A 35-year-old female was presented to the department of neurology at our hospital due to an epileptic attack that preceded a speech disorder. The symptoms occurred a week before admission to the hospital. A closer examination revealed the patient was conscious but not fully alert; she responded very slowly, her speech was incoherent and slurred, which suggested motor and sensory speech disorder. Other symptoms included spastic paresis of the right upper limb, partial blindness of the right side, trouble with left eyeball abduction, feet clonus. The deep reflexes were normal and symmetrical, no meningeal sign was observed.

Before the episode she also complained about hearing loss and Type 1 diabetes. Gynecological history was unremarkable; she had three children and no miscarriage. From the patient family history we knew that her mother also suffered from Type 1 diabetes, hearing loss, and dementia. She died at the age of 60.

Prior to the patient’s current admission, a second epileptic attack took place. As the partial paresis of the right limbs and speech difficulties sustained CT scan of the brain was performed and a large hypodensic area described as ischemic stroke of the left hemisphere was diagnosed.

MRI confirmed the diagnosis of a fresh ischemic stroke that involved temporal, occipital, and parietal lobe as well as left thalamus. In addition to that, a small ischemic area in the right temporal lobe appeared. In USG mean flow velocity in ACA, MCA, PCA and VA was measured and no change was detected. EEG was typical for epilepsy, and the record changed especially in the frontotemporal part of the brain.

Based on these findings and clinical history, epilepsy was diagnosed and valproic acid was administered.

During the patient stay in our department no further epileptic attacks were observed. Patient state stabilized and almost all symptoms except the low grade sensory aphasia disappeared.

Patient was transmitted to the Clinical Department of Neurology were genetic tests were performed and diagnosis of MELAS was confirmed. The mutation MT-TL1 was ascertained in the patient and her son.

DISCUSSION

MELAS is a multisystem disorder with onset typically in childhood. Children develop normally with the exception of height, as they tend to be short for their age. Symptoms like generalized tonic-clonic seizures, recurrent headaches, anorexia, and recurrent vomiting start to appear between the ages of two and 10 years and are caused by lactic acidosis. Also exercise intolerance, proximal limb weakness as a primary syndrome of myopathy, and sensoneural hearing loss as well as retinitis pigmentosa are common. Just as in our patient, seizures are often associated with stroke-like episodes or transient hemiparesis or cortical blindness. Laboratory findings include lactic acidosis and diabetes. In pathologic sample of biopsied muscle ragged red fibers are seen.1 2

MELAS is caused by mutations in mtDNA and is transmitted by maternal inheritance. The mother of a proband usually has the mtDNA mutation and doesn’t have to have...
symptoms but transmits the mutation to all of her offspring. Mutation in mitochondrial DNA gene MT-TL1 is the cause of the disease in 80 percent of cases. Other mutations, like MT-ND5, can also be causative.5-9

Because of the heteroplasmy (e.g., occurrence of regular and mutated DNA in the different mitochondria of the same cell), detection of the mutation may be complicated.4 Hence, the leukocyte may not be good material for examination. The best samples are skeletal muscle tissue, skin fibroblasts, hair follicles, and urinary sediment. No specific treatment for MELAS exists. However L-arginine showed promise in treating stroke-like episodes. Some authors suggest coenzyme Q10 and its analog idebenone as a prevention of primary manifestations.1,7,8

Prenatal diagnosis of MELAS is possible if a mtDNA mutation has been detected in the mother. However, prediction of the phenotype of the offspring is not possible.3

What is important from a physician point of view is valproic acid can increase the frequency of seizures in individuals with MELAS. However, we didn’t observe that fact in our patient. Also, an anticancer drug called DCA should be avoided because of increased risk for peripheral neuropathy. Patients should be examined at annual intervals by an ophthalmologist, cardiologist, and endocrinologist. Furthermore, the patient’s offspring also should be monitored.9

CONCLUSION
Since the mutation MT-TL1 was ascertained in our patient and her son, both of them are being closely monitored in our outpatient clinic. The diagnosis has a great meaning for the patient and her children, especially when it comes to the prevention of the disease for which they are at risk. When young people present with ischemic stroke, the diagnosis of MELAS should always be taken into consideration. Inexpensive and easy tests can ascertain the concentration of lactic acid in blood serum, which is usually much higher than in the normal population.