



# Machine Learning Methods in Medical Diagnosis

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**Abstract.** Incorrect diagnosis can significantly affect outcome of treatment of a patients, which is often caused by cognitive bias of clinicians. This paper summarises development of three common machine learning methods in medical diagnosis -- Artificial Neural Networks(ANNs), Decision Tree and Bayesian Classifier (BC). Development of novel molecular approach can improve the ability of ANN models to accurately classify cancer subtypes such as Small Round Blue Cell Tumors (SRBCTs) . Moreover, new medical equipment such as mass spectrometry can assist ANNs model in analysis of ovarian cancer. In order to achieve higher accuracy of Decision Tree in medical diagnosis, Shouman et al. examined different combination of discretization methods and Decision Tree and found that disequal frequency discretization Gain Ratio Decision Tree achieved highest accuracy. In addition, BC is more interpretable than other two classifier models because it can produce probabilistic outputs of the likelihood of a certain diagnosis or outcome.

**Keywords:** “machine learning”, “Medical Diagnosis”, “Artificial Neural Networks(ANNs)”, “Decision Tree and Bayesian Classifier (BC)”.

## 1 Introduction

The outcome of a treatment can be influenced due to errors made by clinicians in the diagnosis of a patient. Diagnosis error can be divided into two types -- wrong (incorrect diagnosis), missed (no diagnosis has ever made) [1]. Diagnosis errors can lead to unnecessary treatment which might increase medical bills. and deteriorate patients' illness [2]. Cognitive bias can contribute to incorrect diagnosis of a patient. For example, anchoring bias may influence a clinician's decision in diagnosis [3]. They may rely heavily on early information received in decision-making process. Therefore, to minimise diagnosis errors and improve quality of healthcare services, Machine Learning (ML) methods has been emerged as useful tools to help clinicians making decisions [4]. Application of Artificial Neural Networks(ANNs), Bayesian Classifier(BC) and Decision Tree in medical care is well-established. Each method is capable of classifying different types of diseases and assist clinician in medical diagnosis [5]. In addition, ANNs can handle complex multidimensional data, which is common in medical diagnosis [6]. Decision Tree can provide high accuracy of classification but with a

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simplest representation of gathered knowledge, which is easy to understand [7]. BC is easy to understand because it gives detailed explanation of how classification is carried out [8]. This paper aims to summarise development of these three common ML methods which has been used for classification tasks in diagnosis and provide direction for future research.

## 2 Artificial Neural Network (ANN)

ANN is a information processing system which imitates structure of neural network in human brain and can help improve accuracy of medical diagnosis [9]. ANN can be trained to recognise and categorise complex patterns, therefore, ANN can help improve accuracy of identification and categorisation of medical condition.

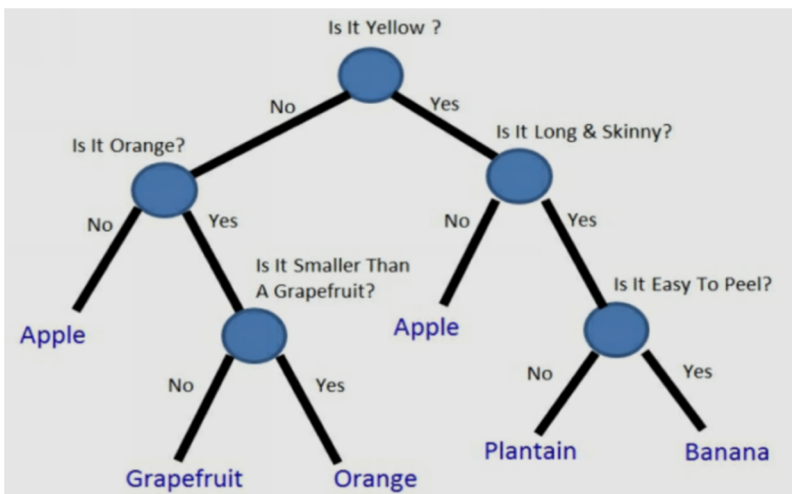
### 2.1 Cancer

Cancer now becomes a global health problem because it is a leading cause of death worldwide, constituting nearly 10 million death in 2020 [10]. Many cancers can be cured if detected early and treated effectively [10]. Therefore, a rapid and correct diagnosis is crucial for treatment of cancer such as selecting suitable treatment strategies. ANNs began to be used in the late 1990s as a computational tool for distinguishing cancer types and predicting development of cancer. Recently, many technological advancements have facilitated the application of ANNs in medical diagnosis. Firstly, the introduction of novel molecular approach such as gene-expression profile, along with use of ANNs model, has been used to broaden possibility of identifying specific types of cancer [5]. For example, Small Round Blue Cell Tumors (SRBCTs) are a group of highly malignant tumors that mainly occur in children [11]. These tumors are characterized by small, round, blue-staining cells when viewed under a microscope after staining with hematoxylin and eosin. There are four types of SRBCTs -- Neuroblastoma (NB), Rhabdomyosarcoma (RMS), Ewing Sarcoma (EWS), Non-Hodgkin's Lymphoma (NHL). However, clinicians often have difficulty in recognising and identifying these cancers [11]. For example, diagnosis of EWS relies on immunohistochemical evidence of MIC2 expression and lack of expression of the leukocyte common antigen CD45. Reliance on evidence of MIC2 can result in misdiagnosis, as MIC2 expression can also be found in other tumor types, such as rhabdomyosarcoma (RMS) and non-Hodgkin's lymphoma (NHL) [9]. Khan et al. trained a ANN model to recognise different cancers within the four SRBCT categories using gene-expression data[9]. This ANN model can correctly identify all samples into one of the four categories and genes most relevant to the classification. Gene-expression data provided detailed molecular profile of a tumor, allowing for more precise identification of sub types of cancer. Moreover, more advanced methods such as mass spectrometry can provide more detailed information about molecular composition in the ANNs analysis [12]. An ANN classifier, when trained on the most informative data points on mass spectrometry curve, can achieved a overall sensitivity of 99.8% and specificity of 96% [12]. While new medical equipment can help improve sensitiv-

ity and specificity, it should be noted that we should develop new philosophy of medical diagnosis and new machine learning algorithms rather than rely on new medical equipment and new technology or approach in future development of machine learning in medical diagnosis [13]. Philosophy of medical diagnosis includes how clinicians process and interpret medical information such as symptoms and patient history. New development of philosophy can possibly reduce diagnosis error. Moreover, new algorithms can be developed to handle complex data patterns and provide highly accurate predictions, enhancing diagnostic precision. Medical equipment and new technologies such as novel molecular approach can just serve as supportive tools.

### 3 Decision Tree

Decision tree is a supervised learning algorithm which inputs a object or situation with a set of properties and outputs a yes/no decision [14]. One of the biggest strengths of decision trees is that Decision Tree can provide very high accuracy on clinical data with simple structure, which is easy to understand [15]. The resulting model can be visualized as a tree structure where each internal node represents a decision based on a feature, each branch represents the outcome of that decision, and each leaf node represents a final classification or prediction. This structure makes it easy for clinicians to understand how decisions are being made. Figure 1 presents a simple example of a Decision Tree, illustrating how it classifies different types of fruits based on their features:



**Fig. 1.** Example of Decision Tree.

Source: <https://pantelis.github.io/cs301/docs/common/lectures/decision-trees/>

However, it should be noted that there are three factors that influence accuracy of Decision Tree. Firstly, Discretisation methods and decision tree types can affect accuracy and performance of Decision Tree model. Shouman et al. examined different

combination of discretisation methods, decision tree types and voting techniques in order to find which combination provide higher accuracy in the diagnosis of heart disease [16]. Three Decision Tree types are: Information Gain, Gini Index, and Gain Ratio. There are four Discretisation methods : chi merge and entropy supervised discretisation, equal-width interval and equal-frequency unsupervised discretisation. The highest accuracy 84.1% was achieved by using equal frequency discretization Gain Ratio Decision Tree [16]. Most previous research use Decision Tree based on Gain Ratio and binary discretisation but this research systematically tested different combination of different combination of discretisation methods, decision tree types.

In addition, choice of input subset of feature also plays a important role in accuracy of Decision Tree. Therefore, features selection becomes a crucial issue in establishing Decision Tree model. Liu & Wu improved Decision Tree model by analysing correlation between features in a breast cancer datasets and testing independence between the features by using chi-square test, which can help to identify the most important features in a breast cancer datasets [17]. Results of their study showed that the Decision Tree model achieved nearly 95% classification accuracy for the four selected subset of features. These four features outperformed other feature subsets in terms of both highest classification accuracy and average classification accuracy. Moreover, integrating features of breast cancer classification such as "tumor thickness", "uniformity of cell shape" by correlational analysis and independence test is useful for diagnosis, which implies that these four features should receive more attention from clinicians during diagnosis [16].

#### **4 Bayesian Classifier(BC)**

Bayesian Network is a statistical model which is based on Bayesian Theorem for classification tasks. It classifies data into categories by calculating the probability that a data point belongs to a particular class, given the observed features of the data [18]. There are three types of BC: Gaussian Naive Bayes, Multinomial Naive Bayes, Bernoulli Naive Bayes and Naive Bayes Classifier. BC is more interpretable than most advanced methods such as Decision Tree, ANNs. For example, Kononenko et al. compared performance of six algorithms on eight medical datasets [19]. Findings of their study suggested that the Naive Bayesian classifier provides higher explanation ability than other algorithms tested, including K-nearest Neighbor Algorithms(KNNs), Decision Tree (Assistant-R, Assistant-I).The reason is that BC can provide probabilistic outputs, which can be understood by clinicians as the likelihood of a certain diagnosis or outcome [19]. ANNs and deep neural networks, are often considered "black boxes.", can make highly accurate predictions but don't provide clear reasoning behind their decisions [19].

#### **5 Challenge and Ethic Consideration**

ANNs, often act as black box, usually showing low explanation ability and making their results difficult to interpret [20]. This lack of transparency and openness may

hinder ANNs' acceptance in clinical setting . For example, through the analysis of patient data, ANNs and Decision Tree can generate patient-specific insights and offer recommendations for patients, enabling tailored treatment plans. However, if the AI tools being used show low explanation ability for the results generated, the recommendations provided by them are unlikely to be accepted by clinicians or healthcare providers. In addition, AI systems often make decision that could be biased or discriminatory [21]. This means that results generated by AI systems can be skewed, leading to unfair treatment of certain patients. Moreover, combining AI algorithms into current clinical workflow can be challenging. Integrating AI algorithms into existing clinical workflows is indeed challenging, encompassing aspects such as workflow disruption, compatibility with existing systems such as detection system of breast cancer [22].

## 6 Conclusion

In conclusion, this paper discusses how ANNs, Decision Tree and BC can improve accuracy of medical diagnosis. They both show high classification accuracy and good performance in medical diagnosis. Firstly, this paper introduces development of application of ANNs in medical diagnosis, including when it is used as a tool in medical diagnosis and how new technologies improve accuracy of ANNs analysis in medical diagnosis. In addition, this paper discuss factors which affect accuracy of Decision Tree and efforts scholars made to achieve higher accuracy. Compared to other advanced ML methods, BC show higher explanation ability as it can show the likelihood of a certain diagnosis or outcome. This study offers a holistic understanding of how ANNs, BC and Decision Tree perform in medical diagnosis and how new technologies developed improve their application in classification tasks of medical diagnosis. In the future, AI developers should train AI algorithms using unbiased data and improve openness and explainability of 'black box' AI algorithms such as ANNs. Healthcare providers and clinicians need to ensure that efficiency is maintained and existing system is not disrupted when integrating AI algorithms into clinical workflow.

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