A causal Bayesian network model for early diagnosis of autism.

by

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Abstract — The aetiology of the condition autism spectrum disorder is suspected to derive from multiple dependent and independent variables, and as such lends itself well to analysis and diagnosis by Bayesian networks. In this project, a state-of-the-art network using Bayesian artificial intelligence and probabilistic reasoning is built for the purpose of aiding accurate early diagnosis of autism spectrum disorder, incorporating some of the growing new body of knowledge.

Keywords — Autism spectrum disorder; diagnostic model; medical idioms; causal Bayesian network model, AI tool; CNV - copy number variation.

I. INTRODUCTION

Autism spectrum disorder (ASD) is a subcategory of Pervasive developmental disorder (PDD), characterised by a range of symptoms and comorbidities (NHS, 2022a) (NAS, 2022a) (Hayes, et al., 2018). Severity varies widely, with capabilities able to evolve over time in diametrically opposed directions, but common to all is the altered impact on the lives of sufferers and their families. Confirmed diagnosis requires psychiatric/psychological assessment by trained professionals using recognised standard custom graded questionnaires. Despite considerable advances in elucidation of the neurobiology of ASD that span genetic factors and markers (Toma, 2020) (Timothy, et al., 2013) to radiomics (Sen, et al., 2018), its method of diagnosis has barely changed since it was devised some five decades ago.

ASD incidence has progressively increased to almost epidemic proportions in the last few decades (Lord, et al., 2018;). Early positive diagnosis can lead to early intervention leading to better outcomes for sufferers and parents, while negative diagnosis enables further assessment and identification of alternative condition. The World Health Organization (WHO) puts the current worldwide base rate at 100 per 10,000 population or 1 ASD person per every 100 people (WHO, March 2022), although reliable global base rates are elusive in the literature due to lack of accurate statistics from low and middle-income countries. Many artificial intelligence (AI) methods have been proposed in recent years for diagnosing ASD, however many lack clarity and reproducibility when given different realistic scenarios of risk factors. Bayesian networks offer many advantages over traditional AI that make their use in modelling and hence diagnosing ASD a preferably choice. They have been described as direct representations of the world that model the "... *top-down (semantic) and bottom-up (perceptual) combination of evidence* ..." (Pearl, 2011). The model built in this research project follows these principles to offer a more accurate and reliable diagnostic tool.

(restricted access - full project and research dissertation paper in restricted repository, freely available to stakeholders and interested relevant parties. Interested? Please contact

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for access. The following are major chapters and some sub-chapters. Thank you.)

II. LITERATURE REVIEW

A. Autism in General

B. Diagnosis of ASD

C. Machine and Deep Learning Methods in Autism Research

D: Bayesian Networks as diagnostic tool

III. METHODOLOGY

A. BN Architecture

B. BN Parameters

IV. RESULTS

V. DISCUSSION

VI. CONCLUSION

From the results obtained, this causal BN has been shown to model real world scenarios with excellent diagnostic performance. It makes an important contribution to the field, which is modelling acquired cultural behavioural mitigation.

VII. FUTURE WORK

REFERENCES