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Some women with autism present differently

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Summary

Autism spectrum disorder is still often insufficiently recognised in women, by professionals and the general public alike. This is partly due to the difference in presentation between genders. In women we see the presence of both camouflaging behaviour that helps cover up autism symptoms and more internalising symptoms such as depression, anxiety and fatigue. A more in-depth approach is necessary during the diagnostic process to determine whether or not all criteria are met. With this case report I would like to illustrate what the line of questioning looks like in clinical practice, leading to the substantiation of an autism diagnosis in a very high-functioning adult who at first glance does not immediately present according to the stereotypical image of autism.

Introduction

Autism is a hereditary, heterogeneous developmental disorder that can co-occur with other conditions (Lord et al., 2020). Its prevalence has risen over the years, partly because of a change in the diagnostic criteria and an improvement in the diagnostic assessment carried out. Research now estimates the prevalence of autism (ASD) in Europe at 1.4%, with a male-to-female ratio of 3.5:1 (Rosemarie et al., 2022).

The question remains whether women are under-represented in this ratio, given that sex differences in presentation and a male bias in ASD are responsible for misdiagnosed or missed diagnoses in women (Ochoa-Lubinoff et al., 2023).

Articles have already been published that make the case for better early diagnosis in girls. In 2023 an article appeared that described the presentation of ASD in girls under the age of 10. The population studied had already come into contact with child and adolescent psychiatry at a young age (M.E. Buruma, 2023). This contrasts with the subpopulation of adult women with ASD on which this article focuses, in whom the burden of distress only prompted a first contact with professional care later in life.

Here we see that, because of the different life stage, the cognitive flexibility they have acquired and their by-then highly developed ability to compensate and camouflage, the features of ASD show themselves in other ways than in a presentation under the age of 18 (Perry et al., 2022).

Up to 80% of adult women with ASD appear to be undiagnosed (McCrossin, 2022). The number of incorrect diagnoses in adult women also underlines the lack of expertise in this area within the care field to date. A recent study showed that, once ASD had been established, a previous different diagnosis turned out to be incorrect in 47% of cases in women, compared with 27% in men. These were most frequently personality disorders (Kentrou et al., 2021). Conversely, ASD is also sometimes wrongly

diagnosed when a different diagnosis actually applies; Tak described several examples of this from practice in 2020 (Tak, 2020). There is a clear need for more expertise and more publications on ASD in adult women who only make a first contact with professional care later in life.

This case vignette examines the picture of ASD specifically in women who, under the age of 18, still 'flew under the radar' and did not meet the criteria for ASD conspicuously enough in childhood to be assessed for it at a younger age. There are also still very few publications on how a psychiatrist using purely process-based diagnostic assessment, and without a multidisciplinary approach, can nonetheless arrive at a diagnosis of ASD in very subtle presentations of autism. This case is an example of how it can be done differently.

First impression

For the patient, the reason for referral to the psychiatrist was fatigue. This is the patient's first contact with psychiatry. She works half-time in an administrative role in a hospital and previously obtained a master's degree in midwifery. In addition, she runs the household and shares the care of 2 children, the youngest of whom was diagnosed with ASD a year ago. Since then, the patient has wondered whether she might also have autism.

At first sight the patient does not stand out socially. She has a long and happy marriage with 2 children and a steady group of friends whom she sees regularly. She receives no complaints from those around her about her communication, and during the first diagnostic interview she also comes across as sufficiently attuned, both verbally and non-verbally. Eye contact and reciprocity go well. However, the patient would describe herself as socially awkward, finds eye contact tiring and experiences that she constantly has to think about how to approach her social interactions, because she lacks a natural feel for them.

In terms of rigidity and restricted interests, too, the patient does not at first sight appear to fall short in her functioning. She does not speak or behave stereotypically during the first contact, and here too she receives no complaints from those around her, with the occasional exception of her partner. She does, however, have a strong preference for predictability and often has fixed routines. Even so, she tries to adapt to the unpredictability of the world here too, and is flexible when necessary. She also sometimes experiences fixated interests, but does not suffer greatly from them or receive criticism about them from those around her.

In addition, with regard to personality, we note that she has noticed all her life that she expects (too) much of herself, finds it hard to let things go, and has negative cognitions and feelings of guilt when she does something incorrectly or not well enough. She also finds it hard to prioritise her own needs or to take enough time to relax when something still needs to be done.

From the above it is clear that the patient does not present according to the stereotypical picture of ASD. What is present, however, are complaints of stress, worrying, anxiety and many periods of fatigue, both physical and mental. These are nonetheless related, among other things, either to social communication and interaction, or to problems around predictability and routine.

Although a few features of ASD can be noted at first sight, more depth is needed to determine whether the patient meets all the criteria.

Going deeper

When **part A of the ASD criteria (persistent deficits in social communication and social interaction)** is examined in more depth, it becomes increasingly clear that the patient experiences distress and a disproportionate loss of energy because social interactions are difficult for her. It also becomes clear that the patient has developed strong skills to camouflage these deficits from the outside world, so that they will not stand out in more superficial contacts. Only her partner regularly sees her get stuck in this; to the outside world she puts on a mask and uses a great deal of energy to appear more cognitively flexible and socially stronger than she actually feels.

- Regarding deficits in social-emotional reciprocity, we find that, although there is no marked impairment, almost all interpersonal interactions take a lot of energy every day. She has learned to make small talk, although she still does not like doing it and experiences it as pointless. In her head she is constantly busy thinking about how she can respond most appropriately to what the other person says.
- Regarding deficits in nonverbal communicative behaviours used for social interaction, we find that the patient also experiences difficulties here, camouflages them and compensates for them cognitively. She consciously works at making and maintaining eye contact, although she does not find it pleasant. She does not always understand whether people mean something as a joke, so she waits to see how those around her react before saying anything herself. With people she knows well, she simply asks. She regularly takes things literally when proverbs, figurative expressions, messages given between the lines and vague statements are used.
- Regarding deficits in developing, maintaining and understanding relationships, we find that she has been able to build lasting friendships and a relationship, but that the process was difficult and tiring. She has also always found getting to know new people hard, because she had no feel for how to make contact, what to ask and how to keep the conversation going.

When **part B of the ASD criteria (restricted, repetitive patterns of behaviour, interests or activities)** is examined in more depth, it becomes clear that the patient meets all the criteria here as well.

- Regarding stereotyped or repetitive motor movements, use of objects or speech, we find repetitive behaviour in the form of fiddling with her fingers and jiggling her legs; as a child she also bit her nails in a way that could not be stopped. In addition, she has been playing the same game for a long time, in which the same mechanism always applies (she is now at level 6089).
- Regarding insistence on sameness, inflexible adherence to routines or ritualised patterns of behaviour, we find difficulty with unpredictability. When plans change, she needs time to get used to it, with an initial resistance to the change, even when the proposed change is a pleasant one and does not disadvantage the planning. She has several fixed daily routines, such as when cooking and going to bed, in which everything is always done in the same order.
  All her life she has experienced a gap between the cognitive flexibility the world expects of her and how difficult and tiring this actually is for her.
- Regarding highly restricted, fixated interests that are abnormal in intensity or focus, we find that the patient has had several obsessions in which she could lose herself in the past. Examples are sewing, crocheting and immersing herself in autism after her son was diagnosed. At such times she is constantly occupied with it, and notices that she also wants to talk about it a lot with those around her. These interests come at the expense of her sleep: she can then no longer stop in time and often loses

her sense of time as well. She has collected many books, podcasts, articles and websites about these "special interests", more than she has time to get through.

- Regarding hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment, we find sensitivity in several areas. Examples are given in the visual, auditory and tactile domains, regarding food, and sensitivity to heat.

Additional information from collateral history

Interview with the patient and her partner

In this interview all the diagnostic criteria are also gone through with the partner, to check whether he too recognises the criteria in the patient. Attention should be paid to whether the patient's picture of herself corresponds with the experience of those closest to her. The partner recognised the various criteria for autism and was able to give additional new examples that the patient had not yet mentioned.

Interview with the patient and her parents

For this, a standardised developmental history questionnaire was used. It is important here to check whether the criteria for ASD were already present in early childhood. It is also striking that several people in the family recognise themselves in these criteria. Finally, this interview provides additional information with regard to differential diagnosis and possible comorbidities.

In summary, the parents confirmed that the criteria were already present in childhood, with new examples from that period that could support the criteria. There was also an emotional moment during the assessment when the patient's mother strikingly recognised herself in several features of ASD as well.

The patient's own perspective

> Since I came into more contact with information about autism after our son's diagnosis, I did recognise some traits. However, I did not recognise myself in the stereotypical picture. On the one hand I felt I had everything under control; on the other hand I felt I was drowning more and more in all the expectations from society and from myself.
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> Through the diagnosis and going through my difficulties, I am getting to know myself. Strange that this has to happen at 39. I now understand better where my behaviour and reactions come from, so I can make more sense of them. I am kinder to myself and am learning to sense and set my limits. By always just carrying on, I was exhausted. The need for help had been high for years, yet it never rang a bell with anyone that there was more going on underneath.
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> I am very grateful that more stories of women with autism are coming to light. The recognition does me good. It would be wonderful if there were more knowledge in society (individuals, employers, care professionals, ...) about the difficulties women face.

Discussion

This case is a clear example in which depth during the diagnostic process proved essential to reaching the correct diagnosis. Neither the objective psychosocial functioning nor the first clinical impression was indicative of ASD, and because of the high level of functioning and masking, an in-depth form of process-based diagnostic assessment was necessary.

Recent research also shows that people with ASD can function well psychosocially. A 6-year longitudinal study of adults with ASD and average to high-average intelligence showed that the majority of the study population functioned well psychosocially, in the domains of work, capacity for independent living and friendships. What also stood out in this study group was how late their ASD diagnosis came, at an average age of 33 (Scheeren et al., 2022). This supports the clinical experience that, to establish ASD in adults, one needs to look further than their objective psychosocial functioning.

The case also underlines that someone who gives an impression of verbal and non-verbal reciprocity during the interview can still meet the diagnosis of ASD, because this is learned behaviour. In the literature this is described with the term “masking” (Alaghband-rad et al., 2023). Masking here covers both suppressing features that are typical of ASD and displaying neurotypically coded behaviour in order to adapt as well as possible to the social context.

Annelies Spek’s DSM-5 interview for autism spectrum disorder (Spek, 2015) was used in this case as a guide for the further, deeper process-based diagnostic assessment. This left enough room for nuance and helped to substantiate the diagnosis of ASD firmly, despite the less typical presentation. The additional input from the developmental history with the parents and the collateral history with the partner was also essential to making the diagnosis.

Although it is not a diagnostic criterion of ASD, the main presenting complaint in this case was fatigue. During the diagnostic process it then became clear that this was strongly associated with the difficulties of autism, such as masking, sensory overload and over-demand on cognitive flexibility. The literature also supports fatigue as a typical symptom in women who mask a great deal. Besides fatigue, anxiety and depressive symptoms are also more often present as comorbidities in women with autism (Hull et al., 2016), and these may be part of the initial presenting complaint.

Conclusion

The case described above shows how high-functioning women with ASD can present with a stable psychosocial life and, during the first contact, give a first impression of adequate social skills and cognitive flexibility. In other words, they will less often bring the “stereotypical” picture of ASD to mind. Yet on closer examination they will meet all the criteria, and they will experience significant distress associated with this. In people with this form of presentation, we see that the psychological distress is just as present, but they come forward with internalising symptoms as their main complaint, such as depressive symptoms, anxiety or fatigue. Be sure to consider a possibly missed diagnosis of ASD if these complaints have proven chronic or treatment-resistant. Men can also present in this way, but according to current science and clinical experience this happens less often than in women. Keep the differences in presentation between genders in mind at your next contact in which ASD is being considered diagnostically. Annelies Spek’s DSM-5 interview for autism spectrum disorder can be a good guide here, in combination with a developmental history with the parent(s) and a collateral history from a partner or close friend.

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