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STUDY ON PRENATAL SCREENING AND DIAGNOSIS FOR CHROMOSOMAL ABNORMALITIES IDENTIFICATION

Jesmitha S[1], Dr. Manjula T R[2] and Dr. C R Byra Reddy[3]

[1]Assistant Professor, Department of ECE, Bangalore Institute of Technology, Research Scholar, Department of ECE, School of Engineering, JAIN University, Bangalore, INDIA

[2]Associate Professor, Department of ECE, School of Engineering, JAIN University, Bangalore, INDIA

[3]Professor, Department of ECE, Bangalore Institute of Technology, Bangalore, INDIA

ABSTRACT

Pregnancy is a time of great anticipation and anxiety. Prenatal testing is carried out for women during pregnancy to find out whether fetus has possibility to be born with genetic condition or birth defect. The prenatal testing determined different options for pregnancy and delivery to improve the outlook for baby. Prenatal screening tests identify that baby have certain birth defects or genetic disorders. The screening tests include blood tests or ultrasound and prenatal cell-free DNA screening. Different prenatal testing is used depending on mother pregnancy trimester. Prenatal diagnosis used different screening methods to determine the health condition of an unborn fetus. It is important one to attain details about unborn baby health. Many researchers carried out their research on prenatal screening and diagnosis methods. But, the diagnosis accuracy was not enhanced and diagnosis time consumption was not minimized by existing methods. In order to address these issues, different existing data pre-processing and feature selection methods for prenatal screening and diagnosis is reviewed in this paper.

Keywords: Pregnancy, Prenatal testing, DNA screening, ultrasound, pregnancy trimester

1. INTRODUCTION

Prenatal screening tests are carried out during first or second trimester. Screening tests performed the definitive diagnosis. The healthcare provider discussed the options for diagnostic test to perform efficient diagnosis. A diagnostic test is carried out for performing efficient diagnosis. The diagnostic tests like chorionic vill us sampling and amniocentesis are carried out to reduce the risk of miscarriage. In first trimester falls in week 1-12, Second trimester falls in week 13-26, Third trimester falls in week 27-40.

During first trimester, the health care provider prescribed the blood test and ultrasound to compute the size of clear space in tissue at back of baby neck. Figure 1 illustrates the types of screening for Prenatal diagnosis

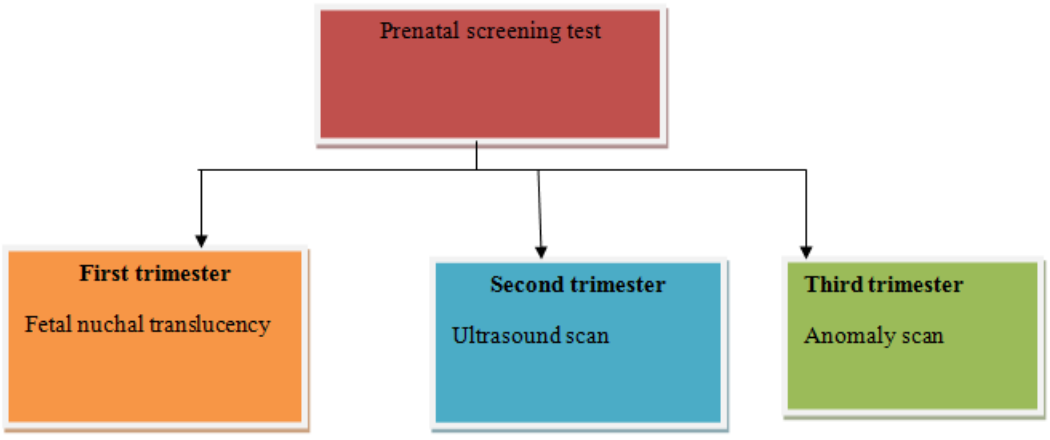

Figure 1 Types of screening for Prenatal diagnosis

From above figure 1 three types of trimester is explained namely first trimester, Second trimester, Third trimester. In Down syndrome, the nuchal translucency measurement is higher than the usual value. The health care provider prescribed blood test termed quad screen. The test determines the four substance levels in your blood. The results represent the risk of carrying baby with the chromosomal conditions like Down syndrome. The test identifies the neural tube defects and brain or spinal cord abnormalities. The blood test determined the fetal DNA in maternal bloodstream to screen for increased chance for particular chromosome issues like Down syndrome. The screening presented the information about the baby sex and Rh blood type. All pregnant women have option to experience the prenatal testing. When women age increased, chance of having baby with chromosomal abnormality gets increased. The Processing step of Prenatal screening is described is figure 2

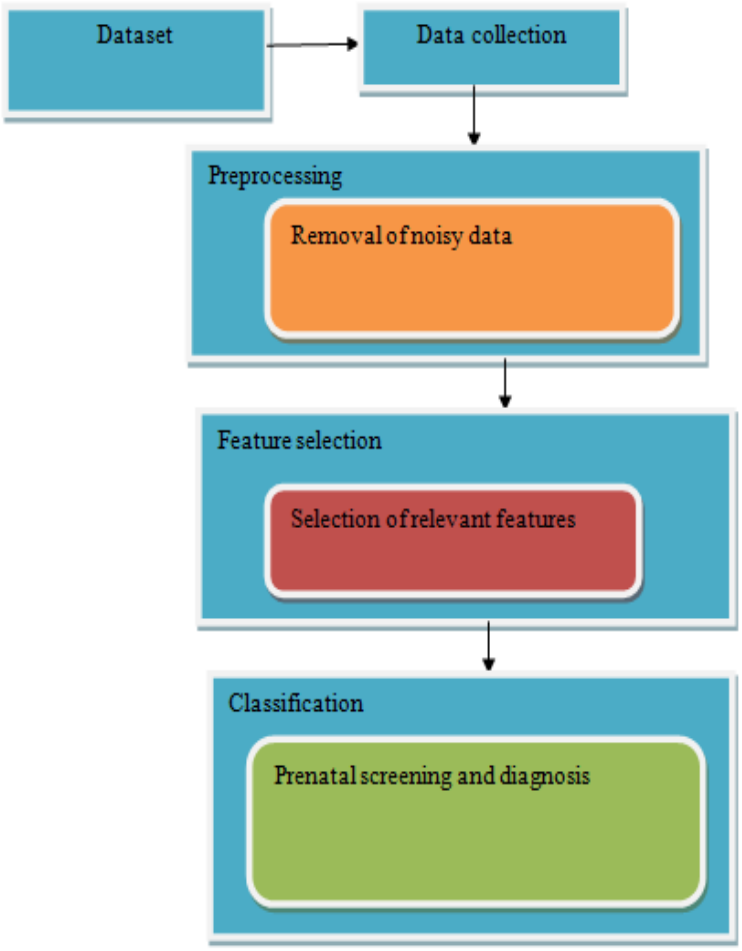

Figure 2 Processing step of Prenatal screening

In figure 2 , data are gathered from dataset and then preprocessing is performed to remove the noisy data. After that Feature selection is carried out to select the relevant features. Finally classification is performed for Prenatal screening and diagnosis. This paper is arranged as: Section 2 reviews the drawbacks on existing prenatal diagnosis and screening methods. Section 3 shows the study and analysis of existing prenatal diagnosis and screening methods. Section 4 identifies the possible comparison of prenatal diagnosis and screening with data pre-processing and feature selection techniques. Section 5 describes the limitations of existing prenatal diagnosis and screening techniques. The conclusion of paper is discussed in the section 6.

2. LITERATURE SURVEY

The naïve bayes classifier approach was introduced in [1] to forecast the person with the cardiovascular disease. The designed approach was employed to attain highest diagnosing accuracy. But, the complexity level was not minimized by designed approach. A genome-wide association study (GWAS) was carried out in [2] for finding genetic diseases by computing the correlation between trait differences and allele frequency of genetic markers. However, the space complexity was not minimized by designed study.

Chromosomal microarray analysis (CMA) was carried out in [3] through array comparative genomic hybridization. CMA provided additional results through sub-microscopic imbalances on G-banded chromosome preparation. But, the preprocessing time was not minimized by CMA. A machine learning-based automated diagnostic tool was discussed for Fetal Acidosis diagnosis. A 1D-CNN model was introduced in [4] to diagnose the Fetal Acidosis into healthy or unhealthy with higher accuracy. Though accuracy level was improved, the time consumption was not minimized by 1D-CNN model.

The hermeneutic phenomenological study was carried out in [5] to provide maternal experience of pregnancy after agenesis of corpus callosum diagnosis. The asynchronous and focus groups with the incidents during pregnancy were discovered. However, the computational cost was not minimized by designed study. But, the relevant features were not selected by using designed study. The visuospatial ability was discussed in [6] for studying individual variations. The individual results were attained from undergraduate respondents. However, the feature selection accuracy was not improved by designed ability.

IUGR carried out in [7] for antenatal diagnosis and treatment with delivery to minimize the risks. The presence of late IUGR was discussed through CTG findings. The feature elimination was carried out to increase the diagnosis accuracy with relevant features in predicting late IUGR using logistic regression and SVM. But, the feature selection accuracy was not improved by IUGR. The single-nucleotide polymorphism–based prenatal cell-free DNA screening was discussed in [8] for 22q11.2 deletion syndrome detection. Patients with single-nucleotide polymorphism prenatal cell-free DNA screening was employed for 22q11.2 deletion syndrome. However, the feature selection time was not minimized by prenatal cell-free DNA screening.

An evidence-based clinical practice principle was employed in [9] for noninvasive prenatal screening (NIPS) with pregnant individuals at risk for fetal trisomy. The designed principle determined the NIPS utility for finding the chromosomal disorders. However, the diagnosis time consumption was not reduced by evidence-based clinical practice principle. A population-based retrospective study was discussed in [10] with linked data. The multiple regression models were used for identifying the maternal age, living area and prenatal screenings. But, the error rate was not minimized by population-based retrospective study.

The prenatal diagnosis and perinatal findings was carried out in [11] in fetus and bilateral hyperechogenic kidneys on fetal ultrasound. The array comparative genomic hybridization (aCGH) analysis was discussed with DNA extracted from amniocytes. However, the computational complexity was not reduced by aCGH analysis. The prenatal diagnosis was carried out in [12] for pregnant woman with intellectual disability. A new heterozygous deletion in TBR1 gene was identified by premature stop. Prenatal diagnosis signified absence of mutation and family decided to preserve the pregnancy after genetic counseling. But, the false positive rate was not reduced by new heterozygous deletion.

A graph-based manifold regularization learning (MRL) framework (GMRLNet) was introduced in [13] for accurate PI diagnosis. An ultrasound and MFI images were considered as input with modality-shared information for efficient multimodal feature representation. A graph convolutional-based shared and specific transfer network (GSSTN) was employed to determine intra-modal feature associations.

A period-aware attention network (PA^2Net) was designed in [14] for FECG detection. FECG period-aware attention module (FPAM) was employed to eliminate the noise interference through modeling periods and signal

features. PA^{2}Net was employed to identify the FECG signals masked in noise. MECG period-aware attention module was created from FPAM by KL-divergence for mixed ECG signals.

An intelligent Fetal Imaging and Diagnosis (iFIND) was carried out in [15] with dual-probe ultrasound robot. The workflow comprised the abdominal surface mapping step to attain the non-parametric spline surface to position every individual joint. The motor synchronization method was employed to attain smooth motion towards target point.

A robotically assisted endoscopic process was carried out in [16] depending on steerable catheters to treat spina bifida defects. The process was carried out in fully remote way with the magnetic guidance and haptic controller. The deep convolutional neural network was designed in [17] to automatically recognize planes of fetal brains. The deep convolutional neural network (DCNN) and CNN-based domain transfer learning was carried out for abnormality detection in fetal brain image recognition.

A new regularized transfer learning approach was designed in [18] with network knowledge for facial recognition. The learning approach regularized the model behavior for adjusting the pre-trained weights. The designed approach increased the model generalizability under diverse training samples.

A single-shot fast spin echo T2-weighted imaging (SSFSE-T2WI) sequence was employed in [19] for constructing the image the fetal body. The fast imaging with steady-state acquisition (FIESTA) sequence was carried out to scan fetal head and ear in axial, coronal and sagittal planes. A 3D-FIESTA sequence with breath-hold was employed to compute the fetal external ear and EAC.

Eligible pediatric patients had different congenital anomalies and neurocognitive disabilities in [20]. The prenatal patients have one or more structural anomalies, disorders of fetal growth or fetal effusions. The prenatal presentation of fetal bradyarrhythmia with postnatal outcome was correlated in [21]. The retrospective analysis of case records was discussed. The fetuses with bradyarrhythmia beyond 11 weeks were discussed in the presentation.

A cascaded machine learning framework was introduced in [22] for down syndrome prediction depending on pre-judgment with isolation forest method, ensemble model through voting policy and final judgment through logistic regression. The designed framework was determined with maternal serum screening data set for different evaluation parameters. But, the diagnosis time was not minimized by designed framework.An automated framework was designed in [23] with model operator clinical workflow from first trimester fetal ultrasound scan videos. 2D+t convolutional neural network-based design was designed for video annotation with transfer learning and spatio-temporal (2D+t) modeling. The automated framework divided the ultrasound video into meaningful temporal segments depending on fetal anatomy detected.

A multiview classification and localization network (MCLN) and improved contrastive learning network (ICLN) was designed in [24] for multi-view fetal heart ultrasound image recognition and quality assessment. A multi-head enhanced self-attention mechanism was employed to build the classification network and identify fetal heart rate.

A deep learning-based brain age prediction model was employed in [25] for preterm infants through neonatal MRI. A particular dataset with MR images of preterm infants are collected. A DeepBrainNet was employed with backbone and transfer learning was used to increase the baseline model through collecting the knowledge from ImageNet dataset.

Deep convolutional neural network (DCNN) architecture was designed in [26] to automatically recognize the fetal facial standard plane. DCNN included the convolutional layers with kernels and fully connected layers. A global average pooling was used in pooling layer to minimize the network parameters for avoiding the over fitting issues and under limited training data.

A differential convolutional neural network (differential-CNN) was designed in [27] to find fetal brain standard planes (FBSPs) from non-standard planes. In designed framework, the differential feature maps were obtained from feature maps in CNN through differential operators.

3. PRENATAL DIAGNOSIS AND SCREENING METHODS

Prenatal testing identifies the birth defects at diverse stages. Prenatal testing comprised the prenatal screening and prenatal diagnosis with prenatal care. Prenatal testing aimed on identifying the issues with pregnancy. Prenatal testing was carried out to address the anatomic and physiologic issues with health of zygote, embryo, or fetus before gestation starts. Screening identifies the issues like neural tube defects, chromosome abnormalities and gene mutations. Prenatal screening aimed on identifying the issues among large population with reasonable and noninvasive techniques. The prenatal diagnosis collected additional information. The screening procedures are routine ultrasounds, blood tests, and blood pressure measurement. The diagnosis process comprised the amniocentesis and chorionic villus sampling. The tests are employed to determine whether the fetus gets aborted or not. The physicians and patients diagnosed high-risk pregnancies early in order that delivery scheduled in a tertiary care hospital.

3.1 An Image Processing Approach for Detection of Prenatal Heart Disease

Prenatal heart disease termed cardiac problems (CHDs) was the collection of ailments that damage heartbeat. Prenatal heart disease linked the plethora of cardiovascular disease risks for early recognition. Data preprocessing was essential method for determining large information in medical business. An investigators employed data mining algorithm to examine large volume of intricate medical information for addressing clinicians forecast heart problems. The system was predicated on classification methods like NB, KNN, DT, and RF algorithms. Prenatal heart disease comprised large number of cardiac disease-related variables. It used large dataset from medical research database of patients with heart disease. The dataset includes 300 instances and 75 attributes. The objective of using naïve bayes classifier was to forecast whether the person develop cardiovascular disease or not with highest accuracy.

3.2 Novel directions in data pre-processing and genome-wide association study (GWAS) methodologies to overcome ongoing challenges

A genome-wide association study (GWAS) was carried out for finding the heritable genetic diseases through finding the correlations between trait differences and allele frequencies of genetic markers. The data pre-processing and GWAS methodologies was carried out through discussing new methods. Data pre-processing with GWAS addressed the challenges in Hardy-Weinberg (H–W) estimation and genotyping. The fast development that addressed the existing challenges was likelihood ratio test for H–W assessment, sequencing for genotyping, and sample structure. GWAS methodologies were reviewed with the statistical method for genetic association analysis. In GWAS method, genotyping cost and capacity gets increased for dataset analysis to observe the tissue-specific signals. The prospective and retrospective association analysis was employed to address the binary traits, non-random ascertainment and to enhance capacity. The rare variants denoted the large portion of genetic markers for computing the disease susceptibility. The fast developments in GWAS data preparation and methodologies addressed current challenges in medical field.

3.3 Prenatal diagnosis by chromosomal microarray analysis

Chromosomal microarray analysis (CMA) was carried out through array comparative genomic hybridization or single nucleotide polymorphism array. In prenatal setting, CMA with karyotyping was employed for chromosomal imbalance detection like aneuploidy and unbalanced reorganization. CMA provided additional diagnostic advantages through sub-microscopic imbalances or copy number changes on standard G-banded chromosome preparation. The submicroscopic imbalances were considered as microdeletions and microduplications in particular genomic regions linked with clinical sequelae. All microdeletion/duplication were not linked with undesirable clinical phenotypes. They were linked with spectrum of clinical phenotypes varies range from benign to severe. The scenarios provided the challenge for prenatal diagnosis and genetic counseling to identify the complex results.

3.4 1D-FHRNet: Automatic Diagnosis of Fetal Acidosis from Fetal Heart Rate Signals

Automated diagnostic technology with artificial intelligence was used by obstetricians in medical decisions. An automatic diagnostic tool was employed for identifying the health care centre and remote areas. A machine learning-based automated diagnostic tool was used for classification and diagnosis of Fetal Acidosis with fetal heart rate. A 1D-CNN model was introduced due to their ability for diagnosing the Fetal Acidosis. The designed model classified the data into healthy or pathological conditions with high accuracy. The signal pre-processing was carried out before classification task to improve accuracy with artifacts in collected signal.

3.5 Improving patient understanding of prenatal screening tests: Using naturally sampled frequencies, pictures and accounting for individual differences

Health professionals communicate the prenatal screening test results efficiently to patients. The experiment results determined the efficiency results of prenatal screening test for Trisomy 21, Trisomy 13, or DiGeorge Syndrome through sampled frequencies. Participants presented the task interpretation with posterior probability of embryo.People obtained better results with sampled frequencies. The visuospatial ability was accounted for individual differences within conditions. Participants not vary the ratings of different presentation formats with the lack of awareness. The results were obtained from the undergraduates respondents or members of population recruited online. All experiments were reviewed through relevant Institutional Review Board through online survey platform. The participants randomly provide the statistical reasoning tasks than key variable manipulation in between-subjects design. An arbitrary medical scenario employed the genetic counseling scenarios and test performance information. Bayesian reasoning task were employed in between-subjects design, realistic genetic counseling contexts with participants receiving percentage information. Participants form different conditions obtained the pictorial of the task in nested tree form.

3.6 Prediction of Late Intrauterine Growth Restriction using Machine Learning Models

Late IUGR was linked with higher risk of perinatal hypoxic events and suboptimal neurodevelopment. Late IUGR resulted in perinatal mortality and suspected in third trimester of pregnancy that confirmed at birth. IUGR in third trimester of pregnancy was linked with low-risk pregnancies. IUGR carried out antenatal diagnosis and treatment with delivery to minimize the risks. The presence of late IUGR was discussed through CTG findings. The seven dissimilar machine learning methods like Naïve Bayes, k-Nearest Neighbors, Support Vector Machine, Logistic Regression, Decision Tree, Random Forest and AdaBoost were discussed for highlighting the preprocessing effects. The logistic regression and support vector machine were introduced to enhance the accuracy, precision, recall and F-score. The designed method increased the robustness and flexibility as a predictive model for the late IUGR. The feature elimination was carried out to enhance the diagnosis accuracy performance with relevant features in predicting late IUGR using logistic regression and SVM.

3.7 Threat and adaptation: The maternal lived experience of continuing pregnancy after receiving a prenatal diagnosis of agenesis of the corpus callosum

A prenatal diagnosis includes uncertainty because of heterogeneous presentation, classification, cause and outcome wastermed as agenesis of corpus callosum. The neuroanatomical anomaly was identified in second or third trimester. The diagnosis caused distress for expectant mothers. The hermeneutic phenomenological study was carried out to explore and provide maternal experience of continuing pregnancy after agenesis of corpus callosum diagnosis. The asynchronous and focus groups lived incidents during pregnancy from diagnosis to birth were discovered with 26 mothers participated in international study. Themes were built through reflexive thematic analysis to describe lived phenomenon experience. Under Threat included Threat to Life of Baby and Threatened Image of Expected Family. Healthcare professionals require the awareness with individual prenatal diagnosis to support mothers through their continued pregnancies. The healthcare professionals employed prenatal diagnosis objective to support maternal preparation. Healthcare service was used to respond in a woman-and family-centred way to minimize the threat and support maternal adaptation after prenatal diagnosis.

3.8 Cell-free DNA screening for prenatal detection of 22q11.2 deletion syndrome

Prenatal screening focused on Down syndrome (T21) screening and aneuploidies (T13 and T18) in fetus. The chromosomal aneuploidies included small proportion of genetic conditions to adverse the infant and childhood results. The noninvasive prenatal screening was carried outdepending on sequencing of circulating cell-free DNA (cfDNA) in maternal blood. The screening has large potential for any genome region with option for subchromosomal variants like chromosomal microdeletions. The prenatal screening was carried out for fetal aneuploidies detection. Cell-free DNA allowed noninvasive screening for subchromosomal copy number variants with 22q11.2 deletion syndrome. The screening resulted in congenital heart defects and neuro-developmental delay.

The screening feasibility was carried out with deletion syndrome and confirmatory postnatal testing to assess test performance. The study assessed single-nucleotide polymorphism and prenatal cell-free DNA screening for deletion syndrome detection. Prenatal DNA samples were requested for genetic confirmation with chromosomal microarrays. The primary outcome attained higher sensitivity, specificity, positive predictive value, and negative predictive value of cell-free DNA screening detection. Noninvasive cell-free DNA prenatal screening for 22q11.2 deletion syndrome identified the affected cases with small nested deletions and lesser false positive rate.

3.9 Noninvasive prenatal screening (NIPS) for fetal chromosome abnormalities in a general-risk population: An evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG)

An evidence-based clinical practice principle was employed for noninvasive prenatal screening (NIPS) with pregnant individuals at risk for fetal trisomy 21, trisomy 18, or trisomy 13. The designed principle was employed to determine the utility of NIPS for additional chromosomal disorders. NIPS Evidence-Based Guideline Work Group was carried out based on American College of Medical Genetics and Genomics (ACMG) systematic review to form the evidentiary basis. Workgroup members employed grading recommendations evaluation, growth and evaluation evidence to perform the draft recommendations. The guideline underwent wide-ranging internal and external peer review with public comment period before approval by ACMG Board of Directors. The evidence increased the accuracy of NIPS for trisomies 21, 18, and 13 in singleton and twin gestations. An autosomal trisomies and microdeletion syndromes with NIPS has increased the area of interest. ACMG recommended NIPS for all pregnant patients with singleton and twin gestations for fetal trisomies 21, 18, and 13. The designed principle recommended NIPS to screen for fetal sex chromosome aneuploidy.

3.10 Prenatal ultrasound screening and congenital anomalies at birth by region: Pattern and distribution in Latvia

The ultrasound scan frequency and congenital malformation rate was discussed between urban and rural areas. A population-based retrospective study was carried out with linked data from administrative data sources and register data. All singleton live births were included in data analysis. The residence place was classified into riga, big cities and rural areas. The multiple regression models were discussed for maternal age, living area and prenatal screenings. The congenital anomalies at birth were carried out with the musculoskeletal system and congenital malformations of circulatory system and genital organs. The association was discussed between rate of foetal anomalies and frequency of prenatal examinations. The number of US examinations per pregnancy was examined in rural regions. Regional variations were carried out with congenital anomalies. Surveillance systems were employed to analyze the US examinations efficiency for early prenatal detection of congenital anomalies.

3.11 Prenatal diagnosis and perinatal findings of 17q12 microdeletion encompassing HNF1B in a fetus with bilateral hyperechogenic kidneys on fetal ultrasound and mild renal abnormality after birth, and a review of the literature of prenatal diagnosis of 17q12 microdeletion

The prenatal diagnosis and perinatal findings of 17q12 microdeletion was carried out with HNF1B in fetus and bilateral hyperechogenic kidneys on fetal ultrasound. A 36-year-old and primigravid woman underwent amniocentesis at 17 weeks of gestation due to advanced maternal age. The simultaneous array comparative genomic hybridization (aCGH) analysis on DNA extracted from uncultured amniocytes. The designed analysis

showed 1.38-Mb 17q12 microdeletion with LHX1 and HNF1B. Prenatal ultrasound showed bilateral hyperechogenic kidneys with normal corticomedullary (CM) differentiation. The parents continued the pregnancy and cross normal 3180-g male baby was delivered at 39 weeks of gestation. At age 2 years and 4 months, the renal ultrasound revealed bilateral increased the renal echogenicity with normal CM isolation and small left renal cysts. The 17q12 microdeletion with LHX1 and HNF1B at prenatal diagnosis presented the variable clinical spectrum with bilateral hyperechogenic kidneys on fetal ultrasound and mild renal abnormality after birth. Prenatal diagnosis of fetal hyperechogenic kidneys increased suspicion of 17q12 microdeletion syndrome.

3.12 Prenatal diagnosis by whole exome sequencing in a family with a novel TBR1 mutation causing intellectual disability

The prenatal diagnosis was carried out for pregnant woman with genetic history of intellectual disability. The chinese pedigree with intellectual disability was gathered. Cytogenetic study, chromosomal microarray analysis (CMA) and completeexome sequencing (WES) followed by Sanger validation were conducted to identify the genetic pathogenesis. A new heterozygous deletion in TBR1 gene was identified resulted in a frame shift mutation by premature stop codon at position 141. Segregation analysis was identified and co-segregated among affected family members. Prenatal diagnosis signified the absence of mutation and family decided to maintain the pregnancy after genetic counseling. The importance of genetic testing was carried out in intellectual disability diagnosis.

4. PERFORMANCE ANALYSIS ON PRENATAL DIAGNOSIS AND SCREENING METHODS

In order to compare the prenatal diagnosis techniques, number of data is considered as an input to conduct experiment. Experimental evaluation of twelve techniques, namelynaïve bayes classifier, genome-wide association study (GWAS), Chromosomal microarray analysis (CMA), 1D-CNN model, visuospatial ability, Late Intrauterine Growth Restriction, neuroanatomical anomaly, Cell-free DNA screening, evidence-based clinical practice principle, population-based retrospective prenatal diagnosis and perinatal findings and prenatal diagnosis are implemented with fetal heart rate features of healthy and late IUGR fetuses Dataset.

URL of the dataset is given as https://ieee-dataport.org/open-access/fetal-heart-rate-features-healthy-and-late-iugr-fetuses. Result analyses of existing disability detection techniques are computed with parameters are,

- Preprocessing accuracy,
- Feature Selection Rate,
- Prenatal Diagnosis Accuracy,
- Feature Selection Time,
- Space Complexity and
- Error Rate

4.1 Result Analysis on Preprocessing Time

The pre-processing time is defined as the product of number of data points and amount of time consumed to preprocess one data point. It is measured in terms of milliseconds (ms). It is calculated as,

$$PT = N * time\ (preprocess\ one\ data\ point) \quad (1)$$

From (1), 'PT' symbolizes the preprocessing time. 'N' symbolizes the number of data points. When the preprocessing time is lesser, the method is said to be more efficient.

Table 1 Tabulation of Preprocessing Time

Number of Data Points (Number)	Preprocessing Time (ms)			
	Naïve Bayes Classifier	GWAS	CMA	1D-CNN model
25	10	14	17	20
50	13	16	19	22
75	15	19	23	25
100	17	21	25	28
125	20	24	28	30
150	22	26	30	33
175	25	29	32	35
200	27	31	34	38
225	30	34	37	41
250	32	37	40	44

Table 1 explains the performance analysis of preprocessing time for four different techniques, namely naïve bayes classifier, genome-wide association study (GWAS), Chromosomal microarray analysis (CMA), 1D-CNN model with respect to different number of data points ranging from 25 to 250. From table value, it is clear that the preprocessing time of Naïve Bayes Classifier is comparatively lesser than other three existing methods. The graphical representation of preprocessing time is shown in the figure 1.

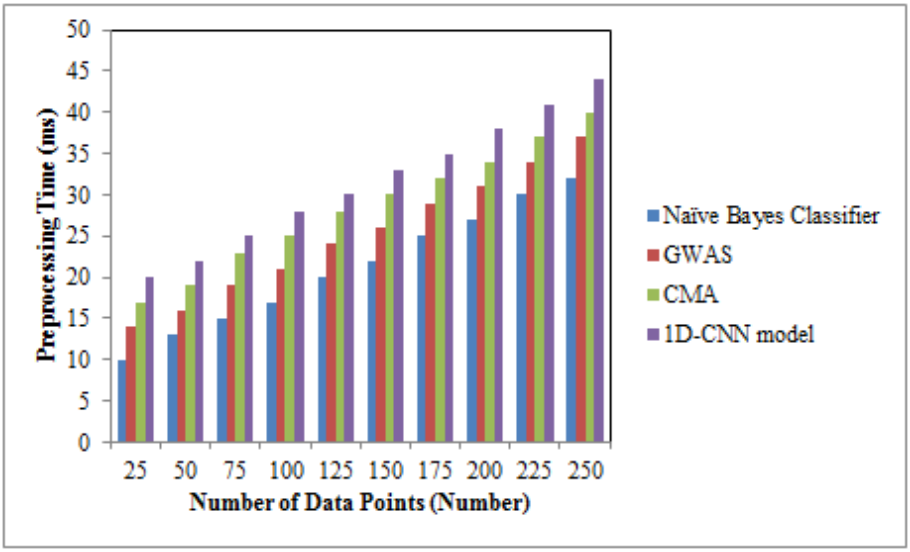

Figure 3 Measurement of Preprocessing Time

Figure 3 shows the preprocessing time comparison on different number of data points. The blue color bar in figure represents the preprocessing time of naïve bayes classifier. The brown color bar and green color bar represents the preprocessing time of GWAS and CMA. The violet color bar in figure represents the preprocessing time of 1D-CNN model. It is clear that the preprocessing time using naïve bayes classifier is higher when compared to the genome-wide association study, Chromosomal microarray analysis CMA and 1D-CNN model. This is due to the application of data mining algorithm to examine the large volume of intricate medical information for addressing clinicians forecast heart problems. The naïve bayes classifier forecast person develop cardiovascular disease or not with lesser preprocessing time. Consequently, the preprocessing time of naïve bayes classifier is reduced by 17% when compared to the GWAS, 28% when compared to the CMA and 35% when compared to the 1D-CNN model.

4.2 Result Analysis on Space Complexity

The space complexity is described as the product of number of data points and amount of space consumed by one data point after data pre-processing. It is measured in terms of megabytes (MB). It is calculated as,

$$SC = N * space\ (preprocess\ one\ data\ point) \quad (2)$$

From (2), 'SC' symbolizes the space complexity. 'N' symbolizes the number of data points. When the space complexity is lesser, the method is said to be more efficient.

Table 2 Tabulation of Space Complexity

Number of Data Points (Number)	Space Complexity (MB)			
	Naïve Bayes Classifier	GWAS	CMA	1D-CNN model
25	22	26	15	17
50	25	28	18	20
75	28	30	21	23
100	30	33	23	25
125	33	36	26	28
150	35	39	29	30
175	37	42	31	33
200	40	44	35	38
225	42	47	37	40
250	44	50	40	43

Table 2 explains the performance analysis of space complexity for four different techniques, namely naïve bayes classifier, genome-wide association study (GWAS), Chromosomal microarray analysis (CMA), 1D-CNN model with respect to different number of data points ranging from 25 to 250. From table value, it is clear that the space complexity of Chromosomal microarray analysis (CMA) is comparatively lesser than other three existing methods. The graphical representation of space complexity is shown in figure 2.

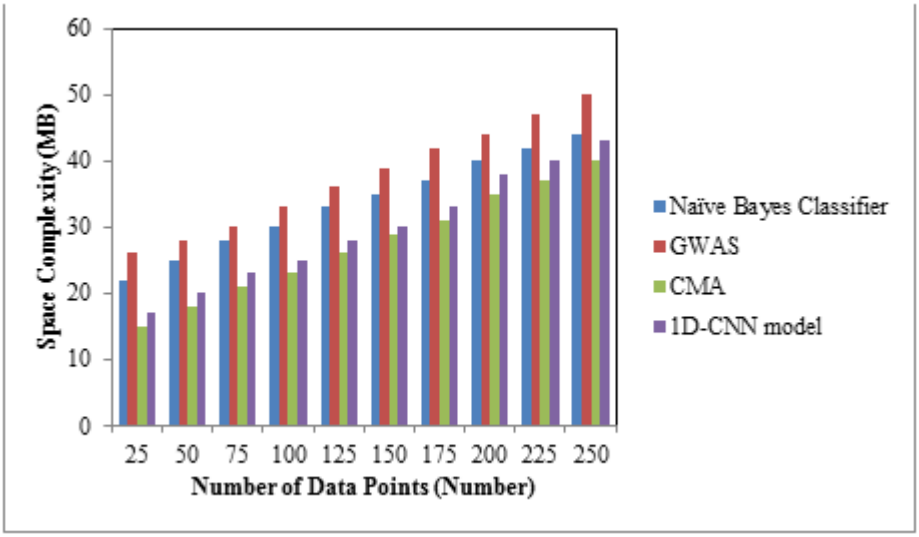

Figure 4 Measurement of Space Complexity

Figure 4 illustrates the space complexity comparison on different number of data points. The blue color bar in figure represents the space complexity of naïve bayes classifier. The brown color bar and green color bar represents the space complexity of GWAS and CMA. The violet color bar in figure represents the space complexity of 1D-CNN model. It is clear that the space complexity using chromosomal microarray analysis is lesser when compared to the naïve bayes classifier, GWAS and 1D-CNN model. This is due to the application of CMA with karyotyping for chromosomal imbalance detection. CMA provided sub-microscopic imbalances on G-banded chromosome preparation with lesser space complexity. Consequently, the space complexity of CMA is reduced by 20% when compared to the naïve bayes classifier, 28% when compared to the GWAS and 8% when compared to the 1D-CNN model.

4.3 Result Analysis on Feature Selection Rate

Feature Selection Rate is described as the ratio of the number of features that are accurately selected to the total number of features. It is measured in terms of percentage (%). It is calculated as,

$$FSR = \frac{Number\ of\ features\ that\ are\ accurately\ selected\ for\ diagnosis}{Number\ of\ features} * 100 \qquad (3)$$

From (3), 'FSR' denotes the feature selection rate.When the feature selection rate is higher, the method is said to be more efficient.

Table 3 Tabulation of Feature Selection Rate

Number of Features (Number)	Feature Selection Rate (%)			
	Visuospatial ability	Late Intrauterine Growth Restriction	Neuroanatomical anomaly	Cell-free DNA screening
3	91	78	82	73
6	89	75	80	70
9	87	73	78	68
12	90	76	82	71
15	92	78	85	73
18	94	80	87	77
21	97	82	89	80

Table 3describes the performance analysis of feature selection rate for four different techniques, namely Visuospatial ability, Late Intrauterine Growth Restriction, Neuroanatomical anomaly and Cell-free DNA screening with respect to different number of features ranging from 3 to 21. From table value, it is clear that the feature selection rate of Visuospatial ability is comparatively higher than other three existing methods. The graphical representation of feature selection rate is shown in figure 3.

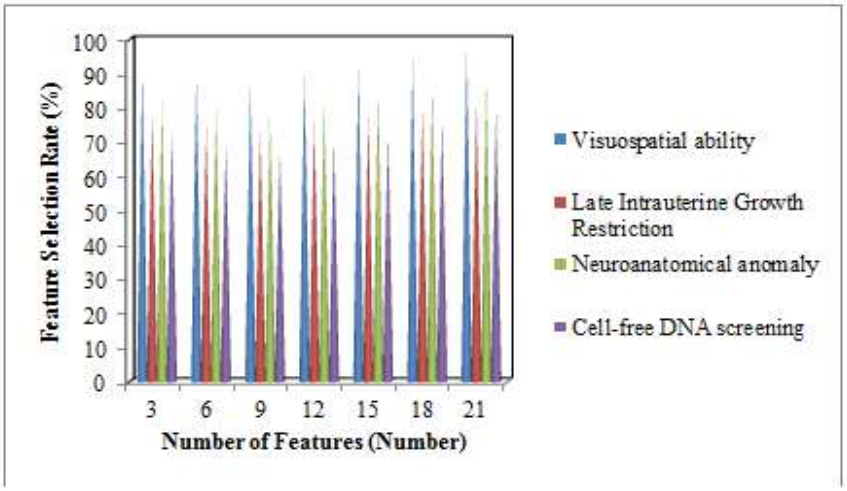

Figure 5 Measurement of Feature Selection Rate

Figure 5 illustrates the feature selection rate comparison on different number of features. The blue color bar in figure represents the feature selection rate of Visuospatial ability. The brown color bar and green color bar represents the feature selection rate of Late Intrauterine Growth Restriction and Neuroanatomical anomaly. The violet color bar in figure indicates the feature selection rate of Cell-free DNA screening. It is clear that the feature selection rate using Visuospatial ability is higher when compared to the Late Intrauterine Growth Restriction, Neuroanatomical anomaly and Cell-free DNA screening. This is due to application of participants with different presentation formats. The bayesian reasoning task were used with realistic genetic counseling contexts with higher feature selection rate. Consequently, the feature selection rate of Visuospatial ability is increased by 18% when compared to the Late Intrauterine Growth Restriction, 10% when compared to the Neuroanatomical anomaly and 25% when compared to the Cell-free DNA screening.

4.4 Result Analysis on Feature Selection Time

The prenatal diagnosis time is defined as the product of number of features and amount of time consumed to select one feature. It is measured in terms of milliseconds (ms). It is calculated as,

$$FST = Number\ of\ features * time\ (select\ one\ feature) \quad (4)$$

From (4), 'FST' represent the feature selection time. When the feature selection time is lesser, the method is said to be more efficient.

Table 4 Tabulation of Feature Selection Time

Number of Features (Number)	Feature Selection Time (ms)			
	Visuospatial ability	Late Intrauterine Growth Restriction	Neuroanatomical anomaly	Cell-free DNA screening
3	30	22	33	38
6	33	27	36	41
9	35	29	39	43
12	38	33	43	46
15	40	35	46	49
18	43	40	49	52
21	45	43	52	55

Table 3 explains the performance analysis of feature selection time for four different techniques, namely Visuospatial ability, Late Intrauterine Growth Restriction, Neuroanatomical anomaly and Cell-free DNA screening with respect to different number of features ranging from 3 to 21. From table value, it is observed that the feature selection time of Late Intrauterine Growth Restriction is comparatively lesser than other three existing methods. The graphical representation of feature selection time is illustrated in figure 4.

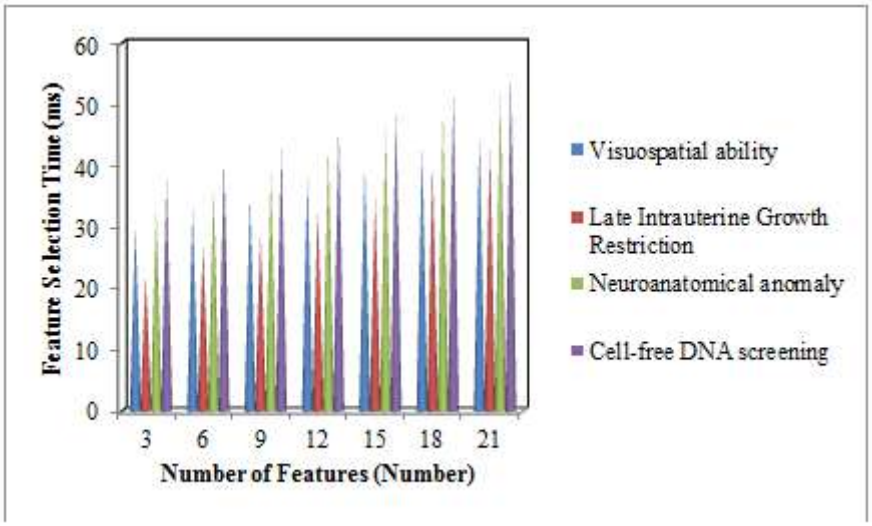

Figure 6 Measurement of Feature Selection Time

Figure 6 explains the feature selection time comparison on different number of features. The blue color bar in figure represents the feature selection time of Visuospatial ability. The brown color bar and green color bar represents the feature selection time of Late Intrauterine Growth Restriction and Neuroanatomical anomaly. The violet color bar in figure represents the feature selection time of Cell-free DNA screening.

It is clear that the feature selection time using Late Intrauterine Growth Restriction is lesser when compared to the Visuospatial ability, Neuroanatomical anomaly and Cell-free DNA screening. This is due to application of logistic regression and support vector machine to enhance the accuracy. The designed method increased the robustness and flexibility. The feature elimination increased the accuracy performance with minimum time consumption. By this way, feature selection time gets minimized. Consequently, the feature selection time of Late Intrauterine Growth Restriction is increased by 14% when compared to the Visuospatial ability, 24% when compared to the Neuroanatomical anomaly and 30% when compared to the Cell-free DNA screening.

4.5 Result Analysis on Prenatal Diagnosis Accuracy

Prenatal diagnosis accuracy is defined as the ratio of the number of data points that are accurately diagnosed to the total number of data points. It is measured in terms of percentage (%). It is computed as,

$$PDA = \frac{Number\ of\ data\ points\ that\ are\ accurately\ diagnosed}{N} * 100 \quad (5)$$

From (5), 'PDA' represent the prenatal diagnosis accuracy. When the prenatal diagnosis accuracy is higher, the method is said to be more efficient.

Table 5 Tabulation of Prenatal Diagnosis Accuracy

Number of Data Points (Number)	Prenatal Diagnosis Accuracy (%)			
	Evidence-Based Clinical Practice Principle	Population-Based Retrospective Model	Prenatal Diagnosis and Perinatal Findings	Prenatal Diagnosis
25	85	74	80	72
50	88	77	82	74
75	90	80	85	77
100	92	83	87	79
125	89	81	84	76
150	87	79	82	73
175	85	77	80	70
200	88	75	83	74
225	91	78	86	76
250	94	80	89	78

Table 5describes the performance analysis of prenatal diagnosis accuracy for four different techniques, namely evidence-based clinical practice principle, population-based retrospective model, prenatal diagnosis and perinatal findings and prenatal diagnosis with respect to different number of data points ranging from 25 to 250. From table value, it is clear that the prenatal diagnosis accuracy of evidence-based clinical practice principle is comparatively higher than other three existing methods. The graphical representation of prenatal diagnosis accuracy is illustrated in figure 5.

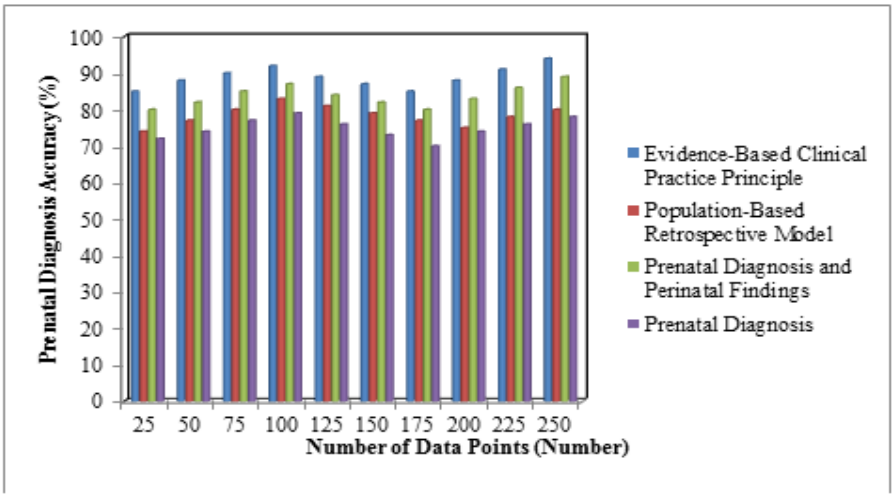

Figure 7 Measurement of Prenatal Diagnosis Accuracy

Figure 7 explains the prenatal diagnosis accuracy comparison on different number of features. The blue color cylinder in figure represents the prenatal diagnosis accuracy of evidence-based clinical practice principle. The brown color cylinder and green color cylinder represents the prenatal diagnosis accuracy of population-based retrospective model and prenatal diagnosis and perinatal findings. The violet color cylinder in figure represents the prenatal diagnosis accuracy of prenatal diagnosis method. It is clear that the prenatal diagnosis accuracy using evidence-based clinical practice principle is higher when compared to the population-based retrospective model and prenatal diagnosis and perinatal findings and prenatal diagnosis. This is due to application of workgroup members with grading recommendation, growth and evaluation evidence. The guideline underwent wide-ranging internal and external peer review with public comment period. As a result, the prenatal diagnosis accuracy of evidence-based clinical practice principle is increased by 13% when compared to the population-based retrospective model, 6% when compared to the prenatal diagnosis and perinatal findings and 19% when compared to the prenatal diagnosis.

4.6 Result Analysis on Error Rate

Error Rate is defined as the ratio of the number of data points that are incorrectly diagnosed to the total number of data points. It is measured in terms of percentage (%). It is computed as,

$$ER = \frac{Number\ of\ data\ points\ that\ are\ incorrectly\ diagnosed}{N} * 100 \qquad (6)$$

From (6), 'ER' symbolizes the error rate. When the error rate is lesser, the method is said to be more efficient.

Table 6 Tabulation of Error Rate

Number of Data Points (Number)	Error Rate (%)			
	Evidence-Based Clinical Practice Principle	Population-Based Retrospective Model	Prenatal Diagnosis and Perinatal Findings	Prenatal Diagnosis
25	24	18	12	30
50	27	20	15	32
75	30	23	17	35
100	33	26	20	38
125	35	29	22	40
150	32	27	19	37
175	30	24	17	35
200	28	21	15	32
225	31	25	19	36
250	33	28	22	39

Table 6explains the performance analysis of error rate for four different techniques, namely evidence-based clinical practice principle, population-based retrospective model, prenatal diagnosis and perinatal findings and prenatal diagnosis with respect to different number of data points ranging from 25 to 250. From table value, it is clear that the error rate of prenatal diagnosis and perinatal findings is comparatively lesser than other three existing methods. The graphical representation of error rate is illustrated in figure 6.

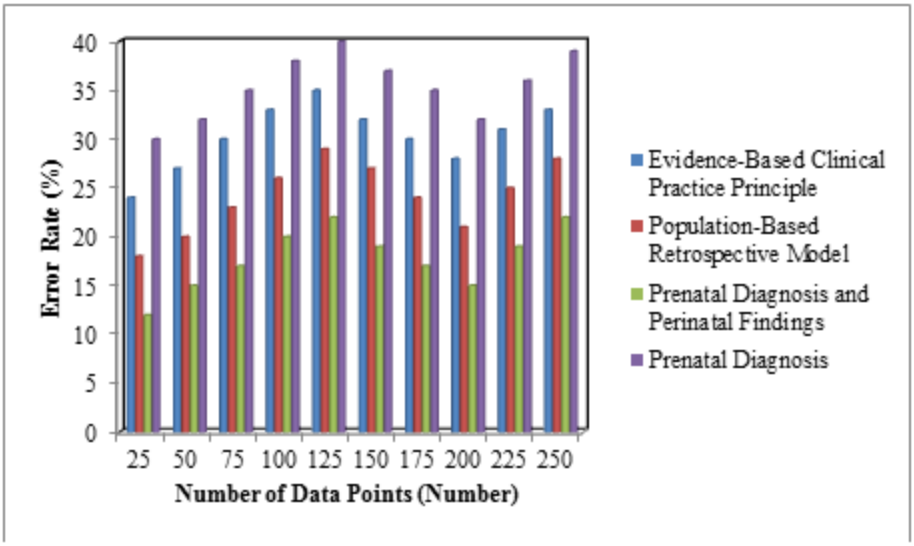

Figure 8 Measurement of Error Rate

Figure 8 illustrates the error rate comparison on different number of features. The blue color cylinder in figure represents the error rate of evidence-based clinical practice principle. The brown color cylinder and green color cylinder represents the error rate ofpopulation-based retrospective model and prenatal diagnosis and perinatal findings. The violet color cylinder in figure represents the error rate of prenatal diagnosis method. It is clear that the error rate using prenatal diagnosis and perinatal findings is lesser when compared to the evidence-based clinical practice principle, population-based retrospective model and prenatal diagnosis. This is because of using simultaneous array comparative genomic hybridization (aCGH) analysis on DNA collected from uncultured amniocytes. Prenatal ultrasound showed bilateral hyperechogenic kidneys with normal differentiation. As a result, the error rate of prenatal diagnosis and perinatal findings is increased by 42% when compared to the evidence-based clinical practice principle, 26% when compared to the population-based retrospective model and 50% when compared to the prenatal diagnosis.

5. CONCLUSION

A comparison of different prenatal screening and diagnosis techniques with data preprocessing and feature selection concepts is discussed. From the study, it is clear that the error rate was not minimized. In addition, the preprocessing time was not reduced to the required level by CMA. In addition, the prenatal diagnosis accuracy was not improved by population-based retrospective model and error rate was not minimized by prenatal diagnosis method. In addition, the space complexity was not minimized by GWAS. The feature selection time was not minimized by Cell-free DNA screening. The wide range of experiment on existing prenatal diagnosis and screening methods determines the data pre-processing and feature selection performance results with its limitations. Finally, from the result, the research work can be carried out using machine learning and deep learning techniques for improving the performance of prenatal screening and diagnosis with higher accuracy and lesser time complexity.

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