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Review article

Clinical characteristics and problems diagnosing autism spectrum disorder in girls

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ABSTRACT

Background: Autism is a neurodevelopmental disorder with various clinical presentations. It has been historically considered a male disorder. An increasing number of authors stress the existence of sex/gender bias in prevalence and the need to define sex/gender differences in the clinical presentation.
Review: Recently, an increasing number of authors have studied the impact of sex/gender on autism's clinical presentation. The sex ratio of four boys to one girl commonly reported in literature is questioned. Sociocultural and familial influences can impact female clinical presentation as well as the way the difficulties of girls with autism are perceived. Issues of autism diagnostic instruments such as sex/gender bias are also studied since they have an impact on the access to diagnosis for girls. Clinical variability is a part of autism spectrum disorder, but some traits appear to be more specific of the female phenotype: existence of a "camouflage" phenomenon and less unusual play or restricted interests.
Discussion: Better understanding and diagnosis of females with autism is required to ensure the access to the support and treatment they need. Professionals must apprehend the sex/gender clinical differences to prevent the frequent misdiagnosis or missed diagnosis of females with autism.
Conclusion: Pursuing research on sex/gender differences seems necessary to ensure appropriate support and diagnosis of undiagnosed females.

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1. Background

Autism, or autism spectrum disorder (ASD), make up a heterogeneous group of neurodevelopmental disorders. ASD is characterized by deficits in socialization and communication as well as restricted interests and repetitive or stereotyped behaviors beginning in infancy or the toddler years (before 3 years of age). Since the initial case series described by Kanner (1943) and Asperger (1944), it has always been considered as predominantly present in males. There are few studies exploring the relationship between gender and ASD. They tend to explore various themes such as genetics, hormonology [1] (testosterone for example), the "female protective effect" (girls should need a greater etiologic load to manifest autistic behavioral impairments [2]), as well as female prevalence of ASD and sensitivity to the female phenotype in the assessment tools [3].

Few studies explore phenotypic differences of ASD between males and females, and the findings are not always related in a straightforward manner. ASD is known to have an impact on socioprofessional insertion and quality of life. An increasing number of studies suggest a delayed diagnosis in females with ASDs. A delay in care for ASD patients is known to be related to more developmental difficulties. Therefore, it appears necessary to study the female phenotype of ASD [4–7].

2. Methods

We searched PubMed for all articles published until August 2017 using search terms "sex OR gender OR females AND autism." Then we screened all articles for relevance along with publications identified via additional literature reviews. A total of 170 articles on ASD symptomatology and diagnosis of girls and boys were extensively reviewed.

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3. Diagnosis problems

3.1. ASD and gender

Most of the research on ASD has been conducted with predominantly male samples. Our current understanding of the pathogenesis and clinical presentation of ASD is therefore about males with autism. Most studies use sex as a variable to control and not as a specific independent aspect of ASD that needs to be studied [8]. General population studies show that male and female brains are distinctive in their way of functioning: distinctive neurological physiology as well as distinctive cognitive profiles [9]. Despite these differences, to reduce experimental variability, studies on autistic behaviors mostly include males [8].

3.2. ASD prevalence

Estimated prevalence of ASD is 1% and about 1 in 68 [10]. The mean male:female ratio most frequently found in the literature is 4:1 [10]. This ratio is not the same within the spectrum. In Asperger autism, the sex ratio the most commonly encountered is 8–14:1. When cognitive impairment is taken into account, the sex ratio usually reported is lower: 2:1 (IQ < 70) to 1.3:1 (IQ < 50), even if some studies show no association between cognitive impairment and sex [11]. Two epidemiological studies [11,12] conducted in the general population found some very different sex ratios. These results were obtained by a two-step screening: use of the Autism Spectrum Screening Questionnaire (ASSQ) first and then both the Autism Diagnostic Interview Revised (ADI-R) and the Autism Diagnostic Observation Schedule (ADOS). This screening allowed Mattila et al. [11] to study the ASD prevalence in a population of 5484 8-year-old Finnish children. They found a male:female ratio of 1.8:1 for ASD and 1.7:1 for high-functioning ASD. Kim et al. [12] found a very similar male:female ratio of 2.5:1 when studying the ASD prevalence in 55,266 7- to 12-year-old South Korean children.

Whatever the pathology in which a gender is known to be predominant, there is a need to mention the possible existence of bias in the diagnosis or in the sampling [13]. Thompson et al. [14] have shown the existence of sampling bias while analyzing qualitatively 392 articles on autism. In these studies, 80% of the patients were males, 5% of the studies (20 studies) analyzed variables considering gender/sex, and three studied the differences in male and female intellectual quotients. Moreover, the vast majority of samples were extracted from health clinics.

Our current understanding of ASD is based on male-centered research [14]. The results presenting a different sex ratio than the classical 4:1 male:female ratio may show that some recent studies might have identified women with ASD more efficiently, particularly in the absence of intellectual disability [7]. Therefore, it seems essential to understand the various factors influencing the diagnosis of ASD in females [7,8].

3.3. Sociocultural influences on ASD diagnosis

Until recently, the influence of sociocultural factors on genetics has not been widely studied, whereas they can have an impact on phenotypic variations between males and females [6]. Some authors suggest that familial and sociocultural factors have an influence on the clinical presentation and on the diagnosis of females with ASD, in particular when there is no intellectual disability (IQ > 70). ASD symptoms in females could then be less clearly recognized because of their differences with so-called typical ASD [3,6,15].

The diagnosis of ASD is based on the evaluation by healthcare professionals, using the ADI-R, which is based on an interview with both parents, and on the ADOS, which is based on observation.

It appears that the diagnosis of ASD depends on different factors: parents'perceived difficulties in their child, the way those difficulties are understood by healthcare professionals, symptoms exhibited by the patient, and finally the initial orientation toward a specialized consultation. This initial orientation can be made by parents, teachers, and healthcare professionals. The diagnosis and more importantly, the care of females with ASD depend on the representations of each actor of this initial orientation of how a girl with ASD should behave.

3.3.1. Parents

Parents' expectations concerning girls and boys commonly differ. Accordingly, they will not have the same education and will act differently. For example, the way a mother talks to her daughter with a large number of emotional references reinforces social abilities and empathy [6]. Games and play are widely influenced by parents: a girl is going to be encouraged to play girls' games (such as dolls) and cooperative pretense play [16]. Parents of ASD children expect from girls more social behaviors than from boys. Therefore, the clinical presentation of girls could be influenced by parenting education (pretense play, games with peers, empathy, social interactions). Moreover, this could have an influence on seeking healthcare (lack of social interactions considered as shyness).

3.3.2. Teachers

School is frequently responsible for the first orientation to specialized consultation as social difficulties are mostly first identified at school. Therefore, teachers are key actors of the diagnosis of ASD.

During school, the subtlety of girls' difficulties makes them less recognized [17,18]. Girls with ASD show fewer problematic behaviors than boys (ADHD, hostility) [19,20]. During class, they behave more discreetly, which is considered shyness [9,21], and therefore do not draw attention.

Two studies [22,23] investigating ASD children's behaviors on the playground highlighted a biased representation on the part of teachers. Teachers expect the same behaviors from girls as boys. Whereas boys with ASD mostly stay alone during recess, girls often have friends of the same age to take care of them. Therefore, they are not as alone as boys on the playground and social deficits can be masked. The camouflaging of symptoms often seen in girls with ASD, by using the example of their peers in games and social interactions, makes their recognition all the more difficult [23].

3.3.3. Doctors

Girls do not always show behaviors identified as typical of ASD in the general population [3]. This might alter the health trajectory as well as the capacity of the clinician to diagnose ASD. When a disorder is considered as occurring more often in one sex than the other, a risk of diagnosis bias exists [13], maintained by today's state of knowledge under-riding medical studies.

It has been shown that three factors are related to the precocity of diagnosis: being male, having an intellectual disability (IQ < 70) and the existence of a developmental regression [24]. Various studies have highlighted that in the absence of intellectual disability or of problematic behaviors, girls with ASD are less likely than boys to be diagnosed, even in the presence of the same level of difficulties [5,9,25,26]. Moreover, diagnosis is often delayed [4,25]. Some authors add that doctors would be more likely to exclude the diagnosis of ASD in the presence of another pathology, particularly intellectual disability [25]. Intellectual disability is then considered as the only diagnosis, even though ASD symptoms are present. In this situation ASD behaviors are considered as being a consequence of intellectual disability and not a comorbidity. This kind of logic is not found for boys [5]. Therefore,

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