

What misdiagnoses do women with autism spectrum disorder receive in the DSM-5?

Liliana Dell'Osso and Barbara Carpita* 

Department of Clinical and Experimental Medicine, University of Pisa, Pisa, Italy

Editorial

Cite this article: Dell'Osso L, and Carpita B (2023). What misdiagnoses do women with autism spectrum disorder receive in the DSM-5? *CNS Spectrums* 28(3), 269–270. <https://doi.org/10.1017/S1092852922000037>

Received: 05 January 2022

Accepted: 14 January 2022

Author for correspondence:

*Barbara Carpita, MD

Email: barbara.carpita1986@gmail.com

Recently, increasing literature is focusing on sex differences in the manifestations of autism spectrum disorder (ASD), highlighting the presence of several possible female-specific features of this condition.¹ Some authors also pointed out that ASD diagnostic criteria would be tailored on the typical male presentations of the disorder, leading to an under-recognition of ASD among females. Subsequently ASD females, in particular those without language or intellectual impairment, would likely receive other kinds of diagnoses, with a consequent negative impact on the course of the disease and on the treatment outcome.^{1,2} One of the first line of investigations in this field focused on anorexia nervosa (AN). Intriguingly, AN shows an opposite gender ratio when compared to ASD, featuring a strikingly higher prevalence among females but also a familiar aggregation with ASD.³ Moreover, the strong interest in diet and weight, together with the ritualized behaviors related to food preparation and consumption typical of AN were noted to resemble an autistic-like pattern of stereotyped interests and behaviors, although focused on food. AN patients were also reported to share with ASD the presence of social difficulties, lack of socioemotional reciprocity, and an altered theory of mind. Several epidemiological studies, including longitudinal ones, highlighted significant overlaps between AN and ASD diagnoses, while more recently the presence of significant autistic traits was reported also in other feeding and eating disorders (FED), such as bulimia nervosa or the emerging condition of orthorexia nervosa.³ These findings progressively provided support to the possibility of a reconceptualization of AN as a female phenotype of ASD, and, in parallel, increased the interest in investigating other sex-specific manifestations of the autism spectrum. Several authors reported possible features of female ASD phenotypes, suggesting that ASD females would focus on different kinds of interests with respect to males, including fictions, celebrities, or fashion.^{2,3} In addition, ASD females would often show a milder impairment in socioemotional reciprocity and would be more oriented toward interacting with others, usually adopting camouflaging behaviors in order to cope with social interactions. ASD females would be more aware of their social difficulties and the need of being, or at least appearing to be, socially integrated: however, the continuous use of camouflaging strategies would result in greater levels of social anxiety, distress, and depression, leading to live every relational situation as a performance.² Noticeably, social anxiety disorder (SAD) is another condition which was described as more common among females and might be considered as one of the possible diagnoses that ASD females could receive: among females without language or intellectual impairment, social anxiety symptoms, together with the eventual use of camouflaging strategies, may mask autistic-like social difficulties as well as the autistic-like tendency toward focusing on lonely activities and interests. Further support to this hypothesis may come from findings that highlighted social brain alterations and impaired theory of mind networks among patients with SAD. It should be noted that SAD itself was reported to be under-diagnosed and often confused with nonpathological shyness, and this fact may have contributed to further preventing women in the autism spectrum from reaching clinical attention.^{2,4,5}

FED and SAD may be not the only diagnoses, which females with ASD are likely to receive. Subjects with borderline personality disorder (BPD), a condition mostly diagnosed among females, may show traits that are also typical of ASD, such as reduced empathy and social-emotional reciprocity, difficulties in regulating emotions, altered reactivity and reactions to stimuli or outbursts of anger, increased self-injuring and/or suicidal ideation or behaviors. An increased prevalence of ASD or subthreshold autistic traits was reported among patients with BPD, while, in turn, ASD patients show a greater frequency of BPD.⁶ To better understand the possible link between these two conditions, the role of trauma and stress-related psychopathology should be specifically taken into account. BPD patients are known to usually report a history of traumatic events, while emerging evidence is suggesting that ASD, or also subthreshold autistic traits, could be considered as a vulnerability factor toward developing trauma and stress-related conditions. It should be noted that, according to the complex Post-traumatic stress disorder (PTSD) (cPTSD) model, subjects with increased vulnerability may develop a peculiar form of PTSD after traumatic events of milder intensity with respect to those described in DSM-5 criterion A for PTSD diagnosis, in particular, if events were repeated or prolonged in time, such as in the case of interpersonal traumatic events. cPTSD features a chronic course, dissociative symptoms and negative alterations in cognition and mood, emotional lability, maladaptive behaviors, instability in self-perception, and interpersonal relationships. The similarities

between the cPTSD clinical picture and BPD have been previously stressed, leading to hypothesize that subjects with cPTSD may often receive a diagnosis of BPD.^{1,6} ASD individuals, due to their social difficulties and altered socioemotional reciprocity, show a higher risk to be exposed to socially stressful or even traumatic events, bullying and rejection, subsequently developing trauma and stress-related conditions.^{1,6} In addition, ASD subjects were reported to show difficulties in recognizing, processing, and externalizing traumatic experiences, with a poorer ability to cope with stressful events. This feature may increase the vulnerability toward developing stress-related symptoms after milder life events, but also the likely that PTSD symptoms remain under-reported and under-recognized in this population.^{1,6} As mentioned above, ASD remains more frequently under-diagnosed among females, while cPTSD may be commonly confused with BPD. Subsequently, it could be hypothesized that female ASD subjects could be more frequently misdiagnosed with BPD when a history of (mild) traumatic events is present.

It is of particular interest to remind that PTSD itself is more frequently diagnosed among females, and that BPD is not the only female-typical disorder, which features a history of trauma: Somatic symptom and related disorders (SSRD) have been associated in the literature with the presence of under-reported traumatic experiences, and show a higher prevalence among females. This diagnostic group includes several clinical pictures that would have previously fallen, together with dissociative disorders, under the broad label of hysteria, a medical condition considered typical of women, starting from its name. In line with a possible link between SSRD and female phenotypes of autism, ASD patients, and in particular females, were reported to show a greater prevalence of somatic symptoms, possibly due to the difficulties in mentalizing and expressing psychological distress.⁷ Moreover, the altered perception and presentation of symptoms typical of this population, ranging from over-sensibility and dramatization to apparent indifference (*“la belle indifférence”*), may be an expression of the hypo/hyperreactivity to sensory inputs of ASD patients. It was recently stressed that ASD, may feature a reduced adaptive interoceptive attention and body awareness, with difficulties in understanding body signals and bodily feelings related to emotions, and, on the other hand, an increased maladaptive awareness toward anxiety-induced somatization.^{2,7} Similarly, it might be hypothesized that the altered, artificial communication style of SSRD patients, may result from an unsuccessful use of social camouflaging strategies in order to overcome the impairment in communication skills and socioemotional reciprocity. The pattern of falsification of symptoms in Factitious disorder should also be considered in light not only of the dissociative symptoms typical of subjects with BPD and/or PTSD, but also of the habit to camouflage typical of females with autism.^{2,7} Noticeably, BPD is known to be frequently comorbid with both FED and SSRD, supporting the inclusion of all these conditions in the range of possible female presentations of the autism spectrum.¹⁻³

These considerations should promote a careful investigation of all the clinical pictures with a typical female prevalence, in order to

evaluate possible differential diagnosis with autism spectrum conditions. Another example may be that of anxiety disorders. Autistic-like features such as difficulties in adjusting to the environment, inflexibility, and hyperreactivity to stimuli may promote the presence of anxiety symptoms and panic attacks. On the other hand, while anxiety and panic disorders were often described in ASD, these conditions show in the general population a higher prevalence among females, where they might mask the presence of an under-recognized ASD or eventually of subthreshold autism spectrum symptoms.^{1,2,4}

Recent research is highlighting the importance of a neurodevelopmental approach to psychopathology, hypothesizing that a neurodevelopmental alteration may be at the basis of different psychopathological conditions: the specific kind, gravity and timing of the alteration may result, interacting with other factors, in different illness trajectories, shaping the clinical pictures as described in the DSM.¹ It is possible that sex, as a genetic and biological factor, may play a major role in influencing the specific kind of psychopathological course starting from a similar neurodevelopmental alteration. On the other hand, gender may influence the kind of environmental factors to which subjects could be exposed during lifetime. Deepening the investigations on female ASD phenotypes may lead to reconsider the sex ratio in ASD prevalence, but also to reach a better understanding of several psychiatric conditions often diagnosed among females and to reconceptualize the whole matter of gender differences in psychiatry, which should be revised in light of a neurodevelopmental approach.

Disclosure. The authors do not have any competing interests to disclose.

References

1. Dell'Osso L, Dalle Luche R, Maj M. Adult autism spectrum as a transnosographic dimension. *CNS Spectr*. 2016;**21**(2):131–133.
2. Dell'Osso L, Lorenzi P, Carpita B. Camouflaging: psychopathological meanings and clinical relevance in autism spectrum conditions. *CNS Spectr*. 2021;**26**(5):437–439.
3. Carpita B, Muti D, Cremonese IM, Fagiolini A, Dell'Osso L. Eating disorders and autism spectrum: links and risks. *CNS Spectr*. 2020;**9**:1–9.
4. Dell'Osso L, Abelli M, Pini S, *et al*. The influence of gender on social anxiety spectrum symptoms in a sample of university students. *Riv Psichiatr*. 2015;**50**(6):295–301.
5. Bas-Hoogendam JM, Westenberg PM. Imaging the socially-anxious brain: recent advances and future prospects. *F1000Res*. 2020;**9**:F1000 Faculty Rev-230.
6. Dell'Osso L, Cremonese IM, Amatori G, *et al*. Investigating the relationship between autistic traits, ruminative thinking, and suicidality in a clinical sample of subjects with bipolar disorder and borderline personality disorder. *Brain Sci*. 2021;**11**(5):621.
7. Trevisan DA, Mehling WE, McPartland JC. Adaptive and maladaptive bodily awareness: distinguishing interoceptive sensibility and interoceptive attention from anxiety-induced somatization in autism and alexithymia. *Autism Res*. 2021;**14**(2):240–247.