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Clinical description of familial cavernous malformations

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Familial cerebral cavernous malformations is a rare inherited disease with a frequency of 1-5/10,000 in the general population. It is inherited in autosomal-dominant pattern through three genes - KRIT1, CCM2 and PDCD10, located respectively in 7q21.2, 7p13, 3q26.1 chromosomes. The disease is diagnosed mostly in Latin Americans. The manifestation is with multiple cavernomas in the head and spinal cord and rarely in retina and skin. Clinical symptoms are determined by localization of the lesions and their size. Most frequently occurring are epileptic seizures, focal neurological deficits, non-specific headache, cerebral haemorrhages, skin and retinal lesions. The disease occurs usually in adulthood and about 40% of cases are asymptomatic. The diagnosis is made with MRI images with multiple vascular malformations of cavernous characteristics and molecular-genetic analysis of the family members. We present a case of familial cerebral cavernous malformations in a father and a son of Armenian origin, diagnosed in the Clinic of Neurology of St. George University Hospital in Plovdiv. In the father, the disease debuted at a young age with epileptic seizures successfully treated with antiepileptic therapy. At age 76, there was abducens nerve palsy as well as a rapidly growing tumor formation in the neck. In the son, a 33-year-old man, the disease is manifested with epileptic generalized tonic-clonic seizures. The MRI performed in both show a number of brain cavernomas of different locations and sizes, some with a typical "popcorn-like" image, as well as deposition of hemoglobin degradation products and calcifications. An adrenal adenoma of the right adrenal gland and two cysts in the left kidney were also found in the older patient. Retinal lesions are not visualized in both. Genetic screening confirms the presence of mutation c.1061_1064dup in the exon 11 of the KRIT1 gene (transcript NM_194455). This is the first family in Bulgaria with familial cerebral cavernous malformations with a mutation in the KRIT1 gene and a typical clinical development.

Biography

M Peicheva is a Bulgarian Neurologist with experience in Vascular Disease. She works in Clinic of Neurology at St. George University Hospital in Plovdiv, Bulgaria. Her interests are connected with ultrasound examination and vascular abnormalities. She is a Volunteer Consultant at Raredis, a part of Bulgarian Institute for Rare Disease. Her last publications are about some rare syndromes in Bulgarian population.

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