Diagnostic Assessment Techniques and Non-Invasive Biomarkers for Autism Spectrum Disorder

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ABSTRACT

Autism spectrum disorder (ASD) is a complex heterogeneous neurological disorder that has led to a spectrum of diagnosis techniques. The screening instruments, medical and technological tools initiate the diagnosis process. Clinicians and psychologists propose therapies depending on the examination done by these methodologies. The literature has accounted dozens of diagnostic methods and alternative and complementary therapies but still lack in highlighting the proper biomarker for early detection and intervention. The emerging multi-modal neuro-imaging techniques have correlated the brain’s functional and structural measures and diagnosed ASD with more sensitivity than individual approaches. The purpose of this review article is: (i) to provide an overview of the emerging ASD diagnosis methods and different markers and; (ii) to present the idea of integrating all the individual methods in to a multi-modal diagnostic system to enhance detection sensitivity. This system possesses the potential to diagnose and predict ASD clinically, neurologically & objectively with high detection sensitivity.

KEYWORDS

Autism, Biomarkers, Correlation, Diagnosis, Integrated, Multi-Modal, Sensitivity

INTRODUCTION

Autism is a perpetual developmental disorder marked by the core symptoms mainly in three domains: (i) social interaction impairment (ii) inability to communicate (iii) repetitive activities and constrained interests. The disorder does not depend upon a single condition but on ‘spectrum’ of the deficits, affecting children in different ways and hence called as Autism Spectrum Disorder (ASD). With the increase in awareness and diagnostic techniques more cases of ASD children are being suspected. The diagnostic label to the phrase “infantile autism” was firstly provided in Diagnostic and Statistical Manual of Mental Disorders, Third Edition (DSM-III; American Psychiatric Association [APA], 1980). Since then, diagnostic criteria have broadened and included the terminologies Autism Disorder (AD), Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS) and Asperger Syndrome (AS) in DSM-IV, Text Revision (APA, 2000). The new edition DSM-5® has combined these three

DOI: 10.4018/IJEHMC.2019070105

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terms under the single diagnosis of ASD (APA, 2013). The complexity of disorder is not limited to diagnosis only but also to intervention. The recommended medicines are used for suppressing disruptive behavioral manifestations but do not play any role in retraining deficits. These alternative & complementary therapies viz. music & sensory integration plays an important role in improving child’s condition by reducing deficits & enhancing his functioning (Höfer, Hoffmann & Bachmann, 2017).

ASD diagnostic methodologies used by clinicians, neurologists/specialists are widely classified in to three domains viz. primary screening instruments, neurological techniques and technology tools. The screening instruments involve clinical observations, behavioral measures, interview and scaling methods. These instruments at level 1 differentiate atypical child from normal & at level 2 distinguish ASD from those with other disorders (Coonrod & Stone, 2005). There are number of screening instruments available such as Autism Diagnostic Observation Schedule (ADOS) and Autism Diagnostic Interview-Revised (ADI-R) & Modified Checklist for Autism in Toddlers-Revised/with Follow-up (M-CHAT/RF). The neurological techniques study neuro-physiological and autonomous activities using different non-invasive methodologies such as Brainstem Evoked Response Audiometry (BERA)/Auditory Brainstem Responses (ABR; Tharpe et al., 2006), Electroencephalogram (EEG; Ekinci, Arman, İşık, Bez & Berkem, 2010), Galvanic Skin Response (GSR; Fukuyama, Kumagaya, Asada, Ayaya & Kato, 2017), Visual Evoked Potential (VEP; Sayorwan, Phianchana, Permpoonputtana & Siripornpanich, 2018). These also involve neuroimaging techniques viz. Magnetoencephalography (MEG; Tsiaras et. al., 2011), Magnetic Resource Imaging (MRI; Katuwal, Cahill, Baum & Michael, 2015), functional Magnetic Resonance Imaging (fMRI; Heinfeld, Franco, Craddock, Buchweitz & Meneguzzi, 2018) and Diffusion Tensor Imaging (DTI; Ingalhalikar, Parker, Bloy, Roberts & Verma, 2011). The technology tools like eye tracker, virtual reality (VR), robots and neuro/bio feedback are used for ASD diagnosis as well as intervention (Papagiannopoulos, Chitty, Hermens, Hickie, & Lagopoulos, 2014; Živoder, Martic-Biocina, Kosic & Bosak, 2015). These tools are non-invasive, easy to use, accurate, and are playing crucial role in ASD diagnosis.

These approaches are good at one point but poor at some other point such as EEG provides good temporal resolution & fMRI provides spatial resolution. The temporal resolution provides better understanding of ASD but at the same time EEG portability is an issue. On the contrary, fMRI has full connectivity & portability but poor temporal resolution. To overcome these problems the diagnostic approaches are shifting to the multimodal/integrated methodologies. The integration of different techniques/tools leads to multi-modal systems that diagnose ASD by correlating its underlying mechanisms (Mutsaerts, Heinrich, Sterkenburg & Sappok, 2016). For instance, integrated neuro-signaling & imaging tools viz. EEG & fMRI have correlated brain’s functional & structural measures & enhanced ASD detection sensitivity (Hames et al., 2016). The EEG & eye tracker combination has correlated the neuro-physiological mechanisms & gaze patterns in ASD (Billeci et al., 2017). These detection methodologies have provided candidate biomarkers viz. biological, neuro-psychological, physiological and neuro-imaging to generalize ASD diagnostic criteria (Anderson, 2015). These biomarkers assist the physicians/clinicians in predicting ASD through behavioral traits and abnormalities. The research in biomarkers has shifted to non-invasive from invasive (blood-based) markers & is still under validation.

There are number of studies reviewing one of the above-mentioned diagnostic approaches and comparing them to find the suitable method. In one such study, the different technological tools for ASD diagnosis have been compared and VR has been proved more advantageous (Liu, Wu, Zhao & Luo, 2017). Since, the disorder is highly complex & selecting a single diagnostic method would not be an efficient solution. With this fact in mind, the present paper’s first objective is to review the different available ASD detection methods & diagnostic markers in single study. Secondly, for further enhancement in diagnostic process, an idea of integrating screening instruments, neurological and technological tools on a single platform is presented in this paper.

The novelty of this paper is that all the diagnostic approaches used by therapists/clinicians have been reviewed in a single study. Also, the novel suggestion of combining all the available
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