Individual Dissertation

AN EXPLAINABLE AI SYSTEM FOR CLINICAL DECISION SUPPORT IN SCHWANNOMATOSIS

(SENTIA-X)

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Abstract

This thesis presents the development of SENTIA-X (Schwannomatosis ENgine for Treatment Insight & eXplainable Analytics), an intelligent Clinical Decision Support System (CDSS), tailored for the management and diagnosis of schwannomatosis, a rare genetic disorder. The system aims to modernize the way patient data is recorded, analyzed and interpreted by moving away from traditional paper-based methods toward a secure and accessible digital platform.

SENTIA-X introduces a standardized process for clinical examinations and data collection, enhancing consistency across different healthcare providers. Leveraging Explainable Artificial Intelligence (XAI), the platform supports early detection and monitoring of medical conditions contributing to timely intervention and improved healthcare outcomes.

Built upon the Argumentation Framework and integrated with the Cognica platform, the system employs Controlled Natural Language (CNL) and XAI to generate transparent, logic-based clinical recommendations. This ensures not only ethical and interpretable decision-making but also builds trust among end-users. Developed through collaboration with medical professionals, the platform also enables knowledge sharing between experienced and junior clinicians.

The system is developed in alignment with the *"ERN GENTURIS Clinical Practice Guidelines for The Diagnosis, Treatment, Management and Surveillance of People with Schwannomatosis* ", ensuring that its design and functionality reflect current best practices. Ultimately, this thesis bridges the gap between health informatics and clinical practice, advancing the quality and consistency of care for individuals affected by schwannomatosis.

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Chapter 1

Introduction

1.1 Problem Statement and Motivation

Schwannomatosis is a rare genetic disorder characterized by the development of multiple schwannomas—benign tumors arising from Schwann cells along peripheral nerves [1], [4]. Despite advancements in medical imaging and genetics, the diagnostic and management processes for this condition remain fragmented, often relying on manual documentation and outdated paper-based systems. These traditional approaches present challenges such as loss or damage of records, limited accessibility, and inconsistency in data collection across clinical settings, and delayed intervention or failure to implement timely and appropriate follow-up actions.

This thesis is motivated by the need to modernize and streamline the handling of clinical data related to schwannomatosis. By transitioning to a digital Clinical Decision Support System (CDSS), this work aims to improve data security, facilitate standardized diagnostic procedures, and support real-time access to patient information. Furthermore, the integration of Artificial Intelligence (AI) enables early identification of complications such as malignancy risk and supports clinical decisions with logic-based recommendations. Importantly, by leveraging XAI, the system fosters trust and transparency between patients and doctors, ensuring that decisions are not only accurate but also interpretable and aligned with clinical reasoning.

A major goal of the proposed system is to shift from outdated, paper-based documentation to a secure, intelligent digital infrastructure. This transition focuses not only on improving accessibility and protection of patient records but also on addressing challenges related to inconsistent formats, potential data loss and limited interoperability across clinical professionals and institutions.

Due to complexity of the disease, the system provides decision support through clear, explainable reasoning. Importantly, it avoids bias in clinical decisions, as its recommendations are not based solely on individual patient records, but on structured logic and predefined clinical rules, ensuring consistency, fairness and transparency in the diagnostic process.

An important feature of the system is its ability to enforce standardization in the diagnostic process, ensuring that data collected across multiple clinicians is consistent, comparable, and useful for future analysis.

Additionally, the platform is designed to support knowledge transfer from experienced clinicians to junior practitioners, promoting consistent quality of care and supporting professional development. This work takes a step toward embedding modern technologies such as XAI into rare disease healthcare practice. It aims to improve efficiency, accuracy, and patient-centeredness in clinical processes, reflecting a shift toward more advanced, effective, and explainable digital tools in medicine.

1.2 Definition and Scope

The focus of this thesis is the development and evaluation of an intelligent, web-based CDSS tailored to the clinical management of Schwannomatosis. The system incorporates AI technologies and logic-based reasoning to enhance diagnostic accuracy, ensure early detection of contraindications, and offer transparent support for clinical decisions. It aims to provide a digital alternative to traditional workflows and promote consistent data handling across clinical settings.

The scope includes the design, implementation, and evaluation of the system in a real-world clinical need. While this project specializes in Schwannomatosis, the underlying architecture and methodology are designed to be adaptable to other rare diseases, serving as a model for scalable decision support solutions.

1.3 Literature Review

Recent literature in the fields of health informatics, XAI in medicine, and CDSS highlights a growing focus on explainability, ethical design, and user-centered development. Argumentation frameworks and logic-based reasoning methods have gained traction for their ability to provide transparent, evidence-based recommendations in complex clinical contexts [2].

In particular, XAI has emerged as a critical area of focus within clinical AI systems, aiming to improve transparency, foster trust and support accountability in healthcare decision making. While much of the discussion around XAI centers on addressing the “black box” problem of machine learning models, this thesis adopts a logic based, rule driven approach to decision support.

By using CNL, the system translates clinical guidelines into structured, machine interpretable rules that mirror human clinical reasoning. This enables the generation of clear, auditable explanations that align closely with clinical logic and can be easily interpreted by healthcare professionals.

Moreover, literature on argumentation based XAI supports the use of frameworks that simulate structured debate presenting both supporting and opposing arguments for a given recommendation. This method is valuable in rare diseases, where uncertainty is common and treatment paths may not be well established. By explicitly presenting the reasoning behind each recommendation, such systems encourage shared decision making and can be used as educational tools [16].

Recent research also emphasizes the need for transparent XAI tools that respect ethical standards, including fairness and traceability [17]. The integration of XAI techniques into rare disease CDSS platforms represents a promising direction for both improving care quality and overcoming the lack of specialized expertise.

1.4 System Overview and Characteristics

The system presented in this study is a modular, web-based platform developed using HTML, CSS, JavaScript, and PHP, with backend support for dynamic data management. It is integrated with the Cognica framework, which employs CNL and logic-based reasoning to generate explainable clinical recommendations.

Key features of the system include:

- Real-time access to patient records.
- Interactive diagnosis selection.
- AI-powered recommendation engine.
- Transparent display of reasoning and clinical logic.
- Modular UI for various use cases (e.g., drugs, pain management).

The system’s design supports uniform data entry and decision-making, ensuring consistent and analyzable medical records across different healthcare providers.

1.5 Objectives and Contribution of the Study

The purpose of this system is to modernize the way clinical examination data related to schwannomatosis is recorded and analyzed, transitioning from outdated paper-based methods to a secure and accessible digital infrastructure. This shift primarily focuses on improving patient data handling by addressing challenges like damage risks and limited accessibility associated with traditional records.

The primary objectives of this thesis are:

1. To digitize and modernize the diagnostic workflow for Schwannomatosis.
2. To implement an AI-enhanced system that provides explainable, evidence-based support to clinicians.
3. To promote knowledge sharing between experienced and junior medical professionals through a transparent and standardized interface.

The main contributions of this study are threefold:

1. It introduces a scalable digital tool specifically designed for the diagnosis and management of rare diseases.
2. It integrates argumentation-based AI methods to provide clinicians with explainable, logic-based support.
3. It delivers a fully implemented proof-of-concept system that demonstrates real-world applicability in clinical environments.

This work addresses a specific clinical challenge while also offering a transferable framework that could benefit other domains of rare disease management.

1.6 Structure of the Thesis

This thesis explores the intersection of rare disease diagnosis and intelligent clinical software design. It begins by examining the background of Schwannomatosis and the potential of AI-powered systems to support clinical workflows. It then presents the technological foundation and methodology adopted for the system's development, followed by implementation details, practical evaluation, and conclusions for future directions.

The thesis is structured as follows:

- **Chapter 2** – Theoretical Background: Introduces foundational concepts in AI, decision support systems, and argumentation theory.
- **Chapter 3 –** Clinical and Guideline Context: Describes the medical background of schwannomatosis, its clinical presentation, and the official diagnostic guidelines.
- **Chapter 4** – Methodology and Development Tools: Details the requirements gathering, system architecture, and the tools and technologies used in implementation.
- **Chapter 5** – System Implementation and Design: Presents the technical components of the system, including frontend/backend integration, UI/UX design, and system logic.
- **Chapter 6** – Evaluation and Results: Analyzes the system's effectiveness through pilot testing, user feedback, and case scenarios.
- **Chapter 7** – Conclusion and Future Work: Summarizes the study's findings and discusses potential improvements and broader applications of the system.

This structure provides a comprehensive framework for examining the development, implementation, and evaluation of an intelligent clinical decision support system for schwannomatosis.

Chapter 2

Theoretical Background in Artificial Intelligence

2.1 Artificial Intelligence in Healthcare

Artificial Intelligence has seen rapid progress in the medical field, especially in domains like patient care, medical imaging and diagnosis. Despite the success demonstrated in retrospective studies, the adoption of these AI solutions in real-world clinical settings remains limited. Key barriers include challenges related to user trust, regulatory compliance, and ethical use of data [2].

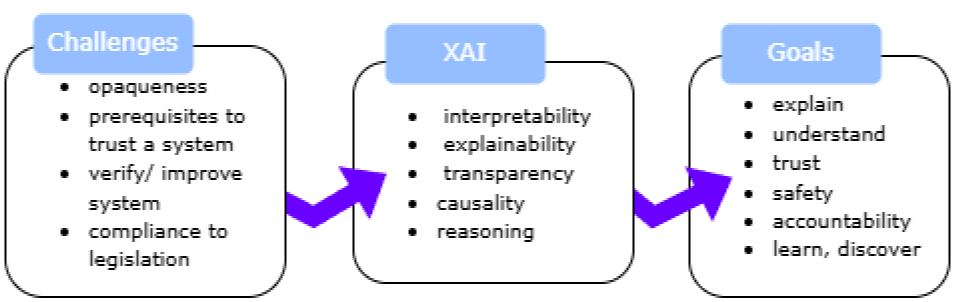


Fig. 1 XAI Related Concepts Summary

Explainable AI aims to bridge this gap by helping humans to understand how AI systems function and by fostering trust in their outcomes [13], [14].

In healthcare, where decisions can have life-or-death consequences, explainability is essential for accountability, clinical validation and shared decision-making.

2.2 Human-Centered and Ethical AI

Human-Centered AI is important in healthcare, where decisions directly affect human lives. AI systems need to be designed as support to the clinicians, not to replace human decision making, in order to humans to trust them. This means that ethical, transparent and accountable design principles must guide development from the ground up [2], [6].

Guidelines from organizations such as the European Commission and IBM outline foundational principles for trustworthy and ethical AI, including [7]:

- Fairness and non-discrimination
- Robustness and safety against failures and biases
- Transparency, including the ability to audit system behavior
- Explainability, so users understand how decisions are made
- Accountability, with mechanisms to assign responsibility for outcomes

TABLE II
TRUSTWORTHY AND ETHICAL AI

Term	Definition
Diversity, non-discrimination, and fairness	- AI systems should use training data and models that are free of bias, to avoid unfair treatment of certain groups. - AI systems should consider the whole range of human abilities, skills and requirements, and ensure accessibility.
Robustness, Safety	- AI systems should be safe and secure, not vulnerable to tampering or compromising the data they are trained on. - Trustworthy AI require algorithms to be secure, reliable and robust enough to deal with errors or inconsistencies during all life cycle phases of AI systems.
Explainability	- AI systems should provide decisions or suggestions that can be understood by their users and developers.
Lineage	- AI systems should include details of their development, deployment, and maintenance so they can be audited throughout their lifecycle.
Transparency	- The traceability of AI systems should be ensured.
Accountability	- Mechanisms should be put in place to ensure responsibility and accountability for AI systems and their outcomes.

Fig.2 Summarized IBM's fundamental pillars and EU guidelines [2]

In medical contexts, these principles relate directly to informed consent, device certification, and legal liability. AI models must not only function accurately but also meet expectations for interpretability, traceability, and compliance.

2.3 Explainable Artificial Intelligence (XAI)

The concept of explainability in AI has been around for a long time. It emerged as a response to the "black box" nature of AI systems, models that provide outputs without clearly showing how those answers were produced. Although these systems may generate accurate predictions, users often struggle to understand the reasoning behind them.

Explainability is closely linked to interpretability, meaning that an interpretable system allows a human to understand how it works either by reading its explanation or by looking inside the model.

As Lipton points out, the term interpretability has a different meaning depending on the person and use case. Some believe that it means seeing the features or rules that the model used and others it means understanding why the model gave a certain result [2].

2.4 Cognitive Argumentation

Cognitive Argumentation provides a framework for modeling human-like reasoning in a structured and explainable way. It extends traditional argumentation theory by introducing cognitive principles derived from philosophy, psychology, and empirical studies of human decision-making [3].

Instead of absolute logical deduction, Cognitive Argumentation views reasoning as the construction and evaluation of arguments and counterarguments. Conclusions are supported not by certainty, but by coalitions of admissible arguments that can defend themselves against opposing claims.

The approach recognizes that human reasoning often operates under uncertainty and incomplete information. Therefore, it supports flexible, context-sensitive decisions that reflect how real humans argue, resolve contradictions, and reach conclusions.

Some of the key ideas in this model include:

- Argument schemes: reusable reasoning patterns (if then statements).
- Cognitive validity: the alignment of reasoning steps with human cognitive expectations.
- Strength relations: hierarchies among given arguments based on relative necessity, sufficiency, or assumptions.

This theory forms the foundation of the Cognica engine used in SENTIA-X, enabling it to provide transparent and human-compatible clinical decision support.

2.5 Admissibility and Explainability in Argumentation

In argumentation theory, admissibility refers to whether a set of arguments is acceptable: it must not be self-contradictory and must be able to defend against all opposing arguments. These admissible sets provide the basis for drawing valid conclusions [3].
There are two main outcomes derived from admissibility [9]:

- **Credulous conclusion**: A conclusion that is supported by at least one admissible set of arguments.
- **Skeptical conclusion:** A conclusion that is supported by all admissible argument sets and is therefore more definitive.

This distinction enables a system like Cognica to classify outcomes as:

- **Yes:** A skeptical conclusion supported by all admissible arguments.
- **Maybe:** A credulous conclusion supported by some arguments but opposed by others.
- **No:** The negation of the statement is a skeptical conclusion.

Explainability is a core feature of this framework. Each decision is not only the result of evaluating arguments but also comes with a traceable explanation.

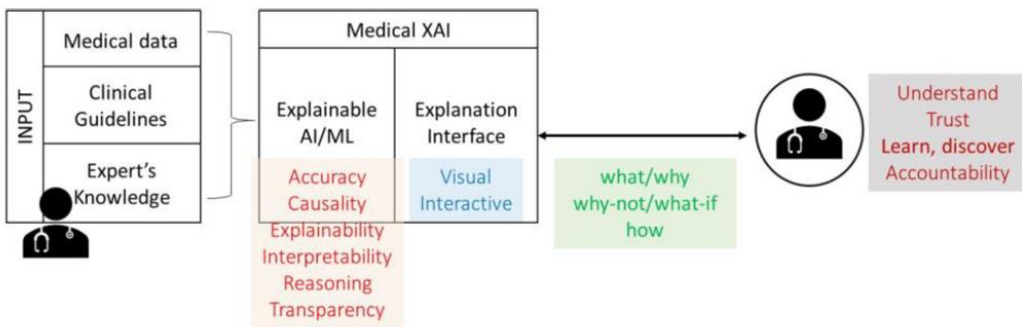


Fig.3 Proposed Medical XAI Design Framework [3]

The Cognica system provides multiple layers of interpretability:

- Verbal summaries tracing the reasoning.
- Graphical diagrams show supporting and opposing arguments.
- Modal dialogs prompting the user to consider additional actions in ambiguous cases.

This structured transparency is what allows SENTIA-X to serve as a truly explainable AI system in a clinical environment, giving medical professionals insight into the reason why a recommendation was made and how to treat uncertain cases.

2.6 Cognica Argumentation Framework

COGNICA is a simulator of human reasoning, built upon the mental models' theory proposed by Johnson-Laird & Byrne, (2002), incorporating preference-based argumentation to formalize and extend this theoretical framework [3].

Mental models are based on the human perception and comprehension of discourse. They represent various possible scenarios based on what an individual has in mind, from which new information can be drawn. It focuses on conditional reasoning, involving statements that are easy to translate into argumentation schemes with the form “If a then b”.

There are two categories of conditional sentences:

1. Sentences that depend on context and the individual’s background knowledge regarding condition and its consequence.
2. Sentences in which the condition and the consequent are semantically independent, aside from their cooccurrence in the conditional form.

Cognica uses nine out of ten categories developed by Johnson-Laird & Byrne: Tautology, Conditional, Enabling, Disabling, Biconditional, Strengthen antecedent, Relevance, Modus Tollens, Modus Ponens. These categories are translated into argument schemes based on the mental model’s theory. Additionally, the system supports a conditional form with necessity modality such as “if a then always b” [3].

The first type of conditionals is translated into argument schemes using contextual reasoning. The second type is interpreted according to whether the condition is necessary or sufficient for the conclusion. When all outcomes are possible, both the conclusion and its negation are supported by hypothetical argument schemes.

To model the relative strength of various conditionals, a priority-based system is used, where argument strength increases from hypotheses(lowest) to statements where the condition is both necessary and sufficient(highest). A special case is introduced for conditionals using “always” which are treated as stronger due to their implication of necessity.

Cognica has been implemented using the Gorgias argumentation framework [15] and is accessible via web at: http://cognica.cs.ucy.ac.cy/COGNICAc/index.php .
To use the system, users must input the policy context, policy options and decision policies, in the form of conditional statements. Upon completing this step, they click the

button “Translate Policy to Cognica DP CNL”, which converts the input into Cognica’s Domain-Preferred Controlled Natural Language. Users are then prompted to review the transformed statements for correctness and if satisfied proceed by clicking “Translate Policy to Cognica CNL”. Next, under the “Prediction” tab, users identify which facts hold or not. From the dropdown menu, they select a conclusion they wish to evaluate and then click “Predict”. Cognica responds with Yes/Maybe/No along with a verbal explanation that justifies its prediction. Beneath Cognica’s explanation, the system also displays a prediction generated by GPT’s 3.5, allowing users to compare symbolic and language model-based reasoning.

Furthermore, the system provides options for viewing and exporting Gorgias Files. Users can access a more detailed explanation by clicking the “Further Explanation” button, which reveals both an extended verbal justification and a graphical representation of the supporting and opposing arguments.

<u>Example Overview: Drug Recommendation Scenario in COGNICA:</u>

In this example, the user defines a policy context related to drug treatment decisions for patients with Schwannomatosis.

The system evaluates two policy options: FirstLineDrugs and SecondLineDrugs.

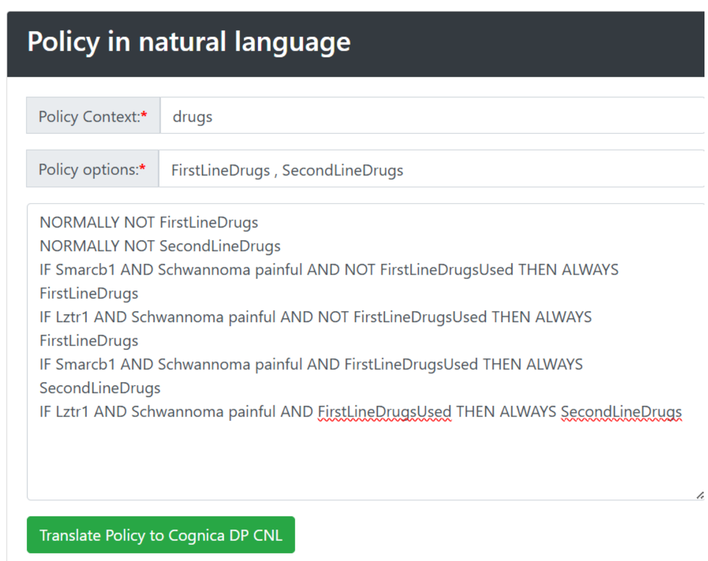


Fig.4 Screenshot of Cognica’s Dashboard while translating policies for Drugs stage to Cognica DP CNL.

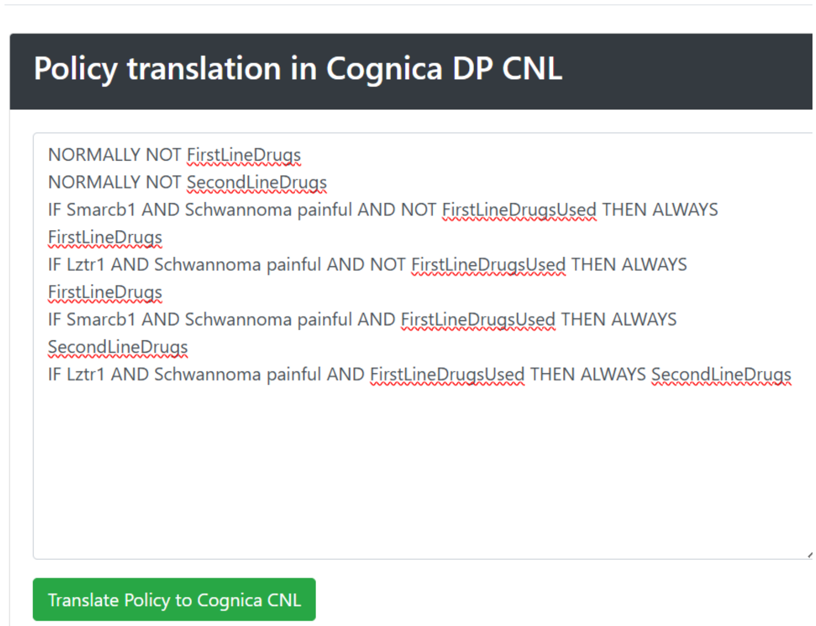


Fig.5 Screenshot of Cognica's Dashboard while translating policies for Drugs stage to Cognica CNL.

These options are encoded as conditional rules using COGNICA's controlled natural language, such as:

- If Smarcb1 and Schwannoma painful and NOT FirstLineDrugsUsed, then ALWAYS FirstLineDrugs.
- If Smarcb1 and Schwannoma painful and NOT FirstLineDrugsUsed, then ALWAYS NOT SecondLineDrugs.

After translating these rules into COGNICA's internal representation (DP CNL), the user tests different combinations of facts to see how the system evaluates which treatment option is appropriate.

Scenario 1: Prediction Yes for First Line Drugs

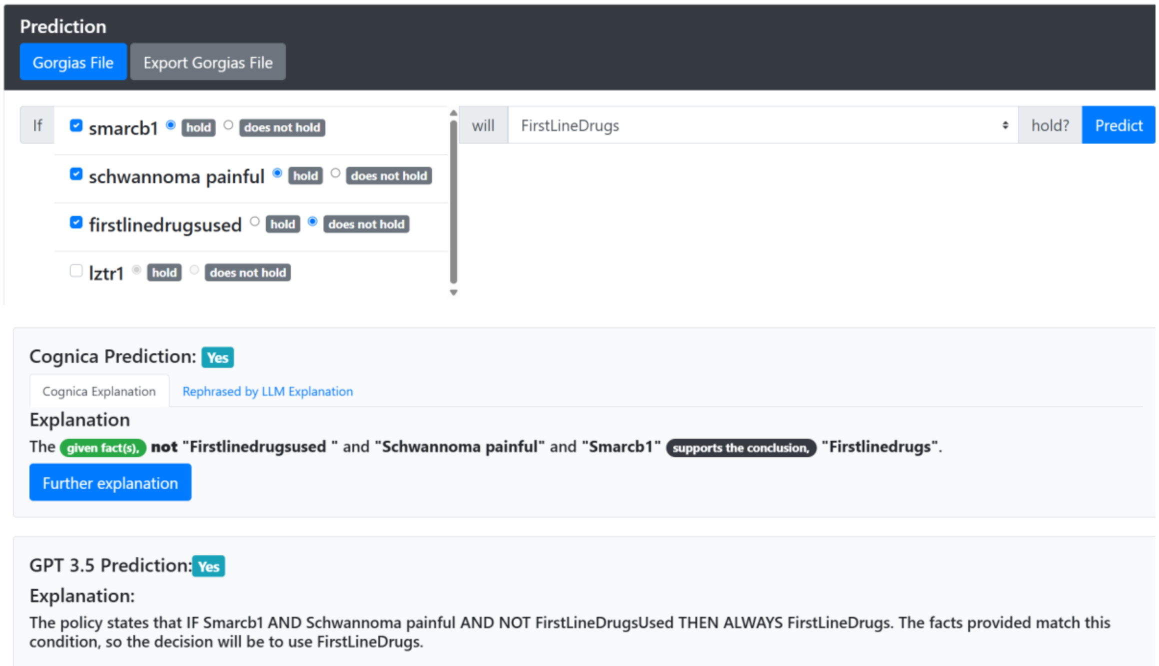


Fig.6 Screenshot of Cognica's Dashboard entering facts that hold or not and the response for First Line Drugs is Yes.

Facts Provided:

- Smarcb1 holds
- Schwannoma painful holds
- First Line Drugs Used does not hold

Prediction:

- COGNICA Prediction: Yes (First Line Drugs should be used)
- Explanation: The facts satisfy a rule that supports First Line Drugs. There are no conflicting arguments with equal strength.
- GPT-3.5 Prediction: Also Yes, explaining that based on the policy and facts, First Line Drugs are required.

This represents a clear decision with no ambiguity, where the necessary condition for using First Line Drugs is fully met.

Scenario 2: Prediction No for First Line Drugs

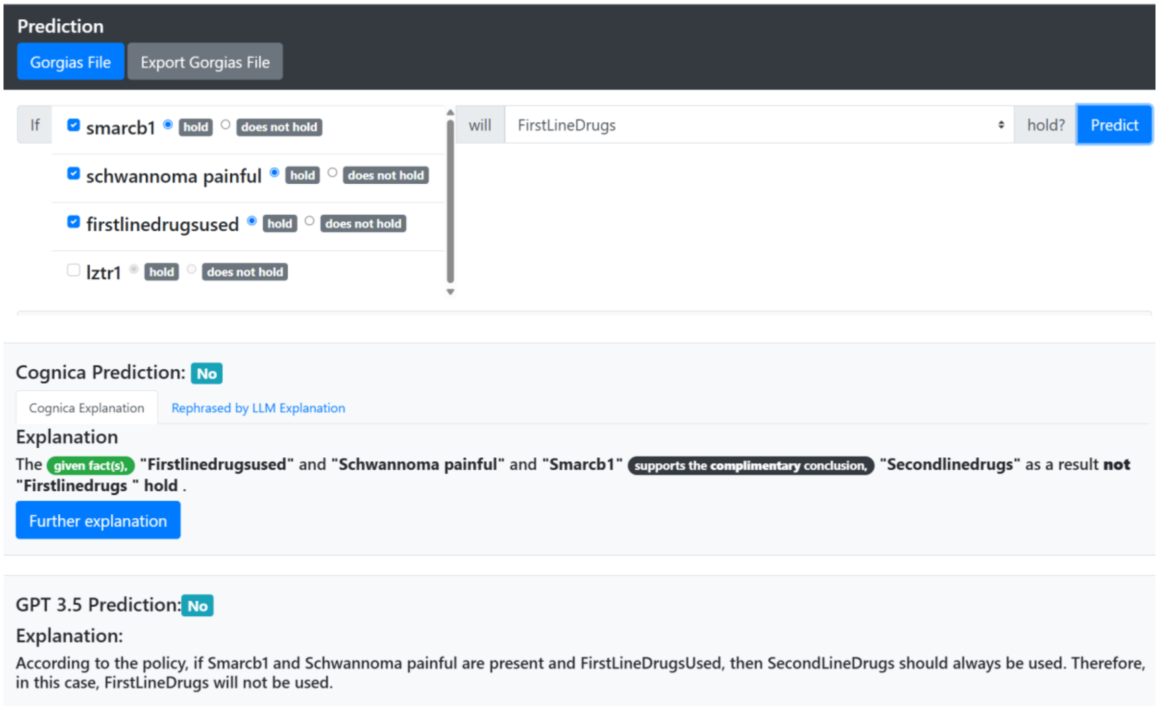


Fig.7 Screenshot of Cognica's Dashboard entering facts that hold or not and the response for First Line Drugs is No.

Facts Provided:

- Smarcb1 holds
- Schwannoma painful holds
- First Line Drugs Used holds

Prediction:

- COGNICA Prediction: No (First Line Drugs should not be used)
- Explanation: The system applies a rule that explicitly denies First Line Drugs when the same conditions apply.
- GPT-3.5 Prediction: Also No, confirming that the policy excludes First Line Drugs in this case.

This result demonstrates how COGNICA can generate mutually exclusive predictions using argument conflict resolution.

Scenario 3: Prediction – Maybe for First Line Drugs

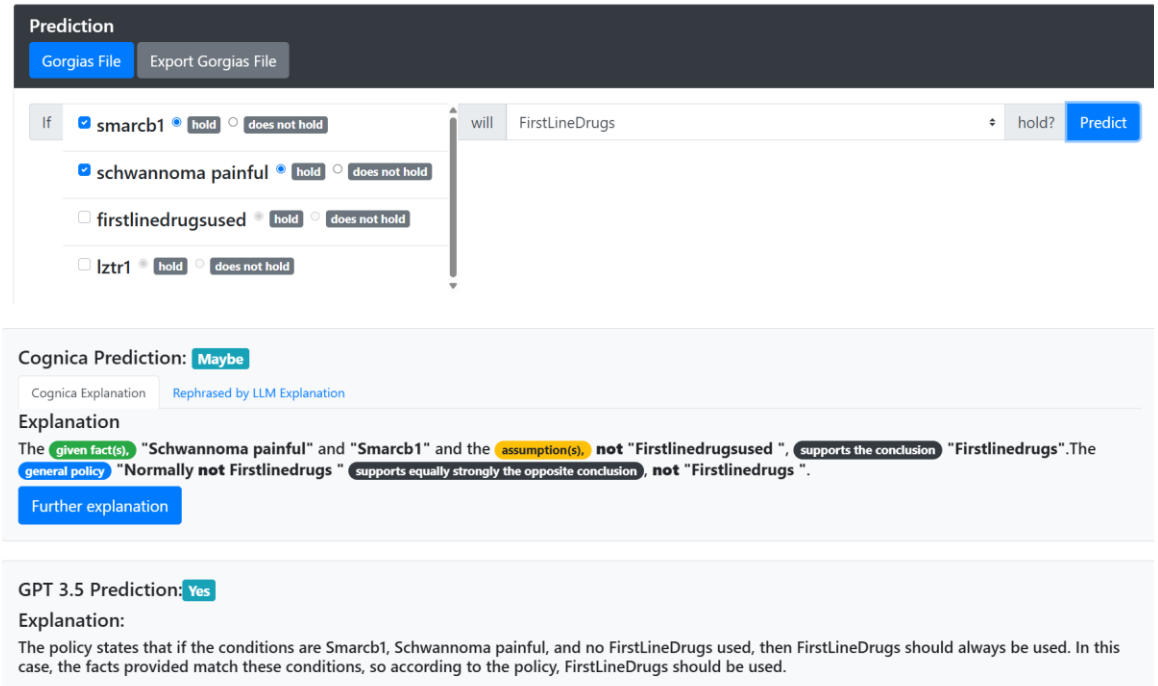


Fig.8 Screenshot of Cognica's Dashboard entering facts that hold or not and the response for First Line Drugs is Maybe (Cognica) but ChatGPT answered Yes.

Facts Provided:

- Smarcb1 holds
- Schwannoma painful holds

However, the key difference is that a general policy (e.g., "Normally not First Line Drugs") is also active, creating a conflict.

Prediction:

- COGNICA Prediction: Maybe (conflicting arguments of equal strength)
- Explanation: One argument supports First Line Drugs, but another equally strong general policy opposes it, leading to ambiguity.
- GPT-3.5 Prediction: Yes – GPT's language model does not consider argument strength/conflict formally, so it concludes based on the stronger semantic interpretation.

This case illustrates conflict handling in COGNICA: when arguments of equal strength exist, the system expresses uncertainty (Maybe).

In addition to using the web interface, COGNICA was also accessed programmatically through its API. This allowed for automated queries and integration into our system's backend logic. The implementation details are presented in Section 5.5.

Chapter 3

Clinical and Domain Background

3.1 Clinical Decision Support Systems (CDSS)

Clinical Decision Support Systems (CDSS) are digital tools designed to assist healthcare professionals to make better clinical decisions by delivering real-time insights and recommendations during patient care. These systems leverage patient data and clinical knowledge to generate case-specific advice, such as diagnostic support, treatment recommendations, drug interaction alerts, and risk assessment tools.

3.1.1 Overview and Use Cases

A CDSS functions like a smart assistant, combining clinical guidelines, patient data, and test results to support clinicians in diagnosing, prescribing, and planning care. It serves as a second expert opinion, providing guidance for decisions that are critical to help to improve outcomes, reduce errors and enhance the overall quality of care [5], [8].

At its core, a CDSS analyzes large volumes of clinical data, such as patient history, test results, and current symptoms and compares them with established medical guidelines. Based on this analysis, it offers suggestions, alerts, or warnings, ensuring that decisions are not only timely but also accurate, consistent and safe [5].

3.1.2 Architecture

A typical CDSS consists of four core components:

- **Knowledge Base**: Stores medical facts, clinical guidelines, treatment protocols.
- **Inference Engine**: Applies logical rules to patient data to generate conclusions and recommendations.
- **Communication Mechanism**: Enables data exchange between the CDSS and external clinical systems, such as Electronic Medical Records (EMRs).
- **User Interface**: Provides an intuitive and accessible interface that allows healthcare professionals to interact with the system efficiently.

These components work together to collect data, compare it to best practices and deliver personalized, real-time guidance. Integration with EMRs ensures seamless data flow and improves efficiency in clinical workflows.

3.1.3 Functionalities and Applications

CDSS offer a broad range of functionalities, from basic alert notifications to advanced diagnostic reasoning engines. Their applications span across clinical workflows and include:

- Enforcement of clinical pathways.
- Prescription safety checks and drug interaction alerts.
- Chronic disease monitoring and management.
- Personalized treatment recommendations based on patient data.

Rare diseases such as Schwannomatosis, CDSS tools play a particularly valuable role. They help standardize diagnostic procedures, reduce variability in clinical decisions, and support less experienced clinicians by providing expert-level recommendations grounded in current medical guidelines.

3.1.4 Guidelines and Design Principles

The effective design of a CDSS requires careful consideration of multiple factors, including usability, explainability, and seamless integration with Electronic Health Records (EHRs). Systems must be designed to align with established clinical guidelines to ensure medical accuracy and relevance.

Human-centered design and XAI principles are particularly important in promoting user trust, interpretability, and clinical adoption. Furthermore, adherence to legal and ethical standards, especially in the context of medical AI, is essential to ensure compliance with data protection regulations, patient safety requirements, and accountability frameworks [12].

3.1.5 Related Work

Existing literature highlights a growing demand for explainable, logic based CDSS platforms across various domains. Argumentation-based frameworks, such as Cognica, have been proposed as promising tools due to their strong emphasis on logic-based reasoning, transparency, and alignment with legal and ethical standards.

CDSS applications have shown significant value in fields where decision-making complexity is high and time sensitive, and where standardized support can significantly improve clinical outcomes. These studies collectively reinforce the importance of developing interpretable and guideline-driven systems capable of supporting real-world clinical practice [11].

3.2 Schwannomatosis: Genetics, Diagnosis, and Management

3.2.1 Definition and Clinical Characteristics

Schwannomatosis is a rare tumor predisposition syndrome characterized by the development of multiple benign nerve sheath tumors (schwannomas) along peripheral nerves. Unlike neurofibromatosis type 2 (NF2), schwannomatosis typically spares the eighth cranial (vestibulocochlear) nerve. Cranial nerves may be affected to a lesser extent, but intradermal schwannomas, characteristic of NF2, are absent. The most common symptom is chronic pain, which may occur even in areas without visible tumors and can be more severe than expected based on tumor size or location [1].

3.2.2 Genetic Basis and Inheritance

Two definitive genes (SMARCB1 and LZTR1) located on chromosome 22q, centromeric to NF2, have been implicated in schwannoma development through a 3-event, 4-hit mechanism that results in complete inactivation of the affected gene and the NF2.
These genes together account for 70-85% of familial schwannomatosis cases and 30-40% of (sporadic) isolated cases with no family history.

3.2.3 Diagnosis and Screening Guidelines

Diagnosis is based on clinical presentation, imaging, pathology and genetic testing. Craniospinal MRI is advised every 2-3 years, beginning:

- at the time of symptomatic diagnosis, or
- if molecularly verified at age 12- 14 in asymptomatic individuals with a relative who has schwannomas.

Whole body MRI may also be deployed and can alternate with craniospinal MRI if available. Particularly in the limbs where a palpable lump is not usually accompanied by pain, ultrasound scanning can be helpful.
The risk of malignancy for the developing Malignant Peripheral Nerve Sheath Tumors should be considered in patients with tumors, that are rapidly growing especially accompanied by functional loss. This risk appears higher in individuals with SMARCB1.

According to ERN GENTURIS [1], the target population includes:

- Individuals with inherited pathogenic variants in SMARCB1 or LZTR1 and a proven schwannoma.
- A healthy carrier of a LZTR1 or SMARCB1 pathogenic genetic variant with a parent with proven Schwannomatosis.

- Analysis of two non-intradermal schwannomas in individuals has revealed that they are not caused by either inherited NF2 or a common NF2 variation between the tumors.
- Presumed schwannomatosis occurs when an individual has multiple confirmed schwannomas or one confirmed plus nerve sheath tumor on an MRI scan does not impact the vestibular nerves on skin itself and no hereditary NF2 variation is detected in the blood.

Annual clinical reviews should include:

- Full pain history and neurological exam
- Psychological assessments
- Quality of life evaluation

Screening typically begins at age 12–14 or at diagnosis and continues every 2–3 years. Routine MRI surveillance is advised, with frequency adjusted based on symptom progression.

3.2.4 Symptoms and Disease Presentation

The most common symptom is chronic pain and often the earliest symptom of schwannomatosis. It can be severe and may occur even in areas where no tumor is visible or detectable, making it particularly challenging to manage with standard neuropathic medications. Additional symptoms include numbness, tingling, muscle weakness, and headaches, depending on tumor location. Symptoms typically emerge in early adulthood.

3.2.5 Management and Treatment Approaches

Management of Schwannomatosis focuses primarily on pain control and surgical intervention. Surgical removal of painful schwannomas is considered first-line treatment when neurologically safe and has shown to be the most effective option for long term symptom relief. However, the risk of functional impairment must be carefully evaluated, especially when tumors are located near critical nerves or structures.

Generally, radiation therapy is discouraged, particularly in SMARCB1 mutation carriers. Regular pain reviews and psychosocial assessments are recommended.

Genetic counseling should be based on both blood and tumor molecular testing to determine inheritance risk and guide family planning. In cases involving germline pathogenic variants, the transmission risk is approximately 50%, although penetrance is variable, particularly in LZTR1 associated cases.

3.2.6 Prognosis, Frequency and Ongoing Research

Prognosis for individuals with Schwannomatosis is generally favorable. Unlike in NF2, life expectancy is typically normal, although many patients experience a significant impact on quality of life due to chronic, often severe, pain and the need for repeated interventions [1], [10].

Schwannomatosis is rare, with an estimated birth incidence of approximately 1 in 60 000 to 70 000 and a prevalence of around 1 in 100 000. These figures may be underestimated due to diagnostic delays and confusion with related conditions, such as mosaic NF2 or isolated schwannomas [1].

Although the condition accounts for only a small percentage of all schwannoma cases, it is increasingly recognized due to advances in genetic testing and awareness of its distinct clinical features.

Current research efforts focus on identifying additional genetic contributors, improving pain management, enhancing diagnostic accuracy and developing personalized care pathways [1].

Chapter 4

Methodology and System Requirements

4.1 Requirement Analysis

Ther motivation for developing the SENTIA-X stemmed directly from the challenges outlined in the ERN GENTURIS Clinical Guidelines [1]. Schwannomatosis is a rare disease, and many doctors may never encounter it in their clinical practice [1]. As a result, they may lack the necessary experience or structured guidance to make informed decisions about diagnosis and treatment, especially in urgent or uncertain situations.

This system was designed to provide both doctors and patients with a clear, step by step decision making tool that reflects expert agreed guidelines [1]. Its goal is to bridge the knowledge gap, reduce uncertainty and ensure that care is consistent and explainable, even for clinicians unfamiliar with the condition.

The core requirements were organized into three key categories:

- Clinical Requirements:

  The system must support decision-making aligned with ERN GENTURIS guidelines for Schwannomatosis [1]. It should allow clinicians to input patient conditions, receive real time recommendations, and understand the logic behind each suggestion.
- Technical Requirements:

  The platform must be lightweight, web based and compatible with modern browsers. It should integrate with a backend reasoning engine, support real time interaction and be usable.
- User Requirements:

  The interface must be intuitive, responsive, and accessible to clinicians regardless of seniority or technical background. It should provide clear visualizations of patient data and recommendations with explanations that support transparency and trust in the system's decision making.

4.2 Functional and Non-Functional Specifications

Functional Specifications

The system was designed to support doctors in diagnosing and managing patients with Schwannomatosis, following the ERN GENTURIS guidelines [1]. Its core functional capabilities include:

- Patient Management:
  - Add and manage patient records.
  - View patient history and previous recommendations.
- Diagnosis and Recommendation Workflow:
  - Select symptoms and action performed.

    - Receive clinical recommendations based on CNL rules processed by Cognica.
    - Accept or deny recommendations and update the patient's status accordingly.
- Explanation and Transparency:
    - Display verbal explanations for each recommendation (Yes, Maybe, No).
    - Handle indefinite results by suggesting additional actions through modals.
- Logging and Historical Tracking:
    - Store all diagnosis actions and explanations based on result (Yes or maybe).
    - Record and display previous decisions and recommendations per patient.

<u>Non-Functional Specifications</u>

To ensure reliability, security and usability in a clinical context, the following nonfunctional specifications were implemented:

- Performance and scalability:
    - Fast response during interaction with Cognica and while rendering results.
    - Modular design allows for future extensions to other rare diseases.
- Security and Privacy:
    - User authentication using PHP.
    - Data protected with server-side controls and access limited to authenticated doctors.
- Usability:
    - Web based interface, compatible with many browsers.
    - Responsive design, usable across devices (laptops, tablets, mobile phones).
    - Clear layout and tooltips to guide clinicians through each step.
- Maintainability:
    - Clean backend logic with well-structured PHP classes.

    - Separation of frontend logic via reusable JavaScript modules.

4.3 Tools and Technologies Used

The following tools and technologies were used in the design and implementation of the system:

- Frontend: HTML5, CSS, JavaScript were used to build a responsive and interactive user interface.
- Backend: PHP for server-side logic and data handling.
- Database: MySQL was used for structured data storage.
- AI/Reasoning Engine: Cognica platform utilizing Controlled Natural Language (CNL) and argumentation-based logic.

4.4 Knowledge Acquisition and Dataset Preparation

The medical knowledge base of the system was constructed using the official clinical guidelines provided by ERN GENTURIS for Schwannomatosis [1]. Key recommendations were manually extracted and organized into structured tables, Appendix A, each covering one of the six main domains: Diagnosis, Pain Management, Drugs, Surgery, MRI after Diagnosis, and Reproductive. Each guideline entry in the tables was associated with a specific clinical context.

This extraction process was iterative and informed by feedback from clinical experts, guideline contributors and professors with expertise in argumentation logic and Cognica framework.

4.5 From Clinical Guidelines to Decision Logic

Once extracted, the clinical recommendations were analyzed to uncover their underlying conditional logic. Each entry was reformulated as a logical rule suitable for computational reasoning. This was done using CNL, which balances human readability with formal structure and is compatible with Cognica.

The logic statements typically follow an "IF... THEN ALWAYS..." structure, where a combination of patient specific conditions leads to a recommended action. These conditional rules form the core of SENTIA-X's decision-making engine.

Below is a representative subset of rules, categorized according to the six main stages:

<u>Diagnosis Rules:</u>

1. **IF** schwannoma is proven **AND** testdna smarcb1 **AND** (Brain Mri **AND** Spine Mri **AND** Whole Body Mri**) THEN ALWAYS** SMARCB1
2. **IF** schwannoma is proven **AND** testdna lztr1 **AND** (Whole Body Mri **AND** Mri With Fine Cuts) **THEN ALWAYS** LZTR1
3. **IF** testdna lztr1 **AND** analysis Two Tumors **AND** (Whole Body Mri **AND** Mri With Fine Cuts) AND (pain low **OR** pain medium **OR** pain severe) **THEN ALWAYS** LZTR1
4. **IF** testdna non mutation **AND** analysis Two Tumors **AND** testrna **THEN ALWAYS** NF2

<u>Pain Management Rules:</u>

1. **IF** pain low **AND** (SMARCB1 **OR** LZTR1) **AND** psychological needs(optional) **AND** assessments **THEN ALWAYS** Drugs
2. **IF** pain medium **AND** (SMARCB1 **OR** LZTR1) **AND** psychological needs(optional) **AND** assessments **AND NOT** Targeting Pain related disability **THEN ALWAYS** Drugs
3. **IF** pain medium **AND** (SMARCB1 **OR** LZTR1) **AND** psychological needs(optional) **AND** Targeting Pain related disability **AND** surgically Removable **AND** assessments **THEN** ALWAYS Drugs **AND** Surgery

4. **IF** pain severe **AND** (SMARCB1 **OR** LZTR1) **AND** psychological needs(optional) **AND** assessments **AND** surgically Removable **THEN ALWAYS** Drugs **AND** Surgery
5. **IF** pain severe **AND** (SMARCB1 **OR** LZTR1) **AND** psychological needs(optional) **AND** disability **AND NOT** surgically Removable **AND** assessments **THEN ALWAYS** Drugs **AND** Radiotherapy

<u>Drugs Rules:</u>

1. **IF** (SMARCB1 **OR** LZTR1) **AND** schwannoma painful **AND NOT** first Line Drugs Used **THEN ALWAYS** First Line Drugs
2. **IF** (SMARCB1 **OR** LZTR1) **AND** schwannoma painful **AND** first Line Drugs Used **THEN ALWAYS** Second Line Drugs

<u>Surgery Rules:</u>

1. **IF** (SMARCB1 **OR** LZTR1) **AND** schwannomas **AND** (pain medium **OR** pain severe) **AND NOT** neurodamage **THEN ALWAYS** Surgery
2. **IF** (SMARCB1 **OR** LZTR1) **AND** schwannoma symptomatic **AND** (pain medium **OR** pain severe) **AND** neurodamage **AND** surgically Removable **THEN ALWAYS** Surgery by Surgeons of Experience Resecting Nerve Sheath Tumors
3. **IF** (SMARCB1 **OR** LZTR1) **AND** schwannoma symptomatic **AND** (pain medium **OR** pain severe) **AND** neurodamage **AND NOT** surgically Removable **THEN ALWAYS** Assessment of Likelihood of Success **AND** Assessment of Risk of Neurological Loss of Experience Surgeons

<u>MRI after Diagnosis Rules:</u>

1. **IF** (SMARCB1 **OR** LZTR1) **AND** Older Than Young Teen **AND NOT** Schwannomas obvious **AND** Symptoms **AND NOT** Symptoms Change **AND NOT** pain **THEN ALWAYS** Full Cranial Spinal Mri **AND** Whole Body Mri **AND** Frequent Mri
2. **IF** (SMARCB1 **OR** LZTR1) **AND** Older Than Young Teen **AND** Schwannomas obvious **AND NOT** Symptoms **AND NOT** Symptoms Change **THEN ALWAYS** Frequent Mri

3. **IF** (SMARCB1 **OR** LZTR1) **AND** Older Than Young Teen **AND** Schwannomas obvious **AND** Symptoms Change **AND** pain **THEN ALWAYS** Full Cranial Spinal Mri **AND** Whole Body MRI **AND** Mri More Frequent **AND** Ultrasound
4. **IF** (SMARCB1 **OR** LZTR1) **AND** Older Than Young Teen **AND NOT** Schwannomas obvious **AND** Symptoms **AND** pain **THEN ALWAYS** Mri with Fine Cuts **AND** Ultrasound

Reproductive Health Rules:

1. **IF** (SMARCB1 **OR** LZTR1) **AND** age reproductive **AND NOT** pregnancy **THEN ALWAYS** Discussion of The Risk of Transmission **AND** Options of Pregnancy
2. **IF** (SMARCB1 **OR** LZTR1) **AND** age reproductive **AND** pregnancy **THEN ALWAYS** Testing in Pregnancy **AND** Pre-Implantation Genetic Diagnosis

These logic-based statements highlight the diversity and complexity of clinical reasoning embedded within the system. They also illustrate how clinical recommendations are conditionally dependent on multiple variables, reinforcing the value of using a structured, XAI framework for personalized decision support.

4.6 Representation in Tables and Conditional Statements

Firstly, to bridge the gap between clinical guidelines and formal logic, the recommendations extracted organized into structured tables. Each table entry mapped a specific combination of patient conditions such as genetic mutation, pain presence, to a corresponding clinical recommendation or action (see Appendix A).

These tables were developed for all major system modules and served as an intermediate, human readable representation before formalization. This step ensured clarity, consistency and traceability of the encoded medical knowledge.

By clearly separating input conditions from resulting actions, the tables simplified the transformation of clinical recommendations into CNL statements, which could then be processed by Cognica.

Chapter 5

System Implementation and Technical Model

5.1 System Architecture Overview

The system follows a modular, web-based architecture composed of three main layers: the frontend (presentation layer), the backend (application logic layer) and the database (data layer). The doctor interacts through a browser-based interface developed with HTML, CSS and JavaScript. On the server side, the backend is developed with PHP, handles requests, interacts with the database and communicates with Cognica framework to generate clinical recommendations based on user input.

A simplified data flow includes:

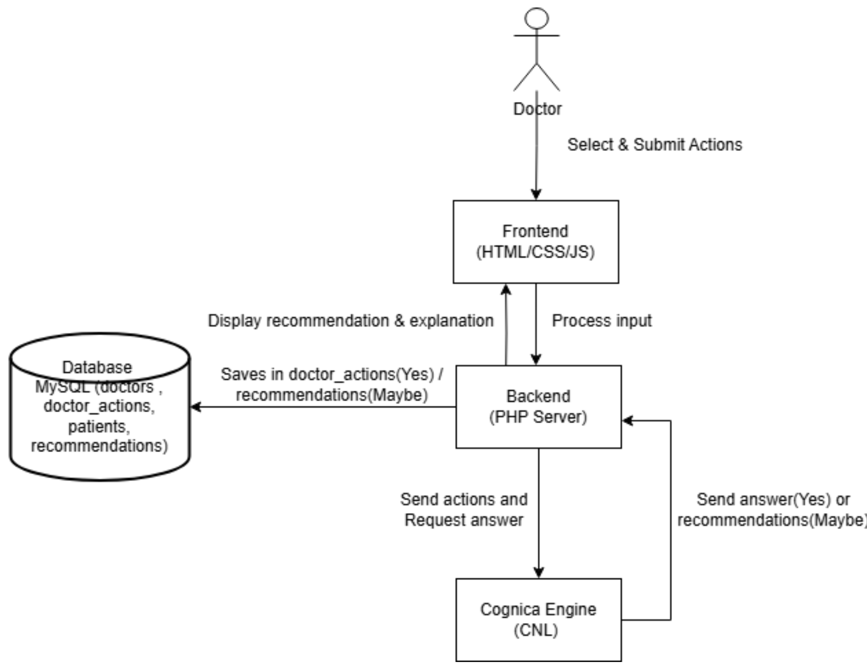


Fig.9 Simplified data flow of SENTIA-X 's system

- Input from doctor
- Server-side processing via PHP
- Response from Cognica with diagnostic output
- Rendering of structured, explainable recommendations in the UI.

5.2 Frontend Development (HTML, CSS, JavaScript)

The Frontend was developed using technologies such as HTML for structure, CSS for styling, while JavaScript was used to add interactivity.

Interactive elements include:

- Toggle buttons that allow the selection of predefined actions.
- Sliders and inputs for Pain levels.
- Modal dialogs for confirming recommendations when results are uncertain (Indefinite Results / Maybe).

- Dynamic rendering of results, which prioritizes ‘Yes’ predictions (Definite Results) and don’t show ‘Maybe / No ’results.
- A patient information sidebar that appears consistently across pages and fetches real-time data using AJAX.

The interface is built with responsiveness and usability in mind, ensuring compatibility with modern browsers and varied screen sizes. Visual hierarchy and consistent layout patterns enhance the experience for clinicians who may not be technically inclined. Additionally, user accessibility and clarity are considered throughout the design, with attention to label clarity, color contrast, and logical grouping of controls.

Overall, the frontend acts as a seamless bridge between the clinical user and the system’s underlying logic, enabling intuitive access to AI-driven decision support without compromising usability.

5.3 Backend Development (PHP)

The language and framework that was used for backend development is PHP, serving as the core of application logic and data flow. It manages all interactions between the frontend, the database and the Cognica decision engine. The backend is designed to ensure consistent and context-aware processing of user inputs and system responses.

A series of modular PHP scripts were developed, and each is responsible for a specific function in the system:

- diagnosisHandler.php , drugsHandler.php , etc : handle the submission of clinical inputs from the doctor, processes them and prepares the query for the Cognica engine using Controlled Natural Language (CNL).
- logDoctorActions.php logs all relevant doctor actions, including diagnoses selected and Accept/Deny decisions made during the workflow.
- update_status.php allows for the updating of diagnosis states in the database, ensuring proper tracking of clinical decision outcomes.

- viewPatient.php is responsible for retrieving and presenting stored patient information and historical data, including diagnoses and system generated recommendations, for clinician review.

Together, these backend components coordinate the flow of clinical data, support explainable AI integration and ensure data persistence and auditability across the platform.

5.4 Database Design and Relationships

The backend of the system uses a relational database implemented in MySQL. The schema is structured to maintain data integrity, support traceability and provide efficient access to patient data, diagnostic actions and Cognica generated recommendations.

Key Tables and their Roles:

- **doctors**: Saves data about clinicians, including first name, last name, email, secure password, phone, qualifications, organization, specialty and experience. Each doctor is uniquely identified by id and can be linked to multiple patients and logged actions.
- **patients**: Contains personal data, such as: First name, last name, email, date of birth and phone. The id is uniquely used throughout the system. Each patient is associated with a doctor id, indicating the doctor that did the examination.
- **doctor_actions**: Logs every action taken during a session, that had led to a definite result(yes), such as doctor id, patient id, Cognica result, actions performed, explanation, action date, status (Accepted, Denied, Pending). Each action is timestamped and linked to both a doctor and a patient.
- **recommendations:** Stores indefinite results(maybe) generated with Cognica. Each record includes patient id, doctor id, date, page name (e.g. Drugs, Pain Management), actions (JSON format) , recommendations (JSON format) and view

status (unread or read). This table allows clinicians to revisit incomplete or uncertain cases and track whether the suggestions have been reviewed.

- **Relationships:**

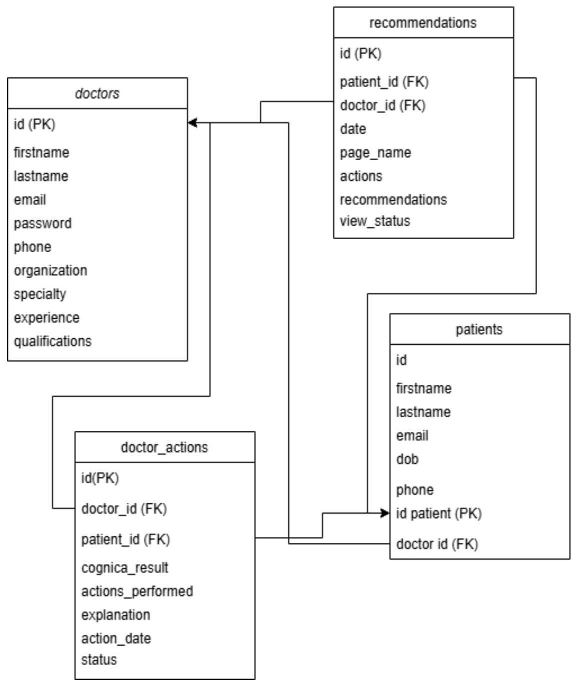


Fig.10 Entity Relationship diagram of the system's database schema

- A one-to-many relationship exists between:
    - doctors and patients (a doctor can manage multiple patients),
    - doctors and doctor_actions (a doctor performs multiple actions),
    - patients and both doctor_actions and recommendations.
- All foreign key constraints are enforced with **ON DELETE CASCADE** to ensure referential integrity. If a doctor or patient is deleted, associated records (actions, recommendations) are automatically removed.

This schema supports robust logging, auditing, and retrieval of medical decisions. The use of JSON in recommendations.actions and recommendations.recommendations enables flexible storage of structured data while preserving queryability.

5.5 Integration with the Cognica Framework

The system integrates the Cognica argumentation framework to enable explainable, logic-based clinical recommendations. Cognica uses CNL to represent medical rules and guidelines in a human-readable yet machine-interpretable format. This allows clinicians to trace and understand the reasoning behind each system generated suggestions, supporting transparency and ethical compliance in AI based healthcare.

From the backend, clinical inputs selected by the doctor are translated into structured CNL queries. These are submitted to Cognica engine through an internal API call.
Once processed, Cognica returns:

- A decision outcome (e.g. Yes, Maybe, No).
- An explanation based on argumentation logic and applicable clinical rules.

The PHP backend receives this response, logs it and sends it to the frontend, where it is displayed alongside the doctor's chosen actions. In cases of Indefinite (Maybe) Results, the system prompts the doctor with further recommendations via a modal dialog. This ensures real time, interactive feedback and maintains clinical transparency.

The integration was implemented in PHP using a custom class named CallCognicaApi. Communication with Cognica's REST API is secure via HTTP Basic Authentication by setting a valid username and password, which allows secure access to the reasoning engine.

The API call required the definition of:

- The medical policy in Cognica DP CNL.
- A query representing the specific conclusion to evaluate.
- A list of complementary options to help the reasoning engine detect and resolve conflicts.
- A fact list, which contained the set of conditions that currently hold or do not hold in the given scenario.

These elements were structured into a JSON payload and submitted to the API endpoint via an HTTP POST request using cURL. A looped API call is used to evaluate multiple queries in a single request cycle, improving performance and maintaining session consistency based on the stage, such as Drugs.

The results, including predictions, explanations and sometimes recommendations when we have maybe, are dynamically rendered on the frontend. Doctors can accept or deny each prediction and interact with follow up prompts, ensuring transparency, interpretability and ethical compliance in AI driven clinical decisions.

5.6 Contraindication Tables and Recommendation Engine

The foundation of the SENTIA-X system was built upon structured medical recommendations derived from ERN Genturis clinical guidelines for Schwannomatosis [1]. These recommendations were initially organized in tabular format, mapping specific clinical scenarios to corresponding treatment decisions. The tables spanned key areas such as:

- Diagnosis
- Pain Management
- Drugs
- Surgery
- MRI after Diagnosis
- Reproductive Planning

Each row encoded a combination of clinical conditions and the corresponding medical recommendation, which were reviewed and validated for consistency.

Following this, the content was translated into Controlled Natural Language (CNL) to be compatible with Cognica argumentation framework. This transformation allowed human readable medical rules to be expressed as machine interpretable logical rules. Each page of the system (e.g. Drugs) was assigned its own CNL rule set.

These rules were sent into the Cognica engine to generate a result or recommendations, based on the user's clinical inputs. The system supports both deterministic ("Yes") results and probabilistic ("Maybe") results, when uncertainty exists. In such cases, it proposes additional recommended actions that can help narrow down the decision.

5.7 Explanation Generation and User Interaction Logic

Once Cognica returns a decision, the explanation associated with the outcome is extracted and shown to the user. This explanation is derived from the logical argument chain evaluated by Cognica, providing transparency into how the system reached its decision.

In the frontend, the system distinguishes between three types of outcomes:

- **Yes:** Only the result and explanation are displayed. Accept/Deny buttons become active.
- **Maybe:** A modal dialog is triggered showing recommended actions. If the doctor selects actions and confirms, the system re-queries Cognica.
- **No:** Only the result is presented, with no further interaction required.

The user interface logic ensures that:

- Maybe-based modals appear only once per session, to avoid repetition.
- If no new actions are selected or the modal is closed , the system stores the previously displayed results.
- Accept/Deny states are updated in the database and visually reflected.

5.8 User Roles and Permissions

SENTIA-X currently supports a single authenticated user role: Doctor.

Upon login, doctors can:

- View and manage only the patients that they have added.
- Perform diagnoses across various models.
- Access patient history and previous recommendations.
- Accept or deny system generated outcomes.

Although multi-role support (e.g., Admin, Researcher) is not implemented in this version, the code structure allows easy extension. All endpoints involving sensitive data are protected by PHP session-based authentication.

5.9 Logging and Historical Tracking

The system maintains detailed logs to ensure traceability and accountability of clinical actions:

- doctor_actions table: Stores every diagnosis interaction that resulted in a definitive answer ("Yes") along with action context and system explanation.
- recommendations table: Stores all Maybe results and recommended actions for review.

Each entry is timestamped and linked to both doctor and patient. This structure allows full traceability of medical decisions.

5.10 User Manual

The full source code is available at: https://github.com/akyria25/sentia-x.git

Interface and Features Overview

- **Name and Logo**

  SENTIA-X : Schwannomatosis ENgine for Treatment Insight & eXplainable Analytics

✓ Logo:

The logo reflects features a modern, abstract design that combines a heart and brain shape, symbolizing the connection between clinical care and intelligent reasoning. The soft gradient colors purple and blue reflect trust, compassion and technology. The lines form a symmetrical, flowing structure, suggesting clarity, balance and continuous care.

- **Login & Registration**
  - ✓ Secure authentication is required for all users.

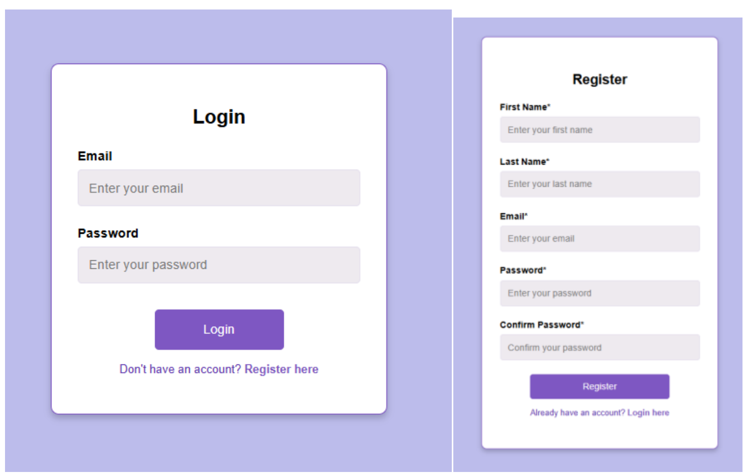


Fig.11 Screenshot of Login and Register page

- **Dashboard**
    - ✓ Upon login, the doctor is directed to the Dashboard, with a navigation bar and shortcuts to key features.

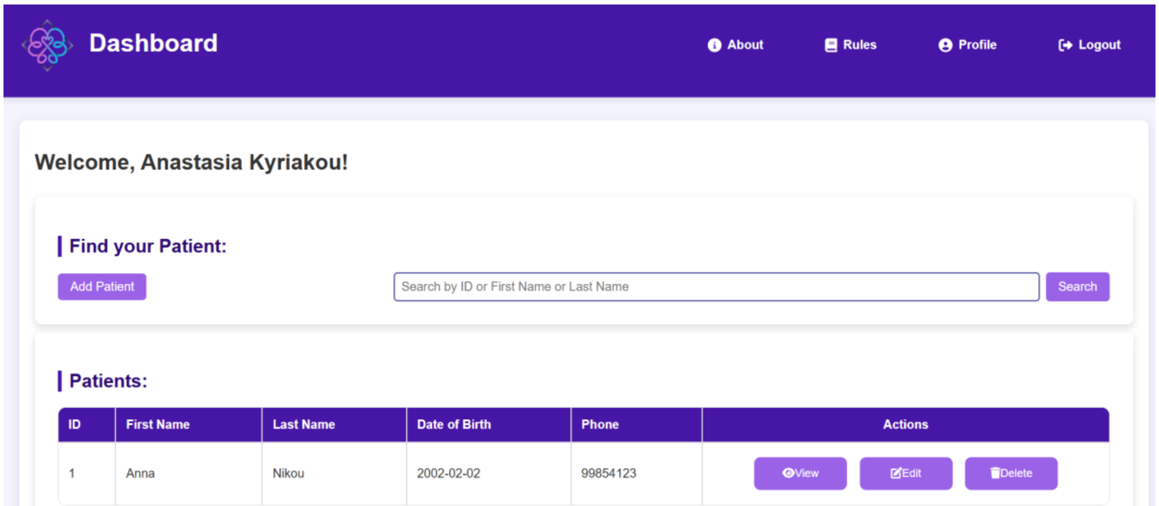


Fig. 12 Screenshot of SENTIA-X's dashboard

- **Navigation Bar**
    - ✓ About: Provides background on Schwannomatosis including disease subtypes, symptoms, and an overview of Cognica and the Explainable AI framework that powers the system.

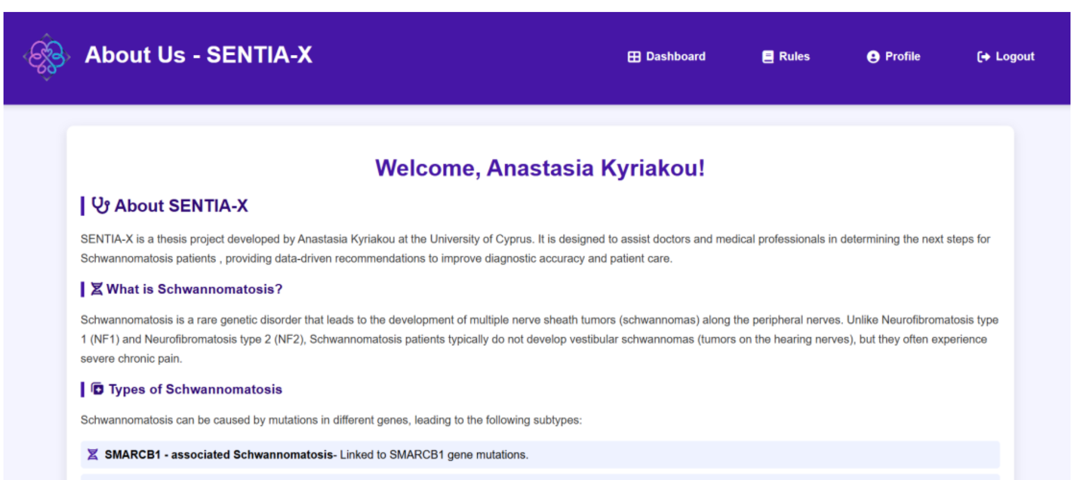


Fig.13 Screenshot of About us page

- ✓ Rules: Displays all the clinical decision rules used across the system, organized by diagnostic stage.

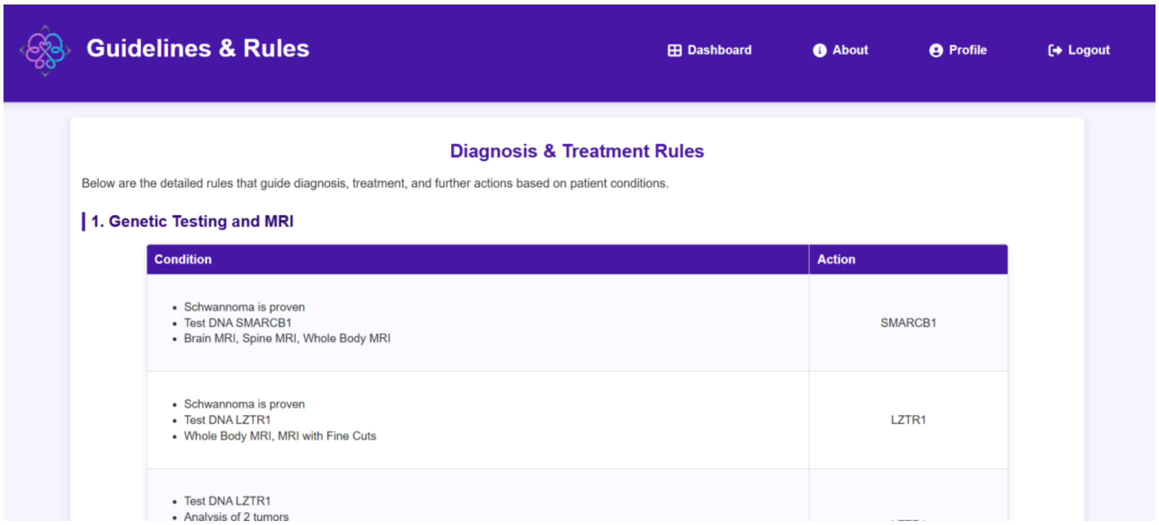


Fig.14 Screenshot of Rules page

- ✓ Profile: Displays doctor's information.

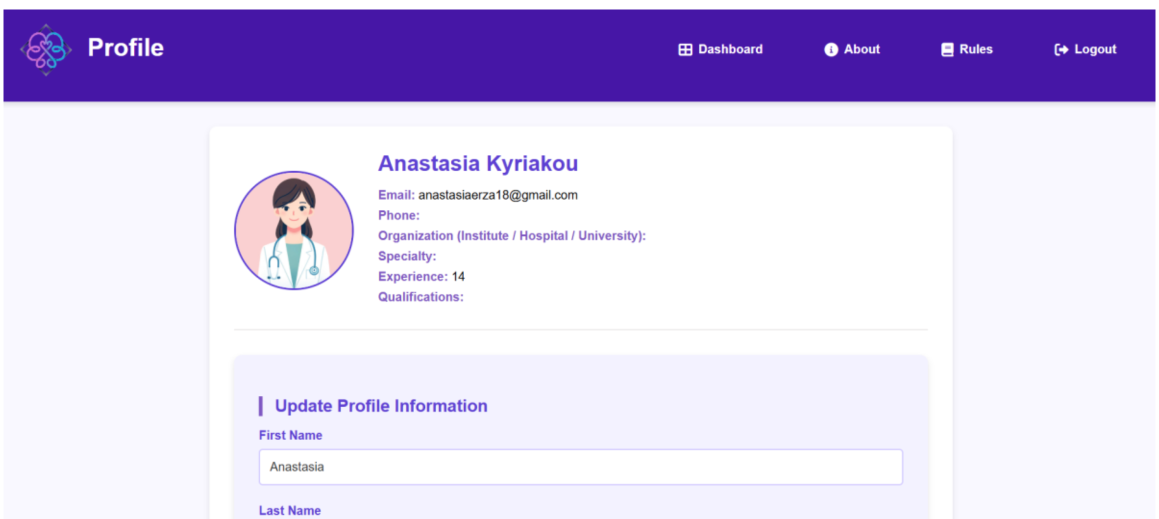


Fig.15 Screenshot of Profiles page

- ✓ Logout: Securely logs out the user out of the session.

- **Patient Management**
  - ✓ Find Your Patient
    - Search for patients by First Name, Last Name, or Patient ID.

Patients:

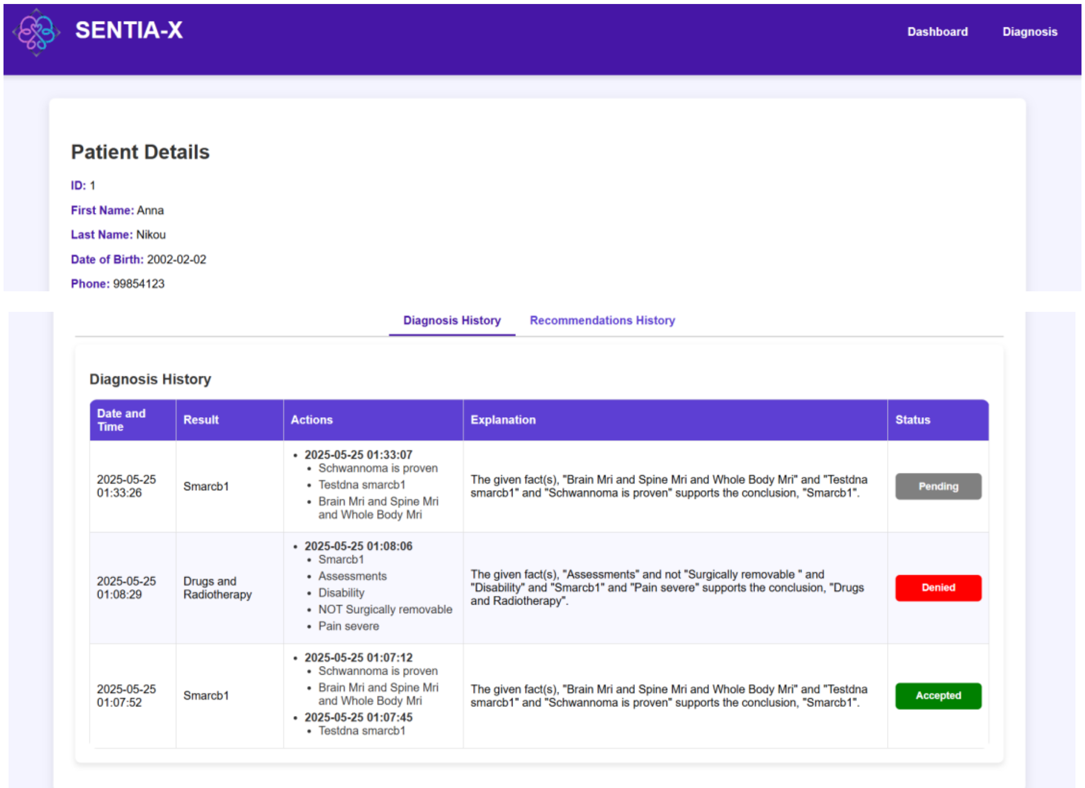
ID	First Name	Last Name	Date of Birth	Phone	Actions
1	Anna	Nikou	2002-02-02	99854123	View Edit Delete

Fig.16 Screenshot of Patient’s table.

    - Add new patient records using the “Add New Patient” form.

  - ✓ Patients List: A searchable and sortable table with:
    - Patient ID
    - First Name
    - Last Name
    - Date of Birth
    - Phone Number
  - ✓ Actions available per patient:
    - View: Access full patient profile.
    - Edit: Modify patient details.
    - Delete: Permanently remove the patient record.

- **Patient View Page**
  - ✓ Patient’s details
    - Displays static information (name, DOB, contact info).
  - ✓ Diagnosis History: Lists all previous diagnoses including:
    - Date
    - Result (Yes/No)
    - Performed actions
    - AI-generated explanation



SENTIA-X

Dashboard Diagnosis

Patient Details

ID: 1
First Name: Anna
Last Name: Nikou
Date of Birth: 2002-02-02
Phone: 99854123

Diagnosis History | Recommendations History

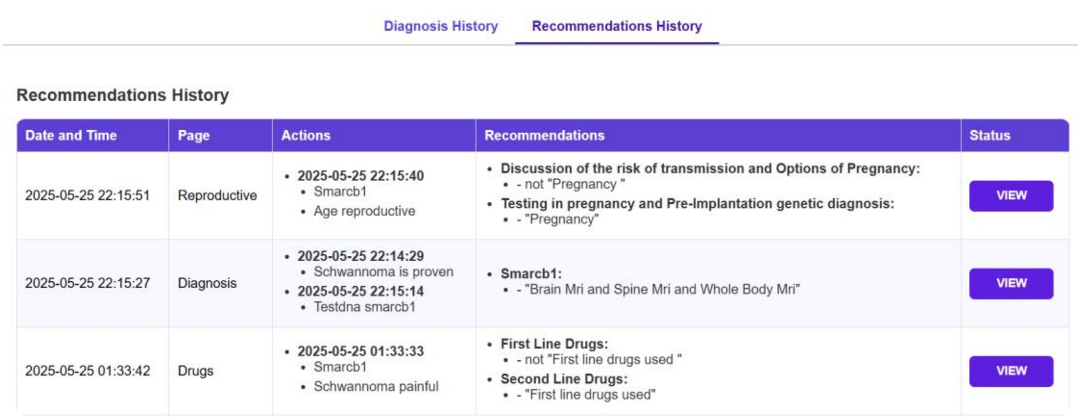
Diagnosis History

Date and Time	Result	Actions	Explanation	Status
2025-05-25 01:33:26	Smarcb1	• 2025-05-25 01:33:07 • Schwannoma is proven • Testdna smarcb1 • Brain Mri and Spine Mri and Whole Body Mri	The given fact(s), "Brain Mri and Spine Mri and Whole Body Mri" and "Testdna smarcb1" and "Schwannoma is proven" supports the conclusion, "Smarcb1".	Pending
2025-05-25 01:08:29	Drugs and Radiotherapy	• 2025-05-25 01:08:06 • Smarcb1 • Assessments • Disability • NOT Surgically removable • Pain severe	The given fact(s), "Assessments" and not "Surgically removable " and "Disability" and "Smarcb1" and "Pain severe" supports the conclusion, "Drugs and Radiotherapy".	Denied
2025-05-25 01:07:52	Smarcb1	• 2025-05-25 01:07:12 • Schwannoma is proven • Brain Mri and Spine Mri and Whole Body Mri • 2025-05-25 01:07:45 • Testdna smarcb1	The given fact(s), "Brain Mri and Spine Mri and Whole Body Mri" and "Testdna smarcb1" and "Schwannoma is proven" supports the conclusion, "Smarcb1".	Accepted

Fig.17 Screenshot of Diagnosis History table.

✓ Recommendations History: Shows all “Maybe” cases:

- Date
- Diagnostic stage
- Actions taken
- Predictions and assumptions
- Doctor’s response and status



Diagnosis History | Recommendations History

Recommendations History

Date and Time	Page	Actions	Recommendations	Status
2025-05-25 22:15:51	Reproductive	• 2025-05-25 22:15:40 • Smarcb1 • Age reproductive	• Discussion of the risk of transmission and Options of Pregnancy: • - not "Pregnancy " • Testing in pregnancy and Pre-Implantation genetic diagnosis: • - "Pregnancy"	VIEW
2025-05-25 22:15:27	Diagnosis	• 2025-05-25 22:14:29 • Schwannoma is proven • 2025-05-25 22:15:14 • Testdna smarcb1	• Smarcb1: • - "Brain Mri and Spine Mri and Whole Body Mri"	VIEW
2025-05-25 01:33:42	Drugs	• 2025-05-25 01:33:33 • Smarcb1 • Schwannoma painful	• First Line Drugs: • - not "First line drugs used " • Second Line Drugs: • - "First line drugs used"	VIEW

Fig.18 Screenshot of Recommendations History table.

- **Diagnosis Page**

  - Step 1: Action Selection

    The doctor selects relevant symptoms, genetic results, imaging findings, or treatment actions using toggle buttons.

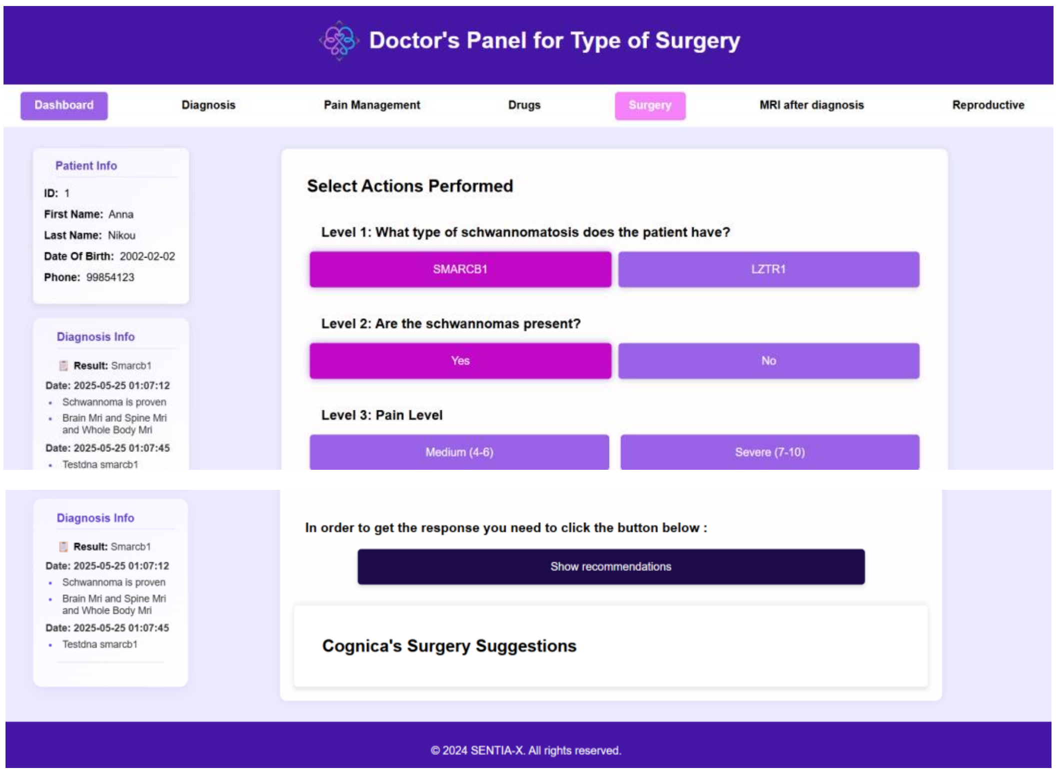


Fig.19 Screenshot of Diagnosis Page - flow.

  - Step 2: Generate Diagnosis

  Click “Show Diagnosis” to submit data.

    ✓ System Response:

    ✓ **“Yes” Result:**

    A confirmed diagnosis is returned with a concise explanation from Cognica. This is logged in the Diagnosis History.

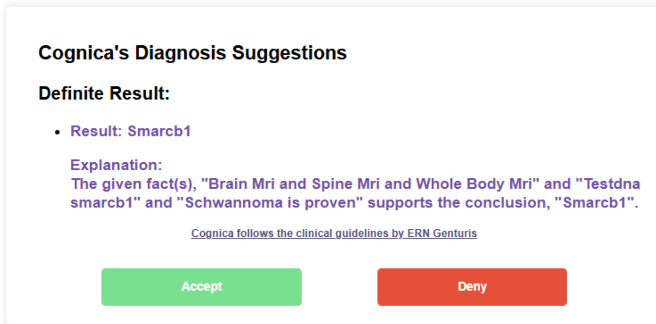


Fig.20 Screenshot of a Definite result displaying prediction and the explanation.

✓ **"Maybe" Result":**

The system presents likely diagnoses and recommended next steps. A modal allows the doctor to select or reject assumptions before proceeding. All entries are stored in Recommendations History.

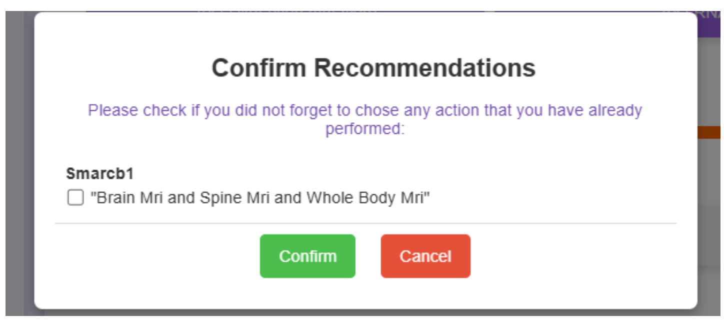


Fig.21 Screenshot of the modal recommendation.

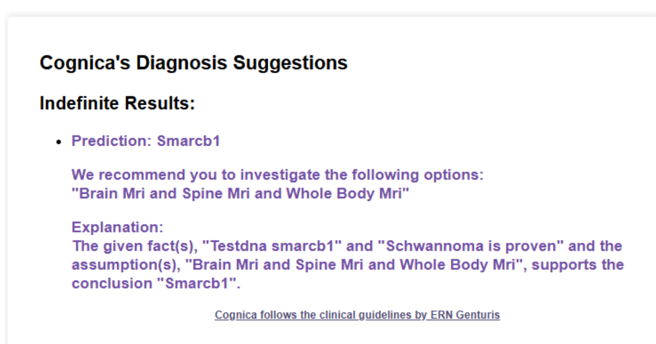


Fig.22 Screenshot of an Indefinite result displaying prediction, recommendation and the explanation.

- **Diagnostic Categories:**
    1. Diagnosis
    2. Pain Management
    3. Drugs
    4. Surgery
    5. MRI After Diagnosis
    6. Reproductive

- **Edit Patient**

  Allow authorized users to update:
    - ✓ First Name
    - ✓ Last Name
    - ✓ Email
    - ✓ Date of Birth
    - ✓ Phone Number

- **Delete Patient**
    - Permanently deletes the selected patient record from the system.
    - This action is irreversible and should be used with caution.

Chapter 6

Evaluation and Results

6.1 Evaluation Setup and Methodology

The evaluation for SENTIA-X CDSS was designed to assess the functionality, usability, and clinical relevance of the system in real-world scenarios, particularly for rare diseases like schwannomatosis. The methodology involves structured testing, simulation with user personas and professional feedback.

Although the full questionnaire-based evaluation is scheduled to take place soon, the system has already been demonstrated to healthcare professionals during the seminar NF Summit 2025 in Barcelona. During that event, Mrs. Melpo presented SENTIA-X to a group of clinicians, researchers and patients.

Early Impressions from Barcelona

During the NF Summit 2025 in Barcelona, the system was introduced to clinicians, researchers and patients. Feedback was encouraging: one doctor expressed interest in future collaboration and patients welcomed the system's potential to make complex information more accessible and understandable.

While informal, this early feedback helped validate the system's conceptual approach and informed refinements to the upcoming structured evaluation.

6.2 Performance and Usability Testing

To simulate realistic use of the system, a user testing scenario was prepared based on a detailed patient persona "Alex P." designed to mimic a typical case of LZTR1 related schwannomatosis.

User Testing Scenario

User Persona:

Alex P., a 34-year-old administrative assistant, has been experiencing persistent, sharp, burning pain in the left thigh for over a year, which they rate as 7 out of 10 on a numerical pain scale. The pain significantly affects their ability to walk, focus at work, and carry out daily activities. They also report intermittent tingling in the right forearm.

Alex has a family history of nerve tumors, their father was diagnosed with multiple schwannomas. Clinical examination and imaging revealed multiple peripheral nerve schwannomas with no vestibular involvement. While mild sensory deficits were present, no motor impairment was observed. Genetic testing confirmed a pathogenic variant in the LZTR1 gene, resulting in a diagnosis of LZTR1-related schwannomatosis.

Current Care Plan:

Alex has recently started gabapentin for chronic neuropathic pain, with dose titration ongoing to balance efficacy and tolerability. The care team is considering the addition of duloxetine if pain persists. Physical therapy and cognitive behavioral therapy (CBT) have also been recommended to address functional limitations and emotional well-being.

Given the severity of pain and its impact on mobility, Alex has been referred to a neurosurgeon. A targeted resection of the most symptomatic schwannoma is planned, with intraoperative nerve monitoring to preserve function. The aim of surgery is palliative, focusing on pain relief and quality of life rather than curative treatment.

Clinicians participating in the future evaluation will use the system to:

- Navigate Alex's medical profile
- Select and assess potential diagnoses
- Review system generated recommendations
- Accept or Reject suggestions
- View past actions and historical decisions
- Continue incomplete diagnosis workflows

This structured interaction will help determine how effectively the system supports real clinical workflows.

6.3 Metrics and System Effectiveness

A comprehensive evaluation was designed, although not yet deployed. It includes validated assessment instruments across usability, satisfaction, and trust. The structured questionnaire is divided into the following parts:

1. Demographics
    - Basic information such as age, gender, professional role and experience.
2. Visual Analogue Scale (VAS)
    - Records participants reactions in different social domains such as usability, acceptability and satisfaction.
3. System Usability Scale (SUS)
    - A 5-point Likert Scale to assess usability.
4. Single Ease Question (SEQ)
    - A 7-point Likert scale on difficulty of engagement with the system.

5. Questionnaire for User Interface Satisfaction (QUIS)
    - Evaluates satisfaction across five Ui domains:
        - Overall reaction to the software
        - Screen
        - Terminology and system information
        - Learning
        - System capabilities
6. User experience Questionnaire (UEQ)
    - A 7-point Likert Scale to measure:
        - Efficiency
        - Perspicuity
        - Dependability
        - Stimulation
        - Novelty
7. Treatment Acceptability Rating Form (TARF)
    - A 7-point Likert scale to assess the acceptability of the use of the holographic solution in a healthcare setting from the perspective of MD's.

These instruments will be used in the upcoming structured evaluation with clinicians, to gather both quantitative and qualitative data on how the system performs in a real clinical environment.

6.4 Results and Observations

While formal testing using structured questionnaires has not yet been conducted, initial feedback from clinical professionals during the NF Summit 2025 seminar in Barcelona was notably positive. During the event, the system was presented to both doctors and patients, offering a preview of its interface and logic-based recommendations.

Clinical users found the system's XAI outputs intuitive and informative. The argumentation-based recommendations were perceived as clear, context-aware, and easy to navigate, with intuitive accept/deny functionality. Several clinicians noted that the system could be especially useful for junior clinicians or in situations where specialist guidance is not always available. One doctor expressed active interest in the system and requested further details for potential collaboration.

Some limitations were noted, such as the need for:

- Broader clinical guideline coverage.
- Better support for multilingual explanations.

Nevertheless, all testers agreed that the system provided meaningful support for managing Schwannomatosis and could be expanded to other rare diseases with minimal architectural changes. Additionally, due to the limited number of specialists in this rare condition, it may be necessary to identify and collaborate with expert doctors abroad.

Chapter 7

Conclusion, Challenges and Future Work

7.1 Conclusion

This thesis presented the design and implementation of SENTIA-X, a Clinical Decision Support System for Schwannomatosis, integrating modern web technologies and XAI-driven logic through the Cognica framework. The system improves traditional patient data recording methods by offering a digital, secure, and standardized platform tailored to rare diseases.

Key features include:

- A modular web interface for inputting clinical information.
- Real-time, transparent recommendations based on argumentation logic
- Support for viewing past decisions and continuing incomplete workflows

Although the formal evaluation has not yet taken place, initial feedback gathered during the NF Summit 2025 in Barcelona was promising. Healthcare professionals found the system's recommendations clear and actionable and several expressed interest in future collaborations. These early insights suggest that SENTIA-X can provide valuable decision support, especially in settings where specialist expertise is limited.

7.2 Implementation and Technical Challenges

Developing SENTIA-X involved integrating a logic-based reasoning engine with a web based clinical interface, which introduced several technical challenges:

- Modular Integrations with the Cognica Framework:
    - One of the primary challenges was translating natural language clinical rules into a format compatible with Cognica’s formal argumentation structure. Ensuring consistency, accuracy and traceability across rules required extensive validation and testing.
- State Management and Workflow Continuity:
    - Enabling clinicians to save, resume and review previous actions introduced complexity in frontend backend synchronization. Careful management of user session data, patient states and action histories were necessary to ensure workflow continuity.
- Frontend Responsiveness and Usability:
    - The interface needed to accommodate complex interactions while remaining responsive and intuitive. Achieving this balance required careful UI design and real time feedback mechanisms. Performance testing helped identify bottlenecks, especially in rendering dynamic diagnosis and recommendation components.
- Data Privacy and Security Considerations:
    - From the early stages, the implementation followed security practices such as session management, server-side validation and secure data handling protocols.

7.3 Limitations in Clinical Usability

While SENTIA-X demonstrates potential as a clinical support tool, several limitations affect its current usability in real world healthcare environments:

- **Limited Scope of Clinical Rules:**
  The system currently supports only schwannomatosis-related decision making. This narrow focus limits its utility in more general clinical workflows, where differential diagnosis may be present. Expansion to related rare conditions would enhance clinical value.
- **Pending Formal Usability Evaluation:**
  Although early feedback from healthcare professionals has been positive, a full scale evaluation using standardized instruments (e.g., SUS, QUIS, UEQ) has not yet been conducted. As a result, user interface improvements are currently based on internal testing and informal feedback.
- **Lack of Role Differentiation:**
  At present, the platform offers single user experience for all clinical users.
- **Language Constraints:**
  All system content is currently in English, which may limit adoption in multilingual or non-English speaking clinical settings.

These limitations will guide future iterations of SENTIA-X with emphasis on improved usability and integration.

7.4 Future Work and Enhancements

Several areas pf improvement have been identified to guide future development

- **Structured Usability Study:**
  The evaluation instruments described in Chapter 6 will be deployed in upcoming studies involving medical professionals. This will provide quantitative data on usability, satisfaction and trust.

- **Role based Multi-User Access:**

  Future iterations will introduce differentiated roles for clinicians, nurses and administrators enabling collaborative workflows.

- **EMR Integration:**

  Interfacing the system with existing hospital infrastructures will be explored to support real time data access and documentation.

- **Multilingual Interface:**

  Support for additional languages will be implemented to ensure broader international usability.

- **Collaborations and Pilot Deployments:**

  Initial interest from clinicians abroad (e.g. at the NF Summit 2025) will be followed up with structured pilots to validate the system in real clinical settings.

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Appendix

Appendix A: Decision policy tables

This appendix contains the complete set of structured tables used for translating clinical guidelines into decision logic. Each table represents one of six main system modules and outlines the condition-action pairs used to generate formal reasoning rules in SENTIA-X.

Diagnosis

Scenario	SWN(SMARCB1)	SWN (LZTR1)	Mosaic NF2-SWN
Schwannoma proven	YES	YES	
Schwannoma proven **AND** Test DNA Smarcb1 **AND** Brain MRI **AND** Spine MRI **AND** Whole Body MRI	YES		
Schwannoma proven **AND** Test DNA Lztr1 **AND** Whole Body MRI **AND** MRI With Fine Cuts		YES	
Test DNA Lztr1 **AND** Analysis Two Tumors **AND** Whole Body MRI **AND** MRI with Fine Cuts **AND** (Pain low **OR** Pain medium **OR** Pain severe)		YES	
Test DNA Non-Mutation **AND** Analysis Two Tumors **AND** Test RNA			YES

Pain Management

Scenario	Drugs	Surgery	Radiotherapy
Pain low **AND** (SMARCB1 **OR** LZTR1) **AND Psychological Needs** (optional) **AND Assessments**	YES		
Pain medium **AND** (SMARCB1 **OR** LZTR1) **AND Psychological Needs** (optional) **AND** Assessments **AND NOT** Targeting Pain related disability	YES		
Pain medium **AND** (SMARCB1 **OR** LZTR1) **AND Psychological Needs** (optional) **AND** Targeting Pain Related Disability **AND Surgically** Removable **AND Assessments**	YES	YES	
Pain severe **AND** (SMARCB1 **OR** LZTR1) **AND Psychological Needs** (optional) **AND Assessments AND Surgically** Removable	YES	YES	
Pain severe **AND** (SMARCB1 **OR** LZTR1) **AND** Psychological Needs (optional) **AND** Disability **AND NOT** Surgically Removable **AND** Assessments	YES		YES

Drugs

Scenario	First Line Drugs	Second Line Drugs
(SMARCB1 **OR** LZTR1) **AND** Schwannoma Painful **AND NOT** First Line Drugs Used	YES	
(SMARCB1 **OR** LZTR1) **AND** Schwannoma Painful **AND** First Line Drugs Used		YES

Surgery

Scenario	Surgery	Surgery By Surgeons of Experience Resecting Nerve Sheath Tumors	Assessment of Likelihood of Success AND Assessment of Risk of Neurological Loss of Experience Surgeons
(SMARCB1 **OR** LZTR1) **AND** Schwannomas **AND** (Pain medium **OR** pain severe) **AND NOT** Neurodamage	YES		
(SMARCB1 **OR** LZTR1) **AND** Schwannoma Symptomatic **AND** (Pain medium **OR** Pain severe) **AND** Neurodamage **AND** Surgically Removable		YES	
(SMARCB1 **OR** LZTR1) **AND** Schwannoma Symptomatic **AND** (Pain medium **OR** Pain severe) **AND** Neurodamage **AND NOT** Surgically Removable			YES

MRI after Diagnosis

Scenario	Full Cranial Spinal MRI	Whole Body MRI	Frequent MRI	More Frequent	MRI With Fine Cuts	Ultrasound
(SMARCB1 **OR** LZTR1) **AND** Older Than Young Teen **AND NOT** Schwannomas obvious **AND** Symptoms **AND NOT** Symptoms Change **AND NOT** Pain	YES	YES	YES			

(SMARCB1 **OR** LZTR1) **AND** Older Than Young Teen **AND** Schwannomas obvious **AND NOT** Symptoms **AND NOT** Symptoms Change			YES			
(SMARCB1 **OR** LZTR1) **AND** Older Than Young Teen **AND** Schwannomas obviously **AND** Symptoms Change **AND** Pain	YES	YES		YES		YES
(SMARCB1 **OR** LZTR1) **AND** Older Than Young Teen **AND NOT** Schwannomas obvious **AND** Symptoms **AND** Pain					YES	YES

Reproductive

Scenario	**Discussion of the Risk of Transmission AND Options of Pregnancy**	**Testing in Pregnancy AND Pre-Implantation Genetic Diagnosis**
(SMARCB1 **OR** LZTR1) **AND** Age Reproductive **AND NOT** Pregnancy	YES	
(SMARCB1 **OR** LZTR1) **AND** Age Reproductive **AND** Pregnancy		YES

Appendix B – Technical Guide

Technologies used

The full source code is available at: https://github.com/akyria25/sentia-x.git

The system was developed using the following technologies:

- PHP: Server-side scripting language for handling backend logic and API calls.
- JavaScript: For frontend interactivity and dynamic behavior.
- HTML/CSS: For structuring and styling the user interface.
- MySQL: For logging history and storing patient data.
- Cognica Framework: An argumentation engine used to generate explainable recommendations based on clinical rules.
- Apache (XAMPP): Local server environment for running PHP code during development.

System Architecture and Module Overview

The system is a web-based CDSS design to assist doctors with diagnosis and treatment recommendations for Schwannomatosis. It consists of:

- **Frontend (JavaScript + HTML/CSS)**
    - Collects user input through buttons, toggles and form fields.
    - Manages modal dialogs for recommendations.
    - Displays results: Yes, Maybe, or No along with explanations and recommendations when the result is Maybe.
    - Files:
        - scriptDiagnosisModule.js: Shared utility functions for diagnosis handling
        - diagnosis.js, drugs.js, etc.: Page-specific logic depends on the stage
- **Backend (PHP)**
    - Handles requests from the frontend and communicates with Cognica.
    - Logs selected actions and stores feedback from users

- Key files:
  - diagnosisHandler.php, drugsHandler.php: Sends facts and queries to Cognica and returns results.
  - saveRecommendations.php: Stores user selections and recommendations in the database.
  - logDoctorActions.php: Stores user actions when the result is definite in the database.
  - update_status.php: Updates the status (accepted/denied) of a recommendation.

- **Cognica Engine**
  - Receives structured facts based on clinical actions.
  - Applies argumentation logic using ERN GENTURIS guidelines.
  - Returns transparent, explainable recommendations.

Appendix C – User Guide

This appendix provides a step-by-step guide for end users (medical professionals) to use the SENTIA-X Clinical Decision Support System for Schwannomatosis diagnosis and treatment planning.

System Deployment and Access

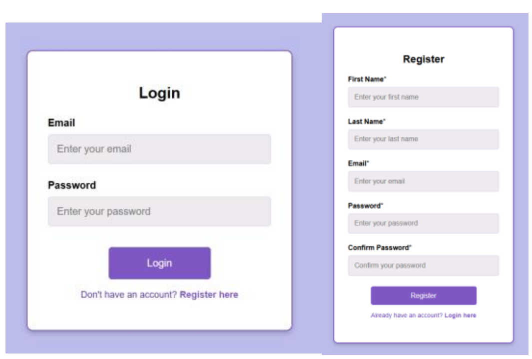
1. The SENTIA-X Clinical Decision Support System is deployed on the Department of Computer Science infrastructure at the University of Cyprus.

- **UCY Development URL:**
  https://thesis.in.cs.ucy.ac.cy/AI_Rare/login.php
  Accessible only within the UCY network or via VPN connection.

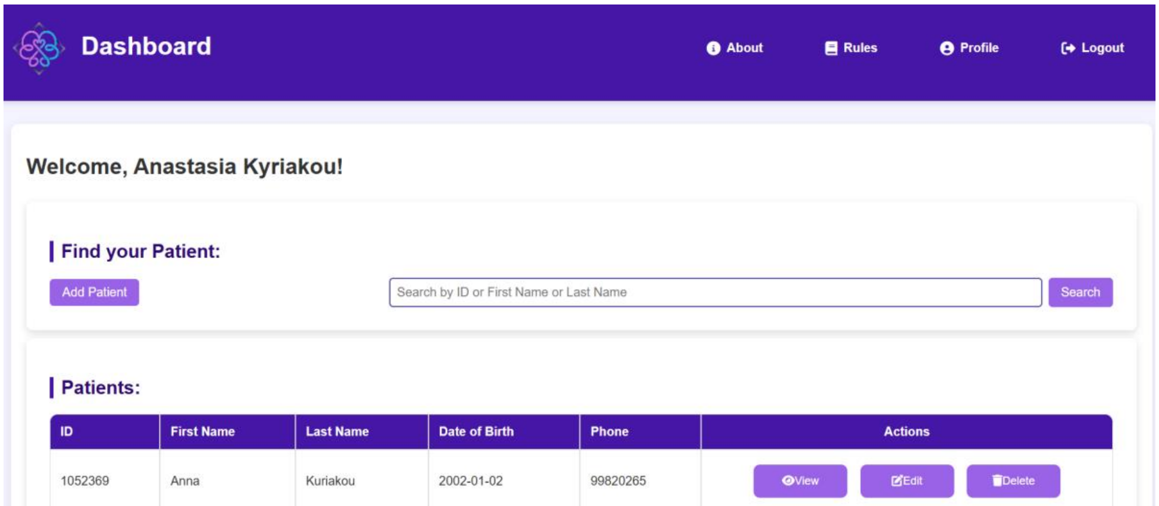
- **Public Deployment:**
  https://ai-rare.rf.gd/
  A publicly accessible version for demonstration purposes.

- **Login / Register:**

Before accessing the system, users must register for a new account or log in with existing credentials via the login page



- **Viewing Patient Information**



From the main interface, users can:

- Select an existing patient
- Manually enter a patient ID
- Add a new patient to the system.

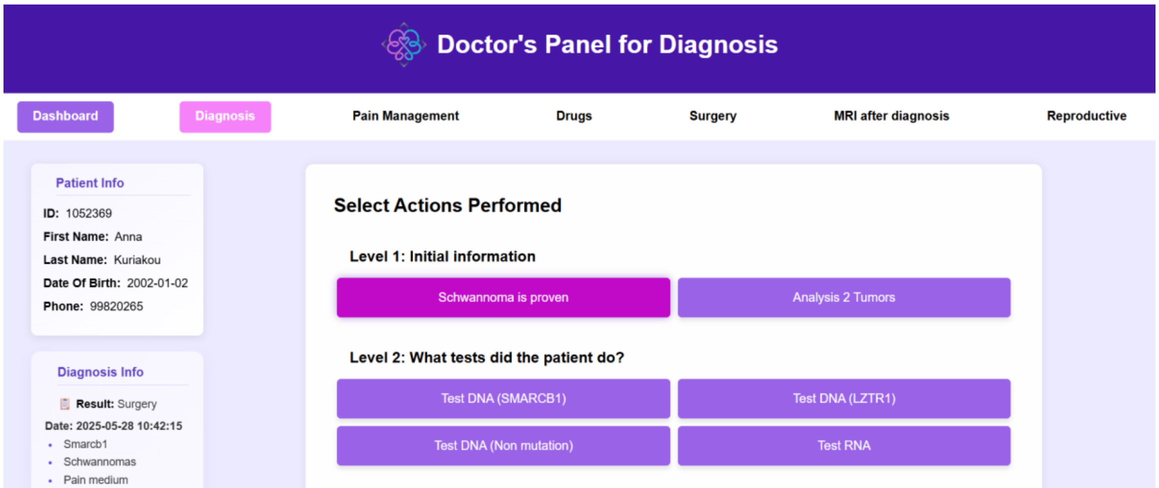
Upon clicking the "View" button for a specific patient:

- The patient's personal information (name, date of birth, phone number, etc.) are displayed.
- A Diagnosis History table is shown.
- A Recommendations History table is shown.

To proceed to the diagnosis process, users can click "Diagnosis" from the navigation bar.

- This section displays the available stages of patient care, allowing users to select clinical actions, receive recommendations, and record decisions.

- **Selecting Diagnoses or Clinical Actions**

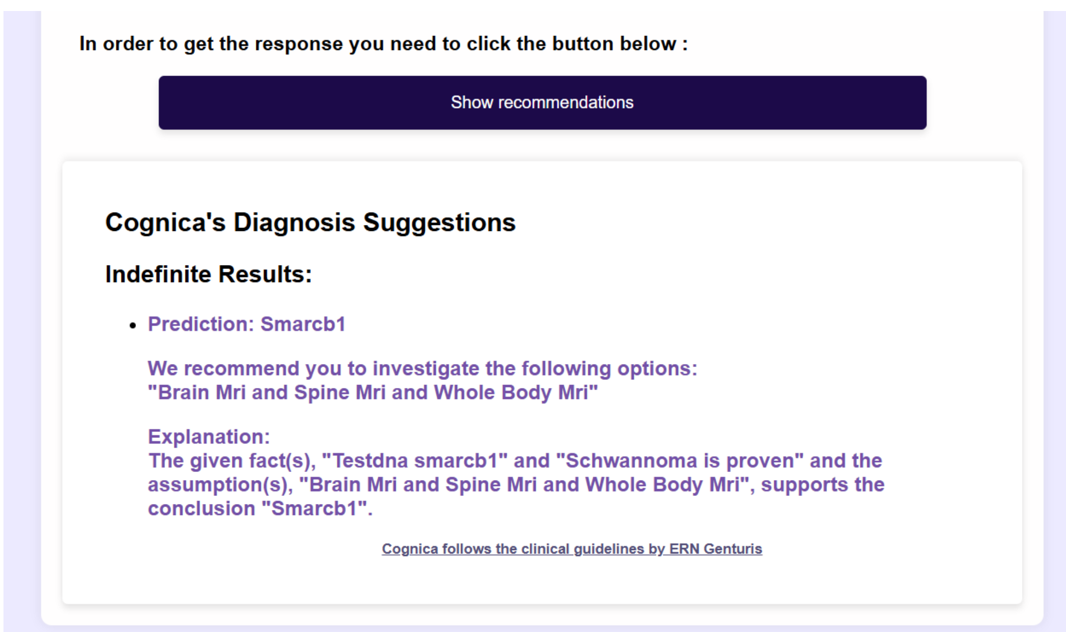


Navigate to the relevant page (e.g., Diagnosis, Drugs, Pain Management).

Use toggle buttons or form inputs to select clinical actions:

- Example: Select "Schwannoma is proven" by clicking it.
- Some pages also include options like pain levels or treatment lines.

- **Generating Recommendations**



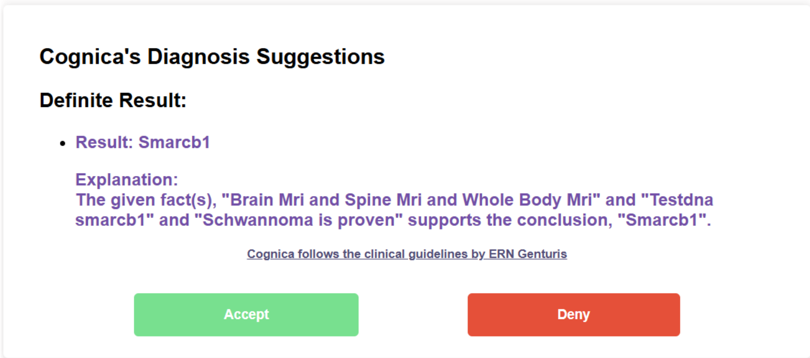
After selecting the desired inputs:

- Click the Show recommendations button.
- The system will query the Cognica engine in the background.

The result will be displayed as:

- Yes – Definite result
- Maybe – Indefinite result
- No result – There are no other actions needed.

- **Accept or Deny a Recommendation**

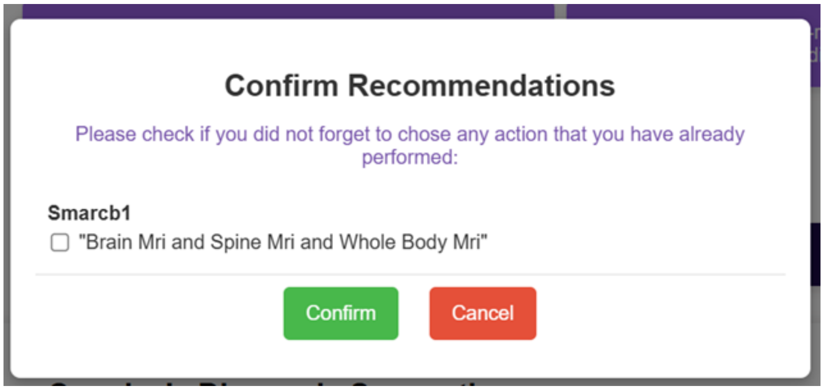


When a result is returned as "Yes", two buttons will be shown:

  - Accept – The doctor agrees with the recommendation as applied
  - Deny – The doctor rejects the recommendation

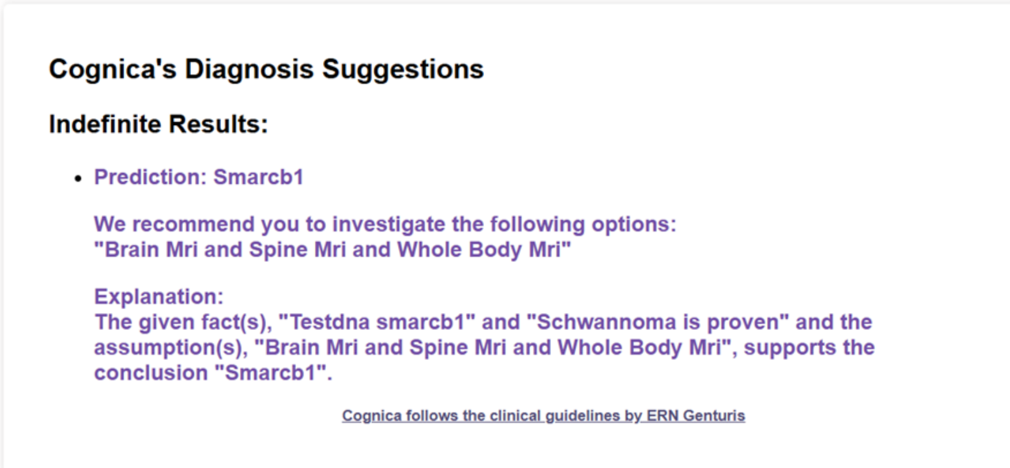
Then, the system will store doctor's decision in doctors_actions table.

- **Handling 'Maybe' Recommendations**



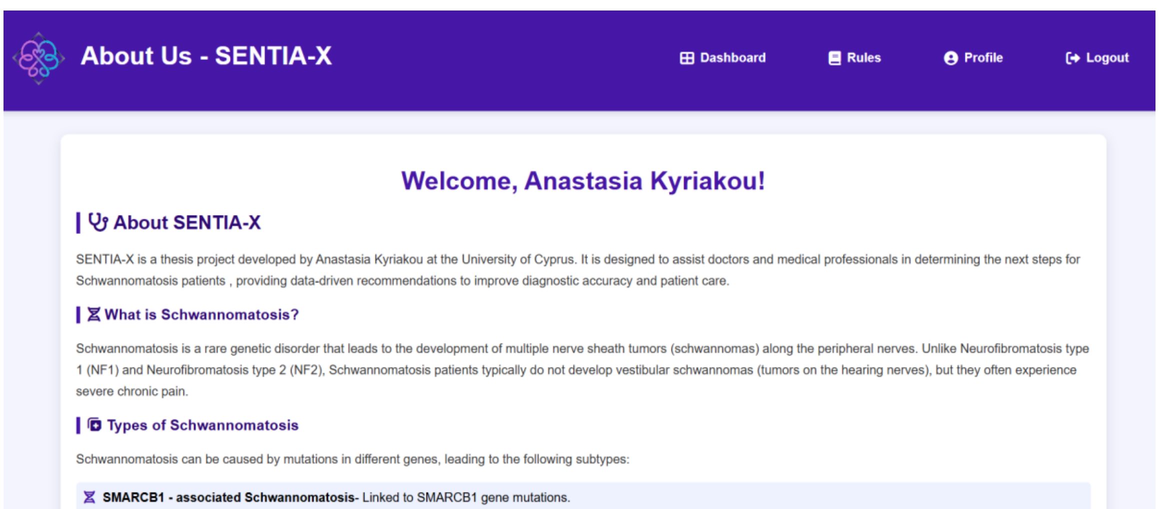
When a result is "Maybe", a modal dialog appears asking the doctor if any relevant action was forgotten, which could help the system conclude a more definitive result.

- If the doctor confirms new doctors, system re-queries Cognica.
    - Shown only per session to prevent repetition
    - If the modal is closed or no additional actions are selected, the previous result is retained, and no further queries are made.



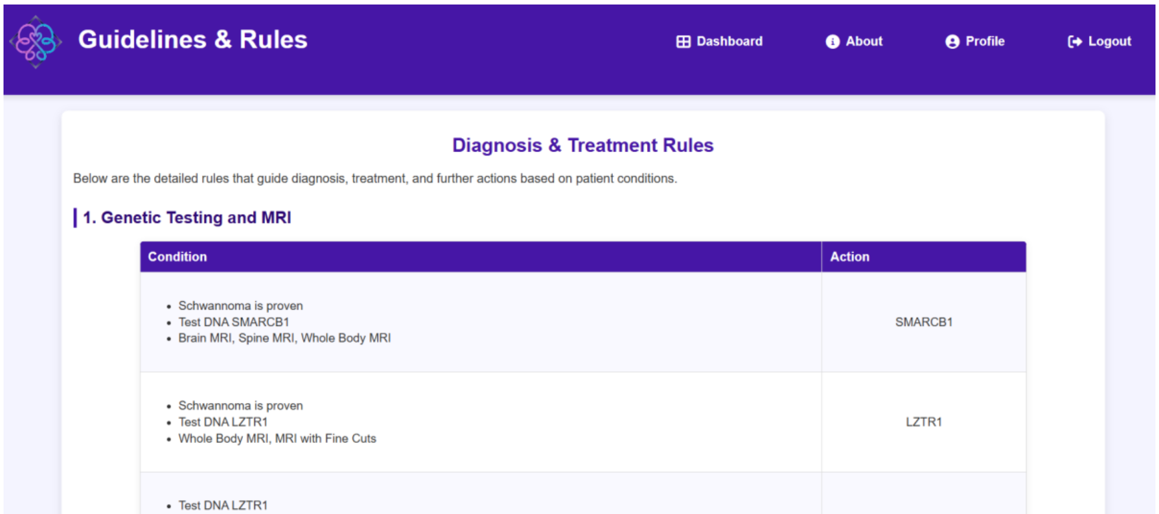
The "Indefinite" result is going to be saved in the recommendations table, including recommendations, the explanation.

- **About Page**



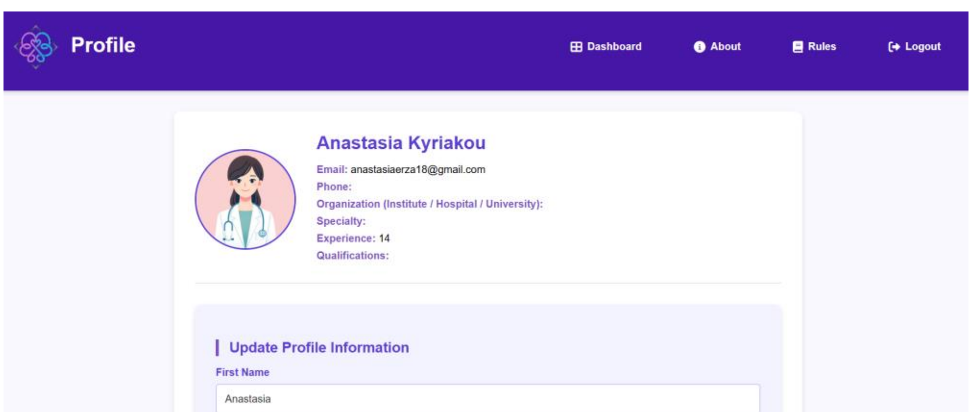
Provides the doctor with background information about Schwannomatosis, including definition, types of Schwannomatosis, common symptoms, Explainable AI, policy translation. It is designed to give quick educational context.

- **Rules Page**



Displays the complete set of clinical rules used by the system, as defined by the ERN GENTURIS guidelines. These rules are grouped by the stage of patient care, allowing doctors to review the logic behind the system's recommendations.

- **Profile Page**



The doctor can see his/her information and add anything about his/her background and experience.