DOI: 10.53469/jcmp.2025.07(01).41

Clinical Analysis of Two Cases of Myotonic Dystrophy Patients Treated with Chinese Medicine

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Abstract: ***Myotonic dystrophy (DM) is a group of multi-systemic autosomal dominant genetic disorders characterized by muscle weakness, myotonia, and muscle atrophy. This study clinically analyzed two patients with myotonic dystrophy, both diagnosed with "Wei" disease in traditional Chinese medicine (TCM), with syndromes of liver-kidney deficiency and spleen-stomach weakness, respectively. The treatment methods included moxibustion, acupuncture at specific points, and a combination of TCM decoctions with modifications. After treatment, the patients showed improvement in their symptoms. This study provides new insights into the TCM treatment of DM.***

Keywords: Myotonic dystrophy, Impotence, Treatment of traditional Chinese medicine**.**

1. Introduction

Myotonic dystrophy (DM) is a group of autosomal dominant inherited disorders characterized by muscle weakness, muscle stiffness, and muscle atrophy, affecting multiple systems. It is similar to the "atrophy disease" in traditional Chinese medicine (TCM) [1]. Atrophy disease refers to the condition where the limbs experience sluggish muscles and tendons, leading to weakness and muscle wasting, progressively impairing voluntary movement. The severity of the disease varies significantly among patients [2-3]. For example, within the same family, one might find asymptomatic adult carriers and severely affected infants. Currently, there is no fundamental treatment for myotonic dystrophy, and treatment is mostly symptomatic. In Western medicine, the primary treatment strategies focus on alleviating muscle stiffness, inhibiting the production of myostatin, and reversing muscle atrophy [4-5]. Some studies suggest that physical therapy has some effect in maintaining muscle function [6], and moderate aerobic exercise can alleviate symptoms of muscle stiffness. However, no obvious clinical efficacy has been reported. On the other hand, TCM has a complete theoretical framework and treatment methods for treating atrophy disease [7]. This paper presents two cases of myotonic dystrophy, a mother and daughter, and analyzes the TCM diagnosis and treatment to raise clinical awareness.

2. Clinical Data

Case 1: The patient, Sun Bangxia, a 61-year-old female, presented with "progressive limb weakness for 14 years, worsening over the past 2 months" and was admitted to the neurology department of our hospital on August 12, 2023, with a suspected diagnosis of "myotonic dystrophy." The patient started experiencing gradual limb weakness, muscle atrophy, difficulty opening the mouth after clenching her teeth, difficulty releasing a handshake, and trouble standing up after squatting 14 years ago. She had not sought formal medical treatment during this time. Currently, her muscle weakness, muscle atrophy, and difficulty lifting limbs have worsened, accompanied by blurred vision and a preference for heat and dislike of cold. Her medical history includes hypothyroidism, hyperlipidemia, and liver dysfunction. She also had a history of gallbladder surgery and cataract surgery on the right eye. Her 37-year-old daughter also presents with similar symptoms, a "hatchet-shaped face," and a gait with crossing steps. No other family history of genetic diseases or similar illnesses was reported.

Physical Examination: The patient was alert and cooperative. She had a crossing gait and her pupils were equal and round, with a normal light reflex. The muscle tone in her limbs was reduced, with muscle strength graded at 4/5 in both upper limbs, 4/5 in the left lower limb, and 3/5 in the right lower limb. Tendon reflexes were hyperactive, and no pathological signs were observed. No edema was noted in the lower limbs. The tongue was red with a thin coating, and the pulse was thin and rough.

Diagnosis and Treatment Progress: After hospitalization, relevant examinations were performed. Main laboratory results included:

- Blood cell analysis: Red blood cell mean volume 103.6fL, mean hemoglobin 33pg;
- Coagulation analysis: Fibrinogen 4.2g/L, D-dimer 2560ng/ml;
- Emergency test results: Glucose 8mmol/L, creatine kinase 219U/L, serum myoglobin 224ng/ml, sodium 148.4mmol/L, chloride 112.8mmol/L;
- Biochemistry results: Cystatin C 1.35mg/L, triglycerides 3.63mmol/L, sodium 148.5mmol/L, chloride 112mmol/L;
- Thyroid function and tumor screening: Total T3 0.79nmol/L, TSH 39.9mIU/L, anti-thyroid peroxidase antibodies 436.6IU/ml, anti-thyroglobulin antibodies 213IU/ml.

Main Examination Results:

- Electrocardiogram (ECG):

1) Sinus bradycardia.
2) First-degree atrioventricular (AV) block.
3) Frequent ectopic ventricular premature beats, abnormal U waves. Blood potassium levels should be checked.

- Thyroid Ultrasound:

1) Coarse echogenicity of the thyroid, further laboratory testing is recommended.
2) No significant abnormalities in bilateral cervical lymph nodes.

- Head MRI + Diffusion Weighted Imaging (DWI):

1) Multiple lacunar infarctions and ischemic changes adjacent to both lateral ventricles and in the centrum semiovale.
2) Brain atrophy. Follow-up is advised.

- Lower Limb Electromyography (EMG):

1) Prolonged motor nerve conduction latency in both tibial nerves and the right common peroneal nerve. Further testing for DM gene is recommended.

Diagnosis:

- **Traditional Chinese Medicine (TCM) Diagnosis: Disease Diagnosis:** Wei disease, a condition characterized by muscle weakness or atrophy. **Syndrome Diagnosis:** Liver and Kidney Deficiency Syndrome.

Case 2: Yang Huijuan, a 37-year-old female and the daughter of the first patient, presented with "progressive instability while walking and limb weakness for 12 years, worsening over the past week." She was admitted on August 12, 2023, with a suspected diagnosis of "muscular dystrophy." She developed weakness in both lower limbs 12 years ago, which caused frequent falls, without any muscle pain, numbness, or swelling. The patient had previously been diagnosed with "myotonic dystrophy" at Beijing 301 Hospital but had not received medication. In July 2017 and July 2021, she had been hospitalized at our hospital for treatment. Over the past week, her symptoms of lower limb weakness have worsened, with muscle atrophy, decreased appetite, loose stools, weaker grip strength, and difficulty squatting or standing.

Past Medical History: The patient had a history of arrhythmia for over 10 years, and she has been taking Shen Song Yang Xin capsules. She was diagnosed with myotonic dystrophy over 10 years ago and has no history of hepatitis or tuberculosis. Her mother also has similar symptoms.

Physical Examination: Physical examination: The patient is conscious, in fair spirits, cooperative during the examination, and responsive to questions appropriately. The patient walks into the room with a crossed-threshold gait, exhibiting a "hatchet-like" face and facial muscle weakness. No jaundice in the sclera, and both pupils are equal in size and round, with sensitive light reflex. No cyanosis in the lips, and the tongue is centered when extended. The neck is soft with no resistance. The sternocleidomastoid muscles appear atrophied. The trachea is centrally positioned, and a soft, well-defined, smooth, non-tender mass approximately 3x3 cm in size is palpated on the left thyroid lobe, which moves up and down with swallowing. Muscle atrophy is noted in the limbs. Muscle strength in both upper limbs is graded 4, with reduced grip strength in the left hand. Muscle strength in both lower limbs is also graded 4, with increased muscle tone in the limbs. Deep tendon reflexes are reduced, and pathological signs are not elicited. Moderate edema is observed on the dorsum of both feet. The tongue is pale red with a thin white coating, and the pulse is deep, thin, and weak.

After admission, the patient underwent relevant examinations, and the main laboratory results are as follows:

- **Complete blood count (five-part differential):** Neutrophil percentage: 77.8% Absolute eosinophil count: 0.04 × 10^9/L Red blood cell distribution width (CV): 14.3%

- **Biochemical panel:** Creatinine: 32.7 μmol/L Glucose: 7.3 mmol/L, Lactate dehydrogenase (LDH): 275.7 U/L, α-hydroxybutyrate dehydrogenase: 220.4 U/L, Serum myoglobin: 289 ng/mL, High-sensitivity C-reactive protein (hs-CRP): 4.5 mg/L, Sodium: 147.2 mmol/L, Chloride: 112.1 mmol/L

- **Neuromuscular enzymes:** Creatine kinase isoenzyme: 14 U/L, Serum myoglobin: 113.1 ng/mL, Lactate dehydrogenase (LDH): 2760 U/L, Creatine kinase (CK): 225 U/L

Main examination results:

- **Bedside Electrocardiogram (ECG):** Shows sinus rhythm with frequent premature beats, possibly premature atrial contractions.

- **Electromyography (EMG):**
  - Motor nerve conduction latency is prolonged in both the right and left median nerves, as well as in both the right and left ulnar nerves.
  - Sensory nerve conduction velocity is slightly slowed in both the right and left median and ulnar nerves.

- **Echocardiography:** Resting state shows a generally normal echocardiogram.

- **24-hour Ambulatory Electrocardiogram (Holter):** Sinus rhythm, with an average heart rate of 65 bpm. Total heartbeats analyzed: 88,061. Fastest heart rate: 121 bpm, occurring at 05:55.Slowest heart rate: 38 bpm, occurring at 01:41.Frequent supraventricular premature beats (11,956, some not conducted), including supraventricular bigeminy (11 episodes) and supraventricular trigeminy.

- **DM Genetic Testing (External):** Chromosome 19q13 3 site, the dystrophia myotonica protein kinase (DMPK) gene shows abnormal CTG trinucleotide repeat

expansion of over 100 repeats (normal value: 5-40). This repeat expansion is associated with the severity of the patient's symptoms.

Diagnosis:

- **Traditional Chinese Medicine (TCM) Diagnosis:** Disease Diagnosis: Wei Disease, a condition characterized by muscle weakness or atrophy. Syndrome Diagnosis: Spleen and Stomach Deficiency Syndrome.

3. Discussion

3.1 TCM Analysis

In Traditional Chinese Medicine (TCM), myotonic dystrophy (DM) falls under the category of "Wei disease" (muscle weakness). The causes of Wei disease can be attributed to both external pathogens and internal factors. Although the disease affects the muscles and tendons, the liver, kidneys, lungs, and stomach are most closely related. According to the theory of the five organs, the liver stores blood and governs the tendons, the kidneys store essence and produce marrow, the stomach generates fluids, and the lungs regulate the body's fluid metabolism. As noted in *The Guidelines for Clinical Diagnosis and Treatment in TCM*, Wei disease is primarily "a disease of the liver, kidney, lungs, and stomach." In *Su Wen: Taiyin Yangming Lun*, it is stated: "When the spleen is unable to nourish the stomach with its fluids, the limbs fail to receive the vital essence from food, and with the decline of qi, the pulse pathway is obstructed. The tendons, bones, and muscles lack nourishment, leading to atrophy." In *Zhengzhi Huibu: Wei Bi*, it is emphasized: "Internal heat leads to Wei disease, which is the root cause of this illness. If there is an external trigger, it must be caused by underlying factors."

Disorder of the nutritive and defensive qi, resulting in deficiency, can also lead to Wei disease. *Yi Zong Bi Du* states: "The spleen and stomach are connected by a membrane and open to the mouth. If the spleen is overheated, it leads to dryness in the stomach and thirst. The spleen governs the muscles, and internal heat depletes essence and qi, leading to muscle numbness and atrophy."

For the treatment of Patient 1, moxibustion was applied to the Baihui acupoint, with acupuncture to Yangming meridian points, complemented by a modified Tiger Subduing Decoction (Hu Qian Tang) [8] and other TCM therapies. Baihui, also known as the "meeting point of the three yang meridians," is the meeting point of the yang qi of the hand and foot yang meridians and the Du meridian. This point helps to boost the yang qi, which is essential for patients with long-term liver and kidney deficiencies, who tend to feel better in warm conditions and dislike cold. Moxibustion at Baihui for 30 minutes is effective in elevating yang qi. Acupuncture was performed at key points of the Yangming meridian: the main points were Jianyu, Quchi, Hegu, Liangqiu, Zusanli, and Yanglingquan, while complementary points were located at Ganshu and Shenshu, with retention of the needles for half an hour. The Yangming meridian is abundant in qi and blood and governs the tendons, helping to regulate yin and yang, tonify qi and blood, and smooth the tendons and meridians. This approach aligns with the principle in *Su Wen: Wei Lun*, which states: "To treat Wei disease, one must target the Yangming meridian." [9] Given the chronic nature of the disease and the liver and kidney deficiencies in this patient, the prescription aimed to "tonify the liver and kidneys, nourish blood, and soften the tendons." The main formula was the Tiger Subduing Pill, with ingredients such as tiger bone and Eucommia entering the liver and kidney meridians to strengthen the tendons and bones. Additional herbs like Rehmannia, Cistanches, and Turtle shell were added to tonify kidney yang, nourish essence, and fill the marrow. Goji berries, Cuscuta seeds, and Sha Yuan Zi were also included to nourish the liver, improve vision, and enhance essence. Although the formula is best as a pill, due to production limitations, it was adjusted to a decoction for oral administration.

For Patient 2, moxibustion was applied to Baihui and Kunlun acupoints, with acupuncture at hand and foot Yangming and Shaoyang meridian points [10], complemented by a modified Shenling Baizhu Powder [11]. This patient exhibited reduced appetite and loose stools, indicating spleen weakness and insufficient spleen yang. Moxibustion at Baihui boosts spleen yang, while Kunlun, an acupoint on the bladder meridian, is the most powerful point for restoring deficient qi. As stated in *Ling Shu: Kou Wen*, "Upper qi...lower qi deficiency leads to Wei and blockage of the heart, which can be remedied by supplementing Kunlun acupoint on the outer ankle." Together, these two points work synergistically to elevate yang qi. Acupuncture points on the Yangming meridian (Zusanli, Zhongwan, Zhangmen, Waiguan, Yanglingquan) and the Shaoyang meridian (Zhongzhu, Yangchi) were selected to harmonize the organs and restore meridian flow. Zhangmen and Zhongwan are the mu points of the spleen and stomach, where the qi of the internal organs converges. Acupuncture at these points helps restore the qi of the spleen and stomach. Yanglingquan, where the tendons converge, is also a Shaoyang point and helps to smooth the meridians. Waiguan connects with the Yangwei meridian and other Yang meridians, acting as a junction point for the Shaoyang meridian to regulate the qi. For this patient, the prescribed formula was based on the principle of "tonifying the middle and benefiting qi, strengthening the spleen, and raising clear yang," using Shenling Baizhu Powder with added Aconite and Cinnamon [12]. The formula included ginseng to tonify the spleen and stomach, Poria and Atractylodes to strengthen the spleen and promote fluid metabolism, as well as Dioscorea, Lotus seeds, and Cuscuta to improve spleen function and alleviate diarrhea. Other herbs like Fructus Psoraleae and Schisandra were included to warm the kidneys, consolidate essence, and restore kidney qi.

Although modern medicine distinguishes between myotonic dystrophy and Wei disease in TCM [13], both patients were diagnosed with Wei disease within the TCM framework. The treatment included methods to regulate the spleen and stomach, tonify the liver and kidneys, nourish qi and blood, replenish fluids, and warm the muscles and tendons [14]. Through a comprehensive diagnosis based on the four diagnostic methods, TCM acupuncture, moxibustion, and herbal decoctions were used to improve the patients' symptoms.

In conclusion, when a family history of myotonic dystrophy is identified, laboratory tests, including genetic testing and electromyography [15], should be completed to consider the possibility of DM. Early diagnosis and intervention are crucial. This article provides a description of the TCM diagnosis and treatment process for two DM patients, aiming to enhance clinical understanding and offer new therapeutic approaches.

Fund Project

Joint Research Project of the China Traditional Chinese Medicine Science and Technology Development Center (CXZH202306); Anhui Provincial Traditional Chinese Medicine Inheritance and Innovation Research Project (20220CYB28); Anhui Provincial Higher Education Scientific Research Project (2022AH040329).

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