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A new complex of phenotype due to heterozygous mutation in the TSC1 gene

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Abstract

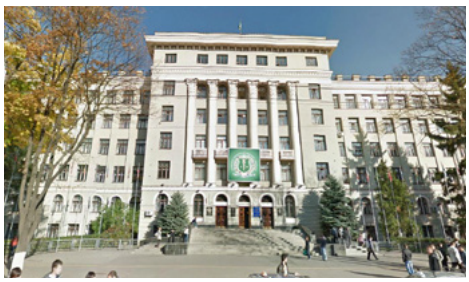
Introduction: Mutations in the TSC1 gene lead to the development of type 1 tuberous sclerosis. Tuberous sclerosis complex (TSC) is a neurocutaneous disorder characterized by multisystem hamartomas and associated with neuropsychiatric features: autism spectrum disorder, attention deficit hyperactivity disorder, cognitive impairment.

Methods: We describe a new complex neurological and skin syndrome characterized by vascular malformation of the lower extremities and Rett –like syndrome.

Results: As a result of exome sequencing (Genotec laboratory, Dr. Natalya Baryshnikova), a mutation was discovered in the TSC1 gene located on chromosome 9 on the 12 exon — a non-synonymous substitution c.1253C> G (p.Pro418Arg) —in a heterozygous state. According to ACMG criteria, the mutation is regarded as probably pathogenic. Predictive programs Polyphen2 and SIFT regarded the identified mutation as pathogenic. The mutation is not described in the databases. The frequency of mutation in the population is unknown. In the genes responsible for Rett syndrome and similar conditions, including the genes CDKL5, FOXG1, NTNG1, pathogenic mutations were not detected. No mutations were found in the PIK3CA and AGGF1 genes associated with Klippel-Trénaunay syndrome. In the genes ACVRL1, AKT1, CCBE1, CCM2, ENG, FLT4, FOXC2, GATA2, GJC2, GLMN, GNAQ, KIF11, KRIT1, PDCD10, PIK3CA, PTEN, PTPN14, RASA1, SMAD4, SOX18, STAMBP, TEK, associated with other vascular malformations - no mutations detected. Skin lesion in our patient with a mutation in the TSC1c.1253C> G gene (p.Pro418Arg) is represented by vascular malformation, and the TSC-Associated Neuropsychiatric Disorder was characterized by Rett-like syndrome.

Biography

Olena P Zdybska graduated from the medical faculty of the state university. In 1991 she graduated from the Department of Clinical Genetics and Ultrasound Diagnostics of the Kharkiv Institute for Advanced Training of Physicians and started working at the Interregional Medical Genetic Center. Since 1992 passed the competition for the position of assistant of the Department of Clinical Genetics and Ultrasound Diagnostics of KMAPE (part-time). In 1996 defended her dissertation on the topic and quot; The role of small developmental abnormalities in the early diagnosis of hereditary and congenital diseases in children and quot; Since 2000 works at the Department of Medical Genetics of Kharkiv National Medical University, in 2004 received the title of Associate Professor. She is author of more than 200 scientific papers and has 2 patents.



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