

Positive and differential diagnosis of autism in verbal women of typical intelligence: A Delphi study

Autism
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Abstract

Diagnostic criteria for autism are relatively vague, and may lead to over and underdiagnosis when applied without clinical expertise. Indeed, autism is best reliably identified by experienced clinicians who take into account qualitative aspects of the condition. When assessing for autism in women, little guidance exists to support clinicians deciding whether to attribute adaptive difficulties to autism, a psychiatric condition, or both. The purpose of this study was therefore to propose guidelines for clinicians assessing for autism in women. To do this, we aimed to describe the clinical expertise involved in making positive and differential diagnoses of autism in adult women of typical intelligence. We interviewed 20 experienced clinicians from seven countries. We then elaborated Delphi statements summarizing participant views on the topic, which our participants rated. We obtained a final list of 37 suggested clinical guidelines to improve specificity and sensitivity of autism diagnosis in women. Participants had developed individual assessment strategies, although much overlap existed across participants. Participants provided insight to differentiate autism from post-traumatic stress disorder and Borderline Personality Disorder, and underlined the importance of being able to make differential diagnoses particularly in cases where non-autistic people had strongly self-identified with the spectrum.

Lay abstract

The diagnostic criteria for autism are relatively vague and can lead to both under- and over-diagnosis if applied as a checklist. The highest level of agreement that a person is autistic occurs when experienced clinicians are able to make use of their clinical judgment. However, it is not always clear what this judgment consists of. Given that particular issues exist when assessing for autism in adult women, we wanted to explore how expert clinicians address difficult diagnostic situations in this population. We interviewed 20 experienced psychologists and psychiatrists from seven countries and discussed how they conducted autism assessments in adult women. We then came up with a list of 35 statements that described participant views. Our participants completed an online survey where they rated their agreement with these statements and provided feedback on how the statements were worded and organized. We obtained a final list of 37 suggested clinical guidelines. Participants agreed that diagnostic tools and questionnaires had to be coupled with judgment and expertise. Participants felt that trauma and Borderline Personality Disorder could be difficult to differentiate from autism, and agreed on some ways to address this issue. Participants agreed that self-identification to the autism spectrum was frequent, and that it was important to provide alternative support when they did not ultimately diagnose autism.

Keywords

adult, autism, camouflaging, differential diagnosis, women

The *Diagnostic and Statistical Manual of Mental Disorders* (5th ed.; DSM-5) diagnosis of autism is categorical and obtained when a clinician determines that a threshold of relatively abstract features which make up autism criteria, such as “deficits in social-emotional reciprocity,” has been reached (American Psychiatric Association, 2013). These features can then vary according to four clinical specifiers: language, intelligence, comorbidity, and support needs,

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despite all cases meeting the same diagnostic criteria. Age and sex represent additional sources of variation that contribute to the widening, and therefore heterogeneity of possible phenotypes consistent with autism diagnostic criteria (Motttron & Bzdok, 2020). Clinical difficulties are inherent to extreme values of the clinical specifiers, as they question the boundaries of the autism phenotype. In this context, positive and differential diagnosis of autism in women of typical intelligence, with no speech-onset delay and relatively low support needs, has been met with much research and clinical interest.

Most standardized autism diagnostic and screening tools are not normed to consider gender differences in the general population (Constantino & Charman, 2012). For example, superior verbal abilities typically found in girls may lead to underdiagnosis of autistic girls (Wing, 1981). Indeed, autistic girls may have more typical narrative skills than autistic boys as measured on the Autism Diagnostic Observation Schedule (ADOS) storytelling task (Boorse et al., 2019; Parish-Morris et al., 2017). They are also rated more positively than autistic boys by naïve confederates, despite experienced clinicians rating their socio-communicative difficulties as equal to autistic boys (Cola et al., 2020). Clinicians may more likely attribute the difficulties of autistic women to other conditions more frequent in women, such as mood, eating disorders, or Borderline Personality Disorder (BPD) (Au-Yeung et al., 2019) or social anxiety (Hull et al., 2019), resulting in under diagnosis of autism. Conversely, some adult women lose their autism diagnosis when re-evaluated by autism experts, in favor of diagnoses such as depression, BPD and Attention Deficit Hyperactivity Disorder (ADHD) (Tak, 2020), but this may cause psychological damage (De Bucy, 2018).

A sex ratio in favor of males is one of the few uncontested markers of autism (Werling, 2016), but where this male:female ratio truly lies is now a contentious topic. Discrepancies in the sex ratio are largely found in autistic people with no speech-onset delay and no intellectual disability, formerly known as Asperger's Syndrome (Loomes et al., 2017). The possibility of an artificially inflated sex ratio due to an under recognition of autistic women and girls has generated a large number of studies over the past decade (Lai et al., 2015). While approximately four boys are diagnosed with autism for every girl, population samples have found lower sex ratios ranging from 2:1 to 3:1 (Loomes et al., 2017), suggesting gender-based diagnostic disparities.

Much research has focused on the mechanisms by which girls on the spectrum may go unnoticed. Heightened social motivation in girls and women (Sedgewick et al., 2016) may facilitate the phenomenon of camouflaging, or the conscious and unconscious "masking" of autism signs.

Camouflaging has been self-reported both in late-diagnosed autistic women, presumably missed in childhood (Bargiela et al., 2016) but also in girls diagnosed in childhood (Tierney et al., 2016). Participants of these accounts consistently detail the great amount of energy they put into trying to appear neurotypical and the mental health cost of attempting to camouflage their autism (Bargiela et al., 2016; Hull, Petrides, et al., 2017; Milner et al., 2019). However, the notion of camouflaging as a clinical indicator of autism, particularly for women, may lack rigor. This may open up the possibility of identifying any psychological suffering associated with social interaction as "autistic camouflaging." For the clinician, it may justify the displacement of the diagnostic threshold up to and including situations where no signs of autism are actually visible, resulting in non-falsifiable diagnoses (Fombonne, 2020). The devaluation of the clinical threshold (Constantino, 2011) and less stigmatizing nature of an autism diagnosis in comparison to some psychiatric conditions like personality disorders (Nylander, 2015) may create a situation of overdiagnosis of autism in adult women of typical intelligence.

The existence of a "female phenotype" is widely cited although little evidence suggests that autistic women differ from men beyond typical sex differences found in the general population. A recent meta-analysis and systematic review of behavioral and cognitive sex/gender differences in autism concluded that "individuals with Autism Spectrum Conditions (ASC) are fundamentally similar to typically developing individuals in regard to their sex/gender variation in core ASC characteristics" (Hull, Mandy, et al., 2017, p. 723).

In the absence of a neurobiological gold standard for diagnosis and related clinical guidelines, one option may be to investigate the decision-making process of expert clinicians who assess adult women of typical intelligence for autism. An existing framework, built by clinicians and researchers seeking to guide research on sex/gender differences in autism, indeed suggests that methods examining endorsement rates from clinicians could be useful in refining autistic phenotypes (Lai et al., 2015). Lai et al.'s framework further identifies two research areas which could benefit from sourcing clinical expertise: (1) defining autism in males and females (nosological challenges) and (2) diagnosing autism in males and females (diagnostic challenges). More specifically, the authors call for qualitative research on behavioral exemplars of autism in women and question how co-occurring conditions or cognitive/temperamental factors may influence the presentation and identification of autism, and whether gender-based interpretation biases may play a role. The Delphi method is a structured process by which a group of participants, selected for their expertise on a particular topic, are presented with statements over several rounds which they review, rate, and offer suggestions on. The Delphi method is frequently used to source expertise from clinicians in

order to develop clinical guidelines and select research outcomes (Boulkedid et al., 2011; Creamer et al., 2012; Spain & Happé, 2020).

Expert clinicians, as defined by extensive exposure to autistic people, have better inter-rater reliability on autism diagnosis when allowed to make use of their clinical acumen, rather than using a standardized checklist of symptoms (Klin et al., 2000). By referring to a behavioral phenotype acquired with experience, expert clinicians incorporate (de Marchena & Miller, 2017) and hierarchize (Muggleton et al., 2019) signs of autism not included in diagnostic manuals, such as gait and prosody (intonation and rhythm of speech).

The purpose of this study was to propose guidance for clinicians assessing for autism in adult, verbal women of normal-range intelligence quotient (IQ). To do this, we aimed to establish areas of consensus for conducting autism assessments in adult women, based on the expertise of clinicians with a large experience of diagnostic assessment of autism in women.

Method

Study design

This Delphi study used content analysis (Stemler, 2000) to analyze semi-structured interviews conducted with clinicians experienced in assessing for autism in adult women and to develop statements to be used in developing a Delphi survey.

Sample

Participants were 20 clinicians from seven different countries with expertise diagnosing autism in adult women of typical intelligence. Sample size was decided by previous literature indicating that the majority of Delphi studies include between 15 and 20 respondents, prioritizing a small group of expert and motivated participants (Hsu & Sandford, 2017). Participants were recruited through Twitter, mailing lists, and word of mouth. Only one participant was recruited through social media, and the vast majority of participants heard about the study through colleague referrals. Participants filled an information and consent form on a secure data collection platform (REDCap), and agreed to be recontacted for validation purposes. Interviews were conducted in French or in English, by phone ($n=2$) or videoconference ($n=18$).

Our inclusion criteria were based on a previously conducted behavioral phenotyping study, in which 151 participants with experience diagnosing autism were asked to estimate the total number of autism diagnoses they had ever given (de Marchena & Miller, 2017). Marchena and Miller found a median of 250 diagnoses across their sample, which was the minimum experience we required for

participation. Furthermore, we added the constraint of having practiced for at least 5 years and assessed at least 100 women in this time. Awareness around gender-based diagnostic disparities has increased over the past years, and we aimed to recruit clinicians with specific experience in this.

Participants practiced in psychiatry, clinical psychology, and speech and language therapy (mandated to conduct autism assessments in their country), and had 5–40 years of experience diagnosing autism (mean (M)=18.3 and standard deviation (SD)=10). The mean number of adult female assessments per year was 35 (SD =19.5). Detailed participant characteristics are included in Supplementary Material.

Procedures

As a first step to developing Delphi survey items, we built a semi-structured interview guide to collect initial participant views, based on the following four main categories: (1) factors of complex assessments in adult women, (2) methods for running such complex assessments, (3) signs indicative of autism, and (4) differential diagnoses and comorbidities. The interview guide (Supplementary Material) was tested and refined by three clinicians external to the research team, and translated into English by J.C. and two bilingual researchers with the use of back-translation (Chen & Boore, 2010).

Interviews were conducted by J.C. and transcribed verbatim by a transcription service. The first half of the interviews were annotated by J.C. and L.M., in order to ensure that material of interest was not missed. Content analysis was used as it allowed us to code our data into four predefined main categories (a priori coding) according to the literature and our research aims (Mayring, 2000). A working analytical framework of subcategories was developed by J.C. based on these preliminary codes (emergent coding) and checked for face validity by the senior author. The second half of the interviews were then coded by a research assistant using the same framework. A consensus approach was used to iteratively refine the subcategories. NVivo software was used to chart subcategory frequencies. These subcategories were then used in the elaboration of a Delphi survey.

Subcategories which were referred to by at least 50% of participants in their interviews were included in the survey as opinion statements that participants could indicate agreement or disagreement with. For example, the subcategory “High empathy of autistic women” became a Delphi statement reading “Autistic women often demonstrate high emotional empathy towards others, but poor understanding/awareness of their own emotions.” A list of 35 statements of opinion was developed across the four main categories. This list was presented as an online survey through REDCap. The 20 participants interviewed for the

study were invited via email to fill out this survey derived from their interviews. Participants were asked to rate each statement on a 4-point Likert-type scale (“Strongly Agree,” “Somewhat Agree,” “Somewhat Disagree,” and “Strongly Disagree”), or could indicate “Not relevant to my practice.” Delphi studies often suffer from high attrition rates over several rounds (Hsu & Sandford, 2017). To address this, we attempted to keep the survey short by only asking participants to comment on items they had rated as “Somewhat Disagree,” to indicate how the statement could be modified to better fit their experience. Half of participants required two or more email reminders in order to fill out the survey.

The results of the Delphi survey were then analyzed. Items were considered to have reached consensus when 70% of the 19 participants indicated that they “Strongly Agreed” or “Somewhat Agreed” (Hsu & Sandford, 2017). Ratings for Round Two are summarized in Supplementary Material. Following this round of feedback from participants, four statements did not achieve consensus. Of these statements, three were reformulated and one was split into two statements according to participant comments. One statement was added according to participant comments on another statement. A final list of 37 agreed upon suggested guidelines was sent out to participants via email. Due to the high number of reminders sent out in the first round, a final survey was not conducted in order to avoid attrition, and participants were instead asked to respond via email indicating whether they wanted to bring any last changes to the final list of guidelines. In total, 14 participants responded with their final approval within the provided timeline, and no further changes were made.

Community involvement

This study was conceived according to research priorities identified by clinicians (colleagues and collaborators). Clinicians working in the field of autism were involved in developing all aspects of the study (interview schedule, Delphi items, and final interpretations). We would like to note that, despite its good intentions, our autistic collaborators have expressed concern about the inclusion of community involvement statements. These may act to pressure autistic people to “out” themselves, and risk restricting their role in research to one of community stakeholders, when in fact autistic people can and do participate as researchers.

Results

The results are summarized below according to the four main categories explored during the interviews and the analyses, namely: (1) factors of complex assessments, (2) managing the complex assessments, (3) signs indicative of autism, and (4) differential diagnosis and comorbidities. A

full list of Delphi statements can be found in Table 1. Of our 20 initial participants, 19 completed Round 2 (95% completion) (Note: quotations which made use of stigmatizing language were reworded for similar meaning, indicated by square brackets.).

Factors of complex assessments

This category sought to identify the factors specific to both women and autism which made assessments more complex for clinicians, such that they may decide to undertake a longer assessment.

Self-diagnosis and history. Participants overwhelmingly agreed that self-diagnosis prior to clinical assessment had increased in recent years, with information about autism increasingly available online. Many participants specified that self-diagnosis was correct in many cases, but participants ultimately reached consensus that extensive research prior to appointments could complicate assessments. More specifically, a clear belief or hope that the assessment would result in an autism diagnosis was seen as a potential source of bias, where the person seeking a diagnosis would describe their life history and behaviors through the lens of the research they had done. Participants indicated high levels of agreement that many women seeking autism assessments had complex psychiatric and life histories, namely, multiple previous psychiatric diagnoses and a high number of Adverse Life Events.

Camouflaging. Autistic women having learned certain social contingencies which could make them appear more neurotypical (camouflaging) made it more difficult to observe whether social difficulties were autistic in nature.

Stigma. A diagnosis of autism can provide a feeling of belonging to a community, and some clinicians felt that the autism as a social identity resonated particularly with their female patients. Many clinicians indicated that autism was seen by their patients and clients as more socially acceptable than a mental health condition, which could complicate the process of making a differential diagnosis and receiving a stigmatizing label.

Negative reactions. Reactions of disappointment, confusion, and/or anger sometimes occurred when assessments did not result in diagnosis of autism. Most clinicians took this possibility into account before and during their assessment, for example, by enquiring about depressive symptoms which could increase after receiving disappointing news. Some clinicians felt that lengthy waitlists exacerbated the issue, with some patients waiting years for an assessment that they therefore placed very high expectations on.

Table 1. Final Delphi statements.

Category	Delphi statements
Factors of complex assessments	<ol style="list-style-type: none"> 1. Autistic women have learned certain social contingencies allowing them to appear more typical 2. Autism is increasingly mediatized and information available online, which has increased rates of self-diagnosis prior to clinical assessment 3. Autism is regarded more positively than most psychiatric diagnoses, it is seen as a social identity which can give access to a community and provide a feeling of belonging 4. Women seeking autism assessment often have complex histories and multiple previous mental health diagnoses 5. Self-diagnosis of autism prior to assessment can sometimes complicate the assessment <ol style="list-style-type: none"> a. <i>Minority opinion: self-diagnosis is often correct</i>
Managing complex assessments	<ol style="list-style-type: none"> 6. Disappointment, confusion, and/or anger can occur when a patient is not given a diagnosis of autism 7. Standardized assessment tools are not equipped to detect autism in adult women of typical intelligence 8. Self-report questionnaires can lack specificity and be biased by the patient's knowledge about autism 9. Self-report questionnaires can provide material to explore in an interview, especially when questionnaires contradict each other or the clinician's observations 10. Diagnostic assessments should ideally be long and run over more than one session, to observe functioning once patient gets tired, and assess several diagnostic hypotheses with relevant tools 11. During the assessment, the person should be challenged with spontaneous interaction to observe how they handle unfamiliar situations 12. The person's difficulties should ideally be corroborated by an external informant who knew them in childhood 13. Asking for specific personal examples can help to confirm that difficulties are based on lived experience rather than patient's research 14. It is useful to manage patient expectations by explaining early on that assessment may not result in diagnosis of autism 15. It is important to draft the final report in collaboration with the patient and share content transparently 16. In cases where autism is not diagnosed, it is important to validate the patient's difficulties and offer other avenues for support or alternative diagnoses 17. The concept of "autistic traits" is useful to explain to patients why a diagnostic threshold of autism was not reached <ol style="list-style-type: none"> a. <i>Minority opinion: this is a possible slip into "we're all a little bit autistic," diagnosis is categorical</i>
Signs indicative of autism	<ol style="list-style-type: none"> 18. Differences can be noticed over time in the more nuanced aspects of social behavior beyond eye contact and prosody, such as topic maintenance, social inferences, and reciprocity 19. Autistic deep interests are ego-syntonic, exhaustive, and cyclical 20. Autistic women often report investing large amounts of energy preparing for social interactions and feeling drained following the interaction 21. Autistic women have often not reached the level of professional/personal achievement expected given their apparent intelligence 22. Autistic women have often made numerous independent attempts to adapt and overcome their difficulties 23. Autistic women often demonstrate high emotional empathy toward others, but poor understanding/awareness of their own emotions 24. Autistic women are often able to recognize their own functioning in that of other autistic people 25. Autistic women are often able to apply their special interests and use them as social currency 26. Autistic women tend to have few or no true peer relationships and to be naïve in their relationships 27. In autistic women, the pursuit of diagnosis is rarely utilitarian but rather about self-knowledge 28. Compared to women who go on to receive other diagnoses, autistic women may require more prompting or guidance to fill out questionnaires and provide information during the assessment 29. In autistic women, gender may be expressed more fluidly, with less attachment to the gender binary, or femininity may appear forced/rehearsed
Differential diagnosis and comorbidities	<ol style="list-style-type: none"> 30. A current acute mental health episode (e.g. depressive episode) can make it difficult to determine baseline functioning to diagnose autism 31. For an experienced clinician, diagnosing autism relies on thorough, reliable assessment, coupled with a "feeling in the room" 32. The chronology of difficulties is extremely important when making differential diagnoses. 33. Most women presenting for an autism assessment have experienced trauma in some form 34. Borderline Personality Disorder is highly present in autism assessment clinics as a past diagnosis and/or a potential differential diagnosis

(Continued)

Table 1. (Continued)

Category	Delphi statements
	35. Autistic women can superficially present with signs resembling Borderline Personality Disorder
	36. Borderline Personality Disorder can be differentiated from autism by exploring the person's understanding of neurotypical social dynamics, and how they describe their emotions
	37. Borderline Personality Disorder can be differentiated from autism by exploring whether attachment difficulties are present

Managing complex assessments

This category addressed how clinicians adjusted their practice during complex assessments including tools and strategies used.

Clinical instruments. Standardized assessment tools were judged as unequipped to detect autism in adult women of typical intelligence. The ADOS was largely seen as inducing false negatives, particularly in girls and women of normal-range IQ. It was also noted by several clinicians that anxiety and mood disorders could skew ADOS results and artificially inflate scores to induce false positives.

Self-report. Self-report measures could lack specificity and be biased by the patient's knowledge about autism. Many participants specifically mentioned that the Autism Quotient (AQ) was widely available online and relatively easy to fill out according to the desired results. However, self-report measures provided useful information when they contradicted each other or clinical observations. A high AQ score with few observable signs of autism could indicate camouflaging or over-reporting of symptoms, and clinicians would explore these discrepancies in their conversations with the person.

Assessment duration and flexibility. Long assessments were judged necessary, to observe functioning in challenging social situations (unfamiliar person, fatigue), and to assess several diagnostic hypotheses. Many clinicians indicated that women having missed out on diagnosis in childhood were often able to present as neurotypical for an amount of time, and that it took several hours or appointments for difficulties to become apparent. These long assessments were also used to rigorously rule out other diagnostic possibilities, even in cases where clinicians were only mandated to assess for autism.

Provoking spontaneity. Creating moments for spontaneous interaction was favored, in order to evaluate how the person coped with unpredictability in social interactions. Many clinicians used humor to see if they could easily elicit a fluid conversational back and forth with the person.

Information sources. The person's present and past difficulties needed to be corroborated by an external source, ideally one having known the person in childhood.

Whether or not a childhood informant was available, participants widely recommended collecting information from third parties. Interestingly, there was no general consensus on the type of information participants were looking for by soliciting third party opinions. Some clinicians indicated that they were looking to understand why the person was considered disabled, as this was crucial to whether or not they could give a diagnosis. For other clinicians, the current presence of a disability was less important, and they specifically sought out early childhood signs of autism. Past healthcare providers were also cited as particularly important pieces of information. For example, clinicians spoke to past therapists to better understand how the person communicated and interpreted emotions.

Providing examples. Clinicians highlighted the importance of validating adaptive difficulties and autistic behaviors by requesting specific personal examples. This allowed clinicians to differentiate lived experience from the product of patient research. Many clinicians spoke of patients giving "textbook answers" (P05) based on their reading, and asked follow-up questions, for example, to enquire about the depth of a topic cited as a special interest.

Facing disappointed patients. When facing a patient disappointed because a diagnosis of autism had not been made, it was crucial to validate the difficulties that had brought them to seek out assessment, "We are never saying to them 'you [have nothing to worry about]', we would be helping them think about what else is going on" (P11). It was important to offer alternative diagnoses where possible and some clinicians indicated that diagnoses such as ADHD and anxiety could be helpful to patients especially when framed as a condition that they could get medication and therapy for. Clinicians indicated that final reports were better received when drafted in collaboration with the patient, and contents shared transparently.

Autistic traits. Invoking "autistic traits" could be helpful to soften disappointment in women who did not obtain the diagnosis: "Sometimes I'll just say 'I think you clearly do have some of these traits' and I'll explain the concept of a bell-shaped population curve and there's always a question of where you draw the line" (P15). However, a minority opinion of participants who strongly disagreed emerged,

explaining that they considered diagnosis to be categorical and that invoking autistic traits risked sounding like a diagnosis of “mild” autism.

Signs indicative of autism

This category explored the specific traits and behaviors clinicians personally ascribed value to when evaluating for autism in women, whether or not these were included in diagnostic criteria.

Subtle understanding of neurotypical socio-communicative rules. Beyond eye contact and prosody, clinicians recommended investigating abilities such as topic maintenance, social inferences, and reciprocity, and exploring the person’s understanding of neurotypical social rules in their interactions. While some participants felt this was not specific to women, others felt that this aspect of meta-communication was crucial due to women’s increased ability to act neurotypical. Participants also agreed it was important to investigate time spent preparing for and recovering from social interaction, but again some participants did not see this as necessarily specific to women.

Deep interests. Participants described autistic deep interests as ego-syntonic, exhaustive, but also cyclical, “it’s not so much that they lose interest, but they move on to something else once they realize they have drawn all possible functional benefits out of the interest” (P02). Clinicians explored the extent to which the interests were truly exhaustive in nature, and saw the person’s ability to elaborate on their interest as crucial to the diagnostic interview. Clinicians also agreed that autistic women often had “useful” deep interests, which they could apply to facilitate social interaction.

Professional accomplishments. Autistic women, despite presenting as intelligent, had often failed to achieve expected levels of personal/professional success. Autistic women had usually taken independent action to compensate for their difficulties, investing a great deal of effort into these attempts. This was particularly significant when considering alternative diagnoses such as personality disorders, in which people may find it difficult to follow through on trainings or therapy. In comparison, a participant noted

autistic women had had CBT for anxiety for many years and it’s never helped, so they got stuck, and they’ve obviously been willing to look at issues and explore ways of improving their lives, but they just can’t seem to get out of it. (P04)

Emotional empathy. High emotional empathy seemed to be a characteristic of autistic women, despite difficulties with cognitive empathy and poor understanding of their own emotions. Many clinicians interestingly noted that autistic

women were often able to recognize their own functioning in that of other autistic people. Compared to men, this was often how they had arrived at self-diagnosis, “I am autistic because I look like this person” (P07).

Requiring instructions. Autistic women required prompting and specific instructions to provide information during assessments. Some clinicians, for example, made use of visual aids like photographs brought in by the person, or homemade composite images, to elicit conversations about emotions when this was difficult to do spontaneously. Dichotomous questionnaires about their own behaviors were often difficult to fill out for autistic women, who tended to circle vague items or leave them blank. Some clinicians, however, pointed out that this was not necessarily specific to women, but that it could be an interesting trait to look for when making differential diagnoses.

Asymmetrical relationships. Clinicians agreed that peer relationships with neurotypical individuals tended to be rare, indicating that they would look for atypical understandings of friendship (e.g. someone working in customer service whom the person regularly saw) or other “mismatches” such as in age. This asymmetry was also identified as one of the mechanisms that could put autistic women at greater risk of being victims of abuse in relationships, as they rarely had a peer group to compare their experiences to.

Purpose of diagnosis. Clinicians agreed that, in women, the purpose of diagnosis rarely seemed utilitarian, but was rather driven by wanting to better understand oneself. One clinician felt that men were more likely to use a diagnosis to try to justify their behavior. Other clinicians indicated that this was a useful theme to explore when making differential diagnoses from personality disorders, where the want of a diagnosis tended to be driven by unstable self-identity.

Gender expression. Participants agreed that gender expression appeared more fluid and androgynous in autistic women. Alternatively, femme-presenting autistic women sometimes exhibited a “deliberately rehearsed femininity” (P02).

Alternative or cumulative diagnoses

This category explored strategies used to disentangle different conditions when clinicians were faced with complex or unclear cases.

Crisis versus baseline functioning. For patients currently experiencing a mental health crisis, it was sometimes difficult to determine the person’s “baseline functioning” (P07). When working with women currently in an acute psychiatric episode, some clinicians felt it was in their

patient's best interest to delay a potential diagnosis of autism, "In depression you have a flat affect, you're not very communicative, and sometimes the priority is to treat the depression, and then when that's lifted, to see what's underneath" (P05).

"Feeling in the room." Coupled with reliable and rigorous assessment, clinicians agreed that they relied to an extent on intuition when making differential diagnoses when assessing for autism. Participants spoke of using their awareness of how the interaction felt, notably when making differential diagnoses of personality disorders. Clinicians indicated that during interviews with people who they suspected were living with a personality disorder, they often felt that the patient was attempting to "crawl under their skin" (P20), which was not the case when interviewing autistic patients. One autistic participant spoke of "the clinical feeling that I'm dealing with someone like me" (P13).

Confounding role of trauma. Clinicians agreed that most adult women presenting for autism assessment had experienced trauma. Childhood trauma was specifically cited as difficult to disentangle from autism, as it could give rise to attachment difficulties that closely resembled certain signs of autism. For example, it was not always clear whether difficulties in relationships could be traced back to a neurodevelopmental difference or were the result of a fear of abandonment.

Chronology. When attempting to differentiate autism from other frequently seen conditions such as post-traumatic stress disorder (PTSD) and personality disorders, clinicians agreed it was imperative to establish when behavioral differences had first been noticed. Trauma-led difficulties could sometimes be pinpointed to the time the trauma had occurred, and personality disorders tended to begin to manifest in the teenage years. Autistic differences, however, were usually noticeable by late childhood.

Differentiating autism from BPD. BPD was highly present in autism assessment clinics as a past and/or differential diagnosis, and clinicians agreed the two conditions bore superficial resemblances. Many clinicians felt that BPD seemed to be a diagnosis reserved for women who self-harmed and had experienced trauma (as is the case for many autistic people). Several clinicians mentioned cases in which autistic women had been misdiagnosed and received therapy for BPD, where they had absorbed therapy vocabulary and now effectively acted in line with certain borderline characteristics. A few clinicians had also noticed that autistic women wrongly labeled as borderline had in fact simply associated with people who also had trouble fitting in to their peer groups, and engaged in risky behaviors. Upon further probing, their own experimentations with substance use were the result of mimicking these peers.

When differentiating the two conditions, clinicians agreed that understanding of neurotypical social dynamics was usually unimpaired in BPD, especially when they were in a phase of emotional stability. The reasons for difficulties with social relationships were also seen as being of a qualitatively different nature. Autistic women usually spoke of "difficulties with needing space" (P10), and ending friendships once social demands became too high, while women with BPD experienced relational difficulties due to a fear of abandonment. Participants agreed that attachment difficulties were important to investigate when deciding between autism and BPD.

The way emotions were described in autism versus BPD facilitated differential diagnosis. While women with BPD tended to explain how they were feeling with relative ease and a varied vocabulary, autistic women often found this verbalization difficult, "The other thing we ask people is 'can you name me five emotions apart from happy, sad and angry?' The average we get is two, women tend to do very badly on that" (P12). The emotions responsible for self-harm were also different. In BPD, self-harm usually followed interpersonal conflict, while in autism, it was often attributed to sensory issues or changes to routines.

Discussion

This study is, to our knowledge, the first to: (1) explore challenges with assessing adult women for autism from the perspective of those performing the assessments and (2) identify the specific tools, methods and behaviors used by expert clinicians when assessing complex cases in adult women. In the following sections, we provide our interpretation of these findings along with potential clinical implications.

Diagnostic strategy

The clinicians surveyed were cautious of overreliance on standardized instruments. Participants largely made use of individually developed techniques, or had individual ways of interpreting standardized scores, in order to reveal signs they saw as most indicative of autism. The limitations of current diagnostic criteria were also underlined—of the statements endorsed by our sample as indicative of autism in women, many were either not present in DSM-5 (gender identity expression, discrepancy between intelligence and professional success, and emotional empathy) or were qualitative evaluations of a DSM-5 trait (nature of social difficulties and interests). This is in line with research suggesting that expert clinicians integrate qualitative, non-verbal information into their assessments (de Marchena & Miller, 2017). Interestingly, these individual approaches ultimately still resulted in a certain convergence of opinion based on common experience, as shown by the agreed upon guidelines. These results make the case for a rebuilding of autism diagnostic criteria using a "bottom-up"

approach based on exposure to many cases (Mottron, 2021).

Expectations placed on assessment

Self-diagnosis is particularly prevalent as an entry point for assessment in adult autism clinics, with some research suggesting that barriers to formal diagnosis of autism, such as fear of not being believed, may bolster self-diagnosis particularly in women and people of color (Lewis, 2017; Sarrett, 2016).

Participants cited numerous examples where they had confirmed autism in women who strongly suspected they were on the spectrum. However, our participants also agreed that self-diagnoses could become problematic when the expectation of diagnosis was not met. Indeed, the general downplaying of women's concerns in medical contexts is well-documented (Chen et al., 2008; Hamberg, 2008) and came up often for our participants, who sometimes expressed concern that denying a diagnosis of autism to someone who had self-identified to the spectrum could cause far-reaching damage to their mental health and trust in the medical system.

Our findings support that autism benefits from relatively positive perceptions in the eyes of patients when compared to psychiatric conditions. An autism diagnosis can legitimize self-identity and sense of belonging to a community; however, this may result in false self-identification to the autism spectrum. Most clinicians mentioned reactions of disappointment when diagnosis was not obtained, which could cause depressive episodes or reactivate mental health symptoms. Many participants in this study invoked "autism traits" and "sub-threshold autism" to mitigate disappointment by validating the person's self-identification. Whether clinicians are able to provide alternative diagnoses greatly depends on their comfort assessing for other conditions, and the mandate imposed by their institution. One clinician hypothesized that self-identification to the spectrum in non-autistic people was not so much indicative of a "conviction of a diagnostic label" as it was "conviction about some difficulties that are not addressed at that point in time" (P18). Seen as such, the ability to propose alternative avenues is of paramount importance when assessing complex adult cases in order to avoid iatrogenic damage. This may be particularly relevant for women given a long legacy of gender biases in healthcare.

Disentangling diagnosis from the person's knowledge of autism

Clinicians identified the ways in which sociocultural perceptions of autism had impacted general diagnosis-seeking behaviors and how this could influence patient expectations. Part of the diagnostic assessment therefore involved

disentangling the person's expectations based on their research, from what clinicians recognized as autism following exposure to hundreds of autism cases. Clinicians and researchers may also be influenced by media coverage of autism. This is demonstrated by the exponential interest in research on camouflaging despite its intrinsic circularity (Fombonne, 2020). Most of our participants mentioned camouflaging as a factor they took into account during their assessment. Interestingly, there was no consensus on whether camouflaging had to be observable by the clinician (e.g. social differences observable once the person is tired) or whether the person's description of their camouflaging sufficed.

BPD, with or without trauma, as a specific issue

The potential phenotypic and life history overlap between autism, trauma, and BPD presented challenges even for experienced clinicians. Self-harm and problems with interpersonal relationships are frequent in both BPD and autism, making the two conditions complex to disentangle (De Bucy, 2018; Ingenhoven, 2020; Trubanova et al., 2014). A large-scale study on the temporal stability of co-occurring psychiatric diagnoses in adult women indeed found that personality disorders were the most commonly removed diagnoses once a diagnosis of autism was obtained (Kentrou et al., 2021). Although research has found heightened self-reported autistic traits in people in BPD (Dudas et al., 2017), another study actually found no incidence of BPD when assessing for personality disorders across a sample of 54 autistic participants with no intellectual disability (Lugnegård et al., 2012). Interesting indicators for differential diagnosis between BPD and autism spectrum were provided, based on the integrity of cognitive empathy in moments of low emotion, the presence or absence of alexithymia, the justifications for interrupting relationships, as well as the different contexts of self-harm and presence of attachment difficulties.

Regarding trauma, cognitive rigidity, and repetitive and avoidant behaviors are found across both autism and PTSD (Haruvi-Lamdan et al., 2017; Stavropoulos et al., 2018). Autistic people are highly vulnerable to trauma, including types of chronic trauma experiences often responsible for long-lasting cognitive and emotional effects, such as sustained bullying (Rumball et al., 2020). While BPD is often considered as an adult manifestation of childhood trauma and conflated with PTSD these conditions are in fact distinct. (Ford & Courtois, 2014). Attachment difficulties can emerge with or without trauma, and clinicians deciding whether to attribute cognitive and behavioral differences to BPD or autism could use tools such as the Coventry Grid (Cox et al., 2019) to determine whether attachment difficulties lie at the heart of these issues.

Limitations

This study involved individual perspectives from multiple cultural perspectives and contexts, different health-care systems, and a range of clinical specialties and environments. Although international, our sample exclusively practiced in Western countries and may not generalize to countries where adult psychiatric care is limited. A semi-structured interview schedule according to a priori defined issues may have led to confirmation bias. However, the triangulation of qualitative and quantitative methodologies as well as multiple coders is a strength of this study and may have helped to limit this source of bias. Acquiescence bias is also a limitation to be considered and may have inflated participant agreement with Delphi statements. This is to be expected from any method seeking to build consensus, and we attempted to highlight areas where disagreements arose. Finally, our participants represented a large range of experience (between 5 and 40 years diagnosing autism) demonstrating that the notion of “expert” remains relatively abstract and ill-defined. We hope further research will clarify this topic.

Clinical implications

This study joins a limited evidence base seeking to refine autism assessment and autistic phenotypes from the bottom up, by suggesting recommendations based on the observations of experts having assessed thousands of autistic women. We confirm that experts diagnose autism based on individual appraisals of very broad constructs, and highlight the frequent presence in specialized autism clinics of conditions superficially similar to autism, such as BPD or PTSD. We urge for further training and exposure to these conditions for clinicians specialized in autism. In cases where diagnostic boundaries appear unclear, as is often the case within extreme values of clinical specifiers, clinicians should build their expertise based on a wide exposure to a large number of male and female autistic people. This allows for the building of a gender-neutral prototype pattern recognition, hierarchizing the weight of different autism signs. It allows the detection of a “familial resemblance” between less evident autistic phenotypes and prototypical adult women diagnosed with high certainty by more than one expert.

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Data accessibility

The verbatim of participants' interviews is accessible through request to the corresponding author.

Supplemental material

Supplemental material for this article is available online.

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