

# World Journal of *Clinical Cases*

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## Contents

Thrice Monthly Volume 9 Number 19 July 6, 2021

## OPINION REVIEW

- 4881** Fear of missing out: A brief overview of origin, theoretical underpinnings and relationship with mental health  
*Gupta M, Sharma A*

## REVIEW

- 4890** Molecular pathways in viral hepatitis-associated liver carcinogenesis: An update  
*Elpek GO*
- 4918** Gastroenterology and liver disease during COVID-19 and in anticipation of post-COVID-19 era: Current practice and future directions  
*Oikonomou KG, Papamichalis P, Zafeiridis T, Xanthoudaki M, Papapostolou E, Valsamaki A, Bouliaris K, Papamichalis M, Karvouniaris M, Vlachostergios PJ, Skoura AL, Komnos A*
- 4939** Enhancing oxygenation of patients with coronavirus disease 2019: Effects on immunity and other health-related conditions  
*Mohamed A, Alawna M*

## MINIREVIEWS

- 4959** Clinical potentials of ginseng polysaccharide for treating gestational diabetes mellitus  
*Zhao XY, Zhang F, Pan W, Yang YF, Jiang XY*
- 4969** Remarkable gastrointestinal and liver manifestations of COVID-19: A clinical and radiologic overview  
*Fang LG, Zhou Q*
- 4980** Liver injury in COVID-19: Known and unknown  
*Zhou F, Xia J, Yuan HX, Sun Y, Zhang Y*
- 4990** COVID-19 and gastroenteric manifestations  
*Chen ZR, Liu J, Liao ZG, Zhou J, Peng HW, Gong F, Hu JF, Zhou Y*
- 4998** Role of epithelial-mesenchymal transition in chemoresistance in pancreatic ductal adenocarcinoma  
*Hu X, Chen W*
- 5007** Insights into the virologic and immunologic features of SARS-COV-2  
*Polat C, Ergunay K*



**ORIGINAL ARTICLE****Basic Study**

- 5019** SMAC exhibits anti-tumor effects in ECA109 cells by regulating expression of inhibitor of apoptosis protein family

*Jiang N, Zhang WQ, Dong H, Hao YT, Zhang LM, Shan L, Yang XD, Peng CL*

**Case Control Study**

- 5028** Efficacy of Solitaire AB stent-release angioplasty in acute middle cerebral artery atherosclerosis obliterative cerebral infarction

*Wang XF, Wang M, Li G, Xu XY, Shen W, Liu J, Xiao SS, Zhou JH*

**Retrospective Study**

- 5037** Diagnostic value of different color ultrasound diagnostic method in endometrial lesions

*Lin XL, Zhang DS, Ju ZY, Li XM, Zhang YZ*

- 5046** Clinical and pathological features and risk factors for primary breast cancer patients

*Lei YY, Bai S, Chen QQ, Luo XJ, Li DM*

- 5054** Outcomes of high-grade aneurysmal subarachnoid hemorrhage patients treated with coiling and ventricular intracranial pressure monitoring

*Wen LL, Zhou XM, Lv SY, Shao J, Wang HD, Zhang X*

- 5064** Microwave ablation combined with hepatectomy for treatment of neuroendocrine tumor liver metastases

*Zhang JZ, Li S, Zhu WH, Zhang DF*

- 5073** Clinical application of individualized total arterial coronary artery bypass grafting in coronary artery surgery

*Chen WG, Wang BC, Jiang YR, Wang YY, Lou Y*

**Observational Study**

- 5082** Early diagnosis, treatment, and outcomes of five patients with acute thallium poisoning

*Wang TT, Wen B, Yu XN, Ji ZG, Sun YY, Li Y, Zhu SL, Cao YL, Wang M, Jian XD, Wang T*

- 5092** Sarcopenia in geriatric patients from the plateau region of Qinghai-Tibet: A cross-sectional study

*Pan SQ, Li YM, Li XF, Xiong R*

- 5102** Medium-term efficacy of arthroscopic debridement *vs* conservative treatment for knee osteoarthritis of Kellgren-Lawrence grades I-III

*Lv B, Huang K, Chen J, Wu ZY, Wang H*

**Prospective Study**

- 5112** Impact of continuous positive airway pressure therapy for nonalcoholic fatty liver disease in patients with obstructive sleep apnea

*Hirono H, Watanabe K, Hasegawa K, Kohno M, Terai S, Ohkoshi S*

**Randomized Controlled Trial**

- 5126** Erector spinae plane block at lower thoracic level for analgesia in lumbar spine surgery: A randomized controlled trial  
*Zhang JJ, Zhang TJ, Qu ZY, Qiu Y, Hua Z*

**SYSTEMATIC REVIEWS**

- 5135** Controversies' clarification regarding ribavirin efficacy in measles and coronaviruses: Comprehensive therapeutic approach strictly tailored to COVID-19 disease stages  
*Liatsos GD*
- 5179** Systematic review and meta-analysis of trans-jugular intrahepatic portosystemic shunt for cirrhotic patients with portal vein thrombosis  
*Zhang JB, Chen J, Zhou J, Wang XM, Chen S, Chu JG, Liu P, Ye ZD*

**CASE REPORT**

- 5191** Myelodysplastic syndrome transformed into B-lineage acute lymphoblastic leukemia: A case report  
*Zhu YJ, Ma XY, Hao YL, Guan Y*
- 5197** Imaging presentation and postoperative recurrence of peliosis hepatis: A case report  
*Ren SX, Li PP, Shi HP, Chen JH, Deng ZP, Zhang XE*
- 5203** Delayed retroperitoneal hemorrhage during extracorporeal membrane oxygenation in COVID-19 patients: A case report and literature review  
*Zhang JC, Li T*
- 5211** Autologous tenon capsule packing to treat posterior exit wound of penetrating injury: A case report  
*Yi QY, Wang SS, Gui Q, Chen LS, Li WD*
- 5217** Treatment of leiomyomatosis peritonealis disseminata with goserelin acetate: A case report and review of the literature  
*Yang JW, Hua Y, Xu H, He L, Huo HZ, Zhu CF*
- 5226** Homozygous deletion, c. 1114-1116del, in exon 8 of the *CRPPA* gene causes congenital muscular dystrophy in Chinese family: A case report  
*Yang M, Xing RX*
- 5232** Successful diagnosis and treatment of jejunal diverticular haemorrhage by full-thickness enterotomy: A case report  
*Ma HC, Xiao H, Qu H, Wang ZJ*
- 5238** Liver metastasis as the initial clinical manifestation of sublingual gland adenoid cystic carcinoma: A case report  
*Li XH, Zhang YT, Feng H*
- 5245** Severe hyperbilirubinemia in a neonate with hereditary spherocytosis due to a *de novo* ankyrin mutation: A case report  
*Wang JF, Ma L, Gong XH, Cai C, Sun JJ*

- 5252** Long-term outcome of indwelling colon observed seven years after radical resection for rectosigmoid cancer: A case report  
*Zhuang ZX, Wei MT, Yang XY, Zhang Y, Zhuang W, Wang ZQ*
- 5259** Diffuse xanthoma in early esophageal cancer: A case report  
*Yang XY, Fu KI, Chen YP, Chen ZW, Ding J*
- 5266** COVID-19 or treatment associated immunosuppression may trigger hepatitis B virus reactivation: A case report  
*Wu YF, Yu WJ, Jiang YH, Chen Y, Zhang B, Zhen RB, Zhang JT, Wang YP, Li Q, Xu F, Shi YJ, Li XP*
- 5270** Maintenance treatment with infliximab for ulcerative ileitis after intestinal transplantation: A case report  
*Fujimura T, Yamada Y, Umeyama T, Kudo Y, Kanamori H, Mori T, Shimizu T, Kato M, Kawaida M, Hosoe N, Hasegawa Y, Matsubara K, Shimojima N, Shinoda M, Obara H, Naganuma M, Kitagawa Y, Hoshino K, Kuroda T*
- 5280** Infliximab treatment of glycogenosis Ib with Crohn's-like enterocolitis: A case report  
*Gong YZ, Zhong XM, Zou JZ*
- 5287** Hemichorea due to ipsilateral thalamic infarction: A case report  
*Li ZS, Fang JJ, Xiang XH, Zhao GH*
- 5294** Intestinal gangrene secondary to congenital transmesenteric hernia in a child misdiagnosed with gastrointestinal bleeding: A case report  
*Zheng XX, Wang KP, Xiang CM, Jin C, Zhu PF, Jiang T, Li SH, Lin YZ*
- 5302** Collagen VI-related myopathy with scoliosis alone: A case report and literature review  
*Li JY, Liu SZ, Zheng DF, Zhang YS, Yu M*
- 5313** Neuromuscular electrical stimulation for a dysphagic stroke patient with cardiac pacemaker using magnet mode change: A case report  
*Kim M, Park JK, Lee JY, Kim MJ*
- 5319** Four-year-old anti-N-methyl-D-aspartate receptor encephalitis patient with ovarian teratoma: A case report  
*Xue CY, Dong H, Yang HX, Jiang YW, Yin L*
- 5325** Glutamic acid decarboxylase 65-positive autoimmune encephalitis presenting with gelastic seizure, responsive to steroid: A case report  
*Yang CY, Tsai ST*
- 5332** Ectopic opening of the common bile duct into the duodenal bulb with recurrent choledocholithiasis: A case report  
*Xu H, Li X, Zhu KX, Zhou WC*
- 5339** Small bowel obstruction caused by secondary jejunal tumor from renal cell carcinoma: A case report  
*Bai GC, Mi Y, Song Y, Hao JR, He ZS, Jin J*
- 5345** Brugada syndrome associated with out-of-hospital cardiac arrest: A case report  
*Ni GH, Jiang H, Men L, Wei YY, A D, Ma X*

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## Collagen VI-related myopathy with scoliosis alone: A case report and literature review

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### Abstract

#### BACKGROUND

Scoliosis is a complex three-dimensional deformity of spine and one of the common complications of collagen VI-related myopathy, caused by mutations in collagen type VI alpha 1 chain (COL6A1), COL6A2, and COL6A3 genes. The typical clinical presentations of collagen VI-related myopathy include weakness, hypotonia, laxity of distal joints, contractures of proximal joints, and skeletal deformities.

#### CASE SUMMARY

A 28-year-old female presented with scoliosis for 28 years without weakness, hypotonia, laxity of distal joints, and contracture of proximal joints. Computed tomography and magnetic resonance imaging revealed hemivertebra, butterfly vertebra, and the missing vertebral space. Patients underwent orthopedic surgery and paravertebral muscle biopsy. The Cobb angle dropped from 103.4° to 52.9°. However, the muscle biopsy showed neurogenic muscular atrophy with myogenic lesions, suggesting congenital muscular dystrophy. Gene analysis indicated that mutations in COL6A1 (c.1612-10G>A) and COL6A2 (c.115+10G>T, c.2749G>A). Immunohistochemistry staining for collagen VI displayed shallow and discontinuous. Eventually, the patient was diagnosed as collagen VI-related myopathy.

#### CONCLUSION

This newly found subtype of collagen VI-related myopathy has no typical



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manifestations; however, it is characterized by severe scoliosis and congenital vertebral deformity.

**Key Words:** Paravertebral pathology; Scoliosis; Collagen VI-related myopathy; Genetic testing; Neuromuscular diseases; Case report

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**Core Tip:** Here, we report a patient diagnosed with congenital scoliosis initially with deformity of the vertebral body. However, gene analyses indicated mutations in *collagen type VI alpha 1 chain (COL6A1)* (c.1612-10G>A) and *COL6A2* (c.115+10G>T, c.2749G>A). Immunohistochemistry of collagen VI displayed shallow and discontinuous staining. This newly found subtype of collagen VI-related myopathy has no weakness, hypotonia, laxity of distal joints, and contractures of proximal joints and is characterized by severe scoliosis and congenital vertebral deformity, indicating that the underlying etiology of congenital scoliosis may be a special type of collagen VI-related myopathy.

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## INTRODUCTION

Characterized by lateral curvature and axial rotation, scoliosis is a complex three-dimensional deformity of the spine[1]. One of the causes is the abnormal development of the vertebral body, called congenital scoliosis[2]. This congenital malformation is mainly caused by asymmetric development of the spine during 4-6 wk of gestation[3]. The incidence rate of malformation in the live fetus is about 1/1000[4], which can be divided into segmental abnormalities, formation abnormalities, or composite abnormalities[5]. Some scholars believe that the occurrence of congenital scoliosis relate to genetic factors (e.g., *T-box transcription factor 6* gene) and environmental factors (e.g., hypoxia)[6,7].

Besides the congenital scoliosis, scoliosis is one of the common complications of neuromuscular diseases, which can be caused by progressive muscle weakness of the paravertebral muscles[8]. It usually appears in the late stage of the disease and is associated with other clinical associated manifestations[8]. Among the neuromuscular diseases, collagen VI-related myopathy is one of the neuromuscular diseases that can lead to scoliosis including Ullrich congenital muscular dystrophy, Bethlem myopathy and various related diseases, caused by mutations in *collagen type VI alpha 1 chain (COL6A1)*, *COL6A2*, and *COL6A3* genes[9]. In a physiological situation, collagen VI (COL6) is widely found in the extracellular matrix and is important for the structure and function of skeletal muscle[10]. As a result, mutation of this gene causes significant damage to muscles and leads to weakness, hypotonia, laxity of distal joints, contractures of proximal joints, and even scoliosis in some severe cases[9].

Here, we report a patient diagnosed with congenital scoliosis initially. Without limb weakness, hypotonia, laxity of distal joints, and contractures of proximal joints, she does not have any typical manifestations. After the spinal surgery, the patient was discharged, and her Cobb angle dropped from 103.4° to 52.9°. To determine the cause, intraoperative paravertebral muscle biopsy revealed muscular atrophy with myogenic lesions. Gene testing showed three mutations in *COL6A1* and *COL6A2*. Immunohistochemistry staining for collagen VI displayed part of the muscle fiber muscle membrane that stained to be shallow and widened, with a few portions of muscle fiber staining to be discontinuous. Eventually, the patient was diagnosed with a new type of collagen VI-related myopathy confined to the paraspinal muscle.

## CASE PRESENTATION

### Chief complaints

A 28-year-old female presented with scoliosis for 28 years and was admitted to the hospital from March 2019 to August 2019.

### History of present illness

The patient was a 28-year-old female who presented with scoliosis for 28 years from birth. An attempt was made to install the external fixation with braces to the patient, but the effect was poor. In recent 4 years, the patient suffered from lower back pain progressively.

### History of past illness

The patient had a free previous medical history.

### Personal and family history

The patient denied any personal history of alcohol and cigarette consumption. Her family history has nothing notable.

### Physical examination

Physical examination showed that the patient did not have obvious weakness, hypotonia, laxity, or contractures of the extremity joint. The mobility of wrist joints, metacarpophalangeal joints, distal interphalangeal joints and metatarsophalangeal joints was normal, and there was no abnormal skin mass. With the exception of superficial sensation abnormality tested by a safety pin, at the proximal lateral front of the left thigh, the rest parts sensation is normal, and the muscle strength of the limbs was classified as grade 5/5[11]. The physiological reflex of the patient was normal, except for the disappearance of abdominal wall reflex, and the pathological signs were negative. Sitting and standing time was not limited, but the patient's walking ability is affected and can only walk 1000 m due to low back pain.

Her menstrual history was generally normal. The menstruation started from age 14. Its period was 5-7 d. Its cycle was 25-28 d. The last menstruation was January 24, 2019. The color was normal without blood clots and without dysmenorrhea.

The patient denied congenital scoliosis teratogenic factors such as hypoxia during pregnancy, vitamin A deficiency, diabetes, and preeclampsia. Family history of scoliosis was denied. Adam's forward bend test was positive, suggesting that the patient had a high likelihood of scoliosis[12].

### Laboratory examinations

Her laboratory examinations have nothing notable.

### Imaging examinations

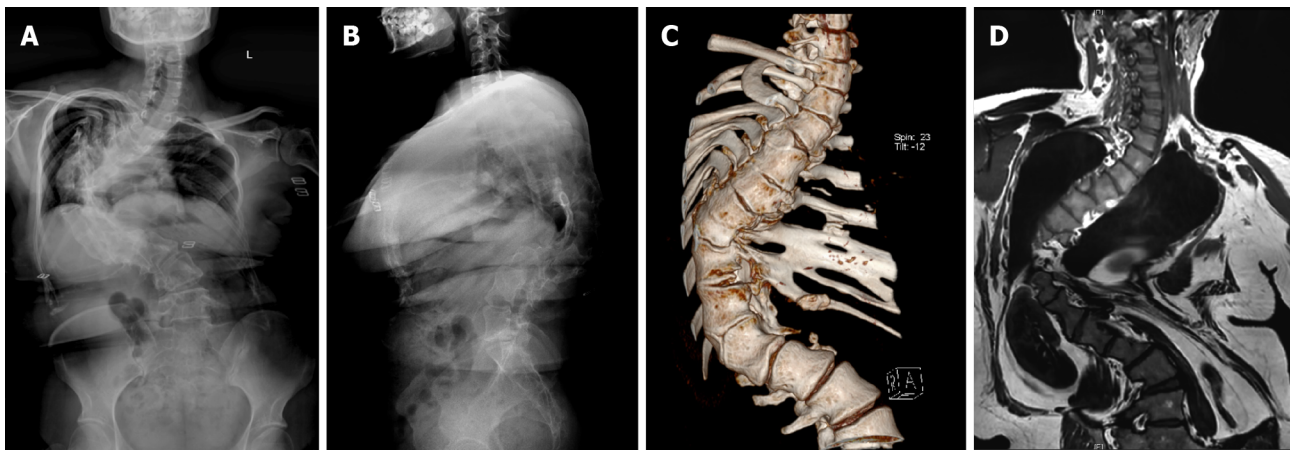
X-ray, computed tomography (CT), and magnetic resonance imaging (MRI) of the whole spine (Figure 1) showed that the Cobb angle was 103.4°, and the T4-6, T8, T12-L1 vertebral body was deformed. At T12-T1, the vertebral bodies fused, the intervertebral space disappeared, and the posterior margin kyphosis. The T11 vertebral body was hemivertebra. There was no obvious herniation of intervertebral disc and no obvious narrowing of vertebral canal. Based on the history, symptoms and signs, the patient was initially diagnosed as mixed congenital scoliosis initially.

### Further diagnostic work-up

The patient and her family members provided informed consent to publish case reports, photographs and carry intraoperative muscle biopsies and 'whole exome sequencing (Slim version)' at KangSo Medical Laboratory in Beijing, which was approved by the institutional review board of the authors' affiliated institutions.

**Histological examination:** Biopsy from the multifidus of the concave and convex sides of the thoracic vertebra was performed. The patients and their families were informed that data from the case would be submitted for publication, and gave their consent.

The process of muscle biopsy was performed as described by Meier *et al*[13]. The biopsy tissue was covered with a semi-moist saline gauze pad and transferred immediately. After gently drying, a small portion of the tissue was separated for electron microscopy, and the other fresh tissue was embedded with tragacanth gum. Then the samples were placed in liquid nitrogen-precooled isopentane for 20 s and then stored in a -20° freezer. 5 µm thick tissue slices were cut in a low temperature



**Figure 1 Preoperative imaging examination.** A: X-ray of the scoliosis - AP view; B: X-ray of the scoliosis - Perfil view; C: Computed tomography three-dimensional reconstruction of the patient; D: Magnetic resonance imaging of the patient's entire spine.

slicer for hematoxylin and eosin staining, special staining: nicotinamide adenine dinucleotide + hydrogen (NADH), Modified Gomori trichrome (MGT) and immunohistochemistry staining: dystrophin-1, dystrophin-2, dystrophin-3, myosin, major histocompatibility complex I, cluster of differentiation 4 (CD4), CD8, CD163, and CD20.

Optical microscope showed that the main part of the biopsy was skeletal muscle tissue, within a few light to medium volume reduced muscle fibers, muscle fiber gap widening, visible minority nuclear ingress, occasional crack, fibrous connective tissue hyperplasia, and a bunch of severe contraction inside muscle fibers, but no obvious degeneration and necrosis were observed. Numerous tendon-like structures formed by dense connective tissue can be seen around the muscle tract (Figure 2).

MGT staining showed a slight increase in reddening particles in some muscle fibers; NADH-tetrazolium reductase staining showed balanced distribution of the two types of fibers, and a small amount of muscle fibers were rotated. Immunohistochemical staining with dystrophin-1 antibody showed positive staining of muscle sarcolemma; dystrophin-2 and -3 antibody staining showed uneven staining of a few muscle sarcolemma (Figure 3). CD4- and CD8-positive cells and other inflammatory cells were occasionally infiltrated around the small vessels (Figure 4).

Electron microscopy showed that most of the muscle fibers were corrugated and the arrangement of extracellular matrix was slightly disordered. Individual myofibers and myoplasm coagulation with focal muscle mass formation. The inner myofibrils of some muscle fibers were arranged neatly, but the a-band structure was unclear (Figure 5).

Biopsy results suggested that neurogenic muscular atrophy with myogenic damage, congenital muscular dystrophy, and another myopathy were more likely (Table 1)[14]. To further investigate the reason for this presentation, patients agreed to have their genes tested for a clear diagnosis.

**Gene testing:** Gene testing resulted in the identification of four heterozygous variants of significance (included in the genetic package for hereditary myopathy) (Figure 6): one *COL6A1* splicing variant (c.1612-10G>A), one *COL6A2* splicing variant (c.115+10G>T), and one *COL6A2* missense variant (c.2749G>A). Of those variants, *COL6A2* missense variant (c.2749G>A) is considered as a disease-causing mutation by 'Mutation Taster', deleterious by 'Sortig Intolerant from Tolerant' and probably-damaging by 'Polyphen2' (Table 2). Therefore, the patient was diagnosed with collagen VI-related myopathy with scoliosis alone from the gene.

**COL6 staining:** Based on the results of gene examination, we carried out *COL6* immunohistochemical staining of the patient (with scoliosis) (Figure 7A), which indicated the low expression of *COL6*, compared with another inflammatory myopathy patient matched by same age and sex (without scoliosis) (Figure 7B).

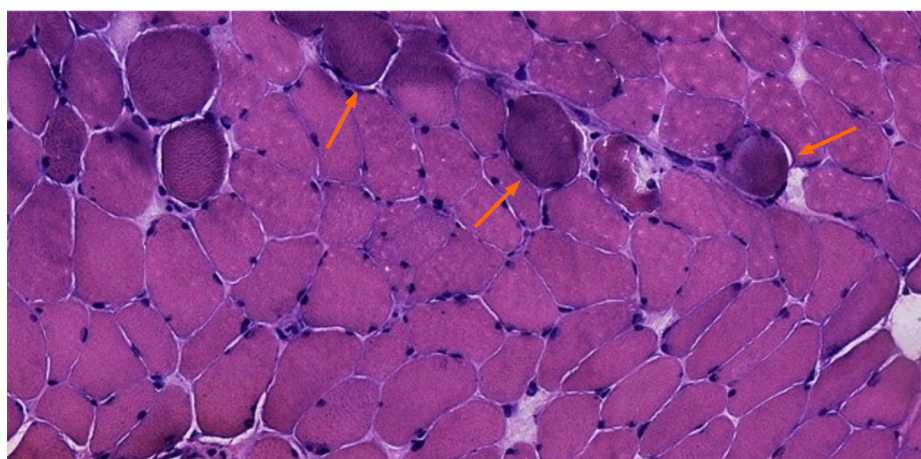
**Table 1 Immunohistochemical results of multifidus muscle**

| Antibody     | Result     | Antibody     | Result                                         |
|--------------|------------|--------------|------------------------------------------------|
| Dystrophin   | +          | Dystrophin-2 | A few sarcolemma staining in different shades  |
| Dystrophin-1 | +          | Dystrophin-3 | A few sarcolemma staining in different shades  |
| Dysferlin    | +          | HLA-DR       | Vascular endothelium and inflammatory cells +  |
| Lambda       | -          | MHC-1        | A few sarcolemma and myoplasm+                 |
| CD20         | Very few + | CD4          | A few vessel wall and interstitial substance + |
| CD163        | -          | CD8          | A few vessel wall and interstitial substance + |
|              |            | CD138        | Interstitial substance visible occasionally +  |

HLA-DR: Human leukocyte antigen DR; MHC-1: Major histocompatibility complex 1.

**Table 2 Genetic tests showed mutations in COL6A1 and COL6A2**

| Gene   | Locus     | Variant      | Consequence  | Location | Type         | Prediction      |
|--------|-----------|--------------|--------------|----------|--------------|-----------------|
| COL6A1 | NM_001848 | C.1612-10G>A | -            | Intron24 | Heterozygote | -               |
| COL6A2 | NM_001849 | C.115+10G>T  | -            | Intron2  | Heterozygote | -               |
| COL6A2 | NM_001849 | C.2749G>A    | P. Val917Met | Exon28   | Heterozygote | Disease causing |



**Figure 2 Multifidus biopsy results of hematoxylin and eosin staining.** A few muscles were slightly reduced in size, round in shape, bundles scattered, widened fiber gap and shrink nuclei, with occasional muscle fissure and connective tissue hyperplasia (orange arrows) (hematoxylin & eosin staining, × 40).

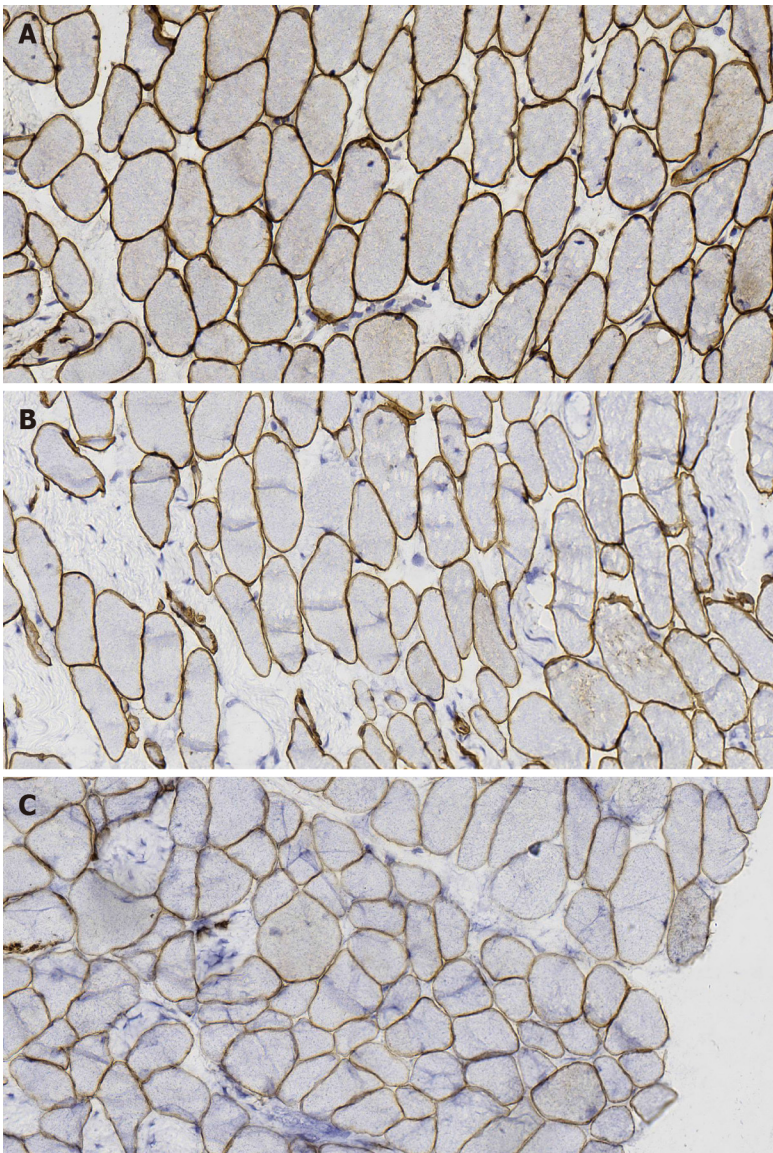
## FINAL DIAGNOSIS

Collagen VI-related myopathy diagnosis was finally affirmed at the gene and protein levels.

## TREATMENT

Because the patient had been born with scoliosis and had a large Cobb angle, we chose the preoperative 'halo-socket distraction' for the patient to enhance spinal flexibility and reduce the risk of nerve injury during surgery, the patient underwent 'halo-pelvic distraction' on March 16, 2019 with 13 cm distraction in 4 wk[15] (Figure 8). On May 10, 2019, the patient underwent an operation to place posterior T2-L4 pedicle screw internal fixation, T9 semi-vertebral resection, T10 total vertebral resection, intervertebral and interarticular bone grafting.





**Figure 3 Multifidus biopsy results of dystrophin.** A: Positive dystrophin 1 staining of sarcolemma (immunohistochemistry [IHC]: dystrophin-1 staining,  $\times 40$ ); B: Partial sarcolemma dystrophin 2 staining was uneven (IHC: dystrophin-2 staining,  $\times 40$ ); C: Partial muscle sarcolemma dystrophin 3 stained unevenly (IHC: dystrophin-3,  $\times 40$ ).

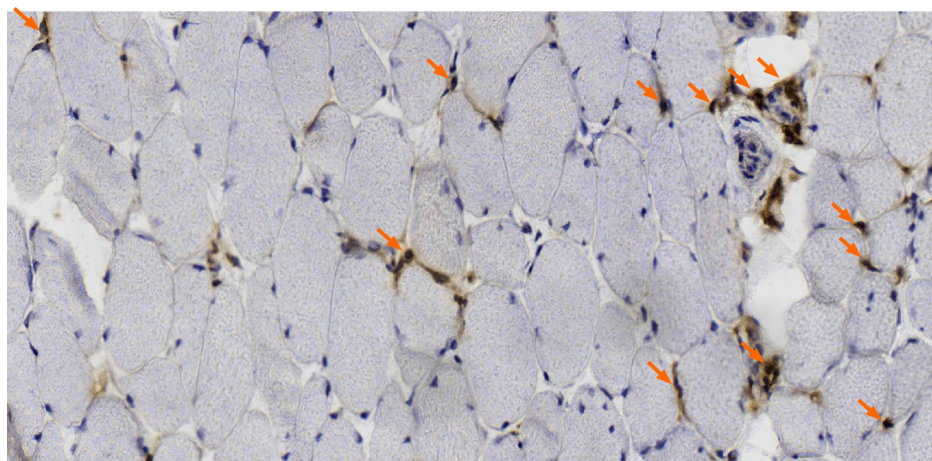
## OUTCOME AND FOLLOW-UP

The operation was successful and the patient discharged. X-ray, CT, and MRI of the whole spine ([Figure 9](#)) showed that the postoperative Cobb angle improvement to  $52.9^\circ$ .

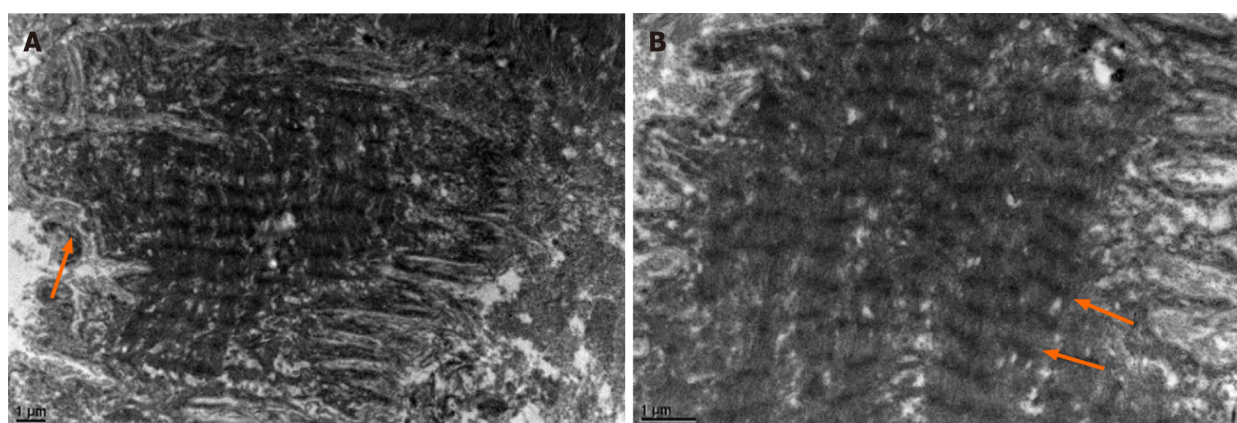
## DISCUSSION

In this case, with the exception of scoliosis and limited walking ability due to low back pain, the patient had no other pathological symptoms and signs in physical examination. CT and MRI imaging revealed complex vertebral development abnormalities such as hemivertebra, butterfly vertebra, and vertebral fusion. Therefore, the patient was diagnosed as congenital scoliosis. However, intraoperative muscle biopsy confirmed the presence of neurogenic muscular atrophy. Gene mutations of *COL6A1* and *COL6A2* were found. *COL6A* staining showed some muscle outside staining becomes shallow and broadening with a handful of muscle fibers staining discontinuous. Based on this the diagnosis of collagen VI-related myopathy was confirmed.

Collagen VI-related myopathy is caused by the mutation of *COL6A1*, *COL6A2* and *COL6A3*, which leads to the structural disorders of the  $\alpha 1$   $\alpha 2$  and  $\alpha 3$  subunits, then



**Figure 4 Multifidus biopsy results of cluster of differentiation 4.** A few cluster of differentiation 4 (CD4)-positive staining were seen in the wall and stroma of focal small blood vessels (orange arrows) (CD4 staining,  $\times 40$ ).



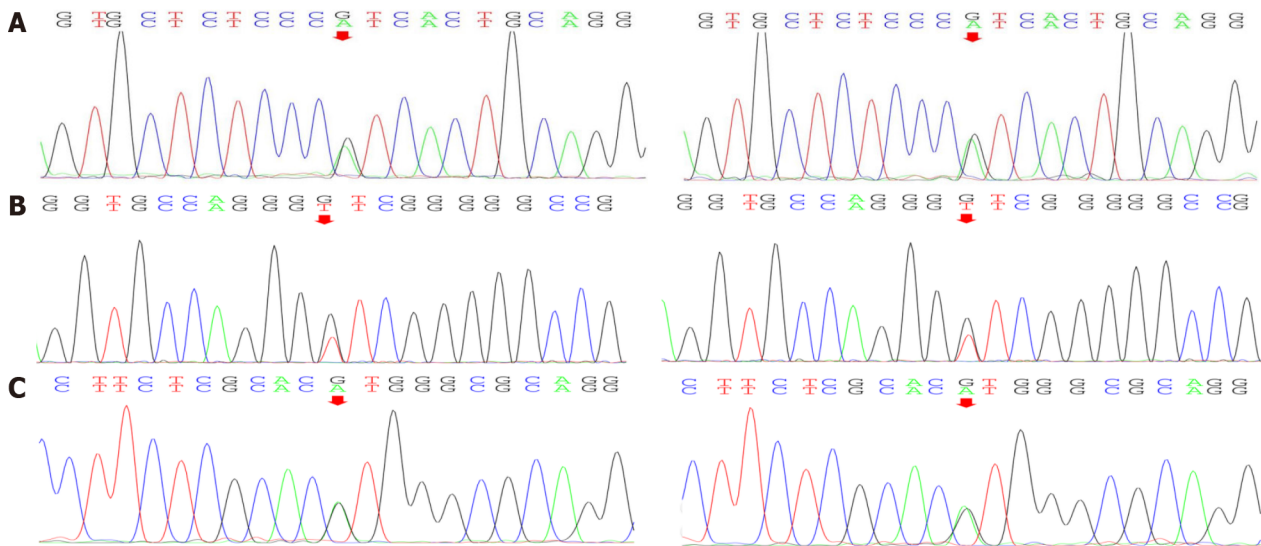
**Figure 5 Electron microscopy results.** A: Some sarcolemma is corrugated, and extracellular matrix arrangement is slightly disordered (orange arrows) ( $\times 6000$ ); B: The inner myofibril arrangement is orderly, but band A is unclear (orange arrows) ( $\times 15000$ ).

affects the formation of tetramers and the following assembly process, resulting in collagen VI fibers abnormality[16]. Since collagen VI fiber is closely related to the functional structure of skeletal muscle, mutation of this gene can cause muscle damage [10]. According to the different lesion site, the disease can be presented as a continuous spectrum of diseases from light to heavy[17]. The relatively mild Bethlem myopathy is characterized by muscular weakness, hypotonia, laxity of distal joints, contractures of proximal joints and bone deformity[18]. Besides the symptoms described above, relatively severe UCMD also manifests a ‘sandpaper’ like rash, congenital hip joint dislocation, torticollis, scoliosis, high arched palate, and prominent heel[19]. In addition, the disease also includes limb girdle muscular dystrophy, muscle sclerosis myopathy[17].

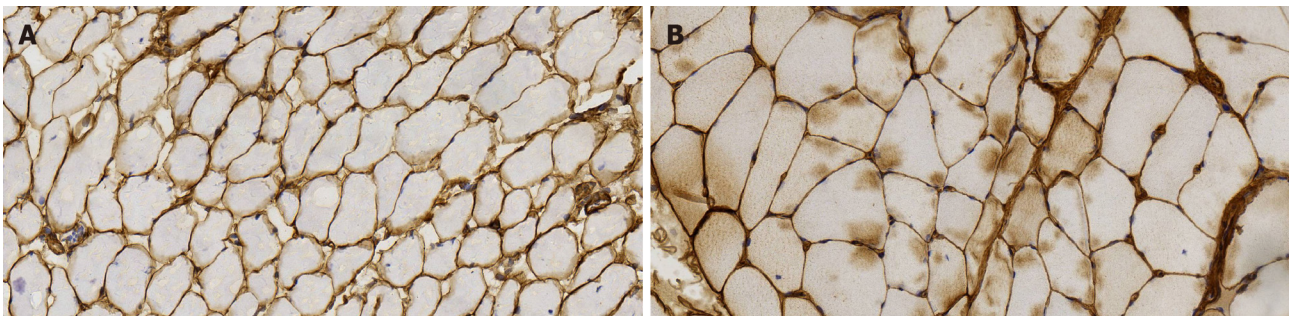
With scoliosis as the chief complaint, the patient was diagnosed with collagen VI-related myopathy by muscle biopsy and gene testing. Considering the diversity of clinical phenotypes of this disease[20], this patient might be a special subtype, called “Collagen VI-related myopathy with scoliosis alone”. In this subtype, scoliosis is the main clinical manifestation, accompanied with spinal congenital malformations such as hemivertebra and paraspinal muscle atrophy and lesions, without limb contracture or hyperextension. However, our report is only an isolated case, and further cases and studies are needed if this subtype is to be included in the disease spectrum.

According to the medical history, the patient has no exposure to teratogenic factors during pregnancy. Therefore, we assume that the congenital abnormality of the vertebral may relate to the gene mutation of *COL6A1* and *COL6A2*. So far, no major susceptibility genes for sporadic congenital spinal malformations in humans have been identified[21]. In zebrafish model, *COL8A1* gene mutation leads to abnormal spinal cord folding in embryos, which leads to spinal deformity, suggesting that extracellular





**Figure 6 Gene mutations in COL6A1, COL6A2.** A: Sequencing of collagen type VI alpha 1 chain (COL6A1) gene revealed splicing mutations; B: Sequencing of COL6A2 gene revealed missense mutations; C: Sequencing of COL6A2 gene revealed splicing mutations.

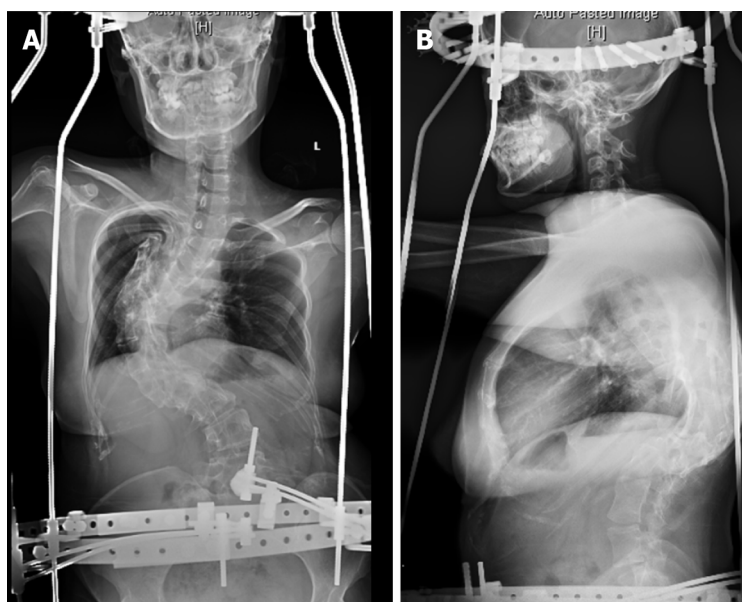


**Figure 7 Collagen VI immunohistochemistry.** A: This patient muscle membrane stained shallow, broaden and discontinuous (immunohistochemistry [IHC]: collagen type VI alpha (COL6A) staining,  $\times 40$ ); B: Another myopathy patient (non-scoliosis) matched by age and sex, staining continuous and dense (IHC: COL6A staining,  $\times 40$ ).

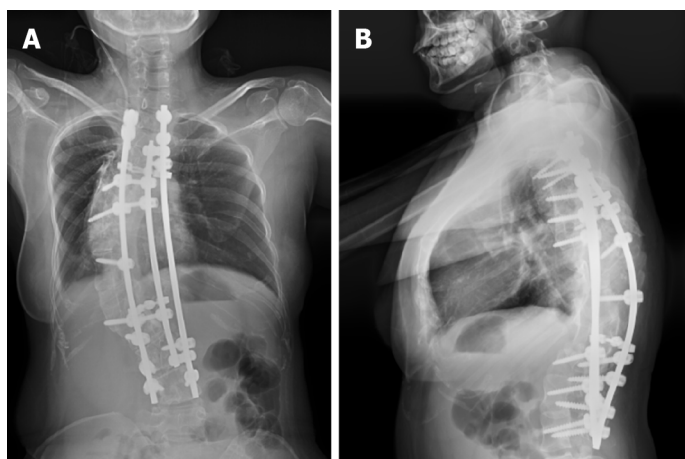
matrix is closely related to notochord normal differentiation[22]. In fact, as a part of the extracellular matrix, various types of collagen are involved in the formation of the spine[23-26]. During the segmentation period, COL15 is mainly expressed in the notochord, and its protein products are only deposited in the basement membrane around the notochord[23]. The expressions of COL2A1 and COL18A1 are also regulated during notochord formation[24]. Clinically, the gene mutation of COL2 can lead to epiphyseal dysplasia in the spine, leading to the occurrence of vertebral deformity [25]. In addition, studies have shown that both human fetal and traditional express COL6 on the surface of the cord, suggesting that COL6 may indeed exist in the human notochord[26]. Similar to the reports above, gene mutations of COL6A1 and COL6A2 in our patient may also affect the differentiation of notochord and the formation of vertebrae, resulting congenital abnormalities of patients. This deformity combined with the lesion of paravertebral muscle finally resulted in severe scoliosis of our patient.

In this case scoliosis was severe and muscle strength was normal, implying the patient lesion was confined to the paravertebral muscle and did not affect the muscles in other parts of body. This location-specific muscle injury confirmed the poor correlation between genotype and phenotype of collagen VI-related myopathy, similarly to the reports of Kim *et al*[27] and Okada *et al*[20]. Also, it suggests that there might be some undetected regulators in collagen VI-related myopathy[27], such as copy number variation, non-coding region variation and pathogenic variation of other genes[28]. These regulators may be missed in PCR-based gene detection, resulting in poor correlation between genotypes and phenotypes[29]. About 25% of the clinically diagnosed gene mutation negative collagen-related myopathy could find a highly repetitive intron mutation in COL6A1 by RNA sequencing[30]. In addition, the





**Figure 8 Imaging of halo-pelvic distraction.** A: Postoperative X-ray of the spine (AP view); B: Postoperative X-ray of the spine (Perfil view).



**Figure 9 Postoperative imaging examination.** A: Postoperative X-ray of the spine (AP view); B: Postoperative X-ray of the spine (Perfil view).

comparative genomic hybridization (CGH) technique based on oligonucleotide arrays is also used in studies to detect copy number variations collagen VI-related myopathy and is considered as a useful complementary diagnostic test[29]. This suggests that RNA sequencing and CGH techniques can be used for further research, which may reveal the intrinsic molecular regulatory mechanism of clinical manifestations of collagen VI-related myopathy in the future.

In addition, this case provides new options for the etiology of scoliosis. Scoliosis without myopathy-related signs might be homologous with collagen VI-related myopathy and could even be classified as a new subtype. Researchers believe that the pathogenesis of scoliosis is related to the deep paravertebral muscle activity[31], and find unbalanced bilateral muscle function in scoliosis[32]. Since then, a large number of researchers have found significant changes in the number, types and sizes of muscle fibers in paraspinal muscle biopsies of scoliosis patients, as well as the muscle lesions such as fat infiltration, atrophic necrosis, fibrosis of bundle membrane and endomysium, infiltration of inflammatory cells, and central core[33-35]. Due to the presence of muscle lesions, some scholars have speculated that the underlying cause of some scoliosis patients may be a special type of neuromuscular disease that affects the paraspinal muscle alone, leading to the decline of the traction and stability of muscle group on the spine, even the scoliosis[36]. Therefore, in addition to careful history collection and physical examination (especially muscle strength examination), the kinases spectrum, electromyography, and nuclear magnetic resonance examination of

muscles can provide hidden evidence for the etiology of scoliosis. In particular, such tests might classify some patients which has diagnosed as congenital scoliosis initially, into a special subtype of myopathy-related disease, leading changes of the treatment plan and prognosis, even providing a new direction for scoliosis etiology.

## CONCLUSION

In this study, we present a report of collagen VI-related myopathy with scoliosis alone. Caused by heterozygous mutation in *COL6A1* and *COL6A2*, the lesion of this subtype is limited to the paravertebral muscle and the development of vertebral body, suggesting patient might be one of the unidentified subtypes of collagen VI-related myopathy with scoliosis alone. On the other side, for patients with scoliosis, the underlying pathogenesis might be a special type of myopathy or neuromuscular disease, which can be further studied by paraspinal muscle MRI, myopathy-related kinases, electromyography and paraspinal muscle biopsy, providing an important entry point for scoliosis etiology.

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