Artificial Intelligent Models for Automatic Diagnosis of Foetal Cardiac Anomalies: A Meta-Analysis

M. O. Divya and M. S. Vijaya

Abstract The foetal anomaly scanning is one of the most challenging areas where accuracy of diagnosis much fluctuating with respect to the expertise of the radiologist and the mental equilibrium of the radiologist at the time of scanning. Amongst the various anomalies, foetal heart anomaly diagnosis expects precise and sensitive intellectual presence since perilous congenital heart diseases are one of the common causes resulting in the major population of infant mortality or into permanent natal faults. The accuracy of manual diagnosis of foetal cardiac abnormalities from the ultrasound scan images vary based on the human expertise and the presence of mind. Therefore, the scope of computer-assisted judgement can produce accurate diagnosis irrespective of the operator's profile. Numerous researches are going on to explore the scope of computer-assisted judgement of abnormalities using ultrasound imaging technique (USIT), specifically using machine learning and deep learning models. This work exploits the opportunities of computer-assisted diagnosis in foetal cardiac anomaly diagnosis as this is one of the most sensitive areas where appropriate diagnosis can save a life and a wrong diagnosis may lose a life unnecessarily.

Keywords Congenital heart disease · Early diagnosis · Ultrasound images · Image processing · Machine learning · Deep learning · Foetal cardiac anomaly

1 Introduction

Ultrasound examination is one of the fundamental medical imaging techniques used in clinics on a regular basis. This is a non-invasive and non-radioactive technique to understand the internal body structure; thus becomes a universally accepted medical

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A. Kumar et al. (eds.), *Proceedings of the International Conference on Cognitive and Intelligent Computing*, Cognitive Science and Technology,
https://doi.org/10.1007/978-981-19-2358-6_18

imaging mechanism for preliminary examination and diagnosis of various biological disorders. The challenge with the ultrasound imaging technique (USIT) is that it purely depends on the operator's perception and expertise.

There has been a recent explosion in the use of artificial intelligence (artificial intelligence), which is now part of our everyday lives. Uptake in medicine has been more limited, although in several fields there have been encouraging results showing excellent performance when artificial intelligence is used to assist in a well-defined medical task. Depending on the problem scenario, data availability and other constraints the researcher can decide which technique or combination of techniques are appropriate. The subsets of AI are mentioned in Fig. 1. In medical field, most of the research has been performed using retrospective data and there have been few clinical trials published using prospective data [1]. This review focuses on identifying the potential uses of machine learning and deep learning models in the field of foetal cardiology.

Congenital Heart Defects (CHD) are one of the most common forms of birth malformations in newborns and infants. They occur at a prevalence of 6–8 per 1000 live births. The prevalence of these defects may be higher in prenatal scans. About 1

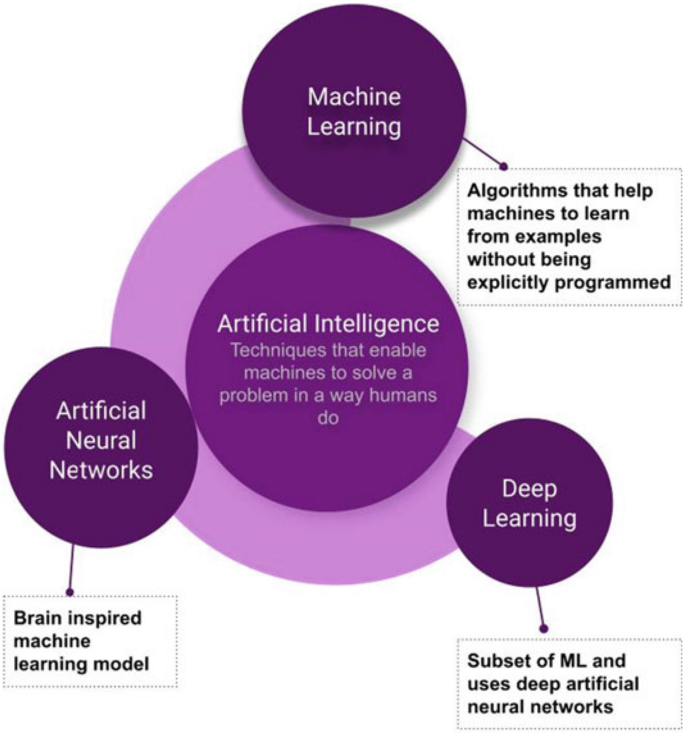

Fig. 1 Relation between AI, ML, DL

in 4 babies with a CHD have a critical CHD. CHDs are a leading cause of birth defect-associated infant illness and death. CHD forms the 5th most common cause of infant deaths on a global level infants with critical CHDs generally need surgery or other procedures while in the uterus or during their first year of life. In India, it is estimated that around 200–250,000 infants are born with CHD every year of which 100,000 are deemed critical requiring early surgical intervention. These surgical procedures are very critical and need thorough setting of environment before the baby is born. Hence identification of these anomalies during the second trimester or third trimester has a value in life.

In the study conducted by Parikh et al. [2] in UK shows that 68.2% of the CHD cases were been diagnosed postnatally and 8.4% of the cases reported with neonatal death. This shows the sensitivity of diagnosis of CHD prenatally and the relevance of USIT. Ultrasound of the foetal heart is highly specific and sensitive in experienced hands, but despite this, there is significant room for improvement in the rates of prenatal diagnosis of congenital heart disease in most countries. USIT can help to improve the present statistics of prenatal diagnosis rate of foetal cardiac anomalies. This research paper projects the possible breakthroughs in automated diagnosing of foetal cardiac anomalies with the help of machine learning and deep learning models.

2 Background Study

Prenatal diagnosis of major forms of CHD is feasible during early and mid-trimester ultrasound scans of pregnancy. The optimal timing of this evaluation is 18–22 weeks of gestation. Several international associations have laid down clear guidelines for the conduct of the foetal heart evaluation during mid-trimester anomaly scans. In the simplest form, this screening involves two views—the four-chamber view and the outflow tracts using the three-vessel view. In the event of a failed screen, the patient needs a more thorough evaluation by an expert for a possible heart defect. Prenatal diagnosis of major CHDs offers more options for management to the expectant families including termination of pregnancy (if diagnosed early < 20 weeks) for complex defects and planned peri-partum care for critical correctable CHDs. Studies from India have shown that prenatal diagnosis of critical CHDs improves the preoperative clinical status and in-hospital outcomes and results in lower costs of in-hospital care due to shorter duration of ICU stay.

However, despite the availability of ultrasound equipment and clear protocols for foetal heart screening, the overall pick-up rates of major CHDs is very low in prenatal scans, especially in the low-middle-income countries (LMICs). Lack of awareness and familiarity with imaging protocols and difficulty in interpretation of images are stated as personal reasons for this. Since the imaging views for the basic screening (two views) are very standard and the image patterns are stereotyped, it is feasible to develop an artificial intelligence (artificial intelligence) based algorithm for a basic level evaluation.

Figure 2, graph explains how minimal is the diagnosis rate of various structural anomalies and the major part of it is unspecified. It shows the importance of intervention of technology so that the device auto diagnoses from the scan image which can very well improve the diagnosis rate. Figure 3 shows the mortality rate due to CHD over the specified period. This is not because of the medical incapability, but it is due to the inaccurate prenatal diagnosis of CHD.

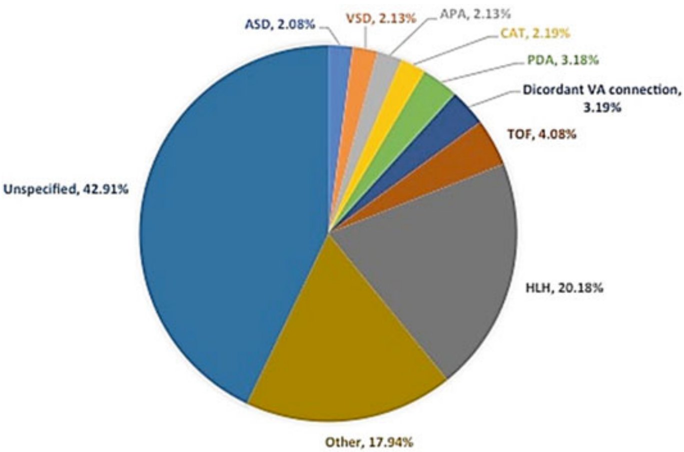

Fig. 2 CHD mortality trends by diagnosis

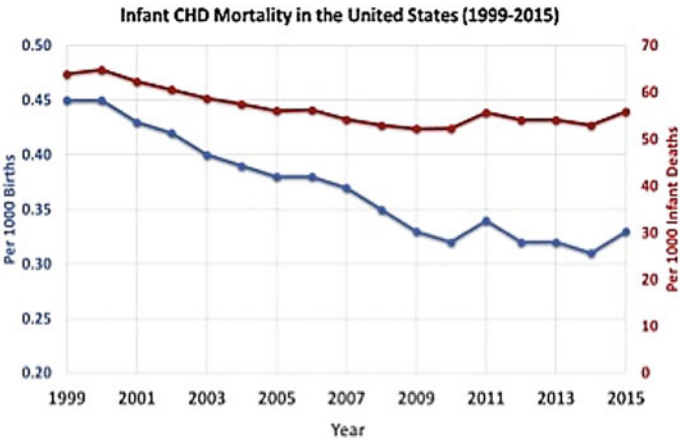

Fig. 3 CHD mortality trends in the US

3 Literature Review

There are numerous researches happening in medical image processing and diagnosing diseases from USIT, with minimum manual interventions. Majority of the work revolves around the opportunities in breast cancer diagnosis and thyroid abnormalities. Those existing research works throw light on the automated diagnosis of diseases affecting other organs as well. For this meta-analysis research, two categories of literatures were taken into consideration. The first set of literatures belongs to the research publication from medical practitioners who have suggested the technological interventions required in foetal cardiac anomaly diagnosis. The second category of literatures constitute the published research work that addresses the diagnosis of various diseases from USIT using machine learning and deep learning [3].

3.1 Medical Literatures Suggesting Technological Interventions

1. Parikh et al. [4] in their research identified how the early diagnosis of CHDs can result in the best-attempted route for the delivery. The research shows that the mode of delivery for a foetus with cardiac structural anomalies is very critical because if it gets unnoticed can cause neonatal morbidity based on the severity of CHD. In the current system, most of the CHDs are diagnosed postnatally. In neonates with connatural defects, 8.4% of 107 neonatal death happened and in 54.2% of 83 neonates, grave respiratory morbidity happened with left ventricular outflow tract defects.
2. Holland et al. [5] The research was a meta-analysis, where the difference in mortality rate was analyzed when the cardiac anomalies were identified prenatally and postnatally. The study indicates that the importance of prenatal diagnosis of CHDs makes a lot of difference and reduces the postnatal mortality rate.
3. Suard et al. [4] did an evaluation on the accuracy rate of prenatal diagnosis of CHDs in South France. This observational study considered 249,070 deliveries into consideration. The diagnosis rate of Group 1 CHD (Where no possibilities of anatomical repair is possible) was 97.8%, for Group 2 (anatomical repair was possible but requires neonatal cardiologic attention and management) was 6.3% and Group 3 (no emergency anatomic procedure was required) was 65.9%. Which shows the Group 2 and Group 3 diagnosis rate could be improved for easy anatomic corrections prenatally and postnatally.
4. Changlani et al. [6], The study shows, short-term consequences of infants where prenatal diagnosis of CHD delivered in a tertiary cardiac care facility. 552 foetuses were diagnosed to have CHD. The study revealed that the prenatal finding of CHD in infants will help for planned delivery in a cardiac facility showed satisfactory fast outcomes, specifically among those receiving dedicated postnatal cardiac care.

5. Vijayaraghavan et al. [7] in their observational study revealed that the planned peri-partum care is an unexplored concept for care of neonates with critical CHDs in low-middle-income countries. The study was a comparison of outcomes of prenatal and postnatal diagnosis of CHDs.

3.2 Review of Articles Related to Diagnosing Diseases Using Machine Learning/Deep Learning and Ultrasound Scan Images

Ding et al. [8], in this research paper explain how the features can be extracted from ROIs. Then these features were used for the diagnosis of breast tumours. As the breast ultrasound images lack clarity due to speckle noise, Multiple-instance learning (MIL) method is more appropriate to classify breast tumours into benign and malignant, using BUS images, for which a novel MIL method is proposed in this research paper. First, a self-organizing map is used to map the instance space to the concept space. The experimental results show better performance, accuracy is 0.9107 and the area under receiver operator characteristic curve is 0.96 ($p < 0.005$).

Wei et al. [9] in their study, texture and morphological features are used for breast tumour classification. This combination is selected because the texture feature is supposed to be very aggressive due to which the low dimensional features will get unnoticed. The study uses, LBP, HOG, GLCM and morphological (i.e. shape complexities) features of breast ultrasound images. To classify, SVM classifier on texture features and a naive Bayes classifier on morphological features is applied. Then the classification results are fused to get the final classification. This method shows accuracy of 91.11%, sensitivity of 94.34% and specificity of 86.49%.

Song et al. [10], in their study, a hybrid multi-brach CNN with feature cropping technique is used for classification of ultrasound images of thyroid nodules. The convolutional neural network for extracting the shared feature maps with a global branch classifier. A feature cropping branch for reducing the impact of similar local features of benign and malignant images, a feature cropping branch also is there. To train the system, a weighted cross-entropy loss function was used. The method has attained accuracy – 96.13%, precision – 93.24%, recall – 97.18% and F_1-measure 95.17% in public and local dataset.

Liyang et al. [11] intended to propose a statical analysis to identify benign and malignant lesions from the breast ultrasound scan images. This research was conducted on 85 cases where 35 malignant and 50 benign ultrasound images were collected. From the images, a fractal dimensional image was produced from the ROI by using box-counting method. From the FD and ROI images, features were extracted including mean, standard deviation, skewness and kurtosis. Statistical tests were conducted on these features. The statistical analysis revealed that the mean texture of images performed the best in differentiating benign versus malignant tumours. The sensitivity, specificity, accuracy, positive predicted value (PPV), negative predicted value (NPV) and Kappa for the mean was 0.77, 0.84, 0.81, 0.77, 0.84 and 0.61 respectively.

Chi et al. [12], their study proposed a deep learning model for classifying the thyroid gland nodule as benign and malignant. They have used a trained GoogLeNet model and the experimental results exhibit excellent performance, accuracy – 98.29%, sensitivity – 99.10% and specificity – 93.90% for the images in an open access database, while classification accuracy – 96.34%, sensitivity – 86% and specificity – 99% for the images in their local region health database.

Abdel-Nassar et al. [13], this research focuses on reducing the speckle noise of breast ultrasound images and thereby increase the accuracy of the model. For this, multiple images of the same targets are used as input to the classifier. The system works in four phases which include, super-resolution computation, extraction of ROI, feature extraction and then classification.

Gao et al. [14] This study explores the eight viewpoints of the heart using the echo videos which intern assist for the diagnosis of cardiologic disorders. The proposed architecture gives the best classification results with 92.1% accuracy rate whereas 89.5% is achieved using only single spatial CNN network.

Reviews on research articles cited in authenticated repositories including Scopus, Web of Science, Medline, PubMed and EMBASE to explore the opportunities in applying artificial intelligence techniques to diagnose foetal cardiac anomalies. The review revealed that a single model might not be enough rather a combination of models are appropriate for accurate diagnosis. The results are based on the guidelines provided by PRISMA (Preferred Reporting Items for Systematic Reviews and Meta-Analyses).

4 Methodology

Congenital cardiac disease is seen in 2–6.5 of 1000 live births and is a major cause of morbidity and mortality, with half of these cases being lethal or requiring surgical correction. Finding of CHDs is very sensitive and can be troublesome. Detection of anomalies alters the obstetric course and outcome, including reassurance, termination, foetal therapy, mode of delivery and postnatal referral to a tertiary care canter with advanced expertise in management of these patients, which is the major difference that could save/change the baby's life after birth. The conventional diagnosis is done manually using the USIT captured by foetal cardiac ultrasound performed between 18 and 22 weeks. This examination is a targeted study where the heart structure and its function. During the first phase (Phase 1), two views of the foetal heart evaluation will be done manually. The four-chamber view and the three-vessel view. In cases where there are critical anomalies found, a detailed echocardiography is performed. Name it as Phase 2 which will include the six views analysis, recommended for an extended basic evaluation of the foetal heart including the situs, four-chamber view, LV outflow tract, RV outflow tract, three vessel and three vessel tracheal view.

The same phases could be implemented with the deep learning/machine learning model as well [3]. There are common classifiers like Support vector machine (SVM), k-nearest neighbours (KNN), etc. in ML and Convolutional neural network (CNN), Recurrent neural networks (RNN), Self-organizing maps (SOM), etc. in deep learning. The most recent research by Liu et al. [15] has proved their research that the deep learning model Stacked Sparce auto encoder (SSAE) based model shows 100% accuracy for classifying medical images.

4.1 Phase 1: Model for Preliminary Classification and Screening

The first phase of proposed system is to develop an algorithm to train the model using labelled images. Here, the model will be trained to evaluate the two views of the foetal heart, the four-chamber view and the three-vessel view. The model will be trained to recognize all possible permutations and combinations of the normal four-chamber and three vessel views (apical, basal and lateral views). This could be done by looking at the patterns (features) extracted from the images and by comparing it to the patterns generated for the normal USIT.

Based on these two views, the model will be trained to report as:

- Normal: If both the views are normal
- Abnormal: If either of the views are not normal

Only the abnormal cases (where CHD is found) need to be taken into consideration for further screening. If the image is diagnosed with anomaly, next-level screening will be done. Here, the model evaluates the six views together with the situs, four-chamber view, LV outflow tract, RV outflow tract, three vessel and three vessel tracheal view. All these will be implemented using the feature vectors extracted from the images. The algorithm will be trained to recognize the patterns [16] in the most common forms of major CHDs found in the UTIs and it will offer a possible diagnosis. Table1 lists the CHDs and gives an overview of the frequency of prevalence of each one.

4.2 Phase 2: Diagnostic Model

The Phase 2, Diagnostic model will be the actual clinical diagnosis system where the live images could be supplied to the trained model and the model will report (Predict) the exact anomaly (ies).

Figure 4 explains the overall workflow of the proposed artificially intelligent system. The major advantage of this automated system will be, once the system is ready after clinical experiments, there might not be changes in the model until unless

Table 1 List of Heart anomalies and its incidence

S. No.	Defect	Incidence	Occurance
1	Ventricular septal defect	Most Common	1.5–3.5 per 1000 live births
2	Atrial septal defect	Fifth most common	1 of 1500 live births
3	AV septal defect	2–7% of All CHD cases	0.19–0.56 per 1000 live births
4	Tetralogy of Fallot	5–10% of All CHD cases	0.24–0.56 per 1000 live births
5	Truncus Arteriosus	1–2% of All CHD cases	NA
6	Transposition of great arteries	1% of All CHD cases	NA
7	Single ventricle	2% of All CHD cases	NA
8	Double outlet RV	less than 1% of All CHD cases	0.08–0.16 per 1000 live births
9	Hypoplastic right heart syndrome	Rare	1.1% of stillbirths
10	Hypoplastic left heart syndrome	2–4% of All CHD cases	0.16–0.25 per 1000
11	Aortic coarctation or hypoplasia	6–7% of All CHD cases	
12	Aortic Atresia or stenosis	Rare	5.2% in newborns
13	Pulmonary stenosis	Rare	7.4% of newborns
14	Ebstein anomaly	less than 1% of All CHD cases	1 per 20,000 live births
15	Ectopia cordis and pentalogy of Cantrell	Rare	14–18 days gestational age
16	Cardiomyopathies	8–11% of All CHD cases	8–11%
17	Arrhythmia	2% of All CHD cases	NA

a new CHD is discovered in the medical field. The logic could be attached with any imaging unit with minor customizations.

5 Results and Discussions

There are many researches happening with regard to medical images majorly for diagnosing, for guiding interventions, for planning a surgery, etc. Table2, lists the latest researches conducted for diagnosing of diseases or structural abnormalities from ultrasound scan images using various ML/DL models. For all the listed researches, the image dataset are ultrasound images.

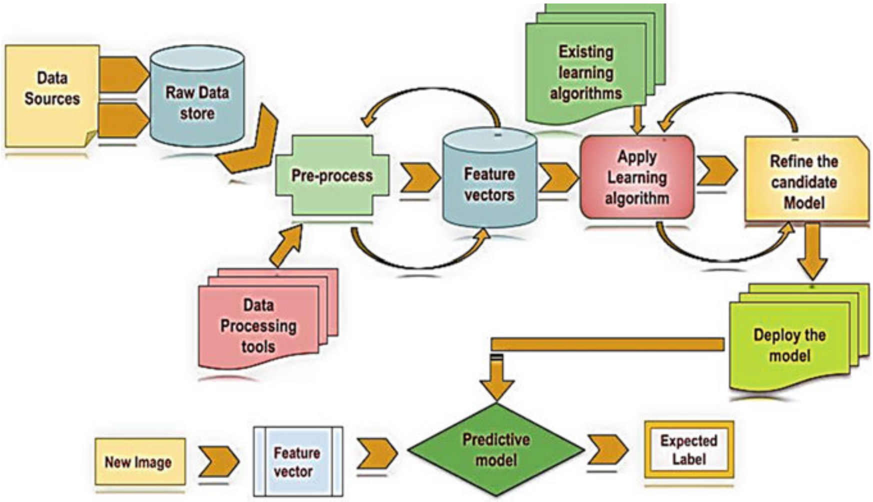

Fig. 4 Proposed CHD diagnosis model building and its implementation

Wei et al. in their research [9], a combination of SVM classifier, naïve bayes classifier and KNN are used which resulted in 91.1% accuracy. Liang et al. [11] using GLCM feature, statistical tastings were done to diagnose breast cancer. The accuracy for the method was 81%. Chollet et al. [17] in their research to detect thyroid nodule, CNN and RCNN were used which has an accuracy of 96.16%. Chi et al. [12] using random forest classifier for thyroid nodules classification got an accuracy of 98.29%. Abdul et al. for classifying breast tumours used co-occurrence matrix which showed an accuracy of 99%. Abdel et al. [13] used GLCM and co-occurrence matrix for classifying breast tumours which showed 99% accuracy. Komatsu et al. [18] in their research used CNN for identifying the structural anomalies with accuracy 70%. Song et al. in his research used CNN for identifying and recognizing thyroid nodules which was showing 98% accuracy. Ding et al. in their research used self-organizing maps along with CNN and bag of words concepts together for breast tumour classification. This showed an accuracy of 91.07%. Virmanj et al. [19] in their research SVM classifier was used with texture feature classifying liver ultrasound images. This showed an accuracy of 88.8%.

The literatures considered for this review includes two categories, the medical articles which prove the requirement of technological interventions in foetal cardiac anomaly diagnosis and the second set where different models were suggested for similar diagnosis using ultrasound scan images. While scanning through published articles that suggests the different models for different diagnosis, most of them fall under the following categories.

- Breast tumour classification
- Thyroid nodule classification

Table 2 The overview of performance of various ML/DL models used in different research used for various diagnosis

S. No.	Date of publishing	Reference index	Title	organ or body location	Application	Technique used	Accuracy
1	Jun-21	[8]	Breast ultrasound image classification based on multiple instance learning	Breast	CNN SOM	Shape	0.9107
2	Jan-21	[18]	Detection of cardiac structural abnormalities in fetal ultrasound videos using deep learning	Heart	CNN	Supervised object detection with Normal Data only	0.7
3	Oct-20	[9]	A benign and malignant breast tumour classification method via efficiently combining texture and morphological features on ultrasound images	Breast tumor diagnosis	Classification LBP, HOG, GLCM, morphological feature	SVM classifier, naïve bayes classifier, KNN, DT, LDA	91.11
4	Apr-20	[11]	Classification of Breast ultrasound tomography by using textural analysis	Breast tumor diagnosis	GLCM, Statical methods, t-test, mean, average etc.	Laplacian of Gaussian (LoG) method, texture images were calculated using box-counting	0.81
5	Mar-20	[17]	Thyroid nodule ultrasound image classification through hybrid feature cropping network	Thyroid	Hybrid feature cropping network, CNN	CNN, RCNN, Boundary feature cropping (RFC),Random feature cropping (RFC)	96.13

(continued)

Table 2 (continued)

S. No.	Date of publishing	Reference index	Title	organ or body location	Application	Technique used	Accuracy
6	Jul-17	[12]	Thyroid nodule classification in ultrasound images by fine-tuning deep convolutional neural network	Thyroid	GoogleNet model	Random forest classifier	98.29
7	Aug-16	[13]	Breast tumour classification in ultrasound images using texture analysis and super-resolution methods	Breast	GLCM, LBP, phase congruency based LBP, HOG, Pattern lacunarity system	Co-occurrence matrix	0.99
8	Aug-15	[1]	Multi task cascade convolution neural networks for automatic thyroid nodule detection and recognition	Thyroid	CNN	Texture	0.98
9	Oct-12	[19]	SVM based characterization of liver ultrasound images using wavelet packet texture descriptor	Liver	SVM	Texture	0.888

- Brain tumour classifications
- Liver-related diagnosis.

Though the medical literatures clearly prove the requirement of technological inventions in foetal cardiac anomaly detection, the interesting part noticed here was after long search for literatures suggesting models/Algorithm which propose any intervention in foetal cardiac anomaly detection were very few. There can be another fact that keep researchers away from this because there is no standard medical database available for this research. Getting the real patient images are also a challenge as there will be long procedure to undergo. The ethical issues related to the confidentiality of patient data can be a hurdle to get data from clinics. Even then it can be collected with not stop efforts and if the researcher could prove his/her genuine interest to solve a social issue. There are concepts like crowdsourcing which could also help developing the dataset. The advancements in IoT can supplement the implementation of the model in smart gadgets for live monitoring of patients. This projects the strong scope of this area.

6 Conclusion

This review paper projects the importance of accuracy in the cardiac anomaly diagnosis and the positive revolutions that artificial intelligence can bring in this field. Any compromise on the prenatal diagnosis may result with the death of the baby or with a permanent damage that affects the whole family throughout their lives. The various research quoted above has modelled several mechanisms for diagnosing various abnormalities in different organs, majorly breast and thyroid. The outcome of these research shows the possibilities of similar models for diagnosing CHDs as well. The outcome is going to be revolutionary because the postnatal treatments can be well planned in advance and will result in promising healthy life for humankind.

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