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Development of a Domain Ontology for Decision Support in the Treatment of Gait-related Diseases

by

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DECLARATION

I, Terrance Marthinus, declare that the contents of this thesis represent my own unaided work and that the thesis/dissertation has not previously been submitted for academic examination towards any qualification. Furthermore, it represents my own opinions and not necessarily those of the Cape Peninsula University of Technology.

Signed **Date:** 19 January 2024

ABSTRACT

The growing complexity in diagnosing and treating gait-related diseases necessitates the existence of a domain ontology that can facilitate intelligent decision support on gait analysis. The study aimed to develop a domain ontology that can support decision-making on the treatment of gait-related diseases. The motivation for this study is deeply rooted in the multifaceted and interrelated challenges presented by gait-related diseases in both the diagnosis and treatment contexts. A coherent and accessible knowledge base stands as a tool to facilitate informed decision-making. This will ensure that gait experts are adeptly navigated through the complexities of numerous gait disorders, enhancing their ability to make precise and consistent clinical decisions. The domain ontology aimed to bridge existing knowledge gaps, streamline the retrieval and application of critical information, and ultimately, enhance the precision and consistency of clinical decisions by providing a unified and comprehensive view of the domain.

The study adopted the Ontology Development 101 methodology to ensure a systematic, structured, and replicable approach to ontology creation. The Ontology 101 methodology guided the research through pivotal stages, including identifying the domain and scope, enumerating crucial terms in the domain, defining classes and properties, establishing class hierarchies, and ensuring detailed documentation. The Protégé ontology editor, a standard open-source ontology development tool, was used to create the gait analysis domain ontology (GADO). The developed domain ontology was evaluated using a combination of ontology verification and ontology validation procedures.

For ontology verification, the study employed the Framework for Ontology Conformance Analysis (FOCA) evaluation methodology, analysed domain task fit using competency questions, and assessed content richness. The ontology verification underscored the GADO's proficiency in domain task fit and content richness, substantiating its potential as a viable tool for supporting clinical decisions in the domain of gait-related diseases. The validation of the GADO was done by utilising ontology reasoners, HermiT, and Pellet to determine structural and logical consistency of its components and its correctness. Description Logic (DL) queries and SPARQL queries were also used to assess the ontology’s aptitude in representing domain knowledge accurately and its ability to answer domain-specific queries. This was accomplished by trying to address some of the competency questions using SPARQL queries to evaluate the GADO's capacity to fetch pertinent ontological instances (individuals). The results from the ontology validation process indicate that the GADO effectively supports the retrieval of domain-specific knowledge.

This ontology, therefore, stands poised to significantly impact the field by enhancing decision-making related to gait-related diseases and paving the way for future advancements in an AI-driven context that can facilitate clinical decision support.

Keywords: artificial intelligence, domain, gait analysis, gait-related diseases, knowledge base, ontology development

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DEDICATION

This thesis is devoted to the memory of my dearly departed loved ones; William Marthinus, Gafsa Ely, and Fatima Heugh who have journeyed on. This also serves as a tribute to those loved ones who continue to walk this path with me; Georgina Marthinus and Russel Marthinus.

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LIST OF ABBREVIATIONS

Abbreviation/Acronym	Description
AI	Artificial Intelligence
AMOD	Agile Methodology for Ontology Development
CQ	Competency Questions
DDS	Decision Support Systems
DL	Description Logics
FOCA	Framework for Ontology Conformance Analysis
FOL	First-order logic
GQM	Goal/Question/Metric
HPO	Human Phenotype Ontology
KIF	Knowledge Interchange Format
KR	Knowledge Representation
MMS	Magnetic Measurement Systems
MCC	Mobile Cloud Computing
MeSH	Medical Subject Headings
MOE	Modular Ontology Engineering
NCBI	National Center for Biotechnology Information
NLM	National Library of Medicine
NLP	Natural Language Processing

OBI	Ontology of Biomedical Investigations
OD-101	Ontology Development -101
OLS	Ontology Lookup Service
OMIM®	Online Mendelian Inheritance in Man
OML	Ontology Markup Language
OSMMI	Ontologie du Systeme Musculo-squelettique des Membres Inferieurs
OWL	Web Ontology Language
RDF	Resource Description Framework
RDFS	Resource Description Framework Schema
RIF	Rules Interchange Format
SPARQL	SPARQL Protocol and RDF Query Language
SQL	Structured Query Language
TOVE	Toronto Virtual Enterprise
W3C	World Wide Web Consortium
XML	eXtensible Markup Language

CHAPTER 1: INTRODUCTION

1.1 Introduction

The rapid surge and growing adoption of innovative technologies, particularly in the biomedical and digital realms, have opened new doors to help and assist humans with a range of diseases and associated treatments. The principal intent of this study is to focus on gait analysis and develop a suitable domain ontology that can be leveraged by decision support systems (DSS) to effectively treat gait-related diseases. Given the intricate nature of gait disorders in both diagnosis and treatment, a structured knowledge base is critical for facilitating informed clinical decisions, enabling domain experts to navigate the complexities of various gait abnormalities and improve the accuracy of their medical findings.

Gait analysis is primarily the study and observation of human locomotive movement(s) that can be examined and measured via computing equipment at varying intervals of an identified individual. These gait impressions and assessment analysis are used to evaluate and treat patients with medical conditions affecting their mobility and ability to walk relatively normally. The continual evolution in technology can generally help improve disease management and treatment for patients with the support of gait analysis and with greater accuracy (Younesi et al., 2015). Leveraging these technological advancements made in mobile cloud computing (MCC), wearable sensors, and body markers that are interlinked to a computer (system), yield accelerated assimilation and analysis of engendered data (Nguyen et al., 2019). The data collected from gait analysis can be used to identify problems with a patient's gait and to develop treatment plans to improve their walking. This can enable gait experts to ostensibly make better-informed decisions, in near real-time, from within any location (Nguyen et al., 2016). Moreover, it can be vital that gait experts detect, treat, and expedite the decision-making in an effort to speed up a patient's well-being and overall health progress (Agibetov et al., 2016).

Historically, the word ontology refers to "the nature of being or existence" as (pro)claimed by philosophers, whose area of knowledge centers on entities and how they are clustered in relation to a similarity's hierarchy (Rapaport, 2005). However, in Information or Computer Science, ontology represents a set of concepts and categories that capture the properties and relations of a domain, allowing for a shared understanding and interoperability. Guarino (2009) emphasises that ontologies in Computer Science are not just hierarchical representations of entities but are explicit specifications of conceptualisations (Guarino et al., 2009). These ontologies, which are integrated and utilised across various applications, serve as knowledge artefacts that define a specific reality. Thus, while the concept of ontology has its roots in philosophy, its application and realisation in Computer Science offer a more structured and operational representation of the nature of things (Lu & Xu, 2017). Diverse knowledge areas or subject fields are categorised in their respective ontologies, which fundamentally represent a knowledge-domain with data, information, and knowledge about that domain. Thus, an ontology can provide a common shared vocabulary and (syntax) rules for publishing data and a semantic depiction of the data. In Computer Science, domain ontologies are used in Artificial Intelligence (AI), natural language processing (NLP), and semantic web applications to create a shared understanding of the meaning and relationships of concepts within a particular domain. One of the main motives for developing an ontology is information sharing, which can hopefully improve problem-solving within a domain. Henceforth, ontology in the ambit of

Computer Science represents a structure for describing concept data and the demonstration of knowledge to aid decision-making and knowledge administration within a community of shared interest. Accordingly, an ontology is a powerful tool that can be used to improve the representation, sharing, reuse, searchability, decision-making, and automation of knowledge (Sanfilippo et al., 2019; Uschold & Gruninger, 2004).

Biology or medical research, commonly referred to as biomedical, is where the conception of gait analysis predominantly exists. Contained by this medical purview, gait experts have their own vocabularies, terminologies and classifications that can be better represented when modelled and structured in a knowledgebase. In this way, an ontology can assist gait experts and patients by delivering adequate decision support in a (community-based) domain ontology. Users of an ontology publish and submit basically two types of data, ontology vocabularies and instance data. Domain ontology offers and provides a structure of the data by illustrating how meaning can be represented and the relationships among entity elements in the data. Therefore, it enables relationships between entities and concepts to be defined. This compulsion to accurately depict facts about gait-related diseases necessitates a proficient strategy to compose, standardise, illustrate, and disseminate domain knowledge via a structured vocabulary and a developed ontology application (Salvadores et al., 2013).

1.2 Background of the study

Implementing an intelligent system using ontologies often involves using the Web Ontology Language (OWL), the standard language semantics adopted by the World Wide Web Consortium (W3C). This allows for seamless procedural information interchange between applications within the semantic web framework. The implementation of an ontology, based on OWL, consists of classes as sets of concepts, individuals as instances of classes, and attributes as relationships between these entities. OWL ontologies can be demonstrated by using foundational ontology editors to infer knowledge about the knowledgebase. The growing expansion of the semantic web makes it easier for users and computers to implement ontology-based knowledge structures that can be useful for all relevant actors or role-players when competently designed and efficiently deployed. The semantic web is basically an extension of the existing web that gives structure and meaning to information, promoting enhanced interoperability between humans and machines (W3C, 2020).

Generally, OWL builds on Resource Description Framework Schema (RDFS), providing a more expressive language for describing classes, individual members of those classes, “object properties” that link sets of individuals, “data properties” that link individuals to exact values, and “annotation properties” for describing any of these. Ontologies can be designed and developed using formal ontology languages such as OWL or RDF (Resource Description Framework). The theoretical foundation for OWL is Description Logics (DL), a decidable section of First-order logic (FOL). The decidability of the underlying logical formalism in OWL ensures that stable and robust DL reasoners can be constructed to check the consistency of the ontology axioms, verifying whether there are any logical contradictions in the ontology. In addition, these reasoners can be used to make inferences from the asserted information, such as determining whether a particular class (concept) is a subclass (sub-concept) of another or if a particular individual belongs to a specific class. OWL reasoners are essential components of the semantic web, providing the capability to process and analyse ontologies automatically and to derive implicit knowledge from straightforward statements (Staab & Studer, 2009).

Computer Science ontologies are based on formal logic and facilitate logic-based reasoning. They represent knowledge through explicit specifications, promoting a shared understanding of a domain's vocabulary, concepts, and relations. In today's interconnected world, ontologies enable a uniform representation of information for effective communication and collaboration between humans and machines. The W3C defines a full stack for the semantic web, which includes eXtensible Markup Language (XML) for document structure and syntax, RDF for concepts, and OWL for inferencing to support DL. OWL ontologies facilitate a shared understanding and knowledge reuse at a universal level. This shift from human-centered to machine-readable content has spawned new methodologies, techniques, and tools for production and transformation into OWL ontology development (Roldán-Molina et al., 2021).

DL benefits knowledge representation in biomedical information processing, the semantic web, and intelligent systems. The Ontology for Biomedical Investigation (OBI), built on W3C principles, can be used with other available data on the semantic web. OBI is an integrated ontology that offers concepts with clearly defined definitions to describe all aspects of how investigations in the biological and medical domains are conducted. This facilitates knowledge-sharing and decision-making in the biomedical field and ultimately warrants patient well-being and general health improvement. Quite a few OWL reasoners are available and have been tested with OBI, each with satisfactory capabilities (W3C, 2020).

Recent years have seen ontologies gain significant attention and use in various research fields. Lately, ontologies have been widely applied in knowledge engineering, AI, e-commerce, digital information sharing, database management, bioinformatics, NLP, expert systems, and decision support. The need for gait experts to have access to and insight into clinical gait analysis and information has become increasingly important, especially with the growth of big data volumes and innovative technologies. Now more than ever, developing and designing a suitable domain ontology to assist gait experts with informed decision-making and advanced treatment ideas is rapidly becoming necessary (Agrawal & Cui, 2020).

In essence, this study transcends the mere development of a technology artefact; it addresses a critical need within a domain-specific context by harnessing the power of converging intelligent reasoning and semantic processing that paves the way for personalised, AI-driven disease management, which can enable decision support in the diagnosis and treatment of gait-related diseases (Atoba, 2022).

1.3 Research problem

Despite the emphasis on gait analysis in the treatment of gait-related diseases, gait experts face challenges in interpreting and drawing inferences from data due to a need for domain ontologies on gait-related disease matters (Cicirelli et al., 2022). This shortfall of a comprehensive and structured system for organising and categorising information about gait-related diseases makes it difficult for gait experts to access and utilise relevant information when making patient treatment and disease management decisions (Liu et al., 2021). Moreover, there is an insufficiency of consistency in the terminology and concepts used to describe gait-related diseases across different research areas, which are mainly derived from disparate sources. This can lead to confusion and misunderstandings when sharing and comparing vast amounts of information in addition to potential misdiagnosis or delayed care (Ajami & McHeick, 2018; Daramola & Moser, 2021).

This has resulted in limited decision support for gait experts, leading to unreliable data analysis, reduced knowledge-sharing, and difficulties in providing personalised treatment solutions and monitoring the progress of patients affected by gait-related diseases (Manosperta, 2019; Lu & Xu, 2017; Daramola & Moser, 2021).

Therefore, decision support for gait experts in the treatment of clinical gait-related diseases and knowledge-sharing has not been maximised due to a lack of a dedicated domain ontology for gait analysis that can also facilitate knowledge-sharing among stakeholders.

1.4 Aim and objectives of the study

This study aimed to develop a domain ontology for gait analysis that can support decision-making in the treatment and management of gait-related diseases.

1.4.1 Objectives

Based on the aim, the objectives of this study are to:

i. Identify the requirements of a domain ontology that can support decision-making in the treatment of gait-related diseases.

ii. Design a domain ontology that can support decision-making in the treatment of gait-related diseases.

iii. Develop a domain ontology to support decision-making in the treatment of gait-related diseases.

iv. Evaluate the domain ontology in terms of domain task fit, content richness, and correctness.

1.5 Research questions

The main research question of the study is:

How can a domain ontology that can support decision support for gait analysis be developed?

This study investigated the following sub-questions:

i. What are the requirements of a domain ontology that can support decision-making in the treatment of gait-related diseases?

ii. What is the conceptual design of a domain ontology that can support decision-making in the treatment of gait-related diseases?

iii. What does the development of a domain ontology that can support decision-making in the treatment of gait-related diseases entail?

iv. How effectively does the domain ontology for gait-related diseases meet criteria for domain task fit, content richness, and correctness?

1.6 Delineation of the study

This study concentrated on the development of a domain ontology specifically tailored for gait-related diseases, emphasising the critical facets of gait analysis. The ontology covers various dimensions of gait analysis, including the biomechanical, physiological, and especially the computational aspects. However, it is important to highlight that the research predominantly delved into the computational perspective, and there was no direct patient or human involvement in the study. Furthermore, certain areas, such as drug design for gait-related diseases and the pharmacological implications, particularly the effects of drugs and chemicals on gait patients, have not been explored within the scope of this study.

1.7 Significance of the study

The benefits of this study are significant because gait experts can reference this shareable domain ontology (knowledge base) to obtain decision support when dealing with patients with gait-related diseases. The ontology, serving as an open and freely accessible knowledge base, will facilitate the standardisation of terms and the shared understanding of concepts related to gait analysis across the healthcare sector.

1.8 Organisation of the thesis

This thesis is structured into six succinct chapters, each focusing on a specific facet of the study on gait-related and ontology development themes.

Chapter one presents an overview of gait analysis, highlighting its importance and the role of ontology in information organisation related to gait, thereby setting the context for the study. Chapter two presents a review of the literature on gait analysis, ontology, and related previous studies. Chapter 3 outlines the methodology adopted for the study, specifically the activities associated with the "Ontology Development 101" methodology, which was adopted for the development of the ontology. Chapter four presents the design and development of the gait analysis domain ontology. Chapter five presents the evaluation of the Gait Analysis Domain Ontology, focusing on its domain task fit and content richness through standard metrics. The chapter also conducts error checking and employs the FOCA methodology, Description Logics, and SPARQL queries to ensure the ontology's robustness, consistency, and applicability in the domain. Chapter six of the thesis presents a summary of the study, contributions, recommendations, and prospects for future work.

CHAPTER 2: LITERATURE REVIEW

This chapter presents a review of the literature aimed at identifying the critical elements for domain ontology development with a special emphasis on gait disorders and the importance of gait analysis. It delves into the theoretical background of gait analysis, ontology engineering, ontology evaluation, and querying. Furthermore, the different tools and languages available to facilitate the development and evaluation processes to assess accuracy and effectiveness are also discussed. The chapter concludes with an overview of related work.

2.1 Gait analysis

Gait analysis is the study of human locomotion, which includes measuring and characterising several aspects of movement, functioning as cadence, speed, stride length, foot impressions, skeletal joint and muscle movements, and other postural functions. In general, gait analysis involves measuring and analysing distinctive features of human motion, like the timing, sequence, and force of movements, to identify any abnormalities or deviations from standard gait patterns. This can be done through a variety of methods, which are video analysis, pressure analysis, and kinematic analysis. Gait analysis is commonly used to diagnose and treat gait-related diseases, as it can provide useful information about an individual's walking pattern and potential areas of improvement. Typically, it entails studying how a person moves while walking and analysing the biomechanical factors that affect their gait. This information can be used to diagnose and treat many gait-related diseases, such as Parkinson's disease, stroke, neuropathy, arthritis, and various other gait-related pathologies (Del Din et al., 2019).

Gait analysis can be performed for different purposes, including diagnosing and monitoring medical conditions that affect mobility and gait, improving athletic performance, and designing and fitting prosthetics or orthotics (Baker et al., 2016). The information elicited from gait analysis can provide important information and be used to diagnose and treat a range of gait-related diseases. Novel advancements in technology have made gait analysis more measurable and quantifiable by utilising computer-aided technologies and sensory configurations. Hence, the ascension of artificial intelligence serves as a method or instrument in gait analysis, enhancing the process by analysing and interpreting data generated by gait analysis equipment. Therefore, using ontologies in healthcare can aid in decision-making by assisting healthcare professionals with customising treatment plans and making decisive advances within the domain (Ajami & McHeick, 2018).

2.1.1 Gait analysis methods

Gait analysis methods embrace the use of electronic sensors or cameras to capture and analyse gait patterns. The instrumented gait analysis method provides valuable information about walking speed, step length, foot placement, and other gait parameters, which can be used to diagnose gait abnormalities and develop treatment plans. Instrumented gait analysis is more objective and accurate compared to observational gait analysis, which relies on visual observation. However, gait analysis is a complex discipline that requires expertise in biomechanical fundamentals, sensor technologies, and advanced data analysis techniques.
Biomechanical principles involve the study of forces and motion in the human body during gait, while sensor technologies include motion capture systems, (force) pressure sensors, and

wearable sensors. Force platforms and motion capture systems are often combined for comprehensive gait analysis, utilising data from both systems. Motion capture systems, known for their accuracy, are optoelectronic marker-based components with cameras and body markers attached to human subjects, which measure spatiotemporal gait parameters. This gait analysis method is typically used in research laboratories or controlled environments to facilitate human detection in marker-based vision systems (Cicirelli et al., 2022). Below in Figure 2.1 is a depiction of gait analysis equipment used in a laboratory setup to illustrate the gait analysis method.

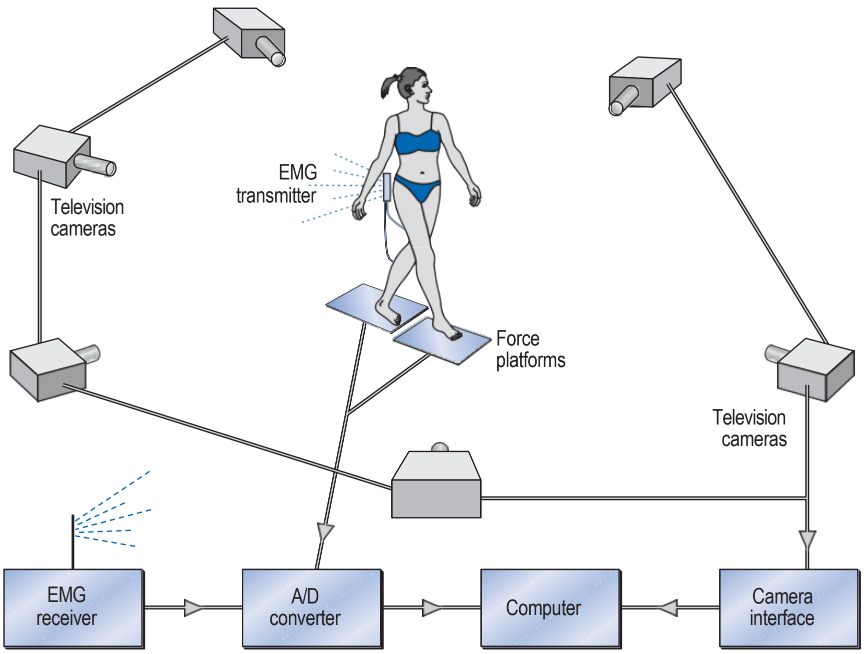


Figure 2.1: Gait analysis equipment used in a gait analysis method (Whittle, 2014)

Advanced data analysis techniques involve the use of statistical analysis and machine learning algorithms to analyse gait data. AI-based gait analysis methods can improve the accuracy, efficiency, and clinical applicability of gait analysis by automating data processing, reducing human error, and providing insights that may not be easily discernible by human observers. These techniques enable gait experts to gain valuable insights into the underlying mechanisms of human walking and develop new interventions to improve gait patterns in individuals with movement disorders. In healthcare, intelligent motion tracking and analysis are progressively being used, particularly in physiotherapy and sports kinematics. Therefore, gait analysis methods are essential for gait experts in diagnosing and treating gait-related diseases (Liu et al., 2021).

2.1.2 Gait analysis equipment

The use of gait analysis equipment has become an essential component of the comprehensive management of gait-related conditions. Significant advancements in gait analysis equipment have occurred over the years, making them increasingly accurate, reliable, and informative (Whittle, 2014). Several forms of gait analysis equipment are available, each with unique advantages in assessing, diagnosing, and managing gait-related matters. Biomechanical equipment, like motion capture systems and pressure sensors, can assess joint angles, muscle function, and foot mechanics during gait. Nonetheless, wearable sensors provide continuous measurements of walking patterns, whereas electromyography measures muscle activity during gait cycles. Magnetometers, when combined with accelerometers and gyroscopes equipment, form Inertial and Magnetic Measurement Systems (MMSs) that offer new possibilities for measuring kinematic force, including position, acceleration, and speed generated by motion. In gait analysis, these systems provide precise information on body segment orientation and movement, enabling researchers and clinicians to assess abnormalities, monitor rehabilitation, and optimise sports performance (Cicirelli et al., 2022).

These devices have been used in clinical rehabilitations for various conditions, allowing gait experts to ascertain a patient's type of gait exhibition, understand musculoskeletal classification, and postulate possible neuromuscular system complications. Progressive technological advances have led to the integration of AI algorithms in gait analysis, enabling the identification of subtle abnormalities that may not be apparent to the naked eye. AI algorithms, particularly machine learning and deep learning techniques can be used to analyse and interpret data collected by gait analysis equipment. These advancements provide gait experts with factual means of assessing and quantifying gait abnormalities, which in turn allows for informed decisions to be made about appropriate treatment options (Liu et al., 2021).

2.2 Gait-related diseases

Geriatrics is the medical field concerned with the physical, mental, and collective health of senior adults, particularly those over 65 who are considered elderly. Gait-related diseases and disorders are more prevalent among the elderly and are linked to loss of autonomy, limited mobility, and a lower quality of life (Auvinet et al., 2017). Cognitive decline, a significant contributor and underlying factor, is directly associated with the severity of gait impairment and abnormality in specifically older and less influential younger demographic groups (Auvinet et al., 2017). While the ageing process continues, several physical variations can profoundly impact an elderly individual's posture stability and gait mechanics. Such changes include, but are not limited to, stiffening of the connective tissue, reduced muscle strength, delayed reaction times, decreased visual clarity, debilitated vibratory and proprioceptive physicality, and aggravated postural guided movements. These age-related deviations can give rise to an array of gait abnormalities, ranging from simple age-related transitions in gait and balance to more complex dysfunctions of the nervous, muscular, skeletal, and respiratory systems or because of physical weakness following an extended period of inactivity. With the accruing of age, the progressive decline in muscle strength and dynamic motor controls required to co-ordinate sensory input and muscle contraction leads to changes in gait patterns over time, resulting in various gait impairments (Anwary, 2019). Studies have also shown that neural connectivity and function, play a crucial role in the mechanisms of gait, making the features and variability of gait more complex than just age-related changes as the elderly population is increasing (Mirelman et al., 2019). Using a domain ontology to treat these disorders can help improve the understanding and management of certain geriatric conditions.

Most gait disorders stem from neurological, orthopaedic, and health issues, such as sensory or motor impairments, osteoarthritis, musculoskeletal deformities, heart failure, cardiorespiratory deficiency, peripheral arterial disease, and obesity. Notwithstanding, psychological factors can also influence gait, with depression correlated through a lingering gait and anxiety linked to an immensely cautious gait. Studies revealed that patients with dementia walk slowly but too fast in relation to their motor and cerebral shortcomings, which subsequently increases their risk of falling. In the event of a risk of falling, relatively healthy individuals prioritise maintaining stability. However, individuals with Parkinson's disease fail this approach, highlighting the impact of the disease on gait and balance control (Pirker & Katzenschlager, 2017).

Daramola and Moser (2021) explored the use of new digital technologies to improve decision-making in the diagnosis, treatment, and rehabilitation of gait-related diseases. One of the key areas of focus was the integration of heterogeneous data (Daramola & Moser, 2021). The book chapter highlighted the importance of accurate diagnosis and treatment in patients with cardiovascular and neurological conditions that affect gait, for example Parkinson's disease, stroke, and other gait-related diseases. In terms of cardiovascular diseases, peripheral arterial disease is a condition that can cause gait impairments. Regarding neurological diseases affecting gait, one prominent example is stroke. Hence, stroke is a neurological condition that occurs when blood flow to the brain is disrupted. This disruption can cause a range of symptoms, including gait disturbance and abnormalities. The use of wearable sensors and mobile devices can be utilised to monitor the gait patterns of patients, which can inform treatment decisions and rehabilitation programmes (Daramola & Moser, 2021).

Through leveraging technologies, gait experts can monitor patients with gait-related diseases, provide personalised treatment plans, and optimise healthcare solutions to improve patient healing (Daramola & Moser, 2021). The leading purpose of the domain ontology is to enable systematic information extraction for decision support in the treatment of gait-related diseases. According to Ajami and Mcheick (2018), domain ontologies are feasible solutions for managing large data volumes in healthcare, particularly for monitoring patients. These ontologies can also assist in decision-making and disease management by providing gait experts with treatment suggestions and progress advancements made within the domain (Ajami & McHeick, 2018). Therefore, categorising and determining specific improper gaits exhibited by a patient, such as Hemiplegic gait, Diplegic gait, Neuropathic gait, Myopathic gait, or Ataxic gait, can ultimately help make better-informed decisions (Manosperta, 2019). Some of these gait dysfunctions are depicted in Figure 2.2 below.

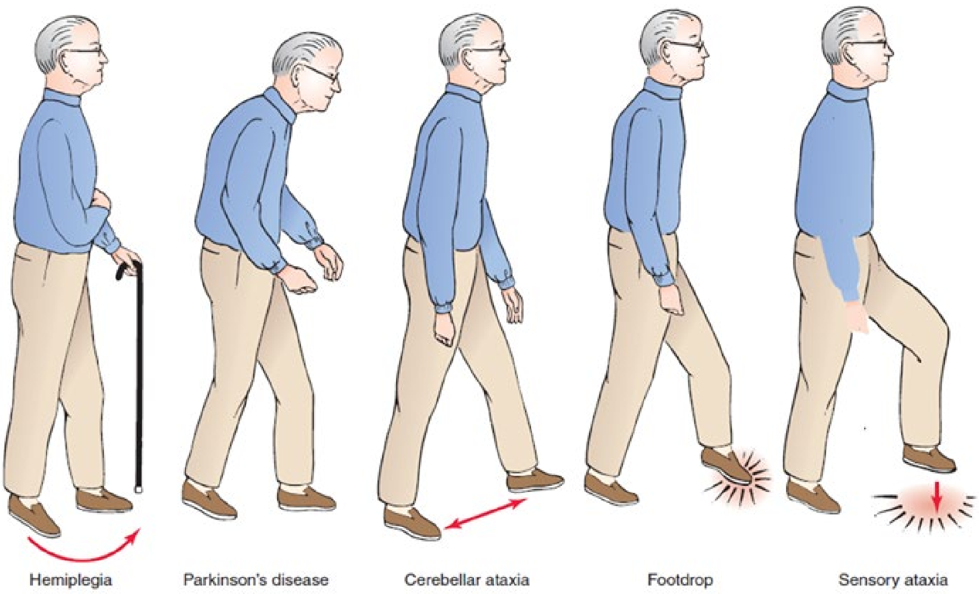


Figure 2.2: Neuro Exam: Post-Stroke Hemiplegic Gait (Cohan, 2019)

2.3 Ontology and ontology types

Ontologies have gained significant relevance across various applications, including knowledge management, artificial intelligence, and natural language processing, due to their ability to enhance system interoperability, improve data accuracy and consistency, and facilitate efficient data searchability. In terms of system interoperability, ontologies establish a shared understanding of terms and concepts within a specific domain, enabling seamless data exchange and effective communication between systems. Moreover, ontologies ensure data accuracy and consistency by precisely defining the meaning of terms and concepts, ensuring uniform interpretation across different systems. Ultimately, ontologies improve data searchability by offering a taxonomy of terms and concepts, enabling efficient information retrieval (Roldán-Molina et al., 2021).

The development of ontologies is an interdisciplinary field that draws on knowledge from Computer Science, Information Science, and Philosophy. In general, ontology is a philosophical discipline that focuses on how a set of thoughts within a subject area relate to each other and their fundamental reason for being. In artificial intelligence and knowledge representation, ontology has become essential for modelling a set of objects or categories, within a domain, and depicting their applicable properties and relationships. This makes domain assumptions explicit and promotes a shared understanding of the knowledge-domain. Subsequently, it constitutes a formal vocabulary to model a set of objects or categories within a domain. In an area where conceptualisation is often the abstract model of a real-world phenomenon, ontology is an explicit and formal specification of concepts and intelligent semantic relationships. This helps to facilitate clear and consistent communication between domain experts and enables the development of knowledge-based systems that can be used to reason about, orchestrate, and interpret data within that domain (Ajami & McHeick, 2018).

The structural constituents of an ontology are for the most part constituted triple, as in individuals, concepts, and relation roles. Individuals refer to the concrete or abstract objects that exist within a specific domain or subject matter, serving as the basic units of description. Concepts, on the other hand, refer to classes or categories of objects in the domain, providing a generalisation of the properties and relationships of the individual objects. Lastly, relation roles represent the relationships that exist between the individuals or concepts within the domain, providing a means to describe how the objects are related to one another. This can typically be achieved with annotations, as they assign semantic meaning to data. Ultimately, annotations provide additional details and clarification about the concepts defined in the ontology. Together, these fundamental components comprehensively represent the objects and their relationships within a given domain, forming the basis for a well-structured ontology. They allow for the representation of complex relationships, facilitating the organisation and retrieval of information, and supporting automated reasoning and decision-making. By providing a shared and agreed-upon understanding of the objects and relationships within a domain, ontologies play a pivotal role in facilitating communication and collaboration among domain experts, as well as enabling the development of applications and services that can utilise and reason about this knowledge. These elements constitute an ontological vocabulary for the domain, and the ontology is a set of statements expressed in this vocabulary, also known as axioms (Moodley, 2015).

2.3.1 Ontology types

Basically, ontologies can be categorised into four primary types: upper-level ontologies, domain ontologies, application ontologies, and task ontologies.

Upper-level ontologies serve as foundational frameworks, providing a common vocabulary and a broad range of concepts that can be reused across multiple domains. These ontologies capture generic knowledge and offer a high-level abstraction of concepts and relationships. They are frequently utilised as a reference for integrating different domain-specific ontologies (Keet, 2020).

In contrast, domain ontologies possess the primary objective of providing a thorough depiction of distinct knowledge domains. Their purpose lies in capturing and representing the knowledge employed by specific systems or applications. Hence, domain ontologies facilitate efficient retrieval of information, logical reasoning, and seamless knowledge dissemination within the confines of a particular domain (Sattar et al., 2021).

Application ontologies are meticulously crafted to cater to the unique needs of individual software applications or systems. These ontologies combine concepts extracted from diverse domain ontologies, forging a customised knowledge representation that seamlessly aligns with the specific requirements of the application domain. Their fundamental aim is to facilitate streamlined information processing and foster interoperability within the context of a particular application (Pittet & Barthèlèmy, 2015).

Task ontologies are geared towards representing knowledge essential for executing specific tasks. Therefore, this type of ontology incorporates concepts from both domain and application ontologies. Task ontologies capture the necessary concepts, relationships, and constraints for modelling and reasoning about tasks or activities within a given domain. In effect, they play a decisive role in facilitating intelligent task planning, process automation, and decision-making (Chavula & Keet, 2015).

Holistically, a domain ontology comprehensively represents the concepts and relationships that comprise a particular knowledge domain. Therefore, it is a type of ontology that is used to describe a specific area of knowledge and its associated terms, relationships, and definitions. Domain ontology provides a shared understanding of a domain and its related concepts, making it easier to share and exchange information between different collaborators. This permits them to define the structure of a given domain in a specific way. Domain ontologies, in particular, are critical for developing AI systems that can reason about specific domains of knowledge. Thus, domain ontologies are used in natural language processing, knowledge engineering, and semantic web development in order to provide a shared understanding of the domain and facilitate the efficient interchange of information. This facilitation of a knowledge-domain enables domain assumptions to be explicit (Abdelghany et al., 2019).

2.4 Ontology development

Ontology development connects the creation and refinement of a standard and machine-interpretable vocabulary that defines the entities, concepts, and relationships within a particular domain. Basically, it involves using ontology development tools and methodologies to create formal naming and definition of the terms within the domain, as well as to define their usage by others in the field. The ultimate goal of ontology development is to facilitate better semantic integration and sharing of information within a domain and across different disciplines. The introduction and inception of the semantic web have made ontology development more achievable as it enables humans to construct interrelated data on the World Wide Web, shape lexicon vocabularies, and formulate rules for data management. Ontology development is essential for building AI applications that can understand and interpret data in a meaningful way. In the semantic web context, ontologies provide a shared vocabulary for describing and exchanging data (W3C, 2020).

The semantic web is essentially a "web of data" that provides a collective framework for sharing and reusing data across applications in a standardised manner, primarily on the World Wide Web. The semantic web is a stack of technologies and language syntaxes that make data and information more machine-understandable on the internet, which is helpful for ontology development in the biomedical domain. For computer-readable ontologies to be practical, they need to be syntactically sound and human-understandable, which has been made possible with the advent of the semantic web (Blondé et al., 2009; Blondé et al., 2011). This can be used to build intelligent agents that automatically gather, analyse, and integrate data from multiple sources to support decision-making processes. The advancements in the semantic web include indexing, annotating, and reasoning using semantically enriched data, where ontology plays a dominant part (Ochs et al., 2016).

2.4.1 Ontology development methods

Ontology development and engineering follow a similar process to software engineering as both result in an artefact after being developed, conform to a system's life cycle, and adhere to a specific methodology. Different methods and methodologies exist for various applications;

some are built from scratch or can be derived from others. Therefore, there is no standardised methodology or one-size-fits-all approach. The technique of ontology development is a meticulous process as it involves systematic specifications and thorough collaboration of information sharing by several role-players engaged in constructing the ontology (Barton et al., 2014). Selecting the most appropriate methodology from proposed methodologies depends on the desired output and expected deliverables. The process of ontology development has become quite significant, in recent years, as ontologies have become the pillar of the semantic web, and the methodologies for ontology building determine which ontology is more suitable over another (De Nicola et al., 2009; Saad et al., 2011). Centered on specific requirements, a hybrid or adapted methodology might be useful in certain instances. Existing methodologies like "Methontology", Toronto Virtual Enterprise (TOVE), Uschold and King Enterprise Ontology, Modular Ontology Engineering (MOE), Ontology Development 101 (OD-101), as well as Agile Methodology for Ontology Development (AMOD), are prime examples of ontology development methods. Based on individual merits, each of these methodologies has its own strengths and weaknesses, and it is essential to consider which is best suited for a particular ontology (Abdelghany et al., 2019).

2.4.2 Ontology languages

Part of the ontology engineering process is utilising a suitable methodology to apply the right tools and techniques by implementing the appropriate ontology language it supports. Several ontology development languages denote reasoning and logic for formal language ontology representation of concepts, taxonomies, relations, and axioms.

Ontology languages are a set of formal languages used to represent the concepts and relationships within a specific domain or subject area. They are used to create ontologies, which are formal representations of knowledge that can be shared and reused across different systems and applications. Numerous ontology languages are widely used, including RDF (Resource Description Framework), OWL (Web Ontology Language) and Manchester OWL Syntax. RDF is a simple, flexible language that allows for creating statements about resources and their relationships. OWL, on the contrary, is a more expressive language that allows for the creation of rich and complex ontologies, including the ability to express complex relationships and constraints between concepts (Yadav et al., 2016). Protégé is an open-source platform that allows developers to create, edit, and visualise ontologies using different ontology languages, including RDF and OWL. Using ontology languages, like Web Ontology Language (OWL), Ontoligua, Ontology Markup Language (OML), Knowledge Interchange Format (KIF), and the standard Rules Interchange Format (RIF) language for the semantic web, can ultimately aid in the representation of concepts, taxonomies, relations, and axioms within the ontology. Development tools, such as Protégé, WebOnto, OWL Editor, Ontolingua and Swoop, can also support the construction and editing of the ontology (Sattar et al., 2021).

2.4.3 Ontology development tools

Ontology development tools are designed to facilitate the creation, editing, and maintenance of an ontology and ontology engineering. In order to achieve this, an editing environment and subsequent deployment for (automatic) updating can be enabled by an ontology editor (Yadav et al., 2016).

Most of these tools are based on existing ontology languages like OWL or RDF. Conventional tools used for ontology development include ontology editors, which provide an editing environment and facilitate the construction and amendment of an ontology. Also, it can be common practice for ontology editors to use one or more ontology languages. In addition, there are ontology visualisation tools, like OntoViz, which help visualise an ontology's structure and make it easier to understand. There are also ontology comparison tools like OntoCompare, which can be used to compare two ontologies to identify their differences and similarities. Other tools, such as ontology merging tools, can combine multiple ontologies into a single unified ontology. Finally, there are tools for evaluating ontologies for metrics-based and rule-based approaches, used to measure an ontology's accuracy, completeness, and consistency. Diverse ontology methodologies, languages, and tools, that may be of purpose, will enable the developer to investigate and identify which are appropriately suited for development purpose tasks (Peffers,Tuunanen & Niehaves, 2018).

2.5 Ontology evaluation

Ontology evaluation is an efficient process in ensuring the quality and sustainability of any ontological knowledge base. Primarily, it covers maintenance concerns for verifying and validating updates, amendments, the integrity of knowledge, consistency, and domain information inclusion (Amith et al., 2018).

Several different types of ontology evaluation tools can be used to assess an ontology’s quality. These include metrics-based approaches that measure an ontology's accuracy, completeness, and consistency. Rule-based approaches use formal rules to check the correctness of an ontology, and usability-based approaches measure the usability of an ontology. Amith et al. (2019) surveyed and evaluated numerous ontology evaluation and software tools like OntoQA, OntoAnalyser, OntoClean, OntoKeeper, OntoGenerator; as well as web-based ontology evaluation tools, against a set of self-imposed criteria, and concluded that OntoKeeper performed slightly better as an application tool for ontology evaluation in biomedical domains (Amith et al., 2019).

These evaluation methods ensure that ontologies are highly competent and embrace system interoperability and ontology alignment. Substantially, most ontology evaluation tools consist of metrics and attributes to determine ontology quality, which can be categorised as "domain task fit", "error checking", "libraries", "metrics", and "modularisation" (McDaniel & Storey, 2019).

2.5.1 Ontology evaluation of classes

2.5.1.1 Domain task fit

The need for ontologies is often closely tied to a specific domain or field. In the biomedical realm, for instance, ontologies are pivotal in supporting clinical decision support systems, data integration systems for bioinformatics, and health analytics. The quality of these resources can significantly impact both patient treatment and scientific research studies. Therefore, it can be challenging to separate the specific task for which an ontology is needed from the broader domain in which it is situated. Moreover, in assessing an ontology’s quality, researchers must consider not only its performance on a given task or set of tasks but also the accuracy and applicability of the ontology to the domain at hand. Thus, ontology task fitness and domain fitness are closely connected and can be considered as part of a single evaluative metric grade (McDaniel & Storey, 2019).

2.5.1.2 Metrics

The systematic evaluation of ontologies requires well-defined and manageable metrics to evaluate specific aspects of an ontology rather than simply assessing its overall effectiveness. Multiple sets of metrics, ranging from specific to complex, have been developed to weigh each aspect differently, thereby providing an overall rating. The completeness of an ontology is a crucial evaluation metric that determines whether it includes all necessary elements, like classes, properties, relationships, and instances, to represent the knowledge domain adequately. Metrics such as coverage, which measures the proportion of real-world entities defined in the ontology, can be used to evaluate completeness. The evaluation of ontologies is better performed using software rather than human intervention and requires objective assessment to translate attributes into numerical quality values. However, the discordance of consensus on quality attributes for different domains and applications presents a moderate challenge (Roldán-Molina et al., 2021).

2.5.1.3 Error checking

Checking ontology correctness involves detecting and correcting various error types, ranging from syntax issues to complex semantic and structural problems. Syntax errors can be detected by examining expression construction rules, while semantic errors require identifying inconsistent or incorrect meanings. Thus, prior research knowledge of evaluated ontologies for completeness, redundancy, and semantic errors using formalised concepts based on accredited literature and data sources, reinforces correctness and by doing so reduces or removes errors (Roldán-Molina et al., 2021).

2.5.1.4 Libraries

Ontology libraries have been developed to decrease the cost of creating new ontologies, simplifying the process for knowledge engineers to integrate applications. These libraries can store domain-specific ontologies, for instance, repositories dedicated to biomedical and upper-level ontologies. The reuse of existing ontologies enhances semantic interoperability, and as the number of ontologies increases, the demand for more libraries will subsequently rise. Evaluating the quality of these libraries is crucial, incorporating automated systems that continuously monitor and assess newly published ontologies conforming to a specified format (McDaniel & Storey, 2019).

2.5.1.5 Modularisation

The modularisation of ontologies is like software engineering and involves dividing ontologies into smaller, independent pieces that are easier to understand, maintain, and reuse. Modular ontologies have several layers of ontology modules, including a top-level ontology, mid-level domain ontologies, and domain-specific segments. Generating extractable parts of an ontology increases scalability and allows for the distribution of effort and greater control over visibility. To achieve these benefits, each module must be of high quality, fit the intended task, and be used independently. Pruning, the process of removing elements outside of a specific application domain, can be used to create a balance between completeness and preciseness. The goal is to create a single ontology or set of ontology units that provide a rich conceptualisation of the target domain while excluding any parts that are outside of its specific focus (McDaniel & Storey, 2019).

2.5.2 FOCA methodology

FOCA (Framework for Ontology Conformance Analysis) is a structured methodology for evaluating ontologies. Typically, it combines a set of criteria, like the number of classes, the use of taxonomies, the use of roles and properties, the use of constraints, and the use of generalisation or specialisation, to evaluate how well an ontology demonstrates a given domain. The FOCA methodology considers the main quality criteria and matches them with the five roles of knowledge representation using the goal question metric (GQM) approach. The GQM approach covers the five metrics roles: completeness, adaptability, conciseness, computational efficiency, and clarity. This approach was decided because it aligns with evaluation goals, metrics, and criteria. The roles of knowledge representation are the goals, while the quality criteria are the metrics. The GQM approach is meant to be cyclical and should be repeated as the ontology evolves to ensure that it continues to meet its goals. FOCA provides several measures and a framework for ontology evaluation, making it accessible to developers of any experience level. In essence, it incorporates a question style for component evaluation and statistical modelling to calculate ontology quality. Beta regression models are used in FOCA to estimate the probability of each ontology element being correct, given its position within the ontology and other factors. By ensuring that the ontology is accurate and complete, FOCA methodology helps to support accurate and reliable knowledge representation and reasoning (Bandeira et al., 2016). Table 2.1 below illustrates FOCA based on GQM, which is meant to be cyclical and should be repeated as the ontology evolves to ensure that it continues to meet its objectives.

Table 2.1: The FOCA methodology GQM (Bandeira et al., 2016)

The GQM of FOCA Methodology		
Goal	**Question**	**Metric**
1. Check if the ontology complies with Substitute.	Q1. Were the competency questions defined? Q2. Were the competency questions answered? Q3. Did the ontology reuse other ontologies?	1. Completeness. 1. Completeness. 2. Adaptability.
2. Check if the ontology complies with Ontological Commitments.	Q4. Did the ontology impose a minimal ontological commitment? Q5. Did the ontology impose a maximum ontological commitment? Q6. Are the ontology properties coherent with the domain?	3. Conciseness 3. Conciseness 4. Consistency.
3. Check if the ontology complies with Intelligent Reasoning.	Q7. Are there contradictory axioms? Q8. Are there redundant axioms?	4. Consistency. 3. Conciseness.

4. Check if the ontology complies with Efficient Computation.	Q9. Did the reasoner bring modelling errors?	5. Computational efficiency.
	Q10. Did the reasoner perform quickly?	5. Computational efficiency.
5. Check if the ontology complies with Human Expression.	Q11. Is the documentation consistent with the modelling?	6. Clarity.
	Q12. Were the concepts well written?	6. Clarity.
	Q13. Are there annotations in the ontology that show the definitions of the concepts?	6. Clarity.

In accordance, ontology evaluation is a comprehensive process involving two key components: validation and verification. The first component, ontology validation, ensures that the ontology is built properly and conforms to the requirements established in the specification phase. This can be solved by conducting an ontology content evaluation, which assesses whether the ontology meets specific criteria, as well as by using competency questions to evaluate the accuracy of the ontology. The purpose of ontology validation is to determine whether the ontology is semantically consistent, complete and meets the intended requirements. In lieu of this, verification ensures that the ontology meets specific quality criteria and is built optimally. This can be done by conducting an ontology taxonomy evaluation and using the FOCA methodology, which introduces a number of steps and provides a framework for evaluating them. This allows developers, of any experience level, to assess their design and ensure that the ontology ideally meets the desired requirements. FOCA is an excellent tool for ontology developers as it provides an awareness of the durability of the ontology (Alsanad et al., 2019).

2.6 Formal languages in ontology development

In the context of knowledge representation and Computer Science, an ontology is a formal, explicit specification of a shared conceptualisation (Gruber, 1995). This conceptualisation can be represented using formal languages, which are foundational tools for ontology development. The choice of formal language is important, as it determines the expressiveness and computational properties of the ontology (Baader et al., 2008). Two prominent formal languages in ontology development are Description Logics (DL) and First-Order Logic (FOL). Particularly, DL and FOL play a decisive role in evaluating the ontology's logical soundness and expressive power, enabling the identification of any inconsistencies or incompleteness in the knowledge representation and the reasoning about the knowledge to make deductions and inferences.

2.6.1 Description logics

Description logics (DLs) are a family of formal knowledge representation languages that are used in artificial intelligence and ontology development to represent a domain's concepts and relationships. DLs provide a set of rules for defining classes and their properties, allowing for the formal specification of knowledge and automated reasoning about the consistency and inferred relationships in the ontology (Keet, 2020).

DLs can express a wide range of knowledge and provide a flexible and intuitive approach for representing and reasoning ability. They have been used in knowledge representation, and their expressive power and efficient decision procedures have made them a popular choice for ontology development. Furthermore, DLs are the basis for the Web Ontology Language standardised by the World Wide Web Consortium (W3C, 2020). DLs are particularly useful for knowledge representation in biomedical, semantic web, and intelligent systems. In biomedical applications, DLs can be used to represent the structure and function of biological entities, which are genes, proteins, and diseases. In the semantic web, DLs are used to represent the meaning of web resources in the manner of web pages, documents, and images. In intelligent systems, DLs are used to represent the knowledge that is used by the system to make decisions and solve problems (Kamide, 2020).

Description Logics are widely used in ontology engineering, which involves representing knowledge and its structure in a computationally understandable format. Moreover, DLs provide a systematic way to describe and define the components of an ontology, serving as concepts, individuals, and relationships. This is achieved by expressing these components and their relationships in the form of logical sentences known as DL axioms. The collection of DL axioms constitutes a DL ontology, which represents a specific knowledge domain. In this context, an ontology can be thought of as a set of assumptions that comprehensively describe a particular subject matter, for example, gait-related concepts. The purpose of DLs is to allow for a precise and unambiguous representation of this knowledge in a format that can be processed by machine reasoning algorithms such as an ontology reasoner. DL Query is a powerful tool that allows users to query a domain ontology using Description Logic expressions (Moodley, 2015).

OWL, developed by the W3C group, is a semantic web ontology language with defined semantics that can be translated into an expressive Description Logic. DL provides a formal language for describing and representing ontologies, which are proper descriptions of the concepts, entities, and relations in a given domain. DL has two main components: syntax and semantics. Syntax outlines the structure of the language, and semantics defines the meaning of the language. Syntactically, DL is composed of a set of logical symbols and operators that can be used to construct logical expressions. Semantically, DL uses an interpretation function to assign meaning to these logical expressions. The interpretation function maps symbols, terms, and formulas to the entities in the domain, enabling the expression to be evaluated accurately. This language was designed to leverage the results of DL inferencing and utilise existing DL reasoners to provide reasoning capabilities for OWL applications. The presence of these tools has facilitated the extensive utilisation of OWL, not only within the realm of the semantic web, but also as a favoured language for ontology creation across a multitude of disciplines, including biology, medicine, geography, geology, astronomy, agriculture, and defence. OWL applications are especially prevalent in the life Sciences, where it has been employed by the creators of numerous large-scale biomedical ontologies (Baader et al., 2008).

Indicative of the abovementioned, an OWL ontology is composed of a collection of axioms. These axioms facilitate the declaration of subsumption or equivalence relationships with regard to classes or properties, the establishment of disjointness among classes, and the identification of equivalence or non-equivalence among individuals (instances). The axioms supported by OWL are consolidated in Table 2.2 below to illustrate examples of DL syntax applications.

Table 2.2: Example OWL axioms and DL syntax (Baader et al., 2008)

Axiom	DL syntax	Example
subClassOf	$C_1 \subseteq C_2$	Human ⊆ Animal ∩ Biped
equivalentClass	$C_1 \equiv C_2$	Man ≡ Human ∩ Male
subPropertyOf	$P_1 \subseteq P_2$	hasDaughter ⊆ hasChild
equivalentProperty	$P_1 \equiv P_2$	cost ≡ price
disjointWith	$C_1 \subseteq \neg C_2$	Male ⊆ ¬Female
sameAs	$\{x_1\} \equiv \{x_2\}$	{Pres_Bush} ≡ {G_W_Bush}
differentFrom	$\{x_1\} \subseteq \neg\{x_2\}$	{john} ⊆ ¬{peter}
TransitiveProperty	$P \in R_+$	hasAncestor$^+$ ∈ R_+
FunctionalProperty	$T \subseteq (\leq 1\ P)$	T ⊆ (≤ 1 hasMother)
InverseFunctionalProperty	$T \subseteq (\leq 1\ P^-)$	T ⊆ (≤ 1 isMotherOf$^-$)
SymmetricProperty	$P \equiv P^-$	isSiblingOf ≡ isSiblingOf$^-$

Description Logics

2.6.2 First-order logic

First-order logic provides a concept of logical consequence and universal truth, which embodies the core of human reasoning. These ideas can be explained using model-theoretic semantics, in which a model of a logical theory explains an event that renders the theory valid. Deduction or inferencing, which is a method of accessing knowledge that is not explicitly offered but is implicitly represented by the theory, can be used to draw logical consequences from a theory. Since the time of the Greek philosophers, deduction has been studied, and proof theory specifies syntactic rules that act on views and allow logical consequences to be inferred without the use of models. This enables automated deduction, the main goal of automated reasoning. In order to fulfil this to be effective, such algorithms need to be solid and, ideally, complete (Grimm et al., 2009).

In the context of domain ontology development, first-order logic is often used as the logical basis for Description Logics, which is a family of knowledge representation languages used to describe and reason about concepts and their relationships in a domain. FOL and DL provide a foundation for developing knowledge-based systems that can reason about the concepts and relationships within a domain. First-order logic, also known as predicate logic, is a formal system used to represent and reason about statements involving quantifiers and predicates. In FOL, variables can take on values from a specified domain, and predicates are used to describe the properties of those variables. Concepts correspond to classes in domain ontology development, unary predicates in logic, or concepts in description logic. Relationships represent the semantic connections between concepts and instances in ontology, binary predicates in logic, or roles in Description Logics (Keet, 2020). Table 2.3 depicts an overview of the corresponding terms used in OWL, DL and FOL.

Table 2.3: Logic terms

OWL	DL	FOL
class	concept name	unary predicate
name class	concept	formula with one free variable binary
object property name	role name	predicate
object property	role	formula with two free variables theory
ontology	knowledge base	theory
axiom	axiom	sentence
vocabulary	vocabulary / signature	signature

Synopsis of the corresponding terms used in OWL vs DL vs First-order logic.

2.7 Ontology querying and reasoning

SPARQL querying and reasoners are both tools that operate on ontologies, but they serve distinct purposes. SPARQL is a query language used to retrieve and manipulate data stored in RDF format. Reasoners, on the other hand, are tools that infer implicit knowledge from explicit facts and relationships specified in an ontology. In general, these tools or mechanisms interact with ontologies post-development.

2.7.1 SPARQL querying

SPARQL Protocol and RDF Query Language (SPARQL) is a powerful query language and protocol designed for the semantic web, which enables users to query and manipulate RDF data. The SPARQL Protocol and RDF Query Language is a W3C standard for querying and manipulating RDF data in a knowledge base, which an ontology is an example of (WC3, 2020).

SPARQL offers distinct advantages when querying ontologies and RDF data compared to other query languages, such as structured query language (SQL) for relational databases or XQuery for XML data. Unlike SQL, which is designed for tabular data, SPARQL is optimised for graph-based data structures, enabling more expressive and flexible querying of complex relationships. Furthermore, to effectively work with SPARQL, it is necessary to delve deeper into the inner workings of OWL. Relational databases use tables to represent data, while OWL uses triples as its fundamental construct. The foundation of RDF graphs is based on triples that consist of a subject, predicate, and object. Whenever a property is defined in OWL, it is essentially describing a predicate. Lastly, individuals can serve as either the subject, the object, or both, further contributing to the complexity of SPARQL querying (DuCharme, 2013).

The SPARQL query language enables retrieving data from RDF graphs, which consists of two main parts, the “SELECT” and “WHERE” clauses. The SELECT clause defines what data to display, while the WHERE clause specifies what to match in the query. The SPARQL dataset comprises subject-predicate-object triples, with each triple representing a statement. The WHERE part of the query can match variables to parts of the triple pattern and constrain the data returned in the SELECT portion. This SELECT part of the query specifies what variables to display, and can also group, order, or filter data. Thus, SPARQL's syntax is based on a set of graph patterns matching these operations to facilitate various queries, ranging from simple value extractions to complex graph transformations (Debellis, 2021).

One can examine the use of SPARQL to test and validate ontologies and how it can be used as a tool to extract meaningful information from ontologies. In ontology development, SPARQL is essential for querying and extracting useful information on top of its compatibility with ontology reasoners, making it a key component to providing inferred knowledge. Hence, it provides a more structured and efficient approach by enabling semantic annotation, query-based retrieval, and integration of multiple data sources. Therefore, SPARQL is reinforced as the standard query language for RDF, allowing users to search, filter, and manipulate data stored in ontologies. Moreover, the semantic web relies on SPARQL playing a pivotal role in its ecosystem (DuCharme, 2013).

2.7.2 Ontology reasoners

An ontology reasoner is a type of software programme used to infer knowledge from a given ontology. Ultimately, it is responsible for searching through the ontology and applying reasoning rules to infer new information from existing knowledge. This allows the reasoner to make logical deductions, answer queries, and find relationships between concepts in the ontology. Ontology reasoners are often used in artificial intelligence applications, such as natural language processing and text mining, to help extract meaning from text. They can also be used to assist in decision-making and problem-solving. Reasoners are also used in the semantic web, where they are used to help machines understand the meaning of web content (Golbreich et al., 2007).

A reasoning support system must have a formal semantic foundation to enable automated derivations that do not need to be manually constructed. This reasoning support is highly beneficial for the development and maintenance of large ontologies, as it allows checks to be made on the consistency of the ontology, any unintended relationships between classes, and the automatic classification of instances. Mapping an ontology language to a general logical formalism and using existing reasoners is the typical approach to providing formal semantics and reasoning support. OWL has been partially mapped to a description logic and utilises existing automated reasoners for this purpose (Staab & Studer, 2009).

OWL reasoners use DL to process ontologies and produce meaningful inferences. For example, suppose an ontology states that all cats are animals. In that case, an OWL reasoner can infer that all cats are mammals since mammals are a superclass of animals. OWL reasoners can also detect inconsistencies in the ontology if the ontology states that cats are both animals and reptiles. Reasoners can perform a variety of tasks, like consistency checking, classification, instance retrieval, and query answering. Some popular OWL reasoners include HermiT, Pellet, JFact, Fact++, and RacerPro. These reasoners differ in terms of performance, functionality, and supported OWL features. In particular, some reasoners are optimised for consistency checking, while others are designed for efficient classification and instance retrieval. Proper semantics and reasoning support for the Web Ontology Language can be provided by mapping the OWL to a recognised logical formalism, such as the use of DLs, and employing existing automated reasoners (Baader et al., 2008).

Computer Science and Mathematics have created methods to express knowledge and rules formally using logical expressions. This allows for the creation of a formal proof that demonstrates how conclusions can be derived from the premises. While this process can be done manually for small theories, it becomes impractical for more extensive theories or axioms. In a bid to overcome this challenge, ontology developers have focused on automating reasoning processes. Thus, ontology enables rigorous reasoning based on semantics and logic, improving the construction of engineering models and linked processes (Keet, 2020).

2.8 Related work

The use of domain ontology in gait and gait analysis has gained widespread recognition and implementation. In a study by Turcin et al. (2013), the authors aimed to demonstrate the application of domain ontology in the construction of a decision-support data warehouse architecture. The focus of the domain ontology in this study was on the musculoskeletal lower limbs and the mapping of this ontology to data warehouse models for generic data mining. The authors explored data mining as a method of foundation during the creation process since the focus area was an ontology of lower limb gait disorders. Contending to the size and complexity of biomedical ontologies, the authors opted to adopt OSMMI (Ontologie du Systeme Musculo-squelettique des Membres Inferieurs) as an ontology for their project. Since the authors based their model on OSMMI, it did not comprehensively satisfy the gait analysis requirements because it was too generic but could highlight other aspects of posture, ligament, and articular contact.

Research on ontology implementation for gait analysis has been an active area of study. One recent trend is the use of fuzzy ontologies and sensor data for gait recognition, as proposed by Huitzil et al. in 2019. The main goal of this study was to demonstrate the interoperability and reuse of applications and data using semantic web technologies for human gait motions. Instead of using traditional ontology methods, the authors suggested using fuzzy ontologies, which can handle non-specific and vague knowledge by substituting exact numerical figures with more flexible fuzzy datasets. This research aimed to gain an insight into human gait by analysing data collected from human subjects in a controlled environment. The authors claimed that some knowledge is not always clear and precise, with fuzzy data or datasets that must be considered and can be better represented in "fuzzy ontologies" instead of "conventional ontologies". Gait data acquisition and extraction procedures were set out to aggregate data values. These data imprecisions were represented in a fuzzy ontology. The authors used a Microsoft Kinect sensor for recording human subjects for gait analysis. They also used an algorithm to determine the gait patterns of human subjects for gait (pattern) recognition that could be useful in security surveillance.

In 2019, Manosperta conducted a study investigating the relationship between neurological and physical disorders and specific gait abnormalities. The study used a combination of wearable sensors, floor sensors, and image processing techniques to gather and analyse gait data. The collected gait data were then compared to existing datasets in three databases to improve the accuracy and precision of the analysis. The research specifically evaluated neurodegenerative diseases such as Parkinson's, Alzheimer's, Huntington's, and Dementia to detect changes in posture during gait observations. This enabled the researcher to enhance the accuracy of the analysis and gain a deeper understanding of the impact of neurodegenerative diseases on gait.

In recent years, there has been a growing interest in the use of ontologies for the representation and organisation of medical knowledge. The methodology for ontology design and construction has been widely discussed in the literature as a key factor in developing effective ontology-based systems for medical diagnosis. One notable study in this area was "Methodology for ontology design and construction" by Bravo, Reyes, and Ortiz (Bravo, Reyes & Ortiz, 2019). This study proposed a methodology for ontology design and construction that is applied in a case study for medical diagnosis. The case study involved designing and implementing an ontology system to support decision-making during the diagnosis of medical diseases. The methodology was based on a three-step process: analyse the domain of the ontology, create the ontology, and validate the ontology. The first step consisted of modelling the domain and identifying the concepts and relationships between them. The second step involved the implementation of the ontology, which included defining the classes, properties, and instances. Finally, the third step was dedicated to validating the ontology, which involved testing the ontology's consistency, completeness, and correctness. The study by Bravo et al. provided valuable insights into the methodology for ontology design and construction in the context of medical diagnosis. The results of the case study demonstrated the potential of ontologies in improving the decision-making process during medical diagnoses and highlighted the importance of a practical methodology for ontology design and construction. The objective of the case study was to support the decision-making process during the diagnosis of medical diseases by providing a structured representation of medical knowledge. The authors concluded that their proposed methodology was effective and efficient for ontology design and construction in the medical domain, but it was too basic.

Despite the abundance of research on assorted gait-related topics, there was a sheer lack of studies that specifically focused on developing domain ontologies for decision support for gait experts who essentially treat gait-related diseases. Many of the proposed approaches concentrated only on gait analysis, without emphasising the importance of creating an ontological knowledgebase to assist with expedited decision making.

More research is needed in this area to develop a comprehensive knowledge base that can help gait experts make accurate and accelerated decisions about the treatment of gait-related diseases. In addition, constructing a domain ontology for decision support with the capabilities to accommodate expansion enables integration for reusability and regular maintenance enhancements.

2.9 Summary

The literature review chapter explored the concepts and methods of gait ontology development and evaluation, emphasising the importance of gait analysis for gait-related diseases and the suitability of ontology development as an approach. The chapter discussed ontology development tools and methodologies, the FOCA methodology for ontology evaluation, and other evaluation techniques, as well as the use of Description Logics, First-order logic formalism, SPARQL querying, and ontology reasoners. The insights gained from the review were instrumental in developing a comprehensive ontology for clinical decision-making.

CHAPTER 3: METHODOLOGY

This chapter presents the research methodology adopted for this study. The discussion covers aspects of the research philosophy, research approach, research strategy, and research design used to execute the study. In order to achieve this, the Ontology Development 101 methodology was rigorously employed to guide the ontology development process. Furthermore, a mixed-methods research design was adopted, combining both qualitative and quantitative approaches for data collection and subsequent analysis.

3.1 Research philosophy

In research, a researcher is guided by a specific worldview, which shapes how the interpretation of research data reflects the research paradigm. This philosophical way of thinking guides a researcher's action and set of beliefs by creating the lens through which the researcher looks at the world (Kivunja & Kuyini, 2017).

The philosophical paradigm that was adopted for this study was pragmatism. Pragmatism enables the researcher to convert observations of reality into theories and then evaluate those theories through practical action (Tran, 2017). The pragmatist considered the most effective methods to use to solve real-world everyday problems with tangible entities of conception. Hence, pragmatism was aptly suited for this study in the development of a domain ontology artefact for gait-related diseases.

Pragmatism prioritises the practical application of ideas and the usefulness of knowledge. This guiding principle is particularly relevant in research endeavours that involve artefact development. In the case of gait analysis, the ontology artefact is a formal account of the domain of knowledge. Pragmatism directed the researcher towards domain ontology development by prioritising practical utility, shared understanding, and flexible adaptation to domain-specific contexts, ensuring a focus on real-world problem solving and robust knowledge representation (Kankam, 2019).

3.1.1 Ontological stance of the study

The ontological perspective adopted for the study was objectivism. This means that the development of the ontology was grounded in observations of the physical world and aimed to create a structured representation of the domain that could provide decision support. In view of the researcher developing an application artefact for a domain ontology for gait-related diseases, it adopted an objectivist perspective. Therefore, it was assumed that the domain had an objective reality and properties that could be comprehensively understood and studied. The focus was on developing a structured domain ontology that would provide decision support and facilitate a gait expert’s ability to make informed decisions when treating patients with gait-related diseases. Hence, the researcher considered that the ontology ought to be a comprehensive and accurate representation of the objective reality of the domain to enable adequate decision support capable of addressing gait-related matters objectively (Weigand, Johannesson & Andersson, 2021).

3.1.2 Epistemological stance of the study

In adopting a pragmatic epistemological perspective, this study posits that knowledge is derived through the active engagement of individuals within specific domains. Anchored in this belief, the primary aim of this study was to enhance the decision-making process of gait experts in the treatment of gait-related diseases.

The domain ontology would function as a focal tool, encapsulating a collective understanding and presenting a standardised terminology for gait experts. Beyond merely providing a structured overview of the domain, this ontology can assist gait experts in efficiently querying and extracting relevant data and information. Throughout its creation, the ontology was iteratively refined to ensure its fidelity to the domain, cementing its practical utility as a critical tool for improved decision support. Upon its complete development, this ontology stands to benefit a broader spectrum of domain experts. The ultimate goal is to establish a unified and standardised vocabulary, thereby fostering a more streamlined and effective approach to the domain. This approach emphasises the introduction of coherent terms and definitions that find pertinence across diverse practical applications. It serves as a testament to the idea that concentrating on the practical implications of knowledge can significantly enhance the understanding and practice within a given domain (Weigand et al., 2021).

3.2 Research approach

The central hypothesis of the study posits that a domain ontology can support decision-making in relation to gait-related diseases. Consequently, all aspects of the study were designed to test and validate this hypothesis. The data collection process, which encompassed both qualitative and quantitative data, aimed to either validate or refute the hypothesis. Thus, while the study employed a mixed-methods methodology, the overall research approach can be classified as deductive reasoning.

This study involves conducting a comprehensive literature review to gather existing knowledge in the field. Furthermore, document reviews, published articles, and online sources were utilised to collect qualitative data, thereby capturing the experiences and perspectives of gait-related subject matters. The information acquired through these qualitative methods served as a foundation for the subsequent development of an ontology artefact that was both scientifically accurate and practical for implementation in medical settings. Subsequently, deductive reasoning was applied during the evaluation of the developed artefact to validate the hypothesis that a domain ontology can support decision-making about gait-related diseases (Miah & Genemo, 2016).

3.3 Research methodology

The study implemented a mixed-methods approach guided by pragmatism, prioritising practical functionality and the iterative refinement of the gait analysis ontology through cycles of qualitative and quantitative analysis (Kankam, 2019). Initially, qualitative data from document reviews, literature, and requirements gathering informed the ontology's core concepts and relationships. This information then steered the development of the domain ontology, which was subsequently evaluated using quantitative data to assess its effectiveness and identify areas for improvement. This cyclical process, grounded in pragmatism's emphasis on practical consequences and abductive reasoning, enabled robust methodological triangulation and enhanced the validity and verifiability of the developed ontology (Kelle et al., 2019).

3.4 Research strategy

3.4.1 Ontology Development 101 activities

This study adopted the Ontology Development 101 (OD-101) methodology, an established and widely recognised ontology engineering methodology, which offers a standardised approach to ontology development, proposed by Noy and McGuinness (2001). Drawing upon the OD-101 methodology presents a comprehensive strategy that encompasses all the essential steps required for the successful creation of the gait analysis ontology, ensuring its quality, reusability, and practicality (Spoladore & Pessot, 2021). Fundamentally, OD-101 in general adheres to the following activities:

i. **Identify problem (Define the domain of the ontology):**
The initial phase involved identifying the problem and defining core requirements. This included establishing the scope of the domain-specific knowledge that the ontology needed to cover, specifically focusing on gait-related matters. By clearly defining the problem and setting the boundaries of the ontology, this stage laid the foundation for subsequent development steps.

ii. **Design (Identify the key concepts in the domain and develop a taxonomy of the concepts):**
Resources such as articles, books, scientific papers, and relevant materials were gathered to acquire in-depth information about the domain. This information was utilised to design an ontology that accurately represented the concepts and relationships within the gait-related domain. Key concepts and terms relevant to the gait-related domain were identified, and a taxonomy was developed to organise these concepts into a hierarchy.

iii. **Development (Define the properties of the classes, define the relationships between the classes, and create instances of the concepts):**
Once the design phase was completed, the development phase commenced. The ontology was developed using the preferred ontology language in accordance with the methodology. The structure of the ontology was defined, and it was populated with relevant data, meticulously aligning with the ontology engineering procedures prescribed by the methodology.

iv. **Evaluation (Evaluate the ontology):**
Following the development phase, the ontology underwent rigorous evaluation. This crucial step aimed to assess the domain task fit, correctness, and content richness of the ontology. Various evaluation techniques were deployed, including competency questions, standard metrics, error checking with automated reasoners, and rule-based query logic. Through a combination of quantitative and qualitative data analysis, the effectiveness of the ontology was thoroughly examined.

v. **Conclusion:**
Finally, the conclusion phase provided valuable recommendations and insights to refine and improve the ontology to ensure possible deployment.

3.5 Research design

A research design defines a logical plan of activities to attain the objectives of a study. The research design of the study is based on the OD-101 methodology, which provided a comprehensive and detailed framework for the development of a domain ontology for gait-related diseases.

Ontology Development 101 (OD-101) was used to guide the ontology development process. This feasible methodology emphasised the importance of domain analysis and the formulation of competency questions, which were critical steps in developing an ontology that met the needs of its expected target group. OD-101 also stressed the importance of ontology design, implementation, and evaluation, as well as iteration and maintenance, to ensure the ontology remained up-to-date and useful over time. This approach to ontology development can adequately facilitate ontology maintenance and evolution (Spoladore & Pessot, 2021).

The ontology was iteratively developed, allowing for user feedback integration and continuous refinement based on evaluation outcomes. Underlining domain analysis, it used competency questions as a framework and followed the systematic OD-101 methodology for its development. This methodology emphasises iterative processes and adjusts to requirements and growing domain knowledge, particularly useful for the ever-evolving data sources in gait-related diseases. With a focus on standard formats and terminologies, OD-101 enhances interoperability and flexibility, adapting to specific domains through usability and performance evaluations. This approach ensured an optimised ontology for specific use cases and promoted a real-world application, fostering ongoing maintenance updates and improvements. The OD-101 methodology offered a flexible way to improve the ontology over time (Aminu et al., 2020). This methodology in general encompasses the following:

3.5.1 Domain Analysis (Phase 1):

The research commenced with a thorough domain analysis to understand the intricacies of gait-related diseases. The first step was to thoroughly analyse the domain, identifying the key concepts and relationships within the domain. The requirements for the domain ontology and identifying the right competency questions on gait-related diseases were carried out by gathering information from numerous secondary sources like websites, scientific journals, published articles, academic books, and topic-related reference ontologies. The researcher also conducted a document review by referencing online documentation and medical publications to define the scope and granularity of the domain. Literature review and analysis were used to enhance credibility, validity, and a broader understanding of the requirements for a domain ontology on gait-related subject matters.

Utilising secondary data for ontology development in gait analysis offers distinct advantages, especially considering its broad domain. The primary merit of secondary data in this context is the establishment of a shared understanding rooted in established knowledge. Peer-reviewed publications, medical databases, and authoritative texts on gait analysis serve as invaluable sources for obtaining this foundational knowledge. These data sources provide a wealth of well-vetted information, ensuring the ontology's credibility and transparency, and eliminating potential biases from subjective views (Nowell et al., 2017).

During the pandemic, acquiring primary data for gait analysis became increasingly challenging due to participants' reluctance, logistical issues, and technical limitations, particularly in regions experiencing frequent power outages. Although modern technologies, like videoconferencing, offer some respite, they are not fool proof solutions. Given these hurdles, secondary data became indispensable. To ensure ethical standards, the research integrated document review analysis, which addressed concerns about participant technology proficiency and affordability that seem crucial amid the prevailing load-shedding power issues.

Furthermore, the discipline of ontology development, specifically in gait-related domains, is supported by an extensive literature foundation, further enriched by advanced data mining techniques. Therefore, emphasising secondary data is not merely a matter of convenience or cost-saving but is fundamental to ensuring the ontology's relevance, reliability, and authority (Morgan, 2022).

3.5.2 Design (Phase 2):

Formulation of Competency Questions: A set of competency questions was formulated based on the domain analysis. These were questions that the ontology should be able to answer once it is developed. This was important to ensure that the ontology would meet the needs of its intended users. These competency questions were focused on gait-related concepts and the relationships between them. They addressed the scope of the domain, the level of detail needed for the concepts and relationships, and the specific requirements of the domain. The competency questions were worded in everyday language and used to evaluate the comprehensiveness and accuracy of the ontology and to determine if the model could effectively represent the concepts and relationships relevant to the domain (Antia & Keet, 2021). Formulating the competency questions was informed by literature. Generally, by forming and answering these questions, the overall quality and validity of the ontology could be established and verified (Suarez et al., 2022).

Ontology Design: The subsequent step was to design the ontology. This included defining the classes and properties of the ontology and the relationships between them. The ontology was designed to accurately represent the concepts and relationships within the domain and must be consistent with existing ontologies and standards. Thereafter, the relevant terms and concepts were identified, and a conceptual model of the domain ontology was developed.

3.5.3 Development (Phase 3):

During this phase of developing the domain ontology, the goal was to engineer an ontology that could provide decision support for decision-making in healthcare. A specific tool, namely the Protégé ontology editor, was employed for the primary development of the domain ontology. Protégé was used to create the ontology from scratch and edit it as necessary. For visualising the ontology in a graphical format, integrated plugins within Protégé, such as OwlViz and OntoViz, were utilised. The Protégé editor was also used to check the ontology for errors and inconsistencies after it was developed. Secondary data sources were crucial in building the domain ontology for gait-related concepts. This process involved extracting data from various sources like medical databases, published articles, relevant books, and web portals into the ontology being developed. This step helped ensure that the ontology was comprehensive and correct and reflected the current state of knowledge in the domain. The process of developing the ontology typically involves the following steps:

Data selection: This involved identifying the most relevant, applicable, and trustworthy sources of data for elicitation.

Data extraction: This involved extracting relevant data from the selected sources, such as concepts, relationships, and instances, and then transforming these ontology components into a format that can be assimilated into the ontology (Yadav et al., 2016).

Data integration: Once the domain knowledge was gathered, the next step was to define the structure of the ontology. This involved incorporating the extracted data into the ontology, typically by adding new classes, properties, and individuals and specifying the relationships between them via properties assertion which is described in Table 3.1.

Table 3.1: Components of an ontology

Ontology Component	Definition
Classes or Concepts	These are the main building blocks of an ontology, representing the objects or ideas in the domain.
Properties or Attributes	These are the characteristics or features of the classes or concepts, describing what the objects or concepts represent.
Instances or Individuals	These are the specific examples or realisations of the classes or concepts in the domain, representing real-world objects or events.
Relationships	These are the connections or links between the classes, concepts, instances, or properties, defining the relationships between them, such as inheritance, association, or aggregation.
Axioms or Rules	These statements or conditions specify constraints, restrictions, or additional information about the ontology's classes, concepts, instances, or properties.

Data population: This refers to the process of adding specific instances to the already-defined ontology. These instances are concrete examples of the classes and their associated properties specified in the ontology schema. Populating the ontology with these instances was essential as it enables the system to perform reasoning and inferencing based on real-world data. In this study, data population was primarily conducted manually. The populated data enriches the ontology, helping it to represent specific entities within the domain and thereby facilitating more accurate and detailed queries and decision-making (Keet, 2020).

Engineering: Intended for ontology development, the initial step was to select an appropriate logic-based language tool. An array of tools is available to help with individual techniques or a specific method. Identifying the best tool for solving a problem is a highly valuable skill and essential for finding an effective solution. The choice was based on the necessary language features and automated reasoning needs, which aligned with the ontology's overall goal (Keet, 2020; Bravo et al., 2019).

Iteration: The ontology development phase relied heavily on iteration to refine and improve the ontology over time. In this context, iteration pertains to the iterative approach of designing, examining, and refining the ontology until it attains the desired degree of rigour and correctness. This process of iteration persisted until the ontology could precisely embody the knowledge domain and fulfill the criteria for its designated use cases. The number of iterations needed to accomplish this goal depends on factors such as the complexity of the domain, the required level of accuracy and precision, and the quality assessment gathered from validation and evaluation procedures (Shimizu, 2020). Moreover, this approach permitted the inclusion of evaluation and resolution outcomes, culminating in a more comprehensive and useful ontology, which ensured its suitability for the envisioned application. This was also where OD-101 methodology came into play, which offered a stepwise and systematic approach to ontology development, enabling the ongoing refinement and improvement of the ontology as new information became available. OD-101 offered an agile avenue to ontology construction, while it also provided a practical, user-centered perspective for ontology development.

Maintenance: The maintenance of the ontology was an essential aspect of ontology development that aimed to maintain its quality and facilitate its use. Once the ontology was developed and implemented, it required regular maintenance to ensure that it remained accurate and up to date with the latest knowledge in the domain. This step involved maintaining and updating the ontology to reflect new knowledge by adding new classes or properties or modifying existing classes or properties as needed. The maintenance process involved regularly updating the evolving modules to keep the ontology current. Furthermore, maintenance consisted of periodically checking the ontology for errors, inconsistencies, or outdated information, addressing any issues that arose and keeping it in sync (Keet, 2020).

3.5.4 Evaluation (Phase 4):

The research design process, culminated in the evaluation phase, ensured the development of a comprehensive and effective gait analysis domain ontology. The last step was to evaluate the ontology to ensure that it met the quality criteria of domain task fit, content richness, and correctness.

Assessing the ontology's ability to answer competency questions correctly ensured that the ontology accurately and completely represented the domain and was fit for its intended purpose. Evaluating the quality attributes of a domain ontology for gait analysis involved assessing if it is fit for the task at hand, its richness of content, and its operational correctness. Next, a check was done to evaluate the content richness based on standard metrics and to check for correctness with the ontology built-in reasoner. Content richness was evaluated using standard metrics acting as the number of classes and properties in the ontology. This helped to ensure that the ontology was detailed enough to be useful for the assigned tasks.

Checking for errors and inconsistencies was important to guarantee the correctness of the ontology.

In this evaluation phase, a multi-faceted approach was adopted to ensure the robustness and accuracy of the developed ontology. Initially, Description Logic queries were crafted to rigorously test the ontology. Leveraging the capabilities of Protégé, these queries were pivotal in determining if the ontology aptly answered domain-specific questions, thereby affirming its accurate representation of domain knowledge.

Following this, the ontology underwent a representation using First-order Logic. This step was crucial in ascertaining the ontology's expressiveness and semantic accuracy, adding an additional layer of validation to the process.

To further enhance the evaluation, SPARQL queries were employed, focusing on the validation of data within the ontology. This meticulous process ensured that the ontology's data remained consistent, precise, and aligned with real-world scenarios. The results derived from these queries were invaluable, providing insights on the ontology's practical applicability and usage.

Lastly, the ontology was subjected to an independent assessment using the FOCA evaluation methodology. This comprehensive evaluation, involving expert evaluators, centered on assessing the ontology's domain task fit, content richness, and overall correctness. The feedback garnered from this phase was instrumental, guiding the refinement of the ontology and ensuring its feasibility and robustness.

This resulted in an ontology that was a machine-understandable model of gait-related information, which can be used in applications for storing, providing automated access, and sharing a domain ontology for information and data dissemination to knowledge and domain experts (Fonou-Dombeu et al., 2021).

3.5.5 Conclusion (Phase 5):

Conclusion was drawn from the outcome of the evaluation of the developed gait analysis ontology in terms of establishing domain task fit, assessing content richness through standard metrics, and conducting error-checking with the built-in reasoner, in addition to using the FOCA methodology, independent evaluator feedback, description logics, and SPARQL queries dimensions that were evaluated.

The research design of this study is based on OD-101 and depicted in Figure 3.3. The mapping between the research objectives and the adopted research design is presented in Table 3.3.

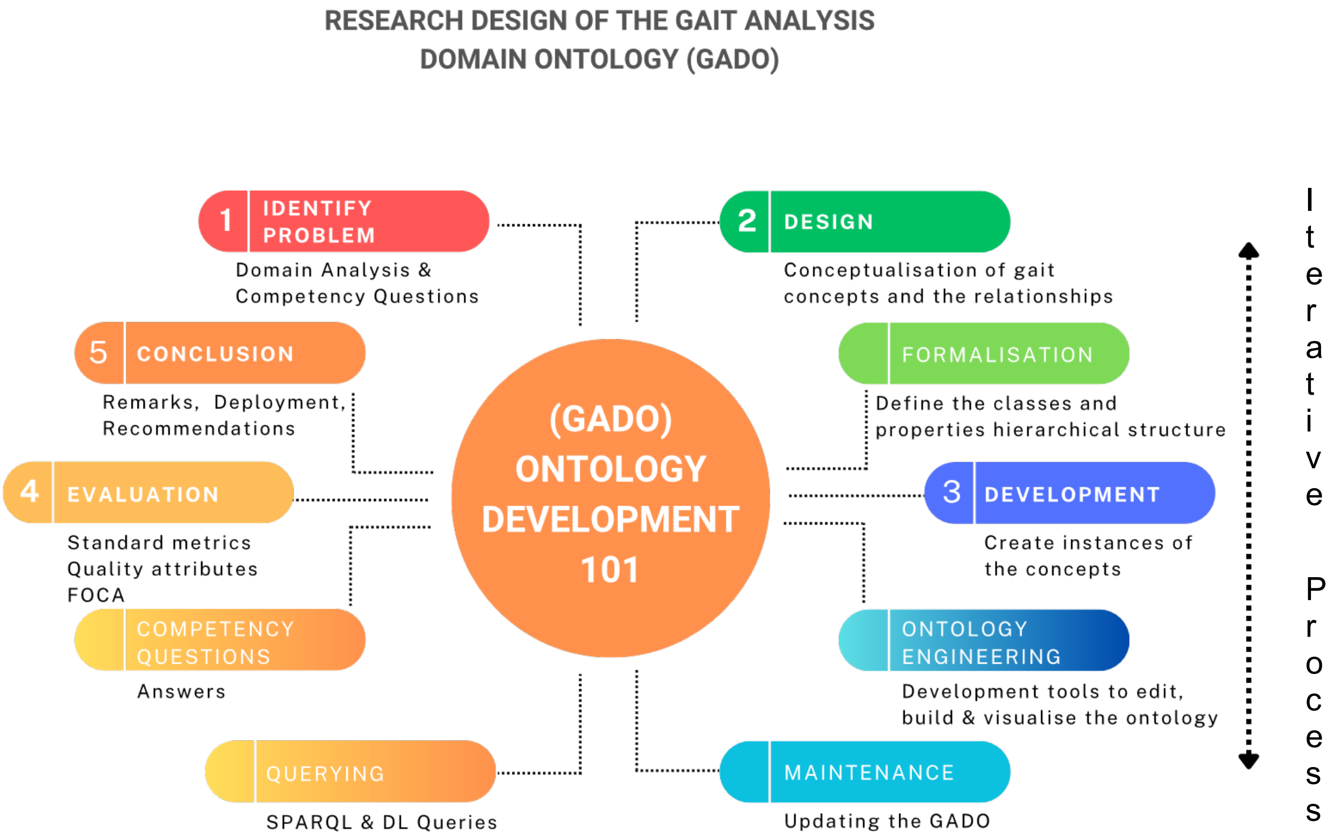


Figure 3.3: Research design process based on OD-101 methodology (Researcher, 2023)

Table 3.3: The research design process mapping

Research Design Phase	Objective	Methods/Activities	Output
Identify problem	i. Identify the requirements of a domain ontology that can support decision-making in the treatment of gait-related diseases.	Elicitation of key concepts & relationships • Online medical database: Web-based • Document Review: Book Publications • Literature review: Scientific journals & articles	**Domain Analysis** Competency Questions
Design	ii. Design a domain ontology that can support decision-making in the treatment of gait-related diseases.	Conceptualise the ontology domain using ontology web language (OWL) to model and formalise the ontology.	**Ontology Design**
Development	iii. Develop a domain ontology to support decision-making in the treatment of gait-related diseases.	OD-101 methodology with ontology developing tool (Protégé) for ontology creation and development.	**Ontology Development**
Evaluation	iv. Evaluate the domain ontology in terms of domain task fit, content richness, and correctness.	Establish domain task fit from defined classes, and the answering of competency questions. Evaluation of standard metrics to determine content richness and conclude error checking for correctness with the built-in ontology reasoner(s). The FOCA methodology was employed to assess the ontology, using feedback from independent evaluators for evaluation and verification. Description logics and SPARQL queries were also used for validation.	**Ontology Evaluation**
Conclusion	Derive conclusion from the artefact.	Final remarks and recommendations were derived for adopting the artefact and research findings to check if the ontology can be improved and refined.	**Artefact, Thesis, Articles**

3.6 Research ethics

The research ethics that were considered in the study included:

Informed consent: Sufficient information and assurances about the study were conveyed, accompanied by a consent form to the participants before data collection. In the evaluation phase of the ontology, participants had the right to withdraw from the study at any time and anonymity was ensured. There was no patient involvement in the study.

Data Privacy and Confidentiality: All third-party and proprietary software licenses, data and information were sourced lawfully and used within the legal parameters.

Dissemination of Research: Non-sensitive data for public use, which was relevant to the study, were obtained from secondary data sources.

3.7 Summary

This chapter presented the OD-101 methodology, the adopted research design for this study's execution. The philosophical foundations, including ontological and epistemological commitments, were outlined as they influenced the conceptualisation of the research problem and interpretation of potential results.

The chapter also described the multi-step process planned for the development of the domain ontology. This process was designed to encompass scope identification, knowledge gathering, structure definition, and the proposed evaluation methods for assessing the ontology's accuracy and effectiveness. A systematic and iterative approach to data selection, extraction, integration, validation, and maintenance was described as part of the OD-101 methodology.

CHAPTER 4: ONTOLOGY DESIGN AND DEVELOPMENT

In this chapter, a comprehensive guide to the process of developing the domain ontology for gait-related diseases is presented. The chapter describes how the steps of the Ontology Development 101 (OD-101) methodology were followed to develop the gait analysis ontology. The chapter begins by discussing the domain analysis, which involves identifying relevant concepts, relationships, and entities in the domain.

The development process was facilitated by:

4.1 Use Case of the Gait Analysis Domain Ontology (GADO)

The GADO aims to facilitate better information sharing and decision support for gait-related issues. Thus, its scope in terms of structure, classes, properties, and individual instances must adequately cover the knowledge that exists in the domain. In order to effectively comprehend the GADO, it is important to understand the various classes, their properties, and how they relate to one another. This includes querying the ontology and extracting relevant information for specific use cases. Figure 4.1 shows the different use cases of the GADO in terms of the intelligent operations that it should support. The description of use cases is shown in Table 4.1.

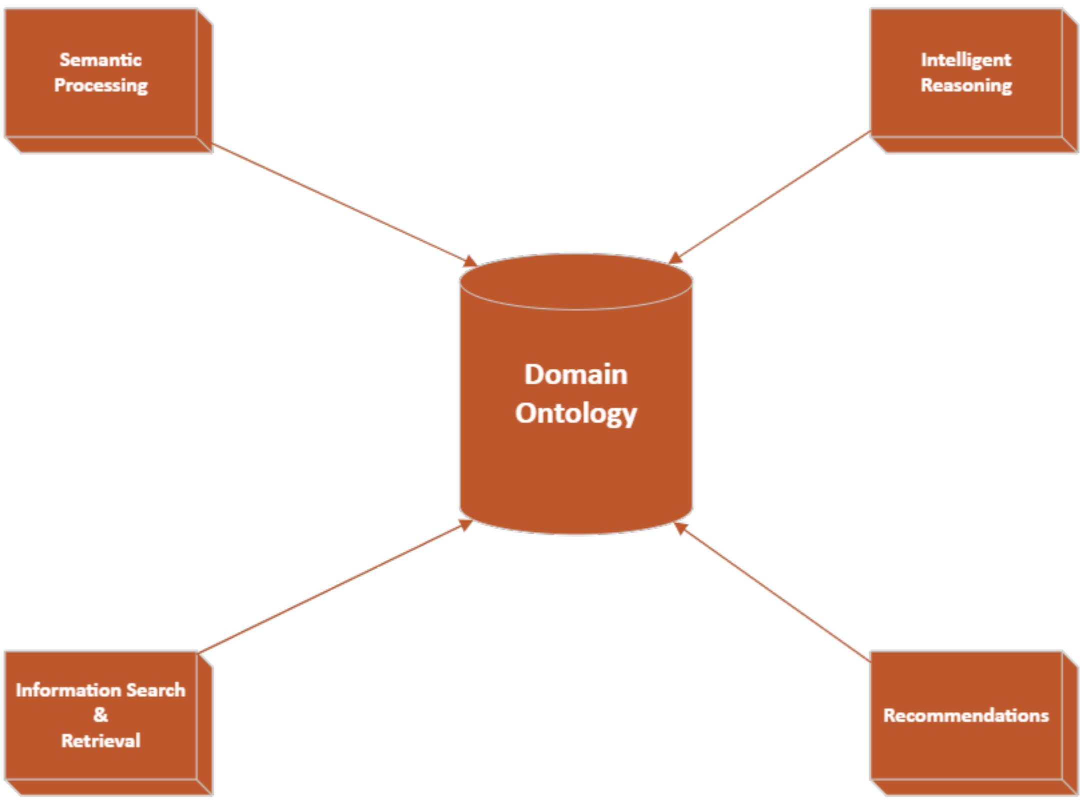


Figure 4.1: Use case of the GADO (Researcher, 2023)

Table 4.1: Typical use cases of the Gait Analysis Domain Ontology

Use case	Narrative
Semantic Processing:	To define and structure semantic relationships in a shared vocabulary, for gait-related diseases, via semantic web technologies to users.
Intelligent Reasoning:	To provide reasoning about facts and inferencing capabilities for clinical gait analysis.
Information Search & Retrieval:	To enable domain searching functionality on gait-related issues and facilitate domain knowledge information retrieval.
Recommendations:	To recommend information based on predefined gait-related data using rules and cases.

4.2 Domain analysis

Through the literature review, the requirements of a domain ontology for gait-related matters were identified. These requirements included the need for standardised terminology and concepts, the ability to support decision-making, and the ability to integrate with other healthcare innovations. The development of the GADO started with domain analysis which consists of steps that are presented in the sequel sections.

4.2.1 Identify the scope

The first step was to identify its scope. This involved defining the boundaries of the ontology, including the concepts and relationships that were included and those that were excluded.

4.2.2 Gather domain knowledge

The next step was to gather domain knowledge from appropriate sources, like scientific literature, medical journals, and existing ontologies. This step was important to ensure that the ontology was comprehensive, accurate and well-suited. The subsequent step involved retrieving the information from the identified and described sources. This included searching for relevant literature, consulting online databases in the field of gait-related subject matters, and reviewing existing ontologies related to the domain. The information gathered during this step was vital in establishing a solid foundation of knowledge upon which to build the ontology. By gathering domain knowledge from various sources, the researcher ensured that the resulting ontology was robust and reflective of the current state of knowledge in the domain.

Historically, achieving a comprehensive understanding of a specific disease or disorder involved referencing medical books. However, this approach was subject to several significant limitations, including outdated information, limited accessibility in non-digital formats, and books being primarily designed for academic study rather than data mining functions. In contrast, the World Wide Web presented a potentially valuable resource for accessing available, reliable, and factual information to anyone with an internet connection.

In the development of the domain ontology, a targeted search of specific web-based ontologies and knowledge sources was conducted to acquire data related to gait-related health matters. These sources were not integrated in real-time but served as valuable repositories from which relevant data was manually extracted and included in the ontology. The following online knowledge portals were consulted:

i. OMIM® An Online Catalog of Human Genes and Genetic Disorders,
ii. PubMed,
iii. The Human Phenotype Ontology, and
iv. EMBL-EBI Ontology Lookup Service.

i. OMIM® (Online Mendelian Inheritance in Man)

This is an online catalogue of human genes and genetic disorders developed and maintained by the National Center for Biotechnology Information (NCBI). The catalogue contained detailed information about known inherited diseases, including information about their allele frequencies, inheritance patterns, associated phenotypes, and links to related medical articles. OMIM® was a valuable resource for researchers and clinicians, as it provided detailed information about genetic diseases that can be used to diagnose and treat patients. See the snapshot example for OMIM® in Figure 4.2 below when searching for gait-related matters.

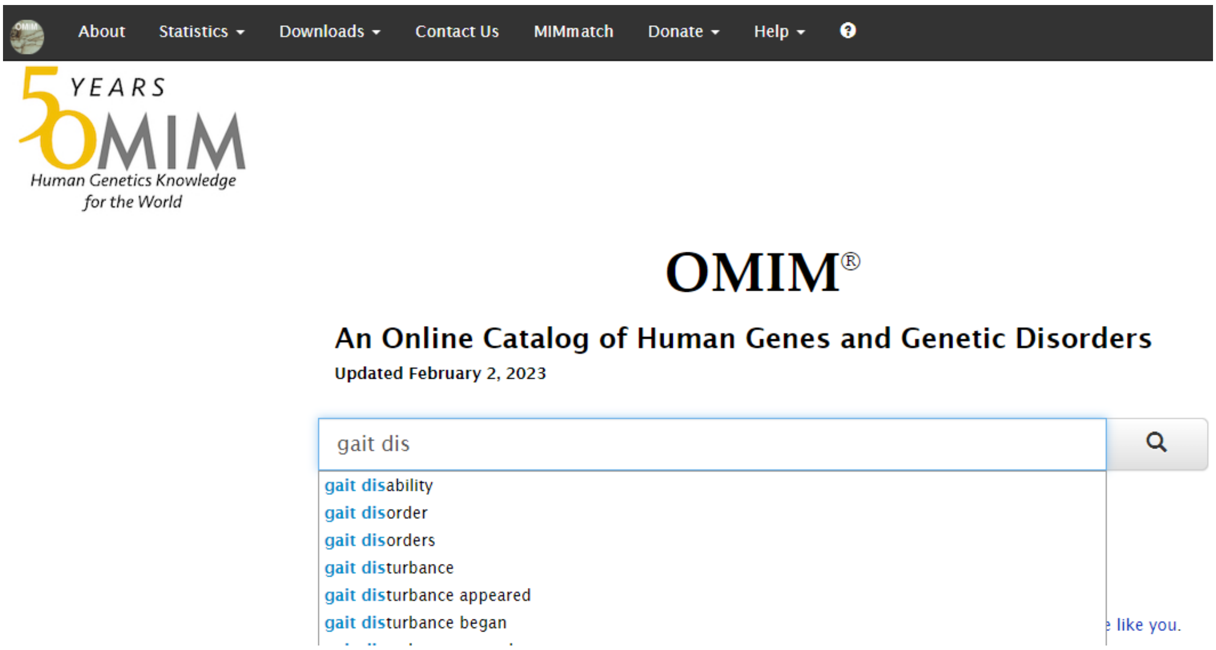


Figure 4.2: OMIM® An Online Catalog of Human Genes and Genetic Disorders (OMIM, 2022)

ii. PubMed

Pubmed is a free online database of biomedical and life Sciences journal articles maintained by the U.S. National Library of Medicine (NLM). PubMed includes over 29 million citations from MEDLINE and other life Science journals, as well as books, reviews, and reports. PubMed is an invaluable resource for researchers, clinicians, and the general public, as it provides access to a vast database of medical literature. The Pubmed online interface is shown in Figure 4.3 below.

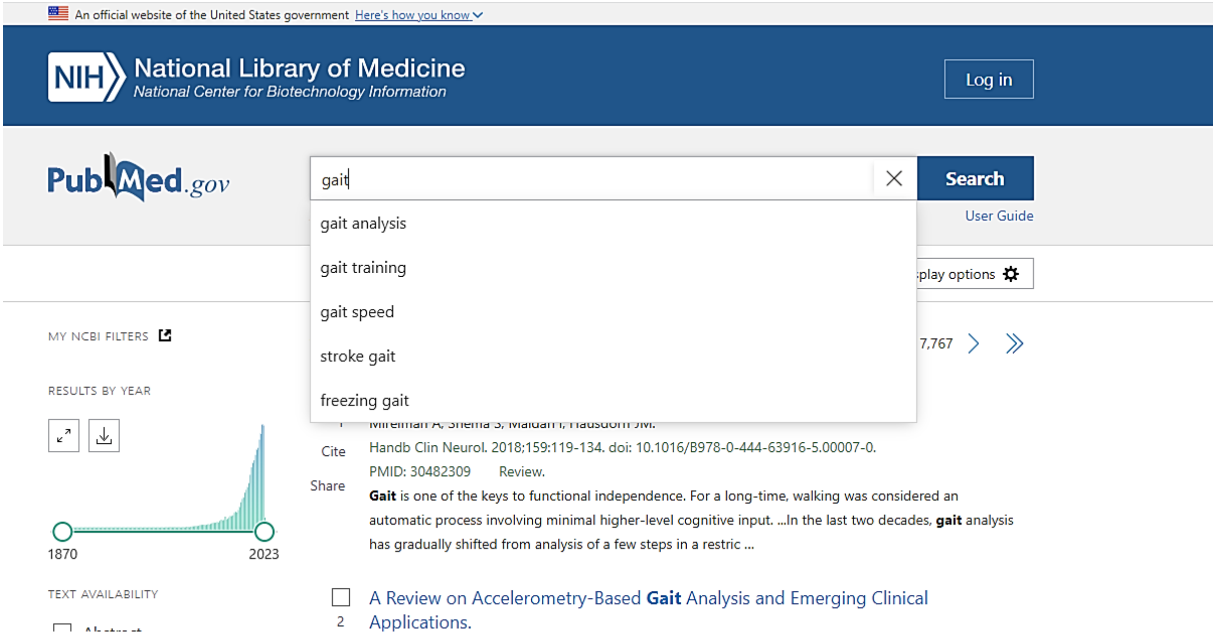


Figure 4.3: Pubmed (Pubmed, 2022)

iii. The Human Phenotype Ontology (HPO)

This is an online database of phenotypic information related to human diseases. This is typically a structured vocabulary used to describe the phenotypic features associated with a disease or disorder. The HPO database contained over 11,000 terms related to human phenotypes, including anatomical, clinical, and pathological features. The HPO database is currently maintained by the Phenotype and Disease Ontology (PDO) project at the University of Edinburgh and was freely available for use in research and clinical practice, as depicted in Figure 4.4 below while searching for gait disturbance(s).

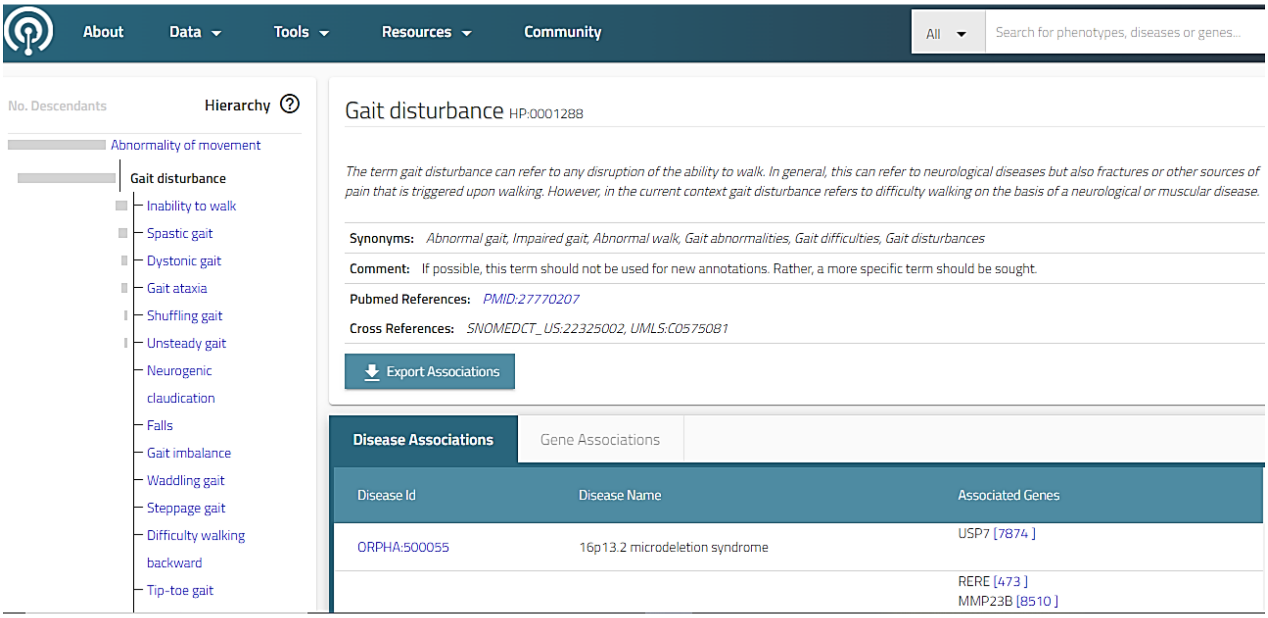


Figure 4.4: The Human Phenotype Ontology (Human Phenotype Ontology, 2022)

iv. The EMBL-EBI Ontology Lookup Service (OLS)

The OLS is an online tool that provides access to multiple ontologies, including the Gene Ontology (GO), the Human Phenotype Ontology (HPO), the Mammalian Phenotype Ontology (MPO), and the Cell Ontology (CL). OLS enabled users to search for terms, view relationships between terms in an ontology, and explore the ontology hierarchy. OLS also provided access to multiple ontology databases, like the UniProt Knowledgebase and the NCBI Taxonomy database. OLS was a valuable source as it provided access to multiple ontologies that can be exploited to understand the biological context of diseases and aid in developing diagnostics and treatments. Figure 4.5 illustrates OLS.

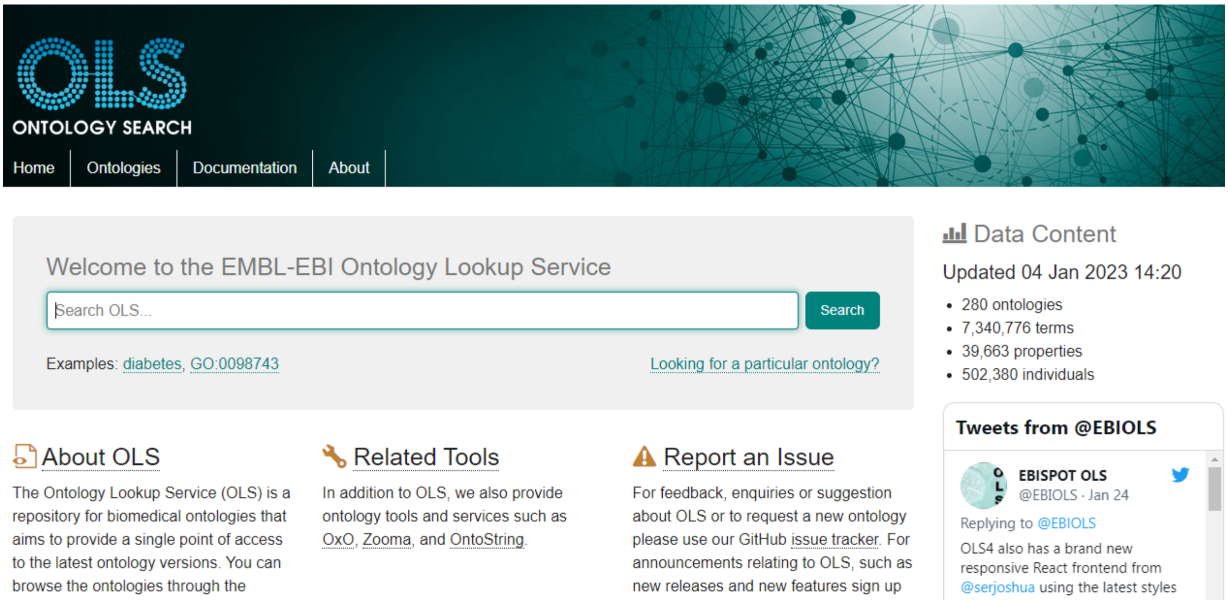


Figure 4.5: EMBL-EBI Ontology Lookup Service (EMBL-EBI, 2022)

In order to identify diseases across different sources of information, it was necessary to map each disease to the Human Disease Ontology in an effort to obtain disease formalities for the vocabularies such as OMIM® or Medical Subject Headings (MeSH). Using multiple vocabularies allowed for the highest amount of means of identification. An example of this was extracting all the 97 PubMed articles associated with each of the identified gait-related diseases. (Refer to **Appendix D**).

MeSH (Medical Subject Headings) is an indexing system used by the National Library of Medicine (NLM) to classify articles in the fields of medicine and health. Principally it is used to find and organise information on medical topics in databases functioning as MEDLINE, PubMed, and the NLM Catalogue. The MeSH system is hierarchical and organised into 15 main categories, each with its own set of subcategories. Each subcategory was further divided into terms and descriptors, and the terms were organised into a hierarchy to help users find the most relevant information. MeSH provides a comprehensive indexing system for medical and health-related topics, making it a useful resource for anyone interested in medical research. See Figure 4.6 below for the online web MeSH interface.

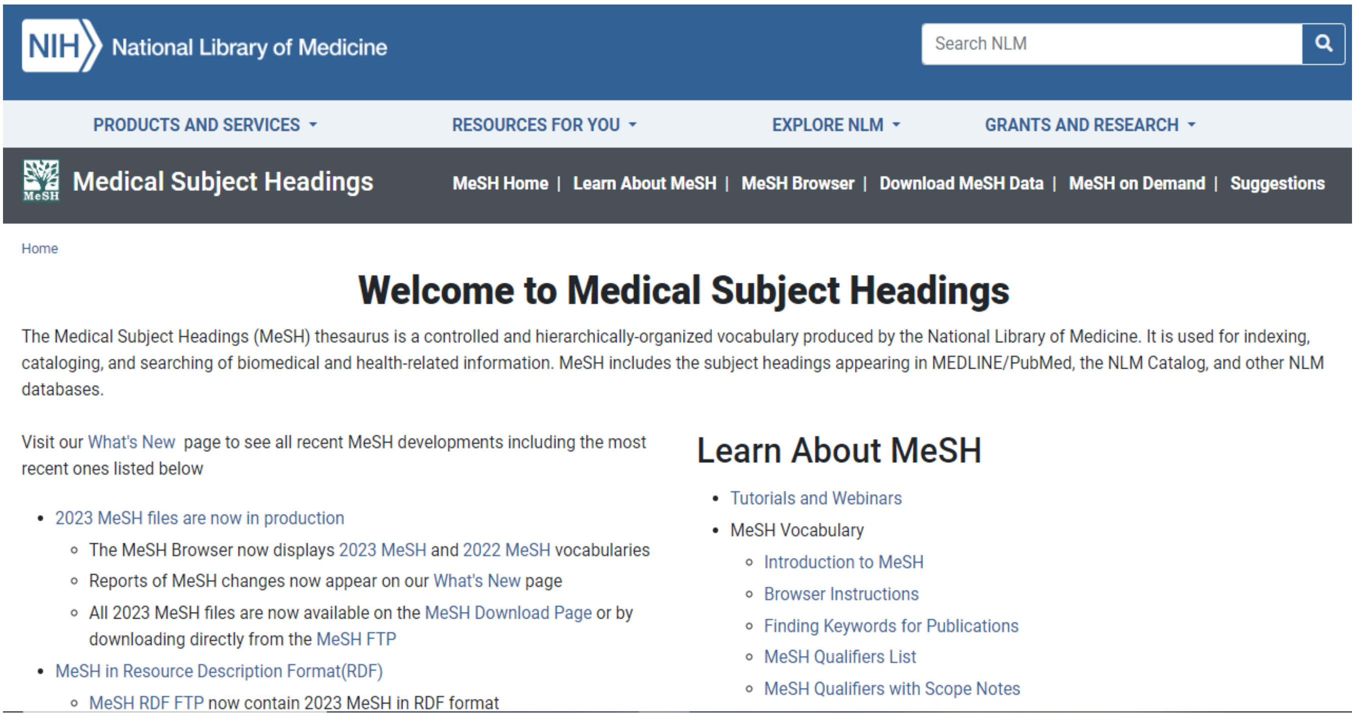


Figure 4.6: MeSH (Medical Subject Headings, 2023)

The "Web Crawler", a bot that is systematically operated by search engines like Google to index content across the internet, was responsible for collecting and extracting specific text and references related to phenotypic manifestations of diseases from the hyperlinks in a results list. This component also endeavoured to gather information regarding disease coding systems; these codes were used to identify diseases in various databases and data vocabularies. The output of this component was a refined set of validated terms that were considered to be the true phenotypic manifestations of diseases. In an attempt to enhance accessibility and usability, the extracted information was made available through the Entrez search interface. This interface offered various options for conducting precise searches, such as configurable facet filters and an advanced search interface and managing results. A facet filter is a type of filter used in search engines or databases that allows users to narrow down search results based on specific attributes or characteristics, known as facets. Entrez is a text search and retrieval system provided by the National Center for Biotechnology Information (NCBI) that integrated the biomedical literature database PubMed, as illustrated in Figure 4.7 below.

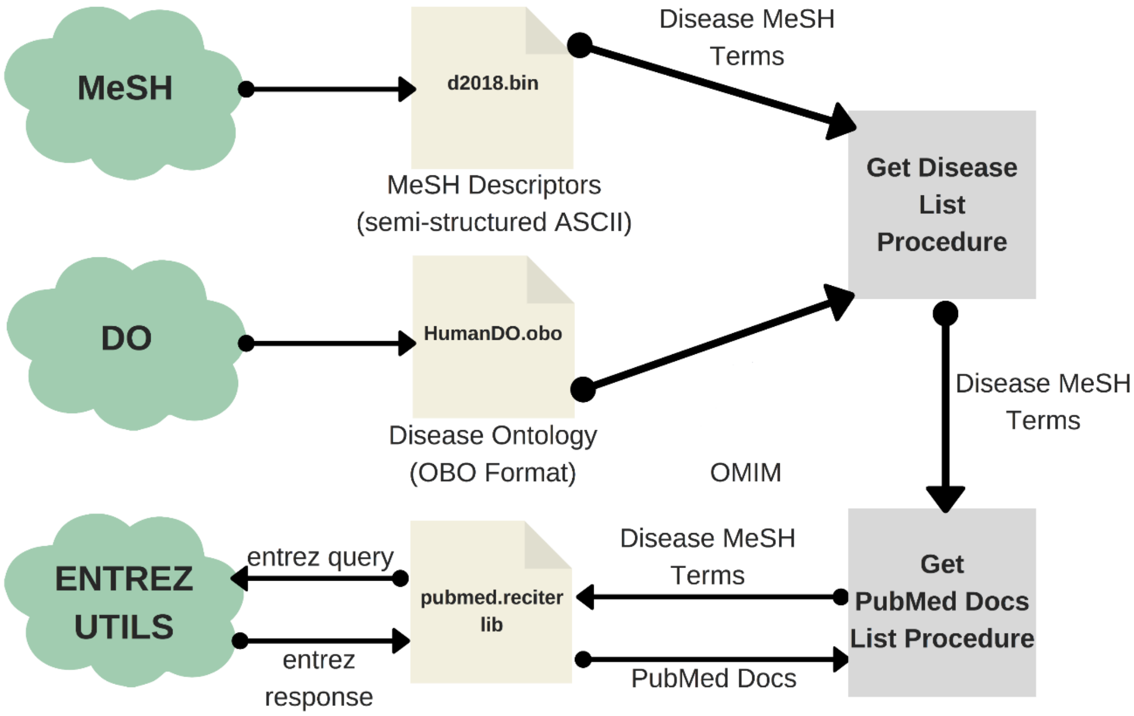


Figure 4.7: (Adapted) Web crawling for gait-related data (Lagunes-Garciá et al., 2020)

In the process of building an ontology for gait analysis, several reputable web sources were consulted to gather comprehensive and authoritative information on the topic of gait-related diseases. Among these, the Human Phenotype Ontology (HPO) emerged as the primary resource, providing a wealth of standardised vocabulary and phenotypic abnormalities related to human diseases. HPO's structured approach to annotating patient phenotypes proved invaluable, facilitating a deeper understanding of gait disturbances specifically, and their underlying genetic and clinical implications. While other sources such as OMIM®, PubMed, and EMBL-EBI Ontology Lookup Service offered significant insights into genetics, biomedical literature, and ontology visualisation respectively, it was the HPO that contributed most of the data used in the domain ontology construction. The depth and specificity of information available in HPO made it an indispensable tool in this endeavour.

In addition to the online web-based data sources mentioned above, three published books were also reviewed to provide universal and scientific information to construct a domain ontology with gait-related themes. These books were remarkably useful and highly instrumental sources, which supplemented and enriched the scope as well as requirements for the domain and ontology development. The literature publications that were reviewed are:

An Introduction to Gait Analysis, 4^{th} edition (Whittle, 2014)
An Introduction to Ontology Engineering v1.5 (Keet, 2020)
Handbook on Ontologies, 2^{nd} edition (Staab & Studer, 2009)

v. An Introduction to Gait Analysis, 4^{th} edition

This book publication provided a comprehensive overview of the fundamental principles and techniques used in analysing human gait. It covered the anatomy and biomechanics of the lower limb and the gait cycle, as well as the methods that were used to quantify and analyse gait, in the manner of motion analysis and gait classification. The book also included descriptions of the latest advances in technology for monitoring gait and discussed how these advances could be used in research and clinical settings. This information was essential for understanding the basics of gait analysis, a crucial aspect of studying gait-related diseases (Whittle, 2014).

vi. An Introduction to Ontology Engineering v1.5

This book provided a detailed guide to ontology engineering, which is the practice of creating and managing ontologies. The book covered the fundamental concepts, methods, and tools used in ontology development and covered extensive topics like ontology design, alignment, and evaluation. It also covered more advanced topics like semantic web technologies, ontology languages, and ontology reasoning and included complete examples of creating, modifying, and publishing ontologies. The book provided an accessible introduction to the field of ontology engineering and was an important data source (Keet, 2020).

vii. The Handbook on Ontologies, 2nd edition

This is an edited volume by Steffen Staab and Rudi Studer. This book contained a comprehensive collection of chapters exploring the use of ontologies in a variety of applications, from the semantic web and knowledge engineering to natural language processing and artificial intelligence. The book included chapters from top experts in the field and provided an in-depth overview of the current state of ontology engineering, as well as the challenges and opportunities of using ontologies in the future. The book also covered a wide range of topics related to ontologies, including their design, development, evaluation, and application. Furthermore, it discussed various ontology languages, tools, and methodologies, as well as emerging trends and challenges in the field (Staab & Studer, 2009).

4.2.1 Formulation of competency questions

Based on the domain analysis, a set of competency questions was formulated. These were questions that the ontology was expected to answer once it was developed. By implication, they also represent the requirements that the domain ontology must satisfy. The questions needed to be specific to the domain of gait-related diseases and were designed to assess the ontology's capability to provide information to support research and clinical decision-making in this field. The competency questions are listed in Table 4.2.

Table 4.2: Competency Questions

Q1	Who are the users of the domain for gait-related diseases?
Q2	What gait-related diseases are linked with geriatric (elderly) patients?
Q3	What are the most common types of gait-related diseases?
Q4	What are the typical neurological gait-related diseases that impact patients' gait?
Q5	Is there sufficient detail to differentiate between different types of gait-related diseases?
Q6	What current gait assistive technology solutions are available for gait disorders?
Q7	What types of non-pharmacological treatments are available for gait-related diseases?
Q8	To what extent can a decision support system for gait-related issues be useful/beneficial?
Q9	Who are the key role players in the execution of a gait-based process?
Q10	What are the types of gait disturbance that are generally associated with children?
Q11	What are the resources that require gait analysis?
Q12	What are the main etiological anatomies for gait pathologies?
Q13	What are the different gait dimensions for the domain?

The first step in establishing a domain ontology for gait-related diseases was to determine the core concepts involved in the biomedical domain. This involved compiling a comprehensive list of important terms in the field and collecting domain concepts, semantics, attributes, and standards. After sorting and refining the collected data and gathered information, a concept review table was established to provide a clear overview of the domain ontology. This was essential to ensure that the core concepts were explicit and covered the entire domain knowledge related to gait-related diseases. The process of eliciting secondary data sources was conducted to identify the key ontology concepts that were relevant to and representative of the development of a gait-related domain ontology.

The activities aligned with common steps in the Ontology Development 101 methodology, which is a foundational approach to creating ontologies in a structured, logical manner. Here is how each part aligns with Ontology Development 101 methodology:

Establishing core concepts

The first step: Determining the core concepts in the biomedical domain of gait-related diseases corresponds to the activity of defining the domain and scope of your ontology. Typically, this involves answering competency questions the ontology should address.

Compiling a list of terms

Collecting important terms, semantics, attributes, and standards corresponds to the identification and collection of terms relevant to the domain. This is often considered an early and crucial stage in ontology development.

Concept review table

Creating a concept review table to sort and refine the data aligns with activities related to documenting the ontology. This is key for ensuring consistency and for facilitating subsequent stages of ontology development, such as formal definitions and relationships among the terms.

Identifying secondary data sources

Eliciting secondary data sources to identify key ontology concepts can be considered part of the research phase, where existing databases, web portals, or medical publications were consulted to ensure that the ontology is robust, accurate, and comprehensive.

Therefore, thirteen (13) main concepts were identified from the requirements and competency questions and were considered necessary for the scope and coverage of the domain ontology for gait-related diseases, as outlined in Table 4.3.

Table 4.3: Gait dimensions

No.	Core Concept	Dimension description
1.	**Gait-based Process**	This concept refers to the process by which movement is executed by the human body during walking or running for assessment.
2.	**Gait-related Disease**	This refers to any medical condition that can affect an individual's gait.
3.	**Gait Analysis Equipment**	This includes the equipment and resources used to analyse and measure the gait-based process.
4.	**Gait Analysis Method**	This refers to the procedures and techniques used to assess and evaluate the gait-based process.
5.	**Gait Assistive Technology**	This includes any devices, treatments, or technologies that are used to support and rehabilitate abnormal gait.
6.	**Gait Classification**	It involves analysing and understanding the distinctive characteristics of a person's gait, such as stride length and cadence, to classify abnormalities or pathologies.
7.	**Gait Cycle**	This concept refers to the complete cycle of movement executed by the human body during walking or running.
8.	**Gait Disturbance**	This refers to any disruption or deviation from normal gait.
9.	**Gait Measurement Point**	Gait measurement points are physical locations that analyse and measure how a person walks or moves.
10.	**Gait Parameters**	This refers to any quantifiable aspect of the gait process, such as speed, step length, and width.
11.	**Gait Pathology**	This refers to any abnormal condition or disease that affects a patient's gait.
12.	**Type of Gait**	This refers to abnormal or improper gait categorised in different type(s) of gait based on the symptoms or appearance.
13.	**Gait Person**	This refers to the individual who is involved in the gait-based process, being a gait patient and a gait expert.

4.3 Design of the ontology

4.3.1 Conceptually define the ontology structure

Once the domain knowledge was gathered, the next step was to define the structure of the ontology. This involved creating classes, properties, and individuals and defining the relationships between them. The design of the ontology was formulated based on the identified requirements.

The core concepts of the ontology offered a complete and systematic representation of the domain for gait-related diseases and laid the foundation for the development of a robust and comprehensive domain ontology as conceptualised in Figure 4.8 below. Every single focus area or concept was typically assigned its own class. The greater knowledge area was defined as the main class, based on its level of abstraction, while the lower-level knowledge point was considered a subclass. Once the set of concepts was identified, they were organised into a structure that reflected their attributes and relationships between them. This was also where concept clustering came in. Concept clustering consisted of grouping related concepts into classes, each representing a group of interrelated concepts. The name of each class was assigned in a singular format and served as the class identification.

Figure 4.8: Gait dimensions covered in GADO (Researcher, 2023)

4.3.2 Conceptual model of the Gait Disturbance class

In an effort to enhance the ontology's conceptualisation, it was beneficial to introduce additional subclasses that capture specific gait disturbances associated with particular medical conditions or injuries. In the context of gait analysis domain ontology design, the conceptual class of ‘Gait Disturbance’ served as a broad category encompassing any deviations from normal walking patterns. The conceptual model provides a high-level representation of abnormal gait in various individuals for example “Gait apraxia”, “Loss of ambulation” and “Gait imbalance” as shown in Figure 4.9. This approach allows for a more comprehensive and detailed representation of gait disturbances within the ontology.

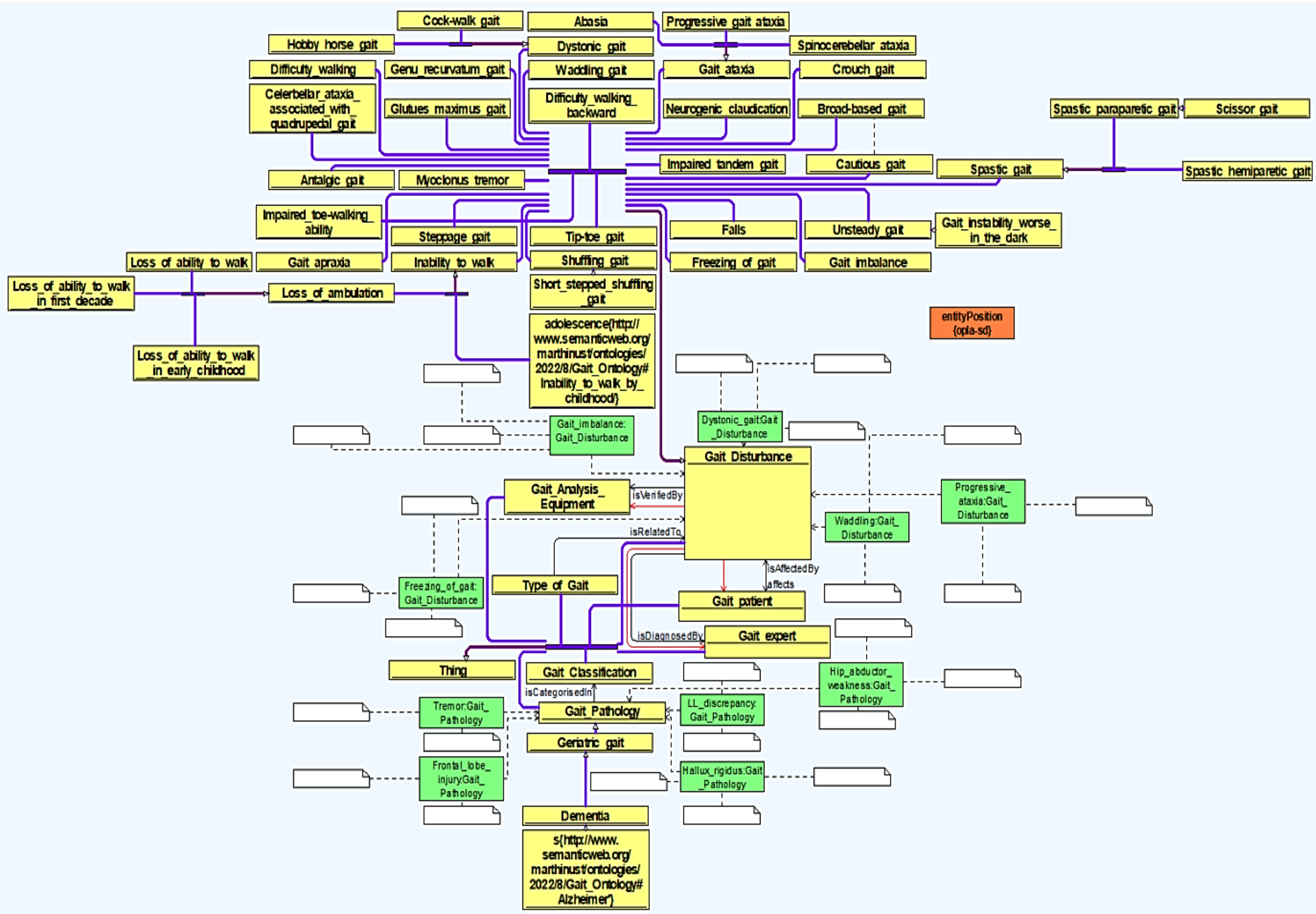


Figure 4.9: Conceptual model of the ‘Gait disturbance’ class

4.3.3 Conceptual model of the Gait expert class

The ontology for gait analysis aims to establish a structured knowledge base that aids in diagnosing and treating gait-related diseases. A pivotal class in this ontology is the 'Gait expert', which acts as an agent interacting with various aspects of the gait analysis domain. The ‘Gait expert’ class is an important concept in the Gait Analysis Domain Ontology because it represents the individuals who have the expertise to diagnose and treat gait-related diseases. The ‘Gait expert’ class includes instances like the Biomechanist, Orthopedist, Healthcare Practitioner, Podiatrist, and Physical Therapist.

The ‘Gait expert’ class exhibits the following key object properties, each defining a specific relationship with another concept in the ontology:

i. **Administers**: Gait expert → Gait-based Process
The Gait expert administers various gait-based processes like gait assessments or analysis.

ii. **Facilitates**: Gait expert → Gait Analysis
Involved in facilitating comprehensive gait analyses, possibly through the use of specific gait analysis equipment and gait analysis methods.

iii. **Analyses**: Gait expert → Gait Data
Responsible for examining gait data acquired from gait analysis activities.

iv. **Observes**: Gait expert → Gait Cycle
The Gait expert observes gait cycles to identify abnormalities or confirm normal gait patterns.

v. **Prescribes**: Gait expert → Gait Assistive Technology
Based on the analysis, the expert may prescribe assistive technologies like braces or walking aids.

vi. **Treats**: Gait expert → Gait patient
Ultimately, the Gait expert is involved in treating patients with gait abnormalities, overseeing their progress, and adjusting treatment plans accordingly.

Figure 4.10 illustrates a conceptual diagram of the 'Gait expert' class and how it relates to other classes within the GADO, such as 'Gait-based Process', 'Gait Analysis', and 'Gait patient' as previously mentioned.

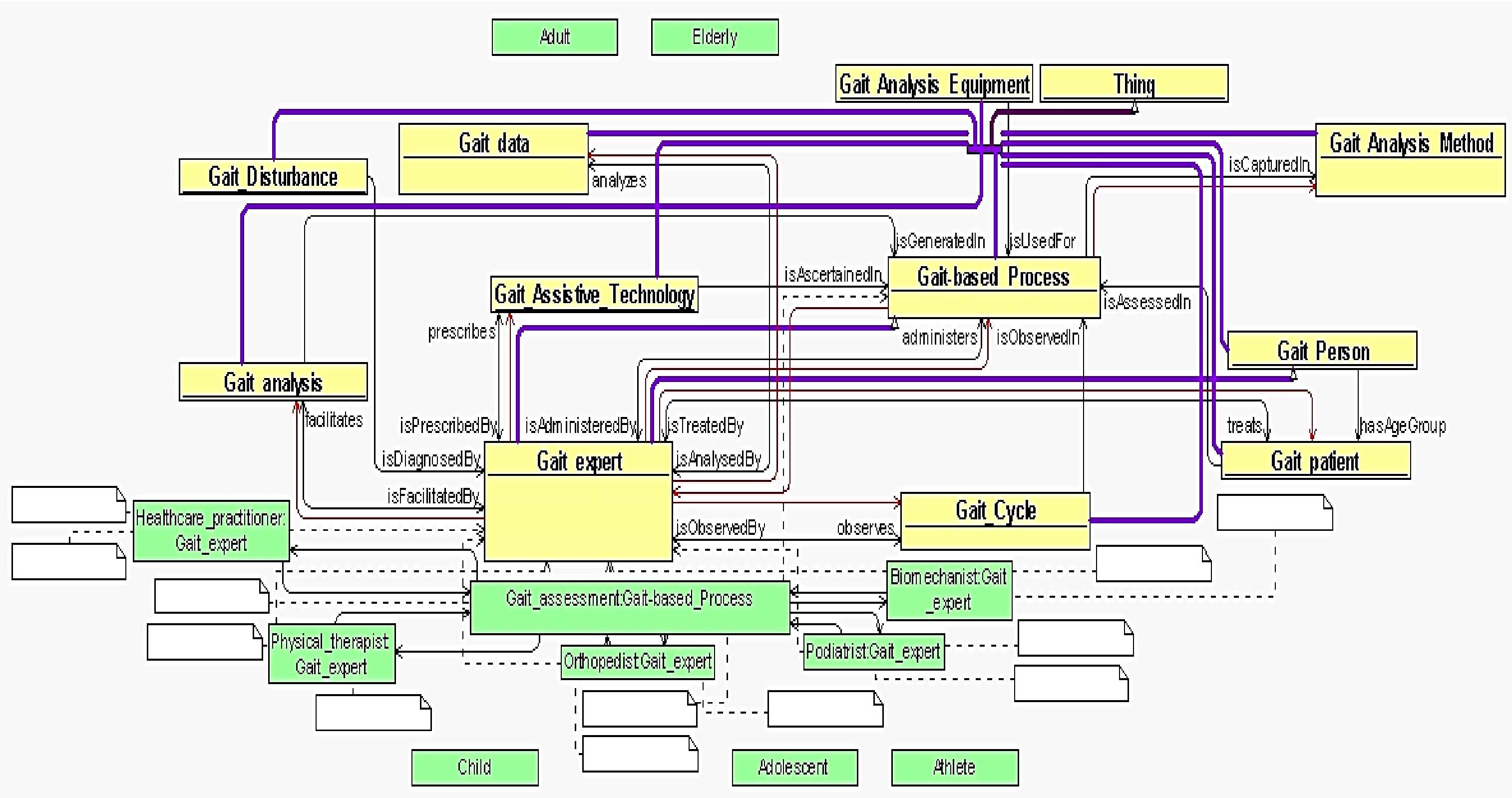


Figure 4.10: Conceptual model of the 'Gait expert' class

4.3.4 Formalising the ontology

Formalising the ontology involved translating the domain-specific conceptualisation into an unambiguous, machine-readable format. The formalisation of the ontology served to delineate explicit definitions for core concepts and to specify the permissible types of relationships among them. Utilising a formal language like the Web Ontology Language (OWL), ensured that the ontology not only provides a common vocabulary but also supports automated reasoning for gait analysis. The resultant formalised ontology facilitates machine-based inferences, thereby enhancing the objectives of the Gait Analysis Domain Ontology. Using OWL to formalise the ontology brings several key benefits:

i. **Clarity:** OWL offers a straightforward way to define concepts and their relationships, reducing ambiguities.
ii. **Consistency:** The ontology can be checked for errors, ensuring its reliability.
iii. **Interoperability:** Being a standard language, OWL ensures the ontology can integrate with other systems.
iv. **Reasoning:** OWL allows for automated insights extraction from the ontology.

Based on the core concepts for the Gait Analysis Domain Ontology, a more enriched formalised ontology involves specifying classes and subclasses as shown in Table 4.4, and then how the object properties relate to these classes.

Table 4.4 Classes and some of the subclasses

Classes	Subclasses
Gait-based Process	Gait analysis, Gait data, etc.
Gait-related Disease	Parkinson's disease, Hemiplegia, etc.
Gait Analysis Equipment	Force plates, Electromyography, etc.
Gait Analysis Method	Motion systems, Kinematics, etc.
Gait Assistive Technology	Braces, Walking stick, etc.
Gait Cycle	Stance phase, Swing phase, etc.
Gait Classification	Musculoskeletal, Neurological, etc.
Gait Disturbance	Gait ataxia, Falls, etc.
Gait Measurement Point	Foot progression angle, Stride length, etc.
Gait Parameters	Speed, Cadence, etc.
Gait Pathology	Pathological gait and Geriatric gait
Type of Gait	Antalgic gait, Ataxic gait, etc.
Gait Person	Gait patient and Gait expert

Below are examples that attempt to articulate the relationships between some of these classes in Table 4.5. These are just snapshot examples of the relationships that exist between some of the classes through object properties in the GADO.

Table 4.5 Some of the object properties relationships

isDetectedIn: Type of Gait → Gait Analysis Method Meaning: A Type of Gait is detected in a particular Gait Analysis Method.
isMonitoredBy: Gait Patient → Gait Analysis Equipment Meaning: A Gait patient is monitored by specific equipment.
affects: Gait Disturbance → Gait patient Meaning: A Gait Disturbance negatively impacts the Gait patient.
isAssignedWith: Gait Person (subtype: Gait Patient) → Gait Assistive Technology Meaning: A Gait Assistive Technology aids the Gait Patient.
isGeneratedIn: Gait Analysis → Gait-based Process Meaning: Gait Analysis is generated in the Gait-based Process.
isAdministeredBy: Gait-based Process → Gait expert Meaning: The Gait expert administers the Gait-based Process.

4.4 Development of the ontology

4.4.1 Ontology development tool

Protégé 5.5.0 was used to create the classes, properties, and individuals representing the concepts, relationships, and instances within the domain of gait analysis.

Protégé is a widely recognised and reliable tool for creating and managing ontologies. Protégé offers a user-friendly graphical interface that simplifies ontologies' creation, editing, and visualisation as an open-source platform. Thus, Protégé was an ideal choice to support OWL as the preferred ontology language and tool for modelling the domain ontology for gait-related diseases. Protégé provided a structured and machine-readable format for knowledge representation, making it well-suited for domain ontology development.

In addition to generating classes, properties, and relationships between classes, Protégé also enabled the inclusion of annotations and labels to enrich the terms and concepts. This feature improved the ontology's readability and supplied supplementary information regarding the terms and concepts.

4.4.2 Creating the ontology class hierarchy

In the context of gait-related diseases, classes for various gait-related diseases, like Parkinson's disease, Neuropathy, and Hemiplegia, were created in the ontology, as well as classes for related core concepts like ‘Gait Cycle’, ‘Gait Disturbance’, ‘Type of Gait’, et cetera. This led to the creation of a hierarchical structure in which each class or subclass embodied a distinct concept or knowledge point associated with the domain.

Figure 4.11 shows the class hierarchy of the core concepts for the domain ontology, with the ‘Gait-related Disease’ class expanded to offer a more detailed view of the sub-concepts (subclasses) for gait-related diseases. This expansion enabled a clearer understanding and easier navigation of the concepts pertaining to gait-related diseases.

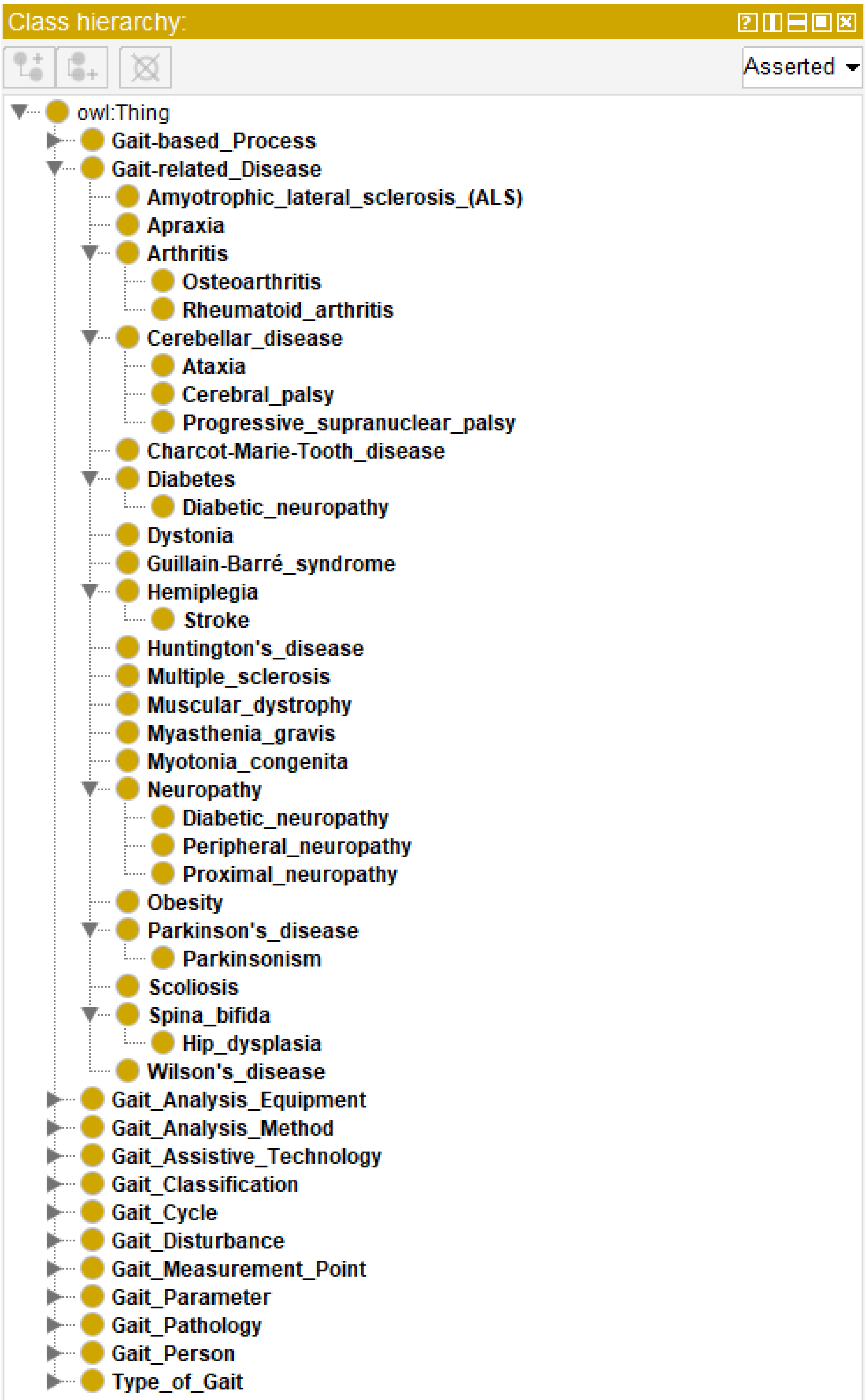


Figure 4.11: Class hierarchy of the domain ontology

4.4.3 Creating ontology properties

The development of a domain ontology for gait-related diseases required a comprehensive representation of the knowledge and relationships between different concepts in the domain. Object properties defined the relationships between entities in the ontology and provided a semantic understanding of how they were related, while data properties were used to represent the data attributes of each concept. By defining these properties, a comprehensive and interconnected representation of the knowledge in the domain was formed, which is an essential aspect for developing a knowledge-based system and application in the treatment of gait-related diseases. This understanding of the knowledge and relationships between concepts provided a solid foundation for the development of a knowledge-based system and applications that could be used to improve the diagnosis, treatment, and overall management of gait-related diseases.

4.4.3.1 Object properties

Object properties represented the relationships between concepts in the ontology. These relations linked semantically related concepts, providing a more comprehensive and interconnected representation of the knowledge in the domain. In contrast to data properties, which related a concept with a data type, object properties defined the relationships between entities or instances in the ontology and provided a semantic understanding of how they are related. This enables a richer semantic understanding of how these entities relate to one another, which is essential for capturing the complexity of the domain. The process of defining and analysing the core concepts within the domain also involved exploring potential relationships between them by explicitly and implicitly including reciprocal cross-connections. Explicit connections are those that are directly specified within the ontology, while implicit connections arise from the inherent relationships between concepts that may not be immediately apparent.

In the ontology, it was important to consider the properties of the ‘object’ properties themselves carefully. For example, in the ‘Type of Gait’ class, the object property "isExhibitedBy" could be defined as functional, meaning that a "Gait patient" could only exhibit one ‘Type of Gait’ at a time, or it could be described as inverse functional, meaning that a ‘Gait patient’ could only exhibit one ‘Type of Gait’. This semantic ability to enrich concepts, in this case, classes, through object property relationships enabled improved knowledge representation and formulation in the domain ontology with powerful, intelligent semantic capabilities. These considerations allowed for creating more accurate and reliable knowledge-based systems and applications for treating gait-related diseases.

Object properties were also essential for representing meaningful relationships between classes in the ontology, making inferring and reasoning about the domain easier. The associated object properties for the ‘Type of Gait’ concept in Figure 4.12 provided better insight into the relationships between related concepts in the domain ontology, allowing for more accurate and efficient knowledge representation, interpretation, and reasoning.

Figure 4.12: Object property relationships for the Type of Gait class in Protégé

Object property: “isDetectedIn”, attributes that a ‘Type of Gait’ is detected in a ‘Gait Analysis Method’.
Object property: “isExhibitedBy”, attributes that a ‘Type of Gait’ is exhibited by a ‘Gait patient’.
Object property: “isInfluencedBy” attributes that a ‘Type of Gait’ is influenced by a ‘Gait-related Disease’.
Object property: “isRelatedTo” attributes that a ‘Type of Gait’ is related to a ‘Gait Disturbance’.

This exploration involved identifying connections between entities and determining if certain relationships were reciprocal. These reciprocal cross-connections serve to reinforce the overall structure of the ontology and have led to a more interconnected and semantically rich representation of the knowledge in the domain of gait analysis.

Figure 4.13 illustrates the ‘Type of Gait’ class and its relevant subclasses with the inverse functional of “exhibits” for the “isExhibitedBy” object property, which is highlighted in red.

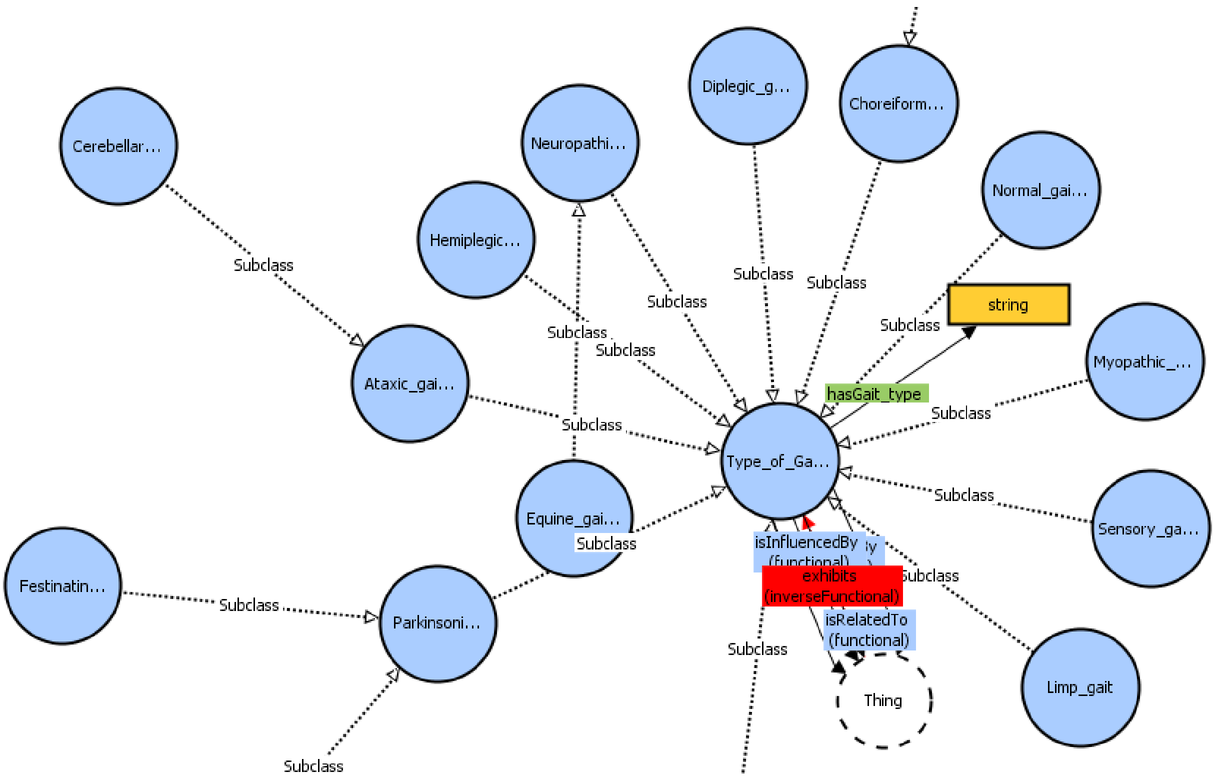


Figure 4.13: Type of Gait class and its associated subclasses in a VOWL graphical view

The modelling of object data properties allowed for the representation and understanding of interactions between entities within a specific domain. These interactions were expressed through concepts, relationship roles, and data types, which provided clear semantic meaning to the ontology. It was crucial to create the connection between different entities and define how each relationship is represented linking them in order for the ontology to make logical sense. In an example to demonstrate this, consider the following: ‘Gait data’ is acquired from the ‘Gait patient’ during ‘Gait acquisition’ in a ‘Gait-based process’ facilitated by a ‘Gait expert’ for ‘Gait analysis.’ This example serves to describe the class of the 'Gait-based Process' within the ontology as shown in Figure 4.14.

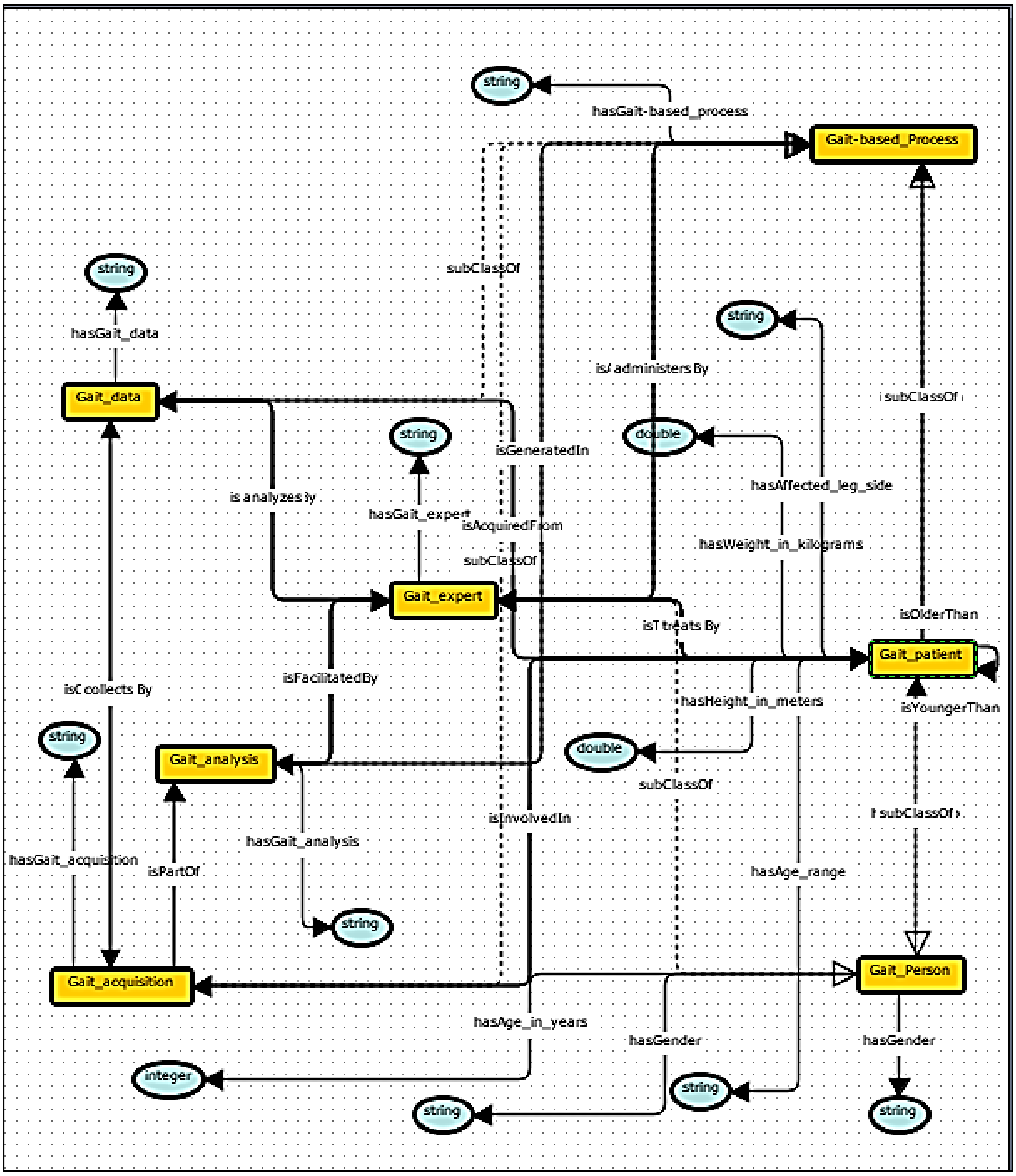


Figure 4.14: Gait-based Process model

Figure 4.15 is a snapshot of the object property type hierarchy for the GADO.

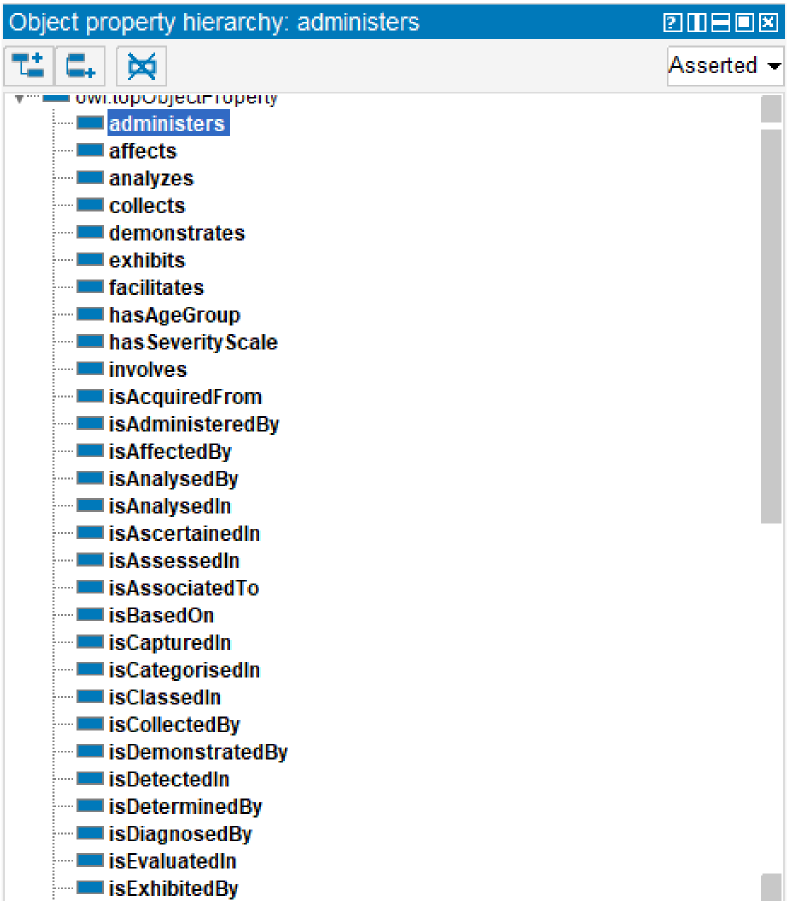


Figure 4.15: Snapshot view of the object property type hierarchy for the domain

4.4.3.2 Data properties

Data properties were used to represent the data attributes of each concept. Data properties are related to individuals or classes, as they were entities that defined or explained them to some degree. In the domain ontology, a 'Person' class has properties for gender, age, etcetera. These properties were used to describe the characteristics of the 'Person' concept and provided additional information about it. The 'Person' class concept consisted of two (2) subclasses representing the 'Gait expert' and 'Gait patient', as portrayed in Figure 4.16.

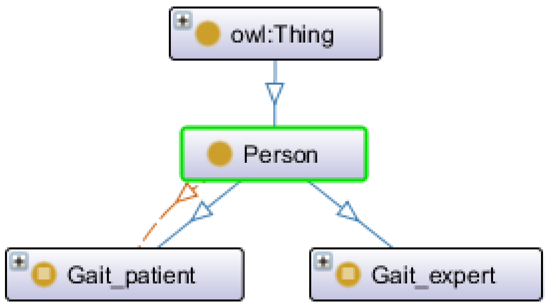


Figure 4.16: Graphical representation of the person concept

Using data properties, a more elaborate and detailed representation of the domain knowledge was achieved, which was necessary for developing a knowledge-based system and application for the treatment of gait-related diseases. Figure 4.17 depicts the data properties for the 'Gait patient', and Figure 4.18 shows the data properties for the 'Gait expert'.

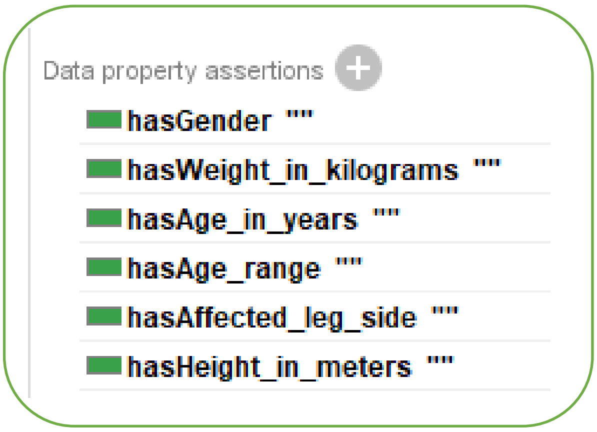


Figure 4.17: Gait patient data properties

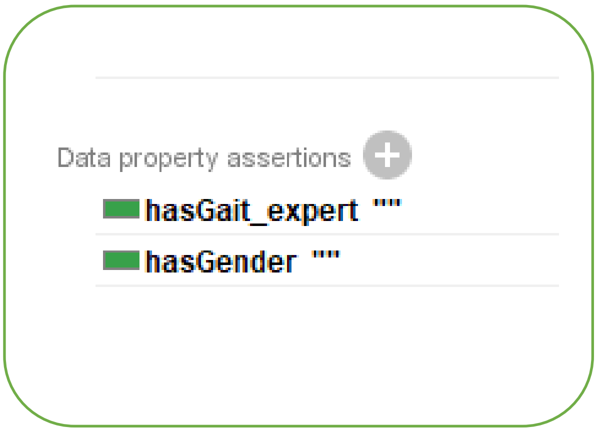


Figure 4.18: Gait expert data properties

By defining these properties, it became possible to build a more vigorous and interconnected ontology that could be used to improve the diagnosis, treatment, and overall management of gait-related diseases. Figure 4.19 shows the complete data property type hierarchy for the domain that was developed and captured within Protégé.

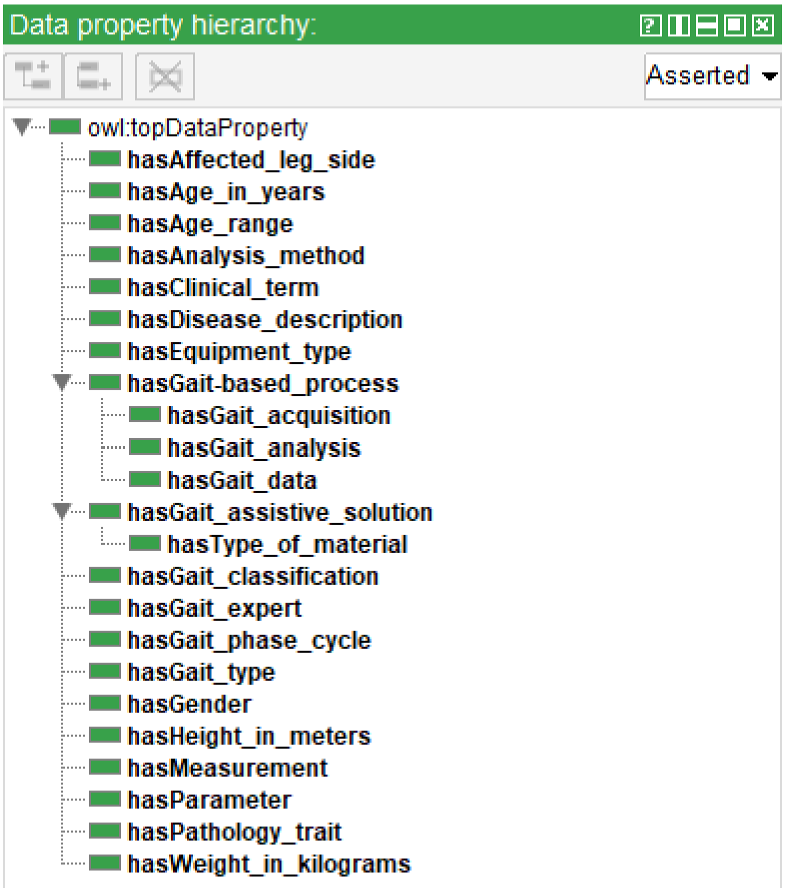


Figure 4.19: Data property hierarchy of the domain

4.4.3.3 Creating individuals or instances

In the development of a domain ontology for gait analysis and related diseases, individuals, also known as instances, were specific objects or entities in the domain being modelled. The individuals were what the ontology was designed to represent and reason about, and therefore it was essential to ensure that they were accurately defined and representative of the real-world entities they intrinsically modelled. In the context of a gait-related diseases domain, these individuals represented gait patient, gait expert, gait disturbance, gait assistive technology, type of gait, and all other relevant entities in the scope of the domain ontology. The ontology defined the classes and properties that the individuals belonged to and have respectively. This made it possible to determine and make inferences about the semantic relationships between individuals and the properties they possessed.

In general, individuals are the basic building blocks of the ontology and can be both concrete and abstract concepts. Concrete individuals refer to physical objects or entities that can be observed or perceived. Inversely, abstract individuals refer to non-physical entities, like ideas, concepts, and relationships. Individuals in this domain included concrete concepts functioning as specific gait diseases with the individual examples of “Parkinson's disease” and associated gait disturbances, such as “Gait imbalance” and “Difficulty walking”. Abstract concepts included the relationships between gait diseases and other medical classifications for gait, essentially Musculoskeletal, Neurological and Neuromuscular, based on the characteristics of the gait-related disease or dysfunction. Individual instances also included the class Type of Gait exhibited for a gait-related disease, being “Ataxic gait”, “Parkinsonian gait”, or “Limp gait”.

It is important to note that while the number of individuals in an ontology can be vast, only a portion of them was typically included in the representation. This was because including all individuals would be computationally infeasible and would lead to a cluttered and difficult-to-understand representation. In turn, only a subset of relevant individuals was included in the ontology to provide a clear and concise understanding of the domain.

All the high-level classes in the ontology must have at least one individual to be considered a valid class. Thus, the top class of ‘Gait-related Disease’ must have at least one individual, acting as either “Ataxia”, “Dystonia”, or “Parkinson’s” disease, as depicted in Figure 4.20. Similarly, the class of ‘Type of Gait’ must have at least one individual, such as “Ataxic gait”, “Parkinsonian gait”, or “Limp gait”, as displayed in Figure 4.21. In this way, the ontology presents a comprehensive representation of the real world by providing various individuals belonging to the different ontology classes.

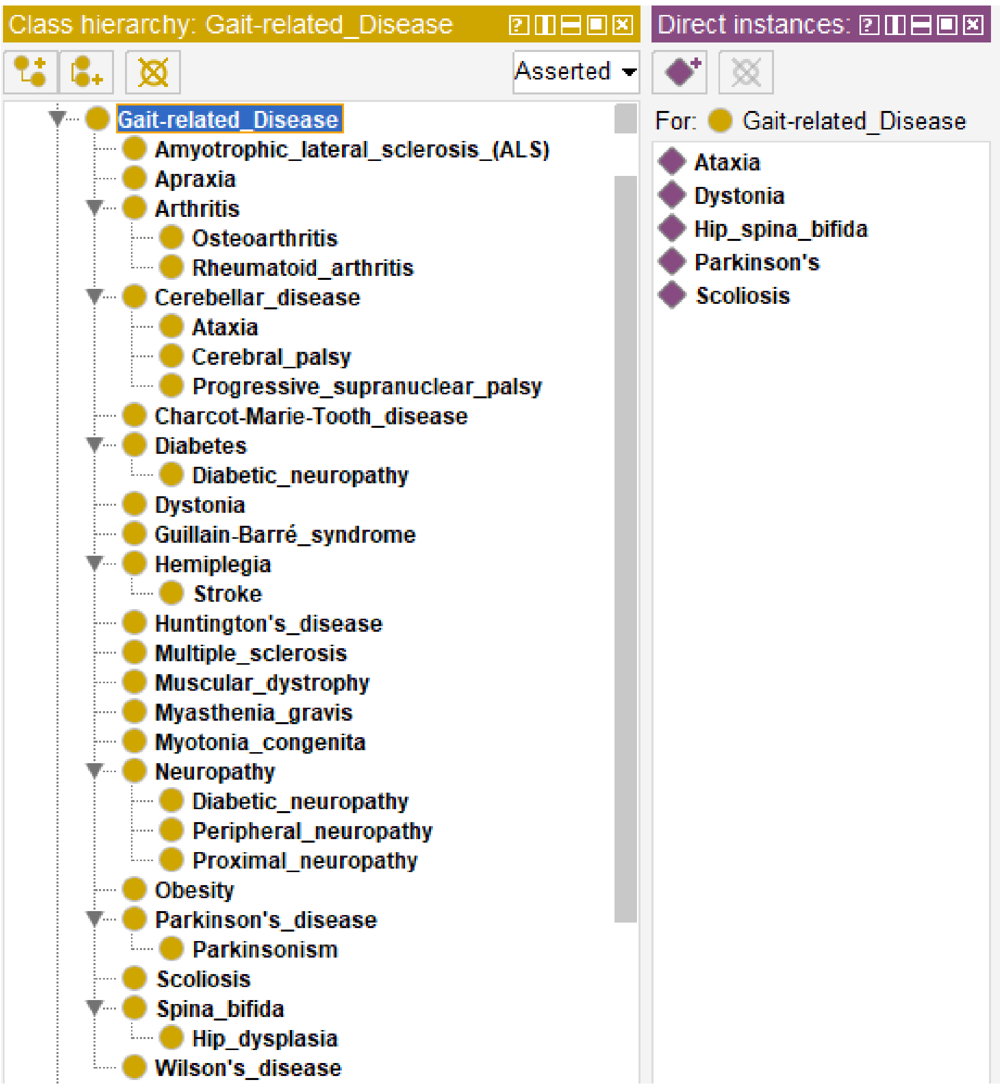


Figure 4.20: Gait-related disease class individuals or instances

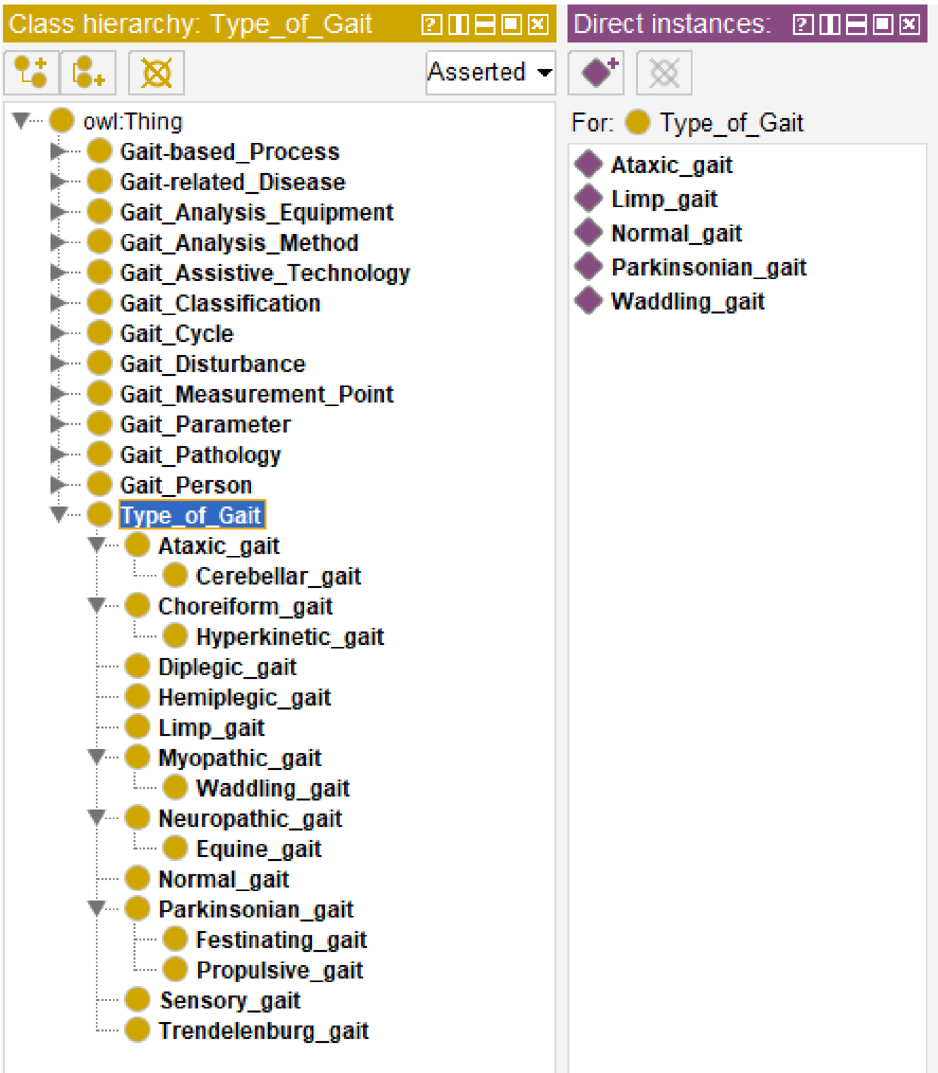


Figure 4.21: Type of gait class individuals or instances

In Figure 4.22 below, a snapshot overview in Protégé illustrates an individual instance, Adult, for the 'Gait patient' concept with its related object and data properties asserted attributes.

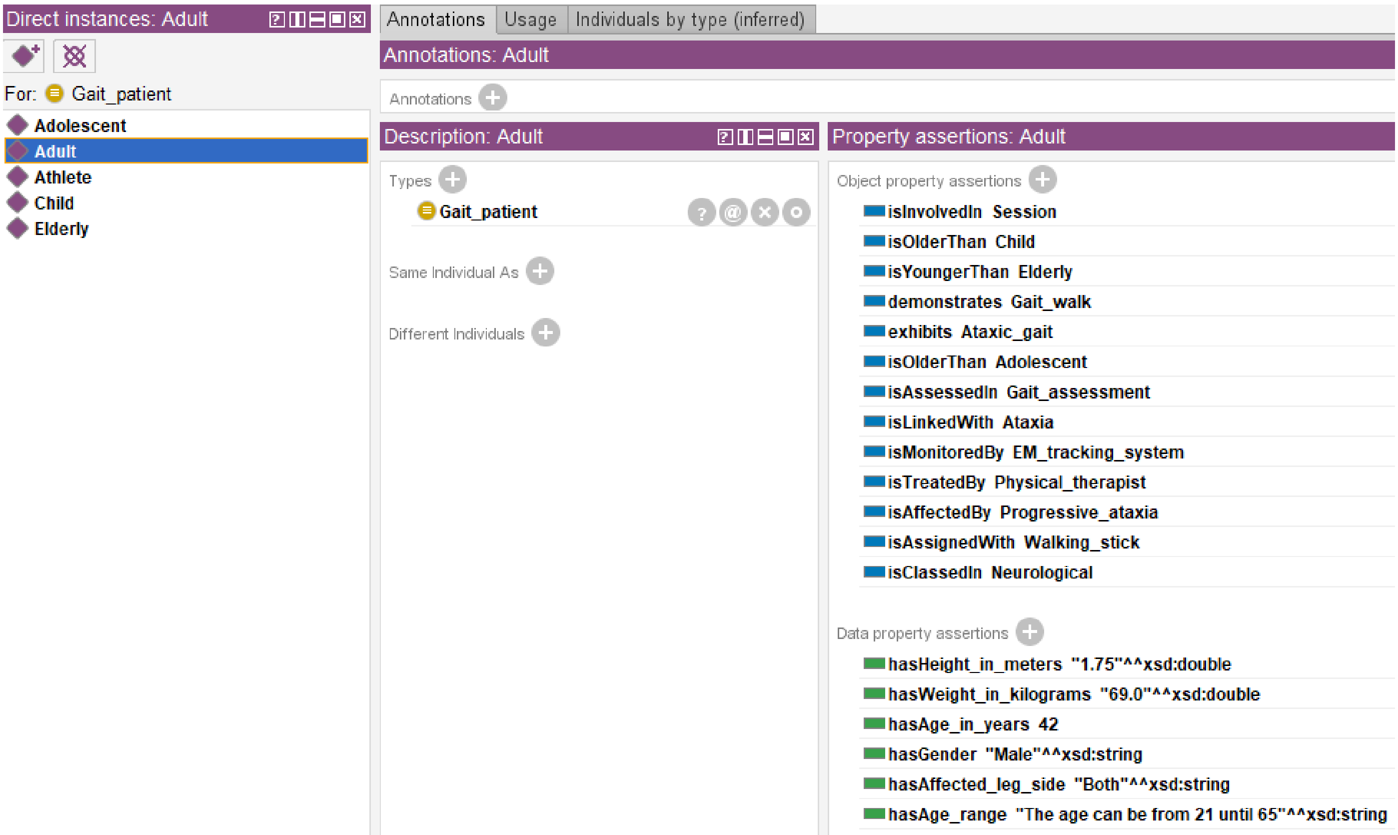


Figure 4.22: Object and data properties for the "Adult" individual/instance

Based on the object property assertions of the instance for the 'Gait patient' class, the object property restrictions for the 'Gait patient' class were defined as depicted in Figure 4.23.

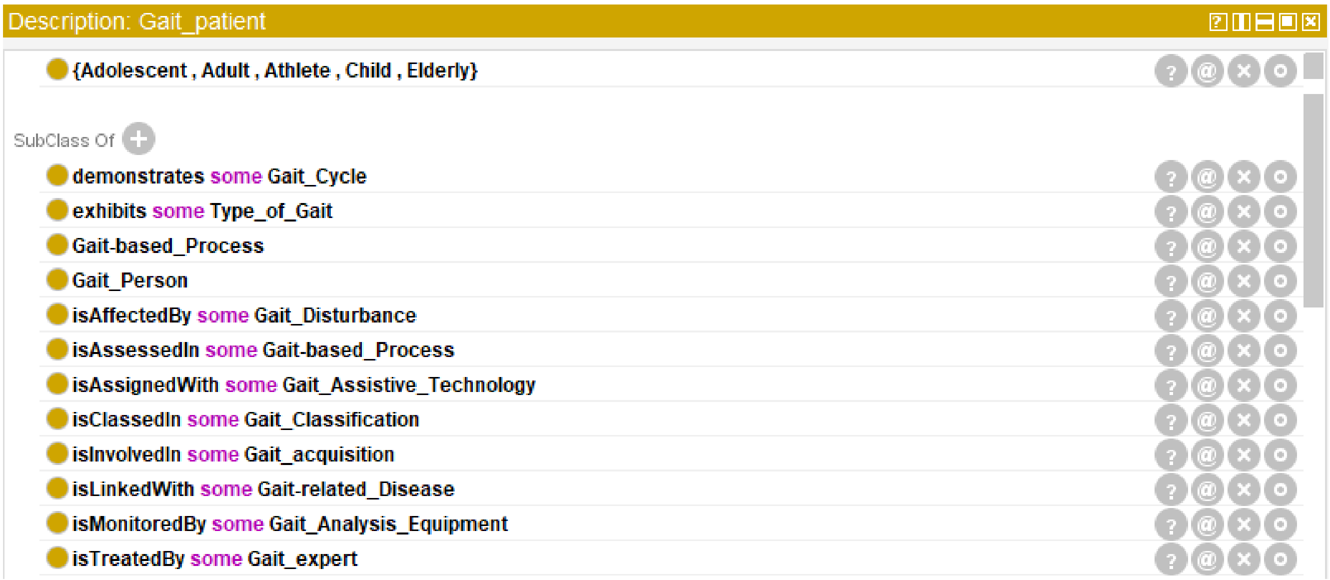


Figure 4.23: Object property restrictions for the 'Gait patient' class

The ‘Gait patient’ concept (with individual instances modelled as Adolescent, Adult, Athlete, Child, and Elderly) was restricted by “some” object property and core concept, as shown in Table 4.6.

Table 4.6: Gait patient individual/instance

Object Property	**Restriction**	**Concept**
demonstrates	*some*	Gait Cycle
exhibits	*some*	Type of Gait
isAffectedBy	*some*	Gait Disturbance
isAssessedIn	*some*	Gait-based Process
isAssignedWith	*some*	Gait Assistive Technology
isClassedIn	*some*	Gait Classification
isInvolvedIn	*some*	Gait acquisition
isLinkedWith	*some*	Gait-related Disease
isMonitoredBy	*some*	Gait Analysis Equipment
isTreatedBy	*some*	Gait expert

Figure 4.24 is a graphic illustration of the 'Gait patient' concept based on attributed object property relationships established in the domain ontology creation.

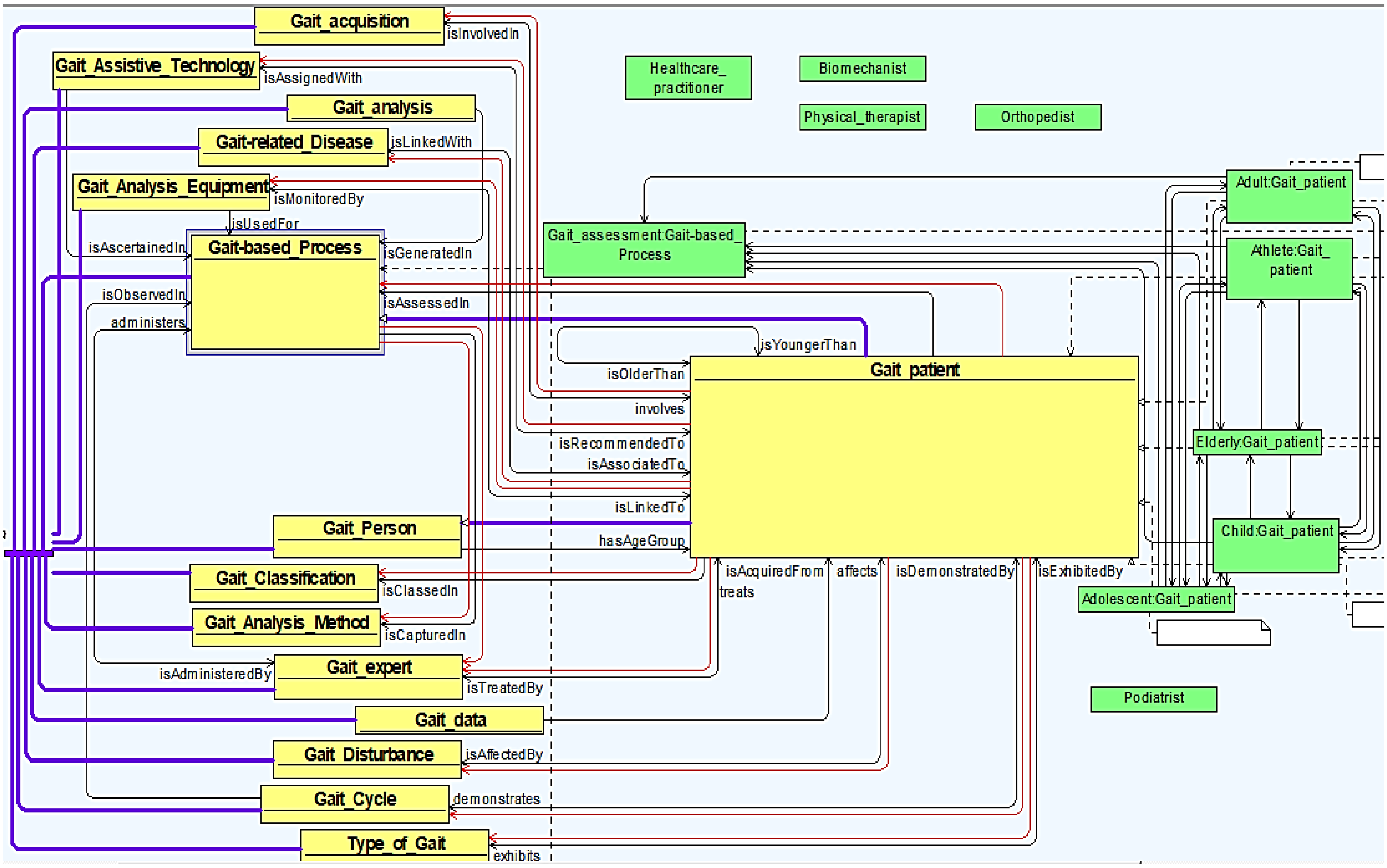


Figure 4.24: The 'Gait patient' class with associated object property relationships

Figure 4.25 presents sample instances for the main classes of the domain ontology.

Class	Members
owl:Thing	
Gait_Measurement_Point	Foot_pronation
Gait_Disturbance	Waddling, Gait_imbalance, Progressive_ataxia, Freezing_of_gait, Dystonic_gait
Gait_Classification	Musculoskeletal, Neurological, Neuromuscular
Gait_Parameter	Pace
Gait_Analysis_Equipment	'Ground Reaction Force sensor', Gonio, Gyroscope, Electromyography, EM_tracking_system
Gait_Person	
Gait_expert	Biomechanist, Physical_therapist, Orthopedist, Podiatrist, Healthcare_practitioner
Gait_patient	Adult, Child, Adolescent, Elderly, Athlete
Gait-based_Process	Gait_assessment
Gait-related_Disease	Parkinson's, Hip_spina_bifida, Ataxia, Dystonia, Scoliosis
Gait_Assistive_Technology	Orthotic, Pharmaceutical, Walking_stick, Braces, Walker
Gait_Pathology	Frontal_lobe_injury, Hip_abductor_weakness, Hallux_rigidus, Tremor, Leg_length_discrepency
Gait_Cycle	Gait_walk
Gait _Analysis_Method	Motion_system, Kinematography, AI_method, Sensory, Image_sensing
Type_of_Gait	Waddling_gait, Ataxic_gait, Parkinsonian_gait, Normal_gait, Limp_gait

Figure 4.25: Individual instance members of the GADO

4.4.4. Annotations

During the development of domain ontology, descriptive information or labels were added to individual classes, properties, or instances of the ontology to provide a detailed annotation (description) of the knowledge within the domain. The purpose of these annotations was to enhance the understanding, meaning and purpose of the components of the ontology by humans and machines. Purposefully, annotations will enable accurate semantic analysis that can aid information retrieval.

In Figure 4.12, the relationship between the 'Type of Gait' class and 'Gait Disturbance' class was expressed as:

Object property: "isRelatedTo", which attributed that a 'Type of Gait' is related to a 'Gait Disturbance'.

This was further asserted by the individuals or instances between these two class concepts as annotated and demonstrated in Figure 4.26 and Figure 4.27, respectively.

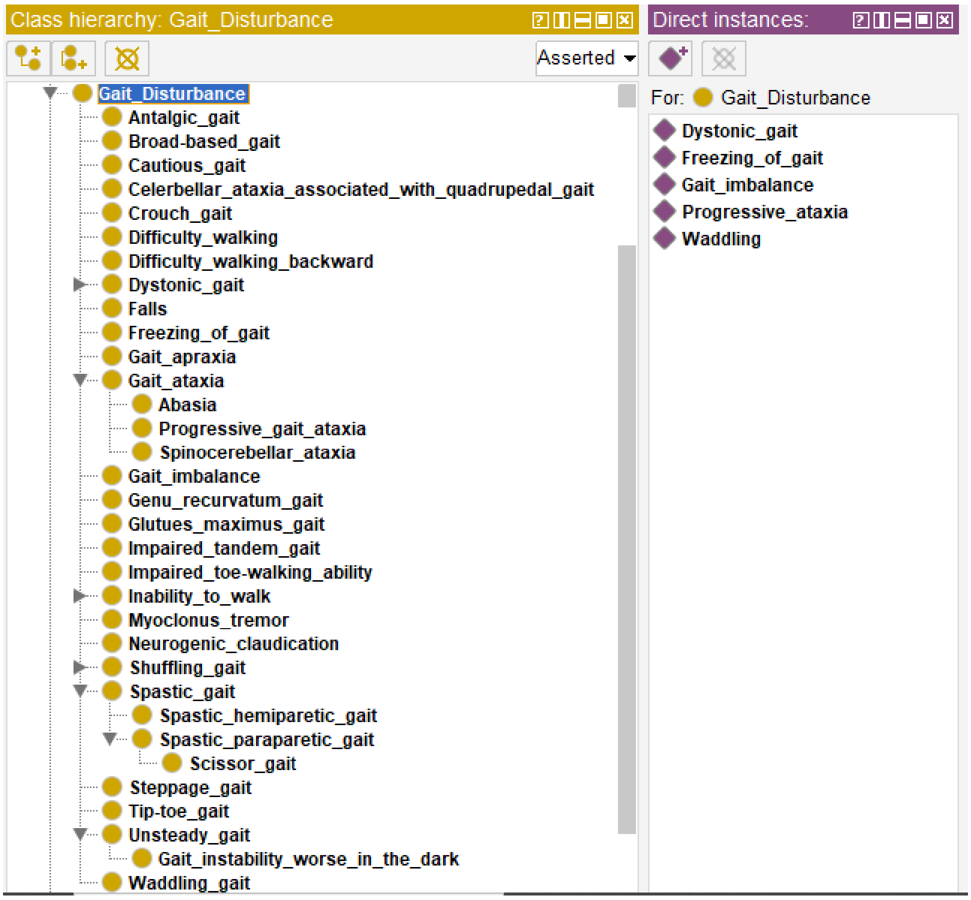


Figure 4.26: Gait disturbance class instances

The core concept, ‘Type of Gait’ with instance “Parkinsonian gait”, is related to the “Freezing of gait” instance of the ‘Gait Disturbance’ concept, as per “isRelatedTo” object property relationship. To further bolster the object property assertion, annotations strongly made this relationship more explicit and descriptive, as revealed in Figure 4.37 below.

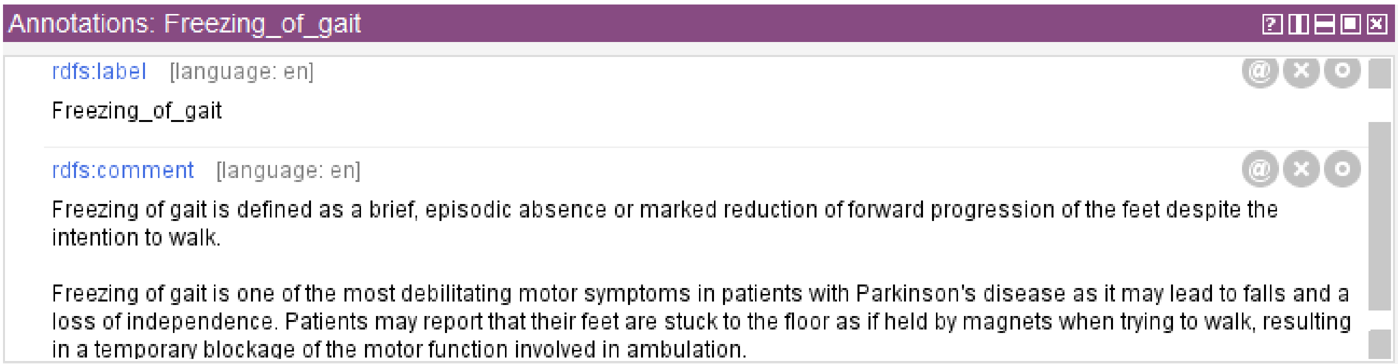


Figure 4.27: Annotations for freezing of gait individual/instance

Based on the abovementioned assumption, it can be inferred that the “Parkinsonian” ‘Type of Gait’, exhibited by a patient, “*isRelatedTo”* the ‘Gait Disturbance’, which in this case was “Freezing of gait” as richly annotated from credible medical data sources.

4.4.5 Iteration

The iterative nature of the OD-101 methodology enabled an incremental improvement of the ontology, as new concepts and relationships could be added, and existing ones refined and fine-tuned. This also inadvertently allowed for a more complete and accurate representation of the domain, as different perspectives and nuances can be considered. Furthermore, the iterative propensity of this methodology facilitates the incorporation of feedback from stakeholders and domain experts to ensure that the ontology accurately reflects the desired needs and requirements.

The development of a domain ontology for gait-related diseases has been updated and modified with several iterations to ensure accuracy and comprehensiveness. While brand-new information becomes available, and as the needs of the domain evolve, the ontology must be updated and modified to reflect these changes. This was and is an ongoing process that requires continuous improvement and refinement. The first iteration of the ontology typically involved the identification of the domain concepts, defining the domain classes, and specifying the relationships between the classes. This was followed by the definition of the properties of the classes, serving as their attributes and constraints.

In subsequent iterations, the ontology was further developed to include more detailed information and relationships. A case in point was with new classes and subclasses added to represent specific types of gait-related diseases, and more specific relationships defined between classes to represent the relationships between different concepts in the ontology. In addition, the ontology was updated with current information, such as new research findings or medical terminology as was the case. Iteration also involved the integration of added information from the literature data collection sources. This ensured that the ontology was kept up-to-date and accurate and could provide accurate and relevant information to users.

The ontology underwent multiple iterations, during which it was updated and revised to incorporate new information and integrate new knowledge, ensuring consistency, coherence, and a well-structured framework. Regular validation and checking were conducted to confirm that the ontology was free of errors and inconsistencies and that it accurately represented the current state of knowledge in the domain. The domain ontology was extended with the inclusion of several classes and subclasses, as exemplified by 'Pathological gait' in Figure 4.28 below. This example illustrates the expansion of the ontology to include various classes and subclasses, being the 'Pathological gait' class and its related subclasses, which defined the core concept or main class, 'Gait Pathology'. The addition of these classes and subclasses to the domain ontology enabled a more precise and comprehensive depiction of pathological gait. This iterative aspect of incremental updating, expanding, and refining was crucial for developing a robust and accurate ontology that supported research and practice in gait-related diseases.

Figure 4.28: Class hierarchy of gait pathology

Gait Pathology main concept consists of two (2) subclasses:

i) Geriatric gait: This subclass was divided into four (4) sub-concepts, which were further divided to categorise the pathologies specific to 'Geriatric gait', namely: Calcification, Dementia, Disability and Morbidity. These subclasses were further divided with their respective associated subclasses, as depicted in Figure 4.29.

ii) Pathological gait: This subclass was divided into two (2) sub-concepts, which were further subdivided to classify the pathologies specific to 'Pathological gait': Musculoskeletal and Neurological, which were then subdivided more with their respective associated subclasses as illustrated in Figure 4.30.

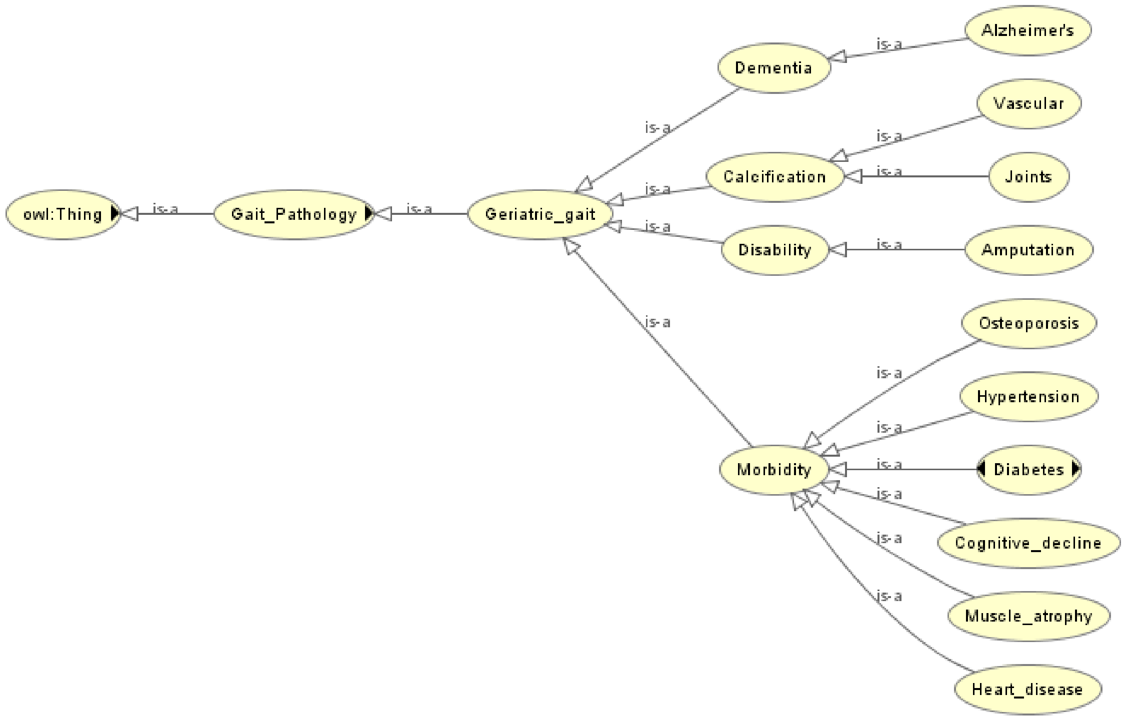


Figure 4.29: Graphical representation of geriatric gait

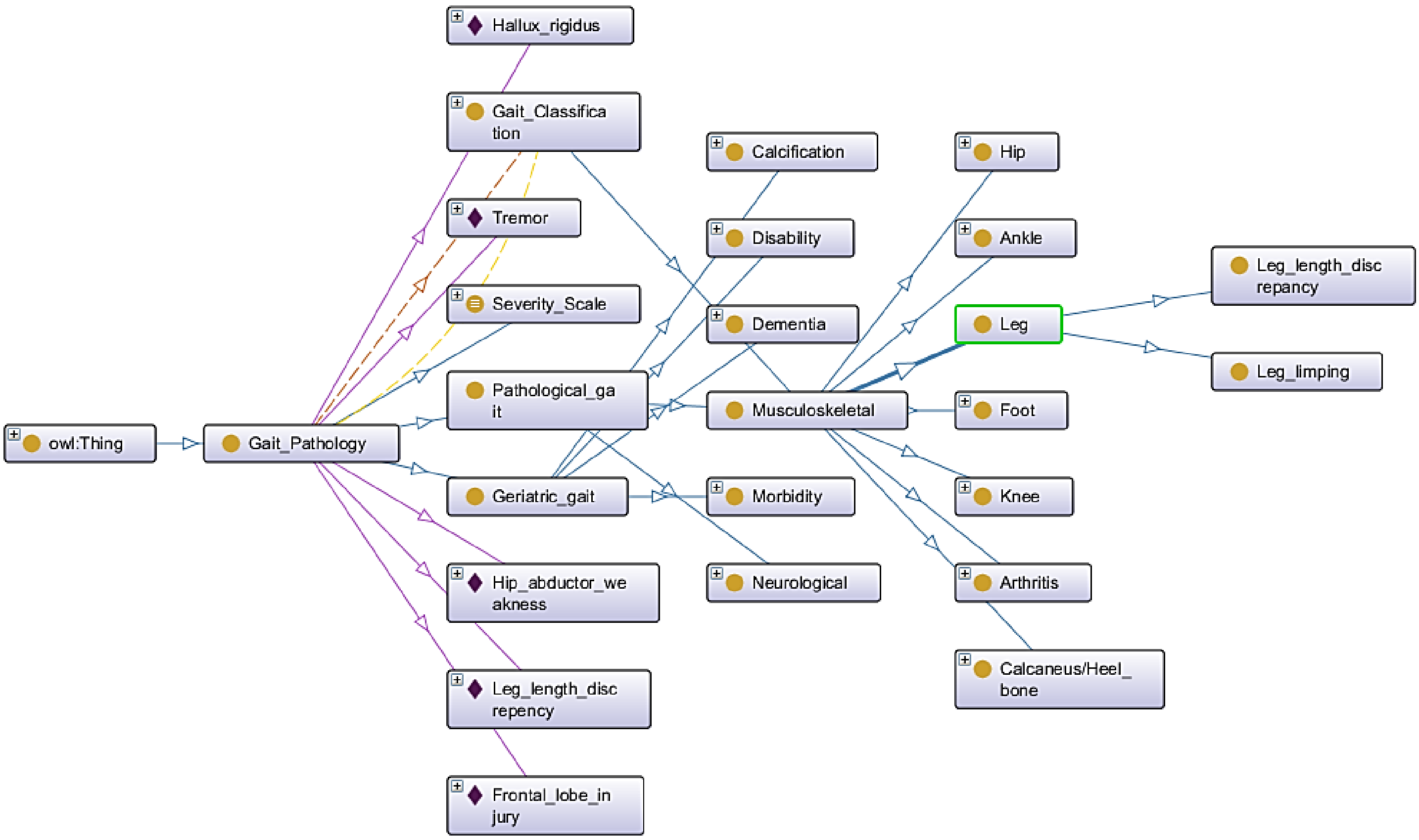


Figure 4.30: Graphical representation of pathological gait

4.5 Summary

The chapter on ontology design and development in the thesis focused on constructing the desired ontology for gait-related matters. The chapter began with a domain analysis, identifying relevant concepts, relationships, and properties in the domain. The requirements and competency questions derived from the domain analysis were then utilised to guide the iterative development process of the ontology. Classes, object properties, data properties, and the importance of maintaining the ontology were emphasised in the development process with the Protégé editing software. The chapter also highlighted the importance of modelling data object properties and instances while discussing the use of annotations to provide metadata and additional information about the ontology. This chapter described the process involved in creating an ontology for gait-related diseases, serving as a blueprint for developing the domain ontology. Overall, the ontology design and development chapter demonstrated the importance of a structured and iterative approach to ontology development.

CHAPTER 5: ONTOLOGY EVALUATION

This chapter presents the procedure for the evaluation of the Gait Analysis Domain Ontology (GADO). The chapter kicks off by applying standard metrics such as "domain task fit," "content richness," and "correctness" to assess the ontology's foundational quality. Following this, the FOCA methodology is employed for an objective evaluation by a panel of experts from diverse backgrounds. To further substantiate the ontology's validity, Description Logic (DL) and SPARQL queries are executed. This multi-faceted approach aims to affirm the ontology's robustness, consistency, and applicability in the Gait Analysis Domain Ontology.

5.1 Evaluation of the GADO

Ontology engineering is a set of tasks related to the development, maintenance, and use of ontologies, which are formal representations of a set of concepts and the relationships between them. During the ontology engineering process, evaluating the ontology is a vital phase to determine the quality and effectiveness of the domain ontology. Hence it is categorised into ontology validation and verification. Both validation and verification are important for ensuring the quality of the GADO ontology. Validation focuses on the ontology's content, while verification focuses on the ontology's implementation.

5.1.1 Verification of the GADO

Ontology verification is the process of ensuring that the ontology is well-constructed, adhering to design criteria, maintaining internal consistency, and is free from errors (Alsanad et al., 2019). It is essential because a verified ontology guarantees the reliability and accuracy of the knowledge it represents, ensuring that it can be confidently used in applications and decision-making processes.

5.1.1.1 Domain task fit

The first step in assessing the quality of the ontology involved evaluating its compatibility with the intended domain task. This was done to determine if the ontology could successfully answer the competency questions and meet the needs of its intended purpose/task. This helped to establish whether the ontology was able to provide the necessary information to support the goals of the specific domain task. This is part of verification because it checks whether the ontology is suitable for the tasks it is intended to support.

The ontology was designed to support gait diagnosis, treatment, and analysis tasks in the context of gait-related diseases. For instance, the ontology was required to support the classification of different types of gait, the identification of gait pathologies, and the selection of appropriate gait assistive solutions.

The competency questions, previously defined in Table 4.1, were meticulously assessed in the domain ontology. This assessment helped ensure the ontology could answer the correlated competency questions, as explained, and detailed in Table 5.1 beneath. Thus, evaluating the ontology's domain task fit confirmed its ability to effectively support gait-related tasks.

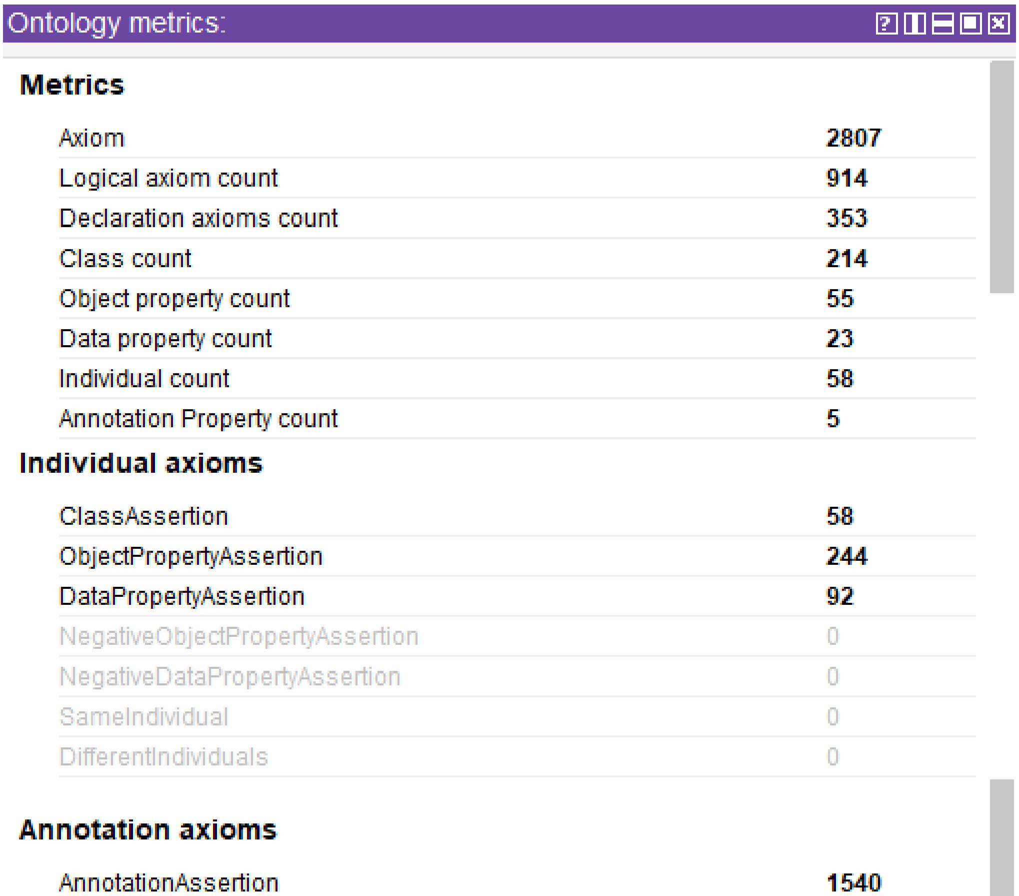
Table 5.1: Competency Questions Answered

Competency Questions		Competency Questions' (Answers)
Q1	Who are the users of the domain for gait-related diseases?	The domain users for gait-related diseases are gait experts, domain experts, and clinicians (all of whom are classes contained in the ontology).
Q2	What gait-related diseases are linked with geriatric (elderly) patients?	Gait-related diseases that are linked with geriatric patients include dementia, osteoporosis, calcification, and muscle atrophy (all of which are linked to the Geriatric gait class in the ontology).
Q3	What are the most common types of gait-related diseases?	The most common types of gait-related diseases are Parkinson's disease, cerebral palsy, multiple sclerosis, and peripheral neuropathy (all of which are classified as Gait-related diseases in the ontology).
Q4	What are the typical neurological, gait-related diseases that impact patients' gait?	Typical neurological gait-related diseases patients endure, include ataxia, dystonia, multiple sclerosis, Parkinson's disease, and stroke (all of which are classified as neurological Gait-related diseases in the ontology).
Q5	Is there sufficient detail to differentiate between different types of gait-related diseases?	Yes, the domain ontology contains up-to-date information on the current state of knowledge on gait-related diseases, including sufficient detail to differentiate between different types.
Q6	What current gait assistive technology solutions are available for gait disorders?	Current gait assistive technology solutions may include orthotics, canes, walkers, and therapy (all of which are listed as gait assistive technology solutions in the ontology).
Q7	What types of non-pharmacological treatments are available for gait-related diseases?	Non-pharmacological treatments available for gait-related diseases include physical therapy, rehabilitation therapy and gait training (all of which are listed as non-pharmacological treatments for gait-related diseases in the ontology).
Q8	To what extent can a decision support system for gait-related issues be useful/beneficial?	A decision support system for gait-related issues can significantly provide healthcare professionals with accurate, evidence-based recommendations for the diagnosis, treatment, management, and informed decision-making of gait-related diseases (because the ontology contains information on all of these aspects).
Q9	Who are the key role players in the execution of a gait-based process?	Key role players in the execution of a gait-based process may include a gait expert, physical therapist, orthopedist, and biomechanist (all of which are listed as key role players in the gait-based process in the ontology).
Q10	What are the types of gait disturbance that are generally associated with children?	Gait disturbances that are generally associated with children include cerebral palsy, muscular dystrophy, loss of ability to walk in early childhood and spina bifida (all of which are classified as Gait disturbances associated with children in the ontology).
Q11	What are the resources that require gait analysis?	Resources that require gait analysis include motion capture systems, infrared cameras, force plates, sensors, and trained experts to perform and analyse the tests (all of which are listed as resources required for gait analysis in the ontology).
Q12	What are the main etiological anatomies for gait pathologies?	The main etiological anatomies for gait pathologies include musculoskeletal and neurological pathological gait (both of which are classified as etiological anatomies for Gait pathologies in the ontology).
Q13	What are the different gait dimensions for the domain?	Gait-based Process, Gait-related Disease, Gait Analysis Equipment, Gait Analysis Method, Gait Assistive Technology, Gait Classification, Gait Cycle, Gait Disturbance, Gait Pathology, Gait Measurement Point, Gait Parameter, Type of Gait & Gait Person (all of which are listed as gait dimensions in the ontology).

5.1.1.2 Content richness

The next step is evaluating the quality of the ontology involved in assessing its content richness. This was done by using standard metrics serving as the number of classes and properties to determine if the ontology was wide-ranging and included all important concepts within the domain. In pursuit of ensuring content richness, the ontology was evaluated to determine if it covered all the relevant concepts related to gait-related diseases. Furthermore, the concepts were described in sufficient detail, achieved through annotations of all the key entities, such as classes, subclasses, instances, objects, and data properties, with the completion of labels and comment assignments in annotations.

The ontology was also required to describe the relationships among these concepts in detail, ensuring that they were accurate, complete, and logically consistent. This was also proven with complete ontology annotations. The content richness of the ontology was evaluated to ensure that it comprehensively covered all the essential concepts within the domain. The ontology was populated and integrated into the developed domain ontology for gait analysis. The logical domain ontology structure in Protégé was constituted of 2807 validated axioms distributed throughout 214 classes with 55 object properties, 23 data properties, and 58 individual instances. The individual instance axioms contained 58 class assertions, 244 object property assertions and 92 data property assertions. The combined total of annotations across the domain ontology was composed of 1540 annotation axioms. The domain ontology metrics are enclosed in Figure 5.1 as generated within the Protégé editor.


Ontology metrics:

Metrics	
Axiom	2807
Logical axiom count	914
Declaration axioms count	353
Class count	214
Object property count	55
Data property count	23
Individual count	58
Annotation Property count	5
Individual axioms	
ClassAssertion	58
ObjectPropertyAssertion	244
DataPropertyAssertion	92
NegativeObjectPropertyAssertion	0
NegativeDataPropertyAssertion	0
SameIndividual	0
DifferentIndividuals	0
Annotation axioms	
AnnotationAssertion	1540

Figure 5.1: Ontology metrics for the domain

The evaluation revealed that the ontology model was consistent, as all reasoning tasks included class hierarchy, object property hierarchy, data property hierarchy, class assertions, and object property assertions. The same individuals returned a consistent ontology model. This indicated that the ontology model was free from logical inconsistencies, and the relationships between the classes, object properties, and data properties were precisely defined with no errors.

5.1.1.3 FOCA methodology

This is a bit of a hybrid but leans more towards verification. It is an objective evaluation to ensure that the ontology meets certain quality criteria. The methodology checks for well-defined goals and whether those goals are met.

5.1.1.3.1 FOCA methodology applied by the researcher

Step 1 - Ontology Type Verification:

The first step in verifying the quality of the ontology was to determine its type. The FOCA approach specified two ontology types: task/domain ontology and application ontology. In this case, since the ontology was a domain ontology, it fell under type 1. According to FOCA, a type 1 ontology should answer Q5 instead of Q4 for goal 2.

Step 2 - Questions Verification:

The second step involved answering 12 out of 13 questions (one question is not answered based on the ontology type selected in step 1) and scoring each answer based on the researcher's assessment. Each question represented one of the ontology quality criteria, and the 12 questions were associated with five goals:

i. **Goal 1**: Competency questions and reuse.
ii. **Goal 2**: Ontology terms satisfy criteria and meet the expected level.
iii. **Goal 3**: Contradictions or invalid (re)use of terms in the ontology.
iv. **Goal 4**: Reasoning and reasoner performance.
v. **Goal 5**: Ontology substantiation and consistency between the model ontology and the design.

The FOCA methodology used a Goals Questions Metrics (GQM) approach, which consisted of goals, their corresponding questions, and metrics. Using these descriptions, the researcher determined the score for each question. The mean score for each goal was subsequently calculated. Gradings of questions were scored on base percentages of 25, 50, 75 or 100. Notably, all goals received a mean score of 100%, except goals 1 and 2. Goal 1 received a mean score aggregate of 58.33% because the domain ontology was a completely new development without reusing other ontologies. Goal 2 received a mean sum of 87.50% as the domain ontology provided reasonable abstraction. The grading of the ontology is depicted in Table 5.2.

Table 5.2: FOCA grading of the domain ontology (refer to **Appendix E**: How to verify FOCA)

Goal	No.	Question (score each question either 25, 50, 75, or 100)	Score	Mean
	Q1.	Were the competency questions well-defined?	75	
Goal 1	Q2.	Were the competency questions answered? Yes = 100 / No = 0	100	**58.33**
	Q3.	Did the ontology reuse other ontologies? Yes = 100 / No = 0	0	
	Q4.	Did the ontology impose a minimal ontological commitment?	_	
Goal 2	Q5.	Did the ontology impose a full ontological commitment?	75	**87.50**
	Q6.	Are the ontology properties coherent with the domain? Yes = 100 / No = 0	100	
	Q7.	Are there contradictory axioms? Yes = 0 / No = 100	100	
Goal 3	Q8.	Are there redundant axioms? Yes = 0 / No = 100	100	**100**
	Q9.	Does the reasoner bring modelling errors? Yes = 0 / No = 100	100	
Goal 4	Q10.	Does the reasoner perform quickly? Yes = 100 / No = 0	100	**100**
	Q11.	Is the documentation consistent with the modelling? Yes = 100 / No = 0	100	
Goal 5	Q12.	Were the concepts well written? Yes = 100 / No = 0	100	**100**
	Q13.	Are there annotations in the ontology bringing the definitions of the concept? Yes = 100 / No = 0	100	

Step 3 - Quality Verification:

The FOCA methodology is a systematic approach that combines various metrics to estimate the total quality of an ontology. This approach involves the utilisation of a beta regression model which integrates various coverage aspects, including structural coverage (CovS), conceptual coverage (CovC), relationship coverage (CovR), and concept property coverage (CovCp), along with the level of expertise (LExp) and normalisation level (Nl). The use of total quality verification was selected in this case because maximum goals were evaluated. This full quality was calculated using the following beta regression model formula:

$$\hat{\mu}_i = \frac{\exp\{-0.44+0.03(CovS\times Sb)_i+0.02(CovC\times Co)_i+0.01(CovR\times Re)_i+0.02(CovCp\times Cp)_i-0.66LExp_i-25(0.1\times Nl)_i\}}{1+\exp\{-0.44+0.03(CovS\times Sb)_i+0.02(CovC\times Co)_i+0.01(CovR\times Re)_i+0.02(CovCp\times Cp)_i-0.66LExp_i-25(0.1\times Nl)_i\}}$$

In order to calculate the total quality, the following variables were considered:

CovS: The mean score of goal 1.

CovC: The mean score of goal 2.

CovR: The mean score of goal 3.

CovCp: The mean score of goal 4.

LExp: The researcher's experience, with 1 representing experienced and 0 not experienced.

NI: If a goal cannot be evaluated or any question cannot be evaluated, NI is set to 1.

Sb: Set to 1 because all goals were weighed.

Co: Set to 1 because all goals were weighed.

Re: Set to 1 because all goals were weighed.

Cp: Set to 1 because all goals were weighed.

Based on these variables, the total quality of the ontology was calculated by substituting the variables with the corresponding values in the equation mentioned above.

$$\hat{\mu}_i = \frac{\exp\{-0.44 + 0.03(58.33 \times 1) + 0.02(87.5 \times 1) + 0.01(100 \times 1) + 0.02(100 \times 1) - 0.66 \times 1 - 25(0.1 \times 0)\}}{1 + \exp\{-0.44 + 0.03(58.33 \times 1) + 0.02(87.5 \times 1) + 0.01(100 \times 1) + 0.02(100 \times 1) - 0.66 \times 1 - 25(0.1 \times 0)\}}$$

$\exp\{-0.44 + 0.03(58.33 \times 1) + 0.02(87.5 \times 1) + 0.01(100 \times 1) + 0.02(100 \times 1) - 0.66 \times 1 - 25(0.1 \times 0)\}$

$= \exp\{-0.44 + 1.75 + 1.75 + 1 + 2 - 0.66\}$

$= \exp\{5.4\}$

$= 221.4$

and

$1 + \exp\{-0.44 + 0.03(58.33 \times 1) + 0.02(87.5 \times 1) + 0.01(100 \times 1) + 0.02(100 \times 1) - 0.66 \times 1 - 25(0.1 \times 0)\}$

$= 1 + \exp\{-0.44 + 1.75 + 1.75 + 1 + 2 - 0.66\}$

$= 1 + \exp\{5.4\}$

$= 222.4$

Hence, the expression becomes:

221.4 / 222.4 = 0.9955 (rounded to four decimal places).

$\hat{\mu} = 0.9955$

Therefore, the result of the expression was approximately 0.9955, which is nearer to 1.

Subjectively, this indicated that the domain ontology is of high quality and fit for purpose.

5.1.1.3.2 FOCA methodology applied with the evaluators

In this research, data collection and analysis processes were implemented by adopting a mixed-methods approach, incorporating both qualitative and quantitative aspects. The goal of this approach was to obtain in-depth insights and comprehensively evaluate the Gait Analysis Domain Ontology from different perspectives. The data collection process encompassed obtaining the background and experience of evaluators, providing an overview of GADO in the form of a document guide, and finally, evaluating the GADO based on the FOCA evaluation methodology. See representation Figure 5.2 below of the GADO overview document that was distributed to evaluators to participate in the evaluation of the domain ontology.

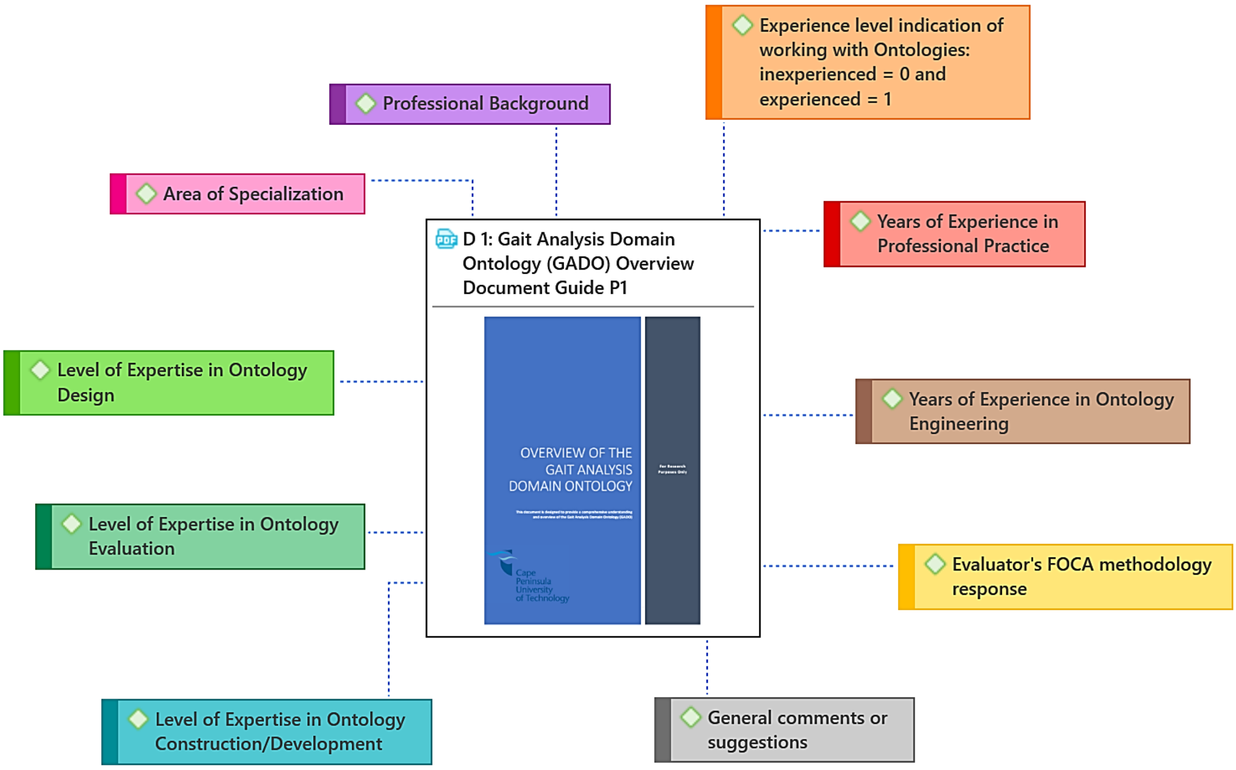


Figure 5.2: GADO overview document guide snapshot

The document guide provided a preparatory overview of the GADO to ensure that evaluators have a common understanding of the ontology they are evaluating. That section of the document is primarily informational, and it aimed to equip evaluators with the necessary knowledge about the structure, classes, properties, individual instances, and querying procedures of the domain ontology. The GADO was evaluated by four experts, indicated as P1, P2, P3, and P4 respectively, each bringing unique expertise and background in ontology engineering and related fields. The composition of the evaluation panel diversely ensured a comprehensive assessment from different perspectives.

Collection of evaluator background information

One of the significant aspects of the evaluation process is understanding the background and expertise of the evaluators. This is vital as the knowledge and experience of evaluators may influence the feedback and evaluation results. In obtaining this information, the evaluators were requested to complete an "Ontology Evaluator Background and Experience Form." This form is qualitative in nature and captures data on the professional background, areas of specialisation, experience in professional practice, and expertise in ontology engineering. Understanding the evaluators' background helps in interpreting their feedback with context and may be used to weigh their inputs based on their relevant experience.

P1 has a professional background in academia and specialises in Artificial Intelligence. They have 2-5 years of experience in professional practice and an equal amount of experience in ontology engineering. Their level of expertise is intermediate in ontology design, ontology construction/development, and ontology evaluation. This evaluator indicated that they are relatively inexperienced in working with ontologies.

P2 hails from industry and is a Systems Developer. With 5-10 years of professional practice and 2-5 years of experience in ontology engineering, they possess advanced skills in ontology design and construction/development and have an intermediate level of expertise in ontology evaluation. This evaluator is experienced in working with ontologies.

P3, with a background in academia, specialises as a Systems Developer. They have accumulated over 10 years of experience in their field of professional practice and 2-5 years in ontology engineering. Like the first evaluator, they have an intermediate level of expertise in ontology design, construction/development, and evaluation. They also indicated being experienced in working with ontologies.

P4 has a research background with a specialisation in Knowledge Representation. They bring over 10 years of experience in professional practice and 5-10 years in ontology engineering. Significantly, they hold expert-level skills in ontology design, construction/development, and evaluation, which is invaluable for the assessment of GADO. This evaluator is also experienced in working with ontologies.

The panel's collective expertise, varying years of experience, and diverse backgrounds contributed to a well-rounded evaluation of the Gait Analysis Domain Ontology. Particularly, the presence of an expert-level evaluator in ontology design, construction, and evaluation ensures that the ontology is assessed against the highest standards in the field. The aforementioned is shown in Table 5.3 below.

Table 5.3: Evaluator profile (refer to **Appendix G**: Evaluators' profiles)

Evaluator Profile	P1	P2	P3	P4
• **Professional Background**	Academia	Industry	Academia	Research
• **Area of Specialization**	Artificial Intelligence	Systems Developer	Systems Developer	Knowledge Representation
• **Years of Experience in Professional Practice**	2-5 years	5-10 years	Above 10 years	Above 10 years
• **Years of Experience in Ontology Engineering**	2-5 years	2-5 years	2-5 years	5-10 years
• **Level of Expertise in Ontology Design**	Intermediate	Advanced	Intermediate	Expert
• **Level of Expertise in Ontology Construction/Development**	Intermediate	Advanced	Intermediate	Expert
• **Level of Expertise in Ontology Evaluation**	Intermediate	Intermediate	Intermediate	Expert
• **Experience level indication of working with Ontologies: inexperienced = 0 and experienced = 1**	0	1	1	1

Evaluation of the GADO by the evaluators

In the closing section of the GADO overview guide, evaluators were asked to provide feedback and general comments on the GADO by employing the FOCA evaluation methodology. This methodology is quantitative and involves assigning grades to twelve questions grouped into five goals. Each evaluator assigned a grade (25, 50, 75, or 100) to the questions, and the means of each goal were calculated to summarise the assessment review.

The FOCA evaluation methodology enables a structured and quantifiable measure of the ontology's fitness for its intended purpose. It allows for a goal-oriented assessment, wherein different characteristics of the ontology such as its structure, consistency, and utility are evaluated against predefined criteria.

Each evaluator had to complete Table 5.4 below, by answering twelve (12) out of thirteen (13) questions and assigned a grade according to their individual assessment as per the FOCA evaluation methodology. The Goal Question Metric (GQM) based on FOCA was used.

Each question represents one of the ontology quality criteria, and is associated with five goals:

i. **Goal 1**: Competency questions and reuse.

ii. **Goal 2**: Ontology terms satisfy criteria and meet the expected level.

iii. **Goal 3**: Contradictions or invalid (re)use of terms in the ontology.

iv. **Goal 4**: Reasoning and reasoner performance.

v. **Goal 5**: Ontology validation and consistency between the modelled ontology and design.

Table 5.4: Evaluators grading of the GADO

Goal	No.	Question (Grade each question either 25, 50, 75, or 100 as per FOCA)	Grade	Mean
Goal 1	Q1	Were the competency questions well-defined?		
	Q2	Were the competency questions answered?		
	Q3	Did the ontology reuse other ontologies?		
Goal 2	Q4	~~Did the ontology impose a minimal ontological commitment?~~		
	Q5	Did the ontology impose a full ontological commitment?		
	Q6	Are the ontology properties coherent with the domain?		
Goal 3	Q7	Are there contradictory axioms?		
	Q8	Are there redundant axioms?		
Goal 4	Q9	Does the reasoner bring modelling errors?		
	Q10	Does the reasoner perform quickly?		
Goal 5	Q11	Is the documentation consistent with the modelling?		
	Q12	Were the concepts well written?		
	Q13	Are there annotations in the ontology bringing the definitions of the concept?		

The FOCA evaluation methodology was employed to calculate the total quality of the ontology based on the evaluators' assessments. The grade scores and mean values provided by the evaluators were used as inputs for the FOCA regression model formula, which respectively produced the estimated total quality for each evaluator's assessment. These values were then plugged into the beta regression model formula to calculate the estimated total quality of the ontology. The respective grades assigned by evaluators are listed below in Table 5.5.

Table 5.5: Evaluator grading scores for the GADO

Evaluator Gradings	P1	P2	P3	P4
Goal 1				
Q1	100	100	75	75
Q2	100	100	100	75
Q3	100	0	100	0
Mean (CovS)	100	66.67	91.67	50
Goal 2				
Q5	100	50	100	75
Q6	100	100	100	100
Mean (CovC)	100	75	100	87.5
Goal 3				
Q7	50	75	100	50
Q8	50	100	75	100
Mean (CovR)	50	87.5	87.5	75
Goal 4				
Q9	0	100	100	100
Q10	100	100	100	100
Mean (CovCp)	50	100	100	100
Goal 5				
Q11	100	100	100	50
Q12	100	100	100	75
Q13	100	75	100	100
Mean	100	91.67	100	75

In order to calculate the total quality, the following variables were considered:

CovS: The mean value of goal 1.

CovC: The mean value of goal 2.

CovR: The mean value of goal 3.

CovCp: The mean value of goal 4.

LExp: The evaluator's experience, with 1 representing experienced and 0 not experienced.

NI: If a goal cannot be evaluated or any question cannot be evaluated, NI is set to 1.

Sb: Set to 1 because all goals were assessed.

Co: Set to 1 because all goals were assessed.

Re: Set to 1 because all goals were assessed.

Cp: Set to 1 because all goals were assessed.

Estimated total quality calculation for P1:

$$\hat{\mu}_i = \frac{\exp\{-0.44+0.03(CovS\times Sb)i+0.02(CovC\times Co)i+0.01(CovR\times Re)i+0.02(CovCp\times Cp)i-0.66LExpi-25(0.1\times NI)i\}}{1+\exp\{-0.44+0.03(CovS\times Sb)i+0.02(CovC\times Co)i+0.01(CovR\times Re)i+0.02(CovCp\times Cp)i-0.66LExpi-25(0.1\times NI)i\}}$$

CovS = 100

CovC = 100

CovR = 50

CovCp = 50

LExp = 0

NI = 0

Sb = 1

Co = 1

Re = 1

Cp = 1

Numerator = exp(-0.44 + 0.03(100 × 1) + 0.02(100 × 1) + 0.01(50 × 1) + 0.02(50 × 1) - 0.66 × 0 - 25(0.1 × 0))

= exp(-0.44 + 3 + 2 + 0.5 + 1)

= exp(6.06)

≈ 428.5 (approximately)

Denominator = 1 + exp(-0.44 + 0.03(100 × 1) + 0.02(100 × 1) + 0.01(50 × 1) + 0.02(50 × 1) - 0.66 × 0 - 25(0.1 × 0))

= 1 + exp(6.06)

≈ 429.5 (approximately)

$\hat{\mu}$ = 428.5 / 429.5 ≈ 0.9977 (rounded to four decimal places)

Hence, the estimated total quality of the ontology is approximately <u>0.9977</u> for **P1**.

Estimated total quality calculation for P2:

$$\hat{\mu}_i = \frac{\exp\{-0.44+0.03(CovS \times Sb)_i+0.02(CovC \times Co)_i+0.01(CovR \times Re)_i+0.02(CovCp \times Cp)_i-0.66LExp_i-25(0.1 \times NI)_i\}}{1+\exp\{-0.44+0.03(CovS \times Sb)_i+0.02(CovC \times Co)_i+0.01(CovR \times Re)_i+0.02(CovCp \times Cp)_i-0.66LExp_i-25(0.1 \times NI)_i\}}$$

CovS = 66.67

CovC = 75

CovR = 87.5

CovCp = 100

LExp = 1

NI = 0

Sb = 1

Co = 1

Re = 1

Cp = 1

Numerator = exp(-0.44 + 0.03(66.67 × 1) + 0.02(75 × 1) + 0.01(87.5 × 1) + 0.02(100 × 1) - 0.66 × 1 - 25(0.1 × 0))

= exp(-0.44 + 2 + 1.5 + 0.875 + 2 - 0.66)

= exp(5.275)

≈ 195.1 (approximately)

Denominator = 1 + exp(-0.44 + 0.03(66.67 × 1) + 0.02(75 × 1) + 0.01(87.5 × 1) + 0.02(100 × 1) - 0.66 × 1 - 25(0.1 × 0))

= 1 + exp(5.275)

≈ 196.1 (approximately)

$\hat{\mu}$ = 195.1 / 196.1 ≈ 0.9949 (rounded to four decimal places)

Hence, the estimated total quality of the ontology is approximately <u>0.9949</u> for **P2**.

Estimated total quality calculation for P3:

$$\hat{\mu}_i = \frac{\exp\{-0.44+0.03(CovS\times Sb)i+0.02(CovC\times Co)i+0.01(CovR\times Re)i+0.02(CovCp\times Cp)i-0.66LExpi-25(0.1\times NI)i\}}{1+\exp\{-0.44+0.03(CovS\times Sb)i+0.02(CovC\times Co)i+0.01(CovR\times Re)i+0.02(CovCp\times Cp)i-0.66LExpi-25(0.1\times NI)i\}}$$

CovS = 91.67

CovC = 100

CovR = 87.5

CovCp = 100

LExp = 1

NI = 0

Sb = 1

Co = 1

Re = 1

Cp = 1

Numerator = exp(-0.44 + 0.03(91.67 × 1) + 0.02(100 × 1) + 0.01(87.5 × 1) + 0.02(100 × 1) - 0.66 × 1 - 25(0.1 × 0))

= exp(-0.44 + 2.75 + 2 + 0.875 + 2 - 0.66)

= exp(6.525)

≈ 680.7 (approximately)

Denominator = 1 + exp(-0.44 + 0.03(91.67 × 1) + 0.02(100 × 1) + 0.01(87.5 × 1) + 0.02(100 × 1) - 0.66 × 1 - 25(0.1 × 0))

= 1 + exp(6.525)

≈ 681.7 (approximately)

$\hat{\mu}$ = 680.7 / 681.7 ≈ 0.9985 (rounded to four decimal places)

Hence, the estimated total quality of the ontology is approximately 0.9985 for **P3**.

Estimated total quality calculation for P4:

$$\hat{\mu}_i = \frac{\exp\{-0.44+0.03(CovS \times Sb)_i+0.02(CovC \times Co)_i+0.01(CovR \times Re)_i+0.02(CovCp \times Cp)_i-0.66LExp_i-25(0.1 \times NI)_i\}}{1+\exp\{-0.44+0.03(CovS \times Sb)_i+0.02(CovC \times Co)_i+0.01(CovR \times Re)_i+0.02(CovCp \times Cp)_i-0.66LExp_i-25(0.1 \times NI)_i\}}$$

CovS = 50

CovC = 87.5

CovR = 75

CovCp = 100

LExp = 1

NI = 0

Sb = 1

Co = 1

Re = 1

Cp = 1

Numerator = exp(-0.44 + 0.03(50 × 1) + 0.02(87.5 × 1) + 0.01(75 × 1) + 0.02(100 × 1) - 0.66 × 1 - 25(0.1 × 0))

= exp(-0.44 + 1.5 + 1.75 + 0.75 + 2 - 0.66)

= exp(5.9)

≈ 365.8 (approximately)

Denominator = 1 + exp(-0.44 + 0.03(50 × 1) + 0.02(87.5 × 1) + 0.01(75 × 1) + 0.02(100 × 1) - 0.66 × 1 - 25(0.1 × 0))

= 1 + exp(5.9)

≈ 366.8 (approximately)

$\hat{\mu}$ = 365.8 / 366.8 ≈ 0.9973 (rounded to four decimal places)

Hence, the estimated total quality of the ontology is approximately <u>0.9973</u> for **P4**.

The following calculations were performed to determine the overall mean value for the GADO from the evaluators' estimated total quality values:

0.9977 + 0.9949 + 0.9985 + 0.9973 = 3.9884

3.9884 / 4 = 0.9971

The overall mean of the estimated total quality for the evaluators is 0.9971 seen in Figure 5.3.

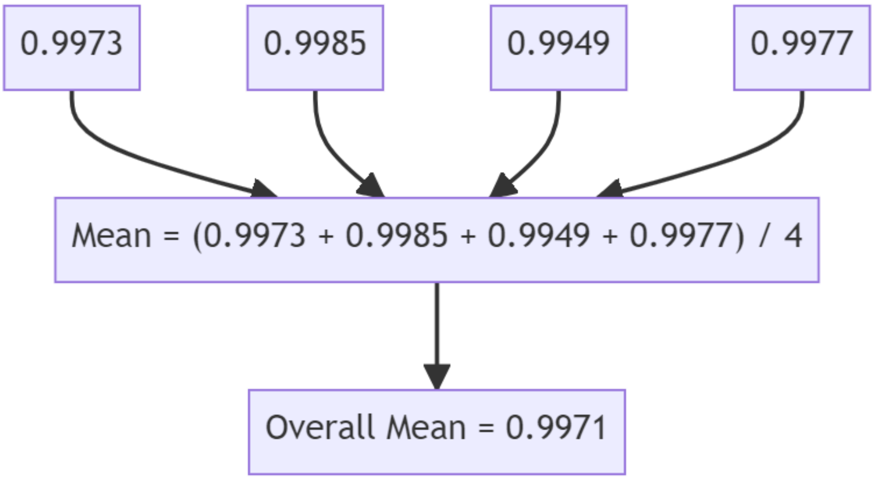


Figure 5.3: The calculated mean of the evaluators' combined estimated total quality

This is the objective evaluation presented in the FOCA methodology by evaluators, and is close to 1, which suggests that the Gait Analysis Domain Ontology is of a very high quality.

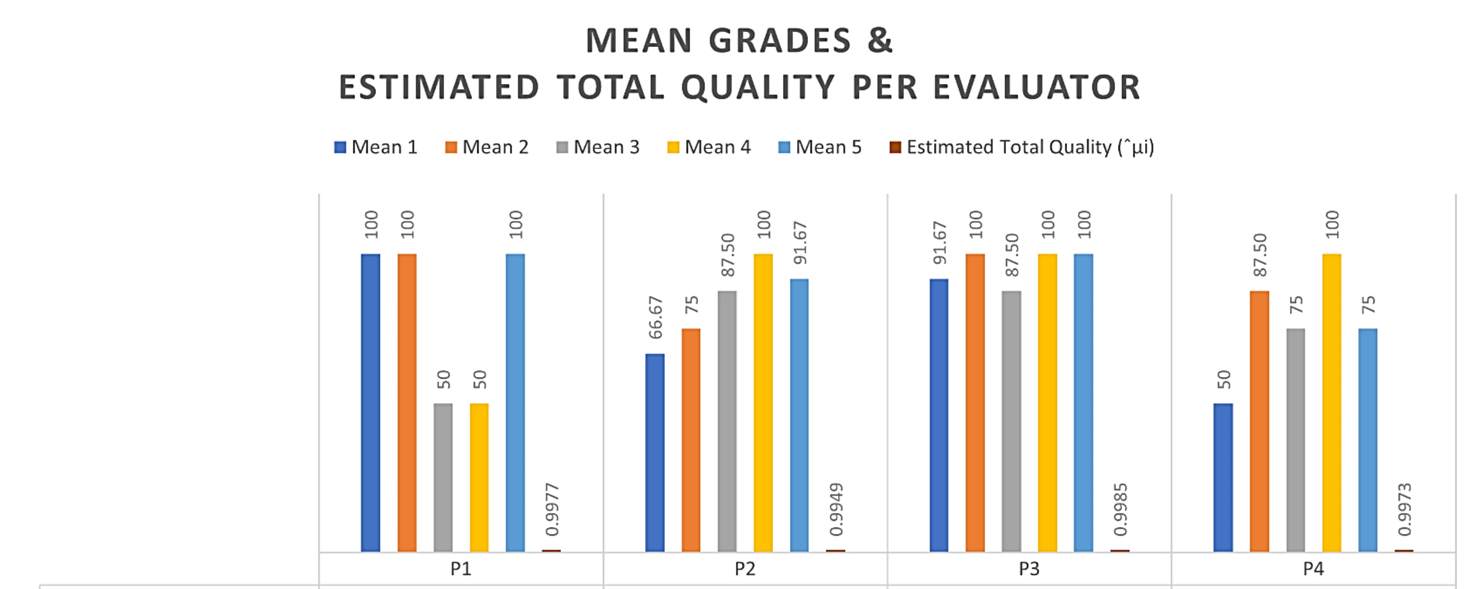
5.1.1.4 Discussion and Analysis

The evaluation of the Gait Analysis Domain Ontology was conducted by a panel of four experts (P1, P2, P3, and P4) from diverse backgrounds. This section provides an analysis of the participants' responses, considering their respective professional backgrounds, areas of specialisation, years of experience, and levels of expertise in ontology engineering.

The following Table 5.6 summarises the mean values and the calculated estimated total quality of the Gait Analysis Domain Ontology for each evaluator and are graphically displayed in Figure 5.4.

Table 5.6: Evaluators mean grade scores and estimated total quality

Evaluator	Mean 1	Mean 2	Mean 3	Mean 4	Mean 5	Estimated Total Quality (ˆµi)
P1	100	100	50	50	100	0.9977
P2	66.67	75	87.50	100	91.67	0.9949
P3	91.67	100	87.50	100	100	0.9985
P4	50	87.50	75	100	75	0.9973



	P1	P2	P3	P4
Mean 1	100	66.67	91.67	50
Mean 2	100	75	100	87.50
Mean 3	50	87.50	87.50	75
Mean 4	50	100	100	100
Mean 5	100	91.67	100	75
Estimated Total Quality (ˆµi)	0.9977	0.9949	0.9985	0.9973

Figure 5.4: Evaluators estimated total quality and mean grades

P1, with an academic background and specialisation in artificial intelligence, showed polarised responses. High scores were given in most questions, but significantly lower ones for Goal 3 and Goal 4. This may be attributed to P1's intermediate level of expertise and relative inexperience in working with ontologies. The lower scores in Goal 3 and Goal 4 could indicate a lack of experience or comfortability in assessing aspects that may pertain to ontology evaluation.

P2, with an industry background and relatively advanced skills in ontology design and construction, displayed more variation in responses, especially in Goal 1 and Goal 5. This variation could stem from a practical industry perspective where certain criteria in these goals may not align with the practical applications or industry standards. With experience in working with ontologies, P2 might be assessing the ontology's applicability and usefulness in real-world scenarios.

P3, having an academic background with over 10 years of experience in professional practice, scored consistently high across all goals. This might indicate that P3, with an intermediate level of expertise in ontology engineering and experience in working with ontologies, found the GADO to be largely compliant with academic standards and criteria.

P4, having a research background with expert-level skills in ontology engineering, gave varied scores across the goals. This variation, especially in Goal 1 and Goal 5, may indicate a more critical evaluation from an expert perspective. Since P4 is well-experienced, the areas with lower scores might require improvements to meet higher standards in knowledge representation.

The panel’s distinct expertise contributed to a well-rounded evaluation of the GADO. The variance in responses across goals suggests that the GADO has different levels of compliance with criteria important to academia, industry, and research. For example, lower scores in Goal 1 by P2 and P4 may imply the need for improvements in aspects related to design, while the lower scores by P1 in Goal 3 and Goal 4 might reflect areas requiring further clarity or development. In addition, consistently high scores by P3 could imply the GADO’s strong alignment with academic standards. The results suggest the importance of incorporating feedback from various stakeholders to enhance the utility and applicability of the Gait Analysis Domain Ontology across different knowledge areas.

Furthermore, the FOCA methodology furnished both objective and subjective facets to the evaluation. The researcher's assessment generated an expression with a resultant value of approximately 0.9955. Since this value gravitates toward 1, it can be subjectively interpreted as an indication of high quality.

Concurrently, a cohort of independent external evaluators undertook an objective assessment. Through their critical scrutiny employing the FOCA methodology, an overall mean of 0.9971 was derived from the estimated total quality values. This score, being close to 1, objectively substantiates the high quality of the Gait Analysis Domain Ontology.

In addition, the evaluators provided general comments and suggestions, which highlight several key areas for improvement and consideration in the GADO. While there is a general consensus on the value and potential of the ontology, it is apparent that documentation, standardisation, and planning for sustainability are critical aspects that require attention that can be synthesised as follows:

i. Refining and standardising the ontology’s construction and documentation.
ii. Developing a comprehensive ontology requirements specification document.
iii. Implementing strategies for long-term maintenance, versioning, and performance optimisation.
iv. Engaging with a diverse group of stakeholders to continuously evaluate and enhance the GADO based on evolving needs and knowledge in the domain of gait analysis.

5.1.2 Validation of the GADO

Ontology validation is the process of ensuring that the ontology correctly represents the knowledge in the domain it is supposed to cover (Alsanad et al., 2019). This is more about the semantic aspects and the ontology's applicability in real-world scenarios.

5.1.2.1 Correctness

The concluding step in evaluating the quality of the ontology meant checking for errors and inconsistencies. This was done by performing error checking, such as consistency checking, to ensure the ontology was accurate and followed the appropriate standards and guidelines. Using automated reasoning tools, HermiT, and Pellet, was crucial for ensuring the ontology's stability, logical consistency, and compatibility. This was important for confirming that the ontology was compatible with other ontologies in the same domain and could be integrated with other systems and tools.

The gait analysis domain ontology was successfully processed by the HermiT and Pellet reasoners, two popular and powerful reasoning engines used in the field of ontology engineering. The HermiT and Pellet reasoners could efficiently handle large and complex ontologies. They have been extensively tested and used in various domains, including health and biomedical applications. These reasoners' successful processing of the domain ontology demonstrated their ability to accurately infer new knowledge from the given ontology for detecting inconsistencies, classifying individuals into different classes, and inferring new relationships between classes and individuals. This provided valuable insights and knowledge that could aid in diagnosing, treating, and managing gait-related diseases.

The HermiT reasoner in Protégé took slightly longer to process due to several factors like the size and complexity of the domain ontology being processed, the type and number of axioms and logical expressions in the ontology, and the computational resources available to the reasoner. This reasoning process is initiated within Protégé as observed in Figure 5.5 below.

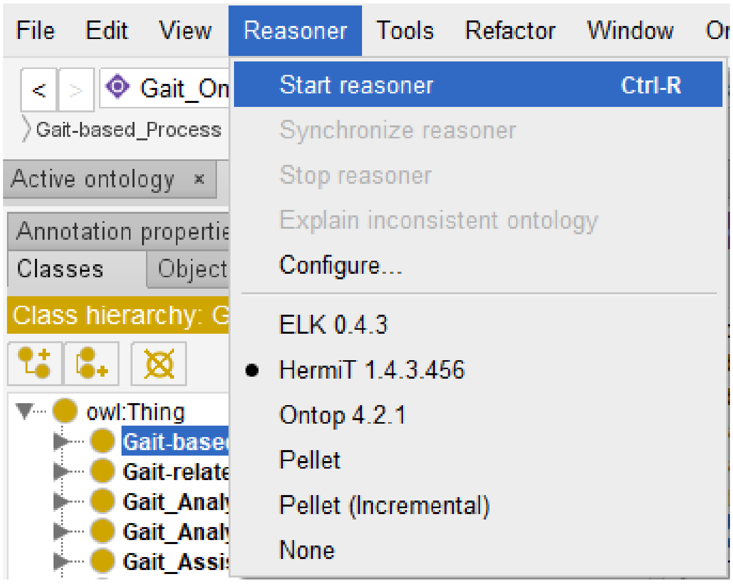


Figure 5.5: Ontology reasoners

If the ontology is large and complex, with many axioms and complex logical expressions, it can take longer for the reasoner to process and draw conclusions. Similarly, it may take longer to complete if the reasoner runs on a computer with limited resources, such as memory or processing power. In addition, some reasoning tasks may be inherently complex and computationally intensive, such as classification or consistency checking. In these cases, it was not practicable to speed up the reasoning process because executing the reasoning tasks was processed in mere seconds and completed consistency checking. The HermiT reasoner processed the ontology in 18708 ms (milliseconds), and the Pellet reasoner finished processing the ontology in 658 ms, respectively, as shown in Figure 5.6.

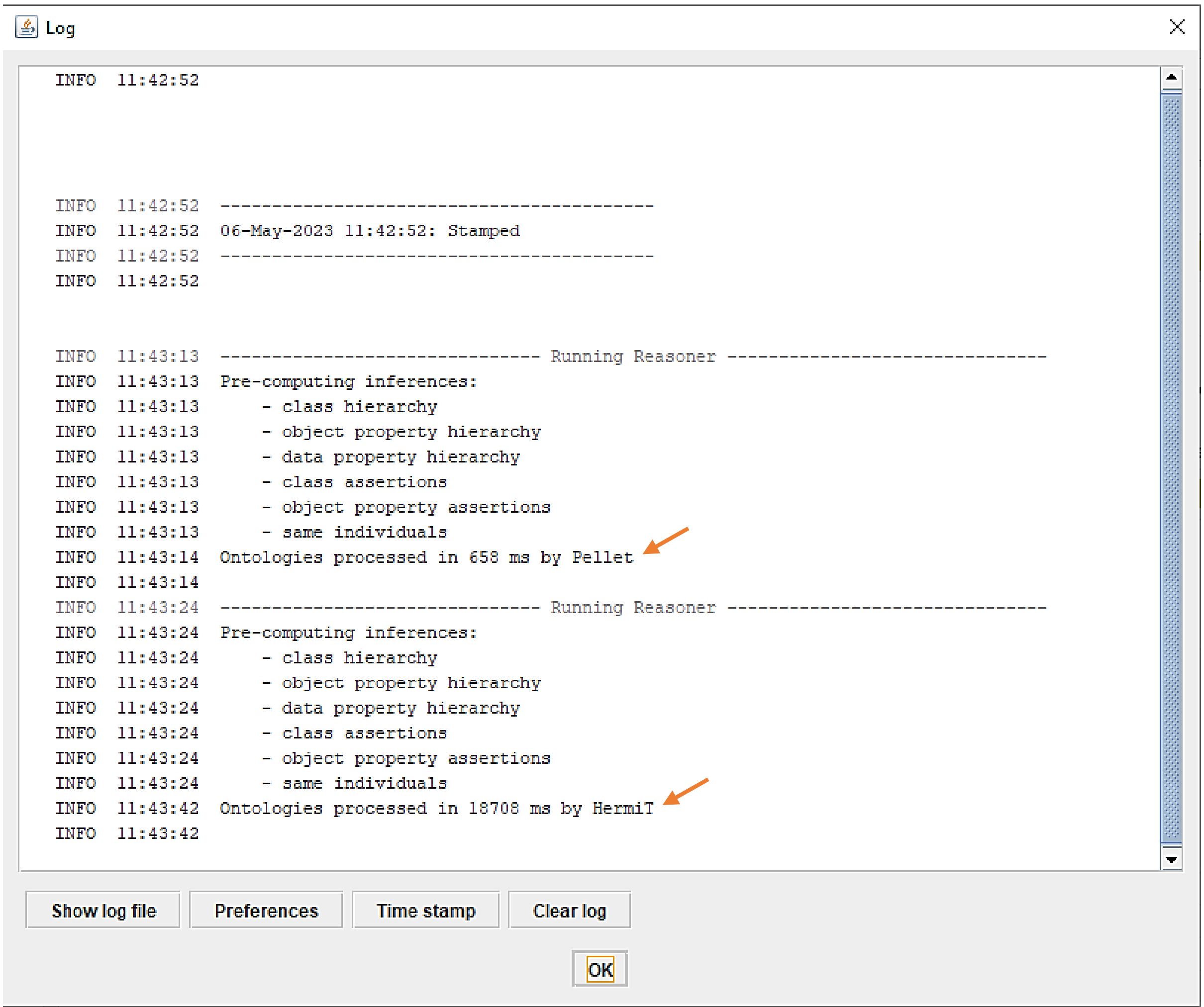


Figure 5.6: Log results of the ontology reasoners

In order to determine how much faster the Pellet reasoner was compared to the HermiT reasoner, the speedup factor was calculated. The speedup factor is the ratio of the time the HermiT reasoner takes to process the ontology to the time the Pellet reasoner takes to process the same ontology.

The speedup factor was calculated as follows:

Speedup factor = Time taken by HermiT reasoner / Time taken by Pellet reasoner

Substituting the given values:

Speedup factor = 18708 ms / 658 ms

Speedup factor ≈ 28.41

Therefore, the Pellet reasoner was approximately 28.41 times faster than the HermiT reasoner in processing the given ontology.

To calculate the difference in processing time between the two reasoners, subtract the time taken by Pellet from the time taken by HermiT:

18078 ms - 658 ms = 18050 ms

Consequently, HermiT took 18050 ms (or approximately 18.05 seconds) longer than Pellet to process the ontology.

5.1.2.2 DL query

Formulating DL queries, one can evaluate and test the domain ontology by checking if the returned results correspond to the expected outcomes. This helped to validate and verify the ontology design and implementation, identify any errors and inconsistencies, and detect any potential omissions. In general, DL query was a valuable technique for the researcher in the ontology engineer's capacity to interact with and evaluate the ontology, as well as to extract meaningful insights and knowledge. This falls under validation as it tests whether the ontology correctly represents the domain knowledge and can answer domain-specific queries.

Querying the ontology enabled the researcher to check if it captured the required knowledge that it should represent and whether it could answer the questions that needed to be answered. Queries can help assess the expressiveness and completeness of the ontology and identify potential inconsistencies, redundancies, or gaps in the knowledge representation. The use of the Description Logic query language ensured affordances for specifying and reasoning about the semantics of the ontology.

The following Protégé DL query was a description logic expression that specified a complex class expression to query the ontology to display the gait patients who also have a gait assistive technology assigned, respectively.

```
Gait_Assistive_Technology and isRecommendedTo some Gait_patient or (hasGait_assistive_solution some xsd:string) or Gait_patient
```

"Gait_Assistive_Technology": This is a concept or class in the ontology. It represents assistive technologies designed to aid patients with gait-related impairments.

"isRecommendedTo": This is a role or property in the ontology. It relates a ‘Gait_Assistive_Technology’ to a ‘Gait_patient’ that it is recommended for. For example, a walker may be recommended to a patient with gait balance issues.

"some": This quantifier in DL specifies that the role "isRecommendedTo" should have at least one filler or value. In this case, it means that a ‘Gait_Assistive_Technology’ must be recommended to at least one ‘Gait_patient’.

"hasGait_assistive_solution": This is another role in the ontology. It relates a ‘Gait_patient’ to a string value that represents the gait assistive solution they require. For example, a patient may require a walker or a cane.

"xsd:string": This is a data type in XML schema definition (xsd) that specifies that the value of "hasGait_assistive_solution" should be a string.

"or": This logical operator in DL specifies that the expression on the left or the expression on the right can be true. In this case, the left expression is "isRecommendedTo some Gait_patient", and the right expression is "hasGait_assistive_solution some xsd:string".

So, the entire expression can be read as follows:

A ‘Gait_Assistive_Technology’ is recommended to at least one ‘Gait_patient’ OR it has a ‘Gait_patient’ who requires a gait assistive solution represented by a string value.

Once queried, the string value was substituted for any ‘Gait_Assistive_Technology’ defined instance and showed the individual instance results such as a walker, orthotic, walking stick, pharmaceutical and braces, along with the individual instance results of ‘Gait_patient’, which is child, adolescent, adult, elderly, and athlete.

This expression defines a class of individuals that are members of the class ‘Gait_Assistive_Technology’ and has the object property “isRecommendedTo” that points to at least one individual that is a member of the class ‘Gait_patient’.

The above DL query was done in Protégé, and the results are expressed in Figure 5.7 below.

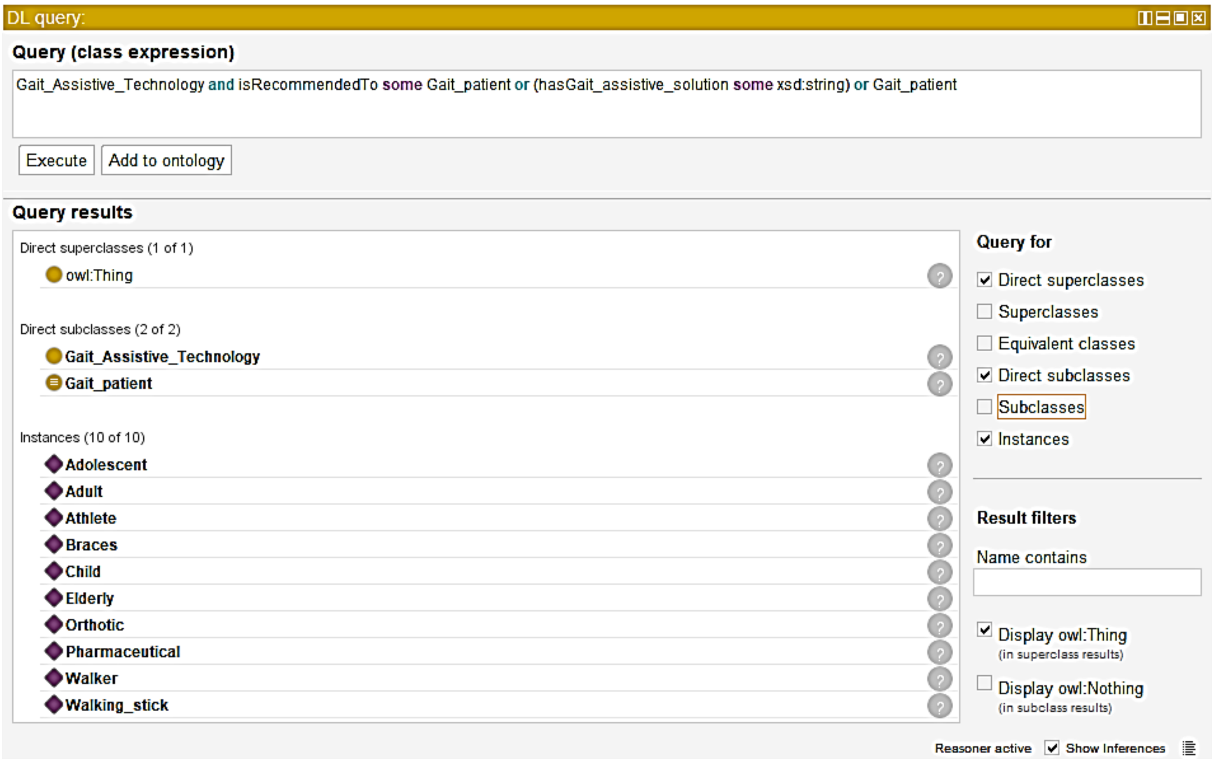


Figure 5.7: DL query class expression

5.1.2.3 SPARQL query

SPARQL queries can be employed to validate the data within the ontology, ensuring that it is consistent and accurate. This is crucial for maintaining the quality and reliability of the knowledge base. Ontologies are used to represent and formalise knowledge in a specific domain, and in this case, the domain is gait analysis for gait-related disorders. SPARQL querying performs a focal role in the development, utilisation, and evaluation of the domain ontology for gait-related matters by enabling efficient data retrieval, analysis, validation, and integration. Conclusively, it allows gait experts and other stakeholders to extract valuable insights from the ontology, ultimately leading to better understanding, diagnosis, and management of gait-related diseases. It checks whether the ontology can answer specific queries and thus validates the ontology's utility in real-world applications.

These query results have the potential to enhance understanding and validate the ability to extract, manipulate, analyse, and retrieve information efficiently. They serve as evidence of the intelligent reasoning capabilities derived from the domain ontology, thereby providing valuable insights to stakeholders. Figure 5.8 below illustrates the GADO architecture used for SPARQL query execution.

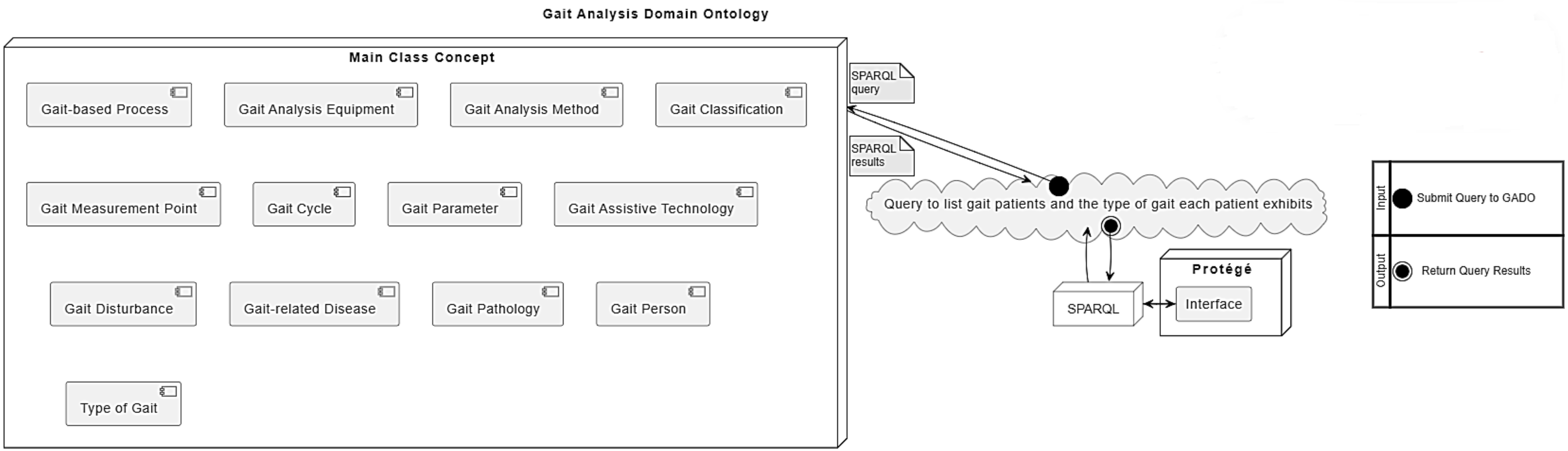


Figure 5.8: SPARQL querying performed by the GADO

The SPARQL query below was executed to search and retrieve information about the gait patients and their associated type of gait exhibition from the GADO.

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX Gait_Ontology:
<http://www.semanticweb.org/marthinust/ontologies/2022/8/Gait_Ontology#>

SELECT ?gaitPatient ?typeOfGait
WHERE {
   ?gaitPatient a Gait_Ontology:Gait_patient .
   ?gaitPatient Gait_Ontology:exhibits ?typeOfGait .
   ?typeOfGait a Gait_Ontology:Type_of_Gait .
}
```

To promote better understanding, the above query is broken into its components below:

PREFIX declarations: These lines define short aliases for commonly used namespaces in RDF and OWL to make the query more readable and compact.

rdf: Namespace for RDF syntax.

owl: Namespace for OWL.

rdfs: Namespace for RDF Schema.

xsd: Namespace for XML Schema data types.

Gait_Ontology: Custom namespace for the specific Gait Analysis Domain Ontology being used.

SELECT clause: Specifies the variables that the query will return. In this case, the query will return two variables: ?gaitPatient and ?typeOfGait.

WHERE clause: Contains a set of triple patterns that must be satisfied for the query to return results. The query looks for the following patterns:

?gaitPatient a Gait_Ontology:Gait_patient: This pattern searches for all individuals (?gaitPatient) that are instances of the Gait_patient class in the Gait_Ontology.

In other words, it looks for all gait patients.

?gaitPatient Gait_Ontology: exhibits ?typeOfGait: This pattern searches for all relationships where a gait patient exhibits a certain type of gait (?typeOfGait).

The relationship is represented by the 'exhibits' property in the Gait_Ontology.

?typeOfGait a Gait_Ontology:Type_of_Gait: This pattern searches for all instances (?typeOfGait) that belong to the Type_of_Gait class in the Gait_Ontology.

In plain English, it looks for all types of gait in the Gait Analysis Domain Ontology. When all three patterns are satisfied for a combination of ?gaitPatient and ?typeOfGait, the query will return the corresponding values as a result. Therefore, the query returns a list of gait patients and the type of gait each patient correspondingly exhibits as displayed in Figure 5.9 below.

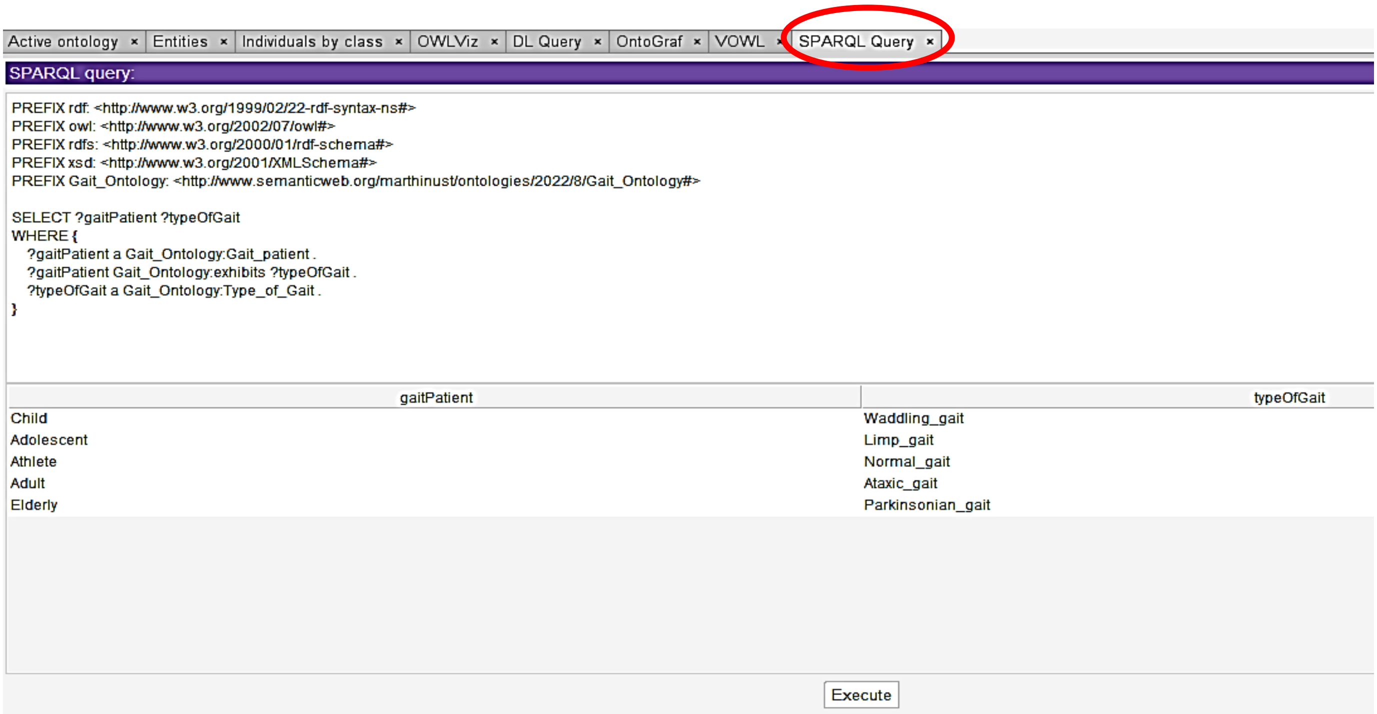


Figure 5.9: SPARQL query for the type of gait exhibited by a gait patient

Another SPARQL query was performed to search and retrieve information listing gait patients and the gait assistive technologies assigned to each.

```
PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>
PREFIX owl: <http://www.w3.org/2002/07/owl#>
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>
PREFIX                                                    Gait_Ontology:
<http://www.semanticweb.org/marthinust/ontologies/2022/8/Gait_Ontology#>
SELECT ?gaitPatient ?assistiveTechnology
WHERE {
   ?gaitPatient a Gait_Ontology:Gait_patient .
   ?gaitPatient Gait_Ontology:isAssignedWith ?assistiveTechnology .
   ?assistiveTechnology a Gait_Ontology:Gait_Assistive_Technology .
}
```

The above query declarations are explained in the components as follow:

PREFIX rdf: <http://www.w3.org/1999/02/22-rdf-syntax-ns#>: This defines the namespace for RDF, a foundational standard for representing data in the Semantic Web.

PREFIX owl: <http://www.w3.org/2002/07/owl#>: This defines the namespace for OWL, the Web Ontology Language.

PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>: This defines the namespace for RDFS, a schema language for RDF.

PREFIX xsd: <http://www.w3.org/2001/XMLSchema#>: This defines the namespace for XML Schema, a language for defining the structure of XML documents.

PREFIX Gait_Ontology: <http://www.semanticweb.org/marthinust/ontologies/2022/8/Gait_Ontology#>: This defines a custom namespace for the Gait Analysis Domain Ontology.

SELECT Clause: SELECT ?gaitPatient ?assistiveTechnology: This indicates that the query will return two types of variables: ?gaitPatient and ?assistiveTechnology.

WHERE Clause: The WHERE clause contains the conditions that the data must satisfy to be returned by the query.

?gaitPatient a Gait_Ontology:Gait_patient : This line finds all instances where a subject is of type Gait_patient. In other words, it is looking for all gait patients.

?gaitPatient Gait_Ontology:isAssignedWith ?assistiveTechnology : This line establishes a relationship between gait patients and some form of assistive technology. It is looking for all gait patients who have been assigned a specific type of assistive technology.

?assistiveTechnology a Gait_Ontology:Gait_Assistive_Technology : This line ensures that the ?assistiveTechnology variable represents instances of the Gait_Assistive_Technology class.

In brief, the SPARQL query was executed in the ontology editor Protégé to retrieve all gait patients and the specific gait assistive technologies assigned to them from the GADO as shown in Figure 5.10.

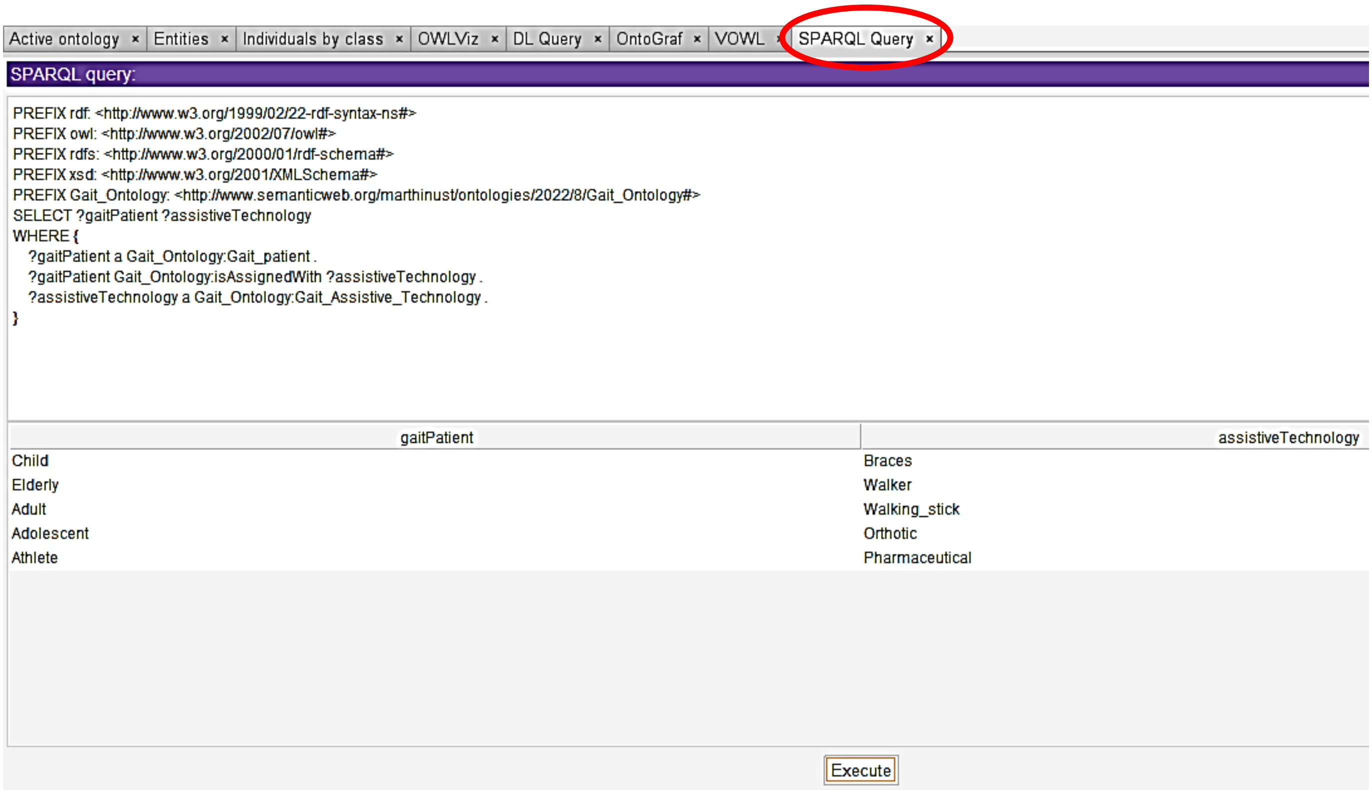


Figure 5.10: SPARQL query listing gait patients and the gait assistive technology assigned

5.2 Summary

In summary, the Gait Analysis Domain Ontology (GADO) underwent evaluation using the FOCA methodology with expert evaluators scrutinising its structure and logic, including querying the ontology. The evaluators affirmed its consistency, well-organised relationships, and domain-specific content, deeming it intuitive and comprehensible for both humans and machines. The evaluation yielded positive outcomes, indicating that the GADO is apt for gait analysis applications, as it satisfies quality criteria including correctness, domain task alignment, and content richness. Furthermore, the evaluation highlighted the ontology's potential to improve data quality, enhance efficiency, and reduce errors, which was showcased through practical use cases.

CHAPTER 6: SUMMARY, CONTRIBUTIONS AND RECOMMENDATIONS

The closing chapter of the thesis provides a comprehensive summary and concluding remarks of the study on the successful development of the Gait Analysis Domain Ontology (GADO) for the treatment of gait-related diseases, discussing the benefits of utilising a domain ontology. The chapter revisits the research objectives, summarises the findings, highlights the contributions, and offers recommendations for proposed future work.

6.1 Summary of the study

The research underscored the transformative potential of a gait analysis domain ontology. By harnessing standardised terminology and concepts, it fosters enhanced communication and data sharing, culminating in more effective treatment strategies and improved patient outcomes. The ontology stands as a pragmatic tool, underpinning decision-making in the treatment of gait-related diseases. Through meticulous research design and execution, the GADO was conceptualised, designed, developed, and evaluated.

At the outset of this research, several objectives were set to guide the study. These objectives are reflected here:

Objective 1: Identify the requirements of a domain ontology that can support decision-making in the treatment of gait-related diseases.

The research journey began with identifying the need for a domain ontology that could support decision-making in the treatment of gait-related diseases. The research successfully identified the requirements through a comprehensive domain analysis that was informed by literature, online web portals and biomedical ontologies. This provided a solid foundation for the subsequent stages of developing the GADO ontology. Applying a domain ontology has several notable benefits, including providing a clear and structured representation of concepts and relationships within the domain. This can help to improve the understanding of gait-related diseases and ultimately aid researchers, gait experts, and patients in identifying underlying causes and effective treatments. In addition, using a shared ontology can enhance communication and information sharing between stakeholders, leading to more effective collaborations and a more efficient dissemination of knowledge.

Objective 2: Design a domain ontology that can support decision-making in the treatment of gait-related diseases.

A structured ontology design was formulated, capturing the essential concepts, relationships, and attributes pertinent to gait analysis. Once the ontology's scope, structure, and data were established, the next step was to formalise it into a machine-understandable format by using OWL. Competency questions and statements of anticipated knowledge were also used to conceptualise the representation of the domain. In analysing the ontology's ability to answer these questions correctly, it tacitly ensured that the ontology was thoroughly representative of the domain and suitably fit for its envisioned purpose.

Objective 3: Develop a domain ontology to support decision-making in the treatment of gait-related diseases.

Protégé was used to develop the ontology, ensuring that it was both comprehensive and aligned with the identified requirements. The ontology, built using Protégé, has proven to be an asset in understanding and managing gait-related issues. Developing a domain ontology required a thorough domain analysis, a well-structured ontology design, and an appropriate implementation and evaluation process. Notably, using Protégé was found to be particularly useful for ontology engineering with its user-friendly features, various functions, handy plugins, and built-in automated reasoners that assisted with ontology building for reasoning, visualising, and interacting with interrelated data. Furthermore, Protégé has proven to be an efficient tool for accomplishing these steps.

Objective 4: Evaluate the domain ontology in terms of domain task fit, content richness, and correctness.

The ontology underwent rigorous evaluation, revealing that it met the criteria for an effective ontology in the domain. The FOCA evaluation methodology further affirmed its quality and applicability. Through the evaluation of the ontology which centered on criteria attributed to domain task fit, content richness, and correctness, it was possible to refine and improve the ontology to ensure it was not only comprehensive and correct but also useful. These improvements led to more efficient analysis and management of gait-related matters, delivering better results more quickly and reliably. This kind of intelligent reasoning was further reinforced semantically by logic querying with the information retrieval of query results. Therefore, it was sensible to use multiple tools and approaches to evaluate ontology quality and validate ontology content against real-world use cases.

The evaluation of the quality attributes of the domain ontology was a pivotal phase in ensuring that the ontology was accurate, robust, and practical. The evaluation required assessing the ontology based on the domain task fit, content richness, and correctness to identify areas for improvement and refinement that could better support the diagnosis and treatment of gait-related conditions. Description Logics and SPARQL querying were respectively deployed to formalise this information, and the resulting ontology was encoded in web ontology language (OWL) using the Protégé software editor. The HermiT and Pellet reasoners, alongside FOCA methodology, were also commissioned to further verify and validate the developed ontology. The evaluators' feedback was instrumental in guiding the refinement and future development of the GADO. Thus, by addressing the identified areas of improvement, it can be optimised for clarity, efficiency, and broader impact in the domain of gait analysis.

6.2 Contributions

This study aimed to create a knowledge-domain ontology that exhibits artificial intelligence reasoning to support gait experts in the treatment of gait-related diseases. The study's contribution is categorised into theoretical, methodological, and practical.

The theoretical contribution of this study lies in its contribution to the existing theory on gait-related subjects and ontology building. The knowledgebase domain ontology artefact created enables intelligent reasoning and semantic capabilities that can be leveraged in decision support for the treatment of gait-related diseases. This theory could form the basis for future research in the area, leading to further development of ontologies for other medical conditions.

The methodological contribution of this study was a sustained methodology for developing a flexible and extensible domain ontology that can be iteratively updated with future knowledge. By adopting the OD-101 ontology engineering approach, the study addressed the challenges of existing ontology development methodologies, making it more effective yet practical for better ontology maintenance and refinement to ensure sustained evolution. This methodology facilitates active evaluation, as it can pinpoint the ontology's strengths and weaknesses.

In terms of practical contribution, the study resulted in a knowledgebase artefact that can infer knowledge and support gait experts in decision-making related to gait-related diseases. The ontology incorporates artificial intelligence reasoning and semantic capabilities, enabling it for intelligent knowledge inferencing and providing dynamic decision support for treating and managing gait-related conditions to advance patient healthcare. The ontology queries, powered by the rich semantic structure of the ontology, can provide insights that traditional database queries may miss, thereby enhancing the overall practical utility of the Gait Analysis Domain Ontology.

6.3 Recommendations

Using a domain ontology in the treatment of gait-related diseases has offered numerous benefits, including improved understanding of the domain, enhanced communication between different stakeholders, and better data integration, which could lead to systematic accuracy and efficiency of these situations, plus the methodical operation of a user-friendly application. The ontology used competency questions to ensure accuracy and completeness while facilitating interchange. Furthermore, it supported the development of a decision support system by providing a solid foundation of domain data and knowledge. Incipiently, the study aimed to develop a domain ontology for gait-related concepts that could provide a basis for improved decision-support to gait experts in treating of gait-related diseases. The study's objectives were to identify the domain ontology's requirements, formulate the ontology's design, develop the ontology, and evaluate its quality attributes. The results of the study showed that the developed ontology met the identified requirements and had high levels of domain task fit, content richness, and correctness. This suggests that the ontology can provide a useful tool for improving the treatment of gait-related diseases.

6.4 Future work

Increased efficiency was another benefit of using a domain ontology. The domain ontology can be used to support the development of decision support systems and other applications for the management of gait-related diseases, which can lead to increased proficiency in diagnosing and treating gait-related diseases. By providing a standardised framework for decision-making, the ontology can assist in reducing errors and streamline processes. The use of a domain ontology can also lead to better data integration. Hence, the ontology can ensure that data from disparate sources are integrated and interpreted consistently, providing a more holistic understanding of the disease and treatment. This can help researchers and gait experts to make more informed decisions and better patient welfare.

Ontology-based data representation and querying promote data interoperability between different systems and applications. This facilitates data sharing and reuse, as well as the development of new applications that build upon existing data and knowledge. Ontology querying is often used in AI-driven applications, like intelligent search engines, recommender systems, and decision support systems. These applications leverage the knowledge stored in the ontology to provide more accurate, context-aware, and customised results. Therefore, these techniques can be applied to the results of ontology queries to enhance decision-making processes as was showcased. Querying the ontology allowed for more sophisticated, context-rich search and retrieval of data based on the semantics encoded in the ontology, thus proving useful in the various use case scenarios.

These results suggest that using a domain ontology in gait-related treatment can have numerous advantages and should be further explored for potential use in healthcare. There is a need to expand the scope of the ontology to include other related domains, such as rehabilitation, physiotherapy, and sporting activities. This would enhance the usefulness of the ontology in a wider range of clinical settings and increase its potential impact on patient health.

Hence, it is recommended to involve various stakeholders in the development of the ontology, including patients, caregivers, and other healthcare professionals. This would ultimately ensure that the ontology reflects a more comprehensive and accurate representation of the domain and promotes better communication and understanding among different stakeholders.

Lastly, there is a need to explore the potential of using the ontology as a basis for developing other decision support systems and healthcare applications that can support the treatment of gait-related diseases. This can include the development of mobile applications that can help patients track their progress and adherence to treatment plans, as well as tools that can help gait experts make more informed and evidence-based decisions. Machine learning algorithms can be used to learn patterns in the data and make predictions or recommendations. These techniques can be applied to the results of ontology queries to enhance decision-making processes. Furthermore, exploring the potential use of the domain ontology in other decision support systems and healthcare applications is also vital. This will require a thorough understanding of the limitations and strengths of the ontology and identifying areas where it can be leveraged to improve decision-making processes and patient health concerns.

All things considered, building on this groundwork and foundation, future work should prioritise the development of an all-inclusive strategy for the continued growth and refinement of the domain ontology. Such an ontology would not only help ensure consistency in applying gait analysis techniques but also facilitate the comparison of data across different studies and institutions. This includes ongoing assessment and evaluation of its efficacy in healthcare settings and identifying opportunities for collaboration and integration with other applicable technologies and platforms.

REFERENCES

Abdelghany, A.S., Darwish, N.R. & Hefni, H.A. 2019. An agile methodology for ontology development. *International Journal of Intelligent Engineering and Systems*, 12(2): 170–181.

Agibetov, A., Gutierrez, K.E.T., Catalano, C.E., Patanè, G., Hurschler, C. & Spagnuolo, M. 2016. GaitViewer: Semantic gait data analysis and visualization tool. *CEUR Workshop Proceedings*, 1748.

Agrawal, A. & Cui, L. 2020. Quality assurance and enrichment of biological and biomedical ontologies and terminologies. *BMC Medical Informatics and Decision Making*, 20(10): 1–4. https://doi.org/10.1186/s12911-020-01342-4.

Ajami, H. & McHeick, H. 2018. Ontology-based model to support ubiquitous healthcare systems for COPD patients. *Electronics (Switzerland)*, 7(12).

Alsanad, A.A., Chikh, A. & Mirza, A. 2019. A Domain Ontology for Software Requirements Change Management in Global Software Development Environment. *IEEE Access*, 7: 49352–49361.

Amith, M., He, Z., Bian, J., Lossio-Ventura, J.A. & Tao, C. 2018. Assessing the practice of biomedical ontology evaluation: Gaps and opportunities. *Journal of Biomedical Informatics*, 80(August 2017): 1–13. https://doi.org/10.1016/j.jbi.2018.02.010.

Amith, M., Manion, F., Liang, C., Harris, M., Wang, D., He, Y. & Tao, C. 2019. Architecture and usability of OntoKeeper, an ontology evaluation tool. *BMC Medical Informatics and Decision Making*, 19(Suppl 4): 1–18. http://dx.doi.org/10.1186/s12911-019-0859-z.

Antia, M.-J. & Keet, C.M. 2021. Assessing and Enhancing Bottom-up {CNL} Design for Competency Questions for Ontologies. *Proceedings of the Seventh International Workshop on Controlled Natural Language (CNL 2020/21)*. https://aclanthology.org/2021.cnl-1.11.

Anwary, M.A.R., 2019. *An automatic wearable multi-sensor based gait analysis system for older adults* (Doctoral dissertation, Bournemouth University).

Atoba, G.M., 2022. *Development of a domain ontology to support information retrieval on the South African informal sector services* (Masters dissertation, Cape Peninsula University of Technology).

Auvinet, B., Touzard, C., Montestruc, F., Delafond, A. & Goeb, V. 2017. Gait disorders in the elderly and dual task gait analysis: a new approach for identifying motor phenotypes. *Journal of NeuroEngineering and Rehabilitation*, 14(1): 1–14.

Baader, F., Horrocks, I. & Sattler, U. 2008. *Foundations of Artificial Intelligence: Handbook of Knowledge Representation*. First. Amsterdam: Elsevier.

Baker, R., Esquenazi, A., Benedetti, M.G. & Desloovere, K. 2016. Gait analysis: Clinical facts. *European Journal of Physical and Rehabilitation Medicine*, 52(4): 560–574.

Bandeira, J., Bittencourt, I.I., Espinheira, P. & Isotani, S. 2016. FOCA: A Methodology for Ontology Evaluation. , (December). http://arxiv.org/abs/1612.03353.

Barton, A., Rosier, A., Burgun, A. & Ethier, J.F. 2014. The cardiovascular disease ontology. *Frontiers in Artificial Intelligence and Applications*, 267(September): 409–414.

Blondé, W., Antezana, E., De Baets, B., Mironov, V. & Kuiper, M. 2009. Metarel: an Ontology to support the inferencing of Semantic Web relations within Biomedical Ontologies. *Nature Precedings*: 2–5.

Blondé, W., Mironov, V., Venkatesan, A., Antezana, E., De Baets, B. & Kuiper, M. 2011. Reasoning with bio-ontologies: Using relational closure rules to enable practical querying. *Bioinformatics (Oxford, England)*, 27: 1562–1568.

Bravo, M., Reyes, L.F.H. & Reyes Ortiz, J.A. 2019. Methodology for ontology design and construction. *Contaduria y Administracion*, 64(4): 1–24.

Chavula, C. & Keet, C.M. 2015. An orchestration framework for linguistic task ontologies. *Communications in Computer and Information Science*, 544: 3–14.

Cicirelli, G., Impedovo, D., Dentamaro, V., Marani, R., Pirlo, G. & D'Orazio, T.R. 2022. Human Gait Analysis in Neurodegenerative Diseases: A Review. *IEEE Journal of Biomedical and Health Informatics*, 26(1): 229–242.

Cohan, R. 2019. Neuro Exam: Post-Stroke Hemiplegic Gait – StrokeSciences. https://strokesciences.com/post-stroke-hemiplegic-gait/ 14 March 2023.

Daramola, O. and Moser, T., 2021. Towards Semantic Data Integration in Resource-Limited Settings for Decision Support on Gait-Related Diseases. In *Advanced Concepts, Methods, and Applications in Semantic Computing* (pp. 236-256). IGI Global.

Debellis, M. 2021. A Practical Guide to Building OWL Ontologies Using Protégé 5 . 5 and Plugins A Practical Guide to Building OWL Ontologies Using Protégé 5 . 5 and Plugins 8 April 2021 Michael DeBellis. , (April).

Del Din, S., Elshehabi, M., Galna, B., Hobert, M.A., Warmerdam, E., Suenkel, U., Brockmann, K., Metzger, F., Hansen, C., Berg, D., Rochester, L. & Maetzler, W. 2019. Gait analysis with wearables predicts conversion to parkinson disease. *Annals of Neurology*, 86(3): 357–367.

DuCharme, B. 2013. *Learning SPARQL, 2nd Edition_ Querying and Updating with SPARQL 1.1*. Second. New York: O'Reilly Media.

EMBL-EBI, O.L.S. 2022. Ontology Lookup Service < EMBL-EBI. https://www.ebi.ac.uk/ols/index 13 March 2023.

Femi Aminu, E., Oyefolahan, I.O., Bashir Abdullahi, M. & Salaudeen, M.T. 2020. A Review on Ontology Development Methodologies for Developing Ontological Knowledge Representation Systems for various Domains. *International Journal of Information Engineering and Electronic Business*, 12(2): 28–39.

Fonou-Dombeu, J.V., Naidoo, N., Ramnanan, M., Gowda, R. & Lawton, S.R. 2021. OntoCSA. *International Journal of Agricultural and Environmental Information Systems*, 12(4): 1–20.

Golbreich, C., Horridge, M., Horrocks, I., Motik, B. & Shearer, R. 2007. OBO and OWL: Leveraging semantic Web technologies for the life Sciences. *Lecture Notes in Computer Science (including subseries Lecture Notes in Artificial Intelligence and Lecture Notes in Bioinformatics)*, 4825 LNCS: 169–182.

Grimm, S., Hitzler, P. & Abecker, A. 2009. Knowledge representation and ontologies. In *Scientific data mining and knowledge discovery: principles and foundations* (pp. 111-137). Berlin, Heidelberg: Springer Berlin Heidelberg.

Gruber, T. 1995. What is an Ontology? *International Journal of Human-Computer Studies*: 907–928. http://www-ksl.stanford.edu/kst/what-is-an-ontology.html 15 December 2020.

Guarino, N., Oberle, D. & Staab, S., 2009. What is an ontology?. *Handbook on ontologies*, pp.1-17.

Huitzil, I., Dranca, L., Bernad, J. & Bobillo, F. 2019. Gait recognition using fuzzy ontologies and Kinect sensor data. *International Journal of Approximate Reasoning*, 113: 354–371. https://doi.org/10.1016/j.ijar.2019.07.012.

Human Phenotype Ontology. 2022. Human Phenotype Ontology. https://hpo.jax.org/app/ 13 March 2023.

Kamide, N. 2020. An extended description logic for inconsistency-tolerant ontological reasoning with sequential information. *ICAART 2020 - Proceedings of the 12th International Conference on Agents and Artificial Intelligence*, 2(Icaart 2020): 313–321.

Kankam, P. K. 2019. The use of paradigms in information research. *Library and Information Science Research*, 41(2), 85–92. https://doi.org/10.1016/j.lisr.2019.04.003.

Keet, C.M. 2020. *An Introduction to Ontology Engineering*. University of Cape Town, pp. 25–158.

Kelle, U., Kühberger, C. & Bernhard, R. 2019. How to use mixed-methods and triangulation designs: An introduction to history education research. *History Education Research Journal*, 16(1): 5–23.

Kivunja, C. & Kuyini, A.B. 2017. Understanding and Applying Research Paradigms in Educational Contexts. *International Journal of Higher Education*, 6(5): 26.

Lagunes-Garciá, G., Rodríguez-González, A., Prieto-Santamariá, L., Del Valle, E.P.G., Zanin, M. & Menasalvas-Ruiz, E. 2020. DISNET: A framework for extracting phenotypic disease information from public sources. *PeerJ*, 8: 1–34.

Liu, X., Zhao, C., Zheng, B., Guo, Q., Duan, X., Wulamu, A. & Zhang, D. 2021. Wearable Devices for Gait Analysis in Intelligent Healthcare. *Frontiers in Computer Science*, 3(May): 1–8.

Lu, J. & Xu, Q. 2017. *Ontologies and Big Data Considerations for Effective Intelligence*. http://www.igi-global.com/book/ontologies-big-data-considerations-effective/171688.

Manosperta, L. 2019. Automatic Gait Analysis in neurodegenerative diseases. , (February).

McDaniel, M. & Storey, V.C. 2019. Evaluating Domain Ontologies. *ACM Computing Surveys*, 52(4): 1–44.

Medical Subject Headings, M. 2023. Medical Subject Headings - Home Page. https://www.nlm.nih.gov/mesh/meshhome.html 13 March 2023.

Miah, S.J. & Genemo, H. 2016. A Design Science Research Methodology for Expert Systems Development. *Australasian Journal of Information Systems*, 20(November): 0–29.

Moodley, K., 2015. *Practical reasoning for defeasible description logics* (Doctoral dissertation, Ph. D. thesis, University of KwaZulu-Natal).

Morgan, H. 2022. Conducting a Qualitative Document Analysis. *Qualitative Report*, 27(1): 64–77.

Nguyen, A., Gardner, L. & Sheridan, D. 2019. Towards Ontology-Based Design Science Research for Knowledge Accumulation and Evolution. *Proceedings of the 52nd Hawaii International Conference on System Sciences*, (April).

Nguyen, T.H., Grundy, J.C. & Almorsy, M. 2016. Ontology-based automated support for goal–use case model analysis. *Software Quality Journal*, 24(3): 635–673.

De Nicola, A., Missikoff, M. & Navigli, R. 2009. A software engineering approach to ontology building. *Information Systems*, 34(2): 258–275.

Nowell, L.S., Norris, J.M., White, D.E. & Moules, N.J. 2017. Thematic Analysis: Striving to Meet the Trustworthiness Criteria. *International Journal of Qualitative Methods*, 16(1): 1609406917733847. https://doi.org/10.1177/1609406917733847.

Noy, N. & McGuinness, D. 2001. Ontology Development 101: A Guide to Creating Your First Ontology. *Stanford Knowledge Systems Laboratory Technical Report KSL-01-05 and Stanford Medical Informatics Technical Report SMI*-2001-0880.

Ochs, C., Geller, J., Perl, Y. & Musen, M.A. 2016. A unified software framework for deriving, visualizing, and exploring abstraction networks for ontologies. *Journal of Biomedical Informatics*, 62: 90–105. http://dx.doi.org/10.1016/j.jbi.2016.06.008.

OMIM. 2022. Home - OMIM. *OMIM*. https://www.omim.org/ 13 March 2023.

Peffers, K., Tuunanen, T. & Niehaves, B. 2018. Design science research genres: introduction to the special issue on exemplars and criteria for applicable design science research. *European Journal of Information Systems*, 27: 129–139.

Pirker, W. & Katzenschlager, R. 2017. Gait disorders in adults and the elderly: A clinical guide. *Wiener Klinische Wochenschrift*, 129(3–4): 81–95.

Pittet, P. & Barthèlèmy, J. 2015. Towards a Task-based Evaluation of Application Ontologies Throughout Their Lifecycle. , 2(Ic3k): 263–268.

PubMed. 2023 *National Center for Biotechnology Information*. Available at: https://www.ncbi.nlm.nih.gov/pubmed (Accessed: 13 March 2023).

Rapaport, W.J. 2005. Philosophy of Computer Science: An introductory course. *Teaching Philosophy*, 28(4): 319–341.

Roldán-Molina, G.R., Ruano-Ordás, D., Basto-Fernandes, V. & Méndez, J.R. 2021. An ontology knowledge inspection methodology for quality assessment and continuous improvement. *Data and Knowledge Engineering*, 133(December 2020): 101889. https://doi.org/10.1016/j.datak.2021.101889.

Saad, S., Salim, N., Zainal, H. & Muda, Z. 2011. A process for building domain ontology: An experience in developing Solat ontology. *Proceedings of the 2011 International Conference on Electrical Engineering and Informatics, ICEEI 2011*, (January).

Salvadores, M., Alexander, P.R., Musen, M.A. & Noy, N.F. 2013. BioPortal as a Dataset of Linked Biomedical Ontologies and Terminologies in RDF. *Semantic Web*, 4(3): 277–284.

Sanfilippo, E.M., Kitamura, Y. & Young, R.I.M. 2019. Formal ontologies in manufacturing. *Applied Ontology*, 14(2): 119–124.

Sattar, A., Ahmad, M.N., Surin, E.S.M. & Mahmood, A.K. 2021. An Improved Methodology for Collaborative Construction of Reusable, Localized, and Shareable Ontology. *IEEE Access*, 9: 17463–17484.

Shimizu, C.M., 2020. *Advances in modular ontology engineering: methodology and infrastructure*. Kansas State University.

Spoladore, D. & Pessot, E. 2021. Collaborative ontology engineering methodologies for the development of decision support systems: Case studies in the healthcare domain. *Electronics (Switzerland)*, 10(9).

Staab, S. & Studer, R. 2009. *Ontology Engineering Methodology*. 2nd ed. B. Peter, B. Jacek, S. Gunter, & S. Michael, eds. Springer.

Suarez, C., Griol, D., Figueroa, C., Corrales, J.C. & Corrales, D.C. 2022. RustOnt: An Ontology to Explain Weather Favorable Conditions of the Coffee Rust. *Sensors 2022, Vol. 22, Page 9598*, 22(24): 9598. https://www.mdpi.com/1993154.

Tran, T.T. 2017. Opción de investigación pragmatismo en la investigación sobre la cooperación entre universidades y empresas en el contexto Vietnamita. *Revista Lusofona de Educacao*, 36(36): 67–80.

Turcin, I., Ergovic, V. & Lackovic, M. 2013. Ontology Driven Decision Support System Architecture for Gait Analysis. In Y. Dekhtyar, A. Katashev, & L. Lancere, eds. *International Symposium on Biomedical Engineering and Medical Physics, 10-12 October, 2012, Riga, Latvia*. Berlin, Heidelberg: Springer Berlin Heidelberg: 78–81.

Uschold, M. & Gruninger, M. 2004. Ontologies and semantics for seamless connectivity. *SIGMOD Record*, 33(4): 58–64.

WC3. 2020. Semantic Web - W3C. *W3C.Org*. https://www.w3.org/standards/semanticweb/.

Weigand, H., Johannesson, P. & Andersson, B. 2021. An artifact ontology for design science research. *Data & Knowledge Engineering*, 133: 101878.

Whittle, M.W. 2014. *Gait Analysis: An Introduction*. 4th ed. Philadelphia: Elsevier Ltd.

Yadav, U., Narula, G.S., Duhan, N., Jain, V. & Murthy, B.K. 2016. Development and visualization of domain specific ontology using protege. *Indian Journal of Science and Technology*, 9(16).

Younesi, E., Malhotra, A., Gündel, M., Scordis, P., Kodamullil, A.T., Page, M., Müller, B., Springstubbe, S., Wüllner, U., Scheller, D. & Hofmann-Apitius, M. 2015. PDON: Parkinson's disease ontology for representation and modeling of the Parkinson's disease knowledge domain. *Theoretical Biology and Medical Modelling*, 12(1): 1–17.

APPENDIX A: OMIM® gait difficulties results

OMIM Search - '"gait difficulties" '		
Downloaded:	Mar 25, 2023	
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MIM Number	Title	Included Titles
#607317	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 4; SCAR4	
#616516	EMERY-DREIFUSS MUSCULAR DYSTROPHY 3, AUTOSOMAL RECESSIVE; EDMD3	
#618655	MYOPATHY, DISTAL, 6, ADULT-ONSET, AUTOSOMAL DOMINANT; MPD6	
#125250	OPTIC ATROPHY WITH OR WITHOUT DEAFNESS, OPHTHALMOPLEGIA, MYOPATHY, ATAXIA, AND NEUROPATHY	
#604187	SPASTIC PARAPLEGIA 10, AUTOSOMAL DOMINANT; SPG10	
#300623	FRAGILE X TREMOR/ATAXIA SYNDROME; FXTAS	
#609161	STRIATAL DEGENERATION, AUTOSOMAL DOMINANT 1; ADSD1	
#609286	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, AUTOSOMAL DOMINANT 3; PEOA3	
#612319	SPASTIC PARAPLEGIA 35, AUTOSOMAL RECESSIVE, WITH OR WITHOUT NEURODEGENERATION; SPG35	
#614298	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 4; NBIA4	
#615290	SPINAL MUSCULAR ATROPHY, LOWER EXTREMITY-PREDOMINANT, 2A, CHILDHOOD ONSET, AUTOSOMAL DOMINANT; SMALED2A	
#601162	SPASTIC PARAPLEGIA 9A, AUTOSOMAL DOMINANT; SPG9A	
#615643	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 6; NBIA6	
#616040	MYASTHENIC SYNDROME, CONGENITAL, 7A, PRESYNAPTIC, AND DISTAL MOTOR NEUROPATHY, AUTOSOMAL DOMINANT; CMS7A	
#616625	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2W; CMT2W	
#616924	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2CC; CMT2CC	
#617284	DYSTONIA 28, CHILDHOOD-ONSET; DYT28	
#617336	NEMALINE MYOPATHY 11, AUTOSOMAL RECESSIVE; NEM11	
#618124	PERIPHERAL NEUROPATHY, AUTOSOMAL RECESSIVE, WITH OR WITHOUT IMPAIRED INTELLECTUAL DEVELOPMENT; PNRIID	
#618129	MUSCULAR DYSTROPHY, LIMB-GIRDLE, AUTOSOMAL DOMINANT 4; LGMDD4	
#618138	MUSCULAR DYSTROPHY, LIMB-GIRDLE, AUTOSOMAL RECESSIVE 23; LGMDR23	
#618369	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 27; SCAR27	
#619052	MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 8; MC4DN8	
#619061	MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 17; MC4DN17	
#619903	PERIPHERAL MOTOR NEUROPATHY, CHILDHOOD-ONSET, BIOTIN-RESPONSIVE; COMNB	
#118700	CHOREA, BENIGN HEREDITARY; BHC	
#603472	NEURONAL INTRANUCLEAR INCLUSION DISEASE; NIID	
#603896	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER; VWM	VANISHING WHITE MATTER LEUKODYSTROPHY WITH OVARIAN FAILURE, INCLUDED
#214400	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4A; CMT4A	
#261640	HYPERPHENYLALANINEMIA, BH4-DEFICIENT, A; HPABH4A	HYPERPHENYLALANINEMIA, BH4-DEFICIENT, DUE TO PARTIAL PTS DEFICIENCY, INCLUDED
#263570	POLYGLUCOSAN BODY NEUROPATHY, ADULT FORM; APBN	
#604360	SPASTIC PARAPLEGIA 11, AUTOSOMAL RECESSIVE; SPG11	
#604484	NEUROPATHY, HEREDITARY MOTOR AND SENSORY, OKINAWA TYPE; HMSNO	
#605361	SPINOCEREBELLAR ATAXIA 14; SCA14	
#606688	SPONGIFORM ENCEPHALOPATHY WITH NEUROPSYCHIATRIC FEATURES	

#607060	PARKINSON DISEASE 8, AUTOSOMAL DOMINANT; PARK8	
#270750	SPASTIC PARAPLEGIA 23, AUTOSOMAL RECESSIVE; SPG23	
#270800	SPASTIC PARAPLEGIA 5A, AUTOSOMAL RECESSIVE; SPG5A	
#300423	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, HEDERA TYPE; MRXSH	
#300986	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, BAIN TYPE; MRXSB	
#607736	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J; CMT2J	
#607831	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2K; CMT2K	CHARCOT-MARIE-TOOTH DISEASE, AUTOSOMAL DOMINANT, TYPE 2K, INCLUDED
#608358	CONGENITAL MYOPATHY 7A, MYOSIN STORAGE, AUTOSOMAL DOMINANT; CMYP7A	
#608703	SPINOCEREBELLAR ATAXIA 25; SCA25	
#608984	ATAXIA, SENSORY, 1, AUTOSOMAL DOMINANT; SNAX1	
#609270	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 7; SCAR7	
#609285	NEMALINE MYOPATHY 4; NEM4	CAP MYOPATHY 2, INCLUDED CAPM2, INCLUDED
#612716	DYSTONIA, DOPA-RESPONSIVE, DUE TO SEPIAPTERIN REDUCTASE DEFICIENCY	
#613280	HYPERMANGANESEMIA WITH DYSTONIA 1; HMNDYT1	
#613287	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2N; CMT2N	
#610250	SPASTIC PARAPLEGIA 31, AUTOSOMAL DOMINANT; SPG31	
#611228	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4J; CMT4J	
#613710	THIAMINE METABOLISM DYSFUNCTION SYNDROME 4 (BILATERAL STRIATAL DEGENERATION AND PROGRESSIVE POLYNEUROPATHY TYPE); THMD4	
#614153	SPINOCEREBELLAR ATAXIA 36; SCA36	
#614409	SPASTIC PARAPLEGIA 46, AUTOSOMAL RECESSIVE; SPG46	
#614898	SPASTIC PARAPLEGIA 53, AUTOSOMAL RECESSIVE; SPG53	
#615009	SCHUURS-HOEIJMAKERS SYNDROME; SHMS	
#615043	SPASTIC PARAPLEGIA 43, AUTOSOMAL RECESSIVE; SPG43	
#615048	SPINAL MUSCULAR ATROPHY, JOKELA TYPE; SMAJ	
#615191	LISSENCEPHALY 5; LIS5	
#605820	NONAKA MYOPATHY; NM	
#137200	NEUROMYOTONIA AND AXONAL NEUROPATHY, AUTOSOMAL RECESSIVE; NMAN	
#145900	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	
#160500	MYOPATHY, DISTAL, 1; MPD1	
#182600	SPASTIC PARAPLEGIA 3, AUTOSOMAL DOMINANT; SPG3A	
#183086	SPINOCEREBELLAR ATAXIA 6; SCA6	
#183090	SPINOCEREBELLAR ATAXIA 2; SCA2	AMYOTROPHIC LATERAL SCLEROSIS, SUSCEPTIBILITY TO, 13, INCLUDED ALS13, INCLUDED
#193003	SPINOCEREBELLAR ATAXIA 27A; SCA27A	
#310200	MUSCULAR DYSTROPHY, DUCHENNE TYPE; DMD	
#310400	MYOPATHY, CENTRONUCLEAR, X-LINKED; CNMX	
#600334	TIBIAL MUSCULAR DYSTROPHY, TARDIVE; TMD	
#601152	NEUROPATHY, HEREDITARY MOTOR AND SENSORY, TYPE VIA, WITH OPTIC ATROPHY; HMSN6A	
#609452	MYOPATHY, MYOFIBRILLAR, 4; MFM4	
#613640	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE IC; HSAN1C	
#613647	SPASTIC PARAPLEGIA 48, AUTOSOMAL RECESSIVE; SPG48	
#615768	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 16; SCAR16	
#615889	LEUKOENCEPHALOPATHY, PROGRESSIVE, WITH OVARIAN FAILURE; LKENP	
#616155	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2S; CMT2S	
#616541	SHORT STATURE, MICROCEPHALY, AND ENDOCRINE DYSFUNCTION; SSMED	

#616668	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2X; CMT2X
#616907	SPASTIC PARAPLEGIA 76, AUTOSOMAL RECESSIVE; SPG76
#617046	SPASTIC PARAPLEGIA 77, AUTOSOMAL RECESSIVE; SPG77
#617225	SPASTIC PARAPLEGIA 78, AUTOSOMAL RECESSIVE; SPG78
#617258	MYOPATHY, MYOFIBRILLAR, 8; MFM8
#617404	MUSCULAR DYSTROPHY, CONGENITAL, WITH CATARACTS AND INTELLECTUAL DISABILITY; MDCCAID
#618135	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE), TYPE C, 8; MDDGC8
#618239	MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 17; MC1DN17
#618418	SPASTIC PARAPLEGIA 80, AUTOSOMAL DOMINANT; SPG80
#618569	NEURODEVELOPMENTAL DISORDER WITH ATAXIA, HYPOTONIA, AND MICROCEPHALY; NEDAHM
#618653	INTELLECTUAL DEVELOPMENTAL DISORDER WITH IMPAIRED LANGUAGE AND DYSMORPHIC FACIES; IDDILF
#618654	CONGENITAL MYOPATHY 8; CMYP8
#618683	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 6; MC5DN6
#618824	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 8, AUTOSOMAL RECESSIVE; IBGC8
#618877	LEUKOENCEPHALOPATHY, DEVELOPMENTAL DELAY, AND EPISODIC NEUROLOGIC REGRESSION SYNDROME; LEUDEN
#618940	OCULOPHARYNGODISTAL MYOPATHY 2; OPDM2
#618960	MITCHELL SYNDROME; MITCH
#619054	MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 11; MC4DN11
#619062	MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 18; MC4DN18
#619090	DEVELOPMENTAL DELAY, IMPAIRED GROWTH, DYSMORPHIC FACIES, AND AXONAL NEUROPATHY; DIGFAN
#619099	INTELLECTUAL DEVELOPMENTAL DISORDER WITH SPEECH DELAY AND AXONAL PERIPHERAL NEUROPATHY; IDDSAPN
#619112	NEURONOPATHY, DISTAL HEREDITARY MOTOR, TYPE VC; HMN5C
#619121	NEURODEVELOPMENTAL DISORDER WITH CARDIOMYOPATHY, SPASTICITY, AND BRAIN ABNORMALITIES; NEDCASB
#619473	OCULOPHARYNGODISTAL MYOPATHY 3; OPDM3
#619661	LEUKOENCEPHALOPATHY, HEREDITARY DIFFUSE, WITH SPHEROIDS 2; HDLS2
#619686	SPASTIC PARAPLEGIA 85, AUTOSOMAL RECESSIVE; SPG85
#619687	DYSTONIA 33; DYT33
#619733	INCLUSION BODY MYOPATHY AND BRAIN WHITE MATTER ABNORMALITIES; IBMWMA
#619764	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1H; CMT1H
#619966	SPASTIC PARAPLEGIA 87, AUTOSOMAL RECESSIVE; SPG87
#620068	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2II; CMT2II
#620158	SPINOCEREBELLAR ATAXIA 50; SCA50
#620161	CONGENITAL MYOPATHY 15; CMYP15
#620221	SPASTIC PARAPLEGIA 79A, AUTOSOMAL DOMINANT, WITH ATAXIA; SPG79A
%607458	SPINOCEREBELLAR ATAXIA 18; SCA18
%300158	ARTHROGRYPOSIS, CONGENITAL, LOWER LIMB, X-LINKED; ACLLX
%612335	SPASTIC PARAPLEGIA 38, AUTOSOMAL DOMINANT; SPG38
%614860	DYSTONIA 23; DYT23
%609454	SUPRANUCLEAR PALSY, PROGRESSIVE, 2; PSNP2
604916	HYDRONEPHROSIS, CONGENITAL, WITH CLEFT PALATE, CHARACTERISTIC FACIES, HYPOTONIA, AND MENTAL RETARDATION
***605713**	SERINE PALMITOYLTRANSFERASE, LONG-CHAIN BASE SUBUNIT 2; SPTLC2
***606598**	GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1; GDAP1
***606983**	DIACYLGLYCEROL O-ACYLTRANSFERASE 2; DGAT2

***300163**	FOUR-AND-A-HALF LIM DOMAINS 1; FHL1	FHL1B, INCLUDED
***608378**	NUCLEAR EXPORT MEDIATOR FACTOR; NEMF	
***609007**	LEUCINE-RICH REPEAT KINASE 2; LRRK2	
***611146**	SOLUTE CARRIER FAMILY 30 (ZINC TRANSPORTER), MEMBER 10; SLC30A10	
***611508**	CALMODULIN-BINDING TRANSCRIPTION ACTIVATOR 2; CAMTA2	
***614297**	CHROMOSOME 19 OPEN READING FRAME 12; C19ORF12	
***129010**	EARLY GROWTH RESPONSE 2; EGR2	
***176980**	PROTEIN KINASE C, GAMMA; PRKCG	
***143450**	HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, BETA SUBUNIT; HADHB	
***182125**	SEPIAPTERIN REDUCTASE; SPR	
***304040**	GAP JUNCTION PROTEIN, BETA-1; GJB1	
***309550**	FRAGILE X MESSENGER RIBONUCLEOPROTEIN 1; FMR1	FRAGILE SITE, FOLIC ACID TYPE, RARE, fraXq27.3, INCLUDED FRAXA, INCLUDED
***590060**	TRANSFER RNA, MITOCHONDRIAL, LYSINE; MTTK	
***601517**	ATAXIN 2; ATXN2	
***617513**	OXOGLUTARATE DEHYDROGENASE-LIKE PROTEIN; OGDHL	

APPENDIX B: OMIM® gait disturbance results

OMIM Search - '"gait disturbance" '		
Downloaded:	Mar 25, 2023	
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MIM Number	Title	Included Titles
#301094	NEURODEVELOPMENTAL DISORDER WITH GAIT DISTURBANCE, DYSMORPHIC FACIES, AND BEHAVIORAL ABNORMALITIES, X-LINKED; NEDGFAX	
%300861	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, CHUDLEY-SCHWARTZ TYPE; MRXSCS	
*300237	TRANSCRIPTION ELONGATION FACTOR A-LIKE 1; TCEAL1	
#616192	ATAXIA, COMBINED CEREBELLAR AND PERIPHERAL, WITH HEARING LOSS AND DIABETES MELLITUS; ACPHD	
#263570	POLYGLUCOSAN BODY NEUROPATHY, ADULT FORM; APBN	
#193003	SPINOCEREBELLAR ATAXIA 27A; SCA27A	
#600116	PARKINSON DISEASE 2, AUTOSOMAL RECESSIVE JUVENILE; PARK2	
#616710	PARKINSON DISEASE 22, AUTOSOMAL DOMINANT; PARK22	
#108600	SPASTIC ATAXIA 1, AUTOSOMAL DOMINANT; SPAX1	
#213600	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 1; IBGC1	
#216400	COCKAYNE SYNDROME A; CSA	
#236690	HYDROCEPHALUS, NORMAL-PRESSURE, 1; HYDNP1	
#241080	WOODHOUSE-SAKATI SYNDROME; WDSKS	
#245200	KRABBE DISEASE; KRB	
#248900	MAST SYNDROME	
#250100	METACHROMATIC LEUKODYSTROPHY; MLD	PSEUDOARYLSULFATASE A DEFICIENCY, INCLUDED
#254780	MYOCLONIC EPILEPSY OF LAFORA	EPILEPSY, PROGRESSIVE MYOCLONIC, 2B, INCLUDED EPM2B, INCLUDED
#604360	SPASTIC PARAPLEGIA 11, AUTOSOMAL RECESSIVE; SPG11	
#300957	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED 12; XLID12	
#607822	ALZHEIMER DISEASE 3; AD3	ALZHEIMER DISEASE, FAMILIAL, 3, WITH SPASTIC PARAPARESIS AND UNUSUAL PLAQUES, INCLUDED
#609286	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, AUTOSOMAL DOMINANT 3; PEOA3	
#612319	SPASTIC PARAPLEGIA 35, AUTOSOMAL RECESSIVE, WITH OR WITHOUT NEURODEGENERATION; SPG35	

#613280	HYPERMANGANESEMIA WITH DYSTONIA 1; HMNDYT1	
#609524	MYOPATHY, MYOFIBRILLAR, 5; MFM5	
#183086	SPINOCEREBELLAR ATAXIA 6; SCA6	
#302800	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED DOMINANT, 1; CMTX1	
#312080	PELIZAEUS-MERZBACHER DISEASE; PMD	
#600142	CEREBRAL ARTERIOPATHY, AUTOSOMAL RECESSIVE, WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY; CARASIL	
#601104	SUPRANUCLEAR PALSY, PROGRESSIVE, 1; PSNP1	
#601419	MYOPATHY, MYOFIBRILLAR, 1; MFM1	
#616779	CEREBRAL ARTERIOPATHY, AUTOSOMAL DOMINANT, WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY, TYPE 2; CADASIL2	
#616859	SPASTICITY, CHILDHOOD-ONSET, WITH HYPERGLYCINEMIA; SPAHGC	
#618387	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE, WITH AXONAL NEUROPATHY 3; SCAN3	
#620098	DEVELOPMENTAL DELAY WITH VARIABLE INTELLECTUAL DISABILITY AND DYSMORPHIC FACIES; DIDDF	
#620221	SPASTIC PARAPLEGIA 79A, AUTOSOMAL DOMINANT, WITH ATAXIA; SPG79A	
#620286	MYOPATHY, SARCOPLASMIC BODY; MYOSB	
%276100	TRYPTOPHANURIA WITH DWARFISM	
*118190	HEAT-SHOCK 60-KD PROTEIN 1; HSPD1	
*300371	ATP-BINDING CASSETTE, SUBFAMILY D, MEMBER 1; ABCD1	
*611146	SOLUTE CARRIER FAMILY 30 (ZINC TRANSPORTER), MEMBER 10; SLC30A10	
*607574	ARYLSULFATASE A; ARSA	
*176640	PRION PROTEIN; PRNP	
*600075	TATA BOX-BINDING PROTEIN; TBP	
*602194	HTRA SERINE PEPTIDASE 1; HTRA1	
#109150	MACHADO-JOSEPH DISEASE; MJD	
#603472	NEURONAL INTRANUCLEAR INCLUSION DISEASE; NIID	
#221770	POLYCYSTIC LIPOMEMBRANOUS OSTEODYSPLASIA WITH SCLEROSING LEUKOENCEPHALOPATHY 1; PLOSL1	
#221820	LEUKOENCEPHALOPATHY, HEREDITARY DIFFUSE, WITH SPHEROIDS 1; HDLS1	
#229300	FRIEDREICH ATAXIA; FRDA	FRIEDREICH ATAXIA WITH RETAINED REFLEXES, INCLUDED FARR, INCLUDED
#230600	GM1-GANGLIOSIDOSIS, TYPE II; GM1G2	GANGLIOSIDOSIS, GENERALIZED GM1, LATE-INFANTILE TYPE, INCLUDED
#231095	GHOSAL HEMATODIAPHYSEAL DYSPLASIA; GHDD	
#231670	GLUTARIC ACIDEMIA I; GA1	
#234500	HARTNUP DISORDER; HND	

#248500	MANNOSIDOSIS, ALPHA B, LYSOSOMAL; MANSA	
#257220	NIEMANN-PICK DISEASE, TYPE C1; NPC1	NIEMANN-PICK DISEASE, TYPE D, INCLUDED
#604484	NEUROPATHY, HEREDITARY MOTOR AND SENSORY, OKINAWA TYPE; HMSNO	
#605280	SPASTIC PARAPLEGIA 13, AUTOSOMAL DOMINANT; SPG13	
#606072	RIPPLING MUSCLE DISEASE 2; RMD2	
#606232	PHELAN-MCDERMID SYNDROME; PHMDS	
#607225	SPASTIC PARALYSIS, INFANTILE-ONSET ASCENDING; IAHSP	
#607250	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE, WITH AXONAL NEUROPATHY 1; SCAN1	
#607259	SPASTIC PARAPLEGIA 7, AUTOSOMAL RECESSIVE; SPG7	
#607459	SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS; SANDO	SPINOCEREBELLAR ATAXIA WITH EPILEPSY, INCLUDED SCAE, INCLUDED
#269600	SEA-BLUE HISTIOCYTE DISEASE	
#270685	SPASTIC PARAPLEGIA 17, AUTOSOMAL DOMINANT; SPG17	
#270700	SPASTIC PARAPLEGIA 15, AUTOSOMAL RECESSIVE; SPG15	
#272800	TAY-SACHS DISEASE; TSD	TAY-SACHS DISEASE, JUVENILE, INCLUDED
#300055	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC 13; MRXS13	
#300423	INTELLECTUAL DEVELOPMENTAL DISORDER, X-LINKED, SYNDROMIC, HEDERA TYPE; MRXSH	
#300894	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION 5; NBIA5	
#607734	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1F; CMT1F	
#609161	STRIATAL DEGENERATION, AUTOSOMAL DOMINANT 1; ADSD1	
#612069	AMYOTROPHIC LATERAL SCLEROSIS 10 WITH OR WITHOUT FRONTOTEMPORAL DEMENTIA; ALS10	FRONTOTEMPORAL LOBAR DEGENERATION WITH TDP43 INCLUSIONS, TARDBP-RELATED, INCLUDED
#612674	POLYNEUROPATHY, HEARING LOSS, ATAXIA, RETINITIS PIGMENTOSA, AND CATARACT; PHARC	
#612989	OPTIC ATROPHY 7 WITH OR WITHOUT AUDITORY NEUROPATHY; OPA7	
#613026	CHROMOSOME 19q13.11 DELETION SYNDROME, DISTAL	
#610250	SPASTIC PARAPLEGIA 31, AUTOSOMAL DOMINANT; SPG31	
#610743	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 8; SCAR8	
#614305	SCLEROSTEOSIS 2; SOST2	
#614409	SPASTIC PARAPLEGIA 46, AUTOSOMAL RECESSIVE; SPG46	
#614458	THIAMINE METABOLISM DYSFUNCTION SYNDROME 5 (EPISODIC ENCEPHALOPATHY TYPE); THMD5	

#614867	PEROXISOME BIOGENESIS DISORDER 5B; PBD5B	
#615085	OSTEOPETROSIS, AUTOSOMAL RECESSIVE 8; OPTB8	
#615483	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 5; IBGC5	
#127750	DEMENTIA, LEWY BODY; DLB	DIFFUSE LEWY BODY DISEASE WITH GAZE PALSY, INCLUDED
#133540	COCKAYNE SYNDROME B; CSB	
#136140	FLOATING-HARBOR SYNDROME; FLHS	
#137440	GERSTMANN-STRAUSSLER DISEASE; GSD	CEREBRAL AMYLOID ANGIOPATHY, PRNP-RELATED, INCLUDED
#168600	PARKINSON DISEASE, LATE-ONSET; PD	
#168601	PARKINSON DISEASE 1, AUTOSOMAL DOMINANT; PARK1	ATYPICAL PARKINSON DISEASE, INCLUDED
#182900	SPHEROCYTOSIS, TYPE 1; SPH1	
#184255	SPONDYLOMETAPHYSEAL DYSPLASIA, CORNER FRACTURE TYPE; SMDCF	
#203450	ALEXANDER DISEASE; ALXDRD	
#311070	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED RECESSIVE, 5; CMTX5	
#600363	SPASTIC PARAPLEGIA 6, AUTOSOMAL DOMINANT; SPG6	
#601042	DYSTONIA 9; DYT9	
#601162	SPASTIC PARAPLEGIA 9A, AUTOSOMAL DOMINANT; SPG9A	
#615558	HYPOBETALIPOPROTEINEMIA, FAMILIAL, 1; FHBL1	LOW DENSITY LIPOPROTEIN CHOLESTEROL LEVEL QUANTITATIVE TRAIT LOCUS 4, INCLUDED LDLCQ4, INCLUDED
#615577	IMMUNODEFICIENCY, COMMON VARIABLE, 10; CVID10	
#615704	POIKILODERMA, HEREDITARY FIBROSING, WITH TENDON CONTRACTURES, MYOPATHY, AND PULMONARY FIBROSIS; POIKTMP	
#615768	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 16; SCAR16	
#616199	POLYGLUCOSAN BODY MYOPATHY 2; PGBM2	
#616291	LICHTENSTEIN-KNORR SYNDROME; LIKNS	
#616541	SHORT STATURE, MICROCEPHALY, AND ENDOCRINE DYSFUNCTION; SSMED	
#616688	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2Z; CMT2Z	
#616878	METABOLIC CRISES, RECURRENT, WITH RHABDOMYOLYSIS, CARDIAC ARRHYTHMIAS, AND NEURODEGENERATION; MECRCN	
#617017	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2T; CMT2T	
#617284	DYSTONIA 28, CHILDHOOD-ONSET; DYT28	
#617882	CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE G; CMTDIG	
#618093	SPINOCEREBELLAR ATAXIA 48; SCA48	
#618170	NEURODEGENERATION, CHILDHOOD-ONSET, STRESS-INDUCED, WITH	

	VARIABLE ATAXIA AND SEIZURES; CONDSIAS	
#618193	POLYCYSTIC LIPOMEMBRANOUS OSTEODYSPLASIA WITH SCLEROSING LEUKOENCEPHALOPATHY 2; PLOSL2	
#618418	SPASTIC PARAPLEGIA 80, AUTOSOMAL DOMINANT; SPG80	
#618430	DEVELOPMENTAL DELAY WITH VARIABLE INTELLECTUAL IMPAIRMENT AND BEHAVIORAL ABNORMALITIES; DDVIBA	
#618564	MICROANGIOPATHY AND LEUKOENCEPHALOPATHY, PONTINE, AUTOSOMAL DOMINANT; PADMAL	
#619051	MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 7; MC4DN7	
#619090	DEVELOPMENTAL DELAY, IMPAIRED GROWTH, DYSMORPHIC FACIES, AND AXONAL NEUROPATHY; DIGFAN	
%118420	CHIARI MALFORMATION TYPE I	CHIARI MALFORMATION TYPE I WITH SYRINGOMYELIA, INCLUDED
%600223	SPINOCEREBELLAR ATAXIA 4; SCA4	
168100	PARALYSIS AGITANS, JUVENILE, OF HUNT	
*104311	PRESENILIN 1; PSEN1	
*114217	CALNEXIN; CANX	
*604824	KLOTHO; KL	
*605262	NMYC DOWNSTREAM-REGULATED GENE 1; NDRG1	
*606352	ALSIN RHO GUANINE NUCLEOTIDE EXCHANGE FACTOR ALS2; ALS2	
*606370	THIAMINE PYROPHOSPHOKINASE; TPK1	
*606686	EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 1; EIF2B1	
*300005	METHYL-CpG-BINDING PROTEIN 2; MECP2	
*607839	GLYCOGEN BRANCHING ENZYME; GBE1	
*608181	ACIDIC CLUSTER PROTEIN, 33-KD; ACP33	
*608272	NEURAMINIDASE 1; NEU1	
*609139	RECEPTOR EXPRESSION-ENHANCING PROTEIN 1; REEP1	
*612641	ANKYRIN 1; ANK1	ANKYRIN-R, INCLUDED ANK, INCLUDED
*614297	CHROMOSOME 19 OPEN READING FRAME 12; C19ORF12	
*605862	RIBITOL XYLOSYLTRANSFERASE 1; RXYLT1	
*607623	NPC INTRACELLULAR CHOLESTEROL TRANSPORTER 1; NPC1	
*138491	GLYCINE RECEPTOR, ALPHA-1 SUBUNIT; GLRA1	
*146920	ADENOSINE DEAMINASE, RNA-SPECIFIC; ADAR	
*174763	POLYMERASE, DNA, GAMMA; POLG	
*191290	TYROSINE HYDROXYLASE; TH	
*314850	KELL BLOOD GROUP PROTEIN, MCLEOD SYNDROME-ASSOCIATED; XK	
*601097	PERIPHERAL MYELIN PROTEIN 22; PMP22	
*601143	DYNACTIN 1; DCTN1	

*601253	CAVEOLIN 3; CAV3
*616244	COILED-COIL-HELIX-COILED-COIL-HELIX DOMAIN-CONTAINING PROTEIN 2; CHCHD2
*616661	MORC FAMILY CW-TYPE ZINC FINGER PROTEIN 2; MORC2
*617342	PEPTIDYL-tRNA HYDROLASE DOMAIN-CONTAINING 1; PTRHD1

APPENDIX C: Extract of HPO for gait-related diseases results

DISEASE_ID	DISEASE_NAME					
ORPHA:500055	16p13.2 microdeletion syndrome					
ORPHA:1606	1p36 deletion syndrome					
ORPHA:261349	2p15p16.1 microdeletion syndrome					
ORPHA:313947	2q23.1 microduplication syndrome					
ORPHA:67047	3-methylglutaconic aciduria type 3					
OMIM:614739	3-Methylglutaconic aciduria with deafness, encephalopathy, and leigh-like syndrome					
ORPHA:65286	3q29 microdeletion syndrome					
ORPHA:168563	46, XY gonadal dysgenesis-motor and sensory neuropathy syndrome					
ORPHA:289494	4H leukodystrophy					
ORPHA:13	6-pyruvoyl-tetrahydropterin synthase deficiency					
ORPHA:75857	6q terminal deletion syndrome					
ORPHA:171829	6q16 microdeletion syndrome					
ORPHA:96121	7q11.23 microduplication syndrome					
ORPHA:531151	9q21.13 microdeletion syndrome					
ORPHA:495818	9q33.3q34.11 microdeletion syndrome					
ORPHA:324708	ABeta amyloidosis, Iowa type					
ORPHA:14	Abetalipoproteinemia					
ORPHA:48818	Aceruloplasminemia					
ORPHA:99736	Acetazolamide-responsive myotonia					
ORPHA:466794	Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome					
ORPHA:98916	Acute inflammatory demyelinating polyradiculoneuropathy					
ORPHA:139417	Acute transverse myelitis					
OMIM:103050	Adenylosuccinase deficiency					
ORPHA:482601	Adenylosuccinate synthetase-like 1-related distal myopathy					
ORPHA:139399	Adrenomyeloneuropathy					
ORPHA:206448	Adult Krabbe disease					
ORPHA:206583	Adult polyglucosan body disease					
ORPHA:99027	Adult-onset autosomal dominant leukodystrophy					
ORPHA:284289	Adult-onset autosomal recessive cerebellar ataxia					
ORPHA:420492	Adult-onset cervical dystonia, DYT23 type					
ORPHA:329336	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy					
ORPHA:329478	Adult-onset distal myopathy due to VCP mutation					
ORPHA:171442	Adult-onset nemaline myopathy					
ORPHA:3385	African trypanosomiasis					
OMIM:218000	Agenesis of the corpus callosum with peripheral neuropathy					
ORPHA:51	Aicardi-Goutières syndrome					
OMIM:225750	Aicardi-Goutieres syndrome 1					
OMIM:615010	Aicardi-Goutieres syndrome 6					
OMIM:617694	Al Kaissi syndrome					
OMIM:616459	Al-Raqad syndrome					
ORPHA:404454	Alacrimia-choreoathetosis-liver dysfunction syndrome					

ORPHA:58	Alexander disease					
OMIM:300523	Allan-Herndon-Dudley syndrome					
ORPHA:93925	Alobar holoprosencephaly					
ORPHA:399058	Alpha-B crystallin-related late-onset myopathy					
ORPHA:280333	Alpha-dystroglycan-related limb-girdle muscular dystrophy R16					
OMIM:203740	Alpha-Ketoglutarate dehydrogenase deficiency					
OMIM:248500	Alpha-mannosidosis					
ORPHA:62	Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3					
OMIM:619268	Alzahrani-Kuwahara syndrome					
OMIM:607822	Alzheimer disease 3					
OMIM:613435	Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia					
OMIM:615515	Amyotrophic lateral sclerosis 19					
OMIM:205100	Amyotrophic lateral sclerosis 2, juvenile					
OMIM:602433	Amyotrophic lateral sclerosis 4, juvenile					
OMIM:608030	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia					
OMIM:608627	Amyotrophic lateral sclerosis 8					
ORPHA:357043	Amyotrophic lateral sclerosis type 4					
OMIM:105830	Angelman syndrome					
ORPHA:72	Angelman syndrome					
ORPHA:411511	Angelman syndrome due to a point mutation					
ORPHA:411515	Angelman syndrome due to imprinting defect in 15q11-q13					
ORPHA:98794	Angelman syndrome due to maternal 15q11q13 deletion					
ORPHA:98795	Angelman syndrome due to paternal uniparental disomy of chromosome 15					
OMIM:206570	Angiomatosis, diffuse corticomeningeal, of divry and van bogaert					
ORPHA:2356	Arachnoid cyst					
OMIM:616268	Arboleda-Tham syndrome					
OMIM:207800	Argininemia					
ORPHA:268882	Arnold-Chiari malformation type I					
ORPHA:1136	Arnold-Chiari malformation type II					
OMIM:300158	Arthrogryposis, congenital, lower limb, X-linked					
OMIM:187370	Arthrogryposis, distal, type 10					
OMIM:617146	Arthrogryposis, distal, with impaired proprioception and touch					
OMIM:208230	Arthropathy, progressive pseudorheumatoid, of childhood					
OMIM:277460	Ataxia with isolated vitamin E deficiency					
ORPHA:96	Ataxia with vitamin E deficiency					
OMIM:616192	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus					
OMIM:208920	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia					
OMIM:609033	Ataxia, posterior column, with retinitis pigmentosa					
OMIM:608984	Ataxia, sensory, autosomal dominant					
OMIM:270500	Ataxia, spastic, childhood-onset, autosomal recessive, with optic atrophy and mental retardation					
ORPHA:1168	Ataxia-oculomotor apraxia type 1					
OMIM:159550	Ataxia-Pancytopenia syndrome					
ORPHA:2585	Ataxia-pancytopenia syndrome					
ORPHA:1184	Ataxia-photosensitivity-short stature syndrome					

ORPHA:1178	Ataxia-tapetoretinal degeneration syndrome					
OMIM:208900	Ataxia-telangiectasia					
ORPHA:100	Ataxia-telangiectasia					
ORPHA:251347	Ataxia-telangiectasia-like disorder					
OMIM:604391	Ataxia-telangiectasia-like disorder 1					
OMIM:615919	Ataxia-Telangiectasia-Like disorder 2					
ORPHA:1192	Atherosclerosis-deafness-diabetes-epilepsy-nephropathy syndrome					
OMIM:209100	Atonic-Astatic syndrome of foerster					
ORPHA:314632	ATP13A2-related juvenile neuronal ceroid lipofuscinosis					
ORPHA:391411	Atypical juvenile parkinsonism					
ORPHA:216873	Atypical pantothenate kinase-associated neurodegeneration					
ORPHA:99750	Atypical progressive supranuclear palsy syndrome					
ORPHA:3095	Atypical Rett syndrome					
ORPHA:169189	Autosomal dominant centronuclear myopathy					
ORPHA:99	Autosomal dominant cerebellar ataxia					
ORPHA:99947	Autosomal dominant Charcot-Marie-Tooth disease type 2A2					
ORPHA:99939	Autosomal dominant Charcot-Marie-Tooth disease type 2E					
ORPHA:99940	Autosomal dominant Charcot-Marie-Tooth disease type 2F					
ORPHA:99944	Autosomal dominant Charcot-Marie-Tooth disease type 2K					
ORPHA:488333	Autosomal dominant Charcot-Marie-Tooth disease type 2W					
ORPHA:435387	Autosomal dominant Charcot-Marie-Tooth disease type 2Y					
ORPHA:466768	Autosomal dominant Charcot-Marie-Tooth disease type 2Z					
ORPHA:98808	Autosomal dominant dopa-responsive dystonia					
ORPHA:98853	Autosomal dominant Emery-Dreifuss muscular dystrophy					
ORPHA:266	Autosomal dominant limb-girdle muscular dystrophy type 1A					
ORPHA:67036	Autosomal dominant optic atrophy and cataract					
ORPHA:98673	Autosomal dominant optic atrophy, classic form					
ORPHA:254892	Autosomal dominant progressive external ophthalmoplegia					
ORPHA:251282	Autosomal dominant spastic ataxia type 1					
ORPHA:100991	Autosomal dominant spastic paraplegia type 10					
ORPHA:100993	Autosomal dominant spastic paraplegia type 12					
ORPHA:100994	Autosomal dominant spastic paraplegia type 13					
ORPHA:100998	Autosomal dominant spastic paraplegia type 17					
ORPHA:100999	Autosomal dominant spastic paraplegia type 19					
ORPHA:100984	Autosomal dominant spastic paraplegia type 3					
ORPHA:101011	Autosomal dominant spastic paraplegia type 31					
ORPHA:320365	Autosomal dominant spastic paraplegia type 36					
ORPHA:171612	Autosomal dominant spastic paraplegia type 37					
ORPHA:171617	Autosomal dominant spastic paraplegia type 38					
ORPHA:320355	Autosomal dominant spastic paraplegia type 41					
ORPHA:171863	Autosomal dominant spastic paraplegia type 42					
ORPHA:100988	Autosomal dominant spastic paraplegia type 6					
ORPHA:444099	Autosomal dominant spastic paraplegia type 73					
ORPHA:100989	Autosomal dominant spastic paraplegia type 8					

ORPHA:447753	Autosomal dominant spastic paraplegia type 9A			
ORPHA:447757	Autosomal dominant spastic paraplegia type 9B			
ORPHA:228169	Autosomal dominant striatal neurodegeneration			
ORPHA:247815	Autosomal recessive ataxia due to PEX10 deficiency			
ORPHA:88644	Autosomal recessive ataxia, Beauce type			
ORPHA:521411	Autosomal recessive axonal Charcot-Marie-Tooth disease due to copper metabolism defect			
ORPHA:324442	Autosomal recessive axonal neuropathy with neuromyotonia			
ORPHA:169186	Autosomal recessive centronuclear myopathy			
ORPHA:453521	Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency			
ORPHA:412057	Autosomal recessive cerebellar ataxia due to STUB1 deficiency			
ORPHA:352641	Autosomal recessive cerebellar ataxia with late-onset spasticity			
ORPHA:404499	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to RUBCN deficiency			
ORPHA:284282	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency			
ORPHA:95434	Autosomal recessive cerebellar ataxia-movement disorder syndrome			
ORPHA:284271	Autosomal recessive cerebellar ataxia-psychomotor delay syndrome			
ORPHA:363429	Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome			
ORPHA:1170	Autosomal recessive cerebelloparenchymal disorder type 3			
OMIM:600142	Autosomal recessive cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL)			
ORPHA:101097	Autosomal recessive Charcot-Marie-Tooth disease with hoarseness			
ORPHA:363432	Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency			
ORPHA:324262	Autosomal recessive congenital cerebellar ataxia due to MGLUR1 deficiency			
ORPHA:357058	Autosomal recessive cutis laxa type 2A			
ORPHA:101150	Autosomal recessive dopa-responsive dystonia			
ORPHA:98855	Autosomal recessive Emery-Dreifuss muscular dystrophy			
ORPHA:2990	Autosomal recessive multiple pterygium syndrome			
ORPHA:319332	Autosomal recessive myogenic arthrogryposis multiplex congenita			
ORPHA:254886	Autosomal recessive progressive external ophthalmoplegia			
ORPHA:98	Autosomal recessive spastic ataxia of Charlevoix-Saguenay			
ORPHA:314603	Autosomal recessive spastic ataxia with leukoencephalopathy			
ORPHA:254343	Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome			
ORPHA:2822	Autosomal recessive spastic paraplegia type 11			
ORPHA:100995	Autosomal recessive spastic paraplegia type 14			
ORPHA:100996	Autosomal recessive spastic paraplegia type 15			
ORPHA:209951	Autosomal recessive spastic paraplegia type 18			
ORPHA:101000	Autosomal recessive spastic paraplegia type 20			
ORPHA:101001	Autosomal recessive spastic paraplegia type 21			
ORPHA:101003	Autosomal recessive spastic paraplegia type 23			
ORPHA:101004	Autosomal recessive spastic paraplegia type 24			
ORPHA:101006	Autosomal recessive spastic paraplegia type 26			
ORPHA:101008	Autosomal recessive spastic paraplegia type 28			
ORPHA:171622	Autosomal recessive spastic paraplegia type 32			
ORPHA:171629	Autosomal recessive spastic paraplegia type 35			
ORPHA:139480	Autosomal recessive spastic paraplegia type 39			

ORPHA:320370	Autosomal recessive spastic paraplegia type 43					
ORPHA:320401	Autosomal recessive spastic paraplegia type 44					
ORPHA:320396	Autosomal recessive spastic paraplegia type 45					
ORPHA:320391	Autosomal recessive spastic paraplegia type 46					
ORPHA:306511	Autosomal recessive spastic paraplegia type 48					
ORPHA:320380	Autosomal recessive spastic paraplegia type 54					
ORPHA:320411	Autosomal recessive spastic paraplegia type 56					
ORPHA:431329	Autosomal recessive spastic paraplegia type 57					
ORPHA:401795	Autosomal recessive spastic paraplegia type 59					
ORPHA:401800	Autosomal recessive spastic paraplegia type 60					
ORPHA:401780	Autosomal recessive spastic paraplegia type 61					
ORPHA:401785	Autosomal recessive spastic paraplegia type 62					
ORPHA:401805	Autosomal recessive spastic paraplegia type 63					
ORPHA:401810	Autosomal recessive spastic paraplegia type 64					
ORPHA:401815	Autosomal recessive spastic paraplegia type 66					
ORPHA:401820	Autosomal recessive spastic paraplegia type 67					
ORPHA:401840	Autosomal recessive spastic paraplegia type 71					
ORPHA:468661	Autosomal recessive spastic paraplegia type 74					
ORPHA:488594	Autosomal recessive spastic paraplegia type 76					
ORPHA:466722	Autosomal recessive spastic paraplegia type 77					
ORPHA:513436	Autosomal recessive spastic paraplegia type 78					
ORPHA:447760	Autosomal recessive spastic paraplegia type 9B					
ORPHA:95433	Autosomal recessive spinocerebellar ataxia-blindness-deafness syndrome					
ORPHA:101010	Autosomal spastic paraplegia type 30					
ORPHA:397946	Autosomal spastic paraplegia type 58					
ORPHA:401849	Autosomal spastic paraplegia type 72					
ORPHA:209004	Axonal polyneuropathy associated with IgG/IgM/IgA monoclonal gammopathy					
ORPHA:352577	Bainbridge-Ropers syndrome					
OMIM:615485	Bainbridge-Ropers syndrome					
OMIM:618218	Baker-Gordon syndrome					
OMIM:619255	Baralle-Macken syndrome					
OMIM:209900	Bardet-Biedl syndrome 1					
OMIM:302060	Barth syndrome					
OMIM:213600	Basal ganglia calcification, idiopathic, 1					
ORPHA:464738	Basel-Vanagaite-Smirin-Yosef syndrome					
ORPHA:98895	Becker muscular dystrophy					
ORPHA:275864	Behavioral variant of frontotemporal dementia					
ORPHA:117	Behçet disease					
OMIM:210000	Behr syndrome					
ORPHA:1429	Benign hereditary chorea					
ORPHA:119	Beta-sarcoglycan-related limb-girdle muscular dystrophy R4					
ORPHA:610	Bethlem myopathy					
ORPHA:363454	BICD2-related autosomal dominant childhood-onset proximal spinal muscular atrophy					
ORPHA:101070	Bilateral frontoparietal polymicrogyria					

ORPHA:268940	Bilateral polymicrogyria					
OMIM:617308	Bile acid synthesis defect, congenital, 6					
OMIM:617595	Birk-Landau-Perez syndrome					
ORPHA:97297	Bohring-Opitz syndrome					
OMIM:215470	Boucher-Neuhauser syndrome					
OMIM:619543	Boudin-Mortier syndrome					
OMIM:271630	Brachyolmia type 1, Toledo type					
OMIM:618476	Brain abnormalities, neurodegeneration, and dysosteosclerosis					
ORPHA:352649	Brain dopamine-serotonin vesicular transport disease					
ORPHA:209905	Brain-lung-thyroid syndrome					
ORPHA:90354	Brittle cornea syndrome					
OMIM:614170	Brittle cornea syndrome 2					
ORPHA:267	Calpain-3-related limb-girdle muscular dystrophy R1					
OMIM:131300	Camurati-Engelmann disease					
ORPHA:1328	Camurati-Engelmann disease					
OMIM:606631	Camurati-engelmann disease, type 2					
ORPHA:171881	Cap myopathy					
ORPHA:97355	Caribbean parkinsonism					
OMIM:607674	Cataract, congenital, with mental impairment and dentate gyrus atrophy					
OMIM:618761	Catifa syndrome					
ORPHA:505652	CDKL5-deficiency disorder					
OMIM:619482	Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction					
ORPHA:504476	Cerebellar ataxia with neuropathy and bilateral vestibular areflexia syndrome					
OMIM:601338	Cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss					
OMIM:619576	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects					
ORPHA:94122	Cerebellar ataxia, Cayman type					
OMIM:601238	Cerebellar ataxia, Cayman type					
OMIM:224050	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1					
OMIM:615268	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome4					
OMIM:614575	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome					
ORPHA:1174	Cerebellar ataxia-ectodermal dysplasia syndrome					
OMIM:618501	Cerebellar atrophy with seizures and variable developmental delay					
OMIM:614756	Cerebellar dysfunction with variable cognitive and behavioral abnormalities					
OMIM:619761	Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism					
OMIM:618479	Cerebellar, ocular, craniofacial, and genital syndrome					
OMIM:616779	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2					
OMIM:125310	Cerebral arteriopathy, autosomal dominant, with subcortical infarctsand leukoencephalopathy					
ORPHA:136	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy					
ORPHA:199354	Cerebral autosomal recessive arteriopathy-subcortical infarcts-leukoencephalopathy					
OMIM:300352	Cerebral creatine deficiency syndrome 1					
OMIM:605388	Cerebral palsy, ataxic, autosomal recessive					
OMIM:213700	Cerebrotendinous xanthomatosis					
ORPHA:909	Cerebrotendinous xanthomatosis					
OMIM:204200	Ceroid lipofuscinosis, neuronal, 3					

OMIM:609055	Ceroid lipofuscinosis, neuronal, 9				
OMIM:118300	Charcot-Marie-Tooth disease and deafness				
ORPHA:101081	Charcot-Marie-Tooth disease type 1A				
ORPHA:90658	Charcot-Marie-Tooth disease type 1E				
ORPHA:101085	Charcot-Marie-Tooth disease type 1F				
ORPHA:98856	Charcot-Marie-Tooth disease type 2B1				
ORPHA:99948	Charcot-Marie-Tooth disease type 4A				
ORPHA:99956	Charcot-Marie-Tooth disease type 4B2				
ORPHA:99949	Charcot-Marie-Tooth disease type 4C				
ORPHA:99950	Charcot-Marie-Tooth disease type 4D				
ORPHA:99953	Charcot-Marie-Tooth disease type 4G				
OMIM:617087	Charcot-Marie-Tooth disease, axonal, autosomal recessive, type 2A2B				

APPENDIX D: Extract of PubMed gait disorders results

PMID	Title	Authors	Publication Year
30704677	Gait Disorders and Falls in the Elderly	Ronthal M.	2019
30482309	Gait	Mirelman A, Shema S, Maidan I, Hausdorff JM.	2018
9929776	Gait disorders	Cantor CR.	1999
21827923	Balance and gait problems in the elderly	Viswanathan A, Sudarsky L.	2012
29220753	A systematic review of the gait characteristics associated with Cerebellar Ataxia	Buckley E, Mazzà C, McNeill A.	2018
29903447	Neurophysiology of gait	Serrao M, Ranavolo A, Casali C.	2018
34939221	Discussion of Research Priorities for Gait Disorders in Parkinson's Disease	Bohnen NI, Costa RM, Dauer WT, Factor SA, Giladi N, Hallett M, Lewis SJG, Nieuwboer A, Nutt JG, Takakusaki K, Kang UJ, Przedborski S, Papa SM; MDS-Scientific Issues Committee.	2022
34358847	Instrumented gait analysis for management of gait disorders in children with cerebral palsy: A scoping review	States RA, Krzak JJ, Salem Y, Godwin EM, Bodkin AW, McMulkin ML.	2021
31502995	From Emotions to Mood Disorders: A Survey on Gait Analysis Methodology	Deligianni F, Guo Y, Yang GZ.	2019
32580330	Gait Analysis in Parkinson's Disease: An Overview of the Most Accurate Markers for Diagnosis and Symptoms Monitoring	di Biase L, Di Santo A, Caminiti ML, De Liso A, Shah SA, Ricci L, Di Lazzaro V.	2020
36617694	Functional Gait Disorders: Clinical presentations, Phenotypes and Implications for treatment	Issak S, Kanaan R, Nielsen G, Fini NA, Williams G.	2023
21753097	Office management of gait disorders in the elderly	Lam R.	2011
35378605	Interventions to improve gait in Parkinson's disease: a systematic review of randomised controlled trials and network meta-analysis	Hvingelby VS, Glud AN, Sørensen JCH, Tai Y, Andersen ASM, Johnsen E, Moro E, Pavese N.	2022
26852960	Poor Gait Performance and Prediction of Dementia: Results from a Meta-Analysis	Beauchet O, Annweiler C, Callisaya ML, De Cock AM, Helbostad JL, Kressig RW, Srikanth V, Steinmetz JP, Blumen HM, Verghese J, Allali G.	2016
33461679	Gait analysis in neurological populations: Progression in the use of wearables	Celik Y, Stuart S, Woo WL, Godfrey A.	2021
18058946	The role of executive function and attention in gait	Yogev-Seligmann G, Hausdorff JM, Giladi N.	2008
33078988	Role of machine learning in gait analysis: a review	Khera P, Kumar N.	2020
32462346	Key gait findings for diagnosing three syndromic categories of dynamic instability in patients with balance disorders	Schniepp R, Möhwald K, Wuehr M.	2020
34963115	Associations of Gait Disorders and Recurrent Falls in Older People: A Prospective Population-Based Study	Marini K, Mahlknecht P, Schorr O, Baumgartner M, De Marzi R, Raccagni C, Kiechl S, Rungger G, Stockner H, Willeit P, Willeit J, Poewe W, Seppi K.	2022
2184358	Geriatrics: gait disorders in the elderly	Sudarsky L.	1990
30636313	The Timed Up and Go test	Browne W, Nair BKR.	2019
29733529	Sensor-based gait analysis in atypical parkinsonian disorders	Raccagni C, Gaßner H, Eschlboeck S, Boesch S, Krismer F, Seppi K, Poewe W, Eskofier BM, Winkler J, Wenning G, Klucken J.	2018
9305281	Neurophysiology of gait disorders: present and future applications	Dietz V.	1997
16796396	The pathomechanics of plantar fasciitis	Wearing SC, Smeathers JE, Urry SR, Hennig EM, Hills AP.	2006
16519061	[Gait and balance disorders. Fall in the elderly]	Taithe F, Durif F.	2005

31677546	Gait deficits and dynamic stability in children and adolescents with cerebral palsy: A systematic review and meta-analysis	Chakraborty S, Nandy A, Kesar TM.	2020
21553270	Quadrupedal coordination of bipedal gait: implications for movement disorders	Dietz V.	2011
33713194	Fall prediction in neurological gait disorders: differential contributions from clinical assessment, gait analysis, and daily-life mobility monitoring	Schniepp R, Huppert A, Decker J, Schenkel F, Schlick C, Rasoul A, Dieterich M, Brandt T, Jahn K, Wuehr M.	2021
36026713	Gait Analysis in Orthopaedic Surgery: History, Limitations, and Future Directions	Hecht GG, Van Rysselberghe NL, Young JL, Gardner MJ.	2022
1871684	[Evaluating gait disorders in geriatrics]	Bopp I, Six P.	1991
36065671	[Adaptive Gait from the Neurorehabilitation Perspective]	Morioka S.	2022
35954925	Research Progress of Music Therapy on Gait Intervention in Patients with Parkinson's Disease	Wu Z, Kong L, Zhang Q.	2022
33349582	Unmet needs in Parkinson disease: Motor and non-motor	LeWitt PA, Chaudhuri KR.	2020
30199607	Toe Walking Assessment in Autism Spectrum Disorder Subjects: A Systematic Review	Valagussa G, Trentin L, Signori A, Grossi E.	2018
35101749	Components of gait in people with and without mild cognitive impairment	Lindh-Rengifo M, Jonasson SB, Ullén S, Stomrud E, Palmqvist S, Mattsson-Carlgren N, Hansson O, Nilsson MH.	2022
35506743	Impairments in ankle range of motion, dorsi and plantar flexors muscle strength and gait speed in patients with chronic venous disorders: A systematic review and meta-analysis	Nepomuceno de Souza I, Fernandes de Oliveira LF, Geraldo Izalino de Almeida IL, Ávila MR, Silva WT, Trede Filho RG, Pereira DAG, de Oliveira LFL, Lima VP, Scheidt Figueiredo PH, Costa HS.	2022
6625987	Gait disorders among elderly patients. A survey study of fifty (50) patients	Sudarsky L, Ronthal M.	1983
35848860	Towards adaptive deep brain stimulation for freezing of gait	Tan H.	2022
21138821	Tiptoeing around gait disorders: multiple presentations, many causes	Wick JY, Zanni GR.	2010
27816899	The developmental dynamics of gait maturation with a focus on spatiotemporal measures	Kraan CM, Tan AHJ, Cornish KM.	2017
36236656	Ground-Reaction-Force-Based Gait Analysis and Its Application to Gait Disorder Assessment: New Indices for Quantifying Walking Behavior	Park JS, Kim CH.	2022
30814368	Virtual reality during gait training: does it improve gait function in persons with central nervous system movement disorders? A systematic review and meta-analysis	De Keersmaecker E, Lefeber N, Geys M, Jespers E, Kerckhofs E, Swinnen E.	2019
35274462	Gait as a quantitative translational outcome measure in Angelman syndrome	Petkova SP, Adhikari A, Berg EL, Fenton TA, Duis J, Silverman JL.	2022
24132841	Moving forward on gait measurement: toward a more refined approach	Lord S, Galna B, Rochester L.	2013
21068447	[Gait and gait disturbance]	Shibasaki H.	2010
34029190	Gait Analysis by Causal Decomposition	Peng X, Feng Y, Ji S, Amos JT, Wang W, Li M, Ai S, Qiu X, Dong Y, Ma D, Yao D, Valdes-Sosa PA, Ren P.	2021
23072364	Geriatric syndromes--vascular disorders?	Strandberg TE, Pitkälä KH, Tilvis RS, O'Neill D, Erkinjuntti TJ.	2013
26818868	Regulation of Gait and Balance: The Underappreciated Role of Neuronal Nicotinic Receptor Agonists	Young MF, Wecker L.	2016
33554744	Gait Speed is independently associated with Depression Symptoms in Mild Cognitive Impairment	Naharci MI, Katipoglu B, Veizi B, Tasci I.	2022

26709688	Evaluation and management of crouch gait	Kedem P, Scher DM.	2016
35987823	The Conventional Gait Model's sensitivity to lower-limb marker placement	Fonseca M, Bergere M, Candido J, Leboeuf F, Dumas R, Armand S.	2022
34846376	Functional Gait Disorder, Before and After Treatment	Friedman JH, Sousa K.	2021
36764083	Dual tasking affects gait performance but not automaticity in functional gait disorders: A new diagnostic biomarker	Gandolfi M, Fiorio M, Geroin C, Torneri P, Menaspà Z, Smania N, Giladi N, Tinazzi M.	2023
29336642	Concurrent Validity of Two Gait Performance Measures in Children with Neuromotor Disorders	Ammann-Reiffer C, Bastiaenen CHG, Klöti C, van Hedel HJA.	2019
34370398	Evaluating Gait and Locomotion in Rodents with the CatWalk	Garrick JM, Costa LG, Cole TB, Marsillach J.	2021
34924400	Troubleshooting Gait Problems in Parkinson's Disease Patients with Subthalamic Nucleus Deep Brain Stimulation	Guimarães TG, Cury RG.	2022
36011667	SANE (Easy Gait Analysis System): Towards an AI-Assisted Automatic Gait-Analysis	Sipari D, Chaparro-Rico BDM, Cafolla D.	2022
28268761	Combined gait asymmetry metric	Ramakrishnan T, Muratagic H, Reed KB.	2016
35278691	Kinetic symmetry indices and standing gait analysis: A review of current methods and data	Adrian D, Brown D.	2022
27516008	Wearable sensors used for human gait analysis	TarniŢă D.	2016
35384055	The Contribution of Noradrenergic Activity to Anxiety-Induced Freezing of Gait	Taylor NL, Wainstein G, Quek D, Lewis SJG, Shine JM, Ehgoetz Martens KA.	2022
34960297	Comparative Study of Markerless Vision-Based Gait Analyses for Person Re-Identification	Kwon J, Lee Y, Lee J.	2021
23706539	Can 4-aminopyridine modulate dysfunctional gait networks in Parkinson's disease?	Luca CC, Singer C.	2013
36054444	Specific Gait Changes in Prodromal Hereditary Spastic Paraplegia Type 4: preSPG4 Study	Laßmann C, Ilg W, Schneider M, Völker M, Haeufle DFB, Schüle R, Giese M, Synofzik M, Schöls L, Rattay TW.	2022
28113185	Toward Pervasive Gait Analysis with Wearable Sensors: A Systematic Review	Chen S, Lach J, Lo B, Yang GZ.	2016
33928809	Three-dimensional gait analyses in dizygotic twin athletes	Aydın CG, Hekim HH, Üçpunar H, Öztaş D, Bayhan Aİ.	2021
34833363	Wearable Sensor for Assessing Gait and Postural Alterations in Patients with Diabetes: A Scoping Review	Brognara L, Mazzotti A, Di Martino A, Faldini C, Cauli O.	2021
30909242	Gait Dysfunction in Motoric Cognitive Risk Syndrome	Ayers E, Verghese J.	2019
35336475	Reliability of IMU-Derived Temporal Gait Parameters in Neurological Diseases	Hansen C, Ortlieb C, Romijnders R, Warmerdam E, Welzel J, Geritz J, Maetzler W.	2022
27986427	Gait and energy consumption in adolescent idiopathic scoliosis: A literature review	Daryabor A, Arazpour M, Sharifi G, Bani MA, Aboutorabi A, Golchin N.	2017
35490252	Stepping up to meet the challenge of freezing of gait in Parkinson's disease	Lewis S, Factor S, Giladi N, Nieuwboer A, Nutt J, Hallett M.	2022
35511847	Validation of a Spatiotemporal Gait Model Using Inertial Measurement Units for Early-Stage Parkinson's Disease Detection During Turns	Yang Y, Chen L, Pang J, Huang X, Meng L, Ming D.	2022
17277257	Evaluation of the elderly patient with an abnormal gait	Lim MR, Huang RC, Wu A, Girardi FP, Cammisa FP Jr.	2007
35219146	Number of synergies impacts sensitivity of gait to weakness and contracture	Kuska EC, Mehrabi N, Schwartz MH, Steele KM.	2022
35221197	Theta rhythms may support executive functions in Parkinson's disease with freezing of gait	Zampogna A, D'Onofrio V, Suppa A.	2022
36332288	Gait and axial postural abnormalities correlations in Parkinson's disease: A multicenter quantitative study	Pongmala C, Fabbri M, Zibetti M, Pitakpatapee Y, Wangthumrong T, Sangpeamsook T, Srikajon J, Srivanitchapoom P, Youn J, Cho JW, Kim M, Zamil Shinawi HM, Obaid MT, Baumann A, Margraf NG, Pona-	2022

		Ferreira F, Leitão M, Lobo T, Ferreira JJ, Lopiano L, Artusi CA.	
33892391	Differences in predictors for gait speed and gait endurance in Parkinson's disease	Shearin S, Medley A, Trudelle-Jackson E, Swank C, Querry R.	2021
36316420	Quantification of pathological gait parameter thresholds of idiopathic normal pressure hydrocephalus patients in clinical gait analysis	Möhwald K, Wuehr M, Decker J, Asch EM, Schenkel F, Illigens B, Schniepp R.	2022
36271367	Efficacy and evaluation of therapeutic exercises on adults with Parkinson's disease: a systematic review and network meta-analysis	Yang Y, Wang G, Zhang S, Wang H, Zhou W, Ren F, Liang H, Wu D, Ji X, Hashimoto M, Wei J.	2022
21626560	Milestones in gait, balance, and falling	Nutt JG, Horak FB, Bloem BR.	2011
35452782	Usefulness of measuring maximal gait speed in conjunction with usual gait speed for risk stratification in patients with cardiovascular disease	Ueno K, Kamiya K, Hamazaki N, Nozaki K, Ichikawa T, Yamashita M, Uchida S, Noda T, Maekawa E, Yamaoka-Tojo M, Matsunaga A, Ako J.	2022
23831298	Contribution of new techniques to study the gait in old populations	Gillain S, Petermans J.	2013
26333073	Short case: Gait examination	Fahey M, Adsett D.	2015
33923809	The Smart-Insole Dataset: Gait Analysis Using Wearable Sensors with a Focus on Elderly and Parkinson's Patients	Chatzaki C, Skaramagkas V, Tachos N, Christodoulakis G, Maniadi E, Kefalopoulou Z, Fotiadis DI, Tsiknakis M.	2021
35458810	Detection of Human Gait Phases Using Textile Pressure Sensors: A Low Cost and Pervasive Approach	Milovic M, Farías G, Fingerhuth S, Pizarro F, Hermosilla G, Yunge D.	2022
15563372	Gait unsteadiness and fall risk in two affective disorders: a preliminary study	Hausdorff JM, Peng CK, Goldberger AL, Stoll AL.	2004
18668618	Gait festination and freezing in Parkinson's disease: pathogenesis and rehabilitation	Morris ME, Iansek R, Galna B.	2008
29031570	Relationship among Depression, Gait Disturbance, Disability, and Neurobiological Abnormalities	Rodakowski J.	2018
19447724	Computational intelligence in gait research: a perspective on current applications and future challenges	Lai DT, Begg RK, Palaniswami M.	2009
11347218	Cerebellar ataxic gait	Hallett M.	2001
34633932	Detection of Unsupervised Standardised Gait Tests from Real-World Inertial Sensor Data in Parkinson's Disease	Ullrich M, Mucke A, Kuderle A, Roth N, Gladow T, Gabner H, Marxreiter F, Klucken J, Eskofier BM, Kluge F.	2021
31416377	The effects of muscle vibration on gait control: a review	Layne CS, Malaya CA, Levine JT.	2019
34833749	Gait Disorder Detection and Classification Method Using Inertia Measurement Unit for Augmented Feedback Training in Wearable Devices	Kim H, Kim JW, Ko J.	2021
31301561	The Primary Gait Screen in Parkinson's disease: Comparison to standardised measures	Schmitt AC, Daniels JN, Baudendistel ST, Okun MS, Hass CJ.	2019
10463015	Gait analysis in the therapeutic environment	Coutts F.	1999

APPENDIX E: How to verify FOCA GQM

Goal	Question	How to Verify
1	Q1	Firstly, check if the document has the ontology competencies defined. If they do not exist, the grade is 0. If they exist, answer three sub-questions: Does the document define the ontology objective? (For example: "This ontology models the domain of…"); Does the document define the ontology stakeholders? (For example: "This ontology should be used by…"); Does the document define the use of scenarios? (i.e., the situations in which the ontology must be used). For each sub-question, give one of these grades: 25,50,75,100. Finally, the mean of the three sub-questions must be calculated.
	Q2	If you established grade 0 in the previous question, the competencies were not defined, and you cannot evaluate this question. Thus, the grade of this question is 0. If the competencies exist, check if the ontology responds to what was defined in the competencies document. Grades: 25,50,75,100.
	Q3	Check if the ontology reuses other ontologies. If it does not, the grade is 0. If it does, the grade is 100.

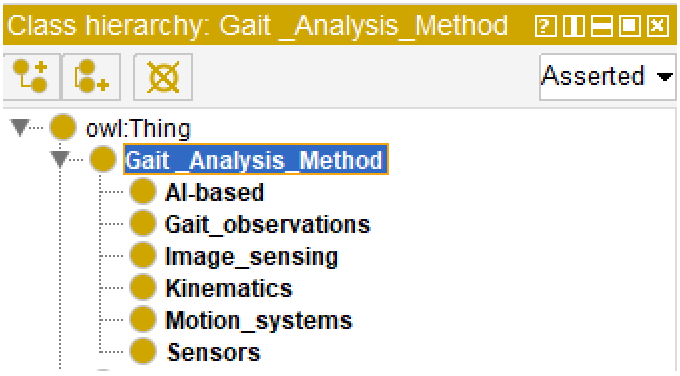
Goal	Question	How to Verify
2	Q4	This question should only be checked if the ontology is type 2. If the ontology is type 1, go to the next question. In this question, check if the ontology does not use much abstraction to define the concepts. If the ontology is full of abstraction (for example: an ontology which models the Facebook site does not need to define what a computer system is, or what a computer is, and other abstraction concepts), the grade is 0. If there are only some abstractions, give a grade between these: 25 (very specific), 50 (moderate abstraction), 75 (many abstractions), 100 (full of abstractions).
	Q5	This question should only be checked if the ontology is type 1. In this question, check if the ontology uses primitive concepts to define the evaluated domain (for example, an ontology which models a person, uses the concepts thing → living being → human being → person to define the person concept). If the ontology does not use abstractions, the grade is 0. If there are only some abstractions, give a grade between these: 25 (very specific), 50 (moderate abstraction), 75 (many abstractions), 100 (full of abstractions).
	Q6	In this question, check if the classes and properties are coherent with the modelled domain. If the ontology is full of incoherences (for example, an ontology which models the concept car has a class lion and the property quantityOfPaws, that is, do not exist in domain), the grade is 0. If there are some incoherences, give a grade between these: 25,50,75. If there is no incoherence, the grade is 100.

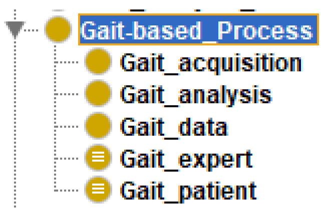
Goal	Question	How to Verify
3	Q7	In this question, check if the classes and properties (functional, transitive, reflexive and others) characteristics contradict the domain (for example: LivingBeing is a subclass of Person in an ontology which models the person concept or socialSecurityNumber is not a functional property, because a person cannot have more than one Social Security Number). If the ontology is full of contradictions, the grade is 0. If there are some contradictions, give a grade between these: 25,50,75. If there are no contradictions, the grade is 100.
	Q8	In this question, check if there are classes or properties which model the same thing with the same meaning (for example, use mouse for hardware and animal). If the ontology is full of redundancies, the grade is 0. If there are some redundancies, give a grade between these: 25,50,75. If there are no contradictions, the grade is 100.

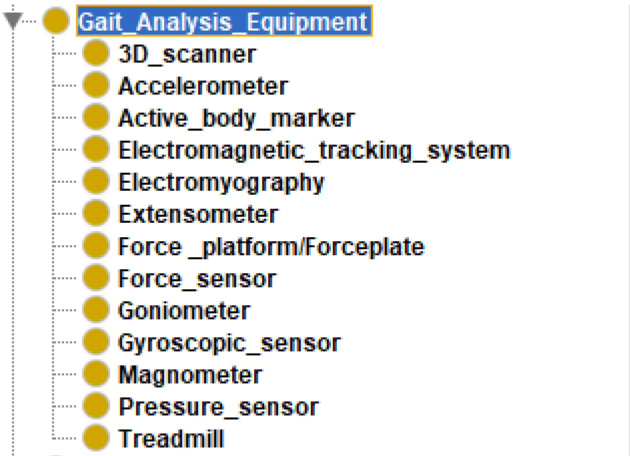
Goal	Question	How to Verify
4	Q9	Save all your records here. In this question, check if, running the reasoner, returns some kind of error. If the ontology is full of errors (or the software stops responding), the grade is 0. If there are some errors, give a grade between these: 25,50,75. If there are no errors, the grade is 100.
	Q10	In this question, check if the reasoner is running quickly. If the reasoner stops, the grade is 0. If there is any delay, give a grade between these: 25,50,75. If it runs quickly, the grade is 100.

Goal	Question	How to Verify
5	Q11	In this question, check if the documentation of ontology exists. If it does not exist, the grade is 0. If the documentation exists, answer two sub-questions: Are the written terms in the documentation the same as the modelling? Does the documentation explain what each term is and does it justify each detail of modelling? For each sub-question, give one of these grades: 25,50,75,100. Finally, the mean of two sub-questions must be calculated.
	Q12	In this question, check if the classes or properties of ontology are written in an understandable and correct form (according to English or another language). If the ontology is difficult to understand or full of poorly written terms, the grade is 0. If there are some errors or a mix of languages, give the grade between these: 25,50,75. If the ontology is well written and one language was used, 100.
	Q13	In this question, check if the existing annotations bring the definitions of the modelled concepts. If there are no annotations, the grade is 0. If there are some annotations, give a grade between these: 25,50,75. If all the concepts have annotations, the grade is 100.

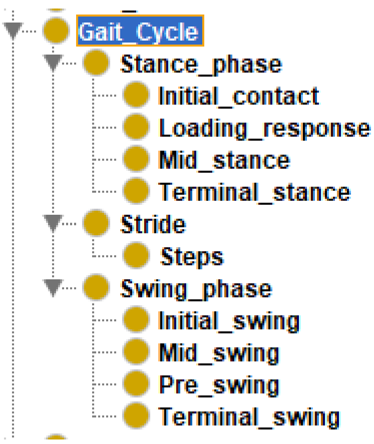
APPENDIX F: Remaining expanded classes that is part of the developed GADO

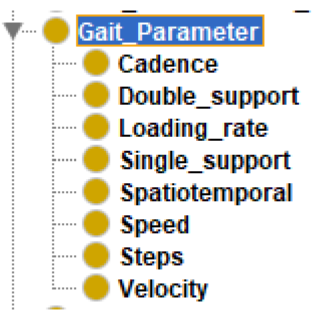


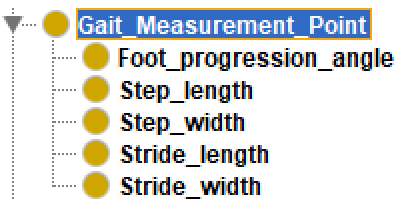





Gait_Assistive_Technology
Actuator
Cane
Crutch
Exoskeleton
Mobility_scooter
Orthotic_device
Prosthetic_leg
Specialised_footwear
Treatment
Pharmacotherapy
Physical_therapy
Gait_training
Rehabilitation_therapy
Surgery
Walker
Wheelchair


Gait_Cycle
Stance_phase
Initial_contact
Loading_response
Mid_stance
Terminal_stance
Stride
Steps
Swing_phase
Initial_swing
Mid_swing
Pre_swing
Terminal_swing


Gait_Parameter
Cadence
Double_support
Loading_rate
Single_support
Spatiotemporal
Speed
Steps
Velocity


Gait_Measurement_Point
Foot_progression_angle
Step_length
Step_width
Stride_length
Stride_width

APPENDIX G: Evaluators' profiles

Project: GADO
Report created by Terrance Marthinus on 22/Jun/23

Quotation Report 1
(4) quotations
Local filters:
Show quotations coded with Professional Background
1:76 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P1
[ ] Academia
2:75 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P2
[ ] Industry
3:75 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P3
[ ] Academia
4:84 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P4
[ ] Research

Quotation Report 2
(4) quotations
Local filters:
Show quotations coded with Area of Specialization
1:77 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P1
[ ] Artificial Intelligence
2:76 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P2
[ ] Systems Developer
3:76 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P3
[ ] Systems Developer
4:86 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P4
[ ] Knowledge Representation

Quotation Report 3
(4) quotations
Local filters:
Show quotations coded with Years of Experience in Professional Practice
1:78 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P1
[ ] 2-5 years
2:77 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P2
[ ] 5-10 years
3:77 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P3
[ ] Above 10 years
4:87 p 3 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P4
[ ] Above 10 years

Quotation Report 4
(4) quotations
Local filters:
Show quotations coded with Years of Experience in Ontology Engineering
1:79 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P1
[ ] 2-5 years
2:78 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P2
[ ] 2-5 years
3:78 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P3
[ ] 2-5 years
4:88 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P4
[ ] 5-10 years

Quotation Report 5
(4) quotations
Local filters:
Show quotations coded with Level of Expertise in Ontology Design
1:80 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P1
[ ] Intermediate
2:79 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P2
[ ] Advanced
3:79 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P3
[ ] Intermediate
4:89 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P4
[ ] Expert

Quotation Report 6
(4) quotations
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1:81 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P1
[ ] Intermediate
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[ ] Advanced
3:80 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P3
[ ] Intermediate
4:90 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P4
[ ] Expert

Quotation Report 7

(4) quotations

Local filters:

Show quotations coded with Level of Expertise in Ontology Evaluation

1:82 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P1

[ ] Intermediate

2:81 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P2

[ ] Intermediate

3:81 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P3

[ ] Intermediate

4:91 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P4

[ ] Expert

Quotation Report 8

(4) quotations

Local filters:

Show quotations coded with Experience level indication of working with Ontologies: inexperienced = 0 and experienced = 1

1:83 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P1

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2:84 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P2

1 [ ]

3:83 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P3

1 [ ]

4:92 p 4 in Gait Analysis Domain Ontology (GADO) Overview Document Guide P4

1 [ ]