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# Electromyoneurography in diagnostic procedures of movement disorders

## Abstract

Electromyoneurography (EMNG) examination is used to diagnose pathology of lower motor neuron, peripheral nerve, neuromuscular junction and muscle. Movement disorders are group of neurological diseases caused with pathology in basal ganglia, thalamus and cerebellum. This paper is a review of movement disorders accompained with periferal nerve or muscle involvement where electromyoneurography should be used in diagnostic procedures. Patients with non-Huntington disease chorea need to be evaluated for neuropathy and myopathy. Movement disorders accompayned with ataxia should also be checked for neuropathy. Diagnostic criteria for stiff person and stiff limb syndrome include electromyoneurography finding of continous MUAP in paravertebral muscles and involved limbs. Finding of interictal myokimia in episodic ataxia/chorea/dystonia serves as a diagnostic marker. Although electromyoneurography is a diagnostic tool for peripheral nerve and muscle disorders, it has a significant role in diagnostic procedures of movement disorders.

**Keywords:** Movement disorders, Electromyoneurography, Neuropathy, Myopathy

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## Manuscript

Electromyoneurography (EMNG) examination is used to diagnose pathology of lower motor neuron, peripheral nerve, neuromuscular junction and muscle. Myography analyze muscle unit action potential (MUAP) discharges during activation and spontaneuos activity while resting. Neurography serves to analyse sensory and motor nerve conduction velocity, distal latence, F wave latence, and amplitude of the nerve potential.[1] Movement disorders are group of neurological diseases presented with hypo- or hyperkinetik movements. Parkinsonism present hypokinetik, while dystonia, chorea, athetosis, balism, ticks and myoclonus present hyperkinetik movements. These disorders appear due to pathology in basal ganglia, thalamus and cerebellum.[2]

This paper is a review of movement disorders accompained with periferal nerve or muscle involvement where electromyoneurography should be used in diagnostic procedures.

Chorea-Acanthocytosis is rear autosomal recesive disorder caused by mutations in the VPS13A gene. This gene encodes for a protein called chorein. It could be presented with the different types of movement disorders (dysonia, chorea, tics and parkinsonism). Dystonia is common and most prominent on oral region. Tipicaly, patients have tongue protrusion dystonia, causing dysarthria and serious dysphagia. Habitual tongue and lip biting are one of the characteristic signs. Beside movement disorder this patients have behavioral and cognitive changes and myopathia. In laboratory work up there are acanthocytes in blood smear and elevated kreatin kinase level. With EMNG myopathic pattern and sensorimotor axonal neuropathy could be detected.[2,3] McLeod syndrome is also autosomal recesive disorder caused by mutation in XK gene. It is presented with movement disorders (chorea, dystonia and tics), cognitive, behabioral changings, myopathy and haemolytic anemia with acanthocytosis. Patiens have absent expression of the Kx erythrocyte antigen and weakened expression of Kell blood group antigens. On EMNG there are sensorimotor axonal neuropathy and myopathy.[2,4]

Spinocerebellar ataxias (SCAs) are a clinically heterogeneous group of disorders primarily presented with ataxia with or without some movement disorders. Some of them has distinguishing clinical features and neurophysiological findings that are compatible with a dying-back axonopathy and/or a neuronopathy. This clinical specificity could help in diagnostic procedures. The hereditary ataxias can be subdivided by mode of inheritance (i.e., autosomal dominant, autosomal recessive, X-linked, and mitochondrial). Among autosomal dominant form there are SCA 1, 2, 3, 25 and episodic ataxia type 1 with movement disorders, peripheral nerve and muscle involvement. SCA-1 is neurododegenerative disorders caused by mutation on gene located on the short arm of chromosome 6. Due to mutation there is pathological ataxin 1 protein. Tipically, patients have ataxia with pyramidal signs and axonal sensorimotor neuropathy. Parkinsonism and dystonia could be also presented.[5-7] In SCA-2 gene locus is mapped on chromosome 12. These gene encode for protein ataxin 2. In this SCA parkinsonism could be rarely presented, while tipicaly patiens has slow saccadic eye movements, decreased tendon reflexes and demenia with axonal sensorimotor neuropathy.[5,6,8] Patients with SCA-3 carry an expanded CAG repeat in the MJDl gene mapped to chromosome 14q32 coding for ataxin 3 protein. It is more common in Portugal, where it was originally described in Portuguese families from the Azores and called Machado-Joseph disease. Dystonia, restless leg syndrome, facial-lingual fasciculation-like movements or myokymia, pyramidal signs, lid retraction nystagmus, decreased saccade velocity, amyotrophy and sensory loss are characteristic of this disorder. Neurography reveale axonal sensorimotor neuropathy.[5,6,9] In a large French family with SCA, Stevanin et al found linkage of the disease locus, designated SCA25, to a 12.6-cM region of chromosome. Facial ticks and myokimia are one of the presenting signs together with the typical finding of sensory neuropathy.[10] In episodic ataxia type 1 (EA1) there is mutation in protein for alpha subunit of sodium channal on KCNA1 gene. Patients can have short attacks of ataxia, chorea or dystonia that last from few seconds to few minutes. It could be provoked as startl phenomen or with movement. One of the diagnostic criteria is finding of interictal myokimia with myography.[2,11,12]

Among autosomal recesive ataxias there are ataxia with oculomotor apraxia type 1, ataxia-oculomotor apraxia type 4, ataxia teleangiectasia, infantile-onset spinocerebellar ataxia (IOSCA) and cerebrotendinous xanthomatosis accompained with neuropathy and some type of movement disorders. Ataxia with oculomotor apraxia type 1 has mutation in aprataxin protein due to involvement of APTX gene. This ataxia is characterized by childhood onset of slowly progressive cerebellar ataxia, followed by oculomotor apraxia, mild intellectual disability, hypoalbuminemia and a severe primary motor peripheral axonal motor neuropathy. Choreathetosis and upper-limb dystonia are common in clinical presentation.[2,11,13] Ataxia-oculomotor apraxia type 4 is is caused by homozygous or compound heterozygous mutation in the PNKP gene on chromosome 19q13. It is characterized by onset of dystonia and ataxia in the first decade. Additional features include oculomotor apraxia and peripheral neuropathy.[11,14] Ataxia-teleangiectasia developed due to mutation in ATM gene. It is also known as Louis-Bar syndrome and could be present in classic or non classic form. Classic form, usualy beginning between age two to four, tipicaly present with teleangiectasies of the conjuctive, frequently infections, increased risk for malignancy and choreatethosis. Non classic form beginning as adult-onset with early onset dystonia. On neurography patients have demyelinating sensorimotor neuropathy.[2,11,15] Mutations in the C10orf2 gene cause infantile-onset spinocerebellar ataxia (IOSCA). It is a severe, progressive neurodegenerative disorder characterized by optic atrophy, deafness, ophtalmoplegia, athetosis and sensory neuropathy.[2,11,16] Cerebrotendionous xanthomatosis a type of lipide storage disease. It is caused by mutations in the CYP27A1 gene that provides instructions for producing an enzyme called sterol 27-hydroxylase. This enzyme is important in the pathway that breaks down cholesterol to form acids used in the digestion of fats (bile acids). It is tipicaly characterized by infantile-onset diarrhea, childhood-onset cataract, adolescent- to young adult-onset tendon xanthomas, and adult-onset progressive cognitive decline with dystonia, myoclonus and sensorimotor neuropathy.[2,11,17]

X-linked spinocerebellar ataxia-1 (SCAX1) is caused by mutation in the ATP2B3 gene on chromosome Xq28. Patients may presents with tremor, parkinsonism and sensory neuropathy.[2,18]

Stiff person syndrome is autoimmune disorder presented with prominent rigidity in trunk musle and painfull spasm. Disorder has progressive character with involvement of proximal part of legs and arms and development of gait disturbances. A patient had fixed hyperlordosis due to rigidity of paravertebral muscle.

One of the diagnostic criteria for stiff person syndrome is finding of continuated muscle unit action potential (MUAP) in paravertebral muscles. If the disease starts and is more prominent in limbs, then it should be diagnosed ad stiff limb syndrome and checked if continuated MUAP is present in involved limbs.[19,20]

## Conclusion

Although electromyoneurography is a diagnostic tool for peripheral nerve and muscle disorders, it has a significant role in diagnostic procedures of movement disorders.

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## Conflicts of interest

None.

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