

Neurology 2020: MRI of the brain, limb muscles and mitochondrial disorders in myotonic dystrophy - Olga Petrovna Sidorova - Moscow Regional Research Clinical Institute by M.F. Vladimirskiy

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Objective: To study changes in the brain and muscles, mitochondrial disorders in patients with myotonic dystrophy.

Material and Methods: 12 patients with myotonic dystrophy were examined. Of these, 1 was diagnosed with type 2 and the remaining 1 type of the disease. The diagnosis is confirmed by DNA diagnostics. MRI of the brain was performed. The MRI of lower and upper limb muscles was done on the scanner built around a 1.5 Tesla (T) magnet (Optima MR450w, GE Healthcare) using a T1- and T2-weighted images. For the muscle damage quantification by using MRI a qualitative muscle grading scale, developed by Mercuri et al. (2002) is used. Muscle atrophy grade evaluated during MRI can be divided according to the following criteria:

Grade 1 – Early moth-eaten appearance with scattered small areas of increased signal.

Grade 2a – Late moth-eaten appearance with numerous discrete areas of increased signal with beginning confluence, comprising less than 30% of the volume of the individual muscle.

Grade 2b – Late moth-eaten appearance with numerous discrete areas of increased signal with beginning confluence, comprising 30–60% of the volume of the individual muscle

Grade 3 – Washed-out appearance, fuzzy appearance due to confluent areas of increased signal.

Grade 4 – End stage appearance, muscle replaced by increased density of connective tissue and fat, with only a rim of fascia and neurovascular tissue distinguishable.

For a quantitative cytochemical study of the activity of mitochondrial enzymes in peripheral blood lymphocytes, the method proposed by A.G.E. Pearse in the modification of P.P. Narcissov. The activity of four mitochondrial enzymes being involved in carbohydrate (lactate dehydrogenase, LDH), amino acid (glutamate dehydrogenase, GLDH), and fat (α -glycerophosphate dehydrogenase, α -GPDH) metabolism, as well as the enzyme of complex II of mitochondrial respiratory chain (succinate dehydrogenase, SDH) were evaluated.

We also measured blood lactate level before and after a carbohydrate loading.

Results: MRI of the limb muscles was performed in 6 patients with myotonic dystrophy. In all patients, the medial head of the gastrocnemius muscle was affected. From 2 to 4 points. In 5 out of 6 patients, the soleus muscles were involved in the pathological process. In 4 patients, a lesion of the intermediate broad thigh muscle, the medial broad thigh muscle, the anterior tibial muscle, the long extensor of the thumb, the long extensor of the toes of the foot, the long fibular muscle, the short calf muscle, and the lateral head of the gastrocnemius muscle were noted. MRI of the brain was investigated in 5 patients. In 4 patients revealed the presence of subcortical foci in the white matter of the brain.

The mean values of the activity of mitochondrial enzymes LDH, α -GPDH, GDH and LDH were statistically significantly reduced ($p < 0.05$). The decrease in the activity of mitochondrial enzymes LDH and α -GPDH was in all the examined patients. The activity of these enzymes was within the limits of normal values in one patient, and LDH - in 7.

Blood lactate before and after meals exceeded normal values (2.2 mmol / l). The median indicator of blood lactate before the meal was 2.8, after the meal - 3.2. The difference is not statistically significant.

Conclusions: Thus, in patients with myotonic dystrophy, not only damage to the muscles of the extremities was revealed, but also damage to the white matter of the brain, probably due to pronounced mitochondrial disorders in these patients. The revealed changes are indications for the appointment of energy-tropic therapy - carnitine and carnosine preparations.