EPP374

Evaluation of NCAM1, NRXN1, NLGN4, N-CADHERİN Levels in Peripheral Circulation of Children with Autism Spectrum Disorder and Their Siblings

A. T. Hira Selen[1]*, N. Uzun[1], I. Kılınc[2] and A. O. Kılıc[3]
[1]Child and Adolescent Psychiatry; [2]Clinical Biochemistry and [3]Department of Pediatrics, Necmettin Erbakan University, Konya, Türkiye
*Corresponding author.
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Introduction: Autism spectrum disorder (ASD) is linked to synaptic function and plasticity, and cell adhesion molecules play an important role in these processes. Dysfunction of CAMs disrupts synaptic activity and plasticity processes.
Objectives: This study aims to investigate NCAM1, NRXN1, NLGN4, and N-cadherin levels in the peripheral circulation of individuals with ASD, their siblings, and healthy controls without any psychiatric diagnosis. We also investigate how these biochemical parameters affect autism severity, behavioral problems, and autistic traits in siblings.
Methods: The patient group of the study consisted of 41 children aged between 18-72 months who were diagnosed with ASD according to DSM-5 diagnostic criteria and 41 healthy siblings aged between 24-72 months of the child diagnosed with ASD (Control Group 1). We assessed the severity of ASD symptoms in participants diagnosed with ASD using the Childhood Autism Rating Scale and the Autism Behavior Checklist. Control Group 2 consisted of 41 children aged between 18-72 months who did not have ASD or any other psychiatric disorder or physical illness (Control Group 2). The parents of the patient group completed the Autism Spectrum Screening Scale for the sibling of the individual diagnosed with ASD. We determined serum NCAM1, NRXN1, NLGN4, and N-Cadherin levels in peripheral blood samples by ELISA.
Results: There was no statistically significant difference in NCAM1, NRXN1, NLGN4, and N-Cadherin levels between all three study groups. There was a statistically significant positive correlation between NCAM1 and NRXN1 levels in the ASD group.
Conclusions: In this study, we investigated the serum levels of NCAM1, NRXN1, NLGN4, and N-Cadherin, which are cell adhesion molecules potentially associated with ASD etiology. Although the proteins we investigated were expressed differently between the groups, we did not find any statistically significant difference between ASD patients, healthy siblings, and healthy groups. More research is needed in this area.

Disclosure of Interest: None Declared

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The use of computer-aided analysis of facial expressions based on the support vector machine in the diagnosis of ASD

K. Kania[1]*, K. Wilczyński[2], D. Pęszor[3], L. Cichoń[2], K. Wojciechowski[3], A. Kawalec[2] and M. Janas-Kozik[2]
[1]Medical University of Silesia, Katowice; [2]Department of Psychiatry and Psychotherapy of Developmental Age, Medical University of Silesia, Sosnowiec and [3]Research and development center, Polish-Japanese Academy of Information Technology , Warsaw, Poland
*Corresponding author.
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Introduction: Autism spectrum disorder (ASD) is characterized by communication challenges, particularly in non-verbal aspects such as facial expressions.Research in this area is limited due to the lack of accurate methodologies. Existing literature generally agrees that individuals with ASD often show a disconnect between verbal communication and emotional expression, with facial expressions being diminished or inappropriate. Most studies have relied on ratings by highly trained observers, which can reduce accuracy and introduce biases, such as confirmation bias.
Objectives: Our goal was to create a model for capturing and analyzing facial expressions using computer algorithms and assess its effectiveness in identifying individuals with ASD.
Methods: The study involved 100 participants, divided into two groups based on ASD diagnosis. The ASD group included 73 individuals, 51 (69.8%) of whom were male, while the control group comprised 27 participants, with 16 (59.2%) being male. ASD diagnoses were made by a specialist child and adolescent psychiatrist using developmental history and mental state examinations, confirmed with the ADOS-2 protocol. In the control group, ASD was ruled out using the same protocol. A significant age difference was found between the ASD group (mean age: 14 years; 95% CI: 13.5-14.5) and the control group (mean age: 16.3 years; 95% CI: 15.2-17.5), according to the Mann-Whitney U test.
All participants completed three tasks: a semi-structured conversation, recognizing facial expressions displayed on a screen, and imitating these expressions. Throughout the tasks, participants' faces were recorded using five cameras positioned around them. Faces were then detected in the images using a sliding window algorithm in a multi-resolution representation of the Gaussian pyramid utilizing a linear classifier based on the Support Vector Machine (SVM) with a classical Histogram of Oriented Gradients (HOG) descriptor. An Ensemble of Regression Trees was applied to these detected faces to model facial landmarks in each frame. Using these landmarks, anthropometric distances and proportions were calculated, which were then used to train the SVM classifier.
Results: The obtained model was able to predict the diagnosis of ASD in the study population with almost 100% accuracy. The mean difference between the probability of the correct class and the probability of the incorrect class determined by the SVM on the test set was 56%.
Conclusions: This method of facial expression analysis using an SVM classifier shows potential as a tool for diagnosing ASD. The technique could be applied using smartphones. However, further research is needed to evaluate its clinical viability, particularly when using non-standard devices. These findings also support the hypothesis that individuals with ASD display facial expressions significantly differently from neurotypical individuals.

Disclosure of Interest: None Declared