Stakeholder-led Study Group Application (N-CODE) - Version 2.

Automated Diagnosis of Autism Spectrum Disorder

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Background

The economic costs of autism in the UK exceed £32 billion per year [1]. Quick, accurate and cost-effective diagnosis can help, but ASD diagnosis is currently none of those things. NHS waiting times are long. Private diagnoses cost in the order of £3000 [2]. The process of diagnosis is time consuming, labour-intensive, and requires the expertise of a very limited group of highly qualified individuals. There are also emerging concerns of the quality of diagnosis and diagnostic overshadowing. This is because ASD is diagnosed by three cardinal symptoms whose aetiology can be diverse, expertise for which is not easily available. Identification of biomarker to robustly screen or diagnose ASD would be a "gamechanger", for patients, clinicians, services and for the health and social care economy.

What's already available?

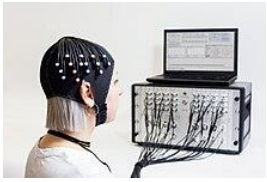
Proof-of-concept research at the University of Plymouth demonstrates that AI-supported analysis of brain scans can be used to diagnose autism with 98% accuracy [3]. This work made use of large, publicly available datasets [4].

Questions to be tackled during the meeting

The top-level question is – *how do we go from proof of concept to a workable, cost-effective, commercial product?* We can see at least four challenges that need to be addressed:
First, the publicly available data, which are essential for training the AI, are licensed for research only. Second, the proof-of-concept system is a binary classifier, i.e. it can only respond 'autistic' or 'neurotypical'. It is an important question whether the system would false-positive on any type of non-neurotypicality (e.g. misclassify dementia as autism). This can only be answered by widening the test set. If the false-positive rate is high, this would have to be addressed by widening the training set. Thus, there are two related questions here – what is and is not permissible use of research-licensed data, and the practicalities of collecting large-sample data sets, in the order of 1000s of scans based on prior research.
The third challenge revolves around issues relating to the fast, cost-effective, and accurate application of the proof-of-concept to real-world clinical settings. The proof-of-concept AI uses resting-state functional MRI (rs-fMRI) as its input. This information is not collected in clinical settings, where almost all uses of MRI are structural scans. rs-fMRI takes longer to perform, requires more analysis / postprocessing and the analysis is rarely supported in clinical MRI settings, which are focussed on fMRI for motor or

language localisation. It is less reliable as the signal is weaker, and scan quality is more affected by patient variables (such as whether they are sedated, on other medication, etc.). It is louder than structural MRI, which may be an issue for anxious patients and other vulnerable groups. Thus, there is a research question about whether our proof-of-concept approach can be developed such that it is robust, quick, and turn-key reliable in a clinical setting, or if the findings can be translated to use structural data which are readily available.

Improving community-based diagnosis



The key challenge here concerns the availability and portability of scanning technology. The first step towards broader accessibility is the introduction of NHS Community Diagnostic Centres [5]; an important development that nonetheless brings its own challenges in terms of translating work so far done in high-field strength scanners with research-active radiographers to the low field-strength, less research intensive, multi-manufacturer context of community diagnosis. But, for wide community-based use, we need to go further. How do we do this? One option would be to use portable MRI [5]. Another, very exciting, option is to explore using FNIRS rather than fMRI. The technology already exists and is already about as portable as EEG [6].

Required research skills

This cross-disciplinary project would benefit from a wide range of expertise. We are interested to talk to people with expertise in large-sample multi-site collection of MRI data. Discussion with researchers who have successfully navigated the translation from proof-of-concept research to clinical use would be valued– as close in area as possible, but we suspect many of the skills are transferable. Also, moving to community or mobile modes brings issues of image quality – expertise in how to improve this would be useful. The input of an epidemiologist would also be valuable, in order to conduct a systematic review of existing literature. To some extent a coalition of the willing, we suspect. We're hopeful that N-CODE is in a uniquely good position to pull a team together!

References

[1] https://pubmed.ncbi.nlm.nih.gov/24911948/ [2] https://www.oxfordcbt.co.uk/how-much-is-a-private-autism-assessment-for-an-adult [3] https://arxiv.org/abs/2409.15374 [4] https://fcon_1000.projects.nitrc.org/indi/abide/ [5] https://www.plymouthhospitals.nhs.uk/community-diagnostic-centre/ [6] https://hyperfine.io/swoop/overview [6] https://en.wikipedia.org/wiki/Functional_near-infrared_spectroscopy