



Sex and gender impacts on the behavioural presentation and recognition of autism

Meng-Chuan Lai^{a,b,c,d,e} and Peter Szatmari^{a,b,c}

Purpose of review

With increasing awareness of potential differences of autism presentation in nonmale versus male individuals, this review summarizes the rapidly evolving literature on sex and gender impacts on autism across nosology, behavioural presentation, developmental change and contextual recognition biases.

Recent findings

Most studies have not differentiated sex versus gender impacts. Regarding behavioural presentation, measurement invariance across sex/gender was found in several standard measures. On this basis, diagnosed females overall showed lower restricted/repetitive behaviour/interests/activities (RRBI) than males, with small and variable effects depending on age, developmental level and kinds of RRBI. Differences insufficiently captured by standard measures may include autistic females displaying female-gender-typical narrow interests, higher social attention, linguistic abilities, motivation for friendship and more camouflaging than autistic males. Regarding developmental change, diagnosed young girls were more likely to have better cognitive development, less intense autistic symptoms and reduction of symptoms over time. Difficulties in adaptive functioning and social challenges, however, may emerge more for females in adolescence. Regarding diagnosis, general expectancy biases and gender-stereotypes may impede timely recognition of autism in females.

Summary

Appreciating the multilevel sex and gender impacts on presentation, development, and diagnosis is key to sex-equitable and gender-equitable care for autistic individuals. A holistic approach to understanding the person in the contexts of sex and gender is essential for timely and accurate diagnosis and support.

Keywords

autism, behaviour, development, diagnosis, gender, sex

INTRODUCTION

There is increasing awareness of how autism presents in nonmale populations. The latest meta-analysis shows that male:female prevalence ratio is 4.56:1 in epidemiological studies identifying cases passively (i.e. from existing medical/educational records), but 3.25:1 in studies actively identifying cases in the general population [1^{••}]. This supports the male-vulnerability of developing autism, but also underscores current under-recognition of autism in females—a predictive model constructed from population-based data estimated that ~39% more girls should be diagnosed [2]. Under-recognition is further supported by findings that, compared to males, autism tends to be diagnosed later in females [3] especially if the diagnosis is made after preschool-age [4]; given comparable autistic traits females are less likely to be diagnosed [5], or they require more behavioural, emotional, cognitive challenges, or even higher autistic traits to be

diagnosed with autism [6,7,8[•]]; autism in females is more likely overshadowed by other prior diagnoses (e.g. ADHD) [9[•]]; and given comparable behavioural presentation on the Autism Diagnostic Observation Schedule (ADOS), females had higher parent-reported autistic traits and adaptive dysfunction [10^{••}]. The under-recognition may stem from

^aThe Margaret and Wallace McCain Centre for Child, Youth & Family Mental Health and Campbell Family Mental Health Research Institute, Centre for Addiction and Mental Health, ^bDepartment of Psychiatry, The Hospital for Sick Children, ^cDepartment of Psychiatry, Faculty of Medicine, University of Toronto, Toronto, Canada, ^dAutism Research Centre, Department of Psychiatry, University of Cambridge, Cambridge, United Kingdom and ^eDepartment of Psychiatry, National Taiwan University Hospital and College of Medicine, Taipei, Taiwan

Correspondence to Meng-Chuan Lai, 80 Workman Way, Toronto, ON M6J 1H4, Canada. Tel: +1 416 5358501 ext. 34050; fax: +1 416 9794996; e-mail: mengchuan.lai@utoronto.ca

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KEY POINTS

- To improve the recognition of autism across sexes and genders, we need to appreciate the complexity of sex and gender impacts across nosology, behavioural presentation, developmental change, and context.
- On conventionally used standard measures, diagnosed autistic females overall show lower RRBI than autistic males, with small and variable effects depending on age, developmental level, and kinds of RRBI.
- Behavioural differences likely insufficiently captured by standard measures may include autistic females displaying female-gender-typical narrow interests, higher social attention, linguistic abilities, motivation for friendship, and more camouflaging than autistic males.
- Developmentally, young girls diagnosed with autism are more likely to have better cognitive development, less intense autistic symptoms, and reduction of symptoms over time than young boys with autism, yet adaptive dysfunction and social challenges may emerge more evidently for autistic females during adolescence.
- General expectancy biases and prevailing gender stereotypes in society may impede timely and accurate recognition of autism in females.

multiple sex-related and gender-related factors affecting behavioural presentation and development of the individual, and biases of the referral sources and diagnostic processes.

With increased awareness of the historically male-based lens on autism, the increasingly popular idea of a ‘female autism phenotype’ [11] may however risk giving an impression that the solution to better recognizing autism in females is simply to revise the diagnostic criteria and instruments to capture such phenotype. The solution is actually more complex—to improve the recognition of autism across sexes and genders, the nuances across nosology, behavioural presentation, developmental change, and contextual biases should all be appreciated.

Autism should be conceptually defined the same way across sexes and genders to provide the abstract-level consensus for diagnosis, based on social-communication-interaction challenges and restricted, repetitive patterns of behaviour, interests or activities (RRBI). This avoids creating ungrounded diagnostic distinctions by sex or gender without clear evidence supporting a sex-stratified or gender-stratified nosological departure from current conceptualization across neurodevelopmental and psychiatric conditions [11]. On this common ground, presentation and developmental differences by sex and gender should be clarified at the level of ‘narrow constructs’

(i.e. subdomains/constructs that are under or beyond the two cardinal domains) or ‘behavioural exemplars’ [11]. It is also important to consider sex and gender impacts on the environmental contexts surrounding the individual.

SEX AND GENDER IMPACT ON BEHAVIOURAL PRESENTATION

Narrow-construct differences

The literature on behavioural sex or gender differences in autism (most studies unfortunately did not differentiate ‘sex’ versus ‘gender’—in these scenarios we used ‘sex/gender’ to be unbiased) mostly used conventional, standard measures. At the narrow-construct level, small numbers of studies examined measurement invariance (i.e. the same construct is being measured across groups) or clinical cut-off across sexes/genders. So far, invariance has been demonstrated for selected Autism Diagnostic Interview-Revised (ADI-R) and Social Responsiveness Scale items [12], Social-Communication Questionnaire [13], Autism Spectrum Quotient-Short [14], Broad Autism Phenotype Questionnaire [15], and Autism Mental Status Examination [16]. However, these studies were done with individuals already clinically diagnosed (based on standard measures), therefore not giving the information regarding the measures’ psychometric properties when used with individuals who are under-recognized or display nontypical phenotypes (see section ‘Behavioural differences likely insufficiently captured by existing standard measures’), or outside the population referred for diagnosis.

Another narrow-construct level difference regards co-occurring health challenges. Conclusions cannot be reached yet, as some studies reported more internalizing symptoms in diagnosed autistic females and more externalizing symptoms in males [17–19] or more executive dysfunction in females than males [20], but others reported no clear differences [21,22]. A latest meta-analysis on psychiatric diagnoses in autism shows studies with more females tend to report more co-occurring depressive disorders [23].

Behavioural differences in standard measures and practice

Using conventional ‘gold-standard’ measures (e.g. ADI-R or ADOS), the most consistent finding is fewer RRBI in diagnosed females than males but comparable social-communication-interaction challenges, shown by meta-analysis of smaller studies [24] and recent large-scale studies with samples amalgamated across clinics or research cohorts in different countries [17,25,26,27*,28–30]. The effect sizes of the

RRBI differences are small, and there are several considerations for interpretation.

First, patterns of sex/gender differences in diagnosed individuals are dependent on age, language level, and intellectual level of the cohort. A lack of difference across social-communication-interaction, RRBI, cognitive, and adaptive functioning was found in minimally verbal individuals [31[¶]], toddlers/preschoolers diagnosed early in life [32–37], and children/youth in large clinical programs [38]. Lower RRBI in females is more likely found in young children without intellectual disability [39] and adults, including those diagnosed later in life [28]. Second, RRBI is a broad construct. Detailed observation shows that sex/gender differences may exist in fine-grained subdomains even when there are no differences on the overall RRBI score. Compared to autistic males, females tend to show reduced restricted, repetitive and stereotyped behaviours and interests, but more self-injurious and compulsive behaviours and sensory challenges [40,41]. These differences were especially evident when item-level data (instead of ‘diagnostic algorithm scores’) of the ADI-R or the Repetitive Behaviour Scale-Revised (RBS-R) were used to discriminate autistic females versus males [42[¶],43[¶]]. Finally, some behavioural exemplars of autism may not be well captured by conventional/standard measures (see section ‘Behavioural differences likely insufficiently captured by existing standard measures’), which contributes to the mixed picture.

Additionally, girls and boys might meet the diagnostic criteria in different ways. Diagnosed girls seem more likely to be able to engage in reciprocal conversations and share interests, to integrate non-verbal and verbal expressive behaviours (but not different from boys in understanding others’ non-verbal behaviours), to show better imagination, to initiate (but not maintain) friendships, and to display less stereotyped use of objects and less (and different contents of) narrow interests [44].

Behavioural differences likely insufficiently captured by existing standard measures

Recent interests in the ‘female autism phenotype’ [11] stem from autobiographical writings of lived experiences, clinical observations, and emerging qualitative research with autistic girls and women, particularly those diagnosed later in life [45]. This field is rapidly evolving to incorporate various research methods to capture potential behavioural differences beyond those conventionally measured. The reported differences may have sex-related biological bases, but more importantly, may also reflect influences of prevailing gender norms and gendered

social–cultural contexts on shaping the autistic person’s behaviour and development [46].

Autistic girls and women may not have ‘typically autistic’ contents of narrow interests (e.g. transportation, numbers, dinosaur, technology, scientific facts, and so on). Narrow interests in some autistic girls and women may involve topics related to people and animals rather than objects and things [11] and are more likely seen by parents as ‘seemingly random’ topics or objects (e.g. animals, rocks, stickers, pens) [44]. If the intensity, exclusivity, and functional impact of their interests were ignored/shadowed by the seemingly gender-typical contents, the presence of autistic narrow interests could be overlooked. This may be an important reason why measured restricted interests on conventional instruments were lower in females.

Autistic girls may allocate their social and non-social attention differently from autistic boys, in a way mirroring normative sex/gender differences. Eye-tracking studies show autistic girls have gender-typical visual attention in circumscribed interests similar to typically developing girls, and different from autistic and typically developing boys [47]. Autistic girls also show more social attention than autistic boys, close to typically developing boys, and approaching typically developing girls [48[¶]].

Autistic girls may show higher linguistic abilities than autistic boys (e.g. telling richer stories, more descriptors of planning or intention, more internal state language, more typical-sounding disfluency patterns) [49–51], again mirroring normative sex/gender differences and putting them closer to typically developing peers, and away from autistic boys.

Autistic girls tend to show social motivation and interests in friendships more so than autistic boys [52[¶]]. In school-age, autistic boys are more overtly socially excluded than autistic girls, who tend to be overlooked rather than rejected [53]. In adolescence, autistic girls reported higher friendship quality than autistic boys, showing a level approaching typically developing girls [52[¶],54[¶]]. They viewed friendship as desired [52[¶]], important and rewarding [55], but difficult (e.g. ‘friends are hard work’) [52[¶]]. This suggests that gender and gendered contexts play critical roles in autistic individuals’ social relational experiences.

Finally, ‘camouflaging’ may be common in autistic females. Camouflaging refers to compensating for inherent social-communication difficulties or masking autistic behavioural appearances [56[¶]]. Increased attention has been paid to camouflaging as a part of the behavioural presentation of some autistic individuals, and its impact on diagnostic accuracy [11], especially considering the under-recognition of autism in females [46]. Qualitative/mixed-methods research has described, primarily in autistic adults, the

presence of camouflaging; the reasons to camouflage (e.g. to function in school/workplace, to make friends); the strategies used; the mental health impacts (e.g. exhaustion, strain, anxiety, depression and suicidal risk, and even a loss of identity); the higher frequency/level of camouflaging in women; and the association with under-recognition or later-diagnosis of autism in females [45,56[■],57,58,59[■]]. Camouflaging has been quantified in adults via a ‘discrepancy’ approach (i.e. the difference between intrinsic autistic status and extrinsic social behaviour) [60[■],61,62] and a ‘reflection’ approach (i.e. self-reflected/reported intention to camouflage) [63], often with higher levels found in autistic women than men [60[■],62,64]. Neuro-cognitively, camouflaging correlates with ventral medial prefrontal cortex neural activation during self-referential cognition in autistic women but not autistic men [65]. Research on camouflaging is still in its infancy—the measurement, multifaceted cognitive and neural mechanisms, developmental trajectories, mental health impacts, and relations with social coping in autistic people need to be further studied.

SEX AND GENDER IMPACT ON DEVELOPMENTAL CHANGE AND OUTCOME

Follow-up studies of ‘high-risk’ infants who have older siblings with autism show early sex/gender differences at 18–36 months. Across low-risk typically developing, high-risk nonautism, and high-risk autism (diagnosed at 3 years old) groups, males had lower developmental functioning (on Mullen Scales of Early Learning) and higher ADOS RRBI scores than females [35]. Sex/gender differences in young autistic children again mirror normative sex/gender differences seen in young children without autism. In an inception cohort with autism diagnosed by age 2–4 years and followed up to 6 years-old, grouped based on developmental trajectories, boys were more likely to be in the group with more intense ADOS autistic symptoms and a stable trajectory, whereas girls are more likely to be in the group with less intense autistic symptoms and with more rapid reduction over time [66]. This may indicate higher likelihood of attenuating autistic symptoms over time in preschool autistic girls than boys.

Cross-sectional data of children aged 7–13 years indicate that despite younger autistic girls showed better adaptive functioning than autistic boys, the opposite was found in older children [67]. Furthermore, developmental trajectories of autistic social traits in the general population were nonlinear (a decline in age 7–10 years, followed by an increase in age 10–16 years), but the increase in females was

steeper, so that females were more likely than males to experience an escalation of autistic social traits during adolescence [68[■]]. Together, these initial data lead to the hypothesis that perhaps some females with diagnosed autism, or mild autistic characteristics, may encounter increasing adaptation challenges and/or social difficulties when they enter late childhood and adolescence, even more so than males. Late childhood and adolescence are developmental periods with increasingly complex social demands and pubertal biological changes. The findings may imply an ‘adolescent-onset’ need for autism diagnosis for some individuals, particularly females, explaining why male:female ratio in referral for autism assessment significantly drops to ~2:1 in teenage and adulthood from the childhood ratio of 5.5:1 [69]. This also supports the importance of adequate health and social support for autistic children and youth, especially females, when they enter adolescence and transition into emerging adulthood.

SEX AND GENDER IMPACT ON RECOGNITION AND DIAGNOSIS OF AUTISM

The under-recognition of autism in females can also result from the way in which behavioural presentations are interpreted by key supporters (e.g. caregivers, teachers, friends) and clinicians screening or diagnosing autism. Recognition and measurement errors in these contexts can originate from general expectancy biases (i.e. because autism is more prevalent in males, associated characteristics are less likely to be interpreted as signposting autism when shown in females) and prevailing gender stereotypes in society. For instance, social withdrawal or passivity may be more likely interpreted as ‘shyness’ in girls but ‘unresponsiveness’ in boys, the latter being more readily considered a red-flag for autism. Anxiety may be normalized more in females, given the higher general population rates in females and potential gender stereotypes (e.g. ‘women are more emotional’), preventing further in-depth investigation of the nature of anxiety, which may in fact be highly relevant to autism (e.g. social overload, fear for uncertainty, resistance to change, sensory challenges).

In preschoolers with autism, prediagnosis concerns of teachers and parents were generally less for girls than boys [44,70,71]; further, parents were more likely to name autism as a concern for boys than girls [72]. In school-age likely-autistic children, co-occurring mental health problems were rated less impairing in girls than in boys [18]. On the playground, gendered contexts could reduce the chance for social difficulties to be recognized: compared to autistic boys, autistic girls were more likely to stay in

close proximity to peers and to weave in and out of activities, so they were less likely to be identified as struggling socially—from a distance they may ‘look like’ neurotypical girls [73[■]].

Empirical research into recognition biases because of gender stereotypes is emerging [74]. Ways to reduce general expectancy biases and gender stereotype-based biases still need to be developed. Longitudinal studies of infants ‘at-risk’ of developing autism reported a male:female ratio of 3.18:1 [35], comparable to the ratio from actively ascertained population data (3.25:1) rather than the male-enriched clinical data (4.56:1) [1[■]]. This suggests that at least for children with older autistic siblings, close follow-up from birth using standard measures may reduce under-recognition of autism in females. Strategies to improve the recognition in other groups of nonmale individuals (e.g. without family history, presenting clinical challenges in older ages) are suggested below.

CONCLUSION: CLINICAL IMPLICATIONS AND FUTURE DIRECTIONS

To achieve sex and gender equitable recognition of autism, one key principle is to appreciate the multi-level sex and, even more importantly, gender impacts on the behavioural presentation (e.g. narrow interests aligned with social and gendered norms) and developmental trajectories (e.g. increasing social challenges in adolescence) of autism. Another key principle is to be aware how general expectancy biases and gender-stereotyped assumptions influence awareness, assessment, interpretation and formulation of autism differently by sex and gender. In short, clinical assessment should aim at (1) recognizing behavioural exemplars of and narrow constructs closely associated with autism that are influenced by sex and gender, and (2) accounting for and minimizing general expectancy biases and gender stereotypes held by informants and diagnosticians. In-depth understanding of the individual’s behaviour across contexts and from multiple sources, instead of solely relying on a cross-sectional assessment and score-thresholds on the ‘gold-standard’ measures, is key to achieving sex- and gender-sensitive clinical recognition of autism [75[■]]. Furthermore, it is crucial to understand the subjective experiences and insights of the individuals themselves (taking into account their self-reflection and communication capacity and development of self-concept). The field of diagnosing autism has historically been heavily based on caregiver report and behaviour observation, which are necessary but not sufficient, and may miss critical information of subtle signs of autism that are more likely to present in females, especially those who are older and not intellectually disabled. Possibilities of

camouflaging should be assessed, and one should avoid quickly ‘ruling out’ autism simply based on the presence of well modulated eye contact, gestures, language fluency, social motivation, friendship or romantic relationship, affective empathy, or imagination [76]. A holistic approach with good awareness of sex and gender impacts is essential.

In addition, our current understanding of autism is not only male-biased but at best sex-based, with insufficient focus on the impacts of gender and understanding of gender diversity. With increasing acknowledgement of heightened gender diversity and sexual identity diversity in autistic individuals [77,78], in-depth understanding of the clinical implications of gender and sexual diversity in autism is needed [79]. Learning from lived experiences of non-male autistic individuals and their families to translate into measurement development will be a useful next step. With improved knowledge, sex- and gender-informed support for autistic people across the life-span will be essential to optimize care and support.

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Conflicts of interest

There are no conflicts of interest.

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