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RESEARCH PAPER

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Expert System for Early Detection of Thalassemia Disease Using Case-Based Reasoning Method

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ABSTRACT

Thalassemia is a blood disorder caused by abnormalities in globin chain formation. In Banyumas Regency, the prevalence of thalassemia continues to increase yearly, while detection processes are often delayed due to limited access to medical experts. This delay impacts the effectiveness of treatment, necessitating a technology-based solution to accelerate early detection. This study aims to develop a web-based expert system for the early detection of thalassemia using the Case-Based Reasoning (CBR) method with the K-Nearest Neighbor (KNN) algorithm. The system is designed to help identify individuals who may carry the thalassemia gene trait, enabling faster and more accurate medical intervention. The system was tested using the black box method to ensure all features function properly across different user roles, including general users, administrators, and experts. Accuracy evaluation was conducted using a confusion matrix, achieving an accuracy rate of 95.23% based on 21 test data samples. These results indicate that the developed system provides highly accurate early detection and supports preventive efforts against thalassemia. Further development is recommended by creating an Android-based application to enhance accessibility for the broader community. Additionally, continuous updates to the knowledge base are necessary to improve the system's accuracy and scope in identifying various types of thalassemia. This study is expected to contribute to the prevention and management of thalassemia, increase public awareness, and support better healthcare services in Indonesia.

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1. INTRODUCTION

Genetics is a branch of science that focuses on the study of inherited traits [1], [2]. These traits are passed from parents to offspring through genes [3], [4]. Genetic testing plays a crucial role for couples who wish to understand potential genetic risks for their future children [5].This test can detect genetic disorders, such as thalassemia, which can lead to serious health conditions [6]. Thalassemia was chosen as the main focus of this research because, according to data from the Indonesian Ministry of Health, the number of thalassemia patients in Indonesia has been increasing annually. In 2012, there were 4,896 recorded cases, and as of June 2021, the number of individuals with thalassemia in Indonesia had risen to 10,973 cases [7]. Thalassemia is a disease caused by a disorder in globin chain formation [8]. This disease is characterized by red blood cells that are fragile and have a lifespan of only about 23 days, significantly shorter than the normal lifespan of 120 days [9], [10].

Based on the Hardy-Weinberg principle regarding allele frequencies from one generation to the next, it is estimated that around 2,500 babies with thalassemia major will be born each year [11]. However, if only 1,500 babies are recorded each year, one can imagine how large the number of children with thalassemia major that the country must support in the next 10 years. Data from various teaching hospitals show that the number of thalassemia major patients registered throughout Indonesia is only around 7,670 people [11]. The distribution of thalassemia major cases in Indonesia can be seen in Figure 1. This number is still much smaller than the actual estimate. This is likely due to the fairly diverse variation of gene mutations in Indonesia, ranging from very severe to mild so that transfusions are not required (asymptomatic), or due to limited knowledge of medical personnel and the lack of diagnostic laboratory facilities that cause many cases to go undetected (under-diagnosed).

Central Java has a high number of thalassemia patients, based on the results of interviews conducted by Banyumas Regional General Hospital, more than 500 patients were recorded undergoing routine treatment at Banyumas Regional General Hospital (RSUD). The high number of thalassemia sufferers is caused by limited access to expert medical personnel and the lack of

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adequate diagnostic facilities [7], [10]. Several specific obstacles that cause delays in early detection of thalassemia include:

1. Geographical Factors : People in rural areas have difficulty accessing specialized health services, because medical facilities that treat thalassemia are generally concentrated in big cities [12].
2. Economic Factors : The cost of laboratory tests and consultations with hematology specialists is still relatively high for most people, especially for families with lower to middle economic levels.
3. Lack of Public Awareness : The lack of education about the risks of thalassemia results in people not realizing the importance of screening before marriage or before having children.

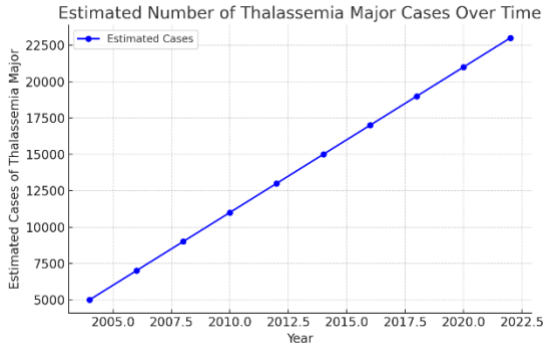

Fig. 1. Estimated number of children with thalassemia major by calculation using the Hardy-Weidberg principle

Previous studies have discussed the use of expert systems in various fields of health and technology with various methods applied. For example, research on expert systems to detect scalp diseases using the Case-Based Reasoning (CBR) method shows that this approach can help medical personnel identify the type of disease, prevention solutions, and optimal care. This study shows that the system built is able to provide early diagnosis and recommendations for effective prevention and treatment for patients [13]. In addition, other research on expert systems in diagnosing malaria and typhoid using the Naïve Bayes method also makes a significant contribution to the health sector. This system is designed to diagnose patients in real time without the need for laboratory tests [14]. In addition to its application in the health sector, the CBR method has also been applied in diagnosing photocopier damage. In this study, an expert system was developed using the PHP programming language and MySQL database to analyze and provide solutions to damage experienced by photocopiers. By adopting the CBR method, the system can compare new cases with old cases that have been stored [15].

In other health sectors, research on the diagnosis of Cerebral Palsy also utilizes the CBR method to support medical decision making. This desktop-based expert system developed using the Java programming language and MySQL database allows patients to interact with the system directly through a question and answer session regarding the symptoms they are experiencing. The diagnostic results produced include the type of Cerebral Palsy that may be suffered and its treatment options, with a certain level of probability based on the similarity of previous cases [16].

Although the various studies above have proven the effectiveness of the CBR and Naïve Bayes methods in expert systems for disease diagnosis and case analysis, this study has differences and gaps that still need to be filled. The research conducted in this study focuses on the design and development of a web-based expert system for early detection of Thalassemia disease using the Case-Based Reasoning (CBR) method. Unlike previous studies that have been more widely applied to scalp diseases, malaria, typhoid, and Cerebral Palsy, this study will develop a system specifically for early diagnosis of Thalassemia by considering differences in symptoms and differential diagnoses of diseases that have similar symptoms. Thus, this study aims to expand the scope of expert systems in supporting medical personnel and the public in identifying the risk of Thalassemia earlier and with a better level of accuracy.

Expert systems can be a technological solution to overcome undetected thalassemia cases in Banyumas. This system is designed to be easy to use and able to provide early detection [17]. Observations at the Indonesian Thalassemia Foundation (YTI) Banyumas show that this foundation is active in educating the community and providing support for patients. However, until now, YTI Banyumas has not utilized expert system technology for early detection, even though it has great potential in helping to reduce the birth of children with thalassemia genes. This study proposes the design of a web-based expert system using the Case-Based Reasoning (CBR) and K-Nearest Neighbor (KNN) methods. Case-Based Reasoning (CBR) is a method that solves new problems by using experience from previous cases [18], [19]. This method consists of four main steps: (1) retrieving similar cases from the database, (2) reusing information from previous cases, (3) revising solutions to suit new conditions, and (4) storing new cases for future use [16], [20]. This approach is effective in detecting thalassemia because it is able to learn previously diagnosed symptom patterns, so it can provide solutions based on existing case histories.

On the other hand, K-Nearest Neighbor (KNN) is an instance learning-based algorithm that classifies new data based on its similarity to existing data. In the context of thalassemia detection, KNN compares the symptoms entered by the user with previous patient datasets to determine the likelihood of someone having thalassemia [21]. The combination of CBR and KNN is expected to improve the accuracy of early detection because these two methods complement each other in handling variations in symptoms and patient characteristics [22], [23]. The purpose of this study is to help break the chain of thalassemia gene inheritance by developing an expert system for early detection, in collaboration with YTI Banyumas. With the implementation of this system, the

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public is expected to have wider access to health information, so that they can take preventive measures earlier. Early detection of thalassemia has a significant impact on public health, because it allows for faster treatment, better disease management, and reduces the birth rate of children with thalassemia. In addition, this system can also help medical personnel in improving the effectiveness of disease management for patients who have been diagnosed, so that it can extend life expectancy and improve the quality of life of thalassemia sufferers.

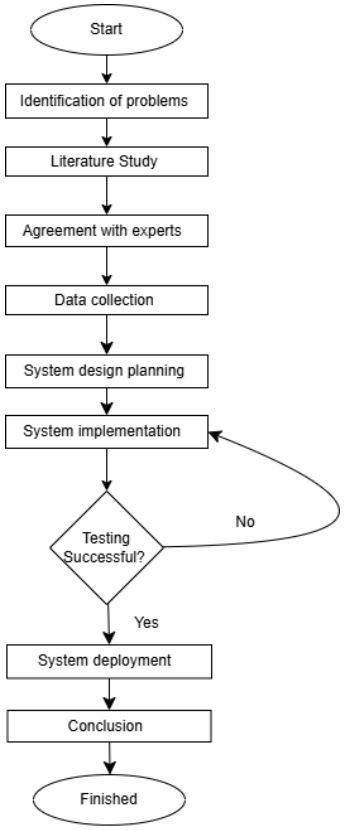

Fig. 2. Flowchart of the research methodology, illustrating the step-by-step process and framework used in this study.

2. MATERIALS AND METHOD

Figure 2 illustrates the research flow for designing and developing a web-based expert system to detect thalassemia. The process begins with problem identification, followed by literature study, expert collaboration, and data collection. The system is then designed, implemented, tested, and iteratively refined until successful. Finally, the system is deployed, and the research concludes with findings and recommendations.

A. Identification of Problem

In this first stage, researchers identify and define problems based on the context of the problems that occur. In this case, there are several main problems related to the high number of thalassemia sufferers in Banyumas Regency, which are caused by the lack of early detection and socialization about this disease. The public is generally unaware of whether they are carriers of the thalassemia gene due to the lack of information received and limited access to genetic testing. In addition, the number of medical personnel who are specially trained in handling thalassemia in this area is very limited, thus hampering the optimal diagnosis and treatment process. This condition is exacerbated by the limited medical facilities that provide early detection services, as well as the social and economic impacts borne by patients and families due to high medical costs. Therefore, this study aims to explore the factors that cause these problems and provide recommendations for increasing public awareness and strengthening the health system in Banyumas Regency.

B. Literature Study

Previous research references and relevant theories were obtained from journals, papers, online articles, and other reliable sources. This literature study was used to explore information related to thalassemia disease, symptoms, risk factors, and early detection methods. This information helps understand the problems raised in the study, especially regarding early identification of thalassemia disease and efforts to mitigate its risks. This study also highlights the application of the case-based reasoning (CBR) method and the k-nearest neighbor algorithm used in the development of a web-based expert system [24].

C. Agreement with Experts

At this stage, the researcher coordinates with an expert as a hemato-oncology specialist, namely dr. Wahyu Djatmiko, Sp.PD., KHOM. The request for cooperation is made to obtain the data needed to strengthen this research and learn more about thalassemia disease, including symptoms, early detection methods, and its treatment. Meetings with the expert will be scheduled according to agreement to ensure that the information obtained is accurate and supports the development of the expert system being developed.

D. Data Collection

Data collection in this study was conducted through face-to-face interviews with dr. Wahyu Djatmiko, Sp.PD., KHOM, a hematology and oncology specialist who has expertise in handling thalassemia. He played a major role in this study because of his competence in diagnosing and treating thalassemia patients. The information obtained includes the type of thalassemia, symptoms, severity, case base, and diagnostic threshold values. The data collected was then stored in Excel format before being integrated into the system database. In addition, this study also received guidance from Mr. Ns. Rozali Arsyad Kurniawan, S.Kep., M.Kep., a member of the Banyumas Hospital Nursing Committee, and dr. Prio Sapto Utomo, who is responsible for the medical services of Banyumas Hospital. Rozali was chosen because of his expertise in thalassemia patient care management, while dr. Prio Sapto Utomo has experience in medical services and managing thalassemia cases in hospitals. The

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combination of insights from these three experts ensures that the system developed is based on accurate medical data and is in accordance with applicable clinical practices. Explains the process of collecting data needed for research and data processing using the Case-Based Reasoning method and calculating similarity using k-nearest neighbor [25]. Table 1 shows the types of thalassemia and differential thalassemia diseases along with disease codes, which in total consist of five diseases.

Table. 1. Classification of Diseases That Can Be Detected Early in Thalassemia Detection

Code	Disease
P001	Thalassemia Mayor
P002	Thalassemia Intermedia
P003	Thalassemia Minor
P004	Anemia
P005	Polisitemia

Table 2 below shows the symptoms of the disease along with the symptom codes and weights obtained from direct interviews with experts.

Table. 2. Symptoms and Corresponding Weights for Early Thalassemia Detection Analysis

Code	Symptoms	Weight
G001	Family history of thalassemia	5
G002	Pale facial appearance	5
G003	Reddish skin on the face or body	5
G004	Cold sensation in the extremities	1
G005	Darkening or bluish skin color (Pigmentation)	3
G006	Itching, especially after a warm bath (Pruritus)	1
G007	Vision problems, such as blurry vision	3
G008	Easily feeling weak/fatigued	5
G009	Susceptibility to infections	5
G010	Frequent dizziness or headaches	3
G011	Decreased appetite	3
G012	Unexplained weight loss	1
G013	Rapid and irregular heartbeat	3
G014	Shortness of breath and chest pain	3
G015	Frequent nosebleeds or other bleeding episodes	5
G016	Tingling or numbness in the hands and feet	3
G017	Brittle and easily broken nails	1
G018	Unexplained bone fractures (Pathological Fractures)	3
G019	Delayed growth and development	5
G020	Short stature	3
G021	Enlarged neck	1
G022	Prominent cheekbones	5
G023	Forehead prominence and flat nose (Mongoloid face)	3
G024	Protruding abdomen	3
G025	Swelling in the upper right abdomen/liver (Hepatomegaly)	5
G026	Swelling in the upper left abdomen/spleen (Splenomegaly)	5

Table 3 below is the formation of a case base that is used as a knowledge base to compile and match facts between symptoms and types of disease.

Table. 3. Case-Based Knowledge Representation for Early Thalassemia Detection Using CBR Method

Code	Symptoms Code Case Base	Disease Code
BK001	G001, G002, G005, G020, G021, G022, G023, G026	P001
BK002	G001, G009, G018, G019, G024, G025	P002
BK003	G001, G010, G011, G012, G017	P003
BK004	G002, G004, G008, G013, G014	P004
BK005	G003, G006, G007, G015, G016	P005
BK006	G001, G002, G005, G023	P001
BK007	G001, G009, G018	P002
BK008	G001, G010, G017	P003
BK009	G002, G004, G008	P004
BK010	G003, G006, G007	P005
BK011	G021, G022, G023, G026	P001
BK012	G019, G024, G025	P002
BK013	G011, G012, G017	P003
BK014	G008, G013, G014	P004
BK015	G007, G015, G016	P005

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BK016	G001, G002, G021, G022	P001
BK017	G001, G009, G019, G024	P002
BK018	G001, G010, G011, G017	P003
BK019	G002, G004, G008, G013	P004
BK020	G006, G007, G015	P005
BK021	G005, G020, G023, G026	P001
BK022	G009, G018, G024, G025	P002
BK023	G001, G011, G012, G017	P003
BK024	G002, G004, G008, G014	P004
BK025	G003, G007, G015, G016	P005
BK026	G005, G021, G022, G023	P001
BK027	G009, G018, G019	P002
BK028	G001, G011, G012	P003
BK029	G002, G008, G014	P004
BK030	G003, G015, G016	P005

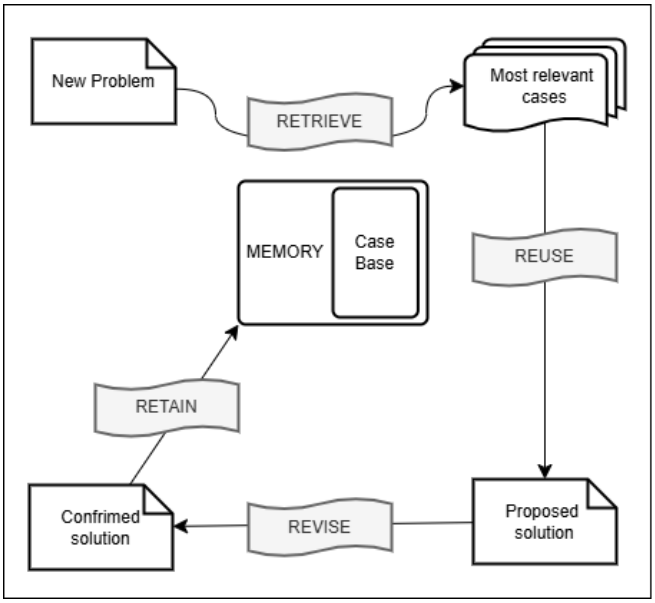

Fig. 3. Comprehensive Case-Based Reasoning (CBR) Life Cycle for Problem-Solving and Learning

E. System Design Planning

This stage involves designing the architecture and components of the system to be developed. Researchers define the structure, functionalities, and interactions between different system components to ensure optimal performance and usability [26].

F. System Implementation

After the system planning stage is complete, the next step is to implement the results of the planning [23], [27]. The PHP programming language and other supporting infrastructure are used in the system implementation process. Figure 3 below explains the system flow.

The flowchart illustrates the Case-Based Reasoning (CBR) expert system flow for detecting thalassemia based on user-selected symptoms [13], [28]. The system calculates case similarity using K-Nearest Neighbor, then applies reuse, revise, or retain before presenting the diagnosis [15].

1. Retrieve

The Retrieve phase in the Case-Based Reasoning (CBR) process involves selecting the most relevant past cases from the knowledge database based on their similarity to the new case. In this phase, similarity is calculated using the K-Nearest Neighbors (K-NN) algorithm, where the system compares the new case (P) with stored cases (Q) and retrieves the most similar ones. The similarity between cases is determined using Eq. (1).

$$Similirity(P,Q) = \frac{S1\ X\ W1+S2\ X\ W2+\cdots+S_nX\ W_n}{W1+W2+\cdots+W_n} \quad (1)$$

Where (P) represents the new case, (Q) denotes previous cases stored in the knowledge database, (W) refers to symptom weights, (S) indicates similarity with a value of 1 if identical and 0 if different, and nnn represents the number of data points. After calculating similarity for all stored cases, the system retrieves the top $Cretrieved$ most similar cases using Eq. (2).

$$Cretrieved = \{Q_i \mid Q_i \in knowledge\ database, sorted\ by\ Sim(P,Q_i)\ in\ descending\ order\} \quad (2)$$

Where $Cretrieved$ is the set of retrieved cases, Q_i represents the previous cases stored in the knowledge database, $Sim\ (P,Q_i)$ calculates the level of similarity between new cases and existing cases. These cases are then sorted based on the similarity value $Sim(P,Q_i)$ in descending order, so that the case with the highest similarity to the new case P will be selected first.

The purpose of this function is to ensure that the system only retrieves the most relevant cases to help the next process in the expert system, namely the Reuse stage, where the solution from the previous case can be reused or modified to solve new cases.

2. Reuse

In the Reuse stage of the Case-Based Reasoning (CBR) process, the system selects the case that has the highest similarity value to the new case being evaluated. This selection is done using Eq. (3).

$$Cselected = \arg_{Qi \in \underset{casebase}{max}} Sim\ (P,Q_i) \quad (3)$$

Where $Cselected$ is selected cases that have the highest similarity to new cases, $arg\ max$ Is selected cases that

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have the highest similarity to new cases, $Sim\ (P, Q_i)\ is$ calculates the level of similarity between new cases P and existing cases Q_i. Which means the system searches for and retrieves cases with the highest similarity value from a collection of cases stored in the casebase. In the formula, Q_i represents the previous case stored in the casebase, while P is the new case being tested. The function $Sim\ (P, Q_i)$ calculates the level of similarity between new cases and existing cases, so that the system can determine the most relevant cases to be reused as references in solving new cases.

3. Revise

The Revise stage in Case-Based Reasoning (CBR) is to compare the Similarity (Sim) value from the best case selection results $(Cselected)$ to the threshold value $(Threshold)$ of 70% which can be written as Eq.(4).

$$Decision\ Result\ = \begin{matrix} AcceptSolustion, if Sim(P, C_{selected}) \geq \text{Threshold} \\ ReviseSolution, if Sim(P, C_{selected}) < \text{Threshold} \end{matrix} \quad (4)$$

Where $Sim(P, C_{selected})\ is$ Similarity value between new cases and selected cases. The threshold value (0.7) determines whether the solution is accepted immediately or needs to be revised. Accept Solution If the similarity value ≥ Threshold the solution from $Sim(P, C_{selected})$ is accepted without changes. Revise Solution If the similarity value < Threshold, solution needs to be revised before use.

If the Sim value is less than 70%, the system will make adjustments by looking for alternative solutions, combining solutions from several cases, or requesting intervention from experts to determine a more appropriate solution.

4. Retain

Retain stage, if the solution obtained has been verified or revised correctly, the system will save the new case into the case base (Q) so that it can be used in the future. The Eq. (5) is the Operation of adding new case (P) to the case base (Q)

$$Q = Q \cup \{(P, Solution)\} \quad (5)$$

Where Q is previous cases stored in the knowledge database, P is new cases under evaluation. $Solution$ is The solution obtained from the results of Accepted Solution or Revised Solution By implementing the CBR method, the system can perform early detection of thalassemia disease automatically, based on experience, and is able to learn from each new case to improve diagnostic accuracy.

G. Testing

System testing is conducted using blackbox testing to ensure that each feature functions according to predetermined specifications. This testing focuses on validating system functionality without looking at the internal structure of the code. In this study, there are three user roles tested, namely User, Admin, and Expert, each with specific features as shown in the use case diagram. The testing process is carried out with the following approach:

1. Determination of Testing Scenarios : Each feature is tested based on normal usage scenarios and error handling scenarios. Scenarios for Users registering with valid and invalid data, performing early detection, and viewing detection history. Scenarios for Admin managing user data and detection history. Scenarios for Experts managing disease data, symptoms, case bases, threshold values, expert revisions, and adding new case data.
2. Testing Process: Each feature is tested directly by entering input according to the scenario.The system is analyzed based on the output produced, whether it is in accordance with expectations or not.
3. Success Criteria:Output Accuracy The results produced by the system must be in accordance with the input provided. System Response There are no errors or system failures when the feature is used. Data Validation Incorrect input must be handled with appropriate notification. System Performance The system response time is still within reasonable limits when processing data.

With this testing method, it can be ensured that the developed system has met user needs and can be used properly according to the expected functionality. To clarify the testing process in this study, in addition to using black-box testing to ensure the system functions according to specifications, accuracy testing is also carried out using a confusion matrix. Accuracy testing with a confusion matrix aims to evaluate the extent to which the system can correctly detect thalassemia based on the case data entered. By combining black-box testing and confusion matrix, system evaluation covers both functionality and diagnostic accuracy, ensuring that the system performs well technically and in disease detection. The evaluation metrics used in the expert system testing with a dataset are as follows:

1. Accuracy

Accuracy measures how often the system classifies cases correctly, as shown in Eq. (6).

$$Accuracy = \frac{TP + TN}{TP + TN + FP + FN}\ X\ 100\% \quad (6)$$

Where (TP) True Positive is the system correctly identifies a positive case. (TN) True Negative is the system correctly identifies a negative case. (FP) False Positive is the system mistakenly classifies a negative case as positive. (FN) False Negative is the system fails to recognize a positive case and instead considers it negative [26], [29].

2. Precision

Precision measures the proportion of predicted positive cases that are actually positive, as shown in Eq. (7).

$$Precision = \frac{TP}{TP + FP} \quad (7)$$

A high precision value indicates fewer false positives in detecting positive cases.

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3. Recall (Sensitivity)

Recall (Sensitivity) measures the system's ability to identify true positive cases. It is calculated using Eq. (8)

$$Recall = \frac{TP}{TP + FN} \quad (8)$$

A higher recall value means the system is better at identifying true positive cases.

4. F1-Score

The F1-Score is the harmonic mean of Precision and Recall, offering a balanced assessment of model performance, particularly in imbalanced datasets, as shown in Eq. (9).

$$F1 = 2\,X\,\frac{Precision\ x\ Recall}{Precision + Recall} \quad (9)$$

A high F1-Score indicates that the system maintains a good balance between precision and recall in detecting thalassemia. The test data used was obtained from observations with experts, resulting in a total of 51 case data. Of these, 30 cases were used as the system's case base, while 21 cases were used as test data using the confusion matrix method.

H. System Deployment

After the system has successfully passed the testing phase, the next step is the deployment process or system launch. At this stage, the expert system website that has been developed and tested for functionality will be implemented for use by users. Researchers ensure that all system components are running well and ready to be widely operated by users [30].

I. Conclusion

In the conclusion stage, the researcher summarizes the main findings of the study, explains the meaning of the results obtained, and relates them to theory or previous research. This section also reveals the impact or implications of the research results, both for practice, policy, or the development of science. In addition, the conclusion mentions the limitations of the study and provides suggestions for further research to deepen understanding or explore unanswered matters.

3. RESULTS

This system focuses on early detection of thalassemia with a case-based reasoning (CBR) approach, which allows early detection based on similarities with previous cases. The process of determining disease data, symptom data, symptom weights, and thresholds is carried out based on expert insight, which can be influenced by their clinical experience. Differences in expert perspectives or interpretations of the significance of a symptom can cause variations in determining the weight, which can ultimately affect the final results of the system. By considering these factors, this system can be continuously refined to improve diagnostic accuracy and expand the scope of its application in supporting early detection of thalassemia.

Table 4 below displays the symptoms entered by the user for a new case. Each symptom is assigned a unique code and a corresponding weight, indicating the significance of the symptom in diagnosing thalassemia. This data serves as the basis for the system's case-based reasoning analysis. Symptom weights are determined based on consultation with medical experts and references from related literature. The higher the weight of a symptom, the greater its influence in determining the likelihood of a thalassemia diagnosis. In the diagnosis process, these weights are used to calculate the degree of similarity between the new case and the existing case base, thereby helping the system to provide more accurate results. Thus, the use of symptom weights allows the system to tailor the diagnosis based on the severity and relevance of the symptoms reported by the user.

Table. 4. New case representation, detailing the input parameters and processing steps used for classification and analysis in the system.

Code	Symptoms	Weight
G001	Family history of thalassemia	5
G002	Pale facial appearance	5
G011	Decreased appetite	3
G024	Protruding abdomen	3

After receiving the rules from the expert and building the case base, the next step is to calculate the similarity level using the k-nearest neighbor method based on new cases inputted by the user.

A. Retrieve

To calculate the similarity of symptoms selected by the user with previous cases in the case base, you can use the K-Nearest Neighbor (K-NN) algorithm. The search process is carried out by comparing the selected symptoms with 30 existing case bases. The following table is a calculation of the sameness of new cases with the case base.

Table. 5. Retrieving a new case associated with BK001, demonstrating the process of case identification and matching within the system.

Case Base		New Case	
Code Symptoms	Weight	Code Symptoms	Weight
G001	5	G001	5
G002	5	G002	5
G005	3	G011	3
G020	3	G024	3
G021	1		

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G022	5
G023	3
G026	5

$$Similirity(P,Q) = \frac{S1\ X\ W1 + S2\ X\ W2 + \cdots + S_n X\ W_n}{W1 + W2 + \cdots + W_n}$$

After comparing the new case with BK001, the similarity value obtained is 0.3333 or 33,33%

Table. 6. Retrieving a new case associated with BK002, demonstrating the process of case identification and matching within the system.

Case Base		New Case	
Code Symptoms	**Weight**	**Code Symptoms**	**Weight**
G001	5	G001	5
G009	5	G002	5
G018	3	G011	3
G019	5	G024	3
G024	3		
G025	5		

$$Similirity(P,Q) = \frac{S1\ X\ W1 + S2\ X\ W2 + \cdots + S_n X\ W_n}{W1 + W2 + \cdots + W_n}$$

After comparing the new case with BK002, the similarity value obtained is 0.3076 or 30,76%.

Table. 7. Retrieving a new case associated with BK003, demonstrating the process of case identification and matching within the system.

Case Base		New Case	
Code Symptoms	**Weight**	**Code Symptoms**	**Weight**
G001	5	G001	5
G010	3	G002	5
G011	3	G011	3
G012	1	G024	3
G017	1		

$$Similirity(P,Q) = \frac{S1\ X\ W1 + S2\ X\ W2 + \cdots + S_n X\ W_n}{W1 + W2 + \cdots + W_n}$$

After comparing the new case with BK003, the similarity value obtained is 0.6154 or 61,54%.

Table. 8. Retrieving a new case associated with BK004, demonstrating the process of case identification and matching within the system.

Case Base		New Case	
Code Symptoms	**Weight**	**Code Symptoms**	**Weight**
G002	5	G001	5
G004	1	G002	5
G008	5	G011	3
G013	3	G024	3
G014	3		

$$Similirity(P,Q) = \frac{S1\ X\ W1 + S2\ X\ W2 + \cdots + S_n X\ W_n}{W1 + W2 + \cdots + W_n}$$

After comparing the new case with BK004, the similarity value obtained is 0.2941 or 29,41%.

Table. 9. Retrieving a new case associated with BK005, demonstrating the process of case identification and matching within the system.

Case Base		New Case	
Code Symptoms	**Weight**	**Code Symptoms**	**Weight**
G003	5	G001	5
G006	1	G002	5
G007	3	G011	3
G015	5	G024	3
G016	3		

$$Similirity(P,Q) = \frac{S1\ X\ W1 + S2\ X\ W2 + \cdots + S_n X\ W_n}{W1 + W2 + \cdots + W_n}$$

After comparing the new case with BK002, the similarity value obtained is 0 or 0%. The summary of the results of the calculation of the similarity of new cases with the case base (BK001 – BK030) is as follows:

Table. 10. Similarity results summary

S (Case, BK001) = 33,33%	S (Case, BK016) = 62,50%
S (Case, BK002) = 30,77%	S (Case, BK017) = 44,44%
S (Case, BK003) = 61,54%	S (Case, BK018) = 66,67%
S (Case, BK004) = 29,41%	S (Case, BK019) = 35,71%
S (Case, BK005) = 00,00%	S (Case, BK020) = 00,00%
S (Case, BK006) = 62.50%	S (Case, BK021) = 00,00%
S (Case, BK007) = 38,46%	S (Case, BK022) = 18,75%
S (Case, BK008) = 55,56%	S (Case, BK023) = 80,00%
S (Case, BK009) = 45,45%	S (Case, BK024) = 35,71%
S (Case, BK010) = 00,00%	S (Case, BK025) = 00,00%
S (Case, BK011) = 00,00%	S (Case, BK026) = 00,00%
S (Case, BK012) = 23,08%	S (Case, BK027) = 00,00%
S (Case, BK013) = 60,00%	S (Case, BK028) = 88,89%

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S (Case, BK014) = 00,00%	S (Case, BK029) = 38,46%
S (Case, BK015) = 00,00%	S (Case, BK030) = 00,00%

The following is a visual display of table 10 regarding the summary of the results of the similarity between the case base and new cases.

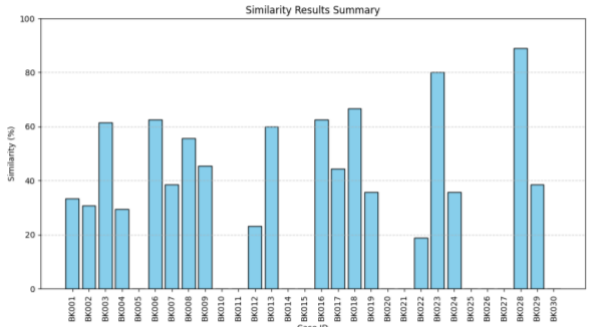

Fig. 4. Summary of similarity results, presenting the calculated similarity scores between the new case and existing cases in the system.

Based on the calculation of the similarity value between the new case and the 30 existing case bases, the next step is the reuse stage. At this stage, the case base with the highest similarity value will be reused as a detection result. However, only cases with a similarity value above the 70% threshold, as agreed with the expert, will be considered as valid detection results.

B. Reuse

In the reuse stage, the case with the highest similarity is selected as the reference for determining the diagnosis. After calculating similarity, case BK028 (Thalassemia Minor) was found to have the highest value of 88.89%, which exceeds the predefined threshold of 70% agreed upon with experts. This indicates that the new case closely matches the characteristics of BK028, making it the most suitable candidate for reuse.

Since the similarity score is significantly high, the system will adopt the diagnostic outcome of BK028 and apply it to the new case. This means the new case is most likely to be classified as Thalassemia Minor. This decision is based on the case-based reasoning (CBR) approach, where previously solved cases are used as a reference to diagnose new cases. The high similarity suggests that the symptoms presented in the new case strongly align with those of BK028, reinforcing the reliability of the diagnosis.

C. Revise

In the revise stage, expert review is required when the highest similarity score falls below the predefined threshold (70%), ensuring that the system's diagnosis is accurate and reliable. If the similarity score is too low, it indicates that the new case does not strongly match any existing case in the case base, requiring expert intervention. Experts analyze the symptoms, compare them with known cases, and determine whether the system's diagnosis should be accepted, modified, or rejected.

In this case, no revision was necessary because the detected Thalassemia Minor case (BK028) had a similarity score of 88.89%, which is well above the threshold. This high similarity indicates a strong match between the new case and BK028, making the diagnosis highly reliable. Since the similarity score met expert-agreed criteria, the system confidently assigned the Thalassemia Minor classification without requiring further expert adjustments. This step ensures that the diagnosis process remains efficient while maintaining accuracy, as only ambiguous or uncertain cases require expert validation.

D. Retain

At the retain stage, various new cases based on user input that have not yet been concluded will be displayed on the new case recommendation page. These new cases will be reviewed and stored in the knowledge case base so that these cases can be used to address other cases in the future.

4. DISCUSSION

A. System Implementation

To increase public awareness about thalassemia and the importance of genetic screening, various campaign strategies can be implemented. One effective approach is through a dedicated website that serves as the main platform for providing comprehensive and easily accessible information about thalassemia, its symptoms, and the importance of early detection before marriage. This website was developed using the Case-Based Reasoning (CBR) method, implemented with the PHP programming language and MySQL database. In addition to functioning as an expert system to assist in diagnosing thalassemia, the site is designed with a CSS framework to ensure an attractive and responsive appearance across various screen sizes, making it easier for users to access the available information and services.

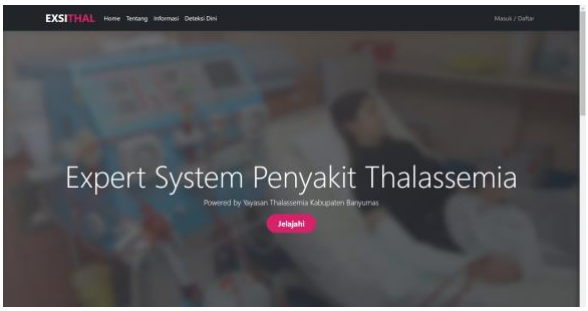

Fig. 5. Landing page displaying the website's main interface for thalassemia awareness and early detection.

Figure 5 is the Home page introducing an expert system for early detection of thalassemia, in collaboration with the Banyumas Thalassemia Foundation. This website provides information, education, and support for affected individuals and families.

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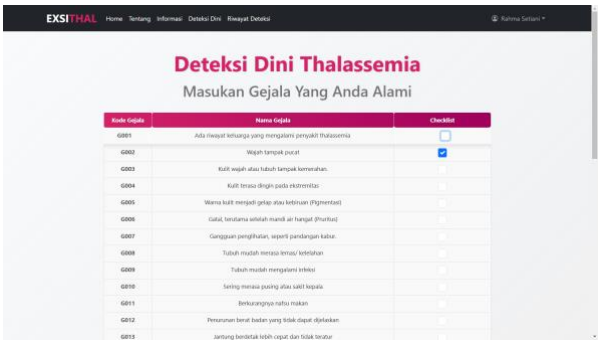

Fig. 6. The Early Detection page provides an intuitive interface to help users select the symptoms they are experiencing.

Figure 6 The Early Detection page displays a symptom form that users can check according to their condition. After selecting the symptoms, users can proceed to the analysis by clicking the "Process Detection" button. This page is designed simply to facilitate early detection of thalassemia.

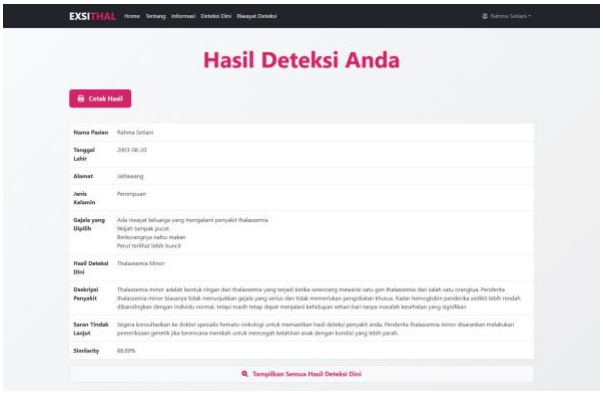

Fig. 7. Early Detection Results displaying the diagnosis outcome based on the selected symptoms and similarity analysis.

Figure 7 shows the results of the same calculations as the manual calculations previously calculated based on user answers in Table 3. The Case-Based Reasoning (CBR) method implemented in the web-based system functions correctly, ensuring accurate detection and classification of cases.

B. Accuracy

After the system is completed and its function is running well, then perform system testing with the aim of evaluating performance in detecting or classifying data accurately. This system testing is carried out using the confusion matrix method, which provides an overview of the number of correct and incorrect predictions from the system. Testing is carried out using 21 test data to measure system accuracy. Accuracy testing using confusion matrix on 21 test data can be seen in the table 11 :

Table. 11. Similarity results summary presenting the comparison between the new case and existing cases based on calculated similarity scores.

	Predicted Positive	*Predicted Negative*
Actual Positive	TP = 20	FN = 1
Actual Negative	FP = 0	TN = 0

In table 11 the results of the Level of Accuracy using 21 test data get accurate results. To find the value of the Level of Accuracy, from testing the test data, you can use the calculation below:

From the results of the tests conducted by experts with 21 trials, an accuracy value of 95.23% was obtained. This shows that the system has excellent capabilities in detecting and classifying cases with a high level of accuracy. Precision is 1, which means that all cases predicted as positive are actually positive cases without any errors. In other words, there are no false positives in the system's prediction results. Recall is 0.9523 (95.23%), which indicates that of all the actual positive cases, the system successfully detected 95.23% of them, but there were still a few positive cases missed (false negatives). By using the harmonic mean formula between precision and recall, the F1-score is 0.9754 (97.54%). This shows that the system has a very good balance between accuracy in predicting positive cases and the ability to capture all existing positive cases. Based on the results of this study, the Case-Based Reasoning (CBR) based system showed an accuracy of 95.23%, which is higher than several previous studies.Compared with studies comparing the CBR and Certainty Factor (CF) methods in diagnosing pests and diseases in rice plants, the CBR system in this study is superior, because previous studies only obtained an accuracy of 89.40% for CBR and 83.48% for CF [31].

Furthermore, research on the application of CBR to an expert system for diagnosing food crop diseases showed an accuracy of 76.74%, which is much lower than the results of this study [32]. Another study on an expert system for diagnosing lung disease using the CBR method recorded an accuracy of 90%, which is still lower than the system developed in this study [33]. In addition, research on the implementation of an expert system to determine the possibility of being infected with the HIV virus using the Forward Chaining and CBR methods showed an accuracy of 82.05%, which is also lower than the results of this study [34]. Another study compared the accuracy between Neural Network (NN) and Case-Based Reasoning (CBR) in diagnosing eye diseases. The results showed an average accuracy comparison, with NN achieving 95.07% accuracy and CBR achieving 94.70% accuracy. The accuracy level is influenced by the network architecture in NN and the similarity function in CBR [35].

Thus, the CBR-based system developed in this study has proven to have excellent performance, even surpassing several previous studies in different fields. This difference indicates that the CBR approach is more

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effective in the medical domain, where previous case experience is a crucial factor in diagnosis. This superior accuracy also strengthens the potential of expert systems in the early detection of thalassemia, assisting medical personnel in making faster and more accurate decisions, and enhancing efficiency in healthcare services. This study still has several limitations, including the relatively small number of case data, which can affect the system's ability to generalize to a wider population. In addition, the data used only comes from one region, so it may not reflect genetic variations and environmental factors in other regions. Because the Case-Based Reasoning (CBR) system relies heavily on historical data, the accuracy of diagnosis can decrease if there are new cases that do not have a match in the database. To overcome these limitations, further research is expected to expand the dataset by collecting data from various hospitals and more diverse populations. In addition, expanding the study to other regions with different prevalences of thalassemia can improve the accuracy of the system globally. More sophisticated approaches, such as combining CBR with machine learning, can also be applied so that the system is able to automatically learn new patterns and improve the accuracy of diagnosis

This research can be further developed by improving the accuracy and coverage of the expert system. One direction of future research is the combination of the Case-Based Reasoning (CBR) method with machine learning so that the system can automatically learn new patterns and provide more accurate recommendations. In addition, the development of prediction features based on genetic data and family history can help users understand their risks more specifically. Furthermore, this system can be expanded by adding a case database from various hospitals and a more diverse population to increase the validity of the diagnosis results. The development of a mobile version and an AI-based chatbot can also facilitate public access to information and early detection services. With these various developments, the thalassemia expert system will not only become a more sophisticated early detection tool, but also contribute to improving the quality of health services and thalassemia prevention policies in the future.

5. CONCLUSION

Based on the findings of this study, a web-based expert system for early detection of thalassemia has been successfully developed with the primary goal of identifying individuals who may carry the thalassemia trait at an early stage. This system aims to enhance early diagnosis, facilitate timely medical intervention, and address the low awareness and limited medical expertise in thalassemia detection. The implementation of the system using the Case-Based Reasoning (CBR) method has been rigorously tested through accuracy evaluation using a confusion matrix. accuracy testing produced a high accuracy rate of 95.23% based on 21 test cases, demonstrating the system's reliability in detecting thalassemia cases. These results indicate that the system outperforms previous studies and offers a more effective approach to early detection. For future development, several enhancements are recommended. Expanding the dataset by incorporating more diverse case studies from various hospitals and regions will improve the system's generalizability. Additionally, integrating machine learning algorithms could enhance the system's adaptability by enabling it to learn from new cases beyond the existing knowledge base. To increase accessibility and usability, the system should be linked with regional healthcare platforms such as community health centers (Puskesmas), public hospitals (RSUD), and digital health applications.

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