

Autism spectrum disorder and personality disorders: differential diagnosis or comorbidity?

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Abstract

Objective: The clinical overlap between autism spectrum disorder (ASD) and personality disorders (PDs) poses significant diagnostic challenges. Shared features – such as social communication difficulties, rigidity, and emotional dysregulation – often lead to misdiagnosis or delayed diagnosis, especially in adults and underrepresented populations (e.g., women).

Methods: This narrative review synthesizes current evidence on the differential diagnosis and comorbidity of ASD and PDs to guide clinical practice.

Results: Findings highlight substantial symptom overlap, particularly in social cognition and emotional regulation. For instance, both ASD and schizotypal PD may exhibit social withdrawal, while ASD and borderline PD share impulsivity and identity disturbances – though with distinct etiologies (neurodevelopmental vs. trauma-related).

Conclusions: Clinicians must adopt a developmental perspective to disentangle ASD from PDs, integrating longitudinal history, neuropsychological testing, and multidisciplinary evaluations. Future research should prioritize biomarker-based diagnostics and tailored interventions for comorbid presentations.

Keywords: Autism spectrum disorder, personality disorders, differential diagnosis, comorbidity, schizotypal personality disorder, borderline personality disorder.

Introduction

Autism spectrum disorder (ASD) is a condition that affects the development of functions related to sensory processing and social interaction. Although functional impairments at the behavioral level typically manifest in early childhood, they can persist throughout an individual's life.^{1,2} Similar to other neurodevelopmental disorders, significant changes in the quality and intensity of the core symptoms of ASD are expected to occur with advancing age, particularly in individuals classified as requiring level 1 support. These changes, which are associated with the natural progression of brain maturation, can vary in degree both within the same individual and among those diagnosed with ASD, leading to the emergence of traits associated with

autism in adolescence and adulthood. In the past, ASD was often viewed as a condition with manifestations limited to childhood and was rarely considered in the diagnostic assessments of adults.

In adults, diagnosing ASD presents a challenge due to its heterogeneity and the overlap of symptoms with other conditions. Making an accurate diagnosis can be difficult and labor-intensive, but it is essential for implementing appropriate adaptations and interventions, thereby avoiding unproductive or harmful treatments. Several factors complicate the diagnosis of ASD in adults, including distorted public perceptions and misinformation disseminated through social media. Existing diagnostic tools often overlook subtle or atypical presentations, particularly in underdiagnosed groups such as women and the elderly.³

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Because they share specific characteristics, such as the persistent nature of their presentation, traits associated with autism are often misinterpreted as fundamental personality traits of the individual. This misinterpretation contributes to the misdiagnosis of personality disorders.⁴ Not surprisingly, a prior diagnosis of a personality disorder is one of the most common mental health issues among adults who receive a late diagnosis of ASD, particularly in women.⁵ Furthermore, in individuals already diagnosed with ASD, many related emotional and behavioral manifestations raise questions and highlight the need for a more comprehensive discussion regarding the pathophysiology and the boundaries that delineate the comorbid presentation of ASD and personality disorders.

Since differentiating between ASD and personality disorders (PD) can be complex in certain clinical settings, there is a recognized need for a deeper understanding and additional markers to enhance diagnostic procedures.⁶ This narrative review aims to explore the relationship between ASD and personality disorders, emphasizing the perspectives of differential diagnosis and comorbidity between these two conditions.

Personality disorders: current perspectives

According to the current version of the Diagnostic and Statistical Manual of Mental Disorders¹ and the 11th Edition of the International Classification of Diseases,⁷ a personality disorder is characterized by a persistent, pervasive, and inflexible pattern of internal experiences and behaviors that significantly deviate from sociocultural norms and expectations. These patterns may be associated with impairments in cognitive, emotional, and interpersonal functioning, as well as impulse control. Manifestations of these disorders typically emerge during adolescence or early adulthood, making it essential to evaluate the individual's long-term patterns of functioning to establish an accurate diagnosis.

The typical personality characteristics of personality disorders can be distinguished from the symptoms and traits that emerge in response to specific situational stressors or from more transient or episodic mental states, as seen in conditions such as mood disorders or substance use disorders. The DSM-5-TR categorizes personality disorders into three distinct groups: Cluster A, which includes schizoid, schizotypal, and paranoid personality disorders characterized by odd or unconventional traits; Cluster B, which encompasses narcissistic, borderline, histrionic, and antisocial

personality disorders, often exhibiting dramatic, emotional, or erratic behaviors; and Cluster C, which includes obsessive-compulsive, dependent, and avoidant personality disorders.¹

The ICD-11, in contrast, offers a fresh perspective on the description and classification of PD. In this latest version, all previously recognized types of PD have been eliminated in favor of a global diagnosis that indicates the presence of deviations in personality development, accompanied by a specification of their severity (mild, moderate, or severe). These changes mean that the diagnosis of a personality disorder is determined by identifying impairments in various aspects related to⁸:

- a) Development of identity (self);
- b) Pattern of interpersonal functioning;
- c) Emotional, cognitive, and behavioral manifestations;
- d) Psychosocial impairment and distress.

In addition, the clinician can specify up to five domains of individual traits that correspond to the predominant psychopathological manifestations. An in-depth discussion of these features is beyond the scope of this review. Having provided an overview of the psychopathological aspects of PD, the following sections will discuss factors common to ASD that are essential for consideration in differential diagnosis.

Personality disorders in the differential diagnosis of ASD

The differential diagnosis is a crucial component that characterizes the semiotic phase of medical investigation. It requires the clinician to determine whether the criteria for a specific condition are met or if an alternative diagnosis may more accurately explain the collection of signs and symptoms observed. This decision also necessitates the identification of potential conditions that may coexist with the primary diagnosis.

Individuals diagnosed with ASD may exhibit affective and behavioral manifestations that often overlap with the psychopathology associated with certain PD. Both conditions are considered pervasive and persistent, leading to various negative outcomes in psychosocial functioning.⁹ The ICD-11 explicitly acknowledges that features of ASD may resemble those of PD and vice versa. As a general guideline for differential diagnosis, ICD-11 states that individuals with ASD should not receive an additional diagnosis of a PD unless there are distinct personality traits that cannot be solely attributed to ASD and its associated psychopathology, such as eating disorders, obsessive-compulsive disorder (OCD), self-injury, depression, or anxiety.

The development of self

Human identity is a broad, multidimensional construct in which self-perception (self-concept) emerges as the central component.¹⁰ Self-perception is defined as an individual's experiences and beliefs about themselves across various domains of life.^{10,11} Concurrently, the development of self-concept relies on insights gained from interpersonal experiences, particularly those related to secure attachments.¹² In this way, behaviors can be influenced, shaped, and even altered by social interactions and surrounding environments, which play a crucial role in the formation of individual identity.¹³

Individuals who exhibit deviations in personality development often experience either an excessively variable or excessively rigid sense of identity. For instance, they may struggle to maintain a coherent understanding of who they are across different social contexts, whether those contexts pertain to themselves or to others. In clinical practice, it is not uncommon to observe failures in the sense of permanence, which is directly linked to the formation of personal narratives and the natural changes that all individuals undergo from childhood to adulthood.¹⁴

Due to the heterogeneous nature of the phenotypic presentation of ASD, self-awareness can be an atypical experience for each individual on the spectrum. Elmore proposed a conceptual framework to understand the characteristics of self-awareness in individuals with ASD, which includes: (1) difficulties in identifying what they do not know, making it challenging for them to determine when and how to seek further knowledge; (2) difficulties in distinguishing between their own preferences and emotions and those of others in social situations; (3) difficulties in relating their own behaviors to environmental and social contexts, as well as to the actions of others; and (4) difficulties in comprehending their own thoughts and feelings, as well as those of others. These descriptions highlight the presence of "fragmented" perceptions of oneself, indicating a loss of certain elements in the differentiation between 'self' and 'other,' along with limitations in the dialectical construction of a self that engages minimally with its surroundings and with others.¹⁵

Empirical data indicate that adolescents with ASD may exhibit an "absent" or diminished psychological self, due to deficiencies in self-referential encoding, autobiographical memory, and future thinking.¹⁶ Autobiographical memories are crucial for constructing a sense of identity and play a directive role in shaping personal values, beliefs, decision-making, and goal-oriented behavior. Evidence suggests that individuals with ASD experience deficits in recalling

autobiographical memories, which may be linked to a lack of self-direction and self-sufficiency – a central feature of PD.^{17,18} Furthermore, low self-esteem, often observed in individuals with PD, can also manifest in many adolescents and adults with ASD.

Interpersonal functioning

Both PD and ASD are characterized by impaired interpersonal functioning, which can manifest in various ways and is associated with several social cognitive mechanisms. Social engagement emerges as a common factor in both diagnoses. In this context, individuals with PD may exhibit a diminished interest in forming relationships due to factors such as generalized avoidance, distrust, or fear of rejection. Conversely, individuals with ASD may primarily struggle with the regulation of social motivation. In both cases, the resulting behavior can present similarly: withdrawal and social isolation.¹⁹ It is important to note that, depending on the type of PD, similar outcomes (e.g., social isolation) can be observed, but with distinct underlying psychopathologies. For instance, individuals in group C may avoid social contact despite desiring and enjoying it from a distance, whereas individuals in group A and those with ASD may not seek social contact because they do not find it enjoyable.²⁰

Mentalizing refers to the domain of social cognition that encompasses an individual's ability to make attributions about their own mental states as well as those of others. It emerges as an essential skill for developing social reflective thoughts and for adjusting behavior in various interpersonal contexts.²¹ As a dynamic skill, mentalization can manifest in two extremes: it may occur excessively (hypermentalization), resulting in an over-interpretation of others' behaviors, or it may occur in a diminished capacity (hypomentalization), which can hinder the processing of diverse social cues and the inference of mental states.²²

Disturbances in Mentalizing skills are a common feature shared by ASD and PD. However, while individuals with PD often experience hypermentalization, ASD is primarily characterized by hypomentalization. This hypomentalization is thought to underlie the social and communication challenges faced by individuals with ASD.²²

Affective and emotional manifestations

Deficits in emotional regulation are a common factor observed in both ASD and PD. Case studies indicate that emotional dysregulation can significantly complicate the accurate diagnosis of PD, often resulting in misdiagnosis in favor of ASD. This misdiagnosis may

occur because ASD is not traditionally associated with emotional dysregulation.²³

Emotional dysregulation is defined as the impairment of the intensity and appropriateness of emotional expression in response to various stimuli and contexts.²⁴ It can manifest as hyperarousal in reaction to stressful stimuli (hyperreactivity) or hypoarousal (hyporeactivity), which is characterized by inexpressive or restricted affect. Both aspects of emotional dysregulation can be observed in individuals with ASD and PD. Common manifestations include anger outbursts, as well as low emotional awareness, recognition, and expression, which are prevalent in both conditions.

Regarding differential aspects, individuals with ASD may experience emotional distress and aggressive outbursts when confronted with specific triggers, such as changes in routine, aversive sensory stimulation, or unexpected events. This phenomenon is classically referred to as Goldstein's catastrophic reaction and Godot anxiety. Additionally, alterations in the affective and the end of a relationship, or the despair stemming from feelings of abandonment, can lead to an emotional and manifest as severe dysphoria in individuals with PD. Similarly, atypical sensory processing in ASD, characterized by hyposensitivity to interoceptive stimuli like pain or temperature, may resemble the inability often seen in PD to recognize and acknowledge difficult or unwanted emotions, a condition known as alexithymia.²²

At the behavioral level, individuals with ASD or a PD may struggle with coping strategies when confronted with stress overload. For instance, it is common to observe inappropriate behavioral responses to intense emotions and stressful situations in individuals with PD, such as self-harm. Similarly, individuals with ASD may exhibit self-injurious behaviors, including more severe actions, in response to emotional dysregulation and "meltdowns."²⁵

Cognitive flexibility and behavioral rigidity

A lack of adaptability to new experiences and circumstances, inflexible adherence to specific routines, persistent preoccupation with one or more special interests, and excessive compliance with rules are some of the central manifestations observed in individuals with ASD. Similarly, in PD, mentalizing emerges as the primary social cognitive function affected, albeit in a qualitatively distinct manner. This impairment leads to reduced accuracy in situational and interpersonal assessments, particularly in contexts of high emotional stress or ambiguity. Consequently, failures in mentalization often result in individuals

with PD exhibiting signs of inflexibility in their beliefs and rigidity in their behavior and thinking, as well as excessive adherence to rules.²⁶ This is especially true for individuals who display prominent anankastic characteristics, which are defined by rigid and systematic daily routines, excessive scheduling, and planning, making them difficult to distinguish from those with ASD. In both ASD and PD, cognitive rigidity is frequently linked to impairments in decision-making during uncertain situations or when confronted with unfamiliar experiences.²⁷ However, individuals with ASD may not only struggle with decision-making in uncertain contexts but may also exhibit resistance to such situations.

The role of neurodevelopmental hallmarks in diagnostic investigation

Although many diagnostic characteristics may overlap between PD and ASD, a comprehensive assessment of the developmental trajectory is essential for accurate differential diagnosis. This consideration is critical, as ASD is classified as a neurodevelopmental disorder. In general, current diagnostic systems indicate that PD are not typically diagnosed in children and adolescents. In contrast, ASD must, by definition, be present from early childhood. Even in cases where atypical personality traits emerge during adolescence, a gradual progression in the intensity of these traits is expected, culminating in a more stable and complete phenotypic expression in adulthood. The onset of ASD typically occurs in early childhood, with a natural tendency for the intensity of core symptoms to diminish as individuals age.

Current literature emphasizes that PD often emerge when individuals' life experiences fail to provide adequate support for typical personality development, particularly in relation to their temperament (i.e., the innate aspect of personality that reflects fundamental genetic and neurobiological processes). Clinicians should, therefore, consider the presence of PD in cases where an individual reports experiencing childhood adversities. It is also important to recognize that many girls who grew up with undiagnosed ASD may feel misunderstood and even neglected by well-intentioned parents and teachers throughout their lives. This issue is closely related to socioemotional development and warrants careful investigation. To enhance diagnostic accuracy, it is crucial to conduct a comprehensive assessment that takes into account current difficulties, developmental history, differential diagnoses, and the identification of therapeutic needs.²⁸

Personality disorders as a comorbidity of ASD

In the past decade, research on individuals with ASD has emphasized the importance of not restricting diagnostic assessments to full clinical presentations. Instead, it is crucial to identify subclinical or milder manifestations of the autistic spectrum. For the first time, subclinical autistic traits were examined among first-degree relatives of patients diagnosed with ASD, a phenomenon commonly referred to as the autism “extended phenotype.”²⁹ The focus on subclinical autistic traits is significant because they appear to adversely affect quality of life and serve as a considerable vulnerability factor for the development of other psychiatric disorders, as well as suicidal thoughts and behaviors.³⁰

Although the relationship between ASD and PD remains poorly understood, recent research has highlighted the similarities in certain symptoms and the potential for comorbid diagnoses. A recent literature review indicates that approximately 50% of individuals diagnosed with ASD may also meet the diagnostic criteria for at least one PD.⁹ Among these disorders, borderline personality disorder (BPD) has been one of the most extensively studied due to its higher prevalence and the associated clinical challenges.

Rinaldi et al. conducted a systematic literature review that focused on analyzing studies evaluating personality disorders as comorbidities of ASD.⁹ Among the studies exploring personality characteristics in individuals with ASD, only a limited number assessed PD as categorical diagnoses. Due to the overlap between autistic traits and various personality disorders, dimensional measures have been favored for assessing personality in adults with ASD. This methodological consideration alone does not clarify whether personality traits are integral to the autistic phenomenology or if they represent distinct categorical factors (comorbidity). Studies evaluating personality disorders as comorbid conditions with ASD indicate that prevalence varies according to the level of functional impairments associated with ASD, with lower prevalence observed in samples with intellectual disabilities. The most common comorbid personality disorders belong to Cluster A or Cluster C, including schizoid, schizotypal, obsessive-compulsive, and avoidant personality disorders. Additionally, significant phenotypic similarities have been noted between high-functioning ASD and personality disorders such as schizoid, schizotypal, and obsessive-compulsive, which may pose challenges in establishing categorical diagnoses. Furthermore, the considerable overlap

of symptoms has led researchers to propose the hypothesis that ASD in adults is linked to a distinct personality profile that may not align with either the diagnosis of ASD or a specific PD. Given the ongoing ambiguity in this area, it is generally recommended that the diagnosis of a personality disorder as a condition associated with ASD be considered a “true comorbidity” if it provides valuable insights into the patient’s functioning and aids in the development of more targeted treatments.

Conclusions and future perspectives

It is now recognized that ASD and personality disorders (PDs) share several common features. However, the precise nature of the relationship between ASD and PDs remains unclear. The overlapping symptom profiles of ASD and PDs can create diagnostic uncertainty; characteristics of ASD and PD may be misattributed, which can easily result in the misdiagnosis of patients with ASD.

PD and ASD are conditions characterized by significant and enduring functional impairments that can often be mistaken for one another, particularly in adolescents and adults, especially females. Both diagnoses exhibit overlapping features related to identity development (self), interpersonal functioning, and cognitive, emotional, and behavioral manifestations. This issue of similarity or overlap is explicitly acknowledged in the most recent version of the International Classification of Diseases (ICD-11). ASD-related features, such as persistent patterns of restricted, repetitive, and inflexible behaviors and/or excessive interests that begin in early childhood, are not characteristics of PD. However, it is important to recognize that females with ASD often exhibit fewer restricted and repetitive interests and behaviors than their male counterparts. Additionally, they frequently make exceptional efforts to compensate for their symptoms throughout childhood, adolescence, and adulthood. Consequently, identifying and differentiating these individuals during clinical assessments can be challenging. This difficulty is further compounded by the tendency of girls to withdraw socially and to exhibit emotional changes in response to their struggles with social adaptation (e.g., displaying heightened emotional reactivity). As a result, this presentation in girls may lead clinicians to mistakenly focus on maladaptive personality functioning or co-occurring mental disorders, rather than on the underlying and often masked atypical neurodevelopmental trajectory. There is an urgent need to investigate, first, how

clinicians make diagnostic decisions, and second, how to effectively address the challenges and difficulties they encounter during this process.⁶

Based on the discussions presented here, several improvements in clinical care, training, and research should be considered: (1) enhancing the techniques for differential diagnosis between ASD and PD during adolescence within the training curriculum for mental health professionals; (2) fostering a deeper understanding of the association with mental health issues, which complicates the differential diagnoses of ASD and PD; (3) studying more representative clinical populations that include adequate sample sizes of both women and men with ASD, with a focus on analyzing the association with PD; (4) improving the understanding and differentiation of social communication aspects between the two diagnoses, along with adjustments in intervention techniques at both behavioral and emotional levels; and (5) increasing clinical expertise in the early detection of ASD in women.

Disclosure

No conflicts of interest declared concerning the publication of this article.

Data availability statement

The data that support this study are available from the authors upon request.

Author contributions

Tiago Figueiredo: Conceptualization, Formal analysis, Investigation, Writing - original draft

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