Myotonic dystrophy type 1 (DM1) is the most common disease causing muscle weakness and atrophy in adults.[1] The prevalence of DM1 in China is not clear. DM1 is an autosomal dominant genetic disorder associated with the cytosine-thymine-guanine (CTG) repeat expansion in the first untranslated region of dystrophin myotonin-kinase (DMPK) gene on chromosome 19q13.3. In DM1, CTG repeat numbers are more than 50. The size of CTG repeat expansion is associated with the time of clinical phenotypes onset and severity.[1‑3] The coexistence of DM1 and syringomyelia is rare. Here, we report DM1 coexisting with syringomyelia in a Chinese male patient.

A 28-year-old man was admitted in Qilu Hospital of Shandong University in December 2013, who had difficulty in flexing his fingers after hands grip, mainly in cold weather, for more than 10 years. Sometimes, he felt tongue stiff and slurred speech. He was shy and never told above symptoms to any relatives. His mother mentioned that he had progressively walking disability, especially with his right leg during the last 2 years. The patient noticed the right calf thinner than the left calf 1 week ago. He received surgery for right parotid benign tumor 9 years ago. His grandfather died of lung cancer at the age of 59, his father died of gastric cancer at 50, and one uncle died of leukemia at 19. Moreover, his aunt was diagnosed with DM1 after a muscular biopsy at the age of 50. The patient’s sister was asymptomatic until now [Figure 1].

His physical examination on admission revealed a typical hatchet face with incipient baldness and weakness of the bilateral eyelid closure but normal ocular movement. Percussion myotonia was present in the tongue, bilateral thenar, and upper arm muscles. He had difficulty in opening the mouth after biting down forcibly and extending the fingers after a hard fist. Right lower limb was thinner than the left by 6 cm, with muscle strength scale 4 and foot drop. Limbs muscular tone was normal. Right achille reflex was absent. There was left semi-jacket-like hypopalgiesia. Bilateral Babinski’s signs were negative. Skin scratch test was positive from T2 to T4 dominated areas.

The laboratory tests showed triglyceride 1.94 mmol/L (normal range: 0.30–1.70 mmol/L), creatine kinase 243 U/L (normal range: 38–174 U/L), serum free thyroxine (FT4) 10.98 pmol/L (normal range: 12.00–22.00 pmol/L), follicle-stimulating hormone (FSH) 19.52 U/L (normal range: 1.50–12.40 U/L), and luteinizing hormone (LH) 14.68 U/L (normal range: 1.70–8.60 U/L). Serum testosterone level and glucose tolerance test were normal. Opacities were present in the center of both lenses at the slit-lamp examination. Thoracolumbar magnetic resonance imaging (MRI) showed syringomyelia between T2 and T5 level [Figure 2a and 2b]. Brain MRI showed multiple Virchow–Robin spaces (VRSs) dispersed in the frontal lobe and patchy lesions in bilateral subcortical and deep white matter [Figure 2c–2e]. Left anterior fascicular block was found in electrocardiogram (ECG). The electromyogram revealed typical myotonic discharges. However, the motor nerve conduction velocity (MCV) of bilateral tibial nerves and left common peroneal nerve, and the sensory
nerve conduction velocity (SCV) of right sural nerve and superficial fibular nerve were all slow, with normal amplitude. H-reflex latency of right tibial nerve extended. During hospitalization, the patient had excessive daytime sleepiness (EDS), several times of nausea, abdominal pain, and emesis. Gastrointestinal tract disease was ruled out after physical examination and computed tomography scan. Histopathological changes of left biceps brachii biopsy revealed ranging muscle fibers and atrophy type I fiber. No abnormality was found in immunohistochemistry. Cytogenetic analysis tested CTG repeat amplification number in DMPK gene was more than 50 times.

Myotonia and muscular weakness are the most typical clinical features for DM1, which primarily affect facial and distal muscles of the extremities. Digit flexors and foot dorsiflexors weakness are very common, and foot drop can lead to disability.[1,2] DM1 causes multisystem disorders. Our patient had the characteristics of myotonia, muscular weakness, muscular wasting in chewing muscles and right lower limb distal muscles. He also had incipient baldness, cataract, and left anterior fascicular block in ECG. This case also presented gastrointestinal symptoms. In addition, psychological defects such as introverted and even paranoid personality traits have received increasing attention in DM1. In endocrine system, our patient had abnormal FT4 and thyroid-stimulating hormone levels. Furthermore, his plasma FSH and LH levels were increased with normal testosterone level indicating relative androgen insufficiency. Sex hormone disorder in male DM1 patients is the main cause for infertility. Endocrine abnormalities increase over time.[4] His endocrine hormones levels should be monitored periodically.

Myotonic discharge is the most prominent or earliest isolated performance in electromyography. Li et al.[5] evaluated electromyograms in 33 DM1 patients and found nerve conduction velocity was all normal. However, besides myotonic discharge, this case also showed slightly slow MCV and SCV with normal amplitude, conformed to demyelinating peripheral neuropathy (PNP). PNP was more frequent in males and patients with a longer duration of the disease.[6]

The patient’s brain MRI showed multiple lesions in periventricular and deep white matter and prominent perivascular spaces inconsistent with the age. The loss of neurons and dilated VRS in cortex or subcortical brain regions have been demonstrated by DM1 autopsies. Using structural brain MRI, Minnerop et al.[7] confirmed visual white matter lesions (WML), especially in frontal brain regions, were more frequent in DM1 than the healthy control. In DM1, WML may reflect underlying small vessel vasculopathy and be associated to psychomotor activity.[8] WML in frontal lobe can explain the presentation of executive dysfunction, such as attention deficit, and indifference, avoidant personality traits. The predominant presence of depressed mood and fatigue or EDS may be because of white matter degeneration in early DM1. It is rare for the patient suffering from DM1 and syringomyelia simultaneously. Levisky et al.[9] reported a sibship with 2 individuals affected by myotonic dystrophy and a third with...
syringomyelia. Mascalchi et al.\cite{9} also reported a DM1 patient with syringomyelia due to spinal hemangioblastoma. No report about DM1 with syringomyelia was documented in China. It is unclear whether the combination of DM1 with syringomyelia is mere coincidence or a new inherited disease derived from DM1 or a concurrent disease from DM1.

The patient and his several relatives had tumors in accordance with recent studies. In a large longitudinal study including 1081 DM patients followed up for 21 years, females had a higher absolute risk of all cancers than males by ages 50 and 60 years, respectively.\cite{10} Brain, colon, endometrial, and ovarian cancers are common malignancies in DM1.

In conclusion, this is a case of DM1 with syringomyelia reported in China. The patient also presented demyelinating PNP and WML. Further study is required to deeper our understanding of this disease and improve the clinical outcome of these patients.

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**Conflicts of interest**
There are no conflicts of interest.

**References**
1. Turner C, Hilton‑Jones D, Lochmüller H, Hanna MG. MRC centre for neuromuscular diseases 1\textsuperscript{st} (1\textsuperscript{st} December 2010), and 2\textsuperscript{nd} (2\textsuperscript{nd} May 2012) myotonic dystrophy workshops, London, UK and the myotonic dystrophy standards of care and national registry meeting, Newcastle, UK July 2011. Neuromuscul Disord 2013;23:1069‑80. doi: 10.1016/j.nmd.2013.07.006.
2. Bouchard JP, Cossette L, Basseez G, Puymirat J. Natural history of skeletal muscle involvement in myotonic dystrophy type 1: A retrospective study in 204 cases. J Neurol 2015;262:285‑93. doi: 10.1007/s00415‑014‑7570‑x.
3. Ørngreen MC, Arlien‑Søborg P, Duno M, Hertz JM, Vissing J. Endocrine function in 97 patients with myotonic dystrophy type 1. J Neurol 2012;259:912‑20. doi: 10.1007/s00415‑011‑6277‑5.
4. Dahlvist JR, Ørngreen MC, Witting N, Vissing J. Endocrine function over time in patients with myotonic dystrophy type 1. Eur J Neurol 2015;22:116‑22. doi: 10.1111/ene.12542.
5. Li M, Wang ZJ, Cui F, Yang F, Chen ZH, Ling L, et al. Electrophysiological features of patients with myotonic dystrophy type 1 (in Chinese). Natl Med J China 2013;93:345‑7. doi: 10.3760/ cma.j.issn.0376‑2491.2013.05.006.
6. Peric S, Stojanovic VR, Nikolie A, Kacar A, Basta I, Pavlovic S, et al. Peripheral neuropathy in patients with myotonic dystrophy type 1. Neur...812Y.0000000144.
7. Minnerop M, Weber B, Schoene‑Bake JC, Roeske S, Mirbach S, Anspach C, et al. The brain in myotonic dystrophy 1 and 2: Evidence for a predominant white matter disease. Brain 2011;134(Pt 12):3530‑46. doi: 10.1093/brain/awr299.
8. Levisky RB, Vianna‑Morgante AM, Frota‑Pessoa O, Scaff M, Tsanaclis AM, Levy JA. Myotonic dystrophy, syringomyelia, and 2/13 translocation in the same family. J Med Genet 1977;14:51‑3.
9. Mascalchi M, Padovani R, Taiuti R, Quilici N. Syringomyelia in myotonic dystrophy due to spinal hemangioblastoma. Surg Neurol 1998;50:446‑8. doi: 10.1016/S0039‑3109(97)00050‑5.
10. Gadalla SM, Lund M, Pfeiffer RM, Görtz S, Mueller CM, Moxley RT 3\textsuperscript{rd}, et al. Cancer risk among patients with myotonic muscular dystrophy. JAMA 2011;306:2480‑6. doi: 10.1001/jama.2011.1796.