The Association of Myotonia Congenita and Ankylosing Spondylitis

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ABSTRACT

This is the first report about the association of ankylosing spondylitis and myotonia congenita. Ankylosing spondylitis is a systemic rheumatologic disease that is characterized by axial skeletal inflammation and accompanied by systemic involvement. The most significant findings of myotonia are the stiffness and the delayed relaxation following the muscle contraction. Both of these pathologies can cause stiffness and also delaying of diagnosis of each other.

Key words: *Ankylosing spondylitis, myotonia congenita, stiffness*

Ankilozan Spondilit ve Myotoni Konjennita İlişkisi

ÖZET

Bu vaka ankilozan spondilit ve myotoni konjenitanın birlikte görüldüğü ilk sunumdur. Ankilozan spondilit aksiyal iskelet inflamasyonu ve sistemik tutulumla karekterize sistemik romatolojik bir hastalıktır. Miyotoninin en belirgin bulgusu tutukluk ve kas kasıldıktan sonra gevşeme gecikmesidir. Tutukluk ve tanı gecikmesi bu iki hastalıkta ortak bulgudur.

Anahtar kelimeler: *Ankilozan spondilit, myotoni konjenita, tutukluk*

INTRODUCTION

Ankylosing spondylitis (AS) is a systemic rheumatologic disease that is characterized by axial skeletal inflammation and accompanied by systemic involvement. Some of the systemic patterns include the uveitis, iritis, aortitis, cardiac conduction disorders and t he pulmonary fibrosis (1). There exist two types of myotonias as dystrophic and non-dystrophic. Dystrophic myotonia manifests with myotonic symptoms and muscle atrophy or dystrophic changes. Due to the different clinical manifestations, the neurologists have classified the non-dystrophic myotonia into two categories as recessive and dominant myotonia congenita. Recessive myotonia congenita was first described by Becker in 1966. In this form of the disease, also known as the Becker Syndrome, the symptoms have an onset at the age of 4 to 12, frequently in the lower extremity and the history usually includes falling episodes (2). The dominant myotonia congenita is known as the Thomsen disease. In this form, muscle hypertrophy is predominant and no transient muscle weakness occurs (3).

In AS, loss of muscle mass also occurs in addition to the other extraarticular involvements. This results from the lesions including the nerve root compression and the fact that the muscles are used less in this disease due to inactivity (4). However, a clinical course primarily involving muscle involvement has not been described in patients with AS. No case with concomitant AS and myotonia congenita has been reported to date. We reported a young female patient diagnosed with myotonia congenita and concomitant AS.

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CASE

The 19-year-old female patient presented to our hospital with the complaints of low back pain and hip pain that have been present for the last 6 months and have aggravated for the last one month. Based on the history, the patient was detected to have an inflammatory type lumbar pain that responsed to nonsteroidal anti inflammatory medicine. The patient had nocturnal pain and the morning stiffness lasted for an hour. She had no history of neck, back pain, arthritis of the peripheral joints, psoriasis, genital discharge, constipation or diarrhea. Her medical history revealed that she had been diagnosed with myotonia congenita when she was 9 years old. The patient's query demonstrated that while she exhibited a normal neurological development in her childhood, she started to experience various complaints including slow walking, difficulty in climbing stairs, falling, and stiffness when she was 7 years old. Her stiffness had been increasing particularly after inactivity. When she was 9 years old, she presented to the hospital upon starting to experience more frequent falling episodes; and she was diagnosed with recessive myotonia congenita by the neurologist based on the electromyography (EMG) performed. EMG demonstrated myotonic discharges. She started treatme nt with carbamazapine 200-400 mg per day. She used the medication regularly and experienced no drug-associated side effects. Her frequency of falling decreased and she started climbing the stairs more easily. However she had no change in her complaint of stiffness. She is currently using her medication. She says that she had difficulty in starting her activities due to stiffness and that she could perform them more easily after warm-up (warm-up phenomenon). Her stiffness further increased upon exposure to cold. She had no complaints of cramps. Her physical examination showed that she had unlimited and painless joint motion in the neck. The chest expansion was 3 cm. The lumbar examination showed unlimited painful joint motion. The Schober test revealed a result of 14.7 cm, the modified Schober test revealed a result of 21 cm and the hand-finger floor distance was 0 cm. The sacroiliac compression tests revealed bilateral positive results. The hip examination was normal. No sensitivity was detected on the enthesis points. The neurological examination detected no atrophy and hypertrophy in the upper and lower extremity. Her muscle strength was complete; the deep tendon reflexes were hypoactive; there was no percussion myotonia. The laboratory results showed a normal blood count (white blood cell 9,8/mm3, hemoglobin 12 g/dl, platelet 245.000/mm3), erythrocyte sedimentation rate of 20 mm/h and C- reactive protein (CRP) of 14,8 mg/l (normal: ≤ 5 mg). The biochemical investigation demonstrated urea, creatinine, SGOT, SGPT, calcium, magnesium, creatinine phosphokinase, lactate dehydrogenase and thyroid-stimulating hormone values that were within the normal limits. Brucella and hepatitis markers were negative. Antinuclear antibody was negative; HLA-B27 was positive.

Lumbar lateral graphy showed square shaping of the ver-

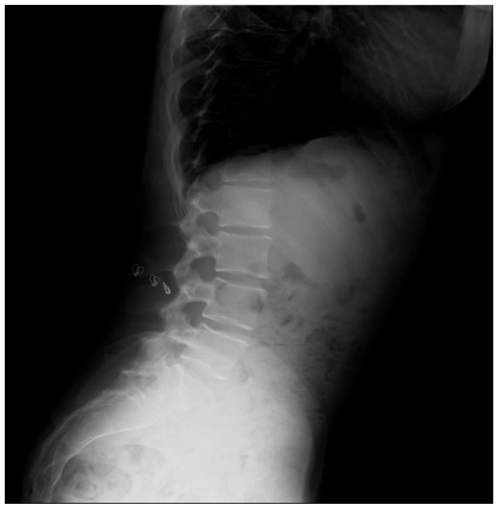

***Figure 1**. Lumbar lateral graphy showed square shaping of the vertebrae and shining corners*

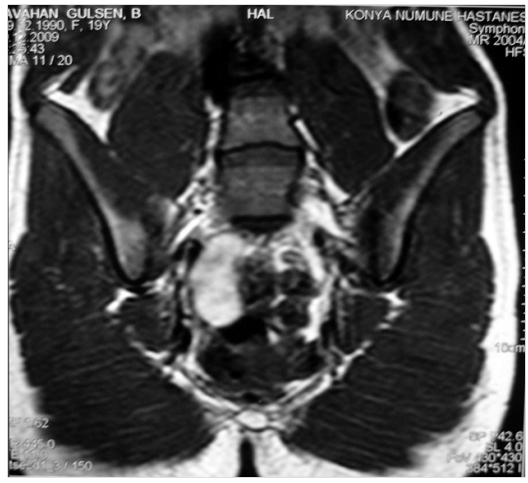

***Figure 2**. The sacroiliac MRI investigation detected decreased signal intensity in T1 series in patches consistent with edema in both sacroiliac joint*

tebrae and shining corners (Figure 1). The lumbar magnetic resonance imaging (MRI) revealed normal values. The sacroiliac MRI investigation detected decreased signal intensity in T1 series in patches consistent with edema in both sacroiliac joints (Figure 2). The patient was diagnosed with AS based on the Modified New York Criteria in addition to the established diagnosis of myotonia congenita (5). The Bath AS disease activity index (BASDAI) was 2.2 and the Bath AS functional index (BASFI) was 2.7.

The patient was admitted to our clinic to receive an exercise program for treating AS. The patient was administered hydrotherapy and conventional transcutaneous electrical nerve stimulation (TENS) and intermittent ultrason for 10 sessions (2.5 w/cm). In addition to the physical therapy, the patient started to receive indomethacin tablet 75 mg/day and sulphosalazine tablet 2 g/day. Upon absence of any regression in the complaints of sacroiliac pain following medical treatment and physical therapy for 10 sessions, 10 mg/2 ml betamethasone diproprionate and 40 mg/4 ml of lidocaine injection was administered to both sacroiliac joints separately by computed tomography guidance. Following injection, the pain in both sacroiliac joints markedly decreased. She was discharged with the recommendations of continuing the medical treatment and exercise program. The follow-up visits performed at 1, 3 and 6 months showed the erythrocyte sedimentation rate and the CRP values to be in the normal limits and no complaints.

DISCUSSION

AS is a systemic rheumatologic disease that is primarily characterized by axial skeletal inflammation and accompanied by extra-articular involvement including the eyes, heart and lungs. Genetic and environmental factors are involved in the pathogenesis of AS. Genetically, the factor that is mostly believed to account for the disease is the presence of HLA-B27 (6). AS is not a disease that primarily involves the muscles. However, reduction in the muscle mass and muscle weakness may occur due to inactivity and nerve compression (4). Our case had AS and concomitant myotonia congenita, which is a primary muscle disease. The most significant findings of myotonia are the stiffness and the delayed relaxation following the muscle contraction. This results from the hyperexcitability of the muscle cell membrane that can be detected by EMG. EMG can detect the myotonic discharges, which can be heard as the sound of a diving plane. The most common form of myotonia is the dystrophic myotonia. In addition, Thomsen and Becker congenital myotonias have also been described, which are nondystrophic myotonias involving a defect of the chloride channels (7, 8). Autosomal recessive myotonia congenita has been first described by Becker in 1966.

Stiffness may occur both in myotonia and AS (9). This may lead to a difficulty in evaluating the disease activity clinically and establishing the differential diagnosis. While the complaint of stiffness is commonly considered to represent inflammatory involvement in the clinical practice, the potential for myotonia should also be considered. Therefore, a comprehensive anamnesis should be obtained for contributing to the differential diagnosis. In case of inflammatory diseases, even if stiffness occurs following daytime inactivity, the morning stiffness is particularly more marked. However, in case of myotonia, daytime stiffness following rest is also marked due to the warm-up phenomenon together with the morning stiffness. In our case, it was difficult to determine the disease that caused the stiffness. If the patient didn't have stiffness secondary to myotonia congenita, she could have been diagnosed with AS earlier and respond to the medical and physical therapy better. In addition, the duration of stiffness used to assess the disease activity during the follow-up of AS patients would not be reliable in this patient. No case with concomitant AS and myotonia congenita has been reported to date. Our patient is the first case in this respect. The delay in the muscle relaxation occurring in cases of myotonic syndromes was observed to cause difficulty in determining the stiffness level of the concomitant AS in this case. This case underlines the significance of considering the potential for other conditions including myotonia or myositis, which may represent the cause for stiffness in a patient with AS. In patients with myotonia congenita, life style changes may facilitate the treatment, particularly by using the warm-up phenomenon. Avoiding excessive rest and keeping the same posture for a long time are useful to manage the muscle stiffness. Flexibility-enhancing exercises prevent damage to the muscles (10). From this point of view, the exercises to be practiced show similarity between the two diseases, AS and myotonia congenita. Flexibility and posture exercises combined with avoidance of inactivity play an important role in the treatment of AS. In our case, we detected a marked reduction in the overall duration of the daytime stiffness secondary to myotonia and the morning stiffness by administering the exercise program

prescribed. In conclusion, this is the first report about the association of AS and myotonia congenita. These pathologies can cause delaying of diagnosis of each other because of the some same clinic manifestation and should be kept on mind.

REFERENCES

1. *Calabro JJ, Dick WC. Ankylosing spondylitis. Lancaster: MTP Press; 1987.*
2. *Becker PE. Syndromes associated with myotonia:clinical and genetic classsification. In: Rowland LP, ed. Pathogenesis of human muscular dystrophies, New York: Excerpta Medica 1977: 699-703.*
3. *Streib EW. AAEE minimonograph ≠ 27: differential diagnosis of myotonic syndromes. Muscle Nerve 1987;10: 603-15.*
4. *Whitfield AGW. Neurological complications of ankylosing spondylitis. In: Vinken PJ, Bruyn PW, eds. Handbook of clinical neurology.Vol. 38, Amsterdam: Elsevier, North Holland Biochemical Press,1979:505-20.*
5. *Van der Linden S, Valkenburg HA. Evaluations of diagnostic criteria for ankylosing spondylitis: a proposal for medication of the New York criteria. Arthritis 1984; 27: 361-8.*
6. *Khan MA. Update on spondyloarthropathies. Ann Intern Med 2002;136: 896-907.*
7. *Koch MC, Steinmeyer K, Lorenz C. The skeletal muscle chloride channel in dominant and recessive human myotonia. Science 1992;257:797-800.*
8. *Zhang J, George A, Griggs RC. Mutations in the skeletal muscle chloride channel gene (CLCN1) associated with dominant and recessive myotonia congenita. Neurology 1996;47:993-8.*
9. *Torbergsen T, Hodnebo A, Brautaset N J, Loseth S, Stalberg E. A rare form of painful nondystrophic myotonia. Clinical Neurophsiology 2003;114:2347-54.*
10. *Conravey A, Santana-Gould L. Myotonia Congenita and Myotonic Dystrophy: Surveillance and Management. Current Treatment Options in Neurology 2010;12:16-2*