Understanding ASD: Design and Development of a Domain Ontology to Assist Professionals In Understanding Autistic Children Based on DSM-5

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Abstract. In recent years, the rate of diagnoses of Autism Spectrum Disorder (ASD) has increased significantly. As a result, ASD has become one of the most studied psychological disorders in various editions of the main literature source for clinical diagnosis, the Diagnostic and Statistical Manual of Mental Disorders (DSM-5). Although it is one of the main problems faced by the public and overall health, there is still a lack of support for the diagnosis, providing a subjective approach, which varies among health professionals. The present work aims to verify the characteristics of autism through an analysis of the DSM-5, to assist health professionals in obtaining answers during the process of identifying factors that allow the diagnosis, in addition to promoting accurate information to researchers regarding people diagnosed with Autism Spectrum Disorder (ASD). Furthermore, this paper describes the project and development of a domain ontology for describing this domain, allowing a more objective way to close an early diagnosis, enhancing the chances of better social insertion of the individual. Adopting the Systematic Approach for Building Ontologies (SABiO), proposed by Falbo [1] we present a domain ontology containing information characteristic of diagnostic criteria A and B to demonstrate clear and structured data, using the DSM-5 and other data sources, providing a reference ontology (for graphical aid) and an operational ontology (for machine-readability). The study presents characteristics according to communication, social interaction, and motor behavior, as well as the level of support indicated for each diagnostic criterion.

Keywords. Domain Ontology, Autism, DSM 5, Diagnosis Criteria, Autism Characteristics, SABiO

1. Introduction

Autism is a neurological condition characterized by affecting social communication, as well as causing repetitive behaviors and restricted interests, often influenced by environ-

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mental and genetic factors, resulting in an increase in its prevalence.[2] ASD - Autism Spectrum Disorder, can be defined as "a pervasive developmental disorder that manifests before the age of 3 and persists throughout life." [3]. Despite all the stereotypes associated with receiving this diagnosis, having autism does not imply being inadequate, but rather the numerous possibilities of working on one's skills and potential. Individuals on the autism spectrum may encounter difficulties when dealing with certain situations. This leads to them exhibiting certain behavior patterns seen as responses to specific occasions. These patterns are reflected in most affected areas: social, communication, and behavioral. Social skills refer to problems related to social interaction relationships that an individual with ASD may experience, as there are children with more complex difficulties who practically isolate themselves in an impenetrable world; different may present difficulties regarding socialization; and those who show very subtle issues, almost invisible to most people, including some professionals [3]. These issues become challenging for those who live with someone diagnosed with the spectrum, as, due to this condition, realizing that isolation may be a trait of ASD and establishing contact can be a rather challenging task. Difficulty in verbal and non-verbal communication is another characteristic area of autism. Some diagnosed children may develop this skill without impairment, speaking clearly and establishing excellent communication. However, some individuals have greater difficulty and tend to produce unilateral language. This type of communication is an interaction "without social reciprocity, seldom used for requesting or labeling rather than for commenting, sharing feelings, or conversing" [4]. Understanding communication patterns makes it easier for healthcare professionals, family members, and educators to adapt intervention strategies and establish communicative acts with autistic individuals [5]. Another point that presents some unique characteristics is behavioral inadequacies, where these behavior patterns vary. This variation is divided into two categories: (1) stereotyped and repetitive motor behaviors and (2) disruptive cognitive behaviors. The former category concerns "jumping, body swinging and hand movements, clapping, shaking or twisting fingers, and making unusual facial expressions" [3]. Generally, these actions are performed repetitively and numerous times at certain moments. The second category consists of responses made by autistic children, it is related to "compulsions, rituals, and routines, insistence, sameness, and circumscribed interests that are characterized by rigid adherence to some rule or the primal need to possess things for own fulfillment" [3]. It can be understood that each person has their specificities and needs. Observing the signs of the spectrum significantly assists in treatment. It is important to note that there are autistic individuals with many abilities and talents. Thus, despite some difficulties, tracing these behavior patterns, can help autistic individuals deal with certain situations and develop their skills. However, it is necessary to consider the needs of each individual, addressing their particularities in a personalized manner. According to McCarty and Richard, 2020 [6] the diagnosis of autism can be challenging since the Diagnostic and Statistical Manual of Mental Disorders (DSM-V) serves as a basis for extracting characteristics based on the criteria, but it does not offer any specific tests. As a result, there are no clinical exams that prove the prevalence of autism in a given individual. The discovery of the diagnosis occurs through questionnaires, interviews, medical opinions, and direct interactions, which can lead to a delayed

In light of these aspects, according to the Diagnostic and Statistical Manual of Mental Disorders, 5th edition [4], a thorough understanding of the characteristic profile of

children or adults within the autism spectrum is essential to identify traits associated with this diagnosis accurately. A detailed understanding of these characteristics is fundamental in the process of identifying and understanding autism, allowing for a more precise and targeted approach to assessment and intervention. Following this perspective, an analysis is proposed to create an ontology based on the DSM-5, which targets the representation of the information present in the manual in a structured manner, especially related to the characteristics linked to communication, social interactions, and behavioral aspects of ASD. The purpose is to expedite the search for knowledge, offering an organized and coherent representation of information, and contributing to a clearer and more accessible understanding of individuals with Autism Spectrum Disorder. According to Turnock et al. (2022), the lack of knowledge about ASD can lead to a reduced quality of life for autistic individuals, as they may feel out of place and end up camouflaging their behaviors. In light of this, a deep understanding of autistic characteristics is necessary to encourage acceptance, care, and the development of individuals within the autism spectrum. [7] To clarify the characteristics that compose the autistic person, this research aims to develop an ontology composed of the characteristics existing in diagnostic criteria A and B of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5), to provide clear information about the autism spectrum. According to McPartland et al. (2012), for the diagnosis of autism, it is necessary to meet at least six attributes from the behavioral criteria, including two from the domain of social deficit, and one prevalent characteristic from each of the areas of communication and restrictive and repetitive behaviors.[8] Furthermore, the present study aims to assist psychologists and psychoeducators in conducting the study during the data collection process to outline the profile of each individual with ASD. The rest of this paper is divided as follows, section 2 provides the details regarding the ontology development methodology, section 3 aligns the design step, allocating the requirements and portraying the referential and operational ontologies, and section 4 provides the conclusive thoughts along with future recommendations and challenges faces throughout the development of this paper.

2. Ontology Development Methodology

To systematically portray the ontology development, we are grounded by the Systematic Approach for Building Ontologies (SABiO) [1]. This methodology provides a 5 steps roadmap toward the systematic development of a domain ontology, i.e., Purpose identification and requirements elicitation, ontology capture and formalization, design, implementation, and testing. The methodology divides the ontology development into two artifacts, the reference ontology, and the operational ontology. Steps one and two are performed to provide the former, a graphical artifact that a foundational ontology must ground. The guideline proposes the usage of the Unified Foundational Ontology (UFO), extended to the OntoUML language ². The use of the OntoUML class diagram becomes necessary for the representativeness of the ontology aimed at assisting healthcare professionals in obtaining early diagnosis, as the environment provided by the tool is fully interactive. It allows for the demonstration of distinctions between types of entities, mereological (part-whole) relationships, and other factors that clearly express the concepts pre-

²https://ontouml.readthedocs.io/en/latest/intro/ontouml.html

Table 1. Stakeholders and Roles Regarding the Ontology Development

Stakeholder	Role
Psychopedagogist 1	Providing the problem landscape, along with questions and approaches toward the characteristics, present on the criteria A and B diagnosis. Those characteristics are aligned with the DSM-5.
Psychopedagogist 2	Clarifying concepts and meaning toward the characteristics of ASD.
Psychologist	Validating and Evaluating the acquired information.

sented in the Diagnostic and Statistical Manual of Mental Disorders - DSM-5. We must employ steps three and four for the operational ontology to provide a machine-readable ontology, e.g., in Web Ontology Language (OWL) ³. Finally, the ontology designers must test, and validate the ontology. In which, after being modeled, the diagram is exported to the Protégé tool so that rules and commands can be applied, with the reasoner executed to ensure consistency and infer new facts capable of addressing competency questions, ensuring that the data obtained is accurate.

3. Ontology Design

Ontology in computer science can be understood as the explicit representation of a set of concepts (classes), attributes (properties), and the relationships that can exist within a specific domain, playing a fundamental role in the semantic web by facilitating interoperability between systems. Antoniou Harmelen (2004) [9] According to Neto (2022) [10], an ontology consists of triples that are constituted of the subject, which is what some information is expressed about; predicate, which constitutes the way two entities are interconnected; and object, which consists of the individual or entity associated with the subject through the predicate. Proceeding from this assumption, an ontology was developed to facilitate studies and support healthcare professionals and society, to promote understanding of individuals with autism, and to disseminate broader knowledge about ASD. Table 1 demonstrates the process of collecting information for the construction of the ontology containing the characteristics of autism according to the diagnostic criteria existing in the DSM-5 manual. Along the way, efforts were made to identify the needs and objectives to be addressed with the ontology, and the information was collected from two psychopedagogists and one psychologist. The former provided and clarified information regarding the discovery process of autistic characteristics in a particular individual, while the latter evaluated the information structured in the ontology constructed in question.

3.1. Ontology Requirements

In the initial phase of gathering relevant information for efficient ontological development, creating the Ontology Requirements Specification Document (ORSD) stands out as one of the most important and promising stages. According to Matrazo Fernando (2022) [11], creating an ORSD is a fundamental piece as it contains all the necessary

³https://www.w3.org/OWL/

information for developing an ontology that stores all the functional and non-functional requirements in the study.

Therefore, the ORSD was based on collecting the necessary information for documentation creation. Thus, an attempt was made to verify the information in the Diagnostic and Statistical Manual of Mental Disorders, as it is a document used by health-care professionals such as psychologists and psychoeducators in their studies regarding ASD. The documentation aims to raise questions that characterize diagnostic criteria A and B according to the DSM-5 manual, to deepen the understanding of these criteria, as indicated in Table 2.

Table 2. Ontology Requirements Specification Document

1. Purpose

The development of the hereby proposed ontology aims at the precise and embracing representation of the factors and particularities related to autism, to provide a solid foundation for research about ASD, as well as providing parental support on the early diagnosis.

2. Scope

The ontology focuses on identifying the level of support, relating to the social condition provided by the DSM-5, employed to criteria A and B.

3. Language

OntoUML with further translation to OWL

4. Intended Users

User 1. Psychologists Psychiatrists, Psychopedagogists, and Physiotherapists provide support in research and facilitate the diagnosis.

User 2. Family members of autistics looking for relevant information about the characteristics of ASD.

5. Intended Uses

Use 1. Graphically represent the ASD domain - Relatives may use this reference in order to better understand autistic behavior.

Use 2. Discovering tendencies and patterns - Researchers may foster on the provided ontology to answer questions regarding criteria A and B.

Use 3. Provide information about the characteristics - Psychologists may use the ontology to find information regarding the level of support.

Use 4. Exhibit the types of language, behavior, social interaction, and characterization that define ASD - Relatives may refer to the ontology to enhance knowledge about the domain.

6. Non-Functional Requirements

NFR1 - The ontology must be grounded by the Diagnostic and Statistical Manual of Mental Disorders [4]

NFR2 - The ontology must be grounded by the book Singular World, Understanding Autism [3]

NFR3 - The ontology must be available on an open-source GitHub Repository

7. Functional Requirements

Competency Question

CQ1 - What characteristics compose criterion A?

CQ2 - What characteristics compose criterion B?

CQ3 - What are the kinds of behavior?

CQ4 - What are the kinds of verbal communication?

Even though we closed our scope into 4 CQs (once we adopt a CQ-driven development) [12], if we would like to parameterize them, we could enhance the knowledge retrieval capabilities, such as *What kinds of support is used in criterion B?*, or even *What are the kinds of non-verbal communication?*.

3.2. Reference Ontology

According to Guizzardi et al. (2022) [13], OntoUML is a language capable of providing a suitable and clear representation of information, allowing for more semantics, that is, communicating the meaning of the entire characteristic context. Hence, the next stage consists of creating an OntoUML model to promote accessible and clear information regarding the characteristics that compose the behavior, socialization, and types of language of individuals with ASD. In accordance with the author, the graphical representation of data in UML class diagrams allows for increased efficiency in communicating and sharing information from a specific area to different groups of people Guizzardi & Wagner (2004) [14]. Thus, we can affirm that the ontological applicability facilitates access for researchers and healthcare professionals who are seeking to investigate specific medical conditions in a more structured and comprehensive manner. During this phase, the final model of the ontology was presented to the two autism specialists who participated in the development, validation, and evaluation process. Tests were conducted to evaluate efficiency, and in this stage, a preliminary model was demonstrated, in which the psychologist evaluated and proposed the application of diagnostic criteria A and B to provide more clarity in the presentation of information. This clarity focuses on usability, allowing individuals with unsatisfactory knowledge regarding autism to understand how the process of identifying autistic characteristics works according to the diagnostic criteria present in the DSM-5.

For the hereupon introduced reference ontology process of development, we provide Table 3 containing the involved agents, and their roles regarding the development, testing, and validation stages.

Stakeholder	Role
Psychopedagogist 1	Specialist, provided the interpretation regarding the usage of simple communication relevant to the populational understanding of individuals diagnosed with ASD.
Psychologist	Specialist, verified and partially validated the final version of the ontology, providing feedback regarding how the information has been structured, and the alignment with the DSM-5.
Jéssica Lima	Ontology Designer, Ontology Engineer, Ontology Tester, Domain Specialist.
Victor Oliveira	Ontology Designer, Ontology Engineer, Ontology Tester.

Table 3. Stakeholders and Roles Regarding the Ontology Ddevelopment

Furthermore, through the acquired information based on the DSM-5, and after being validated by domain experts, we developed a reference ontology focusing on the accessibility and endorsement of knowledge to the general users. Figure 1 showcases the graphical representation of the reference ontology.

Although the explanation of the concepts encompassing the characteristics of the Autism Spectrum is beyond the scope of the present research, some of them will be shortly introduced. It was found that Criterion A is directly related to communication and social interaction, and Criterion B is related to restricted and repetitive patterns of behavior [4]. From collecting this information, an attempt was made to conceptualize the characteristics that define the necessary support for individuals with ASD. Based on this, their use provides a solid and standardized foundation for the classification of the diag-

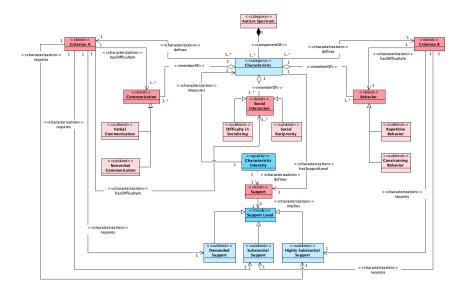


Figure 1. Reference Ontology

nosis, as well as promoting interoperability among different systems and clinical studies. The application of these criteria allows health professionals to easily access the material present in the DSM-5, ensuring a more agile and precise diagnosis. Grounded by the SABiO methodology, we employed the OntoUML language to develop the reference language. As an extension of the Unified Foundational Theory, the classes are further classified as UFO stereotypes. The established classes comprise seven types to identify and categorize the exposed elements. Firstly, we introduce the class *Autistm Spectrum*, which is classified as a category stereotype, holding to its broad nature, it encompasses different identity groups, and it is a rigid stereotype. It is a rather trivial deduction to affirm that the Autism Spectrum is composed of Characteristics, which is also classified as a Category, and it is the main responsible for categorizing the Criterion A, and Criterion B. Furthermore, we introduce the classes Communication, Social Interaction, and Behavior as kinds. A kind, in UFO, provides identity, and it is a rigid FunctionalComplex, i.e., a whole, that bears parts contributing in different ways to its functionality. Those kinds are memberOf the category characteristic, once they hold the same identity provider characteristic, and their existence does not provide an allowance for the characteristic existence, i.e., the characteristic would exist if those kinds do not. Further, The Communication kind is divided into two subkinds, the Verbal Communication, and Nonverbal Communication. Subkinds are also rigid stereotypes, but they do not provide identity, hence, the principle of identity is linked to its superclass. Furthermore, this also happens to the Social Interaction, which is a kind, that is divided into Difficulty in Socializing, and Social Recriprocity. Finally, the class Behavior, introduced as a kind, is also divided into Repetitive Behavior, and Constraining Behavior classified as subkinds.

Furthermore, we introduce the concept of *Characteristic Intensity*, classified as a *quality*. The nature of the stereotype quality is correlational to the definition of *moment*, which in UFO is something that is existentially dependent from its bearer, and it must inhere in from it. In this case, the *Characteristic Intensity* is only enabled by the previous

existence of *Characteristc*. In addition, the *Characteristic Intensity* defines the *Support*, which is a *kind*, and it is characterized (implies) a *Support Level*, which is defined as a *mode*. A mode stereotype, in UFO, is similarly defined as *quality*, however, it must provide a *framework* for how something should happen, in other words, a way something should behave. Finally, the *support level* is divided into three *subkinds*, i.e., *Demanded Support*, which is the minimal support level, *Substantial Support*, and as a higher level of support, *Highly Substantial Support*. The DSM-5 provides the definitions.

Finally, we can characterize each criterion by the three members of *characteristics*, and the *support level* of each. The *Criterion A* is linked through a characterization relation to *Communication* (*hasDifficultyIn*, similarly,*hasDifficultyIn Social Interaction*, and *requires* either a *Demanded Support* or *Substantial Support* level of support (CQ1). Whereas *Criterion B* is solely dependent on *Behavior*, requiring either a *Substantial Support* or a *Higly Substantial Support* level. Those classifications were also provided by the DSM-5 (CQ2). As proposed by the NFR3, the reference ontology, operational ontology, and all supplementary material are available in an open-access GitHub repository ⁴.

3.3. Operational Ontology

With the creation of the graphical representation and generation of classes, it is necessary to describe the internal study in order to provide answers to the competency questions defined in the ORSD document. Thus, after the demonstration of information through OntoUML, a translation is made for the OWL language in which the study will be conducted with the assistance of the Protégé tool. "An image is worth a thousand words," as expressed by Elkashef, et al. (2020) [15], in which he defines OWL as a language designed to objectify ontologies based on the semantic web, to promote explicit representation and specification of a conceptualization. Considering this, when using the Protégé tool, it becomes feasible to manipulate the information previously graphically represented in the class diagram, now converted to the OWL format. Within the tool, it is possible to observe the classes previously built in OntoUML, in which each one is composed of its defined attributes that distinguish them containing information about the characteristics, diagnostic criteria, and level of support belonging to the autistic individual. Figure 2 shows the respective class hierarchy.

As demonstrated, the child classes are characteristics that define the parent classes, such as types of verbal language, non-verbal language, restrictive and repetitive behavior, social reciprocity, and social interactions. To represent the differences between the attributes mentioned here as child classes, the "disjoint with" was assigned so that it would be possible to make distinctions while searching for answers to competency questions.

The Protégé tool uses a property to find relationships between classes, the Object Properties, as depicted by Silva et al. [16]. Therefore, the project defended here utilizes these properties to find relevant information capable of demonstrating the characteristics of the autistic spectrum, as can be observed in Figure 4.

The Object Properties *hasSupport* received the class *criterion* Aas its Domain and the class *characteristic* as its Range. The properties *hasHighlySusbtamtialSupport* and *hasSubstantialSupport* received both the classes *criterion* A and *criterion* B as their Domain, and the class "characteristic" as their Range. As a final step, instances were cre-

 $^{^4 {\}tt https://github.com/jessicaruana/Autism-ontology}$

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Figure 2. Class Hierarchy



Figure 3. Object Proeprties Hierarchy

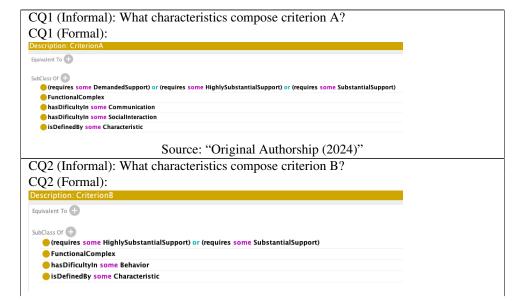
ated in the Individuals tab in the Protégé mechanism, demonstrating the type of support required according to each diagnostic criterion attributed to the autistic individual.

It is worth noting that this ontology is intended for identifying the characteristics contained in diagnostic criteria A and B as advocated in the DSM-5, in order to find attributes that demonstrate the type of support to be used according to the particularities that differentiate them. Therefore, the Object Properties maintain the relationships between the instances of the Support Level class.

3.4. Ontology Validation

According to [12], we must differ in verification, validation, and evaluation processes to build a useful and widely accepted ontology. The former relates to the correctness of the ontology, i.e., to the axioms correlations, classes, and properties structure. The validation of a domain ontology is provided by the level of accuracy it presents its given domain, assessing its reflection of the real world, framed by its level of granularity (one may always take a deeper look, inspiring a more granular approach). Finally, the evaluation metric regards the acceptance of the ontology by a group of active users. In agreement with the author, the validation and verification stage is a crucial part for the ontology to be able to respond to all requirements for which it was designed efficiently. To achieve this, it is necessary for the mechanism to be reviewed by professionals and domain experts for which the ontology tool was designed, in order to ensure the accuracy of the requirements for which it was developed. [17] To verify our ontology, we propose using the previously mentioned reasoner and checking the technical capability of the ontology, by showing no semantic errors. The reasoner can infer from the ontology, which showcases its connection. We propose a twofold approach: answering the CQs by querying the ontology with SPARQL queries and assessing the results for validation, and providing axiomatic classification for concepts. The results were evaluated by the stakeholders, and they showed to be satisfactory, confirming the partial completeness level of validation ([12] defines partial completeness because of the subjective nature of an ontology, which further depends on its granularity, i.e., one could always provide a deeper granularity, depending on its goals). Table 4 provides the Informal Competency Questions from the ORSD, and the Formal Competency Questions, which were translated either to SPARQL queries, or axiomatic representation.

Table 4.: Formal and Informal Questions





4. Conclusion

The main role of the ontology aligns with the provision of structured knowledge and explanation, along with relevant answers. Furthermore, using such methodology in the autism study context becomes fundamental, supplying organized and detailed information that facilitates the clear and efficient comprehension of the characteristics that compose ASD, i.e., communication, social interaction, and behavior. This explanation allows a wider comprehension by society. Moreover, as a subjective matter, the evolution process is always open. This ontology was framed by the requirements proposed by stakeholders, and alignment with literature. The chosen methodology provides a broader application for the ontology, once we could use two artifacts for validation and refinement. For instance, we could partially answer CQ1 with Figure ??, once it provides the relationships for criterion A, allowing a user-friendly visualization of data, hence, more professionals could share their thoughts and evaluate our work.

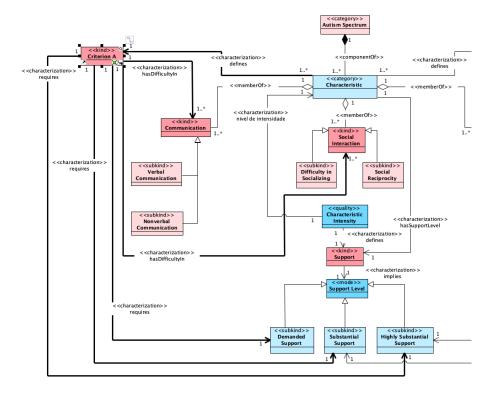


Figure 4. Criterion A characteristics on focus in reference model

We may state a few challenges while developing such an ontology. Work with health professionals always involves an ethical approach, especially when dealing with children observation and data sharing. Furthermore, the encounters with specialists were limited, in average, each specialist have been consulted after a major change in the reference model, hence, 2-3 times per person. Overall, ontologies, especially domain ontologies, are designed to answer specific conceptual questions, defining their granularity. For that, we propose as future work the implementation of case studies, retaining the instantiation

of the ontology, along with the implementation of different observed properties, granting the identification and intensity of the characteristics. Finally, as a technical approach, we aim to develop different SPARQL queries, outgrowing the current knowledge retrieval capabilities, supplemented by SWRL rules, and SHACL constraints, thus, enhancing the supplementary diagnosis capabilities for health professionals.

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