

Automating autism assessment: What AI can bring to the diagnostic process

Yasemin J. Erden PhD¹  | Harriet Hummerstone PhD² | Stephen Rainey PhD³

¹Department of Philosophy, University of Twente, Enschede, The Netherlands

²Head of Teaching and Learning, Priory Lodge School, London, UK

³Philosophy, University of Oxford, Oxford, UK

Correspondence

Yasemin J. Erden, Department of Philosophy, University of Twente, P.O. Box 217 7500 AE Enschede, The Netherlands.
Email: y.j.erden@utwente.nl

Abstract

This paper examines the use of artificial intelligence (AI) for the diagnosis of autism spectrum disorder (ASD, hereafter autism). In so doing we examine some problems in existing diagnostic processes and criteria, including issues of bias and interpretation, and on concepts like the ‘double empathy problem’. We then consider how novel applications of AI might contribute to these contexts. We’re focussed specifically on adult diagnostic procedures as childhood diagnosis is already well covered in the literature.

KEYWORDS

artificial intelligence, autism, diagnosis

1 | INTRODUCTION

The applications of artificial intelligence (AI) in the treatment of autism are already well covered, so this article focuses on its use in the diagnostic process.^{1,2} That said, it can be hard to disentangle diagnosis and treatment where those processes are complex, especially given the array of factors and judgements involved. As Wulff³ notes, what one clinician will consider rational in decision making, another will view quite differently. While this is true of any diagnostic process, a further complexity presents itself where AI is involved. AI, especially machine learning applications, process inputs according to prior held data, and produce outputs deemed appropriate by that synthesis of input and data. In diagnostic settings, AI-enabled devices might be used that (A) provide data relevant to assessment of autism and (B) modify their own behaviour in clinically relevant ways in response to that data. Given the adaptability and responsiveness of AI, this kind of outcome is possible. Indeed, such applications are already planned.

Some diagnostic tools include observation of interaction with robotic playmates for identifying autistic children.⁴ While the robotic devices employed in such scenarios are mainly passive, they can nevertheless be used in a ‘learning mode’, adjusting levels or styles of interaction based on inferences about the child’s behaviour.⁵ Such devices as these may represent ‘theranostic’ tools, in that they deliver

therapeutic intervention in tandem with diagnostic assessments. A learning, AI-enabled robot detecting eye-gaze, affect, movement, and so on, may classify the child it is interacting with as autistic and in so doing alter its mode of interaction seamlessly, in a clinically informed way. Where does the observation end, diagnosis begin, and clinical decision-making occur?

2 | INTRODUCTION TO AUTISM AND DIAGNOSIS

Autism spectrum disorders, as they are referred to in the latest version of the Diagnostic and Statistical Manual,⁶ consist of a broad range of differing experiences and behaviours that are present from birth, with a focus on two main areas: social communication and interaction, and repetitive and restricted behaviours. The process of gaining a diagnosis of autism is difficult, not least because of disagreement about traits, diagnostic processes, as well as about how it overlaps with other conditions such as intellectual disabilities.⁷ Prospects for accurate diagnosis depend on many factors external to the assessment itself, and there is discord as well as confusion about criteria, disputes about terminology, and multiple perspectives on the condition by clinical practitioners.⁸

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Uncertainties partly arise because traits can manifest quite differently between individuals. For these reasons Lorna Wing^{9(p.111)} described an autistic *continuum*, which suggests understanding autism 'on the hypothesis of a continuum of impairments'. However, the latest version of the most recent version of the Diagnostic and Statistical Manual has merged the previous diagnostic terms of Autistic Disorder, Asperger Syndrome and Pervasive Developmental Disorder Not Otherwise Specified into one umbrella term of 'autism spectrum disorders' (ASD), and focuses on only two main behaviours individuals are likely to demonstrate. This adds further complications to the diagnostic process, illustrating how contextual factors (in this case, revisions to the manual) may affect the nature of the diagnosis given. For example, influential changes to the DSM-III in the 1980s led to greater numbers of children diagnosed as autistic.¹ These children are now adults, and some are advocates for their own (neurodiverse) identities,¹⁰ even though they would not receive the same diagnosis using modern classification criteria.

A problem with this diagnostic approach is that it is entirely deficit-focussed, suggesting that these criteria are in danger of over-applying a medical model perspective on disability.¹¹ The neurodiversity movement, which argues that there are natural variations of brain function that can affect beliefs, thoughts, and emotions¹² are opposed to this, with some autistic advocates stressing that autism is inseparable from identity.^{13,14} Consequently, deficits are redefined as *differences*,² representing diverse, yet valid forms of human behaviour, being, and identity.¹⁵ This account fits within a narrative of disability as contingent on the environment, typically referred to as the social model of disability.¹⁶ The neurodiversity view moves away from the medical and deficit-focussed autism narrative that argues for a cure, instead arguing for recognition and acceptance of these neurological differences.¹⁷ This highlights another issue with current autism diagnostic criteria: despite attempts such as Mahdi et al¹⁸ to refine an international measure of functioning for autistic individuals which includes strengths related to autism (such as honesty, attention to detail, advanced memory functions, and expertise in specific areas), the current medical diagnosis for autism negates the strengths and abilities of autistic individuals. It also does not acknowledge that what may be perceived as a difficulty in one circumstance can be useful in another. For example, attention to detail can be debilitating if all-encompassing, but is a strength for proofreading.

In addition to being deficit-focussed, Milton¹⁹ describes the 'double empathy problem' which may also affect the diagnostic process. The double empathy problem suggests that neurotypicals have a responsibility to be aware of a different perspective when communicating with an individual on the autism spectrum. Milton states that '...autistic people often lack insight about non-AS perceptions and culture, yet it is equally the case that non-AS people lack insight into the minds and culture of "autistic people"' (p. 886). Similar social perspectives are reflected in descriptions of autism as a culture that is not accepted by the mainstream.²⁰ Linking closely to the neurodiversity approach, this position suggests that the behaviour of autistic people should not be considered through the lens of neurotypical standards.

During the diagnostic process, the accounts and behaviours of autistic individuals may be rated/judged by a neurotypical clinician – without sufficient understanding and knowledge, it is possible that hetero-normative assumptions will consequently be applied, pathologising individuals.

3 | ASSESSMENT INSTRUMENTS AND AUTISTIC ADULTS

The majority of assessment instruments for autism rely on scaling data, which is combined with clinical knowledge, observations and diagnostic criteria to make a decision. Not only does this necessitate time and expertise, but the supporting psychometric data that contributes to the reliability and validity of these assessment instruments are variable.⁷ In addition, the majority of assessment instruments focus on children in order to identify early interventions – however, this is of little use to adults who are waiting for an autism diagnosis.²¹

Because of the flexibility of age ranges, best fit with the DSM criteria, and international use, the Autism Diagnostic Interview - Revised (ADI-R) is considered the 'gold standard' of standardized interview tools.^{22(p.2)} Individuals or their caregivers are asked 93 questions which cover the individual's full developmental history over the course of 1 to 2 hours. Each question is then given a rating score by the interviewer, and used to calculate scores in three behavioural areas – social interaction, communication and language, and restricted and repetitive behaviours. If the individual's scores meet or exceed the cut-off point for each area, they are given a diagnosis of autism.

However, there are some issues with the ADI-R that remain unresolved. Notwithstanding the training required for a clinician to be qualified in administering the test, the interpretation of the individual's answers and the associated ratings are completed by the interviewer. In addition to the usual issues regarding retrospective recollection of their childhood, autistic adults who are seeking a diagnosis later in life may have developed highly expert and refined coping strategies to allow themselves to 'fit in' to a neurotypical world, referred to as masking or camouflaging.²³ Masking could involve making eye contact, imitating others' behaviour/gestures, and following social scripts to hide social difficulties that the individual experiences – as Lei et al²⁴ observe, 'If the diagnostician further misses signs of camouflaging, superficially "typical" non-verbal skills and social manner may be wrongly taken as evidence to rule out the presence of autism' (p.691). Lei et al²⁴ note that camouflaging occurs in both men and women (slightly more in women), and therefore a 'thorough understanding' (p.700) of it may improve the diagnosis of autism.

In summary, the diagnostic process for autistic adults has two significant issues: the deficit-focussed criteria, and the reliance on others' interpretation of evidence to achieve a diagnosis. AI has been cited as one way to manage the rise in calls for diagnosis in children,²⁵ especially as autistic adults diagnosed later in life report negative



experiences without a diagnosis relating to their identity and a lack of support.²⁶ Removing face to face interaction and programming an understanding of camouflaging, strengths and abilities into AI diagnostic tools may therefore help to alleviate waiting times that a significant number of adults encounter when waiting for a diagnosis, and reduce the number of diagnostic false negatives that may occur from masking and other learnt behaviours.

4 | AI AND AUTISM DIAGNOSIS

According to the latest statistics from NHS Digital (August 2020), individuals over the age of 18 waiting for a referral and diagnosis between April 2019 and January 2020 waited an average of 361 days from initial contact to a diagnosis. The older the individual, the longer this took, with individuals aged 45 to 54 and 55 to 64 waiting for an average of 480 days.

AI is seen by tech enthusiasts as 'a novel way to improve accuracy and effectiveness during the detection of autism', while machine learning (eg, neural networks) can assist in the analysis of large data sets to 'recognize the phenotypes of autism'.²⁷ Researchers also see machine learning methods as a way to cut assessment waiting times.²⁸ Typically both AI and machine learning rely on input about both autism classification as well as existing data about diagnostic processes and cases, which are then measured against independent novel cases so as to measure how effective the technology will be in predicting a diagnostic outcome. Other reasons for the use of AI can include: lack of specialists, as well as clinician uncertainty during the diagnostic process. This, some argue, can be augmented by 'computational systems that have the abilities of a clinician', and which can support existing methods of diagnosis by 'confirming the evaluation decisions of the doctors'.²⁷

AI is also being used in converging technologies that includes brain scanning in diagnostic processes. This is an approach entirely in line with the animating principle of the Research Domain Criteria (RDoC) that guides contemporary US and European thinking in psychiatry.²⁹⁻³¹ A study by Chen et al³² suggests it may be possible to identify 'neuroimaging biomarkers' using MRI and from this to distinguish between those with autism and those without. Their approach included application of 'data-driven artificial intelligence' to help distinguish between various neuroimages. Yes, as the authors acknowledge, these approaches are not unproblematic. On the one hand, they suggest machine learning techniques may offer what they call 'objective tools' (which concept is itself problematic) to 'augment' current diagnostic processes for autism, yet on the other they are clear this is not an effort to replace existing diagnostic criteria.

There is some promise of computer-aided tools that evaluate MRI images with machine learning techniques, yet there are also weaknesses, limitations and hurdles to overcome. It requires sufficient interdisciplinary understanding and dialogue, as well as knowledge that extends beyond disciplinary boundaries. For instance, computer scientists tasked with creating algorithms for diagnosis ought to have sufficient understanding about autism, as well as

about the various facets of the diagnostic process, while it is important that clinicians understand the novel technologies that underpin any automated system.²⁷ The difficulties here also include the depth of understanding. This includes that to know how an algorithm works is not necessarily sufficient to recognize the biases that can be inherent to both the algorithms and the datasets on which they rely. What's more, algorithms require vast troves of data before they can really be effective, which as of now simply do not exist.³³

In these, as well as in the other uses of AI that we note above, we see that the tools being developed are largely complementary to the existing clinician-led diagnostic processes. As such, it is not clear that the use of such systems would in fact resolve the kinds of issues that we note above, that is, as solutions to limited resources or a lack of specialists. Nor is it clear that AI will help to avoid bias, especially if the results of an automated system will require interpretation and/or application by the clinician who has undertaken the assessment. A system cannot prevent a person from using the system to interpret data in a way that replicates their own preferences and biases. As Bone et al note,^{34(p.1123)}

Focusing solely on data processing, but ignoring context, can produce misleading results and conclusions. Conversely, the application of computational methods by researchers outside machine learning communities can be a precarious situation because there are numerous ways to misuse algorithms and misjudge their results.

As Thabtah writes,²⁸ classification is a complex and dynamic process that requires participation of experts who will act as decision makers. Yet studies examining AI in relation to autism assessment tend to 'deal with the problem of ASD classification from a static manner whereas existing machine learning algorithms are merely applied on an historical ASD dataset. Then, a classifier is built without measuring its impact inside the true diagnostic tool'.^{28(p.286)} More than this, there are a number of outstanding, yet important issues that still need to be resolved, including broader ethical implications arising from the use of these technologies, which we have not considered in this article.

In the diagnosis of autism, AI typically relies on data analysis models that operate on established question-based results from diagnostic tools, such as the ADI-R. The statistical analysis therefore relies on good quality data³⁵ and is best suited to diagnosis of individuals who display a certain group of behaviours. As we note above, however, there are not insignificant problems with the ADI-R, especially where there is an overemphasis of certain traits. This includes where AI is underpinned by concepts derived from popular accounts of autism, such as found in Baron-Cohen's³⁶ paper that describes autism as an 'extreme male brain'. This account of autism can and does result in stereotypes about autistic individuals, and where such material is used to guide assessment of autism, whether by clinicians, by AI, or in combination, the quality of such assessment and intervention is affected.^{37,38}

The clinical encounter is, as we note above, structured according to competing and changing norms. For instance, where AI relies on data derived from past cases that, since the time of its collection, has become outdated because wider norms have shifted in terms of how that data ought to be contextualized.³⁹ Past data may have been collected in a paradigm associating behaviours of various kinds as *deficits* requiring remedies. But subsequent deployment of AI assessment on the basis of that data may occur in contexts where neurodiversity is viewed as *difference* rather than deficit. The complexities of a blurred line between observation, diagnosis and treatment are thus complicated yet further in having that blurred line drawn in a space of shifting normative dimensions. Specifically, this affects autonomy both in terms of clinicians and those awaiting assessment. The supposed objectivity of data somehow intrudes upon what might otherwise be a clinical encounter structured dynamically in response to specific judgements made by the people involved, based on negotiated values.⁴⁰ If nothing else, this illustrates that the data upon which AI assessments might be made does not objectively account for the specific case being investigated *here and now*, but instead brings with it an aggregation of prior data, from prior cases. The data is 'objective' in a sense far more general than the given case here and now, and comes embedded in a normative context of some kind relating, for example, to the means and aims of its original collection, mode of processing, storage, federation, and so on.²⁸ The challenge in understanding these multi-dimensional complexities may not be in finding a resolution once and for all, but in finding a mode of continuing critical discussion on what matters without ruling out specific points of view.

Given these complications, a lot of power may be ceded to the programming of the AI device and its capacity to draw robust inferences about behaviour, normal variation and pathology.^{41,42} In this kind of context, a space is opened that would diminish the role of the clinical psychiatrist.⁴³ What's more, as we note above, where assessment of adults is at stake, matters are further complicated in that they are likely to have developed behavioural strategies such that clinically relevant factors (eg, attention, verbosity, affect) might be masked through experiential learning.⁴⁴ This considerably complicates the role of an AI theranostic for general use with adults, unlike the case of a robot interacting with an autistic child, say, who is unlikely to have developed sophisticated masking strategies.

On the one hand, AI can help to avoid some of the biases displayed by clinicians (gender, culture), but on the other it does not have capacity for the kinds of nuance or flexibility that an experienced and knowledgeable clinician can adopt when measuring a person's traits against a particular criteria for assessment. Nor does it have the scope for the kind of discursive, contextual analysis in which to make judgements about what a combination of behaviours means for diagnosis. This includes recognizing that a successful ability does not preclude problematic dimensions, for instance. In this way, it may be difficult for an AI to successfully evaluate a narrow range of tasks that can be considered successful (eg, in measurable outputs) but which have complex experiential qualities. Such as where someone is successful in tasks that make particular use of an autistic trait, like sustained concentration, but which may have some negative experiential qualities.

Where a focus on successful task outcomes is present, concentration is a good thing. Where concentration shades into fixation, or obsession, the potential distress undergone is detrimental to the person. Where task outcomes are evaluated as successful or not, such factors are unaccounted for. Yet these may be essential in assessing for autistic traits.

Compounding all of these complications is the fact that AI tends to produce simplistic classifications. Of the studies that made use of machine learning in autism diagnosis that Thabtah examined, the majority used datasets that 'contained only two class labels, e.g. ASD and Non-ASD'.^{28(p.287)} The problem with this approach to binary classification is that the machine or AI requires sufficient simplicity so as to avoid confusion, yet people being diagnosed are complex beings whose complexity resists such categorization. This is especially true once they are adults with all the training and knowledge that experience can bring. Blurred or fuzzy examples can be confusing to algorithms, which can increase false positives, or indeed result in false negatives.

It seems to us that the use of AI in autism diagnosis for adults requires additional consideration. On the one hand, as we have noted, AI would not necessarily be subject to the kinds of personal biases that can sway the judgements of the physician. On the other hand, AI relies on datasets, and if these datasets are compiled with the contributions of biased physicians then those biases can be further reified. Additionally, an AI could in principle 'keep abreast' of contemporary research in ways not available to the physician, in being updated via continuous data updates. Yet if AI research is underpinned in some way by controversial or misinformed literature, this might not be rectified by additional research accumulation.

5 | CONCLUSION

While AI and machine learning offer much promise for the diagnosis of autism, there are substantial risks that technologies will simply represent and/or rely on the same biases, conceptual flaws, and practical weaknesses evident in existing diagnostic processes. This article has shown what might be some of the positive outcomes from a technological approach to assessment of autism, but we have also shown some of the current limitations. We have not considered whether these limitations are inherent to the technologies, but it seems clear that there is still quite a way to go before even a partially automated system of diagnosis will be reliably effective. A fully automated system, including one that relies on brain data, seems even less likely in the medium term. In any event, it is vitally important that the kinds of issues considered in this article are examined early in the process of planning and designing of technologies, like AI, that could be used for diagnostic processes. To neglect to do so, or to do so only after these technologies are implemented, risks serious long-term problems. Unchallenged biases, including as contained in programming and data sets, can find their way into the status quo for technology, and bad habits and tendencies can lead to reified processes. As Winner^{45(p.126)} explains, knowledge and technological invention, especially as



influenced by factors like corporate profit, can reinforce each other and become entrenched in what we do.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

ETHICAL APPROVAL

No ethical approval required for this study.

DATA AVAILABILITY STATEMENT

Data sharing not applicable to this article as no datasets were generated or analysed during the current study.

ORCID

Yasemin J. Erden  <https://orcid.org/0000-0001-9958-6643>

ENDNOTES

- ¹ Although autism can, in theory, be diagnosed at any time in a person's life, assessments remain more common for children. Barriers to later diagnosis remain, especially for adult women and those with minority ethnic identities (Bargiela, Steward, and Mandy 2016).
- ² An account of neurodiversity and difference does not require a denial of deficit. Instead, the idea of a spectrum points to the complexity of a term like deficit, and how this can play out in different contexts.

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