The female phenotype:

what do expert clinicians think?

By JULIE CUMIN

The issue of autism in women has elicited much clinical and research attention in the past few years. As early as the 90s, researcher-clinicians wondered whether very low rates of autism without intellectual disability or speech-onset delay (formerly Asperger's Syndrome) in women could perhaps be due to a "female version" of autism, more likely to go undetected. Indeed, autistic women may have better imitation skills, allowing them to hide some of their difficulties and remain undiagnosed (camouflaging). Many studies have described the lived experience of women diagnosed later in life (post-adolescence) as well as the consequences of this **underdiagnosis**: missed opportunities for accommodations at school and at work, bullying, abuse, and mental health issues.

30 years on, this debate persists. Is autism in women a truly different manifestation of autism, requiring its own diagnostic tools and criteria, or does it simply reflect differences between men and women found in the general population? A 2017 meta-analysis concluded that, "individuals with Autism Spectrum Conditions are fundamentally similar to typically developing individuals in regard to their sex/gender variation in core autism characteristics" (Hull et al, 2017). However, the "female phenotype" hypothesis has largely been accepted as a fact by the media and much of the autistic community, particularly online (forums, social media). With autism being a less stigmatized and more desirable diagnosis than some psychiatric conditions, some clinicians fear a situation in which **overdiagnosis** would also become possible. This twofold issue (under and over diagnosis) is problematic in that it could prevent access to appropriate care and slow research efforts, which rely on accurate diagnosis.

These are some of the issues addressed by a new study published in *Autism*. The researchers aimed to better understand how clinicians made **differential diagnoses** in adult women and which factors complicated the process.

In the absence of clear diagnostic criteria, the research team decided to source the opinions of **expert clinicians** to better understand how they detected autism in adult women.

The researchers chose the Delphi method, often used to collect the opinions of medical experts and create clinical guidelines over several rounds of revisions. The Delphi method allows for an initial exploration of different themes of interest during interviews, then the creation of clinical guidelines which the same experts revise until they reach a consensus.

The researchers noticed a remarkable level of agreement between the 20 clinicians despite practices located in 7 different countries. The complete list of clinical guidelines having obtained consensus can be found in the original paper. The list is comprised of 37 statements based on clinical observations, two of which are presented below:

Differential diagnosis
Process during which a clinician develops several diagnostic hypotheses which could explain the observed symptoms and eliminates them one by one to make the correct diagnosis.

Expert clinician
The accuracy of a diagnostic tool is determined by inter-rater agreement (how often two clinicians get the same result when using the same tool). For autism, the highest inter-rater agreement (and therefore highest accuracy) is reached when clinicians with a lot of career experience diagnosing autism are allowed to freely use their judgment. The researchers therefore recruited psychologists and psychiatrists having assessed at least 100 autistic women over the past 5 years.

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Original article:
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Self-diagnosis is increasingly present and can be difficult to manage in adult women

Example 1

LClinicians have noticed over the past few years that more and more women have done a lot of research on autism are presenting for assessment. They highlighted that self-diagnosis was often correct, but worried about the impact of refusing an autism diagnosis when a patient had already invested a lot of energy in this hypothesis.

"They've access online resources, maybe joined support groups, and have very strongly identified with the autism spectrum" Participant 4.

"They very often have a specific belief or hope that they have autism rather than something else. And that's what could get in the way of the diagnostic process because we are trying to understand their real experience without being biased by their beliefs" Participant 11.

Many clinicians were worried about potentially activating depressive symptoms when disappointing a patient. They therefore explained to their patients from the very beginning that a diagnosis of autism was not guaranteed.

Example 2

Clinicians also noted that **autism in women could superficially resemble Borderline Personality Disorder (BPD)**. BPD was seen simultaneously as a misdiagnosis that many autistic women received prior to an autism diagnosis, AND a differential diagnosis that many clinicians noticed in their practice.

trigger warning mention of self-harm

"Autistic women have sometimes seen a clinician who told them, "well you have BPD because that's what women who cut themselves have." Participant 15

"Sometimes autistic women have been diagnosed BPD, have had the therapy and learned a lot about BPD and now act borderline because that's what they learned in therapy" Participant 19

Clinicians insisted on the fact that these conditions could be differentiated by the presence (BPD) or lack (autism) of attachment difficulties (emotional instability caused by a fear of abandonment).

This study concludes that the ability to make differential diagnoses is essential when assessing adult women for autism. However, many clinicians are not trained in differentiating autism from social anxiety, ADHD or BPD. Some institutions also limit the role of the clinician to a binary yes or no answer to a diagnosis of autism, without giving the tools to provide the person with alternative answers. Adult women are particularly susceptible to complex psychiatric histories and clinical presentations, making it crucial that clinicians are trained in differential diagnosis.