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A Multilayered Framework for Analysis and Prediction of Haemophilia Using Machine Learning Techniques Based E-Health System

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Abstract

Technology and information-based e-Health systems are essential to the delivery of healthcare; these include teaching and learning programs as well as remote consultation services that have been widely implemented to support many medical specialities and their corresponding remote patient populations. Because of this, e-Health systems are being utilised more and more as a passive as well as an active technique to address issues (such staffing shortages, lack of resources, and lack of knowledge) in the healthcare sector and have been shown to significantly enhance the general health of many nations. While numerous e-Health systems have been created and employed to carry out diverse functions in the medical field, including remote consultation, hospital administration, electronic health record administration, and health awareness, a dearth of research has been found concerning the analysis and prediction of haemophilia in the context of e-Health systems. Because of this, a multilayered framework for the investigation and prediction of haemophilia using machine learning techniques was developed in response to the observation.

***Keywords*:** Haemophillia, e-Health, Machine Learning, Tele-Medicine, Prediction

Introduction

The utilisation of Information and Communication Technologies (ICT) in healthcare, such as networks, the internet, computers, and wireless, has given rise to the wonderful concept of electronic health, or e-Health [1]. The effective adoption of e-Health has revolutionised the delivery of healthcare by making it possible to store medical records, retrieve information and records, and transmit medical services digitally. Furthermore, a number of countries' overall health has significantly improved when e-Health has been used consistently in the healthcare sector [2]. Aside from this, e-Health systems are being used more and more as a passive and active way to reduce various hurdles in the healthcare industry (such a lack of financing, staff, and awareness). All of these reasons have led to the replacement of traditional methods of providing health services in a number of sectors, including medical learning, medical consulting (including patient care, appointment scheduling, and record administration), and health management, by e-Health solutions [3]. Even while e-Health is still a relatively new way of using communications technology to deliver medical services, it has been used for more than thirty years in one way or another.

Early in the 1960s, the National Aeronautics and Space Administration (NASA) coined the phrase "telemedicine" to describe the concept of e-Health [4]. Following that, the first international telemedicine program, "Space Bridge to Armenia/Ufa," was implemented by the National Aeronautics and Space Administration (NASA) in 1989 to conduct telemedicine consultations. The global digitisation of the health sector is now referred to as "e-Health" [5]. Furthermore, the enhanced performance, efficiency, and cost-effectiveness of e-Health is fuelling the growing urge to develop new e-Health systems that will impact every aspect of healthcare and society [5][6]. Although, different kinds of e-Health systems have been developed and utilized to accomplish various tasks in various areas of the medical domain, such as remote consultancy, electronic health record management, hospital management, and health awareness; however, rarity of work has been identified in context of e-Health systems in respect to analysis and prediction of Haemophillia [7][8]. Therefore, the finding served as inspiration for the creation of a multilayered framework that uses machine learning techniques to analyse and forecast haemophilia.

Haemophillia

A mutated gene on the X chromosome causes haemophilia, an X-linked recessive genetic characteristic or disorder that a child may inherit from their parents. Because they have one functioning haemophilia gene and one non-functional gene, females with the XX genotype are carriers of the condition, whereas males with the XY genotype are affected by haemophilia and are referred to as sufferers. However, if there is a mutation on both X chromosomes that results in bleeding symptoms, females may also occasionally be impacted. This condition is known as symptomatic carrier or affected patient of haemophilia [9]. Because males only have one X chromosome, it can be concluded that the condition is caused by a change in the copy of a gene on that chromosome, but females need mutations on both X chromosomes to be impacted. On the other hand, girls who have a single X chromosomal mutation will either be carriers or patients of haemophilia [9]. In addition, it is divided into three subtypes. One of them is haemophilia A (HA), haemophilia B (HB), and haemophilia C (HC), which are brought on by either an excess or a lack of clotting factors VIII, IX, and XI in the blood, together with platelet-rich plasma, or PRP [9]. It has been discovered that, in terms of haemophilia prevalence, haemophilia A (HA) affects one in 5,000 male live births, while haemophilia B (HB) affects one in 30,000 male live births. Sometimes, the severity of different types of haemophilia is further classified into three categories: mild, moderate, and severe, based on the amount of clotting factor activity detected in a subject's blood [9]. The adult classification method states that a factor activity level of 6% to 49% indicates mild haemophilia, a factor activity level of 1% to 5% indicates moderate haemophilia, and a factor activity level of less than 1% indicates severe haemophilia [10]. Abnormal bleeding, such as prolonged bleeding, and frequent spontaneous bleeding in response to minor wounds are common in severe haemophiliacs [10]. While spontaneous bleeding typically occurs in the joints, it is not uncommon for internal bleeding to occur in the kidneys, brain, or digestive system. Chronic joint bleeding can irritate the joint and destroy the cartilage, causing irreversible damage to the joint [10]. The symptoms of small injuries, such as muscle haematomas and internal bleeding, may appear days after the initial trauma. Even though haemophilia is a hereditary condition, approximately one-third of infants diagnosed with the disorder have no known family history [10]. A small number of new cases are also observed in this setting. Despite this, just 33,000 Americans are estimated to be haemophilic, according to a research released in August 2022 by the US Centres for Disease Control and Prevention. As a result, haemophilia is not seen as a common illness. Furthermore, according to the findings, roughly 10 out of 100,000 people have haemophilia A, 3 out of 100,000 have haemophilia B, and 1 out of 100,000 has haemophilia C [11]. Thus, all of the information offers compelling justification for the framework's creation, which will be used to analyse and forecast haemophilia using machine learning techniques.

Machine Learning

Within the field of artificial intelligence, machine learning focuses on developing algorithms and models that enable computers to independently learn from data. It analyses data, makes inferences, and projects results using statistical techniques. Therefore, the models and methods that enable computers to learn from data or past experiences in order to maximise performance criteria are the main emphasis of machine learning. Combining machine learning techniques with new computing technology improves efficiency and scalability. Machine

learning is used in numerous disciplines, including natural language processing, recommender systems, picture and audio recognition, and many more [12]. The ability of contemporary machine learning models to predict anything from stock price fluctuations to disease outbreaks means that machine learning innovations have the potential to revolutionise a number of industries, including the health sector, which significantly improves public health through the use of e-Health [12].

Role of Machine Learning in e-Heath

One important application of machine learning in e-Health is diagnostics. Machine learning is revolutionising the field of e-Health and has opened up various choices and breakthroughs for improving healthcare outcomes. After being trained on enormous datasets of test results, symptoms, and medical records, these models can be trained to identify patterns and make highly accurate diagnosis predictions [12]. This facilitates early disease diagnosis, which may help save lives, and aids physicians in diagnosing patients more swiftly and precisely. Another area of e-Health where machine learning shines is personalised medicine.

By analysing previous patient data, appointment histories, and resource usage trends, machine learning algorithms can efficiently distribute resources, organise appointments, and estimate patient demand [12]. Higher patient satisfaction and cheaper costs are eventually the results of shorter wait times, more staff, and better resource allocation. Machine learning is transforming e-Health by facilitating personalised therapy, streamlining patient monitoring, enhancing diagnosis, and streamlining healthcare processes. Thanks to ongoing advancements in data collecting and machine learning algorithms, e-Health is poised to make significant strides in offering efficient, personalised, and high-quality healthcare services to people worldwide [12]. Because machine learning makes it simpler to conduct complex data analysis, forecast, and make decisions for a range of healthcare applications, such as drug discovery and development, disease prognosis and risk assessment, medical imaging analysis, and health behaviour analysis, the field of e-Health is thereby greatly advanced [12].

Machine Learning and e-Health

Machine learning is revolutionising the field of e-Health and has created new opportunities and breakthroughs for improving healthcare outcomes [13]. Diagnostics is a key use of machine learning in e-Health [13]. After being trained on enormous datasets of test results, symptoms, and medical records, these models can be trained to identify patterns and make highly accurate diagnosis predictions [13]. This facilitates early disease diagnosis, which may help save lives, and aids physicians in diagnosing patients more swiftly and precisely. Another area of e-Health where machine learning shines is personalised medication [13]. Machine learning is investigated in the following fields:

The Discovery and Development of Drugs: Large-scale molecular and genetic data can be analysed by machine learning algorithms to find promising drug candidates, assess their efficacy, and streamline the drug discovery process [13].

Calculating Risk and Prognosis of Disease: In order to estimate patient outcomes, predict the course of diseases, and evaluate the risk of complications, machine learning algorithms can evaluate clinical parameters and patient data [13].

Study of Health Behaviour: In order to evaluate lifestyle characteristics, track treatment plan adherence, and offer tailored recommendations for healthy behaviours, machine learning algorithms can evaluate wearable device data and patient behaviour patterns [13].

Evaluation of the Model: Following training, the effectiveness of the machine learning models is assessed using suitable evaluation measures to make that the models function as intended and have good generalisation to new data [13].

Making Predictions and Decisions: The models can be used to enhance decision-making in a variety of e-Health applications by predicting outcomes and completing assessments. Customising healthcare interventions, finding patterns in medical imaging, and recommending a course of therapy are all examples of e-health [13].

Motivation for Research

Gene inheritance and mutation are the usual causes of haemophilia diseases [14]. Over time, the prevalence of hereditary prenatal illnesses like haemophilia is steadily increasing. According to the World Health Organisation (WHO), there is a need for increased focus to prevent these illnesses because they are common. More than 300,000 newborns are born with serious haemoglobin abnormalities every year [14]. Only 11,586 cases of haemophilia A have been documented, despite the possibility that there are up to 50,000 cases. Additionally, data indicate that 3,000–5,000 babies are born with this problem every year [14]. This makes it a significant global concern in the medical community. Thus, new approaches or instruments are required to deal with this at its most fundamental level. so that initiatives aimed at enabling parents to take charge of the early diagnosis, treatment, and prevention of haemophilia can be undertaken.

Review of the Literature

The use of information and communication technology (ICT) in the healthcare sector for patient care, research, student education, illness tracking, and public health monitoring is known as e-Health, according to the World Health Organisation [15]. Stated differently, it can be argued that e-Health systems perform a variety of healthcare-related functions in spite of insufficient resources (including funding, staff, and infrastructure) [15]. Furthermore, it provides patients with essential medical treatments through electronic media [15]. However, the phrase "e-Health" actually has a wide range of meanings that may be found in older literature. In 2010, Everett J. and Kerr D. defined e-Health as a digital tool that transmits the data required for patient care, health education, and industry administration [16]. In a similar vein, an e-Health system was defined in 2010 by Jim Snaith, Dr. Peter Hayward, Jon Avalon, and Professor Victor Lane as a tool to support healthcare workers by organising and disseminating expert information inside the company [17]. Furthermore, Sharma Kalpa (2012) described e-Health as a helpful tool for improving lives. Based on their observations, other researchers have provided a number of descriptions of e-Health in this manner [18].

The extensive evaluation of the literature on e-Health has revealed a number of environmental issues in the healthcare sector (such as a shortage of staff, resources, and infrastructure), as well as how they affect social health [15][19]. Researchers from all around the world are working constantly to address all of these issues in the healthcare business. Because of their persistent work, e-Health has emerged as a digital instrument for lowering a variety of healthcare issues and raising the standard of care. The literature review affirms that e-Health systems are an acknowledged field of study, but it concentrates on the different e-Health systems that have been created in any format (e.g., mobile applications, websites, portals, models, frameworks, methods, frameworks, and tools) to cater to different healthcare sectors and provide e-Health services to underprivileged communities in far-off places [15]. Consequently, the section addresses the various systems that are mentioned in the literature study [20] and some of which are also shown in Table 1. Aside from this, this section has covered prospective problems and domains where innovative systems could be developed.

Table 1: Systems for Remote Consultation or Telemedicine

S.No	Systems	Year	Services	#Ref
1	Tele-surgery	2005	Provides remote medical staff with access to professional knowledge so they can finish the surgical task more rapidly.	[20]
2	Tele-dermatology	2005	Provides clinical and laboratory data to dermatologists who are located far away, enabling them to utilise it for patient diagnosis and treatment.	[19] [21]
3	Tele-Psychiatric	2008 2014	Provide telepsychiatric services to patients suffering from mental disorders or neurological conditions.	[22] [23] [24]

4	Tele-homecare	2009 2012	In order to help elderly individuals keep an eye on their health at home, the study presents the idea of geriatric healthcare.	[22] [25] [19]
5	Tele-Health	2010 2012	Delivers high-quality medical care to children from Indigenous communities.	[22] [26] [27]
6	Tele-radiology	2013	Sends text and radio graphic images electronically across locations for medical consulting.	[28]
7	eMedical Help	2015	Offers assistance for stress diagnosis.	[29]

All of the systems included in Table 1 electronically give medical professionals in an emergency situation as well as patients in remote areas access to expert knowledge or health services [19, 20, and 23]. All these methods lowered the complexity and amount of time that the public had to spend. Additionally, it has been discovered that telemedicine and e-Health systems that offer remote consultations are more beneficial in regions where there is a known lack of medical resources (e.g., money, infrastructure, manpower). Additionally, it enables online appointment scheduling for medical consultations.

The creation of web-based e-Health systems that aim to shorten patient wait times, such as the Online Registration System (ORS) [31], Health You Card [32], and Practo [33], has made this feasible. Apps such as Practo [33] are growing more and more popular among the general population since they save time, which increases the need for these systems. This fact therefore provided the impetus to discover the unknown region for which a state-of-the-art e-Health system can be built; hence, more investigation was conducted to achieve the objective. As the literature review continues, it is evident that e-Health systems, with their wide range of system types, offer the general public much more in underserved areas in terms of services like online training, hospital administration, digital health record keeping, health education, and resource monitoring [19, 20, and 23]. A portion of them have already been covered in this part; they are tabulated to make things easier to grasp and are arranged or clustered according to the services they offer.

Electronic Health Record (EHR) and Hospital Management Systems [34] constitute the first cluster that was identified during the literature review and is discussed in this section. These systems include all those that offer services such as maintaining patient records and efficiently managing all hospital operations. Thus, it can be concluded that hospital operations and resource management [35] are significantly impacted by Hospital Management Systems (HMS) and Electronic Health Records (EHR) systems [34]. This is seen in several of the systems shown in Table 2, which offers the ability to record and store patient medical data for later examination and care.

Table 2: Systems for Hospital Management and Electronic Health Records

S.No	Systems	Year	Services	#Ref
1	Electronic Health Record System (EHR).	2005	Beneficial in gathering and preserving patient medical records for later use.	[36] [37]
2	Health Vault	2015	Patients may gather store, use, and exchange health information thanks to the system.	[38]

3	Health Memo	2015	Electronically maintained health records are available for users to upload.	[39]
4	Health PIE'	2015	Enables users to upload personal health records and set reminders.	[40]

Additionally, a number of systems, such as Health Net, enable hospital departments to share medical data, such as test results, medications, and medical histories [41]. Additionally, Table 2 illustrates how systems like as Health PIE, Health Vault, and Health Memo facilitate the electronic uploading of personal health records; thus, notable advancements have been noted in the field of these systems. In contrast, Health Awaring e-Health Systems was another class or cluster of systems observed throughout the literature review. The systems that educate the general public about different diseases, their symptoms, accessible treatments, and disease patterns in an effort to empower people to take control of their health are all included in this cluster. Table 3 provides a summary of these systems' descriptions.

Table 3: List of Health Awaring e-Health Systems

S.No	Systems	Year	Services	#Ref
1	Cardio Pulmonary Resuscitation (CPR)	2012	This smartphone app offers resources for averting or recovering from cardiac arrest.	[42]
2	Mswasthya	2014	Offers a variety of services, including hospital searches, immunisation alerts, and diabetes tracking.	[43]
3	Health Central.Com	2015	Provides information to patients and carers to enable improved health outcomes.	[44]
4	Isabel Symptom Checker	2015	List the various illnesses along with their symptoms.	[45]
5	Veegilo	2015	Gives the ability to quickly view disease trends and patterns.	[46]
6	e-medicine	2016	Gives information on medications used to treat illnesses.	[47]

Table 3 presents an examination of each system. It is clear from this study that the systems greatly enhance public knowledge [48] by providing users with comprehensive information about a range of conditions, enabling them to make informed decisions.

Furthermore, e-Health systems provide health professionals with an online platform to access vital knowledge and information in tandem with the creation of new systems. Electronic teaching and learning elements are provided by these systems, which are collectively referred to as Teaching and Learning e-Health Systems. This cluster, as its name implies, houses all the systems that allow personnel, medical students, or remote workers to get online teaching, either fully or partially, in order to expedite their work. A brief overview of the numerous systems that were discovered in this context throughout the literature review is given in table 4.

Table 4: List of e-Health Systems Related to Teaching and Learning Domain

S.No	Systems	Year	Services	#Ref
1	Geochat	2011	Gives people in remote places the opportunity to communicate with community health workers.	[49]
2	Tele- mentoring	2005	Explain the meaning of tele-mentoring as giving a student access to live, interactive teaching methods and strategies in real time.	[50]
3	Mobile Alliance for Maternal Action (MAMA)	2015	Important health data about the behaviour of mothers is given.	[51]

The systems included under the teaching and learning cluster do an amazing job of teaching methods and procedures to remote medical students or interns through communication technology, even in the face of medical professionals' limitations in impoverished areas. The cluster also highlighted initiatives like the Mobile Alliance for Maternal Action (MAMA), which provides mother care and pregnancy-related services in an effort to reduce the number of pregnancies and the health issues that come with it. Under the Mother and Child Care domain, these systems specifically address issues related to pregnancy, childbirth, and newborn care, even though they are connected to the teaching and learning process. This is a serious area of worry since, according to a study of the literature, the rate of maternal or foetal deaths after delivery was greater than anticipated, calling for more research and development. This is what prompted the choice to carry out this field's research. Additional review work has been conducted in order to determine the research gap in this field. During the literature review, a variety of unique e-Health system types that assist both antenatal and postpartum patients were discovered; these are also displayed in table 5. The mother and child care category, which is further separated into two subclasses: e-health systems for prenatal or mother care and e-health systems for newborn care, is where these systems have been gathered.

Table 5: e-Health Systems in the Domain of Mother and Child Care

S.No	Systems	Year	Services	#Ref
e-Health Systems Concerned with Newborn Care				
1	Neonatology	2013	Provides time-sensitive information on neonatal care to a healthcare professional.	[52]
2	Essential Newborn Care	2014	Enhance the critical infant care knowledge and skills of medical practitioners in resource-poor communities.	[53]
3	The Kangaroo Mother Care Initiative (KMC)	2014	Encourage healthcare professionals and parents of low	[55]

			birth weight babies to share their expertise and skills.	
4	Essential Care for Every baby (ECEB)	2015	Enhance the skills and knowledge of medical personnel about critical newborn care in resource-poor locations.	[15]
e-Health Systems Related to Mother or Prenatal Care				
5	a mother-and-child care module	2010	Offers prolonged vaccination campaigns and prenatal care to the underprivileged people in the border region.	[48]
6	OB Insulin	2014	Determine the starting dosage for an insulin-requiring pregnant woman based on user input, such as the mother's weight.	[57]
7	Mobile Alliance for Maternal Action (MAMA)	2015	Important health data about the behaviour of mothers is given.	[51]
8	Verboise	2011	It provides check-up reminders, advice on how to care for your newborn and monthly updates on what to expect when pregnant.	[58]

Therefore, the term "mother and child care" refers to the organisations that offer pregnant women, recently given mothers, and their small children necessary medical care. This divides this sector into sub-parts, prenatal care and post-natal care, each of which is connected to an e-Health system. The treatment provided to pregnant mothers and their unborn children throughout pregnancy, for example, falls under the purview of prenatal care systems. On the other hand, postnatal care systems handle a woman's and her child's postpartum care.

The majority of maternal and newborn mortality occur during the prenatal and postnatal periods, making these periods of care vital areas of concern [59]. Moreover, every year roughly 250,000 babies are born with genetic disorders such sickle cell anaemia, haemophilia, and Thalassaemia since prenatal care is scarce in rural and impoverished areas [59]. The current study aims to develop an innovative system that will electronically provide prenatal services to the underserved public, particularly pregnant women, in the context of inherited disorders, particularly haemophilia. This fact motivated the selection of the prenatal care area, which falls under mother care. Due to the lack of a system in place, inherited illnesses like haemophilia, which is growing more and more frequent, cannot currently be diagnosed during pregnancy. Since severe haemoglobin anomalies affect over 300,000 newborns annually, preventative efforts need to be intensified [59]. Therefore, a number of medical diseases, including prenatal disorders brought on by the inheritance of genetic defects, will benefit from the proposed study. It will also provide parents greater control by making it easier to identify hereditary prenatal illnesses in foetuses, like haemophilia and thalassaemia [59]. Under the supervision of a trained medical practitioner, this will allow decisions to be made about whether or not to become pregnant if there is a possibility that genetic illnesses will be discovered. The data thus led to the design of a planned system that will identify and forecast the likelihood of passing on inherited disorders (such as haemophilia) from parents to offspring at the prenatal stage using the medical, family, physiological, and obstetric history of a couple planning a child. To that end, a framework for implementing the suggested e-Health system has been developed and is discussed in the section that follows in this paper.

Framework for the Prediction of Haemophilia

The framework for a prediction system for haemophilia has been proposed in the current work. The entire project is broken down into phases, which include feature selection, data collection, data prediction, and data visualisation. Since there isn't a secondary dataset on haemophilia, the original dataset has been gathered. The dataset was gathered from a range of physicians, hospitals, and healthcare facilities. The gathered data has undergone data preparation. Duplicate and null values are eliminated. The dataset is subjected to multiple feature selection methods, which divide it into three categories: normal, haemophilia disease, and haemophilia carrier. The impact of haemophilia on the patient is predicted during the prediction phase. Various graphs are used to represent the data in the final phase, the visualisation phase. Figure 1 shows the layered framework of the current work.

1. **Data Collection Phase:** Since the data set was employed to complete the suggested study, the present phase concentrated on the data collection task.In order to conduct additional experimentation through field surveys, the primary data set was gathered from medical colleges and hospitals as well as Non-Governmental Organisations (NGOs), specifically Dayanand Medical College and CMC (Christian Medical College & Hospital), Ludhiana, Punjab, India, and the Haemophilia Society Dehradun Chapter. The 501 patient records in this data collection pertain to the hypothetical disorder "Haemophilia" and include information on the patient's features, medical history, family history, obstetric history, history of symptoms, and three target classes: normal, haemophilia carrier, and haemophilia disease(Haemophilia).
2. **Feature Selection Phase:** Prior to processing, the gathered dataset is cleaned up by removing duplicate values and null values. Out of all the aspects, the relevant features are chosen, and the data is categorised into three goal classes: normal, haemophilia carrier, haemophilia disease, and haemophilia of purposed disorder. The components that have a significant influence on diagnosing haemophilia are the haemophilia carrier and the disease. Normal class, on the other hand, consists of criteria that have little bearing on the diagnosis of haemophilia.

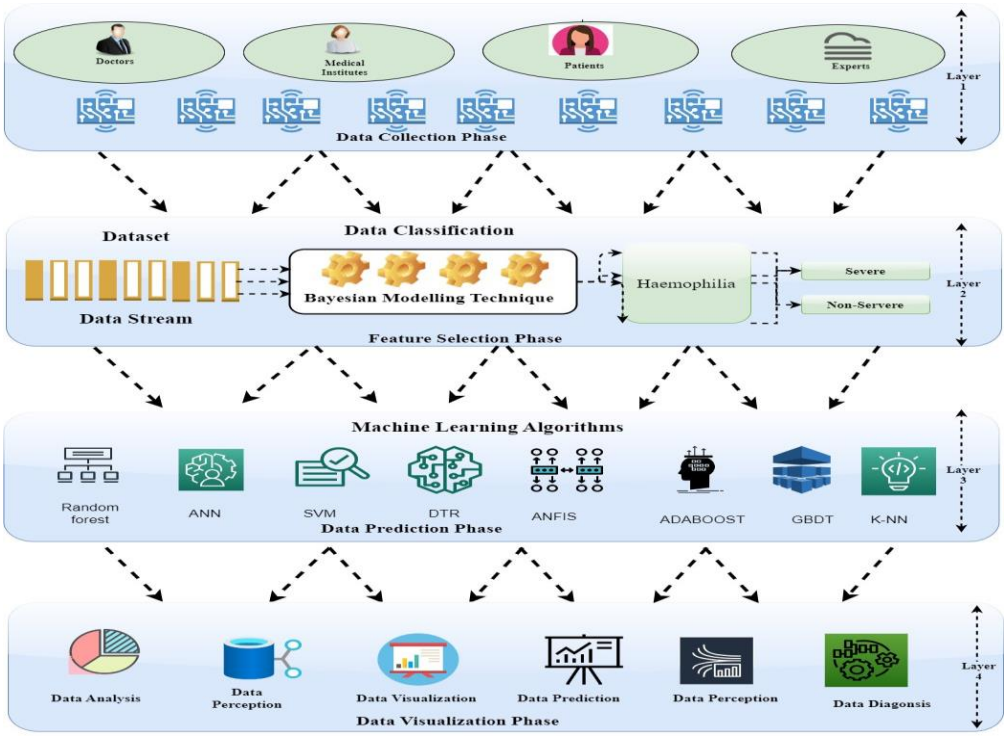

Figure1: A layered framework for the prediction of Haemophilia

Data Prediction Phase: For the purpose of developing a model, several methods, including Gaussian Naive Bayes, Decision Tree, K-Nearest Neighbour (KNN), and Bayesian Network, were implemented separately and with interest. While the model was useful in diagnosing haemophilia and classifying patients into three categories—normal, haemophilia carrier, haemophilia disease, and haemophilia—it was unable to predict the inheritance of haemophilia. Consequently, a rule-based prediction model was created using a Bayesian network, an existing rule set, and a newly developed rule set from the current study. This model was successful in predicting the likelihood of having a child with haemophilia.

Data Visualization Phase: The computed findings have been presented through the use of several pictures and graph formats during the data visualisation phase.

Conclusion

The term "e-Health" describes the digital transformation of the healthcare sector. Additionally, e-Health's increased cost-effectiveness, efficiency, and performance are driving the need to develop new e-Health systems that will impact all aspects of healthcare and society. Gene mutation and heredity are the main causes of haemophilia diseases. Over time, the prevalence of hereditary prenatal illnesses like haemophilia is steadily increasing. Taking into account the intelligent prediction system for haemophilia is crucial. The current research has proposed a tiered framework. in order for the forecast to be made with ease by adhering to the current framework. The researchers may certainly benefit from this framework, which also provides them with new opportunities.

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