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Decreased platelet serotonin level and polymorphisms of serotonin transporter in patients with Sjogren's syndrome

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It is generally thought that fragile X-associated tremor/ataxia syndrome (FXTAS) represents a late-onset neurodegenerative disorder occuring in male carriers of a premutation expansion (55–200 CGG repeats) in the fragile X mental retardation 1 (FMR 1) gene. However, several female patients with FXTAS have also been reported recently. We explore FXTAS in young patents with action tremor, ataxia, emotional disturbances and cognitive dysfunction. Magnetic resonance imaging (MRI) of the brain showed diffuse cortical atrophy, while 1H-MR spectroscopy (MRS) revealed decreased levels of N-acetylaspartate (NAA) in the cerebellum, basal ganglia, and pons. Genetic testing confirmed FMR 1 gene premutation of CGG repeats in the abnormal allele. We concluded that FXTAS may be an under-recognized genetic disorder, particularly in women.

Biography

Helena Sarac completed her doctorate study in the "Serotonin platelet concentration and polymorphism of serotonin transporter in patients's with Sjogren's syndrome" with practical experience in spectrofluorimetric methods and PCR method at the Croatian Institute for Brain Research in Zagreb affiliated to the University of Zagreb, School of Medicine from 1999 and Institute Rudjer Boskovic in Zagreb, Croatia. After her study of serotonin, she jointed the Division of Heredodegenerative Disorders, Department of Neurology, Zagreb University Hospital Centre, University of Zagreb, School of Medicine where she is applying her research on serotonin in various autoimmune and neurological disorders. This method is capable of quantification of the serotonin in platelets as limited peripheral models for serotonergic neurons in the brain. Dr Šarac directed and participate in various clinical research studies in the field of neuroimmunology and movement disorders. It is expected that her work will lead to the explanation of pathogenesis and development of novel biomarkers linked to stress, depression and symptoms of somatic and mental fatigue in various autoimmune and neurological disorders.

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