Overlap of the Pitt–Hopkins and Lennox-Gastaut syndromes

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Pitt-Hopkins syndrome (PTHS) was first described in the literature in 1978 and is clinically characterized by intellectual disability and developmental delay, breathing problems, frequent epileptic seizures and characteristic facial changes. It is thought to be a very rare condition, with approximately 500 affected reported individuals worldwide. PTHS is an autosomal dominant disorder caused by mutations in the TCF4 gene. We present a 24-year-old patient with pharmacoresistant form of epilepsy (absence attacks, atonic seizures, and generalized myoclonic seizures), intellectual developmental disorder and clinical diagnosis of epileptic syndrome (Lennox-Gastaut syndrome). During his life, he repeatedly performed magnetic resonance imaging (MRI) of the brain including MRI 3T, that showed post-ischemic porencephalic changes in the morphology of right occipital cortex. Electroencephalography revealed slow spike-and-wave complexes with diffusely slowed background activity, characteristic for LGS. The examination of the patient showed the development anomalies of the head, foot and spine. Due to pharmacoresistant epilepsy vagal nerve stimulator was implanted in 2009 and reimplanted in 2017.In 2014, because of predominance of atonic seizures, anterior two-third corpus callosotomy was also performed. Clinical improvement has been achieved with the above mentioned procedures, and the frequency of seizures has decreased considerably. In 2016 next-generation sequencing analysis involving 142 genes related to epilepsy was performed which revealed a heterozygotic missense variant in the TCF4 gene, a mutation previously linked to Pitt – Hopkins syndrome.Following the clinical picture and findings of the molecular-genetic testing, it is the first patient with a confirmed diagnosis of Pitt-Hopkins syndrome in Croatia. Considering the importance of counselling and further therapeutic algorithm, genetic counselling should be performed early in the course of the disease. We have described our case report of PTHS that has been published as letter to editor in November 2018 issue of Springer journal Acta Neurologica Belgica (*Acta Neurol Belg. 2018 Nov 13. doi: 10.1007/s13760-018-1045-2).*