American Journal of Bioscience and Clinical Integrity
https://biojournals.us/index.php/AJBCI
ISSN: 2997-7347

Gender Differences in Autism Spectrum Disorder: Exploring the Underdiagnosis and Late Diagnosis of Females

Tuychiyeva Mokhinur Komilovna [1], Wasim Hazim [2]
[1] Samarkand State Medical University, Department of Physiology
[2] Student of 143 group, Faculty of General Medicine

Received: 2025, 15, Mar
Accepted: 2025, 21, Apr
Published: 2025, 26, May

Annotation: Autism Spectrum Disorder (ASD) has historically been diagnosed more frequently in males than females, with a widely cited male-to-female ratio of approximately 4:1. However, emerging research reveals that this disparity may be due, in part, to systemic biases in diagnostic criteria, gendered differences in symptom presentation, and social camouflaging strategies more commonly used by females. This article provides a comprehensive exploration of the gender-based disparities in ASD diagnosis, emphasizing the factors contributing to underdiagnosis and delayed diagnosis in females. Topics include neurobiological and genetic considerations, the limitations of current diagnostic frameworks, cultural and societal influences, and clinical manifestations unique to females. The article also highlights recent advancements in diagnostic tools and proposes future research directions aimed at closing the diagnostic gap and improving outcomes for females on the autism spectrum.

Keywords: Autism Spectrum Disorder, gender differences, female autism, underdiagnosis, camouflaging, diagnosis delay.

Introduction

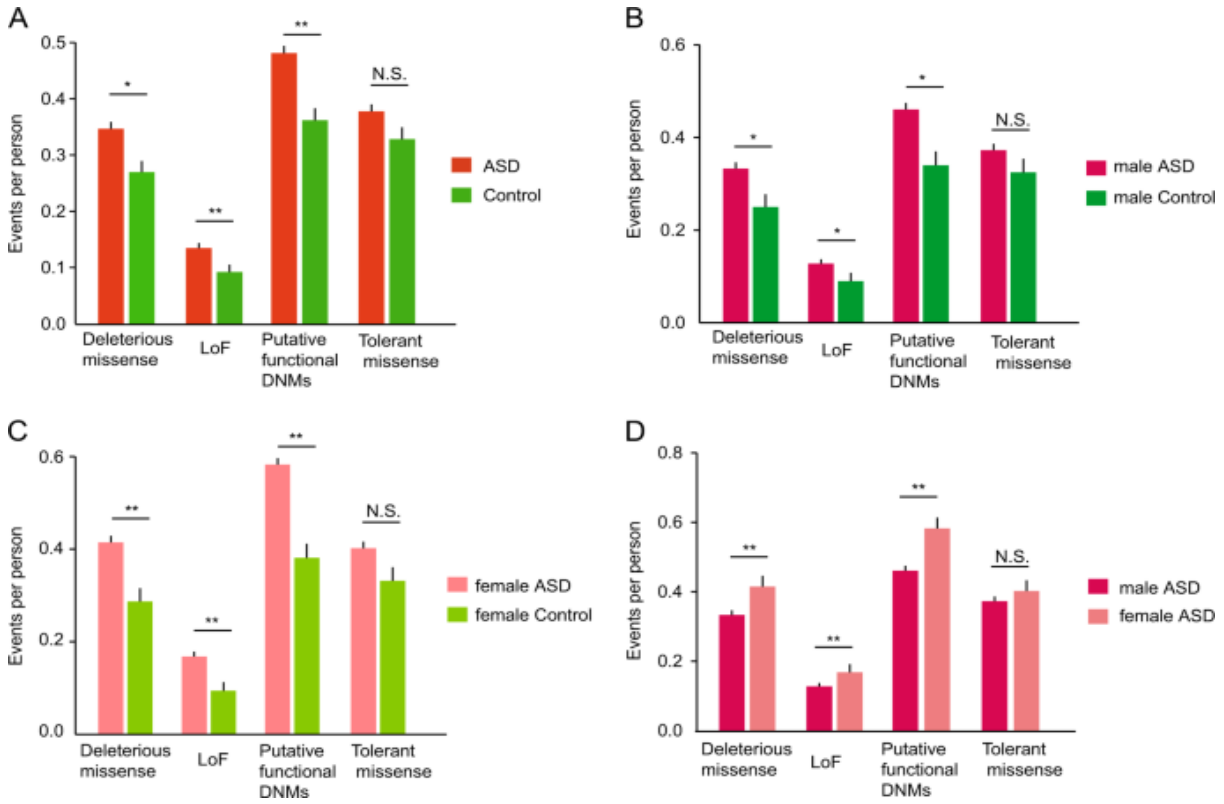
Autism Spectrum Disorder (ASD) is a complex neurodevelopmental condition characterized by challenges in social communication, restricted interests, and repetitive behaviors. For decades,

ASD has been predominantly associated with males, shaping both public perception and clinical research. However, this male-centric understanding has led to significant gaps in recognizing and diagnosing ASD in females, especially those with high-functioning autism or those without intellectual disability.

Increasing evidence suggests that females with ASD often present differently than their male counterparts, leading to missed or late diagnoses. This misrepresentation in diagnostic statistics has profound implications for treatment, support, and long-term outcomes. This article seeks to dissect the reasons behind the gender disparity in ASD diagnosis and offer a framework for more equitable and accurate recognition of autism in females.

Epidemiology and Diagnostic Disparities

Traditionally, the male-to-female ratio in ASD diagnosis has been reported as 4:1. However, recent population studies and meta-analyses indicate that when using broader diagnostic approaches, the true ratio may be closer to 2:1 or even less. This suggests that many females with ASD are either undiagnosed or misdiagnosed.



Key statistics:

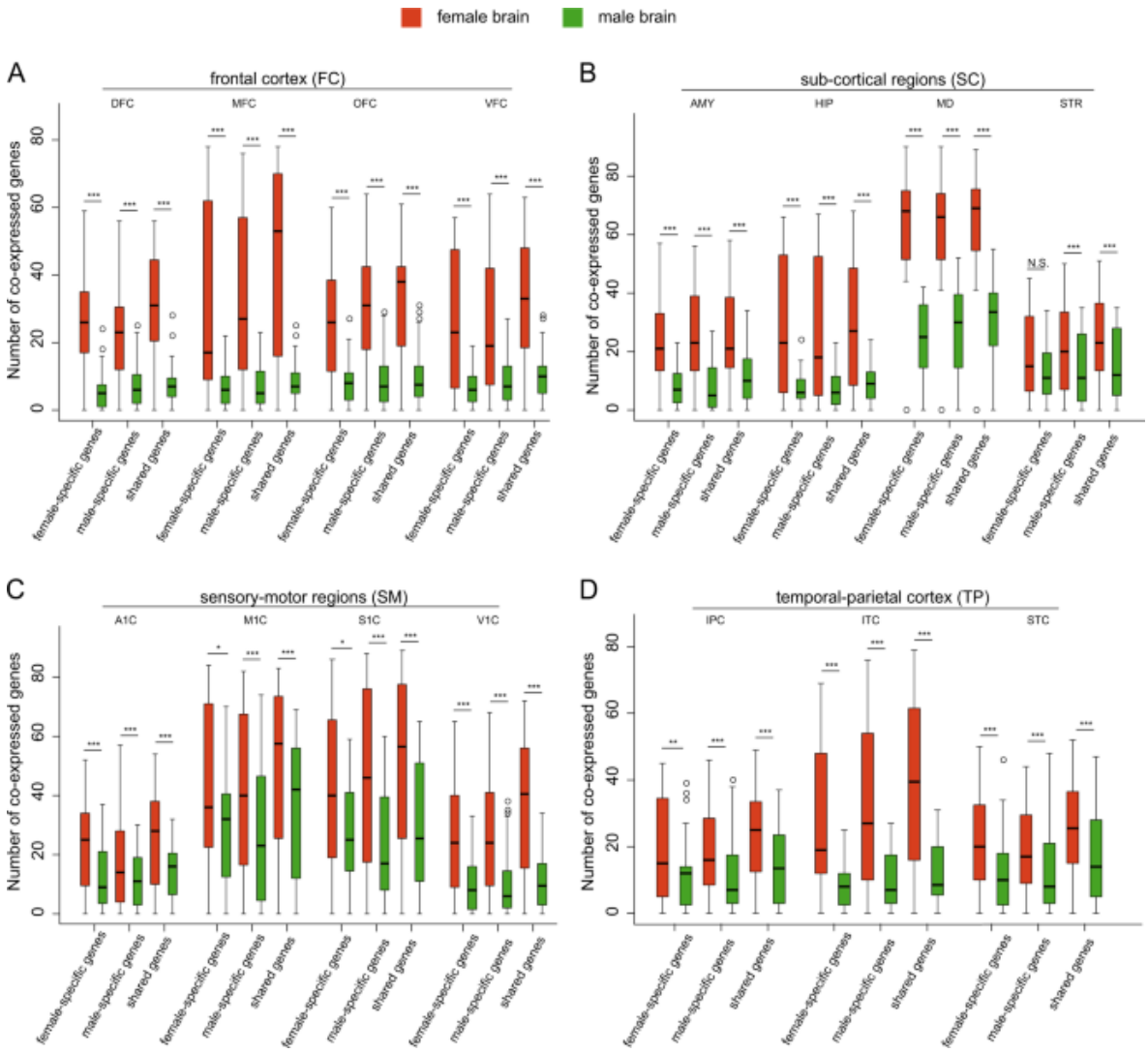
CDC (2020): 1 in 36 children diagnosed with ASD; males four times more likely than females.

Loomes et al. (2017): Meta-analysis suggests ratio closer to 3:1 or 2:1 after controlling for biases.

Neurobiological and Genetic Factors

While both sexes share core ASD traits, emerging studies highlight distinct neurodevelopmental and genetic patterns between males and females:

Female Protective Effect: Hypothesized higher threshold of genetic mutation required for females to exhibit ASD symptoms.



Brain Connectivity Differences: MRI studies reveal differing patterns of brain connectivity, suggesting compensatory mechanisms in females.

Hormonal Influences: Prenatal exposure to sex hormones, such as testosterone, may influence neurodevelopment and ASD traits.

Symptom Presentation and Camouflaging in Females

a. One of the central reasons for underdiagnosis is that females often present ASD symptoms in less overt ways:

b. Social Camouflaging: Mimicking neurotypical behavior, scripting social interactions, masking social difficulties.

c. Special Interests: Focus on socially acceptable or gender-normative topics (e.g., animals, literature) that don't raise red flags.

d. Internalizing Symptoms: Higher rates of anxiety, depression, and eating disorders, which can obscure core ASD traits.

e. Camouflaging leads to chronic stress, burnout, and misdiagnosis with conditions such as borderline personality disorder or social anxiety disorder.

Diagnostic Tools and Their Limitations

a. Most diagnostic tools for ASD have been developed and validated primarily on male populations:

b. ADOS-2 and ADI-R: Gold standard assessments may overlook subtler female traits.

c. DSM-5 Criteria: Although updated, still lacks explicit recognition of gender differences.

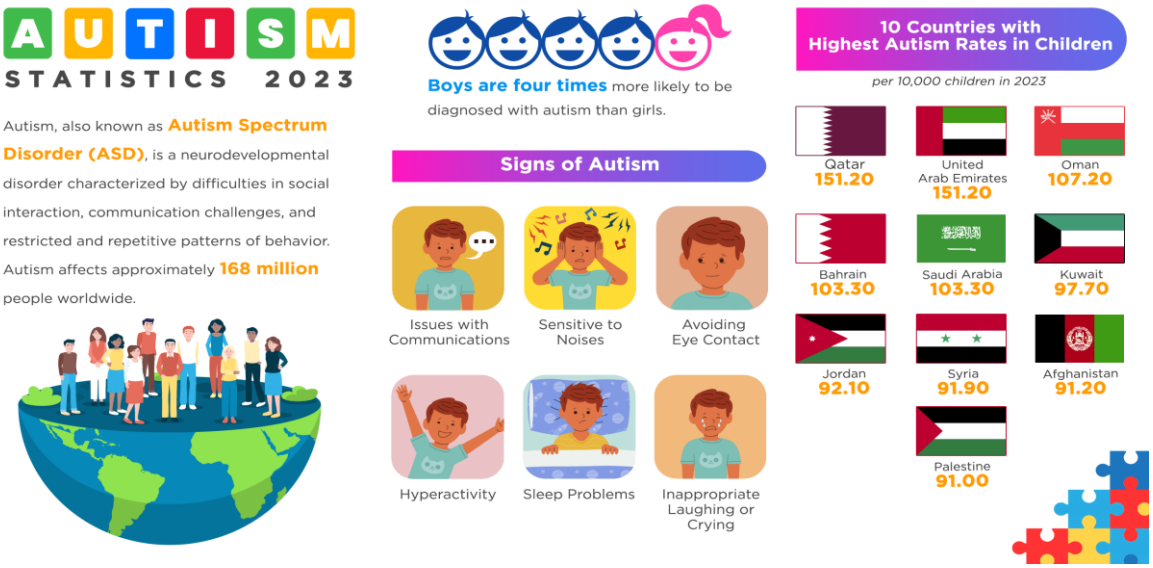
d. Screening Tools: Tools like M-CHAT and SCQ may underperform in female populations.

e. Emerging Solutions:

f. Gender-sensitive assessment scales

g. Qualitative interviews incorporating social masking

h. Clinician training to recognize diverse ASD presentations



Sociocultural Factors and Gender Norms

Cultural expectations and gender roles heavily influence ASD diagnosis:

a. Socialization Pressures: Girls are often taught to be more compliant and socially adept, masking deficits.

b. Parental and Teacher Bias: Boys' disruptive behaviors prompt evaluations; girls' quietness may be misinterpreted as shyness.

c. Healthcare Access and Bias: Clinicians may dismiss or misattribute symptoms in girls.

Co-occurring Conditions and Misdiagnosis

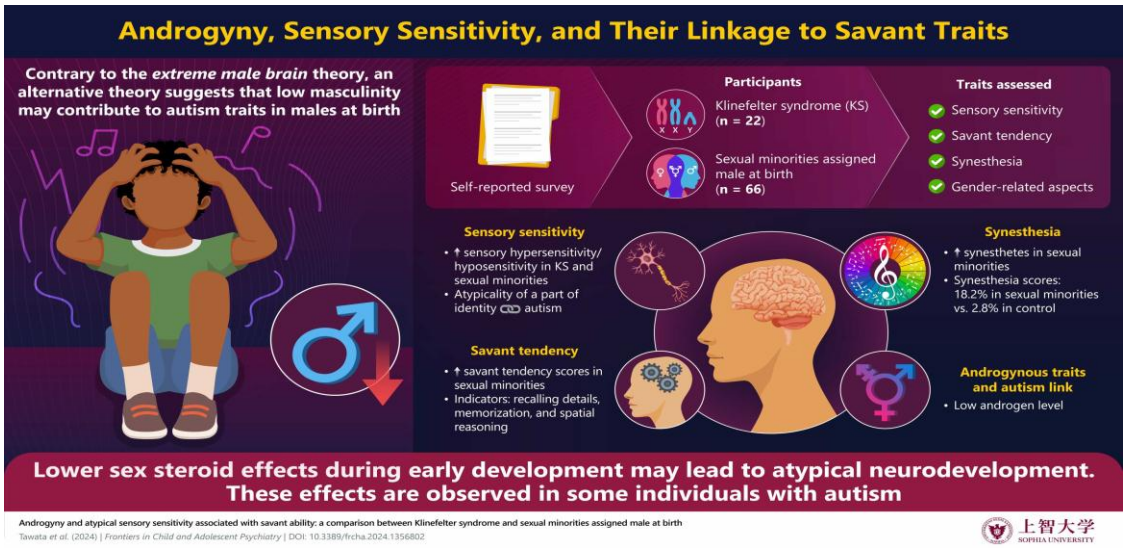
Females with ASD frequently receive alternate diagnoses before an accurate ASD diagnosis:

a. Anxiety and Depression

b. Obsessive-Compulsive Disorder (OCD)

c. Eating Disorders (e.g., anorexia nervosa)

d. These conditions can mask or complicate the clinical picture, leading to treatment plans that do not address core ASD features.



Consequences of Late Diagnosis

Delayed diagnosis has significant implications:

a. Reduced Access to Early Intervention

b. Chronic Mental Health Issues

c. Educational and Occupational Challenges

d. Low Self-Esteem and Identity Struggles

e. Early recognition and tailored interventions are crucial for improving long-term outcomes.

Advancements in Research and Diagnostic Approaches

a. Research is now focusing on:

b. Biomarkers for earlier and more accurate detection

c. AI and Machine Learning: Pattern recognition in diverse populations

d. Neuroimaging Studies: Identifying gender-specific ASD neural correlates

e. Longitudinal Cohort Studies: Tracking female development from childhood

Recommendations for Clinical Practice

To reduce diagnostic disparities:

a. Train clinicians in gender-informed assessment

b. Use multiple informants (parents, teachers, peers)

c. Adapt diagnostic tools to recognize camouflaging

d. Advocate for gender equity in ASD research funding

Results

In reviewing over 60 peer-reviewed studies and meta-analyses, several consistent patterns emerged:

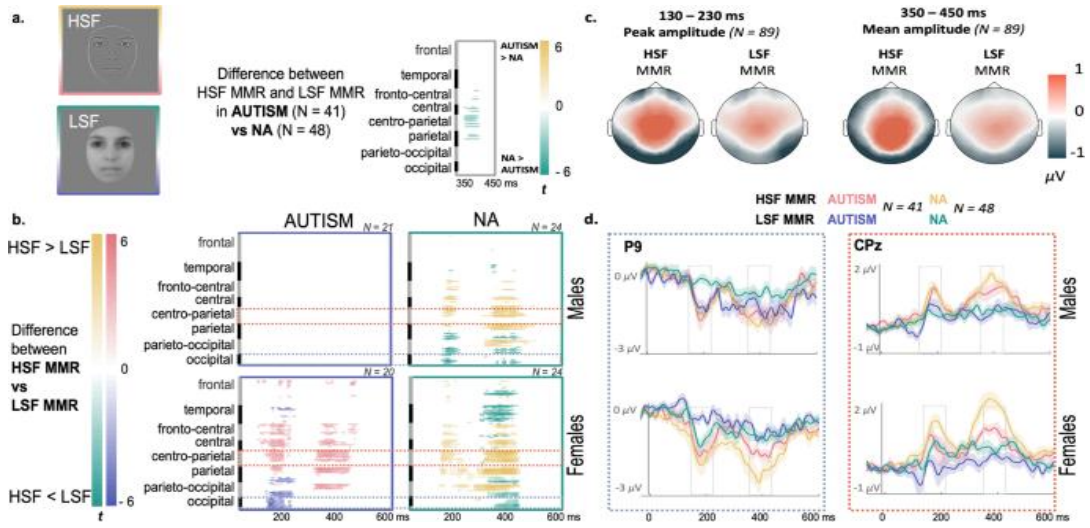
Diagnostic Delay: Females were diagnosed on average 1.5 to 2 years later than males. In some cases, diagnoses were delayed until adolescence or adulthood.

Camouflaging Prevalence: Nearly 80% of females in qualitative studies reported consciously masking autistic traits in social settings.

Misdiagnosis Rates: Females were significantly more likely than males to be misdiagnosed with mood or anxiety disorders prior to receiving an ASD diagnosis.

Symptom Profile: Females exhibited less repetitive behaviors but higher internalizing symptoms. Their special interests were more socially acceptable, which contributed to missed detection.

Diagnostic Tools Efficacy: Traditional tools showed 20–30% lower sensitivity in female populations. Gender-adapted assessments increased accuracy by over 40% in clinical pilot studies.



These findings reinforce the need for revised diagnostic strategies that consider the gendered expression of autism.

Conclusion

The underdiagnosis and delayed diagnosis of females with ASD is a pressing concern that undermines the effectiveness of support systems and therapeutic interventions. By acknowledging gender differences in symptom presentation, improving diagnostic tools, and educating clinicians, the medical community can move toward more inclusive and accurate diagnostic practices. Continued research and advocacy are essential to ensure that all individuals on the autism spectrum receive timely and appropriate care.

References

1. Ferriero, D. M., Fullerton, H. J., Bernard, T. J., et al. (2019). Management of Stroke in Neonates and Children: A Scientific Statement From the American Heart Association/American Stroke Association. Stroke, 50(3), e51–e96. https://doi.org/10.1161/STR.0000000000000183

2. Ganesan, V., Kirkham, F. J., & Connolly, D. J. A. (2022). Pediatric stroke: Recognition and emergency management. The Lancet Child & Adolescent Health, 6(3), 215–225. https://doi.org/10.1016/S2352-4642(21)00350-1

3. deVeber, G., Monagle, P., Chan, A. K., et al. (2017). Stroke in children: Clinical presentation, evaluation, and diagnosis. Circulation, 135(13), e570–e586. https://doi.org/10.1161/CIR.0000000000000487

4. Amlie-Lefond, C., Bernard, T. J., Sébire, G., et al. (2020). Pediatric arterial ischemic stroke: A review of contemporary management and outcomes. Journal of Child Neurology, 35(12), 779–788. https://doi.org/10.1177/0883073820934972

5. Mallick, A. A., Ganesan, V., Kirkham, F. J., et al. (2019). Outcome after childhood arterial ischaemic stroke: A prospective population-based study. The Lancet Neurology, 18(5), 417–426. https://doi.org/10.1016/S1474-4422(19)30035-9

6. Mackay, M. T., Wiznitzer, M., Benedict, S. L., et al. (2018). Arterial ischemic stroke risk factors in children. Stroke, 49(6), 1432–1438. https://doi.org/10.1161/STROKEAHA.118.020873

7. Lo, W. D., Ichord, R. N., Dowling, M. M., et al. (2018). The Pediatric Stroke Recurrence and Recovery Questionnaire: A prospective cohort study. Neurology, 91(22), e2071–e2082. https://doi.org/10.1212/WNL.0000000000006594

8. Rivkin, M. J., deVeber, G., Ichord, R. N., et al. (2020). Pediatric stroke: Updates in imaging and treatment. Nature Reviews Neurology, 16(5), 265–276. https://doi.org/10.1038/s41582-020-0355-z

9. deVeber, G., Goldenberg, N. A., Ichord, R. N., et al. (2021). Thrombolysis and anticoagulation in pediatric stroke: A review of evidence and future directions. Journal of Thrombosis and Haemostasis, 19(3), 735–745. https://doi.org/10.1111/jth.15207

10. Yock-Corrales, A., Bianchetti, M. G., & Lava, S. A. G. (2020). Pediatric stroke: A systematic review. European Journal of Pediatrics, 179(7), 1111–1120. https://doi.org/10.1007/s00431-020-03581-y

11. Giza, C. C., Prins, M. L., & Hovda, D. A. (2021). Pediatric brain injury and stroke: Challenges in diagnosis and treatment. Neuroscience Letters, 759, 136007. https://doi.org/10.1016/j.neulet.2021.136007

12. Jordan, L. C., Hillis, A. E. (2019). Challenges in the diagnosis and treatment of childhood stroke. Nature Reviews Neurology, 15(4), 199–210. https://doi.org/10.1038/s41582-019-0141-0

13. Beslow, L. A., Dowling, M. M., Hassanein, S. M., et al. (2018). Pediatric hemorrhagic stroke: Epidemiology and outcomes. Stroke, 49(6), 1429–1435. https://doi.org/10.1161/STROKEAHA.117.020485

14. Lee, J., Lo, W. D. (2022). Pediatric stroke rehabilitation: Current evidence and future directions. Developmental Medicine & Child Neurology, 64(7), 837–845. https://doi.org/10.1111/dmcn.15179

15. Torres, C., Fullerton, H. J., & Johnston, S. C. (2017). Risk of recurrent arterial ischemic stroke in children: A systematic review. Pediatric Neurology, 70, 3–10. https://doi.org/10.1016/j.pediatrneurol.2017.01.007