

Understanding the Societal, Disciplinary, and Phenotypic Factors Contributing to Hindered Diagnosis of Autism in Women

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ABSTRACT

This systematic literature review investigates the possible factors contributing to the underdiagnosis of autism spectrum disorder (ASD) in women, focusing on three key areas: social masking, gender bias in ASD screening tools, and the unique female phenotype of ASD. By analyzing existing literature from the James E. Walker Library JEWL Search provided by Middle Tennessee State University, I researched how social masking allows women to hide their symptoms, leading to misdiagnosis or late diagnosis. Additionally, I explored how traditional screening tools may be biased toward male presentations of ASD, further complicating accurate diagnosis. The last analyzed topic regards how a possibly unknown and under researched presentation of ASD in females could affect diagnostic probabilities. In summary, research highlights the urgent need for more research on the misrepresentation of ASD diagnosis in women. Though findings were limited and inconsistent, understanding these challenges is crucial for developing effective diagnostic criteria and intervention strategies that carefully consider the distinct presentations of autism in females, promoting improved mental health support and outcomes.

Autism spectrum disorder (ASD) is a neurodevelopmental disorder indicated by obstacles with social communication and interaction, restricted and repetitive behaviors, and sensory integration (American Psychiatric Association, 2022). The condition develops during childhood and lasts throughout one's lifetime (DiRienzo et al., 2023). In America, approximately 1 in 36 kids (Amoakohene et al., 2023) and 1 in 41 adults (Dietz et al., 2020) are diagnosed with autism spectrum disorder each year, and the numbers seem to be increasing. However, there is no clear answer yet as to why (Aylward et al., 2023). Although around 2% of the population is diagnosed with ASD, males are significantly more likely to receive a diagnosis than females and tend to be diagnosed earlier in life (Belcher et al., 2023). In fact, males are two to five times more likely to receive a diagnosis of ASD compared to females (Bishop et al., 2020). The reasons for gender disparity in ASD diagnosis are unclear, but there are many theories. One theory is the presence of a biological sex difference that could explain why more males are diagnosed with ASD. However, a more popular viewpoint is that males and females have an equal likelihood of having ASD, but females experience hindered diagnosis due to camouflaging symptoms, gender-biased screening instruments, varying ASD symptoms, and possibly a combination of these determinants.

The first possible answer to why females are less likely to get diagnosed with ASD is that females are more likely to camouflage their symptoms. Camouflaging is used as a coping mechanism that involves concealing actions or manners associated with ASD, or more specifically, "using explicit techniques to appear socially competent" (Allison et al., 2017b, para 1). In places where the disorder is typically identified in children, such as in the classroom, camouflaging can be a coping mechanism for the person masking their ASD symptoms in the sense that they might feel like they fit in with others more. However, camouflaging also prevents teachers and clinicians from being able to distinguish ASD and work towards early intervention, leading to poorer long-term outcomes for females with ASD (Dean et al., 2017).

The next possible explanation for why females with ASD are overlooked in diagnosis is due to diagnostic screening tools that are possibly biased in favor of males with autism spectrum disorder. Though there is little research on this idea, questions have surfaced about whether the expected male preponderance in ASD has affected what gender-specific symptoms and behaviors clinicians look for in tests (Allison et al., 2017a). This bias can emerge if the screening tools are not gender sensitive, making the instruments questionable in terms of being used for females with autism. For example, many autism diagnostic tools are based on behaviors more commonly observed in males, such as overt social difficulties and restricted interests like an intense focus on objects or patterns. However, females with autism might display more subtle social challenges, or their restricted interests might align more closely with socially acceptable activities like reading or collecting items. If these gender differences are not accounted for, females may be underdiagnosed or misdiagnosed (Kopp & Gillberg, 2011).

One other theory that could explain why females go undiagnosed, misdiagnosed, or have a delayed diagnosis is that there is not enough research on the female phenotype for autism spectrum disorder. Studies, though scarce, have shown that there are sex differences in the behavioral manifestation of ASD symptoms between males and females with ASD, but there is little evidence of whether these differences are specific to the disorder (Bishop et al., 2020; Demetriou et al., 2021). In particular, females with ASD demonstrated higher cognitive performance, although it is not yet proven that this might not also be the case for neurotypical females (Demetriou et al., 2021). Variable aspects of cognitive performance include adaptive behavior, social participation, restrictive and repetitive behaviors (Bishop et al., 2020), verbal learning, memory, and information processing speeds. These variations in cognitive abilities can complicate ASD diagnoses, as they may lead to some individuals being misdiagnosed or overlooked entirely if the diagnostic criteria do not account for this wide range of cognitive functioning (Demetriou et al., 2021).

Though there is still insufficient research to answer why women and young girls with ASD are more likely to receive a delayed diagnosis or no diagnosis at all, the issue is vital to bring awareness to because of the consequences of not being knowledgeable. These consequences could include abnormal levels of anxiety and/or depression for young and adult females without an accurate diagnosis of ASD (Allison et al., 2017b; Haney, 2016). In addition, a delayed diagnosis

can prevent some from receiving early intervention, typically as a child, which has been shown to greatly benefit those with ASD when living a normal life (Haney, 2016). A more thorough understanding of the factors that may lead to misdiagnoses – including social masking, bias in diagnostic tools, and phenotypic variation – could help prevent these poor outcomes.

Methods

Sample of Studies and Inclusion Criteria

Academic sources for this systematic literature review were found using the James E. Walker Library JEWL Search provided by Middle Tennessee State University. The first search in the database was titled “hindered diagnosis in ASD,” and there were 50,571 articles. To determine which articles to analyze further, several inclusion criteria were applied. First, only peer-reviewed academic journals were considered to maintain credibility. Second, all the sources were required to be written in English to ensure accessibility. Third, to prioritize recent, relevant, and up-to-date articles, searches were restricted to publication dates ranging from 2000 to 2024 (when the search was conducted). Finally, the included articles only focused on comparing biological males and females, or cismen and ciswomen, due to a lack of research on autism diagnosis in trans and nonbinary individuals. Participants of any age were considered. After assessing these criteria, a total of 12,259 sources were excluded. Next, advanced searches were conducted for each potential explanation – social masking, bias in diagnostic tools, and phenotypic variation. To determine which articles were relevant to each topic, the titles and abstracts were reviewed. Specific search terms for each topic are described below. Figure 1 displays the search process and the number of articles included and excluded at each stage of the literature review.

Social Masking

For specificity on the tendency for girls to camouflage their symptoms, the following search phrase was used in the JEWL Search: [((hindered diagnosis in ASD)) AND ((women) OR (female) OR (woman) OR (females)) AND ((autism) OR (ASD) OR (autism spectrum disorder)) AND ((social masking)) NOT ((face masks) AND (facemasks) OR (face covering)) OR ((female phenotype autism)) AND ((mood disorders) or (depression) or (bipolar) or (anxiety)) OR ((gender bias in autism diagnosis))]. A total of 45 academic sources written in English surfaced with this search. Of these articles, 31 were deemed irrelevant, based on the title and abstract. Therefore, a total of 14 sources were included in this review (see Table 1).

Gender Bias in Screening Tools

To locate articles dealing with the issue of gender-biased diagnostic tools, the advanced JEWL search included the following phrases: [(gender bias in Autism screening tools)] AND [(sex discrimination)] OR [(autism diagnostic tools)]. A total of 335 sources were identified, and scholarly journals published in or after 2000 were the only materials considered for this analysis. After applying these limiters, a total of 91 sources remained. When adding the phrase [(AND gender)] to JEWL Search, only ten articles appeared, and none were helpful. After removing that phrase and choosing the subjects “autism,” “diagnosis of autism,” “screening,” “questionnaires,” and “diagnostic assessment,” 10 articles surfaced, and all 10 of them seemed to be related to racial discrimination. As a result, I changed the phrases in my search altogether to [(gender bias in Autism screening tools)] AND [(women) OR (female) OR (woman) OR (females)]. I also checked the subject “gender bias” and found 353 articles. Out of the 353 articles, 90 were about gender bias; of these, 10 sources qualified based on an examination of the title and abstract (see Table 2).

The Female Phenotype

The first search in the advanced JEWL search for this topic was phrased as [(female

phenotype in autism spectrum disorder)] OR [(female phenotype theory)] AND [(autism) OR (ASD) OR (autism spectrum disorder) OR (Asperger's) OR (Asperger's syndrome) OR (autistic disorder) OR (Aspergers)] OR [(Sex differences in ASD)], which yielded 843 potential sources. Then, the following subjects were selected: "Sex differences," "female," and "autism spectrum disorder." After applying the decided subjects, 36 peer-reviewed studies remained, and 14 sources were relevant for this project (see Table 3).

Figure 1. JEWL Search Methods

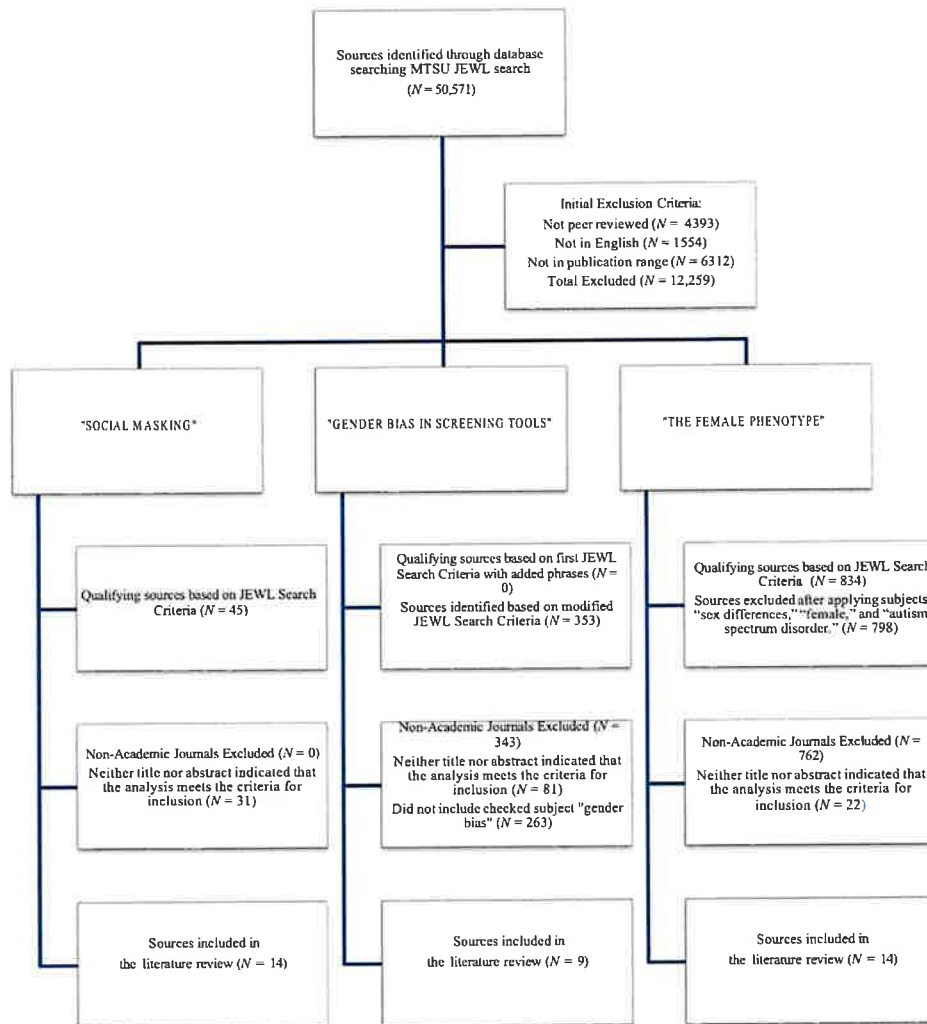


Table 1. Sources on Social Masking

Study	n	Age of Diagnosis (Years)	Research Design and Analysis	Main Themes	Country/Race/Ethnicity	Sex differences	Limitations
(Allison et al., 2017b)	(92) 3 excluded, n = 89	16+	Thematic analysis	Motivation: "I Want to Know and Be Known" Consequences: "I'm Not My True Self"	55% British, 15 nationalities	can only speculate differences in attitudes	only demonstrates ASD in adults (m & f)
(Anderson et al., 2023)	12 (qualitative studies), number of participants not recorded	N/A, age at time of study = 18+	Systematic literature review	"I tend to fall between the cracks" "The hidden hurt" "Having to meet the expectations of others"	N/A	"Increased risks of exploitation for autistic women" "delinquent influences on psychological wellbeing for autistic women"	did not account for adolescents with ASD; did not compare or focus on males; ethnicities not reported
(Burns et al., 2016)	10 (with ASD (15 not recruited))	4-15	Interpretative phenomenological analysis (IPA)	"Overcoming challenges" "Desire for friendships" "Experiences of Social Environment"	NA	most females were motivated to have friendships, with rejection, they began masking	Only includes females with diagnosis based on "male-based criteria"; males were not compared
(Baren-Cohen et al., 2021)	277 (206 diagnosed: 78 females & 128 males)	$\bar{x}$ = 12-14	thematic analysis	Motivations: Lack of awareness and acceptance of ASD " dangers of camouflaging" " positive aspects of camouflaging"	N/A	SD of ASD females' camouflaging scores were higher than males with ASD	Not all participants were diagnosed, but self-identifying; survey only available online (also a strength)
(Cage & Troseel-Whitman, 2019)	262 (135 females, 111 males, 12 other gender identities)	>18, $\bar{x}$ = 33.62	cross-sectional survey	"Falling in and passing in a neurotypical world" "avoiding retaliation and bullying by others" "Internalised stigma"	N/A	both male and female participants rated conventional reasons for camouflaging more highly (an relational reasons; females endorsed conventional reasons more than males	ethnic diversity and education level not accounted for; only represented highly verbal individuals; not generalizable
(Clarke et al., 2021)	24: 8 loads (girl/parent/educator)	7-12	Thematic analysis	4 themes (look at figure 2)	7 white british; 1 white european	women and girls vulnerable to camouflaging and having negative impacts	small sample; only includes autistic girls
(Cook et al., 2021)	29 (studies reviewed): N = 2254 (Participants across all studies) 32: 98 (adult studies)	$\bar{x}$ = 7 (adult studies used in calculations); $\bar{x}$ = 32: 98 (adult studies)	systematic review	Inconsistent results	86.1% white	Out of the 9 studies on sex differences, 7 reported females/identifying girls had increased rates of camouflaging through adolescence	samples differed on key demographics like sex and gender, IQ, and age of diagnosis
(Cook et al., 2024)	133 autistic individuals	3-68	thematic analysis	"embracing diverse communication styles, interests and perspectives" "creating a more inclusive mixed-neurotype social environment together" "minimising and managing mixed-neurotype miscommunication in mutually beneficial ways" "enjoyable interactions involving reduced anxiety and exhaustion as well as genuine connection and rapport"	62.4% white	Sex/gender not considered	sample was mostly white, educated individuals who received ASD diagnosis late in life; not generalizable
(Dean et al., 2017)	96 (24 m with ASD, 24 f with ASD, 24 m without ASD, 24 f without ASD)	(M) $\bar{x}$ = 7.71 (F) $\bar{x}$ = 7.75	ANOVA	Measurement of "Game" "Engagement with Peers" "and Solitary"	NA (assumed all American)	Look at Table 4 (Found significant between-group differences). Girls with ASD have social experiences that differ from those of boys with ASD	size and duration of study was small
(Field et al., 2024)	13 studies	NA	meta-ethnography of 13 qualitative studies	"It's a defense mechanism really" "unintended consequences" "No harm done in my opinion"	NA	Sex/gender not considered	Sex/gender, diverse backgrounds, not considered; sex differences not considered
(Hull et al., 2020)	592 (Participants reported ASD diagnosis, n = 276) (Participants did not report ASD diagnosis, n = 230)	N/A, age at time of study: $\bar{x}$ = 36.8	power analysis	suggestions that camouflaging might be driven by situational pressures, such as stigma or individual differences	N/A	"Camouflaging was negatively related to extraversion, agreeableness, and conscientiousness, while it was positively related to neuroticism"	study conducted online, so participants' diagnoses were not verified, but all participants were required to share healthcare provider that diagnosed them
(Jorgenson et al., 2020)	140 (Males w ASD, n = 55; Females w ASD, n = 23) (Males without ASD, n = 27; Females without ASD n = 35)	N/A; age at time of study = 13-18	ANOVA and MANOVA	Differences in camouflaging by age and developmental stage	N/A	When age was not accounted for, TD females and ASD females had more similar CAT-Q scores than males. When age was accounted for, there were no significant sex differences	smaller samples could affect results, whether age was accounted for or not, no demographic data collected
(Raz & Schneid, 2020)	24 (2 were self diagnosed)	N/A; age at time of study = 16-55 (Mean = 31)	Qualitative analysis	"Passing as normal" "What is wrong with me?"	All Israeli	Sex/gender not considered	intellectual ability not measured, small samples

Table 2. Sources on Gender Bias in Screening Tools

Study	n	Age of Diagnosis (Years)	Research Design and Analysis	Key Findings	Country/Nationality/Race	Limitations	Bias Found?
(Allison et al., 2017)	557 females, 680 males	N/A; age at time of study: $\bar{x} = 35.02$	parallel analysis	No substantial biases were found in the AQ-10	majority White European	Participants were not randomly selected	no
(Atherton et al., 2023)	300 (149 male, 151 female)	(18 to 72, $\bar{x} = 26.79$)	Implicit Association Test	"A significant implicit gender bias towards associating males with autistic traits was found using the IAT."	81.27% White, 2.22% Black or African American, 12.70% Asian, and 3.81% Other	included an online sample; could be argued the study was based on "male-biased" ASD traits	yes
(Auyeung et al., 2019)	1,237 with a clinical diagnosis, 7,356 controls	N/A; age at time of study: $\bar{x} = 33.34$	parallel analysis	No substantial biases were found in the AQ-10 (Replication of study by Murray et al. (2017)): Results showed males having higher levels of ASD traits than females.	NA	Important demographic factors not included: ethnicity, class, education level	no
(Belcher et al., 2023)	5246 women, 1830 men	N/A; age at time of study: $\bar{x} = 32.22$	survey	women had a higher probability of endorsing items relating to social skills and communication	UK	ethnicity was not accounted for; not generalizable	yes
(Gross et al., 2022)	6269 children	Age at time of study = 1.7-17.9	observational study	Findings were inconsistent; "hand mannerisms item demonstrated consistent bias across ADOS-2 modules among female children compared with male children;" females found this item more difficult	1619 Black/African American, 3151 White	lack on info on ethnicity; samples too small to measure bias in other ethnic groups	Yes, but magnitude of bias is small
(Kopp & Gillberg, 2011)	191	6-16 at time of study	correlation	most items were non-significant except the following: "lacks best friend" was high in ASD male group; more rated symptoms in girls were "has a different voice/speech," "avoids demands,"	N/A	small sample from a referred clinic; parental biases to be considered	Predicted with nature of questions, but not tested
(Lonergan, 2021)	7 (results data from 7 diagnostic tools)	NA	systematic review	"Low representation of females in validity samples of tools"	UK	more research is needed	Yes
(Noble et al., 2014)	145 (94% with ASD, 6% with other developmental disorder)	N/A; age at time of study: $\bar{x} = 2.34$ (all under 36 months)	diagnostic assessment analysis	14% of ASD participants passed the screening test; 74% were accurately classified; 86% of ASD children were accurately classified as at-risk	N/A	limited demographic info	Yes, but not directly with gender; could be applied to gender with further research

Study	N	Age of Diagnosis (Years)	Research Design and Analysis	Main Themes	Country/Nationality/Race	Sex differences	Limitations
(Abbo et al., 2022)	81 Females, 156 Males	Males: $\bar{x} = 6.6$; Females $\bar{x} = 7.8$	Statistical analysis	NA	Uganda	females received later diagnosis than males in Uganda	missing data on sex, age of diagnosis, and diagnosis
(Bargiela et al., 2016)	14 women	15+ (10-35 during the study) precise ages not given to protect confidentiality	Semi-structured interview	Pretending to be normal: "You're not autistic"; Forging an identity as a Woman with ASD	UK	Only women are discussed	1st study to focus on late-diagnosed women with ASD (to their knowledge and my own)
(Bishop et al., 2020)	76 females, 471 males	N/A; age at time of study: 14-21	independent sample t tests	teachers and parents reported little differences in social interaction between sexes	45% Non-White and/or Hispanic	standardized scores showed females with ASD being more behaviorally divergent; no significant differences	potential biases in parent and teacher reportings; cannot generalise findings
(Boan et al., 2020)	480	N/A; age at time of study: 10-72 months	quantitative design	significant sex differences in social communication	White 79%, Black 15%, Other 6%, Hispanic ethnicity 5%	girls had bigger social communication deficits than boys, but differences varied by age; no significant sex differences in RRBs	reliance on clinical samples, potential bias in parent-reported data; lack of diversity
(Castrién et al., 2023)	79 studies: Girls/women with only ASD 3131, Boys/men with only ASD 7419, Girls/women with ASD with ID 1253, Boys/men with ASD with ID 2955	N/A; age at time of study: $\bar{x} = 8-15$	meta-analysis	behavior differences and symptom presentation with vs without ID	NA	girls with ASD and ID showed more severe social and repetitive behavior challenges, and poorer language and motor skills than boys	^a ID = intellectual disability
(Chakrabarti et al., 2020)	215 ASD cases	NA	correlation	sex-specific biological differences in how serotonin and 5-HT _{1A} influence brain development and function	NA	males had higher levels of serotonin and 5-HT _{1A} in the blood; females displayed more severe ASD behaviors	^a 5-HT _{1A} is a metabolite for serotonin
(Dermatou et al., 2021)	45 males 34 females	N/A; age at time of study: 8-18	comparative design	females showed some strengths in cognitive tasks compared to males	NA	females with ASD scored higher in verbal learning, memory, and psychomotor speed; neurotypical females also scores higher	narrow age range, small sample size
(Enicoff et al., 2016)	27 w ASD, 23 controls	N/A; age at time of study: 19-56	whole brain analysis with fMRI	highlighted a need to consider sex differences	NA	males with ASD had reduced activity in pSTS when processing social info	small samples
(Hardley et al., 2008)	157 males, 42 females	N/A; age at time of study: 1, 5, 3, 9	MANOVA	sex differences were subtle but noteworthy	97 participants Caucasian, 102 Other	girls had more sleep problems and symptoms of anxiety or depression; boys displayed higher levels of restricted and repetitive behaviors	small sample size, especially females
(Hiller et al., 2014)	69 males, 69 females	N/A; age at time of study: Males: $\bar{x} = 8.76$; Females $\bar{x} = 8.06$	cross-sectional design	Sex differences in overt behaviour presentation, restricted interests and school presentation	NA	some behavioral patterns varied between sexes	small, specific sample
(Kusel et al., 2022)	5 studies, 169 participants	NA	comparative design	biological sex differences affecting ASD symptoms	NA	differences in gene expression, sex-specific alterations in brain regions	small sample size, limited demographic data
(Rippon, 2024)	48	NA	meta-analysis	points out neglect of females in ASD research	NA	vmPFC activation (rewarding stimuli) in females but not males	no new data on sex differences; demographics not mentioned

Social Masking: Results and Discussion

Camouflaging, also referred to as social masking (Allison et al., 2017b; Dean et al., 2017), is used as a coping mechanism that involves concealing actions or manners associated with ASD, or more specifically, “using explicit techniques to appear socially competent.” (Allison et al., 2017b, para 1). In places where the disorder can be identified in children, such as in the classroom, camouflaging can feel like a coping mechanism to the person masking their ASD symptoms in the sense that they might feel like they fit in with others more, but this also prevents teachers and clinicians from being able to distinguish ASD and work towards early intervention (Dean et al., 2017). Masking ASD symptoms makes diagnosing difficult for clinicians because those who mask will observe and mimic social skills deemed socially normal in order to feel accepted by others (Allison et al., 2017b; Clarke et al., 2021; Radulski, 2020).

Results

Some studies on social masking relied heavily on qualitative interviews (Allison et al., 2017b; Anderson et al., 2023; Burns et al., 2016; Clarke et al., 2021; Field et al., 2024; Raz & Schneid, 2020) to explore subjective experiences of camouflaging. Others used more quantitative approaches (Dean et al., 2017; Hull et al., 2020; Jorgenson et al., 2020) to measure personality traits, emotional intelligence, and gender/sex differences associated with camouflaging. Overall, strong effects were found in most of the reviewed sources, but each source had its limitations. See Table 1 for more details.

Discussion

Creating and performing strategies to seem neurotypical is found to be more common in females with autism spectrum disorder than males with autism spectrum disorder (Allison et al., 2017b; Clarke et al., 2021; Hull et al., 2020). In fact, Dean et al. (2017) found that the young males in their research were more likely to have socially unaccepted special interests and repetitive behaviors. In contrast, the young females had more admissible interests and behaviors and were less likely to be noticed and reported by teachers. Further outcomes would include the young males having a higher likelihood for intervention and treatment since school faculty members play a significant role in identifying early signs of ASD (Dean et al., 2017).

In cases where males engaged in camouflaging, they were often more satisfied with themselves than the females, who tended to internalize their actions (Clarke et al., 2021). Hull et al. (2020) and Radulski (2020) discuss how the effects of internalization can lead to negative consequences, including the following: fatigue (Hull et al., 2020), mental health issues (Hull et al., 2020; Radulski, 2020), identity crisis (Hull et al., 2020), loneliness (Schiltz et al., 2021), and suicidal ideations (Hull et al., 2020). Therefore, women and young girls who regularly camouflage and meet ASD criteria are more likely to have lower mental health wellbeing (Allison et al., 2017b; Radulski, 2020) compared to the males who either do not camouflage and receive ASD intervention (Dean et al., 2017), or the males who feel content after camouflaging (Clarke et al., 2021). Due to the included internalized effects and less probability of ASD identification for women and young girls, there is a prospect of many receiving a delayed or wrongful diagnosis, resulting in abnormal levels of anxiety and depression.

Motivations Behind Social Masking

In a study conducted by Burns et al. (2016), 10 adolescent girls aged 13-19 years with a diagnosis of autism spectrum disorder participated in semi-structured interviews, during which they shared their accounts of experiencing hardships in social environments. Before interviews began, the participants were asked to draw a circle on a sheet of paper to indicate themselves. They were also asked to draw a triangle for each friend they have and place it at a distance that they

think signifies how close they are to this friend. Although the results were not included in the final data examination, the pages with the drawings were used to gain an understanding of the social situations that occurred with each individual prior to the interviews.

After the interviews were conducted, interpretative phenomenological analysis (IPA) was used to analyze the collected data. IPA approaches data by using familiarization, which involves identifying key words in the study, recognizing common or related themes in each participant's answers, and grouping these ideas into broader subject matters. In the analysis, input from parents was included in the results section, but they typically only commented when their daughters were unable to verbally communicate their experiences during the interview process. After the interpretative phenomenological analysis was complete, the individuals were given time to review their results and were encouraged to comment on the accuracy of the analysis. When the participants had something to add, their feedback was also included in the study. To reduce bias, a researcher who was not involved in this study reviewed the entire process and concluded that there was no evident bias.

Overall, there were concluded to be three "superordinate themes," with the responses of the 10 adolescent girls with ASD, which include the following: "experiences of social environment," "desire for friendships," and "overcoming challenges."

Experiences of Social Environment. Regarding their feelings and perceptions towards social circumstances, common reactions were narrated as uncomfortable and distressing due to feelings of unsafety and fear of rejection. One participant, under the pseudonym "Emily," stated, "It feels like in my classroom that I'm surrounded by lions... I feel like a mouse, and everyone else is like a giant cat or something." Regarding rejection, Rosie commented, "I would be happy to hang around with them if they would accept me, but they don't." Furthermore, most participants recalled sensory overstimulation in social situations, especially in school when they had to change between classes. Kate said, "I was really sensitive to everything and that was a huge stress cos [I] was like hurting physically as well... loud noises used to really scare me and hurt in my head."

Desire for Friendships. In this theme, participants discussed their experiences with yearning for friendships, their motivations for forming new friendships, and the challenges of creating and maintaining them. Each of the ten individuals expressed the desire to have friendships but felt they had to work extra hard to meet new friends and keep those friendships. They also showed tremendous gratitude for their friends. Rosie explained, "When I'm not with friends... I... think of all the bad things in life and just carry on thinking about them over and over and over and over until I get really depressed."

Regarding group communication, most participants found engaging in groups to be distressing, resulting in withdrawal from group participation. Emily shares her experience by saying, "With a group, there's loads of different people all at different times and it turns into a murder mystery game of Cluedo where somebody made this go wrong, we've got to find out who it is, people blaming each other, splitting each other up and just wrecking everything. I don't like it at all." Another factor contributing to social group withdrawal was social exhaustion, which led to the deterioration of friendships over time. Kate states the following in her account, "I don't really talk to people as much as most other people cos quite often, you know, it's too much effort. So it's a bit stressful cos you have to sort of think about the other person's feelings, it's just a bit too much for me sometimes... but I do worry quite a bit like if the friendship starts to deteriorate and they don't see me as friends any more."

Overcoming Challenges. All ten girls in the study expressed the need for support from others to make new friends, whether it be from their parents or those who initiated interactions first. Five out of the ten participants also stated that they practiced empathy to build social skills. Gemma gives a good example: "I have a very good memory so I can... relate that to a situation the other person's in... I sort of used that memory and just associated with what she knew." Additionally, many participants imitated their peers to minimize the risk of standing out. For example, Laura mentioned feeling the need to copy others, but not in an overt way. Gemma described similar experiences: "I would try and copy them but not look like I was exactly copying them, so if they were playing a game and they moved and they did something, then I would try and copy it but

not exactly like them.” Every girl in the study described feeling “anxious” and “unhappy” in social circumstances and explained how they mask their feelings, whether by putting on an overly joyful face or an excessively blank stare. The interviewer asked Gemma how masking affects her emotionally, and she described how tiring it was to keep up a persona. To cope with the emotional baggage that comes with masking, five out of the 10 participants talked about how they had tried self-harm or had suicidal tendencies. Joëlle said, “I was getting to the point where I actually couldn’t control some urges that I had... I was a little bit suicidal.”

Limitations

This review had several limitations. First, a few sources discussed the personal experiences of individuals diagnosed with ASD who camouflaged (Allison et al., 2017b; Dean et al., 2017). Although there were possible explanations for the differences between females and males, research was limited in explaining why this discrepancy occurred. Smaller samples, which limited the generalizability of findings, were also a common issue. Another significant limitation is the exclusion of research on anyone with ASD who is not a biological male or female. If women with ASD are more likely to camouflage, hypothetically, due to a biological issue, what does this mean for trans and nonbinary individuals? Studies testing this issue are scarce. Cultural factors may also limit research, as most studies typically involve women from Western cultures and countries. Further research is needed to investigate the experiences of autistic women from non-Western cultures and countries.

Clinical Implications

Social masking creates significant challenges when it comes to diagnosing and treating autism, especially in females who tend to mask their symptoms more often (Allison et al., 2017b; Dean et al., 2017). Clinicians need to realize that people with ASD might use intentional strategies to hide their traits in social situations, like copying behaviors that are seen as more socially normal to fit in (Allison et al., 2017b). This might help the person feel more accepted by others, but it can also delay or prevent a proper diagnosis, especially in environments such as schools, where early detection is crucial for receiving the right interventions at the right time (Dean et al., 2017).

Gender Bias in Diagnostic Tools: Results and Discussion

Even though there is not a ton of research on gender bias in autism diagnoses, some studies bring up valid concerns about how the belief that ASD is more common in males may influence the traits clinicians focus on during evaluations (Allison et al., 2017a). Diagnostic tools are often developed using data from predominantly male participants, which tends to emphasize behaviors more typical of males, such as noticeable social issues and narrow interests. Diagnostic tools with a male-focused approach can unintentionally lead to overlooking how autism presents in females, who may express different or more subtle symptoms. For example, girls with ASD might internalize their difficulties or use strategies like camouflaging, making their symptoms harder for clinicians to pick up on (Allison et al., 2017b; Dean et al., 2017).

Results

In terms of finding significant data on gender bias in screening tools, most of the sources did not find substantial results, meaning there was not an alarming amount of bias found in ASD diagnostic tools. Some sources found bias in certain items within the diagnostic tool being analyzed, rather than in the overall test. See Table 2 for more information.

Discussion

To investigate gender bias in autism diagnostic tools, Atherton et al. (2023) examined such biases in a random sample of participants, utilizing both quantitative and qualitative data. They conducted two studies to examine implicit and explicit gender biases. In the first study, implicit association tests (IATs) were used to measure unconscious biases in participants, assessing whether they implicitly associated autism with males more often than with females. IATs are commonly used in psychology to reveal biases that participants might not be consciously aware of, making them a valuable tool for exploring implicit associations. The second study examined explicit biases by presenting participants with hypothetical scenarios describing autistic traits. These scenarios included both male and female characters to see if participants were more likely to associate autistic traits with men than with women. This approach helped the researchers test conscious gender biases in participants' perceptions. Combining implicit and explicit measures allowed the researchers to provide a more complete picture of gender biases in ASD perceptions.

Both studies found a strong gender bias in how people perceive autism. The implicit association tests revealed that participants tended to associate autism more with males than with females. The bias was also seen in the scenario-based study. For example, participants were more likely to associate autistic traits with male characters, even when the descriptions of traits were exactly the same for both genders. These results could point to a deeply rooted belief that autism is primarily a "male" condition, which could impact how autism gets recognized, diagnosed, and supported for different genders.

Limitations

A common limitation across the reviewed studies was the use of small samples, which limited the generalizability of the results. In addition, most of the analyzed sources did not include demographic data, omitting potentially critical information that could impact the interpretations. See Table 2 for the complete list of limitations.

Clinical Implications

Updating screening tools to incorporate the diversity of ASD presentations between males and females, especially by considering factors like camouflaging and internalized symptoms, could help clinicians provide earlier, more accurate diagnoses. Improved tools might also encourage clinicians to address the broader spectrum of ASD experiences, ultimately leading to more comprehensive care (Lai & Szatmari, 2020).

The Female Phenotype: Results and Discussion

The theory of the female phenotype in ASD suggests that females may frequently go undiagnosed, be misdiagnosed, or face delays in their diagnosis because there is not enough research on the female presentation of ASD. Although there is limited research, existing studies have indicated that males and females with ASD show different behavioral presentations of symptoms, emphasizing potential sex differences. However, there is insufficient evidence to definitively determine whether these differences are specific to ASD or also present in the general population (Bishop et al., 2020; Demetriou et al., 2021). For instance, Demetriou et al. (2021) found that females with ASD tend to display higher cognitive performance compared to males. However, it is still unclear whether this difference is exclusive to females with ASD or reflects a broader trend seen in neurotypical females as well. This cognitive advantage could make ASD symptoms less noticeable in females, leading to underdiagnosis. In addition, the cognitive performance of individuals with ASD can vary significantly in areas such as adaptive behavior, social interaction, restricted and repetitive behaviors (Bishop et al., 2020), as well as verbal learning, memory, and processing speed (Demetriou et al., 2021). These aspects of cognitive functioning can complicate the diagnostic process for ASD because diagnostic tools may not sufficiently capture the factors that affect diagnosis. As a result, some individuals, particularly

females, may be overlooked or misdiagnosed. Wrongful diagnosis prioritizes the need for more gender-sensitive research and diagnostic criteria to address these factors more accurately.

Results

Researchers used a range of methods, from neuropsychological testing and behavioral assessments to neuroimaging and transcriptomic analyses, to explore the female phenotype of ASD. Many studies relied on standardized diagnostic tools to assess ASD traits and used teacher and clinician reports to analyze observable behavior differences by sex. Self-report tools, neuropsychological tasks, and executive function tests were also used to examine camouflaging behaviors, particularly among females. Finally, meta-analyses and systematic reviews were included to collect broad data. Overall, the studies presented significant indications that males and females with ASD often present differently in terms of social behavior, cognitive functioning, and neural activity. Refer to Table 3 for more information.

Discussion

In the study conducted by Demetriou et al. (2021), researchers highlight that ASD affects males and females differently, leading to variations in symptoms and coping strategies. Specifically, the study examines both executive and non-executive cognitive functions through a combination of neuropsychological tests and self-report measures. Cognitive functions refer to mental abilities essential for tasks ranging from basic reasoning to complex problem-solving. Executive functions (EF) are a subset of these, encompassing skills such as working memory, flexible thinking, and self-control. Non-executive cognitive functions, on the other hand, include abilities such as basic information processing and sensory perception.

To examine sex-based differences, the researchers used a combination of standardized neuropsychological tests and self-report measures. Neuropsychological tests are structured tasks or activities designed to assess specific cognitive functions. In this study, tests for executive functions likely included tasks measuring working memory, cognitive flexibility, and inhibitory control, all of which are commonly assessed in ASD research. Self-report measures allowed participants to describe their own cognitive and emotional experiences, adding a subjective component to the objective data collected.

Demetriou et al. (2021) found significant differences between males and females with ASD in both executive and non-executive cognitive functions. In executive function tasks, females with ASD generally performed better on measures related to working memory and cognitive flexibility compared to males. Working memory, which is the ability to hold and manipulate information for short periods, is essential for tasks such as problem-solving and decision-making. Cognitive flexibility, or the ability to switch between tasks or thoughts, is crucial for adapting to new situations. These results suggest that females with ASD may have specific cognitive strengths compared to males with ASD, possibly due to their tendency to develop coping strategies that help them navigate social environments.

Limitations

Research on the female phenotype in ASD faces several limitations. First, much of the existing data is based on small sample sizes, which limits generalizability and the ability to capture the full range of experiences among females with ASD. In addition, self-report methods were commonly used among reviewed sources; research could be influenced by personal bias or social desirability. Lastly, many of the qualifying studies had small samples of females compared to males. Refer to Table 3 for further discussion of limitations.

Clinical Implications

The female phenotype often presents with subtle or atypical symptoms, including increased social motivation, camouflaging behaviors, and internalizing difficulties like anxiety and depression (Dean et al., 2017; Hull et al., 2020). These characteristics can lead to misdiagnosis or delayed diagnosis in girls, who might not exhibit the typical ASD signs observed in boys.

Clinicians need to be aware of these differences and consider modified screening tools and diagnostic criteria that account for camouflaging and internalized symptoms in females, ensuring early and accurate identification (Bargiela et al., 2016; Hiller et al., 2016).

It is essential for families, teachers, therapists, and community members to understand how autism shows up differently in girls and women. Learning to recognize these more subtle signs can help create supportive spaces that reduce the pressure to mask and encourage acceptance. Enhanced understanding among those closest to autistic females can also help reduce social stigma and create settings where girls feel safe being themselves (Burns et al., 2016).

General Discussion

In studying the barriers to diagnosing autism spectrum disorder (ASD) in women, it has become apparent that traditional diagnostic criteria and tools are not fully equipped to identify autism in females accurately. Research highlights that ASD often presents differently in women and girls, with more hidden symptoms compared to the typical male ASD presentation, on which diagnostic criteria were originally based (Dean et al., 2017; Hull et al., 2020). Females with ASD are often more socially motivated and engage in camouflaging behaviors, where they consciously mask or adapt their behaviors to fit social expectations. This ability to camouflage can lead to delayed or missed diagnoses because outward signs of autism may be less visible or appear differently than in males (Bishop et al., 2020; Demetriou et al., 2021; Haney, 2016).

In addition, females with ASD often experience internalizing symptoms like anxiety and depression, which are not always associated with autism in standard assessments but are prevalent among autistic women (Allison et al., 2017b; Haney, 2016). This highlights the need for more inclusive, gender-sensitive diagnostic practices and criteria to ensure timely and appropriate support for females with ASD. Limited research involving this field also highlights the need for more studies to better understand why women are overlooked in diagnosis.

Conclusion

The growing awareness of disparities in autism diagnoses between men and women has led to increased focus on three key theories of why females are not being properly diagnosed: higher rates of social masking, gender bias in diagnostic tools, and the theory of a female phenotype for autism. Each of these explanations not only sheds light on the reasons behind underdiagnosis but also highlights the significant mental health consequences for women who remain undiagnosed or misdiagnosed.

Researching and addressing the underdiagnosis of autism in women should be essential for promoting equity in mental health care. By understanding the role social masking plays in hindering diagnosis, challenging gender biases in diagnostic tools, and exploring the female phenotype, healthcare professionals can develop more inclusive diagnostic criteria and interventions. This shift would not only validate the experiences of autistic women but also empower them to access the support and accommodations they need to thrive. Investing in this area of research would ultimately promote a more inclusive and compassionate society that recognizes and meets the diverse needs of all individuals.

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