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Categorical and dimensional approaches in diagnosis using Asperger syndrome as an example

Kategorialne i dymensjonalne podejście w diagnozie na przykładzie zespołu Aspergera

Abstract

In the latest medical classifications (DSM-5 and ICD-11), Asperger syndrome (AS) is no longer a separate nosological entity. It has been included in the category of autism spectrum disorder (ASD), which has significantly affected the lack of differential diagnoses. This is not the best solution for people with ASD, as it does not take into account the clear distinctions between autism and Asperger syndrome despite the presence of similarities. In addition to the similarities, Asperger syndrome is also characterized by numerous differences from autism, and these are not limited to differences of a quantitative nature (the severity of the deficits present). The dominance of the categorical treatment of the phenomena, both in the DSM and the ICD, plays a major role in perpetuating the problem.

Keywords: Asperger syndrome, dimensional diagnosis, categorical diagnosis, autism, autism spectrum disorders (ASD)

Streszczenie

W najnowszych klasyfikacjach medycznych (DSM-5 i ICD-11) zespół Aspergera nie jest już odrębną jednostką nozologiczną. Został on włączony do kategorii zaburzeń ze spektrum autyzmu (ASD), co znacząco wpłynęło na brak diagnoz różnicowych. Nie jest to najlepsze rozwiązanie dla osób z ASD, ponieważ nie uwzględnia wyraźnej odrębności autyzmu i zespołu Aspergera, pomimo występowania podobieństw. Poza podobieństwami zespół Aspergera charakteryzuje się również licznymi różnicami w stosunku do autyzmu i nie ograniczają się one wyłącznie do różnic o charakterze ilościowym (nasilenie głębokości występujących deficytów). Dużą rolę w pojmowaniu ASD odgrywa dominacja podejścia kategorialnego, zarówno w DSM, jak i ICD.

Słowa kluczowe: zespół Aspergera, diagnoza dymensjonalna, diagnoza kategorialna, autyzm, spektrum autyzmu (ASD)

Introduction

The knowledge of Asperger syndrome has been evolving since the 1980s but most extensively since 1994 when Asperger syndrome was codified in the DSM-IV as an autonomous clinical entity. At the time, attempts were made to create separate diagnostic criteria for autism and Asperger syndrome. The failure in this area is due to the lack of a dimensional understanding of both phenomena. Both autism and Asperger syndrome have a strongly differentiated clinical picture and are scalar (have varying degrees of severity of features). Numerous cases of children and adults—men and women—have contributed to the multidimensional picture of AS and the importance of shifting the categorical assessment of disorders towards a dimensional approach.

1. Asperger syndrome and autism

The problem with differential diagnosis of autism and Asperger syndrome has existed ever since the two clinical entities appeared in the diagnostic space. Publications describing both disorders appeared within a one-year interval,¹ but the knowledge of autism did not parallel that of AS. Autism became a recognizable entity in the 1950s, while Asperger syndrome was represented in the DSM-IV over forty years later.

To this day, specialists debate the relationship between the two disorders. They are often viewed as a continuum. In the 2013 DSM (DSM-5) version, Asperger syndrome was included within the autism spectrum disorder. Currently, both the DSM-5 and ICD-11 promote the concept of a “spectrum,” which addresses the strong differentiation between the two phenomena and the overlap between some symptoms/features. The problem of determining the relationship between autism and AS arose from an attempt to establish a categorical classification of diagnostic criteria. According to the theory of categorization formulated by Aristotle, which provided a basis for Western medical diagnostics, as well as psychological and developmental, entities belonging to one set should have “necessary and sufficient properties,” which in diagnostic practice means a list of axial symptoms. This means of interpreting reality is culturally based on analytic thinking, far from the holistic thinking of East Asians (Nisbett, 2015), and has developed into a system of differentiation aimed at creating standardized models. Such standardization, in turn, generates situations where cases that differ from established norms are considered atypical (e.g., the case of autism in DSM-IV).

Autism and Asperger syndrome were described by Leo Kanner and Hans Asperger through the observation of patients with a diagnosis of, *inter alia*, schizophrenia and psychosis, whose features and behaviors did not meet the criteria for axial symptoms of mental disorders. However, as knowledge of these two disorders progressed, they

¹ Leo Kanner described 11 cases of children and coined the term *early infantile autism* for a set of features he observed, but one year later Hans Asperger in his post-doctoral dissertation described individuals who shared many of the same features, but used language communication.

were placed in a taxonomic and categorical diagnostic model of mental disorders. An attempt to establish separate diagnostic criteria failed, and researchers (mainly theoreticians) believed that autism and Asperger syndrome constituted a clinical continuum (Wing, 1988). These opinions still exist in discussions of specialists.

The high heterogeneity in the expressions of two disorders (autism and Asperger syndrome) makes it difficult, but not impossible, to offer early differential diagnoses. Despite having common symptoms, there are no grounds for regarding them differently on a spectrum based solely on the number of demonstrated features. Therapeutic practice provides conclusive evidence that autism and Asperger syndrome do not constitute a continuum because even the most effective therapies are not able to change the diagnosis from autism to AS.

2. Categorical and dimensional understanding of a diagnosis

The case for a categorical diagnosis of Asperger syndrome and autism has its origin both in the history of understanding both disorders and in the dominating role of the DSM (*Diagnostic and Statistical Manual of Mental Disorders*) in the diagnosis of mental disorders, including developmental, personality disorders, psychosocial, and environmental problems. The current DSM concept is based on the work of the German psychiatrist Emil Kraepelin (Trull et al., 2005), whose nosological concepts assumed that mental illnesses have clear boundaries between normality and illness. Accordingly, the DSM defines personality disorders within a categorical, hierarchical taxonomic system (Trull et al., 2005, p. 356). A group of American psychiatrists working on the DSM is referred to as “the neo-Kraepelinian revolution” (Compton & Guze, 1995).

The taxonomic approach to the classification of phenomena, including personality disorders and developmental disorders, is not the only possible diagnostic approach. More and more often, in practice and the literature, voices are heard pointing to the necessity of developing a dimensional approach, “which makes it possible to diagnose a disorder with the use of a continuum of different features. When the comparison is made, the dimensional approach being more reliable shows superiority over the categorical approach” (Nowak, 2015, p. 100). Other experts explain, “there is a clear need for dimensional models to be developed and for their utility to be compared with that of existing typologies” (Rounsavill et al., 2002, p. 12).

The dimensional approach to the diagnosis and the characteristics of developmental disorders makes it possible to present their multidimensionality, rating scales, horizontal intensity (e.g., intensity over time, changeable number of features), and vertical (severity of disorder). It is also possible to show the relationships between the disorders, which significantly changes the picture of a clinical entity.

The dimensional understanding of the disorder leads to the improvement in the diagnosis of less prototypic cases, because “features providing a basis for the identification of disorders are multidimensional and can be moderate or severe. . . . If the feature makes the individual difficult to function . . . the diagnosis is simple, but when the intensity of this feature is less severe a boundary between the norm and pathology is very difficult to draw” (Nowak, 2015, p. 100).

3. Sets of features characteristic of Asperger syndrome

Typical symptoms of Asperger syndrome can be characterized in a few domains, among others, including language communication, topic fixations (interests), social relationships, memory characteristics, manipulative behaviors, and sensory reactions. Heterogeneity of these domains in individuals is significant (Korendo, 2013).² The differences can be understood according to the following:

- a) Language communication. Contrary to what was included as a diagnostic criterion in the DSM-IV and ICD-10³ and the research studies carried out by Hans Asperger, Lorna Wing, and others, even though there may be initial delays in speech development with Asperger syndrome as with autism, the similarity of this symptom does not preclude a differential diagnosis between the two disorders. In addition, persons with AS demonstrate differentiated intensity of other features observed in language communication, such as uneven development of vocabulary (language funnels connected with the child's vague thematic interests), a tendency to quote, language schematisms (repeated, consolidated expressions adequate for a situation, but sometimes used without full understanding), non-linear, associative way of constructing a narrative (which is considered by listeners as a continual change of topic in a story or conversation), understanding words and statements literally, and lack of dialogue skills and listening to the interlocutor's questions and answers. None of these features have to appear in every individual with AS; they are usually of differentiated intensity. For example, the tendency to understand language literally may prevent the understanding of metaphors, including those strongly lexicalized (e.g., dash off for beer), or may influence the preference for precise, prototypic use of vocabulary meaning (e.g., mother to child with AS: "You've got home from school, little snail."; Child: "I'm not a snail, I'm a human.").
- b) Topic fixations (interests). In Asperger syndrome, an individual may exhibit topic preferences, which can range from strong fixations or a deep interest in a specific area of knowledge to slight preferences or slight topic devotion. A lower level of topic fixation is more often present in girls with AS.
- c) Social interactions. A level of difficulty in developing social interactions is significantly differentiated in people with Asperger Syndrome. A child may be on equal terms with adults or totally reject their supervisory role, not accepting the limits they establish and the rules they impose. Social interactions with peers, specifically children of the same age group, play a significant role in diagnosis. In other cases, interactions may be specific; for example, the child may submit to older friends or dominate younger individuals.

² The author described these domains in a monograph "Language interpretation of the world in statements of persons with Asperger syndrome" Kraków 2013 and in the article (Korendo, 2022).

³ Both the DSM-IV and the ICD-10 included a criterion that conclusively indicated the absence of speech delays as a necessary condition for diagnosing Asperger syndrome and differentiating AS from autism.

- d) Memory characteristics. A better-than-average visual and auditory memory often distinguishes persons with AS. This ability is more common in boys than in girls; therefore, devotion to certain topics takes on a different character. However, not everyone with AS demonstrates this ability, nor is it observed at all stages of life.⁴
- e) Manipulative behavior. Children with AS often try to take control over the reactions, behaviors, and emotions of their caregivers who unconsciously or, for the sake of minimizing unwanted results, succumb to manipulations. These are actions with a “control” effect; the child tests the effectiveness and modifies the form accordingly. Forms of manipulation are strongly differentiated, and the degree of intensity depends on the child’s individual predispositions and his or her caregiver’s reaction. Qualitative differences are also observed between the genders; boys more often resort to behavioral manipulations (e.g., screaming, squealing, using vulgar language, hiding under a school bench, or throwing objects), whereas girls look for emotional manipulations (e.g., feigning stomach aches or headaches, crying, or verbalizing fear in uncomfortable situations). Sometimes these behaviors are discrete and difficult to identify by caregivers, but they can also significantly affect daily life at school or home.
- f) Sensory reactions. Similar to other autism spectrum disorders, individuals with AS can experience sensory disorders such as hypersensitivity or hyposensitivity. There is no clear model typical of Asperger syndrome; each of the senses might be heightened to varying degrees. In addition, the tactile sense can be felt differently along all parts of the body (e.g., hyposensitivity of the hand and hypersensitivity of the face). However, people with Asperger syndrome may not experience sensory disorders.

4. Co-occurrence of other disorders

A categorical understanding of disorders also leads to an overly analytical perception of problems that may ignore or isolate peripheral symptoms from those included in the primary diagnosis. In other words, the difficulties a child encounters might only be explained by deficits resulting from the main diagnosis, which can lead to misinterpretation or neglect of the symptoms that are not axial (necessary or sufficient features). For example, this problem may occur in the case of children with AS who struggle with reading skills. Usually, persons with Asperger syndrome, who are associated with better than average memorizing skills, are not included in a group susceptible to dyslexia. Therefore, their aversion to reading might be interpreted as a manipulative behavior, resulting from the urge to evade an unattractive activity. Meanwhile, Asperger syndrome can co-occur with other disorders in the following ways:

⁴ All the boys with AS from my therapy group had better than average auditory and/or visual memory. In the case of Patryk, this skill was significantly weaker, almost imperceptible after the age of adolescence.

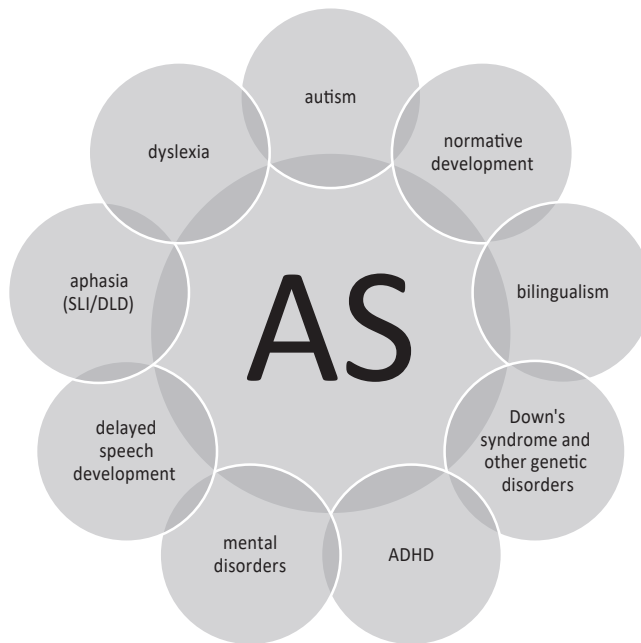


Chart 1. Relationships between AS and other disorders and the norm

- a) AS and the norm (normative development). Due to the multidimensional and strong qualitative heterogeneity of cases (considering all the arguments presented above), it must be emphasized that the boundary between the norm and the disorder is not sharp and cannot be set quantitatively (e.g., on the basis of the number of features). In borderline cases, children often do not receive therapeutic support and diagnosis for a long time, and their behaviors are interpreted as the result of a bad upbringing. This problem is sometimes blamed on parents who are accused of parental incompetence. Children from this diagnostic area often have intensified problems with accepting and respecting social rules. Other symptoms can be discrete, thus not giving a full picture of the disorder.
- b) AS and dyslexia. As noted above, dyslexia and Asperger syndrome can be co-occurring disorders. Statistically, children with AS often learn to read quickly due to better memory skills. However, this sometimes means reading without understanding because memorizing syllables (words) quickly and their sound reproduction does not reflect a similar level of text processing. However, there is a group of individuals with AS who demonstrate typical dyslexia difficulties when acquiring reading and writing skills. They may have a much wider range of dyslectic symptoms, such as problems with timing, a lowered level of gross and fine motor skills (this area is common for both genders), and linear disorder of processing stimuli in time and space. If a child with AS also has dyslectic problems, he or she should have a diagnosis of both dysfunctions.

- c) AS and delayed speech development. Among children with AS, there is a group of individuals with speech delays, which is in contradiction to earlier reports about the absence of these difficulties in AS. Contrary to the conclusions drawn from research carried out by Hans Asperger himself, who did not have any diagnostic experience in a developmental assessment of children under age three, functional, often severe language acquisition impairment may be present in those with AS, but a diagnosis of AS is still possible to establish. Usually, children undergoing speech therapy can quickly build a language system, though some other diagnostic features observed in the language and communication area can also be revealed.
- d) AS and autism. As noted before, difficulties with setting diagnostic criteria for the differential diagnosis of autism and Asperger syndrome have led to an update of the DSM in 2013 and the creation of a new entity, "autism spectrum disorder." Categorical differentiation of both disorders may render problems, but dimensional understanding makes it possible to accurately differentiate autism from Asperger syndrome, which provides the condition for building effective therapeutic and educational programs for children diagnosed with both clinical entities.
- e) AS and aphasia (SLI/DLD). Establishing the relationship between Asperger syndrome and aphasia took quite some time due to there being fewer cases and the initial resistance of diagnosticians and therapists who were using categorical diagnosis and did not acknowledge the possibility of serious, systemic language deficits to be present in AS, thus classifying them as symptoms of autism. Further clinical experience⁵ provided explanations for the relationship between the two disorders. For example, the presence of aphasia and Asperger syndrome may be linked to hereditary factors, while acquired aphasia may be the result of birth injuries. In these cases, apart from prototypic symptoms of Asperger syndrome, a serious, systemic language deficit is present.
- f) AS and bilingualism. The relationship between Asperger syndrome and bilingualism does not have a causal relationship because developing two language systems and functioning in two cultures do not trigger Asperger syndrome, nor do they contribute to the occurrence of symptoms. The relationship between the two, however, is significant in that biculturalism changes how AS may be observed, especially when considering behaviors belonging to two distinct areas of cultural practices. While being tested for Asperger syndrome, a child is assessed on how he or she is able to adapt to generally accepted standards of social behavior. If these rules are different, which does happen in the case of biculturalism, irrespective of whether the child functions in a monocultural family in the host country or in a bicultural family in the heritage country of one parent, the assessment of adaptability to social rules is more difficult. One more aspect should be considered in the relationship between AS and bilingualism; the situation described above may lengthen the diagnosis timeline because all

⁵ For example, boys diagnosed in the years 2019 and 2020; age: 3.2, 3.6; 5.0; 9.6; 10.11.

different forms of behaviors and emotions of a child are usually first attributed to the overlap of cultural models or the difficulties a child has in adjusting to a new environment.

- g) AS and Down syndrome and other genetic disorders. Asperger syndrome can co-occur with Down syndrome. The two phenomena stem from different causes. Down syndrome is linked to the occurrence of trisomy 21 (not inherited) or inherited ZA.
- h) AS and mental disorders. Depression is the most common co-occurring mental disorder in people with ZA. Both children and adults suffer from it. It can be primary or secondary and is often the result of social failure. Other mental disorders may also co-occur with ZA, e.g., psychosis, schizophrenia, and obsessive-compulsive disorders.
- i) AS and ADHD. Both Asperger syndrome and ADHD signify the presence of neuro-atypicality, or a difference of mind. Some features of both phenomena are similar, although the causes are not fully explained. Research into the relationship between ADHD and Asperger's syndrome is still ongoing.

5. Heterogeneity resulting from the overlap of relationships

Co-occurrence of the aforementioned factors makes Asperger syndrome a highly heterogeneous entity. A picture of a child with Asperger syndrome may significantly differ from an accepted prototype in relation to gender, age, and the degree of disorder, as well as the potential overlap of nosological constructs. However, effective therapy depends on the accurate diagnosis, and a better understanding of possible differences also helps children with less common types of the disorder. Distinguishing, for example, between dyslectic reading problems and manipulative behaviors that help to evade an unattractive or must-do activity changes therapeutic effects and the therapist's attitude towards the child's behaviors considerably. Only the right configuration of symptoms and a clear understanding of the origin of deficits will make the therapy most effective.

6. Conclusion

A dimensional approach to diagnosing developmental disorders, so well established in Western culture (Nisbett, 2015), requires breaking categorical, taxonomic preferences, as well as individual and analytical ways of perceiving reality. It requires considerable clinical experience to show the heterogeneity of disorders being assessed. Growing professional experience and evolving neurobiological knowledge should be used to build complex, dimensional models for diagnosing to make assessments more accurate, precise, and sensitive.

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