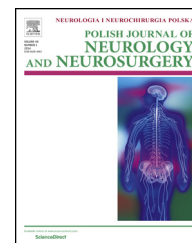


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Case report

Excessive daytime sleepiness in a patient with coexisting myotonic dystrophy type 1, myasthenia gravis and Graves' disease



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ARTICLE INFO

Article history:

Received 3 August 2016

Accepted 19 January 2017

Available online 3 February 2017

Keywords:

Myotonic dystrophy

Myasthenia gravis

Graves' disease

Hypocretin

ABSTRACT

A 41-year-old female with history of Graves' disease, bilateral cataract, paroxysmal atrial fibrillation was admitted because of muscle weakness, daytime sleepiness, fatigability, drowsiness, bilateral eyelid ptosis, descending of head and lower jaw. On neurological examination the patient was presented with muscle weakness, muscle atrophy (in face and sternocleidomastoid muscles), features of myotonia and apocamnosis (orbicular muscles). Electromyography revealed myopathic changes, myotonic and pseudomyotonic discharges, positive repetitive nerve stimulation test in proximal muscles. Myotonic dystrophy (MD) diagnosis was confirmed by genetic testing and myasthenia gravis (MG) by a positive titer of cholinergic receptor autoantibodies. In the CSF concentration of hypocretin was significantly decreased.

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1. Introduction

Myasthenia gravis (MG) and myotonic dystrophy (MD) lead to a dysfunction of motor unit, although they both differ clinically and pathogenetically. Coincidence of MG and MD in one patient is extremely rare [1–6].

Hyperthyroidism may also lead to muscle dysfunction [7,8]. Though endocrine disorders like hypogonadism or glucose intolerance are typical for MD patients, hyperthyroidism is rarely reported in these subjects [7,8]. However in MG, Graves'

disease, which is another autoimmunological disorder, is diagnosed more often than in general population [9].

To the best of our knowledge, coexistence of MD, MG and Graves' disease in one patient has never been reported and it is the first case report describing such a patient.

2. Case report

A 41-year-old female farmer with one-year history of Graves' disease was referred to the Department of Neurology with a

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suspicion of MG. She complained of bilateral eyelid ptosis, decreasing of her head and a lower jaw, and reported also muscle weakness, fatigability and drowsiness making walking long distances impossible. The patient had a history of bilateral cataract (already treated surgically in her both eyes) and biliary calculosis. She complained of cardiac arrhythmia (diagnosed as paroxysmal atrial fibrillation) and frequent respiratory tract infections. She had a history of respiratory insufficiency in the postoperative period after appendectomy. There had been cases of cataract and muscle weakness in the patient's family that were reported by her but not extensively diagnosed: her uncle and aunt had difficulties with walking. They died in their middle age of an unknown reason, leaving no descendants. Our patient's mother was operated on bilateral cataract at the age of 50. She did not complain of any muscle weakness and she died at the age of 76 because of a neoplasm, which might have been a case of a mild type of MD with a leading symptom of bilateral cataract, as it is known that myotonia and muscle dystrophy are not always present in that type of MD [10,11].

Our patient has three children and two of them (a 13-year-old son and 21-year-old daughter) do not report any health problems. Neurologic examination did not revealed any abnormalities. Her 17-year-old son is moderately mentally impaired with a signs of myotonia seen on neurological examination. His EMG examination showed a presence of a myopathic process and myotonic and pseudomyotonic discharges in tibialis anterior and extensor digitorum muscles. Decrement test from both proximal and distal muscles was normal. We diagnosed him with a childhood type of MD, where learning difficulties are the early sign followed by myotonia and muscle atrophy [10,11].

On admittance, the patient was presented with generalized muscle weakness, muscle atrophy – especially in her face and sternocleidomastoid muscles and features of myotonia (difficulty with grip release). There was apocamnosis seen in orbicular muscles of her eyes, but it was difficult to evaluate in hand muscles. She was hyperthyreotic. Her twenty-four hour glycaemic profile, cortisol secretion and synacthen test were normal.

Electromyography revealed myopathic changes (low-amplitude, short-duration, polyphasic motor unit potentials) with significant spontaneous activity consisting of both myotonic and pseudomyotonic discharges (Fig. 1a and b). Repetitive nerve stimulation was negative in distal hand muscles but positive (resulting in significant decrement in successively evoked muscle action potentials) in proximal deltoid muscles (Fig. 2). An edrophonium test was clinically and electrophysiologically positive. Her mediastinum CT did not reveal any changes coherent with a persistent thymus and/or a thymoma.

MD diagnosis was confirmed by genetic testing performed at Department of Human Genetics, University of Ulm, Germany which revealed mean number of 735 (ranging from 535 to 935) CTG triplet copies in locus 13.2 of chromosome 19q. MG diagnosis was confirmed by a positive titer of ACh receptor autoantibodies – 4.03 nmol/l (normal range – below 0.25 nmol/l). Based on her medical history, neurological and electrophysiological examination the patient was diagnosed with Graves' disease, MD and MG.

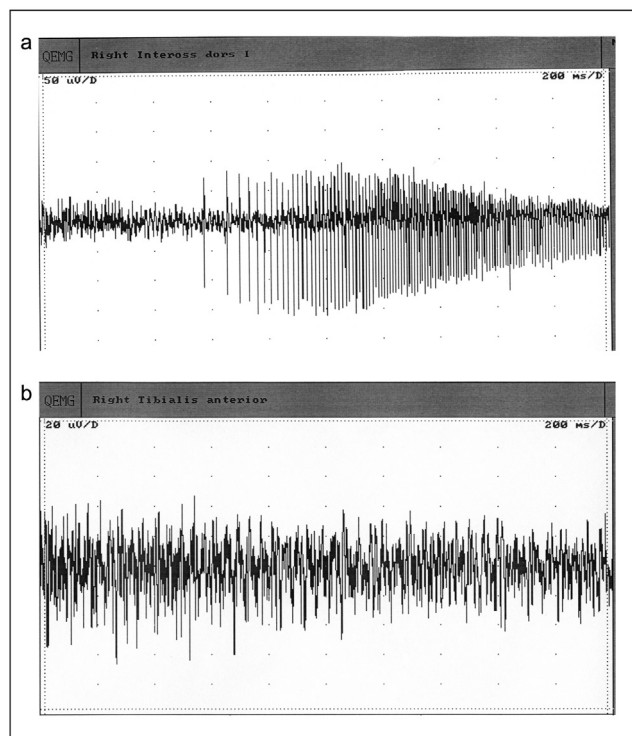


Fig. 1 – (a) Myotonic discharge seen in patient's right first dorsal interosseous muscle. (b) Pseudomyotonic discharge seen in patient's right anterior tibial muscle.

Because of an excessive daytime sleepiness (15 points Epworth scale [12]), CSF/serum analysis was done, showing unaltered total protein concentration normal cell count as well as albumin, IgG, IgA, and IgM CSF/serum concentration quotients. Isoelectrofocusing revealed identical bands pattern, confirming lack of intrathecal IgG synthesis. Nevertheless, decreased concentration of hypocretin concentration (80 pg/ml; normal range >100 pg/ml) was found, which may be connected with excessive daytime sleepiness present in our patient.

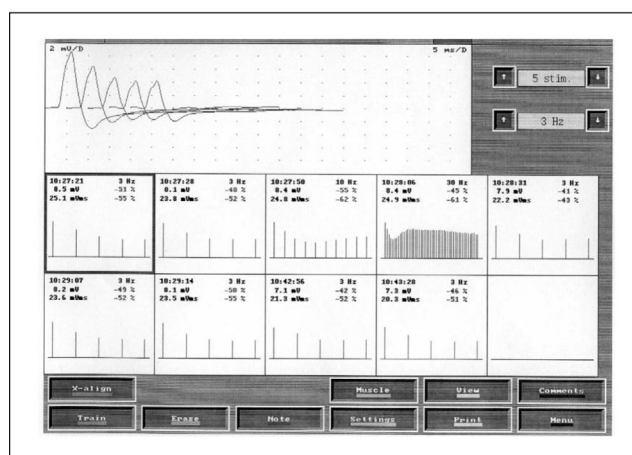


Fig. 2 – Decrement seen on repetitive nerve stimulation test in the patient's right deltoid muscle.

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