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Ontology Based SWRL Rules for Diagnostic of Tumoral Bone Pathologies

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Abstract: **Bone cancer is one of the deadliest cancers in the world. It grows in the skeletal system and destroys tissue. It can spread to adjacent organs, such as the lungs, and occurs when a tumor or abnormal tissue mass forms in a bone. A tumor may be malignant, which means it's growing aggressively and spreading to other parts of the body. This article deals with the diagnostic process of bone tumors. In order to analyze a big volume of medical data, ontologies are the most efficient technique to improve medical image analysis used to detect different tumors and other bone lesions. Therefore. The main objective is to show the contribution of semantic reasoning coupled with the ontological model to detect and diagnose bone cancer disease. The essential characteristics of our approach are the diagnosis of bone tumors through SWRL inference rules. The major advantage of this work is essentially to integrate the reasoning into our Ontobone ontology modeled in a previous work in order to assist in the decision-makinort phase in terms of diagnosis, risk estimation and the proposal of appropriate treatments whatever for the treatment of tumoral bone pathologies or to prevent their risks. The evaluation of our work was based on a set of clinical cases from the medical folder of patients from the radiological service of the CHU Hedi Chaker of Sfax and which have system have correctly diagnosed 27 out of the 40 patients (ratio of correctness is approximately around 90%). These results suggest that the proposed approach could be useful for staging and processing using classification systems. Additionally, we have developed a prototype OntoBone system that demonstrates the effectiveness of our proposed approach.**

Keywords : Semantic representation, Ontology, SWRL rules, diagnosis, tumor bone.

I. INTRODUCTION

Skeletal bones can host numerous types of cancers. They commonly receive metastases from other malignancies, including breast, lung, renal, prostate, and thyroid cancers. Indeed, bone marrow can be the nidus of malignancy in multiple myeloma, lymphoma, and leukemia. In all of these illnesses, however, the malignant cells are not of bony origin. The differential diagnosis between different types of tumoral bone diseases requires the results of several clinical tests. The patient's symptoms alone are not sufficient to give an accurate diagnosis because many types of tumors bone have the same symptoms. Currently there is no specific system in the domain of tumoral diseases. Also, available medical systems do not employ semantic approaches, they are just using database-oriented methodologies and not flexible and adaptable to complex requirements and processes and lack intelligence. This work aims to improve the diagnosis of tumors bone by exploiting Semantic Web technologies. We use ontology and Semantic Web Rule Language (SWRL) to assist the automatic diagnose of tumors bone. Indeed, ontology provides powerful querying and reasoning mechanisms successfully exploited by Decision Support System (DSS) [1]. Also, we have built a domain ontology (OntoBone) that covers domain knowledge of bone diseases. The ontology contains terms, relationship and properties to be used in the approach of diagnosing tumoral bone diseases. SWRL rules are created from valid relationships between ontology concepts to detect the tumor disease and estimate their risk. The rules are used to infer new knowledge from the ontology, knowledge base and patient data. The proposed system was tested using a sample set of patients with tumoral bone diseases provided by a domain expert. Results have shown that the system have correctly diagnosed 27 out of the 30 patients (ratio of correctness is 90%).

II. Tumoral bone diseases

Bone cancers are rare malignant tumors originating in bone and derived from primitive mesenchymal cells. They 2 are frequently aggressive tumors, often occurring in childhood. They require prompt diagnosis and treatment under the care of a specialist bone cancer center. This article reviews primary bone cancer assessment and treatment, highlighting the role of the interprofessional team in the management of this patient group. Indeed, Primary bone cancer (PBC) is a rare malignant tumor of the bone, originating from primitive mesenchymal cells. It accounts for around 0.2% of all malignancies worldwide and is idiopathic in most cases. There are multiple subtypes, with osteosarcoma, chondrosarcoma, and Ewing sarcoma, the most common. Each varies in demographics, imaging appearance, and biological behavior. They are frequently aggressive and require early diagnosis, utilizing imaging and tissue biopsy. Surgical excision remains the mainstay of curative treatment, with chemotherapy and radiotherapy often used in conjunction [2] While primary bone cancer is most often idiopathic, risk factors also play a role in the development of this cancer. Genetic factors are linked. Germline abnormalities in hereditary cancer predisposition syndromes have an increased risk of later developing bone cancer, through downregulation of tumor suppressor genes or upregulation of oncogenes. The tumor suppressor gene is often altered in Li-Fraumeni syndrome, with patients at an increased risk of developing osteosarcoma. Similarly, Werner and Rothmund-Thomson syndromes are also linked to an increased risk of developing osteosarcoma [3].

Previous treatment for cancer with radiotherapy is linked to an increased risk of developing PBC in later life, particularly when exposed to ionizing radiation in childhood [4]. Several benign conditions show the potential to progress to PBC. Paget disease of the bone is a condition characterized by a disorder of bone metabolism, particularly osteoclastic function. These patients are at an increased risk of developing osteosarcoma ; however, it is a rare complication. Enchondromas and osteochondrmas are benign cartilaginous neoplasms that can late [4].

A. Tumoral bones's Risks and evolution

Staging is determined by the size and location of the tumor, and whether or not cancer has spread to other areas. Primary bone cancer is categorized into four stages :

-Stage 1 : The tumor is low-grade, and the cancer cells are still localized.

-Stage 2 : The cancer cells are still localized, but the tumor is high-grade.

-Stage 3 : The tumor is high-grade and cancer has spread to other areas within the same bone.

-Stage 4 : Cancer had spread from the bone to other areas of the body, such as the lungs or liver.

B. Tumoral bone's Treatements

Although the treatment of bone cancers varies depending on the type of modalities used, the main factor in maximizing survival and quality of life is prompt diagnosis and treatment at the time a malignant bone tumor is suspected [5]. Among the appropriate treatments for the management of tumoral bone pathologies, mention is made more particularl :

- Biopsy : A properly performed biopsy starts the process of confirming the diagnosis of bone cancer, establishes tumor grade, and directs treatment [5].
- Chemotherapy : Neoadjuvant chemotherapy regimens aim to cause tumor necrosis and decrease primary tumor size, as well as the number and size of pulmonary metastases. It has increased the feasibility of limbsalvage surgery by reducing the amount of tissue needed to achieve wide margins [5].
- Chemotherapic's medication : Adjuvant chemotherapy has been successful in decreasing postsurgical metastasis. Chemotherapy drugs with proven effectiveness against osteosarcoma include highdose methotrexate, doxorubicin (Adriamycin), and cisplatin [5].
- Surgery : Surgical excision is the definitive treatment for tumor's bone. The goal of resection is to remove primary tumors with clear margins to limit recurrence and metastasis [5].
- Radiation therapy : This treatment shrinks the tumors with high doses of X-rays. Healthcare providers often use radiation before surgery to shrink the tumor so less tissue has to be removed [5].

III. Ontologies in Biomedical domain

Towards the end of the 20th and beginning of the 21st centuries, and especially since the Semantic Web was conceived [6], the term "ontology" (or ontologies) gained usage in Computer Science to refer to a research area in the subfield of Artificial Intelligence mainly concerned with the semantics of concepts and with expressive processes in computer-based communications. In Computer Science, ontologies are a technique used to represent and share knowledge about a domain by 3 modeling the things in that domain and the relationships between those things [7]. Ontologies are represented using standard, machine-processable languages (e.g., RDF [8] and OWL [9]), and they are mainly used for communication between people and organizations by providing a unified terminology that allows to reach a common level of understanding or comprehension within a particular domain. In the biomedical field, ontologies have increasingly become an established method to represent and communicate the huge amount of knowledge about genes, diseases, biomedical processes, and so forth that has been generated during the last years [10].

Biomedical ontologies are considered crucial pieces in the development of informatics applications in several areas, such as knowledge-based decision support, terminology management, and systems interoperability and integration [11]. As a consequence, multiple biomedical ontologies have been developed and maintained, which are stored in large-scale ontology repositories available for researchers. The most popular repository of biomedical ontologies is the NCBO's BioPortal [12], a web-based, open resource that contains more than 300 ontologies with knowledge related to different biomedical topics (anatomy, gene products, immunology, phenotype, etc.) in different organisms (human, plant, mouse, microbe, etc.). In the medical domain, ontologies are key to reuse the large amount of complex information that is involved in many health care activities. They are used to build systems for purposes such as data annotation, information retrieval, and natural-language processing, but they are particularly useful to build knowledge-based systems that provide decision support in health care. This type of systems are generally dependent on large volumes of domain knowledge, which is extremely expensive and difficult to capture and formalize [13]. By means of ontologies, this knowledge can be represented in an application independent manner ; so, it can be reused in new systems without additional knowledge extraction and development effort.

III. Challenges of using OWL and SWRL Languages

The Ontology Web Language (OWL) is a W3C recommendation for an ontology description language that has gained widespread adoption and for which a considerable number of tools have been developed. Many health care processes, such as computer aided decision making or disease diagnosis and treatment, are often best modeled using a declarative approach, leading to a very active interest in rule-based systems [14]. However, interoperability among the multitude of current rule-based systems is limited. The Semantic Web Rule Language (SWRL) has emerged as a first step solution to increase rule-based systems interoperability from the Semantic Web perspective. It is based on a combination of OWL with the Rule Markup Language. The combination of OWL and SWRL provides inference capabilities beyond the classification capabilities built into the description logics [15] implemented by OWL. A SWRL extension to overcome complex scenarios that include mathematical relationships and formulas that exceed current SWRL capabilities is proposed in [16]. In the clinical environment, several kinds of rules can be expressed with this logic.

IV. Semantic Web Rule Language (SWRL)

Semantic Web Rule Language (SWRL) was developed to be the rule language of the semantic web. SWRL allows users to write rules that can be expressed in terms of OWL concepts and that can reason about OWL individuals. One of SWRL's most powerful features is its ability to support custom built-ins, user defined, to extend SWARL's core built-ins so the user can achieve extra extensibility [17] There are several atom types that are supported by SWRL, such as class atoms, individual property atoms, data value property atoms, and data range atoms. The most powerful atoms are built-in atoms, where SWRL provides several types of existing built-ins and allow the user to design and use his own built-ins [18]. SWRL rules are created from valid relationships between ontology concepts to detect and estimate the risk of tumoral bone disease. The rules are used to infer new knowledge from existing ontology knowledge and user input. All rules will be expressed in terms of ontology concepts (classes, properties, individuals). The writing of the semantic rules starts with the concept in which the property belongs, and then chains the concept to other facts in a step-by-step manner until the objective is achieved. OWL 2 language is not able to express all 4 relations, for example, there is no way in OWL 2 to express the relation between individuals with which an individual has relations, the expressivity of OWL 2 can be extended by adding SWRL rules to our ontology

V. Semantic Reasoner

A semantic reasoner, a reasoning engine, a rules engine, or simply a reasoner is a piece of software able to infer logical consequences from a set of asserted facts/axioms. The capabilities of a reasoner depend on the axioms and inference rules that it knows about, which are related to a particular kind of logic. Reasoner is a key component for working with OWL ontologies. All querying of an OWL ontology should be done using a reasoner. This is because knowledge in an ontology might not be explicit and a reasoner is required to deduce implicit knowledge so that the correct query results are obtained. OWL reasoner such as Pellet, FaCT++ and HerMiT would be required for executing SWRL rules and infer new ontology axioms. Pellet has more direct functionality for working with OWL and SWRL rules, it allows to define custom SWRL built-ins. When applying Pellet to reason over ontology with SWRL rules, it takes these rules into consideration, and returns conclusions based on them.

VI. State of the art of on reasoning works for diagnosis of variours diseases

This section presents several related works that use semantic web techniques in similar situations and domains. They focus on various diseases diagnosis using ontology and SWRL rules, ontology and clinical decision support system. There are various works which have explored diseases and related diagnosis systems using different approaches. However, this section focuses only on the approaches that use ontology and SWRL rules. In this context, the authors in [19] have designed system for early diagnosis and treatment of critical diseases which include Heart attack, Stroke, Cancer, Kidney failure and Brain tumour. It consists of knowledge Extraction anddiagnosis & disease prediction modules. In Knowledge extraction module, medical data from various sources is gathered. Based on this knowledge base, a medical Ontology model with Bayesian networks is developed which provides the diagnosis of whether the patient has the potential to have that critical disease. Indeed, they have applied the suggested SWCH by relating web ontology language (OWL) and Semantic Web Rule Language (SWRL) centred on diverse classes associated with numeroussicknesses to acquire a knowledge-based demonstration. In the same way, the authors in [20] have implemented the semantic rule based expert system for diagnosing a kind of blood immune thrombocytopenia disease. They presented an ontology to depict the knowledge domain of this disease, symptoms and its related treatments, to do this, some semantic rules were defined to do the diagnosis process and finally, the diagnosis process is validated by blood specialists. In addition, the authors of [21] have proposed an ontology-Based Framework for Healthcare using the existing ontologies, and also proposes Healthcare Ontology which is a semantic representation of knowledgebase of patients' healthcare information available in the form of Electronic Health Record (EHR). The ontology consisting of systematically generated and exhaustive ontologies may be utilized for predicting semantic inferences related to a patient's medical condition by developing SWRL rules. In this context, authors of [22] have proposed a semantic rule-based modelling and reasoning approach directed towards formalising dengue disease definition in conjunction with operational definitions (semantics) that support clinical and diagnostic reasoning. The operational definitions are incorporated using Semantic Web Rule Language (SWRL) as logical rules that enhance the expressive capability of the knowledge base. A dengue knowledge base has been designed which is extended with International Classification of Diseases (ICD) ontology for associating dengue fever with ICD code. The knowledge base created can be reasoned upon for diagnostic classification that can discover dengue symptoms and predict the possibility of patients to suffer from the disease apart from offering interoperability. 153 real patient cases are classified successfully against the operational definitions incorporated by SWRL rules. In the same context, the authors of [22] presents an expert system to diagnose coronary artery diseases. The design of the system depends on ontology knowledge about the patient's symptoms to build the knowledge base. SWRL rules are used to deduce the suitable medication and the required operation for the patient. The architecture of this system consists of several modules : knowledge base module which consists of the fact base and the rule base, the inference engine module and explanation module. The facts are extracted using the user interface, from the user as the patient's symptoms. The inference engine depends on the facts and the rules to reason the required decision. The final decision results will be introduced to the user through the user interface alongside with the explanation about this decision inferred from the explanation module. This system was tested by several general practitioners using 16 instances to test the validation and the evaluation of the system. This work focuses on diagnosing coronary artery diseases which are considered as part of heart 5 diseases. The diagnosis in this system are based only on the patient's symptoms, without taking into account the diagnosis tests results (i.e. ECG, X-ray, CT scan). The work of [24] presents ontology based expert system for diagnosis of thyroid disease. This system uses ontology to construct the domain knowledge and rules to infer the thyroid disease related diagnosis. It consists of ontology, reasoner, rule base and MySQL Database. Based on the symptoms entered through the user interface, data and SWRL rules get generated for the users. Then, a reasoner called Jena uses these data and rules to infer and make the diagnosis accordingly. This system was implemented based on neural networks and ontology. Total number of instances taken is 60. Neural network considers only TSH, T3, T4 levels in the blood to train the neural network whereas there is no training for the ontology based expert system. The results show that expert system based on ontology gives more accurate results with lesser complexity than the one which uses neural network. In the same context, the authors of [25] present a diagnosis and treatment recommendation system for diabetes. The implemented system relies on two components : a domain ontology which has developed to standardize the domain expertise and a knowledge base system which is includes the necessary rules dedicated to diagnose and propose treatment for the disease. The domain ontology is designed and developed by OWLDL, the rules are constructed by SWRL and executed by JESS inference engine. The system contains four modules : the graphical user interface, the inference engine, the knowledge base and the ontologies. The user interacts through the graphical user interface to test the system or to request any diagnosis for a specific case. The inference engine is the reasoning component which uses the ontologies and the rules in the knowledge base to infer a diagnosis for the specific case. In this context, the authors of [26] develop an ontology based approach to create disease information system. This system consists of three different components such as knowledge base component, rules component and query processing component. The knowledge base consists of the ontology to model diseases and their relationships with symptoms. Rules component contains the semantic reasoner and SWRL Rules. The query processor checks with SWRL rules for relations between the diseases. It returns the diseases associated with the given symptoms. The query processor then returns the output to the user. The predicted human diseases are done with executing rules which extract disease details with symptoms based on the rules specified, then the inferred axioms are reflected in the ontology. This approach is similar to our research in terms of building ontology and using SWRL rules to predict diseases using a knowledge base. But it is different in terms of the field of diseases and diagnosis procedures. In the propostion of [27], the authors propose general knowledge base ontology framework for patient diagnosis based on clinical practice guidelines (CPG) to help the physicians and medical staff to make the right diagnosis decision for the patient situation. This framework is a general base, which can be used with more specialization for quickly modeling a specific clinical practice guideline. The methodology content on four steps. The first is to choose an appropriate clinical practice guidelines resource as the base of this research. The second step and as the domain of the research, 30 different diseases has been chosen from different human organs and have been visualized in tables based on their symptoms, signs, and diagnosis procedures. The third step is capturing and modeling the common symptoms and signs among these 30 different diseases and with the help of the differential diagnosis that will go out at the end, the patient will be successfully diagnosed. The fourth and the last step in this methodology is the transformation of these models into a knowledge base ontology framework for patient diagnosis based on clinical practice guidelines by using Protégé. This approach can be considered as a general knowledge base for various diseases, it lacks further information about the characteristics of each disease in terms of laboratory tests and risk factors, particularly that shares the symptoms and diagnostic methods. In the same context, the authors of [28] propose an ontology based diagnostic method for cancer diseases with knowledge management. The proposed method contains three basic modules namely ; the diagnostic module, the staging module and the treatment recommendation module. All the three modules interact with a database of cancer ontologies through the query module, which maps from the query of the asking module to the structure of the vocabulary of the ontologies stored in the database of cancer ontologies. The database of cancer ontologies describes the different types of cancer diseases in detail. Each cancer ontology describes the cancer in terms of its structure, signs and symptoms, staging and treatment. This method can be applied to help patients, medical students and doctors to decide what cancer type the patient has, what is the stage of the cancer and how it can be treated. The system is used to help doctors to decide what cancer type the patient has. In another medical context, several researches were focused on the Clinical Decision Support Systems (CDSS) which is an application that analyze data to help healthcare providers make clinical decisions. These types of systems require computable medical knowledge, person-specific data, and a reasoning or inferencing mechanism that combines knowledge and data to generate and present helpful information to clinicians [29]. This section 6 presents several ontology-based clinical decision support systems. In parallel, the authors of [30] present an intelligent system to predict the risk of hypertension in three main related areas ; diabetes, cardiovascular problems, and kidney disorders. The system uses ontologies with knowledge base (medical knowledge base), patient medical profile stored in a semantic way and an inference mechanism to extract data in the decision-making process. Predicting the risk of hypertension is performed through three phases. In the initial phase, the user fills adaptive questionnaire. In the second phase, a semantic profile of the patient is generated automatically by the system based on the input (answers) of the questionnaire. The patient profile generated is semantic in nature and is represented in OWL. In the final phase, this semantic patient profile is analyzed by ontology reasoner with the help of clinical guidelines ontology. The output of the reasoner and the rule engine together generates a risk assessment report of hypertension in three main related cases of diabetes, cardiovascular problems, and kidney disorders. In the same context, the authors of [31] presents a clinical decision support system (CDSS) for undergoing surgery based on domain ontology and rules reasoning in the setting of hospitalized diabetic patients. The ontology was created with a modified ontology development method, including specification and conceptualization, formalization, implementation, evaluation and maintenance. Embedded clinical knowledge was elicited to complement the domain ontology with formal concept analysis. The decision rules were translated into JENA format which JENA used to infer recommendations based on patient clinical situations. The evaluation confirms the correctness of the ontology, acceptance of recommendations, satisfaction with the system, and usefulness of the ontology for blood sugar management of diabetic patients undergoing surgery, especially for domain experts. This work is similar to our system in terms of using decision rules. Indeed, this system uses JENA semantic rules to infer recommendations based on patient clinical situations. Our proposed method use SWRL rules to infer correct diagnosis and recommend appropriate treatment. Another work [32] propose an ontology-based decision support system designed to supervise and treat patients affected by acute cardiac disorders. The architecture of the system contains cardiac intensive care units (CICU) devices to connected to a monitor, communication APIs to enable the system's interaction with the CICU devices, Expert System which has 4 essential components knowledge base, fact base, inference engine, and explanation facilities, graphical user interface to enables the communication between the doctor and the expert system and database. The system analyzes the patient's condition and provides a recommendation about the treatment that should be administered to achieve the fastest possible recovery. The knowledge base is consisting of an OWL ontology and a set of SWRL rules that represent the expert's knowledge. This approach provides supervision and treatment of critical patients with acute cardiac disorders. In addition, the author of [33] proposes an ontology based system to collect the patient history to assess the patient risk in diabetes due to smoking history, alcohol history, and cardiovascular history. According to the patient history, a total score is calculated for each of the above factors. Based on the score, the ontology performs the risk assessment on a patient profile and predicts the potential risks and complications of the patient. The system instantiates the questionnaire ontology and stores the corresponding answers in it. The system processes this information and automatically generates a patient ontology instance in the server. Patient medical is an OWL file which encapsulates patient details as entered by the patient, nurse and other users in a web/mobile application. The clinical guidelines are hard coded in Java and the values generated are written back to the ontology. This work is similar to our system in terms of procedures for estimating risk of diabetes. Our system estimates the heart risk according to five factors which are : Age, total cholesterol, HDL, systolic blood pressure and smoking habit. All clinical decision support systems that have been mentioned in this section focused on the diagnosis of different types of diseases such as diabetes, cardiac disorders, breast cancer and hypertension. These works are very helpful in the development of our proposed ontology. We focus in this research on tumoral bone diseases diagnosis, risk estimation and appropriated treatments.

VII. PROPOSED METHOD

In this work, we have based an our modelled OntoBone ontology [34] which modelise the musculoskeletal system and refletes the medical knowledge used by radiologists and doctors to diagnose and treat patients suffring from bone pathologies and to provide decision support about the treatment that should be administered.

A. OntoBone Ontology

At the level of this work we are based on our ontology OntoBone which has already been modeled during a previous work [34]. Indeed, the OntoBone ontology models in a complete way the musculoskeletal system of 7 the human body, namely the composition of the bone system using a model that respects the structuring of the bones as well as their composition (Fig 1)

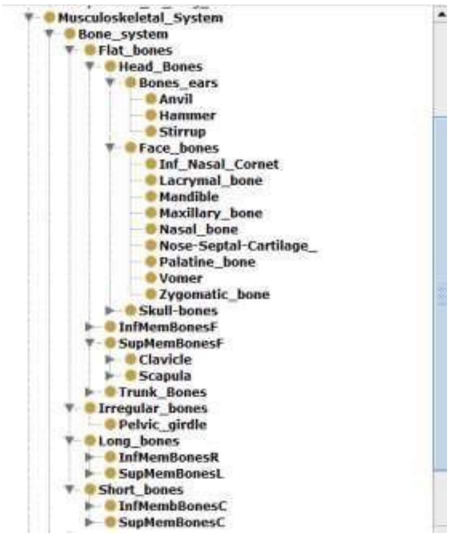

Figure 1. Modeled concepts of the OntoBone Ontology [34]

As an indication, the OntoBone ontology has not only semantically presented the bone system but also the different semantic relationships that exist between the modeled concepts and which implement each of the structuring andarticulation relationships between the bones of the bone system. (Fig2)

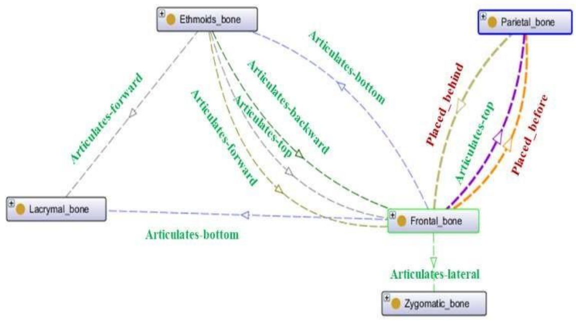

Figure 2. Modeled semantic relations between the concepts on the OntoBone Ontology [34]

In this work, our major contribution revolves around the development of SWRL inference rules for the diagnosis, risk estimation and treatment of tumoral bone pathologies. At this level, we will detail the development process of the SWRL rules that we followed while basing ourselves on medical knowledge in terms of diagnosis of bone tumors within the radiological center of the Hedi Chaker regional hospital in Sfax, Tunisia.

B. SWRL Rules Implementation

The step of the implementation of the SWRL rules consists of calculating or deriving new facts from existing knowledge bases. This is because a rules-based inference engine applies rules with data to reason and derive new facts. When the data matches the rule conditions, the inference engine can change the knowledge base, for example, assert or retract facts, or to perform functions, such as display derived facts. As an indication, these rules will be formalized and represented by the formalization language SWRL (Semantic Web Rule Language) [35] which is the language most commonly used in the semantic web to express the rules of reasoning [36]. By convention, these rules give an additional level of expressiveness which cannot be offered by the OWL language, they improve the ontological language by allowing to describe relations which cannot be described using the description logic used in OWL [37].

SWRL rules were written using the Protégé SWRLTab, which is tightly integrated with OWL, and facilitates the creation of rules that can be expressed using OWL ontology classes to improve reasoning skills [38], [39], incorporating the Pellet rules engine [40] to execute SWRL rules and infer new knowledge about ontology. Indeed, the step of reasoning the ontology through SWRL rules is efficient to allow the bone tumors diagnosis process. The inference rules formalized in our work fall into two main categories. One concerns generic diagnostic rules and detection of bone tumor pathology, in other words, which corresponds to a first level of diagnosis. While the second category, supports more specific and thorough rules in terms of diagnosis, risk and treatments.

C. SWRL Reasonning Process

In order to improve the performance of our inference rule formalization process, we have proposed the implementation of a routing process for inference rules formalization entitled DiagRT (Diagnosis, Risk, Treatement) at the level of the reasoning process which 8 describes the step of detection and diagnosis of bone tumors followed through SWRL rules (Fig 3).

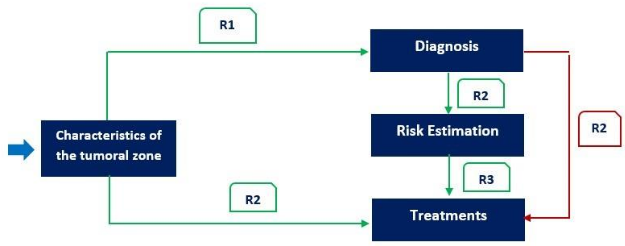

Figure 3. Proposed SWRL Reasonning Process

In such process, the definition of inference rules follows the sequence illustrated by (fig 3). Indeed, the formalization of these rules takes as inputs initially the characteristics of the tumor detected, the symptoms and the information relating to the patient's medical file. This information causes the automatic diagnosis of the bone tumor, namely its type and nature, etc. Consequently, this diagnosis leads in most cases to associated risks which the patient may have. Furthermore, each of these risks and the diagnosis made require treatment to treat and/or prevent them.

Indeed, starting from the diagnosis of the tumoral pathology, we define in some cases directly the rules of inference relating either to the risks if they exist and then to the appropriate treatments. In the same way for the cases of the risks we develop in certain case the rules SWRL corresponding to the treatment necessary to prevent them.

D. Steps for creating SWRL rules via Protégé 2000

The creation of SWRL inference rules follows a certain set of steps at the level of the Protégé2000 ontology editor. After having modeled the concepts, the semantic relations between these different concepts, the data properties and finally starting the instantiation phase of the ontology, the implementation phase of the SWRL rules is put in place. All of the SWRL rules created will then be executed via the inference engine. The (Fig 4) illustrates all the SWRL rules relating to the diagnosis of bone tumor pathologies via the dedicated interface on the Protégé2000 editor.

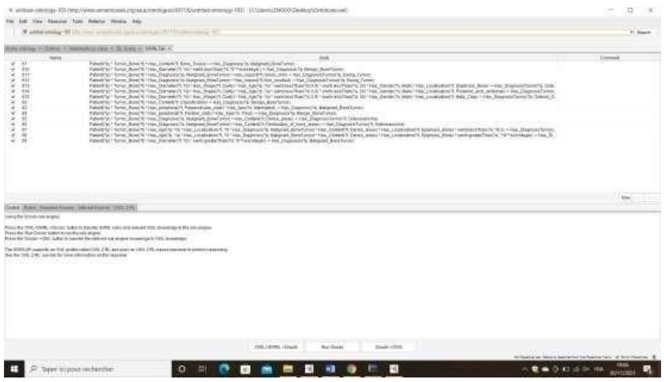

Figure 4. Example of created SWRL Rules via Protege2000 Editor

In what follows, we will present some examples of SWRL inference rules developed relating to the three categories of reasoning, namely, the detection and diagnosis of the tumor, the estimation of the related risks and the proposal of appropriate treatments.

- *Rules for Diagnosis tumoral bone Diseases*

At the level of our development process of SWRL inference rules, we have chosen the approach used in the process of medical diagnosis. Indeed, the first category of developed SWRL rules indeed consists in the detection of the tumor and its diagnosis, namely, its nature, its type, etc., while being based on the adequate and necessary information for the diagnosis. As an indication, this information has two different origins which are both medical imaging from which we will extract the characteristics of the tumor zone, symptoms and the medical file relating to each patient.

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diameter(?t, ?d) ^ swrlb:greaterThan(?d, "6"^^xsd:integer) -> Has_Diagnosis(?p, Malignant_BoneTumor

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diameter(?t, ?d) ^ swrlb:lessThan(?d, "6"^^xsd:integer) -> Has_Diagnosis(?p, Malignant_BoneTumor Patient(?p) ^ Tumor_Bone(?t) ^ Has_Content(?t, Bone_Tissue) -> Has_Diagnosis(?p, Malignant_BoneTumor)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Content(?t, Classification) -> Has_Diagnosis(?p, Benign_BoneTumor)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_peripheral(?t, Perpendicular_slats) ^ Has_type(?s,Interrupted) -> Has_Diagnosis(?p, Malignant_BoneTumor)

- Patient(?p) ^ Tumor_Bone(?t) ^ Slats(?s) ^ Has_Peripheral(?t, Parallel_Slats) ^ Has_Type(?s, Thick) -> Has_Diagnosis(?p, Benign_BoneTumor)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_Content(?t,Dense_areas) -> Has_DiagnosisTumor(?p,Osteosacroma)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_Content(?t, Destruction_of_bone_areas) -> Has_DiagnosisTumor(?p,Osteosacroma)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Age(?p,?a) ^ Has_Localisation(?t,?l) ^ Has_Diagnosis(?p,Malignant_BoneTumor) ^ Has_Content(?t,Dense_areas) ^ Has_Localisation(?t,Epiphysis_Bone) ^ swrlb :lessThan(?a,18) -> Has_DiagnosisTumor(?p,Chondroblastoma)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Age(?p,?a) ^ Has_Localisation(?t,?l) ^ Has_Diagnosis(?p,Malignant_BoneTumor) ^ Has_Content(?t,Dense_areas) ^ Has_Localisation(?t,Epiphysis_Bone) ^ swrlb :greaterThan(?a,18) -> Has_DiagnosisTumor(?p,Giant_cell_tumor)

- Patient (?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p,Malignant_Tumor) ^ Has_Aspect(?t,Onion_skin) -> Has_DiagnosisTumor(?p,Ewing_Tumor)

- Patient (?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p,Malignant_Tumor) ^ Has_Aspect(?t,Not_ossified) -> Has_DiagnosisTumor(?p,Ewing_Tumor)

- Patient(?p) ^ Tumor(?t) ^ Has_Diameter(?t,?d) ^ Has_shape(?t,Cavity) ^ Has_Age(?p,?a) ^ Gender(?s) ^ swrlb :lessThan(?d, 0.9) ^ swrlb :lessThan(?a, 30) ^ Has_ Gender(?s, Male) ^ Has_Localisation (?l, Diaphysis_bone) -> Has_DiagnosisTumor(?p,Osteoid_Osteoma)

- Patient(?p) ^ TumorBone(?t) ^ Has_Diameter(?t, ?d) ^Has_Shape(?t,Cavity) ^ Has_Age(?p, ?a) ^ Gender (?s) ^ Localisation (?l) ^ swrlb:lessThan(?d, 0.9) ^ swrlb:lessThan(?a, 30) ^ Has_Gender(?s, Male) ^ Has_Localisation (?l, Posterior_arch_vertebrae) ^ Has_DiagnosisTumor(?p,Osteoid_Osteoma)

- Patient(?p) ^ TumorBone(?t) ^ Has_Diameter(?t,?d) ^Has_Shape(?t,Cavity) ^Has_Age(?p,?a) ^ Gender (?s) ^ Localisation (?l) ^ swrlb:lessThan(?d, 0.9) ^ swrlb:lessThan(?a, 30) ^ Has_ Gender(?s, Male) ^ Has_Localisation (?l, Meta_Carp) -> Has_DiagnosisTumor(?p,Osteoid_Osteoma)

After having developed the SWRL rules relating to the detection and diagnosis of tumoral bone pathologies, we move on at this level to the development of the SWRL rules concerning to the estimation of risks.

- *Rules for tumoral bone Diseases Risk Estimation*

After the phase of detection and diagnosis of the tumoral bone pathology, one can be faced with risks linked to this pathology, that is to say that this tumoral pathology can over time, whatever in the short or long term, advanced and lead to other tumor problems. On this, we have developed a selection of SWRL rules to deduce the estimation of the risks related to the different bone tumors.

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_RiskEstimation (?p, Lung_Metastases)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_RiskEstimation (?p, Bone_Metastases)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_RiskEstimation (?p, Bone_Infractus)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p, Ewing_Tumor) -> Has_RiskEstimation (?p, Progression_tumor_mass)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p, Ewing_Tumor) -> Has_RiskEstimation (?p, Tumor_spread_to_all_of_the_bone)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p, Chondroma) -> Has_RiskEstimation (?p, Cranial_nerve_problems)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p, Chondrosarcoma) -> Has_RiskEstimation (?p,Various_Metastases)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p, Chondroma) -> Has_RiskEstimation (?p, Cranial_nerve_problems)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p, Chondroma) -> Has_RiskEstimation (?p, Spread_of_tumoral_celles)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p, Bone_lipoma) -> Has_RiskEstimation (?p, Fracture_of_infectedBone)

After having developed the SWRL rules relating to the risk's estimation of tumoral bone pathologies, we move on at this level to the development of the SWRL rules concerning to the differents appropriated treatments.

- *Rules for tumoral bone Diseases treatment*

As in the process followed by doctors, after diagnosing and estimating the risks associated with the detected tumor bone pathology, we have put in place a set of SWRL inference rules relating to the proposal of appropriate treatments for each pathology regardless of for a treatment reason or for a risk prevention reason.

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_Treatment(?p, Chimiotherapy)

- Patient(?p) ^ Tumor_Bone(?t) ^ Chemotherapy (?c) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) ^ Has_Treatment(?p, Chemotherapy) - >Chemotherapy_combinedWith(?c, High_dose_methotrexate_and_cisplatin_and_doxorubici n)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_Treatment(?p, Surgical_intervention_after_10weeks)

-Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p,Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Multiple_Melyoma) -> Has_Treatment(?p, Biopsy)

- Patient(?p) ^ Tumor_Bone(?t) ^ Chemotherapy (?c) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p, Multiple_Melyoma) -> Has_Treatment(?p,BoneAssesment_and_BloodAnalyse)

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Chondroma) -> Has_Treatment(?p, Radiotherapy)

-Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p,Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Ewing_Tumor) -> Has_Treatment(?p, Chemotherapy

- Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor ^ Has_DiagnosisTumor(?p,Chondrosarcoma) -> Has_Treatment(?p, Surgical_removal_tumor)

- Patient(?p) ^ Tumor_Bone(?t) ^ Chemotherapy (?c) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Ewing_Tumor) ^ Has_Treatment(?p, Chemotherapy) –> Chemotherapy_combinedWith (?c,Vincristine_doxorubicin_cyclophosphamide_etoposide_a nd_ifosfamide)

After having formalized the various rules of inference that describe medical knowledge in terms of diagnosis and analysis of bone tumors, a validation phase remains fundamental at this level.

E. Validation of SWRL rules

At the second level, the validation phase concerns the inference rules formalized in the previous step. This step consists of verifying the accuracy and regularity of the medical knowledge represented through the rules of inference with health professionals. In other words, it therefore aims to ensure the compliance of diagnoses and medical decisions taken automatically through these rules with those taken directly by the health specialist. Where 11 appropriate, we try to refine our formalized inference rules as much as possible so that they respond better to medical knowledge in terms of diagnosis and decision-making support by bringing them closer to the decision-making reasoning by radiologists. In what follows, we have opted for the validation of the risks that can be derived from the diagnoses of bone tumors as well as their degrees of severity. In other words, checking whether the preventive decision taken in view of these risks is well carried out or not. Finally, we have started this approval phase by verifying and validating the inferred treatments assigned to the patients.

As a result, we have completed the validation phase of the formalized inference rules with the specialist radiologist. At this level we have a base of validated rules modeling medical knowledge in terms of diagnosis, risk detection and proposal of appropriate treatments. Indeed, the effective use of our domain ontology for purposes of reasoning presupposes that it be added an operational semantics which states the process with which the medical knowledge modeled in our ontology will be used by means of reasoning and generating new knowledge.

The validation of our formalized inference rules was ensured by a health professional, namely Doctor Yosr Hentati, a radiologist in the CHU regional hospital in Sfax. Indeed, this validation phase was carried out during several consultations with the doctor, based on different levels of certification of formalized medical knowledge and which has validated the modeling medical knowledge in terms of detection and analysis of bone tumors, but which in no case entirely replaces health professionals, namely doctors and radiologists.

F. Clinical case study

A 15-year-old boy presented to the emergency department for consultation following acute pain with a functional limitation in the left shoulder. As an indication, no antecedent is noted. After an X-ray examination, the medical image showed a malignant tumor of the Osteosarcoma type. This diagnosis was based on the presence of a tumor with a diameter of 7cm, tumoral consolidation, a lamellar and interrupted periosteal reaction with an extension to the soft tissues. By definition, Osteosarcoma, also called osteogenic sarcoma, is a kind of bone cancer. It happens when the cells that grow new bone form a cancerous tumor. Chemotherapy treatment and surgery to take out the tumor is usually successful when the disease is diagnosed early, before it can spread. Anyone can have osteosarcoma, but it's the most common kind of bone cancer in children and teens. Teenage boys are most likely to get it.

In children and teens, osteosarcoma often happens at the ends of long bones, where bone grows fastest. Indeed, most tumors start around the knee, in either the lower part of the thighbone or the upper part of the shinbone. They also may grow in the upper arm bone close to the shoulder. But osteosarcoma can happen in any bone, especially in older adults, including the Pelvis, shoulder and skull. Indeed, the estimated risks that are linked to this clinical case are the possibility of the generation of pulmonary and bone metastasis and also the possibility of bone infractus. The appropriate treatments to treat osteosarcoma in this case start with chemotherapy combined with high dose methotrexate, cisplatin and doxorubicin, and surgery after 10 weeks of chemotherapy to remove the tumour. Figure 5 below presents the X-ray showing the Osteosarcoma at the level of the left shoulder relating to this clinical case.

Figure 5. X-ray of the knee showing osteosarcoma in the thigh bone

The SWRL inference rules relating to the diagnosis of these clinical cases detecting the presence of the tmours, the estimation of the related risks and the proposal of the appropriate treatments are as follows :

- *Diagnosis SWRL Rules*

Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diameter(?t, ?d) ^ swrlb:greaterThan(?d, "6"^^xsd:integer) Has_Diagnosis(?p, Malignant_BoneTumor)

Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_Content(?t,Tumoral_Consolidation) ^ 12 Has_Periosteal_Reaction(?t,Interrupted_Lamaellar) ^ Has(?t,Soft_Tissue_Extension) -> Has_DiagnosisTumor(?p,Osteosacroma)

- *Risk's estimation SWRL Rules*

Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_RiskEstimation (?p, Lung_Metastases)

Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_RiskEstimation (?p, Bone_Metastases)

Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_RiskEstimation (?p, Bone_Infractus)

- *Appropriated Treatments SWRL Rules*

Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) -> Has_Treatment(?p, Chimiotherapy)

Patient(?p) ^ Tumor_Bone(?t) ^ Chemotherapy (?c) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor(?p,Osteosacroma) ^ Has_Treatment(?p, Chemotherapy) – >Chemotherapy_combinedWith (?c, High_dose_methotrexate_and_cisplatin_and_doxorubici n)

Patient(?p) ^ Tumor_Bone(?t) ^ Has_Diagnosis(?p, Malignant_BoneTumor) ^ Has_DiagnosisTumor (?p,Osteosacroma) -> Has_Treatment(?p, Surgical_intervention_after_10weeks)

IX. Conclusion

Medicine has been always a privileged field of application in order to produce decision support systems in term of medical diagnosis. To this purpose, research in the field of biomedical informatics has been recently developed. In this article, we present a reasoning approach for tumoral bone pathologies diagnosis, risk's estimation and appropriated treatments. The proposed method is based on the OntoBone ontology which describes the musculuskeletal human system and which is readily used in practice. The main objective is to show the contribution of semantic reasoning coupled with the ontological model to detect and diagnose bone cancer disease. The essential characteristics of our approach are the diagnosis of bone tumors through SWRL inference rules. The major advantage of this work is essentially to integrate the reasoning into our Ontobone ontology modeled in a previous work in order to assist in the decision-making support phase in terms of diagnosis, risk estimation and the proposal of appropriate treatments whatever for the treatment of tumoral bone pathologies or to prevent their risks. The evaluation of our work was based on a set of clinical cases from the medical records of patients from the radiological department of the CHU Hedi Chaker of Sfax and which have system have correctly diagnosed 37 out of the 40 patients (ratio of correctness is approximately around 90%). These results suggest that the proposed approach could be useful for staging and processing using classification systems. Additionally, we have developed a prototype OntoBone system that demonstrates the effectiveness of our proposed approach. Several other questions remain to be resolved in our future works. We aim to treat other medical image modalities like CT scans and create an expanded knowledge base with a dynamic ontology. Furthermore, we should include other staging systems and the vascular profile of the MRI sequence which should be automatically calculated.

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