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METABOLOMICS PROFILES OF AUTISTIC CHILDREN'S URINE REVEALS ALTERED DOPA-MINE AND NORADRENALINE LEVELS

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A utism spectrum disorders (ASDs) are a group of neurodevelopmental disorders consisting of a delayed or altered language development and difficulties in social interactions. Beside clinical and social tests, metabolic abnormalities are frequently associated with ASD and their knowledge could help clinicians to provide earlier and more reliable diagnosis. A considerable number (16,704) of urinary altered metabolites were recorded in autistic children, with 222 displaying the largest differences (p-value ≤ 0.05 and fold change ≥ 1.5) between young ASD children and controls. Several altered metabolic pathways have been implicated in the disease, such as tryptophan, purines and pyrimidine's; however, in this paper, altered level of the most important neurotransmitters have been discussed. The highest levels of dopamine and its metabolite, homovanillic acid, indicated a dopamine β -hydroxylase enzyme block, probably due to presence of 4-cresol together with a higher level of vitamin C, all revealed in the urine of autistic children. Moreover, the incorrect synthesis of the active form of vitamin B6, pyridoxal-5'-phosphate was also detected, which is an essential co-factor for the biotransformation of glutamate into GABA. The accumulation of glutamate and lower concentrations of GABA were found in all of the autistic children examined.

BIOGRAPHY

Federica Gevi has completed her PhD in cellular biology at University of Tuscia, Italy. Currently she is a research RTDa of Tuscia University, Italy. She has 10 years of experience in the field of mass spectrometry-HPLC (MALDI TOF, Orbitrap, Q-TOF, amaZon ETD) in protein identification, metabolomic and lipidomic characterizations in various biological samples. She has 23 publications that have been cited over 300 times and her publication H-index is 10.

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