Paramyotonia Congenita and Pregnancy: A Case Report

PARAMYOTONİ KONJENİTA VE GEBELİK: OLGU SUNUMU

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Summary

Objective: To review of the delivery form in a pregnant who has paramyotonia congenita that is a rare state.

Institution: Trakya University Faculty of Medicine, Department of Obstetrics and Gynecology.

Case Report: A 30-year-old pregnant with paramyotonia congenita was admitted to our clinic at 28 weeks'gestation. She was difficulty delivered from vaginal route the five years ago. Her first child had the cerebral palsy. The cesarean section under the epidural anaesthesia was performed, after the patient had been closely followed by the delivery.

Conclusion: We suggest that the paramyotonia congenita is a rare state and the cesarean section under the regional anaesthesia may be preferred to such pregnants, after we review the literature.

Key Words: Paramyotonia congenita, Pregnancy, Regional anaesthesia

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Özet

Amaç: Paramyotoni konjenita tanılı bir gebede doğum şeklinin gözden geçirilmesi.

Çalışmanın Yapıldığı Yer: Trakya Üniversitesi Tıp Fakültesi Kadın Hastalıkları ve Doğum ABD

Olgu Sunumu: Paramyotoni konjenita tanısı alan 30 yaşındaki bir gebe gebeliğinin 28. haftasında kliniğimize başvurdu. O beş yıl önce zor bir vajinal doğum yapmıştı ve ilk çocuğu serebral palsiye sahipti. Hasta doğuma kadar yakın izlendikten sonra epidural anestezi altında sezaryen uygulandı.

Sonuç: Literatürü gözden geçirdikten sonra paramyotoni konjenita'nın nadir bir durum olduğunu ve böyle gebelere rejyonel anestezi altında sezaryen uygulanmasının tercih edilebileceğini öne sürüyoruz.

Anahtar Kelimeler: Paramyotoni konjenita, Gebelik, Rejyonel anestezi

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Paramyotonia congenita (PMC) is a rare, autosomal dominant transmitted congenital disorder (1). It is one of disorders that are collectively called the myotonic dystrophies (2). This disease is connected with paradoxical myotonia. That is, it develops during exercise and worsens as the exercise continues. A widespread myotonia, often coupled with weakness, is induced by exposure to cold (2-3). Paramyotonia congenita represents one of three major variants of hyperkalaemic periodic paralysis. The genetic defect in PMC maps to the same gene locus (SCN4A) on chromosome 17 q as the other hyperkalaemic periodic paralyses (3).

The physiological cause of the paradoxical myotonia is thought to be a defect of the sodium/potassium pump in the sarcolemma and ATPase systems. Exposure to cold produces spontaneous depolarisation of the sarcolemma and contraction of the myofibrils with a very slow relaxation phase. Delayed muscle relaxation after contraction is most evident in skeletal muscle, but some authors consider that smooth muscle may also be involved (2).

Pregnancy-related complications have been reported with myotonic dystrophy (2). These include spontan abortus, hydrops noticeable by difficulty in swallowing, preterm delivery, prolonged labor and postpartum hemorrhage (2,4-7).

Numerous drug therapies have been suggested to control the myotonia. The best current treatment option is avoid the precipitants. The management of a myotonic crisis should include urgent measurement of the serum potassium, sufficiently warm, lower muscle stimulation. If this is inadequate, procainamide, tocainide, mexiletine or other class Ia anti-arrhytmics may be used (3).

We report a pregnant patient with paramyotonia congenita that is a rare state and the approach at the moment delivery.

Case Report

A 30-year-old gravida 2, para 1, woman was admitted at 28 week's gestation. She was known to have paramyotonia congenita with a strong family history of the disease through two generations (her father, two brothers, her uncles and her aunt have paramyotonia congenita). There was point mutation in sodium channel in her DNA analysis that was performed by Ulm University in Germany.

The patient had a history of developing acute cramping of skeletal muscles, especially of the face and extremities, with exposure to cold. Mexiletin had been admitted to patient by neurolog but she hadn't used because of becoming side effects of the drug. She had normally serum creatinine phosphokinase and potassium levels. Electromyographic recordings typically showed shortlasting myotonic runs of spontaneous activity.

She had a difficult vaginal delivery of a male with cerebral palsy the five years ago. This pregnancy progressed normally until the onset of labor (40 week's gestation). We performed tok the elective cesarean section under the epidural anaesthesia using %0.5 bupivakain because of the difficult vaginal delivery previously. The Apgar scores of newborn were 8 and 9 at 1 and 5 minutes, respectively and he had 3650 gram.

Comment

Paramyotonia congenita is a rare, myotonic disorder first described by Eulenberg in 1886. The most typical clinical presentation in paramyotonia congenita is the precipitation of active myotonia by exposure to cold temperatures and prolonged attacks of muscle weakness and stiffness, especially after continued muscle activity (1). The pregnancy, emotional stress, cold and the certain anaesthetics (halothane, succinylcholine) are also precipitating factors (1,4).

During labor, myotonic damage can lead to many problems. Intrapartum complications may occur in all stages of labor. The first stage may be prolonged, most probably due to inefficient uterine contractions, but may also be rapid, due to functional incompetence of uterine muscle caused by myotonia. Maternal fatigue will contribute to prolongation of the second stage, frequently requiring assisted delivery i.e., vacum or forcipal extraction or even cesarean section. The third stage of labor may be complicated by placental retention and postpartum hemorrhage (4-5).

If a cesarean section is required, the anaesthesiological problems may occur (3-4,8-9). Intubation may be difficult, as muscular relaxation is not easily achieved, because of the increase of myotonia induced by the certain anaesthetics. The depolarizing muscle relaxants are contraindicated as they may trigger a myotonic episode, serious cardiac abnormalities (arrythmias and conductance problems) and pulmonary complications (postoperative apnea and aspiration pneumonia) (4,8). Thus the short-acting non-depolarizing agents such as atracurium and vecuronium are preferable (8). Marked respiratory depression may also follow barbiturate administration (4,8). Inhalational agents, such as halothane may be contraindicated, because of the postoperative shivering (8). Also for these reasons, regional anaesthesia may be preferred (2-5,7,9).

We report a pregnant woman with paramyotonia congenita whose pregnancy had progressed normally until the onset of labor. She had a difficult vaginal delivery of a male with cerebral palsy the five years ago. This state may be possible a result of prematurity or a difficult vaginal delivery (e.g, instrumental extraction, birth trauma, etc) that are the obstetrical complications of the PMC. Thus, we preferred to the elective cesarean section under the epidural anaesthesia using bupivakain that has a long effect, after we reviewed the literature.

In summary, the management or delivery of these pregnants require the attention. The serum potassium levels should be measured and the required precautions should be taken against loosing body heat, such as use of a warming mattress, warm intravenous fluids and humidification of anaesthetic gases, since cold can precipitate myotonia the prepartum or preoperative. The body and operating environment temperatures must be closely monitored. When the anaesthesia is required during labor, regional rather than general anaesthesia is preferable because of the dangers of general anaesthetics. The short-acting non-depolarizing agents such as atracurium and vecuronium should be preferred, if the general anaesthesia is selected.

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