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Uncovering biologically relevant Autism subtypes using advanced machine learning techniques

Christopher "Gabby" Vento[1]*, Joseph F. Cubells, MD[2], Larry J. Young, PhD (posthumous)[3], Elissar Andari, PhD[4,5]

[1]College of Medicine and Life Sciences, The University of Toledo, Toledo, Ohio 43614
[2]Department of Genetics, Emory University, Atlanta, Georgia
[3]Department of Psychiatry, Emory University, Atlanta, Georgia
[4]Department of Psychiatry, The University of Toledo, Toledo, Ohio 43614
[5]Department of Neurosciences, The University of Toledo, Toledo, Ohio 43614

*Corresponding author: christopher.vento@rockets.utoledo.edu

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Background: Autism spectrum disorder (ASD) is characterized by main deficits in social interaction and social communication. This vague definition of ASD does not encompass the wide heterogeneity of its phenotypical presentation. It is critical to identify ASD subtypes that are biologically relevant and that respond to personalized treatment.

Objectives: The objective of this study is to use a multimodal approach to create biologically relevant subtypes.

Results: 114 adult men (18-45 years old), including 74 neurotypical (NT) and 40 ASD were recruited and completed a series of behavioral tests such the NEO-PI-R, reading the mind in the eyes (RMET), Symptom Checklist 90-revised questionnaire (SCL-90), intelligence quotient (IQ), and Broader Autism Phenotype Questionnaire (BAPQ). Clinical measurements and fMRI data was collected from ASD subjects for validation. We used a random forest tree algorithm to classify ASD and NT. We included NEO-PI-R and RMET in the main classifier. The random forest tree model classified ASD and NT with an average accuracy of 80%. Top features included personality domains such as extraversion and neuroticism. K-means clustering was used to derive ASD subtypes based on the shapely values; this created 3 subtypes (Subtypes 1, 2, 3). T-tests indicated significant differences in the following measures: ADI-R repetitive behaviors, BAPQ, IQ, SCL-90, neuroticism, extraversion, RMET and rs-FC between the superior temporal sulcus, anterior cingulate, and insula.

Conclusions: Our results suggest that ASD subtype 1 is characterized by high neuroticism, lower warmth, higher scores on RMET, higher IQ, and higher rsFC between STS and salience network. Subtype 2 was found to be close to neurotypicals. Subtype 3 is characterized by high neuroticism, high repetitive behaviors, and lower rsFC between STS and salience network. These results are very promising, and the next step is to examine whether these putative subtypes are biologically relevant and whether specific subtypes respond better to certain pharmacological treatments.

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