Report on a problem studied in the N-CODE Study Group April 2025

Automated Diagnosis of Autism Spectrum Disorder

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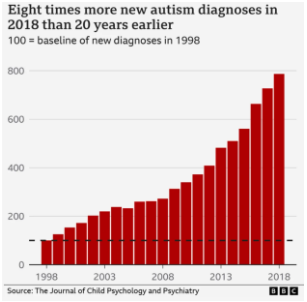
Our group discussed the urgent need for efficient, cost-effective, and accurate diagnostic tools for Autism Spectrum Disorder (ASD), highlighting the challenges of current diagnostic methods, including high costs, long waiting times, and limited specialist availability. We explored the feasibility of automated ASD diagnosis through three promising approaches: functional Near-Infrared Spectroscopy (fNIRS), speech analysis, and sleep/movement data analysis, combined with artificial intelligence (AI). fMRI-based studies previously demonstrated high diagnostic accuracy, but practical implementation barriers necessitated exploration of alternatives. fNIRS emerged as a low-cost, portable technology offering comparable classification potential, achieving approximately 95% accuracy in preliminary studies with children. Similarly, AI-driven speech analysis showed high accuracy (94%) in differentiating autistic from neurotypical individuals based on acoustic properties of speech. Sleep and actigraphy data were also promising. Recognizing the lack of adult-specific data, the study group's planned next step involves a 12-month feasibility study to assess whether these methods can reliably distinguish ASD in high-functioning adults. Successful development and implementation of these approaches would represent a significant advancement, enabling rapid, affordable, and community-based diagnostic support, thereby alleviating pressure on healthcare systems and providing nuanced diagnostic information.

Introduction

Autism spectrum disorder is a neurodevelopmental condition characterized by persistent deficits in social communication and social interaction across multiple contexts, along with restricted, repetitive patterns of behaviour, interests, or activities. The symptoms can vary widely in severity and presentation, but can include (American Psychiatric Association, 2013):

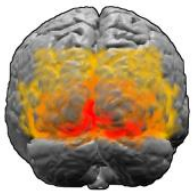
- Failure to initiate or respond to social interactions
- Abnormalities in eye contact
- Trouble making friends
- Stereotyped or repetitive speech or other motor movements
- Inflexible adherence to routines
- Abnormal intensity or focus on a highly restricted range of interests
- Adverse response to certain sounds or textures.

For a clinical diagnosis, the symptoms must be present from early development – although, as we shall see in a later section, increasingly diagnosis occurs in adulthood. For a clinical diagnosis, the symptoms must also cause significant functional impairment (e.g. socially or occupationally).



The economic costs of autism in the UK exceeds **£32 billion a year** (Buescher et al., 2014). Quick, accurate and cost-effective diagnosis can help, but ASD diagnosis is currently none of those things. Private diagnosis costs in the order of £3000 (Oxford CBT, 2023). NHS waiting times are long and increasing; the number of open referrals has risen approximately 10x in five years to 200,000 (NHS England, 2024). This is reflective of a long-term trend; in the period 1998 to 2018, autism diagnoses in the UK increased 8x (Maybin & Blastland, 2025; Russell et al., 2022], see left.

Currently, diagnosis is time-consuming, labour-intensive (at least half a day per client), and requires the expertise of a very limited group of highly qualified individuals. There are also emerging concerns about the quality of diagnosis due to the diverse aetiology, expertise for which is not easily available. **Identification of biomarkers as a clinical support tool in ASD** would be a "gamechanger" for patients, clinicians, services, and for the health and social care economy.



Members of our group recently demonstrated that an **Explainable AI system** could classify individuals as autistic versus neurotypical based on resting-state **fMRI scans**, with **98% accuracy (**Vidya et al., 2025). The system was trained and tested on a large publicly available dataset of such scans (De Martino et al., 2014). The explainable AI approach allowed us to determine which brain areas were critical to this automated diagnosis; they include areas such as the calcarine sulcus (an early visual area; see right, brain viewed from

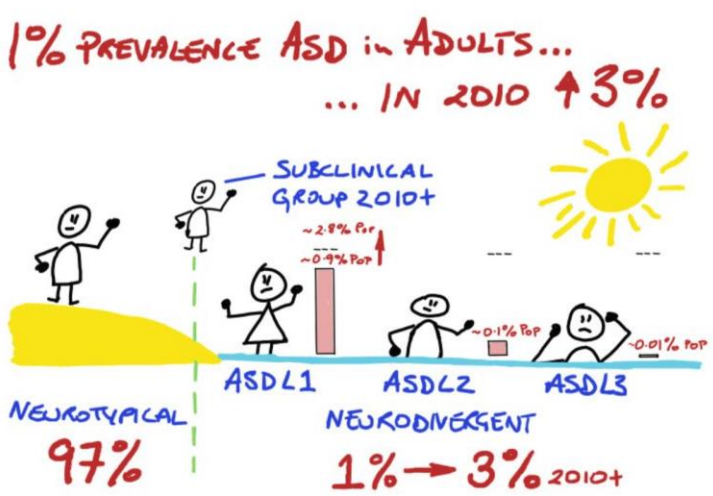
behind); the areas identified are consistent with the underlying neuroscience of the condition (Gandal et al., 2022).

Description of the problem

The N-code workshop was extremely useful in helping us to the refine the clinical problem we were trying to solve. ASD, in terms of a clinical diagnosis, is widely considered to have three levels of severity (American Psychiatric Association, 2013):

At **Level 1**, the patient has clear autistic traits, but they are living a reasonable life. They are known to the primary care system (GPs), but the level of support required post-diagnosis is limited. Level 1 represents 80-90% of those receiving a clinical diagnosis.

At **Level 2,** the patient's autism is a predisposing factor to mental health problems. These patients are known to NHS mental health teams. They are about 10% of the clinically diagnosed population.



At **Level 3**, their autism is the cause of severe disability. They are unlikely to be able to live independently and can require continuous one-to-one care. Behaviours can be challenging (e.g. storing of faeces) and small changes to routine highly problematic (e.g. change in time the post arrives leading towards psychosis). They are about 1% of the clinically diagnosed population.

Taking all these three levels together, clinically diagnosed ASD has historically had an about 1% prevalence rate in the adult population (Brugha et al., 2012). Clinical opinion largely agrees that the increase in prevalence nationally (from 1% in 2010, to 3% now), comes mainly from an additional 2% of the population who are arguably neurodivergent without necessarily meeting the Level 1 criteria above. During the workshop, we coined the term **Level ND** to reflect this group and **Level NT** to reflect the remaining 97% of the adult population, who are neurotypical in this regard.

Given the rapidly increasing waiting lists (see Introduction) and the labour-intensiveness of clinical diagnosis (about half a day clinician time per client, minimum), there is the need for a fast (bio)marker that can inform clinical triage. Most critically, that tool needs to be able to distinguish between the three lowest levels – Levels NT, ND and 1 – as that is where most of the diagnostic need lies. At these levels, initial presentation is often in adulthood, so an adult-focussed clinical diagnosis tool is the focus of this project.

Solutions proposed

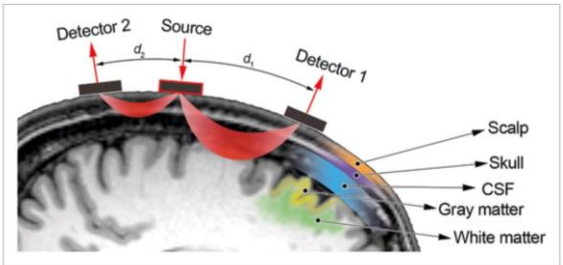
Although we came to the workshop with an in-principle demonstration of high accuracy ASD diagnosis using resting-state fMRI (see Introduction), it became apparent during the workshop that this approach was not promising for the development of a low-cost, community-based tool. We identified two main problems:

1. **Current and likely future capacity and practicality.** fMRI, of any sort, is basically not used in NHS radiography departments, despite its longstanding use in research. Both the radiologist we brought with us (Lucy McGavin) and a neurologist we talked to during the workshop (Brendan McLean) rated the probability of widespread adoption any time soon as very low. This was for a variety of reasons, including the low signal-to-noise ratio, sensitivity to movement, low acceptability to patients (particularly neurodiverse ones) due to loud and potentially claustrophobic conditions, need for training in the complex analysis paths required, and the simple fact that waiting times, even in the case of tumour detection and other major conditions, was already substantial with little hope for additional capacity on the horizon.
2. **Limited potential for community use.** There is a limit to how community-based MRI will ever be. There are basic physics constraints in producing multi-Tesla fields that mean it is hard to ever see this technology becoming available outside of a hospital (or large community diagnosis centre) setting. There are some exciting developments in low-field strength MRI, and these devices are somewhat portable. However, they suffer from some of the same problems are high-field techniques (e.g. movement sensitivity), have worse signal-to-noise ratio, and still are not as practical for community use as some of the solutions we discuss below.

During the workshop, we investigated three different solutions, which we describe in separate sections below.

Functional Near-Infrared Spectroscopy (fNIRS)

Our in-principle demonstration was with fMRI, a technology that detects blood oxygenation level in the brain. This signal is also available, for cortical areas, at the scalp, using near-infrared light, with an arrangement of LEDs and detectors. The technology is known as fNIRS (Pinti et al., 2020), and is portable, wearable, less sensitive to movement and the equipment cost is approximately 100x lower than MRI. One advantage of our Explainable AI-based approach is that it identifies the areas most crucial to diagnosis. These were cortical areas (such as calcarine sulcus, part of early visual cortex) and hence accessible to fNIRS.



Reviewing the existing literature during the workshop, we found encouraging evidence that fNIRS combined with AI could be used for high-accuracy ASD classification. For example, one study was able to distinguish ASD from neurotypical with a 2.5 second sample of resting-state fNIRS recording (Zhang et al., 2023). They used a convolutional neural network to achieve 95% accuracy in classification. Three other studies reported similar results with similar levels of accuracy (Li et al., 2023; Mazziotti et al., 2022; Zhu et al., 2023).

One limitation of the existing research is that it is all done on a child sample (10-year-olds in the case of Zhang et al.). Participants who have been diagnosed by age 10 are likely more severe cases than people diagnosed in adulthood. The latter are our focus for reasons described above. It is also not a foregone conclusion that signals present at 10 years old, are still present in adulthood – brain maturation continues into at least early adulthood (Giedd et al., 1999).

Speech analysis

We were fortunate to be joined during the workshop by Melanie Jouaiti, a computer scientist with experience in the AI-assisted classification of speech data in a healthcare context (Jouaiti & Dautenhahn, 2022). Speech is a very rich data source, containing not only the words that are spoken, but the pronunciation, tempo, rhythm, timbre and so on.

Literature review conducted during the workshop revealed that there are reasons to be optimistic that AI-assisted classification of ASD by speech will work. For example, resting-state fMRI reveals significant differences in speech areas between autistic and neurotypical adults (Li, 2024). The acoustic properties of speech also differ in ASD; for example, in pitch and in vowel formants (Lyakso et al., 2017).

Further review during the workshop revealed several recent promising studies using speech in ASD classification. For example, one study reported 94% accuracy (Alzakari et al., 2025); the latest step in a steady improvement in accuracy from about 70% in 2022 (Chi et al., 2022), and 85% in 2024 (Li, 2024). Some semi-public datasets are also available (MacWhinney, 2019).

However, we then hit the same issue as with pre-existing fNIRS research – it focusses on early diagnosis and hence on data from young children. So, again, we identified a need for an adult dataset.

Sleep and movement data

We were also fortunate to be joined during the workshop by Maia Angelova, an applied mathematician and data scientist with a track record in modelling sleep and insomnia (Rani et al., 2022). We searched the literature, and found good *prima facie* evidence

that sleep data might be usable as a clinical diagnosis tool for ASD. For example, children with autism have a much smaller REM sleep percentage (15%) than neurotypical controls (23%; Buckley et al., 2010) and are about twice as likely to have a sleep disorder (Reynolds et al., 2019). They sleep less and take longer to fall asleep (Elrod & Hood, 2015). They are more likely to have mutations in genes relevant to the regulation of circadian rhythm (Yang et al., 2016). Recently, autism has been classified in children from full sleep EEG data, achieving around 90% accuracy from a random-forest machine-learning approach (Martinez & Chen, 2023).

Overall, the picture is much the same for sleep data as it is for fNIRS and for speech analysis. There are good reasons to be optimistic that there is strong potential for deriving (bio)marker(s) from these data that will allow classification of the studied populations into high-ability autistic, neurodiverse and neurotypical groups. However, pre-existing work is in children, rather than the high-functioning adults who are our focus, and the data is 'research quality' (in this case, full sleep lab studies) rather than what would be available from a cheap, fast, wearable, community-based solution. Hence, a feasibility study meeting those criteria is required.

Relatedly, recent work in Maia's lab indicates that chronic insomnia can be accurately classified from actigraphy (movement) data alone, when collected over a minimum of 5 days (Kusmakar et al., 2021). Although there is less work in this area for ASD, it provides another possible source of signal for ASD classification, given the aforementioned sleep problems.

Future work

In summary, previous research indicates the potential of fNIRS, speech analysis, and sleep/movement data to drive high-accuracy AI classifiers of ASD. The limitation in each case is that this research is on children – in no case has efficacy been demonstrated in adults. So, although the prior research is cause for optimism, it is not a foregone conclusion that the same techniques work in adults – particularly the largely late-diagnosed, high-functioning adults that are source of much of increased diagnosis rate since 2010.

This leads naturally to the first step in our planned future work – we wish to discover whether AI classifiers accurately distinguish between ASD and neurotypical high-functioning adults, using fNIRS, speech analysis, or sleep/movement data? We also want to explore the acceptability of the technologies with patients, and the utility of our approach with clinicians.

During the N-CODE workshop, we planned a 12-month feasibility study along these lines and have now requested follow-on funding to facilitate this. At the end of this feasibility study, we expect to have a better understanding of the potential of fNIRS, speech analysis, and sleep/movement data in the AI-assisted diagnosis of ASD; and we will have submitted our findings for publication in a suitable high-impact journal. We

further expect to have collected some useful PPIE to inform further work in this area, and likewise to publish this.

Our plans beyond the feasibility study obviously depend on how it works out. Nonetheless, we are optimistic that high-accuracy classifiers can be built from at least one of our three modalities – or, if needed, through the multimodal combination of them. If so, this would be the first step towards the development of game-changing technology. Technology that would allow ultimately provide a cheap, fast, community-based, accurate clinical support tool to assist with the diagnosis of ASD.

Assuming the feasibility study works out as planned, the next step is to test robustness of our classification system using a sample of NHS patients who have already received a full clinical diagnosis (versus controls). Assuming that works, the next step would be to test the systems prospectively, bringing them into use clinically for a few hundred patients in a full Randomized Control Trial, comparing against Treatment As Usual (RCT) for both speed and efficacy.

As the efficacy of this technology becomes clearer, there would be an increasing need to develop a commercial, ideally mass-market, product as this is how we lower the cost and hence increase accessibility and benefit.

Discussion and conclusion

We envisage a future where a biomarker-based approach such as ours can bring reality to clinicians and clients of the concept of autism being a spectrum. It is notable that when telling a patient they have high blood pressure, one is also able to quantify how high it is, offer differential support and advice depending on how high it is, and that in general the patient treats it as good news if their blood pressure is only slightly high. None of these things are currently the case for adults who self-refer as possibly autistic – they are seeking a binary classification and are often dissatisfied if that classification is that they are not autistic. Ultimately, a technology that can provide a quick reading that quantifies the level of autistic traits in an individual accurately would give patients a useful measure of the magnitude of their condition; information that would also help stratify further care in an increasingly overwhelmed NHS.

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