Neuropsychiatric Disease and Treatment

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REVIEW

Improving Diagnostic Procedures in Autism for Girls and Women: A Narrative Review

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Abstract: Biases exist in the diagnostic process for autism spectrum disorder (henceforth "autism"), which result in some girls and women being diagnosed later or missed entirely. Current diagnostic tools may not capture the full range of behavioural presentations of autism, leading to under-identification. This review explores why these biases may occur, and how diagnostic procedures could be adapted to better identify autistic girls and women. We recommend that diagnostic assessments are adjusted to capture a broader range of behavioural exemplars of autism; that camouflaging of autistic traits is taken into account; and that care is taken to ensure co-occurring mental health conditions do not overshadow autism diagnosis. We offer recommendations, building on gold-standard diagnostic guidelines, for how diagnostic procedures can be improved for girls and women.

Keywords: autism, girls, women, diagnosis

Autism Spectrum Disorder (hereafter "autism") is a neurodevelopmental condition, that first emerges in infancy and early childhood and persists across the lifespan. It is largely determined by genetic factors, with a heritability estimated between 64% and 91%.[1] Autism is highly heterogeneous both in terms of its presentation and aetiology but, nevertheless, the condition is characterized by a set of core, diagnostic characteristics, namely, atypical social relating and social communication, as well as a tendency towards inflexibility and unusual processing of sensory experience. Most instances of autism should be considered dimensional, rather than categorical: they represent one end of an autistic trait continuum that extends throughout the general population, with no natural "cut point" between autism and non-autism.[2] Autism commonly co-occurs with other neurodevelopmental challenges, in particular difficulties with speech and language and intellectual development. Nevertheless, most autistic people have fluent language and IQs in the normal range.[3]

Once considered a rare condition, autism is now widely diagnosed. Its epidemiological prevalence is estimated as being 1–2%, but evidence is emerging in some industrialized, western countries that actual rates of diagnosis are much higher than this within certain groups, defined by age and sex. For example, in the UK amongst boys aged 5 to 9, reported rates of diagnosis are around 4%.[4] The dramatic growth in the epidemiological and administrative prevalence of autism of recent years started in the 1990s and appears to be ongoing. This phenomenon is not fully understood, but the weight of evidence suggests it is driven by improved recognition, especially amongst those with normal-range IQ, and a broadening of the diagnostic criteria over time.

Autism is not a mental health condition, but autistic people are at high risk of developing mental health problems. One seminal study found that in a community sample of autistic young people, 70% had at least one mental health diagnosis, and 40% had two or more.[5] These very high rates of mental health difficulties persist into adulthood, although the precise nature of risk changes with development. In childhood, anxiety difficulties and emotional and behavioural regulation challenges (eg, Attention deficit/hyperactivity disorder, oppositional defiant disorder) are most common. By adulthood, depression and anxiety problems predominate; and suicide rates are substantially higher for autistic, compared to non-autistic, adults.[6,7]

Received: 23 May 2023
Accepted: 14 February 2024
Published: 7 March 2024

Under current systems of care, outcomes for autistic people are amongst the worst for any psychiatric diagnosis.[8] In addition to the high rates of mental health problems described above, autistic people are at elevated risk of educational under-attainment, unemployment, underemployment, social isolation, poverty, homelessness and low quality of life. Further, compared to non-autistics, autistic people suffer higher rates of morbidity in relation to a range of physical illnesses and higher mortality.[9]

A New Paradigm

Traditionally, understanding of autism has been structured within a "pathology paradigm" (sometimes called the "medical model"), that views autism as a disorder; and which aims to understand autistic behaviours and the challenges faced by autistic people as direct consequences of their individual "deficits" and "impairments". For example, when trying to understand why autistic people experience such high rates of anxiety, a researcher within the pathology paradigm would focus on identifying individual deficits (eg, inability to tolerate uncertainty, brain-based emotion regulation impairments) and investigate whether and how these cause and maintain anxiety problems.

Recently, a new paradigm has become increasingly influential, driven both by scientific advances and the ethical imperative to give autistic people themselves more power over attempts to characterize, understand and help them.[10,11] This is often called the "neurodiversity" paradigm. It views autism not as an inherently disordered developmental state, but rather as a form of difference. Within this way of thinking, in line with the social model of disability, the difficulties faced by autistic people arise due to a misfit between, on the one hand, the autistic individual and, on the other hand, the environments they live in.[12] Work within the neurodiversity paradigm tends to highlight that most autistic people live in unaccommodating, and often outright hostile, environments designed by and for non-autistic people. Logically, this approach tends to place emphasis on changing the environment, not just the person, when thinking about how to improve the lives and functioning of autistic people.[10]

FOOTNOTE: The authors of the current paper seek to work within the neurodiversity paradigm. This is why, in line with the wishes of many autistics, we use identify first language ("autistic person") rather than person-first language ("person with autism"). This is because (i) autism tends to be experienced as central to a person's identity and (ii) person-first language is usually used for negatively valenced conditions, and we seek to convey the idea of autism as a form of difference, rather than an inherently disordered state. For the same reason, we refer to autism, rather than autism spectrum disorder.

The Diagnostic Bias Against Autistic Girls and Women

A striking feature of autism is that more males than females are diagnosed as autistic. Until recently this was widely understood as simply reflecting the nature of the condition, with a consensus that the true male-to-female ratio is approximately four-to-one.[13] However, the accuracy of this male-to-female ratio has been questioned more recently, with some research suggesting that girls and women may be under-represented in clinical studies estimating prevalence, and therefore that the true male-to-female ratio may be close to three-to-one or even two-to-one.[14] If this is the case, it follows that there may be biases in the diagnostic process which particularly impact autistic girls and women. We note here that these biases likely apply to autistic people of all genders, and that these and other biases are also likely to impact other groups of autistic people who are less likely to receive an accurate and timely diagnosis, including ethnic and racial minority groups.[15,16] For the purposes of this review, however, we focus on specific biases which may particularly impact girls and women.

We note here that there has also been some concern reported by clinicians[17,18] and researchers[19] that autism may be over-diagnosed. Rates of autism diagnosis have increased significantly in recent years,[20] and this is partly driven by the relatively high rates of women and girls receiving a diagnosis later in life. We are not aware of any research demonstrating overdiagnosis of girls and women specifically, however, our aim in this review is to promote more accurate diagnosis of autism, including reducing the likelihood of misidentification of autism in women and girls. For an overview of factors which may contribute to over-diagnosis of autism, we point readers to a recent editorial by Fombonne.[19]

One proposed reason for the diagnostic bias experienced by autistic girls and women is differences in autism presentation across genders.[21] Although autism is highly genetically heritable, diagnosis is based on behavioural expression of the core characteristics described above; there are no reliable biomarkers or other biological tests for autism.[13] Variation in expression of these core characteristics across genders may lead to variation in meeting diagnostic criteria.[22] It can be helpful to think of multiple levels of characteristics when considering what behavioural expression of autism actually means. Lai et al[22] refer to three levels of measurement of autism: i) "broad constructs" (ie, the core diagnostic criteria, such as restricted and repetitive

behaviours), ii) "narrow constructs" (ie, subdomains of characteristics, such as repetitive motor actions), and iii) "behavioural exemplars" (ie, specific behavioural examples of characteristics, such as spinning the wheels on a toy car). We propose that gender differences in the expression of autism exist mainly at the level of behavioural exemplars, such that the same characteristics of autism are expressed in males and females, but may be expressed through different behaviours, not all of which are assessed for in current diagnostic tools.

As an example of this, let us take the broad construct of persistent difficulties with "social interaction", of which one narrow construct is difficulties "developing, maintaining, and understanding friendships".[13] Many autism assessment procedures evaluate this using the behavioural exemplar of "difficulty making friends", and so look for evidence for lack of close friendships to support this characteristic. However, research suggests that, while autistic boys may have a tendency towards reduced intimacy in their friendships compared to non-autistic boys, autistic girls report similar levels of close friendships as non-autistic girls.[23] In contrast, autistic girls may experience more conflict within their close friendships compared to autistic boys. These findings suggest that different behavioural exemplars of the same autism construct (in this case, difficulties developing and maintaining friendships) may be relevant for autistic boys and girls. The number of close friendships may be a useful metric to assess friendships in autistic boys, but experience of conflict within friendships may be more useful to assess friendships in autistic girls. We note here that we do not suggest different criteria, or different behavioural exemplars, for different genders. Instead, we recommend a broadening of the behavioural exemplars included in diagnostic assessments to better capture a range of presentations. Many current diagnostic tools do not include specific questions asking about either the number or conflict experience of close friendships,[24] and so it is hard to know to what extent this is being taken into account during the assessment process.

Camouflaging

In addition to differences in the behavioural exemplars of autistic characteristics, there may be additional characteristics associated with autism which occur more often in girls and women, and which impact the diagnostic assessment process. One characteristic which has received substantial interest over recent years is the phenomenon of camouflaging, which encompasses hiding or compensation for autistic characteristics.[25] Examples of camouflaging (also known as masking, compensation, adaptive morphing and various other names)[26] include learning or practicing neurotypical social communication behaviours, stopping oneself from performing obviously autistic behaviours such as hand flapping, and mimicking the behaviours of neurotypical people in order not to stand out.[27] Camouflaging of autistic characteristics has been reported extensively amongst autistic adults without intellectual disability,[26] and has also been observed in autistic children, although it is still unclear exactly when and how camouflaging develops.[28] Some have proposed that camouflaging may represent a distinctively autistic use of more general impression management strategies.[29] It is consistently described by autistic people as being driven by experience of intolerance and stigma for appearing different.[30]

Gender differences in camouflaging have been observed, with autistic women reporting higher levels of camouflaging than autistic men.[31] Autistic women may also demonstrate more behavioural camouflaging, with a greater discrepancy between their self-reported autistic characteristics, and their level of autistic behaviour as observed by clinicians.[32] As camouflaging results in fewer observable autistic behaviours, particularly in relatively short, structured interactions, this may account for the underdiagnosis of some autistic women. Research suggests that autistic females and those undiagnosed but with high autistic traits camouflage more than those with low autistic traits,[28] although there is mixed evidence as to whether camouflaging is associated with later age at diagnosis for females[33,34] While they may experience many autistic characteristics internally, such as difficulty with social communication or specific sensory needs, these may be hidden from the casual observer and so the autistic characteristics of some girls and women may go unrecognised. This is supported by qualitative research with autistic women and girls diagnosed later in life, many of whom report camouflaging at a very high level even before they knew they were autistic, and having to actively stop camouflaging in order to allow for their autistic characteristics to be observed in clinical settings.[35,36] Camouflaging may account for the later age of diagnosis for many girls and women.[37] There may also be gender differences in the developmental trajectories of autistic characteristics, such that some girls demonstrate their autistic characteristics later on than boys.[38] It has been suggested that some autistic girls may either camouflage at a high level, or may have genuinely lower levels of autistic characteristics in early childhood, but that by adolescence the social demands may have increased to the point that their autistic characteristics become more obvious.[2]

It should be noted here that autistic individuals of all genders report camouflaging, and the impact of camouflaging on diagnosis may not be unique to girls and women. However, it may be the interaction between camouflaging, and differential presentation of some autistic characteristics as described above, which leads some autistic women to fall below the diagnostic threshold as some of their autistic characteristics may be hidden at first glance.[21,39]

Co-Occurring Conditions and Differential Diagnosis

Both camouflaging and atypical behavioural presentations may impact the way that women's and girls' autistic traits are evaluated during the diagnostic process. However, there may be additional characteristics which affect the accuracy of diagnosis, without affecting their autistic presentation. These include the co-occurrence of other conditions, particularly mental health conditions, which may overshadow autism diagnoses, or which may be overshadowed by autism and so may themselves be under-identified. Autistic people of all genders experience significantly higher rates of co-occurring mental health conditions than the general population,[7] and are at greater risk of death from suicide.[9] As in the general population there are some mental health conditions which occur more commonly in autistic women and girls than in autistic men and boys. These include anxiety,[40] which is also high in autistic non-binary adults. Anxiety is part of a group of mental health conditions labeled as "internalising" conditions; they represent distress turned inwards, rather than outwards. In the case of anxiety, this may arise partly as a result of being in poorly adapted environments and having autistic characteristicsfor instance, intolerance of uncertainty or anticipation of sensory overstimulation.[41] If such autism-specific risk mechanisms are at play, traditional treatments for anxiety, developed for non-autistic people, may be less effective. Undiagnosed autistic women and girls, who may also be experiencing high levels of anxiety, may find that some of their autistic characteristics are interpreted as anxiety and so they may be less likely to receive a timely and appropriate autism diagnosis. We know of no research directly testing whether anxiety overshadows autism (or vice versa),[42] and therefore join researchers arguing for the need to carefully differentiate characteristics of each, and to consider whether anxiety co-occurring with autism presents similarly to anxiety in non-autistic people.[43]

Another internalising condition which may overshadow autism in women and girls is the presence of an eating disorder. Estimates suggest that 20–30% of women receiving treatment for anorexia nervosa are autistic: in most cases autism is not diagnosed until several years after the eating disorder.[44,45] Often eating disorder patients are not considered eligible for an autism assessment until they are in recovery from their eating disorder. This can have grave consequences for undiagnosed autistic women and girls, whose eating disorders may in fact be related to their autism. For instance, sensory sensitivities and management of emotions may drive restricted eating to a greater extent than concerns around shape and weight for autistic women.[46] Accurate identification of autism is essential to enable appropriate interventions to be developed to support women with eating disorders,[47] and clinicians should be mindful that women and girls assessed for eating disorders should also be screened for autism.

Finally, some expert clinicians have also raised concerns that additional psychiatric conditions, such as borderline personality disorder, may overshadow the diagnosis of autism in some women.[18] While it is worth pointing out the experience of trauma is common in autistic women,[48] and there may be some degree of co-occurrence of autism and borderline personality disorder,[49] it has been suggested that evaluation of understanding of social relationships, and assessment of attachments in particular, can help to differentiate the two conditions and so ensure patients receive appropriate support.[18] This area of research is at an early stage, however, and more information is needed about the impact that earlier adverse life events may have on autistic people. Differential diagnosis between autism and trauma-related conditions can be difficult, due to the overlap between certain characteristics. For instance, case studies suggest that post-traumatic stress disorder in children may be marked by repetitive play behaviours, which can also be considered part of autism diagnostic criteria.[50] However, clinicians should be aware of interpreting autistic-like characteristics within the context of any trauma experienced by the individual and, ideally should seek to determine whether these characteristics were present prior to experiencing trauma. For more detailed guidance on differentiation between autism and borderline personality disorder, we recommend the guidance produced by Cumin et al.[18]

Autism Diagnostic Procedures

Recommendations outlined in the National Institute for Health and Care Excellence (NICE) guidelines are considered best practice for health and care services in the United Kingdom. Two NICE guidelines separately outline recommendations for conducting autism diagnostic assessments with adults and with young people aged under the age of 19.[51] Each guideline provides detailed assessment recommendations tailored to the differing needs of children, adolescents, and adults as well as those with and

without additional language or learning difficulties. According to these guidelines a comprehensive autism assessment should be carried out by a multidisciplinary team of trained professionals and take a multi-factorial approach, gathering detailed diagnostic data about an individual's developmental and behavioural profile from a range of sources. The assessment process should aim to include information about current concerns and background information including an individual's social, educational and/or employment history; assessment of core autism characteristics including the trajectory, pervasiveness and impact of these characteristics via a developmental history and an observational assessment; and finally consideration and assessment of differential diagnoses and/or conditions that commonly co-occur with autism. Clinicians should use all information gathered alongside their clinical judgment and the International Classification of Diseases, 11th Edition (ICD-11) or Diagnostic and Statistical Manual, 5th Edition (DSM-5) criteria to make a diagnostic decision. The current evidence base on autism in girls and women does not indicate that these guidelines need to be radically overhauled but rather fine-tuned, such that additional recommendation could be added whilst certain existing recommendations could be further emphasized or elaborated upon. We offer the following recommendations of how diagnostic assessments could be adapted to more accurately identify autism in girls and women; summarised in Table 1. The recommendations are based on the research and clinical expertise of the authors, however we note the lack of research evaluating any adaptations to diagnosis through experimental paradigms. As such, these recommendations would benefit from thorough empirical evaluation. Further, we also emphasise that the recommendations we

Table 1 Summary of Key Recommendations for Improving Autism Diagnostic Procedures for Girls and Women

Clinicians should be aware of the following:
There is a diagnostic bias against autistic girls and women, whereby they are less likely to receive a timely diagnosis compared to autistic boys and men. As such they are more likely to miss out on the benefits that can derive from an autism diagnosis.
The diagnostic bias is driven by the fact that sex/gender influences how autism presents: current conceptualisations of autism tend to be derived from majority-male samples, such that diagnostic processes are less sensitive to autism in girls and women.
Another likely contributor to the diagnostic bias is camouflaging, which involves learning ways to hide one's autistic characteristics. Autistic people of all genders camouflage, but girls and women do it more than boys and men.
Diagnostic overshadowing also impacts on girls and women's chances of receiving a timely autism diagnosis. Many undiagnosed autistic girls and women develop co-occurring mental health problems, such as anxiety, depression and eating disorders. These can then obscure their autism, with their difficulties being ascribed to their mental health difficulties rather than to their underlying (undetected) autism.
To enhance accuracy of autism assessment for girls and women, clinicians should do the following:
Assessment should be multimodal (ie, encompassing self-report, direct observation and informant report) and include both current and historical information. The collection of developmental, multi-perspective information is especially helpful for overcoming challenges to diagnostic accuracy posed by camouflaging and the co-occurrence of mental health problems.
Autism assessment should always include comprehensive screening for a range of mental health and additional neurodevelopmental difficulties; and formulation of these in relation to any autistic traits.
Assessment and subsequent diagnostic formulation should be broad, rather than simply focusing on core, diagnostic features of autism. It should cover both characteristics of the individual (eg, cognitive capacity, values, hopes) and of their environment (eg, life events including any traumatic experiences, physical environment, social environment). Such assessment supports formulation of the patient's person-environment fit, which will in turn suggest ways of enhancing wellbeing and functioning.
Clinicians should be flexible when looking for behavioural exemplars of autism diagnostic features, considering the ways in which sex and/or gender influence how autism manifests and is experienced.
When standardised quantitative measures are used (eg, the Autism Diagnostic Interview-Revised; the Autism Diagnostic Observation Schedule, Second Edition), clinicians should not make diagnostic decisions based only on quantitative diagnostic algorithms. Rather, they should consider qualitative information collected via the measure, and how this relates to other information from the assessment and to autism diagnostic criteria.
Camouflaging should be assessed, via discussion with the person being assessed and/or use of standardised measures, such as the Camouflaging of Autistic Traits Questionnaire (CAT-Q).

make are not only useful for the assessment of girls and women: they represent our views on good practice and would be appropriate for clients of all genders, especially where presentation is complex and/or subtle.

As outlined in the NICE guidelines, people have the right to be involved in discussions and decisions affecting their care.[51] Thus, during an autism assessment clinicians should always seek to gain a holistic understanding of an individual, incorporating the views and insights of the individual themselves. In the case of women, a general clinical interview should be conducted to gather information about experiences of education, employment, family, social relationships, mental health difficulties and possible trauma as well as areas of strength and difficulty. In the case of girls and adolescents, a similar comprehensive general clinical interview should be carried out with a parent or caregiver. Clinicians should also directly enquire about girls' experiences of family life, education, hobbies, and social relationships as well as their subjective view on their strengths and difficulties.

Although historically downplayed in autism assessments, it is equally important to directly assess an individual's subjective experience of and insight into their (possible) neurodivergence.[52] Many autistic girls and women can provide detailed and insightful accounts of their neurodivergence and during an assessment these can provide invaluable information about core autism characteristics underlying more nuanced or subtle behavioural presentations.

Of course, when determining the best format through which to gain a particular individual's perspective it is important to consider their self-insight, developmental level, social communication abilities and preferences, and possible additional learning needs. For example, younger girls may be asked concrete questions about their social and sensory preferences whilst adolescents and women may be able to discuss past experiences of social confusion or distress in response to change. Some individuals will also require additional processing time, as well as asynchronous and /or non-talking modes of communication (ie, writing, typing, drawing) in order to comfortably and completely express themselves.[53]

This process requires a certain level of trust and rapport and this may be difficult to build with some girls and women who have had previous unhelpful encounters with health professionals in which they felt dismissed, misunderstood or blamed.[35] Clinicians should seek to build a positive working relationship with girls and women during assessments by taking an empathetic stance, validating difficulties experienced and, where necessary, building rapport over multiple sessions.

We do acknowledge the concern held by clinicians that some women may have self-diagnosed or researched autism at length,[18] complicating the assessment process. This is not an issue specific to autism assessments; given the widespread availability of health-related information especially online, it is increasingly common for people to self-diagnose or seek information prior to consulting health professionals.[54] However, autism awareness has increased within the public in recent years, and autism may be viewed as a less stigmatised, and therefore more desirable, condition with which to identify; at least in Western nations.[55] We are aware of some concerns that social media in particular may misrepresent autistic characteristics and therefore encourage inaccurate self-identification.[56] Such difficulties can be overcome by firstly gaining a holistic understanding of an individual including their understanding of autism and motivations for attending a diagnostic assessment and secondly, gathering detailed examples of neurodivergent experience, as discussed above. Moreover, by fostering a positive working relationship, and by identifying specific functional challenges experienced by the individual, clinicians increase the likelihood that an individual feels understood and validated and in turn accepting of the assessment outcome. Finally, researchers and clinicians have a duty to challenge inaccurate representations of autism and to ensure that those they work with are given accurate information about autistic characteristics.

The NICE guidelines recommend clinicians gather diagnostic data from a range of informants (eg, the person being assessed, their family members, and their teachers) using a range of methods (eg, review of previous reports, clinical interview, and observational assessment). A level of discrepancy across informants is to be expected. Indeed, parents, educators, and other professionals may differentially recognise and interpret the social behaviours of autistic girls and women depending on their familiarity with non-prototypical autism phenotypes and their expectancy for autism presenting in girls and women (independent of phenotype).[35,57,58] For this reason, it is important that clinicians systematically gather detailed descriptions of specific behavioural exemplars instead of relying on informants' perceptions of what does or does not constitute autism. Equally, clinicians themselves need to recognise and reduce their own susceptibility to familiarity and expectancy biases.[52,59,60] One way to achieve this is through appropriate training and continuing professional development including dialogue with autistic people of all genders about their experience of neurodivergence. Another way is to engage in reflective practice to better understand how one's experiences, training, and beliefs can lead one to selectively attend to or dismiss particular information during autism assessments. Clinicians should aim to familiarise themselves with a variety of autistic

presentations, including those with intellectual disability and/or minimal spoken language, to ensure that they have an accurate understanding of the core autistic characteristics and the variety of ways these can be expressed.

The NICE Guidelines recommend clinicians consider using autism-specific assessment tools including rating scales (eg, Autism Quotient [AQ]), semi-structured clinical interviews (eg, Autism Diagnostic Interview, Revised [ADI-R], Developmental, Dimensional and Diagnostic Interview [3Di]), and semi-structured observational assessments (ie, Autism Diagnostic Observation Schedule, General or 2nd Edition [ADOS-G/ADOS-2]) in comprehensive autism assessments. Such tools can help clinicians to systematically gather detailed descriptions of specific behavioural exemplars and narrow constructs indicative of broad core autism characteristics as well as information about the trajectory, pervasiveness, and impact of these; essential in the assessment of girls and women with nuanced behavioural presentation and/or co-occuring conditions. However, it is important to note that the use of autism specific-assessment tools alone is not sufficient.[52] In their current form, these tools do not adequately gather relevant background or contextual information nor the subjective views or insights of the individual being assessed and they may also be biased against more nuanced autistic presentations exhibited by some girls and women.[24]

With regard to this last point, most widely used tools, including those considered to be "Gold Standard" (ie, ADOS-2 and ADI-R), were created at a time when a male-centric understanding of autism predominated, and were evaluated with predominantly male samples.[60,61] Consequently, it is possible that these tools do not adequately account for heterogeneity in the behavioural presentation and trajectory of core autism characteristics across autism phenotypes and thus present a barrier to diagnosis for some girls and women.[22,62,63] Large-scale studies suggest small-to-no sex differences in core characteristics when using gold-standard assessment tools, and when controlling for relevant covariates.[64,65] However, a key limitation is that most empirical research in this area has been conducted with individuals who present most similarly to a prototypical male autistic phenotype, that is, those already clinically diagnosed with autism.[66] Promising research suggests gender-specific screening tools may be helpful in identifying autism characteristics more commonly exhibited by girls and women, especially those without additional language or intellectual difficulties.[67–69]

With regard to the use of "gold standard" measures, emerging research has also investigated the usefulness of a female-specific coding framework on the ADOS-2 (the Gendered Autism Behavioural Scale, or GABS).[24] This coding frame proposes additional characteristics which could be captured through the standardised ADOS-2, and which might include a broader range of autistic characteristics expressed by females. However, the GABS is still in development and has undergone only limited psychometric validation, therefore we do not recommend its use in clinical settings at the present time. Thus, when considering all the possible diagnostic data gained from using "gold standard" measures in their current form, it is important not to focus solely on numerical scores. Rather, in most cases it is more helpful to manually map qualitative information about behavioural exemplars indicative of autism characteristics, as well as information about the trajectory, pervasiveness, and impact of these onto the DSM-5 or ICD-11 criteria.[70]

Additionally, it is important to consider and assess characteristics common to autistic girls and women not currently assessed using "gold standard" measures. One such feature is camouflaging. As previously discussed, through camouflaging some autistic girls and women may present with a seemingly neurotypical social presentation in certain environments. It is imperative that at no point during the referral or diagnostic process, autism is ruled out based on the isolated presence of specific social behaviours (eg, eye contact).[71] However, a diagnostic conclusion that an individual camouflages their autistic traits must also be supported by diagnostic data. During an ADOS-2 assessment the social interaction and environment may be optimised in such a way to inadvertently facilitate camouflaging (ie, the assessment lasts approximately an hour or less and involves a structured one-to-one interaction with a trained clinician in a quiet room).[72] Thus, it is important to assess girls and women in additional and/or more complex or naturalistic social environments where their differences or difficulties may be more apparent. For example, clinicians could observe girls in the school playground, view their home videos,[73] or interact with girls and women in the clinic for extended periods of time or over multiple appointments.[18] Equally, if not more importantly, clinicians should directly enquire about girls and women's experiences of camouflaging by asking open ended questions about strategies and behaviours they may have developed to mask or compensate for their social differences or difficulties. The Camouflaging Autistic Traits Questionnaire (CAT-Q),[27] a self-report measure, is also useful in this regard, as it can help individuals identify and reflect on their camouflaging behaviours. However, we note that the CAT-Q does not measure autistic traits, and therefore high scores on the CAT-Q are not necessarily indicative of an autism diagnosis; there are many autistic people who do not camouflage.[25]

Another such feature is mental health conditions. As previously discussed, co-occurring mental health conditions, common amongst autistic girls and women can overshadow signs of autism and complicate diagnostic assessments.[52] For this reason, mental health screening measures should routinely be included in autism diagnostic assessments for girls and women, and processes should be in place for referral to appropriate services and/or treatment as relevant. Positive results need to be followed up with more detailed enquiry and assessment, with particular attention paid to overlapping constructs common to autism and certain mental health conditions. For example, if avoidance of social situations has been endorsed on a mental health screener measure, detailed questioning is required to determine the extent to which this behaviour should be attributed to autism, social anxiety disorder, and a combination of autism and social anxiety disorder.

Summary

There is a diagnostic bias against autistic girls and women, meaning that they are more likely to fly under the diagnostic radar. In practice, this means that, compared to boys and men, they are more likely to be diagnosed autistic late, or not at all. Partly, this reflects the fact that sex and/or gender influences how autistic characteristics manifest. Many autistic girls and women do not fit the common diagnostic consensus about autism which, after all, is largely based on studies in with men and boys predominate. We argue that existing broad definitions of autism, for example, those in DSM-5 and ICD-11, can be used to assess autism in girls and women; and also that existing diagnostic processes and instruments are useful for all genders. As such, an assessment that draws on information from multiple informants (eg, the person being assessed, parents, teachers) and modalities (eg, direct observation, interview, questionnaire); and which centers around multidisciplinary case discussion is the gold standard for girls and women, as well as boys and men. Nevertheless, clinicians must be trained in how autism manifests in different genders. This can help them take a flexible and evidence-based approach to assessing for which specific behavioural manifestations can be characteristic of autism in girls and women. By following the recommendations listed here, clinicians can be more confident in producing an accurate diagnosis which will have clinical utility to the individual, particularly for women and girls.

Disclosure

Julia Cook and Laura Hull are co-first authors for this study. Dr Julia Cook reports funding from UK National Health Service (NHS) for part of her salary for clinical sessions in the NHS, during the conduct of the study. Dr Laura Hull reports grants and fellowship award from Rosetrees Trust & Elizabeth Blackwell Institute, University of Bristol, during the conduct of the study; consulting fees from One Green Bean PR, outside the submitted work. Prof WIll Mandy reports during the writing of this article receiving funding from the NIHR, ESRC, ERC, Dunhill Medical Trust and Autistica. The authors report no other conflicts of interest in this work.

References

1. Tick B, Bolton P, Happé F, Rutter M, Rijsdijk F. Heritability of autism spectrum disorders: a meta-analysis of twin studies. *J Child Psychol Psychiatr*. 2016;57(5):585–595. doi:10.1111/jcpp.12499
2. Pender R, Fearon P, Pourcain BS, Heron J, Mandy W. Developmental trajectories of autistic social traits in the general population. *Psychol Med*. 2021;1–9. doi:10.1017/S0033291721002166
3. Zeidan J, Fombonne E, Scorah J, et al. Global prevalence of autism: a systematic review update. *Autism Res*. 2022;15(5):778–790. doi:10.1002/aur.2696
4. O'Nions E, Petersen I, Buckman JEJ, et al. Autism in England: assessing underdiagnosis in a population-based cohort study of prospectively collected primary care data. *Lancet Reg Health Eur*. 2023;29:100626. doi:10.1016/j.lanepe.2023.100626
5. Simonoff E, Pickles A, Charman T, Chandler S, Loucas T, Baird G. Psychiatric disorders in children with autism spectrum disorders: prevalence, comorbidity, and associated factors in a population-derived sample. *J Am Acad Child Adolesc Psychiatr*. 2008;47(8):921–929. doi:10.1097/CHI.0b013e318179964f
6. Cassidy S, Rodgers J. Understanding and prevention of suicide in autism. *Lancet Psychiatry*. 2017;4(6):e11. doi:10.1016/S2215-0366(17)30162-1
7. Lai MC, Kassee C, Besney R, et al. Prevalence of co-occurring mental health diagnoses in the autism population: a systematic review and meta-analysis. *Lancet Psychiatry*. 2019;6(10):819–829. doi:10.1016/S2215-0366(19)30289-5
8. Howlin P, Moss P. Adults with autism spectrum disorders. *Can J Psychiatry*. 2012;57(5). doi:10.1177/070674371205700502
9. O'Nions E, Lewer D, Petersen I, et al. Estimating life expectancy and years of life lost for autistic people in the UK: a matched cohort study. *Lancet Region Health Euro*. 2024;36. doi:10.1016/j.lanepe.2023.100776
10. Chapman R, Botha M. Neurodivergence-informed therapy. *Dev Med Child Neurol*. 2023;65(3):310–317. doi:10.1111/dmcn.15384
11. Pellicano E, den Houting J. Annual research review: shifting from "normal science" to neurodiversity in autism science. *J Child Psychol Psychiatr*. 2022;63(4):381–396. doi:10.1111/jcpp.13534
12. Mandy W. The old and the new way of understanding autistic lives: reflections on the life of Donald Triplett, the first person diagnosed as autistic. *Autism*. 2023;27(7):1853–1855. doi:10.1177/13623613231194476
13. American Psychiatric Association. Diagnostic and statistical manual of mental disorders (DSM-5®); 2013.

14. Loomes R, Hull L, Mandy WPL. What is the male-to-female ratio in autism spectrum disorder? A systematic review and meta-analysis. *J Am Acad Child Adolesc Psychiatr*. 2017;56(6):466–474. doi:10.1016/j.jaac.2017.03.013
15. Jones DR, Mandell DS. To address racial disparities in autism research, we must think globally, act locally. *Autism*. 2020;24(7):1587–1589. doi:10.1177/1362361320948313
16. Kalb LG, Singh V, Hong JS, et al. Analysis of race and sex bias in the Autism Diagnostic Observation Schedule (ADOS-2). *JAMA Network Open*. 2022;5(4):e229498. doi:10.1001/jamanetworkopen.2022.9498
17. Davidovitch M, Shmueli D, Rotem RS, Bloch AM. Diagnosis despite clinical ambiguity: physicians' perspectives on the rise in autism spectrum disorder incidence. *BMC Psychiatry*. 2021;21(1):150. doi:10.1186/s12888-021-03151-z
18. Cumin J, Pelaez S, Mottron L. Positive and differential diagnosis of autism in verbal women of typical intelligence: a Delphi study. *Autism*. 2022;26(5):1153–1164. doi:10.1177/13623613211042719
19. Fombonne E. Editorial: is autism overdiagnosed? *J Child Psychol Psychiatr*. 2023;64(5):711–714. doi:10.1111/jcpp.13806
20. Underwood JF, DelPozo-Banos M, Frizzati A, John A, Hall J. Evidence of increasing recorded diagnosis of autism spectrum disorders in Wales, UK: an e-cohort study. *Autism*. 2022;26(6):1499–1508. doi:10.1177/13623613211059674
21. Hull L, Petrides KV, Mandy W. The female autism phenotype and camouflaging: a narrative review. *Rev J Autism Dev Disord*. 2020;7(4):306–317. doi:10.1007/s40489-020-00197-9
22. Lai MC, Lombardo MV, Auyeung B, Chakrabarti B, Baron-Cohen S. Sex/gender differences and autism: setting the scene for future research. *J Am Acad Child Adolesc Psychiatr*. 2015;54(1):11–24. doi:10.1016/j.jaac.2014.10.003
23. Sedgewick F, Hill V, Yates R, Pickering L, Pellicano E. Gender differences in the social motivation and friendship experiences of autistic and non-autistic adolescents. *J Autism Dev Disord*. 2016;46(4):1297–1306. doi:10.1007/s10803-015-2669-1
24. Clarke E, Hull L, Loomes R, McCormick CEB, Sheinkopf SJ, Mandy W. Assessing gender differences in autism spectrum disorder using the Gendered Autism Behavioral Scale (GABS): an exploratory study. *Res Autism Spectr Disord*. 2021;88:101844. doi:10.1016/j.rasd.2021.101844
25. Hull L, Petrides KV, Allison C, et al. "Putting on My Best Normal": social camouflaging in adults with autism spectrum conditions. *J Autism Dev Disord*. 2017;47(8):2519–2534. doi:10.1007/s10803-017-3166-5
26. Cook J, Hull L, Crane L, Mandy W. Camouflaging in autism: a systematic review. *Clin Psychol Rev*. 2021;89:102080. doi:10.1016/j.cpr.2021.102080
27. Hull L, Mandy W, Lai MC, et al. Development and Validation of the Camouflaging Autistic Traits Questionnaire (CAT-Q). *J Autism Dev Disord*. 2019;49(3):819–833. doi:10.1007/s10803-018-3792-6
28. Milner V, Mandy W, Happé F, Colvert E. Sex differences in predictors and outcomes of camouflaging: comparing diagnosed autistic, high autistic trait and low autistic trait young adults. *Autism*. 2022;13623613221098240. doi:10.1177/13623613221098240
29. Ai W, Cunningham WA, Lai MC. Reconsidering autistic 'camouflaging' as transactional impression management. *Trends Cognit Sci*. 2022;S1364661322001061. doi:10.1016/j.tics.2022.05.002
30. Pearson A, Rose K. A conceptual analysis of autistic masking: understanding the narrative of stigma and the illusion of choice. *Autism Adulthood*. 2021;3(1). doi:10.1089/aut.2020.0043
31. Hull L, Lai MC, Baron-Cohen S, et al. Gender differences in self-reported camouflaging in autistic and non-autistic adults. *Autism*. 2020;24(2):352–363. doi:10.1177/1362361319864804
32. Lai MC, Lombardo MV, Ruigrok AN, et al. Quantifying and exploring camouflaging in men and women with autism. *Autism*. 2017;21(6):690–702. doi:10.1177/1362361316671012
33. Livingston LA, Shah P, Milner V, Happé F. Quantifying compensatory strategies in adults with and without diagnosed autism. *Mol Autism*. 2020;11(1):15. doi:10.1186/s13229-019-0308-y
34. Perry E, Mandy W, Hull L, Cage E. Understanding camouflaging as a response to autism-related stigma: a social identity theory approach. *J Autism Dev Disord*. 2021;52(2):800–810. doi:10.1007/s10803-021-04987-w
35. Bargiela S, Steward R, Mandy W. The experiences of late-diagnosed women with autism spectrum conditions: an investigation of the female autism phenotype. *J Autism Dev Disord*. 2016;46(10):3281–3294. doi:10.1007/s10803-016-2872-8
36. Bernardin CJ, Mason E, Lewis T, Kanne S. "You Must Become a Chameleon to Survive": adolescent experiences of camouflaging. *J Autism Dev Disord*. 2021;51(12):4422–4435. doi:10.1007/s10803-021-04912-1
37. McDonnell CG, DeLucia EA, Hayden EP, et al. Sex differences in age of diagnosis and first concern among children with autism spectrum disorder. *J Clin Child Adolesc Psychol*. 2021;50(5):645–655. doi:10.1080/15374416.2020.1823850
38. Mandy W, Pellicano L, St Pourcain B, Skuse D, Heron J. The development of autistic social traits across childhood and adolescence in males and females. *J Child Psychol Psychiatr*. 2018;59(11):1143–1151. doi:10.1111/jcpp.12913
39. Lai MC, Lin HY, Ameis SH. Towards equitable diagnoses for autism and attention-deficit/hyperactivity disorder across sexes and genders. *Curr Opin Psychiatry*. 2022;35(2):90–100. doi:10.1097/YCO.0000000000000770
40. Sedgewick F, Leppanen J, Tchanturia K. Gender differences in mental health prevalence in autism. *Adv Autism*. 2021;7(3):208–224. doi:10.1108/AIA-01-2020-0007
41. Ambrose K, Adams D, Simpson K, Keen D. Exploring profiles of anxiety symptoms in male and female children on the autism spectrum. *Res Autism Spectr Disord*. 2020;76:101601. doi:10.1016/j.rasd.2020.101601
42. Hollocks MJ, Lerh JW, Magiati I, Meiser-Stedman R, Brugha TS. Anxiety and depression in adults with autism spectrum disorder: a systematic review and meta-analysis. *Psychol Med*. 2019;49(4):559–572. doi:10.1017/S0033291718002283
43. Lei J, Russell A. I have a fear of negative evaluation, get me out of here! Examining latent constructs of social anxiety and autistic traits in neurotypical and autistic young people. *J Autism Dev Disord*. 2021;51(5):1729–1747. doi:10.1007/s10803-020-04657-3
44. Westwood H, Mandy W, Tchanturia K. The association between symptoms of autism and neuropsychological performance in females with Anorexia Nervosa. *Psychiatry Res*. 2017;258:531–537. doi:10.1016/j.psychres.2017.09.005
45. Westwood H, Tchanturia K. Autism spectrum disorder in anorexia nervosa: an updated literature review. *Curr Psychiatry Rep*. 2017;19(7):41. doi:10.1007/s11920-017-0791-9
46. Brede J, Babb C, Jones C, et al. "For Me, the Anorexia is Just a Symptom, and the Cause is the Autism": investigating restrictive eating disorders in autistic women. *J Autism Dev Disord*. 2020;50(12):4280–4296. doi:10.1007/s10803-020-04479-3
47. Babb C, Brede J, Jones CRG, et al. "It's not that they don't want to access the support it's the impact of the autism: the experience of eating disorder services from the perspective of autistic women, parents and healthcare professionals. *Autism*. 2021;25(5):1409–1421. doi:10.1177/1362361321991257

48. Gibbs V, Hudson J, Hwang YI, Arnold S, Trollor J, Pellicano E. Experiences of physical and sexual violence as reported by autistic adults without intellectual disability: rate, gender patterns and clinical correlates. *Res Autism Spectr Disord*. 2021;89:101866. doi:10.1016/j.rasd.2021.101866
49. May T, Pilkington PD, Younan R, Williams K. Overlap of autism spectrum disorder and borderline personality disorder: a systematic review and meta-analysis. *Autism Res*. 2021;14(12):2688–2710. doi:10.1002/aur.2619
50. Stavropoulos KKM, Bolourian Y, Blacher J. Differential diagnosis of autism spectrum disorder and post traumatic stress disorder: two clinical cases. *J Clin Med*. 2018;7(4):71. doi:10.3390/jcm7040071
51. National Institute for Health and Care Excellence. Autism spectrum disorder in adults: diagnosis and management [Clinical guideline 142]; 2021.
52. Lai MC, Szatmari P. Sex and gender impacts on the behavioural presentation and recognition of autism. *Curr Opin Psychiatry*. 2020;33(2):117–123. doi:10.1097/YCO.0000000000000575
53. Howard PL, Sedgewick F. 'Anything but the phone!': communication mode preferences in the autism community. *Autism*. 2021;25(8):2265–2278. doi:10.1177/13623613211014995
54. Wong DKK, Cheung MK. Online health information seeking and ehealth literacy among patients attending a primary care clinic in hong kong: a cross-sectional survey. *J Med Internet Res*. 2019;21(3):e10831. doi:10.2196/10831
55. Yu L, Stronach S, Harrison AJ. Public knowledge and stigma of autism spectrum disorder: comparing China with the United States. *Autism*. 2020;24(6):1531–1545. doi:10.1177/1362361319900839
56. Haltigan JD, Pringsheim TM, Rajkumar G. Social media as an incubator of personality and behavioral psychopathology: symptom and disorder authenticity or psychosomatic social contagion? *Compr Psychiatry*. 2023;121:152362. doi:10.1016/j.comppsych.2022.152362
57. Tsirgiotis JM, Young RL, Weber N. Sex/gender differences in CARS2 and GARS-3 item scores: evidence of phenotypic differences between males and females with ASD. *J Autism Dev Disord*. 2022;52(9):3958–3976. doi:10.1007/s10803-021-05286-0
58. Whitlock A, Fulton K, Lai MC, Pellicano E, Mandy W. Recognition of girls on the autism spectrum by primary school educators: an experimental study. *Autism Res*. 2020;13(8):1358–1372. doi:10.1002/aur.2316
59. Goldman S. Opinion: sex, gender and the diagnosis of autism—A biosocial view of the male preponderance. *Res Autism Spectr Disord*. 2013;7(6):675–679. doi:10.1016/j.rasd.2013.02.006
60. Rutter M, Caspi A, Moffitt TE. Using sex differences in psychopathology to study causal mechanisms: unifying issues and research strategies. *J Child Psychol Psychiatr*. 2003;44(8):1092–1115. doi:10.1111/1469-7610.00194
61. Lord C, Risi S, Lambrecht L, et al. The autism diagnostic observation schedule–generic: a standard measure of social and communication deficits associated with the spectrum of autism. *J Autism Dev Disord*. 2000;30(3):205–223.
62. Koenig K, Tsatsanis KD. Pervasive developmental disorders in girls. In: Bell DJ, Foster SL, Mash EJ, editors. *Handbook of Behavioral and Emotional Problems in Girls. Issues in Clinical Child Psychology*. US: Springer; 2005:211–237. doi:10.1007/0-306-48674-1_7
63. Kreiser NL, White SW. ASD in females: are we overstating the gender difference in diagnosis? *Clin Child Fam Psychol Rev*. 2014;17(1):67–84. doi:10.1007/s10567-013-0148-9
64. Kaat AJ, Shui AM, Ghods SS, et al. Sex differences in scores on standardized measures of autism symptoms: a multisite integrative data analysis. *J Child Psychol Psychiatr*. 2021;62(1):97–106. doi:10.1111/jcpp.13242
65. Hull L, Mandy W, Petrides K. Behavioural and cognitive sex/gender differences in autism spectrum condition and typically developing males and females. *Autism*. 2017;21(6):706–727. doi:10.1177/1362361316669087
66. D'Mello AM, Frosch IR, Li CE, Cardinaux AL, Gabrieli JDE. Exclusion of females in autism research: empirical evidence for a "leaky" recruitment-to-research pipeline. *Autism Res*. 2022;15(10):1929–1940. doi:10.1002/aur.2795
67. Kopp S, Gillberg C. The Autism Spectrum Screening Questionnaire (ASSQ)-Revised Extended Version (ASSQ-REV): an instrument for better capturing the autism phenotype in girls? A preliminary study involving 191 clinical cases and community controls. *Res Dev Disabil*. 2011;32(6):2875–2888. doi:10.1016/j.ridd.2011.05.017
68. Ormond S, Brownlow C, Garnett MS, Rynkiewicz A, Attwood T. Profiling autism symptomatology: an exploration of the Q-ASC parental report scale in capturing sex differences in autism. *J Autism Dev Disord*. 2018;48(2):389–403. doi:10.1007/s10803-017-3324-9
69. Simcoe SM, Gilmour J, Garnett MS, Attwood T, Donovan C, Kelly AB. Are there gender-based variations in the presentation of Autism amongst female and male children? *J Autism Dev Disord*. 2022. doi:10.1007/s10803-022-05552-9
70. Young S, Hollingdale J, Absoud M, et al. Guidance for identification and treatment of individuals with attention deficit/hyperactivity disorder and autism spectrum disorder based upon expert consensus. *BMC Med*. 2020;18(1):146. doi:10.1186/s12916-020-01585-y
71. National Institute for Health and Care Excellence. Autism spectrum disorder in under 19s: recognition, referral and diagnosis [Clinical guideline CG128]; 2017.
72. Lord C, Rutter M, DiLavore PC, Risi S, Gotham K, Bishop SL. *Autism Diagnostic Observation Schedule, Second Edition (ADOS-2) Manual (Part 1) Modules 1–4*. Western Psychological Services; 2012.
73. Attwood T. *The Complete Guide to Asperger's Syndrome*. Jessica Kingsley Publishing; 2007.