensionality mapped resultant feature images for each true underlying structure as given in equation (8)

Step 8: Return orientation results '$\nabla f[HD_1(SI)]$', '$\nabla f[HD_2(SI)]$', '$\nabla f[HD_3(SI)]$' and '$\nabla f[HD_4(SI)]$'
Step 9: **End for**
Step 10: End

Algorithm 1 Multivariate Logistic Regression Spline-based preprocessing

As given in the above algorithm with the objective of minimizing the training time and improve the overall accuracy, Multivariate Logistic Regression Spline function and intensity gradient function is applied. Initially the raw sample images provided as input is subjected to Multivariate Logistic Regression Spline function that by removing the noise finds the true underlying structure for further processing. Next, the obtained true underlying structure as input is subjected to intensity gradient for generating four distinct orientation results. As a result multiple independent variables account for more characteristics that in turn influence dependent variable, therefore reducing the overall error and bias involved in analyzing the risk factors for Prenatal Diagnosis in chromosomal abnormality identification.

3.3 Quantum Distribution-based Feature extraction

In this section with the computationally-efficient preprocessed images where multiple independent variables account for more characteristics that in turn influence dependent variable in analyzing the risk factors for Prenatal Diagnosis in chromosomal abnormality identification using Quantum Distribution-based Feature extraction model is applied. To be more specific we propose a new correlation based quantum distribution model for precise target measurement that in turn can be further realized by virtue of quantum correlation characteristics. Both variants adaptations (i.e., with respect to x-coordinates and y-coordinates) are analyzed using feasible binary states. The multi-objective optimization (i.e., variants adaptations) is employed for considering two types of variants (i.e., with respect to x-coordinates and y-coordinates) concurrently without redundancy.

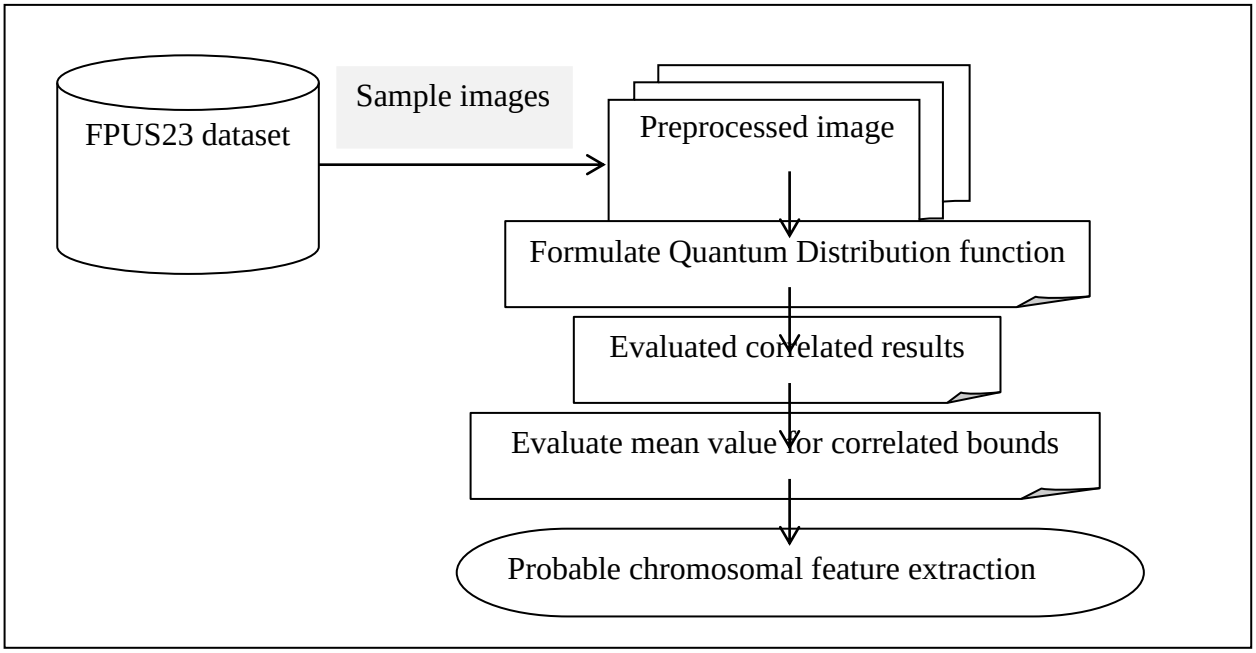

Figure 3 Block diagram of Quantum Distribution-based Feature extraction

To overcome high computational complexity issue with improved precision and recall, problem, we calculate the features-class correlations for extracting the most pertinent chromosomal features from fetal cells instead of straightly obtaining for optimal subset of variants. The proposed Quantum Distribution-based Feature extraction model in our work is applied for extracting the most anticipated chromosomal feature from fetal cells. With this more precise information of fetal disorders indicators can be obtained. Figure 3 shows the block diagram of Quantum Distribution-based Feature extraction model.

As illustrated in the above figure, a Q-bit is the smallest unit of information held in a two-state quantum computer, wither either '100' or '000' state and is represented as given below.

$$|\psi_{state}| = \gamma|0> + \beta|1> \tag{9}$$

From the above equation (), '$|\psi_{state}|$' where 'γ' and 'β' represents the complex integers that describe the probability volumes of the amplitudes of the corresponding states. The probabilities of detecting the Q-bits in the '1' and '0' states are given by '$|\gamma^2|$' and '$|\beta^2|$' respectively. Here, '$|\gamma^2|$' denotes the probability that that qubit will be in the '0' state and on the other hand, '$|\beta^2|$' represents the prospective that it will be in the '1' state.

A single anatomy bound is encoded and denoted by single Q-bits. Each qubit will probably in the '1' or '0' or a superposition of the two states. The data produced by this anatomy bound are not fixed, but they are probable. As a consequence, the straight forward alternative is to utilize the binary coding process in the anatomy bound to code these multistate qubits.

To be more specific, multi-qubits are utilized in denoting the multi-state operator node and is mathematically formulated as given below.

$$Temp_j^t = \begin{bmatrix} \gamma_1 & \gamma_2 & \dots & \gamma_m \\ \beta_1 & \beta_2 & \dots & \beta_m \end{bmatrix}, |\gamma_1|^2 + |\beta_2|^2 = 1 \tag{10}$$

From the above equation (10), '$Temp_j^t$' represents the chromosome of the '$t-th$' generation and the '$j-th$' individual sample with 'm' denoting the Q-bits number. The utilization of Q-bits encoding permits a single individual to influence the superposition of multiple states. Let us further assume with three Q-bits as given below.

$$|\psi_{state}| = \begin{bmatrix} \frac{1}{\sqrt{2}} & \frac{1}{\sqrt{2}} & \frac{1}{\sqrt{2}} \\ \frac{1}{\sqrt{2}} & -\frac{1}{\sqrt{2}} & \frac{\sqrt{3}}{2} \end{bmatrix} = \frac{1}{4}\left|000\right> + \frac{\sqrt{3}}{4}\left|001\right> - \frac{1}{4}\left|010\right> - \frac{\sqrt{3}}{4}\left|011\right> + \frac{1}{4}\left|100\right> + \frac{\sqrt{3}}{4}\left|101\right> - \frac{1}{4}\left|110\right> - \frac{\sqrt{3}}{4}\left|111\right> \quad (11)$$

From the above equation (11), this warrants the three-qubit mechanism for storing extracted features in two eight distinct states (i.e., huvb_head, huvf_head, huvb_abdomen, huvf_abdomen, huvb_arms, huvf_arms, huvb_legs, huvf_legs, hdvf_head, hdvb_head, hdvf_abdomen, hdvb_abdomen, hdvb_arms, hdvf_arms, hdvb_legs, hdvf_legs). Owing to the reason that qubit representation may denote superposition of states, stronger variety quality is said to exist than the conventional methods.

Only one Q-bits chromosome is necessitated to denote eight states, whereas in case of conventional modeling at the minimum eight chromosomes are necessitated. Next, the correlated results to evaluate distinct subsets (i.e., head, abdomen, arms and legs) are measured as given below.

$$FE = \rho PC(p,q) = \frac{\sum_i (p_i - \mu_p)(q_i - \mu_q)}{\sqrt{\sum_i (p_i - \mu_p)^2}\sqrt{\sum_i (q_i - \mu_q)^2}} \quad (12)$$

From the above equation results (12), 'μ_p' and 'μ_q' denote the mean value of image vector 'p' and 'q' respectively. The choice of 'PC' value is '$[-1, +1]$'. The closer to '$+1$', the higher is positive correlation and on contrary the closer to '-1' the higher is negative correlation. Finally, for the anatomy bounds, mean average precision and mean average recall is measured to not label a negative sample as positive and to identify all positive instances of each class respectively. This is mathematically represented as given below.

$$\mu AP = \frac{1}{N}\frac{\sum_{i=1}^{N} pixel_{i,i}}{\sum_{i=0}^{N}\sum_{j=1}^{N} pixel_{i,i} + pixel_{i,j}} \quad (13)$$

$$\mu AR = \frac{1}{N}\frac{\sum_{i=1}^{N} pixel_{i,i}}{\sum_{i=0}^{N}\sum_{j=1}^{N} pixel_{i,i} + pixel_{j,i}} \quad (14)$$

From the above equations (13) and (14), with the total number of output classes being 'N', '$pixel_{i,i}$' denotes the pixels classified as class 'i' and labeled as class 'i'. In a similar manner, '$pixel_{i,j}$' and '$pixel_{j,i}$' denotes the pixels classified as class 'i' and labeled as class 'i. In this manner, most probable chromosomal features are obtained. The pseudo code representation of Quantum Distribution-based Feature extraction is given below.

Input: Dataset 'DS', Sample Images '$SI = \{SI_1, SI_2, \ldots, SI_n\}$', NO_PLANE, FL_PLANE, AC_PLANE, BPD_PLANE $\in SI$
Output: Precise fetal disorder indicators
Step 1: **Initialize**'n', orientation results '$\nabla f[HD_1(SI)]$', '$\nabla f[HD_2(SI)]$', '$\nabla f[HD_3(SI)]$' and '$\nabla f[HD_4(SI)]$'
Step 2: **Begin**
Step 3: For each Dataset 'DS' with Sample Images 'SI' and orientation results '$\nabla f[HD_1(SI)]$', '$\nabla f[HD_2(SI)]$', '$\nabla f[HD_3(SI)]$' and '$\nabla f[HD_4(SI)]$'
Step 4: Formulate Q-bit (i.e., orientation results) held in a two-state quantum computer as given in equation (9)
Step 5: Evaluate chromosome of the '$t-th$' generation and the '$j-th$' individual sample as given in equation (10)
Step 6: Measure three-qubit mechanism for storing extracted features as given in equation (11)
Step 7: Measure pearson correlation to measure distinct subsets as given in equation (12)
Step 8: Evaluate anatomy bounds as given in equations (13) and (14)
Step 9: **Return** most probable chromosomal feature extraction 'FE'
Step 10: **End for**
Step 11: **End**

Algorithm 2 Quantum Distribution-based Feature extraction

As given in the above algorithm, the concept of feasible binary states correlation based feature selection is proposed to extract all the pertinent chromosomal features from fetal cells related with one of the opposing features while keeping away from redundancy using multi-objective optimization. The proposed Quantum Distribution-based Feature extraction model is applied to extract chromosomal abnormalities associated variants. Followed by which pears on correlation function are applied to measure those are highly correlated for extracting the most pertinent chromosomal features from fetal cells. Finally the anatomy bounds results are

determined using the average precision and recall to obtain precise information of fetal disorders indicators.

4. Experimental Setup

This section discusses the parametric analysis for proposed Prenatal Diagnosis in the identification of chromosomal abnormalities with the aid of fetus dataset (i.e., FPUS23) images obtained from https://github.com/bharathprabakaran/FPUS23.

Simulations were performed in Python high-level programming language with Intel Core i7 processor with a 4 GB graphic card, a 64-bit operating system at 1.80 GHz, and 16 GB RAM. Parametric metrics considered for the evaluation of Prenatal Diagnosis in the identification of chromosomal abnormalities consists of precision, recall, training time and diagnosis accuracy. Fair comparison analysis was made using the three methods, where same dataset was used for validating the performance metrics. Figure 4 given below shows the sample training images in each category of diagnostic plane and testing images in each category of diagnostic plane respectively.

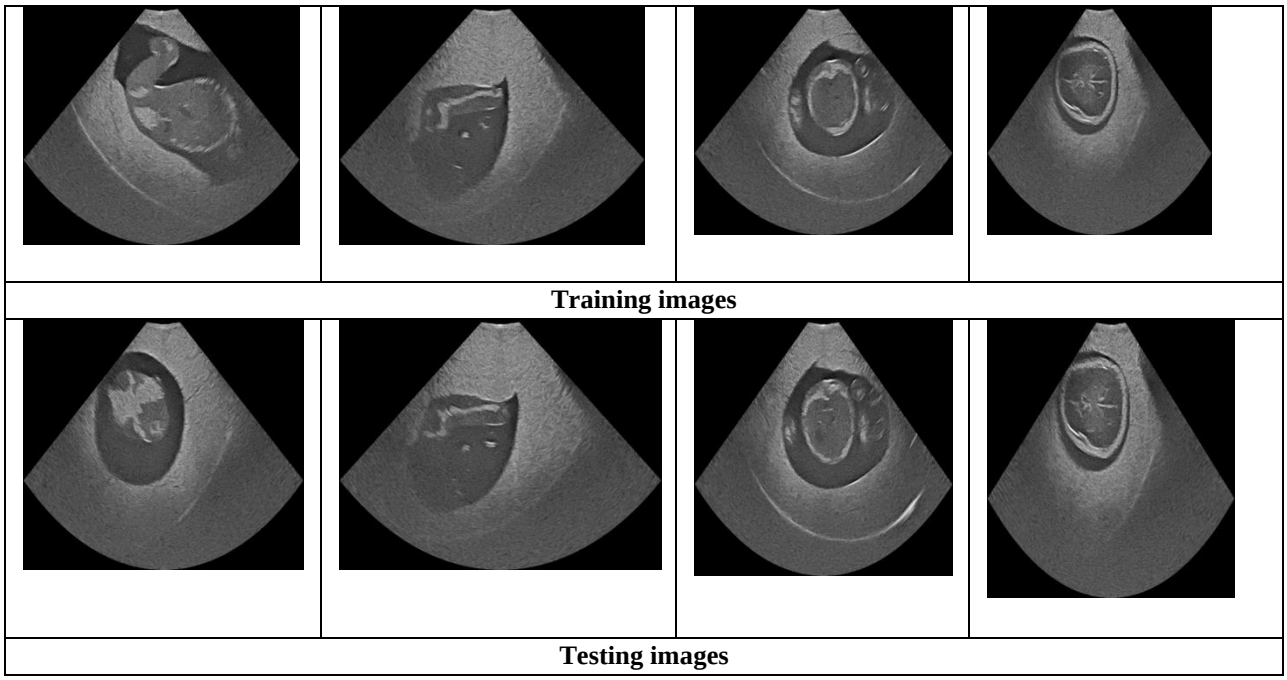

Figure 4 Training images (a) NO_PLANE, FL_PLANE, AC_PLANE and BPD_PLANE (23 week)

The diagnostic performance of the proposed MS-QC method and existing methods deep convolutional neural networks (DCNN) [1] and feature fusion classifier with dynamic weights (FFCDW) [2] are tested under the precision, recall, accuracy and training time factors. The corresponding formulae of these measures are provided in the discussion section.

5. Implementation Details

In this study, a method called Multi-regressive Splines and Quantum Correlated (MS-QC) prenatal diagnosis in the identification of chromosomal abnormalities focusing on the precision, recall, accuracy and training time is designed.

- The MS-QC method comprises of two parts, namely, preprocessing the data and pertinent chromosomal feature extraction preprocessing signals, for prenatal diagnosis in the identification of chromosomal abnormalities.
- The MS-QC method is compared with two existing methods DCNN [1] and FFCDW [2] employing FPUS23 images dataset to validate the results.
- Initially, raw fetus images from FPUS23 images dataset were validated and analyzed to obtain computationally efficient preprocessed images using Multivariate Logistic Regression Spline-based preprocessing algorithm.
- Second feature extraction process was carried out with the computationally efficient EEG signals. In the feature extraction process the preprocessed EEG signals were subjected to Quantum Distribution function for extracting precise fetal disorder indicators. Here, both three-qubit mechanism and pears on correlation function were employed for

preprocessed images to extract probable chromosomal features. According to the above implementation patterns, four different evaluation metrics namely, precision, recall, training time and accuracy are discussed in the next section using table and graphical representations.

6. Discussion

6.1 Scenario 1: Precision

The first metric paramount for prenatal diagnosis in the identification of chromosomal abnormalities is precision. Precision gives the number of chromosomal abnormalities correctly identified among the detected chromosomal abnormalities, which is mathematically stated as given below.

$$Pre = \frac{TP}{TP+FP} \quad (15)$$

From the above equation (15), the precision rate 'Pre' is measured using the true positive rate 'TP' (i.e., samples with chromosomal abnormalities extracted as chromosomal abnormalities) and false positive rate 'FP' (i.e., samples with normal chromosome extracted as chromosomal abnormalities) respectively.

Table 2 Tabulation of precision

Sample images	Precision					
	23 week			**10 to 12 week**		
	MS-QC	**DCNN**	**FFCDW**	**MS-QC**	**DCNN**	**FFCDW**
1000	0.95	0.9	0.87	0.85	0.8	0.75
2000	0.93	0.89	0.85	0.81	0.78	0.73
3000	0.91	0.86	0.82	0.8	0.75	0.71
4000	0.9	0.85	0.81	0.82	0.74	0.7
5000	0.9	0.84	0.8	0.83	0.76	0.75
6000	0.93	0.88	0.84	0.84	0.77	0.73
7000	0.94	0.89	0.85	0. 82	0.78	0.74
8000	0.92	0.85	0.8	0.83	0.75	0.7
9000	0.91	0.86	0.81	0.81	0.76	0.72
10000	0.93	0.87	0.82	0.84	0.79	0.74

Table 2 given below lists the precision rate using the three methods, MS-QC, DCNN [1] and FFCDW [2].

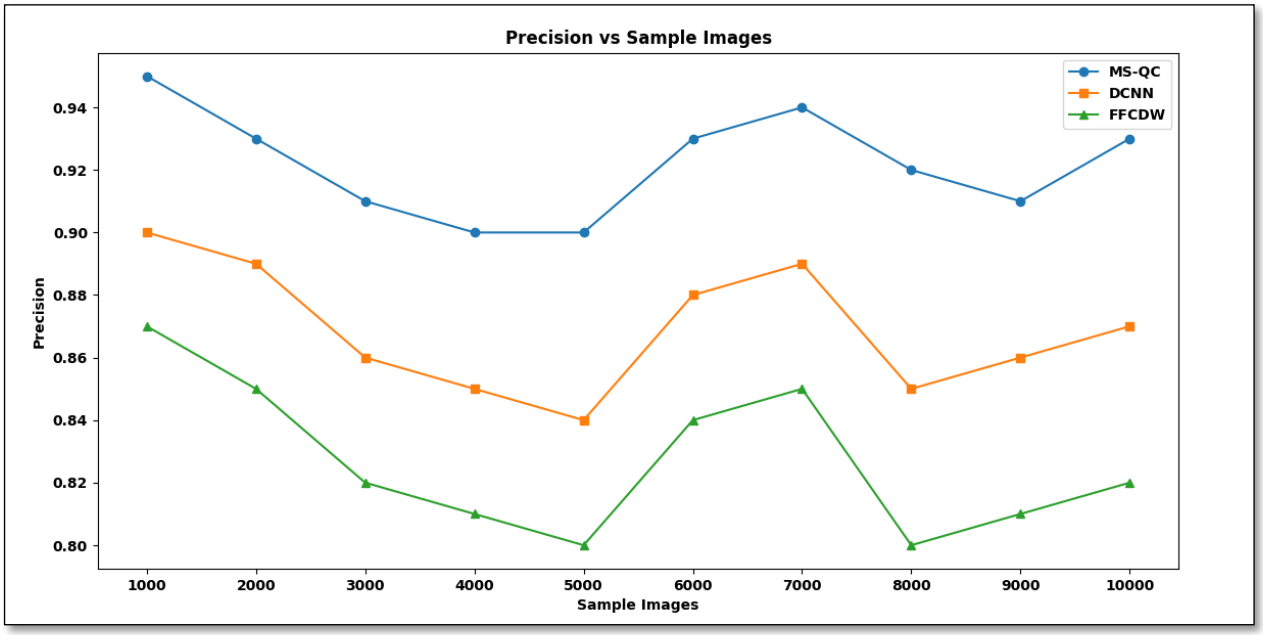

Figure 5 Graphical representation of precision for 23 week

Figure 6 Graphical representation of precision for 10 to 12 week

Figure 5 and 6 given above shows the performance measure of precision in the y-axis with 10000 distinct sample images provided as input in the x-axis. From the above figure the precision rate is not inversely or directly proportional to the number of sample images provided as input.

Also from the above simulations conducted for 1000 sample images, with 300 (chromosomal abnormalities images) and with 700 (chromosomal normal images) observed, the true positive rate and false positive rate using the proposed MS-QC method was found to be 258 and 15, in a similar manner, the true positive rate and false positive rate using the DCNN [1] was found to be 270 and 30, finally, the true positive rate and false positive rate using the FFCDW [2] was observed to be 263 and 37. With this the overall precision using the three methods were found to be 0.95, 0.90 and 0.87 respectively. From this simulation setup the overall precision using the proposed MS-QC method was observed to be comparatively better than [1] and [2]. The reason for improvement was due to the application of Quantum Distribution-based Feature extraction algorithm. By applying this algorithm correlation based quantum distribution for precise target measurement was further realized by virtue of quantum correlation characteristics. By using this Quantum Distribution functions both variants adaptations were analyzed using feasible binary states via multi-objective optimization. This in turn considered two types of variants (i.e., with respect to x-coordinates and y-coordinates) concurrently without redundancy. With this false positive rate was reduced significantly and therefore improved the 23 week of precision rate using proposed MS-QC method by 6% compared to [1] and 12% compared to [2] as well as 10 to 12 week of precision rate using proposed MS-QC method by 7% compared to [1] and 13% compared to [2]

6.2 Scenario 2: Recall

Second in this section the recall rate is measured. The recall rate on the other hand, gives the ratio of chromosomal abnormalities correctly identified among the total detected chromosomal abnormality samples, which is mathematically represented as given below.

$$Rec = \frac{TP}{TP+FN} \tag{16}$$

From the above equation (16), the recall rate 'Rec' is measured based on the true positive rate 'TP' (i.e., samples with chromosomal abnormalities extracted as chromosomal abnormalities) and the false negative rate 'FN' (i.e., samples with abnormal chromosome extracted as normal chromosomes) respectively. Table 3 given below lists the recall rate using the three methods, MS-QC, DCNN [1] and FFCDW [2].

Table 3 Tabulation of recall

Sample images	Recall					
	23 week			10 to 12 week		
	MS-QC	DCNN	FFCDW	MS-QC	DCNN	FFCDW
1000	0.85	0.8	0.76	0.75	0.7	0.68
2000	0.83	0.78	0.73	0.72	0.69	0.66
3000	0.81	0.76	0.71	0.7	0.67	0.65
4000	0.8	0.73	0.68	0.7	0.66	0.63
5000	0.8	0.73	0.68	0.72	0.64	0.61
6000	0.81	0.74	0.69	0.73	0.66	0.64
7000	0.82	0.75	0.7	0.74	0.65	0.64
8000	0.83	0.76	0.71	0.72	0.63	0.62
9000	0.82	0.73	0.68	0.74	0.62	0.61
10000	0.83	0.74	0.69	0.73	0.66	0.63

Figure 7 Graphical representation of recall for 23 week

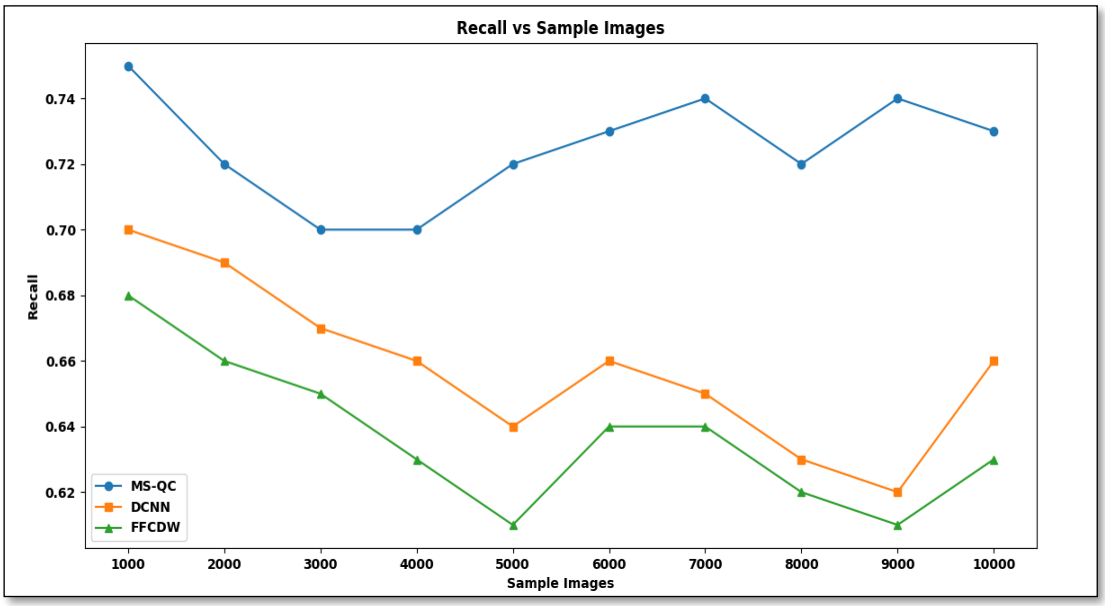

Figure 7 Graphical representation of recall for 10 to 12 week

Figure 7 and 8 given above illustrates the pictorial representation of recall using the three methods, MS-QC, DCNN [1] and FFCDW [2] respectively. Also from the above figure though the recall rate using the first four set of samples saw a decreasing trend however increased for the next four set of samples, therefore corroborating the objective of improving the overall recall.

Also from the simulation with 1000 sample images provided as input, the true positive rate and false negative rate using the proposed MS-QC method was found to be 285 and 50, the true positive rate and false negative rate using the DCNN [1] was observed to be 270 and 65, similarly the true positive rate and false negative rate using the FFCDW [2] was found to be 263 and 80. With this the overall recall rate using the three methods were found to be 0.85, 0.80 and 0.76 respectively. From this simulation results the overall recall rate using the proposed MS-QC method was observed to be comparatively better than [1] and [2]. The reason was by applying the of Quantum Distribution-based Feature extraction model in addition to multi-qubits being utilized in denoting the multi-state operator node correlated results to evaluate distinct subsets (i.e., head, abdomen, arms and legs) using Pearson correlation was used. This in turn extracted highly correlated and most pertinent chromosomal features from fetal cells. As a result the 23 week of recall rate using the proposed MS-QC method was found to be comparatively higher by 9% compared to [1] and 17% compared to [2] as well as 10 to 12 week of recall rate using proposed MS-QC method by 10% compared to [1] and 14% compared to [2]

6.3 Scenario 3: Accuracy

In this section the accuracy involved for prenatal diagnosis in identifying chromosomal abnormalities is measured. The accuracy rate is mathematically formulated as given below.

$$Acc = \frac{TP+TN}{TP+TN+FP+FN} \quad (17)$$

From the above equation (17) accuracy 'Acc' is measured by taking into considerations the true positive rate 'TP', true negative rate 'TN', false positive rate 'FP' and false negative rate 'FN' respectively. It is measured in terms of percentage (%).Table 4 given below lists the accuracy rate using the three methods, MS-QC, DCNN [1] and FFCDW [2].

Table 4 Tabulation of accuracy

Sample images	Accuracy (%)					
	23 week			10 to 12 week		
	MS-QC	DCNN	FFCDW	MS-QC	DCNN	FFCDW
1000	93.5	90.5	88.3	92	90	87
2000	91.45	85.35	84.15	90.65	84.72	83..89
3000	90.35	84.55	83.35	89.42	83.25	82.71
4000	88.65	82.55	81.35	87.31	81.55	81
5000	86.35	82.25	81.05	84.79	81.11	80.43
6000	86	80	79	84.11	79	78.59
7000	87.15	81.05	80	85.68	79.86	78.81
8000	88	82	81	87.20	80	79
9000	89.35	83.25	83.05	88.87	82.45	81.88
10000	90	84	83	89	83.76	82

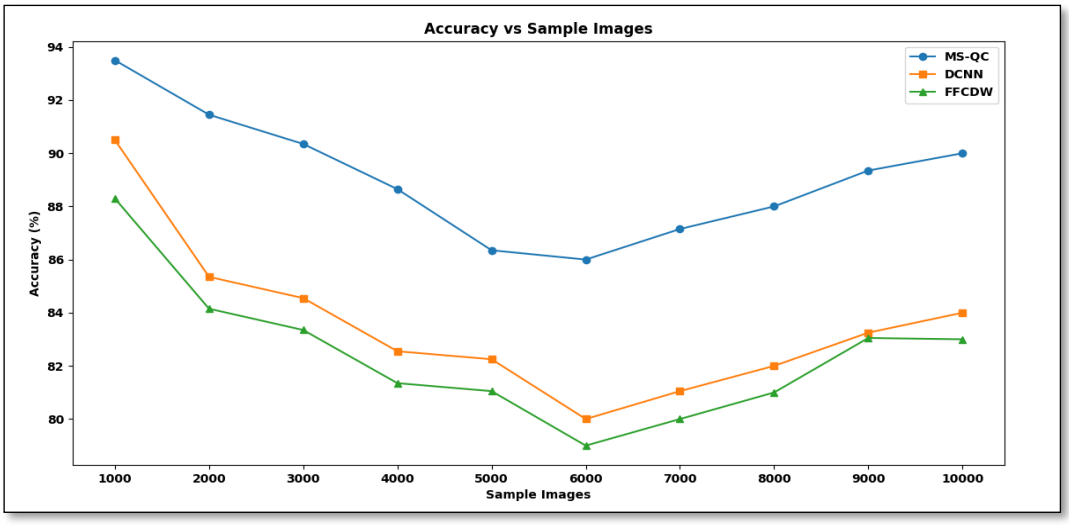

Figure 9 Graphical representation of accuracy for 23 week

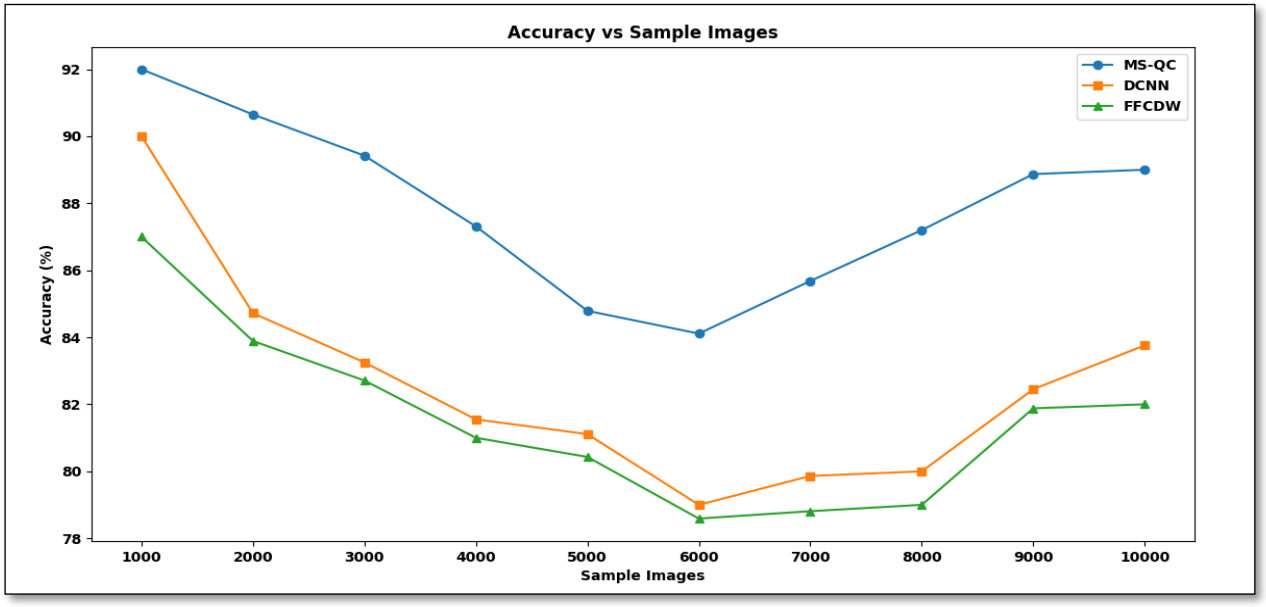

Figure 10 Graphical representation of accuracy for 10 to 12 week

Figure 9 and 10 given above shows the graphical representation of accuracy using the three methods. From the above figure, blue line denotes the accuracy of the proposed MS-QC method, orange line represents the accuracy of the DCNN [1] and finally, the green line denotes the accuracies of the FFCDW [2] method respectively. From the above figure the accuracy rate of the proposed MS-QC method is found to be comparatively higher than [1] and [2]. The reason for improvement in accuracy using the proposed MS-QC method was due to the application of Multivariate Logistic Regression Spline-based preprocessing algorithm. By applying this algorithm, initially dimensionality reduction was performed using the Multivariate Logistic Regression function for the sample images. This in turn reduced the false positive and false negative significantly, therefore improving the overall 23 week of accuracy using the proposed MS-QC method by 7% and 8% as well as 10 to 12 week obtained by 6% compared to [1] and 7% compared to [2]

6.4 Scenario 4: Training time

Finally in this section the training time or the time consumed in measuring prenatal diagnosis in the identification of chromosomal abnormalities is discussed. To be more specific, training time refers to the time consumed in extracting accurate and precise chromosomal abnormalities. Lower the training time involved in extracting accurate and precise chromosomal abnormalities more significant the method is said to be because irrelevant features can be discarded and serves for further identification of chromosomal abnormalities, therefore ensuring earlier remedial actions. The training time is mathematically formulated as given below.

$$TT = \sum_{i=1}^{n} SI_i * Time(FE) \quad (18)$$

From the above equation (18) training time 'TT' is measured by taking into considerations the samples 'SI_i' and the time consumed in extracting the features '$Time(FE)$'. It is measured in terms of milliseconds (ms). Table 5 given below lists the training time using the three methods, MS-QC, DCNN [1] and FFCDW [2].

Table 5 Tabulation of training time

Sample images	Training time (ms)					
	23 week			10 to 12 week		
	MS-QC	DCNN	FFCDW	MS-QC	DCNN	FFCDW
1000	15	23	28	20	26	30
2000	18.35	29.15	35.25	23.45	32.72	36.17
3000	23.55	35.25	45.35	26.15	37.63	47.28
4000	31.45	41.35	50	32.55	43.55	52.43
5000	35	48	61.35	37	49	63.77
6000	37	51	63	39	53	66
7000	38.25	60.35	78.25	40.82	61.73	80.13
8000	45	69.15	80	47	71.15	82
9000	53.15	75	95.35	55.34	77	67.55
10000	60	83.15	105.15	63	84	106.82

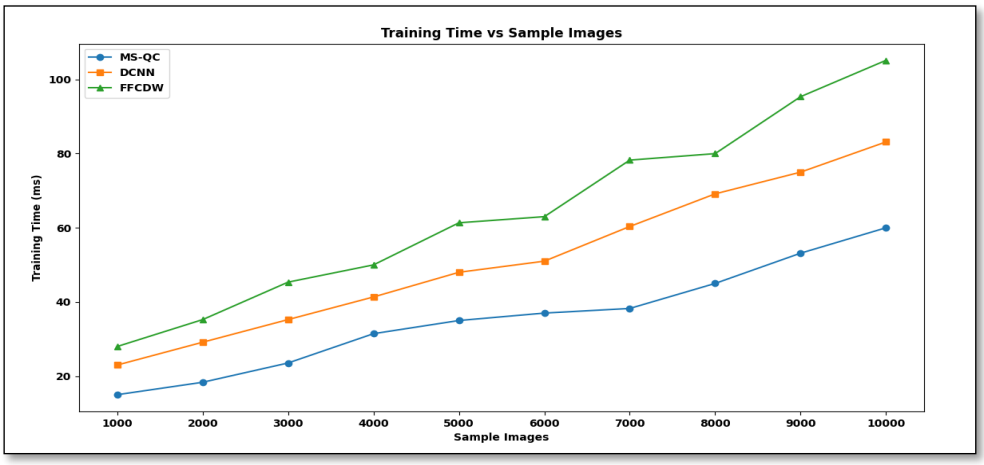

Figure 11 Graphical representation of training time for 23 week

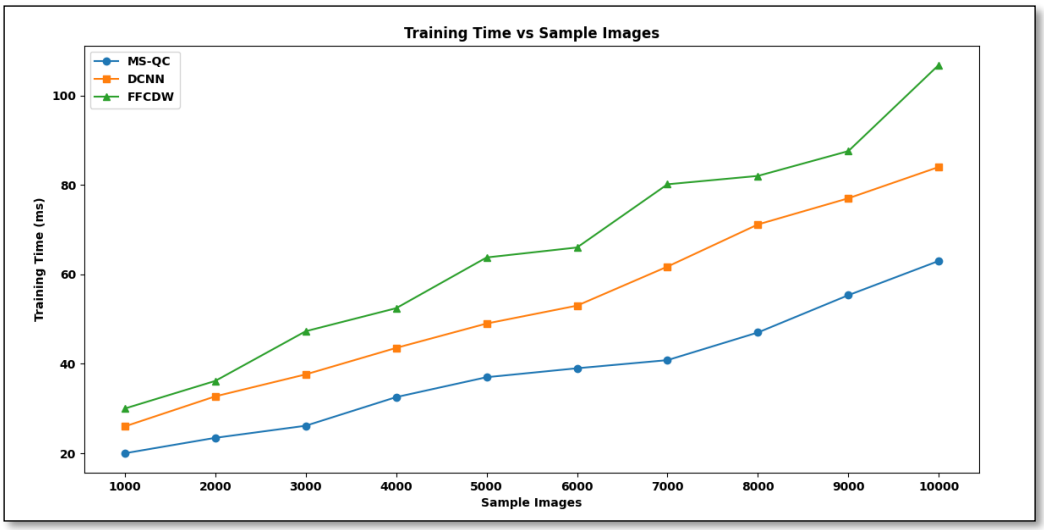

Figure 12 Graphical representation of training time for 10 to 12 week

Finally, figure 11 and 12 given above illustrates the graphical representation of training time. In the x-axis 10000 sample images were provided as input and the training time was measured and plotted in the y-axis for three different methods. From the above figure an increase in sample size saw a subsequent increase in the training time also. This is because with the increase in the sample images also the frequency and size also increases therefore increasing the overall training time of all the three methods. However, simulations performed for 1000 sample images found the training time of MS-QC method to be 15ms, 23ms using [1] whereas 28ms using [2]. From this result it is inferred that the training time consumed using MS-QC method was observed to be comparatively better than [1] and [2]. The reason for the improvement was owing to the application of Multivariate Logistic Regression Spline-based preprocessing algorithm. By applying this algorithm true underlying structure were obtained using piecewise constant function and also gradient of high dimensionality mapped resultant feature images for each true underlying structure that in turn minimizes the time consume din extracting prominent features for prenatal diagnosis in identifying chromosomal abnormalities. With this the 23 week training time using the proposed MS-QC method was reduced by 31% and 45% as well as 10 to 12 week obtained by 28% and 38 % when compared to[1], [2] respectively.

7. Conclusion

It is known that perinatal mortality is caused by inherited anomalies of fetuses and many of these might be explained a condition caused by abnormalities in genes or chromosomes. Several detection mechanisms are said to exist using either invasive or non-invasive techniques for prenatal diagnosis. Also with the aid of machine learning techniques several chromosomal abnormalities are measured in the recent few years. In this work, Multi-regressive Splines and Quantum Correlated (MS-QC) prenatal diagnosis in the identification of chromosomal abnormalities is presented. To begin, Multivariate Logistic Regression Spline function was applied to the raw fetus images. This eventually leads to a decrease in the complexity. The Quantum Distribution function is then used to for extracting the most pertinent chromosomal features from fetal cells.

Experimental evaluation of proposed MS-QC method and existing methods are carried out using FPUS23 Dataset. The experimental results of MS-QC method improve chromosomal abnormal detection accuracy, precision and recall with minimum training time.

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