

Diaphragm-Pacing Stimulation System in McArdle Disease: A New Therapeutic Option

McArdle Kas Hastalığında Diyafragma Pil Uyarı Sistemi: Yeni Bir Tedavi Seçeneği
Anesteziyoloji ve Reanimasyon

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Özet

McArdle nadir görülen bir kas hastalığıdır, yaklaşık 1/100000 oranında görülür. Tip 5 glikojen depo hastalığı olarak da bilinir. Otozomal resesif olarak kalıtılır ve myofosforilaz C enziminin eksikliğinden veya fonksiyon görmemesinden kaynaklanır. 40 yaşında bayan hasta, ateş ve dispne semptomlarıyla acil servise başvurmuş. Toraks bilgisayarlı tomografi (BT)'de lobar pnomoni saptanmıştır. Nörolojik muayenede dört ekstremitenin proksimal kaslarında ılımlı güçsüzlük tesbit edilmiştir. Kas biyopsisi McArdle olarak rapor edilmiştir. Biz burada, McArdle tanısıyla izlediğimiz olguda, diyafragma pil uyarı sistemi (diaphragm-pacing stimulator system (DPSS))'ni uygulamayı planladık. Bu olgu, literatürde McArdle tanısı nedeniyle DPSS uygulanacak olan ilk olgudur.

Anahtar kelimeler: *Diaphragm pil uyarı sistemi, Mekanik ventilasyon McArdle*

Abstract

McArdle disease is a rare skeletal muscle disorder affecting approximately 1 in 100000 people. It is also known as type V glycogen storage disease. It is inherited in an autosomal recessive pattern, caused by a missing or nonfunctioning enzyme called myophosphorylase C. We described a 40-year-old female who was admitted to emergency department with fever and dispnea. Torax BT reported lobar pneumonia. Neurological examination showed slightly weakness in proximal muscles of four extremities. Muscle biopsy specimen revealed McArdle disease. We report planning of an application of DPSS in a McArdle patient. This is the first case in the literature for application of DPSS in McArdle disease.

Keywords: *Diaphragm-pacing stimulation system, Mechanic ventilation McArdle*

Introduction

McArdle, first described the disorder in 1951 in a 30 year old man with lifelong intolerance to exercise^{1,2}. McArdle disease is a rare skeletal muscle disorder affecting approximately 1 in 100000 people. It is also known as type V glycogen storage disease. It is inherited in an autosomal recessive pattern, caused by a missing or nonfunctioning enzyme called myophosphorylase C³⁻⁵. The patients typically show symptoms of myophosphorylase deficiency with exercise intolerance such as early fatigability, myalgia, contractures and sometimes myoglobinuria induced by exertion and second-wind phenomenon³⁻⁶. Persistent and progressive muscle weakness and atrophy may occur in aged patients⁴.

Evolution with rhabdomyolysis may occur and can be complicated with acute kidney injury but rarely, in about 11% of cases, is the initial disease manifestation⁷. No effective treatment for McArdle disease exists. Vitamin B6 supplementation can enhance residual phosphorylase activity and improve insufficient anaerobic glycolysis of skeletal muscle⁸. Other potential therapies that have been tried in patients with McArdle disease include manipulation of diet, moderate aerobic exercise, and supplementation with creatine⁴. Applying diaphragm-pacing stimulation system (DPSS) can return the effectiveness of type 1 fibers which are worsened by respiratory muscle paralysis and atrophic diaphragm by mechanical ventilation. We report our planning of an application

of DPSS in a McArdle patient.

Case Report

A 40-year-old female (Weight 50 kg, height 170 cm) had difficult in walking for fifteen years. She experienced easy fatigability with walking and fall last two years. None of her family members had such symptoms. She was admitted to emergency department with fever and dispnea. Torax BT reported lobar pneumonia. She developed respiratory distress and entubated in emergency department. The she was transferred to intensive care unit (ICU) for close monitoring and treatment. On the 10th day of ICU, tracheostomy was applied due to the continued need for mechanical ventilation. She followed 35 days in ICU.

The patient was transferred to our intensive care unit from another center, for further examination and treatment because of weaning failure. She was conscious. Neurological examination showed slightly weakness in proximal muscles of four extremities. Muscle strenght in extremites were 3/5 in proximal lower extremity, 4/5 in proximal upper extremity and 3/5 in knee extension.

She was consulted by neurology doctors on the 1th day of the ICU stay, muscle biopsy was taken from the biceps brachii on the 60th day of the ICU stay. Periodic acid Schiff (PAS) staining of the muscle biopsy specimen revealed an increased amount of subsarcolemmal glycogen. Genetic analysis was not preferred fo her.

After the diagnosis was made, we started to treat her with pyridoxine hydrochloride (Vitamin B6) supplemet (90mg/day). About three mounths later, she was aware of an improvement of muscle fatigability. Significant improvement was not detected accept prolonged separation times from mechanical ventilation. She was consulted by Thoracic Surgeon and application of DPSS was planned. This is the first case in the literature for application of DPSS in McArdle disease.

Discussion

We described a patient with McArdle disease whom we planned to apply DPSS. Four characteristic clinical features are important for initial suspicion of McArdle disease. These are 1. Exercise intolerance such as easy fatigability, muscle cramps; 2. Elevation of serum CK levels in the absence of exercise; 3. About half of the patients have experienced hyperCKemia or myoglobinuriaafter intensive exerciseand 4. Second wind phenomenon^{4,5}.

Diaphragm pacing is conducted with low frequency electrical stimulation at a slow repetition (respiratory) rate to condition the diaphragm muscle against fatigue and maintain it fatigue-free. Candidates for diaphragm pacing are those with ventilator insufficiency due to malfunction of the respiratory control center in the brain stem (central alveolar hypoventilation) or interruption of the upper motor neurons of the phrenic nerve⁹. The use of DPSS has changed the medical outcome of patients who have long-term dependence on mechanical ventilation¹⁰. Application of DPSS increase the quality of sleep in patients¹¹.

Şanlı et al¹¹ reported DPSS aplication in a tetraplegic patient; Son et al⁹ reported DPSS aplication in a quadriplegic patient. Onders et al¹² reported their trial on the use of diaphragm pacing in patients with unilateral or bilateral diaphragm dysfunction; Onders et al¹³ reported their trial on the use of diaphragm pacing in amyotrophic lateral sclerosis, Tedde et al¹⁰ and Posluszny et al¹⁴ reported their experience in diaphragm pacing in spinal cord injury. Although the DPSS was applied due to different indications, this will be first planned application of DPSS for diagnosis with McArdle.

The implantation of a DPSS is a safe and efficient procedure to improve the quality of life of patients who are dependent to mechanical ventilation (13). This is the first report of DPSS will be used to treat the McArdle disease . This success suggests a potential for a wider use of DPSS and areas for future research.

References

1. DykenML, Smith DM, Peake RL. An electromyographic diagnostic screening test in McArdle's disease and a case report. *Neurology*. 1967;17:1-5
2. Felice KJ, Schneebaum AB, Jones HR. McArdle's disease with late-onset symptoms: case report and review of the literature.*Neurology, Neurosurgery, Psychiatry*. 1992;55:407-8
3. Choleva AJ. Anesthesia considerations in a patient with McArdle disease: a case report. *AANA J*. 2011;79(3): 243-7
4. Izumi R, et al. A case of McArdle disease: Efficacy of Vitamin B6 on Fatigability and Impaired Glycogenolysis. *Inter Med*. . 2010;49:1623-5.
5. Park HJ, et al. The significance of clinical and laboratory features in the diagnosis of glycogen storage disease Type V: A case report. *J Korean Med Sci*. 2014; 29: 1021-4.
6. Witting N, Duno M, Piraud M, Vissing J. Severe axial myopathy in McArdle disease. *JAMA Neurol*. 2014; 71(1): 88-90.
7. Costa R, et al. McArdle disease presenting with rhabdomyolysis and acute kidney injury. *Acta Med Port*. 2013;26(4):463-6.
8. Sato S, Ohi T, Nishio I, Sugie H. Confirmation of the efficacy of vitamin B6 supplementation for McArdle disease by folow-up muscle biopsy. *Muscle Nerve*. 2012;45(3): 436-40.
9. Son B, Kim D, Kim I, Hong JT. Phrenic nerve stimulation for diaphragm pacing in a quadriplegic patient. *J Korean Neurosurg Soc*. 2013; 54: 359-62.
10. Tedde ML, et al. Diaphragmatic pacing stimulation in spinal cord injury: anesthetic and perioperative management. *Clinics*. 2012;67(11):1265-1269
11. Şanlı A, et al. Diaphragm-pacing stimulation system in a tetraplegic patient: a case report and the first Turkish experience. *Türk Göğüs Kalp Damar*. 2014;22(2):439-42.
12. Onders RP, et al. Extended use of diaphragm pacing in patients with unilateral or bilateral diaphragm dysfunction: A new therapeutic option. *Surgery*. 2014;156(4):776-86.
13. Onders RP, et al. Final analysis of the pilot trial of diaphragm pacing in amyotrophic lateral sclerosis with long-term follow-up: diaphragm pacing positively affects diaphragm respiration. *Am J Surg*. 2014; 207(3): 393-7.
14. Posluszny JA, et al. Multicenter review of diaphragm pacing in spinal cord injury:successful not only in weaning from ventilators but also in bridging to independent respiration. *J Trauma Acute Care Surg*. 2014; 76(2): 303-9.