A rare manifestation of Fabry's disease

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Fabry's disease is a recessive X-linked genetic disease, caused by the deficiency of the α-galactosidase-A (GLA) enzyme [1]. Approximately 300 mutations are known to affect the 9 possible isoforms of the GLA gene. Vascular endothelium, the heart, the brain and the kidneys are often involved [2]. Stroke, which is caused by alterations of cerebral vessels, is a frequent complication of the disease [3].

Case report: The patient (male, 42) was observed in 2001 due to hemiparesis. CT analysis led to the diagnosis of ischaemic stroke. Later the patient showed a progressive spastic quadraparesis. Laboratory test for thrombophilia was negative. MRI detected distended cerebral vessels and lesions of the white matter. The CSF test did not prove MS. The patient's neurological state progressed with time. The patient developed psychoorganic syndrome. Histological examination of red papules led to the diagnosis of angiokeratoma corporis diffusum. Genetic examination detected a point mutation (C to T, Arg112Cys) in the GLA gene. The activity of the GLA enzyme was low. In order to trace renal and cardiac defects, the patient was regularly examined. To date these examinations revealed no alterations, and proteinuria did not appear. Symptoms of Fabry's disease also appeared previously in the patient's family.

Our case involving the Arg112Cys mutation is the second in which only the cerebrum and the skin are affected [4]. However, this mutation was already known from a classical Fabry's disease case [5]. In this case the patient carried two mutations in the GLA gene, one of which was the Arg112Cys. Together, this mutation might affect an isoform of the enzyme which exerts its action mainly in the ectodermal tissues. This may be the cause of the limited organic manifestation.

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