

# An Intelligent Diagnostic System for Congenital Heart Defects

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**Abstract**—congenital heart disease is the most common birth defect. The article describes detection and classification of congenital heart defect using classification and regressing trees. The ultimate goal of this research can decrease risk of cardiac arrest and mortality in compared with healthy children. The intelligent system proposed in three stages technique for automate diagnosis:(i) pre-processing(ii), feature extraction, and (iii) classification of congenital heart defects (CHD) using data mining tools. The intelligent diagnostic system has been validated with a representative dataset of 110 heart sound signals, taken from healthy and unhealthy medical cases. This system was evaluated in the test dataset with the following performance measurements global accuracy: 98.18%, sensitivity, 96.36% and specificity 100%. This results show the feasibility of classification based on optimized feature extraction and classifier. This paper follows the Association for the recommendations of the Advancement of Medical Instrumentation.

**Keywords**—congenital heart defects; Heart murmurs; newborns; classification and regression trees;

## I. INTRODUCTION

Physicians use the stethoscope as a device to listen to the acoustic signals which cannot be analyzed by the human ear. The interpretation can provide valuable information regarding the function of heart valves, and is capable of detecting of disorders, especially problems related to the valves. Various techniques have been developed to analyze and classification of the heart sound [1].

Physicians use a physical exam and special heart tests to diagnose congenital heart defects. They often find severe defects during pregnancy or soon after birth. Many congenital heart defects cause few or no signs and symptoms. They are often not diagnosed until children are older.

Congenital heart defects are characterized by anomalies in the structure of the heart and its related valves and vessels; such as holes in the heart, narrowed, leaky valves, malformed, missing vessels or heart chambers ventricular septal defect and coarctation of the aorta are typical examples of CHDs. These defects are the most common type of birth defect and typically present in infancy. In many cases, the cause of the defect in a particular infant is unknown; from an epidemiological standpoint, maternal smoking during pregnancy, genetics, and chromosomal abnormalities have been cited as factors contributing to CHD [2].

Congenital heart problems range from simple to complex. Some heart congenital defects in children can be watched by

the child's physician and managed with medications, while others require surgery, sometimes as soon as in the first few hours after birth. A child may even "grow out" of some of the simpler heart problems, such as patent ductus arteriosus (PDA) or atrial septal defect (ASD), as these defects may simply resolve on their own as the child grows. Other infants will have a combination of malformations and require several operations throughout their lifetime.

Newborns with heart congenital defects experience no symptoms. The heart defect may be diagnosed if the health care provider hears an abnormal sound, called a murmur.

Children with normal hearts also can have heart murmurs, called innocent or functional murmurs. A provider may suggest tests to rule out a heart defect.

Some babies born with a heart congenital defect can appear healthy at first and can be sent home with their family before their heart defect is detected. Congenital heart defects affect approximately 1 in 125 live births. Of these, 30% have extra-cardiac anomalies (such as tracheoesophageal fistula, anorectal anomalies), which might require surgery within the first year of life [3]. These babies are at risk for having serious problems within the first few days or weeks of life and often require emergency care.

Although a normal heart still orders an echocardiogram for reassurance, even though the cost of an echocardiogram is high. The result of this practice is a misallocation of healthcare funds, since echocardiograms are expensive. While it is clearly important to avoid that healthy newborn are sent for echocardiogram, it is also important to avoid that a newborn that has a pathological heart murmur is sent home without proper treatment [4].

Many studies performed on heart congenital defects are concerned with various stages of life, but our dataset included newborns from one day to 2 months after birth [5]. The classification at this stage is very important because pathological heart sounds in newborns are more difficult to diagnose [6]. We achieve a high accuracy result to discriminate CHD by using optimized features and CART [4-8].

In this paper, we present a method for automatically segmentation of phonocardiogram (PCG) data. The proposed methods utilize CART to identify whether a systolic and diastolic pathological murmur holds. Notably, features extraction was very effective to improve experimental results. Our results show an accuracy of 98.18%, which significantly

improves the current state-of-the-art on this specific problem. In fact, other relevant works report accuracies. Improvements have been obtained also in sensitivity and specificity.

## II. METHODS

This phase involves pre-processing and features extraction of the signal that is certain characteristic properties of heart sound that are unique to the signal and are thus suitable for classification purposes. Figure 1 show stages used in the proposed diagnostic system.

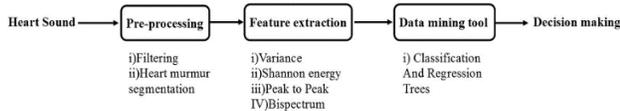


Fig.1. Stages used in an intelligent diagnostic system.

### A. Pre-processing

Pre-processing occurs in two steps that will be described in the following order:

1) *Filtering*: The first step of signal processing is filtering heart sounds, with the goal of removing the unwanted noise. The recording of PCG usually has a sampling frequency higher than 8 kHz. In the event that the recording environment cannot be controlled enough, noise is coupled into the PCG. To avoid unpredictable effects brought by noise, filtering becomes important for later processing. Since the main spectrum of first and second (S1 and S2 respectively) heart sound occurs within the range of 200 Hz, the system filters the original heart sound using a 3rd order band-pass Butterworth filter, with cut-off frequencies at 50 Hz and 200 Hz. An electronic stethoscope has been used to record heart sounds, giving rise to a dataset at 44 kHz and converted to 4 kHz.

2) *Segmentation*: The second step of pre-processing is a segmentation method aimed at identifying the heart sound components S1 and S2 and timing interval between them. Although the detection can also be manual, we used to identify S1 and S2 with an automatic procedure. The segmentation method is based on the timing between high amplitude components. The fact that the time interval that occurs between S1 and S2 (systole) is always less than the one between S2 and S1 (diastole) is the basis for this process. Heart sound signals still have very complicated patterns, with numerous small spikes that have little impact on diagnosis but may influence the location of S1 and S2. Peak conditioning was performed for the obtained peaks using wavelet transform, which enabled the process of cycle detection. We used the Wavelet transform based on Complex Morlet Wavelet (CMW) for finding peak locations. CMW is a kind of Wavelet transform, which are a powerful tool in time-frequency analysis for PCG signals (see figure 2).

K-means uses an iterative method that minimizes the sum of distances from each object to its cluster centroid, over all clusters. Each class of heart murmurs contains distinctive

information in time and frequency domains. This stage involves the extraction of each cardiac cycle of the PCG signal, the formed after the peak detection and conditioning stages. As systolic (S1-S2) and diastolic (S2-S1) murmurs occur within the time intervals that were calculated by the peak conditioning process, these time intervals were clustered into two clusters [9].

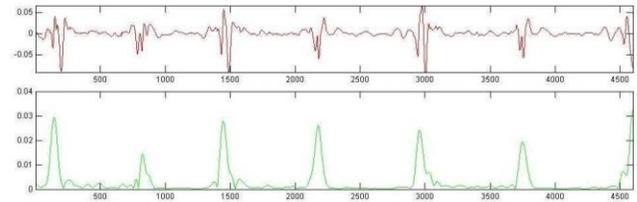


Fig.2. Peak detection: (top) original signal (bottom) coefficients line

Clusters 1 and 2 occur consecutively and indicate a single cardiac cycle. The smaller time interval of each cycle was then identified as systole while the other interval was identified as diastole. After peak detection and condition, cardiac cycle is identified by using k-means, a non-hierarchical clustering algorithm in which observation are divided in k mutually exclusive clusters. We extracted each single cycle of PCG signals using clustering (figure 3).

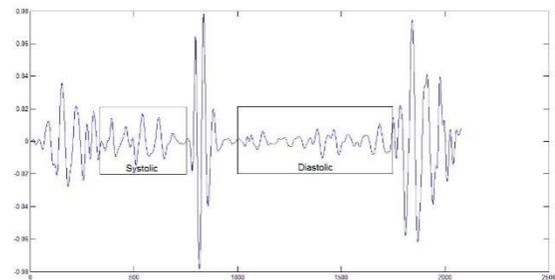


Fig.3. Sample of signal with systolic and diastolic murmur

### B. Feature extraction

This phase is focused on extracting signal features that better highlight the properties of the PCG signal, with the goal of identifying those that are more suitable for classification purposes. It consists of two major steps: feature extraction and feature selection. In the former step we extract several features including Variance, Peak to Peak, Energy Shannon, Bispectrum.

The latter step (i.e. feature selection) was aimed at reducing the size of the feature vector. In particular, we used gains and variable importance in CART to measure the score each of variable. To calculate the importance score of a variable, CART looks at the improvement measure of each variable, in its role as a surrogate to the primary split. The values of these improvements are summed over each node and are scaled according to the best performing variable. In particular, the variable with the highest sum of improvements is scored 100, while other variables have lower scores. Variable importance scores (VIS) are summarized in Table 1. It lists all variables used and not used in the tree building process. A score is attached to each variable, and is based on

the improvement each variable makes as a surrogate to the primary splitting variable. Variable importance measure highlights variables whose significance is masked or hidden by other variables in the tree building process.

TABLE I. SCORE OF VARIABLE IMPORTANCE

No	Feature	Improvement	VIS
7	Variance	0.30724	100.0000
2	Peak to Peak	0.25342	89.0333
4	Shannon Energy	0.04802	53.5714
6	Wigner bispectrum	0.04802	53.3577

As Shannon energy and Wigner distributions have been very important for improving the classification, let us spend a few words on these techniques.

*Shannon Energy:* Shannon energy is another applicable method which we made use of. The calculation of the Average Shannon Energy is based on signal segments. Therefore, here we segment the data, 0.02 seconds and with a 0.01- second signal segment overlapping throughout the signal. The average Shannon energy is calculated as (see [10] for more information):

$$E = -x^2 \cdot \log x^2 \quad (1)$$

$$\text{absolute value } E = |x| \quad (2)$$

*Wigner Bispectrum:* Wigner high-order spectrum is an extension of Wigner-Ville distribution. It keeps the advantages of Wigner-Ville distribution and has also the advantages of High-Order Spectra. High-Order Spectra have been widely used in the non-gauss and non-stationary realm, which is quite applicable to PCG signals. In particular, by combining Wigner-Ville distribution, we could get the time-frequency characters at the same time. The study has proved that under low SNR circumstances, the Wigner bispectrum is better than Wigner-Ville distribution. The second order Spectra of Wigner-Ville Distribution of signal  $x(t)$  is finally defined as follows [1]:

$$W_{2x}(t, f_1, f_2) = \int_{\tau_1} \int_{\tau_2} x^* \left( t - \frac{1}{3}(\tau_1 + \tau_2) \right) x \left( t + \frac{1}{3}(2\tau_1 - \tau_2) \right) \exp(-j2\pi(f_1\tau_1 + f_2\tau_2)) d\tau_1 d\tau_2 \quad (3)$$

### C. Classification and Regression Trees

Classification of congenital heart defect is novel application of CART for clinical and physiological data. CART developed by Breiman et al. (1984), is a nonparametric statistical method that creates binary decision trees. It is a step-by-step process in which a decision trees are constructed by either splitting each node on the tree in two daughter nodes.

The realistic objective of partitioning is to find partition  $s$  of the data such that terminal nodes are as such homogeneous

as possible. The quantitative measure of node homogeneity is called impurity function. The simplest idealization of the impurity function is the number of patients who meet an objective criteria divided by the total number of patients in the node. Ratios close to 0 or 1 are considered more pure.

To partition a node, CART examines all possible splits of the explanatory variables. In general, the number of splits for ordinal or continuous variables is 1 minus the number of distinctly observed values. A potential split is judged by its reduction of the impurity function for both daughter nodes it creates. The partitioning iteratively continues by splitting each node in two daughter nodes and continues until the tree is saturated that is, until no further partitions can be found [11].

The decision tree for predicting heart murmurs is reported in figure 4. We start at the top of the tree and follow different branches, depending on conditions involving the predictor variables. Trees with multiple layers of splits may be conceptualized as describing interactions between predictor variables. Once we arrive at an end-point of the tree, we used 8 nodes and variables classified in two classes (classes 0 and 1 were normal and pathological murmurs respectively [12]).

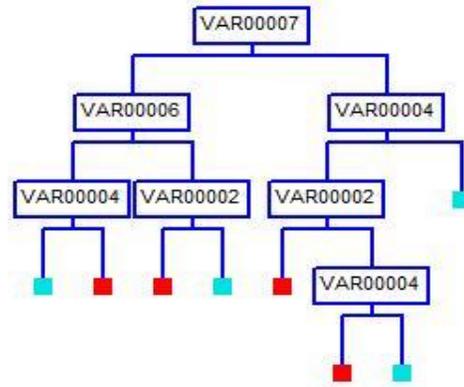


Fig.4. Illustration of decision tree structure.

We calculated the likelihood ratio (LR) to obtain sensitivity and specificity on a tree, defined as follows:

$$LR+ = \frac{\text{sensitivity}}{1 - \text{specificity}} \quad (4)$$

$$LR- = \frac{1 - \text{sensitivity}}{\text{specificity}} \quad (5)$$

The interpretation of likelihood ratios is intuitive: the larger the positive likelihood ratio, the greater the likelihood of heart disease; the smaller the negative likelihood ratio, the lesser the likelihood of congenital heart defects.

### III. EXPERIMENTAL AND RESULTS

In this study, a biomedical system based on variance, peak to peak, Shannon energy and bispectrum was developed in order to diagnose two different heart sounds. A total of 110 heart sounds (normal and pathological) were studied. In this section, we present the results of the application of the above

proposed classification and regression trees technique. K-fold cross validation (K=8) has been used as training and test strategy.

Classification results of the CART are displayed by confusion matrix. Results are shown in Table 2 in the form of a confusion matrix, together with percentage classification accuracy. It can be seen that out of 55 normal signals, 53 were correctly classified as normal, and 2 were misclassified as pathological. Similarly, out of 55 pathological signals, they were correctly classified as pathological without misclassification. A detailed analysis of the misclassified example showed that it was in fact very difficult to classify, even by human experts.

TABLE II. CLASSIFICATION RESULT OF CONGENITAL HEART DEFECTS IN NEWBORNS

Actual Group	Normal	Pathological	Percent Correct
Normal	53 (96.36%)	2 (3.64%)	96.36%
Pathological	0 (0.00%)	55 (100%)	100%
Average/Overall	110		98.18%

Summarizing, 98.18% accuracy, 96.36% sensitivity and 100% specificity were obtained by CART, when used to distinguish between the 110 normal and pathological heart murmurs in newborns.

Let us point out that, for this system, both high sensitivity and specificity are important. In particular, high sensitivity reduces the number of newborns with normal (innocent) murmurs who are identified as pathological murmur and sent to echocardiogram for further testing. More importantly, high specificity reduces the number of newborns with pathological murmurs that are identified as innocent murmurs and have been released with a potentially deadly heart condition. For each fold, learning has been performed in two steps: growing and pruning. It is worth noting that pruning has been performed provided that decision tree error curve did not trespass the threshold of 1%.

The CART decision tree error curve archived automated growing of a too large tree, followed by automated pruning to find the right-sized tree [13]. The rationale for the growing/pruning process is illustrated in the error curve (fig. 5).

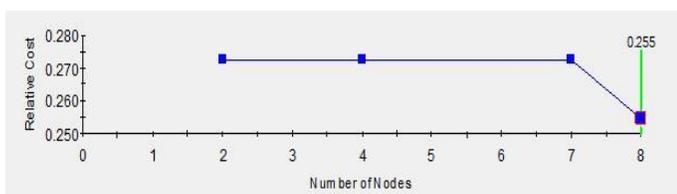


Fig.5. CART decision tree Error curve.

Figure 6 shows a curve which outlines the relationship between classification errors and tree size. The scale is always between 0 and 1, so it is called a relative error curve. A tree with a relative error of 0 or nearly 0 is usually too good to be true. The proposed model shows excellent performance for application of diagnosis of congenital heart defects. In a

Receiver Operating Characteristic (ROC) curve for a binary classification problem, the true positive rate (Sensitivity) is reported as function of the false positive rate (1-Specificity) for different cut-off points. ROC curve is reported in Figure 6 for normal and pathological murmurs.

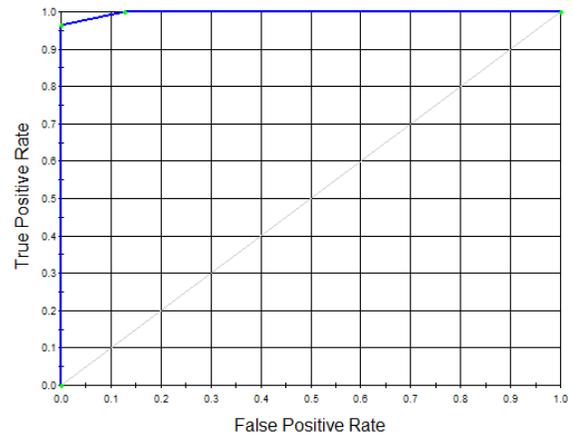


Fig.6. ROC curve of normal and pathological murmurs.

A predictive model with perfect performance has an area under ROC curve equal to 1. We obtained, on average, an accuracy of 0.99 the ROC curve highlights the excellent performance of CART to discriminate of heart murmurs.

#### IV. CONCLUSIONS

In this paper the system which is able to differentiate between normal and congenital heart defects using intelligent techniques is discussed. The intelligent diagnostic system proposes novelties in both segmentation of heart sounds and application of CART. The intelligent diagnostic system not only helps in accurate detection it is also useful for the physician who is in charge to help newborns saving lives of many case of abnormality. This technology is for high-volume screening of newborns suspected of having a heart disease. The software system proposed in this work can be considered the first release of a diagnostic tool able to support physicians in their diagnostic task.

Further work is under way to improve feature extraction and classification and also the diagnostic system can be saved for future use on other data.

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