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# Comprehensive Ontology Design for Autism Spectrum Disorder

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**Comprehensive Ontology Design for Autism Spectrum Disorders**

By

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# Abstract

Ontology is a formal explicit description of concepts in the knowledge domain. In recent years, developing ontology in different domains is a hot topic for many researchers, especially in the medical field because of the benefits offered to users. Using ontology allows sharing and reusing domain knowledge in an efficient and explicit way. In particular, ontology in medical field can facilitate the access to query data, precise knowledge, and seamless sharing of electronic medical records (EMR). Thus ontology increases the accuracy of doctor's diagnostic decision.

Autism Spectrum Disorder (ASD) often presents with difficulties in verbal and non-verbal communication, behavior and social interactions. Autism is difficult to define due to the complex heterogeneous disorders in this domain and to the lack of coherent set of knowledge that deals with all aspects of autism. The purpose of this research was to address these shortcomings by developing a comprehensive ASD ontology that formally conceptualizes both domain and operational autism knowledge, unifies autism terminology, and facilitates access to precise autistic information for both general public and expert users, thus enabling better diagnostic and treatment decisions.

To build such ontology, we investigated many medical research works in the various areas of autism such as disorders, effects and treatments of ASD. The study was done with the purpose of extracting and gathering information from the most trusted sources such as existing ontologies, standard textbooks, relevant articles and clinical studies. These sources were used to build a semantic map linking key concept classes. Mainly we focused on properties and relationships between these classes to formally describe the autistic domain and operational knowledge and to bring the scattered knowledge into the ontological form. Ontology instantiation for each subclass was based on pilot studies and clinical cases.

The system was implemented using Protégé, an ontological framework developed by the Stanford Center for Biomedical Informatics Research at the Stanford University. The ontology was built using the Web Ontology Language (OWL). OWL is a semantic web language designed to indicate the rich and complex knowledge of the domain. Moreover, we developed a basic web query system for the ASD ontology to present the ontology information to different users around the world. The developed system has been evaluated to measure quality of embedded knowledge, ontology correctness and the usability of its web query system.

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# Chapter 1

# Introduction

## 1.1 Autism spectrum disorders

Autism Spectrum Disorders have been a subject that has attracted the attention of researchers, clinicians, and the general public for the past few years. The topic of autism is a huge area in the medical field. Despite much research being done over the last decade there is still a lack of knowledge, evidence, and research compared to other medical subjects. Autism, first described by Leo Kanner in 1943, comprises problems in social interactions, difficulties in communication, and repetitive behavior (L.Kanner, 1943). Autism Spectrum Disorder (ASD) is a range of complex neurodevelopment disorders of brain functions (Maricela Alarcon, 2008). There are many different categories of the ASD disorder. A disorder is a lifelong disability that can be a result of social impairments, communication problems, stereotype repetitive, and restricted behavior. ASD appears in very early brain development, however many scientists believe that the most obvious symptom's and signs of the ASD disorder emerge between 2 and 3 years of age (Gillberg, 1990 ,Charman, 2005). Hence there are various ASD tests and screening tools for those children that can be suspicious of autism in early ages based on their abnormal symptoms.

Autism spectrum disorders symptoms can be present in a range of combinations of disabilities. People with autism spectrum disorders (ASD) have different levels of intelligence and difficulties due to the type of disorder (Teaching Students with Autism Spectrum Disorders, 2003). The exact causes of autism spectrum disorders are still unknown, although there are some factors which are involved in developing ASD such as; genetic condition and environmental factors. Scientists are trying to find the genes that have a role in developing autism spectrum disorders and how these genes are affected and the genetic transmission among them (Teaching

Students with Autism Spectrum Disorders, 2003). There are also some biological causes involved in ASD but none of them are part of the main syndrome. For instance, many researchers present the relation between the fragile X syndrome (FXS) with autism and how can FXS impacts with autism spectrum disorders disabilities (Belmonte, 2006).

American Psychiatric Association (APA) published The Diagnostic and Statistical Manual of Mental Disorders, 5th Edition (DSM-IV) to present all current mental disorders. In DSM-IV, ASD autism spectrum disorder has been grouped in Pervasive Developmental Disorders (PDD) as a disorder in which there is impairments in reciprocal social interaction skills and communication skills, and the presence of stereotypical behaviors, interests and activities.

People with autism spectrum disorder show different quality of social interaction. A lack of interest to interact with others, accompanied with the inability to use communication skills to respond and understand individuals are all causes for people to have limited social interaction, difficulties to establish the conversation, interact with others, and involve into social environment. All people with autism spectrum disorders deal with language and communication difficulties at different levels. Most people with ASD have difficulties in understanding verbal information and long verbal instruction (Teaching Students with Autism Spectrum Disorders, 2003). There are variations of difficulties among the individuals. Some individuals are nonverbal, while others communicate in extensive language or they communicate only with people that they know. There are some individuals with normal intelligence, usually referred to as high-functioning; however, they may have difficulty comprehending verbal information (Teaching Students with Autism Spectrum Disorders, 2003). People with autism spectrum disorders also demonstrate unusual behaviors such as aggression, destruction, screaming, self-injury behaviors or tantrums, and stereotypical behaviors. Studies indicate that autistic people have psycho-educational profiles and deficits in many cognation functions and memory problems are the reason for many learning difficulties in those individuals.

Autism spectrum disorders can be detected at 18 months old or younger by ASD screening instrument, which can be helpful for treatment efficiency (Autism Speaks). With an early screening for ASD this allows many people to get better and quicker treatment and improve the

outcome for young children with autism. Autism screening tools are diagnostic tools for autism spectrum disorders for children and toddlers. These tools can be used by therapists and even parents to diagnose autism. For instance, Social Communication Questionnaire (SCQ) and the Developmental Behavior Checklist–Autism Screening Algorithm (DBC - ASA) use autism screening tools to help diagnose autism at the pre-school phase.

In this research work, autism spectrum disorders domain knowledge has been described using ontology, providing many benefits that can be interdicted to the field for users as human or machine. The main benefits of the autism spectrum disorders ontology will be explained in the thesis contributions section. Ontology is a branch of philosophy that deals with nature and the organization of reality (Obitko, 2007). Ontology consists of classes, concepts, data values, and relations among them to describe the targeted knowledge domain. Additionally, ontology is an important layer of Semantic Web. Semantic Web is an effort to enhance the current web so that computers can process the information presented on World Wide Web (WWW), interpret and connect it, and help users find required knowledge (Obitko, 2007).

Autism spectrum disorders ontology has been divided into three different classes representing disorders, effects and treatments. Each class consists of different subclasses based on the validated data, which is extracted from verified sources. Class of disorders, indicates the knowledge domain of all the available disorders for ASD in social impairment, communication difficulties, and behaviors problems.

The effect class of ASD disorders indicates the effect for autism disorders and each disorder could have one or more effect on autistic people. The treatment class introduces all the available and verified treatment methods and techniques by professionals. Treatment methods consist of therapies for autism, biomedical treatment including dietary and the supplement that can be helpful for autistic people.

## 1.2 Research Contribution

### 1.2.1 Conceptualization of knowledge

The area of research in autism is focused on cognition, clinical phenotype, treatment, social function, brain imaging, and genetics, to name a few (Young, 2009). The autism topic is huge because heterogeneous disorders are available in the field. Therefore, the numbers of individual studies are greater than clinical studies. Although many researches and clinical studies have been done in different countries on a variety of autism spectrum disorders types, such as treatment methods, disorders and effect of treatments, it is still not easy to access all of them for use as a researcher, therapist or parents.

Scattered knowledge is one of the main issues in many fields, especially in autism spectrum disorders that bring difficulties to many users when trying to access the information among many different types of work and research that have been published. Another issue with scattered knowledge is lack of semantics relationships among the available different concepts and information in the domain. With a variety of informational sources with different conceptualization in the same domain and the huge number of work with no semantic relationship between them brings the less likely chance to take advantage of the studies and researches for families and therapist to help autistic people. For instance, it's a pretty hard job for a therapist to access all available successful treatment methods for a specific disorder that has been introduced by multiple researchers in the field at different times. Moreover, it could be a complicated task for those interested users in autism spectrum disorders research or parents of autistic children to go through many articles of different researchers to discover the effect of different therapies on autistic people and their relations.

Consequently, one of the thesis contributions is to present one conceptualization for autism spectrum disorders domain to indicate semantics relationships among the available scattered knowledge from different sources. Hence all the related concepts and information from trusted sources in the field have been gathered in form of ontology in order to make one conceptualization for the autism domain. Ontology is a formal specification of a shared

conceptualization of a domain of interest (Graber. T 1993). The domain of interest here is autism spectrum disorders and the conceptualization is the available information and concepts which have been gathered among the articles and clinical studies.

### 1.2.2 Unification of terminology

Researchers across the world have been participating in various experiments dealing with many topics and concepts related to autism spectrum disorders; hence there is a lot of terminology available for autism. The main issue with some terminology is duplication of data in an altered explanation and indicating the same result. The duplication of data can happen based on two situations; first the researcher has no access to a reliable source of information to find out about the multitude of previous work that have been done by other researchers on the same topic in the field. Second, there are many heterogeneous researches that have been done over the same concept in the field by different people, and the results of those works have been published in different sources. Therefore, due to the use of different terminology, researchers cannot find the various sources that are describing the same concept.

A huge terminology and data duplication resulted in a complex domain, such as that of autism spectrum disorders. Data duplication could cause many problems for both researcher and users. Researching on the same topic that has been already investigated by someone else, is not really appreciated in the academic and research world, unless there is an improvement for a targeted concept. Moreover, it is waste of time and money for something that is not very useful to be used or researched on for academic and research purposes. Additionally, data duplication makes accessing data more complicated for users in term of going through the same data and results but phrased differently. For instance, in the field of treatment for autism spectrum disorders, especially therapies, data duplication has been perceived more in the form of clinical studies. Clinical studies are very common in medical domains and also in autism field to indicate the efficiency of treatments and treatment behaviors during certain period of time and for different cases of patients.

Therefore, another contribution of this thesis is to unify the terminology and semantics that exist in the autism spectrum disorders domain. One of the important benefits of ontology is to describe the knowledge of the domain and merge all related semantics and terminology around agreed-upon concepts. Different data and concepts have been gathered from various trusted sources in form of clinical studies, articles, and books to build ontology without any duplication of data or semantics. This is beneficial for researchers, parents of autistic people, and any other users who are interested to know more about concepts in autism spectrum disorders by experiencing easier access to the unified terminology.

### 1.2.3 Easy access to information

As it is mentioned above, being able to have easy access to data and published documents is one of the main concerns because of the complexity of domain knowledge and related sources that are of different types and sources. Autism spectrum disorders domain includes a huge number of clinical studies and articles that have been published about treatment methods and the effect that treatment has on individuals, as well as the effect of autism disorders and the involved factors that provoke the autism spectrum disorders. Creating ontology based on published information from verified sources provides an environment to share common understanding of the information structure for both people and machines. Additionally, autism spectrum disorders' ontology can be manipulated by humans and systems for better ontology structure in the future.

Ontology is a conceptualization that can be accessed by humans and systems. Hence the target domain knowledge can be reused by users at anytime. Facilitating the access to information by humans and machines is one of the main benefits of using ontology. Ontologies are flexible in terms of updating the concepts and relationships between them, as well as adding new concepts and relations in the future, whenever new knowledge evidence and sources are made available.

Huge amount of data has been gathered for the ontology from a variety of sources on autism disorders such as, the effect of disorders, and autism spectrum disorders treatment, thus bringing them together as a merged source. Duplicated data among the information has been defined as unique concept and relation between the available concepts that have been indicated in the

ontology. Each concept can have one or more relationship with other concepts, and identified relationships assist the users to access precise information and to have a better understanding of the domain knowledge, thus facilitating knowledge analysis.

Consequently, the use of autism spectrum disorders' ontology provides all above mentioned benefits for both users and applications. Moreover, the designed system provides an easy access to the published trusted information and the merging of the information from diverse sources into unified terminology for humans and machines. Represented knowledge in the created ontology includes, in treatment fields, disorders field, and the effect of disorders in autism. The autism spectrum disorders' ontology system can be used by anyone who wants to do a project about autism or who are interested to know more about this field. The created ontology can be updated at any time by researchers and professionals based on new data and concepts in the field. These updates should remain compatible with the recent knowledge and users query.

### 1.2.4 Users query system

One of the main challenges for many query information systems is how easy users can access specific information in large complex repositories. The developed ontology system for autism spectrum disorders allows normal and professional users to inquire about any autism information in terms of autism disorders, effect of disorders on autistic people, and available treatment for those disorders. The system is comprehensive in order to reveal the relationships between disorders, effect and treatments of autism spectrum disorders. The developed system targets two types of users. First, normal users such as parents of autistic children, or any other person with special interests in autism. Second type of users could be professional users for instance, doctors in the field of autism, therapists, researchers, or any other professional that deals with autistic people.

The main beneficial properties for treatment methods of autism have been defined in the ontology and can be accessible by users of the system. The relationship between concepts assists the user to find out about the connections between the disorders, effect of disorders, and treatment for disorders in autism spectrum disorders.

The query system provides a user interface that could be used by any user that deals with autism spectrum disorders for different purposes such as supporting them to make a better diagnostic decision; helping users to find available intervention of autism based on the disorders and effect of physical and mental difficulties in autistic children; facilitating the access to autism data; and demonstrating the relationships between autism concepts for researchers. Therefore, to provide such services for different users, the Web query system allows users to access all ontology information anytime anywhere. The communication portal has been provided beside the autism spectrum disorders ontology for professional and typical users to contribute from different part of the world to improve the system in future based on their experience of using the system and user's observation in autism field.

## 1.3 Summary of the thesis

In Chapter two, different fields of autism spectrum in terms of disorders, treatment and disorders effect of autism spectrum disorders are explained. The role of ontology in the medical domain and the benefits of using ontology for medical purposes have also been discussed. Moreover, work that has been done in this field by different researchers and their main contributions over the last decade are covered in this chapter. Also the problems with past researches for creating autism spectrum disorders ontology have been discussed.

The methodologies of the work have been explained in Chapter three of this thesis. The design of the created autism spectrum disorders ontology is described in terms of classes, their subclasses and concept properties. The structure of each subclass with the sources of the extracted knowledge for each subclass are also shown in Chapter three. The three main subclasses of ontology are disorder class, effect class, and treatment class.

Chapter four presents the experimental result of the system, it shows the usefulness of the created ontology system for different kinds of users and how the designed system helps users accessing the basic precise information and relationship between the concepts of the autism. Chapter four also describes the way users can work with the built system by exhibiting the different features of the system, including the ability to update embedded knowledge with new concepts and

resources about autism. Chapter five presents conclusions drawn from this research and the major benefits of the built ontological system. It also describes future research work.

# Chapter 2

# Background and Literature Review

Before reviewing the past related work and models developed by different researchers, two important knowledge concepts need to be explicit in order to better understand the core of the work presented in the thesis. Autism spectrum disorders (ASD) is the first concept which the thesis is based upon and the main purpose of this research is to help autistic people improve their quality of life by using the knowledge of ontology. In the following section, autism spectrum disorders have been explained from a number of different perspectives. The second required knowledge for better understanding of the thesis methodology is ontology. The basic knowledge of ontology and the benefits of using ontology is explained in the second section of this chapter. Moreover, the role of ontology in the medical field is discussed. In the last section of this chapter, related work has been analyzed in terms of the contributions of the research to the field and limitations.

## 2.1 What is ASD?

ASD stands for autism spectrum disorders. Autism spectrum disorders are any developmental impairment or disability that impacts personal and social activities and can cause brain abnormality. There are many definitions for autism spectrum disorders (ASD). According to the Diagnostic and Statistical manual of mental disorders (DSM), ASD is the presence of markedly abnormal or impaired development in social interaction, communication skills, and can result in a restricted repertoire of activities (Psychiatric, 2013). According to the National Autistic Society (NAS), ASD is understood as a lifelong developmental disability that affects how a person communicates with, and relates to other people (Lambert M Surhone, 2011). ASD also impacts the way in which an individual makes sense of the world around them. Although all people with

ASD share certain difficulties, ASD is a spectrum disorder which means their condition will affect them in different ways (NAS). Although there are many definitions for autism, all of them point to the same idea which is that autism is a disability which impacts an individual's personal activities and the way in which they communicate with others.

Autism is a spectrum disorder because two autistic people do not have the same symptoms and they suffer with different types of disorders compared to each other. Hence, the treatment methods for autistic people are different and the treatment is based on their type of disorders.

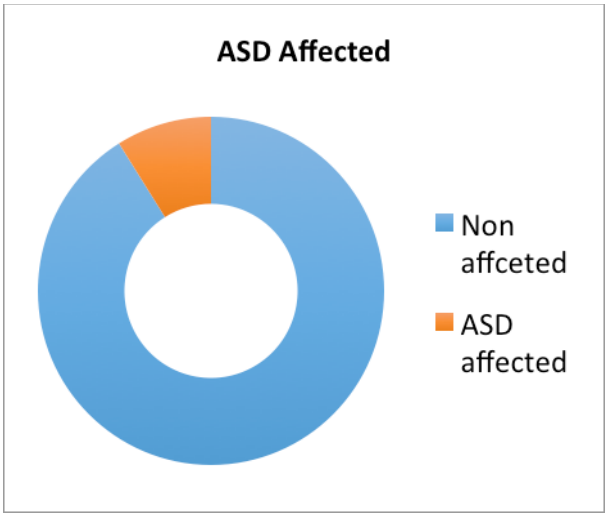


*Figure 1.The ASD Affected popluation is 14.7% based on Centers for Disease Control and prevention (CDC).*

### 2.1.1 Cause of ASD

The prevalence of autism spectrum disorders in children is increasing. Although there are many researchers who are trying to determine the exact cause of autism or find the striking treatment or medicine, the exact cause of autism is still unknown. Based on recent publications, the main involved factor of autism spectrum disorders that has key roles in normal development of brain, memory, motor activity, and regular behavior is neurotransmitters (Paromita Roy Choudhury, 2012). Neurotransmitters impact with brain development by affecting neuronal cell migration, differentiation, synaptogenesis and eventually brain developmental processes (Kwong, 2000) (Chungani, 2011). According to the recent neurotransmitters systems investigation in autism

spectrum disorders pathophysiology, the most commonly associated neurotransmitters with ASD are GABAergic, gultamatergic and serotonogeric systems (Fitzgerald, 2015).

GABA (Gamma Amino Butyric acid) is the main inhibitory neurotransmitter in the brain that helps to balance excitation and inhabitation in order to maintain and homeostasis the central nerves system of the brain (Hübner, 2013). GABA is synthesized from glutamate by the enzyme glutamic acid (Pinal, 1997). The enzyme glutamic acids consist of two informs known as GAD67 and GAD65 and these enzymes are different from each other in terms of enzymatic expression and activity (Buddhala C, 2009). The reduction in the expression of GAD67 and GAD65, which is associated with abnormality in ASD pathophysiology, is because of suppression of GABAergic inhabitation (Hussman, 2001).

Although, GABA has an important role in the regulation of early developmental stages of cell migration, GABA also helps the neuronal differentiation, stages of maturation, and formation of glutamergic system mediated excitatory processes that regulate inhibitory system (Ben-Ari, 2012) (Levitt, 2004). Researchers found that the platelet GABA level of children with autism spectrum disorders is lower than in normal people which is the reason for widespread dysfunction of the GABAergic system of autistic people (Rolf, 1993). Hence, the reduction of GABA in children with autism causes hyperactivity disorder and leads to cognitive dysfunction. Moreover, the prevalence of epilepsy in children with ASD was a good reason for investigating the GABA neurotransmitter system in individuals who have ASD (Han, 2012). Consequently, conducted studies in animal model cases and autistic people have confirmed the hypothesis of decreased GABAgeric transmission in autism spectrum disorders (Fitzgerald, 2015).

The essential excitatory neurotransmitter of the central nervous system is glutamate and it is synthesized from glutamine via glutaminase enzyme (Fitzgerald, 2015). Glutamate has a main role in shaping the architecture of the brain and glutamate transmission level accomplished the maturation and development stage of the brain, cell migration (Mattson, 2008). Moreover, cognitive processes such as memory and learning have a direct connection with glutamate (Manent JB, 2013).

Glutamate receptors associated with autism spectrum disorders are highly expressed in the cerebellum and hippocampus (Ozawa, 1998). Therefore, an ASD patient's glutamatergic system has been investigated to clear the hypothesis of the role of this system in autism spectrum disorders. The first proposed hypotheses are in hypoglutamatergic state (Carlsson, 1998) and the second assumed the depletion of GABAergic excitation / inhibition rate which eventually lead to the hyperglutamatergic state (Rubenstein, 2003). Another hypothesis that has been considered by researchers is hypoglutamatergic state and associated cortical tissue hyper excitability in specific cortical areas (Shinohe, 2006). The incidence of epilepsy for ASD patients is because of an increase in the glutamatergic activity (Fitzgerald, 2015). As a result, it has been confirmed by researchers that there is dysfunction in the glutamatergic system of autistic people and recent studies have been more focused on hyper-glutamatergic state. Researchers believe advanced knowledge in glutamatergic system agents will contribute to ASD pathogenesis in the future (Fitzgerald, 2015).

Serotonin is a neuromodulator that functions as a developmental signal and is synthesized by the enzyme triptophanhidroksilase (Celada, 2013). Serotonin neurotransmitter system has an important role in the regulation of crucial steps of neuronal development such as cell proliferation, migration, differentiation, apoptosis synaptogenesis, and glial and neuronal development (Whitaker-Azmitia, 2001). Additionally, serotonin has a key role in the social skills development in early childhood (Fitzgerald, 2015). The serotonin system in the prefrontal cortex and temporal cortex regulates GABAergic inhibition, thus the serotonin system is involved in many cognitive functions and activities (Yan, 2002). Finally, some studies indicate that children with autism spectrum disorders deal with stimulation deficiency of serotonin at early ages which could be a cause of abnormalities in serotonin metabolism in the future (Fitzgerald, 2015).

Dynamic changes of serotonin levels are impaired in ASD people. For instance, low levels of serotonin have been detected at early ages in ASD people however from the age of 2 to 15 years, serotonin was found to steadily increase until levels reached higher than adult levels (Chugani, 2002). The serotonergic system is the highest level of evidence for autism spectrum disorders relationship with monoamines. The hyposerotonergic and hyperserotonergic hypothesis in people with ASD is because of low levels of serotonin in the brains tissue (Fitzgerald, 2015).

Although there is a great deal of evidence that indicates that the cause of neurotransmitters impairment in people with ASD is genetic, it is important to note that it is not purely genetic. Studies confirmed that environmental factors could also be the cause of ASD. There are many candidate environmental factors for ASD as there are candidate genes for autism (Boucher, 2008). As with susceptibility of genes, different combinations of environmental factors may contribute to ASD (Boucher, 2008). There is less of a chance of a single environmental factor being the main cause of autism in an individual who does not have genetic susceptibility (Boucher, 2008). Hence environmental factors probably have a role as triggers in genetically susceptible individuals (Boucher, 2008).

### 2.1.2 Disorders

Autism spectrum disorders is a heterogeneous disorder. Autistic people have dissimilar disorders and they act different compared to each other. Developmental disorders appear in children when they start showing prevalent impairment in such developmental areas as social skills, communication skills, repetitive or stereotype behavior, and activities and interests (Heather, 2010). Families can notice these changes in their children when compared to other children of the same age. Although children with ASD syndrome can have some mental retardation, many children with mental disorders have normal or above average intelligence (Heather, 2010).

Impairments in social skills and competence in autistic people is one of the main disorders that keeps them from interacting with other people. Socials skills deficits play an important role in the diagnosis of autism spectrum disorders and can influence communication skills. People with autism spectrum disorders have poor emotion recognition and struggle to recognize and identify another person's emotion based on nonverbal cues (Vinood, 2014). Furthermore, facial cues affect autistic children's emotion and they respond differently when they reach four to six months (Poulin-Dubois, 1999). Social gaze and eye contact are also important precursors for typical social skills development. Unfortunately, individuals with ASD participate less in social gaze and they have poor eye contact with others (Vinood, 2014). Autistic children have no way to express their feelings as their facial expressions are either absent or inappropriate to the situation (Ghaziuddin, 1996). Body gestures are another weak point in ASD people towards the

social skills. Children with ASD produce significantly fewer body gestures compared to typically developing children (Attwood, 1988). On another hand, they understand simple body gestures but they have difficulty producing them (Vinood, 2014). Moreover, relationship development and the ability to socially and emotionally grow with others is also a significant challenge for people with ASD. There is also a lack of joint attention that is used to nonverbally communicate with others; social and emotional reciprocity as a social behavior that demonstrates a mutual exchange in interaction with others; imitations of actions of others (Vinood, 2014). Social skills impairment in individuals with autism spectrum disorders influences their ability to maintain relationships with others, live independently, education, and social reciprocity.

Another major disorder in people with autism spectrum disorders is impairment in communication skills. Deficit in language ability is a common problem in communication skills for ASD individuals. Although language difficulties are one of the significant disorders in ASD, language impairment alone does not define the autism (Vinood, 2014). Language difficulties can be demonstrated in the form of delayed language, heterogeneity of language, and repetitive language for different cases of ASD individuals. Several of the diagnostic criteria for ASD symptoms are related to language disabilities including impairment related to conversation and stereotyped, repetitive, and idiosyncratic language (Vinood, 2014). Children with autism spectrum disorders progress from having essentially no expressive and receptive language skills to displaying near normal to above average spoken language skills by age of nine, although some ASD individuals related to how language is used (MacDonald, 2007).

An autistic child's language development is always delayed. There are three common language disabilities in children with ASD; a child with no spoken language and no reciprocity gesture or sign of language, a child with some language but inability to communicate, and a child with language skills but use of language is often bizarre (Vinood, 2014). Some communicative functions are more affected in people with ASD compared to others. For instance, referential deficits is a distinctive communication profile in which shared referencing is more impaired than functions such as requesting (Paparella, 2011). Another language impairment in autism, which is one of the most salient pragmatic difficulties, is echolalia (Tager-Flusberg, 2005). Echolalia is when a child repeats the same words that another person has said. Furthermore, expressive and

receptive language gaps in autistic children may begin as verbal repetition without understanding the words. Sometimes atypical vocalization and odd voice quality or intonation patterns occurs in early ages of ASD children (Vinood, 2014).

Furthermore, certain typical characteristics of word meaning and syntax can be noticed in a significant proportion of ASD individuals' verbal skills (Vinood, 2014). Odd meaning is also another language disability for autistic people in terms of using words with irrelevant meaning to the sentence and difficulty for understanding other's meaning. When the sentence syntax of autistic children is compared with that of other children of the same age, their grammar does not look distinctive (Vinood, 2014). According to studies by Boucher (2001), certain aspects of syntax such as morphology might be more affected in ASD individuals than other word order. Some children with autism spectrum disorders may have problems in extending new words, which might reflect the difficulty in understanding social context about the word extension (McGregor, 2012).

Autistic people face challenges in their physical life and certain health behaviors such as physical activity bring difficulties into their quality of life in several aspects. Physical activity is a complex behavior in ASD in terms of intensity, duration type of activity, activity setting, and reasons for action. Participating in sports and physical education programs is important for young people. Individuals with ASD are more at risk of physical inactivity because of their difficulties in social, behavioral, and motor function (Pan, 2009). Additionally, physical inactivity can be caused by ASD characteristics such as difficulties in making friends, disabilities in making conversation with others, understanding social cues, and displaying stereotypic and self stimulatory behaviors (Vinood, 2014). Autistic people can benefit from physical activity in terms of its positive effect on ASD characteristics such as social cognition, social awareness, social response, and peer relationships which all help to decrease communication deficits.

Autism spectrum disorders is accompanied by complicated disorder conditions that present various forms of challenging behavior such as disruptive behavior, aggression and property destruction (Vinood, 2014). Some individuals with ASD might deal with behavioral and emotional problems including mood swings, tantrums, and self-aggression (Estes, 2009). Hence,

difficulties in behaviors and emotions in ASD children such as aggression can impact significantly on their quality of life because challenging behavior results in social exclusion such as avoiding getting involved in activities and the community. Furthermore, this can limit the person's access to learning opportunities, social participation, and new experiences within environments (Vinood, 2014).

As a result, a variety of characteristics of disorders in autistic children affect their quality of life in terms of social impairments, communication difficulties and challenging behavior they show to others. Characteristics of disorders manifest in different ways for each autism spectrum disorder individual. Additionally, autistic people should be evaluating based on the distinctive difficulties they have with social skills, communication skills, and common behavior to enhance the treatment in each ASD individual. Therefore, the treatment process and techniques are different for each child with ASD.

### 2.1.3 Diagnosis & Assessment

Many people think the term "diagnosis" and "assessment" for ASD are the same because they usually occur together. However, these terms refer to different parts of the process when a professional deals with autistic children at the clinic for the first time (Dawson, 1989). The process of determination of autism spectrum disorders in children is the diagnostic part of the ASD when families bring a child with a problem to a clinic (Dawson, 1989). The diagnosis process identifies whether an individual is appropriately classified as autistic and emphasizes characteristics that autistic people have in common (Eric Schopler, 2013). Assessment includes defining the person's characteristics, traits, individual strengths, weakness and needs from a different perspective that is essential for individualized education and management (Eric Schopler, 2013). The assessment process emphasizes unique characteristics of the individual and indicates the difference between them in order to develop an appropriate individualized program and evaluating its effectiveness in the future (Eric Schopler, 2013) (Dawson, 1989). Additionally, assessment is helpful for research consistency and generalizability which contribute to advancements in treatment and education of people with autism spectrum disorders (Eric Schopler, 2013). Assessment can occur during the initial evaluation of the child.

The importance of early diagnosis for people with ASD has a key role in implementing a better treatment process which enhances the effectiveness of the treatment. Therefore, many researchers have contributed to finding the early syndromes in autistic children for past decades. Such authors included De Sanctis (1906, 1908), Earl (1934), Asperger (1944), Heller (1945), Kenner (1943) and Mahler (1953). Their contribution was related to syndromes in abnormalities and innate disorders of personality in autistic children (Gillberg, 2013). Kenner's syndrome "early infantile autism" became the most widely known and accepted among the other presented syndromes by different researchers for early diagnosis of ASD. Kenner (1943, 1994) selected the following as characteristics and diagnostic: profound lack of affective contact with other people; an anxiously obsessive desire for the preservation of sameness in the child's repetitive activity pattern; a fascination for objects, which are handled with skill in fine motor motivation; mutism, or the kind of language that does not seem to be intended to serve inter-personal communication; and good cognitive potential manifested by feats of memory or skill in performance tasks (Gillberg, 2013). Kenner stated that abnormalities were present from birth or began within the first 30 months of an autistic child's infantile stage (Gillberg, 2013). Asperger's syndrome based on his studies, pointed to repetitive speech; a limited range of circumscribed interest pursued exclusion of other activities; a conspicuous lack of common sense and poor coordination of movement (Gillberg, 2013). Asperger's syndrome was less precise compared to Kenner's syndrome in terms of early age disabilities for children with ASD.

The problem with Kenner's syndrome was the diagnosis can identify the condition for children at age 5 or up but he did not specify clearly enough the sufficient signs and symptoms of autism at early ages and how they were to be defined and measured (Eric Schopler, 2013). Therefore, researchers tried to find another diagnosis technique to identify the ASD which is more precise and ASD can be identified earlier than age 3. Early diagnosis is important because of the benefits that it can have on autism spectrum disorders treatment. Another issue that health specialists complain about is the difficulty to distinguish the autism from other related problems in early ages (Eric Schopler, 2013).

Diagnosis of autism spectrum disorders was based on clinician observation and intuition until the past few years when it began to cause many difficulties for health specialists, including identifying the ASD in autistic children. In recent years many diagnostic instruments have been introduced by different researchers to evaluate the ASD in children based on the score on a standardized instrument. The gold standard diagnosis instruments for clinical purposes and research are the Autism Diagnostic Interview-Revised (ADI-R; Le Couteur, Lord, & Rutter, 2003; Lord, Rutter, & Le Couteur, 1994) and the Autism Diagnostic Observation Schedule (ADOS; Lord et al., 2000; Lord, Rutter, DiLavore, & Risi, 1999). The ability to diagnose children with autism with good reliability has significantly increased as a result of these diagnosis instruments (Elizabeth McMahon Griffith, 2009).

The ADI-R diagnosis instrument focuses on the individual's communication skills, socials skills and repetitive behaviors appropriate to the child between 18 months of age and adulthood. The ADI-R classification for autism spectrum disorders is based on the report of symptoms during the preschool years because diagnostic symptoms change during the lifespan (Elizabeth McMahon Griffith, 2009). ASD symptoms tend to be the most severe and differentiated from other developmental disorders at early ages. ADI-R is a lengthy interview and the shortest version of this interview is an hour and a half. Although many participants find it difficult to involve the ADI-R in their daily practice, it is included in an ASD assessment in a specialized ASD clinical setting (Elizabeth McMahon Griffith, 2009).

Health specialists consider the ADOS as a snapshot of current social and behavioral skills of the autistic child while the ADI-R provides the long view of individuals. ADOS provides a semi-structured play session environment to assess the social skills, communication skills and the restricted or repetitive behavior (Elizabeth McMahon Griffith, 2009). The ADOS interview takes 30 to 40 minutes approximately, which is appropriate for children from age 2 to adulthood. Based on an individual's module level of ASD and language skills of autistic children, there are four ADOS modules (Elizabeth McMahon Griffith, 2009). Module one is appropriate for nonverbal autistic children, module two is for those individuals that use phrase speech, module three is for children and adolescents with fluent language and module four is appropriate for adults with fluent language (Elizabeth McMahon Griffith, 2009).

### 2.1.4 Treatment

Although autism spectrum disorder is a life long disorder, significant improvements in amelioration of ASD symptoms and categorical assessments of independence have been achieved by researchers and health professionals (Vinood, 2014). According to recent studies treatment improvement in core autism symptoms, especially social reciprocity has been reported lately (Levy, 2011). Developmental language and social communication impairments have an important role in life quality of ASD people, thus most educational and treatment programs focus on them (Dawson, 1989).

The treatment for autism typically includes special education services, behavioral programs, complementary and alternative medicine (CAM), dietary, sensory integration therapy, speech language therapy, occupational therapy and several other therapies such as art, music and animal therapy (Strickland, 2009). According to a survey by the Disease Control Prevention (CDC) in 2007, approximately 12 % of autistic people tried complementary and alternative medicine (CAM) which shows that families with autistic children are not aware of the benefits of available treatment and intervention for their children with ASD. The complementary and alternative medicine (CAM) offers different intervention in therapies for individuals with ASD such as acupuncture therapy, yoga, massage therapy, music therapy, herbal medicine, and hypnosis (Wong, 2006). Based on many studies, therapies have a significant role in ASD intervention and improving the quality of life of autistic people. Some therapies include physical activity which can be effective in multiple ways for different types of impairment in autism spectrum disorders such as improving physical and aerobic fitness, muscle strength, and gross motor functioning. Physical exercise can also contribute to a reduction of most commonly reported problems in autistic people such as stereotype and self-stimulatory behaviors (Lang, 2010). Moreover, decreasing the aggression and self-injury and increasing the academic achievement and exercise behavior are the other beneficial effects of physical activity (Lang, 2010).

According to recent studies, diets of people with autism spectrum disorders such as foods, nutrients, vitamins, minerals, antioxidants, herbs, essential fatty acids and pharmaceuticals all

have a key role in maintaining health and help them for amelioration of disorder characteristics (Strickland, 2009). Additionally, diet contributes to sleeping difficulties, behaviors, food allergies, and obesity in children with ASD.

The interventions that address all three core difficulties in autism spectrum disorders are comprehensive educational interventions. On another hand, comprehensive educational interventions improve social skills and social interests, they contribute to communication skills, and reduce ritualistic and stereotype behaviors (Vinood, 2014). Comprehensive educational interventions consist of therapies and educational programs for ASD children that mainly focus on social and communication deficits. There are well known interventions such as: 1) Treatment and Education of Autistic and Related Communication Handicapped Children (TEACCH), (2) Sensory Integration Therapy, (3) Floor-Time, (4) Early and Intensive Behavioral Intervention (EIBI), (5) Pivotal Response Training, (6) Relationship Development Intervention Program (RDI), and (7) the Early Start Denver Model (ESDM). In what follows, each of these interventions will be briefly described, starting with TEACCH (Vinood, 2014). Based on clinical studies and research on comprehensive interventions, Early and Intensive Behavioral Intervention (EIBI) has most significant increases on standard scores in both intellectual functioning and adaptive functioning. Several studies demonstrated that EIBI is the best documented intervention for ASD and reduces the autistic symptoms and abnormal behaviors (Vinood, 2014).

There are many treatment techniques and interventions based on studies in past decades for ASD which can be helpful towards improving the social impairments, communication difficulties and repetitive or stereotype behavior in autistic children. Therefore, families of autistic children have to meet a health specialist or therapist before participating in any interventions for their autistic child in order to implement the most effective treatment based on the child's disorder symptoms and difficulties.

## 2.2 Ontology

Ontology goes back to the forefront of philosophy, science, and technology after a long period of decline (Roberto Poli, 2010). In philosophical discipline, ontology has been a focus area of study for a long time (Raj Sharman, 2007). Ontology comes in two main forms these days: the philosophical understanding of ontology and the computer science based understanding of ontology (Roberto Poli, 2010). The term of ontology in philosophy refers to the question "what kinds of things exist?", while ontology in computer science deals with the question "what kinds of things should we capture and represent?" Research on these questions together yields a broad framework for the analysis of a discourse universe and represents the development of organizations and systems within the universe (Raj Sharman, 2007). The perspective of philosophical framework of ontology refers to the essential properties and relations of all beings in the universe however this idea has been expanded and specialized in the field of computer science and artificial intelligence (Raj Sharman, 2007). According to Marek Obitko 2007 studies for general ontology definition, ontology is a branch of philosophy that deals with the nature and the organization of reality and refers to questions such as "What characterizes being?" and "Eventually, what is being?". Additionally, ontology can be applied independently of the state of the world in practice, as the theory of distinctions. In particular, interested distinctions are among the entities of the world such as physical objects, events, regions, and distinctions among the meta level categories used mode to the world such as concept, property, quality, and role (Obtiko, 2007).

The concept of computer science ontology was taken from the philosophy of science which is a branch of philosophy that looks for the reason and justification of science (Mosterin, 2000). Basically, philosophy ontologies point to questions more focused on general nature and has less in common with ontology in knowledge engineering (Roberto Poli, 2010). The different requirements in various fields such as artificial intelligence, software engineering and database communities made the path followed by ontology concept from philosophy to computer science (Raj Sharman, 2007). In general, ontology in computer science is a way of representing a common understanding of a domain. For instance, although the World Wide Web is used to share the world's knowledge to everyone, all the participants must share a common vocabulary

in terms of an agreement about the meaning of the thing. Thus, ontology is one of the solutions for representing the common understanding.

In 1993, Gruber described the ontology for computer science as an "explicit specification of a conceptualization" (Gruber, 1993). Then in 1997, Borst defined ontology as a "formal specification of a shared conceptualization" (Borst, 1997). This definition emphasized the conceptualization that should be shared between different parties and the conceptualization should be defined as machine-readable format. By merging these two definitions in 1998, Studer et al. put forth a complete form of the definition of ontology in computer science as "ontology is a formal, explicit specification of a shared conceptualization" (Studer, 1998). Hence, conceptualization, specification of conceptualizations, and shared ontological commitments are the three core components of the computational ontologies which are very board categories for any field of application (Guarino, 2009). There are three questions such as "What is conceptualization?", "What is proper formal explicit specification?" and "Why the shared is importance?" which are based on Studer et al. definition for ontology that need to be clear for better understanding the concept of ontology (Guarino, 2009).

Genesereth and Nilsson present a notion of conceptualization. According to their studies, "A body of formally represented knowledge is based on a conceptualization: the objects, concepts, and other entities that are assumed to exist in some area of interest and the relationships that are held among them. A conceptualization is an abstract, simplified view of the world that we wish to represent for some purpose. Every knowledge base, knowledge-based system, or knowledge-level agent is committed to some conceptualization, explicitly or implicitly" (Guarino, 2009).

As it is mentioned in the definition of ontology "explicit specification of conceptualization", conceptualization can be explicitly specifying in two ways: extensionally and intestinally (Guarino, 2009). The extensional method to explicit specification of conceptualization required listing the extensions of conceptual relations in stereotypical world states and correspondence of selected concepts. The intentional way to explicit specification of conceptualization can be given in formal and informal languages but in order to have machine-readable ontology, it must be in formal language, hence natural language is excluded (Guarino, 2009). The definition of ontology

has been defined in other words by Guarino, 2009 as "ontology is just a set of such axioms, i.e., a logical theory designed in order to capture the intended models corresponding to a certain conceptualization and to exclude the unintended ones."

The nature of ontology is to share a set of related concepts for a specific knowledge domain. Ontology can be shared with stakeholders or machine to be reused or extended in the future. One thing that should be considered in ontology that support large scale interoperability is to be well founded in terms of well chosen primitive concepts for better and easier understanding by users (Guarino, 2009). There are many aspects of ontologies that can be settled in different classifications based on the way they are used. For instance, Reference ontology indicates the ontology's structures that are derived from them, to provide a broad view for many different target domain top level ontologies that are suitable (Guarino, 2009). Biomedical ontologies are used to represent biomedical terminology in a common vocabulary and Lightweight Ontologies indicate the backbone taxonomies for any purposed knowledge domain. Therefore, ontology plays an important role in managing the huge heterogeneous knowledge as well as to share them with humans and machines. Ontology is one of the strategies for developing Semantic Web and provides a simpler and transparent codification for knowledge of the objects of the world (Roberto Poli, 2010).

Ontology contributes to different knowledge domain and several field of study. Different fields of computer science such as software engineering, database systems, and artificial intelligence use the ontology to model their knowledge and construct solutions to satisfy their needs (Raj Sharman, 2007). Bouattour et al. (2005) represent the ontology in the architectural designing field to indicate the state of architectural design components by following the user's process decision. Ontology has been used in urban planning by Kaza and Hopkins (2007) to show the different alternatives of decision making in a plan. The healthcare and medical field takes advantage of ontology in different ways such as through the use of medical databases and disease classification (Dieng-Kuntz, 2006). Ontology of gene is one of the popular and well known ontology in the biology field and has been for the past few years (Ashburner, 2000).

Modeling knowledge is the focus area of methodology nowadays compared to developing applications (Rajiv Kishore, 2007). Therefore, using such methodologies are good alternatives for improving and modeling knowledge instead of alternatives for managing an information technology project centered on ontologies (Rajiv Kishore, 2007). Different researchers have developed popular methodologies, such as the following, for creating ontology; Cyc methodology by Lenat and Guha (1990) is based on the experience during the development of the Cyc knowledge base, which contains a good quantity of common sense knowledge. This process contains three basic tasks. The first task is manual extraction of common sense knowledge; the second task is knowledge coding through the use of tools based on stored knowledge in the Cyc knowledge base; and the third task is computer managed to extract the common sense knowledge among the available data. CycL was the language used to implement Cyc and they specified the ontology based on the following two activities:

- Development of knowledge presentation and creating top level ontology with the main concepts.
- Presenting the knowledge for different domains.

There is a variety of ways to apply and use ontology. The following applications are examples for using the ontology in several topics: ONTOLOGER is an application web search optimizing to user profiles (Stojanovic, González and Stojanovic, 2003). OntoWeb uses ontology to exchange information in fields like e-commerce or knowledge management (Oberle and Spyns, 2004). FALAME 2008 is a platform based on ontology to model a system services provider for mobile devices (Weißenberg, Voisard and Gartmann, 2004). OntoVote provides an environment for developing P2P ontologies, and Ontology can be created by the member of P2P application (Ge et al., 2003). EDUTELLA is based on ontology for the P2P network environment for the exchange of educational resources in German universities (Nejdl et al., 2001).

To build ontology like software development, methodology has a key role for obtaining a good result by following a set of step based on practice. There are many methodologies for creating ontology based on the experience of professionals involved in their construction and the way in which a particular ontology was built (Raj Sharman, 2007). Moreover, there are suitable

methodologies represented by different researchers for modeling knowledge in ontology and those most significant methodologies will be covered in chapter three.

### 2.2.1 Ontology in Medical Field

The medical field is a huge and complex domain that consists of different perspectives of modeling and representing intended meaning such as different activity domain (clinical vs. administrative), different user requirements and queries for the same service (physician vs. patient view), different specific granularities (organic detail vs. molecular) and complex terminology (polysemy) (Pisanelli, 2004). Moreover, there are various medical knowledge modalities in terms of tacit knowledge to experimental knowledge and explicit knowledge to data induced knowledge. Therefore, different knowledge modalities within a medical context can be identified as: (a) the problem solving skills, judgment and intuition knowledge of practitioners; (b) consolation and collaborative problem solving issues; (c) recorded and observed clinical experiences; (d) operational knowledge in terms of clinical pathway and protocols; (e) medical literature and medical practice guidelines; (f) medical education contest for patient education; (g) medical education contest for practitioners; (h) extracted knowledge based on data of clinical observations, diagnostic and therapeutic treatments, recorded in medical record and stored in clinical data; and (i) knowledge decision support obtained from domain experts and data decision models (Abidi, 2005). Hence, there is a vast amount of heterogonous knowledge available in medical information systems that can take advantage of using the framework of ontology for providing better services and managing information to facilitate the access to information, clinical data, clinical pathways, and support clinical decision making. Additionally, ontology contributes to medical knowledge management by providing ways to support the creation, sharing and operationalization of medical data. The main beneficial point of using ontology in the medical domain is to represent and recognize the medical terminologies (Liana Stanescu, 2012). Although, building ontology based on medical structure to represent the medical terminology is a difficult task in terms of analysis of medical concepts and available medical terms in the field, but using ontology for medical purposes has many benefits such as (Liana Stanescu, 2012):

- The most significant advantage of using ontology in the healthcare system is to support the integration knowledge and data which is very important.
- Ontologies facilitate the process of transmitting, sharing and reusing the electronic medical records (EMR) to provide better and easier access to them
- They can be used for building a more precise and better interoperable information system in healthcare.
- Ontologies in the medical field can be used for computing different statistical combinations.
- They can be used for better diagnosis decisions made by professionals and doctors in the medical field.

The following examples are the projects that indicate beneficial perspectives of using ontology to solve the challenging points and problems in the medical domain. Generalized Architecture for Language, Encyclopedias, and Nomenclatures in medicine (GALEN) is clinical terminology ontology that facilitates the clinical services in different orders. GALEN replaces the traditional clinical terminologies with description logic to allow clinical data to be represented, captured, manipulated and displayed in better ways. It also provides an environment for the user to reprocess the information and be able to integrate medical records, decision support, and other clinical purposes (Rector, 2003). Gene ontology (GO) is the most developed and widely used ontology. Its great utility to demonstrate annotation and biological interpretations of gene category obtained by high throughput of experiments. Gene ontology provides the annotation of gene sets according to their genetic scale and biological characteristics. It represents biological concepts with different specific levels from general to precise concepts through the GO structure (Martucci, 2004). Foundational Model of Anatomy ontology (FMA) is another example of medical ontology. FMA is used to represent a coherent body of explicit knowledge about the human anatomy and structure of the human body in a form that is easy to be understood and also can be used and navigated by a machine based system (Rosse, 2003). Context based task ontology (CTOs) for clinical guidelines purposes. CTO provides a methodology by means of which task recommendations contain within guidelines can be integrated into the clinical practices of health care sets. CTOs also assist practitioner and patient decision about appropriate

health care for specific clinical circumstances. Clinical care can be improved by better clinical decision making with the contribution of CTOs (Kumar, 2004).

The medical domain is one of the few domains that have extensive operational knowledge and domain knowledge defined through the ontology. Operational knowledge ontologies define the knowledge in terms of a process of objects and terms. In the medical domain, operational ontologies are popular and useful in order to provide better clinical services for patients and easier health care information access for professionals in the field. The examples of operational knowledge ontologies in healthcare are such as patient ontology, clinical pathway ontology, service functionality ontology and service message ontology.

Patient ontology provides the patient information collection, management, and represents the patient's disease and characteristics based on recorded medical information. Computer based patient record (CPR) is a well known patient ontology based on electronic patient records used to support users by providing accessibility to accurate patient's data, reminders, clinical decision support, and links to medical sources. It is also based on direct observation of the patient (Murray, 2002). Clinical pathway ontology describes the process of care of certain medical conditions to enhance the quality of care and decrease the variants of practice (Hurley, 2007). Health care institutions can be evaluated by way of clinical path ontology in terms of availability of medications, procedures, and diagnostic interventions (Hurley, 2007).

Service functionality ontology provides an exchange information environment in an interoperable manner in healthcare institutes by classifying the web services. HL7 classified the service functionality events in the healthcare domain, which reflects the business logic domain. Moreover, using the HL7 in medical institutes has another advantage in that it provides a standards platform to exchange the data over the healthcare computer systems (Dolin, 2001). Service message ontology provides a platform for the users in the healthcare environment to communicate based on the standards meaningful components. It can enhance the accurate communication among users such as doctors, nurses and other professionals in the health care system.

Domain knowledge ontology represents and models the concepts in the specific knowledge domain. Examples of domain knowledge ontology in the medical field are disease and genomic. Pathology ontology describes the phenotypes of disease and expresses the occurrence of disease as individual entities and displays the variability in the constituent entities within a syndrome (Albert Burger, 2007). Additionally, the description of pathology is one of the main features of any disease ontology. Mouse pathology ontology is an instance for disease ontology. Mouse pathology ontology contains full definitions and major classes of lesions generated because of genetic or external damages (Albert Burger, 2007). Genomic ontologies are one of the largest and fastest growing areas in the medical field. For instance, gene ontology (GO) represents genes and their properties and characteristics. Gene ontology is one of the medical ontology topics that have had significant improvements and many groups of researchers have been working on it lately.

Many departments and medical systems in the healthcare field are taking advantage of using the medical ontologies for different purposes these days. In general, medical ontology presents many beneficial features to the medical field as it has been mentioned above and it allows communication between various processing applications in the medical environment (Liana Stanescu, 2012). Moreover, ontologies are fixable in terms of reusing, maintenance, and can be extended based on new achievements and discovery in the medical domain in the future.

## 2.3 Related Work

The contribution of ontology in the medical domain is growing these days. Disease ontology is one of those areas investigated by many researchers. For instance, infection disease ontologies, anatomical disease ontologies, genetic diseases ontologies, cellular proliferation ontologies (cancer ontology), physical disorders ontologies, and mental health disease ontology are all part of the disease ontology family. Autism spectrum disorders is a mental health disease ontology that can be integrated with physical disorders in several individuals with ASD. Although a large amount of work and research has been done for autism spectrum disorders in terms of genetic causes, disorders syndrome, and diagnosis and treatment, the contribution of research on autism spectrum disorders ontology has been less than other ASD aspects in this field. In the previous

section, examples and related work for several fields of medical ontology aimed at different aspects of the healthcare system has been mentioned. The structure and contribution of related research work, which focuses on building autism spectrum disorders ontology is presented in this section. Furthermore, the problems and limitations of these works are discussed as part of this section.

### 2.3.1 Related Work by Lynn young et al.

Lynn young et al. work for building an autism spectrum disorders ontology was based on the phenotypes extracted from literature and instruments of ASD. National Database of Autism Research (NDAR) and UCSD data integration environment were involved in their work for accessing autism studies, information from other labs and the query system to display the requested data to the users. NDAR is an autism database for researchers that provides an environment to facilitate the data access from various areas of autism research. NDAR was built on the original Biomedical Informatics Research Network (BIRN). NDAR allows the researcher and user to submit their achieved data for autism spectrum disorders to be shared with users or integrated with other studies. Integration of data in NDAR is supported by direct submission of information from the researchers or institutes. UCSD is a data integration environment that provides users interfaces for researchers and support collaborative research.

The ontology used NDAR to describe the ASD as a query tool and the high level ontology was based on the phenotypic concepts, which are represented in the research and clinical data abstractions. Phenotypic concepts are the classes and sub classes of the ontology, which have been extracted from literature and autism instruments. UCSD data integration environment provides the user interface for their work and NDAR allows researchers to define the phenotype as elements of the data dictionary for the created ontology. Researchers can submit a query into NDAR to retrieve the phenotypes, then, access the relevant list of subjects. In another word, retrieved data from NDAR by users will be mapped into an ontology. Furthermore, it provides a standard representation of autism phenotypes to benefit the researchers in this field.

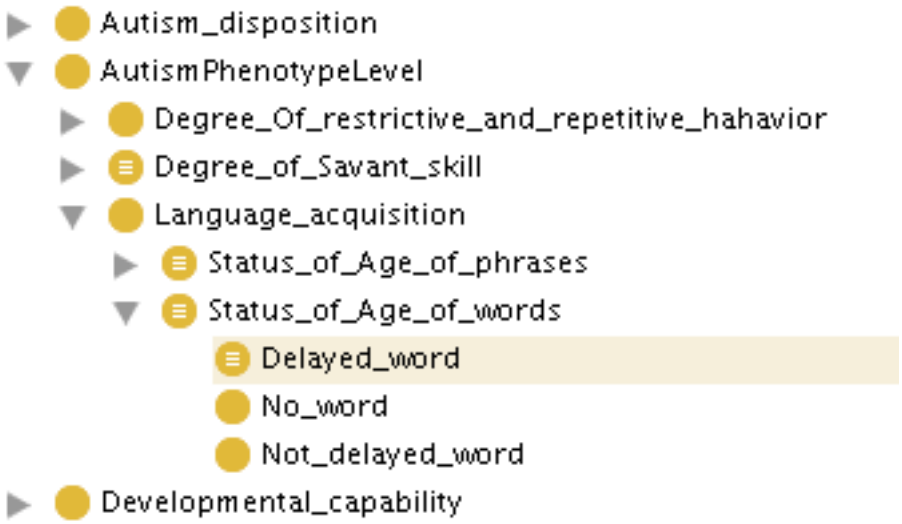


*Figure 2. Part of ontology by Lynn young et al. with delayed words phenotype level.*

According to Lynn's work, the created ontology and query system are mainly for research purposes. The system provides a data query environment for researchers to access the available autism information in NDAR based on concepts of the ontology. Hence, it is not helpful for normal users such as a family with an autistic child or clinicians. The system does not provide precise information for users such as therapists or doctors to assist them with decision making in treatment or diagnosis stage. Alternatively, this approach only retrieves the imported ASD information by different researchers from NDAR database that was included in the ontology. The main goal of this project was to facilitate the access to available autism research and clinical data in NDAR for researchers. Therefore, the proposed ontology was built for research proposes and the classes and subclasses of the ontology were defined from literature abstracts and keywords. It is not an accurate or comprehensive ontology compared to recent versions in terms of classes of disorders and indicting the disorders domain knowledge for ASD.

## 2.3.2 Related Work by Christopher S.G Khoo et al.

Christopher S.G Kho et al. (2011) developed disease treatment ontology to represent the treatment information that can be found in medical articles. The initial version of the ontology created was based on the analysis of 40 medical abstracts and the evaluation of an additional 10 abstracts on colon therapy. The medical abstracts were taken from Medline database. The

purpose of building the disease treatment ontology was to encode the extracted treatment information in the created ontology for knowledge discovery, question answering and information retrieval. The represented information by ontology can support the range and quality of clinical data for doctors to make informed treatment decisions. The top level ontology of disease treatment ontology contains five classes; disease, treatment, condition, effect and evidence. Unified Medical Language System (UMLS) semantic network, the Medical Subject Headings (MeSH), and the NCI (National Cancer Institute) thesaurus are adopted into their ontology design as the base medical ontology and the relations between classes are improved to link the treatment with diseases.

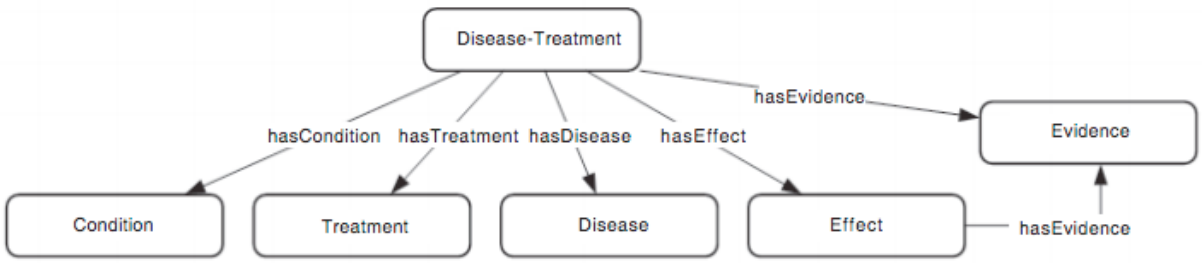


*Figure 3. Top level ontology structure of Disease-Treatment ontology with 5 main classes.*

The ontology was based on existing medical taxonomies and ontologies. Each class of the top level ontology contains other subclasses and related properties. For instance, the treatment class has 3 subclasses such as *therapeutics* and *surgical procedure*, *Drug_food_Chemicals* and *other class hierarchy*. These subclasses have been extracted from Mesh and NCI Thesaurus. Although Disease-Treatment ontology was made based on the abstracts of colon cancer medical articles, the generalized concept of the ontology has made the initial version of the ontology applicable to different medical domains.

The ontology can be considered as a general one with basic structure for medical domain and the designed system can be modified based on the target knowledge domain and variety of concepts in the field. Although the subclasses and properties of each top level class have been defined, the specific concepts and instances of colon cancer have not been imported into the ontology.

## 2.3.3 Related Work by Omri Mozach et al.

Omri Mozach et al. (2013) worked on the exciting autism spectrum disorders ontology by Stanford University. The ontology represents knowledge of the autism phenotypes and autism assessment tools. According to their work, the Stanford University ontology cannot be used for diagnosis of autism based on patient data because it does not include the complex phenotypes of autism as it is defined in the DSM-IV. Hence, they tried to enrich the ontology by using tools such as Diagnostic and Statistical Manual of Mental Disorders (DSM-IV) and Diagnostic Interview-Revised (ADI-R). The ADI-R instrument and DSM-IV have been used as additional knowledge sources of diagnosis of autism in order to support the semantic extraction of autism information from electronic health records and abstract phenotypes related to autism by enriching the ontology.

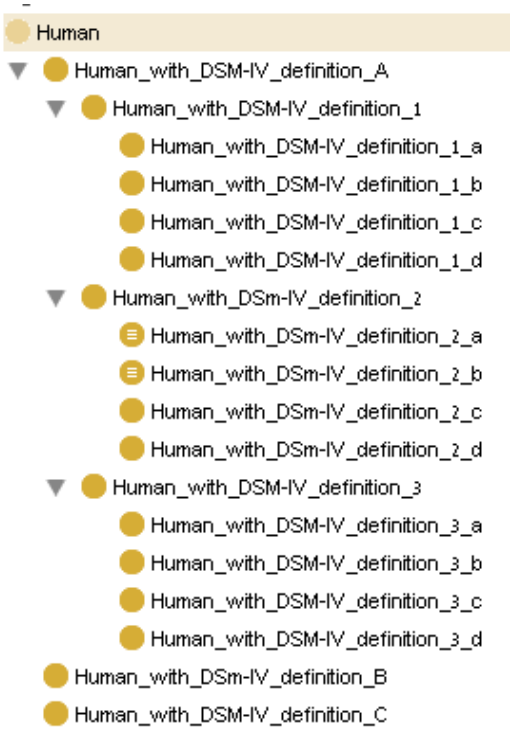


*Figure 4. Hierarchical relations structure of DSM-IV.*

The ontology extended based on DSM-IV to represent all the DSM-IV autism diagnostic criteria and concepts. The modeling knowledge tool for extending the ontology was a protégé and it has

been operated on the OWL format. The limitation of their work was that they used only one ASD instrument (ADI-R) as converted dataset to classes of ontology.

Moreover, the entire system was designed based on ADI-R which is focusing on the disorders in autism spectrum disorders. Alternatively, this ontology does not cover the treatment side of ASD or the effect of disorders and treatment on autistic people and relations between the classes. The main purpose of this extended ontology was to access the related data of ASD among the health record of patients based on the described disorders in DSM-IV.

### 2.3.4 Related Work by Alexa T. Mc Cary et al.

According to the work by Alexa T. Mc Cary and her colleagues, the created phenotype ontology is based on 24 instruments, which are the most significant instruments for autism spectrum disorders in the form of questioners, interviews, and direct assessment. The goal of their work was to build an ontology for providing improved access to data collection of autism spectrum disorders and support the user for assessing the instrument of ASD and compare them for better assessment of disorders in autism. Different screening tools and diagnosis instruments have been gathered for phenotype data collection to define the concepts and terms of the ontology.

The assessment instruments of autism analyzed the nature of the questions asked, the delivery method of the instruments, and the overall scope of the content. The instrument formats used for data collection were: (1) questioners that were completed by families of autistic children (e.g., CBCL); (2) interviews from children with ASD that have been done by professionals (e.g., BPASS); and (3) direct assessments (e.g., ADOS).

Therefore, they have done a comprehensive study for evaluating the selected instrument of ASD to extract the concepts and useful terms of ASD. The collected concepts and terms from different instruments merged together to generate clusters of concepts of instruments. The process involved manual refinement for adding initial clusters, merging the clusters from various instruments, and splitting the clusters based on shared terms. A variety of matrices were applied to develop this merging ontology. For instance, they analyzed the weaknesses of existing

National Center for Biomedical Ontology (NCBO 2013) and additional matrices to analyze the information contact for generating the ontology. The extracted concepts from instruments mapped to the ontology to represent the classes and they used protégé ontology editor as the ontology development environment.

| Instrument | Questions mapped to A | Concepts covered in A | Questions mapped to B | Concepts covered in B | Questions mapped to C | Concepts covered in C |
|---|---|---|---|---|---|---|
| ADI-R | 62 | 25 | 39 | 17 | 10 | 4 |
| ADOS | 31 | 17 | 41 | 15 | 8 | 2 |
| BRIEF | 118 | 38 | 19 | 8 | 1 | 1 |
| BPASS | 10 | 9 | 7 | 6 | 0 | 0 |
| CBCL | 95 | 36 | 113 | 32 | 35 | 20 |
| CCC-2 | 50 | 16 | 34 | 23 | 3 | 2 |
| CELF | 48 | 18 | 48 | 15 | 2 | 1 |
| CTOPP | 13 | 6 | 0 | 0 | 0 | 0 |
| DASII | 11 | 8 | 0 | 0 | 0 | 0 |
| Dean Hand. | 1 | 1 | 0 | 0 | 0 | 0 |
| DKEFS | 299 | 11 | 32 | 1 | 0 | 0 |
| Leiter-R | 23 | 13 | 1 | 1 | 3 | 2 |
| MH | 22 | 3 | 11 | 1 | 1,324 | 93 |
| Mullen | 158 | 22 | 8 | 3 | 0 | 0 |
| PPVT-4 | 8 | 1 | 0 | 0 | 0 | 0 |
| PedsQL | 10 | 7 | 11 | 10 | 6 | 6 |
| RBS-R | 56 | 14 | 0 | 0 | 1 | 1 |
| Rey-O | 22 | 1 | 0 | 0 | 0 | 0 |
| SRS | 30 | 23 | 53 | 29 | 4 | 3 |
| SCL-90 | 49 | 14 | 10 | 6 | 43 | 15 |
| SCQ | 24 | 14 | 16 | 13 | 1 | 1 |
| VABS-II | 216 | 46 | 227 | 52 | 7 | 7 |
| WASI | 18 | 6 | 0 | 0 | 0 | 0 |
| WPPSI-III | 23 | 5 | 0 | 0 | 0 | 0 |

*Table 1. Distribution of concepts in different contributed instruments of ASD (McCray, 2014).*

The created ontology consists of 283 concepts in 3 high level classes of Personal Traits, Social Competence, and Medical History. The Medical History class carries the largest number of concepts and the smallest number of concepts is within the Social Competence class. The ontology was designed for research purposes to allow researchers to pose ontology based questions, provide intelligent and easier access to ASD phenotypic data, and to compare the characteristics and coverage of instruments that were recently used in ASD diagnosis and research.

The phenotypic ontology can be considered as a comprehensive ontology for representing the disorders in autism spectrum disorders. This ontology covers all possible mental and physical difficulties that deal with ASD based on various diagnosis instruments but treatment of autism is not included in the proposed ontology. Treatment of ASD is a heterogeneous and complex domain exhibiting many techniques and methods developed by different researchers. An intervention can be effective for multiple disorders in autistic children, which represent the complex relations between the disorders, treatment methods of ASD, and effect of the treatment on quality of life of the individual with ASD. These relations have not been indicated in this work and their ontology is essentially to represent the ASD disorders phenotype for research purposes.

## 2.4 Contribution of ASD ontology

As discussed in the previous sections, there are many initiatives for building ontologies for autism spectrum disorders by many researchers. These ontologies were built for different purposes and do not cover a comprehensive ontology system for ASD domain. These limitations were discussed in the sections above. The autism spectrum ontology developed in this thesis aims at addressing these shortcomings, thus aiming to build a fully comprehensive ontology and accurate knowledge in field of ASD.

The autism spectrum disorders ontology, which is presented in the next chapter, is created by merging knowledge from many existing ontologies for ASD and other trusted sources such as standard textbooks, related published articles and the clinical studies. These different sources have been used to extract a huge terminology in the field and to avoid the duplications of terms and concept. In other word, using the multiple type of resources allow us to unify the terminology for autism spectrum disorders. Moreover, all ontologies by different researchers have been fully analyzed to create a non-ambiguous ontological structure for ASD, thus, benefiting from strengths of each of these ontologies. Moreover, the intervention knowledge for autism spectrum disorders has also been covered by the proposed ASD ontology. The treatment field of autism spectrum disorders is an important domain in ASD, which has been investigated by many researchers and autism therapies. Additionally, finding the right treatment for families

or caregivers of individuals with ASD is a challenging task in terms of access to information about available treatment methods and relevant interventions for their autistic children. The ASD ontology provides information about many intervention approaches with relevant properties for the intervention to users.

Although most of the pervious work for autism ontology are built based on the hierarchical relationships between the classes and subclasses. The proposed ASD ontology makes use of all of these hierarchical structures to synthesize the most commonly agreed upon classification. For instance, all possible relationships among the treatments, disorders, and effects of the disorders on patients were captured and defined in the proposed ASD ontology.

According to pervious research work, most developed ontologies and their query systems were mainly targeted for research purposes. Technically normal users such families or caregiver of people with ASD are not able to take advantage of these systems. In this study, we propose an ontology and a query system that can be used by both professionals and normal users enabling them to access all kind of information about autism spectrum disorders. The query system displays the basic structure of the ASD ontology based on the user query and provides basic core knowledge as well as advanced knowledge about autism spectrum disorders such as effect of the disorders on autistic people, relevant treatment, and other information for increasing the quality of life for people with ASD.

# Chapter 3

# System design and Methodology

Building ontology for representing knowledge in the medical field and indicating the medical terminology is a difficult task where analysis of structure and the concepts of medical terms should take place carefully before designing the ontology (Stanescu, 2011). The medical field has its own specialized language, lexicons and terms that are used for storing and communicating medical knowledge and patient information in an efficient way. Hence, medical ontologies are optimized for processing and representing a significant amount of knowledge in any specific area of medical science (Stanescu, 2011). Methodology in building ontology is very important and it aids the designed system to be improved in many ways such as using methodology for constructing any piece of software. Methodology supports and allows the system to obtain an efficient result by following a set of steps, which are based on best practices and works by others (Rajiv Kishore, 2007).

The Uschold and King (1995) methodology is a specific proposal for building ontology which they used the proposal to create Enterprise Ontology and a set of guidelines to develop an ontology as described below:

- Identify the purpose of ontology.
- Build ontology based on three activities; first activity is to capture the concepts and relations between the concepts for the ontology; second activity is to code the ontology using formal language such as OWL; third activity is to integrate the ontology with a previous existing one.
- Evaluate the ontology in technical ways to be compatible with software environment and documentation with respect to a frame of reference.

The methodology for this work is based on the ontology proposal by Uschold and King (1995). The purpose of ontology as it has been previously discussed in the introduction chapter (research contribution section) is to conceptualize the available knowledge in different aspects of autism spectrum disorders such as disorders, effect of disorders and treatment in ASD. Furthermore, the goal of this research work is the unification of terminology for duplicated data in this knowledge domain which indicates the same result, and to facilitate access to the basic information of autism spectrum disorders, among the huge amount of available data, for professional users such as clinicians, researchers and normal users (i.e. families of autistic people or who have contact with ASD individuals). The last purpose of the designed ontology system is to provide a query system for users from different parts of the world to access the autism spectrum disorders ontology. Based on the presented ontology proposal by Uschold and King (1995), the second phase of the methodology is building the ontology following three activities described in this chapter, and the last phase of the methodology, which consists of ontology evaluation as described in chapter 4.

Although there are many ontology editors available for creating and editing ontology in Web Ontology Language (OWL), Protégé has been used for creating the autism spectrum disorders ontology as it is one of the most powerful ontology editors. Protégé is freely available and open source and it is the focus of important OWL tutorial material (Mihail Popescu, 2009). Protégé was developed by the Stanford Center for Biomedical Informatics Research at the Stanford University School of Medicine. The Protégé user interface provides the user with access to all of the components of OWL and it focuses heavily on the specification of such OWL ontologies (Mihail Popescu, 2009). Fully integrated access to OWL DL reasoners is an important feature of Protégé that provides many benefits to users through the potential interface.

This chapter contains three main sections, which discuss how to build the autism spectrum disorders ontology. The first section discusses the structure of the top level design for the ontology as lightweight ontology; the second section clarifies the main design and the concepts of the top level classes with their related subclasses; and the third section justifies the relation among the classes and the properties of the classes.

## 3.1 Initial design

As it was discussed in chapter two, domain knowledge and operational knowledge are the two types of ontology that medical and clinical knowledge can be defined through. Operational knowledge ontologies are more complicated to implement compared to domain knowledge because there are many different methods to do the same process and obtain the same result. Moreover, future changes in method development can affect the ontology based on the new techniques and process methods introduced by researchers. Although most of the ontologies for autism are domain knowledge ontology, the created ontology system for autism spectrum disorders, described in this thesis, includes both domain knowledge and operational knowledge ontology. The domain knowledge part represents the information about autism spectrum disorders and the effect of disorders on individuals with ASD, while the operational knowledge signifies the information about the available treatment for autistic people.

The initial design of top level classes of autism spectrum disorders ontology demonstrates the domain knowledge and operational knowledge type of the ontology. In this section, first, light weight ontology knowledge and then purpose of using top level ontology for creating the designed system will be reviewed. Second, the related existing lightweight ontology in the medical field and the influence of the contribution of the existing light weight ontology in this research will be shown. The final part presents the created top level ontology as an initial design for the autism spectrum disorders ontology based on existing designs. Additionally, the changes over the classes, subclasses and properties of existing top level ontology to be compatible in terms of autism spectrum disorders concepts is discussed.

### 3.1.1 Lightweight ontology

The idea of providing one set of defined terms and relations that will precisely represent the concepts of a domain is the goal of lightweight ontology. Light weight ontologies typically consist of a hierarchy of concepts and a set of relations held between concepts (Álvaro Rocha, 2014). In another word, light weight ontology presents the backbone taxonomy where the more specific concept is the child concept rather than parent concept of the domain. Based on the

usage of ontology, there are two types of lightweight ontology; descriptive and classification lightweight ontologies (Farazi, 2010). Descriptive lightweight ontologies are used to describe the meaning of terms and the structure of the knowledge domain in order to detect more information based on natural language usage and human cognition (Álvaro Rocha, 2014). Classification lightweight ontology is used to classify the concepts and terms of domain knowledge in the basic structural form. The lightweight ontology of autism spectrum disorders is based on descriptive lightweight ontologies.

The simplest formalization of the domain can be obtained by lightweight ontologies (Roberto Poli, 2010). Hence, using the lightweight ontology as a basic structure form of the autism spectrum disorders ontology can improve the backbone structure of the concepts in the ontology and it has significant benefits for the system such as:

- Minimizing the difficulty for users to adapt to the ontology (Álvaro Rocha, 2014)
- Classification of lightweight ontology can be used for development of heavy ontology (Roberto Poli, 2010)
- Providing simple structure of concepts in the domain (Álvaro Rocha, 2014)
- Improving the ontology where scale and performance are critical (Roberto Poli, 2010)

Consequently, the lightweight ontology is the top level ontology structure for the created autism spectrum disorders ontology. The top level classes and main concepts of the autism spectrum disorders have been imported in the lightweight ontology. Furthermore, the relations between the main classes of ontology and properties of the classes are part of the lightweight ontology.

### 3.1.2 Related design

Among all the available structures for lightweight ontology, building a suitable structure for the autism spectrum disorders is a challenging task. The lightweight ontology defines the main pattern and enhances the ASD ontology. Hence, the most related lightweight ontology structure, which has been chosen for this work is Disease-Treatment ontology by Christopher S.G Khoo and his colleagues. Disease-Treatment ontology is a general top level ontology for the medical domain which is being developed to model and represent treatment information for disease.

As it was discussed in chapter two, the Disease-Treatment ontology has five classes and each class contains its own subclasses and properties. The concepts for the initial design of the Disease-Treatment ontology have been developed based on an analysis of 50 medical abstracts on colon cancer. The ontology builds on existing medical and taxonomies and available ontology such as UMLS sematic network, Medical subject Heading (Mesh) and the NCI Thesaurus. The top level classes of Disease-Treatment ontology consist of Disease class, Treatment class, Effect class, Condition class, and the Evidence class.

The disease class in Disease-Treatment ontology refer to the available diseases in the interest medical domain. The treatment class provides information about the available treatment for the diseases. The additional conditions or attributes of the patient that may affect the efficacy of the treatment can be map into condition class of the Disease-Treatment ontology. The effect class represents the influence of the disease on patient in long term or short term. Evidence class refers to data reported in the medical article that supports the effect class as its shown in the figure 3.

They specified the subclasses for all the classes in the top level ontology, for instance Disease class has 6 subclasses and one property. However, the top level ontology for ASD does not need to have all those specified classes in Disease-Treatment ontology. Therefore, some classes have been removed and changed based on the concepts and knowledge of autism spectrum disorders. The structure for the lightweight ontology of ASD builds on the Disease-Treatment top level ontology because of the following reasons:

- Disease–Treatment ontology is general medical ontology that can be used for any purpose in the medical field.
- It has the closest pattern structure for the ASD lightweight ontology in terms of main classes and relations between the classes and properties.
- Many medical ontologies are disease based ontology; they represent the knowledge about a specific disease. Disease–Treatment ontology is treatment based ontology and treatment is one of the main classes of the ontology.
- Disease–Treatment ontology is constructed as an enhancement of existing medical ontology such as Unified Medical Language System (UMLS), semantic network 5,6, the

Medical Subject Headings (MeSH)7, and the NCI (National Cancer Institute) thesaurus (http://bioportal.bioontology.org/ ontologies/39478 & http://ncit.nci.nih.gov/ncitbrowser).

- The concepts of Disease–Treatment ontology developed based on 50 medical abstracts.

Therefore, the autism spectrum disorders lightweight ontology pattern is taken from the Disease–Treatment ontology because of the mentioned aims of this design. However, there are some changes that need to be applied to Disease–Treatment ontology in order to make it more fit for the autism spectrum disorders ontology. ASD is a huge field with heterogeneous concepts in terms of disorders, effect, and treatment. Thus, there are many different concepts, relations, and properties compared to other medical fields that need to be considered when creating a comprehensive ontology for autism spectrum disorders. These changes will be discussed in the next section and the initial design for ASD ontology will be shown in the form of lightweight ontology with the top level classes and relations among the main classes. Likewise, the initial design for the ontology will be imported into protégé and subclasses for the top level classes will be indicated.

### 3.1.3 The structure of the design

In the previous section, the related design of lightweight ontology for ASD and the benefits of using the Disease-Treatment ontology for ASD top-level have been discussed. In this section, the main top level classes as a lightweight ontology for autism spectrum disorders based on the Disease-Treatment ontology by Christopher S.G Khoo and his colleagues will be shown. However, there are significant changes that need to be made to adopt the Disease-Treatment ontology for the autism spectrum disorders top level ontology as a main pattern of system.

The following are the applied changes on Disease-Treatment top level ontology:

- Evidence class has been removed from the top level classes. Each class of ASD ontology is based on the information, which has been extracted from trusted and reliable sources. For instance, the sources, which are strong evidence will be indicated for relation of the classes of main ontology design and they are part of the ontology.

- Disease class has been removed from the top level classes of the lightweight ontology. According to the definition by G. Wobeser, disease is any impairment that interferes or modifies the performance of normal functions including responses to environmental factors such as nutrition, toxicants, climate, infectious agents, inherent or congenital defects, or a combination of these factors (Wobeser, 1997). However, autism spectrum disorders deal with disorders in mental and emotional health. Although disorders can be considered as a disease for humans, the word disease has no place in the ASD encyclopedia.
- Condition class also has been removed from the top level classes. Patients with autism spectrum disorders are autistic people who suffer from multiple disorders and the conditions can vary between autistic people because of different difficulties in cognition, emotion regulation, and behavior. The effect of disorders on autistic people can define the condition of individuals with ASD but with different severity.
- Disorder class has been added to the top level classes of ontology to represent all types of disorders in ASD. Autism spectrum disorders are mental disorders. Therefore, the disease class has been replaced by disorder class in the initial ontology design. Mental disorder is a syndrome characterized by clinically significant disturbances in an individual's cognition, emotion regulation, or behavior that reflects a dysfunction in the psychological, biological, or developmental processes underlying mental functioning (Donald W. Black, 2014).

The top level classes of autism spectrum disorders ontology have been defined after applying the above changes to the Disease-Treatment top level ontology. The top level ontology for ASD contains three main classes such as disorder class, effect class, and treatment class as shown in the figure below.

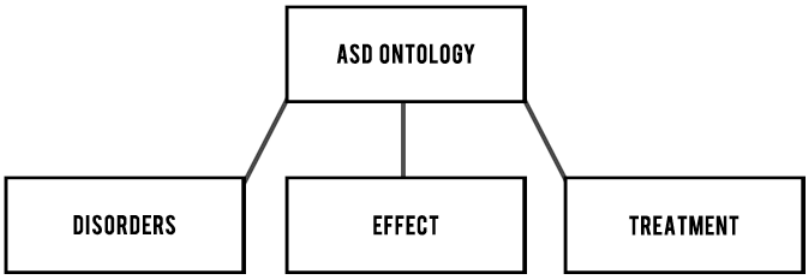


*Figure 5. Top level classes of autism spectrum disorders ontology.*

There is a solid relation between all three main classes of ASD top level ontology. On another hand, we can consider each class as a different ontology with its own subclasses however the strong relation between them is the coherence of the ontology. The mentioned relations among the main classes are discussed below:

- Disorder has effect; the relation indicates that there is one or more effect for every disorder in ASD.
- Disorder has treatment; the disorders can have an intervention in ASD to improve the quality of life of autistic individuals and decrease the difficulties faced by an ASD patient.
- Treatment has effect; treatment of autism has an effect on disorders and mental health and physical situation of autistic people.
- Treatment is part of disorder; there are many available interventions for autistic people with different disorders. This is a symmetric relation with disorder has treatment.
- Effect is part of disorder; there are many long term difficulties that autistic people suffer from during their life. This is a symmetric relation with disorder has effect.
- Effect is part of the treatment; the positive effect of treatment on individuals with ASD. This is a symmetric relation with treatment has effect.

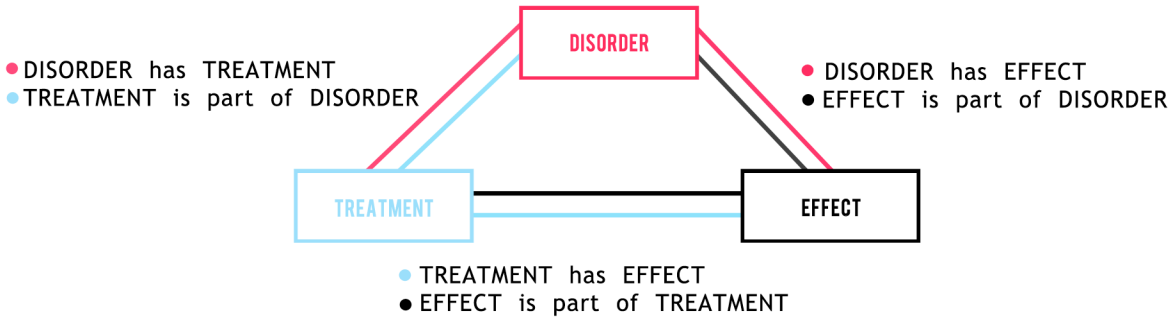


*Figure 6. Relations between top level classes of the ontology.*

The main classes of the lightweight ontology of ASD and the relations between classes have been defined for the initial pattern of ontology. Moreover, the subclasses for each top level class are extracted based on the domain knowledge of autism form, an existing ontology, or other reliable sources.

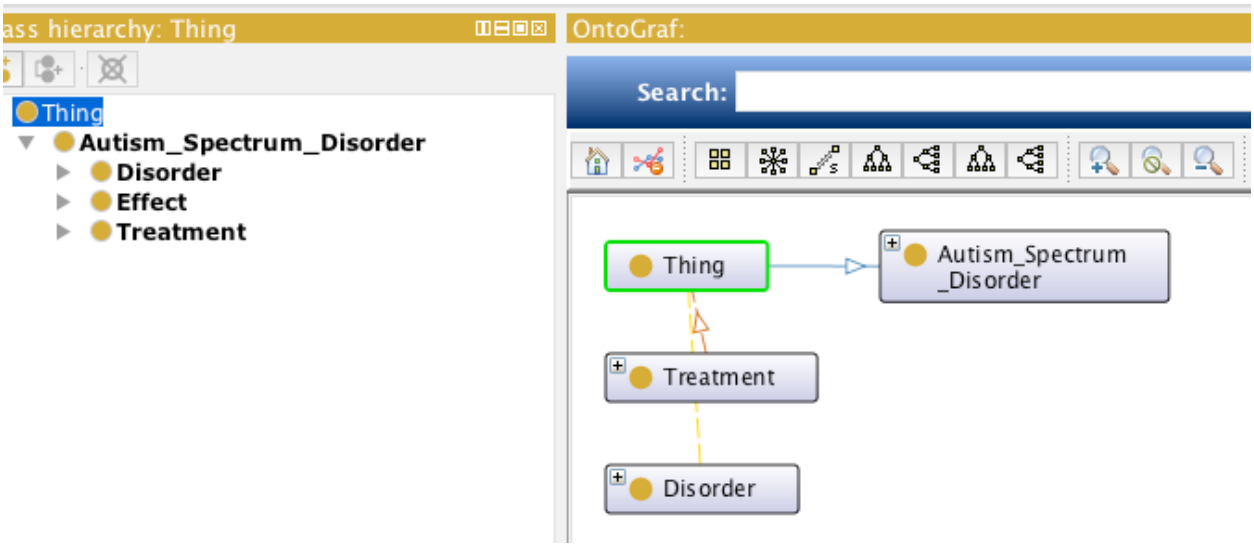


*Figure 7. Top level classes of the ontology imported into protégé (OntoGrph view).*

The disorder class consists of one subclass and one property. Type is the only subclass of disorder, which represents type of disorders in autism spectrum disorders and physical feature is the property of disorder class which indicates the disorders with physical difficulties. The subclass of type is based on the definition of the autism spectrum disorders for existence of different type of disorders in ASD.

According to the book Social Skills and Autistic Spectrum Disorders by Lynn Plimely et al. (2007), autism spectrum disorders represent impairment in three main areas of development where people on the autistic spectrum manifest difficulties in social skills, communication skills, and common behavior (Lynn Plimley, 2007). Additionally, autistic people must have symptoms that belong to the following three main areas of core features of impairment; impairment in language, impairment in social interactions, and the presence of repetitive stereotype behavior or interests (Carol Turkington, 2007). These three main types of disorders have also been recognized by Leo Kanner in 1943 and other researchers as it was discussed in chapter two. Hence, the subclass of the type class is manifested in three subclasses such as social skills class, communication skills class, and common behavior class. After clarifying the subclasses of type class, they have been imported into protégé.

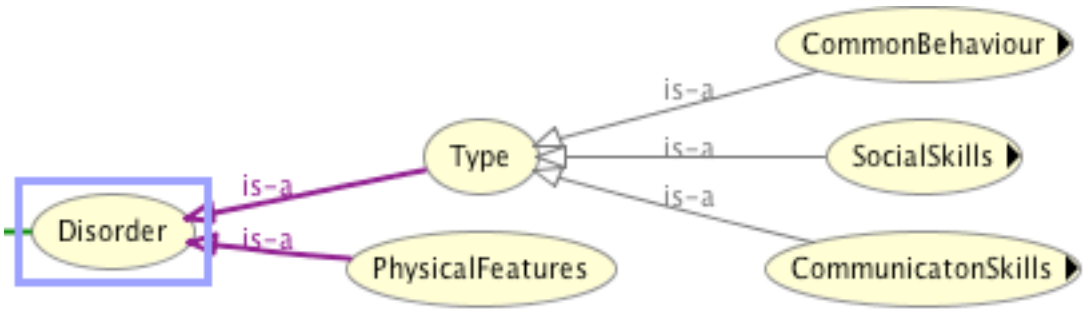


*Figure 8. Hierarchy relation of subclasses of disorder class in protégé.*

The figure above demonstrates the subclasses and the relation of the subclasses of the disorder class in protégé. The hierarchy relation among the subclass and class is represented via the 'is-a' relation in protégé. As an example of an 'is-a' hierarchy relation in protégé, the relation of the type class with the three main subclasses can be expressed as social skills is a type of disorder, communication skills is a type of disorder, and common behavior is a type of disorder. The next main top level class in the ontology is the effect class. The purpose of the effect class is to specify the knowledge of interventions effect and impairment effect in social, communication, and behavior for autistic individuals. Furthermore, it contains two main subclasses such as treatment effect class and disorders effect class. Treatment effect class represents the knowledge

of the effect of available ASD intervention on autistic patients and disorder effect class manifests the effect of disorders in the form of the difficulties that autistic people can experience during their life.

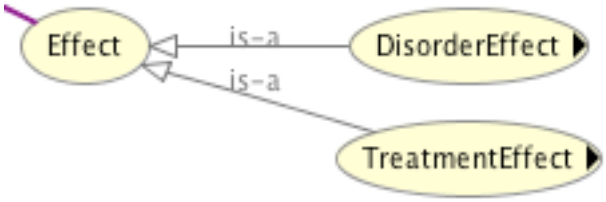


*Figure 9. Two subclasses of effect class shown in hierarchy relation of is-a.*

The last class of the main classes of top level ontology is the treatment class. The purpose of the treatment class is to represent the knowledge of intervention in autism spectrum disorders. According to studies by researchers, effective treatments for ASD mostly target the core area of impairment such as communication, reciprocal social interactions, and atypical behaviors (Carol Turkington, 2007). Furthermore, there is no medication that can cure the ASD completely or treat the core symptom however interventions can have a significant effect on quality of life of autistic patients.

The presented information in this class is helpful and supportive for caregivers and researchers in the field of ASD. Additionally, autistic disorders are recognized as a chronic disorder within which there are changes to the time that various treatments should be applied. However, most treatments are symptom directed at the present time (Michael B. First, 2006). Thus, because similar symptoms are targeted for treatment of disorders, the treatment of other mental disorders is the same as for autistic disorders (Michael B. First, 2006).

The targeted interventions for this ontology are three popular and beneficial types of treatment. The first type is therapies, which has had a big impact on treatment of autism spectrum disorders for years. Therapies include a wide range of tools, services, and teaching methods, all of which help children with autism spectrum disorders potential (Autism speak, 2015). There are many available therapies that are effective for intervention of social and communication difficulties in

ASD. The second type of treatment is biomedical interventions, which is more helpful for problems such as body chemistry, gut, and the immune system of autistic people.

Dietary is one of the very important interventions in biomedical treatment, which is being widely used these days by many ASD individuals. Dietary intervention has major beneficial effects and it is considered as a safe intervention for autistic patient to correct a range of physiological differences (Aitken, 2008). The third intervention is medication, which is not part of this work and it could be part of future work for this project.

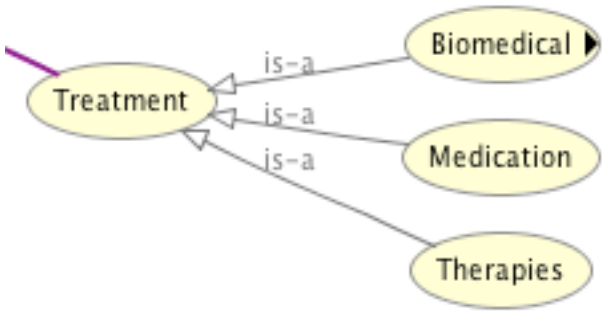


*Figure 10. Treatment class with the three subclasses shown in protégé.*

The treatment class contains three main subclasses based on intervention approaches such as therapies, biomedical, and medication. The most popular treatments are biomedical intervention and therapies because they are considered safe interventions compared to medication. Many of the available medications for ASD are prescribed off label which means that they are not approved by government and health centers (Carol Turkington, 2007). Moreover, medications have side effects on autistic children such as sedation, muscle stiffness, abnormal movements, weight gain, etc. (Carol Turkington, 2007).

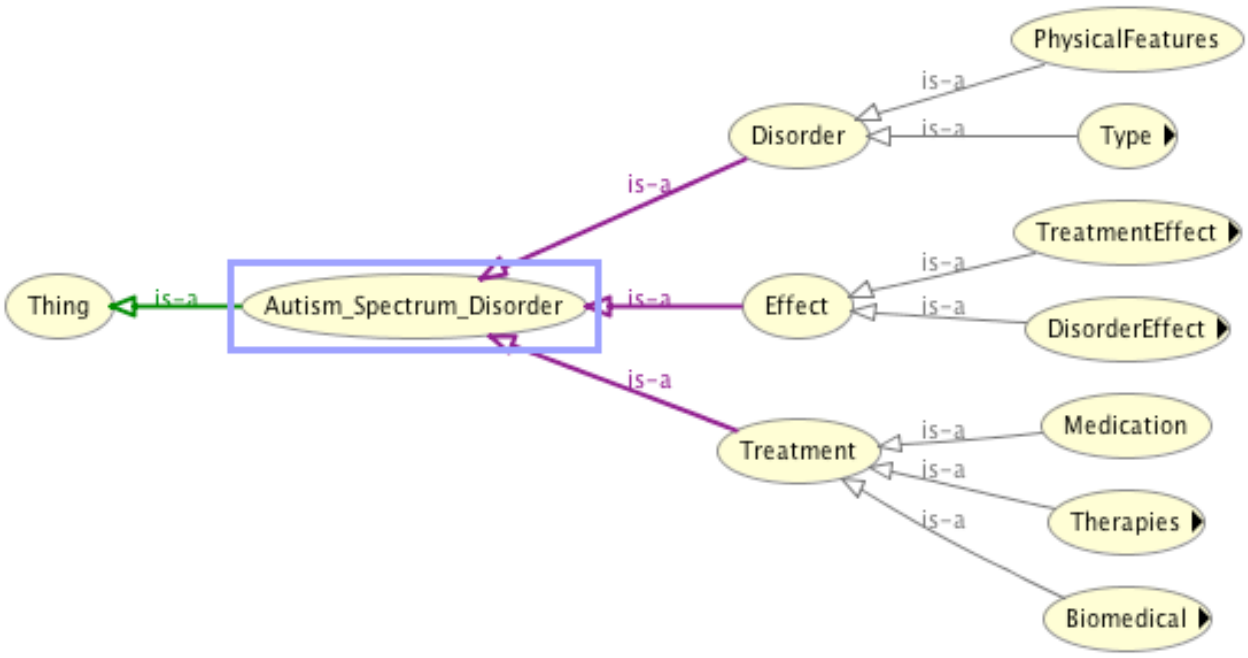


*Figure 11. Top level ontology with their subclasses imported into protégé.*

The top level classes and their subclasses have been added into the ontology as it is shown in figure 12. The core structure of the ontology has been built based on existing ontology such as Disease-treatment ontology and fundamental knowledge of ASD. The lightweight ontology of ASD is mapped into protégé and it has the ability to be a heavy ontology by importing the concepts and terms of each class. As it is manifested in the figure above, the ontology for autism spectrum disorders will cover the domain knowledge of three main aspects of autism which are the core concepts of ASD. In the next section, the heavy ontology will be constructed based on the top level ontology of ASD by adding more concepts and relations between the classes.

## 3.2 Final design

In this section the concepts for each main class of ontology will be introduced and the second version of ASD ontology, which is a heavy ontology with a vast number of concepts will be discussed. It also provides information on the starting point of the second version of ontology development, which allows the ontology to be more flexible with extensive knowledge in different fields of autism such as treatment, effect, and disorders. The starting point for building

the main ASD ontology is to create a set of terms selected from the most commonly used terms in standard textbooks, published articles from high level journals, and relevant domain ontologies.

The ontology development for the main design of ASD ontology was based on avoiding the use of familiar terms with new and different meanings. This was done in order to prevent confusion both in the encoding of information into the ontology and in the interpretation of such information by end users. After extracting the concepts for each main class of the ontology from different trusted sources, all concepts will be imported into the protégé for building a comprehensive ASD ontology.

### 3.2.1 Adding concepts for the main classes

In this section, expanding of ontology for each subclass of the main classes such as disorders class, effect class, and treatment class, will be initiated to bring forward benefits that provide additional justification for its use. For example, adding the concepts for subclasses allows an improved understanding of the domain to generate improvements in the list of terms for the ontology development (Robert Arp, 2015). The purpose of this section is to develop representational information in ASD ontology for the top level classes such as disorder, effect of disorders and available intervention for them.

Although there are many terms for the purpose of successful information exchange, the created set of terms and terminologies for ASD ontology are from the existing relevant ontology such as modeling the autism spectrum disorders phenotype ontology by Alexa T. McCary et al. 2014, which is a commonly used term in standard textbooks and relevant articles published for ASD. Acronyms and abbreviations are avoided to use for formulating terms of ontology for better understanding the terminologies in the created autism spectrum disorders ontology for users. Therefore, the information of ontology becomes more retrievable and combinable in order to overcome the problems caused by multiple conflicting vocabularies (Robert Arp, 2015).

#### 3.2.1.1 Disorder class

Disorder class is one of the top level classes of ASD lightweight ontology which is also an important class for the autism spectrum disorders ontology. Disorder class has a subclass of type and one property. Type class contains three subclasses such as social skills class, communication skills class, and common behavior class as it was discussed in the light weight ontology. In this section, the concepts of each subclass of type will be initiated for the ontology which is extracted from a relevant existing ontology called Modeling the Autism Spectrum Disorder Phenotype by Alexa T. McCary et al. 2014. Modeling the Autism Spectrum Disorder Phenotype ontology is the comprehensive disorders ontology for ASD which is built on 24 autism instrument tools such as questioner, interview, and direct assessment. The created ontology by Alexa T. McCary et al. 2014 has 283 concepts; the concepts for disorders have been extracted from Modeling the Autism Spectrum Disorder Phenotype for ASD ontology on the three subclasses of disorders class.

##### 3.2.1.1.1 Social skills

Social reciprocity deficits among the autistic children are a core feature of the autism spectrum disorders regardless of cognitive or language abilities (Joseph D. Buxbaum, 2012). Moreover, social skills difficulties will not be remitted with development. Indeed, when children with ASD approach adolescence, impairment and distress increase because the social milieu becomes more complex and the child becomes more aware of their social disability (Tantam, 2013). Diverse and involved speech, linguistic conventions, and interpersonal interaction are involved in social impairments in individuals with ASD (White, 2007).

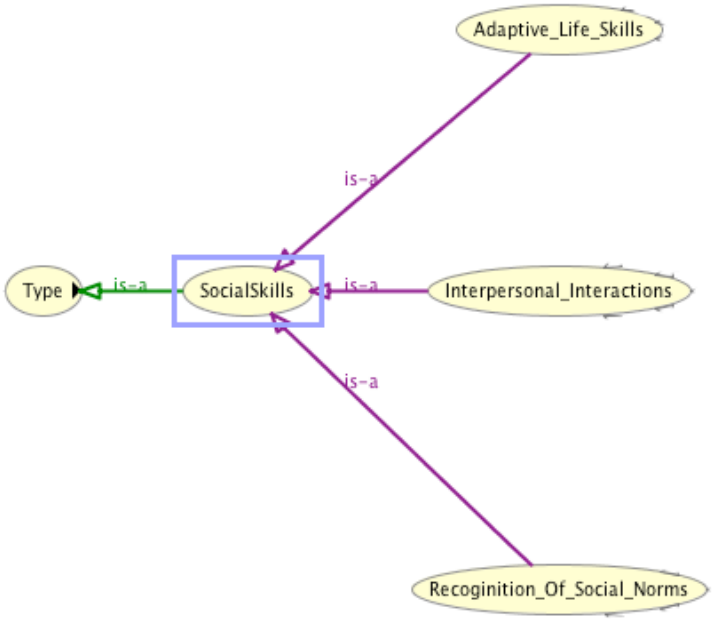


*Figure 12 Subclasses of social skills class shown in protégé.*

Social skills class has three different subclasses such as *adaptive_life_skills*, *interpersonal_interactions*, and *recognition_of_social_norms* as it is shown in figure 13. The relation between the social skills class and subclasses are presented via the 'is-a' relation in protégé which indicates that the adaptive life skills is a social life skill. Thus, social skills impairment in ASD can be defined as difficulties in adaptive life skills, interpersonal interaction, and recognition of social norms. Furthermore, there are subclasses for each subclass of social skills that will be discussed in the section below.

The first class of social skills class is *adaptive_life_skills* class which has three subclasses such as *regression_of_general_skills*, *home_life_skills*, and *community_life_skills.* These sub classes represent the concept of impairment in adaptive life skills for individuals with ASD. Additionally, subclasses can explain the knowledge of the class. For instance, subclasses of *adaptive_life_skills* class explain what the adaptive life skills impairment means and what the categories of this impairment are in autism spectrum disorders.

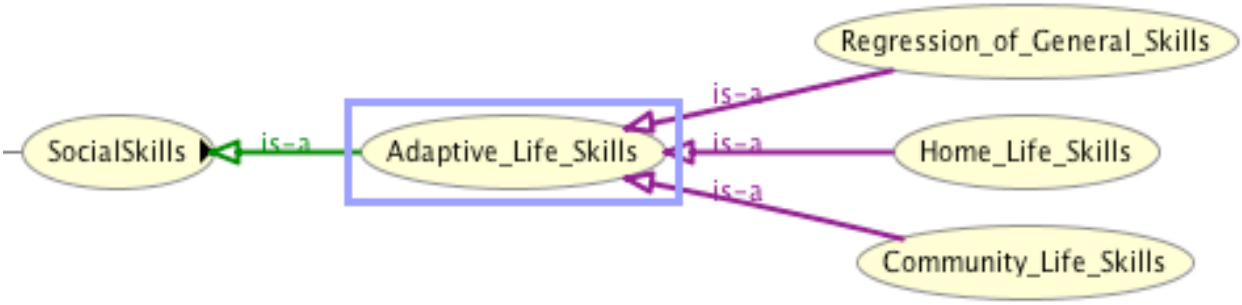


*Figure 13. Subclasses of adaptive_life_skills class.*

The *interpersonal_interactions* is the second subclass of *adaptive_life_skills* class which contains five subclasses such as *social_anxiety*, *interpersonal_awarness*, *reciprocal_social_interaction, socialintrest* and *social_awkwardness.* Subclasses manifest the impairment in interpersonal interaction in autism knowledge.

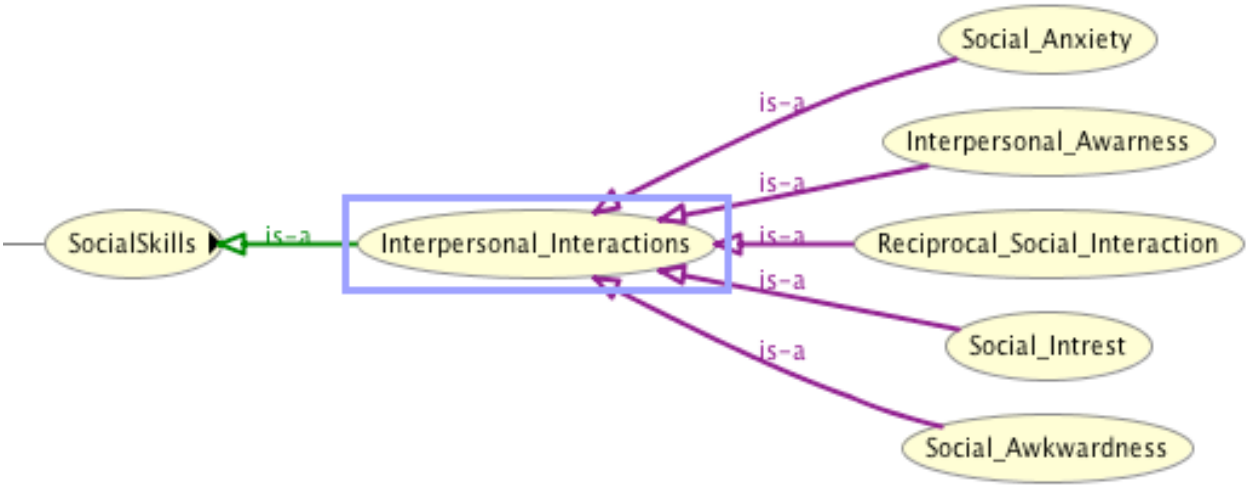


*Figure 14. Subclasses of interpersonal_interactions class.*

The last subclass of social skills class is *recognition_of_social_norms* which presents the difficulties that autistic people face when trying to understand socials norms in society. This concept has four subclasses such as *intentional_bad_behavior*, *awareness_of_social_cues*, *regard_for_customs_and_manners*, and *conversation_skills*.

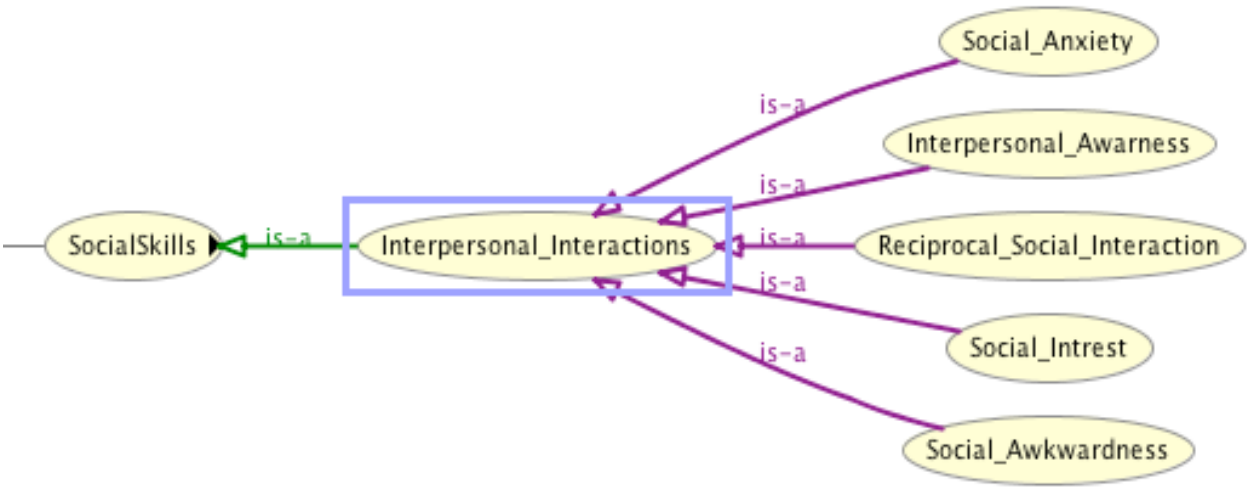


*Figure 15. Subclasses of recognition_of_social_norms class.*

### 3.2.1.1.2 Communication skills

Communication development happens differently and more slowly in children with autism spectrum disorders. Communication skills impairment is more associated with language ability in ASD. However, not every autistic child will have language problems because it depends on his or her intellectual and social development (Gilead, 2011). Although there are some individuals with ASD who are not able to speak, most children with ASD have little or no problem with word pronunciation. The majority of these children deal with difficulties in using language effectively, especially when they talk to other people (Gilead, 2011). Furthermore, problems such as understanding the meaning and rhythm of words and sentences, and understanding body language and the nuances of vocal tones are all common among children with ASD (Gilead, 2011).

The communication skills class is one of the core classes of the disorders type class. This class has one subclass which is *language_ability*. The subclass of communication skills contains seven different classes which are used to describe the challenges that people with ASD face in terms of language ability. The obtained subclasses for *language_ability* are *receptive_language, developmental_or_regression of_language_skills, expressive_language, reading_and_writing,*

*nonverbal_communication*, *integrated_verbal_and_nonverbal_communication*, *and idiosyncratic_routinized_speech*, as it is shown in the figure below.

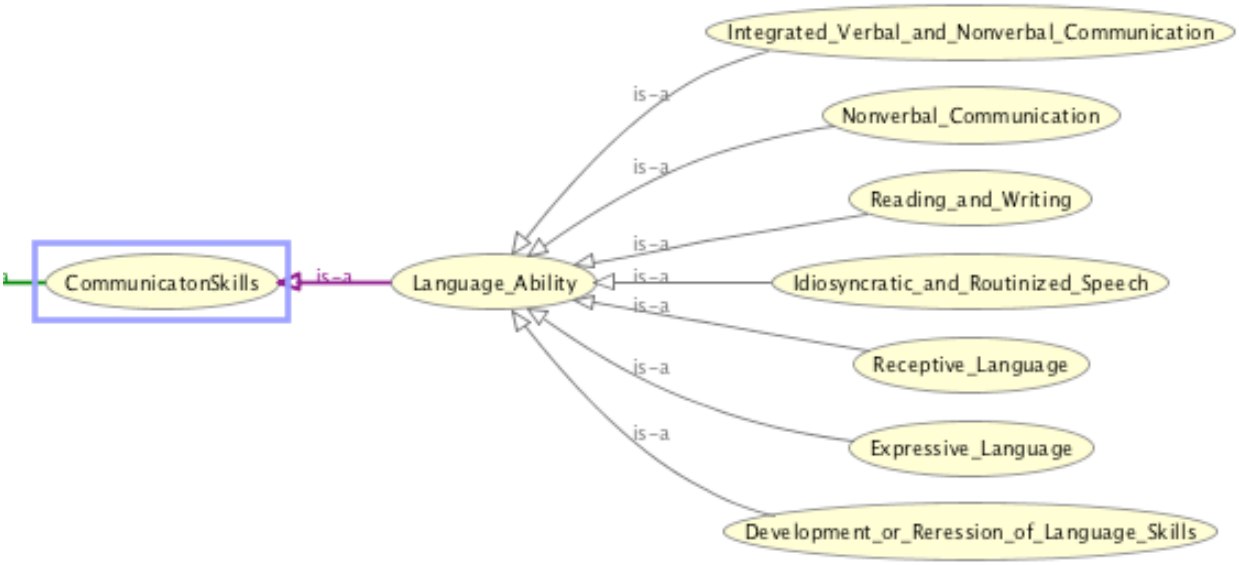


*Figure 16. Subclasses of language_ability class.*

### 3.2.1.1.3 Common behavior

Common behavior is the last type of disorder in autism spectrum disorders which points to behavioral difficulties in autistic children regardless of their age and ability. The best way for helping autistic children with behavioral difficulties is by understanding the condition and the difficulties associated with it (Mark Rosenfield, 2009). The concepts for common behavior class help the user to clarify the difficulties within the common behavior impairment in ASD. The common behavior class has five major impairments in areas such as cognitive ability, emotional traits, executive function, motors skills, and stereotyped restricted repetitive behavior. These five areas of impairment, like other subclasses for disorders type class, are extracted from autism spectrum disorders phenotype ontology by Alexa T. McCary et al. 2014. Once extracted, they are then mapped into the ASD ontology for importing to protégé as subclasses of common behavior.

Cognitive ability is the first subclass for common behavior class. Cognitive ability holds five subclasses such as *abstract_thinking, analytic_capability, IQ, isolated_cognitive_skils,* and *visual_perception.*

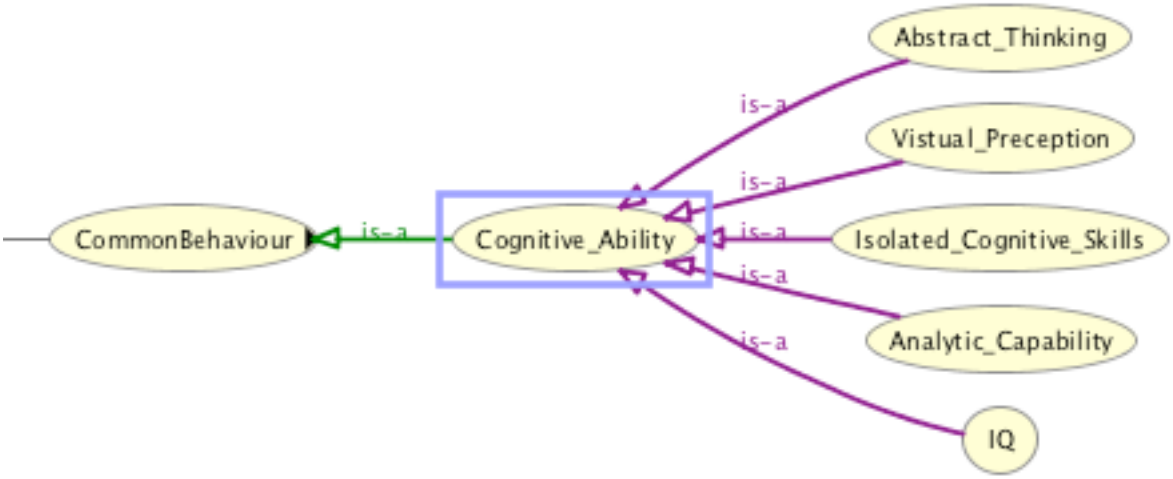


*Figure 17. . Subclasses of cognitive_ability class.*

Emotional traits are another subclass of common behavior class. Emotional traits contain three subclasses which represent the impairment in emotional attributes in mood, self-concept, and affect in people with ASD.

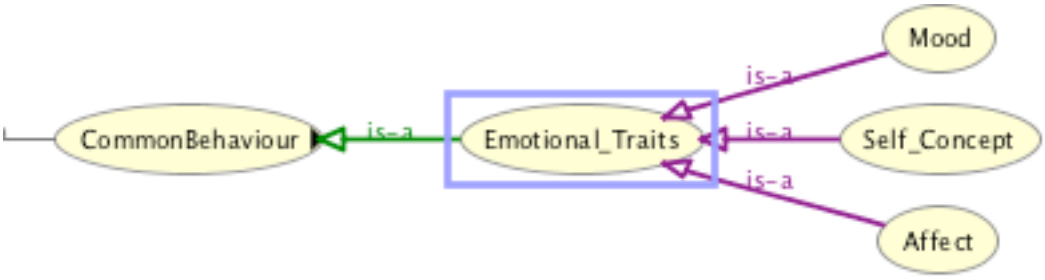


*Figure 18. Subclasses of emotional_traits class imported into protégé.*

The executive function is considered as the third subclass for common behavior and it is a common disorder among individuals with ASD. Executive function is the cognitive construct used to describe goal directed, future oriented behaviors thought to be mediated by the frontal lobes (Duncan, 1986). There are five subclasses for executive function class such as *working_memory, planning, emotional_regulation_and_control, mental_flexibility*, and *response_inhibition.*

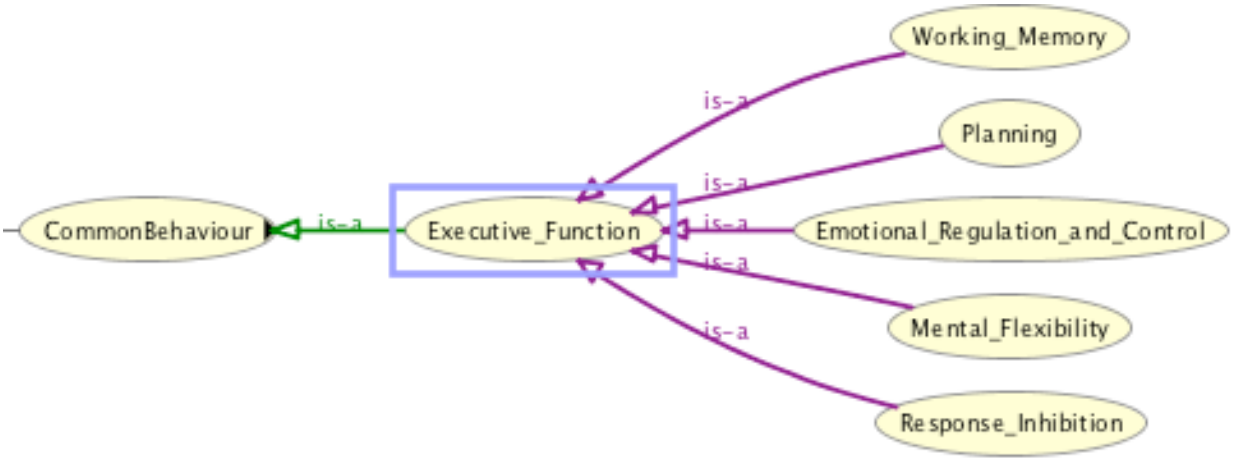


*Figure 19. Subclasses of executive_function class.*

Motor skills is an action or task that has a goal that requires voluntary body or limb movement in order to achieve that goal. Motor skills problems are another impairment of common behavior which has varying degrees of severity among people with ASD. Difficulties in motor skills include subclasses such as *fine_motor_skills, gross_motor_skills*, and *functional_laterality*. The stereotyped restricted and repetitive behavior is the last subclass of common behavior class. Stereotyped and repetitive behavior is one of the three defining features of autism. Although there is extensive published literature on impairment in autism spectrum disorders, this class of behavior continues to be relatively underrepresented in literature (MacDonald, 2007). This subclass of common behavior holds two subclasses such as *restricted_repetitive* behavior and *involuntary_behavior*.

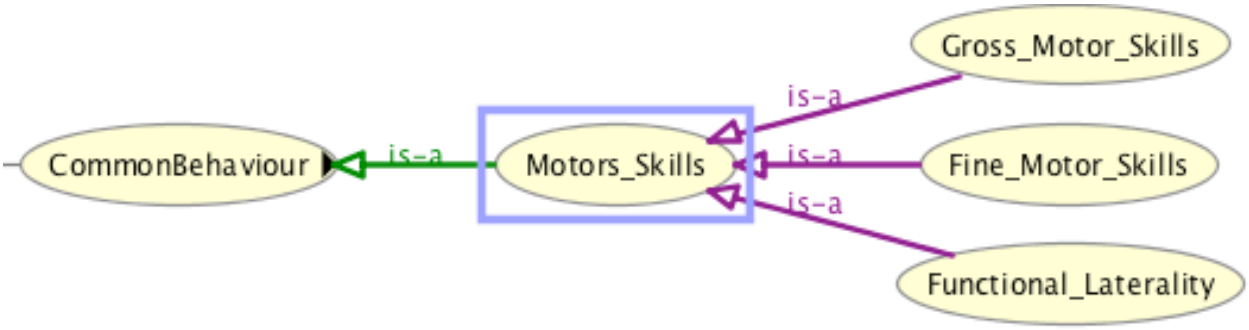


*Figure 20. Subclasses of motor_skills class.*

In this section, all of the concepts for social skills class, communication skills class, and common behavior class have been presented and imported into the ASD ontology. The presented concepts were extracted from an existing relevant ontology such as modeling the autism spectrum disorders phenotype ontology by Alexa T. McCary et al. 2014. The extracted concepts are matched to the pattern of the disorder class and distributed into three categories of disorder.

#### 3.2.1.2 Treatment class

One of the main rules to build a successful and comprehensive ontology is to select terms that are close to the actual usage of a large fraction of those working in the relevant field (Robert Arp, 2015). To achieve this goal for autism spectrum disorders ontology, chosen preferred labels are disseminated by using them in the curtain of large bodies of data which are useful to the wider community such as wide knowledge of ASD treatment.

According to the achieved experience of creating the Gene Ontology (GO) by different researchers, there are three principles that have been used for terminology construction and obtaining terms of ontology which treatment class of ASD ontology is following (Robert Arp, 2015). The three principles are:

- Include in the terminology terms used by influential groups of scientists for the most important types of entities in the domain to be represented.
- Identify areas of disciplinary overlap where terminological usage is not consistent. Look for and keep track of synonyms for terms already in the terminology list from these areas.
- Strive to ensure maximal consensus with the terminological usage of scientists in the relevant discipline. This may well involve working with domain experts, for instance in negotiating terminological compromises.

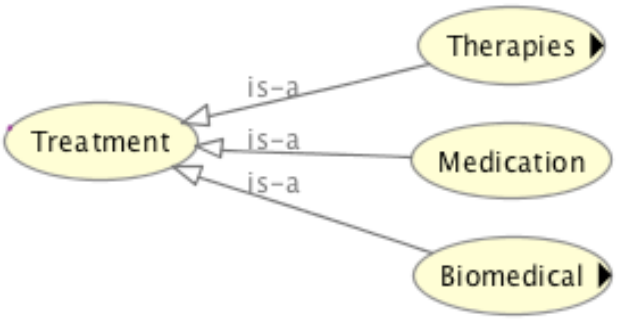


*Figure 21. Subclasses of treatment class.*

The hierarchy relation among the classes and subclasses in treatment class imply an 'is-a' relation in protégé. Treatment class has three top level subclasses which indicate different intervention methods in the autism field. However, the ASD ontology covers the biomedical interventions and therapy interventions in the treatment class. Dietary intervention is one of the popular treatments for children with ASD and has been presented in the biomedical class. To be able to manifest the relations among the disorder class, treatment class, and effect class, there are specific properties for each subclass of treatment class which have been extracted from standard text books for ASD treatment, relevant published articles, and clinical studies which are all based on reliable and trusted sources in the medical field.

### 3.2.1.2.1 Therapies class

Therapies have an important key role in treatment of autism spectrum disorders. Therapies improve function or quality of life of autistic people in such ways as social communication skills, language ability, physical condition, motor skills, and many more. Although there are many introduced therapies for ASD by different researchers, ASD ontology represents the therapies with the highest positive influence in treatment of ASD based on clinical studies and research among autistic individuals. Therapies can be divided into two categories. The first category is anatomy and physiological development which includes physical therapy, massage therapy, holding therapy, etc. The anatomy and physiology development therapies focus more on improvement of physical health of an ASD patient. The second therapy category is communication and social development. This category has a significant influence on social

abilities development, language skills, and behavior such as behavior therapy, speech language therapy, parent-child interaction therapy, etc.

| Therapy | Category |
|---|---|
| Craniosacral therapy | Anatomy and physiology |
| Holding therapy | Anatomy and physiology |
| Physical therapy | Anatomy and physiology |
| Massage therapy | Anatomy and physiology |
| Animal therapy | Communication and social |
| Auditory integration therapy | Communication and social |
| Music therapy | Communication and social |
| Parent-child interaction therapy | Communication and social |
| Sensory integration therapy | Communication and social |
| Art therapy | Communication and social |
| Speech language therapy | Communication and social |
| Occupational therapy | Communication and social |
| Robot therapy | Communication and social |
| LEGO therapy | Communication and social |
| Dance & movement Therapy | Anatomy and physiology |
| Floor time therapy | Communication and social |
| Picture exchange communication therapy | Communication and social |
| Cognitive Behavioral therapy | Communication and social |
| Theatre therapy | Communication and social |

*Table 2. List of standard therapies for autism spectrum disorders intervention.*

The therapies in table 2 are extracted from three standard textbooks for autism spectrum disorders. These are given below:

- Encyclopedia of Child Behavior and Development by Goldstein et al. 2011
- Encyclopedia of Autism Spectrum Disorders by Volkmar et al.2013
- Comprehensive guide to Autism by Vinood B. et al. 2013

These therapies have been confirmed as available interventions for autism spectrum disorders from all textbook sources to be imported into ontology. Some therapies are removed from the therapy table such as Vision therapy because those therapies have not been mentioned as standard therapy for ASD in all textbook sources or published articles. The therapies are extracted based on the three principles of Gene Ontology (GO) to obtain the accurate terms in the knowledge field for the ASD ontology. Hence, the list of therapies has been checked with the clinical studies and published articles of ASD intervention to ensure maximal consensus with the terminological usage of scientists in the relevant discipline.

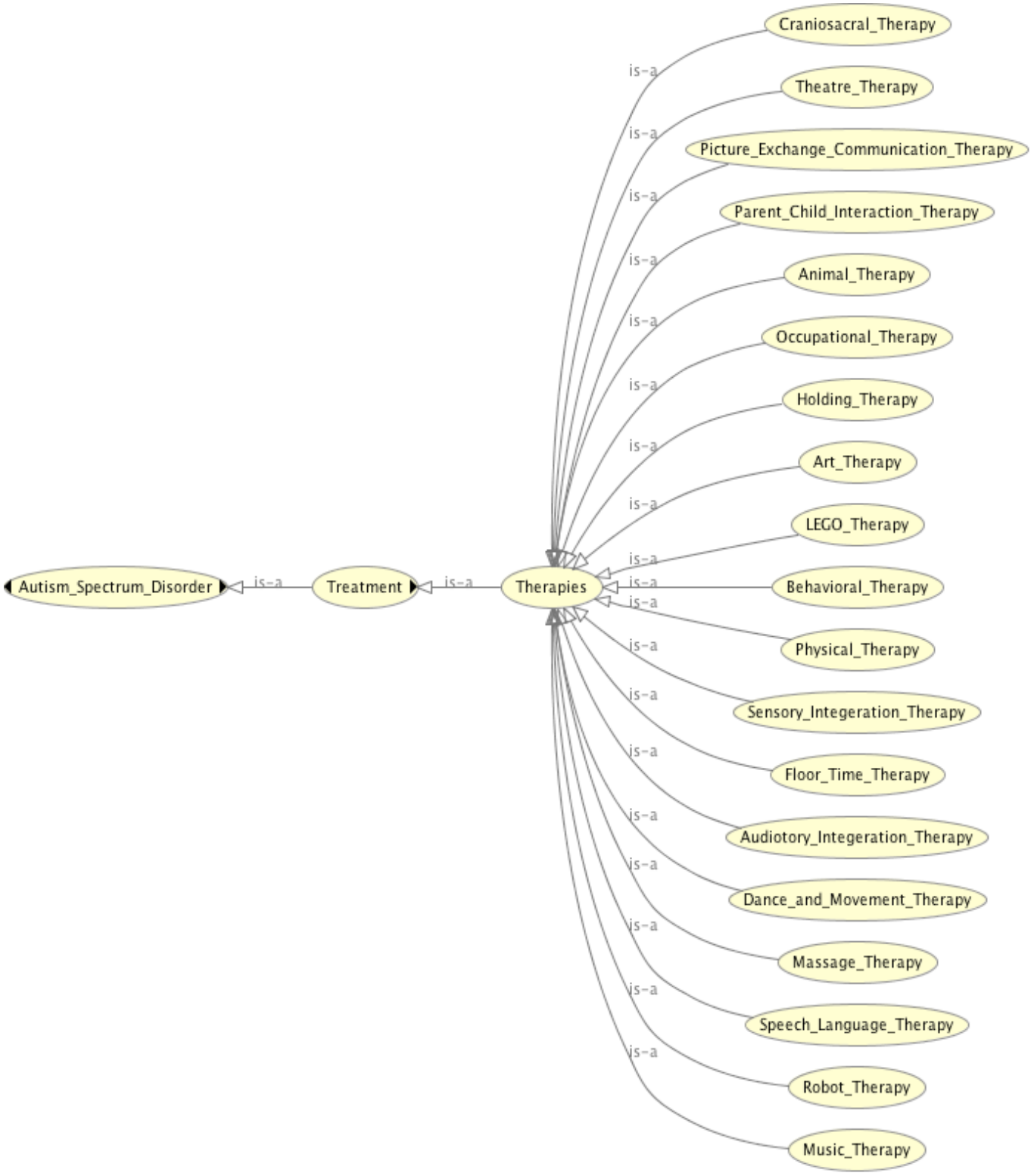


*Figure 22. Therapy classes imported into protégé (OWLviz view).*

Therapies are imported into protégé as subclasses for therapy class and the relation between the different therapies with therapy are indicated with the 'is-a' relation. Therapy class includes 19 subclasses and 10 properties presented for this class. There are a number of properties which were extracted via various sources which is mentioned earlier.

The properties for therapy class are Info, Terminology, Participation, Session, Interventionist, Age, Setting, Equipment, and Sources. The extracted properties for therapies have benefits for the ASD ontology such as explicit knowledge of the purposed therapy, provide more information for users about the intervention, and define the relations of therapy with other top level classes. For instance, 10 extracted properties for Animal therapy (Hippotherapy) are shown below.

- Animal therapy (Hippotherapy)
    - Info: AAI can assist individuals with ASD to develop sensory and social skills, manage problem behaviors, and improve quality of life.
    - Effect: AAI can assist individuals with ASD to develop sensory and social skills, manage problem behaviors, and improve quality of life.
    - Terminology: Animal assisted intervention AAI Dog therapy, Davis (2004) Assistance dog Dolphin therapy, Dilts (2011) Dolphin-assisted therapy Therapeutic horseback, Winchester (2002) Therapeutic horseback riding.
    - Participation: Individual
    - Sessions: 6-48 weeks Duration: 15-60 mins Repeat: None
    - Interventionist: Therapist
    - Age: 4-13
    - Setting: Home, Riding center
    - Equipment: None
    - Sources: (O'Haire, 2013)

The extraction of properties through the different sources has been done for all of the presented therapies of the therapy class as it is exposed in table 3. The sources have also been mentioned as a property of therapy for providing access to the original sources of information to professional users such as doctors, clinicians, and researchers. The feature allows professional users to

investigate and explore more information based on their needs. The effect property has a main key role for delineating the relation among the therapy and the disorder class. The therapy properties have been mapped into ASD ontology via protégé.

| Therapy | Info | Effect | Terminology | Participation | Sessions | Interventionist | Age | Setting | Sources |
|---|---|---|---|---|---|---|---|---|---|
| Animal therapy (hippotherapy) | The inclusion of animals in therapeutic activities, known as animal-assisted intervention (AAI), has been suggested as a treatment practice for autism spectrum disorder (ASD). | AAI can assist individuals with ASD to develop sensory and social skills, manage problem behaviors, and improve quality of life. | Animal assisted intervention AAI, Dog therapy, Assistance dog, Dolphin therapy, Therapeutic horseback, | Individual | 6-48 weeks | Therapist | 4-13 | Home, Riding center | (O'Haire M. E., 2013) |
| Auditory integration therapy | Auditory integration therapy (AIT) was developed as a technique for improving abnormal sound sensitivity in individuals with behavioural disorders including autism. | AIT has improvement on sensory (auditory) processing and communication skills. | Music therapy | Individual | 10-20 days | Therapist, Parents | 4-21 | Home, Clinic | (Edelson, 1999) (Ziring, 1998) |
| Music therapy | a systematic process of intervention wherein the therapist helps the client to promote health using musical experiences and relationships that develop through | Enhance the joint attention behavior, non verbal social communication and social skills. | None | Individual | 10-20 days | Therapist, Parents | 3-40 | Clinic | (James, 2014) (Kim, 2008) |
| Floor time therapy | Floor time therapy is based on applied behavioral analysis and individualized and relationship-oriented. | FTP has effect on social interaction, communication skills, play skills, restricted interest and stereotype behavior. | Developmental Individual-Difference Relation Based, Stanley Greenspan MD Model, PLAY Project Home Consultation (PPHC) | one to one interaction | 3 months | Therapist, Parents | 2-6 | Home | (Dionne, 2011) (Solomon, 2007) |
| Picture exchange communication therapy | (PECS) is a unique communication training program that was developed as a means of circumventing some shortcomings associated with these strategies. It contains six phases. | improvement in ADOS-G rating (language skills) and communication skills. | None | individual | 3months | Therapist | 2-5 | Home Clinic | (Charlop-Christy, 2002) (Bondy, 1997) |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Cognitive Behavioral therapy | The CBT model emphasized behavioral experimentation, | Behavioral therapy remediates the social and adaptive skill and anxiety and depression reduction. | None | individual | 3months | Therapist | 2-11 | Clinic | (Wood, 2009) (McKay, 2015) (Maurice, 1996 |
| Parent-child interaction therapy | PCIT is an evidence-based treatment for disruptive behaviors in typically developing children. PCIT progress is through two phases, Child Directed Interaction CDI and Parent Direct Interaction PDI. | PCIT remediate the significant behavioral problems. | The Child Directed Interaction (CDI)<br><br>The Parent-Directed Interaction (PDI) | individual | None | Therapist | 3-7 | Clinic | (Hatamzadeh, 2010) |
| Sensory integration therapy | SIT providing specific forms of sensory stimulation in the appropriate dosage may improve the nervous system's ability to process sensory stimuli. | improved nervous system and reductions in problem behaviors and more efficient learning. | None | individual | 18 session (45 Min)/Week | Therapist, Teacher | 4-11 | Clinic | (Lang, 2012) |
| Art Therapy | the creative process of art making is healing and life enhancing and is a form of nonverbal communication of thoughts and feelings | AT can be helpful in skill generalization and social skills development. More over AT improve cognitive processing and problem solving. In more details it has positive impact on Cooperation, Assertion, Responsibility, Self-control. | None | Individual, group | Weekly, 8 months | Therapist | 11-18 | Clinic | (Malchiodi, 2003) (Epp, 2008) |
| Speech language therapy | offers numerous approaches to improve a child's language, which is one of the primary problems in autistic disorder. | autism improve their general ability to communicate and interact with others effectively, as well as develop their speech and language skills. | None | Individual | 7 weeks | Therapist | 4-18 | Clinic | (Goldstein, 2011) (Turkington, 2007) (Merrison, 2005) (Jamie B. Schwartz, 2006) |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Occupational therapy | A type of treatment that can help improve the sensory needs of children with autism, who often have lessened or heightened sensitivity to sound, sight, smell, touch, and taste. | to improve problems with fine motor and adaptive skills, such as dressing, eating, and writing, that are sometimes seen in children with autistic disorder. it helps with the behavioral, and cognitive issues typical fragile X syndrome. | None | Individual, group | 10 weeks | therapist | 2-12 | Clinic, Home, School | (Turkington , 2007)<br><br>(Patel, 2014) |
| Robot therapy | using robot as tool for learning a practicing a target skills and behavior, social skills in interactive environment. | improving social skills in individuals with ASD. robot could be used to elicit perseverative speech ,stereo type and repetitive behaviors. | None | Individual, group | None | therapist | 4-11 | Clinic | (Diehl, 2012) |
| LEGO therapy | LEGO therapy is a social skills intervention for school-age children based around collaborative LEGO play. using the child's natural interests to motivate learning and behavior change. | improve the communication and socialization skills. | None | Individual, group | 18 weeks | therapist | 6-11 | Clinic | (Owens, 2008) |
| Theatre Therapy | designed to target the socioemotional challenges of autism by utilizing well-established behavioral intervention paradigms implemented in combination with theatrical techniques. | improve the socioemotional functioning of children with autism. | Social Emotional NeuroScience Endocrinology (SENSE) | group | 3 months | therapist | 6-17 | Clinic | (Corbett, 2011) |
| Dance & Movement Therapy | Dance/movement therapy is a holistic form of therapy, built on the foundation that human reception, processing and response inextricably link the mind and body into a functional whole. | providing physical, social, and cognitive benefits to participants. DMT has intervention in Regulation behavior variability, Imitation disorder, Instict disorder, Emotional disorder and social interaction. | None | group | 2 months | therapist | 4-9 | Clinic | (Scharoun, 2014) (Mateos-Moreno, 2013) |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Craniosacral therapy | CT is a gentle, hands-on method of evaluating and enhancing the functioning of a physiological body system called the craniosacral system - comprised of the membranes and cerebrospinal fluid that surround and protect the brain and spinal cord. | decreasing structural stress and strain on their central nervous system. It has reduction in self injury behavior and hyperactivities as well, and increase in communication. | None | Individual | None | therapist | 5-12 | Clinic | (Hollander, 2003) (Green, 1999) (Upledger, 2001) (Kern, 2011) |
| Holding therapy | The parent tries to make contact with the child by forced holding, a method devised by Columbia University psychiatrist Marth Welch. the parent tries to establish eye contact and share feelings verbally with the child. | Decreasing the aggressive behaviors and delinquency. | None | Individual | 2 weeks | therapist | 5-14 | Clinic | (Myeroff, 1999) (Turkington, 2007) |
| Physical therapy | Physical therapy involves the treatment and prevention of physical disorders or injuries that cause a person to lose normal mobility, strength, range of motion, or quality of their physical functioning. | Improve basic motor skills like sitting, rolling, standing, and playing for children with ASD. Affect on playing skills and their social engagement with other children. | Swimming therapy | Individual | 2 weeks | therapist | 7-12 | Clinic | (Yilmaz, 2004) (Pan, 2011) (Turkington , 2007) |

*Table 3. Extracted properties for therapies classes.*

### 3.2.1.2.2 Biomedical class

Biomedical is the second subclass for the treatment class where dietary treatment class is one of the main subclasses for biomedical class. This type of treatment helps children and adults with autism and Asperger's. Biomedical treatments will not help all autistic people but it has been proven that they have helped most individuals with ASD (Adams, 2007). Furthermore, biomedical interventions are the safe approaches for treating the under issues of ASD inside the body (Lyons, 2015). Moreover, many autism symptoms in behavioral difficulties are treatable and can improve through the proper biomedical treatments (Lyons, 2015). Biomedical class contains 7 classes and two of the classes have their own subclasses. The immune system regulation class has 3 subclasses and the dietary class has 14 subclasses.

The dietary class helps children with ASD who have food sensitivities, due to abnormalities in their immune system or digestive system. Dietary intervention aids problems such as choking/wheezing, diarrhea, vomiting, dizziness/feeling faint, or even severe reactions such as anaphylaxis in ASD individuals (Adams J. , 2013).

| Class | Intervention |
|---|---|
| Biomedical | Vitamin mineral supplements |
| Biomedical | Essential fatty acids |
| Biomedical | Immune systems regulation |
| Immune systems regulation | Actos |
| Immune systems regulation | Intravenous immunoglobulin |
| Immune systems regulation | Low dose naltrexone |
| Biomedical | Melatonin |
| Biomedical | Chelation |
| Biomedical | Glutathione |
| Biomedical | Thyroid supplementation |
| Biomedical | Amino acids |

| Biomedical | Dietary |
|---|---|
| Dietary | Antifungal |
| Dietary | Antioxidant |
| Dietary | Body ecology |
| Dietary | Candida |
| Dietary | Elimination |
| Dietary | Feingold |
| Dietary | Gluten free and Casein free |
| Dietary | Ketogenic |
| Dietary | Low glutamate |
| Dietary | Low oxalate |
| Dietary | Low phenol |
| Dietary | Low phenylalanine |
| Dietary | Rotation |
| Dietary | Specific carbohydrate |

*Table 4. Subclasses of biomedical class with their related classes.*

The subclasses of biomedical classes are extracted from 31 different sources within the field of biomedical and dietary intervention of autism spectrum disorders. Additionally, individual properties for each biomedical intervention such as Info, Type, Effect, Duration, and Sources have been extracted from targeted sources. Each property of biomedical treatment provides a clear side of the treatment for users. For instance, individual property for essential fatty acids is as follows:

- Essential fatty acids
    - Info: Using more essential fatty acids like Omega 3 and Omega 6 food sources supplements.
    - Type: food, supplement.
    - Effect: Depression, gut function, bipolar, increase the language and learning skills
    - Duration: 3-9 months
    - Sources: (Adams J. B., 2007), (Meiri, 2009), (Gilbert, 2008).

| Intervention | Info | Category | Effect | Duration | Sources |
|---|---|---|---|---|---|
| Vitamin Mineral Supplements | Using more vitamin, minerals food sources and supplements. | 1. Juicing<br>making fresh vegetable/fruit juice<br><br>2. Supplements<br>Using Vitamin/mineral supplements | Treatments for guts problem, sleep issues, improve the health and behavior. | 3 months | (Adams J. B., 2015)<br><br>(Adams J. B., 2005)<br><br>(Adams J. B., Summary of biomedical treatments for autism, 2007) |
| Essential fatty acids | Using more essential fatty acids like Omega 3 and Omega 6 food sources supplements. | 1. Food:<br>Fish<br>Flex seed oil<br>Cod liver oil<br><br>2. Supplements<br>Two of the major omega 3 fatty acids are EPA and DHA | Depression, gut function, bipolar, increase the language and learning skills | 3-9 months | (Meiri, Omega 3 fatty acid treatment in autism, 2009)<br><br>(Amminger, 2007)<br><br>(Adams J. B., Summary of biomedical treatments for autism, 2007) |
| Immune System Regulation | Treatment for abnormal immune system in autism | 1. IVIG<br>2. ACTOS<br>3. Low-dose naltrexone | Aberrant behaviors, speech improvement, hyperactivity, Social interaction improvement, agitation, irritability, temper tantrums, social withdrawal, and stereotyped behaviors. | 3-4 months | (Boris M. A., 2005)<br><br>(Gupta, 1996) |
| IVIG | Subcategory of Immune system regulation | None | Aberrant behaviors, speech improvement, hyperactivity, Social interaction improvement | 6 months | (Gupta, 1996)<br><br>(Boris M. A., 2005) |
| ACTOS | Subcategory of Immune system regulation | None | Improvements in irritability, lethargy, stereotype, hyper activities | 3-4 months | (Boris M. C., 2007) |
| Low-dose naltrexone | Subcategory of Immune system regulation | None | attenuate hyperactivity, agitation, irritability, temper tantrums, social withdrawal, and stereotyped behaviors. | 3-4 months | (ElChaar, 2006) |
| Melatonin | Using Melatonin for sleeping problems | None | sleep problems, including falling asleep, nighttime waking, and early waking | None | (Melke, 2008)<br><br>(Adams J. B., Summary of biomedical treatments for autism, 2007) |

| | | | | | |
|---|---|---|---|---|---|
| Thyroid Supplementation | Using thyroid supplement for poor thyroids functioning and sleep problems | None | Treatment for low iodine and poor thyroid function, weight loss | 1-2 months | (Adams J. B., Summary of biomedical treatments for autism, 2007)<br><br>(Adams J. B., 2006) |
| Amino Acids | Involving sufficient amount of protein and amino acids supplements into child's diet | 1. Food<br>2. Supplements | Treatment for low protein and amino acids level.<br>Immune response, weakness, fatigue<br>Stress, Anxiety | None | (Adams J. B., Summary of biomedical treatments for autism, 2007)<br><br>(Tirouvanziam, 2012) |
| Glutathione | Help to bring the active glutathione to sufficient level and normalizing the Glutathione | 1.Supplements:<br>Oral glutathione<br>IV gluthatione<br>Vitamin C<br>TMG<br>DMSA | Protect body from to toxins and toxic metals | None | (Adams J. B., Summary of biomedical treatments for autism, 2007)<br><br>(Adams J. B., 2009) |
| Chelation | Help to bring the active glutathione to sufficient level and normalizing the Glutathione. | DSMA<br>DMPS<br>TTFD | Protect body from to toxins and toxic metals | None | (Adams J. B., 2009)<br><br>(Bradstreet, 2003)<br><br>(Adams J. B., Summary of biomedical treatments for autism, 2007) |

*Table 5. Extracted properties for biomedical intervention.*

| Dietary | Info | Effect | Side effect | Duration | Sources |
|---|---|---|---|---|---|
| **Gluten Free Casein Free** | Expose Your Child to Gluten- and Casein-Free Foods. | Repetitive language subscale, Aggression, Decreased the Hyperactivity and tantrum | Decreased Appetite, Irregular bowel | 3 months | (Johnson, 2011)<br><br>(Elder, 2006) |
| **Elimination** | Eliminates many refined and processed foods, while encouraging a natural, healthy and whole food diet. | Behavioral improvement | Nutritional Deficiencies | 3 months | (Lucarelli, 1995) |
| **Specific Carbohydrate Diet** | The SCD is based on the theory that single sugar unit carbohydrates (monosaccharides) are easily digested and well absorbed whereas carbohydrates containing two or more sugar units (disaccharides and polysaccharides) are difficult to digest. | Less aggressive behavior, eye contact improvement, task attendance Sleeping problem, improvement of the verbal communication for Gastrointestinal problems, restore the digestive system, Periods of diarrhea, fungal overgrowth. | Weight loss | 1-3 months | (Gotschall, 2004) |
| **Rotation** | The rotation diet is based on the theory that the likelihood your child will have an allergic reaction to a food increases over time with exposure. | Better physical and behavioral reactions, eating problem, improve the immune function | None | 3-12 months | (Slimak, 2003)<br><br>(Mary Beth, 2014) |
| **Antifungal** | The diet eliminates foods that contain yeast and foods that supposedly stimulate the growth of yeast. | Improvement in behavior, cure of guts problem, repetitive and bad behavior. | None | 5-14 days | (Adams J. B., Summary of biomedical treatments for autism, 2007) |

| | | | | | |
|---|---|---|---|---|---|
| **Feingold** | It's based on removal of three types of food adjectives, artificial colors, Synthetic flavors and preservative from diet. | hyperactive behavior, reduce impulsivity, compulsions and emotional concerns, improve the attention, reduce seizures, improve toileting, and can improve sleep. | None | 6-8 weeks | (Brenner, 1977) |
| **Low Oxalate Diet** | Remove the high oxalates food to control the oxalates in body | Reduce bad behavior, Improvements in expressive speech, reduced obsessive behavior, motor skills, expressive speech, better sleep, reduced self-abusive behavior. | None | None | (Shaw, 2006) |
| **Low Glutamate Diet** | A low glutamate or glutamate-aspartate restricted diet is one that restricts intake of glutamate and similar molecular compounds such as aspartate. | Improvement in seizure , Headaches, insomnia, anxiety, mood swings, ADHD-like symptoms, problems regulating our appetite, leaky gut, elevated eosinophils, bedwetting, problems focusing eyes, stimming, seizures | None | None | (Shinohe, 2006) |
| **Low Phenylalanine Diet** | The low phenylalanine diet is designed to allow people with an inherited recessive metabolic defect of phenylalanine metabolism. | Help the children with PKU problem | None | None | (de Baulny, 2007) |

| | | | | | |
|---|---|---|---|---|---|
| **Body Ecology Diet** | The approach is low dairy, low carbohydrate and low sugar so adheres to the basic principles that appear to underpin many of the more successful models. | Helpful for guts problem | None | None | (Srinivasan, 2009) |
| **Low Phenol Diet** | A low phenol diet is one, which restricts intake of highly phenolic foods. | Keep the phenol low | None | None | |
| **Ketogenic diet** | A low carb and high fat diet | Reduce the seizure, better cognition functioning and behavior | Fatigue and dizziness, Hypoglycemia (Low blood sugar), Headaches, Constipation. | 6-12 months | (Freeman, 1998)<br>(Hemingway, 2001)<br>(Hallböök, 2012) |
| **Antioxidant diet** | The recommended food groups are fresh vegetables, fresh fruits, cooked legumes, starchy vegetables, and whole grains. | Reduce the Oxidative stress | None | None | (Chauhan, 2004) |
| **Candida Diet** | Control the Candida Albicans of the body, which is part of the immune system and digestive health. | Yeast treatment | None | None | (Richards, 2013) |

*Table 6. Extracted properties for dietary intervention.*

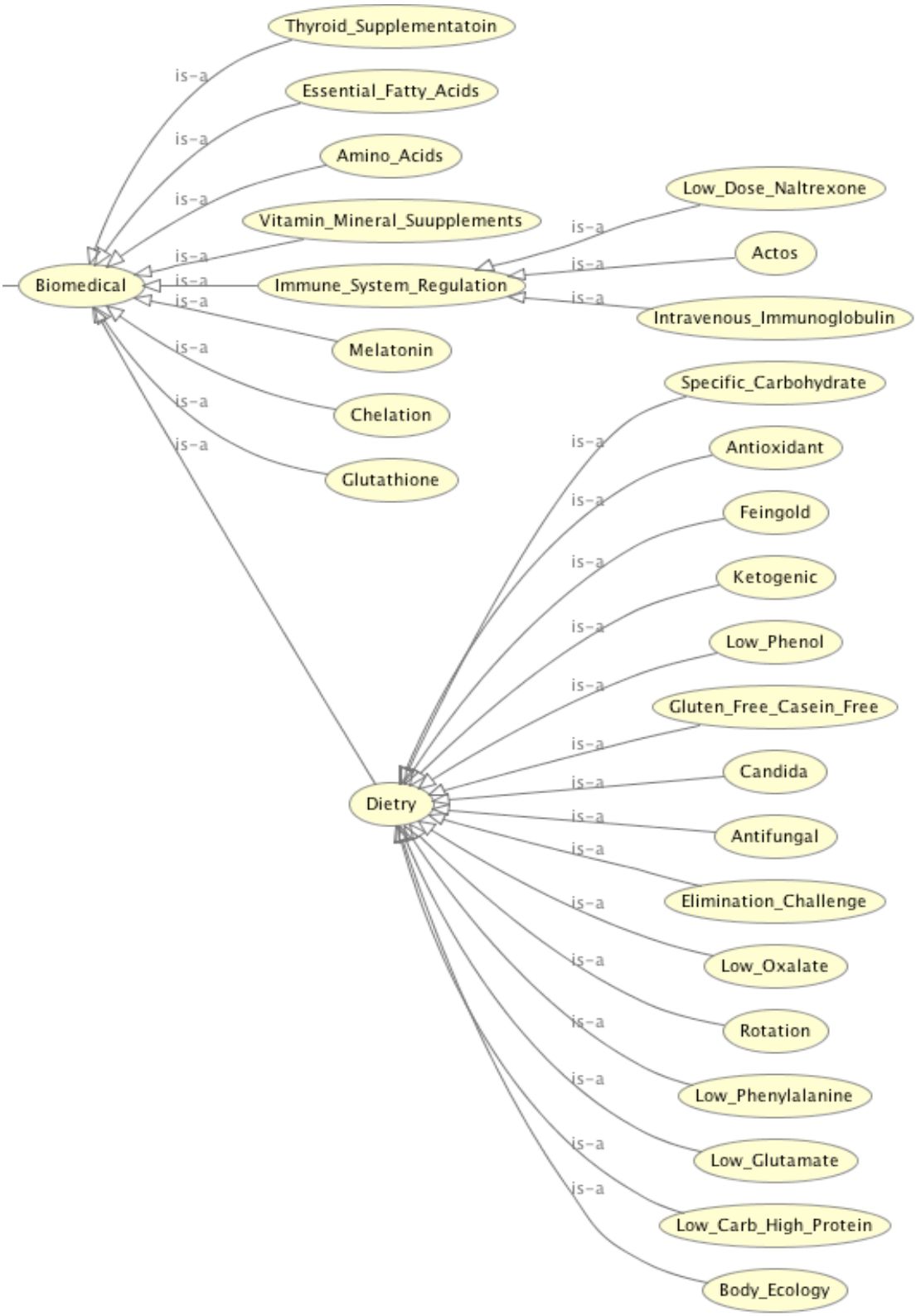


*Figure 23. Biomedical class with subclasses shown in protégé (OWL Viz view).*

The subclasses for biomedical class have been imported into autism spectrum disorders ontology as it is shown in the figure 24. The extracted individual properties are entered as exclusive property for each biomedical treatment and dietary class as they are presented in table 5 and table 6. The properties will provide an easier path for the ASD ontology to define the relations and the effect of treatment in further sections.

### 3.2.1.3 Effect class

The third top level class of the ASD lightweight ontology is the effect class which represents the influence of disorders and biomedical treatment on individuals with ASD. Effect class is divided into two subclasses such as disorder effect and treatment effect. As it was previously discussed, ASD ontology follows a structure where terms are selected from the most commonly used terms in standard textbooks, published articles from high level journals, and relevant domain ontologies in order to obtain the comprehensive ontology as a result of users (Robert Arp, 2015).

Consequently, disorder effect class which is a subclass for effect class is based on an existing ontology of Modeling the Autism Spectrum Disorder Phenotype by Alexa T. McCary et al. 2014. The terms and concepts have been taken from the Modeling the Autism Spectrum Disorder Phenotype ontology to represent the effect of disorders on autistic people in different type difficulties such as social problems and behavioral impairments. Hence, disorders class represents two subclasses such as personal traits and social competence. The treatment effect class builds on the effect property of biomedical treatment which has been extracted from various relevant published articles and textbooks.


*Figure 24. Effect class structure and its two main subclasses.*

Treatment effect class has 15 concepts based on the effect property of interventions which is introduced in the biomedical class. This class defines the influence of represented biomedical

and dietary intervention for people with ASD in biomedical class. Basically, the properties for intervention are extracted manually from the sources to be more accurate and precise in terms of representing the knowledge of the autism spectrum domain. Therefore, the terms and concepts for ontology and possible relations among them will be based on reliable knowledge, solid connections, and standard definitions.

| Effect | Class | Effect | Class |
|---|---|---|---|
| Adherence to rituals and routines | Personal traits | - Impatience | Impulse control and regulation |
| - Insistence on Oder | Adherence to rituals and routines | Language development | Personal traits |
| - Insistence on routine | Adherence to rituals and routines | Motor perseveration | Personal traits |
| - Repetitive actions | Adherence to rituals and routines | Numeracy | Personal traits |
| Anxiety | Personal traits | Pattern recognition and rendering | Personal traits |
| Attention and focus | Personal traits | Preparing for task | Personal traits |
| Awareness and avoidance of danger | Personal traits | Reaction to sensory stimuli | Personal traits |
| Color perception | Personal traits | Reaction to unexpected change | Personal traits |
| Compulsive behavior | Personal traits | Reasoning | Personal traits |
| Control of emotional reactions | Personal traits | Receptive lexicon | Personal traits |
| - Anger control | Control of emotional reactions | Receptive morphology | Personal traits |
| - Emotional outbursts | Control of emotional reactions | Receptive phonology | Personal traits |
| - Managing internal emotion | Control of emotional reactions | Receptive semantics | Personal traits |
| Decision making ability | Personal traits | Receptive syntax | Personal traits |
| Depression | Personal traits | Regression pf language skills | Personal traits |
| Distant or preoccupied affect | Personal traits | Restricted and usual interests | Personal traits |
| Emotional swings | Personal traits | - Intense interests | Restricted and usual interests |
| Expressive lexicon | Personal traits | - Restricted range of interests | Restricted and usual interests |
| Expressive morphology | Personal traits | - Unseal sensory interests | Restricted and usual interests |
| Expressive phenology | Personal traits | Self injury behavior | Personal traits |
| - Prosody | Expressive phenology | Showing emotion | Personal traits |
| - Vocalizations | Expressive phenology | Sorting ability | Personal traits |
| Expressive semantics | Personal traits | Space perception | Personal traits |
| Expressive Syntax | Personal traits | Task performance | Personal traits |
| Facial expression | Personal traits | - Quality of performed task | Task performance |
| Flexibility in problem solving | Personal traits | - Task completion | Task performance |
| Form perception | Personal traits | - Task Initiation | Task performance |
| Gestures | Personal traits | Tics and Mannerisms | Personal traits |
| Imagination | Personal traits | Transitioning ability | Personal traits |
| Impulse control and regulation | Personal traits | Use of free time | Personal traits |
| - Acting before thinking | Impulse control and regulation | Visual Thinking | Personal traits |
| - Immoderate behavior | Impulse control and regulation | | |

| Effect | Class | Effect | Class |
|---|---|---|---|
| Ability to convey information | Social competence | Recognition and responsiveness to voice | Social competence |
| - Ability to convey appropriate level of details | Ability to convey information | Relating to others | Social competence |
| - Ability to convey information | Ability to convey information | Self care | Social competence |
| - Ability to convey feeling | Ability to convey information | - Clothing self | Self care |
| Ability to converse in social setting | Social competence | - Eating ability | Self care |
| - Ability to engage in casual consecration | Ability to converse in social setting | - hygiene | Self care |
| - Using language appropriately | Ability to converse in social setting | - poor eating behavior | Self care |
| Adherence to rules in the community | Social competence | - Toileting | Self care |
| Adherence to rules in the home | Social competence | Sensitivity to conversant | Social competence |
| Aggressive behavior | Social competence | Social dependence on others | Social competence |
| Awareness of harmful social situation | Social competence | Understanding others expectations | Social competence |
| Awareness of social and personal space | Social competence | Understanding the context | Social competence |
| Destructive behavior | Social competence | - Skills in understanding hummer | Understanding |
| Disobedience | Social competence | - Skills in understanding non literal meaning | Understanding |
| Empathy | Social competence | Under sanding the effect the ones actions | Social competence |
| Engagement in social activities | Social competence | Use od everyday household item | Social competence |
| - Participation in athletic activities | Engagement in social activities | | |
| - Participation in play activities | Engagement in social activities | | |
| Engaging in social conversation | Social competence | | |
| Eye contact | Social competence | | |
| Imitation | Social competence | | |
| Initiating and responding to social overture | Social competence | | |
| Intentional cruelty | Social competence | | |
| Interactions with friends and family | Social competence | | |
| - Engaging in social play | Interactions with friends and family | | |
| - Poor treatment by others in social setting | Interactions with friends and family | | |
| - sharing | Interactions with friends and family | | |
| - sharing in interests of others | Interactions with friends and family | | |
| Job performance | Social competence | | |
| Joint attention | Social competence | | |
| Keeps to self | Social competence | | |

| | | | |
|---|---|---|---|
| Lack of appropriate guilt | Social competence | | |
| Maintenance and organization of possess | Social competence | | |
| Money management | Social competence | | |
| Performance in school | Social competence | | |
| Performance of household tasks | Social competence | | |
| - Performance of chores | Performance of household tasks | | |
| - Preparation of food | Performance of household tasks | | |

*Table 7. Subclasses of the effect class with their related classes.*

| Effect | Class |
|---|---|
| Body toxins problems | Treatment effect |
| Chemical sensitivities | Treatment effect |
| - Phenol | Chemical sensitivities |
| - phenylalanine | Chemical sensitivities |
| Constipation | Treatment effect |
| Diarrhea | Treatment effect |
| Food allergies | Treatment effect |
| Gastrointestinal problems | Treatment effect |
| - fungal overgrowth | Gastrointestinal problems |
| - Yeast problems | Gastrointestinal problems |
| Headaches | Treatment effect |
| Hyperactivities | Treatment effect |
| Immune function | Treatment effect |
| Nutritional deficiencies | Treatment effect |
| - Protein | Nutritional deficiencies |
| - Vitamin | Nutritional deficiencies |
| Oxidative stress | Treatment effect |
| Poor thyroid function | Treatment effect |
| Sleeping problem | Treatment effect |
| Weight loss | Treatment effect |

*Table 8. Subclasses of treatment effect class.*

## 3.3 Conceptual model (relation)

As it has been noted in previous sections, terms and concepts of ASD ontology have been gathered and imported into protégé. However, obtaining definitions of terms and concepts of ontology representing universals and defining the classes of autism spectrum disorders ontology is technically not sufficient to capture all of the important information about the ASD domain. The relations between and among the classes need to be distinct and the ontology should represent how specific categories are related to each other. In such cases like autism spectrum disorders ontology that contains multiple ontologies, the corresponding terms are connected together through relations and through corresponding definitions and axioms which need to be defined (Robert Arp, 2015). For instance, treatment class in ASD ontology can be a separated ontology domain however terms and classes of treatment class are related to disorders and effect class that need to be determined in ASD ontology.

The principle of single inheritance in ASD ontology and mentioned definitions drawn in earlier sections are on the central architectural role of the ‘is-a’ relation in ASD ontology construction. For obtaining a comprehensive design for the ontology, there are three basic kinds of relations to be followed and taken into account of the structure of the ontology (Robert Arp, 2015). This helps the ontology for defining the relations that it will represent. The three basic kinds of relations are stated as:

- Relations holding between one universal and another (the relations represented in the ontology itself).
- Relations holding between one particular and another - for example, when asserting that Mary's leg is a continuant part of Mary.
- Relations holding between a particular and a universal - for example, the relationship of instantiation.

These three kinds of relations in the ontology allow the user to use ontology coherently and in conjunction with presented information in the ontology about particulars in the world and to reason about those particulars (Robert Arp, 2015).

The first relation in the autism spectrum disorders ontology, represented in the ASD ontology itself, shows the natural relations among the main top level classes of ontology which represent the definition of the domain knowledge. The first kind of relation is an overall relation which defines the autism spectrum disorder knowledge through the term of the ontology. This relation defines that autism spectrum disorders as ASD contains various disorders for autistic people and disorders have several effects on them which influence their activities in personal and social life. However, there are many interventions for these disorders which can help individuals with ASD enhance their quality of life.

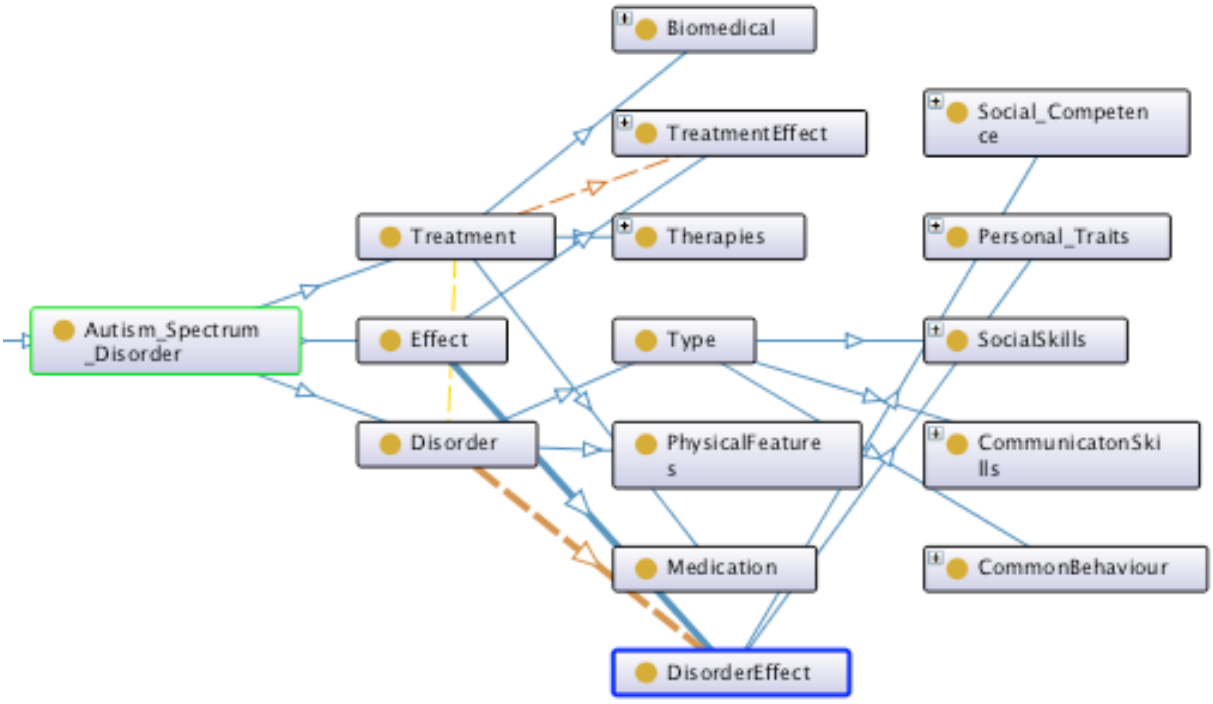


*Figure 25. Relationship among Domain Knowledge ASD ontology Classes.*

The second kind of relation represents the relations between one particular and another particular of classes in the ASD ontology. The relations between the class and the specific subclass of that

class are represented in the hierarchy is-a relation form. For instance, executive function is one of the common behavior disorders in the disorder class. The executive function class has 5 subclasses which these subclasses can represent this class as difficulties such as emotional regulation and control, planning, response inhabitation, working memory, and mental flexibility, all of which are executive function disorders in autism spectrum disorders. The relation between the executive function class with the subclasses reveals the knowledge of the class. This kind of relation in ASD ontology is the second type of relation which is based on the principle of ontology relations by Robert Arp et al. 2015 from Massachusetts institute of technology. The majority of the type of relations have been discussed in earlier sections.

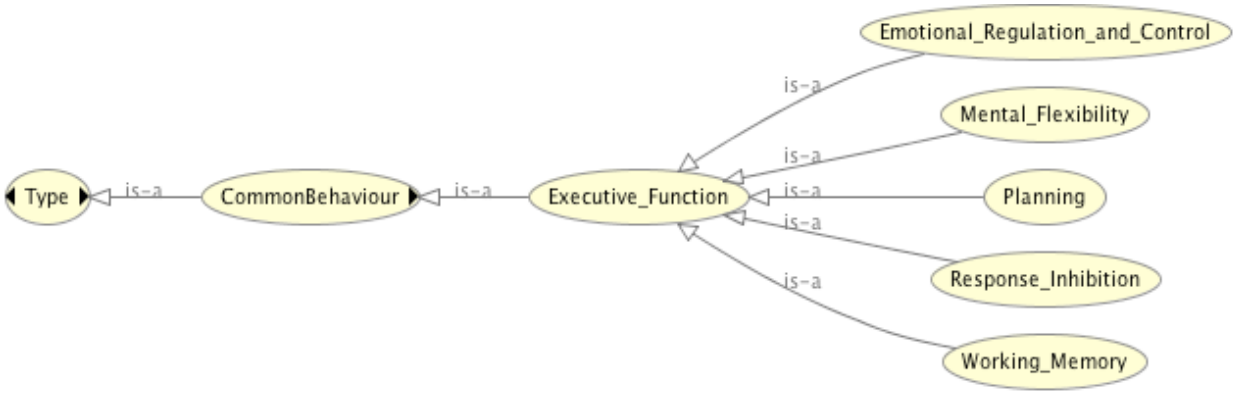


*Figure 26. Second type of relation between the executive function class with the subclasses.*

The third kind of relations has a significant role within the ontology. The third relation reveals the connections between the specific categories and relations among the instances and lower classes in autism spectrum disorders ontology. For instance, there are various relations among the lower classes of disorder class, effect class, and treatment class within the created ontology which indicate the relations between the different knowledge in the domain. Hence there are a number of properties that have been introduced in object properties of protégé for ASD ontology to provide the ability for representing the relations among the different classes with different roots. Furthermore, object properties have two main classes based on the top level classes of the ontology such as treatment class and disorder class. Disorder ingredient and Treatment ingredient are two main object properties for ASD ontology as it is shown in figure 27.

The disorder ingredient object properties represent the possible relations of disorder class with other top level classes such as treatment and effect class. There are *HasDisodersEffect, HasTreatment,* and *HasPhysicalFeature* as three subclasses for disorder ingredient.

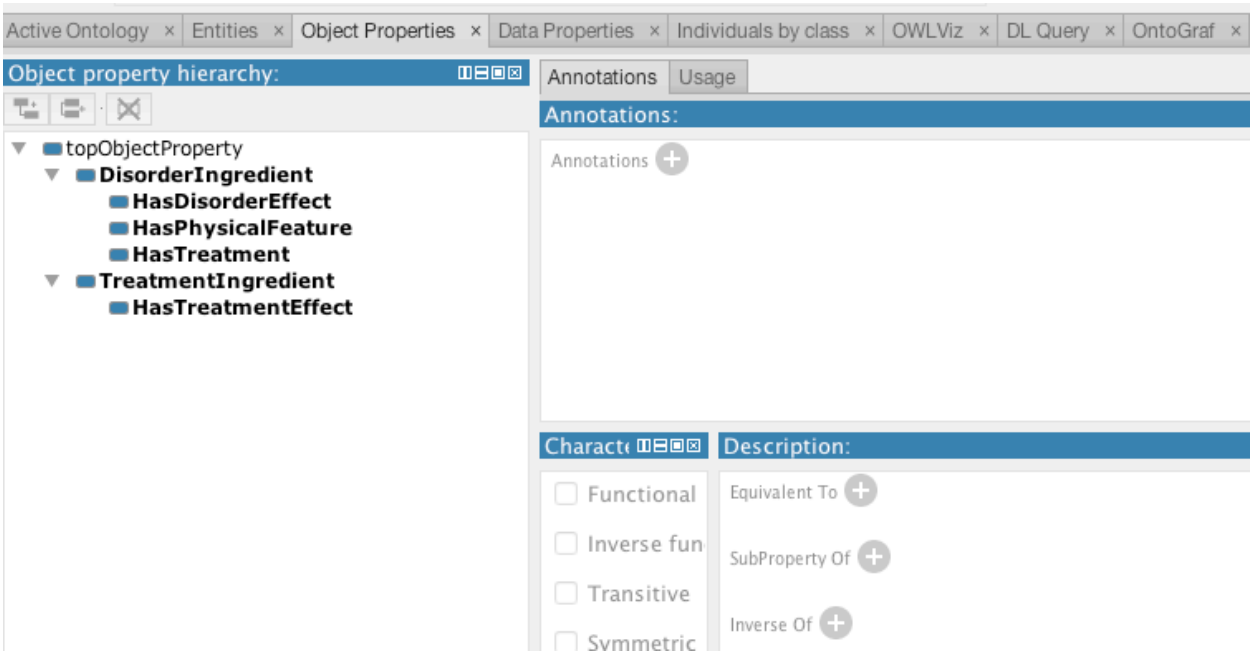


*Figure 27. Defined object properties for ASD ontology in protégé.*

Treatment ingredient contains *HasTretmentEffect* as the only subclass for this object property. This object property defines the relations of treatment class with other top level classes of ASD ontology. For instance, to represent the effect of biomedical intervention of the treatment class which introduces a relation between the treatment class and effect class*, HasTreatmentEffect* object property will be used for the mentioned purpose.

As it has been discussed in the initial design section, there is a solid relation among the top level classes. Thus, the relations among the subclasses of disorder, treatment, and effect class are considered as the third kind of relations. The following are the three categories of relations among the top level classes:

- Relation between subclasses of disorder class with effect class and vice versa
- Relations between subclasses of disorder class with treatment class and vice versa
- Relations between subclasses of treatment class with effect class and vice versa

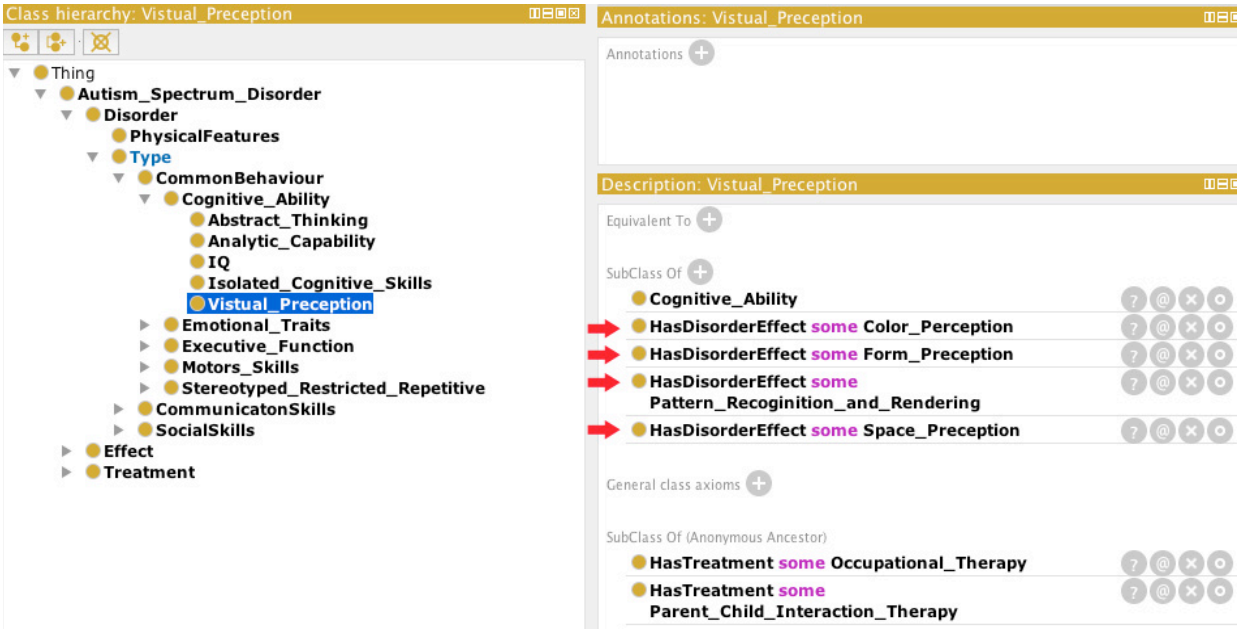


*Figure 28. Third kind of relations for Virtual_perception class with the effect class.*

Figure 29 is an example of the third kind of relation between the subclasses of disorder class with effect class. As it is shown in the figure above, *Virtual_perception* is one of the subclasses of *Cognitive_Abilities* with the root class of disorder. The *Virtual_perception* class is the cause of four effects on autistic people such as difficulties in *Color_preception, Form_Preception, Patten_Recogenition_and_Rendering*, and *Space_Preception.* These relations with effect class are shown with a red arrow in the figure 29. The object property which has been used for defining the relation of disorders with effects is HasDisoderEffect. The same procedure has been used for all of the subclasses of the disorder class in order to define all the possible relations with the effect class.

The third kind of relation between the subclasses of disorder class with treatment class is shown in figure 30. There are three intervention methods which have been displayed for *Abstract_Thinking* disorder based on the relation of this type of disorder with treatment class. However, the relation of *Abstract_Thinking* class with treatment class is represented via HasTreatment object property. This class has three relations with subclasses of treatment class such as *Low_oxlate* in dietary class, *Occupational_therapy*, and

*Parent_child_interaction_therapy* in therapies class as it is shown with red arrows in the figure below.

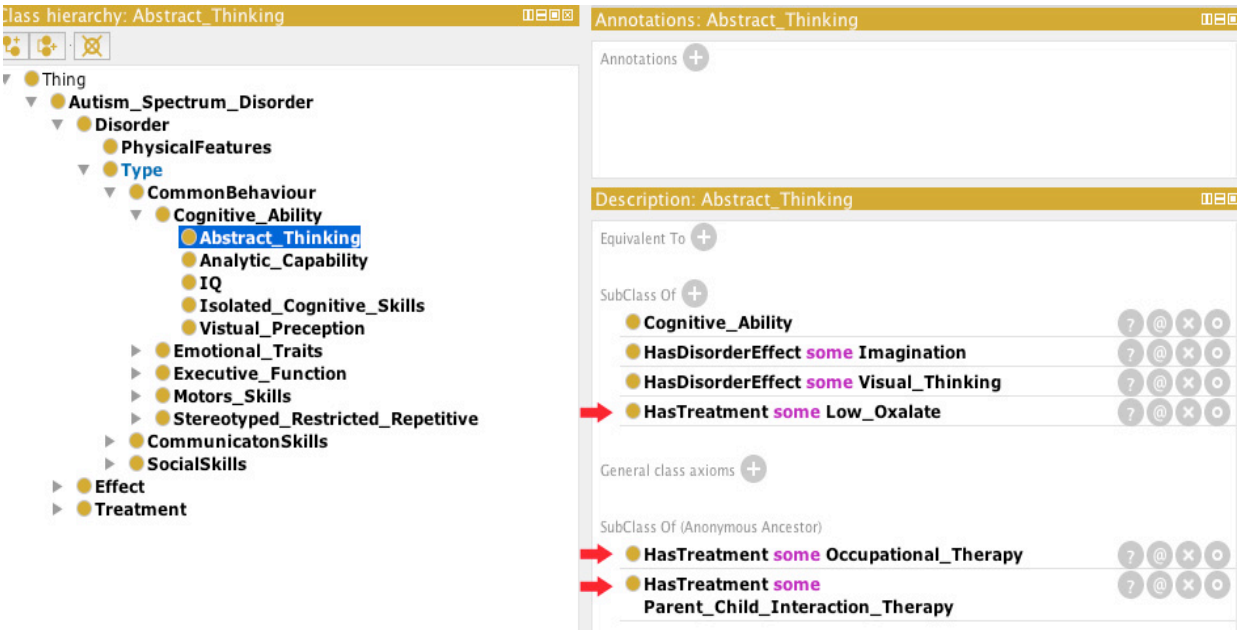


*Figure 29. Third kind of relations for Abstract_Thinking shown with red arrows.*

The relation between subclasses of treatment class with effect class has been shown in the Figure 30. In this example, the influence of using *Vitamin_Mineral_Supplement* for people with ASD is represented through the third kind of relation between the *Vitamin_Mineral_Supplement* and effect class. The object property in use for this type of relation among these two classes is HasTreatmentEffect. The concept of *Vitamin_Mineral_Supplement* subclass has a relation with subclasses of effect class such as *Gastrointestinal_Problems* and *Sleeping_problems*.

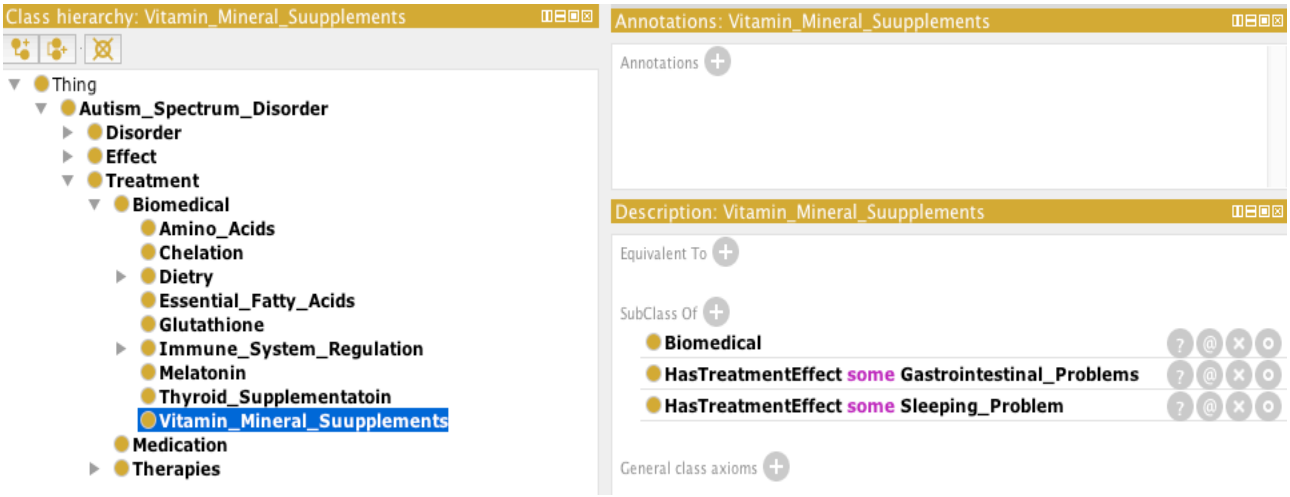


*Figure 30. Third kind of relations for Vitamin_Mineral_Supplement class with the effect class.*

The ASD lightweight ontology has been converted into a comprehensive and heavy ontology after mapping the subclasses to the top level classes and enriching the concepts. The relationship among the imported classes has been defined to support the ontology for representing the domain knowledge and operational knowledge in autism spectrum disorders field.

# Chapter 4

# The Query System

In the pervious chapter the terms and concepts of the autism spectrum disorders ontology have been extracted from reliable sources such as standard textbook, published articles in the field and clinical studies. The classes and their property have been manifested step by step for every top-level classes of the lightweight ontology to build a comprehensive ontology in different domains of autism spectrum disorders such as treatment and disorders. Moreover, the three kinds of different relations in the ASD ontology have been obtained among the classes to complete the steps for developing the ASD ontology.

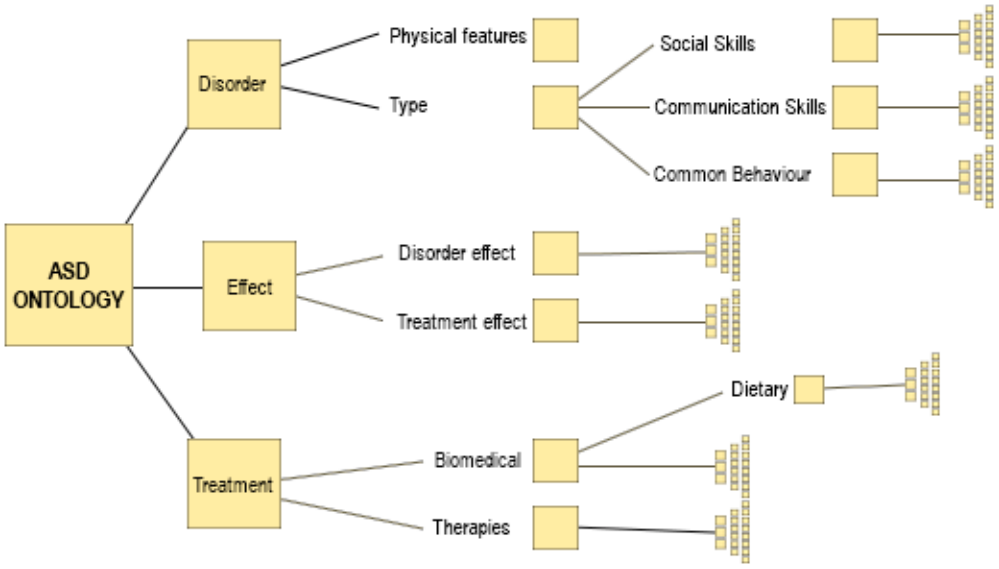


*Figure 31. Complete overview of ASD ontology shown in protégé (OntoGraph).*

The created ASD ontology is ready to be deployed in many specialized applications and to be used by experts for different purposes in the field of Autism disorders. However, the Query

System has been built as part of the thesis to allow the general public extract general and specific information related to Autism. Moreover, the query system provides many other benefits for normal users and researchers. These are discussed below in the chapter. The structure of the Query System along with the tools developed in this research work are presented in the following sections.

## 4.1 System implementation

The Query System of ASD ontology is built on the concept of web based information system. The web based information system delivers services to users online through the Web. For instance, the Query System for ASD ontology provides answers about different aspects of autism spectrum disorders and related them to other more specific aspects of autism disorder disease. To our best knowledge, this is a unique feature that is not available in other developed studies. This section discusses the implementation of the ASD Query System and its structure.

The structure of ASD Query System consists of three parts: database, back-end and the front-end. The most important component is the database, which houses all ASD ontology knowledge. The front-end component allows users interaction while the back-end is responsible for retrieving relative information from the database. The back-end of the ASD Query System has been built using PHP and the front-end is implemented using HTML and CSS.

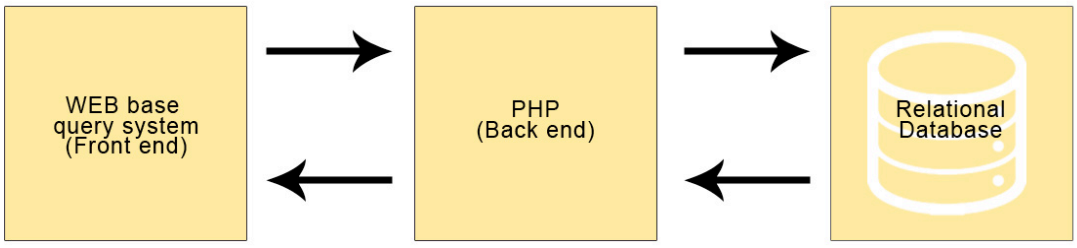


*Figure 32. Structure of the ASD Query System.*

The main parts of the ASD Query System architecture is shown in the figure 33. The relational database contains information of autism spectrum disorders ontology in the form of tables. Technically, users are interacting with front-end part of the system to query needed information. Once user queries are submitted, these passed to the back end for further processing. The back-end structure is based on PHP script to establish a connection with the relational database. Back-end scripts parse user queries; resolve business logic; submits relational queries to the database; and fetch the information from the database. Once the information has been received by the back-end component, it is sent to the front-end for display. The back-end component can be considered as a bridge between the front-end and the database of the Query System.

### 4.1.1 System Design

The database of ASD Query System has a key role for housing and accessing ontology knowledge. The knowledge is organized in tables and related in a meaningful way to allow for access in different logical orders (S. Sumathi, 2007). The ASD Query System uses MySQL for database management system (DBMS). In order to handle the database management system for MySQL over the web, phpMyAdmin platform has been deployed.

The heart of any relational database is the table, which is made by set of related data organized in column/row structure. However, the important characteristic of a relational database is to allow data sets in the tables to associate with each other in significant ways to ensure the integrity of normalized data (Robert Sheldon, 2005). There are three fundamental types of relationships in relational databases: one-to-one relationships, one-to-many and many-to-many relationships (Robert Sheldon, 2005).

Authors

| AuthID | AuthFN | AuthMN | AuthLN |
|---|---|---|---|
| 1001 | Edith | NULL | Wharton |
| 1002 | Kate | NULL | Chopin |
| 1003 | T. | S. | Eliot |
| 1004 | Joyce | Carol | Oates |
| 1005 | Samuel | L. | Clemens |

AuthorsBios

| AuthID | Born | Died |
|---|---|---|
| 1001 | 1862 | 1937 |
| 1002 | 1850 | 1904 |
| 1003 | 1888 | 1965 |
| 1004 | 1938 | NULL |
| 1005 | 1835 | 1910 |

*Figure 33. One-to-one relationship between the authors and authors bios tables.*

The autism spectrum disorder ontology needs to be represented in the ASD Query System in order for autism knowledge to be extracted by humans and computational agents. The latter can directly interact with the database. Ontologies are human readable, comprehensive, sharable and formal. They are expressed in a language that has well-defined semantics. Therefore, the first step is to map the ASD Ontology semantics into a meaningful relational schema, which can then be populated with ontology instances. The structure of the resulting database should faithfully maintain all logical relationships embedded in the developed ASD ontology. It should also allow efficient and flexible information gathering, persistent storage of the ontology and its subsequent retrieval. To achieve this goal, in this study, we have manually built a special purpose relational database tailored to store and retrieve the developed ASD ontology. In this approach the OWL data is mapped into tables of a relational schema and the queries posed are translated into SQL queries. Relational database systems are very mature and scale very well, and they have the additional advantage that in a relational database, ontology data and the traditional structured data can co-exist making it possible to maintain persistent relational storage of OWL ontologies.

The relationships among classes of *treatment*, *disorder* and *effect* are the main features of the ASD ontology that need to be faithfully imported into the database. First, these three top-level classes of the ASD ontology have been placed into different tables. Then, knowledge underneath of each of these classes is organized in other tables that we refer to below as second and third level tables.


*Figure 34 First table of ASD relational database for disorder class.*

The three second-level classes (i.e. subclasses) of disorder type such as common behavior, social skills and communication skills have been placed into the second table of the database for disorder class as shown in figure 35.

| | | | subcat_id | cat_id | subcategory |
|---|---|---|---|---|---|
| ☐ | Edit | Delete | 1 | 1 | Cognitive Ability |
| ☐ | Edit | Delete | 2 | 1 | Emotional Traits |
| ☐ | Edit | Delete | 3 | 1 | Executive Function |
| ☐ | Edit | Delete | 4 | 1 | Motors Skills |
| ☐ | Edit | Delete | 5 | 1 | Stereotyped Restricted Repetitive |
| ☐ | Edit | Delete | 6 | 3 | Language Ability |
| ☐ | Edit | Delete | 7 | 2 | Adaptive Life Skills |
| ☐ | Edit | Delete | 8 | 2 | Interpersonal Interaction |
| ☐ | Edit | Delete | 9 | 2 | Recognition of social norms |

Check All / Uncheck All *With selected:*

*Figure 35. Second table of ASD relational database for disorder class.*

The second table for disorder class contains all the subclasses of the three type of disorders, for instance cognitive ability, emotional traits, executive function, motor skills and stereotypes restricted repetitive. These are the subclasses of common behavior class. They have been imported into the second table of the database for disorders. Similarly, the third-level table contains all subclasses of the classes in table 2. For instance, the classes abstract thinking, analytics capability, IQ, isolated cognitive skills and visual perception represent the subclasses of cognitive ability and are stored in table 3.

| | subcat_id | subcat2 | effect | treatment |
|---|---|---|---|---|
| Edit Delete | 1 | Abstract thinking | Has disorder effect: Imagination, Visual thinking | Has treatment: Occupational therapy, Parent-child ... |
| Edit Delete | 1 | Analytic capability | *NULL* | *NULL* |
| Edit Delete | 1 | IQ | *NULL* | *NULL* |
| Edit Delete | 1 | Isolated cognitive skills | *NULL* | *NULL* |
| Edit Delete | 1 | Visual perception | *NULL* | *NULL* |
| Edit Delete | 2 | Affect | *NULL* | *NULL* |
| Edit Delete | 2 | Mood | *NULL* | *NULL* |
| Edit Delete | 2 | Self concept | *NULL* | *NULL* |
| Edit Delete | 3 | Emotional regulation and control | *NULL* | *NULL* |
| Edit Delete | 3 | Mental flexibility | *NULL* | *NULL* |
| Edit Delete | 3 | Planning | *NULL* | *NULL* |
| Edit Delete | 3 | Response inhibition | *NULL* | *NULL* |
| Edit Delete | 3 | Working memory | *NULL* | *NULL* |
| Edit Delete | 4 | Fine motor skills | *NULL* | *NULL* |
| Edit Delete | 4 | Functional laterality | *NULL* | *NULL* |
| Edit Delete | 4 | Gross motor skills | *NULL* | *NULL* |
| Edit Delete | 5 | Involuntary behaviours | *NULL* | *NULL* |
| Edit Delete | 5 | Restricted repetitive behaviour | *NULL* | *NULL* |
| Edit Delete | 6 | Development or regression of language skills | *NULL* | *NULL* |
| Edit Delete | 6 | Expressive language | *NULL* | *NULL* |
| Edit Delete | 6 | Idiosyncratic and routinized speech | *NULL* | *NULL* |
| Edit Delete | 6 | Integrated verbal and non verbal communication | *NULL* | *NULL* |

*Figure 36. Third table of ASD relational database for disorder class.*

Accordingly, the classes and subclasses of the ontology are represented in different tables based on the leveling of the classes in the ontology. The top-level classes of the disorder type in the ontology are imported into the first table, then the subclasses of the first table are placed in the second table and the third table carries the subclasses of the second table and so forth. For instance, the third table of the disorder class has four columns consisting of subcat_id, subcat2, effect and treatment. The subcat_id column allows the relationship of the second table with the third table and subcat2 is the name of the subclass which is imported from ontology into second table. The effect column represents the effect of the disorder based on existing knowledge in the ontology and the treatment column links to treatment knowledge of the disorder based on the autism spectrum disorders ontology. The same procedure has been applied for treatment class to be mapped into the ASD database. The above-mentioned mapping process has been applied to the remaining ontology classes to map the whole ontology to the database.

### 4.1.2 Back-end Component

The back-end component of the ASD Web Query System is built using PHP. The latter is one of the most used scripting languages that can run by itself in the command line of any computer with PHP installed. However, PHP alone isn't sufficient to build a query system for the ASD

ontology. SQL, HTML and CSS are used in conjunction with PHP to build the ASD Query system. There are three main tasks that PHP handles for the query system:

- Configure the connection between the query system and database to access imported information in database.
- Provide the drop down menu for users to query various ontology knowledge.
- Enable submission of queries and display of fetched information.

Technically, the back-end connects the front-end (user) to the database and establishes access to the information within the database. The back-end component provides three drop down menus for the disorder class based on the three tables for its subclasses in the database. Each drop down menu depends on the pervious selected menu of the web query system. For instance, once the user clicks on the first dropdown menu, information is loaded from the first table of the database to the first dropdown menu, then, the second dropdown will be based on the subclasses of the selected class from the second disorder table in the database. Once the user selects the second dropdown menu, the back-end component retrieves the information from the third table of disorder in the database for the third dropdown menu based on the previous selection. As its shown in figure 38, when the bottom dropdown menu is selected by the user, the final query is made ready for submission to the system in order to retrieve available information for the selected disorder. The information such as treatment for the selected disorder and effect of that particular disorder are imported in the third table for disorder class, which can be displayed to users when the final query is submitted to the system.

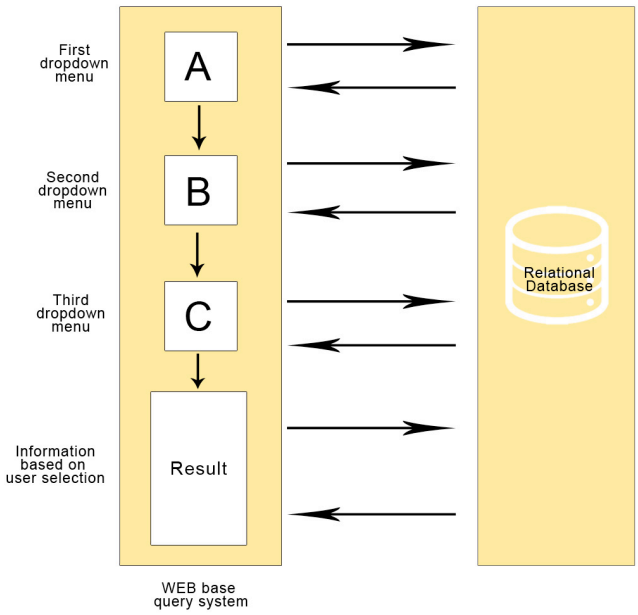


*Figure 37. Process of communication with database for query submission.*

The communication between the ASD web query system and the relational database is shown in figure 38 which enables access to the information for each dropdown menu till submitting the final query for fetching the final result from the ASD database. The drop down strategy has been used for the built query system to demonstrate the knowledge of the classes and subclasses of the autism spectrum ontology. The relationship among the top-level classes of the ontology is established when the final query has been submitted to the system as shown in figure 38.

The users, by selecting the dropdown menus, can browse through the concepts of the ontology. This process allows users to find the query that drives them to the target autism concept(s). For instance, users can select to find out about the intervention and effects for a particular disorder thought the three dropdown menus shown in figure 39.

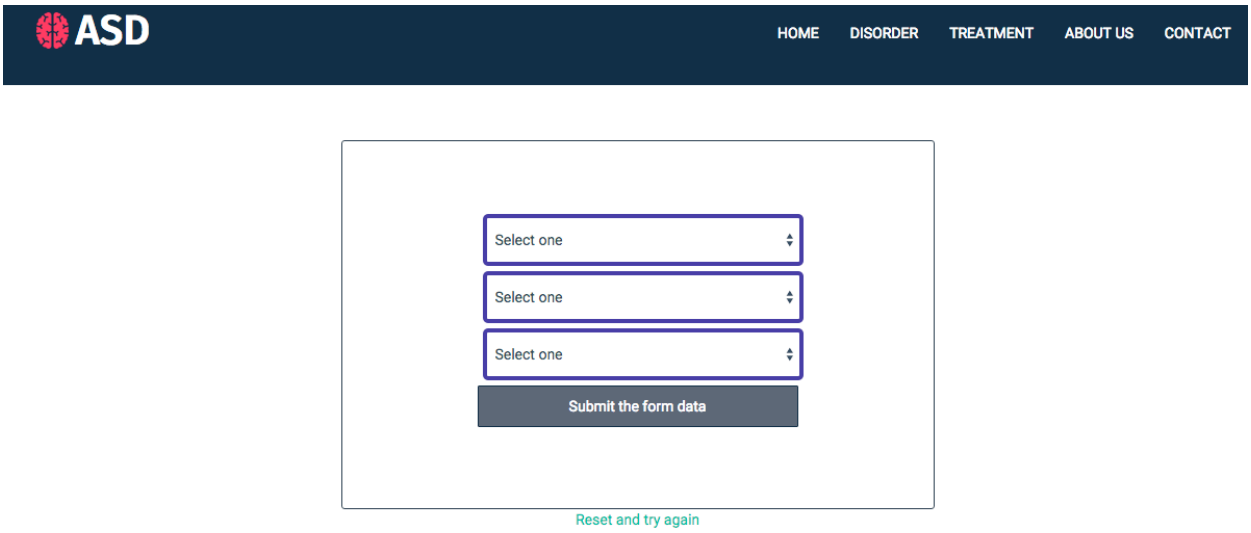


*Figure 38. The three dropdown menus for disorder section.*

The first dropdown menu indicates the top level classes of the disorders class such as: Common behavior, Communication skills and social skills. By selecting one of the disorder types, the query system loads the next menu based on the selected disorder from the database.

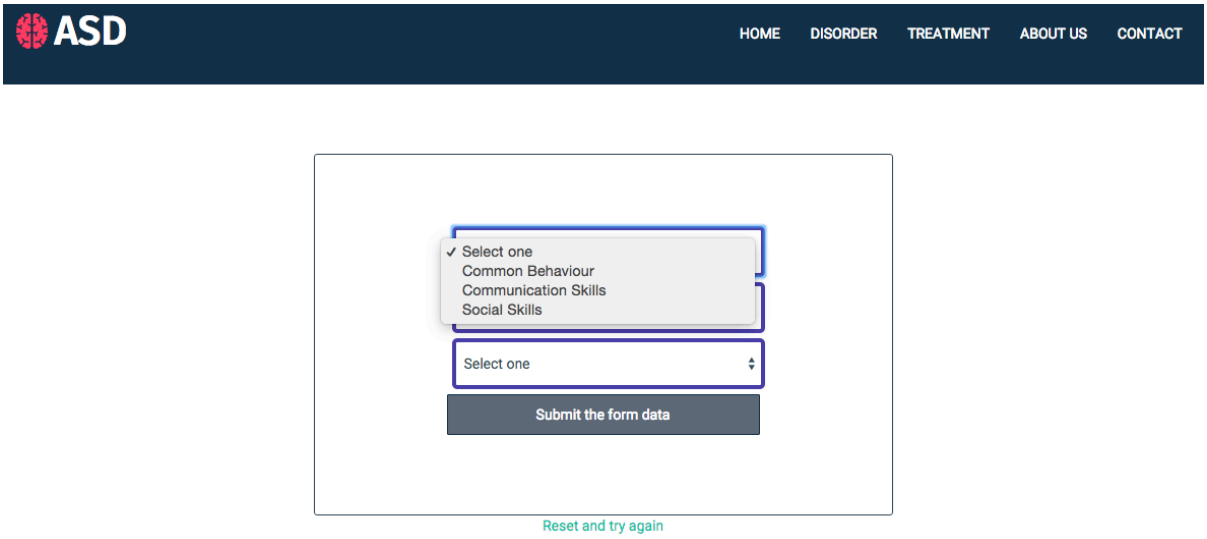


*Figure 39. Selecting the disorder type from first dropdown menu.*

The common behavior is selected by the user as shown in figure 41. The second dropdown shows the subclasses of common behavior based on the ASD ontology from the database.

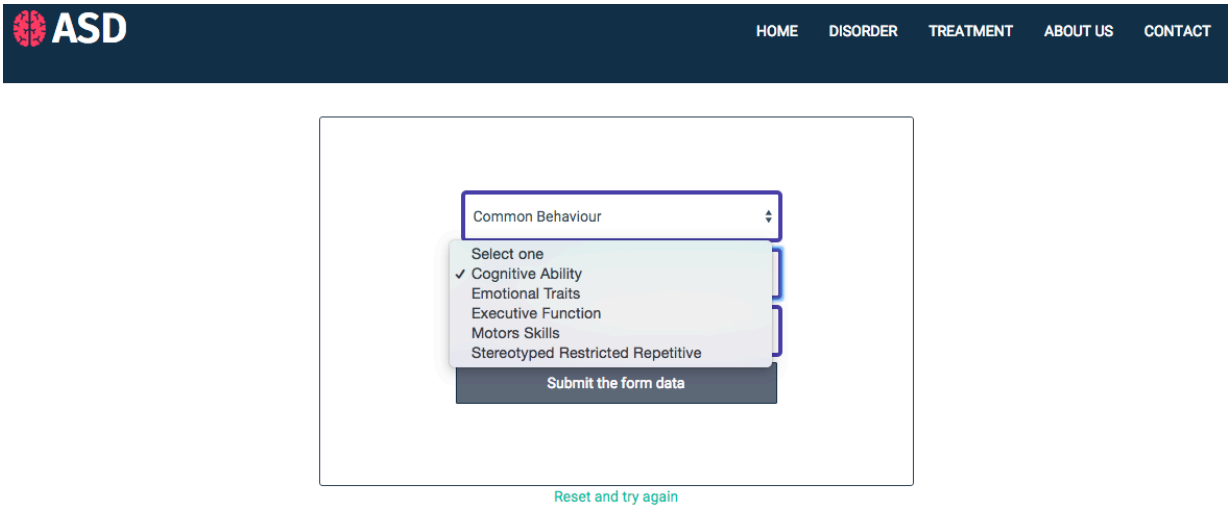


*Figure 40. Selecting the type of Common behviour from second dropdown menu.*

From the second dropdown menu "Cognitive Ability" is selected among the other concepts of Common Behavior as shown in figure 42. The third dropdown menu reveals the concepts for Cognitive Ability as shown in the figure below.

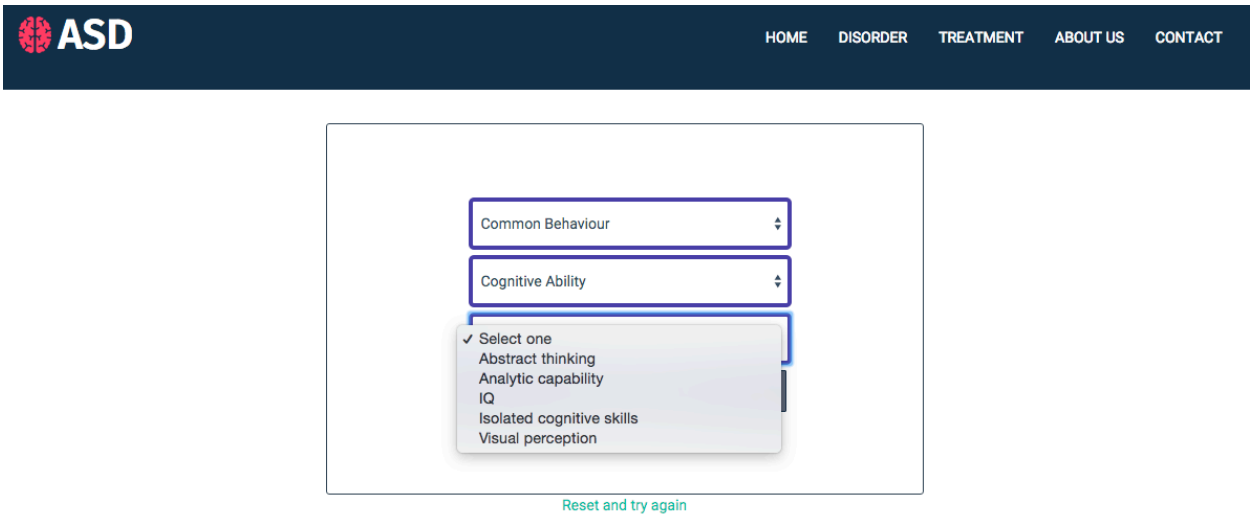


*Figure 41. Selecting the type Cognitive ability from the third dropdown menu.*

Finally, the user selects "Abstract Thinking" and the system provides all relevant ontology knowledge as shown in figure 43.

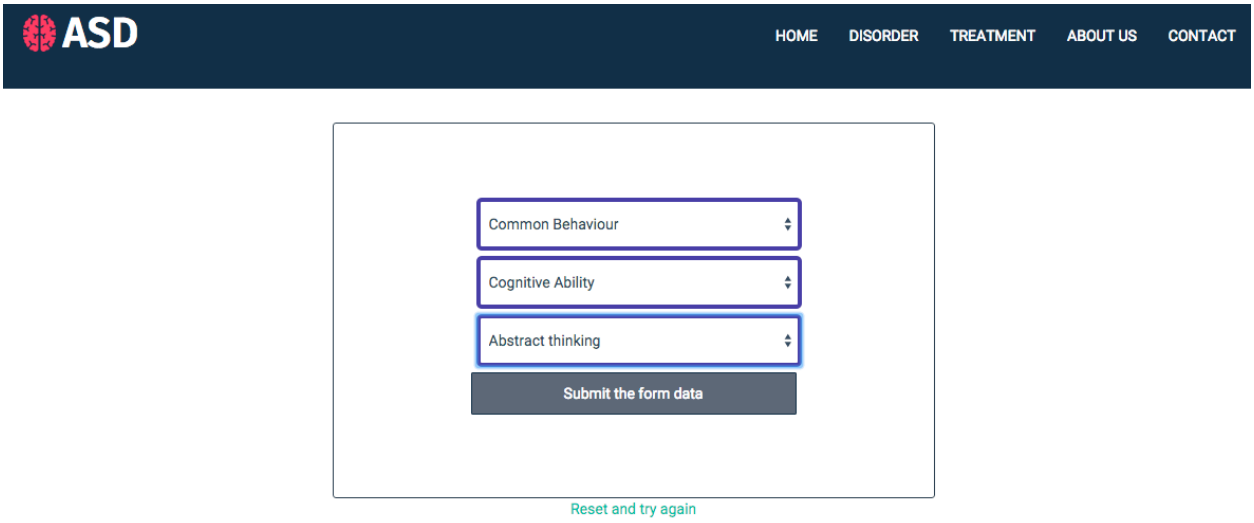


*Figure 42. Selection of all the dropdown menus.*

The figure below shown the results of the submitted query. It provides the effects of abstract thinking on autistic people which are "imagination and visual thinking" and their treatments which are "Occupational therapy, Parent-child interaction and Low oxalate dietary" as shown in figure 44.

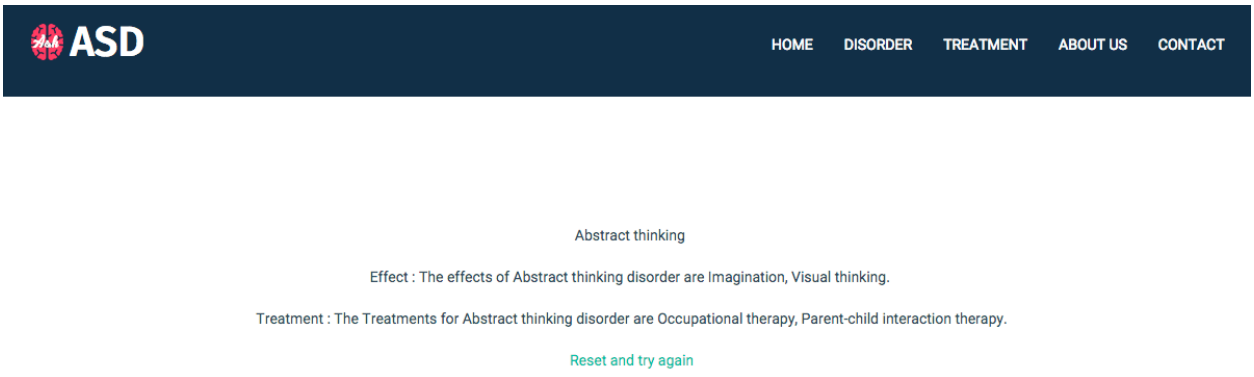


*Figure 43. Information for Abstract thinking displayed to user.*

The web query system also provides the same information via browsing over the treatments for ASD. By choosing any particular intervention method through the treatment section in query

system, users are able to access information such as influence of selected intervention on disorder and their effect in ASD.

## 4.2 Benefits

There are few autism ontologies that have been developed. Most of them have specific focus and purposes, and have been made for target users. So, regular users have no or little access to them. To the best of our knowledge, the ontology developed in this research work is the most comprehensive one. Furthermore, the ontology can be accessed by both expert and normal users. So, the proposed system is of great benefit to experts, researchers and the general public, including family members of autistic people. The latter category will find the proposed system very useful to help support their autistic patients and access technical, clinical and general information about autism.

Users' queries can be submitted by selecting multiple dropdown menus in ASD Query System. The dropdown menus expand the classes and subclasses of the top-level classes of the ontology such as treatment and disorders. Thus, the ASD system provides quick access to the information exported from the ontology from broad to detailed autistic disorders and treatments as illustrated in figure 45.

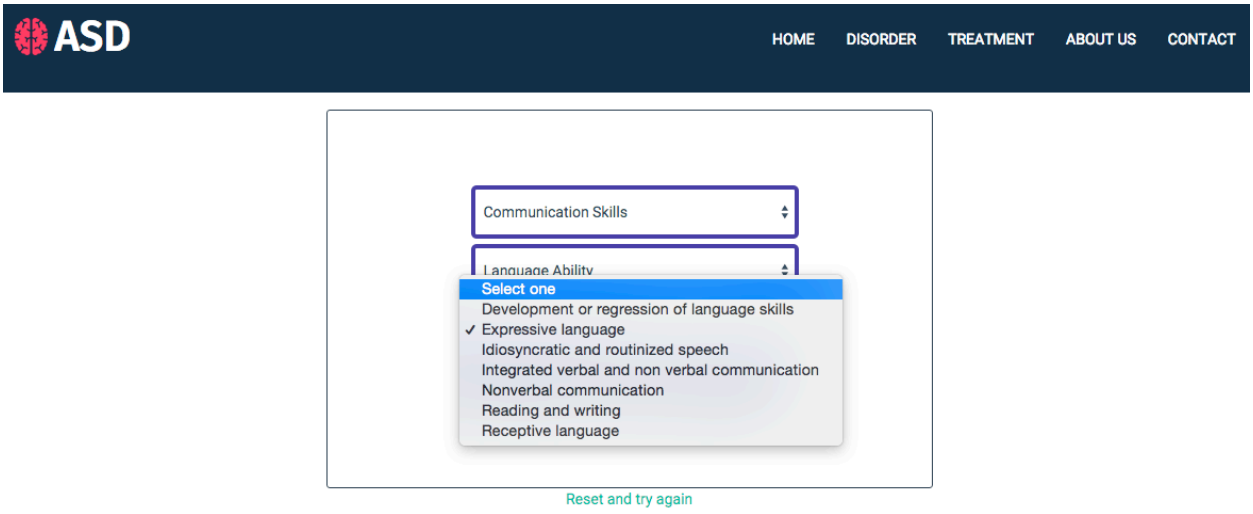


*Figure 44. Dropdown menu system indicating the subclasses options for cognitive ability.*

The users are able to investigate the disorders of autism via query system and select the disorder to find out about the existing treatment. For example, the subclasses of the cognitive ability are manifested to the users in the third drop down menu option in the figure above. Users can understand the knowledge of each term by selecting the disorder displayed in dropdown menu. They can also explore the subclasses of the selected disorder. The subclasses for any class in ASD ontology define the concept of the class and express the definition of the autistic term. Furthermore, query system demonstrates the relationship between the selected disorder and other related concepts (i.e. classes) by the entire dropdown menus for that disorder. Users are also able to figure out the available treatment of a specific disorder as well as the ability to select the treatment method to access more information about the influence of the treatment on disorders and effect of the disorders in ASD. The query system can provide intervention methods for autism spectrum disorders based on the treatment class of the ASD ontology. It can also show the relationship among selected intervention methods.

The ASD ontology is based on trusted knowledge extracted from various expert sources. Users can validate the accessed information by knowing the source from which classes and terms of autism spectrum disorders ontology have been created. Such source could be a textbook, relevant published article(s) or clinical studies. This is a significant feature for researchers, experts and general users to be able to refer to the original source of the provided information for accessing further details, if they wish to do so. Furthermore, as new ASD knowledge becomes available, the proposed ASD System has the ability to expand by integrating the new ontological data. The ASD Query system is built on MySQL tables that can be easily updated through basic SQL commands. Finally, the proposed system is a developed around a Web Query system, making it accessible to users worldwide.

## 4.2 Evaluation

### 4.2.1 The ontology

The developed ontology is based on that proposed by Uschold and King (1995). The aim is to use Uschold and King's ontology as the kernel to build, step-by-step, a comprehensive ontology

based on standards, clinical research findings, and well known therapies in ASD field. The approach is to perform extensive research to capture the standard terms and serve as main ASD ontology class concepts using different sources as textbooks, relevant articles, clinical studies in the domain and existing special purpose ontologies. The second stage is to establish the right relationships amongst the ASD classes, and then finally classify and link available ASD knowledge around ontology classes. The ontology construction process started with identifying three top ASD classed and their relationships. Then, a methodology was adopted to set the full conceptual ontological structure. This has been evaluated to achieve a standard comprehensive ontology for autism spectrum disorders.

Ontology evaluation is concerned with two important aspects, quality and correctness. Quality is mainly referring to conceptual aspects of the ontology such as adoption of standards, terms that describe ontology concepts and classes. Correctness however, refers to ontology conceptual structure in terms of class relationships, class properties, and embedded knowledge.

As discussed in chapter two, the core structure of the created ASD ontology is based on Disease-Treatment Ontology by Christopher S.G Khoo et al. and phenotypic ontology for autism spectrum disorders by Alexa T. Mc Cary et al. The top level ontology for autism spectrum disorders builds on Disease-Treatment Ontology as shown in chapter two. The ontology is being evaluated through series of test sets to ensure that it covers most relevant concepts and semantic relationships in the ASD domain. As part of this process, some new classes and properties have been added to the Disease-Treatment Ontology after the evaluation. The lightweight ontology of ASD was created based on the concepts of Disease-Treatment Ontology. The structure of the classes issues from disorder class and the terminology for these classes have been extracted from phenotypic ontology for autism spectrum disorders by Alexa T. Mc Cary et al, from Harvard Medical School. Terminologies around disorder class of ASD ontology were evaluated in terms of nature of structure and content. Each concept of disorder class has been given both a unique identifier as well as a tree number indicating its place in the hierarchy. Over time, and as more is known about ASD, the tree numbers may change, but the unique identifier will stay constant.

The concepts have been mapped into to standard ontologies, specifically Medical Subject Headings, International Classification of Functioning, Disability and Health, and the Unified Medical Language System to ensure that the ontology can be used to link to other data sources, including the biomedical literature.

Medical Subject Headings (MeSH) come from the National Library of Medicine's controlled vocabulary thesaurus. It contains sets of terms naming descriptors in the form of hierarchical structure that permits searching at various levels of specificity. Description in Medical Subject Headings (MeSH) are categorized in both alphabetic and hierarchical. There are 27,883 descriptors in 2016 MeSH with over 87,000 entry terms that assist in finding the most appropriate MeSH Heading. Mental disorders is one of the most general level of the hierarchical structure heading in MeSH which has been involved for ontology evaluation.

The International Classification of Functioning, Disability and Health (ICF) is a framework for describing and organizing information on functioning and disability. The ICF provides a conceptual basis for the definition and measurement of health and disability and a standard language. Its been approved to be used by the World Health Assembly in 2001. After broad testing across the world involving people with range of disabilities, ICF been approved to be used by the World Health Assembly in 2001. The ICF system designed as a multipurpose classification to provides services for various disciplines and sectors for example in education, health and community services. The major models of disability integrated in ICF which was one of the good sources for evaluating the disorder and effect class in ASD ontology.

Unified Medical Language System (UMLS), is a set of files and software which provides many health and biomedical vocabularies and standards to enable interoperability between computer systems. The main purpose of UMLS is to enhance or develop applications, such as electronic health records, classification tools, dictionaries and language translators. The terms and concepts of disorder and influence of mental disorders of ASD on autistic people have been mapped into UMLS to obtain the accurate terminology for the ontology.

The concepts and the terminologies of the ASD ontology have been gathered from trusted sources such as standard textbooks, published related articles from different journals, relevant clinical studies in autism field to extract valuable concepts among them for building the ontology for autism spectrum disorders. Additionally, the concepts and terms of the ontology have been discussed with a therapist of autistic children from Thunder Bay, Canada, and a specialized doctor in autism from Dubai, UAE to confirm the relationship among the disorder class, treatment class and effect class, which have been in turn defined based on the clinical studies and articles in ASD field.

| | Domain knowledge | | | | | Operational knowledge | | |
|---|---|---|---|---|---|---|---|---|
| **Ontology** | **ASD instruments** | **UMLS** | **ICF** | **MESH** | **Relevant Articles** | **Clinical Studies** | **Encyclopedia of ASD** | **Comprehensive guide to Autism** |
| **ASD ontology** | Yes | Yes | Yes | Yes | Yes | Yes | Yes | Yes |
| **Alexa T. Mc Cary et al. ontology** | Yes | Yes | Yes | Yes | Yes | No | No | No |
| **Lynn young et al. ontology** | Yes | No | No | No | Yes | Yes | No | No |
| **Omri Mozach et al. ontology** | Yes | No | No | No | Yes | No | No | No |

*Table 9. The evolution methods of ASD ontology.*

| **Ontology** | **Disorders** | **Effect** | **Interventions** | **Relation among disorder and effect** | **Relation between domain and operational knowledge** | **Query system for Regular users** |
|---|---|---|---|---|---|---|
| **ASD ontology** | Yes | Yes | Yes | Yes | Yes | Yes |
| **Alexa T. Mc Cary et al. ontology** | Yes | Yes | No | Yes | No | No |
| **Lynn young et al. ontology** | Yes | No | No | No | No | No |
| **Omri Mozach et al. ontology** | Yes | Yes | No | No | No | No |

*Table 10. The difference between ASD ontology and other related ontologies.*

The domain knowledge of ASD ontology refers to disorder and effect class which is evaluated by UMLS, ICF, MESH. Furthermore, the terminologies in the domain knowledge of ASD ontology have been mapped and evaluated by ASD instruments and relevant articles to ensure about the quality of the concepts. The domain knowledge evaluation of ASD ontology is similar to Alexa T. Mc Cary et al. ontology as it's been used as an exacting ontology for our work. The Lynn young et al. ontology and Omri Mozach et al. ontology are not evaluated through the UMLS, ICF and MESH as we can see in the table 9 that brings limitation to the term and concepts of their ontology. Hence, table 9 indicates that the quality of the ASD ontology which refers to terms and concepts in domain knowledge is more improved compare to other ontologies.

The operational knowledge of ASD ontology which refers to treatment class has been evaluated by clinical studies, Encyclopedia of ASD and Comprehensive guide to autism. Although the domain knowledge structure of ASD ontology is similar to Alexa T. Mc Cary et al. ontology, but operational knowledge of ASD ontology is the unique characteristic for our work which makes the ASD ontology different with other related ontologies in the field as its manifested in table 9.

As its been discussed the correctness of ontology is based on the relations and properties of the concepts. The properties and relation between the domain knowledge and operational knowledge are created based on clinical studies, relevant articles and standard text books which evaluated by Encyclopedia of ASD and Comprehensive guide to autism in operational knowledge filed. Additionally, the correctness of the ontology evaluated by experts in the field of autism as been been discussed earlier to ensure the correctness of relations and properties in the piratical side.

The differences of ASD ontology and other related ontologies are displayed in the table 10. As it's been shown in the table 10 ASD ontology is a comprehensive ontology compared to other ontologies in terms of intervention for autism spectrum disorders, clarifying the relation between the domain and operational knowledge by indicating the relationships among the classes of disorder, effect and treatment in autism spectrum disorders. Moreover, ASD ontology has a web query system for regular and professional users to access the ontology information. The web query system enables ASD ontology for better evaluation of relations among the domain

knowledge and operational knowledge from users in practical and experimental concept in the future.

However, the autism spectrum disorder is a heterogonous group of disorders for which the patients have no unique type of symptoms. Autistic people have different type of disorders with different severity, which have important roles in their method of the treatment. Hence the treatment approaches for every child with ASD are different and are based on the disorders of individuals with autism and the severity of their disorders. Consequently, autism spectrum disorders have different influence on autistic people. These can be checked in our system by experiencing specific queries.

Users can participate to evaluate the efficiency of the developed system by providing their feedback about their experience using the ASD web query system. The web query system provides the possibility for users to interact with the system and assess the queried ASD information for different purposes.

### 4.2.2 The query system

The web query system has been evaluated in terms of efficiency and usability. Fourteen users have participated in a usability survey. Many of the participated users have good background of autism (either grasped from the nature of their work or from some related people with autism). The survey consists of a questionnaire of ten questions measuring many aspects of system usability and ease of access to ontology knowledge and embedded information. Usability is not a quality that exists in any real or absolute sense. The usability of any tool or system has to be viewed in terms of the context in which it is used, and its appropriateness to that context (Brooke, 1996). However, it can be best summed up as being a general quality of the appropriateness to a purpose of any particular artefact (Brooke, 1996).

In general, it is impossible to specify the usability of a system without first defining who are the intended users of the system, the tasks those users will perform with it, and the characteristics of the physical, organizational and social environment in which it will be used for (Brooke, 1996).

Usability measurement provides a method to evaluate some general classes of the system. In general, measure of usability should cover the following aspect of the system:

- effectiveness: the ability of users to complete tasks using the system, and the quality of the output of those tasks,
- efficiency: the level of resource consumed in performing tasks, and
- satisfaction: users' subjective reactions to using the system.

The questionnaire is designed based on the System Usability Scale (SUS) and the purpose of the query system in the field of ASD. There are five questions from SUS questionnaire and the rest of the questions are for evaluating the system in terms its efficiency in ASD field. Five scales in the range [1-5] have been used (1: Strongly Disagree – 5: Strongly Agree). The questionnaire for evaluating the web query system is the following:

1. The system was easy to use
2. There were not too many inconsistencies in the system
3. I would imagine that most people would learn to use this system very quickly
4. I felt very confident using the system
5. I found the various functions in this system well integrated
6. I found the system and presented information reliable
7. I found the system accurate based on my query
8. I found the system efficient to use
9. I found the expected information in the system
10. I found the query system user-friendly

The questionnaire has been embedded as a survey section in the web query system to be more accessible for users. Users answered the questions by rating the system from 1 to 5. The questionnaire was designed to provide important information from different type of users to

indicate the positive and negative perspective of the query system in terms of effectiveness of the system, efficiency of presented information and the user's satisfaction.

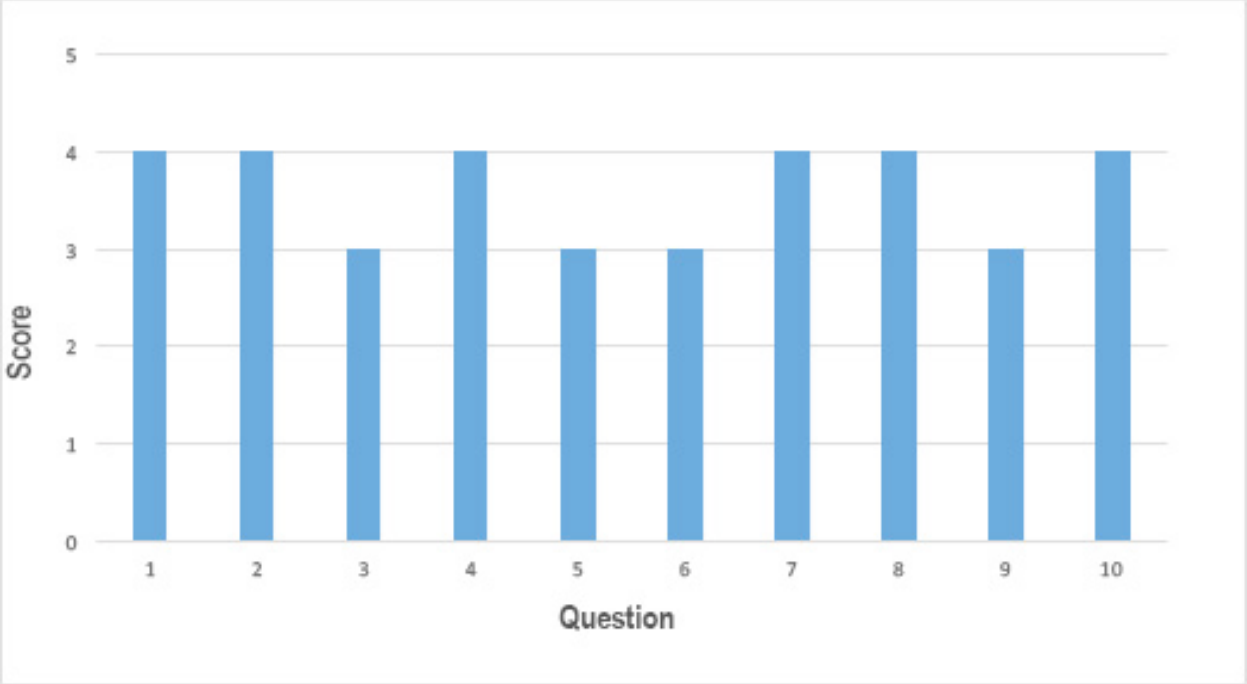


*Figure 45. Average scoring for the questionnaire.*

Fourteen users have participated in the survey. Eight people of the survey takers were caregiver or parents of autistic children that we met them through the teacher for autistic children Ms. Jenn Smith. Other survey takers were professional in field of autism or people with knowledge of autism. The overall score for the questionnaire is 3.6, which is an acceptable score for the ASD query system. We converted the overall score of survey via multiplying by 20 to get the overall score out of 100. The converted score is 72 out of 100, which allows us to compare query system quality with SUS scores and adjective rankings from The Journal of Usability Studies.

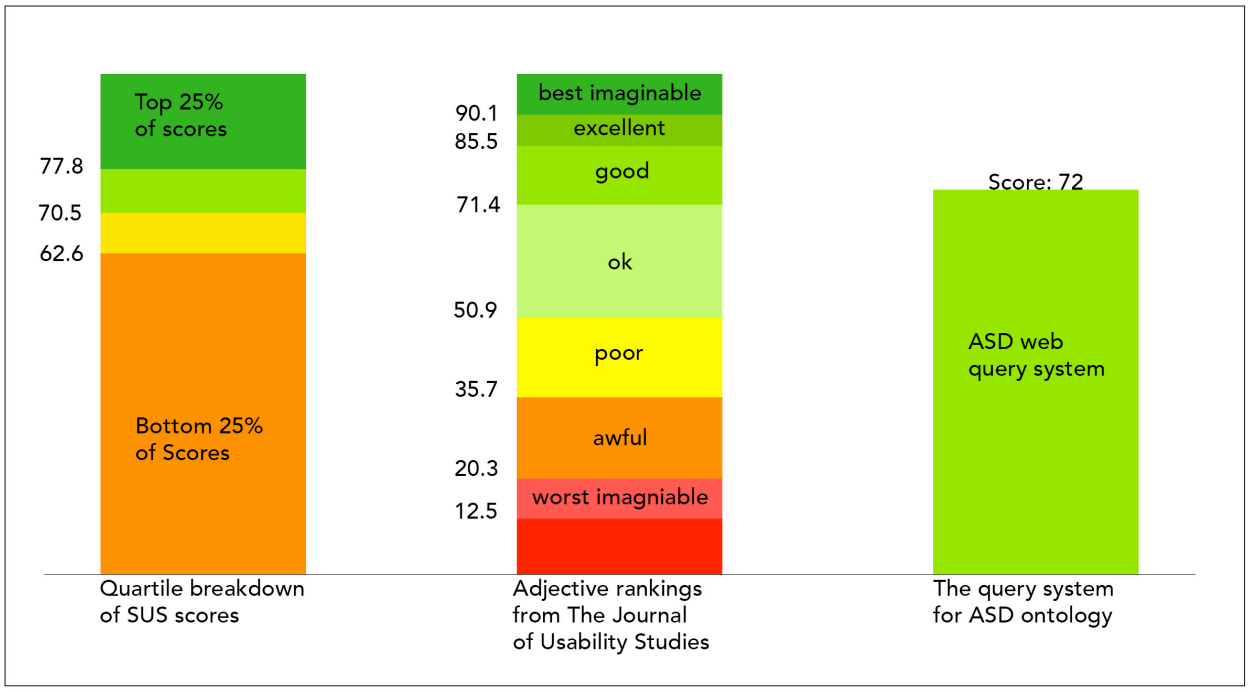


*Figure 46. Evaluation of the ASD query system.*

We obtained a good survey score overall for the web query system compared with quartile breakdown of SUS scores chart and suggested adjective rankings from The Journal of Usability. On the average any system with score above 71 in SUS Scores chart and adjective rankings from The Journal of Usability considered as a good system as its shown in the figure 47.

In general, the overall score of the survey for the query system manifest that the basic query system has a proper interaction with users in terms of effectiveness, efficiency and the user's satisfaction. However, the query system can be improved in the future in order to present additional information of the ontology such as properties of classes and some extra information for intervention and disorder class, thus, the new features can enhance the efficiency of the query system and the user's satisfaction.

# Chapter 5

# Conclusion and Future work

## 5.1 Conclusion

This section reviews all the concepts behind the impetus and contribution of the thesis. It also summarizes the solutions provided in this research work, with special emphasis on the proposed ASD ontology structure and its construction process.

The first and major problem addressed in this research work is linking scattered ASD knowledge into a single knowledge body that is made available to both humans and machines. Scattered knowledge comes as the consequence of lack of information among ASD concepts and limits accessibility to precise information for all community members concerned with autism. The duplication of data is the second problem that has been addressed in this thesis. The main cause of duplicated data in autism spectrum field comes from the heterogeneous research done by different researchers over the same topic, which is usually made for different purposes. Another major problem is difficulties to access accurate ASD information. There are many different sources of information in autism spectrum disorders domain. However, accessing the trusted information is a big challenge for both regular and professional users. The thesis also discussed the limitation of related work developed by other researchers. Such study was very important to understand the limitations of these studies and the necessity of building a comprehensive standard ontology in ASD field that can benefit general users and experts alike.

The ASD ontology aims at representing the knowledge in autism spectrum disorders. The goal is to conceptualize the knowledge and link it to various sources of available ASD information. This

is assured by unifying the terminology around most agreed upon concepts in ASD domain and providing a comprehensive semantic map in terms of a global ontology accessible by both machines and humans, thus facilitating access to both basic ASD information and supporting experts in their clinical decisions.

### 5.1.1 ASD Ontology

The proposed ASD ontology is built on the ontology proposal made by Uschold and King. Their approach consists of three clear steps, which were adopted in this study. The aim is to create a comprehensive standard ontology in terms of structure and concepts. In the first step, the ontology purpose was identified as operational and domain knowledge ontology for describing the whole spectrum of ASD field. To achieve this goal, the concepts and relations among the ASD concepts were extracted and refined from various sources such as standards textbook, published articles and clinical studies. In the second step, ontology concepts were imported into the built ontology and encoded in Web Ontology Language (OWL). In the third and final step, the ASD ontology was heavily reviewed and refined from different perspectives in order to link the whole spectrum of autism disorders, their treatment, and effect of disorders

The core structure of the ASD ontology is based on the top level classes of Disease-Treatment ontology by Christopher S.G Khoo and his colleagues. Disease-Treatment ontology is a medical ontology for general purposes in this field. Although the lightweight ASD ontology is developed based on the Disease-Treatment ontology, but significant changes have been applied over the ontology to be more fit in the field of autism spectrum disorders. The applied changes on the Disease-Treatment ontology made the ASD lightweight ontology more flexible for representing the knowledge in different aspects of ASD such as disorders, treatment and effect of disorders.

### 5.1.2 The ontology design

The ASD ontology is made of two main structural design steps. The first step was initial design to build the basic semantic map skeleton of ASD field, and the second step was final design for obtaining a more comprehensive ontology that can relate concepts to each other, help eliminating

ambiguities in the field, and provides complete linkage between autistic concepts to resources. The initial design aimed at developing the top level ontology with main classes of the ASD field and their relationships, thus representing both the domain knowledge of ASD, such disorder class, and operational knowledge of ASD, such as treatment class and Disorder effect class. The key subclasses of each top level class have been then derived in the initial design of the ASD ontology.

In the final design, the lightweight ontology was converted into a comprehensive ontology by adding the terms and concepts for each main class. The terminologies and concepts for ASD ontology are extracted from existing ontologies in the field, standard textbooks, relevant published articles and clinical studies for autism spectrum disorders. For instance, the terminologies and concepts of disorder class and effect class have been extracted from a relevant exiting ontology of Modeling the Autism Spectrum Disorder Phenotype by Alexa T. McCary et al. (2014). The extracted information has been mapped into the ASD ontology. The terminologies for treatment class are based on various sources such as the standard textbooks, relevant published articles and clinical studies that the intervention has been tested on individuals with ASD.

The ASD ontology maps the concepts and knowledge of autism spectrum disorders. The terms and concepts of the ontology define the domain knowledge of ASD such as disorders and influence of disorders on autistic people. Furthermore, the operational knowledge of ASD in terms of excising standard intervention has been elaborated as part of the ontology. One of the main and unique features of autism spectrum disorders ontology is relationship among the classes of the ontology. As shown in previous chapters, there are three kinds of relationship in ASD ontology. The first kind is the relations holding on between universal and other. The second kind is relations holding between one particular and another. Finally, the third kind of relations shows the coherence among the terms is the relations holding between a particular and a universal. This relation, which is one of the main features of ASD ontology, is based on the revealed information in properties of classes. The properties for each term obtained from various sources to define the relationship among subclasses of disorders, effect and treatment class. It should be noted that the third kind of relation indicates the influences of different aspects such as

disorders, effects and treatments in ASD. For instance, the relation between a particular intervention with disorder and effect of the disorder has been defined.

The Disorder type class contains 23 concepts for common behavior class, 8 concepts for communication skills and 15 concepts for socials skills. The effect class has 63 concepts for personal traits class, 53 concepts for social competence and 20 concepts for treatment class effect. The treatment class of ASD ontology has 47 concepts which biomedical class holds 26 concepts and therapy class contains 19 concepts. In general, the ASD ontology contains 238 concepts in three different aspects of autism spectrum disorders such as disorders, effect and treatment. Putting all this knowledge together, in a comprehensive semantic structure, is the main contribution of this research work.

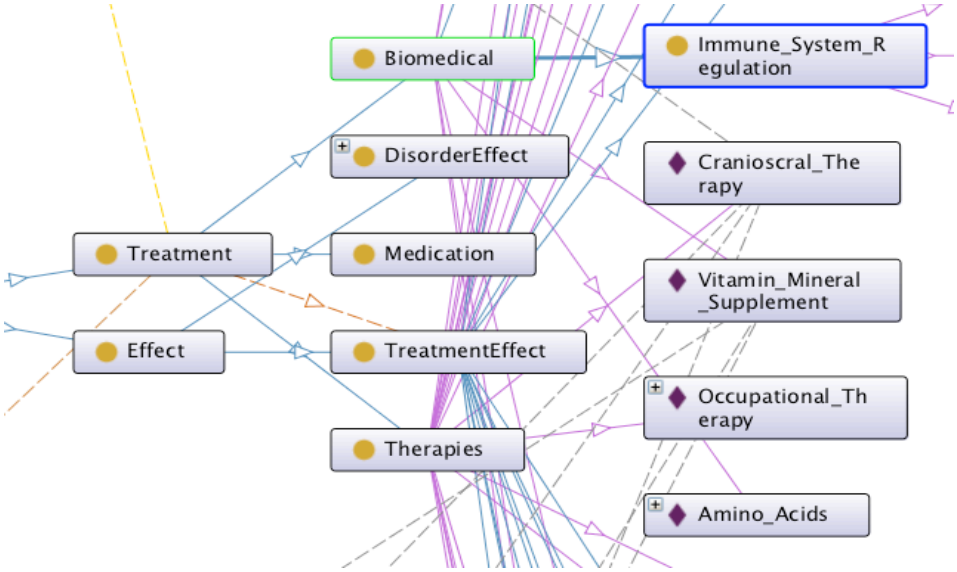


*Figure 47. Relations among the classes of ontology in Protégé shown in colored lines.*

The autism spectrum disorders ontology was built with the aim to support experts in their clinical decisions for treating some aspects of autism. In particular, the indicated relationship among the classes of the ontology can support therapists or doctors for better decision making to find the right intervention based on the disorders and symptoms in the autistic people.

### 5.1.3 Query system

The ASD query system provides dependent, multiple dropdown menus to indicate the classes and subclasses of the ontology. The content of each drop down menu retrieved from the database and the next dropdown menu content will be regained from database based on the pervious selection of the users. This strategy let users to choose the right term among the many terms and concepts in the ontology. The ASD query system is a basic web query system based on relational database for representing the information of the created ASD ontology to regular and professional users around the world. This system has been designed to share the knowledge of autism spectrum disorders with users in order to assist them with better treatment decision making for autistic children, facilitate the access to the ASD information, effect of disorders and available interventions in autism field. Additionally, users can find out about the relationship among the different aspect of autism such as relative effects and interventions for a particular disorder. Moreover, the complexity of the information of the classes and relations in ASD ontology can be delivered in a simple way to regular users such as care givers and parents of autistic children.

The ASD query system is a web based information system that provides an interface for users to interact with autism spectrum disorders ontology. The front-end structure of the system is where users are able to submit their query to the system. The Back-end component, which is written in PHP language connects the user interface to the relational database. The relational database that houses all autism knowledge and information has a key role for the web query system. The information and relations among the classes of ASD ontology has been converted manually and mapped to the tables in the database.

## 5.2 Future work

The ASD ontology represents the knowledge in autism spectrum disorders in different aspects such as various disorders type in ASD, effect of the disorders on individuals with autism and available interventions for these disorders. However, knowledge of health care and medical field

are growing day by day. This section discusses improvements that can be added to the system in the future to provide further facilitates for researchers in the field of ASD in terms of the domain knowledge and operational knowledge. The following are some possible improvements to the system.

The web query system is capable to be updated in future in order to deliver the precise information to the users. The structural design of the query system can be more intelligent based on the users query history to provide the user with any newly added knowledge in the ontology as well as related resources in the database.

The top three level classes of the ontology represent different knowledge fields in ASD such as domain knowledge through the disorders and effect class and operational knowledge via treatment class of ASD ontology. Intervention for autism spectrum disorders, which is considered as operational knowledge of the ontology is an extensive knowledge field. The treatment of ASD is one of the interesting topics for both therapist and researchers for finding efficient way of treatment for ASD. Hence the terms and concepts of treatment can be upgraded from adding the new classes based on the latest intervention methods in therapy, biomedical and dietary.

The subclasses of treatment class such as therapy, biomedical and dietary class contain properties. The properties provide more information about the terms in ASD ontology. Additionally, properties define the relations among the terms of the ontology. Hence, more properties can be extracted from standard sources such as new clinical studies, latest ontologies and can be eventually mapped into the ontology in future for treatment class.

Improving the properties of the ontology in future can reveal more relationships among the subclasses of the top level classes which can be improved by new relations from latest research and achievements in ASD field. This process will upgrade the ontology structure in order to be more accurate and reliable to support the clinical decision by professional users such as therapists and doctors.

One of the most important feature that can be added into the properties of the ontology is the efficiency for a treatment. The presented intervention by ASD ontology can have various efficiency on autistic people in order to improve quality of their life in social, behavior and communications skills. Some clinical studies manifest an approximate efficiency for a few treatments based on period experiment on autistic children for a particular therapy. More clinical studies on therapies and dietary intervention for individuals with ASD can specify the approximate efficiency of treatment. Furthermore, the future interaction between the users and the web query system also can provide useful information about the user's experience for a particular intervention.

The medication for autism spectrum disorders has not been part of the thesis work. Medication for ASD is a very complex topic, which can be done by a specialist or doctors in the ASD field. The medication class can also be mapped as one of the top level classes for treatment class for the future version of the developed ASD ontology. The properties of medication can be obtained and imported into the ontology, which will help provide the relationship between the medication, the disorders and other aspect of autism spectrum disorders.

Finally, the ASD query system can also be improved in terms of providing a visual concept-map (e.g. graph) to display better information to the user about the nature of semantic links between ontology concepts and embedded ontology knowledge in the field of ASD.

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