Abstract

Myoclonic epilepsy with ragged-red fibers: the first described clinical case in Ukraine

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Objective: To provide a description of a case of myoclonic epilepsy with ragged-red fibers (MERRF) diagnosed in Kyiv, Ukraine.

Background: MERRF is a rare mitochondrial disorder that begins in childhood and affects the nervous system and skeletal muscle as well as other body systems. Typical MERRF symptoms include: myoclonus, generalized epileptic seizures, cerebellar ataxia and cognitive decline. Other symptoms may include myopathy, optic atrophy, hearing loss, peripheral neuropathy and cardiomyopathy. Currently there is no effective cure of MERRF (DiMauro S., Hirano M., 2015).

Methods: Clinical description and laboratory testing.

Results: A 36-year-old woman complained of inability to walk, violent muscle jerking of upper and lower limbs, impairment of speech, generalized epileptic seizures and severe general weakness. She described general weakness for 9 years, generalized epileptic seizures and violent muscle jerking of upper and lower limbs for 5 years, walking difficulties for 4 years. She has not been able to walk for six months.

Family history was unremarkable.

On examination, there was mild cognitive impairment. She had the initial stage of optic atrophy, mild dysarthria and mild hearing impairment. There were severe, myoclonic movements in the upper and lower limbs (more pronounced in the left leg), tongue tremor and severe kinetic tremor of both arms. She had mild generalized slowness and mild spasticity on her legs. Tendon reflexes were normal. Babinski reflexes was negative on both sides. Muscle power was 4 points in the upper and lower limbs. There was no rest tremor or sensory loss. She was not able to walk due to unsteadiness and muscles weakness.

Laboratory blood tests were normal; MRI of brain was normal; EEG: epileptiform activity.

DIAGNOSIS of myoclonic epilepsy with ragged-red fibers was confirmed by mitochondrial A8344G mutation.

Conclusions: This case demonstrates the clinical course and progression of a rare neurological disease and is the first case to be diagnosed in Ukraine as genetic testing is now becoming available.

Literature

1. DiMauro S., Hirano M. MERRF. In: GeneReviews®[Internet]. University of Washington, Seattle, 2015.